

An Educator's Guide to Understanding VCFS



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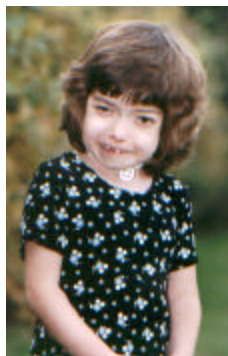
Velocardiofacial syndrome (VCFS) is one of the most common genetic causes of learning disabilities and mild mental retardation known, affecting approximately 1 in 2,000-4,000 people. If a family gave you this booklet, you may already be caring for a child with this syndrome. If not, it is likely that at some point in your career you will work with a child with VCFS. Since a diagnosis of the syndrome is not always apparent, recognized, or revealed, you may not be aware that a child has VCFS.

VCFS is also commonly referred to as 22q11 deletion syndrome, DiGeorge syndrome, or Shprintzen syndrome. Children with VCFS often have a characteristic combination of medical, developmental, cognitive, and behavioral concerns. The potential number of concerns associated with this disorder can be overwhelming to families and providers alike. However, it is important to keep in mind that each child is unique and may have concerns in a few or many of the areas described. Your experience working with one child with VCFS may be very different from your experience with another child with the same syndrome.

Research about VCFS has clearly shown that appropriate educational support is critical to children affected with this condition. Educators are an important part of the team of professionals and family members who provide services to a child with VCFS. The intent of this booklet is to alert educators to possible implications of VCFS so that educators can:

- recommend and implement appropriate educational strategies & special services
- help other educators understand the challenges faced by these individuals
- have further conversations with family members and other professionals
- help the student achieve his/her potential.

When and how a family discusses the VCFS diagnosis with their child(ren) and the school is extremely personal and may differ greatly among families. Therefore, it is important to consult with the family to establish what specific information the student has regarding his/her condition. It is also essential to identify what is appropriate to share with other educators and/or classmates and what should be included/excluded from school records.



What is VCFS?

Every cell in the body has a set of chromosomes that we inherit from our parents. The chromosomes carry genes, which are the instructions for how the body develops and functions. We typically inherit 23 chromosomes from each parent for a total of 46. In a person with VCFS, a small region of one of the two copies of chromosome 22 is deleted (missing). To date, more than 40 genes have been identified in this deleted region. It is unknown which gene or genes are responsible for the characteristics observed in people with VCFS and it is now thought that genes outside of the deleted region also contribute to the syndrome. This region of chromosome 22 is considered a “hot spot” that is more susceptible to genetic changes.

Before a genetic test (florescence in situ hybridization; called FISH) was developed, clinicians made the diagnosis of VCFS based on the constellation of symptoms present. The genetic test revealed that multiple syndromes (VCFS, DiGeorge syndrome, and some individuals with several other syndromes) were usually the result of the same small deletion on chromosome 22 (22q11.2).

VCFS occurs equally in males and females and in all ethnic groups.

Common features of VCFS:

As mentioned previously, individuals with VCFS often have a characteristic combination of medical, developmental, cognitive, and behavioral concerns.

Some of the more commonly recognized features seen in people with VCFS are:

- **Heart defects**
- **Palatal abnormalities** (area separating the mouth from the nasal cavity)
- Subtle **facial features** (smaller appearing eyes, petite mouth, mild variation in ear shape)
- **Learning disabilities** (especially speech and language problems)
- **Social and emotional** challenges
- Compromised **immune system**

The syndrome is extremely variable with more than 180 possible signs and symptoms described in the literature. Some people with VCFS have numerous medical concerns, while others may have very few. However, it is important to note that although it may not be readily apparent, some degree of learning disability or difficulty is found in almost all children (98%) with VCFS.

Development in children with VCFS

In addition to an appreciation of the unique personal attributes of each child, it is often helpful for an educator to be aware of the possible medical and/or developmental challenges a child may experience. Although the specific experiences of each child with VCFS will differ, some of the more common medical, developmental, and behavioral issues are addressed in this section. Cognitive issues and learning styles of children with VCFS are outlined in the next section.

