Part I

Developmental communication disorders

1

Cleft lip and palate and other craniofacial anomalies

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1.1 Introduction

Despite reports from the Centers for Disease Control and Prevention (2006) that cleft lip/palate is the most commonly reported birth defect, clefting remains a low incident disorder. Because it is low incident, there is often little impetus to include velopharyngeal function/dysfunction in educational programmes for speech-language pathologists. There are also limited numbers of patients and clinicians who can provide clinical expertise and training to students and practising clinicians. However, clefting can have a devastating impact on a newborn's ability to feed. Unrepaired or unsuccessfully repaired cleft palate can have a devastating effect on speech development and intelligibility, often preventing successful integration of the affected individual into society.

Velopharyngeal incompetence (VPI) can result from a number of congenital craniofacial anomalies and associated genetic disorders. VPI can also result from acquired neurological disorders such as stroke, head injury and neurological diseases. Also, it can result from ablative surgery from adenoidectomy and head and neck cancer. This chapter will focus on cleft lip and palate. However, understanding principles of evaluating velopharyngeal dysfunction and its treatments can serve clinicians faced with evaluating and treating patients with velopharyngeal dysfunction from other causes.

A cleft lip develops when the prolabium fails to fuse with the lateral lip segments (see Figure 1.1). A cleft palate develops when the palatal segments fail to fuse with the septum in the midline (see Figure 1.2). Clefts of the lip can be unilateral or bilateral. They can affect the lip, the palate or both.

A cleft palate can disrupt the palatal muscles that are responsible for elevation of the soft palate or velum. The levator muscle of the velum

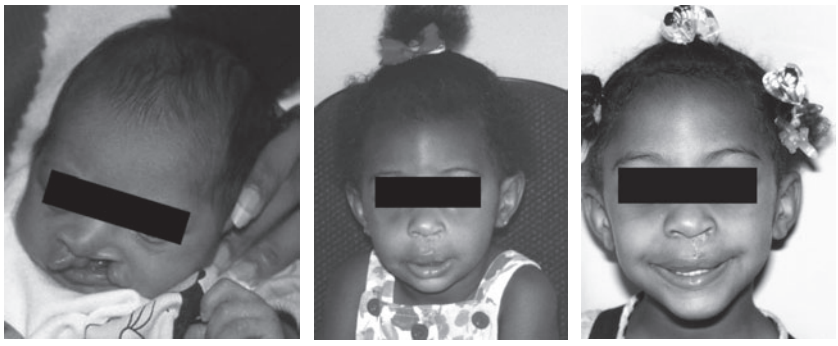

Figure 1.1 Children with cleft lip, unrepaired (left panel) and repaired (middle and right panel). (Permission granted for educational purposes.)

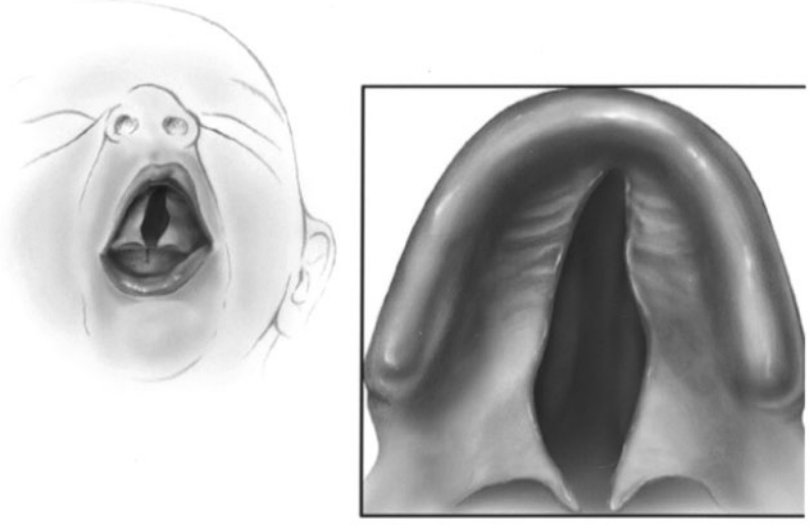

Figure 1.2 Drawing of midline cleft palate. (© 2012 Children's Healthcare of Atlanta, Inc. All rights reserved.)

functions to couple and separate the nasal cavities from the vocal tract during speech (see Figure 1.3). This function determines whether the voiced elements of speech have oral resonance or nasal resonance. It also helps to determine whether oral pressure can be impounded in the oral cavity for plosive, fricative and affricate sounds. Velopharyngeal incompetence or dysfunction of the soft palate often leaves speech unintelligible and hypernasal, lacking in aspiration of pressure consonant sounds.

