INTELLIGENCE AND SPECIFIC COGNITIVE FUNCTIONS IN INTELLECTUAL DISABILITY: IMPLICATIONS FOR ASSESSMENT AND CLASSIFICATION

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ABSTRACT

Purpose of the review: Current diagnostic criteria for intellectual disability (ID) categorise ability as measured by IQ tests. However, this does not suit the new conceptualization of ID, which refers to a range of neuro-psychiatric syndromes that have in common early onset, cognitive impairments, and consequent deficits in learning and adaptive functioning. A literature review was undertaken on the concept of intelligence and whether it encompasses a range of specific cognitive functions to solve problems, which might be better reported as a profile, instead of an IQ, with implications for diagnosis and classification of ID.

Recent findings: Data support a model of intelligence consisting of distinct but related processes. Persons with ID with the same IQ level have different cognitive profiles, based on varying factors involved in aetiopathogenesis. Limitations of functioning and many bio-psychological factors associated with ID are more highly correlated with impairments of specific cognitive functions than with overall IQ.

Summary: The current model of intelligence, based on IQ, is of limited utility for ID, given the wide range and variability of cognitive functions and adaptive capacities. Assessing level of individual impairment in executive and specific cognitive functions may be a more useful alternative. This has considerable implications for the revision of the International Classification of Diseases and for the cultural attitude towards ID in general.

KEY POINTS

- IQ test scores have major limitations as indicators of the complex and dynamic nature of cognitive impairment in persons with intellectual disability (ID), including reasoning in real-life situations and mastery of practical tasks;

- The same IQ score can correspond to very different cognitive and functioning profiles;

- Limitations of functioning and various bio-psychological factors associated with ID are more highly correlated with impairments of specific cognitive functions than with overall IQ;

- New diagnostic criteria for ID should include measures of specific cognitive functions, and contextualised description of consequent learning and adaptive difficulties;

- This approach might bring considerable advantages, with a more inclusive cultural attitude towards ID and other neurodevelopmental disabilities, providing a paradigm shift from "low IQ" to "neuropsychological characterisation".

INTRODUCTION

Measurement of Intelligence Quotient (IQ) plays a central role in the classification of Intellectual Disability (ID), and of all the assessments undertaken with children with ID, IQ is often the only cognitive assessment. This can result in diagnostic labels that may fail to capture performance on different cognitive functions. A lower level of intelligence, approximately set at two standard deviations below the mean IQ of the population, has been the common reference and it is assumed to measure a person's capability in managing environmental demands and producing adaptive behaviours. However, evidence from neuropsychology, genetics, neuroimaging and functional anatomy has recently been growing on a high variability in different cognitive abilities within individuals (Friedman *et al.* 2006; Johnson *et al.* 2008), which challenges the current concept of overall intelligence and the use of IQ. This is particularly relevant at this point in time, given that the ICD-10 is currently under revision (WHO 1992). ICD-10's classification of ID includes a wide range of syndromes as well as non-syndromic conditions, which have only early cognitive impairments in common.

We reviewed the literature in order to evaluate (1.) the utility of the concept of intelligence, and the validity of IQ measurement in the definition of ID, as well as (2.) whether measurement of specific cognitive functions and their bio-psychological correlates are more useful for the diagnosis and sub-typing of ID.

Uni-component models of intelligence and IQ

Historically, Spearman (1927) was the first to propose a uni-component model of intelligence. He observed that test scores on different cognitive tasks correlated with each other and concluded this to be explained by one underlying common factor, i.e. "g factor". The investigation of its neural correlates has received increasing attention by researchers in the last thirty years. Several studies found statistically significant correlations between neural activation, particularly of the frontal area, and performance on a number of tests (Duncan et al. 2000; Gray et al. 2003), as well as between IQ and widespread areas of grey and white matter in several brain regions, including centres of higher cognitive functions such as language, memory, or attention (Jung & Haier 2007; Ramsden et al. 2011). Research on people with focal brain damage described some activation associated with both the g factor (Gläscher et al. 2010) and executive functions, such as working memory, verbal comprehension, and perceptual organisation in the frontal and parietal cortex (Gläscher et al. 2009).

Studies also reported that executive functions underlying the g factor could vary independently of each other, and that IQ score stability could also hide marked variations in

verbal and performance abilities, due to the structural evolution of the sensory-motor areas (Ramsden et al. 2011). Furthermore, grey matter co-varying with g factor not always belongs to those cortical regions suggested to be the seat of general intelligence (Gläscher et al. 2010).

