

**Emotional processing and communication in people
with Huntington's disease: a mixed methods inquiry**

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Declaration

I declare that this thesis is my own work, and has not been submitted in substantially the same form for the award of a higher degree at this institution or elsewhere.

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Date: _____

*The mind is its own place, and in itself
Can make a Heaven of Hell, a Hell of Heaven.*

Acknowledgments

So here we are, Lancaster. Four years later, and I am still getting surprised by how much it rains. However, I feel like I have lost count of the things that have changed in the meantime. I have seen more people come and go in this period of time than I probably did in my entire life, and such experiences do come at the cost of great changes. Luckily enough, I believe in my case the change has been for the better. It has been long, it has not been easy. But it has been a journey that has made me grow definitely more than I expected – and to some extent even more than I was ready to wish for. It has been a humbling experience that has taught me the importance of getting out of my comfort zone, and question what I thought I was, I knew, and was capable of. Despite the many difficulties, and probably more than any career prospects, if this PhD has been worth doing, it has been to make me become (hopefully) a better person.

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Rationale for the Alternative Format

The current thesis is presented in the Alternative Format. As outlined by Lancaster University's Manual of Academic Regulations and Procedures (MARF), this format allows the composition of a thesis incorporating four empirical papers suitable for publication (hereafter referred to as 'publishable papers', PPs), one of which can consist of a literature review in a publishable form. This choice was made in agreement with the candidate's supervisors and the director of the PhD programme within the Division of Health Research, and was based on the nature of the research presented in the thesis (i.e., systematic reviews and empirical studies), with the ultimate aim of maximising the dissemination of findings. As required by MARF regulations, each publishable paper specifies the proportion for which credit is due to the candidate for carrying out the research and preparing the publication.

Thesis abstract

Huntington's disease (HD) is a progressive neurodegenerative disorder caused by the inheritance of the mutation of a protein called Huntingtin. Its typical symptoms include motor impairments, cognitive deterioration, and significant psychological difficulties. All these impairments can have a significant effect on the communication of affected individuals, including nonverbal components such as emotional processing. However, the current literature on HD appears to be particularly characterised by a medical approach to the topic, with little evidence from studies adopting a psychological perspective.

Thus, the overarching aim of the current thesis was to investigate the impact of Huntington's disease on the emotional processing and communication of affected individuals from a health psychology perspective and with the adoption of a mixed-methods approach. After an initial scoping review of the literature, a qualitative study was conducted in the first phase of the research project, with the aim of exploring the perspectives on communication of people with symptomatic HD. In the second phase, two quantitative investigations were carried out, specifically addressing how HD affects emotional processing – in particular emotion regulation and recognition – in symptomatic and presymptomatic individuals.

The results showed that, although emotional processing and communication are affected by HD, the achievement of feelings of control, better

emotion regulation, effective medication regimes, and close interpersonal relationships can play a pivotal role in alleviating the burden of the disease. In addition, emotion regulation and emotional body language (EBL) recognition abilities were both impaired in symptomatic individuals, while evidence with presymptomatic people suggested a relative preservation of these skills. In both cases, no significant relationship was found between these abilities. However, the relationship between depressive symptoms and specific elements of emotion regulation such as emotional awareness should be further explored in presymptomatic participants, as it may play a potential precursory role in the development of emotion recognition impairments in fully symptomatic individuals. The implications of the findings for theory and practice are discussed, and possible directions for future research are provided.

Chapter 1

General Introduction

Huntington's Disease

Historical overview

The history of Huntington's disease (HD), one of the most debilitating and yet currently lesser-known neurodegenerative disorders, appears to be as convoluted as its clinical manifestation. In its earliest depictions, the disease used to be referred to as 'hereditary chorea', from the ancient Greek *χορεία* (*choreia*), literally 'dance'. This is a reference to its characteristic involuntary motor symptoms, which have been historically compared to dancing. Other ancient sources dating back to the Middle Ages refer to it as *Chorea Sancti Viti*, or St. Vitus's dance, although this term has been historically adopted for a broad range of neurological diseases and today is used to refer to Sydenham's chorea (Wexler, 2010). Despite evidence on clinical notes and reports that can be traced back to at least the seventeenth century (Finn, 1970), the recognition of Huntington's disease as a specific clinical entity is only occurred when the American physician George Huntington (1850–1916) published the first comprehensive description of its signs and symptoms in 1872 (Huntington, 1872). Medical interest in the condition was not novel within the Huntington's family: George's grandfather, Dr Abel Huntington, moved to Eastern Long Island at the end of eighteenth century, where he found a significant number of families affected by the disease, which became

the main focus of his clinical work. The same applied for George's father, a physician himself, who was born and raised in Long Island and took on his parent's legacy (Stevenson, 1934). George's first encounter with people affected by Huntington's disease was at the age of eight, when he accompanied his father on one of his clinical rounds; reportedly, the impact of such a condition on his young mind was so strong that he decided to make it the main focus of his clinical education and, eventually, his first contribution to the medical literature (Huntington, 1910). Being raised in close and constant contact with affected individuals –as his father and grandfather before him – George had the unique opportunity to observe the development and evolution of the disease across several individuals and generations. Ultimately he provided a description which was recognised to be among the most accurate, brief and graphic in the whole history of medicine until that point (Osler, 1908). An example of his clearness can be found in the following line, where he delineated the three main features of the disease according to his experience: "There are three marked peculiarities in this disease: 1. Its hereditary nature. 2. A tendency to insanity and suicide. 3. Its manifesting itself as a grave disease only in adult life" (Huntington, 1872, p. 320). Even though he seemed to ignore the existence of a juvenile onset (i.e., before age 20), which is rare but possible (Kremer, 2002), the hereditary nature of the disease and its main psychological and cognitive difficulties appeared to be very clear in his mind. In particular, it is worth noting the sharpness of the description of the genetic transmission of the disease, implicitly outlining an autosomal-dominant mechanism almost 30 years before Hugo de Vries, Carl Correns and Erich von Tschermak made Mendel's laws known worldwide in 1900.

Humbly, George Huntington never omitted mentioning the contribution of his father and grandfather in his success: “as in old Greece the pupil sat at the feet of his teacher, so your essayist sat at the feet of these two, and whatever of honour, whatever of praise, whatever of scientific worth there is, is due much more to them, than to him to whom has come this unsought, unlooked for honour” (Stevenson, 1934, p. 62). Therefore the eponym ‘Huntington’s chorea’, which became very popular among authors after the seminal publication of 1872 (see Figure 1 for an illustration of the original front page) and was later acquired even in more informal registers, can be considered as one created by a whole family rather than a single man. Since approximately the 1970s, however, the term ‘disease’ has started to be preferred to highlight the fact that the motor impairments are not the only feature of the condition, as well as to avoid any possible stigma attached to it (Novak & Tabrizi, 2005; Wexler, 2010).

Aetiology

Despite its clear hereditary aetiology and notwithstanding intense research activity across the whole twentieth century, the specific genetic cause of Huntington’s disease was not identified until 1993, i.e. more than 120 years after the first scientific establishment of the condition. This occurred when the mutation of a protein (from then on named ‘Huntingtin’, or HTT) was recognised to be responsible for an anomalous expansion of CAG trinucleotide (cytosine-adenin-guanin) repeats on the short arm of chromosome 4 (Huntington’s Disease Collaborative Research Group, 1993). The protein is normally present in an individual’s biological make-up, regardless of sex or ethnicity, and it usually expresses a regular number of CAG repeats up to 28; when a mutation occurs,

however, the number of repeats increase, eventually leading to substantial damage to the subcortical regions of the brain called the basal ganglia. Especially affected is the corpus striatum (composed by the caudate nucleus and the putamen), which is involved in the many behavioural, cognitive, and motor tasks that are impaired in Huntington's disease.

As inferred by George Huntington, the mutation is hereditary and the transmission mechanism is autosomal-dominant, meaning that every affected individual has a 50% probability of transmitting it to their children regardless of the condition of the other parent. In most cases the disease is fully penetrant, i.e. all the individuals with the mutant gene will develop the disease at a certain time in their life. More specifically, the probability of developing the disease varies according to the number of CAG repeats on the allele: up to 35 repeats are not associated with the disease, 41 or more repeats are associated with full penetrance (and therefore the individual will surely develop the disease), while a number of repeats between 36 and 40 is associated with a 'grey zone' of incomplete penetrance, in which the individual may or may not develop the disease during his/her lifetime (Walker, 2007). This 'grey zone' is thought to account for 5 to 10% of new cases which are not explained by family history, often due to instable replication on the fathers' side that increases the number of repeats from 28 to 36 or more, thus reaching incomplete or complete penetrance (Semaka, Collins, & Hayden, 2010).

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ORIGINAL DEPARTMENT.

Communications.

ON CHOREA.

By GEORGE HUNTINGTON, M. D.,
Of Pomeroy, Ohio.

Essay read before the Meigs and Mason Academy of Medicine at Middleport, Ohio, February 15, 1872

Chorea is essentially a disease of the nervous system. The name "chorea" is given to the disease on account of the *dancing* propensities of those who are affected by it, and it is a very appropriate designation. The disease, as it is commonly seen, is by no means a dangerous or serious affection, however distressing it may be to the one suffering from it, or to his friends. Its most marked and characteristic feature is a clonic spasm affecting the voluntary muscles. There is no loss of sense or of volition attending these contractions, as there is in epilepsy; the will is there, but its power to perform is deficient, the desired movements are after a manner performed, but there seems to exist some hidden power, something that is playing tricks, as it were, upon the will, and in a measure thwarting and perverting its designs; and after the will has ceased to exert its power in any given direction, taking things into its own hands, and keeping the poor victim in a continual jigger as long as he remains awake, generally, though not always, granting a respite during sleep. The disease commonly begins by slight twitchings in the muscles of the face, which gradually increase in violence and variety. The eyelids are kept winking, the brows are corrugated, and then elevated, the nose is screwed first to the one side and then to the other, and the mouth is drawn in various directions, giving the patient the most ludicrous appearance imaginable.

The upper extremities may be the first affected, or both simultaneously. All the voluntary muscles are liable to be affected, those of the face rarely being exempted.

If the patient attempt to protrude the tongue it is accomplished with a great deal of difficulty and uncertainty. The hands are kept rolling—first the palms upward, and then the backs. The shoulders are shrugged, and the feet and legs kept in perpetual motion; the toes are turned in, and then everted; one foot is thrown across the other, and then suddenly withdrawn, and, in short, every conceivable attitude and expression is assumed, and so varied and irregular are the motions gone through with, that a complete description of them would be impossible. Sometimes the muscles of the lower extremities are not affected, and I believe they never are *alone* involved. In cases of death from chorea, all the muscles of the body seem to have been affected, and the time required for recovery and degree of success in treatment seem to depend greatly upon the amount of muscular involvement. ROMBERG refers to two cases in which the muscles of *respiration* were affected.

The disease is generally confined to childhood, being most frequent between the ages of eight and fourteen years, and occurring oftener in girls than in boys. DUFOSSE and RUFZ refer to 429 cases; 130 occurring in boys and 299 in girls. WATSON mentions a collection of 1,029 cases, of whom 733 were *females*, giving a proportion of nearly 5 to 2. Dr. WATSON also remarks upon the disease being most frequent among children of *dark* complexion, while the two authorities just alluded to, DUFOSSE and RUFZ, give as their opinion that it is most frequent in children of *light* hair. In every case visiting the clinics

Figure 1: Front page of the George Huntington's 1872 research article featuring the first depiction of HD.

Epidemiology

Huntington's disease is a rare condition, showing a prevalence of 5-10 persons per 100,000 in the Caucasian population (Roos, 2010). As far as the UK is specifically concerned, the reported prevalence in 2010 was 12.3 per 100,000 people (Evans et al., 2013). A recent systematic review and meta-analysis of its worldwide incidence and prevalence found an overall prevalence of 2.71 per 100,000 (Pringsheim et al., 2012). More specifically, the overall prevalence was 5.70 per 100,000 in Europe, North American, and Australia, while in Asia it showed a much lower presence, with an overall prevalence of 0.40 per 100,000. Incidence was reported to be 0.38 per 100,000 per year worldwide, again with lower values for Asian countries. This significant geographical difference between Asia and the remaining continents is thought to be due to differences in individuals' haplotypes, i.e. Asian people generally have shorter CAG tracts that may prevent them from developing the abnormal number of repeats typical of the disease.

Genetic testing and diagnosis

Since the discovery of the protein responsible for the disease in 1993, genetic testing is available for individuals with a family history, allowing them to know if they carry the mutant gene even decades before the onset of symptoms. Nonetheless, the number of at-risk people who decide to undertake the test ranges between 3% and 24% (Harper, Lim, & Craufurd, 2000; Laccone et al., 1999), with the lowest uptake observed in Germany, Austria and Switzerland (3-4%) and the highest observed in the UK (18%), Canada (18%) and the Netherlands (24%; Tibben, 2007). With particular regard to the UK, a recent investigation of data

between 1993 and 2012 showed an overall uptake of predictive testing ranging between 15% and 26% (Quarrell & Rosser, 2014).

A positive genetic test, however, has no clinical value *per se* and does not constitute a diagnosis. The full diagnosis of Huntington's disease is based on clinical symptoms and signs along with a familiar history (i.e., proof of an affected parent); more specifically, the current necessary clinical criteria for diagnosis are still motor symptoms, while the presence of cognitive and psychological changes is not necessary (Roos, 2010). People with a positive test for Huntington's but without motor symptoms are usually referred to as being 'gene carriers' or having 'presymptomatic HD' or 'premanifest HD' (Dumas, van den Bogaard, Middelkoop, & Roos, 2013), while individuals with family history of the disease who have not been tested are usually defined as 'at-risk' (Chisholm, Flavin, Paulsen, & Ready, 2013).

Onset and life expectancy

The estimate range of age of onset is 40-50 years. However, juvenile onset (i.e. before the age of 20 years and as early as 2 years) can also occur, as well as a very late onset (up to 80 years; Kremer, 2002). Juvenile Huntington's disease (JHD) is usually characterised by more prominent slowing of movements, lack of muscular tone as well as psychological difficulties such as agitations and irritability (Roos, 2010). The specific age of onset appears to be determined by both genetic and environmental factors, with the number of CAG repeats accounting for approximately the 60% its variation (Walker, 2007). No effective treatment or cure has been found so far and the mean life expectancy after the diagnosis is typically 20 years (Folstein, 1989).

Motor symptoms and signs

The characteristic motor symptom of Huntington's disease is represented by involuntary movements (chorea) that initially start from the distal extremities of the body (fingers and toes) and gradually involve more proximal muscles, including the head and the face. The choreatic movements are not characterised by any specific pattern. They are always present when the affected individual is awake, though tend to disappear during sleep. As the disease progresses, hypokinesia (decreased body movements), akinesia (difficulty in starting movements), bradykinesia (slower movements), dystonia (muscle contractions causing twisting movements and uncomfortable postures) and dysphagia (difficulty in swallowing) appear. As a consequence, walking becomes unstable and normal daily activities such as eating, drinking and talking become progressively arduous (Roos, 2010).

Cognitive symptoms

Huntington's disease is responsible for many cognitive impairments, ultimately leading to dementia. A recent review including studies between 1993 and 2011 found that in manifest Huntington's disease impairments can be expected in memory, psychomotor speed, executive functioning and, in later stages, language (Dumas et al., 2013). In presymptomatic individuals no difference is usually found in terms of linguistic and long-term memory function when compared to control groups, while an early deterioration of executive processes and working memory is sometimes observed (Dumas et al., 2012; You et al., 2014).

Psychological difficulties and well-being

Huntington's disease is also associated with a number of psychological difficulties, of which the most frequent are depression, euphoric or dysphoric mood, lack of inhibition, irritability and aggressiveness, as well as anxiety, agitation, and apathy; more rarely delusions, compulsions, and hallucinations can be observed (Beglinger & Paulsen, 2008; Caletti et al., 2014; Robins Wahlin, 2007; Santacruz, Fenoll, & Munoz, 2014; Vaccarino et al., 2011; van Duijn, Kingma, & van der Mast, 2007; Walker, 2007). Moreover, an increased risk of suicide has often been found in presymptomatic people (Hubers et al., 2012). In particular, along with cognitive impairments, depression has been reported to be a highly significant determinant of the quality of life of affected individuals (Banaszkiewicz et al., 2012), even more than motor symptoms themselves. Another psychological challenging aspect of HD is represented by the impact of predictive testing. With regard to this, as shown by the aforementioned generally low uptake, most at-risk individuals prefer to remain uncertain about their gene status and undergo the test only when they are facing important life choices, such as getting married or having children. On the other side, the studies on those who have undergone the test and received a positive result show a consistent variability: some people show average levels of psychological distress in the long term (e.g., after 1 year) and start to appreciate life and relationships more (Broadstock, Michie, & Marteau, 2000; Duisterhof & Trijsburg, 2001), while others regret being tested and tend to avoid any further investment in education, jobs, family, or long term life plans in general (Hagberg, Bui, & Winnberg, 2011). In some cases a positive test result has been associated with suicidal ideation (Wahlin, 2007). Other psychological difficulties

reported by people with Huntington's disease include genetic discrimination (i.e., being treated unfairly or differently by others due to genetic differences, as opposed to physical ones; Bombard et al., 2011; Williams & Erwin, 2010), family issues due to living with affected relatives and communicating the family history with the disease, especially when young children are involved (Forrest Keenan, van Teijlingen, McKee, Miedzybrodzka, & Simpson, 2009). With regard to this, a framework that has been often adopted to explain psychological difficulties in chronic illnesses is the self-regulation model (SRM; Leventhal, Leventhal, & Brissette, 2003), which identifies patients' perceptions of their chronic condition as a fundamental element that plays a pivotal role in informing the development of coping strategies – which in turn deeply affect the successful operationalisation of psychological well-being (for a review, see Hagger et al., 2017). In the specific case of HD, a more psychological distress is associated with a strong illness identity (i.e., a belief that a high number of symptoms are perceived as attributable to the disease), as well as by a perception of HD as a chronic condition responsible for many negative consequences (Arran, Craufurd, & Simpson, 2014; Helder et al., 2002; Helder & Kaptein, 2002; Kaptein et al., 2006; Kaptein et al., 2007; Klitzman, 2009).

However, despite the research mentioned above, psychological accounts of patients' lived experience are not as evident as those from a more medical, neurological perspective and very little is currently known about how affected people live their lives, manage their emotions, relate and communicate to others, and ultimately make sense of the condition (Auduly, Packer, & Versnel, 2014).

Chapter 2

Overview and Justification of Methods

Multimethod and Mixed Methods Research (MMMR)

General overview

The combination of different types of research methods within the same set of studies has traditionally met with lukewarm approval. Indeed, although the adoption of multiple methods can be traced back to the beginning of the 20th century – especially in the case of social science research – a clear case for its empirical usefulness only occurred in far more recent times (Mark, 2015). One of the possible reasons for this may lie in the variety of definitions that has characterised this approach over the past few decades, and that has led to the development of terms such as ‘mixed methods’ or ‘multimethod’ which, although conceptually very close, can have substantial semantic differences. According to the *Oxford Handbook of Mixed and Multimethod Research Inquiry* (Hesse-Biber & Johnson, 2015), the word ‘multimethod’ refers to the general combination of two or more different methods, whether qualitative or quantitative, in the context of a single study or a series of scientific works. Therefore, it can broadly delineate either the adoption of quantitative and qualitative methods together, or the combination of a series of different quantitative (or qualitative) methods. This can also be expressed with the variation ‘multiple methods’. On the other hand, ‘mixed methods’ is a narrower term that refers to the specific combination of both

quantitative *and* qualitative in the same research context. In order to express this distinction, the collective term ‘multimethod and mixed methods research’ (MMMR) can be adopted (Hesse-Biber & Johnson, 2015).

Philosophical underpinnings of MMMR: the third paradigm

Historically, the dispute between quantitative and qualitative methodologies has been a philosophical one, as their fundamental differences stem from what is considered to be real (ontology), and how reality can be known (epistemology). This has driven the development of two main opposing scientific paradigms. On one side positivism (and later post-positivism), characterised by the belief that reality is unique, objective and knowable through bias-free measures (realism). On the other, constructivism (also known as interpretivism), which entails the belief that there is no unique reality, but rather a number of subjective constructs depending on contexts that can only be interpreted (relativism). As a reflection of these assumptions, positivism has traditionally adopted quantitative methodologies, and constructivism has been characterised by qualitative approaches (Bishop, 2015). While the first half of the 20th century was largely dominated by positivistic science, in the 1970s constructivism and other forms of relativistic paradigms started to challenge the realist approach (Alise & Teddlie, 2010). This quickly escalated into what came to be known as the ‘paradigm wars’ (Mertens, 2014; Oakley, 1999; Tashakkori & Teddlie, 1998), which were driven by the assumption that the paradigms were epistemologically incoherent with one another and therefore could not be combined (the ‘incompatibility thesis’; Howe, 1988). The paradigmatic divide was so prominent that graduate students in those years (and likely still today, in some cases)

reportedly felt as if they were asked to pledge allegiance to one 'faction' or the other in order to work in academia (Johnson & Onwuegbuzie, 2004).

It was not until two decades ago that, in reaction to the paradigm wars, a number of theorists counter-proposed the 'compatibility thesis', leading the way to the first affirmation of MMR (Alise & Teddlie, 2010) under the idea that functional knowledge should be prioritised over philosophical disputes – a position known as 'pragmatism'. The core assumption of pragmatism is that methodological choices do not need to commit to any single ontology and epistemology, but can, rather, be driven by the practical implications of the questions that are being examined and the results that are being sought (Dures, Rumsey, Morris, & Gleeson, 2010; Johnson & Onwuegbuzie, 2004). This enables researchers to draw from a 'toolkit' of both quantitative and qualitative methods and base their rationale on the needs and purposes of the research rather than their philosophical stance (Maxwell & Mittapalli, 2010). As a consequence, due to this shift of focus on purposes, pragmatism has often been regarded as the 'third paradigm' in the philosophical scene (Onwuegbuzie & Leech, 2005), as well as the 'philosophical partner' for MMR (Johnson & Onwuegbuzie, 2004), especially in real world research contexts such as health and social sciences.

MMMR in health research and psychology

Since its initial inception as a scientific discipline, psychology has been largely dominated by quantitative methodological approaches (Alise & Teddlie, 2010). Indeed, its early characterisation as a scientific discipline was clearly reflected in its adoption of a positivist approach. Since then, the strength of the positivist influence has been so significant that, even with the advent of

constructivism, mainstream psychology has always maintained a quantitative stance, with very little (if any) interest in the integration of qualitative components with quantitative research (Frost & Shaw, 2015). This dominion remained unchallenged until at least the 1980s, when the abovementioned paradigm was triggered a renewed interest for qualitative methods, especially in the field of social psychology (Oakley, 1999). Nevertheless, any early attempts at adopting MMMR in psychology were frustrated by the prevailing incompatibility thesis, which led to a fierce entrenchment on paradigmatic and methodological positions (whether post-positivist/quantitative or constructivist/qualitative). This was argued to put at risk the very purpose of scientific inquiry, that is, the advancement of knowledge – a phenomenon that was later defined as ‘methodolatry’ (Curt, 1994).

However, the history of psychology has also been characterised by a rich tradition of methodological experimentation. As a consequence, the psychological panorama has seen a substantial proliferation of methods and techniques, which appears to have reached its peak in the last few decades thanks to the advent of cognitive neuroscience. In addition, as mentioned before, the interest in qualitative methods that characterised the paradigm wars period has continued to favour the compatibility thesis and, now that ‘a fragile peace’ (Bryman, 2006) has been achieved, the time seems ripe for a renewed attempt at exploring the possibilities of MMMR in psychology and related disciplines.

In fact, this appears to be particularly the case in health research (Morgan, 1998). Driven by a more pragmatic logic of inquiry that is focused on the impact of research on practice rather than maintaining a precise methodological stance

(Maxcy, 2003), applied health research has largely embraced the adoption of MMR (Frost & Shaw, 2015). This occurred to the extent of seeing it become one of the dominating methodological approaches in the field (Ó' Catháin, Murphy, & Nicholl, 2010), within the context of what has been defined as a 'quiet revolution' (Halcomb & Andrew, 2009; Tashakkori & Teddlie, 2003). With particular regard to the UK, the recommendation of including qualitative designs along with quantitative studies in health research was initially proposed by the Medical Research Council (MRC, 2000), and has been more recently endorsed by the National Institute for Health and Care Excellence (NICE) in England and Wales (Kelly et al., 2009), as well as the Scottish Intercollegiate Guidelines Network (SIGN; 2008). As a subfield of health research, a similar tendency has been consequently observed in the field of health psychology, in which the adoption of MMR can promote the widening of perspectives in research on clinical practice and outcomes (Mcleod, 2011), as well as the exploration of subjective dimensions and experiences that may otherwise go overlooked (Bryman, 2007). In confirmation of this, in 2005 the *Journal of Counseling Psychology* published a special issue entitled *A Time and Place for Qualitative and Mixed Methods in Counseling Psychology Research* meant as "a call to counseling researchers to increase their dialogue over philosophy of science, research paradigms, and methodological diversity" (Haverkamp, Ponterotto, & Morrow, 2005, p. 123). Similarly, in 2015 *Health Psychology* published a special issue on qualitative research with the aim of "promoting greater uptake and development of qualitative research methods in the field" (Gough & Deatrck, 2015, p. 1). Concurrently, in recent years the psychological literature in the UK has seen a substantial increase of the acknowledgement of the usefulness and value of MMR

(e.g., Bishop, 2015; Bryman, 2007; Cornish & Gillespie, 2009; Dures et al., 2010; Mason, 2006; Todd, Nerlich, & McKeown, 2004), driven by the recognition that “the range of ways in which methods can be mixed, be they with others of the same paradigm or across paradigms, allows the complexity of humanness to be better represented” (Frost & Shaw, 2015, p. 389).

General purposes and rationales for MMMR

Due to its focus research on functional knowledge and practical implications, since the 1980s several models have been proposed to characterise the breadth of aims and purposes that can justify the adoption of MMMR. Despite some variability in terms, a number of common concepts can be recognised in all of them. These include the purpose of triangulating information onto a single answer (e.g., Rossman & Wilson, 1985), widening the range of answers (e.g., Mark & Shotland, 1987), providing different levels of analysis, or enhancing interpretability of the results. From this perspective, the most influential model is the classification proposed by Greene and colleagues (1989), in which they identified five possible purposes for MMMR: a) *triangulation*, that is, searching for convergence and corroboration across methods; b) *complementarity*, that is, enriching the elaboration and clarification of findings; c) *development*, that is, adopting a method to develop or inform the adoption of another one; d) *initiation*, that is, searching for contradictions and generating new research perspectives; e) *expansion*, that is, utilising different methods for different components of a research design. Apart from research purposes, the same author also developed a list of other four of dimensions along which MMMR designs can differ (Greene, 2007). These include the sequence of studies (*timing*), the hierarchical importance

of methods (*status*), whether one study informs any others (*dependence*), and the adopted measures and methods (*methods themselves*). The particular advantage of Greene's model lies in the potential for the researcher to draw a methodological framework based on its dimensions. Thus, this model was adopted for the development of the methodological framework of this thesis.

Philosophical and Methodological Foundations of the Thesis

Rationale for MMR adoption

As previously noted, MMR is a methodological approach that is considered to fit exceptionally well with the needs and purposes of the field of health research and health psychology (Bryman, 2007; Cornish & Gillespie, 2009; Frost & Shaw, 2015; Halcomb & Andrew, 2009; Mason, 2006; Maxcy, 2003; Morgan, 1998; Ó'Catháin et al., 2010; Todd et al., 2004), especially in the UK (Kelly et al., 2009; Medical Research Council, 2000; Scottish Intercollegiate Guideline Network, 2008). In particular, MMR has been reported to benefit considerably the investigation of the complex phenomena related to the interface between health psychology and rare or chronic illness (Bishop, 2015; Dures et al., 2010), in a fashion that cannot be achieved by the adoption of quantitative methods alone (Crossley, 2000). This consideration, combined with the philosophical position of the researcher (see next paragraph), has led to the decision to adopt MMR for the research topic of the current thesis, that is, the exploration of the psychological impact of a rare and chronic condition such as Huntington's disease on patients' emotional processing and communication.

Philosophical stance

As mentioned before, the ideal philosophical stance for the adoption of MMR is often regarded to be a pragmatic one (Johnson & Onwuegbuzie, 2004). Indeed, since its first introduction, pragmatism has been very popular among MMR researchers (Biesta, 2010; Cornish & Gillespie, 2009; Johnson & Gray, 2010; Maxcy, 2003; Morgan, 2014). Nevertheless, when considered from an ontological and epistemological point of view, the pragmatic approach has considerable limitations and shortcomings. In particular, the extent to which it rejects the role of philosophical assumptions has been the subject of growing criticism over the years (Johnson & Onwuegbuzie, 2004), as it may lead to a misleading underestimation of the influence of researchers' ontological and epistemological positions on the chosen methodology, as well as on research questions, purposes and, ultimately, findings (Henry, Julnes, & Mark, 1998; Maxwell & Mittapalli, 2010).

One of the aims of this work was to carry out research that accounts for the heterogeneous and dynamic nature of complex psychological phenomena linked to Huntington's disease *as well as* being explicit regarding the potential influence of the philosophical assumptions of the researcher. As a consequence, instead of a pragmatic approach, a critical realist stance was adopted for the current thesis. In the philosophy of science, critical realism emerged out of the work of Roy Bhaskar (1978). Its distinctive characteristic is that, while it retains a realist ontology (i.e., there is a unique reality that we can investigate), it embraces a relativist epistemology – that is, our knowledge of reality cannot be objective or certain, as it will always depend on our perspectives, values, and contexts. Thus,

on one hand critical realism is akin to pragmatism as it agrees with the justification of methods based on the aims and purposes of research. However, on the other hand, it differs from it by retaining a focus on philosophical assumptions, without having the unrealistic expectation for researchers to dismiss what, in fact, is the subjective lens through which they observe the world and make sense of their findings (Greene & Hall, 2010). From this viewpoint, critical realism not only recognises the importance of physical and behavioural entities, but also considers people's meanings and perspectives as equally real and separate phenomena with explanatory significance that can deepen and expand the interpretation of findings (Sayer, 2000). In terms of methodology, this offers compatibility with both quantitative and qualitative methods. Indeed, more than just representing an effective tool for bridging the two (Mark, Henry, & Julnes, 2000), critical realism also has "important implications for both approaches, ones that push both qualitative and quantitative researchers to examine more closely some issues that they typically dismiss or ignore" (Maxwell & Mittapalli, 2010, p.160).

Therefore, critical realism was considered to be consistent with the research planned for this thesis, as well as the researcher's philosophical assumptions and values. More specifically, a critical realist stance appeared to be particularly indicated for the exploration and integration of qualitative data on the perspectives of people with Huntington's disease (PP2) with cognitive quantitative findings on their psychological difficulties (PP3 and PP4).

Methodological framework

In accordance with Greene's model (Greene, 2007; Greene et al., 1989), a methodological framework was drawn for the current thesis. In particular, MMR

was adopted for two main *purposes*: allowing the initial exploratory studies to inform the methods of the later ones (*development*), and enriching the elaboration and understanding of the psychological influence of HD by diversifying the type of data (*complementarity*). The *methods* included both quantitative and qualitative studies. For this reason, in accordance with the aforementioned distinction between multimethod and mixed methods research (Hesse-Biber & Johnson, 2015), the term mixed methods was chosen to refer to the approach adopted in the present work. As a direct consequence of the development purpose, the *timing* of the methods was sequential and their relationship was characterised by linear *dependence*. Last, despite more quantitative studies, the *status* of the methods was overall equal in standing. Table 1 provides a schematic representation of the methodological framework of the thesis.

Table 1

Methodological framework of the thesis (based on Greene's model, 2007; 1989)

Dimension	Thesis
Purpose	Development and complementarity
Timing	Sequential
Dependence	Linear
Status	Equal
Methods	Quantitative and qualitative

Overview of the mixed methods design

As far as the technicalities and practicalities of mixing methods are concerned, the solution adopted by most authors has been to develop typologies of mixed methods designs. Consequently, the last few decades have seen a

considerable proliferation of models illustrating several typologies and examples (Bishop, 2015). Among these, one of the most recent and influential is the model developed by Johnson and Onwuegbuzie (2004). Inspired by a number of previous works (Creswell, 1994; Morgan, 1998; Morse, 1991; Patton & Quinn, 1990; Tashakkori & Teddlie, 2003), it is based on two primary decisions: the emphasis assigned to different paradigms (intended as qualitative or quantitative components, therefore equivalent to Greene's *status*) and the time order of the studies (equivalent to Greene's *timing*). As a result, the model provides a decision matrix in which nine typologies of mixed methods designs are identified: concurrent equal mixed methods (QUALITATIVE + QUANTITATIVE), concurrent unequal mixed methods (QUALITATIVE + quantitative; quantitative + QUALITATIVE), sequential equal mixed methods (QUALITATIVE → QUANTITATIVE; QUANTITATIVE → QUALITATIVE), and sequential unequal mixed methods (QUALITATIVE → quantitative; qualitative → QUANTITATIVE; QUANTITATIVE → qualitative; quantitative → QUALITATIVE). Table 2 provides an adapted schematic overview of Johnson and Onwuegbuzie's matrix (2004). Due to its compatibility with Greene's methodological framework (Greene, 2007; Greene et al., 1989) and its focus on paradigm emphasis and timing, Johnson and Onwuegbuzie's typology was adopted for the current thesis. In particular, a *sequential equal mixed methods design* (QUAL → QUAN) was selected.

Table 2

Mixed methods design matrix (adapted from Johnson and Onwuegbuzie, 2004)

		Time order (<i>Timing</i>)	
		Concurrent	Sequential
Paradigm emphasis (<i>Status</i>)	Equal	QUAL + QUAN	QUAL → QUAN QUAN → QUAL
			QUAL → quan qual → QUAN
	Unequal	QUAL + quan	
		QUAN + qual	QUAN → qual quan → QUAL

Note. qual = qualitative; quan = quantitative; + = concurrent; → = sequential; upper case = high emphasis; lower case = low emphasis; bold = design typology adopted in the current thesis.

This decision was made for a number of reasons. First of all, as outlined in the thesis methodological framework (Table 1), one of the purposes for the adoption of MMR was to enrich and diversify the type of data. As a consequence, an equal emphasis was given to both qualitative and quantitative methods. Secondly, the choice of developing a sequential design was motivated by the second purpose of the framework, i.e. allowing the initial studies to inform the following ones. This was also compatible with the recommendations of the *ESRC postgraduate training and development guidelines* for PhD students (ESRC, 2009), as well as the guidance for trainee health psychologists (Health Professions Council, 2010), since it allows the development of both quantitative and qualitative research skills. Last, a sequential design better lends itself to a number of separate publications (Bishop, 2015), which benefit the purposes of an alternative format thesis.

Synopsis of studies and methods

As mentioned before, the overarching aim for the current thesis was to investigate the impact of Huntington's disease on the communication of affected individuals. This research question, along with the critical realist position of the researcher, informed the adoption of a mixed methods design, as well as the choice of methods and the sequence of studies¹.

First, a scoping review of the literature on communication in people affected by Huntington's disease (PP1) was conducted, with the aim of identifying the elements of communication that had been investigated, as well as capturing and describing the variety of research perspectives adopted. The scoping approach was chosen as it allowed the use of a systematic and replicable search strategy without specifying a narrowly defined research question, as usually required by systematic reviews (Arksey & O'Malley, 2005; Mays, Roberts, & Popay, 2001) and for which the paucity of research in more specific and narrower areas on communication in HD was problematic. Both quantitative and qualitative studies were included. The results outlined a number of elements of communication that had been particularly neglected in the empirical literature. These included the subjective experiences and perspectives on communication of people affected by HD, as well as some components of emotional processing, such as emotion recognition through non-facial cues (e.g., body language).

¹ For the purpose of this chapter, only a brief overview of methods is provided. The full rationale and justification for each method can be found in the respective publishable papers (PPs).

Informed by the literature review, a study aimed at exploring the perspectives on communication of people with HD (PP2) was then developed. A qualitative design was adopted, based on semi-structured interviews and analysed through thematic analysis (TA). The choice of TA was driven by its recognised usefulness within the field of psychology and communication disorders (Braun & Clarke, 2006), as well as for its flexibility towards both deductive (i.e., theory-driven) and inductive (i.e., data-driven) analyses of the themes (Harper & Thompson, 2011). Apart from the paucity of qualitative literature on the topic that was identified by the review, the decision of starting with a qualitative investigation was motivated by the idea that exploring patients' perspectives would facilitate insights into aspects of communication that may potentially be overlooked in quantitative studies. This was also believed to allow the lines of enquiry to be kept open for a later, more focused quantitative exploration. Indeed, the results provided useful insights around the subjective experience of the communicative issues experienced by people with HD. Among these, one of the most relevant was how HD impaired communication by threatening participants' emotional life and stability, and how one possible solution was represented by improving emotion regulation.

Thus, based on the results of the qualitative study, and co-informed by the observation that no quantitative literature on the topic had been retrieved by the initial scoping review, a quantitative investigation of the impact of HD on emotion regulation was planned. More specifically, two similarly designed yet distinct studies with age-matched controls were developed: an online survey with people with presymptomatic HD (PP3) and a face-to-face between-subject study with

individuals affected by symptomatic HD (PP4). This differentiation was motivated by the characteristics of the research samples. An online survey was considered more appropriate for presymptomatic people, as it allowed for the enrolment of high numbers of participants in different countries thanks to dissemination via email and social media. This was not the case for symptomatic participants, due to the difficulties in using computers and digital devices in general that are often caused by the motor symptoms of the disease. On the other hand, a face-to-face between subject design was considered more suitable for symptomatic individuals, as the presence of the researcher allowed for the arrangement of any required facilitations (e.g., presenting the stimuli on a computer). As a consequence, due to HD's low prevalence, this translated into a much smaller sample compared to the online survey. However, the number of symptomatic participants enrolled (13) was consistent with the average sample size of similar research identified by the initial scoping review. The overarching aim of both studies was to investigate how HD affects emotion regulation, and how that relates to emotion recognition. The latter component was included due to the alterations and impairments of emotion recognition that are known to be present in both presymptomatic and symptomatic individuals (Novak & Tabrizi, 2005). In addition, as one of the most neglected topics identified by the scoping review was emotion recognition via non-facial cues, a test of emotion recognition based on body language stimuli was included in PP4.

The results of PP3 showed that, in presymptomatic people, emotion regulation and emotion recognition are not significantly impaired, and no significant relationship between the two constructs was observed. However, a

specific impairment of the emotional awareness component was found, which appeared to be enhanced by the co-occurrence of depressive symptoms, even at a subclinical level. In addition, this impairment may represent a precursor of emotion the recognition impairment for negative emotions that is typically observed in individuals affected by fully symptomatic HD. On the other hand, the results of PP4 showed that emotion regulation and emotional body language recognition are significantly impaired in people with symptomatic HD. In addition, emotional body language recognition impairments were significantly related to both facial emotion recognition deficits (positively) and the stage of the disease (negatively). However, as in PP3, no significant correlation was observed between emotion regulation and recognition performances.

A visual representation of the overall sequence and logic of the studies is provided by the flowchart depicted in Figure 1. In terms of research design and methods, a similar approach to the current thesis has been adopted before in health psychology for the investigation of the psychological impact of chronic and rare illness. More specifically, it was adopted with people affected by epidermolysis bullosa (EB), where it proved to be a suitable solution for the research questions and purposes (Dures, Morris, Gleeson, & Rumsey, 2010; Dures et al., 2010; Dures, Morris, Gleeson, & Rumsey, 2011). Last, a ‘theoretical integration’ (Moran-Ellis et al., 2006) of the findings of all the studies is provided in the General Discussion chapter.

