Feeding Difficulty in an Infant with Stickler's Syndrome

Abstract

Stickler syndrome or hereditary progressive arthro-ophthalmopathy is an autosomal dominant condition characterized by ocular manifestations, arthritic changes, orofacial features, and deafness, in variable degrees. This is a case report of a 48-day-old infant who presented with severe feeding difficulty. It also details the clinical, diagnostic features, and management of an infant with Stickler syndrome.

Keywords: *Cleft palate, Pierre-Robin sequence, Stickler syndrome*

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Introduction

Stickler's syndrome also known as hereditary arthro-ophthalmopathy is a dominantly inherited disorder of collagen connective tissues, resulting in an abnormal vitreous of the eye, variable degrees of myopia, the risk of retinal detachment, cataract, and glaucoma.[1]

The disease was first described by Stickler *et al*. and subsequently reported in two families with Pierre-Robin sequel as a connective tissue disorder that may affect the formation of the eyes, ears, palate mandible, and joints.[2]

Stickler's syndrome Affects both males and females. Prevalence rates have been estimated at 1–3/1000 births and at 1/7500 births. Most investigators believe that the disorder is highly under-diagnosed making it difficult to determine the true prevalence of Stickler's syndrome in the general population.[3]

Early recognition of the syndrome is important, not only for genetic counseling but also to offer a more precise prognosis and improved treatment of the many serious disorders that may occur in affected children.[2]

Case Report

A 48-day-old infant had reported to our tertiary care hospital with the complaint of feeding difficulty. Clinical examination revealed syndromic facies with retrognathia, micrognathia, cleft palate, and increased work on breathing. Intra-oral examination revealed a secondary cleft palate involving the hard and soft palate [Figure 1].

Detailed case history and genetic evaluation were done which suggested features of syndromic phenotype (Stickler's syndrome). Medical records revealed a large patent ductus arteriosus continuous left-to-right shunt and a refractive error on the myopic side in both eyes. Ear, nose, and throat evaluation revealed decreased hearing with respect to the right ear. Flexible endoscopy showed stridor due to tongue fall and retrognathia. However, she was referred to the Department of Pedodontics due to a severe feeding difficulty.

A putty-based impression was used to record the anatomy of the cleft using a special tray customized for this child. The light-body impression recorded the cleft margins distinctly. A dental stone cast was made out of the impression received [Figure 2].

The cleft margins and area were covered by a wax spacer and an acrylic-based feeding plate was then fabricated on the cast. Once the feeding plate was finished and polished, the wax spacer was removed. Two holes were made on either side of the mesiobuccal and distobuccal alveolar ridge, to insert a dental floss [Figure 2]. This was done to prevent the plate from being aspirated. Excess margins on the palatal aspect were reduced till the child

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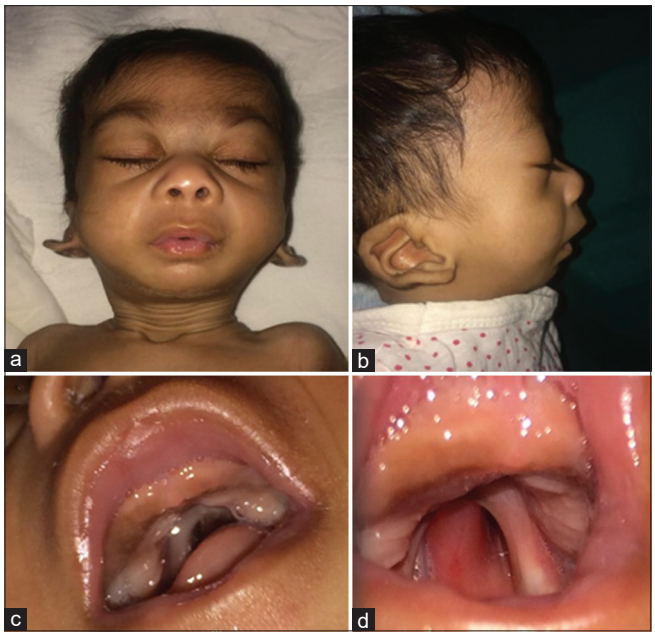

Figure 1: (a-d) Facial features in Stickler syndrome with secondary cleft involving hard and soft palate

could retain the plate in the mouth without triggering a gag reflex. The mother was then asked to feed the child immediately to assess if the child tolerated the feeds well [Figure 3]. As the feeding turned out hassle-free, the mother was instructed to regularly use the plate before every feed. The patient was reviewed periodically every week thereafter for 2 months, and no complaints were reported.

Discussion

Stickler *et al*. described an autosomal dominant connective tissue disorder with hereditary progressive arthro-ohpthalmopathy. Features of the Stickler syndrome include premature osteoarthritis; ocular involvement; sensorineural hearing loss; a characteristic facies with maxillary hypoplasia, midface hypoplasia, long philtrum, and micrognathia; and cleft palate (Pierre-Robin sequence).[4,5] However, most of these features become less distinct as the child reaches adulthood. The facial features are so variable that in isolation they are unreliable for making a diagnosis. The infant, in this particular case, had distinctive facial features such as retrognathia, retroglossoptosis, flattened nasal bridge, hypoplastic maxilla, and shape anomaly of the external ear. A quarter of patients (25% of cases) have some evidence of midline clefting. This can range from the extreme of the Pierre-Robin sequence, through clefting of the hard/soft palate, to the mildest manifestation of the bifid uvula.[6] Affected children may present with speech defects and may require speech therapy. The infant presented here with secondary cleft involving the hard and soft palate. Early diagnosis of such infants can only result in a better prognosis.