In evaluating the utility of psychometric theories supporting a uni-component model, it is necessary to consider whether a deficit in a single cognitive function may have a neuropsychological overshadowing effect, resulting in an artificially lower IQ score; and whether a low IQ score is incorrectly assumed to be the explanation of any anomaly of neuropsychological functioning (Greenspan & Olley 2015). These potential pitfalls arise as the tools for assessing IQ were not originally developed to evaluate below average performance, but to measure a child's abilities predictive of academic achievement, and to identify children in need of additional support in school. For the same reasons such tools cannot measure IQ below 40. Hence this floor effect precludes investigation of how different severities of ID may be associated with distinct and heterogeneous forms of cognitive functioning, as well as the associations with other variables of interest, such as genetic or biological factors.

Multi-component models of intelligence

The most accredited model is the Cattell-Horn-Carroll, upon which the most recent versions of WISC and WAIS have been developed. This model was prompted by Cattell and Horn's model of fluid and crystallised intelligence and subsequently integrated with Carroll's theory of triple stratification. It postulates the existence of nine functions at a broad level, and over seventy more specific, narrow skills. Broad functions include quantitative reasoning, auditory and visual processing, processing speed, reading and writing skills, and long and short term memory (McGrew 2005).

Also Luria (1980) theorized that human cognition couldn't be explained with a unique factor, and postulated the existence of three basic units, resulting from interactions between different brain structures: attentional and arousal unit, integration and sensory input unit, and executive planning and organization unit.

Gardner (1993) questioned the validity of IQ and IQ tests as indicators of cognitive efficiency, claiming that the representation of the human being requires a combination of multiple, specific and different abilities, which he called "talents".

Another alternative to IQ was proposed by the theory of cognitive processing called "Planning, Attention, Simultaneous, and Successive - PASS" (Das *et al.*1975; Das 2000; Naglieri & Das 2002), which describes the existence of interdependent but separate functional systems and aims to develop an assessment of individual differences on the basis of cognitive

processing. This can be used clinically and in research on specific learning disabilities, intellectual disabilities, and attention disorders. On the basis of PASS theory, the Cognitive Assessment System (CAS; Naglieri & Das 1997) has been developed with the aim of assessing cognitive processes in relation to individual biological and socio-cultural characteristics. When administered to people with ID, CAS provided more diverse scores than the sole IQ (Naglieri & Das 1997).

With Goleman's theory of emotional intelligence (1996), the achievement of levels of adjustment and satisfaction in life is also relevant to persons with below average IQ, through a harmonious management of their relationship between themselves and others.

In the Minimal Cognitive Architecture model, (Anderson 1992), cognitive functions represent hierarchically organized and interconnected schemas, according to their role in executing a task or behaviour. Within this frame, individuals who are faster in cognitive processing and in gathering information are more likely to solve problems, have high IQ, and thus to be defined as more intelligent. Positive relationships have been found between IQ level and speed of information processing (Reed & Jensen 1992).

It is clear how theories on intelligence have evolved over the years, from a single factor explaining performance on tests of ability, to more differentiated intelligence structures. At the same time, new instruments for measuring IQ increasingly included scores for sub-indexes, but their utility to describe cognitive profiles of persons with ID across the range of severity of ID has remained questionable.

Specific Cognitive Functions and Executive Functions

There is little consensus on what executive functions actually mean and how they are distinguished from specific cognitive functions. In general, the literature refers to executive functions as higher-order capabilities that are called upon to select, schedule, and monitor appropriate sequences of action, and which encompass a set of more specific cognitive skills.

The International Classification of Functioning, Disability and Health (ICF: WHO 2001) classifies distinct processes of cognitive functioning in the Mental Functions chapter, within the first component of Body Functions, differentiating between global and specific mental functions. A cognitive profile should include functions such as memory, attention, perception, thought, space-time orientation, language or reasoning, and executive functions, such as planning, decision making, inhibition, regulation/correction, and action.

There are many studies reporting correlations between neuroanatomical areas and specific cognitive functions, independent of general intelligence (Johnson et al., 2008). Pascual-

Leone combined various neurophysiological and brain-imaging techniques in order to identify the invariants of functioning with respect to different areas and neural systems, demonstrating that the brain presents a modular structure, and that focal damage causes only a limited impairment of the overall intellectual functioning (Pascual-Leone et al. 1999). The pre-frontal cortex has repeatedly been found to activate during attention switching tasks, and prefrontal cortex injury, especially on the left side, causing impairment in attention switching (Aron et al. 2004; Ruge et al. 2005). Poor performance in switching tasks was also found in patients with lesions in the language cortex (Mecklinger et al.1999) or in the inferior frontal gyrus of the right hemisphere (Dreher & Berman 2002; Brass et al. 2003).

