

Aggenesis of the Corpus Callosum

Jacob K Jacob

a. Department of Medicine, Cooperative Medical College, Kalamassery, Cochin, Kerala*

ABSTRACT

Published on 30th March 2012

Aggenesis of the Corpus Callosum (ACC) is a rare congenital disorder. There is a complete or partial absence of the corpus callosum. Aggenesis of the corpus callosum occurs when the corpus callosum, the band of white matter connecting the two hemispheres of the brain, fails to develop normally, typically in utero.

A 28 year old male patient admitted with headache, giddiness and loss of consciousness was diagnosed after imaging as a case of ACC.

The diagnosis of callosal aggenesis is by neuroimaging. Magnetic resonance imaging (MRI) is currently the imaging procedure of choice in infants and children with ACC.

There are currently no specific medical treatments for callosal disorders, but individuals with ACC and other callosal disorders may benefit from a range of developmental therapies, educational support, and services.

Keywords: Aggenesis, Corpus Callosum

*See End Note for complete author details

Aggenesis of the Corpus Callosum (ACC) is a rare congenital disorder. There is a complete or partial absence of the corpus callosum. Aggenesis of the corpus callosum occurs when the corpus callosum, the band of white matter connecting the two hemispheres of the brain, fails to develop normally, typically in utero. The development of the fibers which would otherwise form the corpus callosum become longitudinally oriented within each hemisphere and form structures called Probst bundles. Other callosal disorders include hypogenesis, dysgenesis of the corpus callosum, and hypoplasia of the corpus callosum.¹

CASE

Here we discuss the case of a Male Patient 28 years who presented to our casualty with history of headache and giddiness followed by loss of consciousness. He gave a history of piercing type of pain left frontal region lasting for about 1-2 hrs. followed by giddiness. The patient fell down with a staring look and body became rigid with tonic movements. After 5min., patient assumed a partial opisthotonic posture for half a minute or so followed by fluttering & watering of eyes. Then consciousness was regained. No h/o fever, vomiting/trauma/ bleeding/neck rigidity. He had four similar episodes in a span of 8 days. In 1996 at 14yrs. of age he had the 1st episode of seizure and was EEG

taken and diagnosed seizure disorder. He started taking Valproate CR (500mg daily) regularly for an year. In 2009 there was recurrence. In 2010 he had two episodes & he discontinued treatment one month before the present admission due to financial problems. Mother had Diabetes and Hypertension. His Sleep, Appetite, Bladder and Bowel normal. Passed 8th standard. Didn't go for further studies due to the seizure disorder, Was an average student. Worked as a Salesman till 3 months back and was dismissed when he had 2 episodes of seizures at work. On physical examination, he his vitals were stable. Conscious and oriented. Memory – Immediate, Recent , Remote – Intact. Intelligence – Subnormal. No delusions / hallucinations. Speech – (N).Motor System – WNL. Reflexes : Normal, Plantar : B/L flexor . Sensory system –WNL. Gait normal. All other systems (CVS, GIT, Respi.) : WNL. Investigations – Hemogram, Renal functions, Liver functions and metabolic parameters were normal. ECG : WNL. Chest X ray was normal. EEG : January 2011 WNL CT Brain elsewhere reported - Lateral ventricles appear widely spread with dilated occipital horns (colpocephaly) Third ventricle is high binding & opening superiorly into the interhemispheric fissure. Corpus Callosal aggenesis present. MRI Brain revealed B/L colpocephaly widely serrated parallel lateral ventricles, non- crossing commissural fibres are seen indenting medial wall of lateral ventricles. Coronal section shows

Corresponding Author:

Dr. Jacob K Jacob, Associate Professor, Department of Medicine, Cooperative Medical College, Kalamassery, Cochin, Kerala.
E-mail: jacobkjacob@yahoo.com

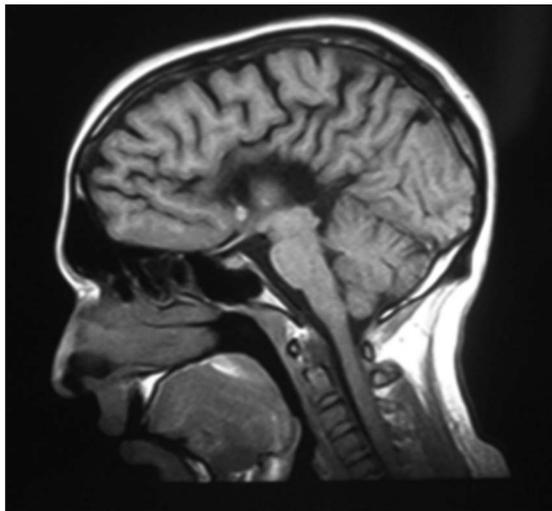


Figure 1. Corpus callosum, agenesis. Sagittal T1-weighted MRI of the brain shows complete absence of the corpus callosum. The cingulate sulcus is absent, and the medial hemispheric sulci reach the third ventricle in a radial fashion

