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Assisting Children with Velocardiofacial Syndrome Who Have Developmental Disabilities and Delays Associated with Speech, Communication, and Education

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ASSISTING CHILDREN WITH VELOCARDIOFACIAL SYNDROME WHO HAVE DEVELOPMENTAL DISABILITIES AND DELAYS ASSOCIATED WITH SPEECH, COMMUNICATION, AND EDUCATION

by

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ABSTRACT

Assisting Children with Velocardiofacial Syndrome Who Have Developmental Disabilities and Delays Associated with Speech, Communication, and Education

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Children with velocardiofacial syndrome (VCFS) have a variety of complex needs. Research shows that VCFS is characterized by a combination of medical problems, developmental delays, and learning disabilities, which vary from child to child. This syndrome also puts adolescents at a higher risk for developing psychiatric and psychotic disorders. The complexity of symptoms that can arise from VCFS can influence the ability of these children to communicate, socialize, and learn effectively. This literature review aims to discuss literature for caregivers, educators, and physicians to aid children effectively and understand their challenges relating to speech, communication, and education. This topic is important to me because my younger brother was diagnosed with VCFS at birth. After understanding the background of the syndrome from the formation of the palate, speech production, communication, and emergence of psychiatric disorders, it is much easier to make the necessary adjustments to assist these children to learn more effectively.

KEYWORDS: velocardiofacial syndrome, education, communication, palate, speech, children, psychiatric disorder, psychotic disorder

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CHAPTER ONE

Introduction

This thesis is a literature review; its purpose is to provide parents, teachers, and physicians with resources and information about how to best assist a child with velocardiofacial syndrome (VCFS) and other related 22q11.2 deletions. Many children may have difficulties with their speech, communication, and education due to both physical and cognitive differences as a result of the deletion. I will begin by giving a brief history and overview of VCFS. The rest of the paper will focus on signs and symptoms of this syndrome, which directly impede the child's ability to speak, communicate, and learn with the same ease other people may take for granted. This syndrome has constructed a different reality for my family as my sibling was diagnosed shortly after birth. It has made us realize the importance of dedication, advocacy, and perspective. My family has been dedicated to making sure all my brother's physical, emotional, and educational demands are being met. We have consistently been an advocate for him by spreading awareness of VCFS to his educators. Lastly, the obstacles associated with his syndrome have given us a different perspective on the ease of living that can be taken for granted.

I will cover anatomical features of the palate and how these differences contribute to difficulties in speech production. Palatal deformities sometimes require surgery or a prosthetic to correct, without correction, the child will experience many challenges during feeding and speech production. Many children with VCFS experience delays in

language and speech production, so early therapy intervention is essential to teach and promote development. I will explain how speech delay influences future communication and interaction with peers. Next, I will describe how communication and developmental difficulties can affect a child's psychosocial and educational well-being. Throughout the paper, as well as a portion at the end, I will discuss approaches for caregivers and educators that will be helpful while raising and teaching children diagnosed with VCFS.

A syndrome is a group of symptoms that consistently occur together or a condition characterized by a set of associated symptoms ("Syndrome," n.d.). VCFS has also been referred to as or in conjunction with Shprintzen syndrome, DiGeorge syndrome, Opitz G/BBB, or conotruncal anomaly face syndrome (CTAFS). The 22q11.2 deletion is thought to be the underlying cause of the medical problems associated with most of the patients with these syndromes (McDonald-McGinn et al., 2000). The diagnostic name given to the patient is often determined by the subspecialist who the patient visited. The book *Faces of Sunshine: The 22q11.2 Deletion* draws a parallel between the discovery of 22q11.2 and an old saying:

> The 22q11.2 deletion story is similar to the adage of a group of blind men trying to identify an elephant by each examining a separate part. Each man was accurate in describing his area of interest, but none was able to see the big picture. So it is that several conditions once thought to be separate are now known to be the 22q11.2 deletion (McDonald-McGinn et al., 2000, p. 9).

For the sake of simplicity, I will be focusing on VCFS, which has a much broader spectrum of symptoms. Reilly and Stedman (2013) described the name velocardiofacial comes from "velum" referring to the palate, "cardia" referring to the heart and "facies" referring to the face. VCFS is a genetic condition that is sometimes hereditary. According to Umlauf (2008) neither parent has the deletion in about 93% of cases.

Lowinger et al. (1999) states that once the deletion is present in a person, there is a 50% chance to pass on the deletion. Since VCFS can be inherited, physicians suggest that parents of children with the deletion should be tested for the deletion. Some people are very mildly affected and therefore, may not have been diagnosed previously.

VCFS is suspected based on clinical examination and the presence of signs and symptoms of the syndrome. Shprintzen (2008) described a fluorescence in situ hybridization, or FISH, a blood test is conducted to look for the deletion, see Fig. 1 below, this diagnostic procedure is highly reliable. More than 95% of individuals with VCFS have a deletion in chromosome 22q11.2. Those that do not have the 22q11.2 deletion by standard FISH testing may have a smaller deletion that can be found using a more sophisticated lab study (Basset & Chow, 2008). VCFS affects about 1:2000 to 1:7000; however, because VCFS is a developmental disorder with clinical findings that may not be evident until later in life, it is probable to assume a prevalence closer to 1:2000 for surviving newborns (Shprintzen, 2008).

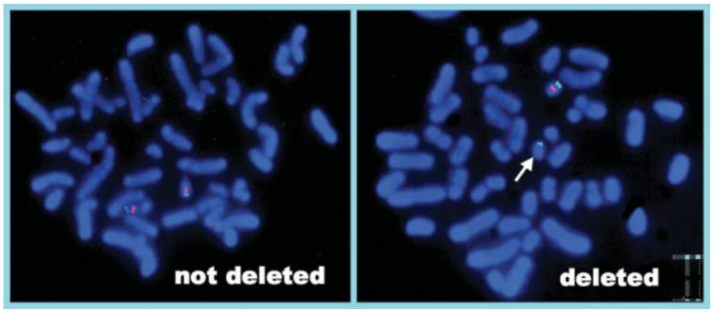

Fig. 1. Fluorescence In Situ Hybridization (FISH) of someone with VCFS (left) compared with a normal inidivual (right). The FISH on the left shows that one copy of chromosome 22 has only one fluorescent signal (the control probe) compared with the normal case that has two signals on each chromosome (control probe and the probe specific to 22q11.2). [Color figure can be viewed in the online issue, which is available at www.interscience.wiley.com.]

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In total, more than 180 distinct clinical phenotypes have been described and reported impacting nearly every organ system and developmental function (McDonald-McGinn et al., 2000). Children who have VCFS often have learning difficulties and developmental delays. De Smedt et al. (2007) describes that many individuals with 22q11.2 deletion have a non-verbal learning disability, when tested, verbal IQ scores were higher than their performance IQ scores. Due to this type of learning disability, these children will have relative strengths in reading and rote memorization but will struggle with math and abstract reasoning. These individuals may also have communication and social interaction problems, such as autism. Martin et al. (2012) states that as adults, these individuals have an increased risk for developing mental illness such as depression, anxiety, and schizophrenia.

VCFS is characterized by a combination of medical problems that vary from child to child. These medical problems include: cleft palate and other palatal differences; heart defects, problems fighting infection; low calcium levels; discrepancies in the way the kidneys are formed or work; a characteristic facial appearance; learning problems; and speech and feeding problems (Reilly & Stedman, 2013). Not all of these identifying features are found in each child. Common clinical findings in children and adults with 22q11.2 deletion syndrome can be visualized in Table 1 below (Solot et al., 2019). The prevalence of speech-language disorders, developmental delay, intellectual disability, learning disability, hearing impairment, and palatal abnormality/velopharyngeal dysfunction are some of the highest percentages. These research findings exemplify why children affected by this syndrome need parental, medical, and classroom intervention to have a more successful social and educational experience. Most babies with VCFS will

live a long life and survive into adulthood, but some will require long-term care by a variety of specialists to manage any health issues.

