Branchial Arch Syndromes

David Alfi, DDS, MD [a,*], Din Lam, DMD, MD [b], Jaime Gateno, DDS, MD [a]

KEYWORDS

• Branchial arch syndrome • Treacher Collins syndrome • Pharyngeal arch • Mandibulofacial dysostosis
• Oculoauriculovertebral dysplasia • Goldenhar syndrome • Möbius syndrome • Congenital facial palsy

KEY POINTS

- Branchial arch syndromes present with variable expressivity.
- Management necessitates a multidisciplinary approach.
- Features corroborate a disorder with first and second branchial arch derivatives.
- Treatment should be performed early for vital functional impairments and delayed for esthetic concerns.
- Möbius syndrome is a rare congenital palsy of the sixth and seventh cranial nerves.

Treacher Collins syndrome

Genetics (including inheritance patterns and frequency)

- Autosomal dominant inheritance pattern (a small percentage has autosomal recessive inheritance)
- Affects 1 in 50,000 live births
- Up to 60% of cases result from new or sporadic mutations
- There are 3 recognized gene mutations:
  - *TCOF1* and *POLR1D*: autosomal dominant
  - *POLRIC*: autosomal recessive
- The result of these mutations is an aberrant production of ribosomal RNA that is important in the structural development of the face, and more specifically the first and second branchial arches

Clinical features

The clinical features of Treacher Collin syndrome (TCS) consist of bilaterally symmetric, abnormal development of the structures arising from the first and second branchial arches. This abnormal development contributes to deficient generation of the lower two-thirds of the face (Fig. 1).

The first branchial arch is involved with the growth of the mandible and the zygomaticomaxillary complex and the second arch or hyoid arch is involved with development of the middle ear. The classically presented patient accordingly has a convex facial profile with gross hypoplasia or incomplete formation of the zygoma, maxilla, and mandible in all 3 dimensions. These individuals also have normal intelligence. These facial features are summarized in Table 1.

Differential diagnosis:

- Craniofacial microsomia: features are asymmetric and usually unilateral
- Pierre Robin sequence: children born with severe hypoplasia of the mandible, high arching cleft palate, and a relative macroglossia
- Stickler syndrome comprises a group of hereditary conditions involving eye, ear, and joint deformities plus Pierre Robin sequence
- Nager syndrome includes malformed upper limbs as well as cleft palate and severe palatal hypoplasia

Treatment considerations for the oral and maxillofacial surgeon

The treatment strategy for patients should prioritize procedures that result in functional improvement while delaying those with cosmetic goals until growth maturity. The following functional categories may necessitate early intervention: airway, feeding, hearing, speech, vision, and socialization.

Airway

Maxillary and mandibular hypoplasia can contribute to a compromised airway. Choanal atresia or stenosis can compound this problem, which may require immediate intervention ranging from observation with pulse oximetry and positioning to mandibular advancement with distraction osteogenesis, or tracheotomy (Fig. 2).

Feeding

An incompetent lip seal or cleft lip and or palate may compromise adequate nutrition. Considerations to cleft treatment algorithms or G-tube placement may be necessary.

Hearing

Pediatric otolaryngology consultation and formal audiology testing is required early in order to establish successful hearing.

Vision

Pediatric ophthalmologist should be consulted to evaluate for any extra ocular muscle (EOM) dysfunction or deficits in visual acuity. When the lateral and inferior orbital supporting

The authors have nothing to disclose.

[a] Department of Oral & Maxillofacial Surgery, Houston Methodist Specialty Physician Group, Weill Medical College Cornell University, New York, 6560 Fannin Suite 1280, Houston, TX 77030, USA
[b] Oral and Maxillofacial Surgery, Virginia Commonwealth University, Richmond, VA, USA
* Corresponding author.
E-mail address: dmalfi@houstonmethodist.org

Atlas Oral Maxillofacial Surg Clin N Am 22 (2014) 167–173
1061-3315/14/$ - see front matter
http://dx.doi.org/10.1016/j.cxom.2014.04.003