In evaluating executive functions in children with ID through the Behavioural Assessment of the Dysexecutive Syndrome (BADS-C) and the Cambridge Executive Functioning Assessment (CEFA), Willner and colleagues (2010) found scores on both tests to be only weakly related to receptive language skill, and even more weakly to IQ. Interestingly, working memory seemed to play a key role in this floor effect.

Several other studies have found significant associations between executive functions, particularly working memory, and general intelligence (Miyake et al 2000; Friedman et al 2006). However, some more precise assessments indicate that only updating working memory correlates with intelligence, whereas inhibiting responses and shifting mental sets does not; these two functions seemed to be related with IQ only via their covariance with updating (Salthouse et al. 1998; Rockstroh & Schweizer 2001; Friedman et al. 2006).Data also suggest that current measures of intelligence do not evaluate all executive functions that are considered to be essential for defining human behaviours as "intelligent" (Friedman et al 2006; Diamond 2013). Gansler and collaborators (2017*) found that assessment of intelligence based on executive functions accounts for more variability in activities of daily living, is better predicted by health status, and less predicted by educational status than the traditional IQ measures.

Using Down syndrome and Fragile-X syndrome as examples to make the point, neuro-imaging studies of persons with Down syndrome or Fragile-X syndrome have focused on specific cognitive or executive functions more than on overall intelligence. Specifically, none of the five studies of persons with Down Syndrome identified in the review (Horwitz *et al.* 1990, Chang *et al.* 1998, Schapiro *et al.* 1999, Jacola *et al.* 2011, Menghini *et al.* 2011) included any overall measure of intelligence. Of the15 studies we identified of persons with Fragile-X, 12 were not based on an overall measure of intelligence (Reiss *et al.* 1994, Guerreiro *et al.* 1998, Cornish *et al.* 2001, Kwon *et al.* 2001, Rivera *et al.* 2002, Cornish *et al.* 2004, Cornish *et al.* 2005, Hessl *et al.* 2006, Hoeft *et al.* 2007, Holsen *et al.* 2008, Hashimoto *et al.* 2011); only one

included an overall measure of intelligence (Hoeft *et al.* 2008); whilst two included less specific measures of intellectual functioning (Kates *et al.* 1997, Hallahan *et al.* 2011). This suggests that researchers have found investigation of relationships between brain structure and function with specific cognitive functions to be more informative, than that with IQ (see Table 1).

The case for using specific cognitive functions in defining ID is also supported by findings of recent research on cognitive and behavioural phenotypes. Our review revealed that different syndromes with comparable IQs were associated with very different cognitive phenotypes, with respect to both relatively intact and impaired functions (see Table 2). Comparing WISC-III scores of children with Williams-Beuren syndrome, Prader-Willi syndrome, and Fragile-X syndrome, with similar sociocultural and socioeconomic backgrounds, Pegoraro and collaborators (2013) found similar general IQ scores but significant differences concerning verbal IQ and verbal and performance subtests. Vocabulary and comprehension subtest scores were significantly higher in Williams-Beuren syndrome in comparison with Prader-Willi and Fragile X syndromes, and block design and object assembly scores were significantly higher in Prader-Willi than in Williams-Beuren and Fragile-X.

The variability between and within phenotypes is also present in autism co-occurring with ID. Of particular interest is the finding that in people with autism, low IQ scores are not necessarily associated with impairment of overall cognitive functioning, but with anomalies of information processing, which in turn have pervasive effects on the overall functioning of the individual (Scheuffgen *et al.* 2000; Anderson 2008).

As indicated, individual differences in specific cognitive functions are highly relevant in differentiating ID phenotypes, and in understanding their biological underpinnings, whereas IQ is not.

Unfortunately, to date knowledge on alterations of very specific cognitive functions in ID of different origin is limited, as well as their impact on 'higher-order' executive functioning abilities. In one of the few studies aimed at this, Scerif and collaborators (2004) compared visual attention in Fragile-X and Williams syndrome, showing that early manifestations of inhibitory deficits affect disengaging and set-shifting abilities in Fragile-X and selective attention in Williams syndrome, with different implications for impulsivity control, executive working memory, and organization of thoughts and behaviour to reach a goal (planning, self-correcting, verifying, and adapting).

