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CASE REPORT

Familial ankyloglossia (tongue-tie)

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Summary 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 author reports a family with isolated ankyloglossia inherited as an autosomal dominant trait. The identification of the defective gene(s) causing ankyloglossia might reveal novel information on the craniofacial embryogenesis and its disorders.

1. 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.

The possible consequences and management of ankyloglossia is controversial. The abnormally short lingual frenulum may result 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]. However, 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 2.3–2.7:1.0 [4–6].

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]. Ballard and co-authors reported a positive family history in 21% of the infants with ankyloglossia [9].

In this case report, I describe a Finnish family with isolated ankyloglossia inherited as an autosomal dominant trait. To my knowledge, there are no previous reports describing pedigrees for familial ankyloglossia in the English written literature. However, Keizer [10] reported (in Dutch) a family

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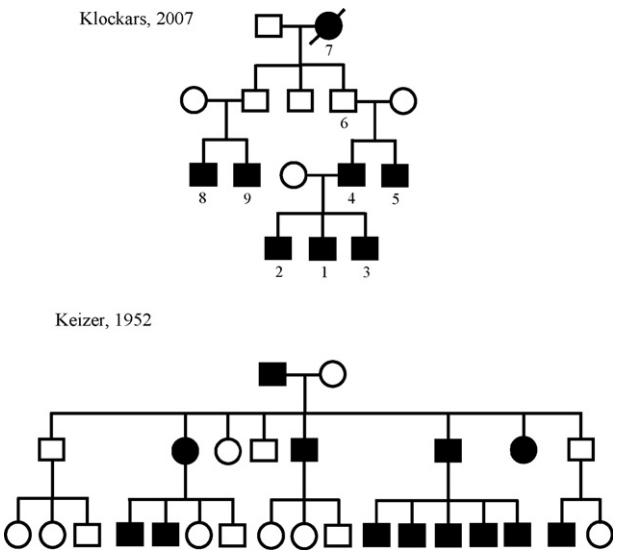

Fig. 1 Ankyloglossia pedigrees.

consisting of 26 members in three generations of which 13 had ankyloglossia inherited as an autosomal dominant trait (Fig. 1) [10].

2. Case report

Patient 1 (Fig. 1), born 2000 was referred to the Department of Otorhinolaryngology, Kymenlaakso Central Hospital (Finland) for surgical treatment of ankyloglossia. According to his parents he had no feeding problems when younger. Despite intensive speech therapy he had difficulties in the production of certain consonants (d, l, alveolar r). The clinical examination revealed an isolated mild–moderate ankyloglossia, the frenulum was thick and short and restricted tongue protrusion and lifting of the tip of the tongue. A frenuloplastia was performed under general anaesthesia.

Patient 2, born 1999, was diagnosed with mild ankyloglossia year 2001 at the Kymenlaakso Central Hospital. He had no feeding problems, but due to speech problems (difficulties in consonants d, l, alveolar r) he had frenuloplastia year 2003. After frenuloplastia and speech therapy the articulation problems have resolved.

Patient 3, born 2004, has no feeding problems. Due to his young age it is difficult to evaluate his speech. Clinical observation revealed mild ankyloglossia with thickened and shortened frenulum. Since asymptomatic, no surgery has yet been performed, but the parents have been informed about the possible future problems and have been advised to contact the hospital if any symptoms of clinical significance arise.

Patient 4, born 1975, came to the Department of Otorhinolaryngology, Kymenlaakso Central Hospital (Finland) to escort his son (patient 1). During his son's preoperative clinical examination a positive family history was obvious and detailed discussions were carried out with the parents to identify affected relatives and the mode of inheritance. His clinical examination revealed an isolated mild ankyloglossia, the frenulum was thick and short and restricted tongue protrusion. Despite history of speech therapy he had difficulties in the production of certain consonants (d, l, r) and could not reach the upper lip with the tip of the tongue. A frenuloplastia was performed under local anaesthesia.

Patient 5, born 1977, came in contact with the author introducted by his brother (patient 4). The clinical examination revealed an isolated mild ankyloglossia, the frenulum was thick and short and restricted tongue elevation (Fig. 2). As his brother,

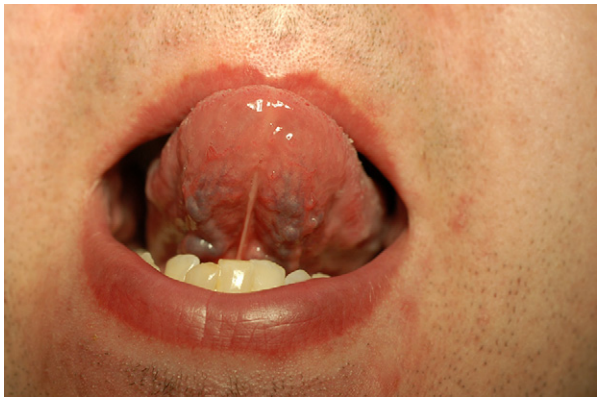

Fig. 2 Patient 5, isolated mild ankyloglossia restricting tongue elevation.

despite history of speech therapy he had difficulties in the production of certain consonants (d, l, r) and had difficulties reaching the upper lip with the tip of the tongue. A frenuloplastia was performed under local anaesthesia.

Individual 6 did not want to meet with the author, but is told to have normal speech and tongue movement. The information on individuals 7, 8, and 9 is based on the information given by patient 4 and they have not been seen or examined by the author. All these individuals were reported to have speech problems with certain consonants and restricted tongue movement.

3. Discussion

In the reported pedigree, ankyloglossia seems to be inherited as an autosomal dominant trait with incomplete penetrance. Based on previous reports ankyloglossia is known to be more common in males [4—6]. Ballard and co-authors reported a positive family history in one fifth of the infants with ankyloglossia [9]. Interestingly, in the pedigree reported by Keizer [10] there are more affected males than females and in the pedigree reported in this paper there is overall male predominance. If one calculates the offspring of the carriers of the defective gene in the two pedigrees there is a clear male predominance: of the affected individuals 18 out of 21 were males (86%).

The positive family history [9] and the pedigrees by Keizer [10] and reported here are suggestive for autosomal dominant inheritance in a significant portion of ankyloglossia patients. In both pedigrees there seem to be unaffected individuals who have passed the condition on to affected offspring. However, it should be noted that in the pedigree described here the patient 6 was not examined by the author but was reported to have normal tongue movement by his son. Thus it is possible that patient 6 could be mildly affected or affected and asymptomatic. The non-affected parents of affected offspring may be explained by incomplete penetrance or variable expressivity, but also by spontaneous or traumatic resolution of ankyloglossia with age.

To identify the defective gene(s) causing alkyloglossia a large number of pedigrees with familial tongue-tie would be needed. A significant percentage of cases of ankyloglossia seem to be familial [9]. Thus finding suitable families for linkage analyses 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 [11] and LGR5, an orphan G protein-coupled receptor, associated with neonatal lethality and ankyloglossia in mice [12].

Due to the limited size, the pedigree reported here and the pedigree described by Keizer [10], do not allow any conclusions about the male to female ratio. However, the pedigrees raise some questions: Is there a male predominance in familial ankyloglossia? Could this predominance explain the male excess seen in ankyloglossia patients in general? To answer these questions larger studies and pedigrees would be needed.

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 disorders associated with its disruption.

Conflict of interest statement

The author has no financial or personal relationships that could influence this work.

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