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“We Live With it” - Portuguese Patients and Relatives’ perspectives of Physiotherapy role in the course of Huntington’s Disease - A Qualitative Study

Dissertação de Mestrado em Prática Avançada em Fisioterapia em Neurologia

Orientador: Prof. João Casaca Carreira

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“Nós vivemos com isto” – Perspetivas dos Pacientes e Familiares de Doença de Huntington sobre o papel da Fisioterapia – Estudo Qualitativo

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Julho 2023

Relatório de Investigação apresentado para cumprimento dos requisitos necessários à obtenção do grau de Mestre em Fisioterapia em Neurologia, realizado sob a orientação científica do Professor Doutor João Casaca Carreira.

Declarações

Declaro que este Relatório de Projeto de Investigação é o resultado da minha investigação pessoal e independente. O seu conteúdo é original e todas as fontes consultadas estão devidamente mencionadas no texto, nas notas e na bibliografia.

O candidato,

_____

Setúbal, 23 de Junho de 2023

Declaro que este Relatório de Projeto de Investigação se encontra em condições de ser apresentado a provas públicas.

O (A) orientador(a),

_____

Setúbal, 23 de Junho 2023

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Abstract

"We Live With it" - Portuguese Patients and Relatives' perspectives of Physiotherapy role in the course of Huntington's Disease - A Qualitative Study

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Background: Huntington's Disease is a hereditary autosomal dominant, and neurodegenerative condition. It combines motor, cognitive and behavioural disturbances. Nowadays, Physiotherapy for Huntington´s encompasses interventions such as exercise, physical activity promotion, balance, task-specific training and education. It seeks to enable patients with impairments, activity limitations and participation restrictions to reach their optimal functional level. However there are just a few articles that relate Huntington's Disease with Physiotherapy, and as far as we know there is none in Portugal that truly evaluates the patient's and families' needs and how Physiotherapy helps.

Purpose: Understand how Physiotherapy helps in the life, challenges and problem-solving of Huntington's Disease patients and their relatives. Additionally, understand what we can change in Physiotherapy to improve the integration of patients in society.

Methods: A phenomenology qualitative work, with semi-structured, audio and video record interviews, made preferentially through APDH and other contacts. Interviews were transcribed and analysed following the theory of thematic analysis to identify salient themes.

Results: A total of 18 participants were interviewed (6 patients and 12 relatives). Five themes derived: Objectives and ICF (Theme 1), Society and HD (Theme 2), Health Care and Needs in HD (Theme 3), Physiotherapy and HD (Theme 4), and Family and HD (Theme 5).

Conclusions: Physiotherapy represents a powerful tool in the lifelong management of Huntington's Disease. It seems to have a role in daily activities, autonomy, self-confidence, family management and strength and walking

capacity. Although there is a need for improving the human-centred approach, to meet patients' and family members' specific objectives in order to reduce their limitations in social participation. Future studies should evaluate the quality and the content of the Physiotherapy sessions.

Keywords: Huntington’s Disease, rehabilitation, Quality of Life, family, society.

Resumo – “Nós vivemos com isto” - Perspetivas dos Pacientes e Familiares de Doença de Huntington sobre o papel da Fisioterapia – Estudo Qualitativo

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Introdução: A Doença de Huntington é uma condição hereditária, autossómica dominante e de carácter neurodegenerativo. Combina alterações motoras, cognitivas e comportamentais. Atualmente a Fisioterapia para a Doença de Huntington engloba intervenções como exercício, promoção da atividade física, treino de equilíbrio e tarefas específicas e educação. Procura desta forma capacitar os doentes que apresentam limitações nas atividades e restrições na participação a atingir o seu estado funcional. Porém a literatura que relaciona a Doença de Huntington com a Fisioterapia é escassa, não sendo conhecido nenhum estudo em Portugal que avalie as necessidades dos doentes e dos seus familiares em relação à Fisioterapia e como esta os ajuda.

Objetivo: Perceber como é que a Fisioterapia tem um papel na vida, desafios e resolução de problemas dos doentes e dos seus familiares. Adicionalmente procuramos perceber o que é que se pode alterar na intervenção da Fisioterapia de forma a melhorar a integração dos doentes na sociedade.

Métodos: Estudo qualitativo fenomenológico com entrevistas semiestruturadas áudio e vídeo gravadas, feitas preferencialmente através da Associação Portuguesa de Doentes de Huntington e outros contactos. Depois de transcritas foram analisadas através da análise temática de forma a identificar os temas relevantes.

Resultados: Um total de 18 entrevistas foram incluídas (6 utentes e 12 familiares). Foram identificados 5 temas: ICF e Objetivos (Tema 1), Sociedade e DH (Tema 2), Necessidades e Cuidados de Saúde (Tema 3), Fisioterapia e DH (Tema 4), Família e DH (Tema 5).

Conclusão: A Fisioterapia tem um forte contributo na gestão da Doença de Huntington. Parece ter um papel ao nível das atividades diárias, autonomia,

confiança, gestão familiar, força e capacidade de marcha. Porém é preciso melhorar os cuidados centrados no utente de forma a alcançar os objetivos dos mesmos e das suas famílias de forma a reduzir as limitações sociais. Estudos futuros devem avaliar a qualidade e o conteúdo das sessões de Fisioterapia.

Palavras-Chave: Doença de Huntington, reabilitação, Qualidade de Vida, família, sociedade;

Index

List of Abbreviations

HD – Huntington's Disease;

PD – Parkinson’s Disease;

ADL – Activities of Daily Living;

QoL – Quality of life;

HTT - Huntingtin Gene;

EHDN – European Huntington’s Disease Network

ICF - International Classification of Functioning Disability and Health;

QR – Qualitative Research;

SRQR - Standards for Reporting Qualitative Research;

APDH – Associação Portuguesa de Doentes de Huntington;

IPS – Instituto Politécnico de Setúbal

IA – Inês Agostinho

JC – João Casaca

MP – Maria Patriarca

GRS – Global Rating Scale

Literature Review

Huntington's Disease - Introduction

Huntington´s Disease (HD) is a hereditary autosomal dominant, progressive and neurodegenerative condition (Ghosh & Tabrizi, 2018; Roos, 2010; Sellers et al., 2020; Teixeira et al., 2020). It was described for the first time by George Huntington in 1872. In 1983 HD was linked to the short arm of chromosome 4 and in 1993 the HD gene was found (Ghosh & Tabrizi, 2018; Roos, 2010).

The prevalence of HD is estimated to be about 5-10:100.000 in Europe and considered less common in southern Europe. In Portugal, it is estimated to be the same, 5-10:100.000 (Barboza & Ghisi, 2018; Carmo Costa et al., 2003; Oliveira et al., 2020). It affects both sexes, and each child of an HD parent has a 50:50 chance of inheriting the mutation (Etchegary, 2011; Layton et al., 2021).

HD combines motor disturbances, cognitive and behavioural symptoms. Sometimes, in the beginning, these symptoms may be easily mistaken with other conditions such as schizophrenia, Parkinson's Disease (PD) or Alzheimer's (Frich et al., 2014; Oliveira et al., 2020). The average age of diagnosis is 40-45 years; however, the symptoms are often present earlier with patients experiencing psychiatric and cognitive symptoms for many years before their official diagnosis (Frich et al., 2014; Ghosh & Tabrizi, 2018; Roos, 2010).

These symptoms display a gradual worsening in mobility and participation of these patients, which directly affects their activities of daily living (ADL) and quality of life (QoL) (Al-Wardat et al., 2022; Busse et al., 2008; Quinn & Busse, 2012). There is no cure for HD, for now, the current treatment of HD consists mainly of symptom management and improving QoL (Al-Wardat et al., 2022; Busse et al., 2008; Frich et al., 2014). The management of HD requires coordination between a lot of health professionals of different domains, including for example Neurologists, Psychiatrists, Psychologists, Physiotherapists, Nurses, Occupational Therapists and Social Services (Baig et al., 2016).

HD is complex, and this complexity leads to a lot of problems and challenges in the management and treatment of this condition, requiring a high level of support from the community, family, and others (Layton et al., 2021).

Pathogenesis of HD

HD is caused by an expanded CAG triplet repeat in the Huntingtin Gene (HTT), which encodes an abnormal polyglutamine expansion (Baig et al., 2016; Barboza & Ghisi, 2018). The gold standard for genetic confirmation is the demonstration of CAG expansion of at least 36 repeats on the HTT gene (Erkkinen et al., 2018). Usually, CAG repeats of 36-39 are considered reduced penetrance and 40 or more are fully penetrant (Erkkinen et al., 2018). One of the most sensitive regions is the striatum, with about 57% loss of cross-sectional area from the caudate nucleus and about 65% loss of the putamen. There is also a tissue reduction in the thalamus, substantia nigra and subthalamic nucleus (Malkani et al., 2018). Studies in cadavers revealed massive neuronal degeneration in the striatum in advanced HD. The GABAergic neurons that project to the globus pallidus and substancia nigra are lost, and there is an atrophy of the cerebral cortex, subcortical white matter, thalamus and hypothalamic nuclei (Ghosh & Tabrizi, 2018).

Symptoms

HD has a progressive dysfunction across multiple neurologic systems including motor, cognitive and psychiatric systems (Erkkinen et al., 2018). It is quite common to have psychiatric or cognitive symptoms and subtle motor signs many years before motor signs (Ghosh & Tabrizi, 2018). The timing of the onset of the symptoms is partially determined by the number of CAG repeats (Erkkinen et al., 2018). Patients have an equivocal onset of HD manifestations when there is the presence of extrapyramidal signs such as chorea, dystonia, bradykinesia and rigidity, and there is no other explanation than HD (Ghosh & Tabrizi, 2018).

One of the major symptoms is the chorea, which is defined as an abnormal and unexpected involuntary movement (involuntary jerking dance), it begins in distal muscles such as fingers, toes and small facial muscles with progression to the proximal ones (Erkkinen et al., 2018; Januário, 2011; Roos, 2010).

Dystonia is also common and an important motor sign in HD, it is characterised by slower movements with sustained muscle contractions and increased muscle tone leading to an abnormal posture with rotation of the trunk or limbs, and it can be one of the first motor signs of HD (Ghosh & Tabrizi, 2018; Roos, 2010). Other motor symptoms such as ataxia, motor persistence, atypical parkinsonism and eye movement abnormalities can appear in HD (Malkani et al., 2018).

All of these motor signs have an impact on gait, motor function and balance, and due to an increased fall risk, they are also associated with a lower QoL (Roos, 2010).

Besides motor manifestations, cognitive and psychiatric are frequent too, they can range from subtle deficits to frank dementia (Ghosh & Tabrizi, 2018; Sellers et al., 2020). Cognitive deficits primarily involve executive functions such as multi-tasking, planning and memory recall (Erkkinen et al., 2018). The most described are depression, apathy, anxiety, obsessive-compulsive symptoms and irritability (Sellers et al., 2020).

The progression of the symptoms can be highly variable ranging from 4 to 38 years. Consequently, about 88% of people with HD require residential care, long-term medical resources, and health support for themselves and their families (Power et al., 2011). Because the mean duration of the disease is 17-20 years, HD leads to more dependency, reduced QoL and finally, death. The most common cause of death is respiratory problems, like pneumonia, followed by suicide (Roos, 2010). Therefore, HD brings an emotional stress and suffering among families; so, there are reports of suicide justified by depression, stress and physical dependence (Stephanie da Silva et al., 2015).

Guidelines for treatment of HD

Since there is no cure for HD, the treatment focuses on managing and alleviating symptoms (Bachoud-Lévi et al., 2019; Erkkinen et al., 2018).

Motor symptoms are the best-known and the most visible symptoms of HD. As they are easily detected, they are often well tolerated in contrast to cognitive and behavioural symptoms (Bachoud-Lévi et al., 2019). Nonpharmacologic

strategies such as Physiotherapy, Occupational Therapy, Speech Therapy, Dietary Consultation, structured schedules and Social Work Services are imperative to HD patients and their families (Erkkinen et al., 2018).

The European Huntington's Disease Network (EHDN) commissioned an international task force to provide a global evidence-based recommendation for everyday clinical practice for HD patients. The objective of this work was to standardize pharmacologic, surgical, and non-pharmacologic treatment (Bachoud-Lévi et al., 2019).

Cognitive disorders, in addition to behavioural symptoms, are the major cause of family disruption and social withdrawal. Based on the present knowledge, no pharmacological treatment is recommended for the treatment of cognitive symptoms. Despite that, the Guidelines suggest multiple rehabilitation such as Speech Therapy, Occupational Therapy, Cognitive and Psychomotricity, to improve or stabilize these symptoms (Grade B) (Bachoud-Lévi et al., 2019).

Regarding motor capacities, in a general way, the Guideline refers that early referral to Physiotherapy is recommended to ensure long-term functional independence. Physiotherapy and/or exercise programs (GRADE B) are beneficial for functionality, motor function and independence in combination with pharmacological treatments (Bachoud-Lévi et al., 2019).

Physiotherapy in HD

The World Health Organisation offers the International Classification of Functioning Disability and Health (ICF), which is a biopsychosocial framework with a person-centred approach (Layton et al., 2021). This tool identifies the aspects of the person, the environment and the task, and enables the person to describe their functional status, and the barriers and facilitators to functioning (Layton et al., 2021). As we already saw, individuals with HD develop physical, cognitive and psychological impairments that affect negatively their participation in life activities and reduce their QoL (Quinn et al., 2020).