A challenge to evaluation is that the aetiology of hypernasality and nasal airflow disorders is often occult or hidden. In reviews of patients receiving surgical correction for hypernasality, approximately 30 per cent did not have a cleft palate (Riski *et al.*1992; Riski 1995). The aetiology in these children was an anatomically deep nasopharynx that can only be

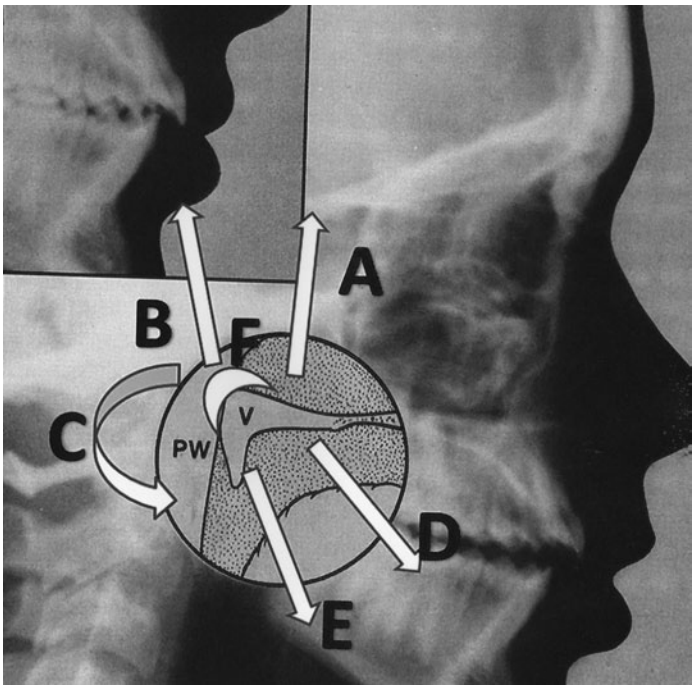

Figure 1.3 Muscles of the velum. The arrows show approximate direction of muscular attachment and direction of movement. A, tensor palatini; B, levator palatini; C, superior constrictor; D, palatoglossus; E, palatopharyngeus; F, musculus uvula; PW, posterior wall of the pharynx; V, velum.

diagnosed accurately by radiographic assessment. Normal velopharyngeal dimensions were described by Subtelny (1957) and highlighted by Zemlin (1997). Also, the anatomical defect of a disproportionately deep pharynx was described by Calnan (1971).

A mistaken concept in speech pathology is that non-cleft hypernasality is erroneously labelled as a 'voice disorder'. Labelling hypernasality a 'voice disorder' is ambiguous (it implies it is a disorder of the larynx), and often hampers successful management of the disorder. Because the physical defect is not recognized, speech therapies are often poorly designed and ineffective (Ruscello 2004). A cleft palate is identified prenatally or perinatally and palate closure is before one year of age (Riski 1995). In stark contrast, the average age of referral to our centre for children with non-cleft hypernasality resulting from 22q11.2 deletion (velocardiofacial syndrome or VCFS) was 9.2 years of age. Children with hypernasality resulting from cleft palate are generally referred to a craniofacial team. In contrast, children with hypernasality with no obvious form of clefting are referred to an ENT specialist, generally in a private office, who may or may not have experience of evaluating and managing velopharyngeal dysfunction. Delayed management of VPI leads to increased failure of surgical intervention and refractory speech deficits. The rate of complete

success from surgery when VPI is managed before 6 years of age is 90.9%. The success rate falls to 73.9% between 6 and 12 years, 70.0% between 12 and 18 years, and 47.0% after 18 years (Riski *et al.*1992).