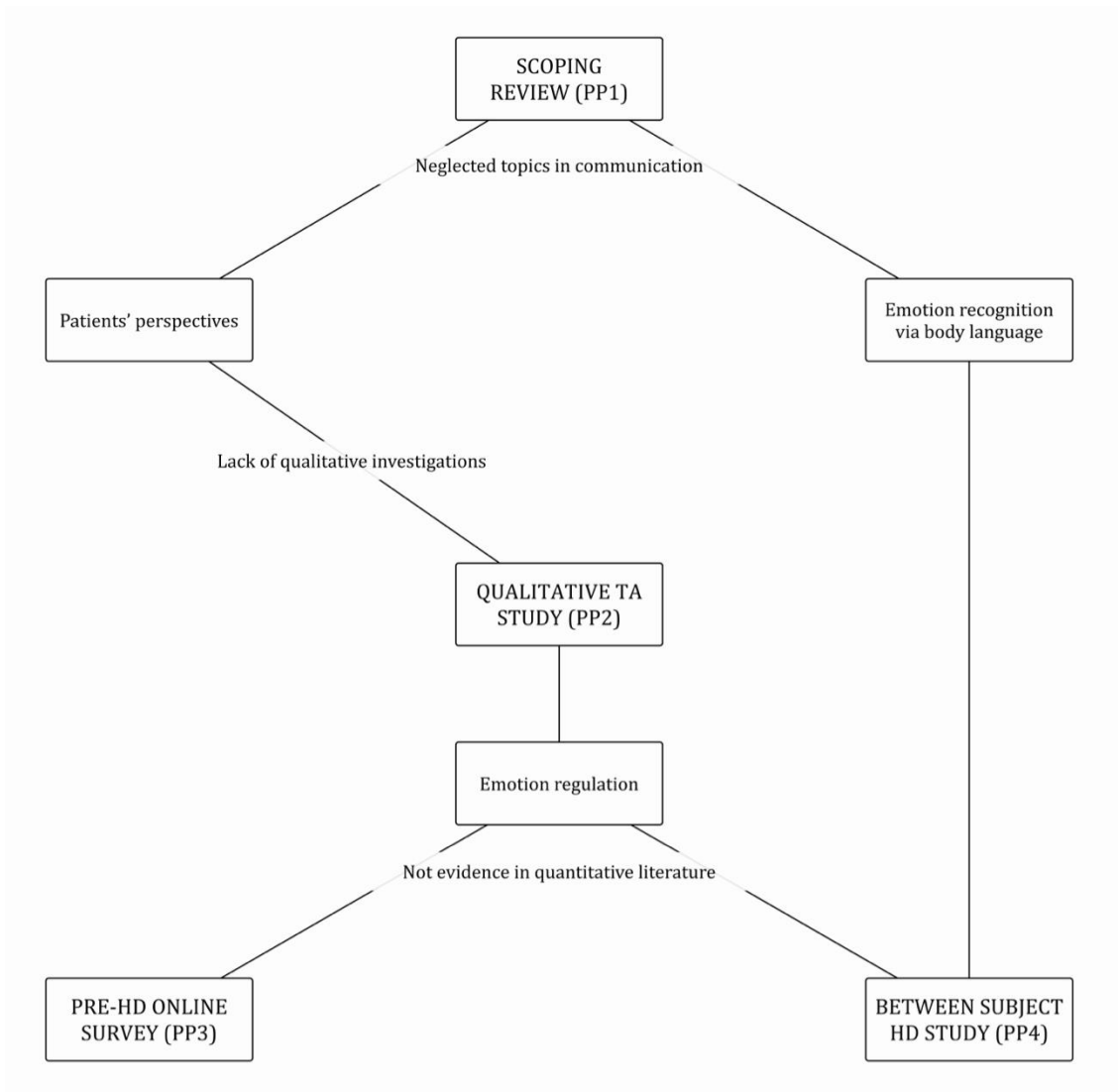


Figure 2: Thesis studies flowchart.

may include infection, medication changes or trauma. In this case, the patient was initially diagnosed with status epilepticus. Continuous EEG monitoring and clinical examination showed that the patient's stereotypic movements were, in fact, due to status dystonicus. This case illustrates the complexity of diagnosis and management of patients with JHD, and can serve as evidence that patients with JHD may be best managed at tertiary care centers where more comprehensive resources, such as continuous EEG and movement disorders consultation, are available.

Poster 35

Attitudes Toward Clinical Trials and Genetic Disclosure in Autosomal Dominant Alzheimer Disease: Implications for Huntington Disease.

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Background: Treatments for autosomal dominant neurodegenerative conditions can be tested in patients who have no symptoms but will develop disease. The design of prevention clinical trials in autosomal dominant conditions, however, may be complicated by low rates of genetic testing among persons at risk to inherit disease-causing mutations.

Methods: We anonymously surveyed participants in the Dominantly Inherited Alzheimer's Network to better understand their attitudes toward genetic testing and clinical trials. Participants were asked whether they knew their genetic status, did not know their genetic status but wished to, or did not know and preferred not to know their genetic status. Participants not interested in genetic testing answered whether they would change their mind if learning that they carried a mutation gave them the opportunity to participate in a trial. Regardless of preference toward genetic testing, participants answered questions related to interest in trials and how placebo ratios and open-label extension studies altered that interest.

Results: Eighty participants completed the survey; 40% knew their genetic status, 15% did not know but wished to learn their status, and 45% did not wish to learn their genetic status. Seventy-two percent of participants who did not wish to learn their genetic status reported that they would change their mind in the setting of a clinical trial. Eighty percent of all participants reported that they were interested in participating in a clinical trial. The proportion interested dropped when there

was a possibility of a placebo. The higher the chance of a placebo, the lower the interest. Nearly all participants (100% of those who knew or wished to know and 96% of those who did not know but would be willing to learn their genetic status in the setting of a clinical trial) would be interested in a trial with an open label extension.

Conclusions: The anonymous nature of the survey and the inability to determine whether differences exist between responders and nonresponders limit interpretation of these results. Nevertheless, the results support the conduct of trials to prevent autosomal dominant Alzheimer disease. Similar data are needed in Huntington disease and may be instructive toward designing feasible and appropriate prevention clinical trials.

Poster 36

Communication in Huntington Disease: An Empirical Review.

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Communication is a multifaceted discipline that includes language, emotion, speech, and proxemics, as well as social and environmental factors. Communication is particularly relevant in the process of adjusting to chronic illnesses such as Huntington disease (HD), with communicative patterns being significantly related to clinical outcomes. To our knowledge, no existing review has been carried out on the range of empirical studies on communication in HD, nor any clear understanding on which components of communication will benefit from further investigation. An empirical review was conducted to identify the elements of communication that have been investigated with people who had symptomatic HD, and the differing methodological approaches. The PubMed, PsycINFO, and Linguistics and Language Behavior Abstracts databases were searched systematically from January 1993 to January 2015, reference lists of included papers were hand-searched, and 49 studies were identified across four topic areas; communicative skills, emotion, language, and speech. The results illustrate that HD impairs language skills, recognition of negative emotions, and speech production when compared with both healthy participants and other neurodegenerative conditions. Preliminary evidence was also found for the significant impact of social and environmental factors on communicative abilities. Areas identified for future research include emotion expression and other nonverbal components of communication, as well as the effect of both impairments and social factors on the functional communicative capacity of people with symptomatic HD.

Poster 37

Healthcare Delivery in Huntington Disease: An Exploratory Global Survey.

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Communication in people with Huntington's disease: a scoping review

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Highlights

- We reviewed communication in people with symptomatic Huntington's disease (HD).
- 49 studies were included.
- 4 topic areas were identified: communicative skills, emotion, language and speech.
- HD primarily impairs language, negative emotions recognition and speech production.
- Patients' perspectives and emotional communication need further investigation.

Abstract

Objectives: Communication is a multifaceted ability that includes language, emotion, speech, and social and environmental factors. It is particularly relevant in the process of adjusting to chronic illnesses such as Huntington's disease, with communicative patterns being significantly related to clinical outcomes. This review aimed at identifying the elements of communication that have been investigated with people with symptomatic Huntington's disease, the breadth of research perspectives, and the differing methodological approaches.

Methods: A scoping review was conducted. Three databases - PubMed, PsycINFO and LLBA - were searched systematically from January 1993 to January 2015, using MeSH and Subject Terms as well as general keywords.

Results: Forty-nine eligible studies were identified across four topic areas; Communicative Skills, Emotion, Language, and Speech.

Discussion: Huntington's disease severely impairs language skills, recognition of negative emotions, and speech when compared to controls. Preliminary evidence was also found for the impact of social and environmental factors on communicative abilities. Directions identified for future research include more comprehensive investigations of patients' perspectives, emotion expression and other nonverbal components of communication, as well as the effect of both impairments and social factors on the functional communicative capacity of people with symptomatic Huntington's disease.

Keywords: Huntington's disease; communication; emotion recognition; language; scoping review.

Introduction

Huntington's disease (HD) is a neurodegenerative disorder caused by the mutation of a protein called Huntingtin, situated on the short arm of chromosome 4. Typical symptoms include motor impairments, cognitive deterioration and significant psychological difficulties (Novak & Tabrizi, 2005). The mutation is hereditary and the transmission mechanism is autosomal-dominant, meaning that every affected individual has a 50% probability of transmitting the mutation to their children, regardless of the condition of the other parent. In the vast majority of cases the disease is fully penetrant, i.e. all the individuals with the mutant gene will develop the disease at a certain time in their life.

HD is considered a rare illness, with a prevalence of approximately 5-10 persons per 100,000 in the Caucasian population (Roos, 2010). A recent systematic review and meta-analysis of its worldwide incidence and prevalence (Pringsheim et al., 2012) found an overall prevalence of 2.71 per 100,000. Genetic testing is available in most medically developed countries for individuals with a family history of the disease, allowing them to know if they carry the mutant gene even decades before the potential onset. People with positive testing for HD without motor symptoms are usually referred to as 'gene carriers' or people with 'pre-symptomatic HD' (Dumas, van den Bogaard, Middelkoop, & Roos, 2013), while individuals with family history of the disease who have not been tested are usually defined as 'at-risk' (Chisholm, Flavin, Paulsen, & Ready, 2013). The estimate range of age onset is 40 to 50 years. However, juvenile onset (before the age of 20 and as early as 2 years) can also occur. No cure has been found so far and the mean life expectancy after the diagnosis is typically 20 years (Folstein, 1989).

The characteristic motor symptom of HD is involuntary movements (chorea) that involve the limbs as well as the face. As the disease progresses, walking becomes unstable and normal daily activities such as eating, drinking and talking become progressively arduous (Roos, 2010). HD is also responsible for many cognitive impairments, which include problems with memory, psychomotor speed, executive functioning and language, and ultimately lead to dementia (Dumas et al., 2013). A number of psychological difficulties can appear throughout the progression of the disease, of which the most frequent commonly reported are depression, euphoria (or dysphoria), lack of inhibition, increased irritability and aggressiveness, tendency to feel anxious, more agitated or apathetic; more rarely, delusions, compulsions, increased sexual drive and hallucinations can be observed (Walker, 2007).

Individual differences in ‘adjustment’ to chronic and disabling illnesses (understood here as the way people adapt “to maintain a positive view of the self and the world in the face of a health problem” (Sharpe & Curran, 2006), p1161) and the causes which may affect them, such as different coping strategies, gender and socio-cultural factors, have long been a focus of investigation in other neurodegenerative conditions (Leventhal, Leventhal, & Brissette, 2003). Among the factors affecting adjustment to chronic illness, a pivotal role is played by interpersonal communication (Stanton, Revenson, & Tennen, 2007), which is defined as the ability that “focuses on how people use messages to generate meanings within and across various contexts, cultures, channels, and media” (Korn, Morreale, & Boileau, 2000) (p44). Indeed communication⁴

⁴ Unless otherwise specified, the general term “communication” has been used in this review to refer to interpersonal communication, including any of the abovementioned aspects.

includes several aspects such as language, speech, and emotional processing, as well as social and environmental factors, and it has proved to be related to clinical outcomes in people with chronic illness in different contexts, such as familial relationships (Rosland, Heisler, & Piette, 2012) or patient-physician interaction (Ong et al., 1995). Moreover, in certain chronic conditions, supportive communication often represents the only possible form of intervention in terms of palliative care (Bury & Wood, 1979).

In this perspective, a positive test for the Huntingtin gene and the development of symptoms leading to a diagnosis of HD certainly necessitate a series of adaptations on behalf of the individual – the successful navigation of which will affect longer term psychological adjustment. However, for people with HD little research concerning this issue is apparent, especially with specific regard to communication, where the literature on people with HD appears to be largely dominated by studies on ‘objectively-observed’ medical and cognitive impairments, i.e. those assessed from a clinician’s perspective, including how they are affected by medical intervention and their neuroanatomical correlates (Henry, Crawford, & Phillips, 2005; Ruzs et al., 2014; Teichmann et al., 2008). In addition, to our knowledge, no existing review has ever been conducted on the range of empirical studies on interpersonal communication in HD, and a clearer characterisation of the current corpus of evidence is warranted. As a consequence, the purpose of this review was to synthesise how communication is empirically conceptualised in research on people affected by HD, as well as provide an initial depiction of the epistemological and empirical heterogeneity of the research in this field, in order to underline the current gaps and inform further investigations.

Methods

Aim and Design

The primary aim of this review was to identify the elements of communication that have been investigated in empirical research with people with symptomatic HD by running a systematic search of relevant databases; the secondary aim was to capture and describe the variety of research perspectives and methods adopted. To fulfil these aims a scoping review was adopted. This allowed the use of a systematic and replicable search strategy without specifying a narrowly defined research question, as is usually required by systematic reviews (Arksey & O'Malley, 2005; Mays, Roberts, & Popay, 2001). The lack of sufficient research in more specific, narrower areas prevented this approach and instead the wider remit of a "form of knowledge synthesis that addresses an exploratory research question aimed at mapping key concepts, types of evidence, and gaps in research related to a defined area or field by systematically searching, selecting and synthesizing existing knowledge" (Colquhoun et al., 2014) (p. 1293-1294) was considered appropriate.

Search Strategy and Inclusion Criteria

A systematic search was performed up to January 31st 2015 using the following databases: PubMed, PsycINFO, and Linguistics and Language Behaviour Abstracts (LLBA). The date range searched started from 1 January 1993 since no genetically confirmed diagnosis of HD could be made before then. This is a common choice in recent reviews involving HD (Dumas et al., 2013; Franciosi, Shim, Lau, Hayden, & Leavitt, 2013; Henley et al., 2012). MeSH and Subject Terms were used for PubMed and PsycINFO respectively. Since no Subject Term for HD was available in LLBA, the general keyword "Huntington's"

was used to run a search through all the database fields. See Table 1 for details of the research terms used with each database.

In order to be included in this review, studies had to be related to any of the different features of interpersonal communication in people with HD. Interpersonal communication was defined as the ability that “focuses on how people use messages to generate meanings within and across various contexts, cultures, channels, and media” (Korn et al., 2000). Therefore, studies referring to types of communication other than interpersonal (e.g., communication between cells) or focused on communication in people without HD (e.g., from a clinician’s or carer’s perspective) were excluded. Given the scoping nature of the review, a variety of methods were included – ranging from cross-sectional and longitudinal designs to qualitative investigations. The participants of the studies had to have a diagnosis of HD confirmed by genetic testing. Since the focus of the review was on symptomatic HD patients, studies focusing only on presymptomatic participants were not included.

Selections of Studies

The initial searches across PubMed, PsycINFO and LLBA identified 478 citations. See Table 2 for details of the citations identified by each of the single databases. 250 citations were subsequently excluded as duplicates or studies published before 1993. The title and the abstract of each of the remaining 228 citations were examined to assess the relevancy of the studies according to the aforementioned inclusion criteria. When the title or the abstract were not sufficient to assess relevancy, the full text of the citations was obtained. The reference lists of the studies included in the final selection were also hand searched, to confirm that no relevant publications were overlooked. Ultimately, 65

studies were identified as relevant for the purposes of this review. See Figure 1 for a flow diagram of the study selection process.

Results

Categorization

Out of the 65 studies that met the inclusion criteria for this review, four main general categories were identified that reflected the main research topics: Communicative Skills (5), Emotion (19), Language (35), and Speech (6). These categories were formulated on the basis of the most common categorisations currently adopted for describing the broad verbal and nonverbal components of communication (Knapp & Daly, 2011). Subcategories were also identified for every topic (see Table 2). The results for each category are discussed below. For full details on all the studies identified by this review please refer to Table 3.

[Figure1 near here]

Communicative Skills

The main adopted methods in the Communicative Skills category included qualitative interviews and videotaped conversations, to investigate the perspective of people with HD on their communicative abilities. A number of social and environmental factors were found to play a relevant role in facilitating or complicating communication. More specifically, negative effects seem to be related to the speed of the conversation or

having fewer people to talk with, while communication appears to be positively affected by sensations of safety, having the opportunity to speak for a longer time and the perception of support and adjustment from the conversation partner (Hartelius, Jonsson, Rickeberg, & Laakso, 2010). Similar results (Power, Anderson, & Togher, 2011) were reported when applying the WHO International Classification of Functioning, Disability and Health (ICF) (WHO, 2001).

The adoption of Talking Mats™ (Murphy & Cameron, 2006) – an augmentation technique based on textured mats and sets of pictures showing different discussion topics – proved to significantly increase the effectiveness of communication in both dyadic (Ferm, Sahlin, Sundin, & Hartelius, 2010) and group interactions (Hallberg, Mellgren, Hartelius, & Ferm, 2013) with people affected by HD. Similar improvements were reported with the adoption of a therapeutic approach based on linguistic and cognitive supplementation strategies (Klasner & Yorkston, 2001).

Emotion

The adopted methods in the Emotion category included the assessment of emotional processing, general neuropsychological evaluations and neuroimaging techniques. A general impairment in emotion expression was found in people affected by HD when compared to controls, especially in the expression disgust, fear, and sadness (Trinkler, de Langavant, & Bachoud-Levi, 2013). This was also highly correlated to an impairment in emotion recognition. A selective deficit for disgust was reported for spontaneous and posed expressions (Hayes, Stevenson, & Coltheart, 2009). The ratings of subjective emotional experiences elicited through affective scenes showed that people with HD had a marginally higher tendency to rate them as more intense, along with a

positive bias for neutral scenes (Ille, Holl, et al., 2011); however, a similar study failed to report any differences with the control group (Ille, Schäfer, et al., 2011).

Labelling or rating tasks based on emotional facial expressions - such as the Ekman Pictures of Facial Affect (POFA) (Ekman, P., Friesen, 1976) - were adopted by the vast majority of the studies assessing emotion recognition, while other methods included self-report emotion ratings (Hayes, Stevenson, & Coltheart, 2007; Sprengelmeyer et al., 1996) and custom recognition tests based on olfactory and auditory tasks (Calder et al., 2010; Hayes et al., 2007; Rees et al., 2014; Robotham et al., 2011; Sprengelmeyer et al., 1996). The majority of the studies on emotion recognition (12/18) found a significant impairment involving the recognition of negative emotions (sadness, fear, anger and disgust). A selectively more severe impairment for the recognition of disgust was also reported in some instances (Hayes, Stevenson, & Coltheart, 2009; Hayes et al., 2007; Sprengelmeyer et al., 1996; Trinkler et al., 2013; Wang, Hoosain, Yang, Meng, & Wang, 2003). Other studies, however, did not find any selective impairment of disgust and observed in some cases that the impairment extended to positive emotions as well as neutral expressions (Labuschagne et al., 2013; Robotham et al., 2011; Snowden et al., 2008). Other findings reported a predominant impairment for negative emotions other than or along with disgust, such as fear (Hayes et al., 2009; Milders, Crawford, Lamb, & Simpson, 2003a; Robotham et al., 2011; Snowden et al., 2008) and anger (Calder et al., 2010; de Gelder et al., 2008; Henley et al., 2008; Robotham et al., 2011; Scharmüller et al., 2013). The neuroanatomical findings associated with such impairments included reduced activity in both subcortical and cortical regions in general (Dogan et al., 2014), and in particular specific involvements of the cuneus, precuneus, precentral gyrus and lingual

gyri (Scahill et al., 2013), insula, prefrontal cortex and hippocampus (Ille et al., 2011), as well as the cerebellum Scharmüller et al., 2013.

The impairment for negative emotions was also found to differ across both different modalities and different emotions (e.g., HD closer to controls' performance in recognizing anger through vocal stimuli; Rees et al., 2014). Moreover, when investigating emotion recognition through nonverbal stimuli based on body language, a predominant impairment with angry and emotionally neutral instrumental body postures (i.e., pouring water into a glass) was found (de Gelder et al., 2008).

Language

The adopted methods in the Language category ranged between general neuropsychological assessments of cognitive functioning, assessments of language alone, and conversation analysis approach, with both cross-sectional and longitudinal designs.

Comprehension appeared to be generally impaired in HD for complex discourses when compared to controls (Saldert et al., 2010), although no correlation was found with the progression of the disease, except for the comprehension of metaphors. Other observed impairments included comprehension of past tense verbs (Longworth, Keenan, Barker, Marslen-Wilson, & Tyler, 2005), comprehension of sentences (Sambin et al., 2012), phoneme discrimination (Teichmann, Darcy, Bachoud-Lévi, & Dupoux, 2009), rule application (Teichmann, Dupoux, Kouider, & Bachoud-Lévi, 2006), as well as word and rule learning (De Diego-Balaguer et al., 2008). When compared with people affected by Parkinson's disease, the comprehension of participants with HD was found to be qualitatively and quantitatively similar in terms of comprehension impairments (Murray & Stout, 1999).

General neuropsychological investigations (which included language subtests) were performed in many studies adopting broad batteries, such as the Repeatable Battery for the Assessment of Neuropsychological Status (RBANS; Randolph, 1998), as well as single function tools like the Boston Naming Test (BNT; Kaplan, Goodglass, & Weintraub, 1983), and phonemic and semantic fluency tasks. Semantic and phonemic fluency, naming, and comprehension were found to be generally impaired in cross-sectional studies (Arango-Lasprilla et al., 2006; Begeti et al., 2013; Duff, Beglinger, Theriault, Allison, & Paulsen, 2010). The impairment on verbal fluency was found to be predicted by structural alterations on the caudate nucleus and the putamen (Backman, Robins-Wahlin, Lundin, Ginovart, & Farde, 1997). The same impairment was also reported at baseline in longitudinal designs; however no significant change in language was found across the follow-ups (Beglinger et al., 2010; Lemiere, Decruyenaere, Evers-Kiebooms, Vandebussche, & Dom, 2004). Moreover, a study adopting event-related potentials (ERPs) found significantly longer latency times in HD when investigating language production (Munte et al., 1997).

In terms of language production, significant impairments were generally found for both phonemic and semantic fluency (Monsch et al., 1994; Rosser & Hodges, 1994; Troster et al., 1998; Chenery, Copland, & Murdoch, 2002; Lepron, Peran, Cardebat, & Demonet, 2009), sometimes with a predominant deficit for the semantic variant (i.e., generation of words from a category) (Barr & Brandt, 1996) or the phonemic one (i.e., generation of words from a letter) (Taylor, Salmon, Monsch, & Brugger, 2005). However, an improved performance was reported with the use of cued stimuli (Christopher Randolph, Braun, Goldberg, & Chase, 1993). The performance on verbal fluency was also found to be significantly correlated with abnormalities in cerebral blood flow (CBF)

velocity Deckel & Cohen, 2000), as well as lack of activation of the left inferior temporal gyrus (Lepron, Peran, Cardebat, & Demonet, 2009).

A number of studies found no overall significant impairment in HD in terms of lexical-semantic descriptive abilities of people with HD, except an increased number of grammar errors (Jensen, Chenery, & Copland, 2006; Murray & Lenz, 2001). However, another investigation did report a deficit of lexico-semantic abilities, along with an impairment on the interpretation of ambiguous meanings (Chenery, Copland, & Murdoch, 2002). Other observed impairments of language production included a significant deficit of application of grammatical rules (Teichmann et al., 2005, 2008), especially in relation to past tense rule use (Longworth, Keenan, Barker, Marslen-Wilson, & Tyler, 2005; Ullman et al., 1997), which appears to be related to specific striatal subregions (Teichmann et al., 2008).

Interestingly, a case study that reported the presence of anomia (difficulties retrieving names), reduced comprehension, and echolalia (automatic repetition of vocalizations) in a participant with HD also found that the latter impairment was actively used by the patient as a compensatory strategy to enhance conversational participation (Saldert & Hartelius, 2011).

Speech

The most adopted method in the Speech category was Acoustic Voice Analysis, i.e. a group of techniques assessing several features of voice such as syllable length, vowel duration, accent production, and phonation time. Observed impairments in people with HD included pitch and loudness (Hertrich & Ackermann, 1993), utterance duration (Ingo Hertrich & Ackermann, 1994), phonation time and movements of the larynx (Velasco

García et al., 2011). Significantly worse impairments were reported in HD ion syllable duration when compared with other neurological conditions, such as Parkinson's disease, Friedreich's ataxia, and pure cerebellar syndrome (Ackermann, Hertrich, & Hehr, 1995). Moreover, as the impairments were in some cases positively correlated with the severity of the disease, the hypothesis of the adoption of a speech acoustic marker to mark the onset of HD was put forward (Velasco García et al., 2011; Vogel, Shirbin, Churchyard, & Stout, 2012). Promising results were also found by a single group pre-test post-test study, based on a month of daily speech therapy sessions on phonation, respiration, and labial and lingual movements that showed to significantly improve cranial nerves assessment scores and swallow functioning (Giddens, Coleman, & Adams, 2010).

Discussion

Summary of Main Findings

In this review empirical studies were searched to identify the features of communication that have been investigated to date. The most adopted investigation methods were visual recognition tasks of emotional pictures, cognitive language examinations, and acoustic voice analysis. Very few studies focused on therapeutic interventions on communication or speech, often with a single case design (Ferm et al., 2010; Giddens et al., 2010; Hallberg et al., 2013; Klasner & Yorkston, 2001). Other much less investigated elements of communication included emotion expression and language comprehension. Despite the general focus of this review, the most neglected category was Communicative Skills, with only two studies focussing on their assessment, primarily through qualitative interviews and focus groups. However, this underrepresentation in the qualitative first person perspective literature on communication appears to be a

general tendency in all those chronic illnesses which feature communicative impairments, such as stroke, Parkinson's disease, and motor neuron disease (Mistry & Simpson, 2013; Thorne et al., 2002). This may be partially explained by the frequent co-occurrence of speech and cognitive impairments, which could confound the patients' understanding of their own communicative abilities.

Many studies compared people with symptomatic HD with people with other neurological disorders (Ackermann et al., 1995; Arango-Lasprilla et al., 2006; Barr & Brandt, 1996; Chenery, Copland, & Murdoch, 2002; Jensen et al., 2006; Longworth, Keenan, Barker, Marslen-Wilson, & Tyler, 2005; Monsch et al., 1994; Murray, 2000; Murray & Lenz, 2001; Murray & Stout, 1999; Peran, Demonet, Pernet, & Cardebat, 2004; Possin et al., 2005; Christopher Randolph et al., 1993; Rosser & Hodges, 1994; Snowden et al., 2008; Taylor et al., 2005; Testa et al., 1998; Troster et al., 1998; Ullman et al., 1997; Wang et al., 2003) or premanifest HD (Begeti et al., 2013; Henley et al., 2008; De Diego-Balaguer et al., 2008; Labuschagne et al., 2013; Lemiere et al., 2004; Milders, Crawford, Lamb, & Simpson, 2003b; Scahill et al., 2013; Velasco García et al., 2011; Vogel et al., 2012). However, only 21 studies out of 65 included specific information concerning the stage of the disease for each of the participants with manifest HD. The results of the comparisons with other neurological conditions normally vary upon the conditions themselves, and can be useful to better understand the role of neuroanatomical structures in the genesis of the disorders. The general conclusion from comparative studies with premanifest HD was that many of the cognitive and communicative impairments observed in symptomatic HD may show a much earlier onset than motor difficulties, thus starting to affect patients' lives long before a formal diagnosis of HD is even formulated. These findings, along with

the results of the PREDICT-HD cohort study, are coherent with the proposal for new broader and more flexible diagnostic criteria for HD (Loy & McCusker, 2013).

In terms of augmentation of communication, the adoption of Talking Mats™ and linguistic and cognitive supplementation strategies have yielded provisional positive results, helping people with HD to create or improve their coping and compensation strategies.

Despite the general lack of literature, the findings of the investigations of communication from the perspective of people with HD looked seminal in shedding a preliminary light on the influence of social and environmental factors on communication, such as the type of interlocutor, the amount of time allowed for conversations and the presence of a strong support from relatives and carers. However, many of these aspects (e.g., the nonverbal features of communication) are yet to be fully investigated.

The inconsistency of the findings on emotion expression makes it difficult to draw any conclusions about the impact of HD on the ability to convey emotional messages. This is likely to be due to the fact that the studies adopted diverging methods to conceptualize emotion expression (e.g., subjective intensity rating VS objective expression judgement). Moreover, the adoption of only pictorial visual stimuli leaves unanswered the question as to whether dynamic images would trigger different performances in emotion expression. Another limit in the results is the absence of stimuli for sadness, surprise and anger in some of the adopted assessment tools, as well as the lack of stimuli for the expression of emotional body language in general.

A deficit of recognition of negative emotions, which has been widely noted in the empirical literature (Bates, Tabrizi, & Jones, 2014), was confirmed in this review.

However, the results are not clear whether the impairment can be selective for disgust, as it has been often reported in previous studies (Henley et al., 2012) since specific deficits were also found for other emotions (e.g., anger and fear), in some cases (Labuschagne et al., 2013; Robotham et al., 2011; Scharmüller et al., 2013; Snowden et al., 2008) even without a concomitant deficit for disgust itself. Despite a general predominance of the use of visual methods for the assessment of emotion recognition and a relative homogeneity between them (i.e., often based on emotional picture labelling), this inconsistency among the performances may be due to the variation between similar tasks involving the same sensorial modality, as well among the few ones investigating different modalities, such as auditory or olfactive tasks. Indeed, certain stimuli may elicit more than one negative emotional response (e.g., both sadness and fear or fear and disgust) as well as elicit ambiguous responses that cannot be categorised properly. This appears to be particularly relevant for emotional responses based on body language, where disgust and fear tend to share the same kind of reaction (i.e., moving backward whilst putting hand palms forward; de Gelder et al., 2008) However, the data on body language responses are still quite sparse, and in this review only one study investigated their recognition.

Impairments of numerous domains of language, such as discourse comprehension, phonemic and semantic fluency, naming, picture description and syntactic abilities, were reported. This was particularly clear across all the cross-sectional studies, while the data appears to be more controversial in the fewer studies that adopted a longitudinal design, which often showed no significant changes in language over the period of years. Moreover, the vast majority of studies on language production focused on word retrieval; this leaves a gap in the investigation of the extent and variability of other aspects of language production. In addition, very little attention was given to the impact

of these impairments on conversational abilities. From this perspective, a single case study appears to suggest that deficits such as echolalia may be occasionally exploited by affected individuals as a compensation strategy to improve participation in conversations. This accords with a recurrent topic in clinical neuropsychology, i.e. that an impaired test result does not always reflect a functional impairment, as it is not often easy to distinguish between signs of impairment and signs of functional compensation (Leiwo & Klippi, 2000).

Finally, the findings showed that HD has a significant impact on features of speech such as pitch, phonation and loudness. However, promising results were reported for speech therapeutic interventions (Giddens et al., 2010). Perhaps most importantly, the observed impairments showed a tendency to correlate with the progression of the disease, leading to the hypothesis of a speech acoustic marker of disease onset. Yet again no study focused on the impact of these impairments on communication of both people affected by HD and close family and friends.

Limitations, Future Directions, and Implications for Clinical and Rehabilitative Practice

When considering the results of the present review, a potential limitation could be represented by the necessity of excluding all the citations published prior to the introduction of genetic testing in 1993. However, this exclusion currently represents a standard in research on HD, as demonstrated by its adoption by a number of recent reviews (Dumas et al., 2013; Franciosi et al., 2013; Henley et al., 2012).

Several directions for future research have emerged from the conclusion drawn by this review. As far as methods are generally concerned, it seems relevant to aim for an

increase in the number of studies adopting longitudinal designs in order to track the changes in communication throughout the whole progression of the disease. This is also compatible with the need for more comprehensive and precise information regarding the stage of disease of the participants included in the samples, as well as a deeper understanding of the several variables that can affect communication directly or indirectly, such as social issues, family support or medical treatment. Moreover, even though many studies included some kind of baseline neuropsychological assessment, the need exists for a more systematic inclusion of assessments of any possible variables that may affect communicative performances due to cognitive deterioration – such as aphasia, working memory functioning and deficits involving attention, visuo-spatial and executive skills – as well as motor impairment.

It is also apparent the need for further research characterised by a deeper focus on the subjective experience of affected people and their close relatives, and a more comprehensive coverage of the multiple features of communication. The same need exists for studies investigating the mechanisms involved in the way people with a diagnosis of HD communicate emotionally, as well as a focus on emotion recognition through different modalities. Moreover, as suggested by the limited evidence retrieved by the review, the heterogeneous nature of this construct is likely to be more efficiently understood through the adoption of diverse methods, ranging from subjective measures of emotional effectiveness, appropriateness, and intensity to objective evaluations performed by independent observers.

Finally, very little is known concerning the effect of the impairments of language, speech or emotion on the functional communicative capacity of people affected by HD and its impact on their quality of life and, not least, their healthcare experience. Indeed, poor

verbal communication can lead to inaccurate clinical assessments due to the inability of the patients to express their needs or comprehend the demands of their clinicians or carers. This can be also exacerbated by the combination of expressive aphasia and dysarthria that is often observed in HD due to involuntary movements and cognitive deterioration. Moreover, difficulties in emotional communication, both in terms of expression and recognition or empathy, can lead to problems similar to those observed with poor verbal communication, such as misunderstandings, feelings of awkwardness, and frustration. Moreover, the subtler and less evident nature of emotional difficulties (as compared to language impairments), may also trigger important social consequences, such as the development of avoidant behaviour, a further overall deterioration of interpersonal interactions – as observed with alexithymia (Spitzer, Siebel-Jürges, Barnow, Grabe, & Freyberger, 2005) – and the potential for social discrimination and stigma. Therefore, a more complete understanding of these constructs is required, possibly through the adoption of a variety of methods – including longitudinal designs and the investigation of a wider range of verbal, emotional and social components of communication – in order to monitor the subjective and objective experience of people with HD over the different stages of the disease. This will bring the overarching potential to help inform new intervention strategies, refine current clinical approaches, as well as improve patients' relationships with their caregivers and clinicians.

Conflict of Interest

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Tables

Table 1

Database search details.

Database	Search terms	Citations
PubMed	"Huntington Disease"[Mesh] AND "Communication"[Mesh]	262
	("Huntington Disease"[Mesh]) AND "Language"[Mesh]	100
PsycINFO	DE "Huntingtons Disease" AND DE "Communication"	16
	DE "Huntingtons Disease" AND DE "Language"	11
LLBA	"Huntington's"	89

Note: No Subject Term for HD was available in LLBA, therefore the general keyword "Huntington's" was searched through all the database fields.

Table 2

Distribution of citations among categories and subcategories.

Category	Subcategory	Partial citations	Total citations
Communicative Skills	Assessment	2	5
	Augmentation	3	
Emotion	Expression	4	19 (3 overlapping)
	Recognition	18	
Language	Comprehension	7	35 (1 overlapping)
	Neuropsychological Investigations	7	
	Production	22	
Speech	Assessment	5	6
	Therapy	1	

Table 3

Details of studies identified by the review.

Study	Category (Subcategory)	Methods	Groups	HD Demographic data			Relevant materials and techniques	Relevant findings
				Stage	Age (SD)	Gender		
Ackermann et al., 1995	Speech (Assessment)	Speech assessment	HD (14) PD (17) FA (9) PCS (13)	1 (2) 2 (3) 2-3 (2) 3 (2) 4 (5)	46.2 (11.8)	F (5) M (9)	Oral diadochokinesis task Acoustic analysis	Significantly lower syllabic rate in people with HD.
Arango- Lasprilla et al., 2006	Language (Neuropsychological Investigations)	Neuropsychological assessment	HD (11) FAD (10) Ctrl (17)	NR	52.4 (11.7)	NR	Semantic fluency Letter fluency Modified BNT	HD impaired vs Ctrl in all cognitive domains; more impaired than FAD only in letter and category fluency tasks.
Backman et al., 1997	Language (Neuropsychological Investigations)	Neuropsychological assessment PET, MRI	HD (5) Ctrl (5)	NR	49.4 (7.6)	F (2) M (3)	Verbal fluency task	HD severely impaired on verbal fluency. Performance on fluency predicted by alterations on caudate and putamen.
Barr & Brandt, 1996	Language (Production)	Language assessment	HD (30) AD (32) VD (23) Ctrl (40)	NR	44.4 (10.9)	F (14) M (16)	Letter and category fluency task	All participants more impaired on semantic rather than phonetic fluency.
Begeti et al., 2013	Language (Neuropsychological Investigations)	Neuropsychological assessment	HD (126) Pre-HD (28) Ctrl (21)	Ear. (49) Mod. (40) Adv. (37)	54.9 (2.9)	NR	ACE-R	HD more impaired on phonemic fluency rather than semantic.

Study	Category (Subcategory)	Methods	Groups	HD Demographic data			Relevant materials and techniques	Relevant findings
				Stage	Age (SD)	Gender		
Beglinger et al., 2010	Language (Neuropsychological Investigations)	Neuropsychological assessment (longit.: 2 visits, 16 mo.)	HD (38)	1 (47%) 2 (38%) 3 (12%) 4 (3%)	49.5 (11.6)	F (45%) M (55%)	Phonemic fluency RBANS	Progressive decline on attention, coding, digit span, list recognition, figure copy and recall.
Calder et al., 2010	Emotion (Recognition)	Neuropsychological assessment Emotional assessment	HD (21) Ctrl (NR)	NR	50.4 (8.71)	F (9) M (12)	BFRT Recognition of famous faces Ekman 60 Faces Test Emotion Hexagon Recognition of non-verbal emotional sounds Emotional vignettes (conceptual)	HD impaired on recognition of anger, fear and disgust across all domains.
Chenery et al., 2002	Language (Production)	Language assessment	HD (13) BD (13) Ctrl (13)	NR	52 (10.6)	F (8) M (5)	WAB BNT TLC-E TWOK TWT-R	HD severely impaired on lexico-semantic abilities, category fluency and interpretation of ambiguous meanings.
De Diego-Balaguer et al., 2008	Language (Comprehension)	Language learning assessment	HD (43) Pre-HD (13) Ctrl (20)	1 (24) 2 (18) 3 (3) 4 (1)	46.6 (8.8)	F (19) M (24)	Artificial language streams (trisyllabic items)	HD impaired on both word and rule learning.
de Gelder et al., 2008	Emotion (Recognition)	Emotional assessment	HD (19) Ctrl (19)	1 (10) 2 (9)	52 (9.1)	F (9) M (10)	Matching of nonverbal whole body instrumental	HD impaired in recognising instrumental and angry body language. Body language deficits

Study	Category (Subcategory)	Methods	Groups	HD Demographic data			Relevant materials and techniques	Relevant findings
				Stage	Age (SD)	Gender		
							and emotional expressions	correlated with measures of motor deficit
Deckel & Cohen, 2000	Language (Production)	Language assessment CBF	HD (9) Ctrl (13)	1 (5) 2 (2) 3 (2)	38.8 (3.3)	F (8) M (1)	Out loud word fluency and word reading tasks	HD impaired on both word fluency and word reading. CBF velocity inversely related to word fluency.
Dogan et al., 2014	Emotion (Recognition)	Emotional assessment fMRI	HD (14) Ctrl (14)	1 (6) 2 (6) 3 (2)	43.9 (8.9)	F (6) M (8)	Video clips displaying emotional facial expressions	HD impaired on recognition of negative emotions. Deficit related to reduced activity in both subcortical and cortical areas.
Duff et al., 2010	Language (Neuropsychological Investigations)	Neuropsychological assessment	HD (75)	NR	46.8 (12.9)	F (39) M (36)	RBANS	HD severely impaired on all cognitive domains measured by RBANS.
Ferm et al., 2010	Communication (Augmentation)	Videotaped conversations Quantitative and qualitative analysis	HD (5)	2-3 (2) 3 (3)	61 (14.8)	F (3) M (2)	TM EFFC	Both HD and partners more involved in conversations when using TM.
Giddens et al., 2010	Speech (Therapy)	Speech therapy Cranial nerves examination	HD (5)	NR	NR	F (4) M (1)	Labial and lingual motion and resistance training Respiratory and glottal exercises	Improvements in respiratory, phonatory, and articulatory functions, plus cranial nerve scores.
Hallberg et al., 2013	Communication (Augmentation)	Videotaped discussion groups	HD (4)	NR	41 (18.9)	F (1) M (3)	TM EFFC	Increased effectiveness and follow-up questions when using TM.

Study	Category (Subcategory)	Methods	Groups	HD Demographic data			Relevant materials and techniques	Relevant findings
				Stage	Age (SD)	Gender		
Hartelius et al., 2010	Communication (Assessment)	Qualitative interviews Focus groups	HD (11) FM (7) CG (8)	1 (1) 1-2 (2) 2 (3) 3 (5)	50.3 (8)	F (7) M (4)	/	Need for richer social life, more conversation partners and conversation adjustments.
Hayes et al., 2007	Emotion (Recognition)	Emotional assessment Neuropsychological assessment	HD (14) Ctrl (14)	NR	54.6 (11.2)	F (6) M (8)	Declarative knowledge of the situational determinants of basic emotions Recognition of vocal emotional expressions Categorisation of emotion words IAPS Disgust Scale Smell and taste test	HD impaired olfactory disgust, vocal disgust expressions, classification of disgusting pictures, and declarative knowledge of disgust elicitors.
Hayes et al., 2009	Emotion (Expression)	Emotional assessment	HD (11) Ctrl (11)	NR	53.4 (11.9)	F (3) M (8)	Spontaneous facial expressions (odorants) Posed facial expressions (induction and imitation)	HD showing fewer disgust-like facial reactions and less accurate facial expressions of disgust.
Hayes et al., 2009	Emotion (Recognition)	Neuropsychological assessment Emotional assessment	HD (14) Ctrl (14)	NR	47.8 (11.7)	F (6) M (8)	BFRT Emotion Hexagon FEEST	HD generally impaired on recognition of negative emotion on group analysis. Specific disgust deficit on individual analysis.