trident shaped frontal horns with elongated foramen of monroe, keyhole temporal horns & vertically oriented hippocampi. Sagittal section shows non-visualisation of Corpus Callosum with absent cingulated gyrus, radially arranged gyri are seen pointed to 3rd ventricle which is high riding & opening in to interhemisphere cyst. The findings are consistent with agencies of Corpus Callosum.

Spectrum of Corpus Callosal abnormalities

ACC may be complete, partial, or atypical. With complete agenesis, the corpus callosum is totally absent.

With partial agenesis (hypoplasia), the anterior portion (posterior genu and anterior body) is formed, but the posterior portion (posterior body and splenium) is not. The rostrum and the anterior/inferior genu are also not formed.

An atypical appearance occurs when the anterior to posterior formation is not respected.

In holoprosencephaly, callosal anomalies are atypical; for example, the splenium may be present without a genu or body. In middle interhemispheric fusion, which is a variety of holoprosencephaly, the genu and splenium may be present without the callosal body.⁷

With pseudo-corpus callosum, which involves conditions of complete or partial agenesis, the hippocampal commissure may become enlarged and appear like the posterior part of the corpus callosum.

Secondary destruction of corpus callosum occurs when the genu and anterior body are destroyed, leaving the posterior portion of the corpus callosum intact. This

may occur secondary to porencephaly or schizencephaly, as a surgical complication in cases involving the transcalsal approach to the lateral and third ventricle, or with hemisection of the callosum for the treatment of seizures.

Other cerebral malformations may coexist with callosal dysgenesis. Examples of these include interhemispheric cysts; intracranial lipomas; and disorders of neuronal migration, such as neuronal heterotopias, lissencephaly, pachygyria, and, as mentioned, schizencephaly.

DIAGNOSIS

The diagnosis of callosal agenesis is by neuroimaging. In the newborn, before closure of the anterior fontanelle occurs, screening Ultrasonography may clearly show the absence of the corpus callosum; it may also show parallel lateral ventricles, interhemispheric cysts, hydrocephalus, and other related anomalies.

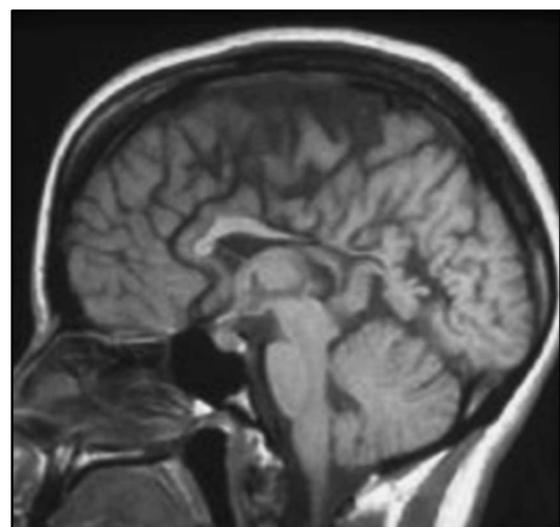


Figure 2. Corpus callosum, agenesis. Sagittal T1-weighted MRI of the brain shows partial agenesis of the corpus callosum. The genu and anterior body of the corpus callosum are visualized, whereas the posterior body, splenium, and rostrum are absent

Antenatal diagnosis of agenesis of corpus callosum (ACC) is possible from about 20 weeks' gestation. Characteristic intrauterine sonologic findings include colpocephaly and parallel ventricular walls. Computed tomography (CT) scan shows Parallel lateral ventricles, colpocephaly, and extension of the third ventricle into the interhemispheric fissure.

Magnetic resonance imaging (MRI) is currently the imaging procedure of choice in infants and children with ACC. Neuronal migration anomalies or atypical forms of holoprosencephaly may be extremely subtle or indiscernible on CT or sonologic images.

CAUSE

Agenesis of the corpus callosum is caused by disruption to development of the fetal brain between the 3rd and 12th week of pregnancy. Other possible causes may include chromosome errors, inherited genetic factors, prenatal infections or injuries, prenatal toxic exposures, structural blockage by cysts or other brain abnormalities, and metabolic disorders.

