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Association of Cleft Palate and Ankyloglossia

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Abstract

Clefts of the lip and/or palate are among the most common birth defects worldwide [1]. The majority are non-syndromic where cleft lip and palate occurs in isolation of other phenotypes. Where one or more additional features are involved, clefts are referred to as syndromic. Collectively cleft palate has a major clinical impact requiring surgical, dental, orthodontic, speech, hearing and psychological treatments or therapies throughout childhood. The etiology of cleft palate seems complex, with genetics playing a major role. Several genes causing syndromic cleft palate have been discovered. The etiological complexity of nonsyndromic cleft lip and palate is also exemplified by the large number of candidate genes and loci. To conclude, although the etiology of nonsyndromiccleft palate is still largely unknown, mutations in candidate genes have been identified in a small proportion of cases. We present a case of syndromic cleft palate with ankyloglossia

Keywords: Cleft Palate; Ankyloglossia; T-box Transcription Factor-22.

Introduction

Development of the head and face comprises one of the most complex events during embryonic development, coordinated by a network of transcription factors and signaling molecules together with proteins conferring cell polarity and cell–cell interactions. Disturbance of this tightly controlled cascade can result in a facial cleft where the facial primordia ultimately fail to meet and fuse or form the appropriate structures. Collectively, craniofacial abnormalities are among the most common features of all birth defects. The most frequent of these are the orofacial clefts, cleft lip and/or cleft palate. Cleft palateresults in complications affecting feeding, speech, hearing and psychological development. Patients will undergo multiple rounds of surgical repair starting in the first year of life and may continue until 18 or 20 years old.

Frequently, extensive dental and orthodontic treatment, speech and hearing therapy may be required as well as referral for psychotherapy and genetic counselling. Occurrence estimates range between 1/300 and 1/2500 births for cleft lip with or without cleft palate and around 1/1500 births for cleft palate alone [2]. It has been reported that cleft palate occurs more frequently in males, while the sex bias is reversed forcleft palate, which is more common in females [3].

Approximately 50% of cleft palate patients also havecleft palate, which is thought to be a secondary effect resulting from the defect in facial prominence fusion that precedes palate formation. Cleft palate occurring alone is therefore considered to be etiologically distinct from cleft lip and palate. The majority of cleft lip and palate (< 70%) are regarded as non-syndromic, where the clefts occur without other anomalies. The remaining syndromic cases have additional characteristic features that can be subdivided into categories of chromosomal abnormalities, recognizable Mendelian single gene syndromes, teratogenic effects and various unknown syndromes.

TBX22 is a recently described member of the T-box containing transcription factor gene family that is

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conserved throughout metazoan evolution. These genes play essential roles in early development and in particular mesoderm specification.

Cleft palate or in some cases bifid or absent uvula, the majority of these patients also display ankyloglossia (tongue-tie). This minor feature is frequently missed or unreported; however, when noted in addition to X-linked inheritance, it is an important diagnostic marker for CPX.

Material and Method

A 5 year old girl presented to the outpatient department of Plastic Surgery, JIPMER, Puducherry in April 2016 with difficulty in speech production. On evaluation patient was found to have bilateral complete cleft palate with ankyloglossia grade II.

Treatement plan for the child included palate repair, release of ankyloglossia along with speech therapy.

Fig.1: Bilateral complete cleft palate

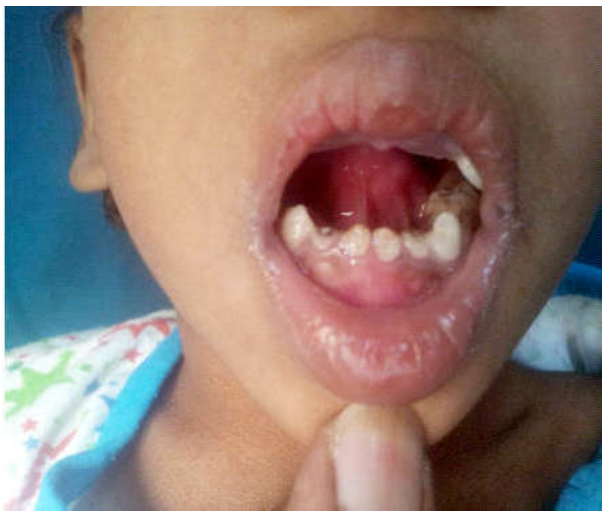

Fig. 2: Ankyloglossia

Discussion

The overall development of the palate involves the formation of the primary palate followed by the formation of the secondary palate. At approximately 30-37 days' gestational age , the primary palate forms by the growth and fusion of the medial nasal, lateral nasal, and maxillary processes. The maxillary process, derived from the proximal half of the first arch, grows to meet and fuse with the nasal processes that have grown and moved in association with the olfactory placode. General opinion holds that mesodermal penetration underlies the formation of the primary palate. Mesodermal reinforcement along lines of fusion is important, as epithelial breakdown and clefting is thought to result from the lack of reinforcement. The secondary palate arises from the 2 palatal shelves, which are initially are in a vertical position because of the interposed tongue. With extension of the head at 7 weeks gestational age and mandibular growth, the tongue is withdrawn, and the palatal shelves can swing into a more horizontal and midline position for fusion and formation of a hard and soft palate. The cleft of the hard palate and soft palate is thought to occur because of the intervening tongue, which impedes elevation of the palatal shelves

Clinically, when cleft lip and palate appears with other (usually two or more) malformations in recognizable patterns, it is classified as syndromic. If it appears as an isolated defect or if syndromes cannot be identified, the term nonsyndromic is used. The number of syndromes is large and still growing.

Ankyloglossia is a congenital anomaly that has a high prevalence rate and may cause difficulty in breastfeeding or speech problems for affected infants. Ankyloglossia often appears as a single anomaly, though it sometimes occurs as a symptom of such rare syndromes as X-linked cleft palate. In patients with X-linked cleft palate, the hereditary trend coincides well with Mendel's laws. In those patients, the mutation of the *TBX22* gene was observed and thus, the *CPX* gene was expressed and ankyloglossia occurred as one of the symptoms of the syndrome. In addition, patients with X-linked cleft palate may present only ankyloglossia or various other physical findings from incomplete to complete cleft palates.

TBX22 is a member of a phylogenetically conserved family of proteins that share a common DNA-binding domain, the T-box [4]. TBX22 is composed of seven exons spanning 8.7 kilobases of genomic DNA in Xq21.1. The TBX22 mRNA is 2099 base pairs long

and encodes a 400-amino-acids protein containing a T-domain in its NH2-terminal region which has the unique feature of missing 20 amino-acids relative to the other known T-domains.

Conclusion

Cleft palate can occur in association with many anomalies co-existing. The association of cleft palate and ankyloglossia though reported in association with other anomalies has not been reported in isolation.

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