

Frontofacionasal Dysplasia: Another Observation

Rabah M. Shawky¹, Doaa I. Sadik²

¹Pediatric Department, Ain Shams University, ²Medical Genetics Center, Ain Shams University

ABSTRACT

Frontofacionasal dysplasia (FFND) is a rare group of disorders, characterized by ocular hypertelorism and frontonasal process anomalies in which clinical and etiological heterogeneity have been recognized since the first review by Gollop 1981.¹ Frontofacionasal dysplasia is inherited as an autosomal recessive genetic trait.

We report on a 10 month old male whose parents are non consanguineous. The patient has severe craniofacial anomalies characterized by: hypertelorism, unilateral (Right sided) malformed eye, lagophthalmos, irregular Sshaped palpebral fissures, deformed nostrils, hypoplastic nasal wing, cleft lip, cleft palate and meningocele. This association of anomalies suggests the diagnosis of frontofacionasal dysplasia and in our case is associated with facial hemangioma. To our knowledge, facial hemangioma in association with FFND have not been described before.

Key Words:

Hypertelorism, facial hemangioma, frontofacionasal dysplasia.

Corresponding Author:

Rabah M. Shawky
E-mail: rabahshawky@hotmail.com

INTRODUCTION

Frontofacionasal dysplasia (FFND) is a term that Gollop used in 1981¹ to describe a constellation of findings limited to the face and head. Frontofacionasal dysplasia (FFND) is considered as subtype of frontonasal dysplasia (FND) which is the hallmark of several syndromes involving the frontonasal process that includes: isolated frontonasal dysplasia (MIM136760 and 305645), acrofrontofacionasal dysostosis 1 (MIM 201180), acrofrontofacionasal dysostosis 2 (MIM 201181), frontofacionasal dysplasia (MIM 229400), oculoauriculofrontonasal

syndrome (MIM601452), among other related conditions.²

In frontofacionasal dysplasia, there is usually marked hypertelorism, a widow's peak, cranium bifidum occultum, a bifid nose and a cleft lip and palate. The nasal alae are hypoplastic and there is often mid facial hypoplasia. However, the eye signs are more marked than in frontonasal dysplasia, in that there are often eyelid colobomas, small palpebral fissures with prominent blepharophimosis, Sshaped palpebral fissures, and a limbic dermoid of the

eye. Other ocular features include small eyes, iris colobomas and cataracts.

A frontally situated lipoma, causing a swelling at the nasion, was reported in one patient and an encephalocele has been described.³ Mental retardation has been reported in one patient with Frontofacionasal dysplasia.⁴

Al Gazali et al.⁵ reported a case with severe frontofacionasal dysplasia associated with multiple skin appendages.

Frontofacionasal dysplasia is inherited as an autosomal recessive genetic trait.⁶

CASE REPORT

We report a case 10 months old male first in order of birth born of a non

consanguineous parents with no other affected family members. The mother's pregnancy and delivery were uneventful and he is born at fullterm by normal vaginal delivery. There was no significant developmental delay. The patient had no history of seizures. The patient's intelligence is normal.

The weight is at the 50th centile and height is between the 5th and 10th centile.

He has severe craniofacial anomalies characterized by, asymmetric face, wide forehead, marked hypertelorism, broad nasal root with a nasal groove associated with absent nasal tip, anteverted nares, hypoplastic alae nasi, narrow right side nasal opening, a unilateral (Right sided) thin upper lip associated with cleft lip operated upon (Figure 1a), cleft palate (Figure 1b), low