The evaluation of oral clefts and hypernasality is conducted by specialists in craniofacial clinics. However, in the United States, Public Laws 94-142 and 99-457, which culminated in the Individuals with Disability Education Act (IDEA, 1990, reauthorized in 1997 and 2004), mandated that special services, such as speech therapy, be provided through specialists in schools and developmental centres. The professionals in these settings usually have limited experience of cleft-related problems because these problems typically form a very small part of their caseload. This separation of evaluation and therapy can lead to poor communication between professionals and therapy plans that do not directly address the therapy needs of the patient. There is an unquestionable need for partnerships between evaluation centres and the settings in which therapy is conducted.

1.2 Epidemiology and aetiology of cleft lip and palate

In the United States, the Centers for Disease Control and Prevention (CDC) conducts a surveillance study of birth defects in 14 states. It is estimated that each year 2,651 infants are born with a cleft palate and 4,437 infants are born with a cleft lip with or without a cleft palate (Parker *et al.* 2010). The incidence and prevalence of clefting vary with epidemiological study. Bister *et al.* (2011) reported the incidence of facial clefts to be 0.127 per cent in a British population. Prevalence rates between 0.97 per 1,000 live births (Golalipour *et al.* 2007) and 1.47 per 1,000 live births (Gregg *et al.* 2008) have been reported. The condition is more prevalent in males than in females. Golalipour *et al.* (2007) reported the prevalence of oral clefting to be 1.08 per 1,000 male births and 0.86 per 1,000 female births. Prevalence rates also vary with the ethnicity of populations. Among Asian populations, Cooper *et al.* (2006) reported the prevalence rate of syndromic plus non-syndromic cleft lip with or without cleft palate to be 1.30 per 1,000 live births (Chinese), 1.34 per 1,000 (Japanese) and 1.47 per 1,000 (Other Asian). Compared to Caucasians, the prevalence of cleft lip with or without cleft palate is lower among Africans, higher among Native Americans and the same among Japanese and Chinese (Croen *et al.* 1998).

The distribution of different types of oral clefts has been examined in several studies. In an investigation of 835 cases, González *et al.* (2008) reported cleft lip and palate in 70% of cases, cleft palate in 21%, cleft lip in 8% and separate cleft lip and palate in 1%. Gregg *et al.* (2008) found a significant left-sided predilection for unilateral clefting of the lip. The aetiology of clefting is still uncertain. Studies from the CDC found that

women who smoke are more likely to have a baby with an orofacial cleft than those who do not smoke (Little *et al.* 2004; Honein *et al.* 2007). Women who have diabetes are at increased risk of having a child with a cleft lip with or without a cleft palate (Correa *et al.* 2008).

Orofacial clefts can sometimes be diagnosed during pregnancy, usually by a routine ultrasound as early as 17 weeks. Riski (2006) found that 50% of families had a prenatal diagnosis of cleft by ultrasound, while Bister *et al.* (2011) reported that 65% of clefts were detected by antenatal ultrasound screening. Isolated clefts of the palate might not be identified until the perinatal period. Submucous cleft palate and bifid uvula might not be diagnosed until later in life. Bifid uvula has been identified in 2.26% of school-aged children and often occurs without any other palatal involvement (Wharton and Mower 1992). However, a bifid uvula highlights the need for a complete assessment of the velopharyngeal mechanism if there is any nasal regurgitation or hypernasality or if an adenoidectomy is planned. There are also children born with VPI without an observable cleft. These children have a deep nasopharynx identified only by lateral cephalometric radiographs (Calnan 1971; Riski *et al.* 1992; Riski 1995).

