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A Rare Case of Acrocallosal Syndrome in a Neonate – A Case Report

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Abstract: ***Acrocallosal syndrome is characterised by classical craniofacial and digital malformations with partial or complete agenesis of corpus callosum. This is considered as a rare disorder and is included in the Rare Diseases list of the National Institute of Health (NIH), which means there are fewer than 200,000 cases).Here we report one such rare case of acrocallosal syndrome diagnosed in a neonate with characteristic clinical features and neuroimaging findings of partial agenesis of corpus callosum and arachnoid cyst in posterior fossa.***

Keywords: Acrocallosal syndrome, Corpus callosum agenesis, Posterior fossa cyst

1. Introduction

Acrocallosal syndrome (ACS) is a very rare genetic disorder and the condition was first described by Schinzel (1). The term “acrocallosal” refers to involvement of “acra”meaning fingers and toes; “callosal” meaning corpus callosum. Although the mode of inheritance is autosomal recessive, cases often occur sporadically (2). The gene for this syndrome was identified by Pfeiffer,et al as chromosome 12p (3).

2. Case Report

A seven day old full term female neonate was referred to Chengalpattu Medical College Hospital with convulsions and respiratory distress on day 7 of life. Baby was clinically stable during the first 6 days of life. The child was second born of a non-consanguineous marriage, delivered by LSCS in a nearby GH. Birth weight was 2.8kg.The perinatal transition was completely uneventful and there was no significant antenatal insult. There was no history of abortions or any birth defects in the family. The elder sibling is normal and there is no family history of mental retardation.

On clinical examination, baby had dysmorphic facies with low set ears, hypertelorism, depressed nasal bridge, high arched palate. The child had frontal bossing and wide open anterior fontannel. The head circumference was 36.5cm which was normal for age. There was postaxial polydactyly of the left hand and postaxial polysyndactyly of the right foot. Cardiovascular examination revealed a systolic murmur with apex on the left. On chest examination, baby had tachypnea with minimal retractions (attributable to aspiration due to seizures). Abdomen and genital examination were unremarkable. Examination of the central nervous system showed sluggish neonatal reflexes.(fig 1,2)

Baby was evaluated with imaging and blood investigations. USG cranium showed dilatation of all four ventricles. ECHO revealed an ostium secundum type of ASD. Xray chest was normal. MRI Brain showed partial agenesis of corpus callosum with arachnoid cyst in the posterior fossa. (fig 3) Septic screening, metabolic and thyroid profile were negative. Ophthalmic and ENT evaluation were normal. Baby was given supportive management, seizures controlled and parents were counselled.

3. Discussion

Acrocallosal syndrome (ACS) is characterised by multiple congenital anomalies that involve the midline facial structures ,central nervous system and skeleton. There is no reported sex predilection. Some of the features of this syndrome are prominent forehead, large anterior fontanelle, hypertelorism, strabismus, broad nasal bridge, high arch/cleft palate, macrocephaly, mental retardation, agenesis of corpus callosum (partial/complete), hand (pre) postaxial polydactyly, feet pre (post)axial polydactyly, syndactyly etc (4).

A diagnostic criteria was proposed by Courtens et al , which requires 3 out of 4 to arrive at a diagnosis(5). These are: 1. Total or partial absence of corpus callosum; 2. Minor craniofacial anomalies (prominent forehead, hypertelorism, short nose with anterverted nostrils, large anterior fontanel; 3.Moderate to severe psychomotor retardation (with hypotonia) and 4.Polydactyly.

We did not have macrocephaly which is usually described in acrocallosal syndrome. Similar to our case , a normal head circumference in a neonate with ACS was reported by Ravish Singhal et al (6), whereas microcephaly was noted by Naresh et al in their case report(7).Our neonate did not have duplication of hallux or thumb. However our case had craniofacial malformations, partial agenesis of corpus callosum, polydactyly and thereby satisfied 3 of the proposed criteria.

The syndrome needs to be differentiated from other conditions with midline and digital anomalies. These include Greig cephalopolysyndactyly, Meckel-Gruber syndrome, Rubinstein Taybi syndrome, oral facial digital syndrome type 2.

