Educating Children with Velo-Cardio-Facial Syndrome, 22q11.2 Deletion Syndrome, and DiGeorge Syndrome

Third Edition

A Volume in the
Genetics and Communication Disorders Series

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Editor

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Preface

In 1994, my fourth-grade child was diagnosed with a genetic deletion syndrome called velo-cardio-facial syndrome (VCFS) or 22q11.2 deletion syndrome. After 10 years of speech therapy, occupational services, and various surgeries and interventions, there finally was a name that explained his difficulties. A blood test called the fluorescence in situ hybridization (FISH) test had just been developed that could positively verify the absence of a piece of DNA on chromosome 22. My husband and I had a genetic explanation and a contact in the United States, Dr. Robert Shprintzen, from New York. No one in Madison, Wisconsin, our hometown, knew much at all about the syndrome. In fact, 5 years earlier, a geneticist had looked at our child and his medical history of a heart defect, palate abnormalities, hypotonia, and learning difficulties and screened him for VCFS/22q11.2DS. He rejected that diagnosis and we spent the next 5 years searching for answers. Now we had a contact that could help us understand the nature of our child's disability.

Thus began our journey. Twenty-six years ago, very little was known about the cognitive/learning profile of children with this genetic deletion. No studies were available that focused on how these children learn or what kinds of interventions worked best. What I knew as a parent and teacher of 20 years was that my child was struggling. He learned very differently from others and had trouble remembering directions, understanding math concepts, or telling about what he had read. He was frustrated, I was stressed, and the school was perplexed. What was wrong? Why did techniques typically used for learning-disabled students fail with him? Why could he memorize with drill and practice, but have difficulty telling me what he did in school that day? How could he sit through an afternoon of school and fail to learn much of anything?

Since 1994, an enormous amount has been learned about the 22q11.2DS or velo-cardio-facial syndrome. Due to the amazing work of the Human Genome Project and scientists at Albert Einstein School of Medicine, the genes that make up the genetic deletion have been identified (Edelmann, Pandela, & Morrow, 1999). Researchers are learning how the genes are expressed and they are beginning to understand why children with the

deletion have specific difficulties. Imaging studies from several medical centers such as Upstate Medical University, Stanford University, Great Ormond Street Hospital, London, University of California's MIND Institute at Davis, and University of Geneva have uncovered abnormalities in the brains of children and adults with the syndrome. These studies are beginning to explain the reasons for the learning difficulties. Behavior studies have documented particular learning strengths and weaknesses with the 22q11.2DS population. Longitudinal studies through centers that specialize in the 22q11.2DS such as Children's Hospital of Philadelphia have highlighted trends that seem to be present and are offering direction for better long-term treatments.

The estimated prevalence of this genetic deletion in the United States and other First World nations ranges from about 1 in 2,000 persons (Robin & Shprintzen, 2005; Shprintzen, 2005a, 2005b) to 1 in 4,000 to 6,000 (Panamonta et al., 2016). This is the second most common genetic deletion syndrome after Down syndrome. Yet, many children go undiagnosed, schools and teachers remain unaware of the learning profiles of these children, and parents complain that the schools are not preparing their child for any meaningful place in society. A 2005 study conducted by the Stanford University School of Medicine surveyed 53 pediatricians and 69 teachers from Northern California as to their knowledge of physical, cognitive, and behavioral features associated with velo-cardio-facial syndrome, Fragile X syndrome (X-linked mental retardation), and Down syndrome. The study concluded that the level of awareness of the physical features of VCFS was only 21% among the teachers and that their understanding of the cognitive and behavioral aspects of the syndrome was 8%. Physicians scored only slightly better with only 32% aware of the physical characteristics, 12% knowledgeable of the cognitive profile, and 16% aware of the behavioral issues associated with VCFS/22q11.2DS (Lee et al., 2005). Clearly, with 92% of teachers surveyed unaware of the learning issues associated with this syndrome, a great deal must be done to educate the general public and medical and learning professionals. It was for this reason that I, along with several other dedicated medical practitioners, decided to write the first edition of this book. It was our attempt to blend what has been learned in the cognitive science labs with learning theory to give practical advice to all persons who are devoting their time and energy to help a child with this syndrome. Now, 13 years later, we are pleased to update the third edition with new research and current information to better assist families and professionals. Through my work of advocacy for children with 22q11.2DS, I have assisted hundreds of families navigate through the educational system and on to adult life. This intimate glimpse into the lives of these families has enriched my understanding of the syndrome and reaffirmed my desire to broaden the understanding of the learning challenges associated with it. The research community has also grown and more studies have been done in the last 5 years that will elucidate the needs of this population as they mature into adulthood. My young child with 22q11.2DS, too,

