
Craniofacial anomalies are anatomical deviations that can affect the oral and facial structures, the cranium, or both. They are often complex and may occur as a feature of a particular syndrome (1). This Communication Facts addresses select craniofacial syndromes that speech-language pathologists and audiologists may encounter in their clinical practice.

Orofacial Clefts

A cleft is an elongated opening, especially one resulting from the failure of parts to fuse or merge early in prenatal development (2).

Demographics

- Orofacial clefts are one of the most common congenital anomalies. One case of orofacial cleft occurs in approximately every 500 to 550 births (3).
- The majority of orofacial clefts are of multifactorial origin and result from the interaction of genetic and environmental factors. Between 20% and 30% of all cleft lip (with or without cleft palate) are associated with a broader pattern of malformations (4, 5).
- The estimated average lifetime medical cost per orofacial cleft is $100,000 per child, amounting to $750 million for all such children born within one year (3).

Communication Manifestations

- The primary communication difficulties affecting individuals with cleft lip or palate relate to language, speech, resonance, and voice production, as well as to hearing; however, mental development is expected to follow the distribution seen in the normal population (6).
- Language abilities, particularly expressive language, may be delayed in children with cleft palate (1).
- Babbling patterns of children with cleft palate reveal delay and differences from the normal pattern of development. Deviant phonetic patterns may persist in the speech of older children with cleft palate (1).
- Middle ear disease is present at birth in most infants with cleft palate, but there is probably no increased risk when only cleft lip is present (2).
- Sensorineural hearing loss has been reported among a small proportion of individuals with cleft palate; however, conductive hearing loss is much more prevalent with cleft palate and cleft lip (6).
**Apert Syndrome (Acrocephalosyndactyly Type 1)**

Apert syndrome is a genetic disorder. It can be inherited or it may occur without a known family history. It is characterized by premature closure of the seams between the skull bones, which results in a peaked head and an unusual facial appearance (7).

**Demographics**

- The incidence of Apert syndrome is estimated to be 1 in 80,000 individuals (8, 9).
- According to one study, advanced paternal age is a factor in most documented sporadic cases of Apert syndrome (8).

**Communication Manifestations**

- There are few studies that report findings on the speech and language characteristics of Apert syndrome, and little is known about the cognitive profile of the syndrome (6, 10).
- Cleft palate may occur in 30% of individuals with Apert syndrome (8).
- There are some indications that many individuals with this syndrome display a conductive hearing loss of sufficient magnitude to interfere with verbal communication (6).
- Fixation of the middle ear bones, as well as other middle ear anomalies resulting in a conductive hearing loss, have been observed among this population (6).

**Crouzon Syndrome**

Crouzon syndrome is a genetic disorder characterized by the premature joining of certain bones of the skull, which affects the shape of the head and face (11).

**Demographics**

- Crouzon syndrome is estimated to occur in 15 to 16 individuals per million births (11).
- Crouzon syndrome constitutes approximately 4.8% of cases where there is a premature joining of certain bones of the skull (12).

**Communication Manifestations**

- The major communication difficulties in Crouzon syndrome lie in the degree of palatal involvement, the severity of the oral cavity misalignment, and the type and degree of hearing loss (6).
- Anomalies of the palate include lateral palatal swellings in 50% of cases, but cleft lip/palate is not common (8).
• It is estimated that 30% to 55% of patients with Crouzon syndrome have hearing loss. The hearing loss is usually conductive in nature (8, 13) and may range from mild to moderately severe (6).

• Acquired ear diseases may be similar to those typically observed among individuals with cleft palate or anomalies in the growth patterns of the skull (6).

Fetal Alcohol Syndrome

Maternal alcohol abuse during pregnancy is associated with several birth defects. A pattern of these defects consisting of prenatal and/or postnatal growth retardation, craniofacial anomalies, and nervous system defects is called fetal alcohol syndrome (F.A.S.) (14, 15).

Demographics

• Estimates on the incidence of F.A.S. vary from 0.2 to 1 per 1,000 live-born infants (15, 16).

• Differences on reported estimates have also been noted among racial/ethnic populations (15).

Communication Manifestations

• Disorders of speech production associated with the syndrome include deficits in fluency, lack of intonation, voice dysfunctions, slurred speech, and poor articulation (14).

