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A Case Report On Multiple Congenital Malformations: Anophthalmia Plus Syndrome

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ABSTRACT: Anophthalmia plus syndrome is a very rare syndrome that involves malformations in multiple organs of the body. Fryns and colleagues in 1995 described the condition first. Since that time, only a few cases have been reported with variable clinical presentations.We describe a term female neonate with congenital malformations including anophthalmia, bilateral choanal atresia, microphthalmia, microcephaly and hypertelorism and fused eyelids. Antenatal period was uneventful. Computed tomography reported absence of left eyeball cribriform plate and bony nasal septum.
KEYWORDS: Anophthalmia, choanal atresia,microcephaly,microphthalmia

I. INTRODUCTION:

Anophthalmia plus syndrome is a very rare congenital anomaly syndrome[1].Fryns and colleagues described initially in 1995Since that time a very few cases were reported[1]. It has been suggested that APS is inherited in an autosomal recessive manner,although the genetic cause has not yet been identified[1]. It involves multiple congenital malformations including eyes, ears, nose, face mouth brain meninges, abdominal wall heart fingers and toes[2].

Most common findings include anophthalmia one or both microphathalmia cleft lip palate others includes hypertelorism low set ears narrowed or blocked nasal passages neural tube defects clinodactyly. Based on the few cases reported[1,4,5,6,7,8,9,10] it appears all affected individuals have had anophthalmia and microphthalmia .

Anophthalmia is described in 80-99%, abnormal nasal morphology, bilateral cleft lip and palate choanal atresia, facial cleft, hypertelorism low set ears in 30-79%. Aplasia, hypoplasia of ears, blepharohimosis, deviation of fingers, coloboma, spina bifida vertebral segmentation defect in 5-29%.[2,3]

A review of the available medical literature does not yield information about specific diagnostic criteria for diagnosis of anophthalmia plus syndrome[1,2,3,7,10].

II. CASE DISCUSSION:

Anonehour old baby was referred from peripheral care centre to our hospital with severe respiratory distress. Term female child was born out of a nonconsanguineous marriage. Mother is a primigravidae of age 20yrs residing in a rural village in marathwada region of Maharashtra. Antenatal history was uneventful regular antenatal check-ups done iron and folic acid supplementation taken. There is no history of any fever with rash, drug intake or radiation exposure in the past, no history of any previous miscarriage. All antenatal sonography was reported normal except for third trimester scan which showed ventriculomegaly. There was no history of prolonged labour and baby was b/o normal vaginal delivery. History of meconium stained liquor was present. Baby had a birth weight of 2.5kg, head circumference of 30cm and length of 49cm.

On admission:

HR 189/min RR 80/min spo2 60% on o2 with cyanosis and severe retractions . Chest air entry was bilaterally equal right sided creps.Cvs s1s2 heard normal no murmur present. Per abdomen soft with just palpable liver. Had severe respiratory distress {Downes score 7].Baby was intubated and put on invasive ventilation cyanosis disappeared and maintained spo2 of 98% on mechanicalventilator