Study	Category (Subcategory)	Methods	Groups	HD Demographic data			Relevant materials and techniques	Relevant findings
				Stage	Age (SD)	Gender		
							Vocal emotion recognition IAPS Situational determinants of emotion	
Henley et al., 2008	Emotion (Recognition)	Emotional assessment VBM	HD (40) Pre-HD (21) Ctrl (20)	1-2 (40)	48.5 (9.6)	F (20) M (20)	Ekman Pictures of Facial Affect Voxel-based MRI	HD and Pre-HD impaired at recognising, surprise, disgust, anger and fear. HD more impaired than pre- HD on disgust and anger. Deficit generally associated with striatal atrophy, plus insula and prefrontal involvement for fear.
Hertrich & Ackermann, 1993	Speech (Assessment)	Speech assessment	HD (5) PD (7) Ctrl (12)	NR	47.4 (10.9)	F (3) M (2)	Assessment of acoustic signals: syllable lengths, vowel durations, sound intensity, pitch accent strength	HD showing reduced durational, pitch, and loudness changes concomitant with accent realization.
Hertrich & Ackermann, 1994	Speech (Assessment)	Speech assessment	HD (13) Ctrl (12)	1 (2) 2 (3) 2-3 (2) 3 (2) 4 (5)	49.8 (11.4)	F (5) M (8)	Assessment of acoustic signals: syllable lengths, vowel durations and VOT	HD with increased variability of utterance duration, impairment of durational parameters of phonetic timing and articulatory accuracy.
Ho et al., 2002	Language (Production)	Language assessment	HD (21)	NR	47.8 (10.6)	F (6) M (15)	Letter and category fluency task	HD producing significantly fewer correct words over time, and significantly

Study	Category (Subcategory)	Methods	Groups	HD Demographic data			Relevant materials and techniques	Relevant findings
				Stage	Age (SD)	Gender		
		(longit.: 3.5 follow- ups per yr)						more word repetitions. Plus significant impairment of phonemic switching for both letter and category fluency. Stable semantic switching.
Ille, Holl, et al., 2011	Emotion (Expression) (Recognition)	Emotional assessment Neuropsychological assessment	HD (28) Ctrl (28)	NR	48.4 (9.4)	F (11) M (17)	QADS Pictures depicting facial expressions of emotions (Karolinska- Set) IAPS	HD giving lower intensity ratings for anger, disgust and surprise, plus reduced classification accuracy for angry, disgusted, sad and surprised faces.
Ille, Schäfer, et al., 2011	Emotion (Expression) (Recognition)	Neuropsychological assessment Emotional assessment VBM	HD (18) Ctrl (18)	NR	51.9 (10.4)	F (8) M (10)	QADS Recognition of facial emotions IAPS	HD with normal affective experience but impaired recognition of disgust, anger, and sadness. Deficit related to atrophy in insula, prefrontal cortex and hippocampus.
Jensen et al., 2006	Language (Production)	Language assessment	HD (6) NS (6) Ctrl (6)	NR	51.8 (9.37)	F (4) M (2)	Picture Description task from the WAB	HD producing significantly more grammatical errors than both NS and controls.
Klasner & Yorkston, 2001	Communication (Augmentation)	Qualitative interviews	HD (1) FM (1)	NR	44 (N/A)	M (1)	Linguistic and cognitive supplementation strategies	Improvements in communication reported by both HD and FM.

Study	Category (Subcategory)	Methods	Groups	HD Demographic data			Relevant materials and techniques	Relevant findings
				Stage	Age (SD)	Gender		
Labuschagne et al., 2013	Emotion (Recognition)	Emotional assessment	HD (113) Pre-HD (115) Ctrl (116)	1 (72) 2 (41)	49.2 (9.2)	F (61) M (52)	Ekman Pictures of Facial Affect	PreHD impaired on recognition of fear, anger, and surprise. HD impaired on all emotions, neutral included. Neuroleptics associated with worse facial emotion recognition, SSRIs associated with better facial emotion recognition
Lemiere et al., 2004	Language (Neuropsychological Investigations)	Neuropsychological assessment (longit.: 3 visits, 2.5 yr)	HD (19) Pre-HD (12) Ctrl (11)	NR	49.9 (12.2)	F (12) M (7)	WAIS BNT Token Test	HD progressively impaired on attention, executive functions, memory, and object and space perception.
Lepron et al., 2009	Language (Production)	Language assessment PET	HD (12) Ctrl (17)	NR	45.9 (9.1)	F (9) M (3)	Word generation task	HD showing higher reaction times and number of errors. Left inferior temporal gyrus activated only in Ctrl and not HD during the task.
Longworth et al., 2005	Language (Production) (Comprehension)	Language assessment	HD (10) PD (15) CBV (7)	Mild (7) Mod. (3)	49.7 (6.2)	F (3) M (7)	Verbal and category fluency Past tense morphology tasks	HD impaired in both production and comprehension of novel past tense verbs.
Milders et al., 2003a	Emotion (Recognition)	Emotional assessment Neuropsychological assessment	HD (20) Pre-HD (20) Ctrl (20)	NR	47.6 (8.4)	F (8) M (12)	Ekman Pictures of Facial Affect Matching facial expression across identity	HD significantly more impaired on negative expressions other than disgust. No impairment in Pre-HD.

Study	Category (Subcategory)	Methods	Groups	HD Demographic data			Relevant materials and techniques	Relevant findings
				Stage	Age (SD)	Gender		
							BFRT - Short Version	
Monsch et al., 1994	Language (Production)	Neuropsychological assessment	HD (42) AD (44) Ctrl (44+42)	NR	48.7 (12.8)	F (19) M (23)	Category fluency task Letter fluency task	HD impaired on both letter and category fluency.
Munte et al., 1997	Language (Neuropsychological Investigations)	Neuropsychological assessment ERP	HD (9) Ctrl (9)	NR	43.6 (11.1)	F (4) M (5)	Word recognition tasks	HD showing a significant longer latency shift.
Murray & Lenz, 2001	Emotion (Recognition)	Conversation analysis Neuropsychological assessment	HD (9) PD (10) Ctrl (17)	NR	42 (11.1)	NR	Conversational discourse activities AIDS	HD using shorter and fewer grammatically complete utterances than Ctrl, but no difference with PD.
Murray & Stout, 1999	Language (Comprehension)	Neuropsychological assessment	HD (9) PD (9) Ctrl (8+8)	NR	41.6 (11.8)	F (4) M (5)	DCT ADP	HD equally impaired as PD on discourse comprehension,
Murray, 2000	Language (Production)	Neuropsychological assessment	HD (10) PD (10) Ctrl (9+9)	NR	42.2 (10)	F (4) M (6)	Narrative tasks Sentence Intelligibility (subtest of AIDS) ADP	HD producing shorter and syntactically simpler utterances than PD, despite similar cognitive and motor performance.
Peran et al., 2004	Language (Production)	Language assessment	ND-HD (17) D-HD (9) Ctrl (26)	NR	51.7 (11.6)	F (9/2) M (8/7)	Noun and verb generation task	No specific deficit for verb production ND-HD. Global language deficit in D-HD, in particular increased difficulties in verb production.

Study	Category (Subcategory)	Methods	Groups	HD Demographic data			Relevant materials and techniques	Relevant findings
				Stage	Age (SD)	Gender		
Possin et al., 2005	Language (Production)	Neuropsychological assessment	HD (10) PD (52) PSP (6)	NR	42.6 (13.4)	F (11) M (8)	Perseveration analysis Letter and category fluency WAIS BNT CVLT	HD presenting recurrent perseverations.
Power et al., 2011	Communication (Assessment)	Qualitative semi-structured interviews Pragmatics assessment Language assessment	HD (1) FM (1)	Adv. (1)	37 (N/A)	M (1)	WHO ICF ICF-based RPS-Form Pragmatic protocol in 1:1 and group situations WAB Modified CETI	Environmental factors such as family support can represent both key barriers and facilitators to communicative participation.
Randolph et al., 1993	Language (Production)	Language assessment	HD (8) AD (11) PD (8) Ctrl (8+11)	NR	43.6 (9.8)	F (3) M (5)	BNT Semantic fluency tasks (cued & uncued)	HD impaired on uncued task, but improved performance on cued one.
Rees et al., 2014	Emotion (Recognition)	Emotional assessment	HD (15) Ctrl (18)	1 (15)	52.3 (9.4)	F (12) M (3)	Static b/n photos (Manchester Face Set) Nonverbal vocal audio clips 1-second film stimuli	HD impaired on recognition of anger, disgust and fear on multiple modalities.
Rich, Troyer, Bylsma, &	Language (Production)	Language assessment (longitudinal: 3 yrs)	HD (72) Ctrl (41)	NR	42.5 (10.6)	F (33) M (39)	Letter fluency task	HD impaired on switching but not clustering. Switching but not clustering related

Study	Category (Subcategory)	Methods	Groups	HD Demographic data			Relevant materials and techniques	Relevant findings
				Stage	Age (SD)	Gender		
Brandt, 1999								inversely with disease severity.
Robotham et al., 2011	Emotion (Recognition)	Emotional assessment	HD (14) Ctrl (15)	Ear. (14)	51.3 (7.7)	F (6) M (8)	Recognition of nonverbal vocalizations of emotions	HD impaired on recognition of both negative and positive emotions.
Rosser & Hodges, 1994	Language (Production)	Neuropsychological assessment	HD (10) AD (10) PSP (10) Ctrl (25)	NR	59.9 (8)	NR	Letter and category fluency task	HD impaired on both letter and category fluency.
Saldert & Hartelius, 2011	Emotion (Recognition)	Conversation analysis	HD (1)	Adv. (1)	NR	F (1)	Sequential analysis of conversational interactions with a personal assistant	Echolalia used by participant as coping strategy to improve conversational exchanges.
Saldert et al., 2010	Language (Comprehension)	Language assessment	HD (18) Ctrl (18)	1 (5) 2 (7) 3 (6)	49.9 (13.4)	F (8) M (12)	Aphasia test (A-Ning) Comprehension of metaphors and lexical ambiguities from BeSS Discourse Comprehension Task	HD impaired in complex discourse comprehension. Impairment related to disease severity.
Sambin et al., 2012	Language (Comprehension)	Language assessment	HD (15) Ctrl (15)	NR	43.7 (9)	F (10) M (5)	Sentence-picture matching tasks	HD showing difficulties in sentence comprehension independently from working memory limitations.

Study	Category (Subcategory)	Methods	Groups	HD Demographic data			Relevant materials and techniques	Relevant findings
				Stage	Age (SD)	Gender		
Scahill et al., 2013	Emotion (Recognition)	Emotional assessment MRI	HD (120) Pre-HD (119)	NR	40.8 (8.9)	F (54) M (66)	Ekman Pictures of Facial Affect	HD impaired on recognition of negative emotions.
Scharmüller et al., 2013	Emotion (Recognition)	Emotional assessment VBM	HD (18) Ctrl (18)	NR	51.9 (10.4)	F (8) M (10)	KDES	HD impaired on anger recognition, but not other negative emotions. Impairment related to atrophy in cerebellar regions.
Snowden et al., 2008	Emotion (Recognition)	Emotional assessment Neuropsychological assessment	HD (10) FTD (12) Ctrl (12)	NR	47 (9)	F (5) M (5)	Definition of emotion labels Multiple-choice comprehension of emotion terms and situations Binary-choice comprehension of emotional situations FEEST Two-choice face-face expression and identity matching	HD impaired on recognition of all negative emotions, with most impaired emotion being anger. Impairment in HD worse than FTD.
Sprengelme yer et al., 1996	Emotion (Recognition)	Neuropsychological assessment Emotional assessment	HD (13) Ctrl (17)	NR	45 (7.6)	F (7) M (6)	Perception of gender, age and gaze BFRT Perception of morphed faces Ekman Pictures of Facial Affect	HD significantly impaired on recognition of negative emotions, with especially severe impairment on disgusts, on both facial and vocal modalities.

Study	Category (Subcategory)	Methods	Groups	HD Demographic data			Relevant materials and techniques	Relevant findings
				Stage	Age (SD)	Gender		
							Vocal emotion recognition Anger Scale Disgust Scale Fear Schedule	
Taylor et al., 2005	Language (Production)	Language assessment	HD (16) AD (20) Ctrl (16+20)	NR	44.3 (10.8)	F (7) M (9)	HCD	HD more impaired on semantic pairings, whereas AD more impaired on phonemic pairings.
Teichmann et al., 2009	Language (Comprehension)	Language assessment	HD (15) Ctrl (15)	1 (15)	48 (7.8)	F (5) M (10)	Phoneme perception and perceptual compensation tasks	HD not impaired on either phoneme perception or perceptual compensation, but reduced ability of phoneme discrimination.
Teichmann et al., 2006	Language (Comprehension)	Language assessment MRI	HD (15) Ctrl (15)	1 (15)	46.9 (9.8)	F (9) M (6)	Rule application and lexical tasks	HD selectively impaired on rule application, but not lexical abilities.
Teichmann et al., 2005	Language (Production)	Language assessment	HD (30) Ctrl (20)	1 (15) 2 (15)	48.9 (6.6)	F (14) M (16)	Conjugation and sentence-picture matching tasks	HD impaired on rule application, but not lexical abilities.
Teichmann et al., 2008	Language (Production)	Language assessment PET	HD (31) Ctrl (20)	1-2 (15)	45.4 (9)	F (13) M (18)	Rule application and lexical tasks	HD impaired on rule application, but not lexical abilities. Both abilities related to striatal activation, but different subareas.

Study	Category (Subcategory)	Methods	Groups	HD Demographic data			Relevant materials and techniques	Relevant findings
				Stage	Age (SD)	Gender		
Testa et al., 1998	Language (Production)	Neuropsychological assessment	HD (18) AD (96) PD (60)	NR	53.5 (12.3)	F (9) M (9)	Supermarket Task (semantic fluency) BNT	HD significantly impaired in terms of semantic cluster size.
Trinkler et al., 2013	Emotion (Expression) (Recognition)	Neuropsychological assessment Emotional assessment	HD (13) Ctrl (13)	NR	54.1 (7.2)	F (6) M (7)	KDEF Ekman Pictures of Facial Affect Videos of basic emotions expression TAS20 IRI BEES	HD significantly impaired on both emotion recognition and expression. No impairment on alexithymia and empathy scores.
Troster et al., 1998	Language (Production)	Neuropsychological assessment (Exp. 3 only)	HD (24) Ctrl (63)	NR	50.2 (12.9)	F (11) M (13)	BNT FAS Animal fluency tests	HD impaired on word generation, switching and phonemic and semantic cluster size.
Velasco García et al., 2011	Speech (Assessment)	Speech assessment	HD (18) Pre-HD (2) Ctrl (20)	NR	47 (12.7)	F (5) M (13)	VHI Analysis of aerodynamic efficiency Acoustic analysis Laryngeal examination	Reduced maximum phonation time and uncontrolled adduction-abduction movements observed in HD. Impairments related with disease severity.
Ullman et al., 1997	Language (Production)	Language assessment	HD (17) PA (8)	NR	45 (NR)	F (5) M (12)	Past tense word production and rule application tasks	HD showing excess motor activity and past tense rule use.
Vogel et al., 2012	Speech (Assessment)	Speech assessment	HD (17) Pre-HD (13)	NR	57 (12.2)	F (8) M (9)	Analysis of timing, frequency and intensity	HD significantly impaired in speech rate. Pre-HD not significantly impaired, but

Study	Category (Subcategory)	Methods	Groups	HD Demographic data			Relevant materials and techniques	Relevant findings
				Stage	Age (SD)	Gender		
			Ctrl (15)					tendency to significance. Hypothesis of speech acoustic marker for diagnosis of HD.
Wang et al., 2003	Emotion (Recognition)	Neuropsychological assessment Emotional assessment	HD (6) WD (32) Ctrl (16)	NR	44.8 (4.2)	F (2) M (4)	Perception of gender, age & gaze direction BFRT Perception of basic emotions	HD generally impaired on emotion recognition, with specific severe impairment for disgust.

Note: ACE-R = Addenbrooke's Cognitive Examination-Revised; AD = Alzheimer's Disease; ADP = Aphasia Diagnostic Profiles; AIDS = Assessment of Intelligibility of Dysarthric Speech; BD = Brain Damaged; BEES = Balanced Emotional Empathy Scale; BFRT = Benton Facial Recognition Test; BNT = Boston Naming Test; CBF = Cerebral Blood Flow; CBV = Cerebrovascular condition; CETI = Communication Effectiveness Index; CG = Caregiver; CVLT = California Verbal Learning Task; D-HD = Demented HD; DCT = Discourse Comprehension Test; EEFC = Effectiveness Framework of Functional Communication; ERP = Event Related Potentials; FA = Friedreich's Ataxia; FAD = Familial AD; FAS = Letter Fluency Test; FEEST = Facial Expressions of Emotion: Stimuli and Tests; FM = Family Member; FTD = Frontotemporal Dementia; HCD = Hat-Cat-Dog task; HD = Huntington's disease; IAPS = International Affective Picture System; fMRI = functional Magnetic Resonance Imaging; IRI = Interpersonal Reactivity Index; KDEF = Karolinska Directed Emotional Faces; MRI = Magnetic Resonance Imaging; ND-HD = Non-Demented HD; NR = not reported; NS = Non-thalamic Subcortical Stroke; PA = Posterior Aphasia; PCS = Pure Cerebellar Syndrome; PET = Positron Emission Tomography; PSP = Progressive Supranuclear Palsy; QADS = Questionnaire for the Assessment of Disgust Proneness; RBANS = Repeatable Battery for the Assessment of Neuropsychological Status; RPS-Form = Rehabilitation Problem Solving-Form; TAS20 = Toronto Alexithymia Scale; TLC-E = Test of Language Competence - Expanded Edition; TM = Talking Mats; VD = Vascular Dementia; TOMWK = Test of Word Knowledge; TWT-R = The Word Test - Revised; VBM = Voxel Based Morphometry; VHI = Voice Handicap Index; VOT = voice-onset-time; WAB = Western Aphasia Battery; WAIS = Wechsler Adult Intelligence Scale; WD = Wilson's Disease; WHO ICF = World Health Organization International Classification of Functioning.

Figures

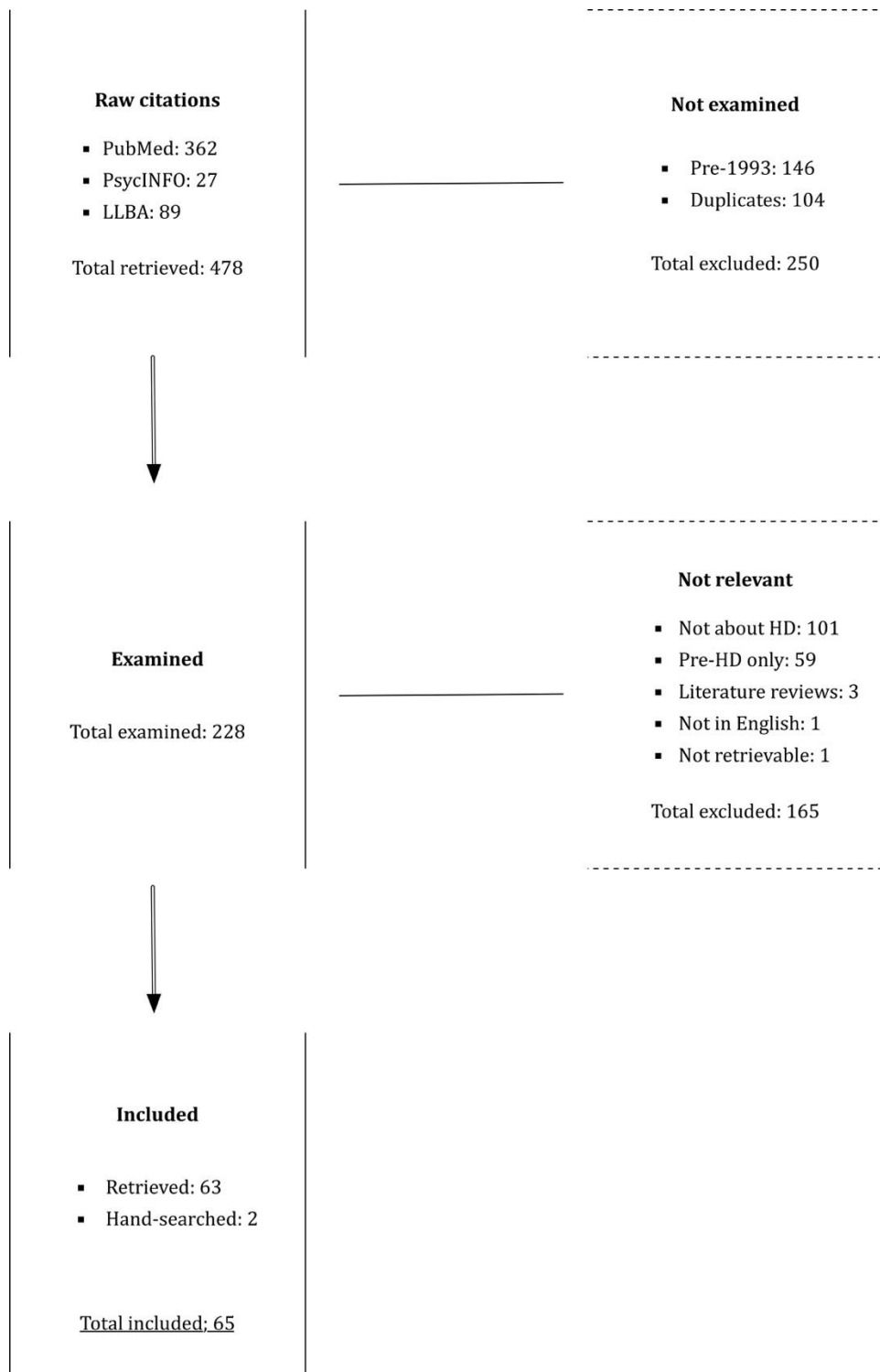


Figure 3: flow diagram of the study selection process.

Chapter 4

Publishable Paper Two (PP2)

“I have a feeling I can’t speak to anybody”: a thematic analysis of communication perspectives in people with Huntington’s disease

Status:

- A. Published as abstract following poster presentation at HSG 2017⁵.
- B. Published.

Journal:

- A. *Neurotherapeutics*
- B. *Chronic Illness*

Statement of authorship:

Mr Nicolò Zarotti: 90%	Signed:
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⁵ Huntington Study Group (HSG) Annual Meeting 2017, November 2-4, Denver (CO), USA. For the relative poster, see Appendix 2.

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Abstract

Background: Synaptic dysfunction is increasingly recognized as one of the earliest pathological events in many neurodegenerative diseases, including Huntington's disease (HD). Despite HD being a monogenic disease with a well-characterized pathology, the molecular mechanisms underlying synaptic dysfunction remain poorly understood. In this study, we show that loss of specific synaptic connections can be observed as early as 3 months in multiple slow progressive HD mouse models prior to the onset of motor or cognitive impairment and that microglia contribute to this via a complement-dependent mechanism.

Methods/results: During development, synapse loss as a result of pruning is a normal and highly regulated process required for correct wiring of the brain. Microglia together with complement proteins, a key component of the innate immune system, play a key role in regulating this process. Here, we provide evidence that aspects of this developmental mechanism are aberrantly reactivated in HD mouse models, leading to the loss of specific synaptic inputs. Using super-resolution imaging, genetic ablation, and viral-mediated labeling, we demonstrate that complement proteins selectively target corticostriatal connections for engulfment by microglia and that this process requires mutant huntingtin expression in both striatal and cortical neurons. Moreover, blocking this process at multiple levels through both genetic- and antibody-mediated approaches is able to prevent synaptic pathology. Notably, we find that aspects of this mechanism may be conserved in human disease, as postmortem tissue reveals evidence of synapse loss, complement deposition at synapses, and phagocytic microglia. Together, these findings demonstrate that elements of the innate immune system previously only thought to be involved in late-stage inflammation mediate the loss of specific synaptic connections early in HD pathology.

Poster #70

"I Have a Feeling I Can't Speak to Anybody": A Thematic Analysis of Communication Perspectives in People with Huntington's Disease

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Abstract

Background: The current literature on communication in Huntington's disease (HD) appears to be mainly characterized by quantitative studies focused on the observation of medical and cognitive impairments, whereas the investigation of the patients' subjective experiences is generally much rarer. This study explored the perspectives of people affected by HD on their own communicative abilities.

Methods: Qualitative semi-structured interviews were carried out with 8 people with early-stage HD. The data were analyzed through thematic analysis. Four themes were constructed from the resulting data, characterized by the following core topics: how HD directs and mediates communication; regaining control to improve communication; emotional outflows into communication and the struggle for separation; and sheltering as a way to boost confidence in communication.

Results: The findings showed that separating patients' identity as individuals from that of a person with a disease can help increase communicative control. Consistent with the general theory and model of self-regulation, patients should be allowed a wider range of choices to regain control over communication. In addition, achieving better emotion regulation is of paramount importance for communication, and factors such as medication regimes, relationships, and existing coping strategies should be strengthened. Consistent with previous research, feelings of safety and the idea of a safe place ("sheltering") represent an effective coping mechanism. Practical implications include the refinement of communication and relationships among clinicians, caregivers, and patients with HD by considering a wider range of medical, psychological, and socio-environmental factors.

Poster #71 Award Winner for Michael Conneally Genetics Abstract

Selection of an Adeno-Associated Virus Gene Therapy Targeting Huntingtin for the Treatment of Huntington's Disease

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Abstract

Background: Studies in animal models of Huntington's disease (HD) support a strategy focused on lowering huntingtin (HTT) mRNA and protein, as partial suppression of HTT in the central nervous system is both safe and

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“I have a feeling I can’t speak to anybody”: a thematic analysis of communication perspectives in people with Huntington’s disease

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Abstract

Objectives: This study explored the perspectives of people affected by Huntington's disease on their own communicative abilities.

Methods: Qualitative semi-structured interviews were carried out with 8 people with early HD. The data were analysed through thematic analysis.

Results: Four themes were constructed from the data, characterised by the following core topics: How HD directs and mediates communication; Regaining control to improve communication; Emotional outflows into communication and the struggle for separation; Sheltering as a way to boost confidence in communication

Discussion: Separating patients' identity as individuals from that of a person with a disease can help increase communicative control. Consistent with the general theory and model of self-regulation (SRM), patients should be allowed a wider range of choices to regain control over communication. Achieving better emotion regulation is of paramount importance for communication, and factors such as medication regimes, relationships and existing coping strategies should be strengthened. Consistent with previous research, feelings of safety and the idea of a safe place ('sheltering') represent an effective coping mechanism. Practical implications include the refinement of communication and relationships among clinicians, caregivers, and patients with HD by considering a wider range of medical, psychological and socio-environmental factors.

Keywords: Huntington's disease; communication; emotion regulation; thematic analysis; patients' perspectives.

Introduction

Huntington's disease (HD) is a hereditary chronic neurodegenerative disorder which affects 10-12 people per 100,000 in the western world.¹ Typical symptoms include involuntary movements (chorea), cognitive deterioration, psychological difficulties, and psychiatric disorders.² Since the transmission mechanism is autosomal-dominant, affected individuals have a 50% probability of transmitting it to their children (usual age of onset is 40-50 years). The mean life expectancy after the diagnosis is typically 20 years³. Genetic testing is available for individuals at risk, allowing them to know if they carry the disease gene before the onset of symptoms. All individuals with the gene without symptoms ('presymptomatic' people) will develop the disease.

Many cognitive impairments have been reported in people with HD, including problems with all aspects of communication. Communication is understood in this context as a multifaceted ability that investigates how people create meanings through messages transmitted across various channels, media and contexts,⁴ and includes elements of language, speech, as well as emotion and social abilities. Regarding more specific aspects of communication, speech production is often impaired and starts to deteriorate before comprehension,⁵ and the spontaneous initiation of conversations is reduced.⁶ Research on nonverbal communication, such as body language and emotional processing, is less frequent in HD, with the exception of emotion recognition.⁷ In this respect studies have indicated that all these components can be affected by the disease.⁸ In addition, HD can also negatively affect the ability to comprehend and reflect on the mental

states of oneself and others (i.e., theory of mind⁹), which plays a fundamental role in communicative competence, especially through the attribution of intentions.¹⁰

Nevertheless, the current literature on communication in HD appears to be mainly characterised by quantitative studies focused on the observation of medical and cognitive impairments,¹¹ with a tendency to prioritise the perspectives of clinicians.¹²⁻¹⁴ The investigation of the perspectives of people with HD on their own communication abilities is generally much rarer and only recently has an interest started to emerge in communication as a phenomenon that embraces social skills and interactions, and not just the traditional underpinnings of language and speech.¹⁵ Indeed, even when patients' perspectives on the general impact of the disease have been investigated, data analysis falls more in the quantitative category, with greater focus given to the frequency of reoccurrence of themes rather than a qualitative exploration of patients' narratives themselves.¹⁶ On the other hand, to our knowledge only two studies have so far used a qualitative approach to investigate communication in HD.

Hartelius and colleagues¹⁷ adopted individual interviews and focus groups to triangulate the information between people with HD, family members, and caregivers, finding that a number of social and environmental factors play a relevant role in complicating communication. These included the speed of conversations or having fewer people to talk with, while a positive impact was reported for sensations of safety, having the opportunity to speak for a longer time, and the perception of support and adjustment from the person with whom they were speaking. However, the thematic analysis was characterised by a very descriptive approach, and the interviews did not include any questions on

nonverbal aspects of communication. In the second study conducted, Power, Anderson and Togher¹⁸ carried out a single case analysis with a man with advanced-stage HD and also found that supporting social and environmental factors can have a positive impact on communication. However, their approach (content analysis) offered a useful but predominantly descriptive report and did not allow for a more in-depth interpretation of meanings, as well as omitting nonverbal elements of communication such as emotional processing.

Therefore, considering the current gap in the literature, the research question addressed by this study was the exploration of how people affected by HD make sense of their communication experience with others.

Methods

Methodological approach

A qualitative design was adopted, based on semi-structured interviews analysed through thematic analysis (TA). We selected TA for its recognised usefulness within the field of psychology and communication disorders,¹⁹ and its ability to allow for the usage of both deductive (i.e., theory-driven) and inductive (i.e., data-driven) analyses of the themes identified within the interviews.²⁰

Sampling

People affected by Huntington's disease were invited via post across the North West of England by the Huntington's Disease Association (HDA). Of the people who expressed their interest, eight participants with symptomatic HD were considered eligible. The inclusion criteria included being aged 18 or more, being

able to be interviewed in English and being symptomatic at an early or moderate stage of disease. This limit was due to the difficulties in attending personal interviews that arise in the later stages of HD. As the study focused on the impact of the disease on patients' communicative experiences, we did not include any pre-symptomatic individuals. See Table 1 for the demographic details of the participants.

Procedure

Individual semi-structured interviews were carried out face to face with the participants at a time and place convenient to them (usually their home). We selected this approach to allow the in-depth exploration of relevant themes as well as to ensure that all participants felt comfortable and had all the assistance they might need.²¹ The length of the interviews ranged from 40 to 60 minutes ($M = 45$). Most of the participants reported to feel more comfortable if a caregiver (e.g., their partner) was allowed to be present during the interviews. Since our main aim during the data collection was to make the participants as comfortable as possible, this request was always facilitated. However, the caregivers were asked not to contribute in any way and any comments they made did not form part of the data collection. Moreover, in order to guarantee a minimum impact on the interviewing process, the caregivers were asked to sit in a position which limited any verbal and nonverbal interferences (e.g., out of the participants' direct eye contact). We structured the interviews according to a framework consisting of four general topics: Verbal Communication, Nonverbal Communication, Mediators of Communication, Contexts of Communication. The topics were based on some of

the most common categorisations of communication dimensions²² and to be as broad and yet comprehensive as possible.

Data analysis

All the interviews were transcribed verbatim and analysed using thematic analysis (TA). The six steps outlined by Braun and Clarke¹⁹ were used as a guide to analysis.

Ethics approval

This study was reviewed and approved by the Faculty of Health and Medicine Research Ethics Committee at Lancaster University (ref: FHMREC14026).

Results

Identified codes and themes

Following familiarisation with the whole dataset, 73 codes were generated. These were then collated to six initial candidate themes. Upon further revision, the final code list was reduced to a total of 36. Out of these, four final themes were identified that were distinctive in their own right, as well as coherent with the broader scheme of the analysis and relevant to the research question. See Table 2 for the final theme list and breakdown into the respective codes.

“You ever wanna say things to people, but you can’t”: how HD directs and mediates communication

All the participants agreed that HD plays an important role in their communication with others. In fact, they made sense of their communicative

difficulties by seeing HD as the main cause and this expressed itself both directly and indirectly. Directly, the participants saw HD as an external agent that actively blocks their communication:

“I think it is a funny illness, but affects you in different ways. [...] you ever wanna say things to people, but you can't. Because it feels there's something wrong, with your illness. [...] You want to say things, but you can't!” (Participant 3)

The effects of HD can present in many different ways, starting from speech and language. Problems with articulating words, controlling voice speed, or respecting conversational roles were frequently reported and disrupted verbal communication at its core:

“I probably speak fast. And also not loud enough. Sometimes I have to speak twice. I probably do articulate a bit harder. [...] I don't use many long words now, where I used to. I used to bullshit to heaven, but now I don't.” (Participant 7)

The cause of these difficulties was entirely attributed to HD, which the participants perceive as the external reason they felt and behaved the way they did. In this perspective, HD acts as director of the participants' communication, taking control over their active role in everyday interactions by drastically decreasing their communicative repertoire. As some of them pointed out:

“You don't speak to people... the illness makes you feel that way, even though you're trying to speak to people.” (Participant 3)

However, the influence of HD on communication is not confined to its most apparent manifestations, such as articulation impairments. Indeed, many related difficulties such as feelings of constant fatigue, memory problems, and attentional drops can indirectly affect the participants' communicative experience. Although not strictly related to the verbal side of communication, these problems ultimately play an important role in participants' everyday life by undermining their ability to interact with people or read situations:

"I like speaking with my son, I just keep... running out of things to say, because I keep forgetting." [...] For example we go to my sister's for Christmas, and I was trying get involved but I just feel so stupid because I can't think, my memory is very random [...] and it just adds more and more pressures because I start getting agitated with myself. [...] It must be to do with HD... [...] I don't know what's going on half of the time."
(Participant 4)

"The other thing that happens sometimes is that I can't think of a word to say next." (Participant 8)

Therefore, HD not only directly influences participants' communication through its characteristic symptoms, but also subtly mediates it via less apparent yet related conditions that pull the strings of nonverbal interactions and greatly increase the effort involved in simple discourses. Just as for their verbal issues, however, the participants made sense of these mediational effects as an external source of discomfort due entirely to HD. Not surprisingly, the combination of these influences eventually led the participants to develop a reluctant attitude towards

communication. This particularly manifested through being quiet or avoiding situations in which communication is required:

"I just can't be bothered, I just bugger off. I got a little scooter, so I just nip off to the quarry. [...] I don't really bother that much talking. If it's not the neighbour then I wouldn't bother. Yeah, it's just a bit of an effort. [...] I just say hello to acknowledge people." (Participant 1)

Again, it is worth noting how these behaviours were seen by the participants as due to the disease, to the extent of listing them among its stages and symptoms:

"I don't really mean [to speak to people], because one stage of HD you don't intermingle with people, leave them on your own, if you will. I don't know if you know..." (Participant 4)

Indeed, one of the participants mentioned being identified with HD by her parents, by having 'the Huntington's face':

"They say... 'you know, you look as you have Huntington's'. I've got the face. The Huntington's face. It's just the expressions that my mum used to do with Huntington's. And they say I look the same." (Participant 6)

Clearly, being identified this way can lead to significant difficulties in communication. From this perspective, the participants' tendency to conceive HD as an external director and mediator may represent one of the causes of the 'fighting' metaphors often used to describe their struggle with symptoms and their

consequent attempts to regain control over communication and prevent it from being incorporated into their own identity.

Becoming a director again: regaining control to improve communication

Of all the effects of HD on participants' communication, the lack of control appeared to be perceived as one of the most substantial and, as mentioned, it manifested in several direct and indirect ways. However, it also seemed to drive the participants' will to develop strategies to reacquire control over communication and become once again the real director of their daily interactions. For instance, all the participants claimed to find using the telephone much harder since the onset of the disease, due to the difficulties people had in understanding them. As a consequence, they started avoiding phone conversations, asking their carers or partners to intercede for them. However, this went beyond a mere avoidant behaviour, since it was often taken by the participants as an opportunity to shift the communication to more comfortable settings and modalities, like meeting in person or replying by email:

"The phone is probably the most difficult one. [...] I talk to people, but it's just the phone sometimes. I prefer to leave it. [...] If somebody phones me, and I have to get back in touch with them, I just use text or an email. That type of thing." (Participant 7)

Texting was a popular choice for most of the participants, as it did not impose the same constraints as other communication methods in terms of time and effort. As Participant 4 noted:

“Texting! That's one thing that I can do! If I text people, they understand what I'm talking about!” (Participant 4)

From this perspective, delegating phone conversations and switching to texting represent a good strategy to shift part of the burden of communication. This was even clearer when some of the participants claimed to prefer listening over talking. Well aware that communication consists of both sending and receiving, they appeared to have developed a preference for the latter:

“I can still listen, you know what I mean. I prefer listening to speaking. I can understand what people are talking about, I can listen. If I'm going to a group or something, I sit and listen, and then... when I feel comfortable with people around me, then I can open up and I think that has to do with HD...” (Participant 4)

This is probably due to how listening allows engagement in conversations without the amount of energy required by speaking. As Participant 5 noted:

“I know when to chill out and I know when to use the energy. Like this is using more energy than I would normally. But tomorrow daytime I can relax and I've got a birthday party tomorrow night and I know I'm gonna use more energy for that, because I'll be seeing a lot of people and I'll be talking to a lot of people. (Participant 5)

In addition, the management of these aspects of communication seems to benefit from an attitude of general openness towards HD and its effects. Indeed, many participants mentioned telling people about their condition, in order to prevent any distressing situations:

"I'm more open. I know I can explain things to people. [...] I do say to people 'look, if I sit with you, I know I might bump into you'. If I'm going out somewhere I'll sit at the end of the table. [...] People understand. If I'm open about it, people understand." (Participant 2)

Openness, however, comes at the cost of potential misunderstandings, as reactions and interpretations may differ significantly among people:

"The worst thing people do, is tell people. As soon as you tell them. That's it. Bloody terrible. I've never done, I'd never tell anybody again, I've never advise anybody to tell anyone. 'Cause once you've told them, then you're not going to get employed or you're going to have a rough ride from then on." (Participant 7)

Thus, trying to be open to regain communicative control can backfire, by turning communication into a source of emotional discomfort.

"One moment I'm fine, the next moment I'm not": emotional outflows into communication and the struggle for separation

One of the most challenging effects of HD was how it made emotions become unstable, and for many this was a new experience:

"One moment I'm fine, the next moment I'm not. Different things upset me... I was at work, and there was this nurse, she was bossing me about, and I said you're being horrible to me [...] and I got angry with her – never been like that before [...] I was thinking 'what's wrong with me?' And she

said to me 'what's wrong with you?', I said "I don't know", I said I'm going to go and get tested. I may have Huntington's." (Participant 6)

Moreover, the emotional aspects varied significantly. For instance, Participant 5 mentioned getting progressively detached from her emotions:

*I used to watch telly and I used to cry at the drop of a hat, and I used to have a box of tissues next to me and be soppy at anything, and then now I'm not. That box of tissues can stay there for months *laughs*. [...] I'm not as sympathetic as I used to. [...] I can still get angry, but not as much. I'm becoming more and more apathetic. Definitely. (Participant 5)*

On the other hand, Participant 6 seemed to experience the opposite situation:

I think I am sort of angry a lot more that I used to be, but I don't mean to be. [...] I knew there was something wrong with me, snappy and shouting at people, and I thought that's not like me, because I don't shout. I do now. It's my Huntington's, I can't help it. (Participant 6)

However, the actual effect perceived by affected individuals can be rather homogeneous, since the participants made sense of it as a general external influence beyond their control. As for speech and language, emotional issues were perceived as a symptom of the disease, rather than part of the psychological adjustment to the illness. As Participant 2 noted:

"I think it was just the fact that... a symptom of the disease. As for depression." (Participant 2)

Difficulties with emotions can become even more problematic when, as previously mentioned, distressing emotional situations may arise while trying to be open about HD and regain control over communication. Although openness and control can promote illness acceptance (which is considered of primary importance in psychological adjustment to chronic illness^{23,24}), those emotional experiences took a major toll on the participants, ultimately leading them to perceive their emotional and communicative life as a source of unsteadiness that frustrated any attempts at accepting their condition:

“With this illness, even though I accept it, it's very hard to accept things. I cannot drive anymore, and that's what I loved. I loved to drive all the time. All the sport's gone...” (Participant 4)

“I'm good at crying... I think I've got a little more sad. I started to be sort of thrown off without sense. I think when you have to be strong every day, sometimes it is hard...” (Participant 8)

One of the ways to manage their communication and ultimately promote illness acceptance was for the participants to learn better how to regulate their own emotions. However, with the participants, emotion regulation seemed to happen at the cost of engaging with extremely difficult and fundamentally disturbing thoughts, which led them to deal with challenging emotional outflows into their communicative experience. As Participant 4 mentioned:

“The thing with HD, I've noticed, if I am aggravated or someone else is aggravated or agitated, it takes some 5 or 10 minutes to me to calm down, but with HD it has a knock-on effect so it's like taking 3 times as

long for me just to calm down because I go up on it, got more and more to think about and it drives me crazy.” (Participant 4)

The process of achieving a better level of separation between communication and emotions clearly plays a pivotal role from this perspective. Yet, this can prove to be a very long and painful path, characterised by pervasive feelings of anxiety and instability that can turn communication into both a benefit and a liability, a ‘double-edged sword’:

“I know it's important to talk to people, but when everybody comes around to see me, I just feel a bit nervous about talking. And I think it must be the illness, Huntington's, making me feel like that way. But I don't mind people talking about my illness to people, so it's a double-edged sword.” (Participant 3)

However, whereas such a separation is achieved, emotion regulation has a clear beneficial effect. In the case of the participants, it promoted healthy grieving, helped them to better accept their condition, and ultimately improved their communication:

“I think I got better, 'cause I've grieved for what I've lost. So I accepted that. 'Cause a couple of years ago, I'd go into a shop and say 'I've got Huntington's' and I'd cry my eyes out. I've come to terms with it.” (Participant 1)

Although long and challenging, the process of regulating emotions was seen to be promoted by a number of medical and psychosocial factors. For instance, all the participants agreed on the paramount importance of medication:

"I was just tired and couldn't be asked to do things, you know. But now, once I started to take Citalopram I felt so much better in myself, it really lifted me up." (Participant 2)

In certain cases, being prescribed the appropriate medications could even benefit communication directly:

"I've got my medications now I'm fine. [...] I wouldn't even be able to speak if I didn't take them." (Participant 6)

A similar beneficial effect was also mentioned for social and environmental factors. In particular, having a close family and friends around allowed the participants to feel more comfortable about communication, as well as safer when coping with daily situations by being able to ask for help when needed:

"I do feel more comfortable here, you know like if you have people around, I'm fine. [...] I'm alright with going to different places. Well, I do try to, I like to try different restaurants and stuff like that, which I've already done. That hasn't gone yet. And we do have people round for meals and stuff like that." (Participant 7)

It is also important to notice how the positive relationship between emotion regulation and communication is bidirectional: just as regulating emotions can improve communication, so communicating better can promote emotion regulation; similarly, better relationships can improve both emotion regulation and communication – and vice versa. Thus, the successful expression

and reinforcement of these factors allowed for a virtuous circle to come into being, which considerably improved the participants' quality of life.