Table 1. Common clinical findings in children and adults with 22q11.2 deletion syndrome.

Finding	Children (%)	Adult[a] (%)
Speech-language disorders	~95	?
Developmental delay	>95	
Intellectual disability	~75–85	92
Learning disability	82–100	
Hearing impairment	6–60	28
Palatal abnormality/VPD	67	42
Laryngotracheal abnormalities	14	?
Congenital heart/cardiovascular disease	64	26
Gastrointestinal/feeding problems	65	40
Immune deficiency	77	?
Hypocalcemia	55	64
Genitourinary anomalies	24	41
Autism spectrum disorder	19	16
ADHD	32–52	16–35
Anxiety disorder	~35	25
Psychotic disorder	15	40–58
Schizophrenia	2	~25

Note. Data based from Bassett et al. (2005, 2011), Campbell et al. (2018), De Smedt et al. (2007), Dyce et al. (2002), Green et al. (2009), Schneider et al. (2014), and Verheij et al. (2017). ? = unknown; VPD = velopharyngeal dysfunction; ADHD = attention deficit hyperactivity disorder.

[a]Many adult study population sizes are small.

Note. Reprinted from "Speech-Language Disorders in 22q11.2 Deletion Syndrome: Best Practices for Diagnosis and Management," by Solot et al., 2019, *American Journal of Speech-Language Pathology, 28,* p. 985. Copyright 2019 by American Speech-Language-Hearing Association.

CHAPTER TWO

Palatal Abnormalities, Diagnosis, and Correction

The 22q11.2 deletion is currently the most common diagnosis associated with palatal problems, and the spectrum of such issues in the deletion are broad (McDonald-McGinn et al., 2000). A cleft is an abnormal separation of parts of the upper lip or the roof of the mouth. Children with VCFS are prone to both cleft lip and cleft palates. However, many children with 22q11.2 deletion exhibit signs or symptoms of palatal problems even in the absence of a palatal cleft, this condition is referred to as velopharyngeal incompetence or insufficiency (VPI) (McDonald-McGinn et al., 2000). According to Jackson and Kaye (2019), VPI is caused by poor movement of the velopharyngeal structures or abnormality of the soft palate. McDonald-McGinn et al. (2000) states "VPI happens when a child's soft palate is too short or too weak to close off the nasal cavity from the oral cavity during speech. VPI may present itself as leakage of air, food, or fluids through the nose, hypernasal speech, and errors in articulation caused by an attempt to compensate for the inability to close off the nasal cavity during speech (pp. 31-32). Velopharyngeal dysfunction (VPD) is a broad term used to describe the nasal tone to a person's voice when too much air flows through the nose due to the failure of the soft palate to fully close the velopharyngeal port (Jackson & Kaye, 2019). Solot et al. (2019) recommends VPD should be suspected in all children with VCFS until sufficient speech has developed to confirm its presence or absence.

These symptoms are not visible until later due to their appearance during speech production. The assessment of velopharyngeal function can be conducted with a variety of tests with a multidisciplinary approach (McDonald-McGinn et al., 2000). A perceptual speech assessment, as well as tests to detect abnormal airflow through the nose, will likely be done (McDonald-McGinn et al., 2000). VPI's unique excessive airflow through the nose can cause nasal resonance during speech known as hypernasality (McDonald-McGinn et al., 2000). A perceptual speech assessment utilizes auditory perception to recognize compensatory errors in articulations. However, in order to evaluate the velopharyngeal function, visualization during speech production is necessary. Visualization occurs by utilizing endoscopy (nasendoscopy) and video x-ray evaluation (Jackson & Kaye, 2019). Solot et al. (2019) states that "Variables assessed during visualization include velar length, pharyngeal width and depth, pharyngeal wall and velar movement, extent and pattern of VP closure, VP gap size, tonsil and adenoid size/position, and timing of VP closure" (p. 990). Clinicians should use a standard speech sample, including the repetition of oral-loaded stimuli at the word, phrase, or sentence level (Solot et al., 2019). If a persistent opening is seen in the velopharynx during speech, the child has demonstrated evidence of velopharyngeal insufficiency (McDonald-McGinn et al., 2000).

According to Bohm et al. (2019), a surgical plan will not be formulated until the diagnostic test with a nasendoscopy or a test to look at the inside of the nose, throat (pharynx), and voicebox (larynx). A comprehensive preoperative medical evaluation should take place to assess cardiac and airway factors, cervical spine stability, and carotid artery position before surgery (Solot et al., 2019). Abnormal placement of the internal

carotid arteries and kinked vertebral arteries are potentially dangerous risks when pharyngeal surgery is being planned (Shprintzen, 2008). Surgical procedures to correct VPD diminish the size of the nasopharyngeal airway and can cause or worsen obstructive sleep apnea (OSA) (Solot et al., 2019). A tonsillectomy or adenoidectomy before VPD surgery should be considered to reduce the risk of postoperative sleep apnea or if the accurate placement of a pharyngeal flap or sphincter pharyngoplasty will be affected (Solot et al., 2019). An adenoidectomy before planned VPD surgery may worsen hypernasality and speech until the pharyngoplasty can be performed (Solot et al., 2019). Since VPD and speech disorders in VCFS are often severe and multifactorial, management in this population is more complicated than others.

Jackson and Kaye (2019) discussed the timing and candidacy for VPD management is a complex decision involving the patient, family, the SLP, and surgeon. Ideal presurgical decision making requires that the child has sufficient speech for the evaluation of VP structure and function (Solot et al., 2019). For this reason, many centers recommend waiting until at least four years old before VPD surgery. Unfortunately, in some patients, complete correction may not be possible without significant risk for OSA (Jackson & Kaye, 2019). Nonsurgical options, such as prosthetics, may be considered for individuals who are not surgical candidates (Solot et al., 2019). These devices typically are affixed to the teeth and can help lift the soft palate (Jackson & Kaye, 2019). The prosthetic will require continual readjustment with the growth of the child or if the child does not tolerate it well (Jackson & Kaye, 2019).

If a patient has a cleft palate or cleft lip, it is usually addressed early on since it causes difficulties with feeding (McDonald-McGinn et al., 2015). All surgical

procedures for the correction of VPI require some palatal and pharyngeal wall movement to assist with velopharyngeal closure (McDonald-McGinn et al., 2000). According to Jackson and Kaye (2019), primary surgeries are less effective at improving speech, and a second surgery is often required. The goal of cleft lip surgery is to restore the standard appearance and function of the upper lip. Cleft lip surgery is recommended at three to four months old ("Cleft Lip and Palate Treatment," n.d.). The purpose of cleft palate surgery is to repair the palate to avoid abnormal speech development; therefore, surgery should be scheduled before the child develops speech usually when infants are 6 to 18 months old ("Cleft Lip and Palate Treatment," n.d.). McDonald-McGinn et al. (2000) states that "When a cleft in the palate is present, separation of the oral and nasal passages does not occur, and fluids can leak through the nose during feeding, and air escapes through the nose during speech" (p. 34).