Physiotherapy interventions such as balance exercises, aerobic exercises, gait training, physical activity promotion, task-specific training, education, strength exercise and respiratory therapy become a fundamental part of managing HD (Al-Wardat et al., 2022; Quinn et al., 2020).

It is important to notice that aerobic exercise is suggested with strong evidence as it should be prescribed at moderate intensity paired with upper and lower body strengthening 3 times per week for a minimum of 12 weeks (Quinn et al., 2020). Additionally and with strong evidence too, is one-on-one supervised gait training in order to improve spatiotemporal measures of gait (walking speed and step length) (Quinn et al., 2020). Therefore, these Guidelines, recommend with a weak recommendation the balance exercises, although, if prescribed, they should be individualise and with moderate frequency and intensity (Quinn et al., 2020).

In point out, there are some limitations to this Guideline such as the small number of available interventions, and the lack of control groups in some studies, which can limit the ability to interpret the results. Other than these, it is important to notice that EHDN is working to review these Guidelines in 2024.

Despite the points presented before, through a partnership with family and community, Physiotherapy seeks to enable patients with impairments, activity limitations and participation restrictions to reach their optimal physical and social functional level and plays an important role to maintain independence (Busse et al., 2008; B. E. Gibson & Martin, 2003; Quinn et al., 2020; Quinn & Busse, 2012).

Family and HD

The time of diagnosis is a very challenging moment for the whole family and patients as everyone has to adjust their lives. Family members report negative emotions including anxiety, depression, fear and loneliness (Etchegary, 2011; Røthing et al., 2015b). HD brings a lot of pressure on the family as well in terms of care and sometimes even to a couple, in which they had to deal with the possibility of transmission of the disease (Etchegary, 2011; Power et al., 2011; Røthing et al., 2015b). Family members play an important role as “caregivers”, and it is important to mention that sometimes their knowledge about HD is limited and only find out about HD when a relative or themselves are diagnosed (Oliveira et al., 2020; Røthing et al., 2015a).

Health professionals have an important role in supporting patients and their families since they can give valuable input to understand the barriers and facilitators of patients to engage in rehabilitation programs (Frich et al., 2014)

Health services are increasingly considering family members to be partners in collaborative teams, to achieve higher quality and effectiveness (Røthing et al., 2015b). Additionally, health professionals are working on taking active steps to involve the families even more and improve the QoL of HD patients (Røthing et al., 2015a)

So, the primary objective of this study is to understand, from the point of view of patients and their families how Physiotherapy helps in the life, challenges and problem-solving of HD patients and their relatives. Additionally, we seek to understand what we can change in Physiotherapy to improve the integration of HD patients in society.

Methodology

Type of Study

Qualitative Research (QR) is a valuable source of clinical information and evidence (B. E. Gibson & Martin, 2003; Korstjens & Moser, 2017; Moser & Korstjens, 2017). The QR methodology is informed by epistemology, which shapes the researcher's approach in terms of the role of the investigation, analytical procedures, measures, results and writing (Cristancho et al., 2018). For our study, constructivism was selected because it has as a principle that society and people exist mutually in a social world and allows an understanding of the meanings assigned (Kelly et al., 2018). There are several methodological approaches in QR, among which there is, Phenomenology (Moser & Korstjens, 2018). Phenomenology seeks to understand the subjective lived experience of the phenomenon in this case the HD disease diagnosis and Physiotherapy (Cristancho et al., 2018). In our study, our purpose is to understand the importance of Physiotherapy in HD patients' lives, social and personal challenges by interviewing and video record a family member (appendix 1) and HD patients (appendix 2).

To ensure the complete and clear reporting of the QR we use the Standards for Reporting Qualitative Research (SRQR) (Jette et al., 2019) The SRQR has 21 items that are essential to establish a consistent report of the data in QR (Jette et al., 2019) (appendix 3).

Sampling

Sampling is the process of selecting the participants and the context where we are going to collect the data of interest to the investigation (Moser & Korstjens, 2018). The main strategy used in Phenomenology is criterion sampling in which the participants meet predefined criteria (Moser & Korstjens, 2018). We looked for participants who have shared an experience (in this case HD patients who have Physiotherapy or have done Physiotherapy, and a family member of a person with HD doing or have done Physiotherapy but varied in their personal experience (Moser & Korstjens, 2018). The sample must be large enough to assure that all of the important perceptions are covered, but at the same time, it should not be too large because it becomes repetitive (Mason, 2010). Saturation is defined as the point at which the data collection process no longer offers any new or relevant data (Dworkin, 2012). So, for a Phenomenologic study, it is estimated that we should need 10 to 20 interviews, but at least 6 (Mason, 2010; Moser & Korstjens, 2018).

The study was conducted in Portugal and the participants were recruited and received an explanatory letter (appendix 4) through *Associação Portuguesa de Doentes de Huntington (APDH).* The contacts were through their mailing list and telephone contacts, as well as other contacts (such as patients and family members of colleagues or other institutions), therefore the sample was non-probabilistic for convenience. In order to characterise the participants, an initial socio-demographic questionnaire was used.

Eligibility Criteria

The inclusion criteria for patients were: (1) Portuguese adults, (2) diagnosed with HD,(3) they had to have the capability of answering simple questions (personal information to the socio-demographic questionnaire), (4) they had to be currently or had to have Physiotherapy, (5) be able to understand the goal of the study, (6) be able to give their informed consent (appendix 5). Because we want the double perspective we recruited family members of HD patients. The inclusion criteria for the family members were: (1) Portuguese adults, (2) family of a HD patient, who does or did Physiotherapy, (3) be able to understand the goal of the study and give their informed consent.

Data collection

Interviews are one of the most powerful tools to understand human beings and explore topics in-depth (McGrath et al., 2019). The participants can provide an insight into their lived experiences, and how they view and interpret the world around them (Carter et al., 2014; Cristancho et al., 2018). The interview allows spontaneity, flexibility and responsiveness to the individuals (Carter et al., 2014). In this work the interviews were semi-structured. These sort of interviews are designed with a list of open-ended questions that serve as a guide, but do not constrain the interview (Cristancho et al., 2018; Moser & Korstjens, 2018). The importance of accurate preparation of the interviewer should not be underestimated, so the interviewer has to be prepared to conduct a qualitative interview, and that is important to the success of the interviews (McGrath et al., 2019). For our work, a pilot test with a preliminary interview was done with 2 intentions: the first was to guide and confirm that all the relevant content is queried and if needed, reformulate some questions (Moser & Korstjens, 2018) and the second has to do with the "skills training" where the skills like, exploring language, clarity on questions or active listening were trained as an interviewer (McGrath et al., 2019).

The interviews were audio and video recorded, preferentially we tried face-to-face interviews. However, due to geographic constraints, they were also done via ZOOM, in a comfortable and pleasant place for the participants, the interviews were also carried out individually and separated, except when the caregiver's collaboration was requested due to difficulties in understanding the patient. They were done between December and March (appendix 6) (after Instituto Politécnico de Setúbal (IPS) Ethical approval). After that, they were transcribed verbatim, with all personal identifiers eliminated. Non-verbal actions, such as cough, pauses, tone of voice and others were described in detail (Moser & Korstjens, 2018). To ensure that the transcription was well done and to increase consistency, JC and MP verified the IA's transcription.

The video record allows observation-based research that can provide a powerful insight into the feelings and way of being of the individuals (Cristancho et al., 2018)). With that, we have a mix of data sources, such as interviews and

the video record. This will help us review the data and analyse it with more precision of the verbal and nonverbal interactions (Cristancho et al., 2018)

The materials used were, an interview guide, a camera, an audio record device, one computer, a phone and a data storage device.

Data Analysis Methods – Codification and Thematic Analysis

The audiotape interviews and field/observation notes are our major data sources (Moser & Korstjens, 2018). When analysing the interviews, we looked for the uncovered meaning, and we tried to develop an understanding and discovery of the insights that were relevant to the research question (Cristancho et al., 2018). An inductive content analysis involves the observation of the data and breaking it down into smaller units of analysis, coding and naming those units according to the content and grouping the coded material based on shared concepts (Moser & Korstjens, 2018). The coding is the identification of the segments that are responsive to the research question. Then we compared one segment with another one looking for a recurring pattern (Cristancho et al., 2018). After, we analysed to see if those categories identified continue to exist or not, or if there are new categories arising (appendix 7). Then, we finalised understanding of how the categories interrelate and, finally, interpreted their meaning and their interrelations (Cristancho et al., 2018).

Trustworthiness

Each QR approach has specific techniques for conducting, documenting, and evaluating the data analysis process, although it is the researcher's responsibility to assure trustworthiness (Nowell et al., 2017). Trustworthiness relies upon four general criteria approach: credibility, transferability, dependability and confirmability (Norman A. Stahl and James R. King, 2020).

Credibility is determined by how congruent the findings are with reality. Triangulation is one of the techniques that can be addressed to credibility (Norman A. Stahl and James R. King, 2020; Nowell et al., 2017). It refers to the use of multiple methods or data sources and it helps to explore and explain human behaviour, offering the readers a more detailed and balanced explanation (Noble & Heale, 2019). In this study, 3 types of triangulation were used: the

investigator triangulation (1), which combines the participation of 2 or more researchers in the same study, providing multiple observations and different conclusions, in this case with the participation of JC, IA and MP; the methodological triangulation (2), because it involves the use of multiple methods of data collection, is present in our study through the use of interviews and video records and the combination of qualitative and quantitative data, by using the Global Rating Scale (GRS) to evaluate the impact of Physiotherapy in HD patients and family members; lastly, (3) the data triangulation, since we collected information/opinions about the importance of Physiotherapy to the patients and families and thus earning more perspectives and validation of the data (Carter et al., 2014).

The transferability refers to the generalizability of surveys (Nowell et al., 2017). In this way and to achieve transferability, although this is not one of the objectives of QR, all information about interviews was detailed throughout this work and the interviews are also available in appendixes 1 and 2.

To ensure dependability, the research process must be logical, traceable and well-documented. To ensure this, all the research members participated in the analysis process after the transcription. The analyses were done by IA, but JC and MP gave their opinion and helped to achieve an agreement. Additionally, study recruitment and interviews were conducted by the same investigator, in this case, IA, who had undergone soft skills training in qualitative methods and in-depth interviews, as already explained in the data collection topic.

Finally, confirmability is concerned with the interpretations and findings, and if they are derived from data and not from the researcher's motivation. Confirmability is established when credibility, transferability and dependability are achieved (Nowell et al., 2017).

Ethical Considerations

Throughout the work, the health, dignity, integrity, privacy and confidentiality of the person's information were ensured (Korstjens & Moser, 2017). All participants gave their informed consent and were informed that they could withdraw at any time without any consequences. The researchers were always available throughout the process to answer any question and great care and effort

were made to make sure that all the participants understood the aim and implications of the study. An Ethical Approval for the study was carried out by IPS Ethical Committee (CE-IPS – PI nº 23A/2022).

At the beginning of the interviews, each participant received a code, so that they could be identified without providing his data. Through the interviews, data processing, thematic analysis or any other necessary reference to that participant were carried out through their code. The interviews that were conducted face-to-face were recorded using a tape recorder, excluding the participant's demographic data collection. For the interviews that were carried out via video call, an individual meeting was will be created in advance via Zoom platform, where only the interview content was recorded, excluding the recording of the participant's demographic data collection. The personal information, such as the name of each participant, was encoded in an Excel document and that document has a key-word which only the first and second researcher have. These same data were kept from the beginning of the study, until the date of delivery of the Master's Dissertation and publication of the research products.

Results

A total of 21 interviews was made, with a final number of 18 included, in which 12 participants are relatives and 6 are patients. 3 patients were excluded since they did not comply with the inclusion criteria, since 2 of them, despite having the ability to answer sociodemographic questions, did not have enough verbal ability to carry out semi-structured interviews. The other one noticed during the interview that it had not done Physiotherapy but instead it had Physiatry consultations. Table 1 represents the sociodemographic data.

Due to the emotional burden, and cognitive-behavioral alterations that come from this disease and as mentioned in Methodology, the video recorded of the interviews allowed us to follow up the interviews in a detailed way, allowing the visualiation of non-verbal information.

The results will be followed by illustrative excerpts of each theme. Due to the coding being carried out in Portuguese, the acronym “U” corresponds to “utente” (patient) and the “C” corresponds to “cuidador” (caregiver).

Table 1 – Sociodemographic data of relatives and patients;

*n.d (not discriminated)

	Family Members n=12	Patients n=6
Age		
18 – 65y	9	5
> 65y	3	1
Gender		
F	11	3
M	1	3
Family Relations		
Mother	2	n.d
Daughter	6	n.d
Uncle	1	n.d
Sister-in-Law	1	n.d
Wife	2	n.d
Years of care		
<10y	5	n.d
10-20y	7	n.d
Years of disease		
1-5y	n.d	2
6-10y	n.d	3
>10y	n.d	1

There were 5 core Themes (Table 2). One is related to Objectives and ICF (Theme 1) which takes into account the patients and their problems and changes in everyday life. The second theme is related to Society and HD (Theme 2) and refers to changes in society's behaviour towards HD. A third one is related to Health Care and Needs in HD (Theme 3) and is about the healthcare needs of these patients related to the disease progression and their symptoms. The fourth is about Physiotherapy and HD (Theme 4), and refers what has been done related to Physiotherapy and its results. The last Theme is Family and HD and shows us the perspective of living with a neurodegenerative familiar disease (Theme 5).