“I go back into my little world”: sheltering as a way to boost confidence in communication

Another beneficial factor that emerged from some interviews was the idea of having a personal safe place where the participants could take shelter. Initially, this idea appeared to be mainly identified with a feeling of preference and protection for their own home:

“I think, if I'm here, just sitting here, it feels comforting. I feel better at home.” (Participant 8)

“I don't know, I just... I feel my Huntington's. [...] I just need to get in the house, get in my comfy house.” (Participant 6)

However, as a couple of participants further elaborated, the idea reached a deeper level of meaning which transcended the geographical space and embraced a more abstract concept of safety. For example, Participant 4 talked of his 'little world':

*“I go back into my little world, you know what I mean [...] I just shut down, I don't know how I'm doing it *laughs*” (Participant 4)*

Similarly, Participant 5 mentioned her 'little bubble':

"I know that sounds so ignorant, but I can just sort of getting into my little bubble and just completely close down if I need to, you know."

(Participant 5)

This mental safety net appeared to have developed to help the participants deal with their everyday emotional and communicative life: when fatigue kicked in, emotions became overwhelming, or the circumstances got too demanding they could return to their shelter - of which their house in some cases only seemed to constitute a physical representation:

"I just seem to be able to switch off because I don't know what we are talking about. [...] It's quite hard to explain... I just shut up... I just shut down and ignore people and then I look back here [at home]."

(Participant 4)

From this perspective, the idea of sheltering appeared to provide the participants with an effective coping mechanism to boost their self-confidence. By knowing that they had a safe shelter, they could feel safer while dealing with everyday life's demands. Indeed, as consequence of the process of taking shelter, of 'feeling like in their own home', they could also feel comfortable enough to open up again and communicate:

"I do try to talk to people. Sometimes when I just feel... like in my own home. Yeah, my own home. So I invite people to come around here, to chat with people." (Participant 6)

Therefore, along with the aforementioned beneficial effects of medications and close relationships, sheltering seemed to play an important role in maintaining or restoring participants' communicative abilities

Discussion

Summary of main findings

The four themes identified by our study showed substantial areas of relevance to current models and theories of communication and psychological adjustment to chronic illness. The first theme concerned how HD acts as both director and mediator of the participants' communication, affecting their linguistic abilities by making them feel blocked and not able to speak as they used to, or pulling the strings of a number of collateral conditions that deeply affected their communicative experience, such as chronic fatigue and memory problems. According to the self-regulation model (SRM; ²⁵), patients' representations and beliefs regarding their chronic illness have a substantial effect on the successful development of coping strategies and ultimately their psychological well-being. The SRM has been specifically adopted with HD,^{26,27} finding that the perceptions of the disease are often characterised by strong illness identity due to its overwhelming perceived effect on all domains of their life. From this perspective, by conceiving HD as an external, separate agent the participants seemed to lay the foundations to fight for a fundamental domain of their own life by taking back some control over communication, which in turn can promote an identity less characterised by their illness.

The importance of control was emphasised in the second theme: retaking control over the features of communication appeared to represent an effective coping strategy aimed at improving the participants' communicative skills. Whether it was the possibility to avoid phone conversations, the decision to prioritise listening over speaking, the management of scarce energy resources, or being open about HD, regaining aspects of control affected the participants positively by giving them a chance to break free from the pervasive nature of HD, better accept their condition and become the director of their communication once again. This view fits with the general theory of self-regulation,²⁸ which sees self-regulatory skills as a limited resource that can be exhausted – a phenomenon which has been named self-regulatory fatigue or ego depletion²⁹ and that plays a pivotal role in quality of life and coping in chronic illness^{30,31}. Thus, regulating energy and fatigue by having a choice can represent an effective strategy to deal with the demanding nature of communication.

The third theme focused on how HD threatened participants' emotional life and stability. Presentations of this included inconsistent feelings of anger and sadness, apathy, and considerably longer emotional 'cool-down' times, which are not uncommon in HD¹. These ultimately led the participants to perceive their emotions and communication as a source of unsteadiness. One possible solution was represented by improving emotion regulation, which is a concept that refers to the process of influencing which emotions one experiences, as well as when and how such experiences occur,³² and whose importance in mental health and well-being has been widely recognised over the last decade.³³⁻³⁵ However, for the participants emotion regulation required engaging with difficult and disturbing

thoughts, leading to challenging emotional outflows into their communicative attempts, which caused pervasive feelings of anxiety that could turn communication itself into “*a double-edged sword*”. Nonetheless, where a better level of separation between emotions and communication was achieved (and also thanks to the medication and supportive social relationships), emotion regulation allowed for a substantial improvement in their willingness to communicate.

Last, the fourth theme explored a coping strategy adopted by many participants: the idea of having a personal safe place where they could take shelter. This was initially identified with their home, but later exceeded the physical dimension and extended to a more abstract idea of safety, a “*little bubble*” where they could switch off. The idea of sheltering provided the participants with a feeling of safety that helped them deal with stressful situations, and eventually shaped an effective mechanism to boost their self-confidence in regulating emotions and regaining control over communication. This finding seems consistent with the positive effect of feelings of safety reported by Hartelius and colleagues.¹⁷

While a number of the overall features of the communication impairments found in our study – such as general difficulties of emotional processing – are often observed in a variety of neurodegenerative conditions (e.g., Alzheimer’s disease and Parkinson’s disease^{37–40}), some of the more detailed aspects are more likely to be specific to HD. In particular, in HD’s case the difficulty in reading or understanding situations during communication is likely to be a direct consequence of the inability to recognise emotions efficiently, which is known to be characteristically part of the cognitive manifestations of the disease.⁷ Moreover,

given the loss of control on body movements that is characteristic to HD's symptomatology, it could be hypothesised that the particular emphasis on the theme of control, mentioned by the participants, might represent a way to cope with the threats posed by this condition in particular.

Limitations and future directions

A number of limitations should be considered with our results, such as the inclusion of people at the early to moderate stages only, In this study the SRM was adopted as a general theoretical framework to interpret the findings, since it has produced meaningful conclusions in previous studies with people affected by HD.^{26,27} However, other theoretical models could also be useful – such as Sharpe and Curran's hierarchical model on world and self views³⁶ – and future studies could take advantage from their adoption. It should also be remembered that communication is only one aspect of a number of challenges which these participants were managing. Seeing this in the context of their more general illness experience is also important theoretically. Future research should also aim to find ways to investigate the personal experiences of people at later stages of disease, using more adapted communication methods such as LiteWriters™.

Conclusion and implications for clinical practice

Our study has helped shed new light on multiple factors that have the potential of informing clinical communicative strategies between clinicians, caregivers, and patients with HD. First, our results suggest that patients could be encouraged to develop an identity which is less focused on the belief that the biomedical manifestations of HD are responsible for all the difficulties that they

experience. Stigmatising language (e.g., 'the Huntington's face') should be avoided. Secondly, patients should be allowed to regain actively some control over various features of communication, by, for example, having a choice on avoiding phone conversions, or managing their own energy in a way which is not then pathologised.

Finally, more effort should be put into helping patients reach a better level of emotion regulation. This could be achieved through the combination of a wide number of factors, including the review of medication regimes, the promotion of close relationships, the refinement of current coping strategies (such as sheltering), and their inclusion in new therapeutic interventions.

Declaration of Conflict of Interests

The authors declare that there is no conflict of interest.

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Tables

Table 1

Participants demographic data.

Participant	Gender	Age (yrs)	Diagnosis (yrs)	HD Stage
1	F	53	5	Moderate
2	F	53	7	Early
3	M	53	9	Moderate
4	M	41	5	Moderate
5	F	50	5	Early
6	F	62	5	Early
7	M	57	8	Early
8	F	49	6	Moderate

Table 2

Final list of identified themes and breakdown of relative codes.

Theme	Codes	
“You ever wanna say thing to people, but you can’t”: how HD directs and mediates communication	Articulation is harder	Not understanding what is going on
	Chronic fatigue makes communication harder	Not willing to go out
	Fear of saying or doing something wrong	Public places are uncomfortable
	Interrupting or jumping into a conversation	Speaking is harder
	Memory has changed	The HD face
	Not into communication anymore	Writing is harder
Becoming a director again: regaining control to improve communication	Being open about HD helps communication	New or unexpected things are harder
	Communication with familiar people is easier	New technologies can be helpful
	Communication with more than one person is harder	Not using the phone anymore
	Communication with strangers is harder	Texting helps communication
	Getting used to new people helps communication	Physical contact and proximity are harder
	Listening rather than speaking	Retiring from work is helpful
“One moment I’m fine, the next moment I’m not”: emotional outflows into communication and the struggle for separation	Acceptance helps with emotions	Having a close family is helpful
	Emotions have changed	Having relatives or friends around is helpful
	Feeling discriminated	Medications help with emotions
	Feeling ignored	
	Feeling lonely	
	Feeling misunderstood	
“I go back into my little world”: sheltering as a way to boost confidence in communication	Home is a safe place	My little world
	My little bubble	

Chapter 5

Publishable Paper Three (PP3)

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Exploring emotion regulation and emotion recognition in people with presymptomatic Huntington's disease: the role of emotional awareness

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Abstract

Interest in the role of both emotion regulation and recognition in our understanding of mental health has been steadily increasing, especially in people with chronic illness who also have psychological difficulties. One illness which belongs to this category is Huntington's disease. Huntington's disease (HD) is a chronic neurodegenerative disorder that can cause a number of cognitive and psychological difficulties, including emotion recognition deficits, even before the onset of the symptoms required to make a formal diagnosis. Despite the lack of definite evidence, recent studies have suggested that deficits of emotion regulation and recognition may be expected to play a pivotal role in the early cognitive manifestations of HD.

In this study, we hypothesised that the ability to regulate emotions can be impaired in people with presymptomatic HD, and that such impairment may be associated with a deficit of emotion recognition. To test this, an online survey was carried out with 117 English and Italian-speaking people with presymptomatic HD, compared to 217 controls matched for age and education.

The results suggest that, in presymptomatic participants, emotion regulation and emotion recognition are generally not significantly impaired, and no significant relationships between performances on the two constructs were observed. However, a specific impairment in emotional awareness (a subscale on the Difficulties in Emotion Regulation Scale, DERS) was observed, which appears to be enhanced by the co-occurrence of depressive symptoms, even at a subclinical level. Consequently, it is suggested that difficulties in emotional awareness may

represent a precursor of more general emotion recognition impairments, which only become apparent as the disease reaches a more symptomatic level. Clinical implications of the findings are discussed and directions for future research are proposed.

Introduction

Emotion regulation

Emotion regulation is defined as the process of managing the emotions that one has, as well as when and how one experiences or expresses them (Gross, 1998; Mayer, 2001). It involves both negative and positive emotions and its successful operation for good mental health has been widely recognised, with a substantial increase in the number of empirical investigations addressing this broad research area in the last two decades (e.g., Eftekhari, Zoellner, & Vigil, 2009; Gross, 2013; Gross & Muñoz, 1995). The most popular framework that has been developed to explain its functioning is the Process Model of Emotion Regulation (Gross, 1998), which identifies five fundamental families of regulatory emotional processes: a) *Situation selection*, i.e. taking necessary actions to approach or avoid situations potentially involving emotional responses; b) *situation modification*, i.e. modifying a situation in order to affect its emotional impact; c) *attentional deployment*, i.e. deploying or distracting attention in a situation to alter the emotional response; d) *cognitive change*, i.e. changing point of view or perspective on a situation in order to change the emotional response; e) *response modulation*, i.e. taking direct action on managing the behavioural and physical components of the emotional response. The latter two processes are responsible for the two main strategic outcomes of emotion regulation: *reappraisal*, which originates from cognitive change and involves actively rethinking a situation to alter the emotional response, and *emotion suppression*, which belongs to the response modulation family and promotes the decrease of emotion expression (Gross, 2013). While

reappraisal is a cognitively-oriented strategy that has proven to be particularly beneficial for the regulation of negative emotions and the promotion of positive experiences, the evidence on emotion suppression (which, by contrast, is a behaviourally-oriented) shows limited benefits and suggests the potential for mainly detrimental effects such as increased negative experiences, and memory difficulties (for a meta-analysis, see Webb et al., 2012).

Regardless of their specific efficacy, however, the successful implementation of emotion regulation strategies is based on the accurate functioning of a number of fundamental physiological, behavioural, and cognitive mechanisms that are known to be involved in emotional processing and response (Mauss et al., 2005). These include the ability to recognise emotions in other people, which represents a key cognitive and social skill and whose impairment is likely to cause emotional dysregulations. This is especially true in clinical conditions where emotion recognition is frequently impaired, such as in neurodegenerative disorders (Löffler et al., 2015).

Emotional difficulties in neurodegenerative disorders: the case of Huntington's disease

Huntington's disease (HD) is a hereditary progressive neurodegenerative disorder which affects 5-10 people per 100,000 in the Caucasian population (Roos, 2010). Typical symptoms include involuntary movements (chorea), involuntary abnormal postures (dystonia), cognitive deterioration (dementia) and significant psychological problems (Novak and Tabrizi, 2005). Since the transmission mechanism is autosomal-dominant, every affected individual has a

50% probability of transmitting it to their children. The estimate range of age of onset is 40-50 years, and the mean life expectancy after the diagnosis is typically 20 years (Folstein, 1989). Genetic testing is available for individuals at risk, allowing them to know if they carry the mutated gene. Individuals with this gene mutation will develop the disease at a certain time in their life, and are defined as 'presymptomatic'. The full diagnosis of Huntington's disease is based on the development of motor symptoms along with a familiar history (i.e., proof of an affected parent). Cognitive and psychological changes, including emotional problems, do not have to be present for a diagnosis to be made, even though they can arise much earlier than motor impairment, thus still affecting 'presymptomatic' individuals.

An impairment in the ability to recognise emotions has been widely noted in empirical studies in people with symptomatic HD, with both cross-sectional and longitudinal investigations consistently reporting evidence of early deterioration in the facial recognition of negative emotions, more specifically anger, fear, and disgust (e.g., Bates et al., 2014; Dumas et al., 2013; Henley et al., 2012; Johnson et al., 2007; Robotham et al., 2011). Evidence in people with presymptomatic HD for the same set of deficits is more sparse and contrasting, with some within subjects studies reporting specific impairments for disgust and negative emotions and others reporting no significant impairment at all (for a systematic review, see Henley et al., 2012). On the other hand, very little is currently known about emotion regulation in people with neurodegenerative disorders, and HD in particular. For example, a recent review (Löffler et al., 2015) was able to retrieve only one study, which found no differences between people with symptomatic HD

and controls in the self-reported usage of emotion suppression and reappraisal (Croft et al., 2014). Moreover, no studies were retrieved on emotion regulation in people with presymptomatic HD, and to our knowledge none has ever been carried out.

However, as previously anticipated, emotion recognition plays a pivotal role in the successful implementation of emotion regulation, and impairments in emotion recognition are likely to contribute to the development of emotion regulation difficulties, especially in clinical populations (Cecchetto et al., 2014; Gray and Tickle-Degnen, 2010; Löffler et al., 2015). In particular, according to emotional intelligence theory (Mayer, 2001; Salovey and Mayer, 1989), emotion regulation can only occur after emotions have been recognised (Izard et al., 2001; Yoo et al., 2006). With specific reference to the abovementioned Process Model, the inability to recognise emotions effectively seems likely to impair the regulatory processes that are based on the accurate emotional assessment of people and situations. Such processes could include situation selection (e.g., not being able to recognise those that are potentially negative), attentional deployment (e.g., not recognising relevant emotional cues), and cognitive change (e.g., basing change on misrecognised emotions). This is further corroborated by current evidence of a significant correlation between deficits of emotion recognition and emotion regulation difficulties in other clinical populations, such as people with anorexia nervosa (Harrison, Sullivan, Tchanturia, & Treasure, 2009), bulimia (Harrison et al., 2010), and borderline personality disorder (Domes et al., 2009). Moreover, recent evidence on anorexia nervosa has also shown associations between specific components of emotion regulation and

psychological difficulties such as depression and anxiety (Racine and Wildes, 2013). Thus, in the particular case of HD, it would be expected that emotion regulation and emotion recognition could play a major role in the early cognitive and psychological difficulties that occur prior to the formal diagnosis. This in turn represents the main argument of a currently ongoing debate on whether more comprehensive diagnostic criteria should be considered for the disease (Loy and McCusker, 2013; Paulsen, 2011; Reilmann et al., 2014).

Consequently, considering the evidence of potential early emotion recognition deficits in presymptomatic people (Henley et al., 2012) as well as the link between emotion recognition deficits and regulation impairments observed in other clinical populations (Harrison et al., 2010; Harrison et al., 2009; Racine and Wildes, 2013), the overarching aim of this study was to investigate the hypothesis that emotion regulation abilities can be impaired in presymptomatic HD gene carriers, and that such impairment may manifest in association with a deficit of emotion recognition. More specifically, the following hypotheses were formulated for this study: a) People with presymptomatic HD were predicted to report significantly more emotion regulation difficulties when compared to controls; b) performance on emotion recognition was predicted to be worse in people with presymptomatic HD when compared to controls; c) a significant positive relationship was predicted between difficulties in emotion recognition and emotion regulation, with more difficulties in emotional recognition correlating with more difficulties in emotional regulation. Moreover, in order to make sure to explore possible associations between emotion regulation and

recognition with psychological difficulties, depression and anxiety were also measured.

Materials and Methods

Design

This study adopted an online survey to explore emotion regulation and recognition in people with presymptomatic Huntington's disease with age-matched (non-affected) controls. The survey was developed with the Qualtrics® software, and included measures for emotion regulation, emotion recognition, as well as anxiety and depression. Both English and Italian versions of the survey were developed. The Italian version was developed via an ongoing collaboration between the Division of Health Research (DHR) at Lancaster University and the Italian League for Research on Huntington and Related Disease (LIRH Foundation) at the Mendel Institute of Human Genetics in Rome. The aim was to expand the sample size and was possible due to the availability of standardised translations of all the included measures that report the same validity as the English version. Separate links were generated to facilitate the dissemination among the target populations.

Participants

In total, 334 participants took part in the present study. The power calculation showed that, assuming a small effect size ($d = .2$), a minimum sample of 188 participants (99 for each group) was required to achieve a minimum statistical power of .8 using a probability value of 0.05.. The first group (Pre-HD)

consisted of 117 people with presymptomatic HD, of which 83 were English-speakers and 34 Italian-speakers. The second group (Ctrl) consisted of 217 age-matched controls, of which were 69 English-speakers and 148 Italian-speakers. An initial self-report question was included for the Pre-HD versions of the survey in order to control the participants' presymptomatic status. The presymptomatic participants and the controls did not present any significant differences in terms of age [$t(305.767) = 1.789, p = ns$], years in full-time education [$t(189.749) = -1.864, p = ns$], and gender [$\chi^2(2, N = 334) = 1.606, p = ns$]. See Table 1 for the full demographic details of the participants.

Table 1

Demographics of the participants.

	Pre-HD		Ctrl	
	N	Mean (SD) Min-Max	N	Mean (SD) Min-Max
Language (EN/IT)	83/34		69/148	
Gender (M/F)	35/82		59/158	
Age (yrs)	117	37.38 (11.06) 19-70	217	40 (15.39) 18-74
Education (yrs)	117	14.49 (2.77) 11-21	217	13.94 (2.11) 11-17
Test-time (yrs)	117	5.09 (5.34) 0-30		

Note. Ctrl = control group; F = female; M = male; Min-Max = minimum-maximum value; N = count; Pre-HD = presymptomatic group; SD = standard deviation; yrs = years.

The participants were enrolled across the UK and other English-speaking countries (e.g., USA, Canada, Australia and New Zealand), as well as Italy and San Marino via social media and with the help of local and international Huntington's disease associations. All the participants reported to be native speakers of the respective languages.

Measures

Emotion recognition measures

Reading the Mind in the Eyes test

The Reading the Mind in the Eyes test (RME; Baron-Cohen et al., 2001) is a test consisting of 36 still pictures of the eye regions within faces expressing different emotional states. The participant is asked to match a list of provided emotional tags to the emotions displayed in the pictures. The test yields a total score out of 36, and higher scores equal higher recognition performance. The RME is used worldwide and has been adopted with many clinical conditions, including schizophrenia (Kettle et al., 2008), autism (Baron-Cohen et al., 2001) and anorexia nervosa (Harrison et al., 2009), as well as Huntington's disease (Allain et al., 2011). It has previously shown acceptable construct validity when compared to other emotion recognition tasks (Alaerts et al., 2011), as well as acceptable internal consistency (Cronbach's $\alpha = .63$ for men, $.60$ for women; Voracek and Dressler, 2006). The Italian translation by Vellante and colleagues (2013) was used for the Italian version of the survey, which has shown good construct (discriminant and convergent) validity and acceptable internal consistency (Cronbach's $\alpha = .60$).

Emotion regulation measures

Difficulties in Emotion Regulation Scale

The Difficulties in Emotion Regulation Scale (DERS; Gratz & Roemer, 2004) is a validated self-report questionnaire consisting of 36 items rating emotion regulation on a 5-point Likert scale. It includes 6 subscales: non-acceptance of emotional responses (NONACCEPT), difficulties engaging in goal

directed behaviour (GOALS), impulse control difficulties (IMPULSE), lack of emotional awareness (AWARE), limited access to emotion regulation strategies (STRATEGIES), lack of emotional clarity (CLARITY). It yields a subscore for each subscale, as well as total score (SUM) out of 180. As the focus of the test is on difficulties, higher scores equal poorer emotion regulation. To our knowledge, the DERS has never been adopted with HD; however, it has been used with several other clinical populations, including participants with both psychological (Fowler et al., 2014) and physical conditions (Kököneyi, Urbán, Reinhardt, Józán, & Demetrovics, 2014). The DERS has previously shown good construct validity (Kököneyi et al., 2014), even when adopted across different cultural and ethnical groups (Ritschel et al., 2015). It also showed very good internal consistency, with a Cronbach's α of .93 for the total score (SUM), and figures ranging between .80 and .89 for the subscales (Gratz and Roemer, 2004). The Italian validation by Sighinolfi and colleagues (2010) was adopted for the Italian version of the survey, which has showed psychometric properties comparable to the English version.

Mood and anxiety issues measures

Hospital Anxiety and Depression Scale

The Hospital Anxiety and Depression Scale (HADS; Zigmond & Snaith, 1983) is a validated self-report questionnaire consisting of 14 items rating anxiety and depression symptoms on a 3-point scale. No unified score is provided at the end of the test. Instead, individual scores on a scale out of 21 are provided for anxiety and depression. The HADS represents one of the most adopted measures in clinical populations and it has been specifically validated with people affected

by HD (De Souza et al., 2010). A review of its psychometric properties (Bjelland et al., 2002) reported good construct validity when compared to other common clinical measures, as well as good internal consistency (mean Cronbach's $\alpha = .83$ for anxiety, $.82$ for depression). The same study identified a recommended cut-off point of 8/21 to yield both good sensitivity (anxiety = $.90$; depression = $.83$) and specificity (anxiety = $.78$; depression = $.79$). The HADS was incorporated in this study to control for the potential confounding effect of depression and anxiety levels on the ability to regulate and recognise emotions. For the Italian version of the survey, the validation by Costantini and colleagues was adopted (1999), which showed comparable high construct validity and internal consistency (Cronbach's $\alpha = .89$ and $.88$ for anxiety and depression respectively).

Statistical analysis

Data were analysed with IBM SPSS Statistics® programme v23 (Armonk, NY: IBM Corp). Independent-sample t-tests and one-way ANOVAs were performed to compare means across the participants groups. Considering the number of repeated comparisons, in order to control for family-wise error-rate (FWER) the Bonferroni correction was applied and the significance level was adjusted from $.05$ to $.005$. The relationship between the participants' demographic and clinical characteristics and the outcome variables (i.e., emotion regulation, emotion recognition, depression and anxiety) was investigated through Pearson's correlations (two-tailed).

Ethics approval

This study was reviewed and approved by the Faculty of Health and Medicine Research Ethics Committee at Lancaster University (ref: FHM REC16015).

Results

Despite the differences in language and culture between English and Italian-speaking participants, a whole pool of presymptomatic individuals (Pre-HD) and a whole pool of controls (Ctrl) were considered for the purpose of the analysis. This decision was taken on the basis of the widely confirmed observation in the literature that the expression of emotions is a universal ability which is not affected by language or culture (Ekman, 1993; Ekman & Cordaro, 2011; Ekman & Friesen, 1986; Fritz et al., 2009; Keltner, Ekman, Gonzaga, & Beer, 2003; Wells, Johnson, Ekman, Sorenson, & Friesen, 1969). Therefore, the implicit assumption of the present study was that English and Italian-speaking participants would not differ significantly regarding the constructs evaluated by the adopted measures.

The mean scores of the participants of the Pre-HD and Ctrl groups on the outcome variables are shown in Table 2. Based on the recommended cut-off values of skewness and kurtosis (West et al., 1995), the scores were normally distributed. Of the participants of Pre-HD group, 48.7% and 23.1% showed clinical levels of anxiety and depression respectively, by scoring above the recommended clinical cut-off for the HADS (i.e., 8/21; Bjelland et al., 2002). The participants of the Ctrl group showed similar figures, with 46.5% and 23.5% above the cut-off point. In

terms of levels of emotion regulation difficulties, the Ctrl group showed total scores (SUM) comparable to the reported data with general adult populations (e.g., 77/180; Ritschel et al., 2015), while the Pre-HD group scored marginally higher, meaning that more emotion regulation difficulties were reported. The mean emotion recognition performance of both groups was very similar and slightly below the reported normative data for general adult populations (e.g., 26/36; Baron-Cohen et al., 2001). In terms of measure reliability, high levels were shown by the HADS (Cronbach's $\alpha = .84/.78$ for anxiety/depression) and the DERS (Cronbach's $\alpha = .94$ for the SUM score, .83 to .89 for the subscales). The RME showed a level of reliability (Cronbach's $\alpha = .55$) comparable to previously accepted figures in the literature (Voracek and Dressler, 2006).

The group comparison showed that the Pre-HD and Ctrl groups did not differ significantly in terms of total score of emotion regulation difficulties (DERS SUM; [$F(1, 332) = 1.939, p = ns$]), emotion recognition (RME; [$F(1, 332) = 1.291, p = ns$]), as well as anxiety (HADS-A; [$F(1, 332) = 1.472, p = ns$]) and depression (HADS-D; [$F(1, 332) = .393, p = ns$]). When comparing the subscales of the DERS, significant differences were observed only on the AWARE score [$F(1, 332) = 9.359, p = .002$]. Effect size analysis indicated no relevant effects of any of the other variables, except for a small effect of the IMPULSE score ($d = .220$). The full details of the results of the comparisons are shown in Table 2.

In addition, no significant correlation was found between the total level of emotion regulation difficulties (DERS-SUM) and emotion recognition performance (RME) in the overall presymptomatic group ($r = -.030, N = 117, p = ns$). Similarly, no

significant correlation was observed between the same constructs when assessed separately for participants' language, within either the presymptomatic (English: $r = -.114, N = 83, p = ns$; Italian $r = -.077, N = 34, p = ns$). Table 3, 4, and 5 show the full details of Pearson's correlation coefficients across all the variables for the overall, English, and Italian Pre-HD participants' scores respectively.

Table 2

Participants' scores across the outcome variables.

		Pre-HD	Ctrl	Between-group comparison			Reliability
		Mean (SD)	Mean (SD)	<i>F</i>	<i>p</i>	<i>d</i>	α
HADS	HADS-A	7.85 (4.78)	7.24 (4.16)	1.472	ns	.136	.84
	HADS-D	4.77 (3.911)	5.04 (3.61)	.393	ns	-.071	.78
	SUM	80.91 (27.59)	76.97 (22.93)	1.939	ns	.155	.94
DERS	NONACCEPT	12.98 (5.59)	13.26 (5.83)	.174	ns	-.049	.87
	GOALS	13.12 (5.30)	12.95 (5.05)	.079	ns	.032	.87
	IMPULSE	11.93 (5.33)	10.86 (4.35)	.946	ns	.220	.85
	AWARE	15.77 (5.91)	13.83 (5.31)	9.359	.002	.345	.83
	STRATEGIES	16.90 (7.54)	16.42 (6.79)	.348	ns	.066	.89
	CLARITY	10.21 (4.28)	9.65 (4.14)	.1335	ns	.132	.84
RME	Total Score	24.07 (3.863)	24.58 (3.97)	1.291	ns	-.130	.55

Note. Clinical cut-off for the HADS: 8/21. Significance level = .005; α = Cronbach's alpha; AWARE = lack of emotional awareness; CLARITY = lack of emotional clarity; Ctrl = control group; DERS = Difficulties in Emotion Regulation Scale; *F* = ANOVA *F* value; GOALS = difficulties engaging in goal directed behaviour; HADS-A = HADS anxiety score; HADS-D = HADS depression score; IMPULSE = impulse control difficulties; NONACCEPT = non-acceptance of emotional responses; Pre-HD = presymptomatic group; RME = Reading the Mind in the Eyes test; SD = standard deviation; Sig. = significance; Size = effect size; SUM = DERS total score.

Table 3

Correlation coefficients for pre-HD participants' scores (overall; $N = 117$).

	1	2	3	4	5	6	7	8	9	10	11	12	13	14
1. Gender														
2. Age	-.157													
3. Education	-.080	-.001												
4. Test	-.036	.286**	.057											
5. HADS-A	.157	-.098	.030	-.130										
6. HADS-D	-.002	-.006	-.093	-.146	.669**									
7. DERS SUM	.104	-.065	.055	-.133	.758**	.760**								
8. DERS NONACCEPT	.141	-.006	.067	-.070	.572**	.501**	.796**							
9. DERS GOALS	.147	-.040	.117	-.094	.667**	.628**	.826**	.570**						
10. DERS IMPULSE	.110	.019	.072	-.131	.618**	.580**	.803**	.561**	.619**					
11. DERS AWARE	-.055	-.072	-.085	-.148	.400**	.513**	.662**	.423**	.384**	.391**				
12. DERS STRATEGIES	.115	-.074	.093	-.057	.764**	.745**	.925**	.709**	.786**	.762**	.434**			
13. DERS CLARITY	.040	-.153	-.016	-.181	.643**	.727**	.841**	.587**	.655**	.547**	.606**	.751**		
14. RME	.153	-.026	.194*	.012	-.058	-.184*	-.030	.061	.058	-.058	-.155	-.018	-.030	

Note. * = $p < .05$; ** = $p < .01$; AWARE = lack of emotional awareness; CLARITY = lack of emotional clarity; DERS = Difficulties in Emotion Regulation Scale; GOALS = difficulties engaging in goal directed behaviour; HADS-A = HADS anxiety score; HADS-D = HADS depression score; IMPULSE = impulse control difficulties; NONACCEPT = non-acceptance of emotional responses; RME = Reading the Mind in the Eyes test; SUM = DERS total score; Test = time since predictive test.

Table 4

Correlation coefficients for pre-HD participants' scores (English; N = 83).

	1	2	3	4	5	6	7	8	9	10	11	12	13	14
1. Gender														
2. Age	-.118													
3. Education	-.096	-.081												
4. Test	-.074	.251**	.107											
5. HADS-A	.222*	-.137	-.102	-.211										
6. HADS-D	-.075	-.033	-.262*	-.196	.634**									
7. DERS SUM	.152	-.135	-.087	-.180	.725**	.789**								
8. DERS NONACCEPT	.178	-.074	.039	-.051	.612**	.577**	.821**							
9. DERS GOALS	.244*	-.087	.029	-.178	.567**	.579**	.799**	.579**						
10. DERS IMPULSE	.134	.043	.038	-.205	.589**	.614**	.785**	.582**	.589**					
11. DERS AWARE	-.093	-.086	-.220*	-.194	.349**	.540**	.652**	.451**	.302**	.356**				
12. DERS STRATEGIES	.194	-.164	-.011	-.079	.750**	.764**	.924**	.732**	.770**	.735**	.429**			
13. DERS CLARITY	.067	-.139	-.166	-.205	.604**	.724**	.837**	.592**	.622**	.522**	.621**	.738**		
14. RME	.091	-.198	.241*	.084	-.111	-.227*	-.114	.008	.015	-.116	-.261*	-.068	-.112	

Note. * = $p < .05$; ** = $p < .01$; AWARE = lack of emotional awareness; CLARITY = lack of emotional clarity; DERS = Difficulties in Emotion Regulation Scale; GOALS = difficulties engaging in goal directed behaviour; HADS-A = HADS anxiety score; HADS-D = HADS depression score; IMPULSE = impulse control difficulties; NONACCEPT = non-acceptance of emotional responses; RME = Reading the Mind in the Eyes test; SUM = DERS total score; Test = time since predictive test.

Table 5

Correlation coefficients for pre-HD participants' scores (Italian; N = 34).

	1	2	3	4	5	6	7	8	9	10	11	12	13	14
1. Gender														
2. Age	-.242													
3. Education	-.091	.175												
4. Test	.051	.394*	.111											
5. HADS-A	.061	-.022	-.148	-.063										
6. HADS-D	-.323	-.083	-.256	-.002	.695**									
7. DERS SUM	.109	-.136	.225	-.069	.718**	.507**								
8. DERS NONACCEPT	.009	-.182	.202	-.199	.250	.981**	.631**							
9. DERS GOALS	.222	-.093	.106	-.139	.846**	.697**	.790**	.327**						
10. DERS IMPULSE	.015	.171	.200	-.046	.598**	.352**	.851**	.394*	.635**					
11. DERS AWARE	-.037	-.053	-.074*	-.055	.282	.176	.507**	.104	.340*	.336				
12. DERS STRATEGIES	-.178	.193	-.192	-.053	.673**	.532**	.877**	.502**	.694**	.814**	.148			
13. DERS CLARITY	-.104	-.053	-.215	-.181	.617**	.633**	.779**	.420*	.600**	.508**	.366**	.682**		
14. RME	.299	-.298	-.064	-.310	-.226	-.312	-.077	-.112	-.195	-.074	-.046	-.154	-.020	

Note. * = $p < .05$; ** = $p < .01$; AWARE = lack of emotional awareness; CLARITY = lack of emotional clarity; DERS = Difficulties in Emotion Regulation Scale; GOALS = difficulties engaging in goal directed behaviour; HADS-A = HADS anxiety score; HADS-D = HADS depression score; IMPULSE = impulse control difficulties; NONACCEPT = non-acceptance of emotional responses; RME = Reading the Mind in the Eyes test; SUM = DERS total score; Test = time since predictive test.

Table 6

Standardised coefficients for the multiple regression models of the AWARE subscore.

	Pre-HD		Ctrl	
	β	p	β	p
HADS-A	.128	ns	.129	ns
HADS-D	.669	.000	.221	ns

Note. AWARE = lack of emotional awareness; β = standardised coefficient; Ctrl = control group; HADS-A = HADS anxiety score; HADS-D = HADS depression score; p = significance; Pre-HD = presymptomatic group.

Despite not being initially hypothesised but in order to explore this result further, multiple regressions were conducted to assess the contribution of anxiety and depression to the AWARE subscale, as it was the only one to show a significant difference across the groups. The standardised regression coefficients are summarised in Table 6. Using the enter method, it was found that the model (which consisted of two variables) explained a significant amount of the variance of the AWARE score in both the Pre-HD ($F(2, 114) = 20.947, p < .001, R^2 = .269, R^2_{Adjusted} = .256$) and Ctrl group ($F(2, 214) = 5.623, p = .004, R^2 = .050, R^2_{Adjusted} = .041$). The analysis in the Pre-HD group showed that the AWARE score was not significantly predicted by anxiety ($\beta = .128, p = ns$), but it was significantly predicted by depression ($\beta = .669, p < .001$). On the other hand, in the Ctrl group neither anxiety ($\beta = .129, p = ns$) nor depression ($\beta = .221, p = ns$) significantly predicted the AWARE score.

Discussion

To our knowledge, the present study has been the first to investigate emotion regulation abilities in a population of participants with presymptomatic Huntington's disease (HD) by comparing these with those of age-matched

controls. In addition, it has also been the first to explore the relationship between emotion regulation and emotion recognition, as well the potential association with depression and anxiety, in this type of population. The results showed no significant differences between the two groups in terms of general difficulties in regulating emotions and performance on an emotion recognition task. Moreover, no significant correlation was found between emotion regulation and emotion recognition scores in both groups. As a consequence, none of our initial hypotheses was confirmed by the results, suggesting that the ability both to regulate and recognise emotions does not deteriorate early in non-symptomatic carriers of the gene Huntington's disease, and these two abilities are not significantly correlated. This latter result appears to be in contradiction with previous findings in other clinical populations. However, it may be explained by the specific measure of emotion regulation used in this study as the DERS focuses specifically on difficulties rather than other components such as the use regulatory strategies. Thus, the lack of correlation in this study may be at least partially due to the specific adoption of the DERS, and the way it relates with the RME.

When analysing more specific components of emotion regulation, a significant difference was found between the two groups on one subscale, with the presymptomatic participants reporting significantly greater lack of emotional awareness (DERS-AWARE), i.e. difficulties in the ability to attend to and acknowledge emotions. A possible explanation of this could lie in the development of some of HD's early cognitive symptoms. Indeed, general difficulties of awareness and emotional processing have been linked to poorer functioning of prefrontal brain regions such as the anterior insular (AIC) and anterior cingulate

(ACC) cortices (Craig, 2009; Lane et al., 1998; Phillips, Drevets, Rauch, & Lane, 2003), which are known to be affected by HD (Dogan et al., 2014; Gray et al., 2013) and are often responsible for a wide range of emotional difficulties in both symptomatic (Craufurd et al., 2001; Ho et al., 2006; Hoth et al., 2007; Mörkl et al., 2016) and presymptomatic patients (Kipps et al., 2007; Klöppel et al., 2010). Therefore, the difference observed for the presymptomatic participants on the AWARENESS subscale of the DERS may represent an expression of the many subtle biological and cognitive changes that presymptomatic individuals can experience before a formal diagnosis of HD is made based on motor manifestations (e.g., Tabrizi et al., 2011).

In particular, it could be interpreted in light of the early development of the well-established impairments in emotion recognition in people with symptomatic HD. This appears to be supported by multiple evidence of a significant predictive role played by alexithymia (a condition characterised by pervasive deficits of emotional awareness; Lane et al., 2000) in emotion recognition impairments both when it manifests alone (Lane et al., 2000, 1996), as well as when it is part of other clinical conditions, such as autism (Cook et al., 2013) and eating disorders (Brewer et al., 2015). Moreover, alexithymia measures often show high correlation levels with many of the DERS subscales, including the AWARE one (Ghorbani et al., 2017; Stasiewicz et al., 2012). In contrast, current evidence on alexithymia in HD is extremely limited, as a recent systematic review identified only one study that reported no significant impairment in symptomatic individuals (Ricciardi et al., 2015). Therefore, the significantly greater level of difficulties observed with the presymptomatic participants might represent a

precursory gateway to the development of the emotion recognition impairment found in fully symptomatic individuals, which in turn is likely to affect the other components of emotion regulation. This hypothesis requires further exploration, as it is currently uncertain whether emotional awareness difficulties may lead to a fully alexithymic condition in people with symptomatic HD. In addition, in this study the particular structure and sensitivity of the RME might have prevented the observation of any significant correlations between the specific AWARE subscale and emotion recognition performance (see Limitations section).

Moreover, a considerable contribution to poorer emotional awareness may come from social and environmental factors, and more specifically from the type of family context, as many gene carriers often grow up in contact with a parent affected by symptomatic HD. Indeed, current evidence suggests that the family environment and the emotional climate in which a child is raised are deeply related to the successful development of emotional processing skills, and especially emotion regulation (for a review, see Morris et al., 2007). In particular, parents' emotional responses to their children's emotions have been linked with emotional awareness (Eisenberg et al., 1998; Schultz et al., 2001; Sim et al., 2009). Thus, the fact of living in a family context characterised by challenging emotional responses due to close contact with a symptomatic parent may have hampered the successful learning of the ability to acknowledge emotions in the presymptomatic participants of this study, ultimately contributing to the development of a deficit of emotional awareness in particular.