It is part of ciliopathies. The underlying cause may be a dysfunctional molecular mechanism in the primary cilia structures of the cell organelles which are present in many cellular types throughout the human body. The cilia defects adversely affect "numerous critical developmental signaling pathways" essential to cellular development and thus offer a plausible hypothesis for the often multi-symptom nature of a large set of syndromes and diseases. Known ciliopathies include primary ciliary dyskinesia, Bardet-Biedl syndrome, polycystic kidney and liver disease, nephronophthisis, Alstrom syndrome, Meckel-Gruber syndrome and some forms of retinal degeneration.³

SIGNS AND SYMPTOMS

Signs and symptoms of ACC and other callosal disorders vary greatly among individuals and include vision impairments, low muscle tone (hypotonia), poor motor coordination, delays in motor milestones such as sitting and walking, low perception of pain, delayed toilet training, and chewing and swallowing difficulties. They difficulty transferring more complex information from one hemisphere to the other.⁴ They also have some cognitive disabilities (difficulty in complex problem solving) and social difficulties (missing subtle social cues), even when their Intelligence Quotient is normal.⁴ Specific social difficulties may be a result of

impaired face processing.⁵ The unusual social behavior in childhood is often mistaken for or misdiagnosed as Asperger syndrome or other autism spectrum disorders. Other characteristics sometimes associated with callosal disorders include seizures, spasticity, early feeding difficulties and/or gastric reflux, hearing impairments, abnormal head and facial features, and mental retardation.¹

Associated syndromes and conditions

Aicardi syndrome is a rare genetic malformation syndrome characterized by the partial or complete absence of the corpus callosum, the presence of retinal abnormalities, and seizures in the form of infantile spasms

Menkes disease (MNK), also called Menkes syndrome, copper transport disease, steely hair disease, kinky hair disease, or Menkes kinky hair syndrome, is a disorder that affects copper levels in the body, leading to copper deficiency. It is an x-linked recessive disorder.

Foetal alcohol syndrome (FAS) is a pattern of mental and physical defects that can develop in a fetus when a woman drinks alcohol during pregnancy. The timing and frequency of alcohol consumption during pregnancy are major factors in the risk of a child developing fetal alcohol syndrome.

Acrocallosal syndrome (also known as ACLS) is a rare autosomal recessive syndrome characterized by corpus callosum agenesis, polydactyly, multiple dysmorphic features, motor and mental retardation, and other symptoms.

Septo-optic dysplasia (SOD), also known as de Morsier syndrome is a congenital malformation syndrome made manifest by hypoplasia (underdevelopment) of the optic nerve and absence of the septum pellucidum (a midline part of the brain Mowat-Wilson syndrome This autosomal dominant disorder is characterized by a number of health defects including Hirschsprung's disease, mental retardation, seizure disorder, delayed growth and motor development, congenital heart disease, genitourinary anomalies and absence of the corpus callosum.

The Shapiro syndrome is a rare disorder consisting of paroxysmal hypothermia (due to hypothalamic dysfunction of thermoregulation), epilepsy, and agenesis of the corpus callosum with onset typically on adulthood.

TREATMENT

There are currently no specific medical treatments for callosal disorders, but individuals with ACC and other callosal disorders may benefit from a range of developmental therapies, educational support, and services. It is important to consult with a variety of medical, health, educational and social work professionals including neurologists, neuropsychologists, occupational therapists, physical therapists, speech-language pathologists, pediatricians, geneticists, special educators, early intervention specialists, and adult service providers.

Prognosis

Prognosis varies depending on the type of callosal abnormality and associated conditions or syndromes. It is not possible for the corpus callosum to regenerate or degenerate (i.e., the corpus callosum will not regrow or diminish). Although some individuals with callosal disorders 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 ACC.

END NOTE

Author Information

Dr. Jacob K Jacob,
Associate Professor, Department of Medicine,
Cooperative Medical College, Kalamassery,
Cochin, Kerala.
E-mail : jacobkjacob@yahoo.com

Conflict of Interest: None declared

Cite this article as: Jacob K Jacob. Agenesis of the Corpus Callosum. Kerala Medical Journal. 2012 Mar 30;5(1):30-33

REFERENCES

1. University of Maine.
2. National Organization for Disorders of the Corpus Callosum.
3. Badano JL, Mitsuma N, Beales PL, Katsanis N. The Ciliopathies: An Emerging Class of Human Genetic Disorders. *Annual Review of Genomics and Human Genetics*. 2006;7(1):125–48.
4. Agenesis of the Corpus Callosum”. Travis Research Institute.
5. “Corpus Callosum Research Program”. California Institute of Technology.