International Journal of Research in Medical Sciences
Sharma R et al. Int J Res Med Sci. 2017 Oct;5(10):4640-4642
www.msjonline.org
pISSN 2320-6071 | eISSN 2320-6012

DOI: http://dx.doi.org/10.18203/2320-6012.ijrms20174612

Case Report

Frontonasal dysplasia- a rare case report

Reena Sharma[1], Poojan Dogra[1], Kapil Malhotra[1]*, Vivek Kaushal[2]

[1]Department of Obstetrics and Gynaecology, SLBS GMC Mandi at Nerchowk, Himachal Pradesh, India
[2]Department of Obstetrics and Gynaecology, Dr. RPGMC Tanda at Kangra, Himachal Pradesh, India

Received: 02 August 2017
Accepted: 07 September 2017

***Correspondence:**
Dr. Kapil Malhotra,
E-mail: kapilmalhotra119@gmail.com

ABSTRACT

Frontonasal dysplasia (FND) is a rare malformative complex affecting the frontal portion of the face, the eyes and the nose; it may occur singly or associated with other clinical signs. We report here a rare case of a full-term male baby who presented with features of FND. There was no history of consanguinity, no positive family history. Antenatal ultrasonography was normal. Though this baby did not survive because the defects were not compatible for the survival. But the developing nations still have handicap in the management of such cases in term of fiancés, surgical correction of such major defects, education and social support in these patients.

Keywords: Frontonasal dysplasia, FND, Facial cleft, Neonate

INTRODUCTION

Frontonasal dysplasia (FND) is a congenital malformation of the midface.[1] For the diagnosis of FND, a patient should present at least two of the following characteristics: hypertelorism (an increased distance between the eyes), a wide nasal root, vertical midline cleft of the nose and/or upper lip, cleft of the wings of the nose, malformed nasal tip, encephalocele (an opening of the skull with protrusion of the brain) or V-shaped hair pattern on the forehead.[1] The cause of FND remains unknown. FND seems to be sporadic (random) and multiple environmental factors are suggested as possible causes for the syndrome. However, in some families multiple cases of FND were reported, which suggests a genetic cause of FND.[2,3]

Midfacial malformations can be subdivided into two different groups. One group with hypertelorism, this includes FND. The other with hypotelorism (a decreased distance between the eyes), this includes holoprosencephaly (failure of development of the forebrain).[4] In addition, a facial cleft can be classified using the Tessier classification. Each of the clefts is numbered from 0 to 14. The 15 different types of clefts are then subdivided into 4 groups, based on their anatomical position in the face.[5] There are multiple classification systems for FND.[6] None of these classification systems have unraveled any genetic factors as the cause of FND. Yet, all of them are very valuable in determining the prognosis of an individual.

Sedano classification

This is a classification based on the embryological cause of FND.

De Myer

This classification is based on the morphologic characteristics of FND, which describes a variety of phenotypes.

Both classifications are further described in Table 1. This table originates from the article 'Acromelic frontonasal dysplasia: further delineation of a subtype with brain

malformations and polydactyly (Toriello syndrome), Verloes et al.[6]

Table 1: Phenotypic classifications of the face in frontonasal dysplasia.

De Myer classification (slightly expanded)	Characteristics
Type 1	hypertelorism, cranium bifidum, median cleft nose, and cleft prolabium
Type 2	hypertelorism, cranium bifidum, and cleft nose but intact prolabium and palate
Type 3	hypertelorism, median cleft nose, and median cleft of notched lip
Type 4	hypertelorism and median cleft nose
Each type may be then subdivided in:	
Subtype a	the two sides of the cleft nose are set apart
Subtype b	the two sides of the nose remain continuous. The cleft of the nose involves the nasal septum and extends to the tip of the nose
Subtype c	the cleft does not reach the tip of the nose. hypertelorism is borderline
Sedano-Jirásek classification	**Characteristics**
Type A	hypertelorism, median nasal groove, and absent nasal tip
Type B	hypertelorism, median groove or cleft face, with or without lip or palate cleft
Type C	hypertelorism and notching of alae nasi
Type D	hypertelorism, median groove or cleft face, with or without lip or palate cleft and notching of alae nasi

CASE REPORT

We are reporting here a case of frontonasal dysplasia (FND). A male neonate of 2.8Kg, delivered normally at term by 22 years old primigravida. There is no history of consanguineous marriage and no family history of any genetic disease or such disorders. Antenatal period was uneventful.