has grown into a mature 36-year-old man with a young child of his own. His journey has afforded me a continued connection to the world through the eyes of one grappling with this syndrome. His continued growth, his tenacity, and his desire to partake fully in the community are truly inspirational and proof that with hard work, interventions, and the right supports, there is the chance of a full and productive life despite the VCFS diagnosis.

The overriding purpose of this book is to educate the public and professional communities about this syndrome and improve the lives of those touched by this deletion and other complicated learning challenges. It is my hope, and the desire of the other contributing authors, that bringing cutting-edge research into the classroom will brighten the lives of the many children with the 22q11.2DS worldwide who struggle to learn and will serve as a model for educating children with other genetic syndromes. Although the 22q11.2 DS offers a unique set of characteristics, the information in this volume on teaching methods, identification, application of research, and advocacy is transferrable to a wide range of children with complex medical needs. It is my hope that the suggestions and insight offered will assist parents and school districts with crafting programs that optimize these students' potential and quality of life. Only with collaboration, experimentation, and reflection will progress occur. We are all continuing to learn.

INTRODUCTION

The Name Game: A Lesson in Confusion

(Velo-Cardio-Facial Syndrome, velocardiofacial syndrome, VCFS, 22q11.2DS, DiGeorge, Conotruncal Anomalies Face Syndrome, CATCH 22)

The syndrome discussed in this book has undergone several name changes over time that can be extremely confusing for the reader. Therefore, it is important to understand that the information presented pertains equally to any child diagnosed with a 22q11.2 deletion **regardless of the label assigned by the geneticist or physician**. The underlying genetic deletion of the 22nd chromosome (confirmed by a blood test) is the root cause of the educational challenges, and the research studies or interventions discussed are applicable to all children with **any of the diagnoses listed above**. The issue of what to call this syndrome currently is a hotly debated topic in the scientific community. The fact that one name has not been formally agreed upon is confusing for parents, educators, and the public, who may not realize that the information they are seeking is available under multiple, different names. **Therefore, the terms *VCFS*, *22q11.2DS*, and *DiGeorge* are used interchangeably in this book and any scientific research done under one name is applicable to a child who was diagnosed by a physician using a different label for the deletion.**

The first edition of this book was entitled *Educating Children with Velo-Cardio-Facial Syndrome*. I have expanded the title to include the 22q11.2DS and DiGeorge labels to help clarify this point. I hope that in the near future, scientists will agree on a universal name to call the syndrome so that dissemination of information will be less confusing to the public. In the meantime, parents, educators, and professionals can choose the label they feel most comfortable using, with the understanding that deleted genes on the 22nd chromosome are the underlying reason for the learning challenges discussed.

This book is divided into two parts. The first deals with the research on VCFS/22q11.2DS that has been done in several cognitive science labs both in the United States and abroad. Chapters have been contributed by leading VCFS/22q11.2DS specialists in the areas of speech and language, neurology, psychology, immunology, and cognition. The second section is a practical handbook designed to apply the research to the classroom setting.

Although several scientifically controlled studies have been done in neuroscience labs on VCFS/22q11.2DS and cognition, virtually no research has been completed on children with VCFS/22q11.2DS and teaching interventions. Until recently, very few students were diagnosed with VCFS/22q11.2DS, and unfortunately education programs in universities have not embraced this syndrome as a topic for research. There are no specialized schools that group these students together, and those children identified with the syndrome usually do not live in close proximity to each other. Setting up controlled learning environments and testing interventions is a future goal, but it poses many challenges. In the meantime, this book relies on case study data, personal consulting experience with over 400 families, anecdotal reports from teachers and parents, and educational practice techniques from related studies in special education.

The interventions are grouped according to age level to take into account the unique situations that occur as a child matures. There is, however, a great deal of overlap in appropriate interventions and accommodations. In planning a program for an older child, it will be helpful to read the information for earlier age levels to understand what previous remediation strategies were recommended, as well as the section on optimal classroom environments. Many of these early suggestions can also be applied to older students. Also, the book contains appendixes of possible accommodations for specific needs that can be used at any age.