Infants

Medical concerns:

Some children with VCFS are very ill as infants, while others have no apparent health issues. Infants diagnosed with VCFS may have heart malformations requiring surgery, kidney problems, palate abnormalities, low calcium levels, and/or immune system problems. Feeding problems are common and may include nasal regurgitation, choking, gagging, and low oral muscle tone. In addition, there may be abnormal movement in the back of the throat and/or palatal abnormalities. Recurrent ear infections and constipation are common.

Developmental concerns:

Slow growth and weight gain (failure to thrive) and/or low muscle tone (hypotonia) are common in infants with VCFS. Babies with VCFS are often late in reaching motor milestones such as crawling and walking. Speech and language delay is present in almost all children with VCFS. Often, babies have relative weakness in speech sound and speak their first words later than average. Children may be eligible for early intervention services that can include speech, occupational, physical, and family therapy.



Toddlers

Medical concerns:

The majority of toddlers diagnosed with VCFS experience feeding problems. Recurrent ear infections may persist. Hypernasal speech (too much air escaping through the nose) and velopharyngeal insufficiency or incompetence (VPI) are two common features of VCFS. VPI is caused by the failure of the velopharyngeal valve (the region of the pharynx, or throat, that opens and closes during swallowing and speech) to completely close during speech. Some children have a cleft palate (opening in the roof of the mouth) that may require surgical correction. VPI and clefting are associated with speech delays including problems with articulation and production of speech sounds. Many toddlers benefit from speech therapy designed specifically for their needs.

Developmental concerns:

Toddlers with VCFS usually show mild to moderate delays in all areas of development: language, gross motor (walking, jumping, and running), fine motor (manipulation with fingers and hands), and general cognition (thinking, imagining, and learning). The use of

short phrases and sentences is often delayed until 2 or 3 years of age, even in children who do not have a palatal abnormality or VPI. Toddlers may show significant speech and expressive language delays, especially when compared to their receptive language abilities and overall development.

Behavioral concerns:

Children with VCFS often have less mobility in their faces and may show fewer of their emotions through facial expressions. Therefore, it may be more difficult for peers and adults to read and respond to the child's social and emotional cues. These cues can be easily misinterpreted as disinterest and/or unresponsiveness. At this age, many children begin to experience separation anxiety and/or shyness beyond age-appropriate measures. Delayed expressive speech may cause additional frustration in a child struggling to communicate his or her needs.

Preschoolers

Medical concerns:

By preschool age, the majority of significant medical concerns particular to VCFS have usually presented themselves. At 4-5 years old, some children have surgical correction of VPI. Some children have problems with their kidneys and/or urinary tract that can lead to urinary tract infections, bedwetting, and frequent urination.



Developmental concerns:

Developmental delays usually continue in the preschool years, although some differences are less visible in children of this age when compared to their peers. Delays often become more apparent as children get older. Whether developmentally delayed or not, children with VCFS continue to progress and attain skills, though at their individual pace. Low muscle tone may continue to affect gross and fine motor skills. Speech and language skills are particularly challenging for many preschool children with VCFS. Delays in sound making skills, hearing impairment, attention deficits, and cognitive delays can all contribute to a delay in

language development.

Behavioral concerns:

Preschool children with VCFS may be more socially isolated than peers and engage in less cooperative or interactive play with children their age. Some children tend to be shy, fearful, and/or impulsive.

Early school-age

Medical concerns:

With the exception of severe and uncorrectable heart problems, many school-aged children with VCFS do not display signs of major health problems. A small proportion have voice disorders, motor speech disorders, and/or are hearing impaired in one or both ears. Some children have an unusual quality to their voice due to hoarseness,

hypernasality, and/or poor articulation. VPI can be a continuing issue (see: Toddlers-medical concerns).