In general the most frequently studied functions in ID are working memory and executive functions, and more specifically orientation response and attention switch.

Integration of cognitive and emotional processes

IQ does not include emotional skills, while the reciprocal influence between emotion and cognition has received considerable attention, with very interesting models being proposed, like Ciompi's, Plutchik's or LeDoux's ones. It has been recently suggested that an emotionally charged activating stimulus or a condition of emotional distress may affect the quality of an individual's cognitive performance (LaBar, 2010), particularly memory recall (Savtchouk & June Liu 2011).

The investigation of the interplay between emotional and cognitive processes has been fostered by the new conceptualisation of mental disorder proposed in the project named Research Domain Criteria (RDoC). The RDoC defines the relationship between behaviours and brain activities, and correlates clinical phenomena to the functioning state of neurobiological circuits (Insel *et al.* 2010). The RDoC framework proposed the "construct" as the basic unit of analysis, that summarises all data related to a specific domain of functioning, i.e. genetic, molecular, anatomical, behavioural, and symptomatological. It gives particular attention to the study of emotion, motivation and social processes for their relationship with cognitive functioning. Five constructs were defined, such as negative affectivity, positive affectivity, cognition, social behaviour and arousal /regulator system. The "cognition" construct includes all the cognitive functions indicated by the prevailing literature as the neural basis of behaviour, i.e. attention, perception, working memory, declarative memory, language, and cognitive control.

CONCLUSION

The review identified many limitations of using IQ score as an indicator of the complexity and dynamic nature of human intellectual functioning (Greenspan et al. 2015), and pointed out the need for a common and comprehensive model of intelligence.

Even though there is evidence for both the uni-component model, and multi-component models of intelligence, the latter appear to be more appropriate for explaining the high variability of cognitive functioning in ID. Indeed, experimental data indicate that the same IQ score can correspond to very different cognitive and functioning profiles.

The neuropsychological studies reviewed in this paper indicate that traditional intelligence tests have notable limitations, not capturing those intellectual functions and sub-functions which the literature showed as independent from overall IQ. The most frequently used assessment tools may provide spurious data, which are scarcely usable in the clinical practice with persons with ID. Cumulative scores refer to macro-areas of cognitive functioning and fail to

capture effectiveness of single skills. The DSM-5 indicates that "IQ test scores are approximations of conceptual functioning but may be insufficient to assess reasoning in real-life situations and mastery of practical tasks" and that consequently "a person with an IQ score above 70 may have such severe adaptive behaviour problems" . . . that their "actual functioning is comparable to that of individuals with a lower IQ score."

Full-scale IQ is a metric that many researchers and clinicians consider to be outmoded and ready to be replaced by more meaningful indicators (Flanagan & Harrison 2012, Greenspan & Woods 2014, Harris & Greenspan 2016**). The WPA-SPID and the first working group for ICD-11 proposed a diagnostic approach complementing measurement of IQ with assessment of specific cognitive functions, and a contextualised description of consequent adaptive and learning difficulties (Salvador-Carulla *et al.* 2011; Bertelli *et al.* 2014; Bertelli *et al.* 2016**). This approach was adopted also by the DSM-5; in the chapter on diagnostic features of ID (Intellectual Developmental Disorders) it states that "Individual cognitive profiles based on neuropychological testing are more useful for understanding intellectual abilities than a single IQ score. Such testing may identify areas of relative strengths and weaknesses, an assessment important for academic and vocational planning. "

Within this approach, cognitive skills should be assessed in the most comprehensive way as possible, through direct clinical examination, semi-structured observations, and tests, referring to complex executive functioning, including perceptual reasoning, processing speed, verbal comprehension, as well as to very specific cognitive functions, such as attention orientation, attention switch, visual-spatial perception, or working memory phonological loop.

The evaluation should aim to identify the neuropsychological characteristics that have the greatest impact on the person's quality of life, not only via cognitive skills but also associated behaviours, personal skills, adjustment, and autonomy. This would require professionals to be familiar with several instruments, in order to quickly select the most appropriate ones on the basis of the person's characteristics and the evaluation context, and cultural background.

This approach may favour the understanding of the link between cognitive alterations and psychopathological vulnerability across the life span, as well as bring enormous advantages to a more inclusive cultural attitude towards ID and other neurodevelopmental disabilities, providing a paradigm shift from "intellectually below average IQ" to "neuropsychological characterisation". According to this paradigm, every person would have their own neuropsychological characteristics, in terms of weaknesses and strengths, and some persons

with and without ID could even share one or more specific cognitive dysfunctions and be distinguished only by their severity and their impact on individual functioning.