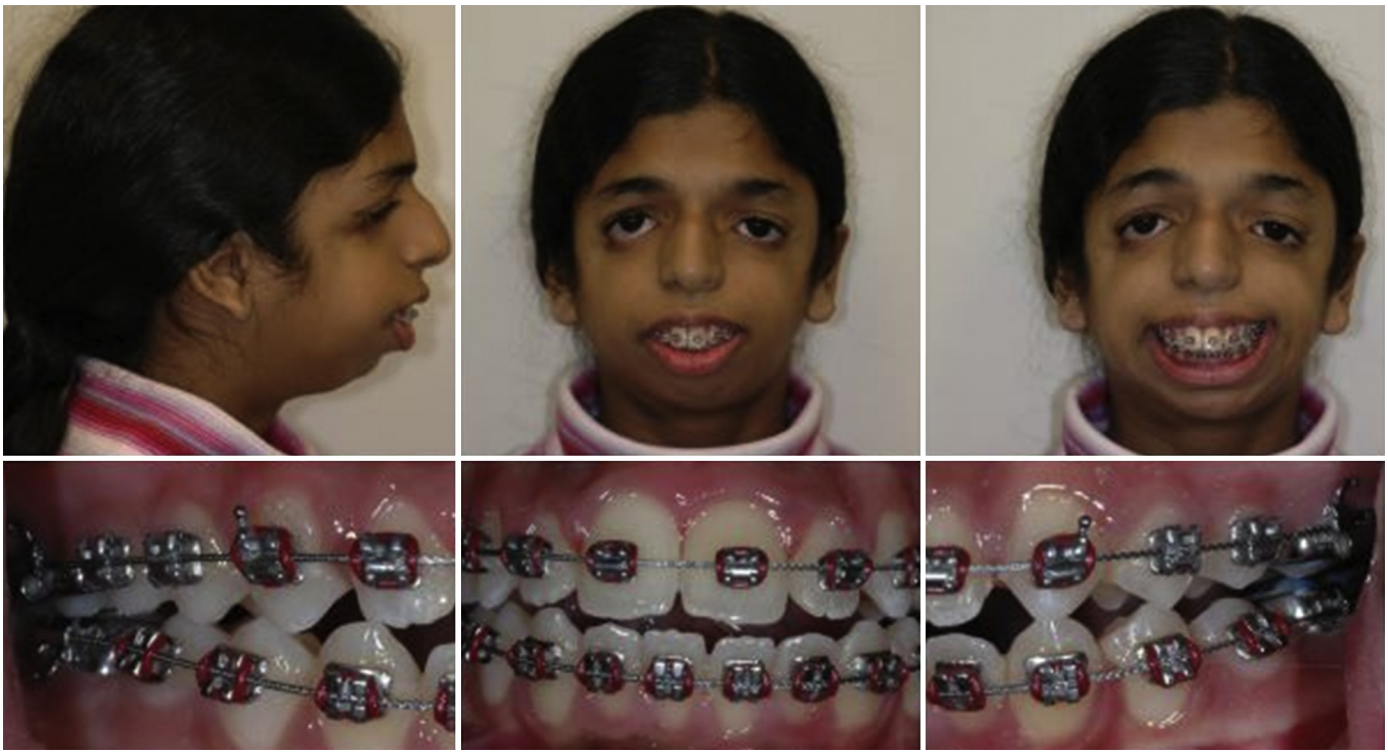

Fig. 1 A 6-year-old girl with TCS. Note the facial features of TCS: colobomata, lower lid hypoplasia, downward slanting of palpebral fissures, prominent nasal dorsum, malar and zygomatic hypoplasia, mandibular hypoplasia, microgenia, Angle class 2 open bite. (*Courtesy of* Dr. Sidney Eisig.)

Table 1 TCS facial features

Eyes	Downward slanting of palpebral fissures Colobomata Lower lid hypoplasia and partial absence of eyelid cilia Dystopia	
Ears	Microtia or absent external ear Middle ear or ossicle hypoplasia Conductive deafness Atresia	
Nose	Prominent nasal dorsum Choanal atresia or stenosis	
Mouth	Cleft palate ± lip Malocclusion (Angel class II, anterior open bite, steep clockwise rotation of the maxillomandibular complex) Dental abnormalities (enamel opacities, dental agenesis, eruption disturbances)	
Musculoskeletal	Skeleton	Mandibular hypoplasia Microgenia Hypoplastic glenoid fossa Maxillary hypoplasia
	Muscles	Muscular hypoplasia (muscles of mastication)
	Joint	Absent, hypoplastic, or deformed TMJ

Abbreviation: TMJ, temporomandibular joint.

structures are hypoplastic or missing the corneas are unprotected, which may necessitate earlier reconstruction of the orbital and zygomatic structures.

Speech

Correction of a cleft palate should follow the standard cleft treatment recommendations.

Secondary treatment strategy should take place at the end of skeletal maturity and include orthognathic surgery and zygomatic-orbital reconstruction.

Malar and orbital reconstruction uses a full-thickness calvarium bone graft through a coronal incision and exposure. It is recommended that this procedure be performed after the age of 6 years, for skeletal maturity of the midface, as well as the ability to reconstruct the donor calvarium site with a local split-thickness calvarium graft.

Maxillomandibular reconstruction is best approached with traditional orthognathic surgery at 13 to 15 years of age, at the time of early skeletal maturity. Le Fort I and rami osteotomies with sliding genioplasty are usually indicated.

When the deformity results in absent ramus, condyle, or glenoid fossa, then reconstruction may be performed at the time of malar and orbital reconstruction. The use of costochondral bone graft to reconstruct the ramus-condyle is advocated. A second reconstruction with conventional osteotomies is almost certain to be warranted after skeletal maturity.

As with conventional orthognathic surgery, nasal reconstruction should be performed as the final reconstructive procedure when indicated.

Goldenhar syndrome

Hemifacial microsomia (HFM), Goldenhar syndrome, and oculoauriculovertebral dysplasia have been used interchangeably.