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Exploratory study on cognitive abilities and social responsiveness in children with 22q11.2 deletion syndrome (22q11DS) and children with idiopathic intellectual disability (IID)

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ABSTRACT

Background: Development of cognitive skills and social responsiveness are areas of concern in children with 22q11.2 deletion syndrome (22q11DS). It remains unclear if the cognitive and social profiles and trajectories are syndrome-specific or similar to those of children with idiopathic intellectual disabilities (IID) with or without comorbid autism spectrum disorder (ASD).
Aims and methods: In this exploratory study, we examined and compared five broad cognitive abilities (BCAs) and the social responsiveness in primary school-aged children with 22q11DS (age 6–13, $n = 21$) and IQ-matched peers with IID ($n = 21$). The relative strengths and weaknesses of both groups were re-evaluated after 19 to 30 months.
Outcomes and results: Four different cognitive trajectories (i.e. absolute progress, stability, growing into deficit, and absolute decline) were demonstrated in both groups. Most children showed combined types of trajectories across BCAs resulting in a complex changing cognitive profile. In the 22q11DS group, social responsiveness problems increased, whereas no significant change was observed in the IID group.
Conclusions and implications: Results reflect similar cognitive and social responsiveness profiles and trajectories across groups with children with 22q11DS being more at risk for growing into a social deficit. We recommend repeated monitoring of social skills development to adapt the environmental demands to the child's individual social capacities.

What this paper adds

To our knowledge, this is the first study to compare the cognitive and social responsiveness profiles and trajectories of primary school-aged children with 22q11DS to those of age- and IQ-matched children (IID comparison group). We examined (a) whether cognitive and social responsiveness profiles are syndrome-specific for 22q11DS, and (b) how these profiles develop over a two-year period.

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1. Introduction

The 22q11.2 deletion syndrome (22q11DS) is the most common deletion syndrome, affecting approximately 1 in 4000 live births (Óskarsdóttir, Vujic & Fasth, 2004). The syndrome is caused by a 1.5–3 MB heterozygous microdeletion in the long arm of chromosome 22 (Yamagishi & Srivastava, 2003). It has an autosomal dominant inheritance pattern, but 85–90% of all cases is a *de novo* deletion (Digilio et al., 2003). Cardinal features of 22q11DS are medical complications (e.g. congenital heart defects and palatal problems), developmental delay or learning disorders, and behavioural or psychiatric problems (Óskarsdóttir, Persson, Eriksson & Fasth, 2005; McDonald-McGinn et al., 2015; Swillen, Vogels, Devriendt & Fryns, 2000).

1.1. Cognitive profiles in primary school-aged children with 22q11DS

Approximately 50% of the children with 22q11DS have an IQ-score below 70 (De Smedt et al., 2007; Furniss, Biswas, Gumber, & Singh, 2011) and learning difficulties are present in 80 to 100% (Swillen, Devriendt, Ghesquière & Fryns, 2001). Studies examining the cognitive phenotype of children with 22q11DS have consistently documented wide cognitive variability (Campbell & Swillen, 2005). During primary school age, full scale IQ (FSIQ) scores can vary from 50 to 100, with a median of about 75 (De Smedt et al., 2007). A subgroup of primary school-aged children with 22q11DS shows significantly better verbal (VIQ) than visuospatial abilities (i.e., performance intelligence, PIQ) (Bearden et al., 2001; De Smedt et al., 2007; Swillen et al., 1999). Despite (severe) limitations in visuospatial skills (Bearden et al., 2001; Swillen et al., 2000; Woodin et al., 2001), processing speed (PS) has been reported to be least impaired (Campbell et al., 2009; Simon et al., 2005) on intelligence tests. Goldenberg et al. (2012) and Gur et al. (2014) demonstrated no significant difference for cognitive flexibility and non-verbal reasoning between children with 22q11DS and chronological age-matched typically developing (TD) children. While Shapiro, Tassone, Choudhary & Simon (2014) found opposite results. When measured with single-unit verbal tasks, short-term memory has been reported to be relatively intact in children with 22q11DS (Majerus, Van der Linden, Braissand & Eliez, 2007). In contrast, visuospatial memory scores are significantly lower in children with 22q11DS compared to TD children (Bearden et al., 2001; Sobin et al., 2005).

In the aforementioned studies different instruments were used for measuring subparts of the cognitive profile, thereby failing to capture the specific cognitive strengths and weaknesses characterizing primary school-aged children with 22q11DS. The Cattell-Horn-Carroll (CHC) theory, which differentiates 10 broad cognitive abilities (BCAs, see Appendix A for definitions) and 72 narrow cognitive abilities (NCAs) in a three-stratum model (see Fig. 1), can allow a more focused and detailed investigation of cognitive abilities in individuals with 22q11DS.

Currently, the overall concept of 'intelligence' is reconsidered and several broad cognitive abilities are described in the Cattell-Horn-Carroll (CHC) theory (Schneider & McGrew, 2012). This theory has become the new framework for assessing cognitive abilities in a more reliable and comprehensive way. Departing from the hypothesis of Detterman (1999) that intellectual disability originates from one or more impaired cognitive abilities with a central role influencing the function of the entire cognitive system, the CHC theory provides valuable insights when characterizing cognitive profiles of diverse groups of children with developmental disabilities. Bergeron and Floyd (2006) reported that in general children and adolescents (ages 8–18) with idiopathic intellectual disability (IID) show poor performance on fluid reasoning (Gf) and crystallized intelligence (Gc). These BCAs were found to be the best indicators for general cognitive functioning (G-factor). The authors also demonstrated that, compared to TD children, children with IID have different cognitive strengths and weaknesses. Visual processing (Gv) and auditory processing (Ga) appeared to be relative strengths for the IID group, and long term memory and retrieval (Glr) and crystallized intelligence (Gc) were relative weaknesses. They concluded that although the group means of children with IID on the seven measured BCAs were within the normative low to very low range, there was a wide range of performance across the BCAs. Not all cognitive abilities need to be impaired in one individual and impaired abilities are not consistent across individuals (Bergeron & Floyd, 2006).

The assessment of BCAs in primary school-aged children with 22q11DS will allow us (1) to look in depth into different facets of intelligence, and (2) to better interpret the complex cognitive profile based on different cognitive skills rather than on single score representations (i.e., VIQ, PIQ and FSIQ).

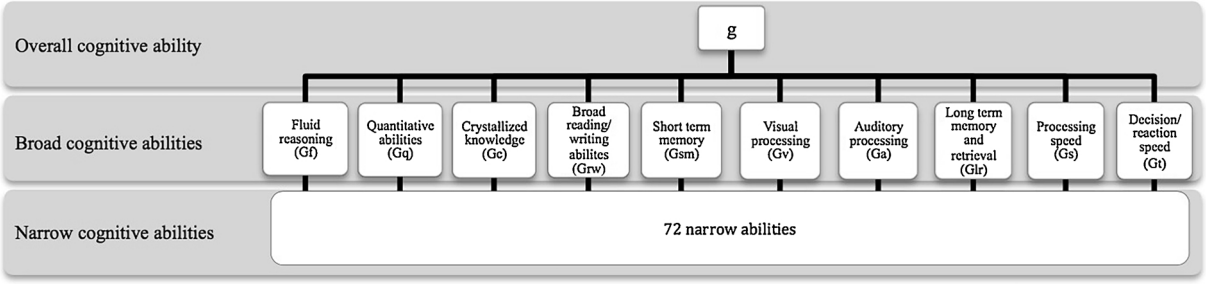

Fig. 1. Schematic overview of the three level CHC model, after Schneider and McGrew (2012).