Table 2 – Themes and Sub-Themes

Core Themes	Sub-Themes
Theme 1 - Objectives and ICF	Life prospects and goals after diagnosis Structures and functions – symptoms and problems derived from the disease Activities – day-to-day limitations; implications of symptoms/disease in daily life
Theme 2 – Society and HD	Society's behaviour towards HD Society knowledge of HD Role of APDH Importance of normalising the disease
Theme 3 – Health Care and Needs in HD	Rehabilitation in HD Implications and limitations on access to therapies Multidisciplinary team and relation between them Lack of knowledge on the part of health professionals
Theme 4 – Physiotherapy in HD	Physiotherapy Sessions Physiotherapy Results Relation between Physiotherapists and patients and families What if HD patients did not have Physiotherapy?
Theme 5 - Family and HD	Impact of HD on families and family members Family members burden Concerns about heredity

Theme 1 - Objectives and ICF

HD patients and their families have their own vision and unique experience about the disease, their symptoms, daily impact and hope for the future. Due to the hereditary course of the disease and although they already know a lot about the disease and how to manage it, there is an important and common landmark that they remember in almost all interviews, the diagnosis.

- **Life prospect and goals after diagnosis**

HD is a complex disease which changes patients' life goals. It is after the diagnosis that patients and their families can explain, understand, and manage the disease.

U_02: *"My goals were to keep my health up to date…"*

U_06: *"(thoughtful) It changed everything because I was not expecting this…"*

C_03: *"From the moment the diagnosis was made, at least my family knew what could happen (pause) from then on, in terms of care and in terms of consequences of the disease itself."*

C_07: *"Of course, everything changed, but the truth is that there was now a justification for what we were already experiencing, (…) suddenly we can give them a name and we can understand what's happening."*

- **Structures and functions – symptoms and problems derived from the disease**

This sub-theme is related to symptoms, and body structures changes that are identified by patients and family members. The most identified ones are speech problems, motor disturbances, balance problems and cognitive and psychological impairments.

U_02: *"it's too much movement… I keep falling…"*

U_07: *"(…) lack of balance (…) anxiety (…) problems from time to time with memory, small lapses and then attention span (…)"*

U_09*: "(…) and I'm afraid of falling, afraid of walking at home alone;"*

***C_01**: "(…) speech problems ahhh locomotion problems, these are the most (emphasizes) obvious; (…) have some breathing problems and problems of attachments (…);"*

***C_03**: "(…) more difficulty moving, walking, holding cutlery was at the same time that he also began to have more difficulty speaking (…)"*

***C_11**: "(…) my mother has serious balance problems, she has swallowing problems, ahh language and have some cognitive problems";*

- **Activities – day-to-day limitations; implications of symptoms/disease in daily life**

Extremely linked with the sub-theme presented before, here, their view about the impact of these body alterations in patients' and families' activities is expressed. Almost all of them are related to hygiene care, motor skills like walking or manual dexterity, feeding difficulties and work maintenance.

***U_02:** "I stopped working, I stopped having a life as it should be, my life was… has been a disappointment…" (…) I've been having a bad time… now it's even worse before I still hung out clothes, I still put the washing machine, I still did my things, but now I don't do anything anymore (…)"*

***U_04**: "(…) I can't do nothing!" (it refers to limitations at home)*

***U_06**: "(…) I didn't shake, I walked, I exercised, I rode a bike, I went to the gym, I did all of those things… I worked… (…)" (referring to activities that stopped doing because HD)*

***C_02**: " (…) she can't hold the trunk anymore (…) her problem is the dipper changing… she starts to be aggressive, giving, giving her the food she is also aggressive… hygiene, that's it no, no, she doesn´t like."*

***C_04**: "(…) they lose their own autonomy (…) even to do their hygiene when they go to the bathroom (…) even in the meal, she still eat but I have to help her eat because the food ends up being spread out on the table because she insists on eating alone (…)"*

***C_07**: " (…) at this moment my mother is already in a wheelchair, she can get up, and take a few steps, a little thing. (She) helps to go to the bathroom, helps to get dressed, helps to put the diaper at bedtime, so she can help, but now, I have to be there always (…)"*

Theme 2 - Society and HD

Society is commonly seen as a group of individuals who share their cultural and ethical values. Behaviour changes towards HD patients, society's lack of knowledge about this disease and the importance of normalising the disease are the sub-themes of this topic.

- **Society's behaviour towards HD**

When someone does not fit in or seems different from what we idealise, it generates behaviours that can vary between fear and indifference or after one gets used to the difference it can also build respect.

***U_02**: "(…) people treat me there with respect!" (referring particularly to where it lives)*

***U_02**: "Society isn't prepared yet, these things haven't even taken into account yet. There are a lot of people who laugh and think it's just that I had a stroke…So people sometimes laughed(…)"*

***U_06**: " (…) for me people are good (…)" (referring particularly to where it lives)*

***C_01**: "(…) People tend to ahhhh shy away from people with these kinds of illnesses like it's something hummm ahhhh contagious (smiles) like it's some kind of contagious disease; other people who think it's some junkie or some drunk or something…"*

***C_03**: "I think that a large part of society does not accept the unknown very well and when it does not accept it, it also tends to not want to know more (…)"*

***C_05**: " (…) and more and more there is a very great indifference"*

- **Society knowledge of HD**

As a consequence of being a rare disease, which affects families in particular way, HD is little known and little talked about. HD patients and families are aware of this gap and it has an impact on their lives.

U_04: "must not know the disease…"

U_05: "Society doesn't know what Huntington's disease is… (pause) we have to… we have to, when we say we have the disease we have to explain everything and anything else;"

U_07: "(…) I think Huntington's disease is still… it's still very invisible…"

***C_05**: "(…) ahh, society has no knowledge, (…) once I was going with an aunt with non-compliant movements in the street and there was a lady who passed by and said, ""first thing in the morning and already with such a drink?""*

***C_09**: "(…) the issue of having a disease that few people know about, that few people know how to deal with either in social terms, in terms of family, or terms of health, and the implications that this has…"*

***C_10**: "(…) the Society doesn't know… most people don't know about this disease… they get like that… people ask me… (…)"*

- **Role of APDH**

APDH is the HD Patient Association in Portugal. Patients and caregivers identify its importance in terms of information, however, difficulties in reaching everyone are mentioned as a limitation.

***C_04:** "In the information, in the sharing, in the meetings that are held and each one shares, what goes on or has gone through and the sharing of knowledge (…) so this sharing ends up helping each other, even in terms of knowledge about what Huntington's disease is, I was only able to find out what Huntington's disease is through association."*

***C_10:** " (…) even I signed up for the… the… the Huntington in what… in the Portuguese and There's nothing either, I don't see anything new."*

- **Importance of normalising the disease**

The families, even with the disease and its symptoms, try to explain and normalize the disease, after all, for them, despite HD, the patients continue to be their loved ones.

***C_03**: "I think that talking about this type of situation or in this case, a rare disease ends up normalizing;"*

***C_04** " (…) When I have the opportunity, I say that she has a degenerative disease, which is Huntington's disease"*

***C_11**: " I think that everyone can play a role in this disease, let alone explain to people what Huntington's disease is (…) I think we all have a role, ahh and it's up to us, even all the physiotherapists and everyone who deal with this (…) we have to normalize the disease (…) and explain to people that it's a result of a disease that in my mother's case is called Huntington's chorea in another case it could be something else."*

Theme 3 - Health care and needs in HD

Given the progression of the disease, which impacts the activities and daily functionality of patients and families, health needs tend to grow.

- **Rehabilitation in HD**

HD combines motor disturbances, cognitive and behavioural symptoms. These symptoms display a gradual worsening in mobility and participation of these patients, which directly affects their ADL, so there is a lot of therapies and medical follow-up that patients need.

***U_05**: "I also have Psychology (pause) and… neurology in the Hospital"*

***U_06**: "(…) one of psychiatry and one of neurology, and that's why I have support from those parts, and also the medications (…)"*

***U_07**: "Ahhh, I have about 2 neurology appointments per year and at least 2 psychiatry appointments a year (…)"*

***C_01**: " We have to provide them as many as therapies we can, from Speech Therapy to help them speak, Physiotherapy (…) Occupational Therapy, all kinds of therapies that, Psychologist (…)*

***C_06**: "(…) 6 months in 6 months, which in my opinion is very little and there is physiotherapy twice a week in a private clinic;"*

***C_07**: "My mother ahhh fortunately is monitored in several areas, so ahhh as soon as there was the diagnosis of Huntington's disease it was neurology right? Ahhh…it was speech therapy, it was gastro because of the swallowing part; (…) psychology (…) Physiotherapy (…) occupational therapy (…)"*

- **Implications and limitations on access to therapies**

Concerning care needs, there are a few problems identified by HD relatives and patients regarding access to therapies and healthcare. There is a lack of response and vacancies at the level of national health service.

***U_02**: "(…) I'm afraid to go, I'm afraid to go to Physiotherapy alone because I fall… and break something, and then I'm afraid…"*

***C_01**: " All private, all paid for me."*

***C_04**: " I think it was important that all patients had access, whether they were able or not, because maybe the majority don't have that possibility, of ahhh paying for these treatments... (…) without having to move because moving also involves costs, even for the National Health Service and it is, ahhh having ahhh vacancy to have these resources, because the list is immense…"*

***C_06**: "yes, physiotherapy is not daily because of the costs";*

***C_07**: " (…) what I was told was that ahhh they have few vacancies and so they give priority to those who have chances of improvement, that is, someone who is to maintain or to try to delay a disease it's not a priority"*

***C_12**: (…) everything else was paid for outside, because if I was waiting for someone to get me, a physiotherapist, to come here or, no way, I mean, I would get a year or so later. I bought the wheelchair, and all the equipment, I didn't even wait because there was no possibility of having what you're waiting for right? That, for those who don't have it in terms of economic possibilities then becomes much more complicated."*

- **<u>Multidisciplinary team and relation between them</u>**

Such a complex disease leads to the need for close professional relations between different professionals. When this happens, there is a feeling of relief and support, but when this does not happen, they feel the need to emphasize *this problem.*

***U_06**: "I don't think there is much contact…"*

***U_07**: " (…) then she wrote me a letter from ahhh explaining my situation, for me to deliver there to a physiotherapy clinic… that's it… that's what I did;"*

***C_02**: " they are separated because they are… ahhh they are from different places (…) there is not much connection; (…) of course they ask what the other said, yes there was the concern but there was no meeting or discussion of ideas (…);*

***C_07**: " No, there is not, because they are not from the same hospital. So no, none of these therapists have contact with each other."*

***C_11**: " (…) it seems to me that they do not communicate, the only thing that seems to me is that they receive a prescription, and they try to comply and make sure that what the doctor prescribed (…)"*

***C_12**: " She (physiotherapist) made a report that she sent to the doctor, she even sent emails to the doctor and the doctor for her, therefore for the evolution of the XXX, what did she notice, it was something I really liked."*

- **Lack of knowledge on the part of health professionals**

The lack of knowledge and sensitivity towards HD disease from the health care professionals was also reported and noticed, particularly by family members.

***C_04**: (…) "if we, if we say that, even in a hospital environment, most doctors and nurses did not know (emphasizes) how to deal with Huntington's disease, let alone the common population."*

***C_05**: " Do you even know that there are doctors who don't know what Huntington's disease is?"*

***C_10**: (…) "even people linked to health; there are many people connected to health who don´t know."*

Theme 4 – Physiotherapy in HD

The fourth theme is about Physiotherapy and what has been made to HD patients. In this theme, knowledge about Physiotherapy sessions is given. There were considered aspects such as the results, the impact of Physiotherapy and the relation between patients and families and their physiotherapists.

- **Physiotherapy sessions**

We tried to understand better what consists a Physiotherapy session with these patients; Most of them gave importance to functional exercises and gait training;

***U_02**: "I used to do 15 minutes on treadmill, then do a few things there, I used to move my hands and do massage… foot massage;"*

***U_04**: " (…) I move my legs; (…) I move my arms; (…) I do difficult things; (…) having to walk (…) doing things related to the arms (…)"*

***U_06**: "(…) I do different exercises that help me to coordinate and improve and maintain what I have, what I'm doing, walking, dressing, washing (…) I do bike sitting down, arm exercises to pull, and sometimes I put on shin guards to walk,*

then the machine sitting on to my legs strength, and then walk in line with one foot in front of the other;"

***U_09:** " Walk back and forth";*

- **<u>Physiotherapy results</u>**

Patients and family members confer importance to Physiotherapy and perceive its results. Besides functional results, there are also mentioned the results in the psychological domain. As referred before a methodological triangulation was made; a GRS was used, and the results are presented in Table 3.

