New dental findings in the median cleft facial syndrome


The term “median cleft face syndrome” was introduced in 1967 by DeMyer. Other terms used to describe this disorder are “frontronasal syndrome,” “frontronasal dysplasia” and “frontronasal malformation.”

The main features of this syndrome are hypertelorism, cranium bifidum occultum, widow’s peak, and midline clefting of the nose, upper lip and palate. Primary telecanthus, ocular colobomas, microphthalmia and clefting of the alae nasi also can be present; in fact, the alae nasi may be affected in both vertical and oblique clefs.

The presence of hypertelorism is necessary to diagnose this syndrome. The next most frequent characteristic is a true clefting of the nasal midline. Other anomalies that may be present are encephalocoele, hydrocephalus, holoprosencephaly, macrocephaly, agenesis of the corpus callosum, duplication of the pituitary gland, cleft mandible, and maxillary cervical abnormalities or tetralogy of Fallot. The intelligence of patients with median cleft facial syndrome can range from normal to severe mental retardation; nevertheless, the intelligence quotient does not appear to be related to the severity of facial clefting.

Other oral abnormalities that can be found in patients with median cleft facial syndrome are micrognathia, highly arched hard palate, hypoplastic maxilla, abnormalities of the development of the tongue, conical teeth and hypoplastic enamel.

Different authors have classified this syndrome according to the facial clefting present in the patient. Nevertheless, Sedano and colleagues classification is used often. It identifies four basic facies that are ranked according to severity from least to most:

- facies A includes ocular hypertelorism, broad nasal root and median nasal groove with absence of the nasal tip;
- facies B includes ocular hypertelorism, broad nasal root and deep median facial groove or cleft that affects the nose or both the nose and the upper lip, and the palate also may be cleft;
- facies C comprises ocular hypertelorism, broad nasal root, unilateral or bilateral notching and the nasal ala;
- facies D is a combination of facies B and C.
pronounced the facial changes are. Nevertheless, Guion-Almeida and colleagues considered this to be a pathogenetically heterogeneous syndrome that represented a regional defect that might not be isolated to a single development field.

The cause of the syndrome still is unknown, though a possible relationship with certain teratogenic drugs cannot be disregarded. Nearly all cases of median cleft facial syndrome occur sporadically with no evidence for a genetic basis; however, a few familial cases have been reported. Ultrasonography is the only efficient technique for diagnosing this syndrome prenatally.

Since the first description of median cleft facial syndrome in 1967, many cases have been reported in the literature; however, no researchers have studied the resulting dentomaxillary anomalies in depth.

In this article, we present the case of a young girl who had a clinical diagnosis of median cleft facial syndrome. Specifically, we analyzed her oral alterations, which had not been studied in detail.

**CASE REPORT**

The girl was 4 years and 7 months old when she arrived at our clinic. She was born after a full-term pregnancy without complications. Her weight and Apgar scores were normal. The psychomotor development of the patient at the time we initially saw her and at follow-up visits was normal for her age. There was no history of maternal exposure to teratogenic drugs during pregnancy. Family history did not reveal any ocular alterations or any similar developmental defects in other family members. We found no extrafacial anomalies.

The patient had ocular hypertelorism, a broad nasal root with a grooved midline, absence of the nasal tip and low-set ears (Figure 1). The patient did not have a cleft palate or lip; however, the intraoral examination revealed other anomalies such as fusion of a primary maxillary incisor with a supernumerary tooth (Figure 2), a supernumerary primary mandibular incisor (Figure 3) and areas of hypomineralization on the vestibular surfaces of both primary maxillary canines. A panoramic radiograph revealed a supernumerary permanent mandibular incisor, ectopic eruption of permanent maxillary first molars, and agenesis or delayed formation (because of her age, we could not be sure) of permanent maxillary second molars (Figure 4).

**Figure 1. A girl aged 4 years and 7 months who had a clinical diagnosis of median cleft facial syndrome, as exhibited by ocular hypertelorism, a broad nasal root with a grooved midline, absence of the nasal tip and low-set ears.**

**Figure 2. Fusion of a primary maxillary incisor with a supernumerary tooth.**

Anterior cranium bifidum occultum can be present in all four types of facies.

