Dysgnathia Complex: A Case Report and Review of the Literature

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Abstract

Introduction
Dysgnathia complex is a rare craniofacial anomaly with a reported incidence of 1 in 70,000 births. We report a case associated with choanal stenosis, microstomia, aglossia and laryngeal stenosis.

Study Design: Case report with review of the literature

Methods/Case Description:
Our case refers to a 5-year-old boy born at 31 weeks via caesarean section. Mandibular agenesis was first identified during prenatal ultrasound. Computed tomography (CT) revealed a horseshoe-shaped bone representing a diminutive mandible or hyoid bone, as well as, left choanal stenosis and right choanal atresia. Chromosome analysis and microarray were normal.

Direct laryngoscopy and bronchoscopy were performed which revealed a narrowed oropharynx with a shelf of tissue posteriorly and a laryngeal inlet with no discernable opening. No anatomic variations of the distal trachea or bronchi were noted.

The child is tracheostomy and PEG dependent. To date he has undergone multiple surgeries for treatment of his microstomia (4 mm) and agnathia including insertion of a submental tissue expander, construction of mandible with a rib graft, and multiple rhomboid flaps.

Results:
Dysgnathia complex involves disrupted development of the first pharyngeal arch during weeks 4-7 of gestation. Historically, prognosis is extremely poor and those that survive beyond 2 months represent <5% of the documented cases. However, due to advancements in prenatal diagnosis and post-delivery management the prognosis is increasingly optimistic.

Conclusion:
Tracheotomy is currently the definitive airway management for these patients. However, with a greater understanding of the anatomic variations present in these patients other treatment options can be explored.

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Introduction

Dysgnathia complex, or Agnathia-Otocephaly Complex, is an extremely rare, and historically lethal disorder characterized by micrognathia or agnathia, microstomia, hypoglossia or aglossia, and variable ear anomalies.

It is caused by disrupted development of the first pharyngeal arch during weeks 4-7 of gestation, the time period during which facial development normally occurs.

There is a reported incidence of 1 in 70,000 births, and it has been linked to genetic and teratogenic factors[1, 2]. More recently, molecular genetic studies have been used to identify mutations in the PRRX1 and OTX2 genes[3-6].

At this point in time, there is still no genetic test available to screen for this developmental disorder, and is most commonly identified via ultrasonography.

There is little documentation in the otolaryngology literature on the associated upper airway findings in these patients, which we will now discuss.

Case Report

***History*:**
5-year-old boy with normal birth history born at 31 weeks via caesarean section to G2P1 mother. (Birth weight: 2 lbs, 11 oz).

Prenatal History:
- Mandibular agenesis was first identified during prenatal ultrasound.
- Chromosome analysis and microarray were normal.

Early History
Patient underwent tracheotomy at birth and remained in the NICU for 1 month.

Since then he has undergone multiple reconstructive surgeries with plastic surgery including:
- Multiple microstomia repairs.
- Insertion and removal of a neck tissue expander
- Rib graft to mandible

He remains PEG dependent. At age 3, he underwent a swallow evaluation which showed a strong oral aversion and oral hypersensitivity. Thus no assessment of pharyngeal swallow could be performed.

Imaging:
Initial Computed tomography (CT) revealed a horseshoe-shaped bone representing a diminutive mandible or hyoid bone, as well as, left choanal stenosis and right choanal atresia.

