

# Acrocallosal Syndrome: A Case Report With Review Of The Literature

P Swain, A Thaplial

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## Abstract

Acrocallosal syndrome is a rare genetic disorder that is apparent at birth inherited as an autosomal recessive disorder. It is usually seen in consanguineous parents. The main characteristics of this condition consist of mental retardation, hypotonia, agenesis of the corpus Callosum and pre-axial polydactyly involving both feet and the facial features consist of a prominent broad forehead and hypertelorism. Prognosis is dependent on the degree of hypotonia and early onset of epilepsy rather than the degree of craniofacial and limbs malformations. Neonatal respiratory distress & intercurrent infections are the leading cause of early death in these children. We have reviewed the literature to delineate the syndrome in detail. In our case inguinal hernia was a new finding to this syndrome.

Antenatal diagnosis is possible for better prevention of this genetic disorder. Acrocallosal syndrome should be suspected in any child with polysyndactyly, absence of corpus Callosum, mental retardation and hypotonia.

## INTRODUCTION

The Acrocallosal Syndrome was first described in 1979 in a child with macrocephaly, craniofacial anomalies, hallux duplication, postaxial polydactyly, absence of the corpus callosum, and mental retardation, by Schnizel<sup>(1)</sup> is a rare genetic disorder that is apparent at birth inherited as an autosomal recessive disorder. But some cases often appear to occur sporadically. The main characteristics of this condition consist of mental retardation, agenesis of the corpus Callosum and pre-axial polydactyly involving both feet. Retardation is usually severe, and the facial features consist of a prominent broad forehead and hypertelorism (not usually marked). Post-axial polydactyly of both hands is not unusual and the same abnormality may occur in both feet. Prognosis is dependent on the degree of hypotonia and early onset of epilepsy rather than the degree of craniofacial and limb malformations.

This study was carried out to present & highlight the features of Acrocallosal syndrome with reporting a case with a new clinical feature (inguinal hernia) and review of literatures. This is the first case to be reported from Nepal. Approximately 68 cases have been reported worldwide till date.

## CASE REPORT

A 18 month old male child born to a consanguineous

parents, the mother having polysyndactyly of the left great toe and the father having a cleft lip, from a remote area in western Nepal was admitted to Manipal Teaching Hospital with the following features:

1. cough for 3 days
2. fever for 3 days
3. vomiting for 3 days
4. loose motion for 2 days

Baby was home delivered with birth weight of 3 kg. One of polysyndactyly of left upper limb was operated at the age of 9 months in a private nursing home. There was no H/O seizure. At the time of admission weight was 7kg, length 68cm, mid arm circumference –13cm, head circumference-50cm,pulse –140/min, respiratory rate-42/min, temp- 103F, no signs of dehydration & moderate pallor. Head to toe examination revealed:

1. Open anterior fontanelle (2.0 x 1.5cm)
2. Frontal bossing
3. Macrocephaly
4. Hypertelorism

5. Medial Epicanthal fold
6. Small nose
7. Depressed nasal bridge
8. Bifid tragus
9. Lowset ears
10. High arched palate
11. Both the Upper limbs have 6 fingers (post- axial polydactyly).
12. Both the lower limbs have 6 toes (pre-axial polydactyly).
13. Syndactyly of toes & fingers.
14. Left inguinal hernia (operated)
15. Pallor

Systemic examination showed pan-systolic murmur at lower left sternal border, (16) generalized hypotonia, (17) severe mental retardation, (18) delayed developmental milestones & (19) optic atrophy on fundoscopy. Both the tonsils are Congested & enlarged. All other systems revealed no abnormality.

Investigation showed:

Hb %-6.1gm/dl, TLC -8600/cmm, ESR -03/1st hour, platelet-2.39000/cmm, reticulocyte -4%, serum sodium - 140meq/L, serum potassium - 3.1meq/L, stool & urine within normal limits, no growth on blood culture, no growth on throat swab culture, moderate (20) membranous VSD on 2D-echocardiography, (21) absence of corpus Callosum in CT Scan of brain. The child was managed as a case of acute gastroenteritis, URTI with a provisional diagnosis of Acrocallosal syndrome .Our case satisfies the criteria of the Courten used for the diagnosis of this syndrome (all the 4 criteria's are present).

The patient was treated for acute gastroenteritis & URTI with I.V. fluids, inj. Ampicillin, antipyretic & cough syrup for 5 days in pediatric ward. The child was discharged after 5 days of stay in hospital with out any fresh complaints in a stable condition. The parents were counseled for the above congenital features & also regarding its way of inheritance in future pregnancies.

**Figure 1**

Photo 1: showing hyeprtelorism, depressed nasal bridge& medial epicanthal fold.



**Figure 2**

Photo 2: showing polysyndactyly(6 toes) of both the feet.



### Figure 3

Photo 3: showing polysyndactyly (6 fingers) of both the hands & operated left inguinal hernia.