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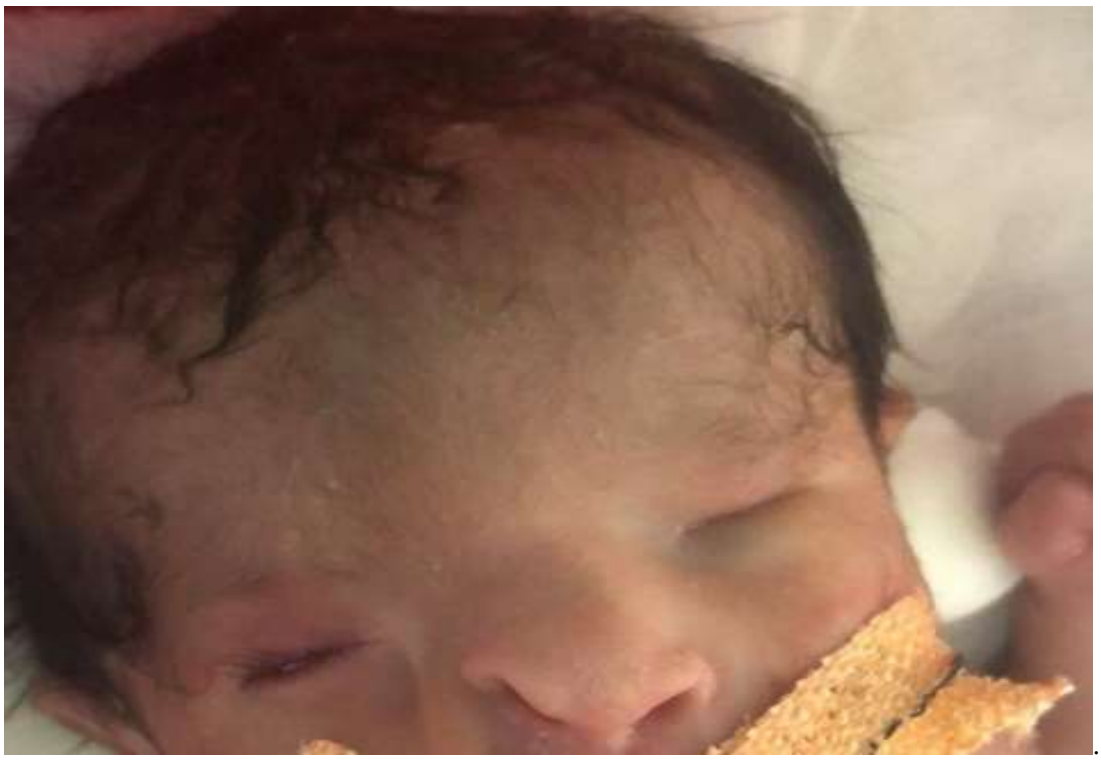

Fig 1- Deformed nose, hypertelorism seen.

Baby had deformed nose was not able to pass feeding tube through both nostrils .CT showed bilateral narrowing of posterior cavity with moderate tissue opacification in bilateral nasal cavities. Bony nasal septum and cribriform plate was not visualised and suggested bilateral choanal atresia.

Fig 2 -Microcephaly and deformed nose evident.

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Hypertelorism was present with anophthalmos on left side and right side eyelids which are inadequately open .USG B SCAN showed right eye globe with axial length of 13mm posterior chamber well visualised with hypoechoic structure with echogenic structure suggestive of lens and left eye globe was not visualised well defined hypoechoic content seen . CT reported with absent left eye globe.

Examined for other morphological features spine, genitalia, external ear, all limbs were normal.

