Purpose: Speech and language disorders are hallmark features of 22q11.2 deletion syndrome (22qDS). Learning disabilities, cognitive deficits, palate abnormalities, velopharyngeal dysfunction, behavioral differences, and various medical and psychiatric conditions are also major features of this syndrome. The goal of this document is to summarize the state of the art of current clinical and scientific knowledge regarding 22qDS for speech-language pathologists (SLPs) and provide recommendations for clinical management.

Method: Best practices for management of individuals with 22qDS were developed by consensus of an expert international group of SLPs and researchers with expertise in 22qDS. These care recommendations are based on the authors’ research, clinical experience, and literature review.

Results: This document describes the features of 22qDS as well as evaluation procedures, treatment protocols, and associated management recommendations for SLPs for the often complex communication disorders present in this population.

Conclusion: Early diagnosis and appropriate management of speech-language disorders in 22qDS is essential to optimize outcomes and to minimize the long-term effects of communication impairments. Knowledge of this diagnosis also allows anticipatory care and guidance regarding associated features for families, health care, and educational professionals.

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2q11.2 deletion syndrome (22qDS) is a contiguous gene deletion syndrome resulting in the loss of approximately 50 genes (McDonald-McGinn et al., 2015), leading to aberrant embryonic development, multiple congenital anomalies, and later onset conditions. Speech and language disorders are a hallmark of 22qDS, with the majority of children demonstrating communication delays and/or disorders (D’Antonio, Scherer, Miller, Kalbfleisch, & Bartley, 2001; Golding-Kushner, Weller, & Shprintzen, 1985; Persson, Lohmander, Jönsson, Öskarsdóttir, & Söderpalm, 2003; Persson et al., 2006; Solot et al., 2000, 2001). Learning disabilities, cognitive deficits, behavioral differences, and psychiatric illness are also major features of this
syndrome. Common medical conditions include cardiac, immunologic, endocrine and gastrointestinal disorders (Bassett et al., 2011; McDonald-McGinn et al., 2015).

Speech-language pathologists (SLPs) may be among the first professionals to be consulted in children with 22qDS because of the high prevalence of feeding difficulties, speech-language delays, and disorders in infants and young children. Velopharyngeal dysfunction (VPD) and associated compensatory misarticulations are frequently observed early in development. In other cases, SLPs may be consulted later as children present with communication disorders in the preschool or school-age years. Therefore, it is important for SLPs to be able to recognize and treat individuals with 22qDS at each stage of development.

This document provides a general description of 22qDS, as well as evaluation procedures, treatment protocols, and associated management recommendations for the SLP. It was developed by an international panel of SLPs and researchers with expertise in 22qDS and is based on research and clinical experience as well as review of the literature. Where evidence is not yet available, the authors provide expert consensus. As these recommendations were developed for international application, the author panel acknowledges that some assessments or therapies may be affected by financial or insurance limitations and varying practice patterns regionally, nationally, and internationally.

Overview of 22qDS

Genetics

22qDS is the most common microdeletion syndrome with an estimated prevalence of approximately 1:3,000–6,000 live births (Du Montcel, Mendizabai, Aymé, Lévy, & Philip, 1996; Goodship, Cross, LiLing, & Wren, 1998; Oskarsdóttir, Vujic, & Fasth, 2004) and 1:1,000 fetuses (Grati et al., 2015). Those with milder phenotypic features or atypical nested deletions may not present until later in life or only following the identification of a child with congenital heart disease. As identification improves through prenatal and newborn screening, true incidence figures in the pediatric population may well increase. The vast majority (93%) of cases arise as a de novo event, but once the 22q11.2 deletion is present, there is a 50% recurrence risk in subsequent births (McDonald-McGinn et al., 2015).

There is wide variability in the range and severity of clinical manifestations. Table 1 outlines common findings in children and adults with 22qDS.

The syndrome has been identified in the majority of individuals with DiGeorge syndrome, velocardiofacial syndrome, congenital anomaly face syndrome, Cayler cardiofacial syndrome, and in some with Opitz G/BBB syndrome (McDonald-McGinn et al., 2015). Each syndrome was originally described by clinicians concentrating on specific areas of interest, such as endocrinology with DiGeorge syndrome or speech pathology with velocardiofacial syndrome. Subsequently, the diagnosis is now referred to collectively by the cytogenetic etiology, 22qDS (McDonald-McGinn, LaRossa, et al., 1997).

Facial characteristics that may aid in identification of 22qDS include mild dysmorphic facial features, such as malar or midfacial flatness; hooded eyelids with upslanting palpebral fissures ± epicanthal folds and hypertelorism; auricular anomalies including thick overfolded helices, pre- and postauricular tags/pits, microtia, and anotia; a bulbous nasal tip with hypoplastic alae nasi ± a nasal crease or dimple; and a small mouth and micrognathia.

