Anophthalmia Cleft Lip & Palate Plus Syndrome: Case Report

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ABSTRACT

Objective: To report a rare case of newborn male presented with right anophthalmia and left unilateral complete cleft lip and palate.

Key words: Anophthalmia, Cleft lip and palate, Porencephalic cyst.

JRMS March 2016; 23(1): 60-63 / DOI: 10.12816/0023362

Introduction

Anophthalmia - plus syndrome is rarely reported syndrome with anophthalmia or severe microphthalmia, cleft lip / palate, facial clefting and choanal atresia.

Anophthalmia and microphthalmia describes respectively the absence of an eye and the presence of a small eye within the orbit. The birth prevalence of anophthalmia and microphthalmia has been generally estimated to be 3 and 14 per 100,000 populations respectively. Both anophthalmia and microphthalmia may occur in isolation or as part of a syndrome as in one-third of cases. [(1)]

Anophthalmia/ microphthalmia have a complex etiology with chromosomal, monogenic and environmental causes. Identified factors include gestational-acquired infections, maternal Vitamin A deficiency, exposure to x -rays, solvent misuse and thalidomide exposure.[(1)] Rarely, isolated anophthalmia may be inherited in autosomal dominant, autosomal recessive or X linked manner.

Assuming that about half of the cases are sporadic and the other half inherited, the empiric risk for siblings without clear etiology or family history is 10%-15%.[(2)]

Case Report

A new born male, product of normal vaginal delivery (cephalic presentation) with a birth weight of 2.7 kg. The physical examination disclosed an active, vigorous not cyanosed baby with an absent right eye and a left cleft lip and palate, otherwise the cranium was normal, left eye was normal in size (Fig.1).

Fig.1: A baby with an absent right eye & left cleft lip & palate .

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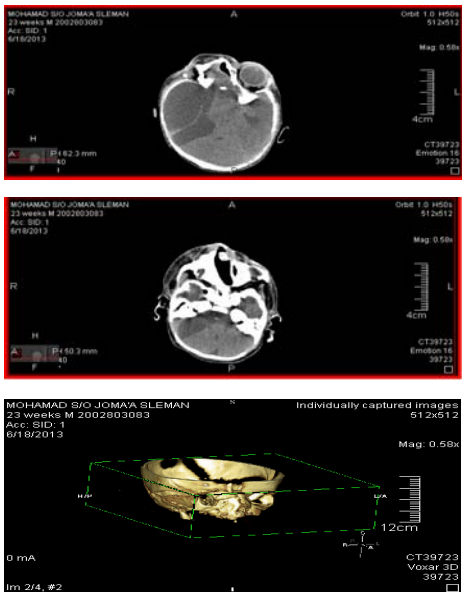
Manuscript received September 4, 2014. Accepted December 4, 2014.

No chest deformity, good air entry bilaterally, normal S1/S2 and no added sounds. Abdomen, back, upper and lower limb examination was unremarkable. The baby was feeding well. The mother is a 31 year old previously healthy lady, G_5P_4 , with no abortions. She is not diabetic nor hypertensive & gave no history of drug exposure during this index pregnancy, there was no history of consanguinity. This baby has three sisters and one brother who are all healthy and product of uneventful pregnancies and normal vaginal deliveries.

Investigations include a skull x-ray which revealed a small right orbital cavity. Orbital US showed absence of the right eye. The abdominal US and the 2.D.Echo were normal. Brain C.T scan revealed a very small deferred right orbital fossa. Absent crista galli, cribriform plate, vomer bone, right great wing of sphenoid bone, and Rt. coronal suture. The Rt. side of the cranium and the frontal bone were small & hypoplastic. There are large two porencephalic cysts communicating with the right lateral ventricle. The larger cyst is Rt. parietal, the other one is Rt. frontal. There is dilatation of occipital horns of both lateral ventricles **(Fig. 2).**

Fig. 2: Brain CT scan revealed a very small right orbital fossa. There are two large porencephalic cysts. The larger cyst is right parietal, while the other one is right frontal



Discussion

Anopthalmia with contralateral cleft lip and palate is an extremely rare anomaly and has never been mentioned in the literature before. We had looked up all the reported cases and compared their physical findings to ours.

