



International Journal of Medical Research & Health Sciences

www.ijmrhs.com

Volume 3 Issue 4

Coden: IJMRHS

Copyright ©2014

ISSN: 2319-5886

Received: 25th May 2014

Revised: 30th June 2014

Accepted: 28th July 2014

Case report

ISOLATED COMPLETE CORPUS CALLOSAL AGENESIS

*Jaiganesh S¹, Venkateshwaran A¹, Naresh Kumar C², Rajasekhar KV³

¹Assistant Professor, ²Post Graduate student, ³Professor & Head, Department of Radiodiagnosis, Meenakshi Medical College and Research Institute, Kanchipuram, Tamilnadu, India

*Corresponding author email: drjenesh@gmail.com

ABSTRACT

Isolated complete corpus callosal agenesis is a rare entity. Usually this condition will be an associated finding in other syndromes. 3 month old male child came with complaints of deformed foot on both sides, not having a social smile and neck holding. Patient referred to the Radiology department for MRI brain which showed complete absence of corpus callosum, widely separated and parallelly placed lateral ventricles, colpocephaly, high riding of 3rd ventricle and absence of cingulate gyrus and radial arrangement of gyri along the interhemispheric fissure. Hence it was reported as isolated complete corpus callosal agenesis and this article describes the Embryogenesis, anatomy, developmental anomalies and its clinical manifestations & prognosis.

Keywords: Corpus; Callosum, Colpocephaly, Agenesis

INTRODUCTION

Corpus callosum is the largest commissure in the central nervous system which connects the both cerebral hemispheres¹. Agenesis of corpus callosum is a rare disorder that is present at birth (congenital). Corpus callosal agenesis is found in about 5 per 1000 births.¹ Agenesis of the corpus callosum is an uncommon cerebral malformation that has been reported in 1 in 19,000 unselected autopsies and 2.3% of children with mental retardation.^{2,3} Its absence may be partial or complete, depending on the stage at which callosal development is arrested.⁴ Corpus callosal agenesis can occur as an isolated abnormality or it can be associated with central nervous system or other abnormalities. Prognosis depends on the extent and severity of malformations. Prenatally diagnosed, isolated agenesis of the corpus callosum is usually associated with a favorable outcome.⁵ Mental retardation does not worsen in Corpus callosal agenesis. Isolated corpus callosal agenesis is infrequent with sulcal and infratentorial abnormalities

as common findings.⁶ Sulcation delay was present in most fetuses with corpus callosal agenesis, including those with good neurodevelopmental outcome and suggests a more global white matter dysplasia⁶. Although many children with the disorder have average intelligence and lead normal lives, neuropsychological testing reveals subtle differences in higher cortical function compared to individuals of the same age and education without Agenesis of Corpus Callosum. Here this article describes a newborn having complete corpus callosal agenesis as an isolated abnormality. Unfortunately the mother of this child didn't undergo antenatal ultrasound; hence this abnormality was not diagnosed early.

CASE HISTORY

3 month old male child came to the Paediatric outpatient department our college with complaints of deformed foot on both sides, not having a social smile and neck holding (usually occurs at 3 months of age).

Patient referred to the Radiology department for MRI brain which showed complete absence of corpus callosum, widely separated and parallelly placed lateral ventricles, colpocephaly (narrow frontal horns and wide occipital horns), high riding of 3rd ventricle and absence of cingulate gyrus and radial arrangement of gyri along the interhemispheric fissure. Hence it was reported as isolated complete corpus callosal agenesis.

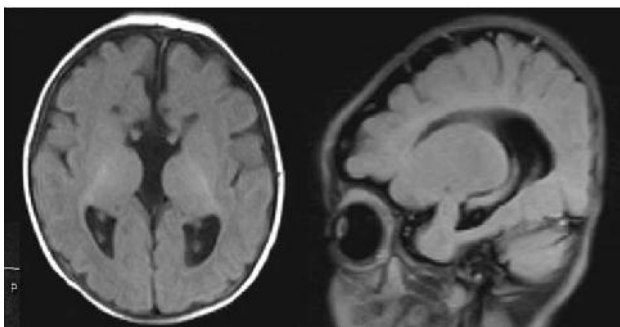


Fig.1: T1W axial and parasagittal image shows dilated occipital horn with pointed frontal horns (colpocephaly)

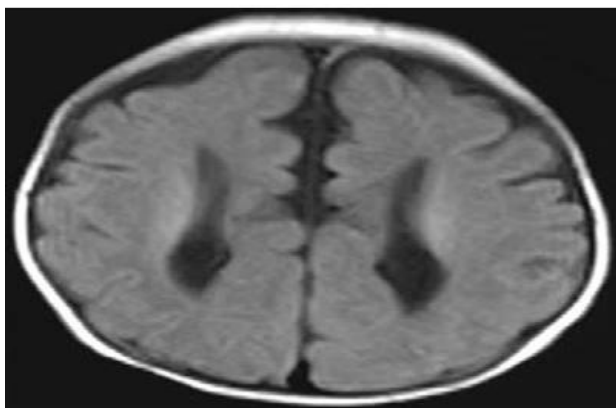


Fig 2: T1W axial image shows widely spaced parallel lateral ventricles and no corpus callosum is seen.