### DISCUSSION AND REVIEW OF LITERATURE

This child was diagnosed as AGE, URTI with Acrocallosal syndrome by features described earlier in the article. Genetic studies were not done because of non-availability of facility in the region. This child was operated at the age of 9 months for left inguinal hernia.

Review of literatures showed a range of clinical manifestations in this syndrome:

### DEFINITION

The main characteristics of this condition consist of mental retardation, agenesis of the corpus Callosum and pre-axial polydactyly involving both feet, Severe retardation, and the facial features consist of a prominent broad forehead and hypertelorism (not usually marked). Post-axial polydactyly of both hands & feet may be present.

### SYNONYMS

- Hallux duplication,
- Postaxial polydactyly,
- Absence of corpus callosum and
- Schinzel syndrome.

### DESCRIPTION

Schnitzel described Acrocallosal syndrome in 1979. The term Acrocallosal refers to the involvement of the acra (fingers & toes) and corpus Callosum, the thick band of fibres joining the hemispheres of the brain. Reported in both males & females, the cause of the disorder is not known.

### SIGNS & SYMPTOMS

Clinical features as described in textbook(1) are as follows:

### TYPICAL FEATURES

1. Reduced / absent corpus Callosum
2. Severe mental retardation
3. Seizure
4. Strabismus
5. Hypotonia
6. Macrocephaly
7. Prominent forehead
8. Large anterior fontanelle
9. Hypertelorism
10. Medial Epicanthal fold
11. Downslanting palpebral fissure
12. Small nose
13. Depressed nasal bridge
14. Malformed ear
15. Short philtrum
16. Post axial polydactyly of upper & lower limbs
17. Pre-axial polydactyly of feet
18. Syndactyly of toes
19. Tapered fingers
20. Fifth finger clinodactyly
21. Optic atrophy
22. Decreased retinal pigmentation
23. Cardiac defects-septal defects
24. Abnormal pulmonary valves
25. Umbilical hernia
26. intracranial cyst
27. delayed developmental milestones(hypotonia)

## **OCCASIONAL FEATURES;**

1. micropolygyria
2. prominent occiput
3. hyperreflexia
4. nystagmus
5. cleft lip
6. cleft palate
7. supernumerary nipples
8. rib hypoplasia
9. prenatal overgrowth
10. post natal growth deficiency
11. anecephaly.

## **NATURAL HISTORY**

Prognosis is dependent on the degree of hypotonia and early onset of epilepsy rather than the degree of craniofacial and limb malformations<sub>(2)</sub>. Neonatal respiratory distress & intercurrent infections are the leading cause of early death in 15 % of cases.<sub>(1)</sub>

## **ETIOLOGY**

Acrocallosal Syndrome is an autosomal recessive inherited disorder<sub>(3)</sub> Chromosomal location is 12p13.3-p11.2<sub>(4)</sub>. Linkage studies suggest that this disorder is not allelic to Greig Syndrome, despite the similarity in facial appearance and the pattern of polysyndactyly.<sub>(5)</sub>

## **DIAGNOSIS**

In 1997 W. Courtens et al defined minimal diagnostic criteria for Acrocallosal Syndrome<sub>(6)</sub> :

1. Total or partial absence of the corpus callosum;
2. Minor craniofacial anomalies (prominent forehead, hypertelorism, short nose with anteverted nos-trils and large anterior fontanelle);
3. Moderate to severe psychomotor retardation (with hypotonia);
4. Polydactyly.

The presence of 3 of the 4 criteria together with other associated findings could lead one to suspect the diagnosis of Acrocallosal Syndrome. All the 4 criteria's were present including other associated features in our case.

## **THERAPY**

No specific therapy is available. Symptomatic approach & multispeciality care is needed along with genetic counseling. Surgery may improve thumb opposition and shoe wear.<sub>(7)</sub> We observed an atypical finding in our case not reported in this syndrome was inguinal hernia.

## **CONCLUSION**

Acrocallosal syndrome is an autosomal recessive disorder having a wide spectrum of clinical manifestations involving most of the systems. Following this pattern, each child born will have a 25% risk of being affected. It can be diagnosed antenatally & can be prevented. Any child with polysyndactyly and absence of corpus Callosum should be examined in detail to rule out this syndrome.

## **CORRESPONDENCE TO**

DR. PRADIPTA KUMAR SWAIN ASSOCIATE  
PROFESSOR, DEPT. OF PAEDIATRICS, MANIPAL  
COLLEGE OF MEDICAL SCIENCES, MANIPAL  
TEACHING HOSPITAL, PHULBARI, POKHARA-11  
NEPAL. Ph = +977-61-526416, ext-186@, 119(O). Fax =  
+97761-527862 E-mail- pkswain1@rediffmail.com

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**Author Information**

**Pradipta Kumar Swain, M.D. Paediatrics**

Associate professor, Manipal college of medical science

**Anna Thaplial, M.D. Paediatrics**

Prof. & HOD, Paediatrics, Manipal college of medical sciences