These features are variable and less frequently identified in non-Caucasians (McDonald-McGinn et al., 2015). Reduced facial affect (Gerdes et al., 1999) and/or animation are often seen as well as asymmetric crying facies (Pasick et al., 2013). For photographic examples of dysmorphic facial features in 22qDS across age and ethnicity, see McDonald-McGinn et al., 2015.

Feeding and Swallowing

Feeding and/or swallowing disorders are common and have been reported in 35%–68% of children (Eicher et al., 2000; Habel et al., 2014; Rommel et al., 1998). Problems include disordered and/or slow feeding, which may be interrupted by gagging or nasal regurgitation. Recurrent vomiting, gastroesophageal reflux disease (GERD), dysmotility, and constipation are also often present (Eicher et al., 2000). Nasal regurgitation tends to improve within the first year. With advancement to chewable foods, gagging or food refusal can develop and may be related to an immature oral transport pattern, sensorimotor dysfunction, and/or GERD. Videofluoroscopic swallow studies have

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

| Finding                        | Children (%) | Adulta (%) |
|--------------------------------|-------------|-------------|
| 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               | 36          | 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.

*Many adult study population sizes are small.
demonstrated pharyngeal hypercontractility, cricopharyngeal prominence, and/or diverticula. These problems are independent of cardiac or palate conditions (Eicher et al., 2000). Some children with more severe dysphagia may require supplemental tube feedings, such as temporary nasogastric or gastrostomy tube placement. Behavioral feeding problems may develop as a consequence of complex feeding issues or may be a manifestation of the behavioral phenotype of the condition (Forsyth & Morrison, 2009). It is important that children with 22qDS with feeding or swallowing disorders be evaluated by an interdisciplinary team of physicians and therapists because of the interrelationship of symptoms and the complexity of management (Bassett et al., 2011).

**Hearing**

Children with 22qDS, with or without obvious palatal anomalies, are at higher risk for otitis media with effusion and eustachian tube dysfunction with resulting conductive hearing loss. Immune deficiency, commonly seen in this population, can lead to more frequent ear infections, persistent middle ear effusion, and subsequent conductive hearing loss as well. While middle ear fluid is the most common cause of a conductive hearing loss, ossicular chain abnormalities have also been reported (Loos et al., 2016). Inner ear malformations in the cochlea and vestibule are also seen (Loos et al., 2016). A meta-analysis of children with 22qDS revealed a prevalence of hearing loss ranging from 6.0% to 60.3% (Verheij, Derks, Stegman, & Thomeer, 2017). Conductive hearing loss occurred in 5.6%–55%, sensorineural hearing loss in up to 19.4%, and mixed hearing loss in up to 28.2%.

**Airway and Voice**

Disorders of the upper and lower airway are often seen in children with 22qDS and include vocal nodules, unilateral vocal fold paralysis, laryngeal web, subglottic stenosis, laryngotracheomalacia, and vascular ring, among others (Dyce et al., 2002; Sacca et al., 2017). Some children with more severe airway conditions may require tracheotomy. Voice disorders, such as decreased loudness, hoarseness, breathiness, tension, vocal fatigue, strained–strangled voice, and high pitch, are relatively common (Solot et al., 2001). Whereas some voice problems are related to the laryngeal conditions described above, compensatory responses to VPD, motor speech disorders, vocal hyperfunction, and GERD also can affect vocal production. Dysphonic characteristics, such as hoarseness, breathiness, and soft volume, may confound the listener’s ability to perceive and/or rate the child’s resonance.

**Cognition and Learning**

Developmental delays in infancy and early childhood are common (Gerdes, Solot, Wang, McDonald-McGinn, & Zackai, 2001; Roizen et al., 2007), with three quarters of preschool children presenting with mild to significant delays in cognitive development, social skills, play and behavior, and almost all children demonstrating delays in gross motor, fine motor, and speech-language skills (Gerdes et al., 2001). Significant differences between performance IQ and verbal IQ (VIQ), almost always favoring VIQ, have been found in preschool children (Gerdes et al., 2001). Hypotonia is also often present, further impacting development (Gerdes et al., 1999).

The neurocognitive profile of school-age children reveals wide variability with full-scale IQ scores ranging from moderately deficient to average, with a significant difference in IQ scales, once again, often favoring VIQ (Swillen & McDonald-McGinn, 2015). VIQ scores do not always predict language skills, with group performance on specific language measures significantly lower than VIQ (Moss et al., 1999; Solot et al., 2001). Furthermore, full-scale IQs may not reflect overall function, as approximately 65% have a > 10-point discrepancy between verbal and performance IQ. Severe intellectual disability is uncommon unless the child has a primary brain malformation (e.g., polymicrogyria) or a secondary insult such as a hypoxic ischemic event, prolonged hypocalcemia, or neonatal seizures (McDonald-McGinn et al., 2015). Longitudinal studies have shown that cognitive development is variable with divergent trajectories. IQ is not necessarily stable across the life span of those with 22qDS; some individuals show cognitive decline, whereas others make progress (Swillen & McDonald-McGinn, 2015; Swillen, Moss, & Duijff, 2018).