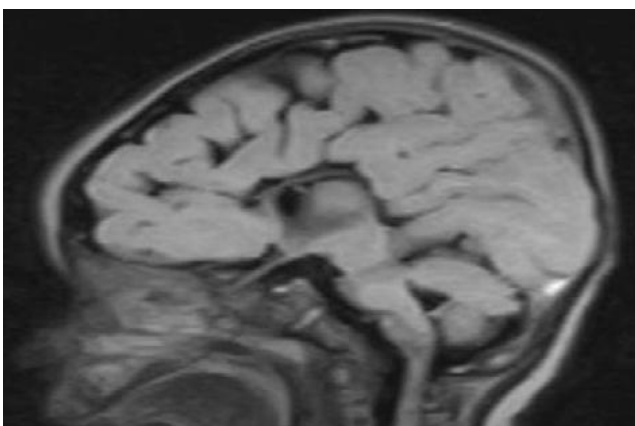


Fig 3: Sagittal FLAIR image shows absent corpus callosum, cingulate gyrus and radial arrangement of gyri (sunburst pattern)

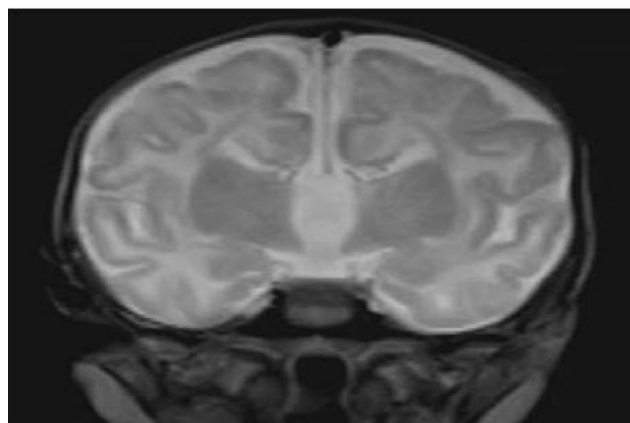


Fig 4: T2W coronal image shows high riding of 3rd ventricle.

DISCUSSION

Embryology: Corpus callosum is the largest neural commissure connecting the two cerebral hemispheres. The development of the corpus callosum occurs between the 12th and 16-20th weeks of gestation.^{7,8} At 12 weeks gestation, it starts to develop from the lamina terminalis near the anterior end of the 3rd ventricle as a bundle of fibers connecting both hemispheres. It begins with the *genu* and then continues posteriorly along the body to the *splenium*. The *rostrum* is the last part to be formed. In primary agenesis parts of the corpus callosum which form before the insult will be present whereas later parts will be absent. Presence of the rostrum essentially excludes primary agenesis.

Normal corpus callosum measures 17 mm at 18 weeks and 44 mm at term.⁷ Callosal agenesis can occur either due to inflammatory or vascular insults around 12-18 weeks.⁸ Agenesis can be partial or complete depending on the time of insult to the development of corpus callosum. Earlier insult leads to complete agenesis while later causes partial agenesis especially affects the posterior part and rostrum. The mechanisms leading to Agenesis of corpus callosum is not very clear; however defective migration of the callosal axons or an abnormality of the callosal neurons has been suggested.⁹ Isolated Agenesis of corpus callosum can be sporadic, autosomal recessive or dominant, or X-linked.⁹ Though multiple chromosomes are responsible for the development of the corpus callosum (1, 8, 13, 15, 18, 21, and X), no definite gene was identified yet for isolated Agenesis of corpus callosum.⁹ Even the condition can be diagnosed during antenatal period,

there is no treatment available to correct this anomaly during antenatal period. Clinical features include intellectual deficit (80%) and/or easily controlled epilepsy and/or behavioural disorders⁹. However, clinical findings vary widely, ranging from asymptomatic cases with normal intellectual capacities to those with severe intellectual deficit⁹. CCA is generally discovered when the child starts school and although febrile seizures appear to be more frequent in CCA patients than in the general population, epilepsy is rare. Behavioural troubles seem to be frequent and may be the first sign of the malformation.