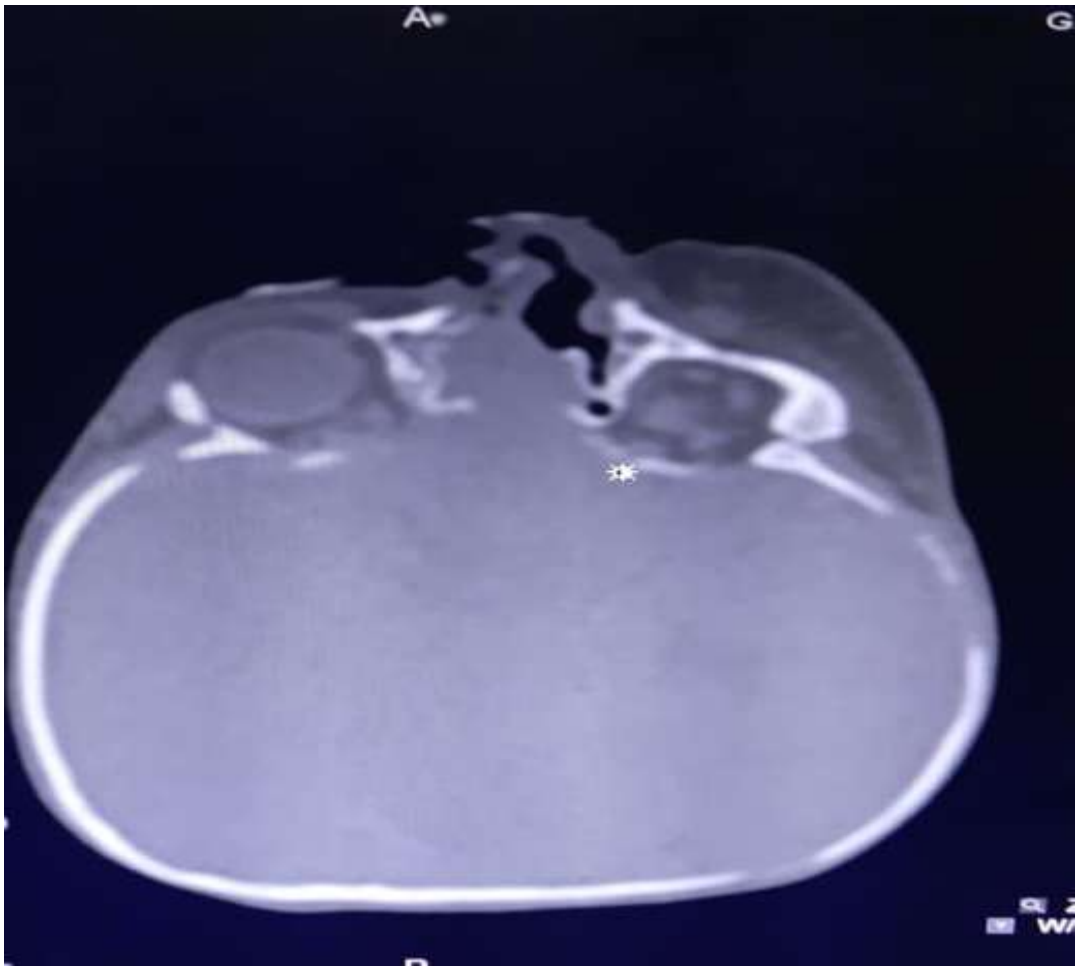

Fig: 3-CT image showing absent turbinates,soft tissue opacification in nasal cavity, choanal atresia.

Chest Xray, USG abdomen and pelvis, spine reported normal . Laboratory evaluation on admission showed complete blood count with leucocytosis { wbc :27000/cumm,N70 L20M4E0} and subsequent cbc showed normal leukocyte count and platelet counts were falling . C reactive protein (32mg/dl) was positive on 3rd day of admission. D-dimer - 4091ng/ml, IL-6 -1845pg/ml, S.ferritin -255.8ng/ml,SGOT -44U/l, SGPT 29U/L, S. Bili total 3.7 and direct 1.1,S. albumin 3.6,S.creatinine 1.1 and S.urea 23mg..