***U_02**: "It says it helps, it improves walking, but I left there tired (…) and it didn't improve at all… nothing… (referring to day-to-day activities)";*

***U_04**: "(…) gives me more strength (…) to go to the bathroom" (…) I think so… it relieves… helping me helps them… (referring to the impact of Physiotherapy on family members)"*

***U_05**: "I feel better and more… with less involuntary movements and more forcefully";*

***U_06**: "Because it helps me not to decline so much and to keep the good thing I have";*

***U_07**: " (…) I think it was very good for the muscles, I felt very good, I haven't felt like this for a long time on muscular level, it was really good; then I also gained some self-confidence, I gained a bit more… a little more self-confidence, more confidence walking (…) exercises walking and going up and down stairs and in the street (…) we go up and down and up and down the stairs, and that did me a lot of good, I gained confidence for that (…)";*

***U_09**: "No, nothing happened, it didn't do anything for me, no, it's not well adapted to my disease, it didn't do anything for me…";*

***C_01**: "So give me a part of quality of life…"*

***C_03**: " (…) it was noted that she was happy and well when the Physiotherapy took place"*

***C_06**: "It has effect… but the effect is short-lasting…"*

***C_07**: " (…) with much higher self-esteem, because now… she is back to being able to do some things that she wasn't (…) she is more satisfied, she is happier, obviously she is not so distressed and the behavioural part is calmer…"*

***C_09**: " So, ahhh increased strength, stability, balance, learned how to fall, protect oneself from falls (…) above all the routine issue has also improved not only the physical part but also the ahhh behavioural part the day-to-day part the routines."*

Table 3 – GRS results (Global Perception of Change score between 1-7)

	Relatives (n=12)	Patients (n=6)
Average	5.42	4.33
Median	5	5
Max Score	7	6
Minimum Score	3	2

- **<u>Relation between Physiotherapists and patients and families</u>**

Due to the complexity of HD, occasionally, some questions and problems can be discussed namely how Physiotherapist can help. Furthermore, it is important to design an intervention centred on the HD patient and his/her objectives, for this, a relation between the various actors in the rehabilitation process is important;

***U_07**: "(…) The physiotherapist had no idea what Huntington's disease was and I explained it to her (…) it helped, it helped a lot because then, due to these symptoms and these things, she did the exercises, she also started to fine-tune the exercises that were most suitable for me (…)"*

***U_09**: " no, no, the physiotherapist never spoke to me…"*

***C_03**: "(…) without a doubt we felt integrated, some exercises were even explained to us that we could ahhh go on doing throughout the day…"*

***C_06**: "Not by chance, not by chance… there's not much talk about that";*

***C_07**: "I never attended my mother's sessions. Ahhh I don't even have contact with the physiotherapist"*

- **What if HD patients did not have Physiotherapy?**

A reflection on the need for Physiotherapy and on the possibility of its absence shows how the participants perceived its gains.

***C_02**: " (…) it delayed her stopping a little, the motor part having stopped so late right?"*

***C_03**: "I think she died much earlier (…)"*

***C_06**: "She would probably be bedridden"*

***C_07:** "oh, much worse, much worse without a doubt. And I'm sorry she didn't start sooner"*

***C_10**: "It was much worse, it was much worse in all aspects"*

Theme 5 – Family and HD

The last theme identified in interviews was the impact of HD on families and the relation with the burden of this disease.

- **Impact of HD on families and family members**

In HD, most of the time, family members become the pillars and active members in the care of these patients, so all their lives and objectives change as well.

***C_01**: "(…) because the objective now is solely and exclusively to take care of my daughter"*

***C_02**: " (…) and so, basically I lost my youth, my freedom, I didn't dedicate myself to my studies as I might have wanted"*

***U_05**: "It had a lot of impact, ahhh ok, I even ended up getting divorced and everything…"*

- **Family members burden**

There is a need for patients to be monitored by family members who end up being burdened with tasks and situations linked to the progression of the disease.

***C_02**: "(…) I had to start going with her, I came running from work and had to stay there at the door waiting, (…) it wears me out and then I waste my whole life out there and, and I leave everything behind;"*

***C_05**: "(…) I'm giving her much more support, but it gets to the point where I'm also saying "pah, I have to take a little bit for myself, don't I?" despite the fact that I do a lot of things and I'm involved in a lot of things, but also if I not in certain things that I am… ahhh I go crazy…"*

***C_10**: "(…) for a person alone it is very complicated, very complicated, and I can't do it anymore (…) How complicated it was for him to go to the institution, I also suffered a lot for him, when he went to the institution because I thought I had abandoned him;"*

- **Concern about hereditary**

HD is a hereditary autosomal dominant, progressive and neurodegenerative condition, which means that children of a person with HD have a 50% chance of having the disease. After the diagnosis, the burden and concern with the possibility of transmission of the disease is something that has a representation in the lives of these families.

***C_03**: "(…) brought an added load, difficult in the sense that it is possible me and in this case, my brother has this disease, ahhh and being there to see what could happen to us;"*

***C_04**: " (…) I just thought, "I don't believe that this could be happening again in the second daughter", with the second daughter it wasn't easy… (…) because seeing a daughter already dying with this disease (…) then starting to relive everything again with the second daughter… no, it is not easy, it is not, it is not easy;"*

***C_05**: "Because I saw ahhh what was happening to my sisters-in-law and I didn't want that happen to my wife (..) it's not for nothing but because, because they suffer isn't it?"*

Discussion

This work aimed to understand, from the point of view of patients and their families, how Physiotherapy can help in the life, challenges and problem-solving of HD patients and their relatives, according to their perception. Additionally, we tried to understand what we can change and progress in Physiotherapy to improve the integration of HD patients in society.

Our work is closely related to what ICF conceptualises since it advocates that a person's level of functioning is a dynamic interaction between health conditions, environmental and personal factors (Center for Health Statistics, n.d.).

With that in mind and taking into account our primary goal, it is crucial to notice that effectively symptoms (health conditions), society and rehabilitation care (environmental factors), and patients themselves and their families (personal factors) have an important interaction and are all related.

The first theme of our work is related to HD itself and the impact that it has on patients' and relatives lives. Regarding symptoms, previous studies suggest that, their management and the progression of the disease impact the ADL and functionality of the patients. As we know, it is quite common to have psychiatric or cognitive symptoms and subtle motor signs many years before marked motor signs in HD (Ghosh & Tabrizi, 2018), and a recent systematic review found that the increase of neuropsychiatric symptoms is related to the decrease of functional capacity in HD (Sellers et al., 2020). These results are in line with ours since motor and balance impairments are reported by patients and relatives as important symptoms, yet they also report anxiety, lack of memory, and depression as a major problem that also impacts day-to-day life. Additionally, due to the hereditary path and symptomatic progression of the disease, there is an important and common landmark that they remember in almost all interviews - the moment of diagnosis.

Related to diagnosis and how HD patients and families manage the information, a qualitative Portuguese study shows that getting the diagnosis is a turning point for patients and their families (Oliveira et al., 2020). The same study demonstrates that there is a feeling of relief in knowing that they have HD, however, at the same time, they have to deal with the heredity, disease progression and lack of effective treatment and knowledge (Oliveira et al., 2020). These results are in line with those obtained in our study, where having a name for the disease helps to manage and understand it, however, since there is no cure the disease brings other concerns such as heredity, the progression of the disease itself and a concern about life goals.

The first theme, especially diagnosis, disease progression and daily life limitations, are also related to our second theme - Society. In a recent review which identifies the social withdrawal of HD patients, it is reported that social withdrawal is common and evident across disease stages and associated with

symptomatic and physiological markers of disease progression. Additionally, it was also found that this withdrawal causes a burden on HD relatives (J. S. Gibson & Springer, 2022). In our results, two perspectives emerge. The first is that, effectively, there is a report of social withdrawal due to the fear of falling, walking alone in the street, and the incapability of working or going to the gym and other social activities, since disease progression has an impact on motor function, however, secondly, our results also show that, despite the decrease in social activity, in their house area, patients feel included due to the long coexistence and follow-up by the neighbourhood of the disease progression, therefore, those who most refer to this social exclusion and impairments with social behaviours are the family members.

Related to Society, J.S. Gibson & Springer also identified that social limitations are often related to the progression of the disease (J. S. Gibson & Springer, 2022). In our case, patients and family members do not specifically relate these social impairments with the progression itself, but with a lack of knowledge that leads to a change in society's behaviours including fear, indifference and rude behaviour.

Associated with the lack of knowledge and visibility of the disease in Society, our interviews led to the emergence of themes such as APDH and its role and support. In Portugal, generally, an Association has the important role of defining common goals and aims to overcome difficulties and generate benefits for its associates. A previous study shows that patient organizations have become important influencers in matters related to health, illness and health care policy, overall the work of the organizations can be seen as the “official” work to improve aspects and knowledge related to the disease (M. Jones et al., 2021). Another study shows that patient organizations should also provide information tools and the active involvement of patients and patient organizations is also a valuable process to ensure helplines in terms of rehabilitation access (Adachi et al., 2023). In Portugal, as described in the previous studies, APDH has the main objective to provide support and guidance to families with HD, in addition to make the disease known and make the people involved closer together to exchange experiences and mutual help. This support provided by APDH seems to be essential for some patients and their families due to information access and

support, however, as many associations in our country due to proximity, financial and social constraints APDH also has difficulties in reaching everyone and some patients feel the need to mention that they need more information as well as social visibility.

The theme “Health Care and Needs in HD” is also closely related to the others, since it presents the health needs that arise due to the symptomatology and limitations caused by the disease. HD has no cure, therefore, treatment (pharmacologic and non-pharmacologic) only alleviates symptoms (Erkkinen et al., 2018). Regarding nonpharmacologic strategies and according to this review, interventions such as Physiotherapy, Occupational Therapy, Speech Therapy, Psychology, structured daily schedules, and Social Support are extremely important for patients and their families and should be particularly considered as the disease progresses (Erkkinen et al., 2018). Our results confirm this information since the needs presented are mentioned by participants, especially Psychology and Psychiatry for cognitive/behaviour impairments, Speech Therapy for speech and ability to swallow disorders and Physiotherapy for motor and balance disorders. Moreover, it is important to give special focus to the follow-up and monitoring by the hospital staff specifically by Neurologist, however, for some of them appointments every 6 months seem to be too spaced outgiven the progression of the disease.

Despite these needs being identified in literature there are several problems and implications in accessing these therapies. A previous study about rare diseases shows that there is a large economic impact of rare diseases due to the direct costs of treatment, drugs, care costs and an overall cost of lost productivity for the patient and their caregivers. Evidence also suggests that often rare disease patients struggle to access treatment due to disparities between rural and urban settings. Moreover, patients with rare diseases often struggle to find healthcare professionals knowledgeable about their conditions leading to subsequential adequate treatment and care (Adachi et al., 2023). These problems also emerged in our interviews, with HD patients and family members identifying problems in accessing rehabilitation related to treatment costs, and difficulties associated with travelling to treatments, lack of vacancies and costs of experienced professionals and equipment.

Other aspects related to health needs and closely related also to the Theme Physiotherapy, are the knowledge sharing, communication and interaction between family members and health professionals. A previous study shows that there is a lack of knowledge and experience among health professionals, and this may present a challenge since poor communication and lack of interaction are problems for these families (Røthing et al., 2015b). Their results also show that this paucity of communication represents an additional burden for families since it may lead to inappropriate care, as it does not take into account the goals and personal particularities of patients and their families (Røthing et al., 2015b). Moreover, another study highlighted that HD is a complex journey which can affect different generations, so it is important to continue efforts to improve pre and post-diagnosis support for HD disease families. Therefore they also refer to the need for education of health professionals, media and society (Oliveira et al., 2020). These results highlight what some participants in our study report, by mentioning that the fact that health professionals lack specific knowledge about HD leads them to not integrate families into clinical decisions and, therefore, the family members are not even aware of what is done in a Physiotherapy session. On the other hand, some participants report that they feel integrated as their family is in Physiotherapy sessions, and there is even an explanation and prescription of activities in order to help all families and integrate ADL.

Despite the need to improve communication and specific knowledge of HD among health professionals, our results on the theme of Physiotherapy demonstrate that the most recent recommendations concerning Physiotherapy in HD are considered most of the time. A previous study suggests that exercise and physical activity may be beneficial for HD patients in terms of motor function, gait speed and balance (Fritz et al., 2017). In general, their results also show that aerobic exercises and strengthening exercises are safe and feasible in this population and besides motor gains, they also show improvements in QoL (Fritz et al., 2017). Additionally, the latest Guidelines show that Physiotherapy interventions such as balance exercises, aerobic exercises, gait training, physical activity promotion, task-specific training, education, strength exercise and respiratory therapy become a fundamental part of managing HD (Al-Wardat et

al., 2022; Quinn et al., 2020). In our results, patients report that their sessions include strategies such as aerobic exercises such as treadmills and cycloergometer, walking training, and strengthening and functionality exercises. Also, patients and their relatives report some benefits of Physiotherapy, mainly about the impact of Physiotherapy in helping their life at home, in confidence and in the feeling of well-being, because they relate their strength and motor capacities with being useful and less dependent.

Despite that, the results of Physiotherapy and its benefits are not unanimous as we can see in GRS, with some patients and family members saying that Physiotherapy has no results. A recent study aimed to understand the facilitators and barriers to implementing the Guidelines for Physiotherapy treatment in HD (U. Jones et al., 2022). The support from colleagues, the individualised physiotherapy plan and the physiotherapist's expertise in HD were reported as the key facilitators to implementing the Guidelines. The barriers identified were HD-specific characteristics, cognitive and behavioural dysfunction and low motivation (U. Jones et al., 2022). These results are also in line with ours since during our interviews numerous factors were reported by patients and family members as an obstacle to their results such as inadequate Physiotherapy context and lack of physiotherapist expertise, the non-definition of objectives individualised taking into account their needs, the lack on prescribing individualized exercises, considering each person and their specific impairments and lack in the humanization of care;

The last theme of our work is concerned with family and its burden with HD. As with all aspects of our work, this theme is also related to the other themes, namely with diagnosis, hereditary concerns and disease progression. An ethnographic study done in Portugal shows that family members become caregivers of their relatives most of the time and lose their free time and physical and emotional health due to the demanding task that is taking care of patients (Teixeira et al., 2020). In our interviews, the participants highlight the change in their life and family goals related to the disease progression and decrease of autonomy, which increase the family burden and the loss of family members' "freedom".