Solot et al. (2019) recommends that parents should be counseled about the anticipated postoperative speech outcome and advised that surgical intervention affects resonance, nasal emission, intraoral pressure, and oral airflow. Learned compensatory articulation and other errors of placement, manner, and voicing require ongoing speech therapy before and after surgery (Solot et al., 2019). Postoperative recovery may pose challenges depending on the child's learning differences or cognitive impairments. If this is the case, the correction of articulation errors and speech improvement may occur more slowly (Jackson & Kaye, 2019). Palatal function plays an essential role in the development of speech; therefore, palatal problems can affect a child's communication with others (McDonald-McGinn et al., 2000).

CHAPTER THREE

Speech Formation and Therapeutic Methods

Solot et al. (2019) explains, "Delayed emergence of speech with restricted and atypical phoneme repertoires are frequent in children with VCFS" (p. 987). Phonemes are units of speech or a distinct unit of sound to distinguish words ("Phoneme," n.d.). Articulation is the formation of clear and distinct speech sounds with the use of the lips, tongue, teeth, etc. (McDonald-McGinn et al., 2000). Research by McDonald-McGinn et al. (2000) found that due to their cleft and pharyngeal abnormalities, many children will alter their typical articulation patterns to compensate for the inability to generate adequate air pressure needed to produce certain sounds. Compensatory errors may be present as distortions, substitutions, omissions, or the addition of sounds during speech sounds (McDonald-McGinn et al., 2000). McDonald-McGinn et al. (2000) found that hoarseness, reduced loudness, grimacing, low tone, and compensatory errors can worsen speech intelligibility. Velopharyngeal dysfunction can lead to maladaptive articulation patterns with unintelligible speech, causing frustration that can affect a child's psychosocial development (McDonald-McGinn et al., 2015). Frequently, speech cannot improve until a secondary surgery has been completed. Surgical intervention allows the child to have anatomic structures that allow adequate air pressure needed for speech production. Although progression and improvement in speech occur in school-age children, speech sound deficits may persist into late childhood and adolescence (Solot et al., 2019). Features of VPD include weak pressure consonants, nasalization of

phonemes, and audible nasal air emission during the production of oral pressure consonants (Solot et al., 2019). These obligatory errors occur because the anatomic structure is not adequate for speech production. See Table 3 below for common speech disorders in children with VCFS (Solot et al., 2019)

Solot et al. (2019) considers speech-language pathologists (SLPs) to be one of the first professionals consulted because of the high prevalence of feeding difficulties, speech-language delays, and disorders in infants and young children with 22q deletion syndrome. Services of a speech and language pathologist can be found in most hospitals, schools, and specialized clinics (McDonald-McGinn et al., 2000). There are also private practices in speech-language pathology, but the individual should be certified by the American Speech, Language, and Hearing Association and licensed by the state (McDonald-McGinn et al., 2000). In other cases, SLPs may be consulted later as children begin to present communication disorders in school-age years (Solot et al., 2019). Early intervention should focus on strategies to improve expressive communication and prevention or remediation of compensatory articulation patterns (Solot et al., 2019). Solot et al. (2019) recommends a variety of approaches to improve speech sound production skills will be used depending on the type and severity of the speech disorder, the child's stimulability, and the development, and the developmental/cognitive status. During the infant and preschool years, intervention should focus on parent counseling, facilitating communicative intent, and social interactions (Solot et al., 2019).

As a parent, knowing how to assist your child developmentally is an empowering ability. Family members are typically the most influential people in a child's life before

attending school. While a parent's efforts cannot replace the expertise and training of an SLP, encouraging and facilitating extra practice outside of therapy could positively influence a child's progress. It is well recognized that more treatment time translates into better outcomes, especially before the child begins attending school (Solot et al., 2019). In school-age children, language intervention should focus on the deficits causing a significant impact on functional communication, social interaction, and academic performance (Solot et al., 2019). McDonald-McGinn et al. (2000) say parents who know about these challenges can provide experiences and guide activities to assist in difficult areas while encouraging their child's strengths. See appendices A-D for best practices for evaluation and management of language for infants, preschool-aged children, early school-aged children, and late school-aged children with VCFS (Solot et al. 2019).

Table 3. Common speech disorders in children with 22qDS.

Speech domain	Common deficits in 22qDS
Resonance	• Hypernasality secondary to velopharyngeal dysfunction and/or submucous cleft palate
Voice	• High pitch • Dysphonia (decreased loudness, hoarseness, breathiness, tension, vocal fatigue, strained–strangled voice) secondary to velopharyngeal dysfunction, dysarthria, laryngeal anomalies, for example, laryngeal web, vocal fold paralysis/paresis, vocal misuse, or gastroesophageal reflux disease
Articulation	• Restricted and delayed speech sound acquisition • Speech sound disorders including articulation impairments characterized by compensatory misarticulations, phonological disorders, and motor speech disorders • Obligatory or passive errors secondary to velopharyngeal dysfunction including weak pressure consonants, audible nasal emission/turbulence, and/or nasalized plosives • Developmental immaturities • Abnormal speech prosody
Motor speech	• Childhood apraxia of speech (CAS) • Dysarthria • Mixed features of CAS and dysarthria • Speech motor delay

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CHAPTER FOUR

Communication Delays: Learning, Comprehension, and Effectiveness

According to Crerand and Rabkin (2019), "Children with VCFS frequently experience difficulties with social functioning likely due to developmental, cognitive, and behavioral problems, as well as risks for speech and language problems because of velopharyngeal dysfunction and other neurophysiological deficits" (p. 635). According to Vollmer (2020), if a person has trouble understanding others or sharing thoughts, ideas and feelings, the person may have a language disorder which can be receptive or expressive. Receptive language is the ability to understand and comprehend the language you hear or read. Children who have difficulty understanding language may struggle following directions, understanding the meaning of gestures, answering questions, identifying objects and pictures, understanding a story, or with reading comprehension (Vollmer, 2020). Expressive language is the ability to express your wants and needs through either verbal or nonverbal communication. These children who have trouble producing language may struggle to ask questions, name objects, use gestures, use facial expressions, and make comments, with vocabulary, syntax (or grammar rules), semantics (word/sentence meaning), and morphology (forms of words) (Vollmer, 2020). Solot et al. (2019) says that the majority of these children experience significant receptive and expressive language impairments.

Infants often have absent or reduced babbling, with their first words not emerging until two years old; however, some children may remain nonverbal until four years old

(Solot et al., 2019). Language difficulties such as reduced grammatical complexity, vocabulary and concept development, abstract reasoning, word-finding, and pragmatic language continue once the child is of age to go to school (Solot et al., 2019). In preschool, evaluation of expressive language must account for cognition, VPD, and speech sound disorders associated with velocardiofacial syndrome (Solot et al., 2019). Social communication skills should also be routinely assessed at all ages (Solot et al., 2019).

Solot et al. (2019) recommends, "In school-age children, basic language skills, as well as higher-level language functions and pragmatic language should be assessed (p. 987). Higher functioning children may do well on tests of basic language skills but have difficulty with narrative, abstract, figurative, or inferential language (Solot et al., 2019). Some children may not display early language deficits, but will demonstrate them later as linguistic demands increase with age; therefore, reevaluation at regular intervals over their lifetime is essential (Solot et al., 2019). Most children with velocardiofacial syndrome will develop oral communication, but due to significant and persistent early language delay, language therapy is recommended. Crerand and Rabkin (2019) observed that children with VCFS are often motivated to engage socially but more often with adults or younger rather than same-age peers.