TEACHER AWARENESS QUESTIONNAIRE

The following questionnaire is a self-test designed to assess your knowledge of the cognitive features associated with Down syndrome, Fragile X syndrome, and 22q11.2. It is adapted from the same questionnaire used by Stanford University School of Medicine to test phenotypic trait

awareness of neurogenetic syndromes mentioned earlier (Lee et al., 2005). See how well you do! The answers are in Appendix B.

Teacher Awareness Questionnaire (Marks of an "X" Are Correct)

Please indicate which of the following cognitive features are associated with each disorder (check all that apply):

	Down syndrome	Fragile X (male)	22q11.2
Arithmetic as a relative weakness	☐	☐	☐
Relative strength in verbal-based learning	☐	☐	☐
Ave IQ 70	☐	☐	☐
Ave IQ 60	☐	☐	☐
Ave IQ 50	☐	☐	☐
Short-term memory deficit	☐	☐	☐
Perseveration on word, thought, or task	☐	☐	☐
Sequencing deficit	☐	☐	☐
Expressive language stronger than ability to understand	☐	☐	☐

Please indicate which of the following behavioral features are associated with each disorder (mark with an "X" all that apply):

	Down syndrome	Fragile X (male)	22q11.2
Attention deficit/hyperactivity	☐	☐	☐
Hypernasal speech	☐	☐	☐
Gaze avoidance	☐	☐	☐
Depression	☐	☐	☐
Anxiety	☐	☐	☐
Relative preservation of social skills	☐	☐	☐
Schizophrenia/bipolar disorder	☐	☐	☐
Multiple autistic-like features	☐	☐	☐
General happy temperament	☐	☐	☐
Tactile defensiveness	☐	☐	☐

Please indicate which of the following physical features are associated with each disorder (mark with an "X" all that apply):

	Down syndrome	Fragile X (male)	22q11.2
Large or prominent ears	☐	☐	☐
Vision impairments	☐	☐	☐
Cleft palate	☐	☐	☐
Delayed motor development	☐	☐	☐
Upslanting eyes	☐	☐	☐
Hearing problems/deficits	☐	☐	☐

If you had difficulty with this questionnaire, you are not alone. We hope this book will delineate how 22q11.2 DS differs from both Down syndrome and Fragile X and offer insight into educational interventions that will make learning more productive for these children.

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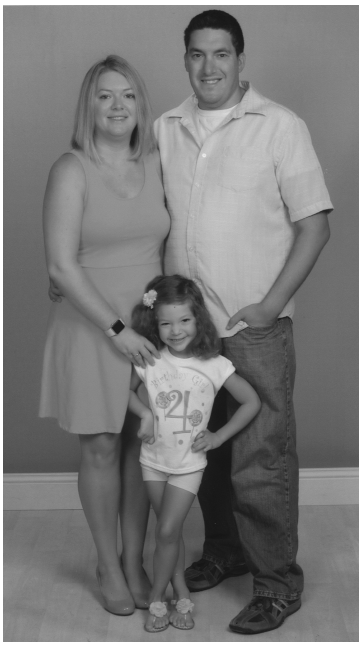
This book is dedicated to my husband, family, and, most of all, to my son, Michael, who has taught me patience, understanding, and courage.



Michael, his wife Jamie, and their daughter Breeja celebrating her fourth birthday.

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CHAPTER 2

Introduction to Education and the Neurocognitive Profile

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Virtually all children with a neurodevelopmental disability will have challenges learning as quickly or efficiently as a typically developing child. Children with 22q11.2DS (also known as velo-cardio facial syndrome or DiGeorge) often present with significant developmental delays in the speech, cognition, and motor domains. Most children with 22q11.2DS will require some type of special education services. Many will need assistance throughout their school years in the areas of academic growth, social relationships, and life skills development. This chapter explores the research studies that have been completed with children diagnosed with 22q11.2DS. From these studies, a clearer picture emerges of a typical profile of strengths and weaknesses in intellectual and achievement domains. These findings help determine whether a child with this syndrome has the ability to process information and complete academic tasks. Again, it should be emphasized that there is a wide variability within the syndrome, and every child must be carefully screened to create his or her individual profile. Nonetheless, there seem to be areas of impairment that are found in the majority of children with 22q11.2DS who have been cognitively assessed.