Median cleft facial syndrome results from arrested development of the normal frontonasal process as a form of field defect. This alteration most likely occurs between days 21 and 70 of gestation. The earlier the alteration occurs, the more
DISCUSSION

Although the majority of the cases of patients with this syndrome are sporadic, autosomal dominant inheritance in some families has been suggested,\textsuperscript{17,18} while other studies have suggested multifactorial transmission (genetic susceptibility plus exogenous factors in utero equal median cleft facial syndrome)\textsuperscript{21} or polygenic mechanism of inheritance.\textsuperscript{16} The familial occurrence of the full syndrome is quite rare; however, individual features of the syndrome do tend to be more frequent in various members of the same family.\textsuperscript{1} In our case, we did not find that any other members of the family were affected.

The symptoms of the patient we described corresponded with facies A, the least severe form of the syndrome. The patient was not mentally retarded, which corroborates DeMyer's\textsuperscript{1} theory that when oculocutaneous hypopigmentation is combined with one or more of the median facial anomalies of median cleft facial syndrome and there are no extracranial anomalies, the probability of the patient's intelligence being normal or near normal increases.

The case of median cleft facial syndrome we present was mild. In some cases, the patient may have a cleft of the nasal midline and a cleft lip and palate. In the most severe cases, a deficit in midline frontal bone (cranium bifidum occultum) may be present.\textsuperscript{26} Although encephalocele and teratoma may be associated with this syndrome,\textsuperscript{26,27} only the failure of the midline bony elements to fuse does not cause the soft tissues to protrude. Apparently, if no pressure or active vector exerts itself, the skin alone confines the normal meninges and brain. In severe cases, a gap of several centimeters may exist between the medial margins of the frontal bones. In these cases, the brain and meninges, covered only by skin, usually do not protrude to form a meningoencephalocele, though sometimes it does happen.\textsuperscript{28} Also, a report of a rare variant of this syndrome associated similar craniofacial anomalies with central nervous system malformations.\textsuperscript{29}

In relation to soft tissues, palpebral fissures and eyelid colobomas have been described.\textsuperscript{30} There is a large variability in nose and lip anomalies, from notched broad nasal tip to completely divided nostrils, with hypoplasia to absence of the prolabium and premaxilla with a median cleft lip.\textsuperscript{22} When the intermaxillary segment (prolabium and premaxilla) is entirely absent, the resulting defect, which is roughly rectangular in outline, has been called a "pseudomedian cleft lip." In these cases, the lateral lip segments have the same configuration as in ordinary cases of bilateral cleft lip. When the prolabium and premaxilla are present in part or whole and the cleft is between the medial nasal processes, the defect is triangular, with the apex oriented toward or even reaching the columella.\textsuperscript{1} In this sense, our patient's case was a mild one, as she did not even have a cleft lip; this confirms Tessier's\textsuperscript{2} finding that a continuous vertical cleft does not always lead to the formation of a cleft lip.

Intraoral anomalies accompanying this pathology have not been studied in detail. Castillo and colleagues\textsuperscript{21} referred to enamel hypoplasia in one case they reported, but they did not present details about its extension or localization. Therefore, we were unable to compare their case with the hypoplastic areas of the enamel in the patient in our case, which appeared only on the
vestibular surfaces of the primary maxillary canines. We have not found any other cases of median cleft facial syndrome with alterations in the number of teeth (for example, fusions, supernumeraries or agenesis) or ectopic eruption of some tooth described in the literature. This could be caused by the absence of detailed studies about dental anomalies that can appear in this syndrome.

CONCLUSIONS

Median cleft facial syndrome is a rare pathology that has been studied little at the oral level. The case we described concerning this pathology showed alterations in the number of teeth and ectopic eruption of some teeth.

We believe that dentists should know the possible oral alterations that can appear in conjunction with this syndrome and be involved in the treatment of these patients, forming part of the multidisciplinary team that treats them.

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