

CASE REPORT

THE ACROCALLOSAL SYNDROME: CLASSICAL IMAGING FINDINGS: A CASE REPORT

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ABSTRACT: We reported a case of two days old baby with typical features of Acrocallosal syndrome. The characteristic features of this syndrome are craniofacial abnormalities, distinctive digital malformation and corpus callosum hypoplasia/agenesis. USG cranium and NECT of brain reveals characteristic features of corpus callosum agenesis and posterior fossa arachnoid cyst.

KEYWORDS: Acrocallosal syndrome, corpus callosum hypoplasia/agenesis, posterior fossa arachnoid cyst.

INTRODUCTION: In 1979 Schinzel^[1] described a condition characterized by postaxial polydactyly, hallux duplication, absence of the corpus callosum, macrocephaly, and severe mental retardation, which was subsequently designated 'acrocallosal syndrome' (ACS).^[2]

Acrocallosal syndrome (ACS) is a rare autosomal recessive genetic disorder with hypoplasia/agenesis of corpus callosum, moderate to severe mental retardation, characteristic craniofacial abnormalities, distinctive digital malformations and growth retardation.

The inheritance is autosomal recessive based on the reports of recurrences in families and parental consanguinity. 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.^[3]

CASE SUMMARY: Two days old baby presented with craniofacial malformations and polysyndactyly of foot. Patient was referred to radiology department for further evaluation.

Radiograph of skull AP/lateral view was taken and revealed: Hypertelorism macrocephaly and widened anterior fontanelle.

Radiograph of both the foot taken and Revealed: Preaxial polysyndactyly of both the foot.

USG Cranium was done and revealed: Features of corpus callosum agenesis like widely spaced, parallel oriented lateral ventricles, dilated and elevated 3rd ventricle.

On sagittal midline image the medial cerebral sulci are typically radially arranged, perpendicular to the expected course of the corpus callosum giving characteristic "sunburst" sign.

There is Arachnoid cyst in the posterior fossa pushing cerebellum anteriorly.

NECT BRAIN Revealed: Widely spaced parallel oriented lateral ventricles rather than normal "bow tie" configuration giving characteristic racing car appearance on axial image.

Coronal section shows dilated high riding third ventricle.

Sagittal midline section shows radially arranged gyri perpendicular to expected course of corpus callosum and large CSF density cystic lesion in posterior fossa pushing cerebellum anteriorly-Arachnoid cyst.

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DISCUSSION: The Acrocallosal syndrome is a true multiple congenital anomaly/mental retardation autosomal recessive syndrome, whose pleiotropic effects mainly involve the central nervous system/facial midline structures and skeleton. Main manifestations include macrocephaly, large anterior fontanelle, prominent forehead, hypoplasia/agenesis of corpus callosum, hand (pre) postaxial polydactyly, feet pre (post) axial polydactyly, syndactyly, mental retardation, hypertelorism, strabismus, small nose, broad nasal bridge, high arch/cleft palate.^[4] There is no reported sex predilection.^[5]

Initially it was thought to be an autosomal dominant condition but subsequent reports of consanguinity as well as affected ACS sibs born to unaffected parents, have suggested autosomal recessive inheritance.^[6] The inheritance has been suggested to be autosomal recessive, thus there is a 25% chance of the next child being affected.^[3]

In India, first ACS was reported in 2003.^[3] Pfeiffer et al (1992) reported de novo inverted tandem duplication of 12p11.2 -p13.3 in a child with ACS.^[3] ACS may represent a heterogenous group of disorders that, in some cases, may result from mutation in GL13 and represent a severe, allelic form of Greig's Cephalopolysyndactyly syndrome.^[6]

The differential diagnosis ACS includes Greig's Cephalopolysyndactyly syndrome, Oro-facial-digital syndromes Type I and II, Meckel-Gruber syndrome, Smith-Lemlioptiz syndrome, Rubinstein-Taybi syndrome.^[7]

Management of ACS includes surgical correction of polydactyly, cleft palate, hernia, 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.^[3]

CONCLUSION: ACS is very rare anomaly and so far very few cases are reported. Imaging plays an important role in the diagnosis of corpus callosum agenesis which is distinguishing feature of ACS. Imaging also helps in diagnosis of other features of ACS like arachnoid cyst of brain and polydactyly.