Further insight was also provided by the results of the regression models, which showed that depression explained a considerable portion of variance on the AWARE subscale in the Pre-HD group only. This differential effect of depression on presymptomatic participants could again represent an expression of the early cognitive manifestations of the disease. In particular, this may limit patients' coping abilities and overall resilience in the face of depression, even when they have not reached clinical levels yet, in a way that is common to many neurodegenerative conditions (for a review, see Baquero, 2015). The predictive role of depression is also corroborated by findings from studies with clinically depressed individuals; these often show problems with many components of emotion regulation (Ehring et al., 2008; Loas et al., 1997), and in particular emotional awareness (Boden and Thompson, 2015). Thus, due to the concurrent development of psychological difficulties within the context of suboptimal psychological resilience due to HD's early cognitive symptoms, depression might show a disproportionate effect on presymptomatic people even at subclinical levels. This may contribute to a greater lack of emotional awareness, and eventually lead to the development of emotion recognition impairments which, as previously mentioned, may in turn affect the other components of emotion regulation, thus establishing a symptomatic vicious circle. Figure 1 shows a schematisation of the hypothesised relationship.

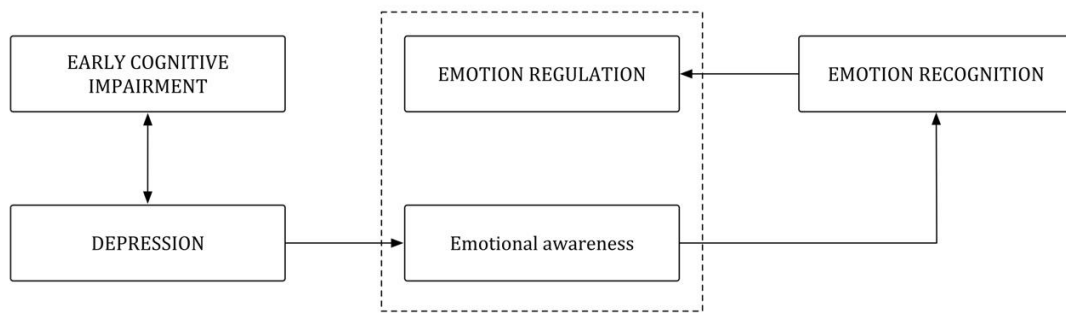


Figure 4: hypothesised relationship among the discussed constructs. Arrowed lines identify direct influence. Upper case identifies main constructs, lower case subcomponents. Dotted lines identify inclusion within the same construct.

Limitations and future directions

Despite allowing for a large sample, the online nature of this study carries the intrinsic limitation of lacking direct contact between the researcher and the participants. This includes the inability to obtain some important clinical details about the participants (e.g., pharmacological therapies). In addition, the responses to all questions were made mandatory to proceed throughout the survey. While this eliminated the need to control for missing data, it might have also limited the sample size due to participants dropping out before completing their participation. To control for this issue, the number of measures was limited to keep the overall time of the survey below 30 minutes. In this perspective, the RME was chosen in this study as its brevity fit particularly well with an online design. However, despite being widely regarded and utilised as an emotion recognition test (Guastella et al., 2010; Harrison et al., 2010; Harrison et al., 2009; Quintana et al., 2012; Vellante et al., 2013), the measure was originally created as a test of theory of mind to assess the recognition of mental states through eye expressions (Baron-Cohen et al., 1997). As a consequence, it may be possible that the RME was

not sensitive enough to detect subtle differences in emotion recognition in the specific population of this study, nor may it be able to show potential correlations with emotional awareness.

Future research should aim at adopting more diversified measures of emotion regulation and recognition in face to face studies. In particular, the adoption of emotion recognition tasks with more comprehensive stimuli based on both faces and body language – such as the recent Bochum Emotional Stimulus Set (BESST; Thoma, Soria Bauser, & Suchan, 2013) – may yield different results in terms of comparison of recognition performance and correlation between emotion recognition and emotional regulation. Moreover, more in-depth measures for emotional awareness and alexithymia should be used to investigate further the precursory role of emotional awareness difficulties hypothesised in this study, and further exploration is warranted on emotion regulation in people affected by HD with the adoption of measures focused on both difficulties and regulatory strategies.

All these suggestions could generally benefit from the inclusion in large-scale longitudinal clinical trials, which would allow an increase in our understanding of emotional processing in Huntington's disease over its full clinical course.

Conclusion

This online study offered some preliminary insight into emotion regulation in people with presymptomatic Huntington's, as well as further insight into emotion recognition. The findings suggest that presymptomatic individuals show a wide range of normal abilities, as emotion regulation and emotion

recognition were not significantly impaired when compared to controls, nor did the shared significant relationships with one another. However, one specific emotion regulation component, emotional awareness, was significantly impaired. This could be due to HD's early cognitive manifestations, and a catalytic role may be played by their co-occurrence with psychological difficulties such as depression, even at a subclinical level. Moreover, the greater level of difficulties in emotional awareness shown by presymptomatic people may represent a precursor of the development of the emotion recognition impairments that are often observed in fully symptomatic individuals and that may in turn have a detrimental effect on the other components of emotion regulation.

These findings can have important implications for clinical practice, as a better management of depression could lead to increased levels of emotional awareness, better emotion recognition performance, eventually better emotion regulation, as well as everyday communication and quality of life. In addition, the possible precursor role of emotional awareness shows the potential to help amend the current diagnostic criteria by shedding new light on early cognitive difficulties in HD, as well as inform new therapeutic protocols and interventions tailored around the emotional and communicative needs of the people affected by this condition.

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Chapter 6

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New perspectives on emotional processing in people with symptomatic Huntington's disease: impaired emotion regulation and recognition of emotional body language

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New perspectives on emotional processing in people with symptomatic Huntington's disease: impaired emotion regulation and recognition of emotional body language

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Abstract

Emotion regulation and emotional body language (EBL) recognition represent two fundamental components of emotional processing that have recently seen a considerable surge in research interest, especially due to the role they play in optimal mental health. This appears to be particularly true for clinical conditions that can profoundly affect emotional functioning. Among these conditions is Huntington's disease (HD), a neurodegenerative disorder that is associated with several psychological difficulties and cognitive impairments, which include well-established deficits in facial emotion recognition. On the other hand, the current evidence in this population on other components such as emotion regulation and EBL recognition is still sparse, and it is unsure whether they are also affected by the condition.

In this study, it was hypothesised that emotion regulation and recognition of emotional body language are impaired in people with symptomatic HD, and that these impairments share a significant relationship with one another. Thus, a between-subjects design was adopted to compare 13 people with symptomatic HD with 12 non-affected controls matched for age and education.

The results corroborated the first hypothesis, that emotion regulation and EBL recognition were significantly impaired by HD. Moreover, a significant positive correlation was observed between facial and EBL recognition impairments, and the EBL performance was negatively related to the stage of the disease. However, emotion regulation and recognition performances did not share

a general relationship with one another. Clinical implications of the findings are provided, and indications for future research are proposed.

Introduction

In the past few decades psychological research into human emotions has seen a surge of interest, especially due to the comprehensive conceptualisation of constructs such as emotional intelligence. Emotional intelligence is defined as the set of cognitive processes that allows the accurate expression and appraisal of emotions in others and the self (Goleman, 1995; Salovey & Mayer, 1989). In particular, the identification, understanding, facilitation, and management of emotions have been recognised as the four fundamental areas required for the successful processing of emotions. Within this framework, a pivotal role in social and affective functioning is played by emotion recognition and emotion regulation (Ochsner, 2009).

Emotion recognition can be defined as the process of correctly perceiving and identifying emotions in other people, as well as in artificial representations such as drawings or music (Mayer, Caruso, & Salovey, 1999). Historically, the most researched medium of emotion recognition is whole facial expression, such as pictures of faces of actors expressing basic emotions such as anger or fear (Henley et al., 2012). However, emotion recognition is a process mediated by a number of different features other than facial clues, and recognition via eyes, voices, and body language have also been investigated (Baron-Cohen, Wheelwright, Hill, Raste, & Plumb, 2001; Beatrice de Gelder & Van den Stock, 2011; Lima, Castro, & Scott, 2013). The latter medium in particular is gaining increased attention, since

emotional body language (EBL) recognition has so far been neglected, despite being deeply involved in fundamental social cognitive skills such as empathy and decision-making (de Gelder, 2006; de Gelder & Hortensius, 2014).

On the other hand, emotion regulation is defined as the “processes by which individuals influence which emotions they have, when they have them, and how they experience and express these emotions” (Gross, 1998; p. 275). More specifically, it involves the processes of selecting and modifying potential emotional situations, deploying attention, changing one’s perspectives on emotions, and modulating emotional responses (Gross, 1998; 2015). In the last 20 years this area has seen a considerable increase in interest due to the recognition of its importance for psychological resilience and mental health (Aldao, Nolen-Hoeksema, & Schweizer, 2010; Ghorbani, Khosravani, Sharifi Bastan, & Jamaati Ardakani, 2017; Gross & Muñoz, 1995; van der Meer, van Duijn, Giltay, & Tibben, 2015).

Based on the theory of emotional intelligence, emotion recognition and emotion regulation are deeply interconnected processes, since emotions need to be correctly recognised before being regulated (Izard et al., 2001; Mayer, 2001; Salovey & Mayer, 1989; Yoo, Matsumoto, & LeRoux, 2006). This is supported by evidence on the neural bases of both processes, which involve similar subcortical structures such as the limbic system and the basal ganglia (Gross, 2013). Not surprisingly, deficits of emotional processing are observed in many neurodegenerative conditions that involve a damage to those structures, such as Parkinson’s disease, Alzheimer’s disease, multiple sclerosis, and – with a

particularly well-established impact on emotion recognition skills – Huntington’s disease (Löffler, Radke, Morawetz, & Derntl, 2015).

Huntington’s disease (HD) is a genetic neurodegenerative disorder whose typical symptoms include involuntary movements (chorea), cognitive deterioration (dementia), and considerable psychological problems (Novak & Tabrizi, 2005). Its prevalence in the Caucasian population is 5-10 people per 100,000 (Roos, 2010). The transmission mechanism is autosomal-dominant, meaning that affected individuals’ children have a 50% probability of inheriting the gene, and genetic testing is available to ascertain gene status (in which case the term ‘presymptomatic’ is used). The onset is usually around age 40, and disease progression can be divided into five stages, starting with mild motor symptoms, cognitive impairment and relative independent functioning (Stage I), and ending with a need for full-time care due to severe motor impairment and dementia (Stage V; Shoulson & Fahn, 1979).

One of HD’s most frequently observed cognitive impairments is a deficit of emotion recognition, which especially involves negative emotions such as anger, fear, and disgust (Bates, Tabrizi, & Jones, 2014). However, while the evidence on this set of impairments is well established, it has been traditionally investigated only through tests based on facial expressions, with very few studies based on different stimuli such as emotional body language (see Henley et al., 2012 for a review). In fact, to our knowledge only two studies have investigated EBL in HD, showing preliminary evidence that a deficit of EBL recognition can also be part of the manifestations of the disease (de Gelder et al., 2008), although it may not be

observed in presymptomatic individuals (Aviezer et al., 2009). In addition to the sparseness of evidence on EBL, very little is also known about emotion regulation in HD. Indeed, a recent review (Löffler et al., 2015) identified only one study that specifically addressed it, and that found no differences between people with symptomatic HD and controls (Croft, McKernan, Gray, Churchyard, & Georgiou-Karistianis, 2014). However, the measure adopted by this study – the Emotion Regulation Questionnaire (ERQ; Gross et al., 2003) – only assesses the use of two regulatory strategies (suppression and reappraisal), and does not allow for the exploration of any other specific components of emotion regulation, such as impulse control or emotional awareness.

As mentioned previously, emotion recognition (both facial and EBL) and emotion regulation are likely to influence each other (Ochsner, 2009), and play an essential role in the successful operation of social skills as well as psychological resilience (de Gelder & Hortensius, 2014; Ghorbani et al., 2017; Gross & Muñoz, 1995). With specific regard to HD, a deeper understanding of how the disease affects these cognitive components would allow for a refinement of current cognitive and behavioural approaches to care and treatment. Moreover, this carries the potential for shedding new light on the neural bases that characterise them and the relationship between cognition and neurobiology, in particular in relation to EBL recognition (de Gelder, 2006). Both these implications have in turn the potential of contributing to a currently ongoing debate which focuses on whether the current diagnostic criteria for HD, which are based on motor manifestations only, should include early signs of cognitive impairment. (Loy & McCusker, 2013; Paulsen, 2011; Reilmann, Leavitt, & Ross, 2014)

Consequently, the aim of this study was to investigate the assumption that emotion regulation and both facial and EBL recognition are impaired in a population of people affected by symptomatic Huntington's disease, and that such impairments may show a significant relationship with one another. In particular, the assumption was based on a comparison to non-affected age-matched controls with the use of more comprehensive tests of emotion recognition and regulation. More specifically, the following hypotheses were formulated: a) People with HD were predicted to report significantly more emotion regulation difficulties than the control group when assessed on a number of different emotion regulation components; b) emotion recognition was predicted to be significantly impaired in people with HD when compared to the controls on both facial and EBL tasks; c) a significant relationship was expected to be observed between emotion regulation difficulties and emotion recognition impairment. In addition, due to the evidence of relationships between psychological difficulties and emotional processing (Cisler, Olatunji, Feldner, & Forsyth, 2010; Joorman & Gotlib, 2010; Martin & Dahlen, 2005), depression and anxiety measures were also included.

Materials and Methods

Design and participants

This study adopted a 2-group between-subjects design with age-matched controls. In total, 25 participants took part, split across two groups (HD and Ctrl) consisting of 13 symptomatic individuals (four male, nine female) and 12 age-matched non-affected controls (five male, seven female). The sample size of the HD group was comparable to the majority of studies investigating emotion

recognition in HD that have been identified by a recent systematic review (i.e., six to 40; Henley et al., 2012). For the HD group, participation was limited to individuals in early to moderate stages of the disease (i.e., I-III). This was decided due to the difficulties in undertaking cognitive tasks that are likely to arise in the later stages of the condition. HD stage was screened through the Total Functional Capacity scale (TFC; Shoulson & Fahn, 1979). More specifically, one participant (7.7%) belonged to stage I, seven (53.8%) to stage II, and five (38.5%) to stage III. The two groups did not present any significant differences in terms of age [$t(23) = .490, p = ns$], education [$t(23) = -1.023, p = ns$], or gender [$\chi^2(1, N = 25) = .322, p = ns$]. See Table 1 for the full demographic details. The participants of the HD group were recruited across the North West of England with the help of the Regional Care Advisory Service of the Huntington's Disease Association (HDA). The participants of the Ctrl group were recruited from partners and caregivers of the participants of the HD group, in order to reflect similar demographic and social characteristics. This study was reviewed and approved by the Faculty of Health and Medicine Research Ethics Committee at Lancaster University (ref: FHMREC15043).

Table 1

Demographics of the participants.

	HD			Ctrl		
	Mean	SD	Range	Mean	SD	Range
Age (yrs)	53.46	5.109	42-63	52.17	7.907	33-63
Education (yrs)	12.92	2.66	11-18	14	2.594	11-18
Diagnosis time (yrs)	5.54	1.713	3-9			
TFC score	6.92	2.139	13-0			

Note. Ctrl = control group; HD = Huntington's disease group; SD = standard deviation; TFC = Total Functional Capacity; yrs = years.

Measures

Total Functional Capacity Scale (Shoulson & Fahn, 1979):

The TFC is a standardised tool that assesses everyday functional capacities such as working, handling money, taking care of domestic chores, performing self-care tasks, and living independently. It is part of the larger Unified Huntington's Disease Rating Scale (UHDRS; Huntington Study Group, 1996). The total score ranges from 13 (normal capacity) to 0 (severe disability) and its intervals can be used to determine the stage of the disease: 13-11 = Stage I, 10-7 = Stage II, 6-3 = Stage III, 2-1 = Stage IV, 0 = Stage V. The TFC is characterised by excellent internal consistency (Cronbach's $\alpha = .95$) as well as high interrater reliability (Huntington Study Group, 1996).

Bochum Emotional Stimulus Set (BESST; Thoma, Soria Bauser, & Suchan, 2013):

The BESST is a validated set of 4490 emotional stimuli consisting of pictures of both male and female facial expressions and emotional body language (EBL). It investigates the recognition of six emotions (fear, disgust, happiness, sadness, surprise and anger) plus neutral expressions. The facial expressions are computer-generated, while the EBL stimuli are based on photographs of actors and actresses. The stimuli feature multiple ethnic groups, and the option to include averted pictures to allow for increased complexity. For this study, 10 frontal stimuli from the BESST were randomly selected for each emotion and each expression modality (facial or EBL), half male and half female, to a total of 140 stimuli for two trials (70 + 70). Thus, the test in this study yielded a total score out of 70 for each modality, as well as a subscore out of 10 for each emotion. The BESST

features excellent norms (Abramson, Marom, Petranker, & Aviezer, 2017), with overall high recognition rates for the whole corpus (83.3/80.3% for frontal/averted faces, 85.5/87% for frontal/averted bodies; Thoma et al., 2013). Other measures of EBL recognition are available in the literature, such as the Bodily Expressive Action Stimulus Test (BEAST; de Gelder & Van den Stock, 2011). However, the latter only consists of the body language component and does not include stimuli for disgust. Therefore, as the recognition of all negative emotions plays a particularly important role in HD (Bates, Tabrizi, & Jones, 2014), the BESST was preferred in this study due to its inclusion of disgust, as well as for being currently the only test to include both facial and EBL stimuli within a single set.

Difficulties in Emotion Regulation Scale (DERS; Gratz & Roemer, 2004)

The DERS is a self-report questionnaire based on 36 items rated on a 5-point Likert scale. It explores emotion regulation on the basis of 6 subscales: non-acceptance of emotional responses, difficulties engaging in goal directed behaviour, impulse control difficulties, lack of emotional awareness, limited access to emotion regulation strategies, lack of emotional clarity. A subscore is yielded for each subscale, which can then be summed to create a total score out of 180 for the whole questionnaire. Higher scores correspond to more difficulties in emotion regulation. To our knowledge, the DERS has never been adopted with people affected by symptomatic HD, but it has been utilised with a number other clinical conditions (Fowler et al., 2014; Kökönyei, Urbán, Reinhardt, Józán, & Demetrovics, 2014), showing very good construct validity (Ritschel, Tone, Schoemann, & Lim,

2015) and internal consistency (Cronbach's $\alpha = .93/.89$ for total score/subscales Gratz & Roemer, 2004).

Hospital Anxiety and Depression Scale (HADS; Zigmond & Snaith, 1983)

The HADS is one of the currently most adopted measures of mood and anxiety symptoms in clinical populations and consists of a self-report questionnaire based on 14 items rated on a 3-point scale. The outcome consists of individual scores out of 21 for both anxiety and depression. The HADS has been previously validated with people affected by HD (De Souza, Jones, & Rickards, 2010), and features good construct validity and internal consistency, with a Cronbach's $\alpha = .83/.82$ for anxiety/depression respectively (Bjelland, Dahl, Haug, & Neckelmann, 2002). The suggested cut-off point for clinical depression and anxiety is 8/21, which guarantees good sensitivity (anxiety/depression = $.90/.83$) and specificity (anxiety/depression = $.78/.79$).

Procedure

In general, all the questionnaires were filled in by hand by the participants directly. However, in case of difficulties due to motor impairments, the questions were read out to the participants and their responses were recorded by the experimenter on their behalf. The two trials of the BESST were administered by the experimenter on a 15-inch laptop. Each stimulus was presented singularly on a black background in an 834x834 pixel format along with seven emotional labels on the right corresponding to the emotions investigated by the test. The participants were asked to name the label corresponding to the presented stimulus. This kind of multiple-alternative forced choice task has been adopted

before with the BESST (Abramson et al., 2017; Soria Bauser, Thoma, & Suchan, 2012) and it currently one of the most widely utilised paradigms in the assessment of emotion recognition (Baron-Cohen et al., 2001; Calder et al., 1996; Beatrice de Gelder & Van den Stock, 2011; Ekman, P., Friesen, 1976; Sahakian & Owen, 1992). No direct interaction was required between the participants and the laptop. A practice session consisting of seven stimuli (one for each emotion) was administered prior to the beginning of each trial, to allow for familiarisation with the task. The order was kept constant among the participants, with the face trial being administered prior to the body language trial. No time limit was set for responses. However, the participants were asked to perform the tasks as quickly as possible. Figure 1 illustrates examples of neutral, positive, and negative stimuli administered via the BESST.

Statistical analysis

All the statistical analyses were performed with IBM SPSS Statistics® programme v23 (Armonk, NY: IBM Corp). On account of the relatively small sample size, non-parametric statistics were adopted. This is a common choice when working with small sample of symptomatic HD participants, and especially when investigating emotional processing (e.g., Croft et al., 2014; Snowden et al., 2008; Trinkler, de Langavant, & Bachoud-Levi, 2013). Mann-Whitney tests were performed to make comparisons between the two participant groups, while two-tailed Spearman's correlations were utilised to investigate the relationship between the two main outcome variables. Effects sizes were calculated with Cohen's *d*. In order to avoid loss of power due to the adoption of conservative

corrections with a small sample size, significance levels were conventionally set at $p = .05$ with no correction for multiple comparisons. This was in line with several previous studies on emotion recognition in small samples of people with HD (Croft et al., 2014; Ille, Holl, et al., 2011; Ille, Schäfer, et al., 2011; Novak et al., 2012; Robotham et al., 2011; Snowden et al., 2008; Sprengelmeyer, Schroeder, Young, & Eppelen, 2006; van Asselen et al., 2012), as well as in other rare clinical populations (e.g., frontal FTD; Keane, Calder, Hodges, & Young, 2002).

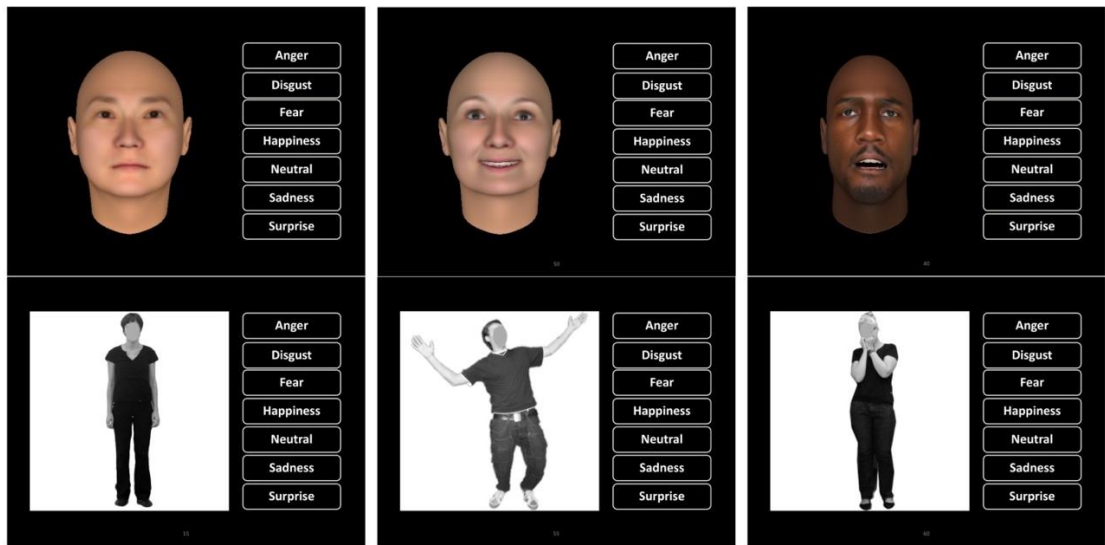


Figure 1: Example of neutral, positive, and negative emotion stimuli administered via the BESST. On the top are stimuli for the face trial, on the bottom are stimuli for the body one. The left column shows neutral stimuli; the central column shows stimuli for happiness; the right column shows stimuli for fear. Each stimulus was presented separately to the participants.

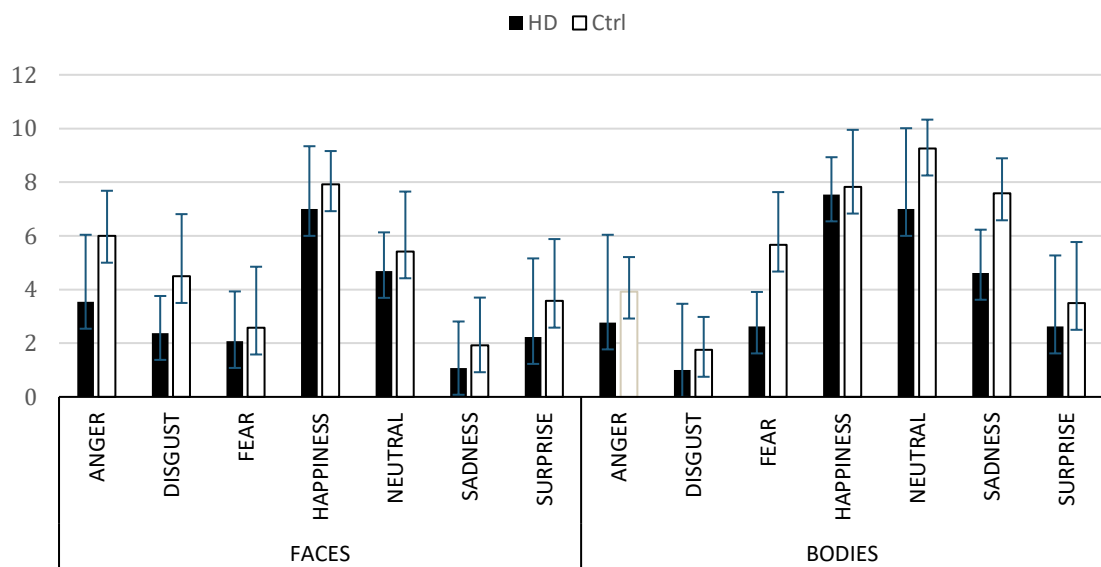


Figure 2: Participants' results on the emotion recognition tasks. Mean (and standard deviation) of correct responses for each of the six emotion category on the BESST, across both the facial and body language modality (max score = 10).

Table 2

Participants' scores across the outcome variables.

		HD	Ctrl	Between-group comparison			Reliability
		Mean (SD)	Mean (SD)	<i>U</i>	<i>p</i>	<i>d</i>	α
HADS	HADS-A	7.00 (5.80)	5.58 (2.46)	72.00	ns	.318	.808
	HADS-D	8.00 (4.49)	3.00 (2.697)	27.00	.005	1.350	.805
	SUM	90.92 (28.85)	67.00 (16.92)	29.50	.008	1.011	.941
DERS	NONACCEPT	12.69 (5.76)	11.42 (6.08)	63.00	ns	.214	.855
	GOALS	14.00 (5.71)	10.92 (2.91)	60.00	ns	.679	.814
	IMPULSE	14.54 (5.14)	9.33 (3.65)	28.50	.007	1.168	.808
	AWARE	17.54 (5.44)	14.33 (4.27)	44.50	ns	.656	.763
	STRATEGIES	18.38 (6.37)	12.92 (4.14)	43.00	ns	1.016	.819
	CLARITY	13.77 (5.08)	8.08 (2.02)	30.50	.008	1.471	.775
	TOTAL	22.85 (7.06)	31.92 (4.33)	16.50	.000	-1.54	.758
BESST FACES	NEUTRAL	4.69 (2.50)	5.42 (1.68)	69.00	ns	-.342	.634
	FEAR	2.08 (1.38)	2.58 (2.31)	71.00	ns	-.262	.567
	DISGUST	2.38 (1.85)	4.50 (2.27)	34.50	.016	-1.023	.638
	HAPPINESS	7.00 (2.34)	7.92 (1.24)	63.50	ns	-.491	.670
	SADNESS	1.08 (1.44)	1.92 (2.23)	62.00	ns	-.447	.616
	SURPRISE	2.23 (1.73)	3.58 (1.78)	44.50	ns	-.769	.531
	ANGER	3.54 (2.93)	6.00 (2.30)	40.00	.037	-.933	.756
	TOTAL	28.15 (11.08)	39.58 (3.85)	32.00	.012	-1.378	.863
BESST BODIES	NEUTRAL	7.00 (3.27)	9.25 (1.29)	40.50	.032	-.905	.896
	FEAR	2.62 (2.47)	5.67 (1.23)	25.00	.004	-1.563	.680
	DISGUST	1.00 (1.29)	1.75 (1.96)	62.50	ns	-.452	.638
	HAPPINESS	7.54 (1.39)	7.83 (2.12)	58.00	ns	-.156	.627
	SADNESS	4.62 (3.01)	7.58 (1.08)	33.00	.013	-1.309	.815
	SURPRISE	2.62 (1.61)	3.50(1.31)	54.50	ns	-.599	.352
	ANGER	2.77 (2.65)	3.92 (2.27)	55.50	ns	-.466	.740

Note. BESST TOTAL max score: 70. BESST single emotion max score: 10. Clinical cut-off for the HADS: 8/21. AWARE = lack of emotional awareness; CLARITY = lack of emotional clarity; Ctrl = control group; *d* = Cohen's *d* effect size; DERS = Difficulties in Emotion Regulation Scale; GOALS = difficulties engaging in goal directed behaviour; HADS-A = HADS anxiety score; HADS-D = HADS depression score; HD = symptomatic HD group; IMPULSE = impulse control difficulties; NONACCEPT = non-acceptance of emotional responses; SD = standard deviation; STRATEGIES = limited access to emotion regulation strategies; SUM = DERS total score; *U* = Mann-Whitney's *U*.

Results

Table 2 shows the mean scores of the participants of the both the HD and Ctrl groups for the outcome variables, and Figure 2 provides a graphical illustration of the scores. All the adopted measures generally showed good levels of reliability. More specifically, high internal consistency was shown by HADS for both anxiety (Cronbach's $\alpha = .808$) and depression (Cronbach's $\alpha = .805$), as well as the DERS, with a Cronbach's α of .941 for the SUM score, and figures ranging from .763 to .855 for the subscales. For both measures, the reliability results were comparable to the levels reported in the literature (Bjelland et al., 2002; Gratz & Roemer, 2004). With specific regards to the BESST, the total scores showed high internal consistency in both the facial (Cronbach's $\alpha = .758$) and emotional body language (Cronbach's $\alpha = .863$) modalities. The single emotions scores generally showed acceptable figures, with Cronbach's α ranging between .616 and .896. However, low levels were found for the fear facial score (Cronbach's $\alpha = .567$), and for both the facial and emotional body language scores for surprise (Cronbach's $\alpha = .531$ and .352).

According to the recommended clinical cut-off for the HADS (8/21; Bjelland et al., 2002), six of the participants of HD group (i.e., 46.15%) showed clinical levels of anxiety, while seven (i.e., 53.8%) showed clinical levels of depression. On the other hand, only three of the participants of the Ctrl (i.e., 25%) group showed clinical levels of anxiety, and only one (i.e., 8.3%) reported clinical levels of depression. With regard to emotion regulation difficulties, the total score (SUM) of the HD group was significantly higher than the available data with

general adult populations (e.g., 77/180; Ritschel et al., 2015), meaning that considerably more emotion regulation difficulties were reported by the participants. Instead, on average the Ctrl group scored rather lower (67/180) compared with the normative data.

The general results for emotion recognition showed a slightly better performance on the body language modality compared to the facial one in both groups. However, in this study the BESST constituted quite an arduous task for all the participants, as rather low overall recognition rates were observed for both the HD group (32.6% for faces, 40.2% for bodies) and the controls (45.6% for faces, 56.5% for bodies). These represented lower rates compared to the ones reported by the validation study (i.e., 83.3/87%; Thoma et al., 2013), but were in line with those reported in studies that adopted the BESST with a multiple forced-choice paradigm (e.g., 50%; Abramson et al., 2017). In terms of specific emotions, in the face task the least recognised emotion in both groups was sadness (HD: 10.8%, Ctrl: 19.2%), while the most easily identified was happiness (HD: 70%, Ctrl: 92.5%). The results on these two emotions were in line with the findings of the validation study. On the other hand, in the body language modality the lowest scores were observed on disgust for both groups (HD: 10%, Ctrl: 17.5%), while the highest were again on happiness (HD: 75.4%, Ctrl: 78.3%), along with neutral stimuli (HD: 70%, Ctrl: 92.5%). Contrarily to the facial modality, this result was opposite to the validation data, which found happiness body stimuli to be least recognised.

The group comparison analysis showed that the participants affected by HD had a significantly greater level of depression when compared to the controls ($U = 27.0, z = -2.787, p = .005$); however, no significant difference was found for anxiety levels ($U = 72.0, z = -.328, p = ns$). In terms of emotion regulation, a significantly greater level of total difficulties (DERS SUM) was reported by the HD group ($U = 29.5, z = -2.639, p = .008$). In this regard, the effect size analysis showed a very large group effect on the overall scores for emotion regulation ($d = 1.011$), facial emotion recognition ($d = -1.54$), and emotional body language recognition ($d = -1.378$). When comparing the specific components of the DERS, significant differences were observed on impulse control difficulties (IMPULSE; $U = 28.5, z = -2.730, p = .007$), and lack of emotional clarity (CLARITY; $U = 30.5, z = -2.595, p = .008$). With regard to emotion recognition, the overall performance of the HD group on the BESST was significantly poorer for both the facial ($U = 16.5, z = -3.352, p < .000$) and body language ($U = 32.0, z = -2.510, p = .012$) modalities. In terms of specific emotions, the facial modality revealed specifically greater impairments in the HD group for disgust ($U = 34.5, z = -2.402, p = .016$) and anger ($U = 42.0, z = -2.082, p = .012$), while the body language modality yielded poorer performances for fear ($U = 25.0, z = -2.914, p = .004$), sadness ($U = 33.0, z = -2.481, p = .013$), and neutral stimuli ($U = 32.0, z = -2.144, p = .012$). However, in spite of a lack of statistical significance, several medium to large effect sizes were observed for specific components of the outcome variables, thus showing group effects at a trend level. These included limited access to regulation strategies (STRATEGIES; $d = 1.016$), difficulties in engaging in goal directed behaviour (DERS GOALS; $d = .679$), lack of emotional awareness (DERS AWARE; $d = .656$), facial recognition of

happiness ($d = -.491$), sadness ($d = -.447$), and surprise ($d = -.769$), as well as body language recognition of disgust ($d = -.452$), surprise ($d = -.599$), and anger ($d = -.466$).

Table 3

Spearman's correlation coefficients for the HD group (N = 13) across the main variables.

	1	2	3	4	5	6	7	8	9	10	11
1. Gender											
2. Age	.112										
3. Education (yrs)	.342	-.127									
4. Diagnosis (yrs)	-.272	.121	-.073								
5. TFC Score	.271	.326	.092	.125							
6. HD Stage	-.501	-.323	-.325	-.025	-.902**						
7. HADS_A	.022	-.032	-.220	-.204	-.301	.260					
8. HADS_D	-.201	.170	-.135	-.222	-.593*	.558*	.510				
9. DERS_SUM	.089	.132	-.316	-.168	-.421	.371	.905**	.629*			
10. BESST_F_SUM	.157	-.213	.296	-.345	.393	-.533	.025	.015	-.168		
11. BESST_B_SUM	.491	-.077	.320	-.238	.521	-.675*	-.207	-.259	-.300	.739**	

Note. * = $p < .05$; ** = $p < .01$; BESST = Bochum Emotional Stimulus Set; BESST_B_SUM = BESST Bodies total score; BESST_F_SUM = BESST Faces total score; DERS = Difficulties in Emotion Regulation Scale; DERS_SUM = DERS total score; HADS = Hospital Anxiety and Depression Scale; HADS_A = HADS anxiety score; HADS_D = HADS depression score; HD = Huntington's disease; TFC = Total Functional Capacity; yrs = years.

In light of the significant differences observed in symptomatic individuals on the group comparison, a correlation analysis was carried out to explore whether the impairments on emotion regulation and recognition in the HD group were correlated with the demographic characteristics and the measures of psychological difficulties. Table 3 illustrates Spearman's coefficients for the correlation analysis of the HD group among the main variables; Table 4 instead illustrates the correlation coefficients among all the variables, including the

respective subscales. The results showed that the overall level of emotion regulation difficulties (DERS_SUM) shared a very strong positive correlation with levels of anxiety (HADS_A; $r_s = .905, p < .001$), as well as a strong correlation with levels of depression (HADS_D; $r_s = .629, p = .021$). In particular, the two components that were specifically impaired in the HD group, IMPULSE and CLARITY, were respectively related to anxiety ($r_s = .675, p = .011$) and depression ($r_s = .717, p = .006$). With regard to emotion recognition, the overall performance for the facial modality of the BESST (BESST_F_SUM) was strongly correlated with the overall performance for the body language modality (BESST_B_SUM; $r_s = .739, p < .001$), confirming the relationship between the two emotion recognition components. This was also confirmed by the observation of significant relationships across the modalities between the single scores for neutral stimuli ($r_s = .606, p = .028$), disgust ($r_s = .582, p = .037$), and anger ($r_s = .589, p = .034$), as well as linear trends close to significance for fear ($r_s = .526, p = .065$) and surprise ($r_s = .499, p = .082$). In addition, the total score for the body language modality (BESST_B_SUM) shared a strongly significant negative relationship with HD stage ($r_s = -.675, p = .011$), meaning that the recognition of emotional body language of the participants affected by HD deteriorated in line with disease progression. The total score for the facial modality (BESST_F_SUM), showed a similar trend towards HD stage ($r_s = -.533, p = .060$).

Discussion

This aim of this study was to investigate whether emotion regulation and emotional body language (EBL) recognition are impaired in people with

symptomatic HD when compared to age-matched controls, and whether such impairments share a significant relationship to one another. In addition, to our knowledge this was the first study with this specific population to explore emotion regulation along with emotion recognition, as well as both facial and EBL recognition modalities together. The results showed significant impairments for the HD group in emotion regulation, as well as emotion recognition in both the facial and EBL modality. This was in line with our initial predictions and confirmed our first two hypotheses.

In terms of specific components of emotion regulation, significant results were found for impulse control difficulties (DERS IMPULSE) and lack of emotional clarity (DERS CLARITY). This appears to be consistent with several previous observations of impulse control and executive functioning deficits in people with HD (Duff et al., 2010; Galvez et al., 2017; Gray et al., 2013; Mörkl et al., 2016), that are often due to the impact of the disease on prefrontal brain areas (Dogan et al., 2014; Gray et al., 2013) and are likely to play a pivotal role in the clarity and control of emotional experiences. No significant differences were observed for the remaining components of emotion regulation, including DERS STRATEGIES. This particular finding was in line with the only other study on emotion regulation in people with HD, which only explored the use of regulatory strategies and found no significant differences with age-matched controls (Croft et al., 2014). In addition, since no authors have previously carried out a comprehensive investigation of emotion regulation in HD which includes all its components, the significant difference on the DERS SUM observed in this study represents the first evidence of a general impairment of emotion regulation in this specific population.

The observed impairment for facial emotion recognition adds further confirmation to the already well-known deficit reported in the literature (for a review, see Bates, Tabrizi, & Jones, 2014). Moreover, the results on the single emotion scores also confirmed the known specific deficit of negative emotions such as disgust and anger, even though no specific difference was found for the facial recognition of fear and sadness. The impairment on the EBL modality was partially in line with the only other study that investigated this construct in people with symptomatic HD, that found a significant impairment for the recognition of anger and emotionally neutral instrumental stimuli, but no deficit for fear and sadness (de Gelder et al., 2008). Indeed, a specific impairment for neutral (yet not instrumental) stimuli was found in the present study too. However, the comparisons on the single emotion scores yielded almost opposite results, with a significant impairment for fear and sadness, but no significant deficit for anger. As the above mentioned study did not include stimuli for happiness, surprise, and disgust, it is not possible to know whether other emotions were impaired, and to what extent our results differ. As a consequence, the finding of the present study also represents the first evidence of an impairment of emotional body language (EBL) recognition in people with symptomatic HD through a comprehensive assessment that includes both positive and negative emotional stimuli.

With regards to the relationship between emotion regulation and emotion recognition, the correlation analysis showed that the observed impairments did not significantly relate with each other. This result went against our predictions, and contradicted our third hypothesis. Moreover, it was also inconsistent with what was previously reported in other clinical populations, such as anorexia

nervosa (Harrison et al., 2009). On the other hand, the overall level of emotion regulation difficulties shared a significant correlation with anxiety and depression. In addition, impulse control difficulties and lack of emotional clarity – the two emotion regulation components that were specifically impaired in the HD group – shared significant relationships with anxiety and depression respectively. These findings suggest that, in the participants of the HD group, anxiety and depression might have played a pivotal role in the operationalisation of emotion regulation. Moreover, they are consistent with previous reports of associations between mood and anxiety problems and deficits of emotion regulation (e.g., Ehring et al., 2008; Loas et al., 1997; for a review on anxiety, see Cisler, Olatunji, Feldner, & Forsyth, 2010), and in particular between impulse control and anxiety (e.g., with Parkinson's disease; Voon et al., 2011), and emotional clarity and depression (Dixon-Gordon et al., 2014; Thompson, Boden, & Gotlib, 2017). Thus, the significant difference on the DERS in the HD group when compared to the Ctrl group may represent a reflection of the significantly higher level of depression reported by the symptomatic participants.