In 1995, Frynas [(3)] reported two siblings with previously undescribed features of anophthalmia-plus syndrome. The first case was a 17 weeks gestation female abortus with bilateral anophthalmia, bilateral cleft lip and palate, bilateral lateral facial clefting, nacrotia, open sacral neural tube defect and mullerian duct anomaly. The second was a 2 year old male who had bilateral anophthalmia and an abnormal left ear. The eldest child in this family was 4 year old boy and was morphologically normal. Frynas suggested a possible autosomal recessive pattern of inheritance. This pattern of inheritance could not be assumed in our patient's family as our patient is the only affected member among 3 healthy sisters and one healthy brother.

In the same year Samson and Vilijon mentioned a female infant with unilateral anophthalmia, lateral facial, cleft lip and palate, microtia, clavicular agenesis and asternia.[(4)]

Warburg *et al*, reported a female infant with bilateral extreme microphthalmia, bilateral oblique facial clefts, bifid uvula, bilateral congenital glaucoma, bilateral lower lid colobomas, low set ears, bilateral choanal stenosis, frontal encephalocele and craniosynostosis.[(5)]

Wiltshire et al documented in the literature a boy with bilateral extreme microphthalmia, left facial cleft, bifid uvula, cataracts with anterior segment disorganization, right choanal atresia and an abnormal nose.[(6)]

Akalin *et al* reported a male neonate with anophthalmia- plus syndrome with congenital hypothyrodism. [7] Our patient's thyroid screen yielded an euthyroid status with no proof of thyroid dysfunction. Makhoul reported an infant boy born with bilateral cleft lip and palate, bilateral microphthalmia with posteriorly located lens, a split vitreous body, iris coloboma and glucoma in the right eye and blepharophimosis in left eye, spina bifida, agenesis of sacral vertebrae and coccyx, hypoplasia of corpus callusum with mild dilatation of the lateral ventricles.[8]

Ozcelik reported a 4 year old boy with unilateral complete right cleft lip and palate, absent vomer bone, right anophthalmia, left congenital nystagmus and mental-motor retardation, which are clinically features similar to Fryns anophthalmia- plus syndrome. These combination of findings have not been described together before, so this is either a clinical variation of Fryns anophthalmia- plus syndrome or a new syndrome.[9]

Huang XS reported a 2 year old boy with developmental delay, mild mental retardation and severe craniofacial malformation, including facial asymmetry with hypoplasia of the left zygoma, maxilla, and mandible, and left anophthalmia and anotia.[10]

Günes N reported an 18-day old boy with bilateral cervical cutaneous defect in the retroauricular region, low-set and posteriorly rotated ears, bilateral microphtalmia and bilateral pseudocleft of the upper lip. Clinical findings led to the diagnosis of Branchio-Oculo-Facial syndrome, characterized by branchial defects, ocular anomalies (microphthalmia, anophthalmia, lacrimal duct obstruction) and facial defects (cleft lip and/or palate, pseudocleft or abnormal philtrum).[11]

All the above mentioned anomalous cases share the presence of anophthalmia or microphthalmia and cleft lip and or palate, but most of them are bilateral and if it was unilateral, it was epsilateral, but in our patient the anophthalmia and the facial arch anomaly were contralateral. These features, to the best of our knowledge, were not reported in the medical literature. In addition, the concomitant central nervous system described malformation in this case makes it unique among other similar reported cases worldwide.

The diagnosis of this syndromatic neonate is more in keeping with a variant of Fryns' anophthalmia-plus syndrome. Although the oculocerebrocutaneous syndrome and the rare cerebro-oculo-nasal syndrome are in the differential diagnosis. They are refuted due to the lack of focal dermal hypoplasia and lack of abnormal appearance of the nose and ocular cyst respectively.[12,13]

Conclusion

Combination of right anophthalmia with contralateral left cleft lip and palate with porencephalic cyst is not matched with other reported cases in literature, and may be a new variant of anophthalmia- plus syndrome.

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