Cognitive strengths include rote processing, verbal memory, reading decoding (but not reading comprehension), and spelling (Swillen et al., 1999; Wang, Woodin, Kreps-Falk, & Moss, 2000). Deficits are seen in visual–spatial processing and memory, working memory, arithmetic, executive functioning, attention, abstract thinking, processing new and complex information, and psychosocial functioning (Moss et al., 1999; Swillen et al., 1997).

**Psychological and Psychiatric Disorders**

Children and adolescents with 22qDS may struggle with impulsivity, inattention, and difficulty in social relationships (Gerdes et al., 1999; Swillen et al., 1999). Autism spectrum disorder and subthreshold autistic symptomatology also show increased prevalence ranging from 20% (Fine et al., 2005) to 50% (Vorstman et al., 2006). Using strict autism spectrum disorder diagnostic criteria, a prevalence of 17.9% was found with a majority of children showing some level of social communication impairment (Ousley et al., 2017). Frequently associated conditions include attention-deficit/hyperactivity disorder, anxiety, obsessive compulsive disorder, and later onset psychosis, including schizophrenia, which has been reported in up to 25% of those with 22qDS (Bassett et al., 2005, 2011; Green et al., 2009; McDonald-McGinn et al., 2015; Schneider et al., 2014). Cognitive decline and a decrease in VIQ have been shown to precede the onset of psychosis in some individuals, in addition to symptoms of anxiety or depression (Gothelf et al., 2007; Vorstman et al., 2015). A decline in
scores should alert the clinician to refer the child to behavioral and mental health specialists. Therefore, it is very important that language and cognitive testing continue through the school years.

**Speech and Language Features of 22qDS**

**Language**

The majority of children with 22qDS experience significant receptive and expressive language impairments. Expressive language delay in early childhood often exceeds what would be predicted from cognitive and receptive language levels (Scherer, D’Antonio, & Kalbfleisch, 1999; Solot et al., 2001). Infants often present with absent or reduced babbling. First words may not emerge until after 2 years of age, and phrases may not emerge until after 3 years of age (Solot et al., 2001). The emergence of sentences is also delayed (Swillen et al., 1997). Notably, some children are still nonverbal at the age of 4 years (Solot et al., 2001). These marked delays in language are not correlated with palate function/VPD or cardiac status (Gerdes et al., 2001; Solot et al., 2001). Children often demonstrate a significant increase in expressive output between ages 3 and 5 years (Gerdes et al., 1999; Solot et al., 2001).

Most young children have very limited expressive vocabulary (Scherer et al., 1999). Delays in language use, such as poor responsiveness to simple questions, reliance on nonverbal communication, reduced utterance length, reduced structural complexity, and concrete utterances are common (Golding-Kushner et al., 1985). Of note, early receptive language skills have been shown to be more predictive of intellectual functioning in middle to late childhood than expressive skills, particularly in girls (Roizen et al., 2007).

Language deficits continue into school age, though the receptive–expressive language discrepancy may become less clear-cut in older children (Glaser et al., 2002). Mean scores on language tests in school-age children have been found to be significantly lower than those obtained during the preschool years (Solot et al., 2001). Language difficulties in school-age children include difficulties with reduced grammatical complexity, vocabulary and concept development, abstract reasoning, word finding, and pragmatic language (Golding-Kushner et al., 1985; Persson et al., 2006; Solot et al., 2001). In addition, difficulties are reported with narrative and descriptive language (Persson et al., 2006; Solot et al., 2001). Cross-linguistic, syndrome-specific deficits in perspective and role taking have been found in Dutch- and English-speaking children with conversational skills characterized by poor cohesion and information transfer (Van den Heuvel et al., 2017).

Assessment of receptive and expressive language should be undertaken immediately following a diagnosis of 22qDS. In the preschool years, evaluation of expressive language must take into account the impact of cognition, VPD, and speech sound disorders on verbal output and intelligibility. Assessment of social communication skills should be included routinely as part of clinical assessment at all ages. In school-age children, basic language skills, as well as higher level language functions and pragmatic language, should be assessed. Higher functioning children may do well on tests of basic language skills but have difficulty, for example, with narrative, abstract, figurative, or inferential language. There is a subset of children who do not display early language deficits but may demonstrate them later as linguistic demands increase with age. As skills change over time, reevaluation at regular intervals is particularly important.

There are few evidence-based studies of therapeutic interventions for children with 22qDS. Many of the therapy procedures utilized are based on evidence-based approaches used with children with similar disabilities without 22qDS. For a review of evidence-based interventions with the general population of language delayed 0- to 3-year-olds, see Gladfelter, Wendt, and Subramanian (2011).