As mentioned above, some of the findings of this study appear to contradict what has been previously reported in the literature. In particular, the results on the single emotion scores of the BESST showed no deficits for the facial recognition of fear and sadness, and the EBL recognition of anger. While some of these inconsistencies may be due to the use of different measures – especially as the BESST has never been adopted before with HD and may not represent the most well-fitting measure with this population – as well as differences in the way the stimuli were administered, an important caveat to be noted is the potential effect

of the relatively small sample size. Indeed, the effect size analysis showed that most of those inconsistent results in fact represented differences at a trend level characterised by medium to large effect sizes ($d = -.452 - 1.016$). Moreover, this could be also applied to some of the results of the correlation analysis that were approaching significance, such as the correlation between facial emotion recognition and HD stage. Thus, it could be hypothesised that the adoption of a larger sample would yield significant differences on fear, sadness, and anger in line with the findings in the previous literature, as well as significant correlations in line with the ones that were found in this study.

Limitations and future directions

A number of limitations should be considered along with the results of this study. First, as the data were collected through single sessions at the participants' home, it was not possible to perform any cognitive screening prior to the administration of the research materials. While this allowed the cognitive load to a comfortable level, it also prevented a more precise understanding of the participants' level of cognitive functioning, which would have allowed for a better clinical depiction of the stage of disease. Moreover, some participants required the experimenter to record their responses on their behalf, thus adding a possible source of misunderstanding. Secondly, the participants in the control group were recruited from partners and caregivers of people with HD. While this facilitated the selection of a sample from a population matching the HD group in terms of age and education, it did not support the assumption that the observations were completely independent. Thirdly, the generally low recognition rates on the BESST

showed that the emotion recognition tasks were rather difficult for both the HD and Ctrl group as compared to the available normative data. This is likely due to the differences in the way the tasks were administered compared to the validation study (Thoma et al., 2013), which was based on a two-alternative forced choice task with a 3000ms limit, while the present study featured a six-alternative forced choice task with no time limit. Indeed, lower recognition rates were reported when adopting the BESST with tasks based on four or more alternatives (Abramson et al., 2017), which currently represents the golden standard of some of the most widely adopted facial and EBL emotion recognition tests (e.g., Reading the Mind in the Eye test – RME, Baron-Cohen et al., 2001; Bodily Expressive Action Stimulus Test - BEAST, de Gelder & Van den Stock, 2011; CANTAB Emotion Recognition Task, Sahakian & Owen, 1992; Ekman 60 Faces Test, Ekman, Friesen, 1976; Emotion Hexagon Task Calder et al., 1996). In addition, the general better performance observed in both groups on the EBL recognition component as opposed to the facial one may be due to an effect of the order of presentation of the tasks (facial first, EBL second), which was kept constant among the participants. Last, despite being in line with most of the current studies on emotion recognition in HD (Bates, Tabrizi, & Jones, 2014), the sample size in this study was relatively limited. This translated into a number of variables that showed differences between the groups at a trend level, but that were not statistically significant. Moreover, the small sample size did not allow to control whether depression levels predicted emotion regulation difficulties. Thus, the conclusion that HD directly impacts emotion regulation should be taken cautiously until additional evidence is obtained with larger samples.

Future research should aim at further exploring emotion regulation and emotion recognition in larger samples of people with symptomatic HD, as well as obtaining a better understanding of the potential relationship between these two constructs. Other measures of EBL recognition should also be adopted with HD populations, in order to control for the external validity of the BESST and to build a comprehensive corpus of data similar to the one currently available for facial stimuli. In particular, more data are warranted on the optimal use of the BESST stimuli when based on a multiple-choice forced task, in order to avoid potential floor effects on the participants' performances. In this perspective, the adoption of EBL measures would benefit from the inclusion in large multi-centre studies, which would also allow to integrate in-depth general cognitive screenings.

Conclusion

This study shed new light on emotional processing in people with symptomatic Huntington's disease by providing multi-componential evidence that emotion regulation and emotional body language (EBL) recognition are significantly impaired in this population, and that the latter is negatively related to the stage of disease. It also provided the first evidence of a significant direct correlation between deficits of facial and body language emotion recognition in HD, although emotion regulation and emotion recognition were not related.

These findings have important implications for clinical practice, as a more in-depth understanding of emotional processing in HD has the potential to revise current therapeutic and communicative protocols, as well as informing new ones. More specifically, better insight into emotion regulation issues in this population,

along with their connections to mood and anxiety disorders, would allow the development of psychological and pharmacological interventions that are tailored around the emotional needs of each patient.

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Table 4

Spearman's correlation coefficients for the HD group across all the variables.

	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21	22	23	24	25	26	27	28	29	30	31		
1 GENDER																																	
2 AGE	.112																																
3 EDUCATION (YRS)	.342	-.127																															
4 DIAGNOSIS (YRS)	-.272	.121	-.073																														
5 TFC SCORE	.271	.326	.092	.125																													
6 HD STAGE	-.501	-.323	-.325	-.025	-.902**																												
7 HADS_A	.022	-.032	-.220	-.204	-.301	.260																											
8 HADS_D	-.201	.170	-.135	-.222	-.593*	.558*	.510																										
9 DERS_SUM	.089	.132	-.316	-.168	-.421	.371	.905**	.629*																									
10 DERS_NONACCEPT	-.067	.029	-.568*	-.017	-.441	.484	.739**	.404	.878**																								
11 DERS_GOALS	.291	-.035	-.218	-.267	-.355	.279	.867**	.436	.919**	.770**																							
12 DERS_IMPULSE	.067	.191	-.396	-.267	-.214	.223	.717**	.457	.866**	.857**	.787**																						
13 DERS_AWARE	.134	-.059	.163	-.548	-.453	.334	.383	.825**	.437	.124	.358	.265																					
14 DERS_STRATEGIES	-.067	.170	-.425	.109	-.550	.558*	.765**	.632*	.916**	.861**	.823**	.724**	.301																				
15 DERS_CLARITY	-.112	.250	.109	-.371	-.459	.491	.351	.687**	.456	.313	.309	.456	.611*	.412																			

Chapter 7

General Discussion

Overview

This chapter serves to present an integration of the findings of the studies of the present thesis, as well as to highlight theoretical and practical implications and limitations. The overall aim of this research project was to investigate the impact of Huntington's disease (HD) on the communication of affected individuals. Communication was here intended and operationalised as the heterogeneous ability that "focuses on how people use messages to generate meanings within and across various contexts, cultures, channels, and media" (Korn, Morreale, & Boileau, 2000, p. 40), thus including nonverbal components such as emotional processing and body language. Based on the research question, and in accordance with the critical realist position of the researcher, it was reasoned that the adoption of a mixed methods design would allow for a better investigation of the heterogeneity of the construct of communication (Frost & Shaw, 2015). This in turn informed the choice of a methodological framework characterised by a sequence of studies that, although equal in terms of methods importance, were designed with the purpose of allowing the initial investigations to inform the later ones (i.e., *development*; Greene, 2007; Greene et al., 1989). As a consequence, within the context of the

abovementioned overall aim of the research project, each study (or *publishable paper*, PP) led to the development of further, more specific research aims.

Review of research aims and summary of main findings

Research aim 1: to identify the elements of communication and methodological approaches investigated in symptomatic HD

The first specific research aim was to explore the breadth of elements of communication that had already been investigated in people with symptomatic HD, along with the diversity of adopted methodological approaches. This was addressed through a scoping review of the empirical quantitative and qualitative literature (PP1). The search identified 49 eligible citations, which were then divided into four categories: Communicative Skills, Emotion, Language, and Speech. Out of these categories, the most investigated elements of communication were facial emotion recognition, the assessment of linguistic productive abilities, and speech assessment. As a consequence, the most adopted methods were visual recognition tasks of emotional facial pictures, as well as cognitive language and speech examinations. The result was in line with a systematic review of the literature of emotion recognition in HD (Henley et al., 2012), which identified an underrepresentation of methods aimed at exploring non-facial features of emotional processing, such as body language.

Much less investigated topics identified by the scoping review included emotion expression, language comprehension, and therapeutic interventions on communication or speech. However, the topic that was by far recognised as the

most neglected in the literature was the subjective perspectives of people with Huntington's disease on their communicative skills and functioning, with only two retrieved studies addressing them. This confirmed an already known underrepresentation of first person perspectives explorations in all chronic illnesses which cause communicative impairments, such as stroke, Parkinson's disease, and motor neuron disease (Mistry & Simpson, 2013; Thorne et al., 2002). Moreover, with specific regard to HD, this finding was in accordance with a general lack of qualitative investigations in Huntington's that was recently observed by a review of the qualitative literature in neurodegenerative conditions (Auduly et al., 2014). However, the results of the two identified studies on communication perspective in people with HD shed important preliminary light on the importance of many social and environmental factors on communication (Hartelius, Jonsson, Rickeberg, & Laakso, 2010; Power, Anderson, & Togher, 2011). In addition, they showed the potential for a qualitative subjective approach to communication in HD to highlight aspects and issues that may go overlooked with the adoption of quantitative methods alone, thus informing the development of the following study of this thesis.

Research aim 2: to explore the perspectives on communication in people with symptomatic HD

The initial scoping literature review (PP1) highlighted the need for further explorations of subjective experiences and perspectives of people affected by Huntington's disease on their communicative skills. As a consequence, this topic was addressed with a qualitative study based on semi-structured interviews

analysed through thematic analysis (PP2). The results identified four main recurring themes among the participants' accounts. The first theme concerned the directing and mediating role of HD within the context of the participants' everyday communication, by blocking their speaking abilities directly or causing conditions that influence their experience in more indirect ways (e.g., chronic fatigue). The identification of HD's influence led the way to the second theme, which was characterised by the need to retake control over the features of communication as an effective coping strategy. Examples of this included having a choice over demanding things that require particular energy, such as answering the phone or participating actively in conversations.

The third theme concerned the impact of HD on the emotional life of the participants, and how it makes it unstable. This included inconsistent emotional states characterised by anger or sadness, often accompanied by considerably longer recovery times, which ultimately caused a pervasive feeling of emotional unsteadiness. With regard to this, an improvement in emotion regulation was identified as a potential solution. Indeed, despite being a challenging process that often required engaging with difficult and disturbing thoughts, the achievement of a better regulation of emotional responses led to considerably increased willingness to communicate. Moreover, this process was reported to be particularly enhanced by the development of a supportive network consisting of close relationships with family member and friends, as well as by the implementation of effective medication regimes.

The fourth theme concerned the in-depth exploration of a particular coping strategy that was mentioned by many participants, and that consisted in the idea of owning a personal safe place (*sheltering*). Although being initially identified with their home, this idea later extended to a more abstract conception of safety, a mind 'shelter' where the participants could feel safe to switch off from the world. In particular, this not only seemed to help them deal with everyday challenges, but also appeared to boost their self-confidence in the process of managing emotions observed in the previous theme, further underlining the importance of the successful operationalisation of emotion regulation.

Research aim 3: to investigate how HD affects emotion regulation abilities and how they relate to emotion recognition

As outlined by the findings of the qualitative study (PP2), the ability to regulate emotions efficiently can be of paramount importance in the psychological adaptation to HD, in particular within the context of communication. However, the results of the initial scoping review had shown no quantitative investigations on whether emotion regulation is affected by HD, nor whether this construct relates in any way to the deficit of emotion recognition caused by the disease. Thus, this aim was addressed with two parallel quantitative studies, in order to target both presymptomatic (PP3) and symptomatic (PP4) individuals. The latter study also included a body language emotion recognition task, in order to address one of the neglected areas in emotional processing in HD identified by PP1.

In general, the results showed that emotion regulation and recognition were not affected by HD in presymptomatic individuals, while significant

impairments were found for both in symptomatic people. This included a deficit of EBL recognition in PP4, which was significantly related to the impairment on the facial tasks. This not only confirmed the well-known facial recognition impairment which is observed in symptomatic HD (Henley et al., 2012; Novak & Tabrizi, 2005), but also confirmed the close connection between these two components of emotional processing (de Gelder & Hortensius, 2014). Within both populations, however, no significant correlation was observed between emotion regulation and recognition. This appeared to be inconsistent with the current evidence with other clinical populations such as anorexia nervosa, in which a significant relationship was reported (Harrison, Sullivan, Tchanturia, & Treasure, 2009). Nonetheless, the findings from PP2 showed that presymptomatic individuals had a specific impairment of the emotional awareness component of emotion regulation originating from the association with subclinical depressive symptoms, and this was proposed to represent a precursor of the emotion recognition impairment later found in fully symptomatic HD. Within this perspective, it could be hypothesised that the relationship between emotion regulation and recognition in Huntington's disease may not consist of a presentation of linearly associated impairments as in other populations. Instead, it may be acting as an earlier and more cyclical process, with some deficits on specific elements of emotion regulation playing a pivotal role along depressive symptoms in the development of emotion recognition impairments, which in turn affects other elements of emotion regulation in the long term.

Theoretical integration

Levels of integration

Within the context of mixed-methods research, a particularly debated subject is at what point different methods should be actually integrated to achieve a logic and consistent account of the research findings (Fetters, Curry, & Creswell, 2013). A possible solution is the categorisation outlined by Moran-Ellis and colleagues (2006). In particular, the authors argue that the concept of integration (as opposed to simple combination) of methods requires that the *status* of all utilised methods is equal in standing, and that they all aim at a common goal while retaining their paradigmatic features. This leads to three possible levels of integration: a) *integrated methods*, i.e. when integration occurs at the design (or methods) level; b) *separate methods, integrated analysis*, i.e. when the integration occurs during the analytical process; c) *separate methods, separate analysis, theoretical integration*, i.e. when the data of the studies are analysed within the parameter of their respective paradigms, and later integrated within a single explanatory or interpretive framework. In consideration of the abovementioned *purpose, timing, dependence, status, and methods* of the studies (Greene, 2007; Greene et al., 1989), a *theoretical integration* was reasoned to be most adequate for the work of the present thesis, as it would allow to reflect the heterogeneity of the different methods and designs involved in the research process (Moran & Butler, 2001). In particular, in the view of integrating the present findings at a theoretical level within the field of health psychology, an interpretive framework that proved to fit especially well with the diverse range of methods of this work is

represented by the already mentioned 'common sense' self-regulation model (SRM; Leventhal, Leventhal, & Contrada, 1998).

The self-regulation model (SRM)

According to the SRM, an individual's psychological adaptation to health threats – and especially chronic conditions – is based on their own lay ('common sense') perspectives about illnesses (Leventhal, Leventhal, & Brissette, 2003). More specifically, adjustment is seen as a function of cognitive and emotional representations that, working as parallel processes, lead to the adoption (or avoidance) of specific coping strategies, which in turn interact with adjustment itself (Hagger et al., 2017).

In particular, the content of cognitive representations can be divided into six dimensions (Moss-Morris, Humphrey, Johnson, & Petrie, 2007): a) *identity*, i.e. the illness 'label' and the number of symptoms attributable to it; b) *timeline*, i.e. perceptions on the time, onset, duration, and development of the condition; c) *cause*, i.e. the individual's perception on what caused the illness; d) *consequence*, i.e. individuals' beliefs on the impact the condition on their everyday life; e) *perceived control*, i.e. perceptions on individuals' capacity to influence the course of the illness; f) *illness coherence*, i.e. individuals' understanding of their own condition. On the other hand, emotional representations are related to the adoption of coping procedures aimed at managing the psychological distress related to process of becoming ill (e.g., anxiety and/or depression), as well as any psychological difficulties directly caused by the illness itself (Hagger et al., 2017).

In fact, a key dimension of the emotional representations arm of the model is represented by emotion regulation, whose successful operationalisation is essential for the management of illness-related distress (Leventhal et al., 2003). In this perspective, an expanded version of the SRM has been proposed, which underlines the importance of the emotion regulation strategies outlined by Gross (1998; 2015) by integrating them into the SRM (Cameron & Jago, 2008). As Figure 3 illustrates, the manifestation of illness stimuli leads to the development of the two types of illness representations, cognitive and emotional, which in turn manage the adoption of cognitive and emotional regulatory (ER) coping strategies. Both processes end with an appraisal of the coping success, which then leads to a reappraisal of the illness stimuli. It is important to note that, despite being parallel processes, the cognitive and emotional arms of the model can affect each other in a number of ways, both positive and negative. For example, the development of coherent cognitive representations and strategies may help lower the impact of excessive emotional representations (e.g., excessive anxiety or fear) and prevent the adoption of counterproductive emotional regulatory strategies, such as emotion avoidance and suppression. Similarly, the successful implementation of emotional representations and regulatory strategies may help prevent incautious cognitive representations leading to dangerous coping strategies (e.g., minimization of symptoms leading to avoidance of treatment). It is also essential to note that the whole system is in constant interaction with the social setting of the individual, which has the potential of affecting deeply the choice of coping strategies, as well as their success, thus mediating the outcome of both the abovementioned processes (Leventhal, Diefenbach, & Leventhal, 1992).

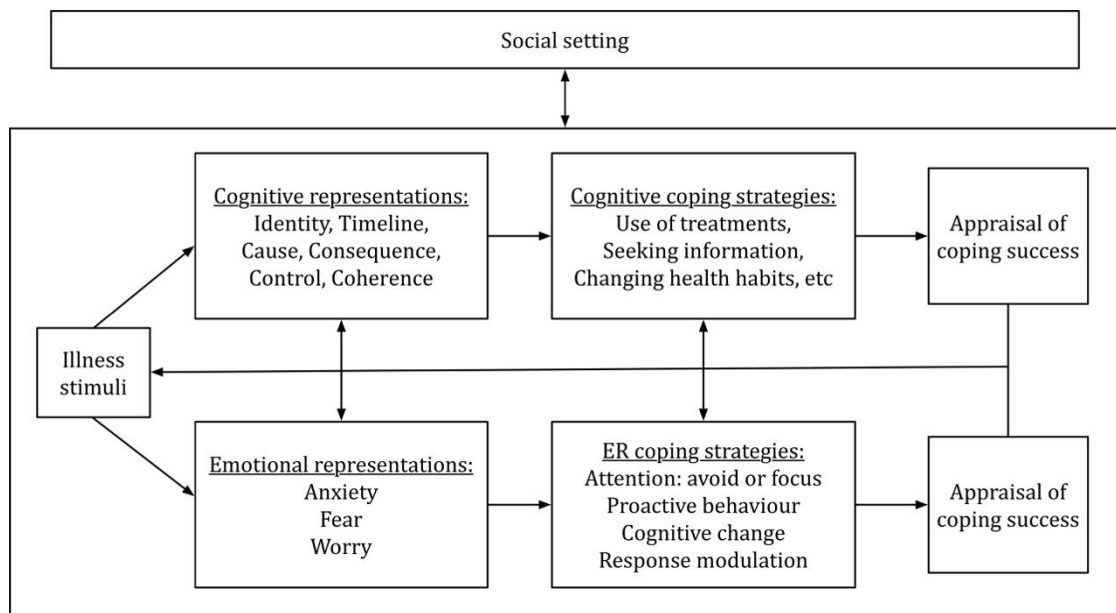


Figure 5: Expanded SRM including emotion regulation (ER) strategies (adapted from Cameron & Jago, 2008).

Theoretical integration within the expanded SRM

Due to its focus on emotion regulation, the expanded version the SRM (Cameron & Jago, 2008) appears to represent a particularly well-fitting interpretive framework for the integration of the findings of this thesis, even more than Leventhal's original model (1998). In this perspective, the findings of each of the PPs following the scoping review (PP1) can be related directly with the components of the model, allowing them to be interpreted altogether despite the different methodologies of the studies. More specifically, they can be viewed as providing insight into specific components that, if successfully operated, can support the development and maintenance of two adaptive circles involving cognitive and emotional coping strategies, as well as social and environmental elements (see Figure 4).

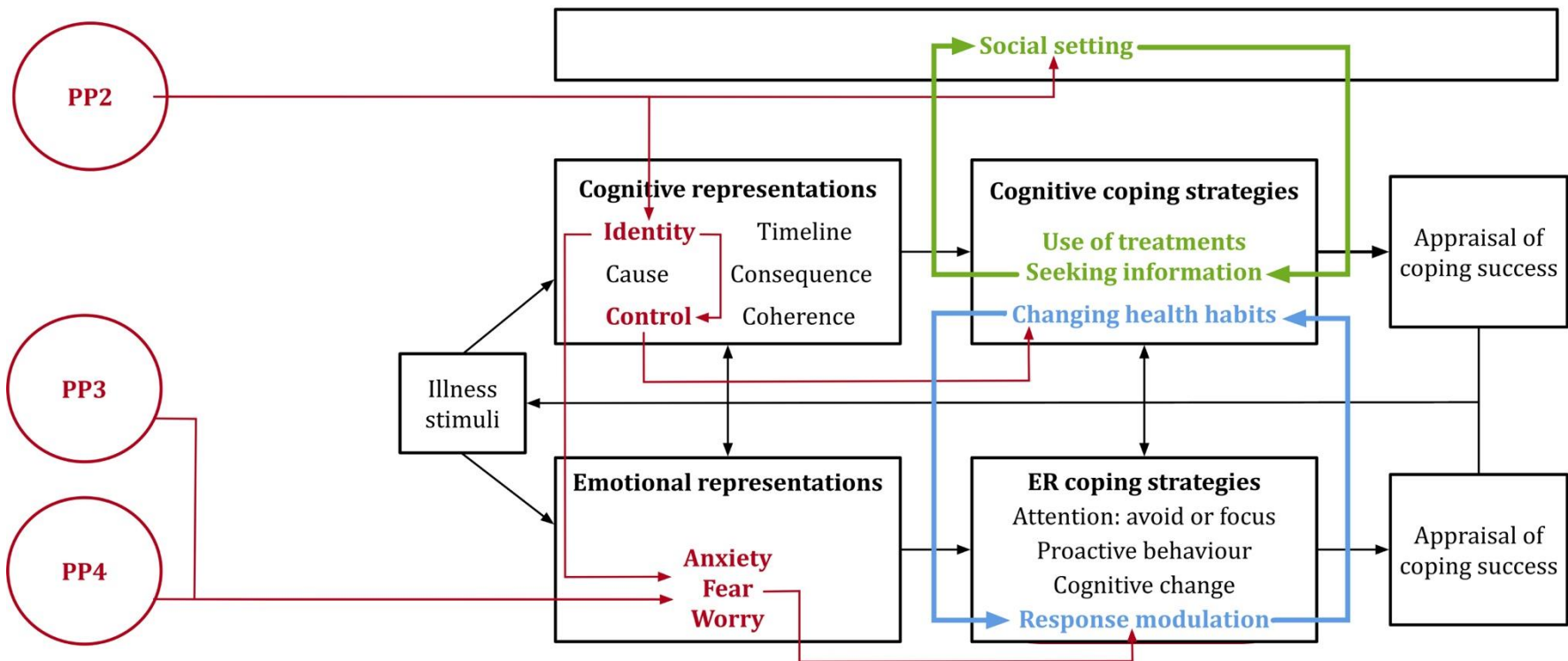


Figure 6: Theoretical integration of the findings of the publishable papers (PPs) within the framework of the expanded SRM (adapted from Cameron & Jago, 2008). The SRM is shown in black, while the PPs integration is shown in red. The proposed adaptive circles are indicated in blue (cognitive coping strategies ↔ ER coping strategies) and green (cognitive coping strategies ↔ social setting).

On one side, the perspectives of people with Huntington's disease outlined in PP2 shed important light on how HD's symptomatology (*identity*) has a severe impact on every aspect of communication, from speech to memory and fatigue, also including emotional processing (*emotional representations*), and in particular aspects such as emotional and mood reactions (*anxiety, fear, worry*). This brought to attention a need for regaining control over communication (*control*) that, if successfully implemented, could lead to the development of efficient cognitive coping strategies such as sheltering (*changing health habits*). In turn, these showed the potential to help enhance emotion regulation strategies, and in particular the management of emotional reactions (*response modulation*), which most of the participants found particularly challenging due to the emotional instability caused by the disease. As a consequence, success at regulating emotions also allowed for the reinforcement of those healthier behavioural and communicative habits, thus enabling an adaptive circle between *cognitive coping strategies* and *ER coping strategies*, in particular between the *changing health habits* and *response modulation* components.

Similarly, a parallel contribution towards this circle was provided by the findings of PP2 and PP3, which shed new light on the impact of HD on emotional processing (*emotional representations*) at both the symptomatic and presymptomatic stage. In particular, they also showed how the disease may affect emotions not only by disrupting their stability directly, but also through the association with concurrent depressive symptoms (*worry*), which can help disrupt specific components of emotional processing (e.g., emotional awareness). As a

result, the combination of these associations will eventually lead to a greater impairment of emotion regulatory skills (*response modulation*). Therefore, the findings of PP2 and PP3 suggest that, by targeting interventions at the specific elements (*emotional representations*, and in particular *worry*) that have shown to contribute to the emotional upheaval caused by HD, the detrimental effect on *response modulation* may be reversed. As observed with the results of PP2, this has in turn the potential to help towards the development of an adaptive circle between cognitive and emotional coping strategies.

On the other side, the findings of PP2 were also helpful in underlining the importance of social and environmental factors (*social setting*), and their effect on communication and emotional processing in people with HD. In particular, many participants recognised the pivotal role played by the support of family members and friends in helping them deal with the everyday challenges of the disease. This translated into a beneficial effect on the motivation to search and ask for support when needed (*seeking information*), as well as on the uptake of effective medication regimes (*use of treatments*). In turn, this had the potential to improve communication and thus interpersonal interactions in general, with an overall beneficial effect on the social and environmental context. As a consequence, this may help the development of a second adaptive circle, this time between *cognitive coping strategies* (in particular the *seeking information* and *use of treatments* components) and *social setting*.

Chapter 8

Conclusion

Theoretical importance

As outlined in the previous chapter, the theoretical integration of the present findings relies on the prospective development of two adaptive circles based on specific cognitive, emotional, and social components of the expanded SRM. This carries the potential to deepen our theoretical understanding of how cognition and emotion work in people affected by Huntington's disease. In particular, the adoption of a mixed-methods approach has helped to unearth and highlight the importance for mental health of the relationship between specific elements of emotional processing and communication that have been so far neglected in the empirical literature, especially the combined role of emotion regulation and emotion recognition. This answered a current general need for investigations focused on the relationship between different emotional components – as opposed to traditional studies targeting them independently – in order to achieve a better understanding of their combined effects on health (see Pandey & Choubey, 2010 for a review). More specifically, the present findings also offer additional theoretical insight and renewed momentum into the currently recognised need for further explorations focused on understanding the psychological issues and difficulties of living with HD (Audulv et al., 2014).

Implications for clinical practice

The results of the present research also feature practical implications for the field of health psychology, as they carry the potential to inform the development of interventions tailored around specific cognitive and emotional needs of patients with HD. Indeed, the possibility of enhancing patients' cognitive reserves through cognitive training interventions has proven to be a very promising approach to delay or control the onset of symptoms in neurodegenerative diseases (Papoutsis, Labuschagne, Tabrizi, & Stout, 2014). With regard to this, however, the current literature seems to be characterised by a significant paucity of studies addressing cognitive interventions in HD, and there is currently a strong need for further investigations in this area (for a review, see Andrews, Dominguez, Mercieca, Georgiou-Karistianis, & Stout, 2015). For instance, recent preliminary evidence suggests that addressing emotion recognition impairments at both presymptomatic and early stage via self-guided computerised training can lead to significant improvement in accuracy, opening up new avenues for innovative methods of intervention (Kempnich, Wong, Georgiou-Karistianis, & Stout, 2017). Moreover, the case of HD represents an ideal model for the exploration of this kind of interventions, particularly thanks to its genetic nature, the availability of predictive testing, and the consequent well-established underlying pathological mechanisms (Papoutsis et al., 2014).

However, in addition to individual cognitive training, the present findings can help focus on other promising areas of development. For example, the observation of coping strategies such as *sheltering* – as well as the importance of

environmental and relational factors such as effective support from friends and family members – carry the potential to help inform a more systemic approach to intervention. Indeed, current evidence shows that interventions based on social models of disability aimed at reducing negative interactions (such as those characterised by stigma) may contribute considerably to preventing negative emotions and tendencies to reduced social participation (Simpson, McMillan, & Reeve, 2013) – which represent the basis of a phenomenon that has recently been defined as *psychoemotional disablism* (Reeve, 2012). With specific regards to the current result, examples of this kind of interventions may include the enhancement of *sheltering* through group meditation (e.g., mindfulness-based programs; Chan, Churcher Clarke, Royan, Stott, & Spector, 2017), the adoption of less stigmatising language (e.g., avoiding terms such as ‘the HD face’), as well as the design and implementation of dementia-friendly environments and, ultimately, communities (Davis, Byers, Nay, & Koch, 2009; Lin & Lewis, 2015; Swaffer, 2014).

Therefore, due to their focus on specific cognitive, emotional, and social components affected by the condition – and in particular their integration into the development of potential adaptive circles – the present findings have the important clinical implication of carrying a revived incentive towards the development of both individual and systemic psychological interventions that may help delay or control symptom onset in people affected by Huntington’s disease.

Limitations and future directions

A number of limitations should be acknowledged when considering the findings of the present research. First, the initial scoping review of the literature (PP1) was limited by the heterogeneity that characterises the terms and definitions of communication, and that might have limited the number of retrieved citations. Furthermore, despite being functional for the aims and purposes of the current thesis, the scoping methodology did not allow for the development of more specific research questions. Thus, future literature reviews would benefit from a narrower focus on specific elements of communication in HD, and possibly from the inclusion of evidence from neuroimaging data.

The qualitative study on communication perspectives (PP2) was characterised by the limit of including only participants from early to moderate stage of disease. Moreover, interpretive frameworks other than the SRM may be utilised to interpret the results. Future research on perspectives of people with HD should aim at involving participants at later stages of disease (possibly adopting innovative communication methods such as LiteWriters™), as well as other theoretical frameworks. Moreover, further explorations on the concept of *sheltering* are advised, as well as potential implementations of this coping strategy into approaches to intervention.

The online survey with presymptomatic individuals (PP3) carried the intrinsic limitation of online studies, namely the lack of direct contact with the participants. In addition, the need for brevity led to the adoption of the RME as a measure of emotion recognition, which may have not been sensitive enough to

detect subtle differences or potential correlations within this specific population. Therefore, future research should aim at developing more sensitive measures that may be included in online surveys without hampering the brevity required by this methodological approach.

Due to HD's low prevalence and despite being in line with most investigations on the topic, the between-subjects study with symptomatic participants (PP4) was characterised by a relatively small sample size. This led to limitations in terms of complexity of data analysis and generalizability of the findings. Moreover, due to the practical requirements of the data collection sessions, no cognitive screening assessment could be performed before administering the research materials. The adoption of the BESST with a multiple choice forced paradigm also produced generally low emotion recognition rates among all the participants. Thus, future investigations on emotion regulation and recognition in HD should be included in large multi-centre clinical studies, in order to allow for the enrolment of higher numbers of participants, as well as the adoption of cognitive screening batteries and more comprehensive measures of emotional processing.

Finally, more research is needed on HD's impact on sensory and emotional processing based on modalities other than the visual one. Indeed, few studies have reported impairments in people affected by HD on abilities such as odour identification and general olfactory functioning (Bylsma, Moberg, & Doty, 1997; Moberg & Doty, 1997; Pirogovsky et al., 2007), as well as emotional processing through the auditory (Calder et al., 2010; Hayes, Stevenson, & Coltheart, 2007;

Snowden et al., 2008; Sprengelmeyer et al., 1996), olfactory (Sprengelmeyer et al., 1996; Scahill et al., 2013), and gustatory (Mitchell et al., 2005) modalities. Considering the paramount importance of all sensory processes in social and communicative contexts (Knapp & Daly, 2011), it is possible to assume that a deficit in any of these abilities (e.g., the inability to identify unpleasant smells in the surrounding environment) may have significant repercussions on patients' interpersonal relationships. Thus, future studies should investigate the potential impact of these impairments on patients' communicative experiences and daily quality of life.

Statement of contribution

In the past four years, the opportunity to carry out the investigations at the basis of the present thesis has represented to me an outstanding experience of personal and professional development as a researcher. In particular, coming from a mainly quantitative neuropsychological background developed during my foundation degrees, the occasion to adopt a mixed-methods approach has been seminal in improving my knowledge of the breadth of methods available to psychological research, as well as the importance of subjective experiences in the characterisation and understanding of psychological difficulties.

Moreover, the opportunity to carry out qualitative audio-recorded interviews in a language different from my own native has greatly increased my understanding of the importance of nonverbal aspects of communication. Indeed, by re-listening to my audio recordings in English during the transcription work for PP2, I had the chance to realise for the first time how hard it can be to understand

verbal messages fully when no nonverbal cues (e.g., facial expressions and body language) are provided. This was especially clear to me due to the fact that, as evidenced by the recordings, during the interview I did understand the same messages without major difficulties. Needless to say, considering the topic of the current thesis, this also helped me from an empirical perspective, allowing me to make more informed choices when developing studies focused on nonverbal communication.

The occasion to carry out the data collection at the participants' home also played a pivotal role in teaching me the importance of contextual variables in cognitive research. Indeed, since most of my previous clinical experience in neuropsychology was based in hospital environments, this kind of activity allowed me to observe how deeply cognitive abilities can vary when the participants are in a familiar or comfortable place. This was also helpful in the development of the concept of *sheltering* that emerged from the findings of PP2, as well as for the observation of the profound beneficial effect that strong familiar or friends' support can have on patients' mental health and functioning.

Finally, I probably cannot express how grateful I am for the opportunity I have had to work with the HD community. Rarely in any of my previous clinical or research experiences have I found such a keen and supportive group of people – whether affected individuals, caregivers, or clinicians – who taught me so much about the value of generosity, mutual help, dignity, and resilience. If this PhD has also contributed to my growth in terms of compassion, I certainly owe it to them.

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Appendix 1
Ethics Approval Letters

Applicant: Nicolo Zarotti
Supervisors: Dr Ian Fletcher, Dr Jane Simpson
Department: DHR

16 March 2015

Dear Nicolo, Ian and Jane,

Re: An exploratory investigation of communication in Huntington's disease (HD)

Thank you for submitting your research ethics application for the above project for review by the Faculty of Health and Medicine Research Ethics Committee (FHMREC). The application was recommended for approval by FHMREC, and on behalf of the Chair of the University Research Ethics Committee (UREC), I can confirm that approval has been granted for this research project.

As principal investigator your responsibilities include:

- ensuring that (where applicable) all the necessary legal and regulatory requirements in order to conduct the research are met, and the necessary licenses and approvals have been obtained;
- reporting any ethics-related issues that occur during the course of the research or arising from the research to the Research Ethics Officer (e.g. unforeseen ethical issues, complaints about the conduct of the research, adverse reactions such as extreme distress);
- submitting details of proposed substantive amendments to the protocol to the Research Ethics Officer for approval.

Please contact the Research Ethics Officer, Debbie Knight (01542 592605 ethics@lancaster.ac.uk) if you have any queries or require further information.

Yours sincerely,

S.C. Taylor

Sarah Taylor
Secretary, University Research Ethics Committee

Cc Fiona Aiken, University Secretary, Professor Roger Pickup (Chair, FHMREC); Prof Stephen Decent (Chair, UREC).

Lancaster University
Research and Enterprise
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Bowland Main
Lancaster, LA1 4YT, UK
T: +44 (0)1524 592 002
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Applicant: Nicolo Zarotti
Supervisor: Jane Simpson
Department: Health Research
FHMREC Reference: FHMREC16015

01 November 2016

Dear Nicolo,

Re: Emotion recognition and regulation in people with pre-symptomatic Huntington's disease: an online survey

Thank you for submitting your research ethics application for the above project for review by the **Faculty of Health and Medicine Research Ethics Committee (FHMREC)**. The application was recommended for approval by FHMREC, and on behalf of the Chair of the Committee, I can confirm that approval has been granted for this research project.

As principal investigator your responsibilities include:

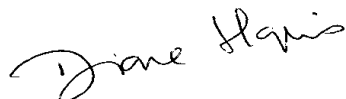
- ensuring that (where applicable) all the necessary legal and regulatory requirements in order to conduct the research are met, and the necessary licenses and approvals have been obtained;
- reporting any ethics-related issues that occur during the course of the research or arising from the research to the Research Ethics Officer at the email address below (e.g. unforeseen ethical issues, complaints about the conduct of the research, adverse reactions such as extreme distress);
- submitting details of proposed substantive amendments to the protocol to the Research Ethics Officer for approval.

Please contact me if you have any queries or require further information.

Tel:- 01542 592838

Email:- fhmresearchsupport@lancaster.ac.uk

Yours sincerely,



Dr Diane Hopkins
Research Integrity and Governance Officer, Secretary to FHMREC.

Applicant: Nicolo Zarotti
Supervisor: Ian Fletcher
Department: Health Research
FHMREC Reference: FHMREC15043

29 February 2016

Dear Nicolo,

Re: Emotion recognition and regulation in Huntington's disease: a cognitive investigation

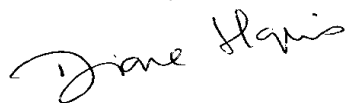
Thank you for submitting your research ethics application for the above project for review by the Faculty of Health and Medicine Research Ethics Committee (FHMREC). The application was recommended for approval by FHMREC, and on behalf of the Chair of the University Research Ethics Committee (UREC), I can confirm that approval has been granted for this research project.

As principal investigator your responsibilities include:

- ensuring that (where applicable) all the necessary legal and regulatory requirements in order to conduct the research are met, and the necessary licenses and approvals have been obtained;
- reporting any ethics-related issues that occur during the course of the research or arising from the research to the Research Ethics Officer (e.g. unforeseen ethical issues, complaints about the conduct of the research, adverse reactions such as extreme distress);
- submitting details of proposed substantive amendments to the protocol to the Research Ethics Officer for approval.

Please contact the Diane Hopkins (01542 592838 fhmresearchsupport@lancaster.ac.uk) if you have any queries or require further information.

Yours sincerely,



Dr Diane Hopkins
Research Development Officer

CC Ethics@Lancaster; Professor Roger Pickup (Chair, FHMREC)

Appendix 2

Research Posters

Communication in Huntington's disease: an empirical review

Nicolò Zarotti, Ian Fletcher, Jane Simpson

Division of Health Research, Lancaster University, UK



Introduction



Communication is a multifaceted discipline that includes language, emotion, speech, as well as social and environmental factors. It is particularly relevant in the adjustment to chronic illnesses such as Huntington's disease (HD), as communicative patterns are significantly related to clinical outcomes and the clinical manifestations of HD are likely to impair several components of communication across all its stages.

An empirical review was conducted to identify the elements of communication that have been investigated with people who had symptomatic HD, as well as the differing methodological approaches.

Methods

A systematic search was performed in the databases PubMed, PsycINFO, and Linguistics and Language Behaviour Abstracts (LLBA) from January 1993 to January 2015.

MeSH Terms were adopted on PubMed and Subject Terms were adopted on PsycINFO. Since no Subject Term for HD was available on LLBA, the general keyword "Huntington's" was used through all the database fields.

The initial search across all the databases identified 478 citations.

Database	Search terms	Citations
PubMed	"Huntington Disease"[Mesh] AND "Communication"[Mesh]	262
	"Huntington Disease"[Mesh] AND "Language"[Mesh]	100
PsycINFO	DE "Huntingtons Disease" AND DE "Communication"	16
	DE "Huntingtons Disease" AND DE "Language"	11
LLBA	"Huntington's"	89

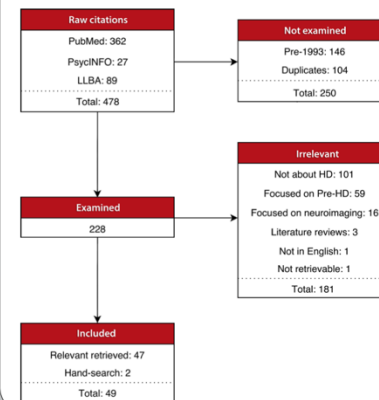
Selection

To be included in this review, the studies had to be related to any of the different features of communication in people with a diagnosis of symptomatic HD confirmed by genetic testing.

The studies with the following characteristics were excluded:

- Publication prior to the introduction of genetic testing in 1993 (Huntington's Disease Collaborative Research Group, 1993).
- Focus on types of communication other than interpersonal.
- Focus on people without HD or with pre-symptomatic HD only.
- Neuroimaging as main or only method.
- Main focus on the neural correlates of cognitive processes.

At last, 49 studies were included. The selection process is detailed below.



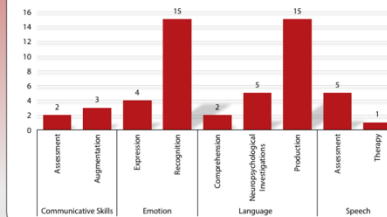
Categorization

Four general categories were identified to reflect the main research topics:

Communicative Skills | Emotion | Language | Speech

Subcategories were also identified for every topic.

The distribution of the citations among the categories is detailed below.



Discussion

The most investigated elements of communication and the related methods were the following:

- Emotion recognition (visual recognition tasks of emotional faces)
- Language production (cognitive testing)
- Speech (acoustic voice analysis)

Other much less investigated elements of communication included:

- Emotion expression
- Language comprehension
- Therapeutic interventions

The most neglected topic was the assessment of communicative skills, with only two studies (Hartelius et al., 2010; Power et al., 2011) investigating it through qualitative interviews and focus groups. Their results showed preliminary evidence on the significant impact of social and environmental factors on communicative abilities.