Another Portuguese study shows that knowing the diagnosis of HD is a complex event in families, because it is a neurodegenerative familiar disease that has no cure, so it brings the problem of dealing with heredity (Oliveira et al., 2020). As we got from our study, heredity is a problem, mostly for family members, since they negatively anticipate the problems that their relatives may experience due to what they already have seen previously in other family members.

In this study, they also analyse two coping strategies used by families, closedness and openness. Closedness seems to be adopted to face shame and stigmatisation concerning society, the openness seems to be a recent mode in which families are involved in accepting and normalising the condition (Oliveira et al., 2020). Our results are in line with this study, as in different themes these aspects are addressed. At some point, we have reports of behaviour changes and societal impairments such as social withdrawal – closedness, but on the other hand and especially in family members' interviews, there is a need to normalise the disease – openness.

As far as we know, this study is the first that seeks to understand how Physiotherapy can help in the life, challenges and problem-solving of HD patients and their relatives, according to their perception. In addition due to the work that has been developed, it is expected that more visibility on the needs and problems of these patients will be taken into account.

However, this work has some limitations. Although we reached the minimum number considered by the literature, we can consider the reduced size of the sample our main limitation, specially concerning patients. We had a good response to the call for participation made through the APDH, but many of the patients did not or had never attended to Physiotherapy and therefore did not meet the inclusion criteria. Turns out, that this limitation can be interpreted as a result itself since it gives an indirect conclusion that access to Physiotherapy by HD patients is still limited.

Another limitation that our work had was the fact that, during the interviews and analyses, the stage of the disease was not taken into account, as we know,

the progression of the disease is characterized by different stages that can change the patients' and families' perceptions.

Conclusions

It is becoming increasingly clear that rehabilitation, specially Physiotherapy may represent a powerful tool in the lifelong management of HD. Physiotherapy, in patients' and family members' perspectives, seems to play a role in ADL, autonomy, self-confidence, family management and strength and walking capacity.

With regard to what we can change in Physiotherapy to improve the integration in Society, there is a need to improve the human-centred approach, so that Physiotherapy can meet patients' and family members' specific objectives in order to reduce their limitations in social participation.

Being a rare disease and although there are already some studies, some of them even carried out in Portugal, there is still a need to carry out more studies and talk more about the disease. So as the next studies, we suggest studies that evaluate the quality and the content of the Physiotherapy sessions. Also, future studies should try to understand the context in which Physiotherapy is carried out, whether this context makes a difference and which factors play a role.

As implications for clinical practice, this work highlights the important effort that has been done by APDH in terms of support and making the disease known. However, it is important to notice that there is still a scarcity of knowledge about the disease in terms of Society and health professionals. This lack of knowledge leads to a problem; as there are health professionals who do not know HD, patients and their families do not know the health care responses that exist, which leads to access difficulties specially with regard to Physiotherapy. It is therefore important to make known, raise awareness and normalise this disease that affects patients and their families and impacts their lives.

Dissemination

The plan for dissemination is structured in 3 phases.

First, and since that we made the interviews with the collaboration of APDH, and because this is the place where the different stakeholders act together, the results will be disseminated at internal events of the Association.

Secondly, and because the study is about Portuguese patients and it tried to complement the clinical reality of Portuguese HD patients and Physiotherapy, we already disseminated preliminary results of the final work in the Portuguese congress - SPDMov – Sociedade Portuguesa de Doenças do Movimento in March 2023 with a Poster (appendix 8).

Finally, while we were developing the final work of the thesis, we are currently in the process of submitting this work to *Physiotherapy* (impact factor 10). If the answer is negative, we propose to submit it to the following journals, the *European Journal of Neurology* (impact factor 6,2) or the *European Journal of Human Genetics* (impact factor 5,31).

Additionally, and as a result of being in SPDMov congress, I started working as a member of EHDN (appendix 9). EHDN believes that facilitating research to advance knowledge is essential as we strive to develop effective therapies for HD, so with my work and results and trying to give more visibility to HD, their patients and the importance of Physiotherapy in this disease, I hope to share, even informally, my results in International Congresses, such as EHDN - European Huntington's Disease Network or the MDS – Movement Disorder Society Congress.

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Appendix 1 – Familiar Member Interview

Introdução (já a gravar): Boa tarde,

Desde já agradeço a sua participação nesta entrevista, cujo o objetivo é perceber o papel da na vida dos Doentes de Huntington. Por favor, responda às questões da forma mais sincera possível. Relembro que entrevista vai ser gravada em forma de áudio e vídeo. Vamos começar então:

Saúde e Huntington – Principais problemas

- O que mudou na sua vida desde do diagnóstico da DH?
- Já tinha tido contacto com DH anteriormente (Frich et al., 2014)?
- Na sua perspetiva quais são os principais problemas do seu cuidando?
- Atualmente, em relação à utilização de serviços de saúde quais são as necessidades do seu cuidando (Etchegary, 2011)?
- Daquilo que me identificou o que acha que ainda está inacessível (Etchegary, 2011)? E porquê?
- Como têm vivido com a doença nestes últimos anos?
- Em relação à Sociedade, como acha que a mesma vê e se sente em com a doença de Huntington (Etchegary, 2011)?

Fisioterapia e Huntington

- O seu cuidando realiza Fisioterapia há quanto tempo?
- O que é que a Fisioterapia adiciona no seu dia-a-dia? E no do seu cuidando?
- Qual o impacto da Fisioterapia na sua vida (adap. Frich et al., 2014)?
- Acha que a Fisioterapia ajuda no seu contexto familiar e social com o seu cuidando?
- Pode-nos descrever um pouco da sessão (adap. Busse et al., 2008)?
- Sente-se integrado durante a sessão de Fisioterapia? O Fisioterapeuta fala consigo e mantêm-no como membro ativo do processo de reabilitação?
- Acha que a Fisioterapia vai de encontro aos objetivos do seu DH (adap. Frich et al., 2014)?

- Sente que a Fisioterapia ajuda a solucionar os problemas do seu cuidando?
- Referiu-me que os problemas são X, Y, Z… sente que através do (treino de equilíbrio, treino de força, treino de AVD's, ou outras), a Fisioterapia está a ajudar o seu cuidando?
- E problemas como as alterações cognitivas, comportamentais, motoras, de equilíbrio, acha que Fisioterapia poderia ajudar a resolver?
- Sente que a Fisioterapia é devidamente adaptada para uma pessoa com Huntington?
- O Fisioterapeuta ajudou-o a perceber a doença e a geri-la?
- Olhando para trás, como acha que estaria se o seu cuidando não realizasse Fisioterapia?
- O seu cuidando realiza outras terapias?
- Sente que médicos, fisioterapeutas e outros intervenientes na reabilitação comunicam entre si em prol do utente?
- Recomenda a Fisioterapia para pessoas com Huntington?
- Tem alguma sugestão para o(s) Fisioterapeuta(s) que trabalham com Doentes de Huntington (Etchegary, 2011)?

Sociedade e Huntington – Necessidades

- Sente que a Fisioterapia ajuda a resolver alguns dos problemas de XYZ do seu cuidando?
- Alguém tratou de forma diferente o seu cuidando por ter Huntington (Etchegary, 2011)? Se sim, o que aconteceu?
- Acha que a Fisioterapia o podia ter ajudado nessa situação (ou em que situações especificas no seu dia-a-dia)?
- Acha que a Fisioterapia poderia ajudar na integração social destes doentes?
- Por fim, existe mais alguma questão, preocupação adicional em relação à sua Saúde, e em especial à Fisioterapia que queira referir?

Obrigada pela sua participação e disponibilidade!

Appendix 2 – HD Patient Interview

Introdução (já a gravar): Boa tarde,

Desde já agradeço a sua participação nesta entrevista, cujo o objetivo é perceber o papel da Fisioterapia na sua vida. Por favor, responda às questões da forma mais sincera possível. Relembro que a entrevista vai ser gravada em forma de áudio e vídeo.

Principais problemas

- Quando foi diagnosticado com Huntington em que é que isso mudou a sua vida?
- Atualmente quais são os seus principais problemas?
- Como têm vivido com a doença estes anos?
- Atualmente, em relação à utilização de serviços de saúde quais são as suas necessidades (Etchegary, 2011)?
- Daquilo que me identificou o que ainda está inacessível para si (Etchegary, 2011)?
- Em relação à Sociedade, como acha que a mesma vê e se sente em com a doença de Huntington (Etchegary, 2011)?

Fisioterapia e Huntington

- Há quanto tempo realiza Fisioterapia?
- O que é que a Fisioterapia adiciona no seu dia-a-dia?
- Pode-nos descrever um pouco a sua sessão (adap. Busse et al., 2008)?
- Acha que a Fisioterapia o ajuda no seu contexto familiar e social?
- Como é a sua experiência na mesma (adap. Frich et al., 2014)?
- Acha que a Fisioterapia vai de encontro aos seus objetivos (adap. Frich et al., 2014)?
- Acha que a sua Fisioterapia é devidamente adaptada para a sua condição?
- O Fisioterapeuta ajudou-o a perceber a doença e a geri-la?
- Em que é que a Fisioterapia o ajudou?

- Referiu-me que tem dificuldades em X, Y, Z, sente que através da Fisioterapia os conseguiu resolver?
- Além da Fisioterapia realiza outras terapias? Se sim, quais?
- O seu fisioterapeuta contactou com o seu médico?
- Se não fizesse Fisioterapia como se imaginaria atualmente?
- Acha que a Fisioterapia ajuda a sua família e aqueles que o rodeiam?
- Recomenda a Fisioterapia para pessoas com Huntington?
- Tem alguma sugestão para o(s) Fisioterapeuta(s) que trabalham com Doentes de Huntington (Etchegary, 2011)?

Sociedade e Huntington – Necessidades

- Sente que a Fisioterapia ajuda a solucionar os seus problemas?
- Qual o impacto da Fisioterapia na sua vida (adap. Frich et al., 2014)?
- Alguém o tratou de forma diferente por ter Huntington (Etchegary, 2011)? Se sim, o que aconteceu?
- Acha que a Fisioterapia o podia ter ajudado nessa situação (ou em que situações especificas no seu dia-a-dia)?
- Acha que a Fisioterapia poderia ajudar na sua integração social?
- Por fim, existe mais alguma questão, preocupação adicional em relação à sua Saúde, e em especial à Fisioterapia que queira referir?

Obrigada pela sua participação e disponibilidade

Appendix 3 – SRQR

Title and Abstract	Page
Title - Concise description of the nature and topic of the study Identifying the study as qualitative or indicating the approach (e.g., ethnography, grounded theory) or data collection methods (e.g., interview, focus group) is recommended	i
Abstract - Summary of key elements of the study using the abstract format of the intended publication; typically includes background, purpose, methods, results, and conclusions	vi - vii

Introduction	
Problem formulation - Description and significance of the problem/phenomenon studied; review of relevant theory and empirical work; problem statement	1-6
Purpose or research question - Purpose of the study and specific objectives or questions	6

Methods	
Qualitative approach and research paradigm - Qualitative approach (e.g., ethnography, grounded theory, case study, phenomenology, narrative research) and guiding theory if appropriate; identifying the research paradigm (e.g., postpositivist, constructivist/ interpretivist) is also recommended; rationale**	6
Researcher characteristics and reflexivity - Researchers' characteristics that may influence the research, including personal attributes, qualifications/experience, relationship with participants, assumptions, and/or presuppositions; potential or actual interaction between researchers' characteristics and the research questions, approach, methods, results, and/or transferability	7-10
Context - Setting/site and salient contextual factors; rationale**	7- 8
Sampling strategy - How and why research participants, documents, or events were selected; criteria for deciding when no further sampling was necessary (e.g., sampling saturation); rationale**	7 - 8
Ethical issues pertaining to human subjects - Documentation of approval by an appropriate ethics review board and participant consent, or explanation for lack thereof; other confidentiality and data security issues	11
Data collection methods - Types of data collected; details of data collection procedures including (as appropriate) start and stop dates of data collection and analysis, iterative process, triangulation of sources/methods,	7-10

and modification of procedures in response to evolving study findings; rationale**	
Data collection instruments and technologies - Description of instruments (e.g., interview guides, questionnaires) and devices (e.g., audio recorders) used for data collection; if/how the instrument(s) changed over the course of the study	8-9
Units of study - Number and relevant characteristics of participants, documents, or events included in the study; level of participation (could be reported in results)	11-12; 21
Data processing - Methods for processing data prior to and during analysis, including transcription, data entry, data management and security, verification of data integrity, data coding, and anonymization/de-identification of excerpts	8 - 9
Data analysis - Process by which inferences, themes, etc., were identified and developed, including the researchers involved in data analysis; usually references a specific paradigm or approach; rationale**	9-13
Techniques to enhance trustworthiness - Techniques to enhance trustworthiness and credibility of data analysis (e.g., member checking, audit trail, triangulation); rationale**	9-10

Results and Findings

Synthesis and interpretation - Main findings (e.g., interpretations, inferences, and themes); might include development of a theory or model, or integration with prior research or theory	11-24
Links to empirical data - Evidence (e.g., quotes, field notes, text excerpts, photographs) to substantiate analytic findings	11-24

Discussion

Integration with prior work, implications, transferability, and contribution(s) to the field - Short summary of main findings; explanation of how findings and conclusions connect to, support, elaborate on, or challenge conclusions of earlier scholarship; discussion of scope of application/generalizability; identification of unique contribution(s) to scholarship in a discipline or field	24-30
Limitations - Trustworthiness and limitations of findings	30

Other

Conflicts of interest - Potential sources of influence or perceived influence on study conduct and conclusions; how these were managed	n.d

Funding - Sources of funding and other support; role of funders in data collection, interpretation, and reporting	n.d.

