Unusual Presentation of Macroglossia in Patient with Costello Syndrome

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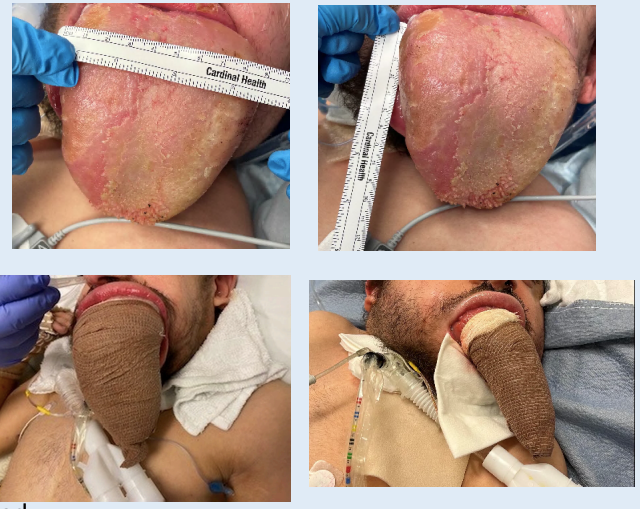
Introduction

Costello syndrome (CS) is a very rare genetic condition that occurs with a mutation in the HRAS gene. Symptoms include developmental delay, intellectual disability, distinctive facial features, short stature, loose folds of extra skin, unusually flexible joints, heart abnormalities and reduced growth hormone levels. CS affects 200 to 300 people worldwide with an estimated prevalence range from 1 in 300,000 to 1.25 million people. The cause of CS is a mutation in the HRAS gene which makes proteins that help control cell growth and division. The craniofacial features common in CS include macrocephaly, bitemporal narrowing, convex facial profile, full cheeks, and large mouth. Additionally, CS patients have a characteristic dental phenotype that includes malocclusion with anterior open bite and posterior crossbite, enamel hypo-mineralization, delayed tooth development and eruption, gingival hyperplasia, thickening of the alveolar ridge, and high palate.

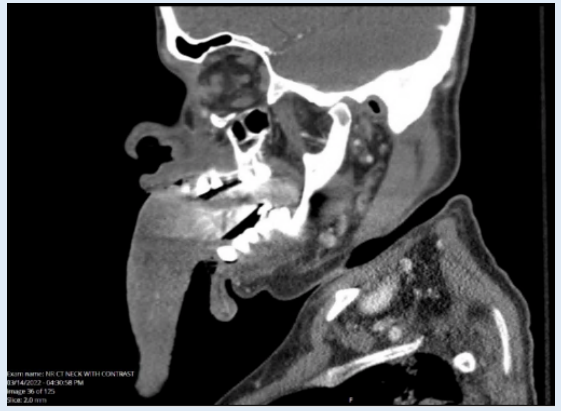
Case Report

This report discusses a 26-year-old male with Costello Syndrome and other associated medical complications including anemia, hypotonia, dysphagia, G Tube, tachycardia, global developmental delay, subaortic stenosis, hypertrophic cardiomyopathy, and trach-dependency. This patient has no known allergies and was taking Benadryl, Clonidine, Ferrous Sulfate, Fluocinonide, Hydroxyzine, and Griseofulvin. There was no significant dental history to be recorded. This patient presented to Rainbow Babies and Children's Hospital for prolonged hospitalization in the PICU with a chief complaint of respiratory failure in the setting of macroglossia and multifocal pneumonia. Repeated intubations led to profound macroglossia necessitating tracheostomy, ventilator dependence and prolonged sedation. The ENT team attempted serial tongue wrapping for 9 days in combination with bilateral bite blocks, Botox and steroid injections with little reduction. Lack of reduction was likely secondary to fibrosis from prolonged edema of the tongue. The patient also developed facial cellulitis thought to be odontogenic in origin with very poor oral hygiene. A multidisciplinary approach was taken by the medical, ENT, and dental teams to evaluate the patient and collaborate on a treatment plan.

Clinical Presentation



Radiographic Findings



Treatment

Due to the extreme nature of the macroglossia, there were concerns of the medical team of re-traumatization the tongue, causing tongue to swell and grow. After discussion with patient's family and other disciplines, it was determined that all teeth were to be extracted with partial tongue resection. Upon clinical exam under general anesthesia, all mandibular teeth were buccally and facially luxated due to weight of the tongue. Teeth were 3+ mobility. Maxillary teeth had multiple fractures and wear due to bruxism. Fragments of fractured teeth were found in and around the tongue, which were possible aspiration risks. The OMFS department completed extractions on Teeth #2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 13, 14, 15, 17, 18, 19, 20, 21, 22 23, 24, 25, 26, 27, 28, 29, 30, and 31. Teeth #16 and 32 were left due to full bony impaction. The case was transferred to ENT team who completed partial resection of the tongue. The tongue was measured 12cm in length and 22cm across at the oral aperture. An anterior resection of 6 cm was completed and a midline wedge resection of 4 cm.

Discussion

Traumatic macroglossia is a rare yet serious condition that requires rapid intervention to help secure the upper airway and protect as much tissue as possible from the tongue. Multiple treatment modalities have been suggested including bite block, tongue massage, wet dressings, intramuscular steroid administration, and tracheostomy in severe cases with or without glossectomy. In this case, many treatment modalities were applied with no success leading to an invasive treatment option. Early detection and intervention will favorably influence the outcome, thereby enhancing the patient's quality of life.

References

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