Greig cephalopolysyndactyly has facial dysmorphism, polydactyly, syndactyly, big toe etc. It is usually associated with hydrocephalus .

Meckel-Gruber syndrome is characterised by polydactyly, occipital encephalocele, along with multi-system involvement. Oral facial digital syndrome type 2 is associated with cleft lip/palate, frenula, polydactyly, syndactyly, clinodactyly or brachydactyly.

All the above said syndromes have facial dysmorphism along with digital malformations but are not associated with corpus callosal agenesis.

Rubinstein Taybi syndrome has dysmorphic facies, along with broad thumbs and big broad toes which is a characteristic finding in this syndrome. Cranial malformations with agenesis of corpus callosum is a very rare association.

The condition requires supportive management, with surgical correction of associated malformations like cleft palate, polydactyly, brain cyst/tumors and congenital cardiac defects. Prenatal diagnosis with antenatal ultrasound and mutation analysis can be done but it is limited due to the variability in presentation. Genetic counselling is most important.

4. Conclusion

Acrocallosal syndrome, a condition included in rare disease list has very few reported cases. Recent research has identified homozygous p.N1060S missense mutation in KIF7(15q26.1), which is a regulator of ciliary Hedgehog signaling as a cause of acrocallosal syndromes, Joubert syndrome, fetal hydrolethalus [8]. During embryogenesis, this mutation affects the early development of midline structures. This case is reported for its rarity and for classical clinical and neuroimaging presentation of Acrocallosal Syndrome in a neonate.

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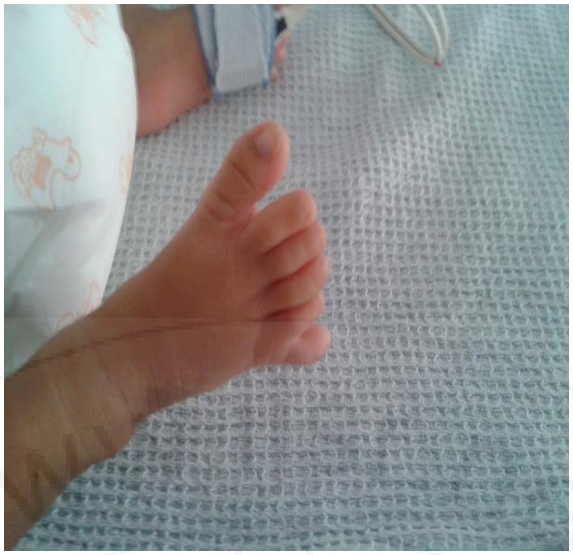
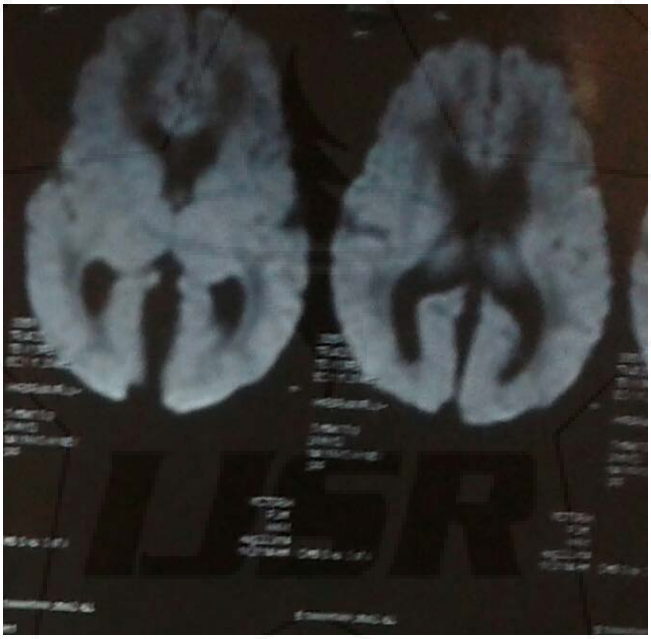


Figure 1: Facial dysmorphism Fig 2: postaxial polysyndactyly



Corpus callosal agenesis