Cognition and learning in 22q11.2 deletion syndrome (VCFS)

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The deletion 22q11.2 syndrome (22q11.2 DS) or velocardiofacial syndrome, is one of the most common genetic causes of learning disabilities and mild intellectual disability (ID). Although there are some similarities and tendencies concerning cognition and learning in persons with 22q11.2 DS, there is a considerable variability in the intellectual capacities and learning styles of persons with the condition, and this seems to change over time. It is important to keep in mind that each infant/child/adolescent/adult with 22q11.2 DS is unique, and that they may have concerns in a few or in many of the areas described.

Developmental and educational concerns are frequently reported in 22q11.2 DS. Many infants with 22q11.2 DS exhibit developmental (or early cognitive and learning) problems that present as expressive language delays, gross motor difficulties, visual-motor and visual-spatial deficits, and/or attentional difficulties during the first few years of life. These early deficits are believed to be associated with later academic and learning problems.

What do we know about the intellectual abilities and cognitive profile in 22q11.2 DS?

Intellectual abilities of children with 22q11.2 DS are generally lower than average: the mean full scale IQ is in the mid-seventies (70-75) (the mean FSIQ in the normal population is 100) with about 60% having a borderline to normal intelligence (FSIQ > 70) and about 40% having (mild) mental retardation (MR) or intellectual disability (ID) (FSIQ < 70).

This cognitive profile on IQ tests seems to change with development: cross-sectional studies in children with 22q11.2 DS indicate in an important subgroup higher verbal IQ (VIQ) than performance IQ (PIQ), whereas in adults, this VIQ > PIQ profile is less common. Also, during adolescence, there is a decline in VIQ scores in a subgroup of individuals with 22q11.2 DS.

Besides a general intellectual delay and slow maturing, many children and adolescents with the 22q11.2 DS exhibit a cognitive profile of strengths and weaknesses. Areas of relative strengths are reading (decoding), spelling, and (auditory/verbal) rote memory. Areas of relative weaknesses are reading comprehension, arithmetic, visual-spatial memory, attention and executive skills (planning, problem-solving, cognitive flexibility, monitoring).

What are the specific cognitive deficits and underlying neural circuits?

Cognitive and neuroimaging studies of children with 22q11.2 DS conducted in the last decade have provided information on the specific cognitive deficits and their possible underlying neural circuits: the frontostriatal and frontoparietal neural networks seem to be particularly affected.

Children and adolescents perform worse (than would be expected by their cognitive level) on tasks requiring shifts of attention, cognitive flexibility, and working memory

(frontal cortex and caudate nucleus) and on tasks involving visuospatial and numerical abilities (posterior parietal cortex).

What does this mean for the learning process in school?

Many children and adolescents with the 22q11.2 DS experience problems in the domain of reading comprehension, mathematics (especially with representation of magnitude, and with mathematical reasoning) and abstract reasoning.
Therefore, many of them will need:

- many learning experiences with concrete materials and experiences
- a highly structured learning environment
- a step by step approach with a lot of repetition and rehearsal
- pre-teaching (for learning new material)
- instructions on how to learn and how to memorize
- an encouraging and reinforcing learning environment with clear learning goals and frequent feedback

Depending on their overall cognitive capacities (borderline intelligence vs. intellectual disability), children and adolescents with 22q11.2 DS will follow normal school with additional learning and educational support (starting from an individualized educational plan (IEP)), or they will need special education with IEPs that are adapted to the individual needs (at a regular basis) of the child/adolescent.

Conclusion

It is clear that appropriate educational support from a very young age on is critical to children affected with this condition. Early educational intervention is strongly recommended, and should include training of both verbal (language, articulation, reading comprehension) and non-verbal areas (motor skills, visual-spatial skills and

memory, mathematical training, attention, and social skills). Educators are an important part of the team of professionals who provide services to a child with 22q11.2 DS. Recognizing areas of strengths and weaknesses may help to guide educational approaches and identify additional resources that can effectively support the learning process. Additionally, a better understanding of the challenges these children and adolescents face, will lead to appropriate intervention and proactive treatment that will help children, adolescents and adults achieve their full potential.