Communication through pointing, gestures, and formal American Sign Language helps bridge the gap before verbal speech develops (McDonald-McGinn et al., 2000, p. 78). When your child is delayed in language and speech, it is essential to keep in mind the larger goal of communication: showing your child how to communicate their thoughts, feelings, and needs successfully, even if it is through gestures or an

approximation of the desired words (McDonald-McGinn et al., 2000). Children with VCFS have deficits in processing and understanding nonverbal information, causing difficulty with social and pragmatic communication (McDonald-McGinn et al., 2000). McDonald-McGinn et al. (2000) report, as a result, these children and adolescents may misread nonverbal cues and signals. Their understanding of social distance may be limited, and they may misinterpret meanings of reinforcements or punishments by caregivers or educators.

Because of their nonverbal processing deficits, children and adolescents with VCFS are at a much greater risk for experiencing social-emotional and mood-based disorders (McDonald-McGinn et al., 2000). According to McDonald-McGinn et al. (2000), their adaptive skills are delayed relative to their peers. They appear to have the most difficulty dealing with everyday life changes, daily living skills, and communication abilities necessary to interact with others (McDonald-McGinn et al., 2000). Crerand and Rabkin (2019) state, "Children with VCFS may have problems understanding metaphors, irony, and idioms, deficits that may have direct impacts on communication with peers and set the stage for social challenges, including bullying" (p. 636). Negative social interactions may contribute to anxiety and withdrawal from social situations, further limiting opportunities for positive interactions and social skill development (Crerand & Rabkin, 2019). Boyer et al. (2012) discovered that socially, children with VCFS demonstrate less interaction with peers, less imaginative play, and less emotional expression. McDonald-McGinn et al. (2000) explain that for the development of social skills, children should be enrolled in either extracurricular activities, sports programs, social networks, or youth groups found in places of worship whenever possible. Boyer et

al. (2012) credits fewer social initiations to be attributed to speech and language delay in childhood. Language is critical because it is one of the ways we regulate our behavior, communicate with each other, and learn in school (McDonald-McGinn et al., 2000). See Table 2 for common language deficits and management strategies found in children with VCFS (Solot et al., 2019)

Table 2. Common language deficits children with 22qDS and management strategies.

Language domain	Common deficits in 22qDS
Vocabulary and concepts	Topic-based and/or abstract vocabulary and concepts Difficulties with terms with multiple meanings Superficial, concrete word/concept knowledge *Vocabulary may need to be taught in each context.*
Structural language	Delayed development of syntax and sentence construction Lack of complexity of verbal output *Syntax may have to be taught directly in therapy.*
Abstract/nonliteral language	Idioms, sarcasm, ambiguity, humor, and nonliteral use of language *These need to be explicitly explained and taught.*
Narrative comprehension and generation	Extracting salient points from verbal or written narratives Understanding implications, making inferences, predictions Disorganized, terse, ambiguous, or verbose narratives *Provide structured support in story and narrative construction.*
Processing speed	May be slower Difficulty understanding long, complex sentences. *Additional time and repetition may be required.*
Classroom or academic language	Educational staff may need instruction regarding: *Modifying and simplifying their language* *Providing repetition as needed* *Ensuring instructions are understood* *Cueing the child on the ending of one task and the commencement of another* *Being alert to the possibility that the child may not seek clarification if tasks or instructions are unclear* *Providing scaffolding to assist the child with expressive language*
Pragmatics	There is increased impact of pragmatic deficits with age secondary to the added sophistication and demands of social communication. Assist with: *Topic introduction, maintenance, shifting* *Use of appropriate tone of voice* *Understanding the perspective of the listener* *Interpreting nonverbal communication (body language/facial expression)*

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CHAPTER FIVE

Education and Learning Disabilities

Most children with velocardiofacial syndrome should be placed in the regular classroom to allow proper socialization and daily academic challenges (McDonald-McGinn et al., 2000). According to McDonald-McGinn et al. (2000), many children will require learning support services or tutoring in elementary school. When additional assistance is needed in certain subjects, special education services can be provided in a resource room or by an in-class aid (McDonald-McGinn et al., 2000). Assistance is typically necessary in the area of math; mathematical learning disabilities (MLD) frequently occur in children with specific genetic disorders, including VCFS (De Smedt et al., 2009). Impairments in understanding and representing quantities and numbers are a likely reason for explaining the MLD associated with VCFS (De Smedt et al., 2009). De Smedt et al. (2009) reported that these children might benefit from interventions that foster the development of quantity representations and the association between numbers and the quantities they represent via number board games or specific remediation programs. The weakness in math in children with VCFS is more pronounced in areas of abstract reasoning, converting language into mathematical expressions, telling time, using money, and problem-solving (Reilly & Stedman, 2013).

Many school-aged children perform close to or in the average range on phonological tasks such as sentence repetition, reading, and metaphonolgical awareness; however, reading comprehension may be a relative weakness in these children (Reilly &

Stedman, 2013). Reilly and Stedman (2013) explain, "They seem to be adept at 'learning to read' but have more difficulty with comprehension or 'reading to learn'" (p. 117). Reilly and Stedman (2013) reported, based on parent and teacher reports, three-quarters of children with VCFS were able to read at an age-appropriate level. However, only 2% were age-appropriate in their understanding of what they were reading (Reilly & Stedman, 2013). Reilly and Stedman (2013) found that school difficulties may become particularly noticeable in the upper primary years as the curriculum begins to focus on cognitive skills such as working memory and more abstract areas in mathematics and literacy.

Children with VCFS may have difficulties with nonverbal processes, visual-spatial skills, visual-spatial memory, and facial processing. In the educational or home environment, try to limit or modify the information, so a strong visual-spatial demand is not present (McDonald-McGinn et al., 2000). Suggested classroom accommodations for this include simplifying the number of questions or problems on each page, cutting back on the number of notes that need to be copied from the textbook or whiteboard, and helping them keep school materials organized in a binder (McDonald-McGinn et al., 2000). Their verbal strengths should be encouraged whenever possible by asking them to "talk through" or "think aloud" about steps needed to get through problems (McDonald-McGinn et al., 2000). "Verbal IQ" was generally higher than their "Performance IQ," which is rare in the general population and often associated with difficulty in math (McDonald-McGinn et al., 2000). McDonald-McGinn et al. (2000) found that after an examination of test scores, math scores were lower than reading and spelling, which is surprising since many have language and speech delays as young children. Longitudinal

studies have shown that cognitive development is variable, and IQ is not necessarily stable across the lifespan of those with VCFS; some individuals show cognitive decline, whereas others make progress (Solot, 2019).

Additional problem areas occur with their attention and working memory (McDonald-McGinn et al., 2000). They have the most difficulty with tests, which often come with a time restraint (McDonald-McGinn et al., 2000). Giving preferential seating near the teacher and controlling auditory distractions can assist the child in maintaining their focus (Reilly & Stedman, 2013). Their working memory skills, which involve keeping information active in the memory so it can be remembered short term, is a significant weakness (McDonald-McGinn et al., 2000). Sanders et al. (2017) reports that anxiety disorders and ADHD are two common diagnoses in children with VCFS, and children with ADHD commonly show working memory deficits. Supplementary notes to support the student's own notes and simplified worksheets, as well as bulleted lists of instructions or checklists, can help students understand an activity (Reilly & Stedman, 2013). Reilly and Stedman (2013) recommend scheduling activities using visual or written time-tables as well as giving plenty of notice that an activity will change using verbal and visual cues can assist a child with their ability to follow directions and remember instructions. There is likely an expectation that children can follow multiple oral instructions and work efficiently and independently, which may be difficult for some of these children. Some children will excel in particular areas, but in order for others to "keep up" with their peers, some will benefit from extra attention in problematic areas (Tobia et al., 2017).

