FAMILIAL ANKYLOGLOSSIA [TONGUE – TIE] – A RARE CASE REPORT
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ABSTRACT: Ankyloglossia or tongue tie is a congenital anomaly with a prevalence of 4-5% and characterized by abnormally short lingual frenulum. For unknown reasons the abnormality is 2 times more common in males. The pathogenesis of Ankyloglossia is not known. The authors report a family with isolated Ankyloglossia inherited as an autosomal dominant trait in four consecutive generations. The identification of the defective gene (s) in these patients might reveal novel information on the pathogenesis of this disorder. This case report elucidates a specific inheritance pattern of tongue tie.

KEY WORDS: Ankyloglossia, tongue tie, autosomal dominant inheritance.

INTRODUCTION: Ankyloglossia commonly known as tongue tie is a congenital anomaly, characterized by abnormally short lingual frenulum. There is no uniform definition or grading system to describe tongue tie. The condition varies from absence of clinical significance to rare complete Ankyloglossiawhere the ventral part of tongue is fused to the floor of mouth.

Tongue tie may result in varying degree of decreased tongue mobility. Tongue tie has been suggested to cause breast feeding difficulties (sore nipples, poor infant weight gain, early weaning), speech disorders (impaired articulation), and problems with deglutition, dentition, and social issues related to limited function of the tongue. Management of ankyloglossia is controversial. There is no common opinion regarding the indications, timing or method of surgical repair for ankyloglossia.

Tongue tie can be considered a relatively common anomaly with a prevalence of approximately 4 – 5%. For unknown reasons the abnormality seems to be more common in males, with male to female ratio 2.5:1. The pathogenesis of ankyloglossia is not known. Ankyloglossia can be a part of certain rare syndromes such as x- linked cleft – palate and Vander woude syndrome. Most often Ankyloglossia is seen as an isolated finding in an otherwise normal child. Maternal cocaine use is reported to increase the risk of Ankyloglossia to more than three fold.

In this case report, we describe a family with isolated Ankyloglossia inherited as an autosomal dominant trait. This family in which 9 individuals spread over 4 generations have Ankyloglossia, inherited as an autosomal dominant trait. As the exact pathogenesis is not known yet and the limited availability of case reports, studies regarding genetic predisposition of tongue tie, the authors present this as an affirmative case report for explaining genetic inheritance pattern of tongue tie.

CASE REPORT: Our cases were diagnosed in Vinayaka Missions Medical College, Hospital, Karaikal, Puducherry(U.T). Two boys of 6 and 11 years each who are brothers presented with slurring of speech and inability to protrude the tongue. Both were diagnosed to have tongue tie. Family history revealed the presence of Ankyloglossia in 4 consecutive generations from maternal side. Patients’ great grandfather, mother, 2 maternal uncles and their children had Ankyloglossia. Except patients...
great grandfather all of the above were examined and confirmed to have Ankyloglossia. Both patients were treated in E.N.T. department of Vinayaka Missions Medical College, Karaikal, by Frenotomy.

**Fig.1: Pedigree of patients affected with familial Ankyloglossia**

**DISUSSION:** Ankyloglossia or tongue tie is a relatively common finding in the newborn population and represents a significant proportion of breast feeding problems. This anomaly is characterized by the attachment of under surface of tongue to the floor of mouth. This condition is the result of failure in cellular degeneration leading to a tongue tie. For this condition treatment options include surgical procedures as frenotomy, frenectomy and frenuloplasty, though its spontaneous resolution is also possible in some cases. No widely accepted criteria have been established for the surgical indications and the selection of surgical procedures.

The exact pathological mechanism of ankyloglossia remains unclear and its conclusive hereditary nature is yet to be elucidated. In the pedigree chart of above mentioned case there seem to be affected individuals who have passed the condition onto affected offsprings. In the reported pedigree ankyloglossia appears to be inherited as an autosomal dominant trait, unless all the spouse of affected individuals in first three generations were carriers, which is highly unlikely. This observation is coinciding with the conclusion made by Klockars as ankyloglossia being inherited as an autosomal dominant disorder.

Except for tongue tie and defect in articulation, we didn’t find any other symptoms or anomalies in our cases. So, our report differs from previously described cases which had fibrous bands associated with congenital abnormality such as anencephaly, tracheo-oesophageal fistula or cleft palate.

To identify the defective gene(s) causing ankyloglossia in the patients, linkage analysis is feasible. Obvious candidate’s genes causative for non syndromic ankyloglossia include TB X 22 gene [a T- box transcription factor gene] which is mutated in x- linked cleft palate and ankyloglossia.
LGR5 gene [an orphan G-protein–coupled receptor gene] is found to be associated with neonatal lethality and ankyloglossia in mice.

**CONCLUSION:** Although in this pedigree the number of patients is limited, the male predominance is established. However, the identification of defective genes causing ankyloglossia might reveal novel information about abnormal craniofacial embryogenesis and the pathogenesis of this particular disorder.

**REFERENCES:**


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