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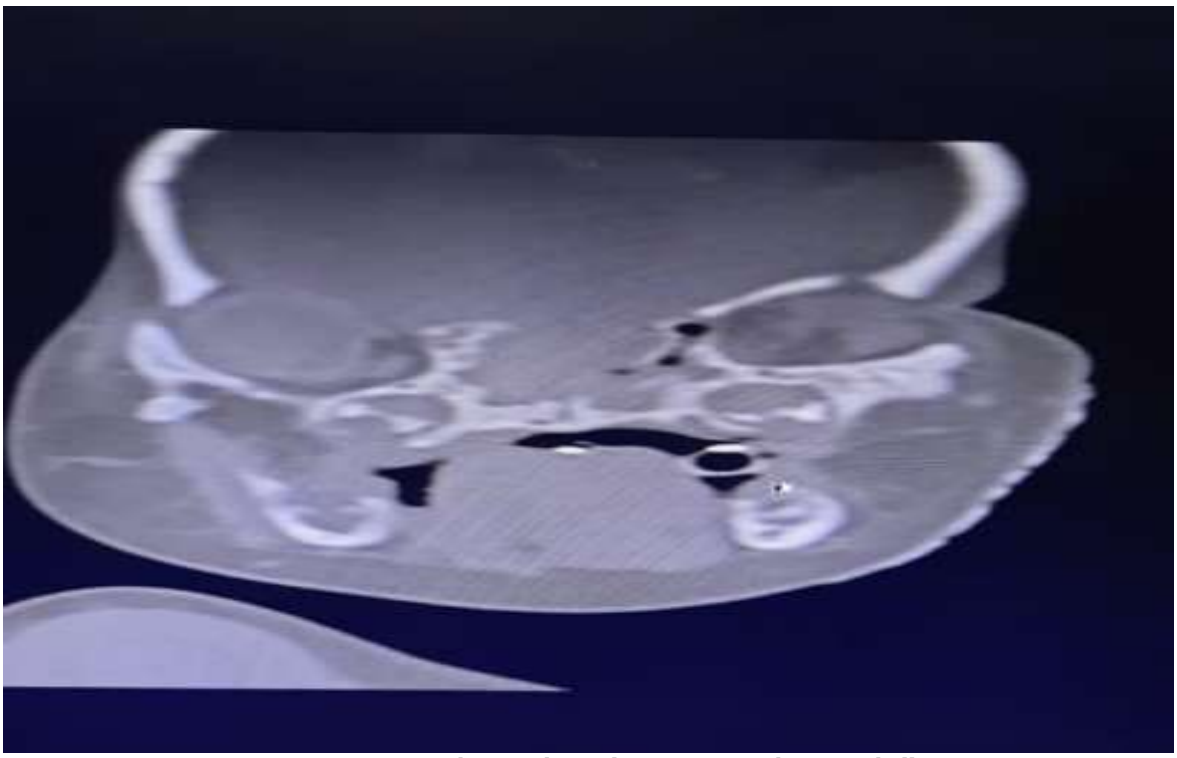

Fig 4- CT image showing hypoplastic sinuses ,absent eyeball

Fig 5 -CT showing absent cribriformplate

III. DISCUSSION.

Neonate described here is a female baby born from NCM presented with multiple congenital malformations consistent with APS including anophthalmia, microphthalmia, hypertelorism, bilateral choanal atresia, fused eyelids, deformed nose and microcephaly.

Imad Makhoul etal described a male baby born from NCM with anophthalmia microphthalmia coloboma fusion of eyelids cleft

lip and palate nasal deformity agenesis of corpus callosum neural tube defects[1]

Fryns etal described a female and male from NCM,normal karyotype with anophthalmia, hypertelorism cleftlip palate neural tube defect and uterus unicornis[4]. Fryns etal suggested that Among all 9 studies published seven studies done karyotyping all of them were normal[4].It has been suggested that APS is inherited in an autosomal recessive manner although genetic cause has not been identified

Akalin etal described a male born from NCM with anophthalmia, microphthalmia fused eyelids cleft lip plate nasal deformity clinodactyly[5]. Arnold etal described a male baby with anophthalmia microphthalmia hypertelorism cleft lip and palate facial cleft low set ears choanal atresia and nasal deformity, microcephaly, vermis hypoplasia, hypoplastic nails midline abdominal wall defects[6]

Leichtnan etal described a case born from NCM and female gender, normal karyotype with anophthalmia, hypertelorism, cleft lip palate hydrocephalus dextrocardia and clinodactyly[7]. Anophthalmia occurs in many known syndromes as reviewed by Leichtman et al[7].

Samson and Viljoen etal described a case with multiple malformations including anophthalmia cleft lippalate facial cleft and midline abdominal wall defect[8].

Warburg etal described a female baby with normal karyotype having anophthalmia microphthalmia eyelid blepharosis cleft lip palate facial cleft low set ears choanal stenosis frontal encephalocele and craniosynostosis[9].

Wiltshire etal described a male baby born out of NCM with normal karyotyping having anophthalmia, microphthalmia, bifid uvula, facial cleft, low set ears ,choanal atresia and nasal deformity[10].

Covid IgG was positive for this baby with raised inflammatory markers including c-reactive protein, D-dimer, IL-6 but there is insufficient data to comment on the status of covid infection. We need further research regarding teratogenicity due to covid infection.

CONCLUSION: Thus we conclude this case as anophthalmia plus syndrome which is a very rare condition.

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