As a group, children with VCFS have relative strengths and weaknesses, but it is a spectrum condition, and thus it is vital to assess each child on an individual basis (Reilly & Stedman, 2013). According to De Smedt et al. (2007), the intelligence quotient (IQ) of 103 children with VCFS were collected and found to have a mean full-scale IQ (FSIQ) of 73.48 (range: 50-109). There were no effects relating to sex, the presence of a heart defect, and psychiatric condition, but the inheritance of the deletion affected cognitive performance (De Smedt et al., 2007). De Smedt et al. (2007) found that children with familial deletions had a significantly lower FSIQ than children with a de novo deletion. About 60% of the children showed borderline to average intelligence (FSIQ >70), whereas an intellectual disability (FSIQ < 70) was noted in 40% (De Smedt et al., 2007).

McDonald-McGinn et al. (2000) states that "Relative to their overall intellectual performance and nonverbal skill, VCFS children and adolescents show a unique ability to learn and retain information taught to them through cultural opportunities, experience at home, or through instruction at school" (p. 99). Children with this syndrome have a remarkable ability to learn and recall rote or repetitive verbal information (McDonald-McGinn et al., 2000). McDonald-McGinn et al. (2000) reported they also show strength in establishing their initial auditory attention span; therefore, teaching methods that utilize rote forms of auditory presentation may provide more success for learning over time. These children and adolescents do better on tasks that are brief and highly structured. They can be expected to perform within normal limits and will benefit from immediate feedback and reinforcement regarding their performance (McDonald-McGinn et al., 2000).

Teaching within the home or school setting by segmenting learning into smaller time components and providing constructive reinforcement and feedback is the best way to facilitate their education (McDonald-McGinn et al., 2000). According to Crerand and Rabkin (2019) "Children with VCFS should undergo a comprehensive neuropsychological evaluation including standardized assessment of cognitive abilities (IQ), attention and executive functioning, visual-spatial and language skills, learning and memory, fine motor control, academic skills, and emotional/behavioral functioning" (p. 637). The child's pattern of strengths and weaknesses can be used to make recommendations for educational and treatment planning (e.g., need for Individualized Education Plans, psychological or psychiatric treatment) (Crerand & Rabkin, 2019). Developmental disabilities in children are often accompanied by high rates of behavioral problems and psychiatric disorders.

CHAPTER SIX

Psychiatric Disorders and Psychotic Disorder Onset

While parents of children affected by VCFS typically receive information about risk for non-psychiatric health concerns, they frequently lack sufficient information about the chance for psychiatric illness in their child (Hart et al., 2015). Parents were more likely to receive information about psychiatric manifestations of the condition through the internet than from clinical sources (Hart et al., 2015). The findings support the need for improved access to medical expertise in psychiatric disorders and a potential role for genetic counselors to provide support resources and referrals (Hart et al., 2015). Gothelf et al. (2009) reports that children with developmental disabilities share common risk factors such as social isolation and rejection, impairments in social and daily living skills, low self-esteem, and overprotection by parents. Common psychiatric disorders reported in a high proportion of children and adolescents with VCFS include attention-deficit/hyperactivity disorder (ADHD, 35-45%), oppositional defiant disorder (16-43%), specific and social phobias (23-61%), generalized anxiety disorder (17-29%), separation anxiety disorder (16-21%), obsessive-compulsive disorder (4-33%), major depressive disorder and dysthymia (10-20%), and autistic spectrum disorders (14-45 %) (Gothelf et al., 2009, p. 154). According to Gothelf (2009), the increased rate of most psychiatric disorders in children with VCFS is similar to that in children with other developmental disabilities. However, by late adolescence and early adulthood, up to one-third of the

patients with VCFS develop psychotic disorders resembling schizophrenia and schizoaffective disorder (Gothelf et al., 2009).

There are early psychiatric signs before the emergence of a full-blown psychotic disorder that seems to predict the later development of schizophrenia in VCFS (Gothelf et al., 2009). The presence of subthreshold psychotic symptoms (STPS) occurs in about one third to one-half of children with the syndrome (Gothelf et al., 2009). Rosengard et al. (2018) states that "STPS include suspiciousness or odd ideas of reference, strange or bizarre ideas that are not delusional, unusual or eccentric behavior, unusual perceptual experiences that are not clearly psychotic, disorganized or odd speech, inappropriate affect, hallucinations or delusions (sub-threshold), and passivity experiences (p. S185). Anxiety, depression, and obsessive-compulsive disorder are also predictors for the onset of psychotic disorders (Gothelf et al., 2009). Gothelf et al. (2009) explains that although ADHD is the most common psychiatric disorder in VCFS, its presence does not seem to place children at a greater risk for developing a psychotic disorder.

One of the best-known risk factors for developing schizophrenia is having VCFS (Schneider, 2019). The relative risk for schizophrenia in a patient with VCFS is about twenty to twenty-five times the lifetime general population risk of 1% (Bassett & Chow, 2008). However, families should be aware that just because the risk of developing a psychiatric illness is high, does not mean every child with VCFS child will develop it. Families should be informed bout practical steps to avoid exposure to triggers of psychiatric illness, such as stress management, proper nutrition, avoiding alcohol, ensuring adequate sleep, avoiding cannabis, and methamphetamine (Hart et al., 2015). Focusing on developing social skills is also valuable in reducing risks. Providers should

communicate that these factors may or may not reduce the risk for particular individuals. According to Hart et al. (2015), it is important for family members to realize that the development of a psychiatric illness does not mean they have failed to prevent the condition. Most genetic counselors agree that the elevated risk of developing a psychiatric illness for VCFS patients should be disclosed to parents; however, a significantly lower proportion of genetic counselors would disclose this information compared to other health related information in an initial session (Martin et al., 2012).

Psychosis and schizophrenia are often confused, but the key difference is that the term schizophrenia is normally used for psychotic states that last longer in duration (Mediavilla, 2019). In fact, psychosis is a symptom of schizophrenia. Psychosis can be classified by the type of difficulties experienced, such as positive or negative symptoms (Mediavilla, 2019). Positive symptoms "add" new experiences to what a person might be perceiving, such as delusions, hallucinations, disorganized speech, disorganized behavior, and agitation (Mediavilla, 2019). Negative symptoms make it difficult to do many activities of daily living; these symptoms include: lack of interest, difficulties organizing, concentration difficulties, social isolation, lack of emotions, and lack of energy (Mediavilla, 2019). Schneider (2019) reports that the presence of negative symptoms should be a primary target for therapeutic intervention in VCFS to improve long and short-term functional outcomes. Impairments in both experience of emotion and self and personal hygiene may increase the ability to predict the emergence and persistence of psychosis (Schneider, 2019).

Discussing the benefits of early detection may be helpful for patients at risk for psychiatric illness, as earlier detection has been found to be associated with improved

prognosis (Hart et al., 2015). Hart et al. (2015) says that families should be provided with internet resources with clinical and scientific information that has been appropriately evaluated, but these resources should not replace interactions with health care providers. Families affected by VCFS need expertise in a range of psychiatric illnesses not only psychosis, but also childhood-onset disorders such as ADHD, autism spectrum disorders, and anxiety/depressive disorders (Hart et al., 2015). According to Hart et al. (2015), "Genetic counselors should also recommend psychiatric, behavioral and emotional assessments be carried out at each stage of development, including at the time of diagnosis, preschool age, school age, adolescence, and adulthood" (p.11).