*The authors created the SRQR by searching the literature to identify guidelines, reporting standards, and critical appraisal criteria for qualitative research; reviewing the reference lists of retrieved sources; and contacting experts to gain feedback. The SRQR aims to improve the transparency of all aspects of qualitative research by providing clear standards for reporting qualitative research. 3

**The rationale should briefly discuss the justification for choosing that theory, approach, method, or technique rather than other options available, the assumptions and limitations implicit in those choices, and how those choices influence study conclusions and transferability. As appropriate, the rationale for several items might be discussed together

Reference: O'Brien BC, Harris IB, Beckman TJ, Reed DA, Cook DA. Standards for reporting qualitative research: a synthesis of recommendations. Academic Medicine, Vol. 89, No. 9 / Sept 2014 DOI: 10.1097/ACM.0000000000000388

*n.d.- not discriminated

Appendix 4 – Explanatory Letter to Participants

"We Live With it" - Portuguese Patients and Relatives' perspectives of Physiotherapy role in the course of Huntington's Disease - A qualitative Research

"Nós vivemos com isto" – Perspetivas dos Pacientes e Familiares de Doença de Huntington sobre o papel da Fisioterapia – Estudo Qualitativo

A DH combina alterações ao nível motor, cognitivo e comportamental (Frich et al., 2014; Oliveira et al., 2020). Este sintomas podem afetar a capacidade do individuo de participar em atividades do seu dia a dia em trabalhar, em participar na comunidade e por último reduzir a sua qualidade de vida (Busse et al., 2008; Quinn et al., 2020). Não existe cura para a DH, contudo existem alguns tratamentos que podem ajudar os pacientes e os seus cuidadores. Deste modo, o tratamento atual consiste maioritariamente na gestão dos sintomas e na melhoria da qualidade de vida (Busse et al., 2008; Frich et al., 2014).

Atualmente, a Fisioterapia para DH junta um conjunto de intervenções como o exercício, a promoção da atividade física, treino de equilíbrio e de tarefas especificas, educação e Fisioterapia Respiratória (Quinn et al., 2020). Através de uma parceria com a família, cuidadores, e comunidade, a Fisioterapia procura capacitar os pacientes a alcançar o seu nível ótimo de funcionalidade física e social, tendo um papel importante na manutenção da independência (Busse et al., 2008; Gibson & Martin, 2003; Quinn et al., 2020; Quinn & Busse, 2012). Deste modo a Fisioterapia tenta assim facilitar e por vezes resolver os problemas e desafios desta população.

Por tudo isto, o objetivo deste Estudo Qualitativo é perceber o papel da Fisioterapia e como é que esta ajuda no dia-dia, nos desafios e na resolução dos mesmos dos pacientes e das suas famílias.

O presente estudo desenvolve-se no âmbito da Dissertação da Tese de Mestrado, do Mestrado em Prática Avançada em Fisioterapia Neurológica da Escola Superior de Saúde do Instituto Politécnico de Setúbal (IPS), da aluna Inês Agostinho sob orientação do Professor João Casaca Carreira. A participação no estudo é totalmente voluntária. É totalmente livre de desistir a qualquer momento. A decisão de desistir ou de não participar não acarreta nenhuma consequência.

Caso decida aceitar participar, além das entrevistas, serão recolhidas várias informações da sua vida pessoal e social, no entanto, todos os dados serão recolhidos ao abrigo do Regulamento Geral de Proteção de Dados em vigor no IPS e assim, a informação recolhida será confidencial e anónima. Ser-lhe-á atribuído um código, que só o investigador principal terá acesso, o que assegurará a confidencialidade dos seis dados e informações fornecidas, uma vez que, na Dissertação e documentos subsequentes a este trabalho a

referência virá sempre pelo código sem qualquer informação pessoal associada. Se em algum momento decidir deixar de participar, os seus dados será imediatamente apagados, se for essa a sua decisão. As entrevistas irão decorrer através da Associação Portuguesa de Doentes de Huntington ou de outros contatos específicos. As mesmas poderão ter uma duração variável, com o tempo estimado de 45-90 minutos.

Agradecemos desse já a sua participação,

Para mais informações, não hesite em contactar,

Inês Agostinho - 969892345

210537008@estudantes,ips.pt

Prof João Casaca Carreira

joao.carreira@ess.ips.pt

Appendix 5 – Informed Consent

CONSENTIMENTO INFORMADO, ESCLARECIDO E LIVRE PARA PARTICIPAÇÃO EM ESTUDOS DE INVESTIGAÇÃO (de acordo com a Declaração de Helsínquia e a Convenção de Oviedo)

Título do estudo: "We Live With it" - Portuguese Patients and Relatives' perspectives of Physiotherapy role in the course of Huntington's Disease – A qualitative Research

"Nós vivemos com isto" – Perspetivas dos Pacientes e Familiares de Doença de Huntington sobre o papel da Fisioterapia – Estudo Qualitativo

Enquadramento: A Doença de Huntington (DH) é uma doença hereditária autossómica dominante, de carácter progressivo e neurodegenerativo (Frich et al., 2014; Oliveira et al., 2020; Roos, 2010; Røthing et al., 2015). A prevalência da DH na Europa está estimada em 5-10:100.000 na Europa, em Portugal é igualmente estimada uma prevalência 5-10:100.000 (Carmo Costa et al., 2003; Oliveira et al., 2020). Afeta ambos os sexos e cada filho de um parente com DH tem 50.50 chance de herdar a mutação (Etchegary, 2011).

A DH combina alterações ao nível motor, cognitivo e comportamental (Frich et al., 2014; Oliveira et al., 2020). A média de idade de diagnóstico é entre os 40-45 anos, porém os sintomas podem aparecer mais cedo (Frich et al., 2014; Roos, 2010). Este sintomas podem afetar a capacidade do individuo de participar em atividades do seu dia a dia, em trabalhar, em participar na comunidade e por último reduzir a sua qualidade de vida (Busse et al., 2008; Quinn et al., 2020). Não existe cura para a DH, contudo existem alguns tratamentos que podem ajudar os pacientes e os seus cuidadores, assim, e uma vez que os sintomas alteram a vida dos DH, o tratamento atual consiste maioritariamente na gestão dos sintomas e na melhoria da qualidade de vida (Busse et al., 2008; Frich et al., 2014).

Neste momento, a Fisioterapia para DH junta um conjunto de intervenções como o exercício, a promoção da atividade física, treino de equilíbrio e de tarefas especificas, educação e Fisioterapia Respiratória (Quinn et al., 2020). A Fisioterapia tenta assim facilitar e por vezes resolver os problemas e desafios desta população. Por vezes apenas o pacientes e as suas famílias sabem pelo que estão a passar, e até os Fisioterapeutas e outros profissionais de Saúde precisam de ajuda para entender.

Objetivo: Perceber como é que a Fisioterapia é relevante e ajuda no dia-dia, nos desafios e na resolução dos mesmos dos pacientes e das suas famílias.

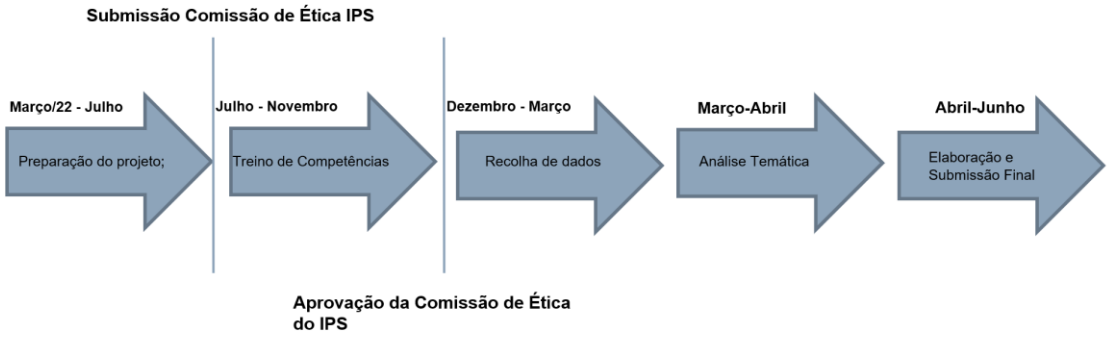
Local do estudo e pessoal responsável: O Estudo desenvolve-se no âmbito da Dissertação da Tese de Mestrado, do Mestrado em Prática Avançada em Fisioterapia Neurológica da Escola Superior de Saúde do Instituto Politécnico de Setúbal (IPS), da aluna e Inês Agostinho sob orientação do Professor João Casaca Carreira.

Explicação do estudo: As entrevistas irão decorrer através da Associação Portuguesa dos Doentes de Huntington ou de outros contatos específicos. As mesmas poderão ter uma duração variável, com o tempo estimado de 45-90 minutos. Prevê-se a recolha de dados demográficos tais como a idade, o género, as habilitações literárias, o estado civil entre outras. Todas as entrevistas serão gravadas através de imagem e som, e transcritas manualmente pela aluna Inês Agostinho. Todos os dados pessoais serão eliminados e a confidencialidade dos participantes assegurada através de uma codificação. Codificação esta que será realizada num documento Exel à parte, devidamente seguro através de uma palavra-passe de acesso ao mesmo;
Os resultados deste estudo irão potencialmente contribuir para melhorar o conhecimento sobre a relevância da Fisioterapia na vida das pessoas com DH e suas famílias, perceber o que ainda se pode melhorar/modificar, a fim de assegurar uma melhor integração das pessoas com DH na sociedade.
Além de integrarem a tese da aluna Inês Agostinho, adicionalmente os resultados obtidos poderão vir a ser publicados em estudos científicos ou usados para materiais em congressos nacionais/internacionais, sem que haja qualquer quebra na confidencialidade/anonimato.
Condições e financiamento: A participação no estudo é totalmente voluntária. Se decidir participar ser-lhe-á pedido que assine este consentimento informado. É totalmente livre de desistir a qualquer momento. A decisão de desistir ou de não participar não acarreta nenhuma consequência para o participante. Não se antecipam inconvenientes ou efeitos secundários à participação no estudo. Não estão comtempladas quaisquer compensações monetárias ou outras pela participação no estudo.
Confidencialidade e anonimato: Todos os dados serão recolhidos ao abrigo do Regulamento Geral de Proteção de Dados em vigor no IPS. Assim, a informação recolhida será confidencial e anónima. A cada participante será atribuído um código que será utilizado em todos os processos de armazenamento e tratamento dos dados;

Muito obrigada pela sua leitura, para quaisquer esclarecimentos adicionais, por favor contacte: Inês Agostinho – 969892345, email 210537008@estudantes.ips.pt ou o Prof. Orientador João Casaca Carreira – joao.carreira@ess.ips.pt ;
Se concorda com a proposta que lhe foi feita, queira, por favor, assinar este documento.
Assinatura de quem recolhe o consentimento:
… … … … … … … ..... … … … … … … … … … … … … … … … … … … ..... … … … ..... … … … …

☐ Declaro ter lido e compreendido este documento, bem como a informação patente na carta explicativa e as informações verbais que me foram fornecidas pela pessoa que acima assina. Foi-me garantida a possibilidade de, em qualquer altura, recusar participar neste estudo sem qualquer tipo de consequências. Desta forma, aceito participar neste estudo e permito a utilização dos dados que de forma voluntária forneço, confiando em que apenas serão utilizados para esta investigação e nas garantias de confidencialidade e anonimato que me são dadas pelo/a investigador/a. Desta forma assino livremente este documento original, sendo-me fornecido um duplicado também ele devidamente datado e assinado.
Nome: … … … … … … … ..... … … … ..... … … … … … … … … … … … …
Assinatura:… … … … … … … ..... … … … … … … … … … … … … … … … **Data:** ..... /.....

