

## CASE REPORT

## A Rare Case of Median Cleft Face Syndrome: A Case Report

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## ABSTRACT

**Introduction:** Median cleft face syndrome, also known as frontonasal dysplasia, is a very rare disorder characterized by abnormalities affecting the head and facial (craniofacial) region. People with frontonasal dysplasia have at least two of the following features: widely spaced eyes (ocular hypertelorism); a broad nose; a slit (cleft) in one or both sides of the nose; no nasal tip; a central cleft involving the nose, upper lip or roof of the mouth (palate); incomplete formation of the front of the skull with skin covering the head where bone should be (anterior cranium bifidum occultum); or a widow's peak hairline.

**Case Report:** This is a case report of a 14-year old partially blind boy showing characteristic features of median cleft face syndrome.

**Conclusion:** Although no particular cause of development of median cleft face is known, appropriate genetic counselling procedures coupled with symptomatic treatment required for the child maybe given.

**Keywords:** Median Cleft Face, partial Blindness, rare disease

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## INTRODUCTION

Midline facial defects with hypertelorism (MFDH) is the name suggested for a rare and heterogeneous group of craniofacial disorders mainly characterized by ocular hypertelorism and bifid nose.<sup>1-3</sup> Several denominations have been used for this condition, such as *median* cleft face syndrome, frontonasal syndrome, frontonasal dysostosis; and malformative frontonasal sequence. Frontonasal dysplasia is the name most commonly accepted.<sup>4</sup>

There are two leading theories of facial cleft formation. The

classic theory, championed by Dursy and His, holds that clefts are caused by the failure of fusion of the facial processes. In this theory, the face forms as the finger-like ends of the maxillary processes meet and coalesce with the united paired globular processes beneath the nasal pits. Once epithelial contact is established, mesenchymal penetration completes the fusion and the lip and hard palate are formed. When the sequence is disturbed, clefting occurs. The mesodermal penetration theory was propounded by Pohlmann and Veau and was later advocated by Stark and Saunders. Proponents of the mesodermal penetration theory think that free-end facial processes do not exist and the face consists of a bilaminar ectodermal membrane, with epithelial seams demarcating the major processes. The mesenchyme migrates into this double wall of ectoderm, penetrates it, and smooths out the seams. If mesenchymal penetration fails, then the unsupported epithelial wall dehisces and a cleft is produced. The severity of the cleft is inversely proportional to the success of mesodermal penetration, with different degrees of incomplete and complete clefts.<sup>3</sup>

This case, a boy of 14 years, presented with extra-oral features of mid facial clefting along with partial blindness. The clinical features combined with his medical history led to the diagnosis of median cleft face syndrome.

## CASE REPORT

During a regular screening and treatment camp undertaken at a blind school, the Poona School and Home for the Blind Boys, a 14 year old boy presented features of a dysmorphic face. On taking the detailed history of the boy, it was found that the boy had been born by a normal delivery at a hospital in his hometown of Jalgaon. The parents were not consanguineously married. The mother received antenatal care at the hospital and was registered at two months of gestation. The mother did not give any history of fever or abdominal pain or any other medical complaint during the gestational period. The mother also did not give any significant history of intake of any form of medication during the gestation period- allopathic or herbal.

The boy is the first child in a monogamous setting; mother is unemployed while the father is a daily wage labourer. There is no history of similar features in the other younger sibling. The boy had obvious facial deformity, widow's peak with divided nostrils and a bifid nose.

The inter-canthal distance was more (32.1mm) as compared to the normal range for children of that age (28mm). The boy weighed 35kgs and had normal height- 130cms. The gait was normal. He had micro-ophthalmos along with partial loss of

vision in the right eye and complete loss of vision in the left eye. He showed an inability to open his left eye. (Fig.1) The oral features showed the presence of a unilateral masticatory chewing habit- chewing only on the left side was seen. There was presence of severe supra-gingival calculus seen on the upper and lower right jaw region which was then cleared with thorough scaling. Labially erupted right maxillary central incisor and right and left maxillary canines were seen. (Fig.2) Partially erupted maxillary second premolar was seen, indicative of delayed eruption. Over-retained deciduous maxillary second molar on the left side of the jaw was seen. (Fig.2) The boy showed the presence of slightly carious pits and fissures in relation to the maxillary and mandibular permanent molar teeth. (Fig.2) There was no frank carious lesion reported.