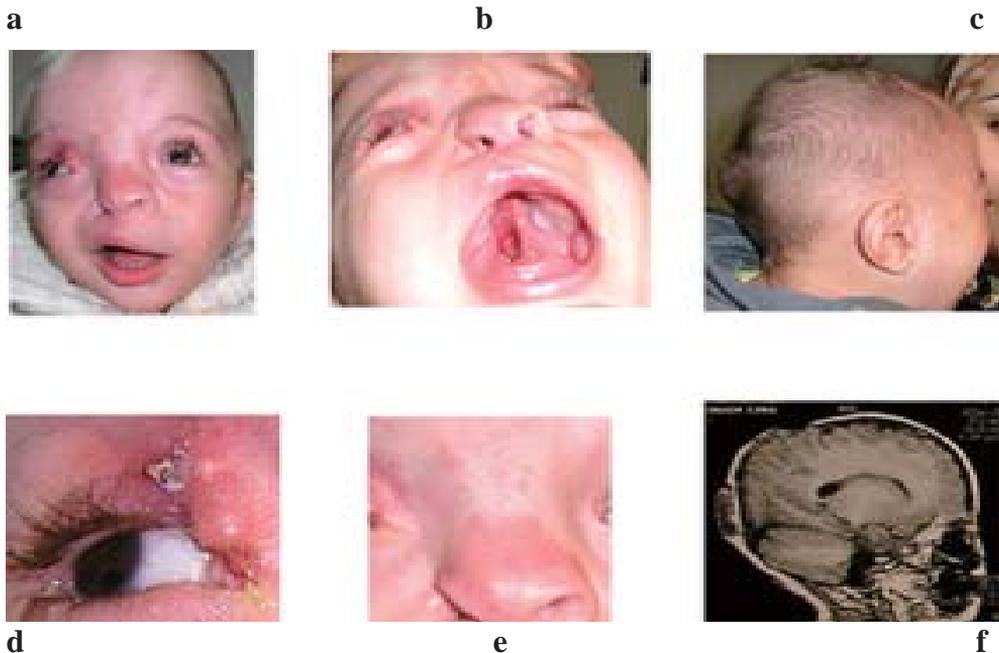


Fig. 1: Clinical features of FFND. (a) Facial view showing marked hypertelorism divergent squint, and central nasal groove. (b) Cleft palate. (c) Meningocele. (d) Eyelid and iris coloboma. (e) Facial haemangioma. (f) MRI showing bony defect, and meningocele.

set ears, prominent tragus of right ear and microbrachycephaly, bony scalp defects and a posterior meningocele (Figure1c).

The ophthalmic manifestations are unilateral (Right sided) microphthalmia, downward slanted irregular S-shaped palpebral fissures, eyelid ptosis, lagophthalmos, divergent squint, microcornea and sparse eyelashes. There are coloboma affecting three areas of the right eye; in the eyelid, in the iris; in the inferonasal quadrant of the eye (Figure1d), and in the choroid (Sector 37). There was right sided dermoid cyst removed from periorbital area.

There is also superficial facial haemangioma extending from lower forehead downward to cover nose, (Figure1e).

CT scan and MRI of the head and facial bones showed posterior parietal meningocele (Containing only CF inside) is seen measuring 2cm in diameter with underlying midline bony defect 1cm, widened anterior fontanel, and defective upper alveolar ridge at right para median location about 1cm width associated with defective soft tissue formation of adjacent lip and nose, and narrow nasal fossa anteriorly. There is no paranchymatous brain focal lesions, or shift of midline structures, otherwise normal ventricular system and other CFS spaces (Figure1f).

Findings from a skeletal survey were unremarkable, and karyotype was normal.

DISCUSSION

Frontofacionasal dysplasia is a rare cause of facial clefts that is apparent at birth. The syndrome is characterized by paramedian facial clefts which involve the nose and palpebral fissures resulting in defects of the alae nasi and blepharophimosis, lagophthalmos, and Sshaped palpebral fissures. In addition affected children have ocular malformations such as epibulbar dermoids and colobomata of the iris or optic disk and may have a posterior encephalocele; these features distinguish this condition from frontonasal dysplasia and early amnion rupture sequence.⁷

Taking into account the above considerations, as well as and the number of reviewed papers, the clinical delineation of the present condition can be established as frontofacionasal dysplasia.

The main features of the present case were severe hypertelorism, downward slanted palpebral fissures, a grossly deformed nose, cleft lip and palate in association with facial haemangioma.

Our case has unilateral features on the right side as microphthalmia, coloboma of the eyelid, iris, and chroid, divergent squint, microcornea, dermoid cyst narrow nasal opening, thin upper lip, and cleft lip. Unilateral craniofacial clefts are usually assumed to have a low recurrence risk, however frontofacionasal dysplasia is an autosomal recessive condition and must be considered in any child with paramedian facial clefts.⁷

Facial haemangioma has not been reported previously in association with frontofacionasal dysplasia, and this suggests the diagnosis of a «New» type of frontofacionasal dysplasia.

The absence of parental consanguinity in the present case cannot exclude autosomal recessive inheritance. Frontofacionasal dysplasia appears to be inherited as an autosomal recessive trait. It is suggested that a more likely candidate for mutations of human TBX15 (T box 15) will have frontofacionasal dysplasia.⁵

Karyotype was normal in our case. Habecker et al.⁸ described a newborn boy, whose karyotype was 46, XY, t (8;12) (q22; q21). Prenatal diagnosis of multiple craniofacial anomalies had been made. Following delivery, the patient was thought to exhibit findings consistent with a diagnosis of frontofacionasal dysostosis. They hypothesized that one of the break points of this translocation may involve a gene essential to craniofacial development.

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