Appendix 6 - Timeline



Appendix 7 – Thematic Analyses

DH – Theme 1 – Objectives and ICF

Perspetivas de vida e objetivos após o diagnóstico	**Estruturas e função (sintomas); problemas que advém da doença**	**Atividades (limitação no dia-a-dia); Implicações dos sintomas/doença no dia-a-dia**
U_02: Os meus objetivos era manter a minha saúde em dia… manter a minha saúde em dia, não estar tão perra como… *(objetivo do dia-dia);*	**U_02:** Não… foi quedas (enfatiza), que eu caia muito, eee dores no corpo sim… *(primeiros sintomas)* (…) **U_02**: é muito muito movimento… ando sempre a cair…	**U_02:** Mudou tudo… infelizmente… tudo (enfatiza) (impercetível).. deixei de trabalhar, deixei de ter uma vida como devia ser, a minha vida foi, tem sido um desânimo (…) Tenho vivido mal… agora até está pior, porque antigamente ainda estendia uma roupita, ainda punha a máquina a lavar, ainda fazia as minhas coisas, só que agora já não faço nada, infelizmente (enfatiza), nada a haver… nadinha…
U_06: (pensativo) mudou tudo, porque não estava a espera disto…	**U_07: (…)** falta de equilíbrio (…) pa além ahh palém de da da ansiedade (…)	**U_04:** é para me vestir, para ir à casa de banho; *(principais problemas)*

	problemas de vez em quando de memória embora a memória não não me afete ainda muito muito mas comecei a ter assim coisas que nunca tinha não é? ahh pequenos lapsos e depois a capacidade de atenção.	(…) **U_04:** muito muito, tem… eu tremo muito… eu não consigo fazer nada! *(limitação em casa)*
C_03: partir do momento que foi feito o diagnóstico au(ou) menos a minha família soube o que poderia vir (pausa) dali para a frente, de cuidados e em termos consequências da própria doença.	**U_09**: ahhh agora, ando muito pouco, pouco ando, já cai por casa, dei duas quedas, e e tenho medo de de cair, medo de andar sozinha em casa… *(referindo-se aos principais problemas)*	**U_06**: (…) tinha poucos sintomas, não tremia, caminhava, fazia exercício, andava de bicicleta, ginásio, fazia essas coisas todas… trabalhava… e assim… *(referindo-se a atividades que deixou de fazer)*
C_09: Na realidade a mudança não foi muita, porque nós já sabíamos a patologia, todos nós achávamos que já existia esta patologia de base, na pessoa em questão, portanto nós já tínhamos um conhecimento, antes do diagnóstico nós já achávamos que era ahhh portanto a patologia em questão e tínhamos um	**C_01**: ahhh tem problemas de fala ahhhh problemas de locomoção, estes são os mais (enfatiza) óbvios. Ahhh tem problemas deeee deglutição (…) tem alguns problemas respiratórios pequenos problemas comportamentais de fixações, (…) fica sempre naquela, naquele registo e não consegue desligar daquele	**C_02:** porque ela ainda não está totalmente acamada, faz o levante (…) já não aguenta o tronco, mas pronto, (…) ahhh, o problema da minha mãe é o, o, o mudar a fralda que ela começ... é agressiva, a dar, a dar a alimentação que ela pede também é agressiva, a higiene, a al..., pronto, não, não, não gosta (…)

diagnóstico clínico, portanto na realidade foi apenas a confirmação daquilo que nós já tínhamos, que nós já sabíamos. Portanto a mudança não foi muita	registo (…) mas por enquanto vamos tentando minimizar ahhh os efeitos de de desse tipo de comportamento; (…) pronto ela neste momento já tem uma PEG;	
	C_03: começou a ganhar mais dificuldade a movimentar-se, a andar, a segurar nos talheres foi na mesma altura que também começou a ter mais dificuldade na fala.	**C_04**: (…) \vão perdendo a autonomia da sua própria vida… e terem que depender das tarefas mais simples que nós automaticamente fazemos… e elas acabam por pouco a pouco ir perdendo essa capacidade… mesmo de fazer a sua higiene quando vão à casa de banho (…)mesmo na própria refeição já vai comendo, mas eu tenho que depois ajuda-la a comer porque a comida depois acaba por estar, por espalhar-se na mesa porque e ela insiste ainda em comer sozinha e eu acabo por deixar (…) Na e e quando vai a casa de banho ou tomar banho já tenho, são tarefas que já têm de ser ajudadas por mim…

	C_11: a minha mãe apresenta graves problemas de equilíbrio, tem uma, problemas de deglutição; tem problemas de de de nitidez de de de linguagem ahhh e tem alguns ahhh problemas cognitivos que podem ou não estar associados à doença, pode ser também associado ao avanço da idade (…)	**C_07**: (…) E neste momento a minha mãe já tá numa cadeira de rodas, ela consegue levantar-se, mas só para dar alguns passinhos, uma coisa pouca. Ajuda a ir à casa de banho, ajuda a vestir-se, ajuda a, a pôr a fralda quando é a hora de ir dormir, portanto ela consegue ajudar, mas já, mas já tem que ser… eu tenho que lá estar sempre, não é?… o banho, é tudo com ajuda

Sociedade e DH – Theme 2- Society and HD

Comportamento da Sociedade perante a DH	**Conhecimento da Sociedade sobre a DH**	**Papel da APDH**	**Importância de normalizar a doença**
U_02: não, onde é que eu moro não… as pessoas tratam-me lá com respeito!	**U_04:** não devem conhecer a doença…	**C_04**: Na informação, na partilha, nas reuniões que são feitas e cada um partilha do… daquilo que que passa ou que passou e da partilha de	**C_03**: eu acho que o falar sobre este tipo de situações ou neste caso uma doença rara acaba por normalizar ahhh o que é os sintomas e o que é as consequências da doença, eu

		conhecimentos (…) aquilo que resulta e partilha com outros (…) então esta partilha acaba por ajudar uns aos outros, mesmo em termos de conhecimento o o o oque é a doença de Huntington, eu só consegui saber o que é a doença de Huntington através da associação. Através da literatura que foi disponibilizada, os sintomas e que muitas vezes aquilo que o doente diz ou faz não é ele próprio é a doença, a doença é que faz com que ela aja ou diga quilo que diz naquela altura.	acho que é difícil mudar uma sociedade que é… (pensativa) antiquada talvez em termos de diferença… eu acho que essa é a única questão (…)

U_06: para mim as pessoas são boas, não tenho problema com sociedade!	**U_05:** A sociedade não sabe o que é que é a doença de Huntington… (pausa) temos que… têm que se, quando dizemos que temos a doença temos que explicar tudo e mais alguma coisa, e mesmo assim não… não… não acreditam, pronto nunca vêm que há realmente tá/(está) a acontecer.	**C_10:** (…) até mesmo eu inscrevi-me no no no no Huntington no que… nos Portugueses e também não há nada, não vejo nada de novo;	**C_04**: Não não não me perguntaram, não não me perguntaram, eu eu é que quando tenho oportunidade, digo que ela têm uma doença degenerativa, que é a doença de Huntington
U_02: A sociedade não está preparada ainda, essas coisas, ainda nem entraram em conta. Há muita gente que ri e pensam que é que é um AVC que me deu. Então as pessoas ás vezes riam (impercetível) e na na Fisioterapia faziam…	**U_07:** (…) acho que a doença de Huntington ainda… ainda está muito invisível…		

C_01: (…) as pessoas tendem a aaaa afastar-se um pouco deee de pessoas com este tipo de doenças como se fosse uma coisa hummm ahhhhh (pensativa) contagiosa (sorri) como se fosse alguma doença contagiosa (…), há pessoas que pronto detetam podem perceber que que sim que é doença conforme o tipo de doente que aparece pela frente e afastam-se tipo (faz gesto) vamos afastar antes que se pegue alguma coisa, há outras pessoas que pensam que é algum drogado ou algum bêbado ou coisa assim.	**C_05**: ahhh, a a sociedade não tem conhecimento (…) uma vez ia com uma tia ou estava com os movimentos desconformes na rua e houve uma senhora que passou e diz assim "logo de manhã e já com uma bebedeira destas (…)		**C_11**: eu acho que que que toda a gente pode ter um papel nesta doença, quanto mais não seja para explicar as pessoas que, o que é a doença de Huntington e toda gente a respeita muito bem, porque percebe, podem não perceber o que é uma doença de Huntington, mas sabem que a minha mãe tem uma doença e que é por isso que precisa de ajuda, pronto, acho que todos nós temos um papel ahhh ee cabe-nos a nós, todos a a aos fisioterapeutas e a todos, que lidamos com com com isto (…) e quando quando a minha mãe toma um café e

			pode entornar um bocadinho ou qualquer coisa, explicar que é normal, que é normal, pronto e que que temos que normalizar as as as doenças pronto e explicar às pessoas que é fruto do que é pronto, uma doença que se chama no caso da minha mãe é coreia de Huntington noutro caso poderá ser outra coisa qualquer!
C_03: acho que grande parte da sociedade tem, não aceita muito bem o desconhecido e quando não aceita também tende a não querer saber mais e a querer melhorar,	**C_09**: a questão de ter uma doença que pouca gente conhece, que pouca gente sabe lidar, quer em termos sociais, quer em termos de família, quer em termos de saúde, e as implicações que isso tem		

C_05: porque o resto a maioria das pessoas… e cada vez mais ahh se nota uma indiferença muito grande…	**C_10**: não sei, a Sociedade não conhece… a maior parte das pessoas não conhecem esta doença… ficam assim…as pessoas perguntam-me… (…)		

Cuidados de Saúde – Theme 3 – Health Care and Needs in HD:

Reabilitação na DH	**Implicações e limitações no acesso às terapias**	**Equipa Multidisciplinar e relação entre eles**	**Desconhecimento dos profissionais de saúde**
U_05: também tenho psicologia (pausa)…. E… neu… neurologia no… no hospital…	**U_02:** (…) tenho medo de ir, tenho medo de ir sozinha para a Fisioterapia por cair… e partir alguma coisa, e então tenho medo…	**U_06**: acho que não existe muito contacto… *(relatórios entre intervenientes)*	**C_04:** (…) se nós, se nós falarmos que, até em meio hospitalar a maior parte dos médicos e enfermeiros não sabiam (enfatiza) lidar com a doença de Huntington

			quanto mais a população comum
U_06: vou às minha consultas em Coimbra ahh, até tenho uma marcada para amanhã, tenho duas consultas este mês; uma de psiquiatria e uma de de de neurologia, e por isso tenho apoio dessas partes, e também os medicamentos e ahhh	**C_01:** Tudo particular, tudo pago por mim.	**U_07: (…)** depois escreveu-me uma carta da da ahh a explicar a minha situação para eu entregar ai numa numa clinica de fisioterapia pronto e foi o que fiz (pausa) ahhhh e eles seguiram mais ou menos penso eu aquilo que eles diziam (…)	**C_05**: até sabe que há médicos que não sabem o que é a doença de Huntington?
U_07: (…) olhe eu tenho todo o apoio do hospital hm universitário de Coimbra. Por acaso tenho eles ahh nesse aspeto não me deixam não me deixam sozinho. Tenho sempre consultas ahhh marcam-me sempre consultas quer de neurologia quer de psiquiatria por exemplo nas duas áreas principalmente estas. Ahhh tenho prai 2 consultas por ano de neurologia e também 2 de de psiquiatria pelo menos.	**C_06**: sim a fisioterapia não é diária por causa dos custos...porque isto , nós indo para uma clinica privada tem sempre um custo mais acrescido.	**C_02:** São separadas por que são… ahh são de sítios diferentes ahhhh pronto… há… não há muita ligação… (...) é claro que perguntavam o que é que não sei quantos disse, o que é que, sim, havia essa preocupação mas não havia… uma reunião… uma discussão de ideias ou de pareceres e…	**C_10**: as pessoas mesmo… mesmo mesmo pessoas ligadas à saúde; há muita gente ligada à saúde que não conhece;

C_01: temos que lhe proporcionar o máximo de terapias que conseguimos, desde Terapia da Fala para ajudar a manter a fala, Fisioterapia, ahhhh, portanto (pensativa), ahhhh Terapia Ocupacional, todo o tipo de terapias que que, Psicóloga, ahhhh todo o tipo de terapias que que possamos proporcionar é hummmm, penso que serão uma ajuda…	**C_04:** Eu acho que isso era importante que todos os doentes tivessem acesso, quer com possibilidades ou quer não, porque se calhar a maioria não tem essa possibilidade, de ahh pagar esses tratamentos… era o o Serviço Nacional de Saúde, ahhh permitir… e não era só aquelas sessões mínimas e depois olhe, vá para casa e continue a fazer, faça assim com a bola ou faça assim não sei o quê, isso não é suficiente…	**C_07:** Não, não há, até porque não são do mesmo hospital. (…) Portanto não, nenhum destes terapeutas tem, tem contacto uns com os outros.	
C_06: ela é acompanhada na ..no Santa Maria de 6 meses em 6 meses, que na minha opinião é pouquíssimo ...e tem fisioterapia duas vezes por semana numa clinica privada;	**C_07**: A parte da fisioterapia e terapia ocupacional e tudo… ahhh, aquilo que me foi dito foi que ahhh, eles têm poucas vagas e então dão prioridade a quem tem hipóteses de melhoria, ou seja, alguém que é para manter ou para tentar	**C_11**: a mim parece-me que não comunicam, a única coisa que me parece é que eles recebem um um receituário, e tentam cumprir e fazer chegar	

	atrasar uma doença ahhh, não é prioritário, pronto. Ahhh… Terapia da fala penso que também foi um bocadinho por aí, portanto, ahhh (…)	ao ao ao doente aquilo que o médico receitou (…)	
C_07: A minha mãe ahhh felizmente é acompanhada em várias áreas, portanto ahhh assim que houve um diagnostico de doença de Huntington foi a neurologia, não é? Ahh… Foi terapia da fala, foi gastro portanto por causa da parte da deglutição .. ahh .. (pensativa) (…) Psicologia também estavam constantemente a falhar eeee… e optámos por fazer no particular, portanto a minha mãe anda em psicologia, mas no particular. Fisioterapia a mesma coisa, tivemos 2 anos à espera e, e depois acabámos por ir para o particular. Fez terapia	**C_12**: Agora, tudo o resto era pago por fora, porque se eu estivesse à espera que alguém me arranjasse, uma fisioterapeuta, para vir cá ou…, nem pensar, quer dizer, arranjava um ano e tal depois. A cadeira de rodas fui eu que comprei, todos os equipamentos, nem esperei porque não havia possibilidades de ter o que está à espera, não é? Isso, para quem não tem em termos de possibilidades económicas, torna-se então muito mais complicado.	**C_12**: ela *(fisioterapeuta)* fazia um relatório que enviava para a médica, ela aliás chegava a mandar e emails à doutora (…) e a doutora (…) para ela, portanto para a evolução do XXX, o que é que ela notava, era uma coisa que me agradava bastante	

ocupacional, mas foram uma série de 12 sessões, acho eu, ou… ou.. eu sei que deu cerca de... (pensativa) 2 meses talvez, e entretanto também teve alta, já não a vão chamar mais (risos) Portanto … Mas pronto, mas de resto, a parte neurológica sim, está a ser bem acompanhada. Há aqui vários serviços que que a minha mãe conseguiu ter acesso.			