• Language acquisition and comprehension are influenced by both hearing and cognitive functions. Verbal learning and memory deficits have been found in children with F.A.S. (14).

• Expressive and receptive language delays are present in 86% of patients with F.A.S. (17).

• The act of involuntarily imitating words and phrases spoken by others has been observed in some children with F.A.S. (14).

• At least four types of hearing disorders result from prenatal alcohol exposure: (a) delayed maturation of the auditory system, (b) sensorineural hearing loss, (c) intermittent conductive hearing loss secondary to recurrent serous otitis media, and (d) central hearing loss (14).

Treacher Collins Syndrome

Treacher Collins syndrome is an inherited disorder characterized by distinctive abnormalities of the head and facial area resulting from underdevelopment of certain portions of the skull. Although the symptoms and physical characteristics associated
with the syndrome may vary greatly in severity from case to case, craniofacial abnormalities tend to involve the cheekbones, jaws, mouth, ears, and/or eyes. Cognitive skills are usually normal (18).

**Demographics**

- The estimated incidence of the syndrome is 1 in 50,000 live births (19-22).
- In approximately 40% of the cases, Treacher Collins syndrome is thought to be inherited as an autosomal dominant genetic trait. However, in 60% of those cases, a positive family history is not found. Research suggests that such cases represent new genetic changes that occur randomly, with no apparent cause (18, 19, 21).

**Communication Manifestations**

- Deformities that affect speech and language include underdevelopment of the facial bones and abnormal dentition with malocclusion (6).
- One-third of patients have cleft palate or velopharyngeal insufficiency (8).
- A congenital bilateral conductive hearing loss is most common, although the presence of sensorineural impairment has been reported in some isolated cases. Presence of unilateral conductive hearing loss is unlikely (6).
- The *auricles* (outer ear appendages) are malformed and may lead to conductive hearing loss (19); *microtia* (congenitally, abnormally small auricles) is reported in 60% to 77% of cases (8).
- To promote normal language development, cleft palate repair as well as evaluation by an otolaryngologist, audiologist and speech-language pathologist are recommended (21).

**Down Syndrome**

Named after John Langdon Down, the first physician to identify the syndrome, *Down syndrome* is the most frequent genetic cause of mild to moderate mental retardation. Down syndrome is a chromosomal disorder caused by an error in cell division that results in the presence of an additional third chromosome 21 or “Trisomy 21” (23). Characteristics of individuals with Down syndrome include facial profile, small nose, a tendency toward protrusion of the tongue, and small ears (6).

**Demographics**

- Down syndrome affects people of all ages, races, and economic levels. The estimate on the incidence of the condition varies from 1 in 800 to 1 in 1,000 births (23-27).
• The incidence of Down syndrome rises with increasing maternal age. The likelihood that a woman under 30 who becomes pregnant will have a baby with Down syndrome is less than 1 in 1,000; chances increase to 1 in 400 for women at age 35 (23).

Communication Manifestations

• Of the manifestations of the syndrome, the cognitive developmental delay appears to be the most deleterious to speech and language acquisition and usage (6).
• The speech of children with Down syndrome is often unintelligible, unlike many other children who have an intellectual disability. However, the nature of their speech disorder is controversial (28).
• Studies of articulatory development after the onset of meaningful speech suggest that children with Down syndrome have a slower rate of development than other children, and that they also make more inconsistent errors (29).
• Vocal quality disturbances emanate from both laryngeal and velopharyngeal levels. In general, individuals with Down syndrome appear to have more breathy, husky, and perceptually lower pitched and more acoustically variable voices than do other individuals (6).
• Studies report a 38% to 78% incidence of hearing loss in children with Down syndrome (30, 31). Congenital otologic anomalies and acquired conditions, such as otitis media and impacted wax, are also frequently observed in this population (31-34).
• Little is known about the processes underlying literacy skills in individuals with Down syndrome. Phonological awareness contributes to literacy development in typically developing children, however, there is inconclusive evidence about these skills in younger children with Down syndrome (35).
• Sensorineural hearing loss is observed among some individuals with Down syndrome as the result of long-standing middle-ear disease (6).

References


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