Most children with 22qDS do develop oral communication. However, because of significant and persistent early language delay, referral for language therapy is recommended at the time of diagnosis. In the infant and preschool years, intervention should focus on parent counseling, facilitating communicative intent, and social interactions. When verbal output is limited or absent, or compromised by severe speech disorders, a total communication approach is recommended. Use of total communication from the time of diagnosis, even in infancy, can aid development of early communication. Some children may benefit from other augmentative and alternative communication approaches. Where social communication impairment is present with significant expressive language delay, a picture exchange communication system (Bondy & Frost, 1994) may also be utilized.

In school-age children, language intervention should focus on those deficits having the greatest impact upon functional communication, social interaction, and academic performance. Table 2 provides a summary of common language deficits and some suggested management strategies. For additional reading, see Cutler-Landsman (2012) and Reilly and Stedman (2013) for practical guidance for the classroom.

**Speech Sound Disorders**

Delayed emergence of speech with restricted and atypical phoneme repertoires are common findings in this population (D’Antonio et al., 2001; Scherer et al., 1999; Solot et al., 2001). Many children have complex speech disorders characterized by features of any or all of the following: VPD, dysarthria, childhood apraxia of speech, and developmental/phonological disorders (Kummer, Lee, Stutz, Maroney, & Brandt, 2007; Persson et al., 2003; Solot et al., 2001). These speech sound disorders often result in poor speech intelligibility, particularly in younger children (Persson et al., 2003; Rommel et al., 1998; Solot et al., 2000). Although there is progression and improvement in speech in school-age children, speech sound deficits may persist into late childhood and adolescence (Persson et al., 2003;
Speech sound production skills have been shown to be worse in children with 22qDS compared to children with isolated cleft palate (Baylis, Munson, & Moller, 2008; D’Antonio et al., 2001; Kummer et al., 2007; Scherer et al., 1999) or Trisomy 21 (Scherer, D’Antonio, & Rodgers, 2001), suggesting that the presence of palate anomalies/VPD or developmental delay alone does not account for the severity of the speech deficits in 22qDS. A syndrome-specific profile emerges that differs in pattern and severity from non-22qDS children with cleft palate/VPD (D’Antonio et al., 2001; Kummer et al., 2007; Scherer et al., 1999).

Speech sound disorders characterized by developmental errors are often observed (Persson et al., 2003; Solot et al., 2001). Obligatory features of VPD are common and include weak pressure consonants, nasalization of phonemes, and audible nasal air emission during the production of oral pressure consonants. Children frequently demonstrate compensatory misarticulations (D’Antonio et al., 2001; Persson et al., 2003; Solot et al., 2001). Glottal stops, pharyngeal fricatives/stops, nasal fricatives, laryngeal fricatives, and, less frequently, clicks are observed. Features of dysarthria and childhood apraxia of speech have been reported in children and in some adults with 22qDS (Kummer et al., 2007; Mills, Gosling, & Sell, 2006; Persson, Laakso, Edwardsson, Lindblom, & Hartelius, 2017; Solot et al., 2001). Speech motor delay has also been described in some children with delay in speech motor development who do not meet criteria for childhood apraxia of speech or dysarthria (Baylis & Shriberg, 2019). See Table 3 for a summary of speech disorders in children with 22qDS.

**Velopharyngeal Dysfunction (VPD)**

VPD and submucous cleft palate (SMCP), overt and occult, are seen in 67% of children with 22qDS (Campbell et al., 2018). In fact, 22qDS is the most common cause of syndromic palatal anomalies and VPD (McDonald-McGinn, Driscoll, et al., 1997). Overt cleft palate (approximately 11%) and cleft lip and palate (1%–2%) occur less frequently (Bassett et al., 2011; McDonald-McGinn et al., 2015). VPD is caused by structural and/or functional abnormalities of the soft palate and pharynx. VPD should be suspected in all children with 22qDS until sufficient speech has developed to confirm its presence or absence.

Symptoms of VPD may include hypernasality, nasal emission, and resultant obligatory and compensatory misarticulation, as noted above. Syndrome-specific differences in velopharyngeal structure and function in 22qDS include hypoplasia and hypotonia of the velopharyngeal muscles, a wide and/or deep pharynx, platybasia (obtuse anterior cranial base angle), cervical spine abnormalities, reduced tonsil and adenoid volume, asymmetric muscle

| 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) |
function, and cranial nerve abnormalities (Chegar, Tatum, Marrinan, & Shprintzen, 2006; Park, Ahn, Jeong, & Baek, 2015; Ruotolo et al., 2006). The timing of velopharyngeal closure may also be slower or poorly coordinated (Baylis, Watson, & Moller, 2009). In the majority of cases, surgical intervention is required for management of VPD. Because of the complexity and risks of VPD in 22qDS, all children should be referred to a cleft palate–craniofacial or 22qDS team for evaluation and long-term monitoring.

As soon as intentional vocal output emerges, assessment of resonance, nasal emission, voice, and speech should begin and continue at regular intervals. In the first few years of life, the SLP should watch for the emergence of obligatory and compensatory features of VPD and for other types of speech sound errors. Intraoral structural indicators of VPD (e.g., signs of SMCP) should be identified as early as possible. Signs of SMCP include bifid uvula, zona pellucida (translucent central zone secondary to muscle diastasis), and a notch of the posterior border of the hard palate (detectable on digital palpation). However, the absence of these findings does not rule out an occult SMCP or VPD.