There have been two main avenues that have been explored to test mental functioning of individuals with 22q11.2DS: neuropsychological testing and cognitive experimentation studies. This chapter deals with the neuropsychological testing results, and the next chapter is devoted to brain imaging and cognitive experimentation studies. Neuropsychological testing is the widely accepted approach that schools use to determine if a child is in need of special education services. A battery of standardized tests that focus on the intellectual, academic, and behavior domains is administered. The tests are given by evaluators who are trained to closely follow test protocol and are skilled in interpreting the results. Standardized test scores are normed with respect to the general population at the same chronological age. From this, standard scores and percentile ranges are generated. Schools use these scores to determine if children are developing at a rate significantly below or above what would be expected for their age and grade level. Test scores can be compared both between individual students and within the child to see if any patterns of strengths or weaknesses occur. Often, this battery of tests is administered within a short time period, such as over one to two sessions. This can be advantageous in that a single testing session can generate a great deal of information. The drawback, however, is that the testing session is just a snapshot of how the child performs on a daily basis. With children who have health challenges, caution should be taken regarding extended test-taking sessions. Because these students tire easily and often have chronic health issues, prolonged testing periods may not be reliable. A more accurate assessment of ability would be obtained from testing sessions spread over several days, observations of parents and teachers, and, most important, day-to-day performance in the classroom. If accommodations during testing are necessary, it is likely that the same supports would be needed on a regular basis in the classroom. Therefore, if tests are administered in shortened sessions, that fact should be taken into account when determining eligibility for services. It would be important to get an accurate assessment of potential ability and cognitive strengths and weaknesses, but not to overestimate a child's ability to function given the demands of a typical school day.

Although neuropsychological testing can offer assistance with planning an Individual Education Plan (IEP), the results should be understood as descriptive and should not be interpreted as necessarily indicating the underlying cognitive and neurobiological impairments. The tests can indicate a student has strengths and weaknesses in particular areas, and this is crucially important for identifying which areas of a student's functioning are causing the greatest difficulties, and thus require some kind of remediation, and which are the functions the student may be able to use in order to support weaker areas of ability. Reducing the gap between a student's abilities and the demands on those abilities (both within and outside of the classroom) can be done by both enhancing those abilities where possible and/or adjusting the requirements for learning in order to establish and

maintain a balance between the two. This process can be an extensive one that will require constant adjustment. Nevertheless, successful calibration offers a real, tractable approach for optimizing learning, motivation, and self-esteem while likely reducing stress and anxiety on the part of the student. Ongoing research is aiming to explain the mental processes being used, the brain circuits activated, or the neurotransmitters involved in order to be able to create novel, highly targeted treatments in the future that will reduce the underlying causes of learning difficulties. An increasing number of cognitive experimentation studies have been and are being carried out to try to begin to answer these questions. The results of some of these experiments are discussed in Chapter 3, "Cognition and the 22q11.2DS Brain."

GENERAL COGNITIVE ABILITY

The range of neuropsychological impairments seen in 22q11.2DS is variable, but numerous studies have identified a pattern of difficulties that seems to be consistent across the 22q11.2DS population. On measures of general intelligence or IQ, children with 22q11.2DS score lower than would be predicted by their chronological age and lower also than unaffected family members such as parents and siblings. Studies have consistently measured IQ in the low-average to borderline range. Although typical IQ is considered 100 ± 15 standard deviation points, verbal IQ for children with 22q11.2DS usually ranges from 75 to 80 and is often (although not always) higher than performance (nonverbal IQ), which often falls from 70 to 75 (Moss et al., 1999). A study of 103 children from 4 to 16 years in age (De Smedt, Devriendt, et al., 2007) found the mean total IQ of the group to be 73, with scores ranging from 50 to 109. The study found no difference in IQ in children with the concurrent diagnosis of either a heart defect or attention-deficit disorder (ADD). The study did find, however, that children who had a diagnosis of autism as well as 22q11.2DS had significantly lower IQ scores. This study also looked at whether there was a difference in IQ levels for children born to parents who did not have the 22q11.2DS deletion (de novo) compared to children born to a parent who also had 22q11.2DS (familial). In this study, 93 children were de novo compared to 10 who were born to an affected parent. The average IQ for the de novo group was 74 compared to 63 for the familial group. Although there is likely to be some genetic component to the measured intellectual abilities, it is much harder for children to excel intellectually and academically in a home where at least one parent's intellectual capacities are also impaired. A more recent study also confirmed the association of lower FSIQ scores in familial versus de novo deletion and further studied whether the sex of the parent impacted the scores. The results indicated that if the

deletion is inherited from the mother, there was a poorer cognitive outcome (McGinn et al., 2018).

