ABSTRACT: Agenesis of corpus callosum can have various neuropsychiatric and developmental manifestations. Following case report highlights the case of a young female child presenting with features of not ability to hold head and delayed speech. The child showed no other positive findings and is conscious and oriented at the time of examination. Following this complains the patient was subjected for a non-contrast MRI. Neuroimaging revealed agenesis of corpus callosum. There were no other findings which could explain the current situation experienced by the patient.

INTRODUCTION: Corpus callosum dysgenesis is the most common CNS malformation and is found in 3-5% of individuals with neurodevelopmental disorders. It has a prevalence of at least 1:4000 live births. Non-syndromic corpus callosum dysgenesis is found in patients of all ages.[1] Agenesis of corpus callosum (ACC) is mostly a congenital anomaly that occurs predominantly in males, either isolated or in combination with other CNS or systemic malformations.[2] The occurrence of ACC is usually sporadic. Disturbance of embryogenesis in the first trimester of pregnancy by an unknown insult is sometimes put forward as a probable antenatal cause.[1] If the anterior portion of the corpus callosum (Genu and Splenium) is small or absent, then one may conclude that there is some disturbance in normal unfolding of events along the axis of passage of the callosal fibers. This state represents true callosal hypogenesis. If the anterior portion of corpus callosum is small or absent and posterior portion is present, then, with certain exceptions, there has most likely been secondary destructive injury to corpus callosum rather than defect in development.[3]

CASE REPORT: A 2 yrs old girl resident of Mumbra of non-consanguineous marriage presented with complains of inability of head holding and delayed speech (Inability to utter any monosyllable) and no other positive complains. The patient had an uneventful full term normal delivery. Immunization history was uneventful. Her other motor and sensory milestones were unremarkable. The girls general and systemic examination is unremarkable.

Following this she was subjected for a routine non contrast MRI to rule out any findings of periventricular leukomalacia or any foci of chronic ischemic insult.

Axial T2 and FLAIR images showed dilated occipital horns of both lateral ventricles. [asteriks] Anterior horns of both lateral ventricle could not be visualized. Giant cisterna magna is seen.

Fig. 1
Sagittal T2 and T1 images showed complete absence of genu and body of corpus callosum with preservation of splenium. Typical 'spoke-wheel' appearance of sulci and gyri was also well appreciated. Another variant of anterior cerebral artery in the form of 'Azygous anterior cerebral artery' was also well appreciated on T2 images.

Coronal T2 and Sagittal T1 images showed lateral displacement of anterior horns of both lateral ventricles with 'Viking head' appearance with hypo intense 'Probst bundles' which are the remnants of corpus callosum noted medially to anterior horns of lateral ventricles.
DISCUSSION: Agenesis of corpus callosum is a rare condition (With US frequency rates reported to be 0.07% to 5.3%) which usually presents with clinical features such as seizures, feeding problems, developmental delay, impaired hand-eye coordination, impaired visual and auditory memory and hydrocephalus. In agenesis of corpus callosum, commissural fibers do not cross the midline instead thick bundles of intersecting fibers called Probst bundles which lie along the super medial aspect of the lateral ventricles and the third ventricle may sometimes be displaced upward. Complete and partial agenesis of corpus callosum can result from genetic, infectious, vascular, or toxic causes. Current evidence suggests that a combination of genetic mechanisms, including single-gene Mendelian, single-gene sporadic mutations and complex genetics (Which may have a mixture of inherited and sporadic mutations) may be involved in the etiology of agenesis of corpus callosum. For approximately 30–45% of individuals with agenesis of corpus callosum, the cause is identifiable (~10% have chromosomal anomalies) and the remaining 20–35% have recognizable genetic syndromes.

Children with callosal conditions experience motor impairments such as hypotonia, spasticity, poor motor coordination and cerebral palsy. Epilepsy and seizures are more common in these children and adolescents, with the reported prevalence varying from 27 to 86% depending on the population studied. Researchers have also reported early sucking, chewing, and swallowing difficulties and esophageal reflux. Besides, developmental delays are quite common among children with callosal conditions, with the reported prevalence ranging from 60 to 80% of those studied. Some children may exhibit delays in achieving motor, language and cognitive milestones and often accomplish toilet training at a much later age than their typically developing siblings. In our case the patient complains were only delay in head holding, further developmental delays may be recognized later as time proceeds.

Another noticeable characteristic of most individuals with callosal problems is the diminished comprehension of humor (Especially certain types of more abstract humor, such as irony or word play) and also the impairment in the verbal expression of their emotional experience (Alexithymia). According to these findings, in the absence of corpus callosum, neither hemisphere alone possesses an adequate comprehension of humor. It is also worth mentioning that some of the deficits seen in individuals with callosal disorders (Diminished comprehension of humor, proverbs, prosody) are similar to deficits seen in right hemisphere damaged patients.

Corpus callosum is the main path for coordinating syntactic and prosodic information and information about negative emotions must be transferred from the right hemisphere to the left in order to produce accurate verbal descriptions about stimuli that involve negative emotions. It is clear that callosal disorders greatly reduce capacity for transferring this complex information between the hemispheres. In general, clinical signs and symptoms in corpus callosum structure disorders vary widely and their severity depends strongly on the presence of other malformations of CNS. Children and adolescents with isolated corpus callosum disorders (Without other associated brain anomalies) can exhibit any of the clinical features described below, but their global functioning and future prognosis are much better.
CASE REPORT

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# CASE REPORT

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