SLPs are encouraged to start direct speech sound intervention early, bearing in mind the effects of VPD on consonant production. Early intervention should focus on developing strategies for expressive communication and the prevention or remediation of compensatory articulation patterns. SLPs may also work to expand the phonetic inventory by first targeting vowels, low-pressure or nasal phonemes, and then working up to high-pressure consonants such as oral stops and fricatives.

A variety of approaches to improve speech sound production skills may be considered depending on the type and severity of the speech disorder, the child’s stimulability, and the developmental/cognitive status. If there are frequent compensatory articulation errors, these are usually addressed first with the goal of establishing accurate oral placement using specialized treatment approaches designed for individuals with cleft palate/VPD. Because compensatory productions negatively impact velopharyngeal closure for speech (Henningsson & Isberg, 1986), their remediation can have a substantial positive effect on intelligibility and on the velopharyngeal (VP) function. Furthermore, use of compensatory errors can interfere with the diagnosis and decision making regarding surgical management of VPD.

Strategies such as whispering or producing sustained /h/ before or after oral pressure consonants may be effective in eliminating glottal articulation. Nares compression/nasal occlusion may also facilitate oral consonant production in children with VPD by allowing the child to experience improved intraoral pressure. This maneuver also provides auditory and tactile–kinesthetic pressure cues for practicing target sounds, simulating the effects of successful VP surgery. Anterior oral pressure consonants (e.g., /p, b, f/), which permit visual and tactile–kinesthetic cues, are recommended as early speech sound targets for therapy. Voiceless consonants are also appropriate early targets and often easier to establish than their voiced counterparts in the presence of VPD. It is important to be alert to and remediate compensatory coproductions—where the child simultaneously produces speech sounds with accurate oral place and compensatory production, such as glottal stops. For a review of cleft palate speech therapy techniques, see Golding-Kushner (2001) or Peterson-Falzone, Trost-Cardamone, Karnell, and Hardin-Jones (2017).

Treatment decisions should be individualized based on each child’s speech sound disorder profile using evidence-based treatment approaches. For children with childhood apraxia of speech and/or dysarthria, incorporating motor learning principles is suggested (Caruso & Strand, 1999; Maas et al., 2008; Strand, Stoeckel, & Baas, 2006). It is important to emphasize that, regardless of the type of speech
disorder, there is widespread consensus and evidence that oral–motor exercises are not effective for improving speech in children and are, in fact, contraindicated (Lof & Watson, 2008; McCauley, Strand, Lof, Schooling, & Frymark, 2009; Ruscello, 2008).

It is well recognized that more treatment time translates into better outcomes, especially in the prekindergarten years (American Speech-Language-Hearing Association, 2009). Expert consensus is that speech therapy time/intensity should be maximized for children with 22qDS with significant speech sound disorders. Articulation-focused therapy should begin as soon as the child is able to be engaged in direct articulation instruction, modeling, and imitation tasks. Individual therapy is the preferred approach for articulation treatment, as it has been shown to yield greater measurable functional progress than those who receive group treatment (American Speech-Language-Hearing Association, 2009). The unique and complex speech deficits in 22qDS make this especially important. It is recognized, however, that individual therapy may not be feasible in some settings/regions of the world, and therapy will need to utilize available resources. Clinicians and families must be patient and persistent with the treatment process. Given the combination of attention and learning difficulties and the severity of speech disorders, children need sufficient time for carryover of newly acquired skills to conversational speech.

Surgical Management of VPD

Preoperative Speech Evaluation and VP Imaging

When VPD is suspected and sufficient speech output is present, instrumental evaluation and VP imaging should be completed. Velopharyngeal imaging is primarily used to guide decisions regarding surgical intervention. Acoustic and/or aerodynamic measures of nasality or velopharyngeal function are a helpful adjunct to the perceptual speech assessment. An informal, yet helpful, method for assessing nasal air emission is placement of a mirror under the nares and observing the presence of fogging during repetition of oral pressure consonants.

Direct visualization of the VP mechanism during speech includes videofluoroscopy and nasopharyngoscopy, each providing unique yet complementary information. Variables assessed 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. Clinicians should use a standard speech sample, including repetition of oral-loaded stimuli at the word, phrase, or sentence level. The sample should consist of the child’s maximal attempts to produce oral pressure consonants with accurate placement, even if productions are weak and have accompanying audible nasal emission. A speech sample that consists of only compensatory articulation, such as glottal stops, is insufficient to diagnose VPD. It is often helpful to provide some degree of training for the child prior to these studies in order to optimize performance during the procedure.

The diagnosis of VPD in 22qDS can be challenging. The presence of delayed language or severely disordered speech, as well as behavioral challenges such as anxiety, may delay or, rarely, preclude completion of VPD studies.