Several studies have suggested that children with 22q11.2DS have more developed verbal than nonverbal abilities. In one study of 33 children with 22q11.2DS, Moss and colleagues (1999) found that full-scale IQ was 71.2 ± 12.8 (mean ± standard deviation), verbal IQ was 77.5 ± 12.8 (mean ± standard deviation), and performance IQ was 69.1 ± 12.0. This pattern of performance IQ being significantly lower than verbal IQ, indicative of a profile resembling that of a nonverbal learning disability, seems to be true for many 22q11.2DS children, but not all (Campbell & Swillen, 2005; Moss et al., 1999; Wang, Woodin, Kreps-Falk, & Moss, 2000). Another study of 103 children with 22q11.2DS found the average verbal IQ to be 78 compared to a 72 performance IQ. In addition, three out of four children tested had a verbal IQ higher than their performance IQ and 22% of these children had discrepancies over 15 IQ points (Swillen, 2006). This finding may, however, be a function of the age of the child tested. In a study of 172 individuals ages 5 to 54 years, the verbal IQ scores were negatively correlated with age. By adolescence and in older adults, the difference in scores between mean performance in verbal and performance measures diminished (Green et al., 2009).

Consistently, however, neuropsychological or psychometric test results show general intelligence is lower than average, with most IQ scores in the 70 to 85 range. Thus, most children with 22q11.2DS will have difficulties across both performance and verbal domains in comparison to typically developing peers. Nevertheless, it appears that such IQ scores are not predictive of real-world Adaptive Functioning abilities. As Angkustsiri et al. (2012) state, Adaptive Function is "a separate, but related, construct to IQ [providing] more ecologically valid metrics of development that reflect age-appropriate expectations of one's ability to independently function and communicate in practical and social environments, such as at home, school, in the community, or on the job." Their study of 7- to 14-year-old children with 22q11.2DS found that "the often-observed relationship between IQ and adaptive functioning is not observed in children with 22q11.2DS, perhaps because of their significant anxiety symptoms," and they suggest that reducing anxiety, which can partly be done by optimizing balance between abilities and demands, as described above, is likely to have a positive effect on Adaptive Functioning.

There are also preliminary indications that there may be a difference in cognitive functioning between boys and girls. A study (Niklasson, Rasmussen, Oskarsdottir, & Gillberg, 2006) of 100 children with 22q11.2DS showed girls outscoring boys with average IQ scores of 74 compared to 65 for the boys. A 2006 study by Swillen, however, found no difference between IQs of males versus females (Swillen, 2006). Another large study of 90 children (50 boys and 40 girls) found that boys with 22q11.2DS were more cognitively impaired than girls (Antshel, AbdulSabur, Roizen,

Fremont, & Kates, 2005). Their average IQ scores on the Wechsler Intelligence Scale for Children (WISC III) were 68.9 ± 12.8 for the boys and 76.3 ± 11.7 for the girls. This study also found that boys with 22q11.2DS scored significantly lower than girls with 22q11.2DS on measures of communication, daily living skills, and socialization on the Vineland Adaptive Behavior Scale. In addition, boys also scored significantly lower than girls on the Wechsler Individual Achievement Test–Second Edition (WIAT-II) in the areas of reading, math, written language, and oral language. In addition, this study noted a negative association between age and cognitive functioning with girls and 22q11.2DS, in that their scores did not keep up with the expected improvement with age, but this was not the case with boys. It should be noted that although girls in this study did better than boys, their scores were still in the low-average range and at levels that would necessitate special education intervention. A longitudinal follow-up of 70 of these same 90 children with 22q11.2DS three years later found that females' cognitive scores across multiple psychological measures declined more their male counterparts. Accordingly, as the females in this study moved into adolescence, there were no longer significant differences in cognitive functioning compared to the same boys in the study. This suggests that the age of participants with 22q11.2DS may impact whether sex differences in the area of cognition are found in a particular study. This may account for why studies of youth with 22q11.2DS have been inconsistent in the extent to which sex differences in cognition are observed and reported (Antshel et al., 2010).