Kates et al. (2018) describes a cross-sectional study of 1402 individuals with VCFS; it was reported that ADHD dropped significantly from 37% in childhood, to 24% in adolescence, and 16% in adulthood. Anxiety disorders dropped less dramatically, from 36% in childhood to 26% in adulthood (Kates et al., 2018). In contrast, mood disorders increased from 3% during childhood to 18% in adulthood (Kates et al., 2018). Lastly, schizophrenia spectrum disorders increased significantly with age from 2% during childhood, over 10% in adolescence, to 24% between 18 and 25 years old, and over 41% in adults over the age of 25 years (Kates et al., 2018, p. 1914). Despite the high prevalence of psychiatric disorders in VCFS, individuals with this syndrome are reported to be undertreated (Kates et al., 2018). Reported medication use was deficient during childhood and early adolescence, except for stimulants (Kates et al., 2018). However, as patients moved into late adolescence and young adulthood rates of anti-anxiety and anti-depressant medication increased significantly, about 25% (Kates et al., 2018). The goal of this study was to report longitudinal trajectories of psychiatric disorders, predictors of

persistence, and medication usage throughout four time points in youth with VCFS (Kates et al., 2018).

As noted previously, VCFS is associated with an increased risk of various disorders, including autism spectrum disorder (ASD) and attention deficit hyperactivity disorder (ADHD) (Hidding et al., 2015). According to Fullman and Boyer (2020), children who are eventually diagnosed with ADHD often display the characteristics of impulsivity, overactivity, and developmentally inappropriate levels of attention in early childhood. Fullman and Boyer (2020) state that "As a parent, it is important to establish a positive relationship, give clear instructions and boundaries, and provide positive reinforcement for appropriate behaviors for children with ADHD" (p. 23). The rate of comorbidity of VCFS and ASD varied by investigation, but all physicians agreed that screenings should take place for ASD as early as possible to provide prompt referrals if any diagnostic indicators would be found (Fullman & Boyer, 2020). Children with both VCFS and ASD demonstrated impairments in nonverbal social interactions and pretend play in combination with ritualistic, repetitive behaviors and motor stereotypes (Fullman & Boyer, 2020). In contrast, children with only VCFS did not display repetitive, ritualistic behaviors or motor stereotypes (Fullman & Boyer, 2020).

CHAPTER SEVEN

Advocates for VCFS: Caregivers, Educators, and Physicians

Crerand and Rabkin (2019) say, "Parents may experience a range of feelings including being overwhelmed, sad, anxious, angry, and even relieved due to having an explanation for their child's symptoms (p. 634). Psychiatric risks associated with VCFS have been rated as the greatest sources of worry for parents (Crerand & Rabkin, 2019). Learning about the risk for psychosis decades before symptom onset may contribute to this anxiety and stigma associated with psychiatric diagnoses (Crerand & Rabkin, 2019). Luckily, most of the medical problems associated with 22q can be managed; it is the cognitive, psychological, and social difficulties that present more of a challenge, especially in the classroom ("22q Educational Tools," 2017).

Crerand and Rabkin (2019) report that screenings for infants and young children can occur through early intervention programs, school districts in the child's community, or with psychologists or developmental pediatricians who specialize in working with children with autism, developmental delays, or complex medical conditions. Early identification of delays and enrollment in developmental therapies can help optimize outcomes (Crerand & Rabkin, 2019). Ongoing surveillance of mental health is recommended throughout childhood, adolescence, and adulthood (Crerand & Rabkin, 2019). According to Crerand and Rabkin (2019), "For school-age children, screening should focus on symptoms of anxiety, ADHD, and cognitive abilities in order to set children up for success at the time of school entry and to facilitate appropriate service

provision in educational settings (p. 637). Social skill deficits also warrant identification; therefore, routine screening for anxiety and depressive symptoms and monitoring for symptoms of schizophrenia, especially in adolescence and beyond, is important (Crerand & Rabkin, 2019). Children with VCFS may have an increased need for sameness, structure, and certainty; therefore, environments that support this need may improve the child's functioning and limit stress (Crerand & Rabkin, 2019).

One study performed focused on unaffected siblings of children with VCFS. In this study, it was found that unaffected siblings reported negative and positive feelings related to the impact of the affected child (Okashah, 2014). Negative feelings were related to the affected child's behavior and restrictions placed on the family due to illnesses or hospitalizations (Okashah, 2014). Okashah et al. (2014) reports that positive impacts were related to developing an increased tolerance, patience, compassion, and understanding of differences. It was also common for siblings to show concerns about people staring or ridiculing their brother or sister (Okashah, 2014). This study also assessed parents' tendencies to share genetic, behavioral, and medical information about the affected sibling with unaffected children, which were generally quite high (Okashah, 2014). Having knowledge about a sibling's medical condition, especially one that's not well known, could be monumental for spreading awareness. Since many children with VCFS can face struggles socially and educationally, it is pertinent that educators are informed of possible challenges and assistance that may be necessary.

In one study, over 50% of teachers of children with VCFS rated themselves highly on knowledge about the syndrome, while 30% felt they had inadequate knowledge (Reilly & Stedman, 2013). Teachers indicated they would benefit from more information

on both the physical and educational aspects of VCFS (Reilly & Stedman, 2013. Crerand and Rabkin (2019) state, "School systems frequently lack awareness of VCFS and its risks for intellectual and learning disabilities, as well as behavioral risks that may require attention in the school setting" (p. 637). Families may encounter barriers to accessing needed special education services for their children (Crerand & Rabkin, 2019). Given some of the known barriers to obtaining support and education, families, as well as other medical and school professionals who are unfamiliar with VCFS, can be direct to reputable online resources for support and information (Crerand & Rabkin, 2019). Two highly recommended websites include The 22q Family Foundation (http://www.22qfamilyfoundation.org) or The International 22q11.2 Foundation Inc. (http://www.22q.org). "22q Deletion Syndrome in the Classroom: A Teachers Reference" is a shortened, but informative, resource of many topics covered in this paper https://22qfamilyfoundation.org/sites/default/files//22q-for-teachers.pdf ("22q Educational Tools," 2017).

Early recognition of VCFS is imperative, so treatment can be initiated, thus preventing or lessening problems for the patient and family members. McDonald-McGinn et al. (2000) voices the importance for a parent to have access to resources regarding VCFS and to establish a professional care provider who can coordinate and orchestrate the care of the child. Establishing an interdisciplinary team for anyone diagnosed with VCFS is essential since the syndrome affects nearly every aspect of their life. Early developmental delays require early intervention services from professionals, including speech-language pathologists, developmental therapists, or child development specialists, physical therapists, occupational therapists, and behavioral specialists (Boyer

et al., 2012). One primary role of a care provider is to be knowledgeable and comfortable enough to talk about all aspects of the syndrome. This person will be vital in interpreting and communicating results from sub-specialists to the parents. This includes informing families about the psychiatric conditions that occur more commonly in patients diagnosed with VCFS than the general population, regardless of the potential stigma that surrounds mental illnesses.

CHAPTER EIGHT

Conclusion

Since VCFS affects up to 1:2000 for surviving newborns, it is surprising the syndrome is not more well-known (Shprintzen, 2008). In total, more than 180 distinct clinical phenotypes have been described and reported impacting nearly every organ system and developmental function (McDonald-McGinn et al., 2000). VCFS is characterized by a combination of medical problems that vary from child to child including cleft palate and other palatal differences; heart defects, problems fighting infection; low calcium levels; discrepancies in the way the kidneys are formed or work; a characteristic facial appearance; learning problems; and speech and feeding problems (Reilly & Stedman, 2013). This literature review aimed to discuss literature for caregivers, educators, and physicians to aid children effectively and understand their challenges relating to speech, communication, and education. The prevalence of speech-language disorders, developmental delay, intellectual disability, learning disability, hearing impairment, and palatal abnormality/velopharyngeal dysfunction are some of the highest percentages. These research findings exemplify why children affected by this syndrome need parental, medical, and classroom intervention to have a more successful social and educational experience.