Results

➤ **Communicative Skills (5):** Two studies addressed the assessment of communicative abilities through qualitative interviews, while three investigated augmentative and alternative communication (AAC) strategies through qualitative interviews and videotaped conversations.

➤ **Emotion (16):** Fifteen studies concerned the assessment of emotion recognition; of these, three were also focused on the emotion expression, therefore falling into both subcategories. Only one exclusively addressed the assessment of emotion expression.

➤ **Language (22):** Two studies were related to language comprehension, six consisted of studies focused on a general neuropsychological assessment that included language and 15 concerned the assessment of language production. Two used a conversation analysis approach and four adopted a longitudinal design.

➤ **Speech (6):** Five studies were related to the assessment of voice through acoustic analysis and one was focused on speech therapy through labial and lingual motion and resistance training.

What next?

Topics that would benefit from future research in people with HD include:

- Emotion expression and nonverbal components of emotion recognition.
- Patients' perspectives on their communicative skills and efficiency.
- The effect of impairments and social factors on patients' functional communicative capacity.

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“I have a feeling I can’t speak to anybody”

A thematic analysis of communication perspectives in people with Huntington’s disease

Nicolò Zarotti, Jane Simpson, Ian Fletcher

Division of Health Research, Lancaster University, UK

Introduction

Huntington’s disease (HD) is responsible for many impairments affecting language, speech, emotions, and social abilities. All these can ultimately lead to significant deficits of communication, intended as the ability to create meanings through messages transmitted across various channels, media, and contexts.

However, the current literature on communication in HD is mainly characterised by quantitative studies focused on medical and cognitive impairments from the perspectives of clinicians, with little attention for patients’ perspectives.

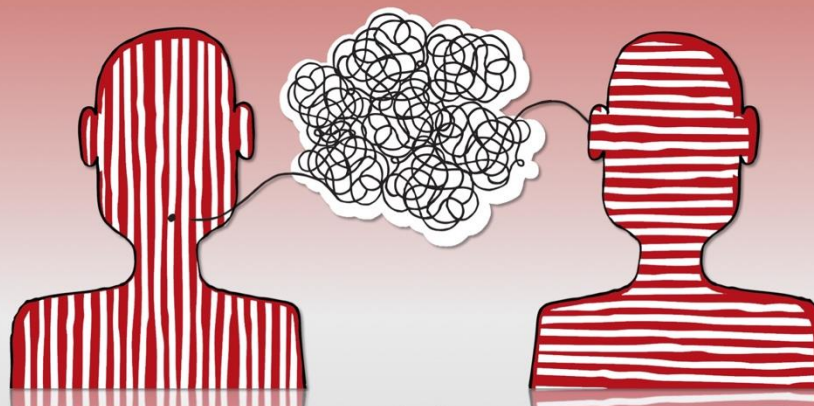
Thus, the present study aimed at exploring how people with HD conceptualise and make sense of their communication experience with others.

Methods

Qualitative semi-structured interviews were carried out with 8 people with early to moderate HD (stage I-III) across North West England. All interviews were audio-recorded and transcribed verbatim. The transcripts were analysed through thematic analysis (TA), adopting the 6 steps outlined by Braun and Clarke (2006) as a guide.

A list of 73 initial codes was generated from the data. Upon revision, the final list was reduced to 36. Out of these, 4 main themes were identified:

1. How HD directs and mediates communication.
2. Regaining control to improve communication.
3. Emotional outflows into communication and the struggle for separation.
4. ‘Sheltering’ as a way to boost confidence in communication.



Theme 1: HD’s Influence

HD acts as both external *director* and *mediator* of patients’ communication, by blocking it *directly* (e.g. language impairments), or by *mediating it* through several collateral conditions such as chronic fatigue or attention or memory problems.

*“You ever want to say things to people... but you can’t!
There’s something wrong with your illness!”* (Pt. 3)

Theme 2: Regaining Control

An effective coping strategy that improves patients’ communication is regaining control over features of communication, such as avoiding phone conversations, saving energy or using texts.

“Texting! That’s one thing that I can do! If I text, they understand what I’m talking about!” (Pt. 4)

Theme 3: Emotional Outflows

HD threatens patients’ emotion regulation with inconsistent feelings of anger and sadness, apathy, and considerably longer ‘cool-down’ times. These emotional difficulties ultimately outflow into participants’ communication making it unsteady.

“One moment I’m fine, the next moment I’m not. [...] It’s my Huntington’s, I can’t help it.” (Pt. 6)

Theme 4: ‘Sheltering’

A coping strategy adopted by many patients is the idea of owning a physical or abstract personal place (*‘sheltering’*). This allows them to feel safe and boost their self-confidence in communication:

“I go back into my little world, you know. [...] I just shut down, I don’t know how I’m doing it.” (Pt. 4)

Clinical Importance

A number of suggestions for clinical practice can be drawn from the present results:

- Patients should be allowed and encouraged to regain actively some control over a number of elements of communication, such as opting for texting rather than phone conversations.
- More effort should be put into helping patients reach a better level of emotion regulation, in order to tackle potentially dangerous emotional outflows into communication and other daily life activities, and ultimately improve their social relationships.
- Spontaneous and more tailored coping strategies, such as *sheltering*, should be strengthened and considered for inclusion within therapeutic protocol and interventions.

Full Text

Zarotti, N., Simpson, J., & Fletcher, I. (2017). “I have a feeling I can’t speak to anybody”: a thematic analysis of communication perspectives in people with Huntington’s disease. *Chronic Illness*. Advance online publication. DOI: 174239531773379.



Appendix 3
Conferences Attendance



CERTIFICATE *of* ATTENDANCE

THIS ACKNOWLEDGES THAT

Nicolò Zarotti

ATTENDED

HSG 2015: BUILDING OUR FUTURE
Oct. 21-24, 2015
Tampa, FL



RAY DORSEY, HSG CHAIR

***"Seeking treatments that make a
difference"***



EUROPEAN HUNTINGTON'S DISEASE NETWORK

Attendance Certificate

We herewith confirm the attendance of

NICOLÒ ZAROTTI
UNITED KINGDOM

at the 9th Plenary Meeting of EHDN in The Hague, The Netherlands
September 16-18, 2016

on behalf of the European HD Network

Prof. Dr. med. Jean-Marc Burgunder
Chair of the EHDN Executive Committee

Ulm, 21 September 2016

The meeting has been accredited by the European Accreditation Council for Continuing Medical Education (EACCME) to provide CME activity for medical specialists. It is designated for a maximum of **15 European CME credits (ECMEC)**. Each medical specialist should claim only those credits that he/she actually spent in the educational activity.

The EACCME is an institution of the European Union of Medical Specialists (UEMS), www.uems.net. Through an agreement between the European Union of Medical Specialists and the American Medical Association, physicians may convert EACCME credits to an equivalent number of AMA PRA Category 1 Credits™. Information on the process to convert EACCME credit to AMA credit can be found at www.ama-assn.org/go/internationalcme. Live educational activities, occurring outside of Canada, recognized by the UEMS-EACCME for ECMEC credits are deemed to be Accredited Group Learning Activities (Section 1) as defined by the Maintenance of Certification Program of The Royal College of Physicians and Surgeons of Canada.

Oberer Eselsberg 45/1 · 89081 Ulm · Germany
Phone +49 {0}731 500-63100 · Fax +49 {0}731 500-63082 · www.euro-hd.net

Appendix 4
Publications

‘I have a feeling I can’t speak to anybody’: A thematic analysis of communication perspectives in people with Huntington’s disease

Chronic Illness

0(0) 1–13

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DOI: 10.1177/1742395317733793

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Nicolò Zarotti, Jane Simpson and Ian Fletcher

Abstract

Objectives: This study explored the perspectives of people affected by Huntington’s disease (HD) on their own communicative abilities.

Methods: Qualitative semi-structured interviews were carried out with eight people with early HD. The data were analysed through thematic analysis.

Results: Four themes were constructed from the data, characterised by the following core topics: How HD directs and mediates communication; Regaining control to improve communication; Emotional outflows into communication and the struggle for separation; Sheltering as a way to boost confidence in communication.

Discussion: Separating patients’ identity as individuals from that of a person with a disease can help increase communicative control. Consistent with the general theory and model of self-regulation, patients should be allowed a wider range of choices to regain control over communication. Achieving better emotion regulation is of paramount importance for communication, and factors such as medication regimes, relationships and existing coping strategies should be strengthened. Consistent with previous research, feelings of safety and the idea of a safe place (‘sheltering’) represent an effective coping mechanism. Practical implications include the refinement of communication and relationships among clinicians, caregivers, and patients with HD by considering a wider range of medical, psychological and socio-environmental factors.

Keywords

Huntington’s disease, communication, emotion regulation, thematic analysis, patients’ perspectives

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Nicolò Zarotti, Division of Health Research, Faculty of Health and Medicine, Lancaster University, Lancaster, LA1 4YG, UK.

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Introduction

Huntington's disease (HD) is a hereditary chronic neurodegenerative disorder, which affects 10–12 people per 100,000 in the western world.¹ Typical symptoms include involuntary movements (chorea), cognitive deterioration, psychological difficulties, and psychiatric disorders.² Since the transmission mechanism is autosomal-dominant, affected individuals have a 50% probability of transmitting it to their children (usual age of onset is 40–50 years). The mean life expectancy after the diagnosis is typically 20 years.³ Genetic testing is available for individuals at risk, allowing them to know if they carry the disease gene before the onset of symptoms. All individuals with the gene without symptoms ('presymptomatic' people) will develop the disease.

Many cognitive impairments have been reported in people with HD, including problems with all aspects of communication. Communication is understood in this context as a multifaceted discipline that investigates how people create meanings through messages transmitted across various channels, media and contexts,⁴ and includes elements of language, speech, as well as emotion and social abilities. Regarding more specific aspects of communication, speech production is often impaired and starts to deteriorate before comprehension,⁵ and the spontaneous initiation of conversations is reduced.⁶ Research on nonverbal communication, such as body language and emotional processing, is less frequent in HD, with the exception of emotion recognition.⁷ In this respect, studies have indicated that all these components can be affected by the disease.⁸ In addition, HD can also negatively affect the ability to comprehend and reflect on the mental states of oneself and others (i.e. theory of mind⁹), which plays a fundamental role in communicative competence, especially through the attribution of intentions.¹⁰

Nevertheless, the current literature on communication in HD appears to be mainly characterised by quantitative studies focused on the observation of medical and cognitive impairments,¹¹ with a tendency to prioritise the perspectives of clinicians.^{12–14} The investigation of the perspectives of people with HD on their own communication abilities is generally much rarer and only recently has an interest started to emerge in communication as a phenomenon that embraces social skills and interactions, and not just the traditional underpinnings of language and speech.¹⁵ Indeed, even when patients' perspectives on the general impact of the disease have been investigated, data analysis falls more in the quantitative category, with greater focus given to the frequency of reoccurrence of themes rather than a qualitative exploration of patients' narratives themselves.¹⁶ On the other hand, to our knowledge only two studies have so far used a qualitative approach to investigate communication in HD.

Hartelius et al.¹⁷ adopted individual interviews and focus groups to triangulate the information between people with HD, family members and caregivers, finding that a number of social and environmental factors play a relevant role in complicating communication. These included the speed of conversations or having fewer people to talk with, while a positive impact was reported for sensations of safety, having the opportunity to speak for a longer time, and the perception of support and adjustment from the person with whom they were speaking. However, the thematic analysis was characterised by a very descriptive approach, and the interviews did not include any questions on nonverbal aspects of communication. In the second study conducted, Power et al.¹⁸ carried out a single case analysis with a man with advanced-stage HD and also found that supporting social and environmental factors can have a positive impact on communication. However, their approach (content analysis) offered a useful but predominantly descriptive

report and did not allow for a more in-depth interpretation of meanings, as well as omitting nonverbal elements of communication such as emotional processing.

Therefore, considering the current gap in the literature, the research question addressed by this study was the exploration of how people affected by HD make sense of their communication experience with others.

Methods

Methodological approach

A qualitative design was adopted, based on semi-structured interviews analysed through thematic analysis (TA). We selected TA for its recognised usefulness within the field of psychology and communication disorders,¹⁹ and its ability to allow for the usage of both deductive (i.e. theory-driven) and inductive (i.e. data-driven) analyses of the themes identified within the interviews.²⁰

Sampling

People affected by Huntington's disease were invited via post across the North West of England by the Huntington's Disease Association (HDA). Of the people who expressed their interest, eight participants with symptomatic HD were considered eligible. The inclusion criteria included being aged 18 or more, being able to be interviewed in English and being symptomatic at an early or moderate stage of disease. This limit was due to the difficulties in attending personal interviews that arise in the later stages of HD. As the study focused on the impact of the disease on patients' communicative experiences, we did not include any pre-symptomatic individuals. See Table 1 for the demographic details of the participants.

Procedure

Individual semi-structured interviews were carried out face to face with the participants

Table 1. Participants demographic data.

Participant	Gender	Age (yrs)	Diagnosis (yrs)	HD Stage
1	F	53	5	Moderate
2	F	53	7	Early
3	M	53	9	Moderate
4	M	41	5	Moderate
5	F	50	5	Early
6	F	62	5	Early
7	M	57	8	Early
8	F	49	6	Moderate

at a time and place convenient to them (usually their home). We selected this approach to allow the in-depth exploration of relevant themes as well as to ensure that all participants felt comfortable and had all the assistance they might need.²¹ The length of the interviews ranged from 40 to 60 min ($M = 45$). A caregiver (e.g. participant's partner) was often present during the interviews, but was asked not to contribute in any way and any comments they made did not form part of the data collection. We structured the interviews according to a framework consisting of four general topics: Verbal Communication, Nonverbal Communication, Mediators of Communication, Contexts of Communication. The topics were based on some of the most common categorisations of communication dimensions²² and to be as broad and yet comprehensive as possible.

Data analysis

All the interviews were transcribed verbatim and analysed using thematic analysis (TA). The six steps outlined by Braun and Clarke¹⁹ were used as a guide to analysis.

Results

Identified codes and themes

Following familiarisation with the whole dataset, 73 codes were generated.

These were then collated to six initial candidate themes. Upon further revision, the final code list was reduced to a total of 36. Out of these, four final themes were identified that were distinctive in their own right, as well as coherent with the broader scheme of the analysis and relevant to the research question. See Table 2 for the final theme list and breakdown into the respective codes.

'You ever wanna say things to people, but you can't': How HD directs and mediates communication

All the participants agreed that HD plays an important role in their communication with others. In fact, they made sense of their communicative difficulties by seeing HD as the main cause and this expressed itself both directly and indirectly. Directly, the

Table 2. Final list of identified themes and breakdown of relative codes.

Theme	Codes	
'You ever wanna say thing to people, but you can't': How HD directs and mediates communication	Articulation is harder	Not understanding what is going on
	Chronic fatigue makes communication harder	Not willing to go out
	Fear of saying or doing something wrong	Public places are uncomfortable
	Interrupting or jumping into a conversation	Speaking is harder
	Memory has changed	The HD face
	Not into communication anymore	Writing is harder
Becoming a director again: Regaining control to improve communication	Being open about HD helps communication	New or unexpected things are harder
	Communication with familiar people is easier	New technologies can be helpful
	Communication with more than one person is harder	Not using the phone anymore
	Communication with strangers is harder	Texting helps communication
	Getting used to new people helps communication	Physical contact and proximity are harder
	Listening rather than speaking	Retiring from work is helpful
'One moment I'm fine, the next moment I'm not': Emotional outflows into communication and the struggle for separation	Acceptance helps with emotions	Having a close family is helpful
	Emotions have changed	Having relatives or friends around is helpful
	Feeling discriminated	Medications help with emotions
	Feeling ignored	My little world
	Feeling lonely	
'I go back into my little world': Sheltering as a way to boost confidence in communication	Feeling misunderstood	
	Home is a safe place	
	My little bubble	

participants saw HD as an external agent that actively blocks their communication:

I think it is a funny illness, but affects you in different ways. [...] you ever wanna say things to people, but you can't. Because it feels there's something wrong, with your illness. [...] You want to say things, but you can't! (Participant 3)

The effects of HD can present in many different ways, starting from speech and language. Problems with articulating words, controlling voice speed, or respecting conversational roles were frequently reported and disrupted verbal communication at its core:

I probably speak fast. And also not loud enough. Sometimes I have to speak twice. I probably do articulate a bit harder. [...] I don't use many long words now, where I used to. I used to bullshit to heaven, but now I don't. (Participant 7)

The cause of these difficulties was entirely attributed to HD, which the participants perceived as the external reason they felt and behaved the way they did. In this perspective, HD acts as director of the participants' communication, taking control over their active role in everyday interactions by drastically decreasing their communicative repertoire. As some of them pointed out:

You don't speak to people... the illness makes you feel that way, even though you're trying to speak to people. (Participant 3)

However, the influence of HD on communication is not confined to its most apparent manifestations, such as articulation impairments. Indeed, many related difficulties such as feelings of constant fatigue, memory problems and attentional drops can indirectly affect the participants' communicative experience. Although not strictly related to

the verbal side of communication, these problems ultimately play an important role in participants' everyday life by undermining their ability to interact with people or read situations:

I like speaking with my son, I just keep... running out of things to say, because I keep forgetting." [...] For example we go to my sister's for Christmas, and I was trying get involved but I just feel so stupid because I can't think, my memory is very random [...] and it just adds more and more pressures because I start getting agitated with myself. [...] It must be to do with HD... [...] I don't know what's going on half of the time. (Participant 4)

The other thing that happens sometimes is that I can't think of a word to say next. (Participant 8)

Therefore, HD not only directly influences participants' communication through its characteristic symptoms, but also subtly mediates it via less apparent yet related conditions that pull the strings of nonverbal interactions and greatly increase the effort involved in simple discourses. Just as for their verbal issues, however, the participants made sense of these mediational effects as an external source of discomfort due entirely to HD. Not surprisingly, the combination of these influences eventually led the participants to develop a reluctant attitude towards communication. This particularly manifested through being quiet or avoiding situations in which communication is required:

I just can't be bothered, I just bugger off. I got a little scooter, so I just nip off to the quarry. [...] I don't really bother that much talking. If it's not the neighbour then I wouldn't bother. Yeah, it's just a bit of an effort. [...] I just say hello to acknowledge people. (Participant 1)

Again, it is worth noting how these behaviours were seen by the participants as due to the disease, to the extent of listing them among its stages and symptoms:

I don't really mean [to speak to people], because one stage of HD you don't intermingle with people, leave them on your own, if you will. I don't know if you know... (Participant 4)

Indeed, one of the participants mentioned being identified with HD by her parents, by having 'the Huntington's face':

They say... 'you know, you look as you have Huntington's'. I've got the face. The Huntington's face. It's just the expressions that my mum used to do with Huntington's. And they say I look the same. (Participant 6)

Clearly, being identified this way can lead to significant difficulties in communication. From this perspective, the participants' tendency to conceive HD as an external director and mediator may represent one of the causes of the 'fighting' metaphors often used to describe their struggle with symptoms and their consequent attempts to regain control over communication and prevent it from being incorporated into their own identity.

Becoming a director again: Regaining control to improve communication

Of all the effects of HD on participants' communication, the lack of control appeared to be perceived as one of the most substantial and, as mentioned, it manifested in several direct and indirect ways. However, it also seemed to drive the participants' will to develop strategies to reacquire control over communication and become once again the real director of their daily interactions. For instance, all

the participants claimed to find using the telephone much harder since the onset of the disease, due to the difficulties people had in understanding them. As a consequence, they started avoiding phone conversations, asking their carers or partners to intercede for them. However, this went beyond a mere avoidant behaviour, since it was often taken by the participants as an opportunity to shift the communication to more comfortable settings and modalities, like meeting in person or replying by email:

The phone is probably the most difficult one. [...] I talk to people, but it's just the phone sometimes. I prefer to leave it. [...] If somebody phones me, and I have to get back in touch with them, I just use text or an email. That type of thing. (Participant 7)

Texting was a popular choice for most of the participants, as it did not impose the same constraints as other communication methods in terms of time and effort. As Participant 4 noted:

Texting! That's one thing that I can do! If I text people, they understand what I'm talking about! (Participant 4)

From this perspective, delegating phone conversations and switching to texting represent a good strategy to shift part of the burden of communication. This was even clearer when some of the participants claimed to prefer listening over talking. Well aware that communication consists of both sending and receiving, they appeared to have developed a preference for the latter:

I can still listen, you know what I mean. I prefer listening to speaking. I can understand what people are talking about, I can listen. If I'm going to a group or something, I sit and listen, and then... when I feel comfortable with people around me,

then I can open up and I think that has to do with HD... (Participant 4)

This is probably due to how listening allows engagement in conversations without the amount of energy required by speaking. As Participant 5 noted:

I know when to chill out and I know when to use the energy. Like this is using more energy than I would normally. But tomorrow daytime I can relax and I've got a birthday party tomorrow night and I know I'm gonna use more energy for that, because I'll be seeing a lot of people and I'll be talking to a lot of people. (Participant 5)

In addition, the management of these aspects of communication seems to benefit from an attitude of general openness towards HD and its effects. Indeed, many participants mentioned telling people about their condition, in order to prevent any distressing situations:

I'm more open. I know I can explain things to people. [...] I do say to people 'look, if I sit with you, I know I might bump into you'. If I'm going out somewhere I'll sit at the end of the table. [...] People understand. If I'm open about it, people understand. (Participant 2)

Openness, however, comes at the cost of potential misunderstandings, as reactions and interpretations may differ significantly among people:

The worst thing people do, is tell people. As soon as you tell them, that's it. Bloody terrible. I've never done, I'd never tell anybody again, I've never advise anybody to tell anyone. 'Cause once you've told them, then you're not going to get employed or you're going to have a rough ride from then on. (Participant 7)

Thus, trying to be open to regain communicative control can backfire, by turning communication into a source of emotional discomfort.

'One moment I'm fine, the next moment I'm not': Emotional outflows into communication and the struggle for separation

One of the most challenging effects of HD was how it made emotions become unstable, and for many this was a new experience:

One moment I'm fine, the next moment I'm not. Different things upset me... I was at work, and there was this nurse, she was bossing me about, and I said 'you're being horrible to me' [...] and I got angry with her – never been like that before [...] I was thinking 'what's wrong with me?' And she said to me 'what's wrong with you?', I said 'I don't know', I said 'I'm going to go and get tested. I may have Huntington's'. (Participant 6)

Moreover, the emotional aspects varied significantly. For instance, Participant 5 mentioned getting progressively detached from her emotions:

I used to watch telly and I used to cry at the drop of a hat, and I used to have a box of tissues next to me and be soppy at anything, and then now I'm not. That box of tissues can stay there for months *laughs*. [...] I'm not as sympathetic as I used to. [...] I can still get angry, but not as much. I'm becoming more and more apathetic. Definitely. (Participant 5)

On the other hand, Participant 6 seemed to experience the opposite situation:

I think I am sort of angry a lot more that I used to be, but I don't mean to be. [...]

I knew there was something wrong with me, snappy and shouting at people, and I thought that's not like me, because I don't shout. I do now. It's my Huntington's, I can't help it. (Participant 6)

However, the actual effect perceived by affected individuals can be rather homogeneous, since the participants made sense of it as a general external influence beyond their control. As for speech and language, emotional issues were perceived as a symptom of the disease, rather than part of the psychological adjustment to the illness. As Participant 2 noted:

I think it was just the fact that... a symptom of the disease. As for depression. (Participant 2)

Difficulties with emotions can become even more problematic when, as previously mentioned, distressing emotional situations may arise while trying to be open about HD and regain control over communication. Although openness and control can promote illness acceptance (which is considered of primary importance in psychological adjustment to chronic illness^{23,24}), those emotional experiences took a major toll on the participants, ultimately leading them to perceive their emotional and communicative life as a source of unsteadiness that frustrated any attempts at accepting their condition:

With this illness, even though I accept it, it's very hard to accept things. I cannot drive anymore, and that's what I loved. I loved to drive all the time. All the sport's gone... (Participant 4)

I'm good at crying... I think I've got a little more sad. I started to be sort of thrown off without sense. I think when you have to be strong every day, sometimes it is hard... (Participant 8)

One of the ways to manage their communication and ultimately promote illness acceptance was for the participants to learn better how to regulate their own emotions. However, with the participants, emotion regulation seemed to happen at the cost of engaging with extremely difficult and fundamentally disturbing thoughts, which led them to deal with challenging emotional outflows into their communicative experience. As Participant 4 mentioned:

The thing with HD, I've noticed, if I am aggravated or someone else is aggravated or agitated, it takes some 5 or 10 minutes to me to calm down, but with HD it has a knock-on effect so it's like taking 3 times as long for me just to calm down because I go up on it, got more and more to think about and it drives me crazy. (Participant 4)

The process of achieving a better level of separation between communication and emotions clearly plays a pivotal role from this perspective. Yet, this can prove to be a very long and painful path, characterised by pervasive feelings of anxiety and instability that can turn communication into both a benefit and a liability, a 'double-edged sword':

I know it's important to talk to people, but when everybody comes around to see me, I just feel a bit nervous about talking. And I think it must be the illness, Huntington's, making me feel like that way. But I don't mind people talking about my illness to people, so it's a double-edged sword. (Participant 3)

However, whereas such a separation is achieved, emotion regulation has a clear beneficial effect. In the case of the participants, it promoted healthy grieving, helped them to better accept their condition, and

ultimately improved their communication:

I think I got better, 'cause I've grieved for what I've lost. So I accepted that. 'Cause a couple of years ago, I'd go into a shop and say 'I've got Huntington's' and I'd cry my eyes out. I've come to terms with it. (Participant 1)

Although long and challenging, the process of regulating emotions was seen to be promoted by a number of medical and psychosocial factors. For instance, all the participants agreed on the paramount importance of medication:

I was just tired and couldn't be asked to do things, you know. But now, once I started to take Citalopram I felt so much better in myself, it really lifted me up. (Participant 2)

In certain cases, being prescribed the appropriate medications could even benefit communication directly:

I've got my medications now I'm fine. [...] I wouldn't even be able to speak if I didn't take them. (Participant 6)

A similar beneficial effect was also mentioned for social and environmental factors. In particular, having a close family and friends around allowed the participants to feel more comfortable about communication, as well as safer when coping with daily situations by being able to ask for help when needed:

I do feel more comfortable here, you know. Like if you have people around, I'm fine. [...] I'm alright with going to different places. Well, I do try to, I like to try different restaurants and stuff like that, which I've already done. That hasn't gone yet. And we do have people round for meals and stuff like that. (Participant 7)

It is also important to notice how the positive relationship between emotion regulation and communication is bidirectional: just as regulating emotions can improve communication, so communicating better can promote emotion regulation; similarly, better relationships can improve both emotion regulation and communication – and vice versa. Thus, the successful expression and reinforcement of these factors allowed for a virtuous circle to come into being, which considerably improved the participants' quality of life.

'I go back into my little world': Sheltering as a way to boost confidence in communication

Another beneficial factor that emerged from some interviews was the idea of having a personal safe place where the participants could take shelter. Initially, this idea appeared to be mainly identified with a feeling of preference and protection for their own home:

I think, if I'm here, just sitting here, it feels comforting. I feel better at home. (Participant 8)

I don't know, I just... I feel my Huntington's. [...] I just need to get in the house, get in my comfy house. (Participant 6)

However, as a couple of participants further elaborated, the idea reached a deeper level of meaning which transcended the geographical space and embraced a more abstract concept of safety. For example, Participant 4 talked of his 'little world':

I go back into my little world, you know what I mean [...] I just shut down, I don't know how I'm doing it *laughs*. (Participant 4)

Similarly, Participant 5 mentioned her ‘little bubble’:

I know that sounds so ignorant, but I can just sort of getting into my little bubble and just completely close down if I need to, you know. (Participant 5)

This mental safety net appeared to have developed to help the participants deal with their everyday emotional and communicative life: when fatigue kicked in, emotions became overwhelming, or the circumstances got too demanding they could return to their shelter – of which their house in some cases only seemed to constitute a physical representation:

I just seem to be able to switch off because I don't know what we are talking about. [...] It's quite hard to explain... I just shut up... I just shut down and ignore people and then I look back here [at home]. (Participant 4)

From this perspective, the idea of sheltering appeared to provide the participants with an effective coping mechanism to boost their self-confidence. By knowing that they had a safe shelter, they could feel safer while dealing with everyday life's demands. Indeed, as a consequence of the process of taking shelter, of ‘feeling like in their own home’, they could also feel comfortable enough to open up again and communicate:

I do try to talk to people. Sometimes when I just feel... like in my own home. Yeah, my own home. So I invite people to come around here, to chat with people. (Participant 6)

Therefore, along with the aforementioned beneficial effects of medications and close relationships, sheltering seemed to play an important role in maintaining or restoring participants' communicative abilities.

Discussion

Summary of main findings

The four themes identified by our study showed substantial areas of relevance to current models and theories of communication and psychological adjustment to chronic illness. The first theme concerned how HD acts as both director and mediator of the participants' communication, affecting their linguistic abilities by making them feel blocked and not able to speak as they used to, or pulling the strings of a number of collateral conditions that deeply affected their communicative experience, such as chronic fatigue and memory problems. According to the self-regulation model (SRM²⁵), patients' representations and beliefs regarding their chronic illness have a substantial effect on the successful development of coping strategies and ultimately their psychological well-being. The SRM has been specifically adopted with HD,^{26,27} finding that the perceptions of the disease are often characterised by strong illness identity due to its overwhelming perceived effect on all domains of their life. From this perspective, by conceiving HD as an external, separate agent the participants seemed to lay the foundations to fight for a fundamental domain of their own life by taking back some control over communication, which in turn can promote an identity less characterised by their illness.

The importance of control was emphasised in the second theme: Retaking control over the features of communication appeared to represent an effective coping strategy aimed at improving the participants' communicative skills. Whether it was the possibility to avoid phone conversations, the decision to prioritise listening over speaking, the management of scarce energy resources, or being open about HD, regaining aspects of control affected the participants positively by giving them

a chance to break free from the pervasive nature of HD, better accept their condition and become the director of their communication once again. This view fits with the general theory of self-regulation,²⁸ which sees self-regulatory skills as a limited resource that can be exhausted – a phenomenon that has been named self-regulatory fatigue or ego depletion²⁹ and that plays a pivotal role in quality of life and coping in chronic illness.^{30,31} Thus, regulating energy and fatigue by having a choice can represent an effective strategy to deal with the demanding nature of communication.

The third theme focused on how HD threatened the participants' emotional life and stability. Presentations of this included inconsistent feelings of anger and sadness, apathy, and considerably longer emotional 'cool-down' times, which are not uncommon in HD.¹ These ultimately led the participants to perceive their emotions and communication as a source of unsteadiness. One possible solution was represented by improving emotion regulation, which is a concept that refers to the process of influencing which emotions one experiences, as well as when and how such experiences occur,³² and whose importance in mental health and well-being has been widely recognised over the last decade.^{33–35} However, for the participants emotion regulation required engaging with difficult and disturbing thoughts, leading to challenging emotional outflows into their communicative attempts, which caused pervasive feelings of anxiety that could turn communication itself into '*a double-edged sword*.' Nonetheless, where a better level of separation between emotions and communication was achieved (and also thanks to the medication and supportive social relationships), emotion regulation allowed for a substantial improvement in their willingness to communicate.

Last, the fourth theme explored a coping strategy adopted by many participants: the idea of having a personal safe place where

they could take shelter. This was initially identified with their home, but later exceeded the physical dimension and extended to a more abstract idea of safety, a '*little bubble*' where they could switch off. The idea of sheltering provided the participants with a feeling of safety that helped them deal with stressful situations, and eventually shaped an effective mechanism to boost their self-confidence in regulating emotions and regaining control over communication. This finding seems consistent with the positive effect of feelings of safety reported by Hartelius et al.¹⁷

Limitations and future directions

A number of limitations should be considered with our results, such as the inclusion of people at the early to moderate stages only. In this study, the SRM was adopted as a general theoretical framework to interpret the findings, since it has produced meaningful conclusions in previous studies with people affected by HD.^{26,27} However, other theoretical models could also be useful – such as Sharpe and Curran's hierarchical model on world and self-views³⁶ – and future studies could take advantage from their adoption. It should also be remembered that communication is only one aspect of a number of challenges, which these participants were managing. Seeing this in the context of their more general illness experience is also important theoretically. Future research should also aim to find ways to investigate the personal experiences of people at later stages of disease, using more adapted communication methods such as LiteWritersTM.

Conclusion and implications for clinical practice

Our study has helped shed new light on multiple factors that have the potential of informing clinical communicative strategies

between clinicians, caregivers, and patients with HD. First, our results suggest that patients could be encouraged to develop an identity which is less focused on the belief that the biomedical manifestations of HD are responsible for all the difficulties that they experience. Stigmatising language (e.g. 'the Huntington's face') should be avoided. Secondly, patients should be allowed to regain actively some control over various features of communication, by, for example, having a choice on avoiding phone conversations, or managing their own energy in a way which is not then pathologised.

Finally, more effort should be put into helping patients reach a better level of emotion regulation. This could be achieved through the combination of a wide number of factors, including the review of medication regimes, the promotion of close relationships, the refinement of current coping strategies (such as sheltering), and their inclusion in new therapeutic interventions.

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Communication in Huntington's disease: What do we know?

Nicolò Zarotti

In a society characterised by a progressive improvement in life expectancy and a consequent increase in population ageing, working with older people often means having to deal with chronic health conditions, which present challenges from both a medical and psychological perspective. Although often overlooked in place of more physical indices, among the many factors affecting psychological adjustment to chronic conditions a major role is played by interpersonal communication (Stanton, Revenson, & Tennen, 2007).

COMMUNICATION is a broad area which includes several disciplines. It concerns cognitive and clinical psychology – with the behavioural study of language, emotion, signs, gestures and management of space – as well as disability studies, given its particular focus on the social, environmental and cultural factors of interpersonal interaction. It also includes concepts deriving from social cognition, such as empathy and theory of mind, which cannot be operationalised and investigated without the adoption of a social perspective. All these aspects can also be included in the broad cluster of interpersonal communication, which is defined as the discipline that ‘focuses on how people use messages to generate meanings within and across various contexts, cultures, channels, and media’ (Korn et al., 2000).

Embracing such a broad perspective, communication has proved to be related to clinical outcomes of people with chronic illness in different contexts, such as familial relationships (Rosland et al., 2012) or patient-physician interaction (Ong et al., 1995). Moreover, with people affected by non-treatable or life-limiting conditions, communication often represents the only possible form of intervention in terms of palliative care and support (Bury & Wood, 1979). The importance of communication appears even more relevant when chronic

health conditions are non-treatable and progressive, as in the case of Huntington's disease.

Huntington's disease

Huntington's disease (HD) is a hereditary progressive neurodegenerative disorder which affects around 5–10 people per 100,000 (Roos, 2010). It is caused by the mutation of a protein – called huntingtin or HTT – that triggers substantial damage to the basal ganglia. Typical symptoms include involuntary movements (chorea), involuntary abnormal postures (dystonia), cognitive deterioration characterised by problems with memory, psychomotor speed, executive functioning and language, ultimately leading to dementia (Dumas et al., 2013).

HD also poses considerable psychological challenges: the most commonly reported problems include depression, euphoria (or dysphoria), lack of inhibition, increased irritability and aggressiveness, and the tendency to feel anxious, more agitated or apathetic. More rarely, delusions, compulsions, increased sexual drive and hallucinations can be observed (Walker, 2007). Moreover, an increased risk of committing suicide has been documented (Novak & Tabrizi, 2005).

Since the transmission mechanism is autosomal-dominant, every affected individual has a 50 per cent probability of transmitting it to their children. The disease is also

almost fully penetrant, meaning that almost all the individuals with the mutant gene will develop the disease at a certain time in their life. The mean age of onset is 40–50 years. However, juvenile onset (i.e. between age 2 and 20 years) can also occur, as well as very late onset (up to 80 years; Kremer, 2002). The mean life expectancy after the diagnosis is typically 20 years (Folstein, 1989). Genetic testing is available for individuals at risk, allowing them to know if they carry the mutant gene before the onset, but not when the disease will strike.

Current research perspectives on communication in HD

The current literature illustrates that HD has a detrimental impact on many aspects of communication. These include linguistic skills (especially language production), recognition of negative emotions (disgust in particular) and speech articulation. This has proved to be a relatively constant finding among studies that compared people with HD to other neurodegenerative conditions (e.g. Parkinson's disease) and healthy age-matched participants, and should therefore be taken into account in any contexts when supporting people suffering from HD.

However, research on communication in HD appears to be largely dominated by studies on medical and cognitive impairments as assessed from a clinician's perspective (Henry et al., 2005; Rusz et al., 2014; Teichmann et al., 2008). Accordingly, it is not surprising that most studies mainly assess language production, emotion recognition, or speech abilities. Along with these aspects, a fewer number of studies also explored emotion expression, language comprehension and therapeutic interventions. Although certainly important, these are just one part of the elements involved in communication. In particular, the investigation of the perspectives of people with HD on their own communication abilities and functioning is much rarer. Only recently has empirical research started to conceive com-

munication as a broader phenomenon that also embraces social interaction, as opposed to an interest for language and speech alone (Heemskerk & Hamilton, 2010). The very few studies which have assessed communication from such a perspective have reported that communication was negatively influenced by a number of psychosocial factors, including isolation, lack of support, lack of discourse initiative, depression and personality changes (Hartelius et al., 2010; Power et al., 2011).

Clinical implications

Not surprisingly, the clinical implications of the above-mentioned impairments are of considerable importance. As far as language and speech deficits are concerned, decades of studies in the field of neuropsychology – and aphasiology in particular – have largely demonstrated the massive impact they have in the clinical setting. Deficits of language production and articulation, as well as comprehension, can cause misunderstandings and frustration that make communicating verbally much more arduous. Consequently, this can have a negative impact on social and familial interactions, as well as the clinical relationship. Moreover, poor verbal communication can lead to inaccurate clinical assessments due to the inability of the patients to express themselves, understand questions or comprehend the demands of a cognitive task. This is particularly important in the case of Huntington's disease, as the frequently-observed cognitive deterioration and involuntary facial movements often tend to trigger a combination of expressive aphasia and dysarthria. In this regard, as is so often the case in people with a dementia, the information provided by family members and caregivers during the clinical interview is frequently essential to fully appreciate the impact of language impairments on the actual communicative capacity of patients. It is also important to remember that an impaired result on a cognitive test does not always reflect a functional impairment, as

it is not often easy to distinguish between signs of impairment and signs of functional compensations (Leiwo & Klippi, 2000). Indeed, there is evidence of deficits such as echolalia being occasionally used by individuals with HD as a compensation strategy to improve participation in conversations (Saldert & Hartelius, 2011).

The study of facial emotions and their recognition is part of a very long empirical tradition that dates back to the 19th century with Charles Darwin (Sheaffer et al., 2009) and their importance in social cognition and human interactions is widely recognised and listed among the essential components of communication (Frith, 2009). It is therefore not surprising that an impairment of emotion recognition can have severe clinical implications. The inability to understand and correctly respond to emotions, as well as empathise with them, can lead to the same kind of issues observed with poor verbal communication, such as misunderstandings, feelings of awkwardness, and frustration. Moreover, being subtler and less apparent than language impairments, a deficit of emotion recognition may also lead to avoidant behaviour and a further overall deterioration of interpersonal interactions (as observed in the case of alexithymia; Spitzer et al., 2005), as well as a potential for social stigma. This is also likely to be true for the non-facial aspects of emotion recognition such as emotional body language and auditory cues, as well as emotion expression – all aspects that are far more neglected in the empirical literature and would benefit from further attention from both researchers and clinicians.

Finally, as shown by the aforementioned preliminary evidence, a number of psychosocial factors appear to have a considerable impact on communication in HD and should therefore be taken into account whenever possible. In particular, the dynamics of patients' interactions with family members, caregivers and friends should be monitored when assessing communication, as any possible deficits may be mediated by lack of

initiative and support, or by isolation. This also applies to the patients' own ideas about their communicative functioning, as communication may benefit from addressing relatively simple and yet often underrated requests, such as being given more time to formulate sentences or having the interlocutor speak more slowly and clearly (Hartelius et al., 2010). Moreover, the evidence shows that therapeutic interventions may also benefit from the adoption of linguistic and cognitive supplementation strategies (Klasner & Yorkston, 2001), as well as augmentative and alternative communication (AAC) techniques. Among these, Talking Mats (Murphy & Cameron, 2006), a set of textured mats and pictures showing different discussion topics, proved to significantly increase the effectiveness of communication in both dyadic (Ferm et al., 2010) and group interactions (Hallberg et al., 2013) with people affected by HD.

What next?

Overall, the current management of the psychological consequences of HD could greatly benefit from a renewed focus in the clinical setting on the issues involving interpersonal communication and communicative skills. Furthermore, a number of topics have been particularly neglected so far in the empirical literature and would benefit from future attention from both researchers and clinicians. These include emotion expression and non-facial components of emotion recognition, patients' perspectives on their communicative skills and efficiency, and the effect of impairments and social factors on patients' functional communicative capacity. All these aspects of communication are likely to have relevant clinical implications, and if addressed correctly have the potential of refining current therapeutic approaches, as well as informing new interventions aimed at improving communication effectiveness and ultimately quality of life.