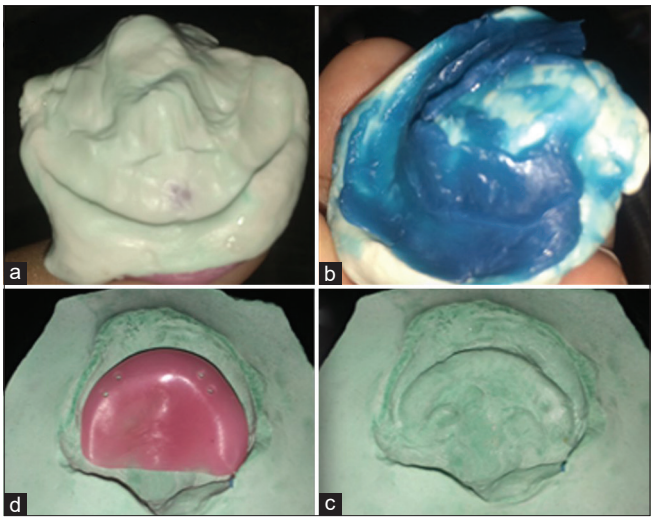

Figure 2: (a-d) Step-wise procedure of fabrication of feeding plate

Of babies born with Pierre-Robin sequence, 30%–44% are subsequently diagnosed with Stickler's syndrome. For the same reason, if a child presents with Pierre-Robin sequence, the ophthalmic investigation should be performed to rule out Stickler's syndrome.[7]

Feeding problems leading to failure to thrive in babies with cleft conditions were recorded as early as the 1600s. It has been accepted that babies with nonsyndromic oral clefts have feeding difficulties related to the structural malformation, which causes abnormal suction and compression during nutritive sucking. Glass and Wolf in 1999 suggested that in cases where the cleft occurs with multiple congenital anomalies, the feeding difficulty is more complex and thought to relate to the neurological status of the baby and/or the presenting medical condition as well as the structural anomaly.[8]

For optimal suction, while feeding, a baby must have intact oral structures, especially lip and palatal structures, and functional competence of relevant musculature including the lips, cheeks, tongue, velum, and pharyngeal walls. Given their abnormal oral anatomy, it is not surprising that babies with CL/P are reported to have difficulty creating the oral pressure gradients necessary for the bottle- or breast-feeding. Choi *et al.* in 1991 documented that the degree of impairment expected as a consequence of particular cleft malformations remains controversial, but the extent, place, and width of the defect may be important factors in influencing the outcome.[8] The infant presented in this case had secondary cleft palate which failed to create the necessary oral seal to help the child be able to feed, which in turn failed to thrive. Thus, the main objective here was to fabricate a feeding plate, so the infant can have proper nourishment and subsequent weight gain to proceed with other surgeries.[9]

The management of patients with Stickler's syndrome is more complex than a simple cleft repair or airway

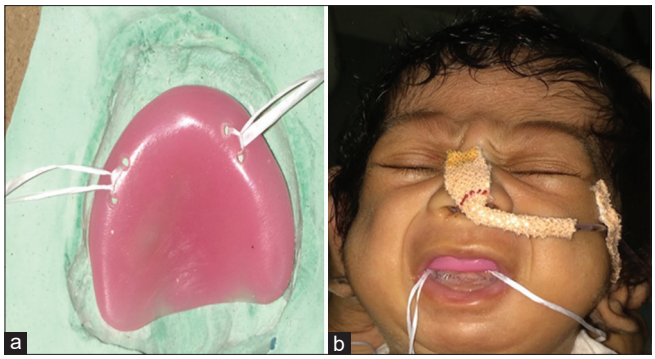

Figure 3: (a and b) Dental floss knotted for ease of handle

management. Successful long-term outcomes depend on individualized, fully integrated, long-term treatment provided in an effective and coordinated manner by a multidisciplinary team of experts in the field from early infancy, through adolescence.[10]

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

References

1. Stickler GB, Hughes W, Houchin P. Clinical features of hereditary progressive arthro-ophthalmopathy (Stickler syndrome): A survey. Genet Med 2001;3:192-6.
2. Stickler Syndrome. Genetic and Rare Disease Information Centre; March, 2018. Available from: https://www.rarediseases.info.nih.gov/diseases/10782/stickler-syndrome. [Last accessed on 2018 Aug 30, 20:37 pm].
3. Popkin JS, Polomeno RC. Stickler's syndrome (hereditary progressive arthro-ophthalmopathy). Can Med Assoc J 1974;111:1071-6.
4. Francomano CA, Liberfarb RM, Hirose T, Maumenee IH, Streeten EA, Meyers DA, *et al.* The stickler syndrome: Evidence for close linkage to the structural gene for type II collagen. Genomics 1987;1:293-6.
5. Kimura D, Sato T, Oosuka S, Kohmoto R, Fukumoto M, Mimura M, *et al.* Case report of a family affected by stickler syndrome in which rhegmatogenous retinal detachment occurred in five eyes of three siblings. Case Rep Ophthalmol 2018;9:1-8.
6. Snead MP, Yates JR. Clinical and molecular genetics of stickler syndrome. J Med Genet 1999;36:353-9.
7. Webb AC, Markus AF. The diagnosis and consequences of stickler syndrome. Br J Oral Maxillofac Surg 2002;40:49-51.
8. Choi BH, Kleinheinz J, Joos U, Komposch G. Sucking efficiency of early orthopaedic plate and teats in infants with cleft lip and palate. Int J Oral Maxillofac Surg 1991;20:167–169.
9. Reid J, Reilly S, Kilpatrick N. Sucking performance of babies with cleft conditions. Cleft Palate Craniofac J 2007;44:312-20.
10. Wellens W, Vander Poorten V. Keys to a successful cleft lip and palate team. B-ENT 2006;2 Suppl 4:3-10.