### Investigations

CBCT investigations done revealed a maxillary deficiency. The maxillary sinus on the left side appears to be obliterated. There is a deviation of the nasal septum seen in the axial section. There is also an irregularity in the cranial vault seen. (Fig.3)

### DISCUSSION

Frontonasal dysplasia (FND) is a rare developmental defect of craniofacial region where the midface does not develop normally. The exact cause of FND is unknown. Anomalies can be explained by single malformation, though most cases are sporadic. Certain studies link the occurrence of this disease to be autosomal recessive. Certain others claimed that the disease was related to a defect in the ALX gene. It has also been suggested that a defect involving chromosomes 3q23, 3q27, 7q21 and 11q21 might play a role.<sup>1</sup>

In our case, genetic testing of the patient's parents was done when the child was born. Both the parents showed a normal karyotype, thus ruling out the possibility of the disorder being transmitted genetically. It is found that during the 3<sup>rd</sup> week of gestation, two areas of thickened ectoderm, the olfactory areas appear immediately under the forebrain in the anterior wall of the stomodeum. By the up-growth of the surrounding parts, these areas are converted into pits, the olfactory pits, which indent the frontonasal prominence and divide into a medial and two lateral nasal processes. Frontonasal dysplasia is due to deficient remodelling of the nasal capsule, which causes the future fronto-nasoethmoidal complex to freeze in the foetal form.<sup>1</sup>

There is no single strategy for treatment of facial clefts, because of the large amount of variation in these clefts. Which kind of surgery is used depends on the type of clefting and which structures are involved. Patient was scheduled for a surgical nasal reconstruction along with a fixed orthodontic treatment.

In 1976, Tessier described an anatomical classification system in which a number is assigned to each malformation on the basis of its position relative to the sagittal midline. This



Figure-1: Clinical extra-oral images of the patient



Figure-2: Clinical intra-oral images of patient

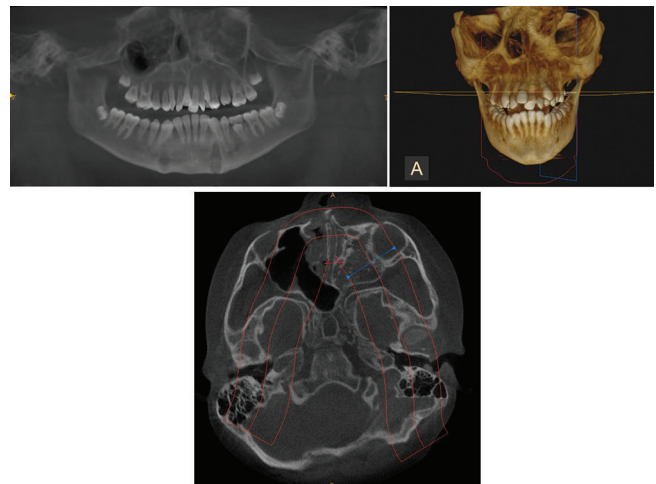


Figure-3: Radiographic and CBCT images of the patient

system has become internationally accepted and allows concise effective communication among clinicians. According to Tessier's classification, the cleft of this patient falls under the cleft no.14 classification (median cleft running through the midline of the face).<sup>6</sup>

According to Van der Meulen classification, that divides different types of clefts based on where the developmental arrest occurs in embryogenesis, this patient shows nasal clefting (development arrest of the lateral side of the nose, resulting in a cleft in one of the nasal halves. The nasal septum and cavity can be involved).<sup>7</sup>

Prevention: Due to the exact cause of facial clefting being unclear, it is difficult to say what may prevent children being born with facial clefts. However, it seems that folic acid contributes to a lower risk of a child being born with a facial cleft.<sup>4</sup>

Genetic counselling may be of benefit for affected individuals and their families. A team approach for infants and children with this disorder may be of benefit and may include special social, educational, and medical services. Other treatment is symptomatic and supportive.

## CONCLUSION

In conclusion, median cleft face syndrome is a very rare disorder whose management requires a multidisciplinary approach. Lack of appropriate diagnostic facilities, socio-economic and cultural factors may prove to be a problem in our setting. We suggest reduction in cost of treatment of children diagnosed with such kind of a rare craniofacial disorder so that morbidity and mortality in such children can be reduced.

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