

Familial Ankyloglossia (Tongue-tie): A Case Report

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Abstract- Ankyloglossia (tongue-tie) is a congenital anomaly with a prevalence of 4–5% and characterized by an abnormally short lingual frenulum. For unknown reasons the abnormality seems to be more common in males. The pathogenesis of ankyloglossia is not known. The authors report a family with isolated ankyloglossia inherited as an autosomal dominant or recessive trait. The identification of the defective gene(s) in these patients might reveal novel information on the pathogenesis of this disorder.

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Key words: Ankyloglossia; tongue-tie; inheritance patterns

Introduction

Ankyloglossia, commonly known as tongue-tie, is a congenital anomaly characterized by an abnormally short lingual frenulum. The phenotype varies from absence of clinical significance to rare complete ankyloglossia where the ventral part of the tongue is fused to the floor of the mouth (1). There is no uniform definition or grading system to describe tongue-tie.

Management of ankyloglossia is controversial. The abnormally short lingual frenulum 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), problems with deglutition and dentition, oral-motor dysfunction and social issues related to the limited function of the tongue (2). There is no consensus regarding the indications, timing or method of surgical repair for ankyloglossia (3).

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 of 2.5:1.0 (4, 5 and 6).

The pathogenesis of ankyloglossia is not known. Ankyloglossia can be a part of certain rare syndromes

such as X-linked cleft palate (OMIM 303400) (7) and van der Woude syndrome (OMIM 119300) (8). 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 threefold (4).

In this case report, we describe a family with isolated ankyloglossia inherited as an autosomal dominant or autosomal recessive trait. This family consisting of five generations in which five individuals had ankyloglossia inherited as an autosomal dominant or recessive trait. Except a report by Keizer (1952) who described a Dutch family in which 13 persons in 3 generations had ankyloglossia and there were many instances of male-to-male transmission, there is no other report of familial ankyloglossia in the OMIM.

Case Report

Our cases were diagnosed in Baqiyatallah Subspecialty Hospital in Tehran. As shown in the pedigree (Figure 1), in the fourth generation there are 4 offsprings (3 females and one male) of which 2 females and the male are affected by congenital ankyloglossia. The history shows that they all underwent frenuloplasty procedure and now they talk normally without any articulation problem.

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Familial ankyloglossia (tongue-tie)

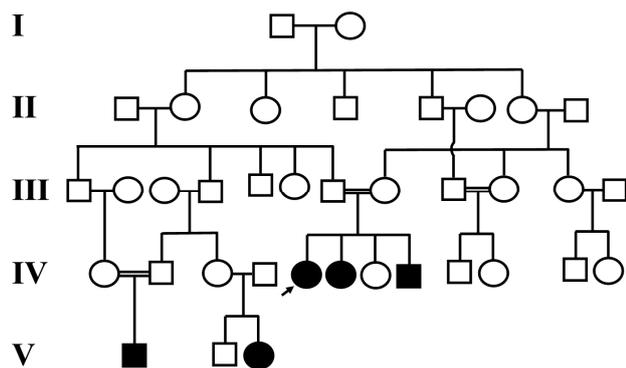


Figure 1. Pedigree of patients affected with familial ankyloglossia

Discussion

Ankyloglossia or 'tongue-tie' is a relatively common finding in the newborn population and represents a significant proportion of breastfeeding problems. This anomaly is characterized by the attachment of the tongue to the floor of the mouth. The condition is the result of a failure in cellular degeneration leading to a much longer anchor between the floor of the mouth and the tongue. In the reported pedigree, ankyloglossia seems to be inherited as an autosomal dominant trait with incomplete penetrance or due to an autosomal recessive trait. Based on previous reports ankyloglossia is known to be more common in males (4- 6). In our pedigree there seem to be unaffected individuals who have passed the condition on to affected offspring. The non-affected parents of affected offspring may be explained by incomplete penetrance, variable expressivity or due to a recessive trait.

Except tongue-tie and defect in articulation we didn't find any other symptom or anomalies in our cases so our report differs from previously described cases which had fibrous bands associated with congenital abnormalities such as anencephaly, tracheoesophageal fistula, patent foramen ovale or strong family histories of cleft palate.

To identify the defective gene(s) causing ankyloglossia in these patients linkage analysis should be feasible. Obvious candidate genes causative for non-syndromic ankyloglossia include TBX22 gene "a T-box transcription factor gene" mutated in X-linked cleft palate and ankyloglossia (9) and LGR5 gene "an orphan G protein-coupled receptor gene" associated with

neonatal lethality and ankyloglossia in mice (10). Although in this pedigree the number of patients is limited but here in contrast to previous reports the number of female patients is more than males.

Tongue-tie is in most cases a relatively harmless condition and the treatment (if even needed) is often relatively simple and safe. However, the identification of the defective gene(s) causing ankyloglossia might reveal novel information about craniofacial embryogenesis and the pathogenesis of this disorder.

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