Palatal function plays an essential role in the development of speech; therefore, palatal problems can affect a child's communication with others (McDonald-McGinn et al., 2000). According to Solot et al. (2019), speech-language pathologists (SLPs) may be

one of the first professionals consulted because of the high prevalence of feeding difficulties, speech-language delays, and disorders in infants and young children with 22q deletion syndrome. Parents who know about these challenges can provide experiences and guide activities to assist in difficult areas while encouraging their child's strengths (McDonald-McGinn et al., 2000). Crerand and Rabkin (2019) describe children with VCFS to frequently experience difficulties with social functioning likely due to developmental, cognitive, and behavioral problems, as well as risks for speech and language problems secondary to velopharyngeal dysfunction and other neurophysiological deficits. Some children may not display early language deficits, but will demonstrate them later as linguistic demands increase with age; therefore, reevaluation at regular intervals over their lifetime is essential (Solot et al., 2019).

Most children with velocardiofacial syndrome will develop oral communication, but due to significant and persistent early language delay, language therapy is recommended. McDonald-McGinn et al. (2000) states "They appear to have the most difficulty dealing with everyday life changes, daily living skills, and communication abilities necessary to interact with others" (p. 111). Children with VCFS may have problems understanding metaphors, irony, and idioms, deficits that may have direct impacts on communication with peers, and set the stage for social challenges, including bullying (Crerand & Rabkin, 2019). McDonald-McGinn et al. (2000) reports that "For the development of social skills, children should be enrolled in either extracurricular activities, sports programs, social networks, or youth groups found in places of worship whenever possible" (p. 111).

Most children with velocardiofacial syndrome should be placed in the regular classroom to allow proper socialization and daily academic challenges (McDonald-McGinn et al., 2000). Many children will require learning support services or tutoring in elementary school (McDonald-McGinn et al., 2000). Reilly and Stedman (2013) describe that as a group, children with VCFS have relative strengths and weaknesses, but it is a spectrum condition, and thus it is vital to assess each child on an individual basis. Children with VCFS should undergo a comprehensive neuropsychological evaluation, including a standardized assessment of cognitive abilities (IQ), attention and executive functioning, visual-spatial and language skills, learning and memory, fine motor control, academic skills, and emotional/behavioral functioning (Crerand & Rabkin, 2019). The child's pattern of strengths and weaknesses can be used to make recommendations for educational and treatment planning (e.g., need for Individualized Education Plans, psychological or psychiatric treatment) (Crerand & Rabkin, 2019).

Common psychiatric disorders reported in a high proportion of children and adolescents with VCFS include attention-deficit/hyperactivity disorder, oppositional defiant disorder, specific and social phobias, generalized anxiety disorder, separation anxiety disorder, obsessive-compulsive disorder, major depressive disorder and dysthymia, and autistic spectrum disorders (Gothelf et al., 2009). According to Schneider (2019), one of the highest known risk factors found for developing schizophrenia is having VCFS. The relative risk for schizophrenia in a patient with VCFS is about twenty to twenty-five times the lifetime general population risk of 1% (Bassett & Chow, 2008). Discussing the benefits of early detection may be helpful for patients at risk for psychiatric illness, as earlier detection has been found to be associated with improved

prognosis (Hart et al., 2015). Hart et al. (2015) says that families should be provided with internet resources with clinical and scientific information that has been appropriately evaluated, but these resources should not replace interactions with health care providers. Hart et al. (2015) states that "Genetic counselors should also recommend psychiatric, behavioral and emotional assessments be carried out at each stage of development, including at the time of diagnosis, preschool age, school-age adolescence, and adulthood" (p.11).

Parents may experience a range of feelings, including being overwhelmed, sad, anxious, angry, and even relieved due to having an explanation for their child's symptoms (Crerand & Rabkin, 2019). It is important for a parent to have access to resources regarding VCFS and establish a professional care provider to coordinate and orchestrate the care of this individual (McDonald-McGinn et al., 2000). This person will be key in interpreting and communicating results from sub-specialists to the parents. In order for teachers to start becoming more informed, parents should take it upon themselves to begin providing information about their child and the syndrome. My mom has a binder that she gives to my brother's teacher every year. This was his first year of middle school, so the binder was given to his resource teacher, whom he goes to for assistance in specifically math and science, but also for homework assistance in general. In one study, over 50% of teachers of children with VCFS rated themselves highly on knowledge about the syndrome, while 30% felt they had inadequate knowledge (Reilly & Stedman, 2013). According to Reilly and Stedman (2013), teachers indicated they would benefit from more information on both the physical and educational aspects of VCFS.

I felt fortunate to be provided with this opportunity to write about a syndrome that has affected my family immensely. My brother certainly began life with many adversities, but with the help of my family, educators, and physicians, he has been able to maintain a relatively healthy and happy life. Many of his teachers previously had no knowledge about the syndrome, and my mom took it upon herself to create a binder with specific excerpts from websites, books, medical notes, and previous school reports in order for his teachers to best understand the challenges that we have and will have to overcome. I have always been reasonably knowledgeable about this topic, but I still learned an extensive amount of new material researching VCFS. I did not realize the total extent of the difficulties these children may have with school and how difficult it would be to assist them academically without knowing the background about the syndrome. Assisting children with developmental disabilities and delays associated with speech, communication, and education indeed requires an interdisciplinary approach and more resources for parents and educators to facilitate the highest quality of life among children with VCFS.

APPENDICES

APPENDIX A

Best Practices for Infants (Age 0-12 Months) with VCFS

Appendix A

Best Practices for Infants (Age 0–12 Months) With 22DS

Diagnosis in the infant period provides the opportunity to initiate early intervention to assist with feeding, development of communication strategies and parent counselling.

Domain	Evaluation	Management
Palate	• Evaluate structure for overt cleft, submucous cleft palate; assess for nasopharyngeal reflux • Referral to Cleft Palate Team	• Monitoring by Cleft Palate Team and 22qDS specialty clinic where possible
Speech-language development	• Referral to speech-language pathologist (SLP) • Assess prelinguistic communication skills (eye gaze, facial expression, joint attention, intentional communication, early signs of comprehension, production of vocalizations and proto words, gestures, babbling, early speech, and vocabulary development)	• Assess for risk of speech/language disorder or velopharyngeal dysfunction • Parent education for speech and language stimulation • Educate parents to recognize emergent communicative acts/early speech attempts • Referral to parent support networks • Therapy for early intervention strategies • Implement Total Communication early, at time of diagnosis • Frequent re-evaluation to monitor progress and adapt therapy plans
Hearing	• Newborn hearing screening • Audiologic evaluation by 12 months (sooner if recommended or a change in hearing is suspected)	• Identify sensorineural and/or conductive hearing loss • Referral to otolaryngologist (ear, nose, and throat physician [ENT]) as needed • Collaborative management by otolaryngologist (ENT) and audiologist
Voice and airway	• Assess quality of cry and voice • Assess for stridor	• Seek ENT evaluation of airway or vocal fold anomalies, if needed
Feeding and swallowing	• Referral to SLP to evaluate feeding difficulties, dysphagia or aspiration • Videofluoroscopic swallow study, fiberoptic endoscopic evaluation of swallowing, and/or multidisciplinary feeding evaluation as needed • Refer to 22qDS growth charts (Habel, McGinn, Kackai, Unanue, & McDonald-McGinn, 2012)	• Feeding therapy as indicated • Specialized child-specific bottle/nipple systems based on needs • Tube feeding may be required • Referral to pediatrician or gastroenterologist as required for management of gastroesophageal reflux, or other complex feeding issues

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APPENDIX B

Best Practices for Preschool-Aged Children (1-5 years) with VCFS

Appendix B

Best Practices for Preschool-Aged Children (1–5 years) With 22qDS

This age range is a critical time in the development of communication skills, as most children with 22qDS are already demonstrating significant delay. Velopharyngeal dysfunction (VPD) becomes noticeable, and frequent assessment and therapeutic intervention is of paramount importance.