Preoperative Medical Evaluation

A comprehensive preoperative workup, including cardiac and airway factors, assessment of cervical spine stability, and carotid artery position, is paramount (Kirschner & Baylis, 2014). Surgical procedures to correct VPD diminish the size of the nasopharyngeal airway and can cause or exacerbate obstructive sleep apnea (OSA). Preoperative polysomnography (sleep study) is recommended to screen for OSA (Kennedy et al., 2014). Tonsillectomy and/or adenoidectomy prior to VPD surgery should be considered to reduce the risk of postoperative OSA or if adenotonsillar hypertrophy interferes with accurate placement of a pharyngeal flap or sphincter pharyngoplasty (Chegar, Shprintzen, Curtis, & Tatum, 2007; Heike et al., 2007). Families should be counseled that adenoidectomy (with or without tonsillectomy) prior to planned VPD surgery may worsen hypernasality and speech during the interim time until the pharyngoplasty can be performed.

Other than in cases when adenoidectomy is performed in preparation for VP surgery, it is prudent to avoid adenoidectomy in children with 22qDS as it can induce or exacerbate VPD (Perkins, Sie, & Gray, 2000) by unmasking a previously unrecognized SMCP and/or VPD. If adenoidectomy is required to treat recurrent ear/nasal infections or severe airway obstruction, a “partial” or “superior” adenoidectomy should be considered.

Surgical and Nonsurgical Management Approaches

Because VPD and speech disorders in 22qDS are often severe and multifactorial, management of VPD in this population is more complex than in other cleft/VPD populations (Kirschner & Baylis, 2014; Nayak & Sell, 1998). There is ongoing debate regarding the optimal surgical approach for individuals with 22qDS and VPD (Mehendale, Birch, Birkett, Sell, & Sommerlad, 2004; Spruijt et al., 2012). In some instances, more than one surgical procedure may be required (Losken, Williams, Burstein, Malick, & Riski, 2003; Pryor et al., 2006; Witt, Cohen, Grames, & Marsh, 1999).

Surgical management may be with a single procedure or a two-staged approach (Kirschner & Baylis, 2014; Mehendale et al., 2004; Sie et al., 1998; Witt et al., 1999). Palatoplasty is performed for overt cleft palate and for SMCP. However, palate repair alone for SMCP is less likely to achieve VP closure, and rates of secondary surgery for persistent VPD are higher in this population (Bezuhl, Fischbach, Klaiman, & Fisher, 2012). VPD is most often managed by posterior pharyngeal flap (Kirschner & Baylis, 2014) or sphincter pharyngoplasty (Sie et al., 1998; Witt et al., 1999). Choice of technique is surgeon specific, taking into account the individual’s structural and neuromuscular characteristics visualized on preoperative imaging.
Timing and candidacy for VPD management is a complex decision involving the patient/family, the SLP, and the surgeon and should take into account medical, speech-language, developmental, and psychosocial factors. Optimal presurgical decision making requires that the child have sufficient speech for the evaluation of VP structure and function as well as the maturity to comply with assessment procedures. For this reason and perioperative airway concerns, many 22qDS centers recommend waiting until at least 4 years of age to perform VPD surgery.

Nonsurgical options, such as prosthetic management (e.g., palatal lift or speech bulb/obturator), may need to be considered in a small number of individuals with VPD who are not surgical candidates. Regardless of the selected management approach, treatment decision making and planning is best conducted in an interdisciplinary setting such as a cleft palate–craniofacial and/or 22qDS team with knowledge and experience with this syndrome and VPD.

**Postoperative Considerations**

Airway symptoms should be monitored closely. Because there is a higher prevalence of OSA in this population and because VPD surgery reduces airway size, some centers routinely perform postoperative sleep studies (Kennedy et al., 2014). At 6–12 months postsurgery, evaluation should include the SLP’s assessment of speech outcome, speech therapy needs, and instrumental assessment (e.g., nasometry). Protocols for postoperative videofluoroscopy and/or nasopharyngoscopy vary. Some centers perform these studies routinely, whereas other centers do so only when there are concerns about persistent VPD at a significant interval after surgery (e.g., >1 year). In some cases, speech outcome may not be fully optimized until several years after surgery (Spruijt et al., 2012), as these children may have slower adaptation to a new VP mechanism as a function of motor deficits, cognitive difficulties, and other factors. Possible slower adaptation is important when considering the need for revision surgery and emphasizes the need for long-term follow-up in this population. In addition, resonance can worsen with facial growth and/or adenoid involution. Follow-up speech evaluations by the cleft team should be conducted as needed until facial growth is complete, beyond the age of puberty.

Individuals with 22qDS are recognized as a particularly difficult group in which to achieve optimal speech outcomes. Data suggest that the more severe the preoperative hypernasality, the more likely there may still be some degree of residual hypernasality or audible nasal emission after surgery (Losken, Williams, Burstein, Malick, & Riski, 2006; Mehendale et al., 2004). Hyponasality following VPD surgery can also be seen in some individuals. 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. See Appendices A–D for a summary of best practices for assessment and management of speech and language by specific age groups.