Fisioterapia na DH – Theme 3 – Physiotherapy in HD: …

Sessões de Fisioterapia	**E se não realizasse Ft?**	**Relação Ft/Doente e Ft/Cuidador**	**Resultados of Physiotherapy**
U_02: Fazia 15 minutos de passadeira depois fazia lá umas coisinhas, agora fazia	**C_02**: siiim, retardou um pouco a paragem dela, a parte motora ter parado tao tão tão tarde não é?, se ela	**C_07**: Eu nunca assisti às sessões da minha mãe. Ahhh, eu nem tenho contacto com a fisio, fisioterapeuta. (…) Ahhhh, e falei uma vez com	**U_02:** Tinha impacto… (…) **U_02:** Positivo…

movimento às mãos e fazia massa… massagens pés;	não tivesse se calhar feito isso já estava há mais tempo há mais tempo debilitada… *mais debilitada* (enfatiza) sem dúvida	uma das fisioterapeutas porque, por acaso, fui ao centro de dia na mesma altura em que ela lá estava e ela pediu pa falar comigo. Ahhh, mas de resto não há, não há grande contacto.	(…) **U_02:** Diz que ajudava, melhora o andar, mas eu saia de lá cansada (…) **U_02**: era… era… era… e não melhorou em nada… nadinha… *(referindo-se às atividades do dia-a-dia);*
(…)**U_04:** mexo as pernas; (…)**U_04:** mexo os braços… (…)**U_04:** faço coisas difíceis; (…)**U_04:** ter de andar (…)**U_04:** fazer coisas relacionadas com os braços… **U_04:** sim… *(referindo-se a exercícios funcionais)*	**C_03**: Acho que tinha falecido muito mais cedo, porque também temos conhecimento de outras histórias de pessoas com a mesma doença que simplesmente são internadas em casas de cuidados continuados e que na verdade não tem acesso ahum, a este tipo de serviços e acabam por deteriorar- se muito mais rapidamente e acabam por falecer.	**U_07**: (…) Fisioterapeuta não tinha muita noção o que era a doença de Huntington e eu fui-lhe explicando (…) **U_07:** ajudou ajudou muito ajudou muito ajudou ajudou, porque ela depois em função desses sintomas e dessas coisas que… ela fazia os exercícios, lá está também começou a afinar melhor os exercícios mais adequados	**U_04**: (…) dá-me mais força **(…)** **U_04**: para ir à casa de banho (…) **U_04:** sim… *(referindo-se à ajuda nas AVD's)* **U_04**: eu acho que sim… alivia… (impercetível) ajudando a mim ajuda a eles…*(referindo-se ao impacto da Fisioterapia nos familiares)*

		para mim; ahhh com…. Ela fez isso eee eu notei, por acaso notei, notei que ela de facto tinha esse cuidado, o exercício ser exatamente para o individuo é para, é para individuo com esta característica e aquela e a outra, pronto foi…era uma Fisioterapeuta 5 est… uma Fisioterapeuta… pois… *(referindo-se à ajuda da Fisioterapeuta na gestão da doença)*	
U_06: pronto…e e era assim uma sessão típica era isso às vezes tinha exercícios novos e diferentes, como esse de	**C_06**:provavelmente já estaria acamada. Provavelmente já estaria. Ela não tem nenhum tipo de exercício a não ser o da fisioterapia. Mas ….nós por mais que tentemos que ela faça connosco, lá é sempre	**U_09:** não, não o Fisioterapeuta nunca falou comigo…	**U_05:** Sinto-me melhor e mais… com menos movimentos involuntários e… (pausa) com mais força (impercetível)…

sair à rua e tal, e dar uma volta e subir e descer escadas mas começámos a fazer isso levantar pesos, pôr pesos nas pernas levantar as pernas, esse tipo de exercícios, era uma sessão era mais ao menos isso;	uma obrigação, cá ela é: "não consigo, não quero!" não dá para forçar ...(ri)		
		U_09: não… ele pouco falou com ela… *(interação Fisioterapeuta-cuidador)*	**U_06**: a Fisioterapia ahhh ahhh adiciona positivamente ahhh, por que me ajuda a não declinar tanto, e a manter o bom que eu tenho;
	C_07: Ai, muito pior. Muito pior sem dúvida. E lamento que não tenha começado mais cedo. Porque, eu acho sinceramente	**C_03:** mas sem dúvida que nos sentíamos integrados inclusivamente eram nos explicados alguns exercícios que poderíamos ahhh ir fazendo ao longo do dia, ela não tinha Fisioterapia todos os	**U_07:** nível muscular já não me senti assim há muito tempo, foi mesmo muito bom; e e pronto também ganhei alguma

	que, por exemplo o andar, o andar podíamos ter ahh atrasado a cadeira de rodas, se houvesse fisioterapia.	dias, tinha, numa fase inicial tinha dia sim dia não e nós fazíamos alguns exercícios com ela nos outros dias	auto confiança, ganhei um boca…um pouco também de autoconfiança mais confiança a andar, porque também fizemos… muitos exercícios a andar e a subir e a descer escadas e ahhh ahhh, na rua, mesmo na rua, e a… ahhh Fisioterapeuta leva-me lá para fora, e vamos subir e vamos descer e vamos subir e vamos descer as escadas, e isso fez-me bastante bem, ganhei confiança para isso porque porque para subir e descer escadas também já tenho de pensar duas vezes onde é que vou pôr o pé;

	C_10: ai estava muito pior, estava muito pior em todos os aspetos… (prossegue) **C_10**: acho que ele não se mexia…	**C_06**: por acaso não, por acaso não…não há muita conversa nessa vertente. É que o que foi falado é que ela tinha essa doença (…) e eles fizeram um plano especifico, entretanto quem ficou, quem falou sobre esse plano até foi o meu tio mas entretanto não… foi uma boa questão... tenho de lá falar com elas... (…) sobre isso mas não estou ativa dentro do plano;	**U_09:** não, por acaso nada, não me fazia nada, não, não está bem adaptada à minha doença, não me fazia nada… *(referindo-se ao impacto da Fisioterapia no dia-a-dia)*
U_09: hum… não, não fazia mais nada… andar lá num corredor só… num corredor pequeno… (…) **U_09**: andar para trás e para diante… **U_09:** era toda manhã… *(referindo-se à duração da Fisioterapia)*			**C_03**: notava-se que ela estava contente e e bem quando quando acontecia a Fisioterapia.

			C_07: Eu acho que sim, mas de uma forma muito suave, ou seja, melhorando a autoestima, claro que vai mexer com o resto. Portanto, a parte comportamental vai ter alguma influência aqui, sim. Ahh… Pode… Eu, eu acho que tudo tem a ver com a autoestima, não é? (…) Como eu senti que ela, que ela melhorou na, no inicio, ahhh, a parte comportamental vai acalmar, porque fica, fica mais satisfeita, fica mais, mais feliz, como é obvio não fica tão angustiada e a parte comportamental fica mais tranquila mais, mais calma. Acho que sim.
			C_06: Tem um efeito só...é pouco duradouro o efeito
			C_07: A ela, além disto, além de sentir que é capaz, é a autoestima, porque finalmente é capaz, não é? E… e notámos

			muito isso quando ela começou a fazer, no início, quando começou a fazer terapia, coincidiu com a fase de terapia ocupacional, ela começou as duas mais ou menos na mesma altura, ahhh, e ela estava com a autoestima muito mais elevada, porque já... voltou a ser capaz de fazer algumas coisas que já não era e… e isto acho que é ótimo para, para as duas ou para todos cá em casa. (risos)
			C_09: Então ahh aumento de força, de estabilidade, equilíbrio, aprendeu como cair, proteger-se das quedas, ahhh, sobretudo também melhorou a questão da rotina porque são doentes que ficam muito tempo na cama, podem ficar a dormir, e portanto obrigava o meu pai a sair de casa, não só a parte física, mas também a parte ahh

			comportamental, a parte do dia a dia, das rotinas.

Cuidador e Família – Theme 5 Family and HD

Impacto na vida e dos cuidadores e familiares	**Sobrecarga dos familiares**	**Preocupação com a Hereditariedade**
C_01: (…) porque o objetivo agora é única e exclusivamente cuidar da, da minha filha portanto tudo o resto de perspetivas diárias, de de de, diárias a curto, longo médio prazo ahhh, deixam de de fazer sentido, porque é tudo planeado em função ahhh da doença e daquilo que a doença permite.	**C_02:** eu antes de ela desistir da Liga por exemplo eu tinha que às vezes todos os dias ou porque a mãe chegava lá e adormecia ou porque a mãe não queria fazer nada quer dizer eu fui ouvindo, uma duas… até que tive começar a ir com ela, vinha a correr do trabalho tinha que ficar lá à à porta à espera, depois entrava é cansativo é… é um desgaste e depois perco toda uma vida lá fora e e e deixo tudo para trás;	**C_03:** (…) pronto e trouxe uma carga acrescida, difícil no sentido em que é possível, eu e neste caso o meu irmão termos essa doença, ahh e estarmos alia ver o que nos poderia acontecer.
C_02: Pronto, e então, basicamente perdi a minha juventude, a minha liberdade, não me	**C_05**: (…) porque me pedem para ir, ou ir buscar um medicamento ou porque	**C_04**: (pensa) Olhe, eu quandoo, quandoooo ahhh comecei a perceber na

dediquei aos meus estudos como se calhar gostaria, mas não me posso queixar porque, de, do, perante o panorama todo ainda consegui ter uma formação. Ahhh, mas por outro lado também ganhei outras coisas que, que me fizeram ver as coi..., a vida de outra maneira. Eeee a responsabilidade, ainda mais.	tenho que ir buscar um medicamento ao médico, ou porque tenho que ir marcar uma consulta, ou porque tenho queeee, ir a qualquer lado, (…) , mas quando andava íamos lá todos os sábados e fazíamos uma pequena caminhada com ela, para ela não, não perder os movimentos, chegou a um ponto que já não não é? (…) e é e é e é aquele apoio sempre que é que é preciso dar à família não é? (…) tou-lhe/(estou-lhe) a dar muito mais apoio, mas chega a um ponto que eu também já estou a dizer assim, *"pah, eu tenho tirar um bocadinho para mim não é?"* apesar de eu de eu fazer muita coisa e de estar em muita coisa, mas eu também se não tiver em certas e determinadas coisas que sou eu ahhh dou em doido…	XXX, porque já tinha passado pela primeira, pela, eu, eu acabei por ee… ee… quando eu vi que ela tinha alguns, começou a mostrar alguns movimentos que eu já tinha visto na YYY, eu só pensei, "*eu eu não acredito que, que isto possa estar a acontecer outra vez nesta segunda"*, na segunda filha, não foi fácil… muuuuuito, mas muito, muito complicado, muito complicado, ahhh porque ver uma filha já a morrer com esta doença, que o ultimo ano, ela esteve (…), amarrada a uma cama, já com a PEG, ahuuum e estar a ver… depois a começar a reviver tudo novamente na segunda filha… não, não é fácil, não é, não é fácil…

	C_10: para uma pessoa sozinha é muito complicado muito complicado eee eu já não consigo… não estava a conseguir… e e depois foi muito, foi muito complicado para mim, e eu e eu e isto sozinha, como foi complicado ele ir para a instituição, eu depois também sofri muito por ele, quando foi para a instituição, porque eu achei que o tinha abandonado	**C_05**: Porque via o ahhh o que é que tava a acontecer às minhas cunhadas "epah" e e não queria que isso acontecesse à minha mulher, nem quero que isso aconteça à minha mulher, não é pelo trabalho que vou ter não é…. se se cá estiver, não é? não é por nada, mas é porque… porque sofrem não é?
U_05: Teve muito impacto ahhh pronto, até acabei por me divorciar e tudo…		

Appendix 8 – SPDMov Congress

Appendix 9 – EHDN Member

EHDN

EUROPEAN **HUNTINGTON'S DISEASE** NETWORK

MEMBERSHIP CERTIFICATE

Inês Agostinho

is as of

16 April 2023

a Regular Member of EHDN

Prof. Anne Rosser
Chair EHDN Executive Committee

PD Dr. Patrick Weydt
Co-Chair EHDN Executive Committee