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Table 1. Indicators of cognitive functioning in neuroimaging studies on Down syndrome and Fragile-X syndrome.

	Number of articles		Indicator of cognitive functioning					
	Keywords match	after article reading	unspecific		overall intelligence		specific cognitive/executive functions	
Down syndrome	151	5	0		0		5	Jacob et al., 2011 Menghini et al., 2011 Schapiro et al., 1999 Chang et al., 1998 Horwitz et al., 1990
Fragile-X syndrome	119	15	2	Hallahan et al., 2011 Kates et al., 1997	1	Hoeft et al., 2008	12	Klabunde et al., 2015 Hashimoto et al., 2011 Holsen et al., 2008 Hoeft et al., 2007 Hessl et al., 2006 Cornish et al., 2005 Cornish et al., 2004 Rivera et al., 2002 Kwon et al., 2001 Cornish et al., 2001 Guerreiro et al., 1998 Reiss et al., 1994

Table 2 - Level of Intellectual Disability and cognitive phenotypes in main genetic syndromes

Genetic Syndrome	Level of Intellectual Disability	Specific Cognitive Phenotype	
		Strenghts	Weaknesses
Down Syndrome	mainly mild and moderate (American Academy of Pediatrics Committee on Genetics, 2001)	visuospatial short-term memory, associative learning, and implicit long-term memory (Lott & Dierssen, 2010)	expressive language, syntactic/morphosyntactic processing, and verbal working memory (Silverman, 2007) receptive vocabulary, verbal short-term memory, and explicit long-term memory(Lott & Dierssen, 2010; Naess et al., 2011)
Prader-Willi Syndrome	mild and moderate (Cassidy et al., 2012)	visual processing for shape identity (ventral stream) (for deletion but not for disomy) (Woodcock et al., 2009), object assembly (only for deletion) (Copet et al., 2010)	phonological loop (particularly for deletion), emotion modulation (Walley & Donaldson 2005), attention/task switch (Woodcock et al. 2009; Woodcock et al., 2010), digit span, digit symbol coding (Copet et al., 2010)
Williams Syndrome	mild and moderate (Donnai & Karmiloff-Smith, 2000)	concrete and receptive language, vocabulary and expressive language, verbal short-term memory, grammatical abilities (Morris, 2010; Mervis & John, 2010), sustained attention (Atkinson & Braddick, 2011)	visuospatial construction (Morris, 2010), working memory, arithmetic skills, planning and inhibition (Menghini et al. 2010), relational/conceptual language (Mervis & John, 2010), selective attention (Fung et al., 2012)
X Fragile Syndrome	mainly mild and moderate (McLennan et al., 2011)	sequential processing, short-term memory, gross and fine motor skills, coordination (Loesch et al., 2003; Wilding et al., 2002; de Vries et al., 1996; Cornish et al., 2009)	verbal labelling and comprehension, visuospatial processing, writing and mathematics (Roberts et al., 2005; Freund & Reiss, 1991), disengaging attention and set-shifting, response inhibition (Cornish et al., 2001; Fung et al., 2012).
Klinefelter's Syndrome	absent or borderline (Boada et al., 2009)	language, comprehension, reading, auditory and verbal memory, attention, and motor functions (Boada et al., 2009; Bender *et al.* 2001)	visual memory, classification, cooperation, problem-solving (Boada et al., 2009; Bender et al., 2001)
Turner's Syndrome	absent or borderline (Hong et al., 2009)	visual-spatial and visual-perceptual skills, executive skills, working and non verbal memory, attention, difficulties in social cognition and emotional tasks (Hepworth & Rovet, 2000; Skuse et al., 2005; Skuse et al., 2005; Ross et al., 2002; Murphy & Mazzocco, 2008)	receptive language, memory (Ross et al., 2002)
Phenylketonuria	Moderate to severe (Blau et al., 2010)	Storage component of working memory, distractor interference and proactive interference components of inhibitory control (in early-treated PKU) (Christ et al., 2010)	executive, manipulation or monitoring components of working memory, distractor and proactive interference (Christ et al., 2010; Huijbregts et al., 2013), information processing speed, perception and visual-spatial abilities (Janzen & Nguyen, 2010), prepotent response inhibition (Huijbregts et al., 2013)