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CLINICAL PHOTOGRAPH

Frontonasal Dysplasia

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A 34+6 week gestational age female was born via C-section due to fetal distress and admitted to the neonatal intensive care unit endotracheally intubated for respiratory failure. At that time she was noted to have abnormal facies and a malformed right lower extremity. The author's service was consulted for her respiratory failure and evaluation of her facial deformities. Physical examination revealed a broad nasal root and widely spaced eyes, a midline deficit of the frontal bone, lack of a bony or cartilaginous nasal dorsum, completely divided nostrils, and the absence of a nasal tip (Fig 1). A pediatric nasopharyngoscope could not be passed through either nostril beyond 25 mm. A 3-dimensional reconstructed computed tomography scan revealed marked frontonasal dysplasia with orbital hypertelorism, near complete absence of the frontal bone, flattening and partial absence of the nasal bones, a deficiency of the maxilla, a posterior encephalocele, and enlargement of the posterior and anterior fontanelles. In addition, her nasal vault was not patent. Magnetic resonance imaging showed a midline intracranial lipoma; the brain was otherwise normal. She underwent a tracheostomy on postdelivery day 11 for ongoing respiratory support. In addition, she required placement of an orogastric and subsequently an open gastrostomy tube to compensate for feeding difficulties related to poor oromotor function. High-resolution cytogenetic analysis revealed a female karyotype with an apparently normal banding pattern. Her echocardiogram was also normal. Radiography of her right lower extremity confirmed absence of the tibia on that side. Procedures to repair her facial deformity were deferred until a later time.

DISCUSSION

Frontonasal dysplasia (FND), also referred to as median facial cleft or frontonasal dysostosis or malformation, is an unusual congenital anomaly of unknown etiology, typically with a sporadic occurrence; however, it can show a familial pattern. This condition may also be seen as part of a broader syndrome, such as oculoauriculofrontonasal syndrome, and its recurrence risk in such cases would be that of the underlying syndrome.[1,2] Clinically, a constellation of anomalies, which can range in severity and affect the eyes, forehead, and nose, can be identified. These include orbital hypertelorism, a widow's peak, and cranium bifidum occultum (deficit in the midline frontal bone), as well as broad nasal root.[1,2,3] Numerous facial cleft classification systems exist, but the Tessier classification is most widely referenced. The abnormality illustrated represents a Tessier 0/14 cleft.[4] An unknown midfacial primary defect appears to be responsible for this condition. When the nasal capsule fails to properly develop, the primitive brain vesicle fills the space it normally occupies. This produces the anterior cranium bifidum occultum, the arrest in positioning of the eyes, and the lack of formation of the nasal tip. The relationship between the abnormalities seen in the frontal area and those in the nose appears to result from disruption of mesenchymal flow to the nose as it passes through the frontonasal process. The widow's peak scalp-hair anomaly is believed to be the result of the ocular hypertelorism because the 2 hair-growth suppression fields are further apart than usual.[3]

Other nasal findings may range from a notched broad nasal tip to completely divided nostrils with hypoplasia and even absence of the prolabium and maxilla with a median cleft lip. In addition, variable notching of the ala is described. Occasionally associated abnormalities include accessory nasal tags, low-set ears, conductive hearing loss, mild to severe retardation, basal encephalocele, and agenesis of the corpus callosum. Importantly, a high incidence of ocular abnormalities is described. More distant anomalies include tetralogy of Fallot, absence of the tibia, and others. When hypertelorism is severe or when extracephalic anom-

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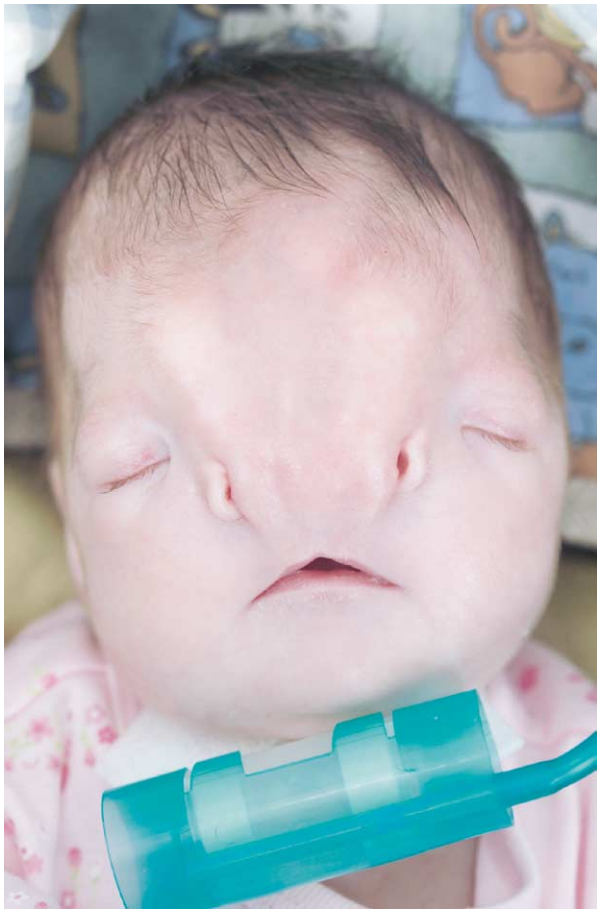

Figure 1 Frontonasal dysplasia.

alies occur, mental deficiency appears to be more likely and more severe.[1,2,3]

Because several of the component anomalies, such as hypertelorism, median cleft lip, and anterior encephalocele, can be detected by prenatal ultrasound, it is possible to diagnose this condition prenatally.[3]

Many conditions, such as craniofrontonasal dysplasia, nasopalpebral lipoma-coloboma, Opitz syndromes, and others, display hypertelorism. In addition, a bifid nose may occur without hypertelorism. A facial phenotype similar to FND may be seen with a large anterior abnormality affecting the frontonasal region, such as an intracranial cyst or hamartoma. It is important for physicians to consider these in their differential diagnosis.[3]

Since there is a significant chance of associated findings, a careful, comprehensive physical examination is important. Because this condition may follow a familial pattern and chromosomal abnormalities in those with FND have been reported, consultation from a geneticist can be valuable. Early assessment by an ophthalmologist is also indicated because of the high incidence of ocular abnormalities in these patients.[3]

Surgical management occurs in stages. Establishing an airway is the priority since newborns are obligate nasal breathers. Computed tomography allows visualization of the full extent of the deformities and is critical to avoiding surgical complications. The facial bipartition technique is widely used to correct the hypertelorism and the broad nasion. There does not appear to be a consensus on the best timing for surgical correction; however, many feel that 6-8 years is a suitable age because the cranial vault and orbits are about 90% of their final size and the child can enter grade school with an improved body image. Thereafter, rhinoplastic surgery is performed to address any remaining nasal abnormalities. Finally, many children will also require orthognathic surgery for maxillary hypoplasia at an age of skeletal maturity (13-16 years old).[5] Because all parts of the face are present in this anomaly, a good functional and morphologic result is obtainable.

In summary, FND is a congenital condition with a broad spectrum of phenotypic features and is associated with significant malformations and genetic anomalies. The range of systems and structures affected requires a multidisciplinary approach. Short-term management focuses on the airway and feeding. Surgical correction of this condition is arduous but possible.

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