Findings observed such as:¹⁰

- Disproportionate enlargement of occipital horns (colpocephaly).
- Pointed anterior ends of lateral ventricle.
- Lateral displacement of both medial and lateral walls of lateral ventricles.
- Absent cingulate gyrus and sulci.
- Radiating gyri and sulci (sunburst appearance) along the interhemispheric fissure.
- High riding of 3rd ventricle.
- On coronal CT and MR scans the medial borders of the parallel lateral ventricles appear concave and indented by the longitudinally oriented fiber tracts (Probst bundles).
- Sometimes in partial agenesis undeveloped corpus callosum is replaced by fat.

It can be associated with some central nervous system (CNS) abnormalities such as Chiari malformations, arachnoid cyst, heterotopias, cephaloceles, anomalies of neuronal migration including lissencephaly, schizencephaly, pachygyria and polymicrogyria, encephaloceles, Dandy-Walker malformations, holoprosencephaly, and olivopontocerebellar degeneration.¹¹

Or may be associated with some chromosomal aberrations (8, 11, 13-15, 18 and rarely chromosome 6)¹², inborn errors of metabolism or genetic syndromes such as Andermann Syndrome (peripheral neuropathy with corpus callosum agenesis), Aicardi Syndrome (infantile spasms, ocular anomalies and agenesis of corpus callosum), Shapiro's syndrome (paroxysmal hypothermia with agenesis of corpus callosum) and sporadically with foetal alcohol syndrome, Leigh's syndrome.¹³

CONCLUSION

Prognosis for isolated Corpus callosal agenesis is good and have normal developmental outcome but 15% are handicapped. Isolated Agenesis of corpus callosum can even be an occasional finding in the investigation of children with mental retardation or microcephaly. Treatment is symptomatic and consists of physiotherapy, speech therapy, antiepileptic drugs and psychotherapy. Isolated CCA appears to be related to a better prognosis than associated CCA, with up to 80% of isolated CCA cases having a normal outcome. Nevertheless, parents should be informed that learning difficulties (associated with slowness, distractibility and attention deficit) may develop and require appropriate rehabilitation.

Conflict of interest: Nil

REFERENCES

1. Pulu G, Nicolaidis KH. Diagnosis of Fetal Abnormalities. The 18-23 week scan. London: Parthenon Publishing; 1999: 9-10.
2. Grogono JL. Children with agenesis of the corpus callosum. *Dev Med Child Neurol.* 1968; 10: 613–16
3. Freytag E, Lindenberg R. Neuropathic findings in patients of a hospital for the mentally deficient: A survey of 359 cases. *Hopkins Med J.* 1967; 121: 379–92.
4. Sangram Singh and Saurabh Garge. Agenesis of the corpus callosum. *J Pediatr Neurosci.* 2010; 5(1): 83–85.
5. Sotiriadis A, Makrydimas G. Neurodevelopment after prenatal diagnosis of isolated agenesis of the corpus callosum: an integrative review. *Am J Obstet Gynecol.* 2012 Apr 206(4): 337. e1-5.
6. Tang PH, Bartha AI, Norton ME, Barkovich AJ, Sherr EH, Glenn OA. Agenesis of the Corpus Callosum: An MR Imaging Analysis of Associated Abnormalities in the Fetus. *AJNR.* 2009;30: 257-63.
7. Carol MR, Ants Toi Diagnostic ultrasound.. Elsevier Mosby; 2005; vol2, 3rd edition 1132, 1258-60
8. Luis F. Goncalves, Maria Veronica Munoz Rojas, Florianopolis, Agenesis of the Corpus Callosum. Brazil – 2000-08-11-14 www.thefetus.net Updated 08.22.2007 by Frantisek Grochal, MD.

9. Marie-Laure Moutard. Isolated Corpus Callosum Agenesis. June 2007. URL - http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=GB&Expert=200
10. Osborn AG. Diagnostic neuroradiology. St Louis: Mosby. 1994; 1st edition: 29-35.
11. Barkovitch AJ, Norman D. Anomalies of the corpus callosum: correlation with further anomalies of the brain. AJR AM J Roentgenol 1988; 151:171-79.
12. Desai AK, Bhide AG, Bhalerao SA. Agenesis of corpus callosum - a rare case. Journal name. 1999;45(1):20-22
13. Gelot A, Lewin F, Moraine C. Agenesis of corpus callosum - Neuropathological study and physiopathologic hypothesis. J Neurochirurgie 1998; 44:74-84.