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.

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