**Communication Disorders in Adults With 22qDS**

Few studies have focused on speech and language in adults with 22qDS; consequently, our knowledge of the communication profile of this group is limited. In one report of adults with 22qDS, the older they were at the time of assessment, the more noncongenital features were found, indicating an ongoing emergence of conditions that may affect communication skills (Bassett et al., 2005). Examples of this include early onset Parkinson’s disease (Butcher et al., 2013; Zaleski et al., 2009), schizophrenia, and anxiety disorders (Philip & Bassett, 2011). Multiple studies have found cerebral alterations in adults with 22qDS (Gothelf, Schaer, & Eliez, 2008; Shprintzen, 2008). In addition, the prevalence of hearing loss has been estimated to be between 30% and 40% (Bassett et al., 2005; Persson, Friman, Óskarsdóttir, & Jónsson, 2012).

Residual VPD of varying degrees has been reported in two thirds of adults, but with only 8% of the sample with persistent articulation errors (Persson et al., 2012). Problems with respiration, phonation, and oral–motor function with signs of motor speech disorders have also been reported (Persson et al., 2017). Together with the findings of structural central nervous system malformations and the suggested early onset of progressive neurodegenerative disorders, it is reasonable to suspect that, at least, some adults will present with residual speech deficits or, possibly, deteriorating speech.

The highly complex nature of speech disorders, language impairment, and cognitive–behavioral difficulties, together with the variable availability of therapeutic resources, all contribute to ongoing speech and language difficulties across the life span. Further investigations in larger samples are needed to determine if findings to date are representative of the broader adult 22qDS population. Long-term speech outcome studies are also needed to assess the effectiveness of surgical intervention and speech therapy. See Appendix E for a summary of best practices for adults.

**Future Directions**

An ongoing challenge facing health care and educational providers is the phenotypic heterogeneity of 22qDS and how to best individualize care for each affected person. The spectrum of 22qDS is wide, making it difficult to make long-term predictions of function. Robust longitudinal data from infancy through adulthood is one of the highest research priorities. Existing research in 22qDS has highlighted a complex communication profile encompassing structural, neurological, and developmental speech and language disorders and social communication deficits. Interdisciplinary research in communication disorders in 22qDS has primarily been directed to the management of...
VPD and associated speech sequelae. A greater understanding of the linguistic and social communication difficulties is also greatly needed. Collaborative research among SLPs, audiologists, surgeons, psychologists, and psychiatrists will be helpful in gaining a better understanding of the interaction of the deficits. Studies of the efficacy and effectiveness of speech-language interventions are needed to help practitioners provide appropriate and effective management.

Summary

22qDS is a common genetic condition, and speech-language disorders are one of its most prevalent features. SLPs can help identify affected individuals and are instrumental in the management and health surveillance of those with 22qDS. Knowledge of 22qDS will allow the SLP to provide syndrome-specific care across the life span. SLPs can serve as liaisons to other health care professionals and educators working with children with 22qDS, as well as initiate referrals to appropriate medical, educational, and behavioral health care providers. The combination of persistent language deficits, social communication impairment, speech sound and motor speech disorders, VPD, and cognitive and behavioral disorders makes the communicative profile of this syndrome unique and treatment especially challenging. The care pathways and best practices summarized here are intended to inform and assist SLPs about 22qDS in the formulation of the most appropriate treatment plans based on currently available literature and expert opinion. Application of these recommendations must be made by each clinician, taking into account the individual’s clinical needs, health care and educational systems, and available family and community resources. As additional research becomes available, current practices may change to reflect advances in understanding this complex syndrome.