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Figure 1: Two days old baby with craniofacial malformations like hypertelorism, depressed nasal bridge and frontal bossing in addition right hand shows polydactyly.



Fig. 1

Figure 2: Typical pre-axial polydactyly of both foot.

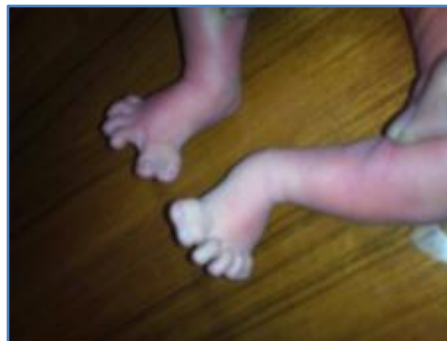


Fig. 2

Figure 3: AP and lateral skull radiograph showing Hypertelorism, macrocephaly and widened anterior fontanelle.

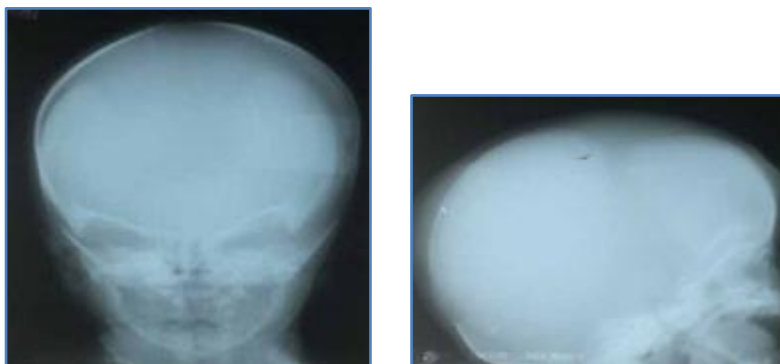


Fig. 3

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Figure 4: Radiograph of both foot showing preaxial polysyndactyly.



Fig. 4

Figure 5: USG Cranium showing features of corpus callosum agenesis like widely spaced, parallel oriented lateral ventricles.



Fig. 5

Figure 6: USG Cranium midline sagittal image showing features of corpus callosum agenesis: the medial cerebral sulci are typically radially arranged, perpendicular to the expected course of the corpus callosum giving characteristic “sunburst” sign. There is arachnoid cyst in the posterior fossa pushing cerebellum anteriorly.

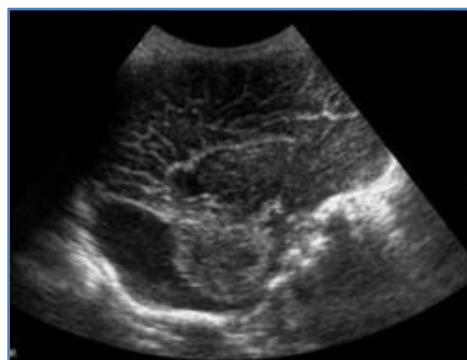


Fig. 6

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Figure 7: NECT Axial image of brain showing widely spaced parallel oriented lateral ventricles rather than normal “bow tie” configuration giving characteristic racing car appearance.



Fig. 7

Figure 8: NECT Axial image of brain showing arachnoid cyst in the posterior fossa pushing cerebellum anteriorly.

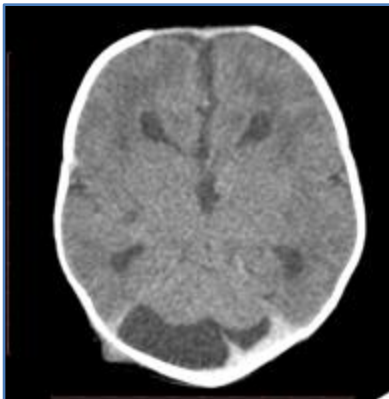


Fig. 8

Figure 9: Coronal NECT image of brain showing dilated and high riding third ventricle.



Fig. 9

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Figure 10: NECT Sagittal image of brain showing radially arranged gyri perpendicular to expected course of corpus callosum and large CSF density cystic lesion (Arachnoid cyst) in posterior fossa pushing cerebellum anteriorly.



Fig. 10

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