Developmental concerns:

Usually, children with VCFS are able to speak by the time they enter school and often speech development improves during the school-age years. However, many children continue to have difficulty with receptive and expressive language skills. A typical profile of learning strengths and areas of concern is often seen in school-age children with VCFS (see next section). It is common for children with VCFS to have problems understanding social rules and cues because they tend to have difficulty deciphering non-verbal communication. Since 65% of human communication is non-verbal (such as facial expressions and body language), a child who has trouble understanding these cues can be at a disadvantage in social situations. In this case, social skills and non-verbal cues need to be taught directly. Low muscle tone may affect a child's stamina during the school day as well as his/her writing ability.

Behavioral concerns:

Some children with VCFS are diagnosed with ADD or ADHD; others may have more complex concerns such as PDD or similar diagnoses. Often, children with VCFS are more withdrawn and shy than their peers. An impaired ability to communicate with others can, at least in part, contribute to poorer social interaction skills and withdrawal.

Late childhood & adolescence

Medical concerns:

Although few adolescents have major medical concerns, many have a complex medical history that may have affected learning and social development.

Developmental concerns:

Transitions from class to class and subject to subject can be particularly challenging. Attention problems are common. Many children with VCFS struggle with social relationships. Older children are often keenly aware of their cognitive differences and that they don't fit in with their peers. This realization can be painful and lead to further social isolation or depression. Some children have unrealistic ideas and expectations, extremely low self-esteem, and/or mood fluctuations. Continuing issues with voice quality, smaller size, stamina, and athletic skills can also contribute to social rejection and isolation.



Behavioral concerns:

In the adolescent years, and sometimes earlier, children with VCFS may be at risk for developing mental health problems including anxiety, depression, phobias, obsessive-compulsive disorder, and schizophrenia. It is thought that hormonal changes of puberty, increasing social demands, and possibly a distinct genetic predisposition may all play a role in the development of mental

health problems. Although researchers are still trying to understand possible connections between VCFS and an increased risk of mental illness, it is important to be alert for behavior and emotional changes in children and teens. This is especially important for an educator to keep in mind because you may be in the best position to first recognize thought or mood disorders.

Children with VCFS face specific challenges that significantly influence their school experience. While there is little published data regarding what the future typically holds for children with VCFS as they mature, it is our hope that developing a better understanding of these challenges will lead to appropriate intervention and proactive treatment that will help children with VCFS achieve their full potential.

Cognition and Learning styles

Children with VCFS often have a unique profile of learning strengths and challenges. They typically follow the same sequence of learning as others, but often reach the same points later than average. Some children with VCFS may perform well in preschool, kindergarten, and first grade, but begin to show evidence of learning problems in second grade and up. This is usually the time when learning involves less rote memorization and is based on more complex concepts, comprehension, and critical thinking.

Recognizing areas of relative strength and weakness may help to guide educational approaches and identify additional resources that can effectively support the learning process. Early Intervention evaluations should include assessment of sensory acuity, auditory and visual perception, motor development, concept development, language skills, and behavior, as many children with VCFS have difficulties in some or all of these areas. Appropriate learning supports throughout a child's educational career might include speech and language therapy, physical therapy, occupational therapy, education specialists, tutors/specialists in math and reading, and psychological support. A neuropsychological evaluation can provide valuable information about a student's individual learning style.

Common areas of relative strength:

- Simple focused attention & initial auditory attention
- Verbal comprehension, word reading & decoding
- Auditory perception & memory
- Rote verbal learning & memory
- Arithmetic
- Willingness to learn

Relative to their overall intellectual functioning and nonverbal skills, many children with VCFS show a strong ability to learn and retain repetitive verbal information. Their performance often improves when they receive frequent feedback and are asked to focus on tasks that are brief and highly structured. Their verbal strengths often include memory

of factual information, knowledge of words, and an understanding of practical/concrete situations.