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**Appendix A**

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

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 | • Monitoring by Cleft Palate Team and 22qDS specialty clinic where possible |
|                             | • Referral to Cleft Palate Team                                          |                                                                            |
| **Speech-language development** | • Referral to speech-language pathologist (SLP)                         | • Assess for risk of speech/language disorder or velopharyngeal dysfunction |
|                             | • 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) | • Parent education for speech and language stimulation                     |
|                             | • Referral to speech-language pathologist (SLP)                         | • Educate parents to recognize emergent communicative acts/early speech attempts |
|                             | • 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) | • Referral to parent support networks                                      |
|                             | • Referral to speech-language pathologist (SLP)                         | • Therapy for early intervention strategies                                 |
|                             | • 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) | • Implement Total Communication early, at time of diagnosis                 |
|                             | • Audiologic evaluation by 12 months (sooner if recommended or a change in hearing is suspected) | • Frequent re-evaluation to monitor progress and adapt therapy plans        |
| **Hearing**                 | • Newborn hearing screening                                              | • Identify sensorineural and/or conductive hearing loss                     |
|                             | • Audiologic evaluation by 12 months (sooner if recommended or a change in hearing is suspected) | • Referral to otolaryngologist (ear, nose, and throat physician [ENT]) as needed |
| **Voice and airway**        | • Assess quality of cry and voice                                        | • Collaborative management by otolaryngologist (ENT) and audiologist       |
|                             | • Assess for stridor                                                     |                                                                            |
| **Feeding and swallowing**  | • Referral to SLP to evaluate feeding difficulties, dysphagia or aspiration | • Seek ENT evaluation of airway or vocal fold anomalies, if needed         |
|                             | • Videofluoroscopic swallow study, fiberoptic endoscopic evaluation of swallowing, and/or multidisciplinary feeding evaluation as needed | • Feeding therapy as indicated                                              |
|                             | • Refer to 22qDS growth charts (Habel, McGinn, Kackai, Uhanue, & McDonald-McGinn, 2012) | • Specialized child-specific bottle/nipple systems based on needs            |
|                             | • Videofluoroscopic swallow study, fiberoptic endoscopic evaluation of swallowing, and/or multidisciplinary feeding evaluation as needed | • Tube feeding may be required                                              |
|                             | • Refer to SLP to evaluate feeding difficulties, dysphagia or aspiration | • Referral to pediatrician or gastroenterologist as required for management of gastroesophageal reflux, or other complex feeding issues |
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     | • Determine need for VP surgical intervention based on individual profile, imaging results and medical history               |
|          | • Imaging studies (nasendoscopy or multiview videofluoroscopy) when SMCP/VPD is suspected and sufficient speech is present for testing | • Speech therapy to address non-obligatory speech sound errors (e.g., compensatory, phonological, motor speech misarticulations) |
|          | • Counseling on risk of VPD with adenoidectomy (consider partial superior adenoidectomy to minimize risk)                      | • Routine re-evaluations with cleft/craniofacial team and 22qDS team where possible                                           |
| Language | • Comprehensive evaluation of language and social/pragmatic skills           | • 1:1 therapy to address language and communication skills                                                                   |
|          | • Annual or biannual re-evaluation to assess progress and current needs     | • Group therapy for social skills training                                                                                   |
|          | • Evaluation for autism spectrum disorders as indicated                     | • Communication among treating speech-language pathologists (SLPs) and other providers                                       |
|          | • Assess speech at the word level and in a connected speech sample.         | • Utilization of Total Communication principles and therapy early/from time of diagnosis                                     |
|          | • Acoustic measures of speech (voice/resonance)                            | • Provide home program for language goals                                                                                   |
|          | • Evaluations of feeding and swallowing as needed                          | • 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 | • Regular, consistent therapy                                                                                               |
|          | • Assess speech at the word level and in a connected speech sample.         | • Intensive therapy is often indicated in the preschool years to establish consonant repertoire and plan for VPD surgery   |
|          | • Acoustic measures of speech (voice/resonance)                            | • 1:1 speech therapy to address deficits                                                                                     |
|          | • Evaluations of feeding and swallowing as needed                          | • 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 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. | • 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 |
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             | Comprehensive evaluation of core language and higher level communication skills | Continued collaboration of speech-language pathologists (SLPs) and educational providers on treatment plans |
| communication                   | Re-evaluation every 2–3 years                                               | Language therapy as needed, with particular reference to core and higher level language skills, educational targets, life-skills and independence |
|                                 | Monitor for significant changes in cognitive-linguistic functioning, onset of language characteristics consistent with psychiatric conditions, and change in affect/mood and behavior | Participation in social skills therapy |
|                                 | SLP must be vigilant to possible drop in language scores with referral to mental health practitioner if noted | 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 |
Appendix E
Best Practices for Adults With 22qDS

VPD and speech-language concerns may have a life-long effect on speech and resonance. Mild speech differences may be present, such as abnormal articulation, problems with prosody, resonance and voice disturbances. The highly complex nature of the speech mechanism together with cognition and behavior all contribute to ongoing speech-language difficulties. Lack of necessary therapeutic resources during childhood may also affect the speech-language profile in adults. Late onset deficits may emerge in adulthood.

The adult should play a central role in decisions regarding the desired treatment plan.

- Adults should be evaluated for persistence of symptoms of velopharyngeal dysfunction (VPD).
- Surgical or prosthetic management for VPD can be performed, but, typically, is less effective than in childhood.
- Speech therapy may have some limited effect on intelligibility and acceptability, depending on the type and severity of speech disorder and other factors (e.g., cognitive level).
- If emergence of new speech difficulties, (progressive) neuromuscular disorders should be considered.
- Hearing testing should be performed when there is known hearing loss, or suspicion of change in hearing or onset of new hearing loss.
- Monitoring for potential changes in language that may signal cognitive decline or psychosis
- Monitoring for obstructive sleep apnea
- Referral to psychologist, neuropsychologist, neurologist, and/or psychiatrist for assessment of cognition, learning and development, behavioral health
- Referral to 22qDS team where possible, in particular for recurrence risk counseling