COGNITIVE DECLINE OVER TIME

There is some evidence to suggest a drop in IQ scores from the preschool level of the mid-80s to the mid-70s in elementary school years (Golding-Kushner, Weller, & Shprintzen, 1985; Shprintzen, 2000). The former scores were obtained using the Leiter and Stanford-Binet tests and the second set of scores used the Wechsler. Golding-Kushner et al. (1985) suggested that this reduction could be due to the nature of the tests administered rather than a drop in global intelligence. The Wechsler test for older children involves more abstract reasoning and higher-order thinking skills, an area of relative weakness for the 22q11.2DS population. A 2001 study of 112 children with 22q11.2DS under the age of 6 used the Wechsler Preschool and Primary Scale of Intelligence (WPPSI-R) and the Bayley Scale to test for IQ (Gerdes, Solot, Wang, McDonald-McGinn, & Zackai, 2001). This study found 34% of the preschool children tested in the average range with IQ scores in the average range (FSIQ > 85), 32% in the mildly delayed range (FSIQ 70–84), and 33% in the significantly delayed range (FSIQ < 70). A small longitudinal study of 24 children found a drop in verbal IQ as

children mature with declines in the areas of similarities, vocabulary, and comprehension. This study also reported a decline in expressive language abilities (Gothelf et al., 2005). A larger longitudinal study involving 70 youth with 22q11.2DS indicated that although there were declines in cognition over time, not all cognitive functions were equally affected. Those areas impacted were the Wechsler Full Scale IQ, Processing Speed, and Freedom from Distractibility indices. Additionally, scores on the California Verbal Learning Test and math academic skills also significantly decreased. In contrast, improvement was seen in the areas of perseverative errors (i.e., ability to shift attention), planning, and reading ability (Antshel et al., 2010). Another study of 172 individuals with 22q11.2DS also found cognitive scores on IQ measures inversely associated with age (Green et al., 2009). It is important to note here that standardized measures are age adjusted to norms and it is those scores and not the raw, unadjusted scores that are reported and analyzed. This means that a person's scores can, and frequently do, rise between Time 1 and Time 2 in such studies, indicating that their intellectual development is advancing. The reduction in IQ scores means that they are simply not advancing as fast as those in the typical population do and so the age-adjusted score is lower. Therefore, reduced scores on these tests rarely indicate that individuals are *losing* skills or competence rather than indicate that they are not gaining competence as fast as unaffected agemates typically do.

More recent studies have focused on whether a drop in IQ scores in this population is a precursor to psychiatric difficulties. In a collaborative study of more than 100 scientists, 829 patients ages 8 to 24 were assessed for cognitive development. This study found three cognitive trajectories: a relatively stable IQ over time, a modest drop of IQ points, and a steeper decline for a subset of students (in verbal IQ) that preceded psychotic illness. On average for the group, there was a cognitive decline of 7 FSIQ points or 9 VIQ points longitudinally (Vorstman et al., 2015). In 2018, the International Brain and Behavioral Consortium on 22q11.2DS expanded the sample reported on by Vorstman and colleagues and compiled cross-sectional with longitudinal IQ data from 1,871 individuals to construct a normative chart for this population for Full Scale, Verbal, and Performance IQ. These data also confirmed a decline for this population in all three measures with the steepest drop observed in the youngest (ages 6–12) and oldest (35+) age ranges. In addition, individuals who went on to develop significant mental health issues did show a negative deviation from their expected trajectory for verbal IQ (Fiksinski, Breetvelt, Bassett, & Vorstman, 2018). This finding puts educators, who often do repeat testing for special education students, in a unique position to monitor a change in cognitive capacity over time and inform families when a significant drop in verbal IQ occurs. Armed with this knowledge, families can be more vigilant and proactive in monitoring for a potential downturn in mental health status.