1.3 Effects of cleft palate

1.3.1 Feeding

An open cleft palate is a detriment to feeding in a newborn and can compromise nutrition. Furthermore, newborns with micrognathia, such as in Pierre Robin syndrome, will also have a compromised airway that complicates the normal suck–swallow–breathe coordination required for successful feeding. Craniofacial clinics should incorporate feeding specialists to evaluate and treat feeding problems found in newborns. The specialists should include speech-language pathologists who specialize in feeding of newborns, nutritionists, lactation consultants and nurses. They will first establish that the child has an adequate airway. There should be no sternal retractions or rapid respiratory rate. The effectiveness of feeding is established by measuring the volume of formula taken within a specific timeframe. Newborns with isolated cleft lip(s) might be able to breast-feed successfully if they have adequate tongue protrusion under the nipple to gain suction. Newborns with a cleft palate are difficult to breast-feed unless the mother hyperlactates and milk flow is rapid and requires little/no suction or compression. Failure to provide adequate nutrition to the newborn can lead to failure to thrive and more aggressive feeding options such as nasogastric tubes or gastrostomy tubes.

For all infants, feeding specialists will help ensure that the child is in a mostly upright position and will assess feeding effectiveness with various bottles and nipples. There are many specialty nursers and nipples available. Commonly used specialty bottles are the Mead Johnson Cleft

Palate Nurser, the Haberman Bottle and the Pigeon Bottle. The nutritionist is vital in documenting weight gain and recommending formulas that can increase the nutritional value of each feeding. The Cleft Palate Foundation (2012) has produced an instructional video for feeding newborns with cleft palate.

1.3.2 Articulation and resonance

Speech and resonance are affected by velopharyngeal incompetence, dental arch malformations and hearing loss. We will discuss some of the common articulation and resonance qualities associated with velopharyngeal function/dysfunction and discuss treatment of these anomalies in section 1.6.

Oral pressure is required for the production of stop-plosive, fricative and affricate sounds. Some languages have trills such as the trilled Spanish 'rr' that also requires oral pressure. Nasal pressure loss through an oral-nasal fistula or VPI can undermine that pressure. The lack of pressure for these sounds can render speech unintelligible. Typical compensations are the use of nasal substitution (e.g. [m] for /b/) or the development of maladaptive articulation errors such as use of the glottal stop or pharyngeal fricative.

Nasal air emission is the quality of non-acoustic sounds and is mostly easily perceived on unvoiced consonants. This quality results when the speaker is attempting oral pressure but it is leaking through a fistula or VPI. Nasal air emission may be inaudible in patients with patent nasal cavities when the air passes through the nasal cavity without creating any audible turbulence. The sound of posterior nasal frication (nasal air leak) in conjunction with oral airflow generally represents touch velopharyngeal contact. Velopharyngeal closing force is not maintained and the air leak through the port creates the posterior nasal frication. There is airflow simultaneously through the oral and nasal cavities.

Oral-nasal resonance is the balance of oral and nasal acoustic (voiced) energies. It is achieved by the appropriate coupling and isolation of the nasal cavities from the remainder of the vocal tract during speech by the movements of the velopharyngeal valve. Three English sounds require the nasal cavities to be coupled with the vocal tract (i.e. /m/, /n/, /ŋ/). All other sounds require the velopharyngeal valve to isolate the nasal cavities from the vocal tract. Hypernasality is the quality perceived by the listener when there is inappropriate nasal coupling with the vocal tract during speech. It is mostly easily perceived on vowel sounds. In contrast, hyponasality is perceived as inadequate coupling or obstruction of the nasal tract during production of those sounds normally associated with nasal energy. The obstruction may be posterior (e.g. hypertrophied adenoids) or anterior (e.g. hypertrophied turbinates, deviated septum). A speaker may also demonstrate mixed hyper-hyponasality when

velopharyngeal closure is incomplete, but the nasal cavity is occluded anteriorly.

Compensatory articulations can develop in response to VPI or dental arch malformations. These articulations not only reduce the intelligibility of speech but have been found to be related to delays in language development in cleft patients (Pamplona *et al.* 2000). The classic compensatory misarticulations of glottal stops and pharyngeal fricatives have been expanded by Trost (1981) to include pharyngeal and midpalatal stops. In a pharyngeal stop, the point of stop is the tongue base to the posterior pharyngeal wall. This articulation is used as a substitution for /k/ and /g/. In a midpalatal stop, the point of stop is midpalate between the position of /t/ and /k/. This articulation is used as a substitution for /t, d, k, or g/. Other compensatory articulations include the use of clicks (Gibbon *et al.* 2008).