Figure 1: A 2.8 Kg Male neonate, case of FND having a split forehead; two eye anlagen; ocular hypertelorism; one nostril; median cleft nose; bilateral choanal atresia and a median facial cleft affecting the upper lip and palate.

No history of any teratogen exposure. On examination, he was found to have a split forehead; two eye anlagen; ocular hypertelorism; one nostril; median cleft nose; bilateral choanal atresia and a median facial cleft affecting the upper lip and palate (Figure 1). No other congenital anomaly was seen on gross examination. In our case, due to inability to do postmortem we were unable to look for detailed structural anomalies of central nervous system and other organs. Soon after birth baby developed respiratory distress and required resuscitation so shifted to nursery. Poor prognosis of neonate and treatment options were explained in detail to the parents but they decided to give consent for do not resuscitate (DNR). Baby died within 24 hours of life. Genetic counselling done but chromosomal studies were not done due to absence of facilities.

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. It has also been suggested that a defect involving chromosomes 3q23, 3q27, 7q21 and 11q21 might play a role. In our case, chromosomal studies were not done due to absence of facilities. The parents of an affected child can expect the risk to be 25% for the next child.[7] The embryonic origin of FND is in the period prior to the 28-mm crown-rump length stages. During the 3rd 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-naso-ethmoidal complex to freeze in the fetal form.[8] Prenatal diagnosis is important with ultrasound observation of craniofacial anomalies (holoprosencephaly). At birth presence of two or more of the following symptoms is considered positive for FND: A skin covered gap in the bones of the forehead (anterior cranium bifidum occultum); hypertelorism; median cleft lip; median cleft nose; and/or any abnormal development of the centre (median cleft) of the face. Diagnostic evaluation ranges from a simple X-ray of the skull to genetic characterization. Computed tomography is the standard study for the evaluation of these patients.[9] Genetic counselling of the parents is an important part of the management strategies. Cosmetic surgery to correct the facial defects is recommended. In severe cases, additional facial surgeries may be required. These include reformation of the eyelids (canthoplasty), reformation of the orbits (orbitoplasty), surgical positioning of the eyebrows, and rhinoplasty. In FND, early and continuing intervention programs are necessary to assist the affected individual.[10]

CONCLUSION

FND is known for its rarity. The exact aetiology is unknown. Lack of adequate diagnostic and treatment facilities, socio-economic factors and cultural taboos make the management difficult in developing nations.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Lenyoun EH, Lampert JA, Xipoleas GD, Taub PJ. Salvage of calvarial bone graft using acellular dermal matrix in nasal reconstruction and secondary rhinoplasty for frontonasal dysplasia. J Craniofac Surg. 2011;22(4):1378-82.
2. Wu E, Vargevik K, Slavotinek AM. Subtypes of frontonasal dysplasia are useful in determining clinical prognosis. Am J Med Genet A. 2007;143A(24):3069-78.
3. Fryburg JS, Persing JA, Lin KY. Frontonasal dysplasia in two successive generations. Am J Med Genet. 1993;46(6):712-4.
4. Vaccarella F, Pini Prato A, Fasciolo A, Pisano M, Carlini C, Seymandi PL. Phenotypic variability of Pai syndrome: report of two patients and review of the literature. Int J Oral Maxillofacial Surg. 2008;37(11):1059-64.
5. Fearon JA. Rare craniofacial clefts: a surgical classification. J Craniofacial Surg. 2008;19(1):110-2.
6. Verloes A, Gillerot Y, Walczak E, Van Maldergem L, Koulischer L. Acromelic frontonasal dysplasia: further delineation of a subtype with brain malformation and polydactyly (Toriello syndrome). Am J Med Genet. 1992;42(2):180-3.
7. Bello M, Garandawa H, Mustapha Z, Isa A, Tahir C, Ngamdu YB, et al. Frontonasal dysplasia Sequence: a case report. Nigerian J Paediatr. 2014;41(2):151-3.
8. Median Facial Cleft Syndrome. Available at http://www.mypacs.net/cases/median-facial-cleft-syndrome.
9. Taybi H. Radiology of syndromes and metabolic disorders. Pediatric Surgery. 2nd ed. Chicago, IL: Year Book Medical; 1983:235.
10. Fox JW, Golden GT, Edgerton MT. Frontonasal dysplasia with alar clefts in two sisters. Genetic considerations and surgical correction. Plast Reconstr Surg. 1976;57:553-6.

Cite this article as: Sharma R, Dogra P, Malhotra K, Kaushal V. Frontonasal dysplasia- a rare case report. Int J Res Med Sci 2017;5:4640-2.