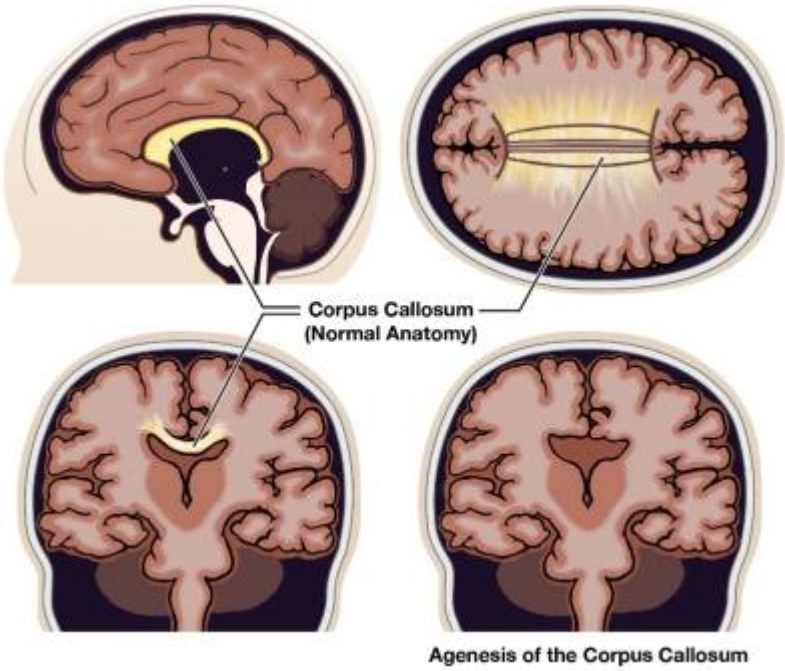
Agenesis of the corpus callosum



Agenesis of the corpus callosum (ACC) is a rare congenital disorder in which there is a complete or partial absence of the corpus callosum. It occurs when the corpus callosum, fails to develop normally, typically during pregnancy. The development of the fibers that would otherwise form the corpus callosum become longitudinally oriented within each hemisphere and form structures called Probst bundles.

Agenesis of the corpus callosum was first recognized and documented in 1887 by John Langdon Down, a British physician best known for his description of the common genetic disorder that is now called Down syndrome

General information

A failure of commissuration occurring ≈ 2 weeks after conception. Results in an expansion of the third ventricle and separation of the lateral ventricles (which develop dilated occipital horns and atria, and concave medial borders).

The corpus callosum (CC) forms from rostrum (genu) to splenium, in agenesis there may be an anterior portion with absence of the posterior segment (the converse occurs infrequently). The absence of the anterior CC with the presence of some posterior CC is indicative of some form of holoprosencephaly.

In addition to agenesis of the corpus callosum, other callosal disorders include hypogenesis (partial formation), dysgenesis (malformation) of the corpus callosum, and hypoplasia (underdevelopment) of

the corpus callosum.

Differential diagnosis

May occasionally be associated with hydrocephalus, but more often merely represents an expansion of the third ventricle and separation of the lateral ventricles.

Case reports

Kho et al., report a patient with Parkinson's disease in whom imaging revealed a complete agenesis of the corpus callosum. Although this co-occurrence is probably coincidental, this finding suggests that the bilateral degenerative changes in Parkinson's disease may occur independent of the interhemispheric connections [1)].

Agenesis of the corpus callosum and Chiari Malformation

- Grey matter hypertropia in a child with recurrent seizure: A case report
- Supratentorial Intracranial Anomalies in Myelomeningocele Patients
- Segmental Agenesis of the Corpus Callosum With Pituitary Hypoplasia
- Atretic Parietal Cephalocele With First Trimester Chiari Malformation and Sinus Pericranii Companion Case
- Corpus callosum disorders and associated malformations in paediatric epilepsy: MRI analytic study
- An Adolescent with a Rare De Novo Distal Trisomy 6p and Distal Monosomy 6q Chromosomal Combination
- Abnormalities of the Fetal Central Nervous System: Prenatal US Diagnosis with Postnatal Correlation
- Myelomeningocele with Associated Anomalies – Case Report and Literature Review

Tijssen et al. describe the neuroimaging findings of an 11-year-old boy who presented with mild occipital headache and precocious puberty. This child was found to have a combination of various midline anomalies including a Chiari type 1 malformation, corpus callosum agenesis and patent craniopharyngeal canal with adjacent intracranial dermoid cyst [2)].

Agenesis of the corpus callosum Chiari Malformation and Hydrocephalus

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- Cervicothoracic Spinal Dysraphism: Unravelling the Pandora's Box
- Loss-of-function variants in NFIA provide further support that NFIA is a critical gene in 1p32-p31 deletion syndrome: A four patient series
- Fetal Cerebral Ventricular Dilatation: Etiopathogenic Study of 130 Observations
- Interfrontal encephalocele: a rare feature of forehead in hydrocephalic myelomeningocele patients. Clinical feature, probable mechanisms, and management

1)
Kho KH, Leijten QH, Dorresteijn LD. A Parkinson's Disease Patient without Corpus Callosum. J Parkinsons Dis. 2019 Mar 21. doi: 10.3233/JPD-191599. [Epub ahead of print] PubMed PMID: 30909250.
2)
Tijssen MP, Poretti A, Huisman TA. Chiari type 1 malformation, corpus callosum agenesis and patent craniopharyngeal canal in an 11-year-old boy. Neuroradiol J. 2016 Oct;29(5):307-9. doi: 10.1177/1971400916656487. PubMed PMID: 27329972; PubMed Central PMCID: PMC5033089.