Case Report

SEVERE FORM OF FRONTONASAL DYSPLASIA (FND) WITH BILATERAL ANOPHTHALMIA
A Case Report with Review of Literature

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ABSTRACT
Frontonasal Dysplasia (FND) is a rare disorder characterized by deformities of variable severity affecting the craniofacial region. FND with bilateral anophthalmia has not been reported yet in the literature. Maxillofacial surgery should be considered for all patients for whom improvement is possible. But in countries like Pakistan where there are considerable limitations in provision of social services, with economic and educational constraints, correction of such major defects remains a challenging task. In this report in an infant a very severe form of frontonasal dysplasia with bilateral anophthalmia with review of literature is presented.

KEY WORDS: Severe frontonasal defects, Bilateral anophthalmia

INTRODUCTION
The frontonasal dysplasia has variously been termed as median cleft face syndrome, inter-nasal dysplasia, doggennase, bifid nose or as No. 0 cleft.1 FND is a non-uniform malformation complex with a spectrum of anomalies. Abnormalities can be nasal, ocular, facial and others.2 Unilateral absence of eye ball has been reported.3,4 But bilateral anophthalmia is not reported yet with frontonasal dysplasia and our report appears to be first such case. Central nervous system involvement occurs only in extreme cases and it is associated with mental deficiency. There is no coronal craniosynostosis in pure FND.5 Herein we are reporting a very severe form of midline facial cleft with bilateral anophthalmia and central nervous system defects. We were unable to find such a severe anomalies spectrum anywhere. Therefore we can safely label it as new syndrome.

CASE REPORT
A two days old female neonate of consanguineous marriage with no family history of frontonasal dysplasia presented to us with multiple birth anomalies. On examination she was found to have grotesque midline craniofacial defects from absence of premaxilla to central nervous system anomalies.

The craniofacial anomalies included midline defects with absent prolabium and bilateral cleft lips, bilateral cleft palate, and complete nasal coloboma with absence of columella and upper part of nasal septum. Inferior part of
nasal septum and inferior turbinates were visible on gross examination (Fig 1). There was hypertelorism with bilateral anophthalmia. Bony orbits were shallow and had bulging skin covered cystic swellings filled with transilluminant fluid. There were no noticeable ocular movements. Because of inability to do postmortem we were unable to look for detailed structural anomalies of optic discs and eyes. There were bilateral frontal encephaloceles and hydrocephalus. The patient had syndactyly of the first three toes of the left foot (Fig 2). Ultrasound examination of the brain revealed absence of corpus callosum, single frontal lobe and single ventricle.

Because of inadequate diagnostic and treatment facilities and poor prognosis, the child was not offered any form of surgical treatment. Parents were counseled and they decided to take the child home. Baby died after a fortnight.

DISCUSSION

Frontonasal dysplasia complex can be graded from mild to severe. The exact cause of Frontonasal Dysplasia is not known. Females are affected more frequently and severely than males and our patient was also a female. It is believed to be transmitted as an X-linked trait with marked female expression and a milder male expression. The exclusively sporadic occurrence of FND is indicative of unlikely hereditary pathomechanism. Consequently, there is no recurrence risk. However, in families with an affected child, generally malformations tend to occur a little more frequently.6

The embryological origin of this syndrome is in the period prior to the 28 mm crown-rump length stage. It is due to deficient remodeling of the nasal capsule, which causes the future fronto-naso-ethmoidal complex to freeze in the fetal form. Experiments have shown that a reduction in the number of migrating neural crest cells results in these multiple defects. The depth and width of the vertical groove may vary greatly.1,6

Anophthalmia occurs when the neuroectoderm of the primary optic vesicle fails to develop properly from the anterior neural plate of the neural tube during embryological development. The more commonly seen microophthalmia can result from a problem in development of the globe at any stage of growth of the optic vesicle. A balanced translocation t (10;22)(q24;q13) has been reported.1 Proper growth of the orbital region is dependent on the presence of an eye, which stimulates growth of the orbit and proper formation of the lids and ocular fornices. Commonly, a child born with anophthalmia has a small...
orbit with narrow palpebral fissure and shrunken fornices. Our case also had extremely shallow bony orbits. (Fig 1)

Frontonasal dysplasia comprise of a variety of low frequency anomalies of craniofacial region, which include agenesis of corpus callosum, anterior encephalocele or meningocele, bifid nasal tip, choanal atresia, midline cleft upper lip, cleft palate, clefting of nose, hypertelorism, hypoplastic alae nasi, holoprosencephaly, arhinencephaly or hydrocephaly. Short palpebral fissures, fallot’s tetralogy, ectopic kidneys, renal agenesis, hypoplastic or absent tibia, widow’s peak are several other defects that may occur in a patient of FND.\(^6\) The majority of affected individuals have normal intelligence. Unilateral anophthalmia has been reported in few cases.\(^7,8,9\) In addition, an abnormal skin-covered gap in the front of the head (anterior cranium occultum) may also be present in some cases.

Several syndromes include frontonasal dysplasia as one of their manifestation. 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.\(^2\)

Cosmetic surgery is usually justified, depending on the severity of the disorders. Maxillofacial surgery should be considered for all patients for whom improvement is possible. Correction soon after birth is not advisable.\(^10\) Treatment is directed at moving the bony orbits and eyes back together again, and reconstructing the nasal and forehead clefts. These complex procedures require a surgical team composed of a pediatric craniofacial plastic surgical experts, oculoplastic surgeon and neurosurgeon. In developing countries like Pakistan, there are still considerable limitations in provision of social services. These are influenced by the economic and educational constraints. Because of lack of adequate tertiary care services, even common problems like cleft lip and palate sometimes remain unattended until adulthood. Therefore, innovative minor corrections may prove adequate and are readily acceptable by the parents. Still the children with severe deformities continue to suffer and remain neglected in our society.\(^11\)

Religious factors and social customs prevent detailed postmortem to study the internal associated abnormalities in cases of various malformations. This is a severe handicap in learning and understanding the entire spectrum of embryological and structural defects. Sophisticated investigations like C.T and M.R.I. are expensive both for the family as well as for the health service providers for routine use. These factors will continue to limit our research abilities in the conceivable future.

REFERENCES