Research Paper Medical Science

Acrocallosal Syndrome- A Case Report

*** Dr.Naresh Kumar Meena**	PG Resident, Deptt. Of pediatrics, GMC Kota * Correspond
Dr.Nishant Aswani	PG Resident, Deptt. Of pediatrics, GMC Kota
Dr.R.K.Gulati	HOD Deptt. Of pediatrics, Govt. Medical College Kota

ABSTRACT

Acrocallosal syndrome was diagnosed in a 9 months female child who presented with, polydactyly and duplication of hallux bilaterally ,microcephaly, low set ears, developmental delay, intracteble seizures.her CECT head revealing partial agenesis of corpus callosum.additionally the index case hadinterhemispheric cyst, right choroid plexus cyst in brain and cortico medullary cyst in right kidney.

KEYWORDS acrocallosal syndrome, corpus callosum,polydactyly.

INTRODUCTION

Acrocallosal syndrome (ACS) is a rare autosomal recessive genetic disorder with partial formation / agenesis of corpus callosum accompanied by craniofacial, digital malformation. Term acrocallosal refers to involvement of acra(fingers and toes) and corpus callosum(1) The gene responsible for this disease has not yet been identified, although Pfeiffer et al suggested that the gene for ACS may be situated on chromosome 12p(2)

Case report

A nine months female child(fig:1) weighing 5 kg presented with a history of fever for 5 days , vomiting for 3 days and intractable convulsions for 1 day which were generalized tonic-clonic in nature. Her mother had no history of medication or exposure to radiation during pregnancy.She was born at term by normal vaginal delivery,birth weight 2.3 kg, 2nd child of non consanguinuos marriage and only affected case in family. Her clinical examination revealed the following:

- Generalized growth retardation with no primary dentition
- Neurological Signs: hypotonia,plantar- extensor
- Craniofacial abnormalities: neurocutaneous marker(multiple tiny hamartomas on right side of face and over right ear).Microcephaly(42 cm,<3SD) with frontal bossing ,large anterior fontanalle, posteriorly angulated malformed ears, small nose with broad nasal bridge, low set ears.
- Limb abnormalities(fig:2): Long tapered fingers, postaxial polydactyly of hands with clinodactyly, 5th finger clinodactyly , hallux valgus duplication of left foot, pre and post axial polydactyly of right foot.

No cardiovascular and respiratory system abnormality seen.

Figure 1

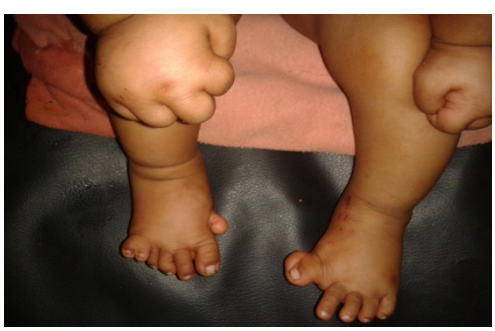
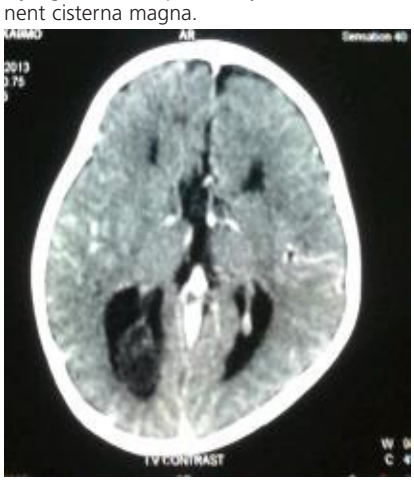


Figure 2

USG findings: Right kidney showing small cortico medullary cyst seen at mid polar region.

Neuroimaging by CECT Head: CECT head revealed partial agenesis of corpus callosum with interhemispheric cyst(size 3.1×2.3×3.0 cm) and hypoplastic interhemispheric fissure ,mild dilated ventricular system with more prominent bilateral occipital horn s/o mild ventriculomegaly with colpocephaly,Right choroid plexuxs cyct(size 1.6 ×2.0 ×1.8) with prominent cisterna magna.



Discussion

The Acrocallosal syndrome is a multi system congenital anomaly involving mainly central nervous system and skeleton. Main manifestations include macrocephaly, large anterior fontanelle, prominent forehead, hypoplasia/agenesis of corpus callosum, post and preaxial polydactyly in hand and feet, syndactyly, mental retardation, small nose, broad nasal bridge, high arch/cleft palate(3) The other less frequent signs such as seizures, cardiovascular and other cerebral anomalies.(3) There is no reported sex predilection.(4) Diagnosis is based on physical examination and, given the high variability of phenotypes, a consensus on minimal diagnostic criteria has been established, with 3 of the 4 following criteria being necessary to suspect the ACS diagnosis: (a) total or partial absence of the Corpus callosum, (b) craniofacial anomalies, (c) moderate to severe psychomotor retardation with hypotonia and(d)polydactyly(2). In India, first ACS was reported in 2003.(5) The patient being reported had partial agenesis of corpus callosum, craniofacial anomalies,hypotonia, polydactyly involving both hands with syndactyly of feet and had following additional features: interhemispheric cyst, right choroid plexus cyst in brain and cortico medullary cyst in right kidney.

The differential diagnosis of ACS includes Greig's Cephalopolysyndactyly syndrome, Orofacial -digital syndromes Type I and II, Meckel-Gruber syndrome, Smith-Lemli-optiz syndrome, Rubinstein-Taybi syndrome.(6)

Management of ACS includes surgical correction of polydactyly, cleft palate, brain cyst/tumors and congenital cardiac malformations. Genetic counseling is of prime importance and antenatal diagnosis can be attempted by mutation analysis and antenatal ultrasound.(7)

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