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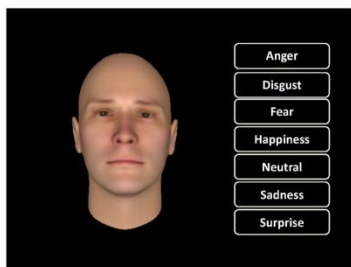
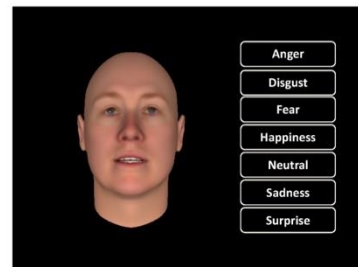
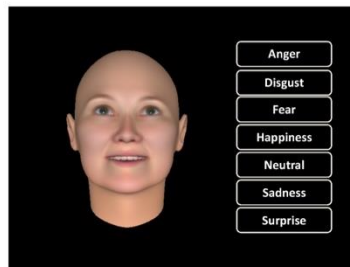
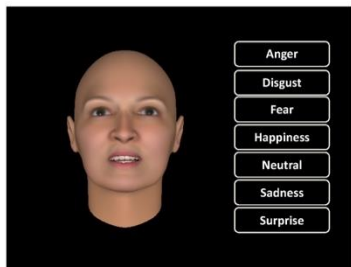
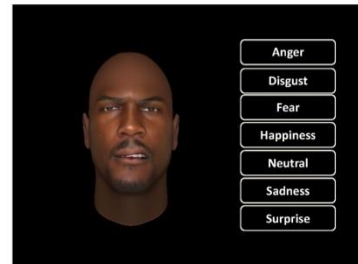
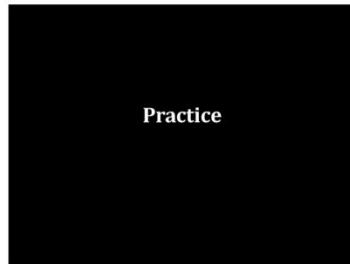
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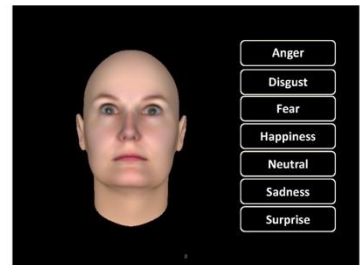
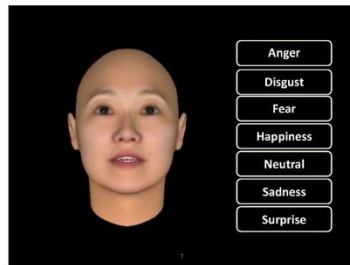
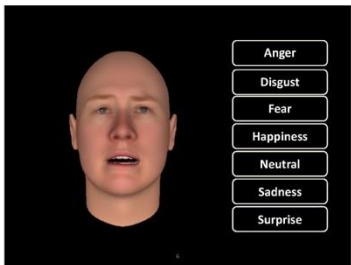
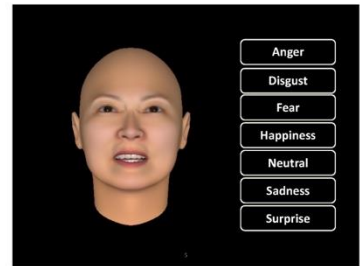
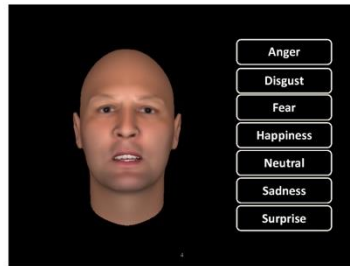
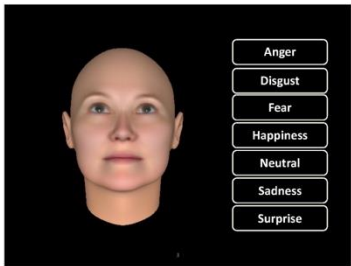
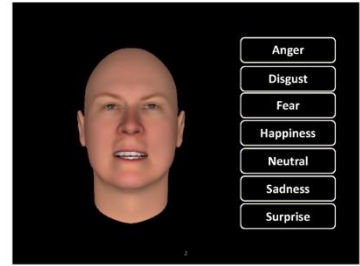
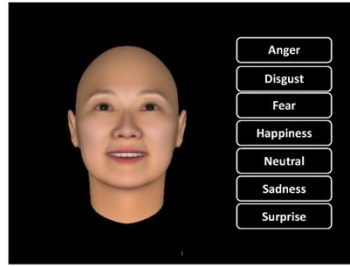
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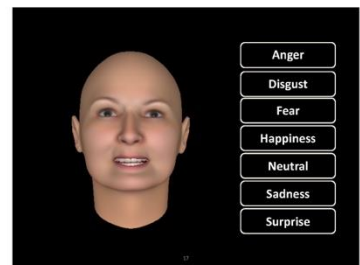
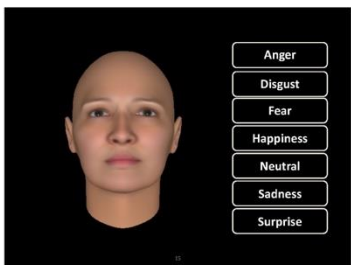
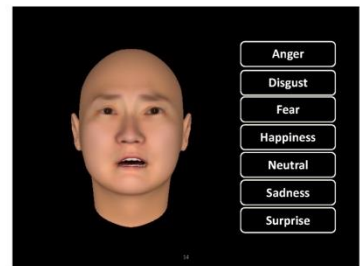
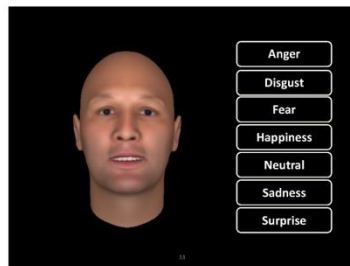
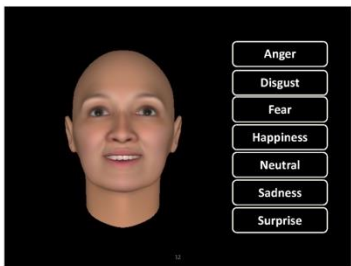
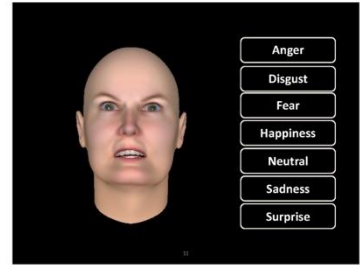
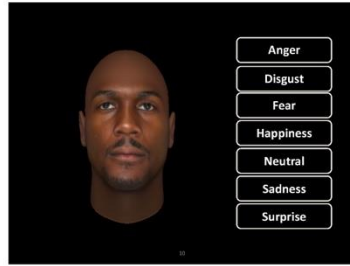
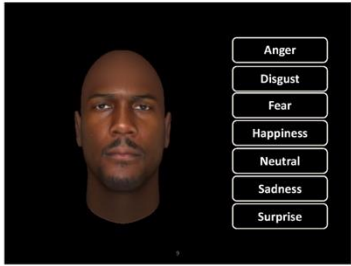
Appendix 5

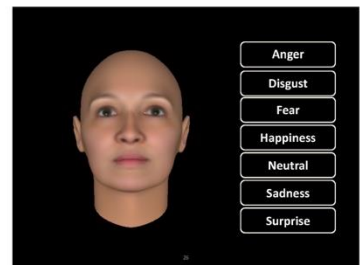
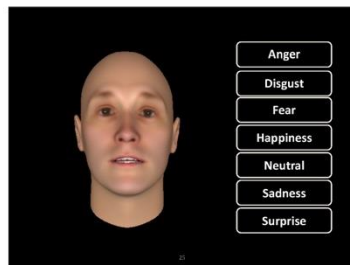
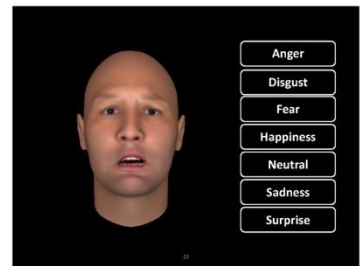
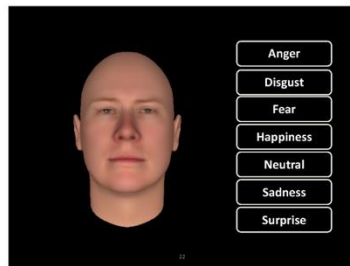
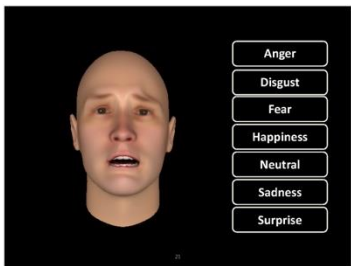
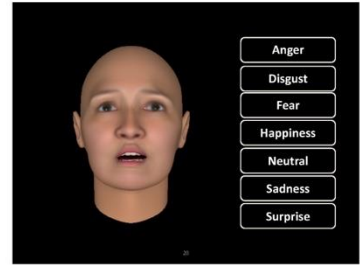
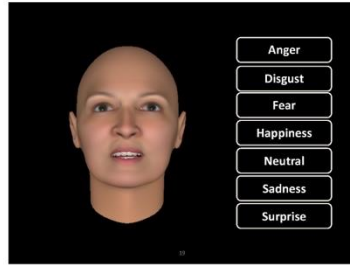
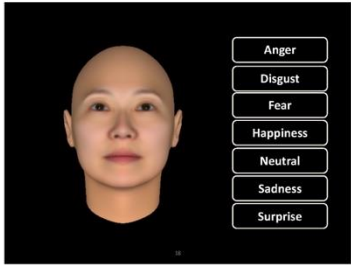
Research Tests and Questionnaires

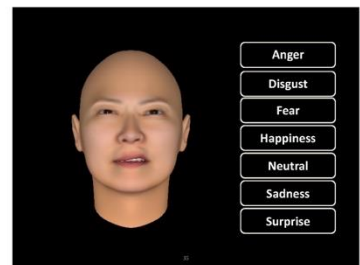
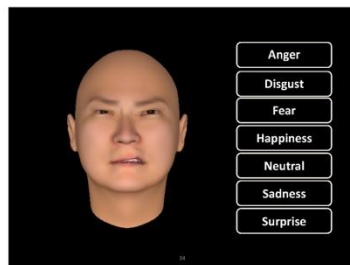
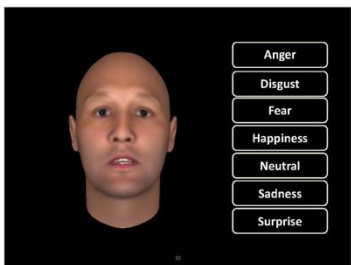
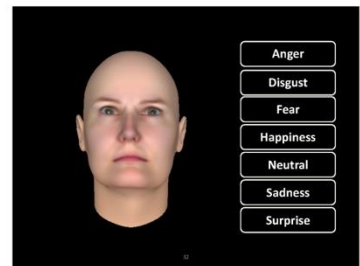
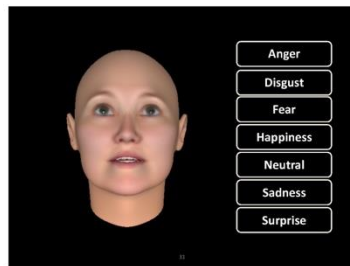
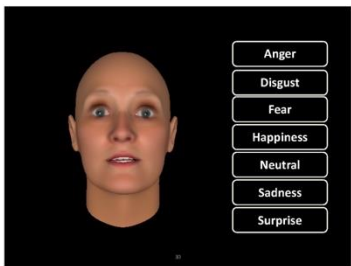
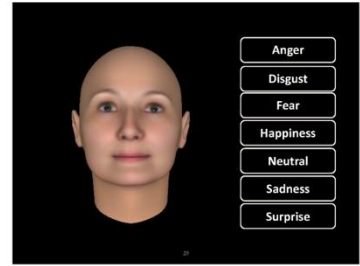
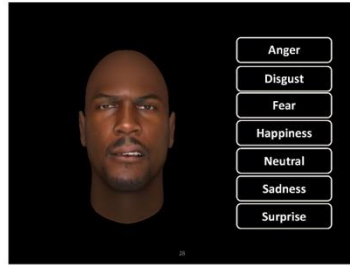
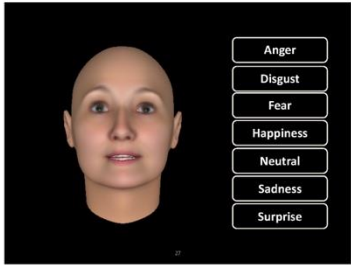
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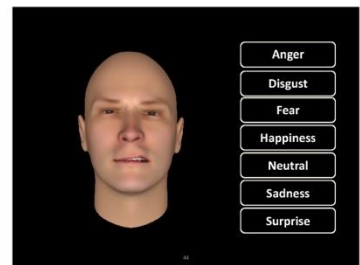
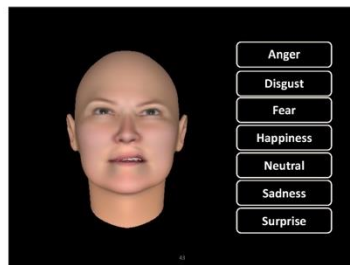
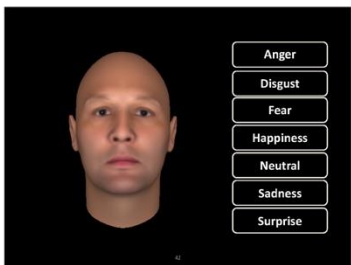
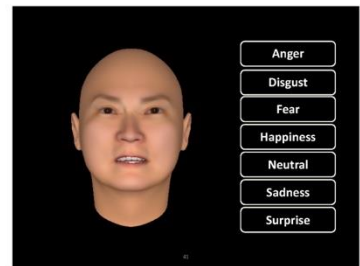
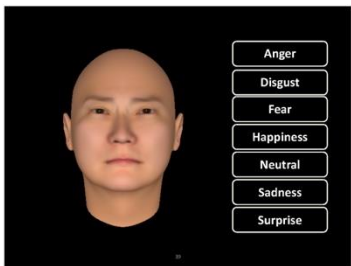
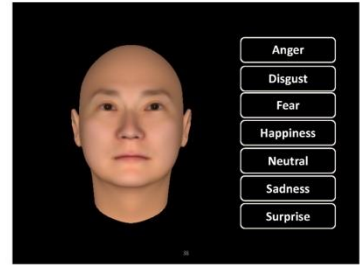
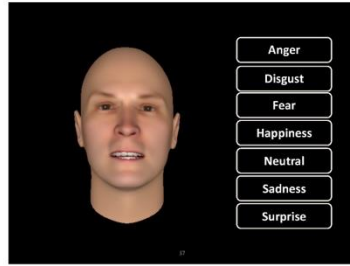
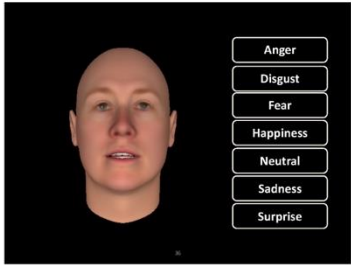


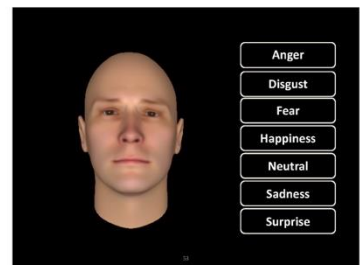
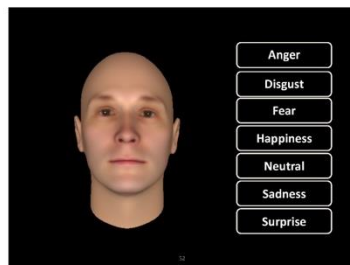
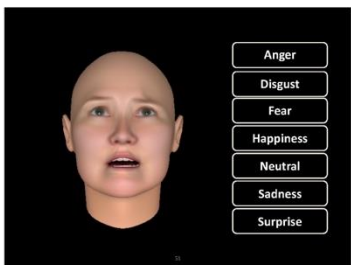
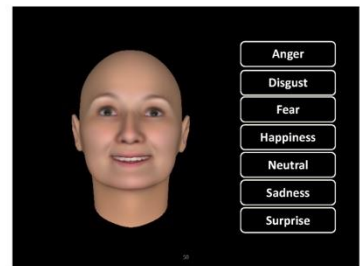
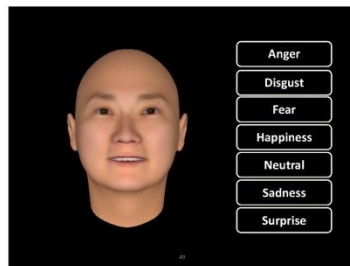
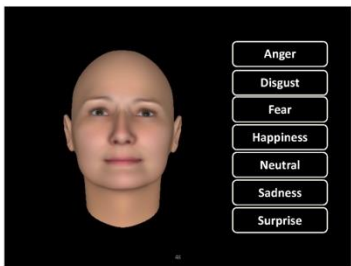
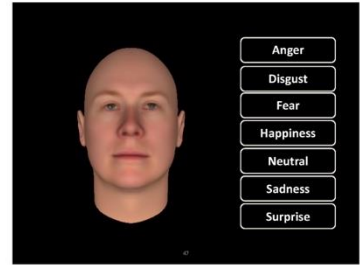
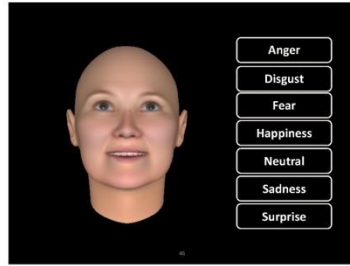
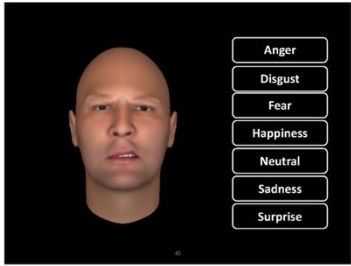


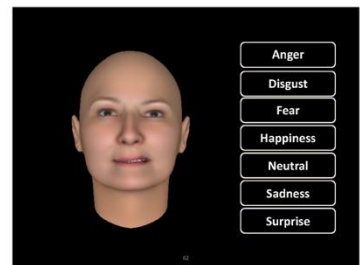
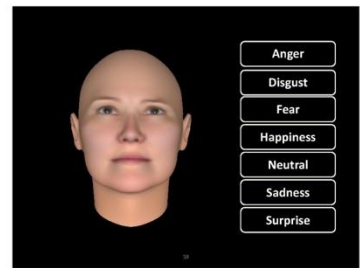
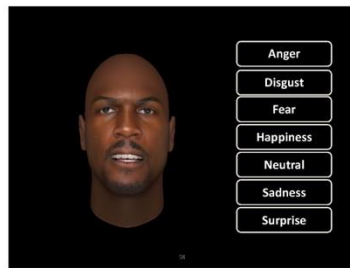
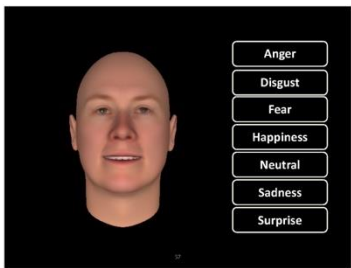
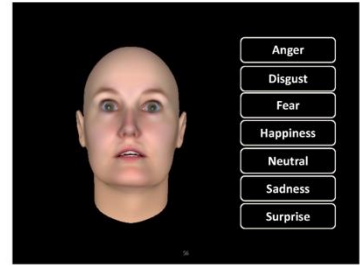
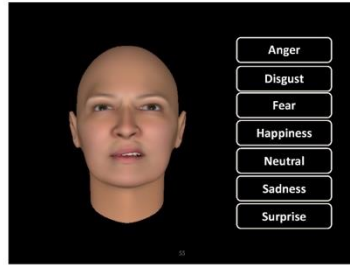
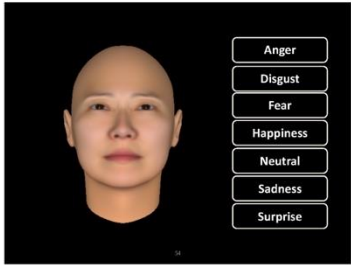


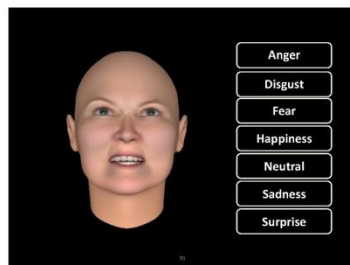
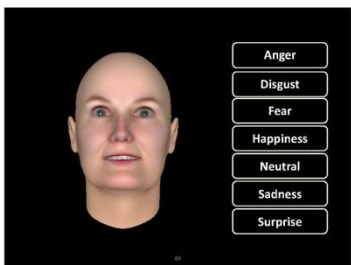
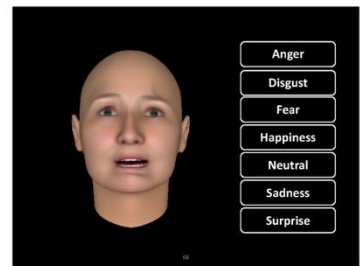
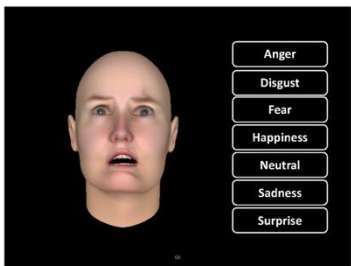
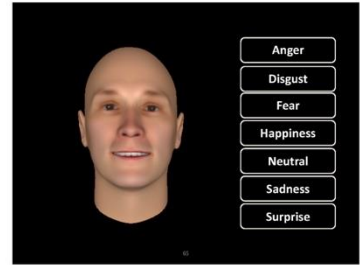
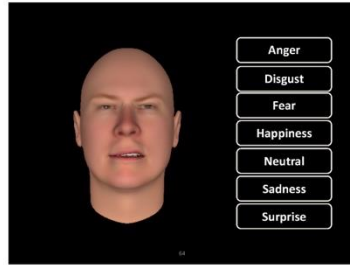
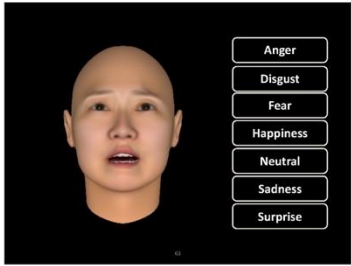













Body Trial

Practice




Anger
Disgust
Fear
Happiness
Neutral
Sadness
Surprise




Anger
Disgust
Fear
Happiness
Neutral
Sadness
Surprise



Anger
Disgust
Fear
Happiness
Neutral
Sadness
Surprise



Anger
Disgust
Fear
Happiness
Neutral
Sadness
Surprise



Anger
Disgust
Fear
Happiness
Neutral
Sadness
Surprise



Anger
Disgust
Fear
Happiness
Neutral
Sadness
Surprise



Anger
Disgust
Fear
Happiness
Neutral
Sadness
Surprise

Trial



- Anger
- Disgust
- Fear
- Happiness
- Neutral
- Sadness
- Surprise



- Anger
- Disgust
- Fear
- Happiness
- Neutral
- Sadness
- Surprise



- Anger
- Disgust
- Fear
- Happiness
- Neutral
- Sadness
- Surprise



- Anger
- Disgust
- Fear
- Happiness
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- Surprise



- Anger
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- Fear
- Happiness
- Neutral
- Sadness
- Surprise



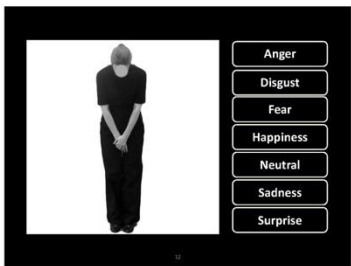
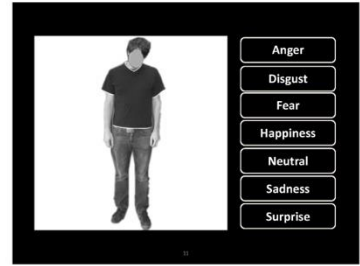
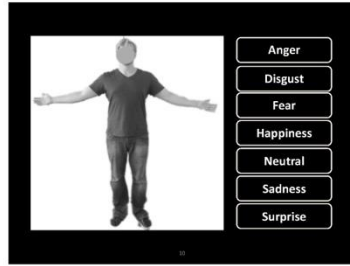
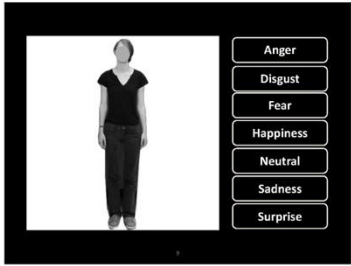
- Anger
- Disgust
- Fear
- Happiness
- Neutral
- Sadness
- Surprise

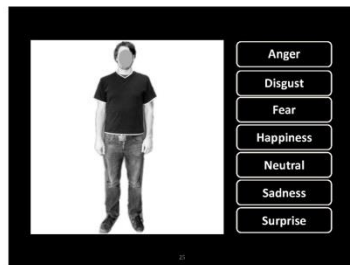
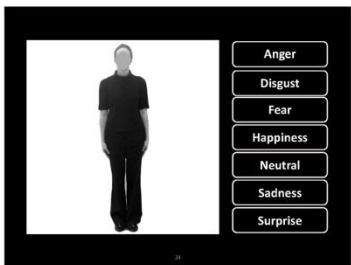
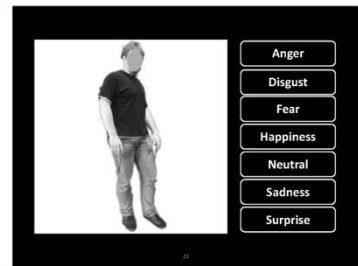
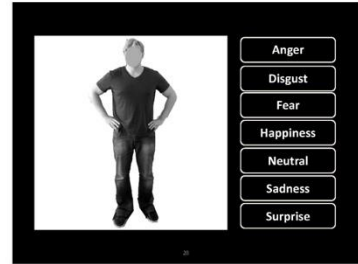
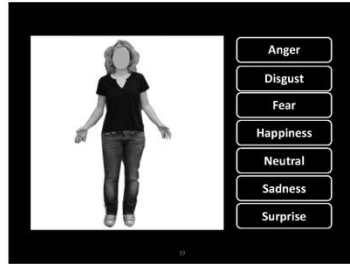
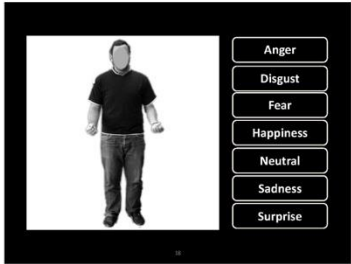


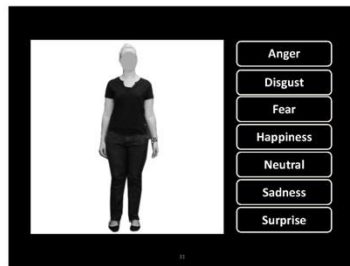
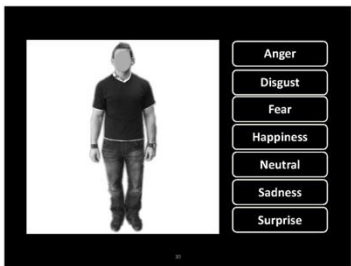
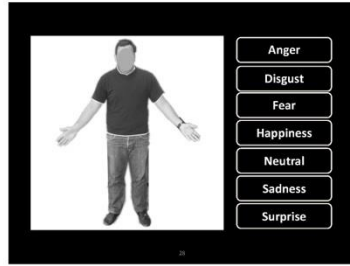
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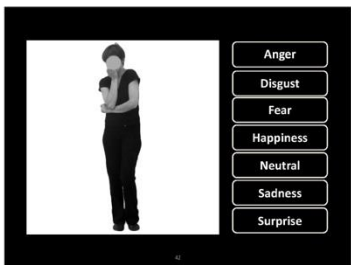
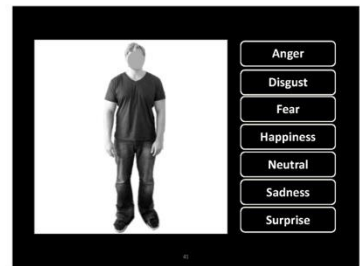
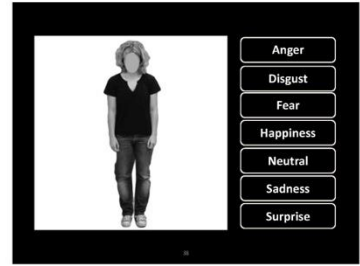


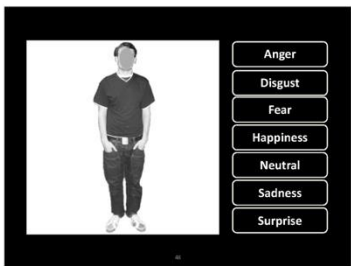
- Anger
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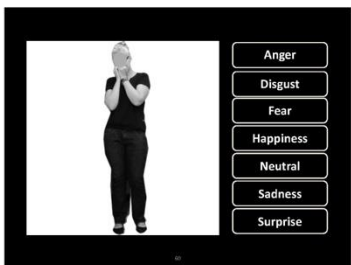
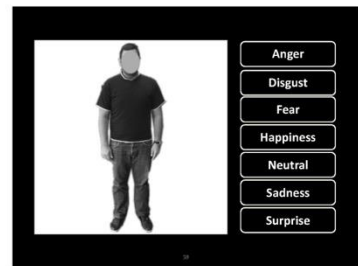
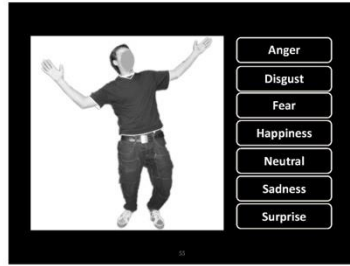


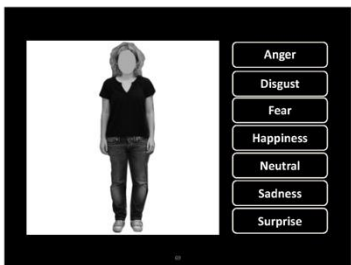
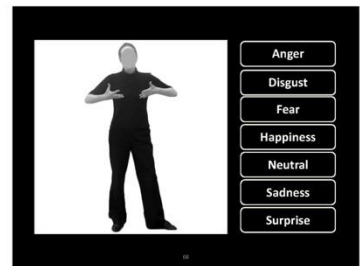












Difficulties in Emotion Regulation Scale (DERS; Gratz & Roemer, 2004)

The next statements are also about you. Please choose the response that best fits for you.

(DERS)

		Almost never	Some times	About half the time	Most of the time	Almost always
1	I am clear about my feelings.	1	2	3	4	5
2	I pay attention to how I feel.	1	2	3	4	5
3	I experience my emotions as overwhelming and out of control.	1	2	3	4	5
4	I have no idea how I am feeling.	1	2	3	4	5
5	I have difficulty making sense out of my feelings.	1	2	3	4	5
6	I am attentive to my feelings.	1	2	3	4	5
7	I know exactly how I am feeling.	1	2	3	4	5
8	I care about what I am feeling.	1	2	3	4	5
9	I am confused about how I feel.	1	2	3	4	5
10	When I'm upset, I acknowledge my emotions.	1	2	3	4	5
11	When I'm upset, I become angry with myself for feeling that way.	1	2	3	4	5
12	When I'm upset, I become embarrassed for feeling that way.	1	2	3	4	5
13	When I'm upset, I have difficulty getting work done.	1	2	3	4	5
14	When I'm upset, I become out of control.	1	2	3	4	5
15	When I'm upset, I believe that I will remain that way for a long time.	1	2	3	4	5
16	When I'm upset, I believe that I will end up feeling very depressed.	1	2	3	4	5
17	When I'm upset, I believe that my feelings are valid and important.	1	2	3	4	5
18	When I'm upset, I have difficulty focusing on other things.	1	2	3	4	5
19	When I'm upset, I feel out of control.	1	2	3	4	5
20	When I'm upset, I can still get things done.	1	2	3	4	5

		Almost never	Some times	About half the time	Most of the time	Almost always
21	When I'm upset, I feel ashamed with myself for feeling that way.	1	2	3	4	5
22	When I'm upset, I know that I can find a way to eventually feel better.	1	2	3	4	5
23	When I'm upset, I feel like I am weak.	1	2	3	4	5
24	When I'm upset, I feel like I can remain in control of my behaviours.	1	2	3	4	5
25	When I'm upset, I feel guilty for feeling that way.	1	2	3	4	5
26	When I'm upset, I have difficulty concentrating.	1	2	3	4	5
27	When I'm upset, I have difficulty controlling my behaviours.	1	2	3	4	5
28	When I'm upset, I believe that there is nothing I can do to make myself feel better.	1	2	3	4	5
29	When I'm upset, I become irritated with myself for feeling that way.	1	2	3	4	5
30	When I'm upset, I start to feel very bad about myself.	1	2	3	4	5
31	When I'm upset, I believe that wallowing in it is all I can do.	1	2	3	4	5
32	When I'm upset, I lose control over my behaviours.	1	2	3	4	5
33	When I'm upset, I have difficulty thinking about anything else.	1	2	3	4	5
34	When I'm upset, I take time to figure out what I'm really feeling.	1	2	3	4	5
35	When I'm upset, it takes me a long time to feel better.	1	2	3	4	5
36	When I'm upset, my emotions feel overwhelming.	1	2	3	4	5

Hospital Anxiety and Depression Scale (HADS; Zigmond & Snaith, 1983)

Instructions: Please read each item and circle the reply which comes closest to how you have been feeling in the past week. Don't take too long over your replies: your immediate reaction to each item will probably be more accurate than a long thought out response.

I feel tense or 'wound up':	A	I feel as if I am slowed down:	D
Most of the time	3	Nearly all of the time	3
A lot of the time	2	Very often	2
Time to time, occasionally	1	Sometimes	1
Not at all	0	Not at all	0
I still enjoy the things I used to enjoy:	D	I get a sort of frightened feeling like 'butterflies in the stomach':	A
Definitely as much	0	Not at all	0
Not quite so much	1	Occasionally	1
Only a little	2	Quite often	2
Not at all	3	Very often	3
I get a sort of frightened feeling like something awful is about to happen:	A	I have lost interest in my appearance:	D
Very definitely and quite badly	3	Definitely	3
Yes, but not too badly	2	I don't take as much care as I should	2
A little, but it doesn't worry me	1	I may not take quite as much care	1
Not at all	0	I take just as much care as ever	0
I can laugh and see the funny side of things:	D	I feel restless as if I have to be on the move:	A
As much as I always could	0	Very much indeed	3
Not quite so much now	1	Quite a lot	2
Definitely not so much now	2	Not very much	1
Not at all	3	Not at all	0
Worrying thoughts go through my mind:	A	I look forward with enjoyment to things:	D
A great deal of the time	3	A much as I ever did	0
A lot of the time	2	Rather less than I used to	1
From time to time but not too often	1	Definitely less than I used to	3
Only occasionally	0	Hardly at all	2
I feel cheerful:	D	I get sudden feelings of panic:	A
Not at all	3	Very often indeed	3
Not often	2	Quite often	2
Sometimes	1	Not very often	1
Most of the time	0	Not at all	0
I can sit at ease and feel relaxed:	A	I can enjoy a good book or radio or TV programme:	D
Definitely	0	Often	0
Usually	1	Sometimes	1
Not often	2	Not often	2
Not at all	3	Very seldom	3

Reading the Mind in the Eyes test (RME; Baron-Cohen et al., 2001)

For each set of eyes, choose and select which word best describes what the person in the picture is thinking or feeling. You may feel that more than one word is applicable but please choose just one word, the word which you consider to be most suitable.

Before making your choice, make sure that you have read all 4 words. You should try to do the task as quickly as possible, but you will not be timed.



Jealous

Panicked

Arrogant

Hateful



Playful

Comforting

Irritated

Bored

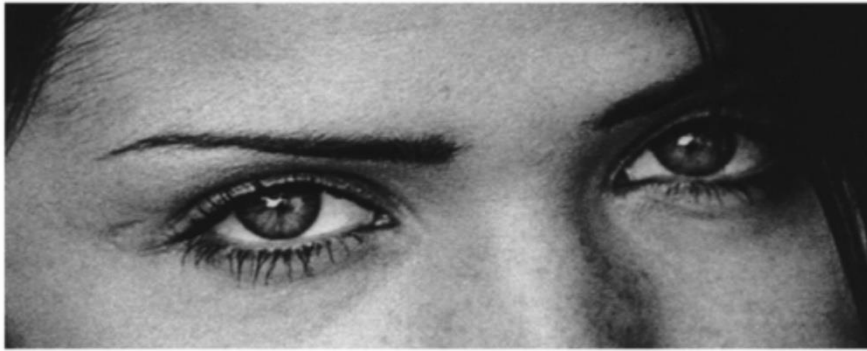


Terrified

Upset

Arrogant

Annoyed



Joking

Flustered

Desire

Convinced



Joking

Insisting

Amused

Relaxed



Irritated

Sarcastic

Worried

Friendly



Aghast

Fantatising

Impatient

Alarmed



Apologetic

Friendly

Uneasy

Dispirited



Despondent

Relieved

Shy

Excited



Annoyed

Hostile

Horrified

Preoccupied



Cautious

Insisting

Bored

Aghast



Terrified

Amused

Regretful

Flirtatious

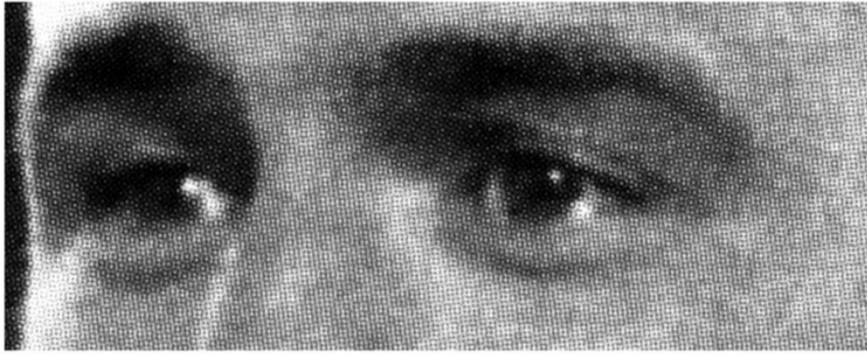


Indifferent

Embarrassed

Skeptical

Dispirited



Decisive

Anticipating

Threatening

Shy



Irritated

Disappointed

Depressed

Accusing



Contemplative

Flustered

Encouraging

Amused



Irritated

Thoughtful

Encouraging

Sympathetic



Doubtful

Affectionate

Playful

Aghast



Decisive

Amused

Aghast

Bored



Arrogant

Grateful

Sarcastic

Tentative



Dominant

Friendly

Guilty

Horrified



Embarrassed

Fantatising

Confused

Panicked



Preoccupied

Grateful

Insisting

Imploring



Contented

Apologetic

Defiant

Curious

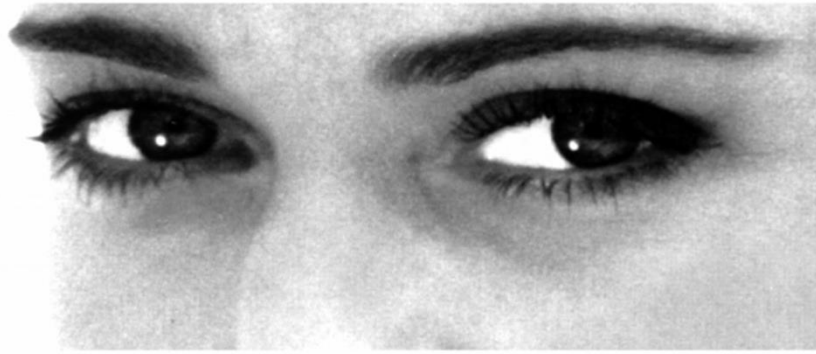


Pensive

Irritated

Excited

Hostile



Panicked

Incredulous

Despondent

Interested



Alarmed

Shy

Hostile

Anxious



Joking

Cautious

Arrogant

Reassuring



Interested

Joking

Affectionate

Contented



Impatient

Aghast

Irritated

Reflective



Grateful

Flirtatious

Hostile

Disappointed



Ashamed

Confident

Joking

Dispirited



Serious

Ashamed

Bewildered

Alarmed



Embarrassed

Guilty

Fantatising

Concerned



Aghast

Baffled

Distrustful

Terrified



Puzzled

Nervous

Insisting

Contemplative



Ashamed

Nervous

Suspicious

Indecisive

Total Functional Capacity Scale (TFC; Shoulson & Fahn, 1979)

STANDARD [STUDY NAME]
 UNIFIED HUNTINGTON'S DISEASE RATING SCALE '99
 (TFC)

00 00

SUBJECT ID VISIT NO

INITIALS SITE NO VISIT DATE
 MM DD YYYY

VI. FUNCTIONAL CAPACITY

70. OCCUPATION 70.
 0 = unable
 1 = marginal work only
 2 = reduced capacity for usual job
 3 = normal

71. FINANCES 71.
 0 = unable
 1 = major assistance
 2 = slight assistance
 3 = normal

72. DOMESTIC CHORES 72.
 0 = unable
 1 = impaired
 2 = normal

73. ADL 73.
 0 = total care
 1 = gross tasks only
 2 = minimal impairment
 3 = normal

74. CARE LEVEL 74.
 0 = full time skilled nursing
 1 = home or chronic care
 2 = home

INFORMATIONAL SOURCES

75. Was the Functional Capacity information obtained from: 75.
 1 = Subject only
 2 = Subject and family/companion

76. Functional Examiner 76.
 STAFF CODE