Domain	Evaluation	Management
Palate	• Cleft palate team to assess for submucous cleft palate (SMCP) and VPD • Imaging studies (nasendoscopy or multiview videofluoroscopy) when SMCP/VPD is suspected and sufficient speech is present for testing • Counseling on risk of VPD with adenoidectomy (consider partial superior adenoidectomy to minimize risk)	• Determine need for VP surgical intervention based on individual profile, imaging results and medical history • Speech therapy to address non-obligatory speech sound errors (e.g., compensatory, phonological, motor speech misarticulations) • Routine re-evaluations with cleft/craniofacial team and 22qDS team where possible
Language	• Comprehensive evaluation of language and social/pragmatic skills • Annual or biannual re-evaluation to assess progress and current needs • Evaluation for autism spectrum disorders as indicated	• 1:1 therapy to address language and communication skills • Group therapy for social skills training • Communication among treating speech-language pathologists (SLPs) and other providers • Utilization of Total Communication principles and therapy early/from time of diagnosis • Provide home program for language goals • Enrollment in structured preschool • Refer to psychologist/developmental pediatrician/neurologist for assessment of development, behavior, cognition, learning
Speech	• Annual or biannual speech evaluation to assess compensatory misarticulations, motor speech and phonological disorders and features of VPD • Assess speech at the word level and in a connected speech sample. • Acoustic measures of speech (voice/resonance) • Evaluations of feeding and swallowing as needed	• Regular, consistent therapy • Intensive therapy is often indicated in the preschool years to establish consonant repertoire and plan for VPD surgery • 1:1 speech therapy to address deficits • Use of specialized speech therapy techniques for compensatory articulation associated with cleft palate/VPD • Utilize principles of motor learning for motor speech deficits • Use of phonological principles for phonological speech disorders • Oral motor exercises are contraindicated • Ongoing feeding management • Communication among treating SLPs and other providers • Provide daily home practice program
Hearing	• Regular hearing test, tympanometry and/or otolaryngology (ear, nose, and throat) [ENT]) evaluation every 6 months if pressure equalization tubes are present	• Ongoing ENT management • Utilization of hearing amplification (FM system, hearing aid) as needed
Sleep apnea	• Screen for obstructive sleep apnea and monitor following VP surgery	• Obtain sleep history, consider polysomnography/referral to sleep center

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APPENDIX C

Best Practices for Early School-Aged Children (6-11 years) with VCFS

Appendix C

Best Practices for Early School-Aged Children (6–11 Years) With 22qDS

Most children are still experiencing significant difficulties in this period. Language difficulties often contribute to learning disabilities. Speech sound disorders are typically resolving, but may persist to some degree. Language and social communication impairments may continue to affect communication skills and school performance. Communication deficits may contribute to social difficulties and reduced self-image. Hypernasality often persists.

Domain	Evaluation	Management
Language and social communication	• Comprehensive evaluation of language and communication skills, including higher level and social/pragmatic language • Testing should consider the impact of communication deficits on school performance and peer relationships • Annual evaluation to chart progress, timing may vary by institution, state or insurance • Speech-language pathologist (SLP) must be vigilant to possible drop in language scores/ performance with referral for further evaluation (particularly if associated with increased anxiety or other changes in mood or behavior)	• Language therapy, as indicated • Specialized educational management/support with modified classroom placement, if needed • Referral to psychologist, neuropsychologist, neurologist, and/or educational specialist for assessment of cognition, learning and development, behavioral health • Collaboration among all providers • Provide home program for language goals • Participation in social skills therapy • Referral to 22qDS team
Hearing	• Hearing testing/tympanometry conducted every 6 months, if pressure equalization tubes are present • Repeat audiograms as needed	• Utilization of preferential seating, hearing amplification (FM auditory system, hearing aid) as needed in classroom • Ongoing otolaryngologic (ear, nose, and throat [ENT]) management
Speech	• Annual or biannual re-evaluation of speech • Assess speech at the word level and in a connected speech sample.	• Continue 1:1 speech therapy to address compensatory misarticulations using specialized cleft palate speech therapy techniques before and after velopharyngeal surgery • Continue therapy for phonological speech disorders • Continue 1:1 therapy for motor speech deficits using principles of motor learning • Provide daily home practice program for speech targets • Consider nasopharyngoscopic biofeedback to target remaining compensatory misarticulations
Palate	• SLP to evaluate for velopharyngeal dysfunction (VPD) • Imaging studies if VPD surgery considered • Post-surgical assessment to determine effectiveness of VPD surgery and speech therapy needs • Annual or biannual cleft palate team visits	• Surgery or, rarely, prosthetic management
Sleep apnea	• Screen for obstructive sleep apnea and monitor post VPD surgery	• Obtain sleep history, consider polysomnography/referral to sleep center

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APPENDIX D

Best Practices for Late School-Aged Children (12-18 years) with VCFS

Appendix D

Best Practices for Late School-Aged Children (12–18 years) With 22qDS

Children in this age range often experience ongoing challenges with language, learning and social communication. Speech disorders are often resolved but may persist to some degree. During this period, children begin preparing for transition to advanced education or to vocational training.

Domain	Evaluation	Management
Language and social communication	• Comprehensive evaluation of core language and higher level communication skills • Re-evaluation every 2–3 years • Monitor for significant changes in cognitive-linguistic functioning, onset of language characteristics consistent with psychiatric conditions, and change in affect/mood and behavior • SLP must be vigilant to possible drop in language scores with referral to mental health practitioner if noted	• Continued collaboration of speech-language pathologists (SLPs) and educational providers on treatment plans • Language therapy as needed, with particular reference to core and higher level language skills, educational targets, life-skills and independence • Participation in social skills therapy • Specialized educational supports, transition services and/or vocational training for individuals with significant learning and/or cognitive deficits • Referral to psychologist, neuropsychologist, neurologist, psychiatrist and/or educational specialist for assessment of cognition, learning and development, behavioral health • Referral to 22qDS team
Speech	• Re-evaluation of speech as needed	• Continue speech therapy as indicated • Consider nasopharyngoscopic biofeedback to target remaining compensatory misarticulations • Provide daily home practice program for speech targets
Palate	• Annual or biennial evaluations with cleft team, although this may vary according to international protocols	• Surgery or, rarely, prosthetic management to improve velopharyngeal closure
Hearing	• Regular audiometric assessments due to ongoing risk of hearing loss into adulthood	• Utilization of preferential seating, hearing amplification (FM auditory system, hearing aid) as needed in classroom • Ongoing otolaryngologic (ear, nose, and throat [ENT]) management
Sleep apnea	• Important to screen for obstructive sleep apnea and monitor post velopharyngeal dysfunction surgery	• Obtain sleep history, consider polysomnography

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