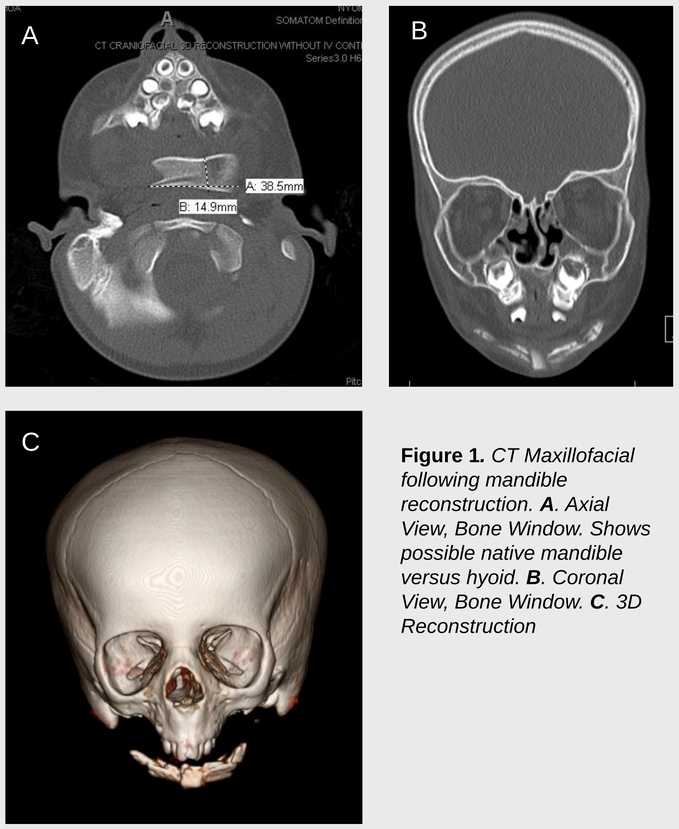

Figure 1. *CT Maxillofacial following mandible reconstruction. **A**. Axial View, Bone Window. Shows possible native mandible versus hyoid. **B**. Coronal View, Bone Window. **C**. 3D Reconstruction*

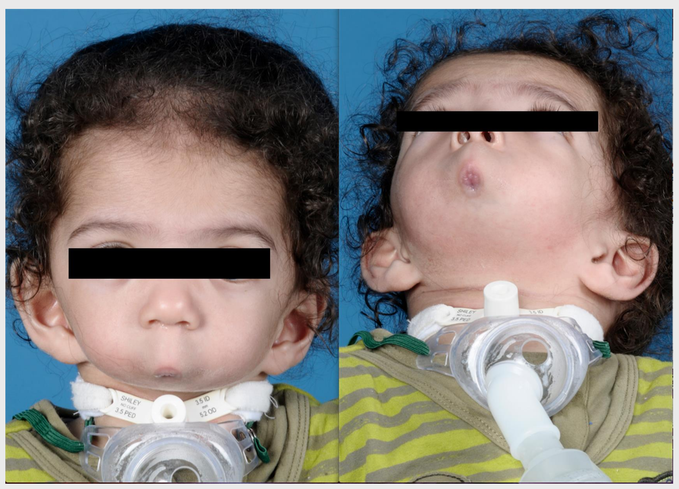

***Figure 2.** Pre-Operative Photos*

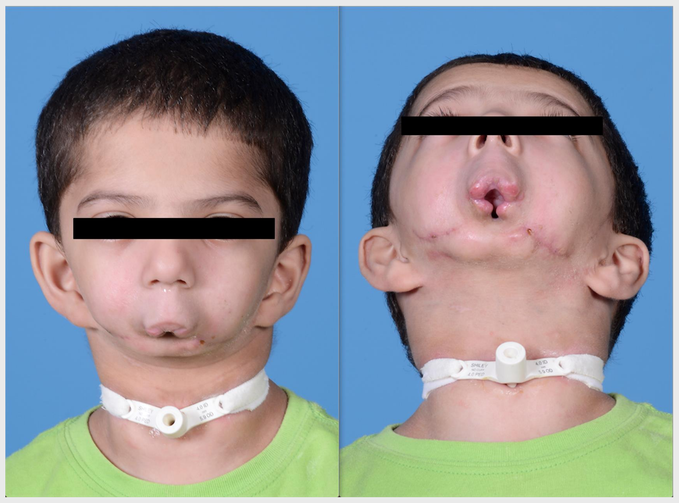

***Figure 3.** Post-operative Photos*

Aerodigestive Tract Findings

Nasal Cavity:
- Left choanal atresia
- Right choanal stenosis

Oral Cavity:
- Severe mandibular micrognathia and microstomia.
- No mandibular teeth.
- Some maxillary teeth.
- Aglossia

Oropharynx:
- Narrowed oropharynx with shelf of tissue posteriorly

Larynx:
- Laryngeal inlet with no discernible opening

Trachea and Bronchi:
- Normal airway caliber, no further anatomic variations or abnormalities.

Conclusions

Dysgnathia complex is a rare disorder characterized by multiple craniofacial and upper aerodigestive tract anomalies.

Tracheotomy is currently the definitive airway management for these patients. However, with a greater understanding of the anatomic variations present in these patients, other treatment options can be explored. This is particularly important given the increasingly optimistic prognosis for these patients.

References

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