Common areas of concern:

- Language & phonological processing
- Visual-spatial skills & memory
- Complex verbal memory
- Working & encoding memory
- Reading comprehension
- Non-verbal processing
- Abstract reasoning
- Fine & gross motor skills
- Executive & adaptive functioning
- Social & emotional functioning

Children with VCFS often experience language-based difficulties, such as weaknesses in comprehension of instructions, ability to access and retrieve language, receptive language, and oral expression. Children with poor visual-spatial skills may have trouble with forming images in their mind, recalling images, spatial perception, and/or spatial relationships. Some children may not be able to recall complex information without learned cues, repetition, and reinforcement to help them. Many children have difficulty figuring out and reasoning nonverbal information, which can lead to problems in social and pragmatic communication. Abstract reasoning can be difficult and children may be more successful with concrete materials and experiences. Students with VCFS may have trouble putting ideas together and making inferences, which can make school assignments in the higher grades particularly challenging. Children may struggle with impulsivity, distractibility, and/or an inability to "shift gears" easily.

Educational considerations and strategies:

The following insights and suggestions¹ can serve as a starting point for an individualized educational approach.

Students with VCFS usually have more success with:

- A direct approach to learning rather than a discovery-based approach.
- Rote memorization, repetition, and concrete examples.
- Instruction on how to learn, including memory techniques and test taking skills.
- Small group instruction or assistance within a large group regular classroom.
- Several small tasks rather than multiple directions or large assignments.
- A structured environment with clear rules to follow.
- Additional opportunities for participation in individual and group physical activities.

Additional strategies:

- Drill and practice activities.

¹ Adapted from presentations by Donna Landsman, M.S. Ms. Landsman is both an educator and parent of a child with VCFS.

- Computer assisted learning and keyboarding.
- Reading aloud and books on tape.
- Additional handouts or notes to assist with difficulties with note taking and large group presentations.
- Study packets prior to a test.

Since educational strategies and interventions targeted at children with VCFS have not yet been thoroughly explored or evaluated, educators need to be creative and flexible with their approach as they discover what works, and what doesn't, for a particular student. Communication between the team of educators, family members, and other service providers, along with continuous assessment of the student's progress, can promote an optimal learning environment.

Suggestions for further reading

Researchers, parents, and educators have worked to increase our collective knowledge of VCFS and to create literature that can assist with the medical management, educational success, and family support of children with VCFS. While the child and his/her family are the most valuable resource available to help an educator understand the particular challenges being faced by a student with VCFS, you may also want to take advantage of the literature and other resources on the subject.

Websites:

An interactive guide to **learning disabilities** for parents, teachers, and children at <http://ldonline.org>

The **VCFS Educational Foundation** website at <http://www.vcfsef.org> (See the VCFSEF newsletter for a three part series on speech and language problems and Donna Landsman's articles on schools and education issues.)

The **22q11 Group** website at <http://www.vcfs.net> (Maintained by an organization of parents in the UK.)

Books and Articles:

Dykens, E., R. Hodapp, B. Finucane. (2000) Genetics and Mental Retardation Syndromes: A New Look at Behavior and Interventions. Paul H Brookes Publishing Co. (Discusses behavioral characteristics and genetics of nine syndromes including VCFS.)

Golding-Kushner, K. (2001) Therapy Techniques for Cleft Palate Speech and Related Disorders. Singular Publishing Group, Inc. (Chapter 10: Velocardiofacial syndrome and other special groups.)

McDonald-McGinn, D., B. Finucane, and E. Zackai. (2000) Faces of Sunshine, the 22q11.2 Deletion Syndrome: A Handbook for Parents & Professionals. The Children's Hospital of Philadelphia.

Shprintzen, R. (2000) "Velo-cardio-facial syndrome: a distinctive behavioral phenotype." *Mental Retardation and Developmental Disability Research Reviews* 6:142-147.

Solot, C., et al. (2000) "Communication disorders in the 22q11.2 microdeletion syndrome." *The Journal of Communication Disorders* 33: 187-204.

Swillen, A., A. Vogels, K. Devriendt, and J. Fryns. (2000) "Chromosome 22q11 deletion syndrome: update and review of the clinical features, cognitive-behavioral spectrum, and psychiatric complications." *The American Journal of Medical Genetics* 97:128-135.

Wang, P., et al. (2000) "Research on behavioral phenotypes: velocardiofacial syndrome (deletion 22q11.2)." *Developmental Medicine & Child Neurology* 42:422-427.