Although more scientific research regarding IQ decline over time is needed to truly understand this aspect of the syndrome, experience gained

in practice and through research demonstrates that, over time, most students with 22q11.2DS have educational success that is noticeably different from unaffected peers, and the gap in functioning widens with time. In early elementary school, many students with 22q11.2DS test similar to their unaffected peers and, with support, function in general education classes (Swillen & McGinn, 2016). However, as they age, the vast majority of affected students are unable to keep pace without increasing dependence on special education staff and parents. Their ability plateaus around a fourth-grade level for generalization of mathematical concepts and reading comprehension skills. Some are able to *participate* in high school courses with a great deal of support (and to memorize content for tests), but these students rarely can generalize learned skills. While many are able to engage somewhat using better-developed verbal skills, lack of progress in cognitive ability impacts their proficiency with life skills that are imperative to independence outside of school. For example, in math, most cannot count change after making a purchase, problem solve, understand a paycheck, budget, or balance a checking account. High school teachers wrongly assume students with 22q11.2DS have mastered these skills. In reality, when they reach age 18 and qualify for graduation, most students are not ready for independent adult life or postsecondary training. Many schools, however, are eager to reward their effort, graduate them, and send them unprepared into the adult world. This contributes to a great deal of stress on the student who is ill-prepared for success at the college level and for families who are faced with dealing with their young adult at home with no support.

MATHEMATICS

Academic impairments are very common in 22q11.2DS and are most pronounced in math. This can be due to a combination of difficulty with visuospatial tasks, working memory impairments, and weaknesses in problem-solving abilities. An early study using the Wide Range Achievement Test (WRAT) demonstrated that math scores among 6- to 11-year-olds with 22q11.2DS ranged from 81 to 90 (population mean = 100, SD = 10) and from 74 to 86 among affected adolescents (Golding-Kushner et al., 1985). Many more recent studies have found results consistent with this initial report (Chow, Watson, et al., 2006; De Smedt et al., 2009). The weaknesses in math seem particularly pronounced in the areas of abstract reasoning, converting language into mathematical expressions, telling time, using money, and problem solving (Kok & Solman, 1995). Another study of 33 individuals with 22q11.2DS found lower composite math achievement scores in comparison to scores of reading and spelling (Moss et al., 1999). A 2006 study looked at 27 children with 22q11.2DS aged 6 to 12 and found 19 out of 25 performed at an abnormally low level on at least one

of the math variables tested (De Smedt et al., 2006). The children in this study could read numbers accurately and could retrieve number facts but had difficulty with such things as understanding number magnitude, identifying and ignoring irrelevant information in story problems, and with accuracy on multiplication with more than single-digit numbers. The older students in this study also worked more slowly than age-matched controls, which may have educational implications. An additional study of 36 children with the deletion sought to shed light on the underpinnings of the arithmetic impairment typically seen in the 22q11.2DS population. Results from this research indicated that the math impairment stemmed from deficits in short-term visuospatial memory skills. Two-thirds of the children tested scored higher in number recall than in the spatial memory test on the Kaufman battery. The mean score for the spatial memory test was 7 with 10 as considered average. Furthermore, the data supported the hypothesis that visuospatial skills are particularly critical for developing early numeracy skills and early math competence. This may account for the lingering number sense and problem-solving deficits despite the development of calculation competence with enough drill and practice (Wang et al., 2000). Another study further examined whether the math impairment in 22q11.2DS is influenced by visuospatial deficits and did find that those with 22q11.2DS had poorer numerical acuity in numerical comparison tasks that have high visuospatial demands, had lower skills in processing length visually but not in auditory tasks, and had a reduced ability to quickly extract numerosities in the subitizing range (Attout, Noel, Vossius, & Roussell, 2017). Impairments in subitizing (the rapid, accurate judgment of quantity without counting) was also found in children with 22q11.2DS in a study that examined nonsymbolic numerical processing while comparing a collection of dots (Oliveira et al., 2014).

The math impairment in the 22q11.2DS deletion is an extremely common aspect of the syndrome and one that persists into adulthood. Despite intensive interventions, many adults with the syndrome continue to have difficulties handling money, understanding math concepts, and becoming fiscally independent. Generalization of learned math strategies into real-world applications is particularly challenging and impacts the young adult's transition into the community. Most continue to need ongoing supervision and support in this aspect of independent life. A more comprehensive discussion on the math impairments associated with 22q11.2DS is found in Chapter 3.

READING

In a study of 50 children with 22q11.2DS aged 6 to 17, reading, decoding, and phonological abilities were found to be stronger than comprehension skills (Woodin et al., 2001). Many children with 22q11.2DS reportedly do