A unique, maladaptive articulation is the posterior nasal fricative. It is typically used as a substitution for sibilant and sometimes affricate and fricative sounds. During production of this fricative, the tongue is used to obstruct oral airflow. The airflow is forced through the constricted velopharyngeal valve which creates frication. There is no oral airflow. This is often seen in children without any cleft. Riski (1984) attributed this compensation to conductive hearing loss. Whereas the sibilant sound /s/ is one of the softest sounds that we produce at 20 dB, the posterior nasal fricative creates a relatively loud bone-conducted signal that bypasses fluid. We might also consider the anterior nasal fricative. This is similar to the posterior nasal fricative, but the point of frication is the anterior nostrils. Nasal grimacing may accompany this substitution.

The unique nature of compensatory or maladaptive articulation errors has led to speculation as to why these develop. The point of articulation is universally below the VPI and typically occurs at the larynx. Morr *et al.* (1988) speculated that these occur in response to certain respiratory receptors in the trachea, larynx and nasopharynx and operate to regulate vocal tract pressures and resistance during speech. In contrast, Netsell (1990) suggested that compensatory articulation is an attempt to generate acoustic distinctions that cannot be produced above the VPI. Additional study of speech compensations to VPI may provide greater insights into speech motor control.

1.3.3 Language

For some time, we have known that there are differences in language and reading abilities between children with isolated cleft palate (CPO) and children with cleft lip, with or without cleft palate (CLP). These differences were first brought to light by Richman and Eliason (1984). They reported significant differences on language measures, reading comprehension and type of reading errors between children with CLP and CPO. Their results suggested that children with CPO constitute a language

disordered group with more severe reading disabilities. In contrast, they found that children with CLP were more likely to have verbal expressive deficits and milder reading problems.

In a follow-up investigation with a larger sample, Richman *et al.* (1988) reported that approximately 35% of students with cleft displayed a moderate degree of reading disability and 17% exhibited severe reading disabilities. Younger children were more likely to have reading disability than older children. However, when the two groups (CLP vs. CPO) were compared, older children with CLP had an incidence of reading disabilities similar to the general population or 9%. In contrast, the incidence of reading disabilities in children with CPO was 33%. They found that there were no differences in gender in the prevalence of reading disability.

Additional research by Broder *et al.* (1998) found that 46% of children with cleft had a learning disability, 47% had deficient educational progress and 27% had repeated a grade (excluding kindergarten). Males with CPO had a significantly higher rate of learning disability than other subject groups. Males with CPO and females with CLP were more likely to repeat a grade in school than were females with CPO and males with CLP.

1.4 Evaluation of velopharyngeal function and articulation

1.4.1 General considerations

The task of evaluating velopharyngeal function may be approached as a multi-level problem. The first level should include the perceptual assessment of resonance, nasal air escape and articulation. The trained ear is still the 'gold standard' of the evaluation. Resonance should be neither hypernasal nor hyponasal. The second level is the screening of velopharyngeal closure. This step uses inexpensive tools and is under-utilized. Patients who fail these two steps should undergo the third step of objective assessment with computerized instruments for voiced and unvoiced speech components. Finally, imaging should account for the three-dimensional nature of the velopharyngeal port and should include some combination of direct visualization using flexible fibreoptic nasendoscopy, radiography or fluoroscopy during speech.

Velopharyngeal function impacts speech proficiency. However, speech proficiency is not an adequate measure of velopharyngeal function (Riski 1979). It is possible to have severely defective speech and a competent velopharyngeal mechanism. However, normal speech usually cannot be produced without a competent velopharyngeal mechanism. Articulation should be evaluated separately with special attention to any compensatory misarticulations and the age-appropriateness of articulation. Also, longitudinal study of velopharyngeal port function has demonstrated its instability in children as their phonological system develops and