



## An Infant with Complete Corpus Callosum Agenesis: Case Report

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### General Note

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## ABSTRACT

Corpus callosum is an important commissure, which connects the right and left hemispheres. Corpus callosum agenesis is a relatively uncommon malformation of the brain in our society of KSA. Its diagnosis is based on brain imaging and sometimes asymptomatic. The Following case report highlights a case of a newborn with ambiguous genitalia, associated with complete agenesis of the corpus callosum discovered at birth, with wide cisterna magna and small cerebellum, multiple congenital anomalies, and dysmorphic features.

### Keywords

Corpus callosum, agenesis, cerebellum, genetic factors, dysmorphic facial features.

## 1. INTRODUCTION

The largest commissure in the brain is called the corpus callosum. This binds the two cerebral hemispheres together. It is divided into the rostrum, the genu, the body, and the splenium (Snell & Richard, 2010). This part of the brain is typically composed of transverse fibres (Aboitiz and Montiel, 2003).

Formation of the corpus callosum takes place usually in the 6th weeks of gestation, and that occurs by destined of axons to cross the midline medially to the other hemisphere (Schell et al., 2008).

Corpus Callosum Agenesis (ACC) is a rare (congenital) condition that occurs at birth. It is distinguished by a partial or complete lack (agenesis) portion of the brain that links the two hemispheres of the brain. The cause of agenesis of the corpus callosum is usually not known, but it can be inherited as either an autosomal recessive trait or an X-linked dominant trait. An infection or injury can also cause it during the twelfth to the twenty-second week of pregnancy (intrauterine), leading to developmental disturbance of the fetal brain (NODCC, 2010).

In some cases of Agenesis of corpus callosum (ACC), it may be associated with other structural defects or anomalies such as Chiari II malformation with characterized by abnormal development of the cerebellum and medulla oblongata, displacement of one or both cerebellar tonsils which tend to descend into the foramen magnum, and usually accompanied by myelomeningocele (Chiappedi et al., 2012).

Its incidence varies based on diagnostic techniques and populations of samples: in the general population, Its prevalence is estimated to be 3–7 per 1,000 births, compared to 2-3 per 100 for children with a developmental disability (Chiappedi et al., 2010). Patients with CCA have a clinical syndrome which had initially been thought to be a consequence of hemispheres disconnection.

Nevertheless, recent studies have shown that in addition to the sulcation abnormalities already described, CCA patients have an abnormal microstructure and a reduced volume of the ventral cingulum bundle, suggesting that defects in intrahemispheric white matter tracts may be an important factor (Nakata et al., 2009).

Corpus callosum disorders may also be observed in conjunction with major embryonic forebrain malformations before the corpus callosum anlage (e.g., holoprosencephaly, HPE) is formed (Schell et al., 2008).

Another interesting finding recently is a reduction in the number of von Economo neurons, large spindle-shaped neurons localized to the anterior cingulate cortex, and frontoinsular cortex in patients with CCA; this is considered another consequence of the genetic disruption that caused agenesis (Kaufman et al., 2008).

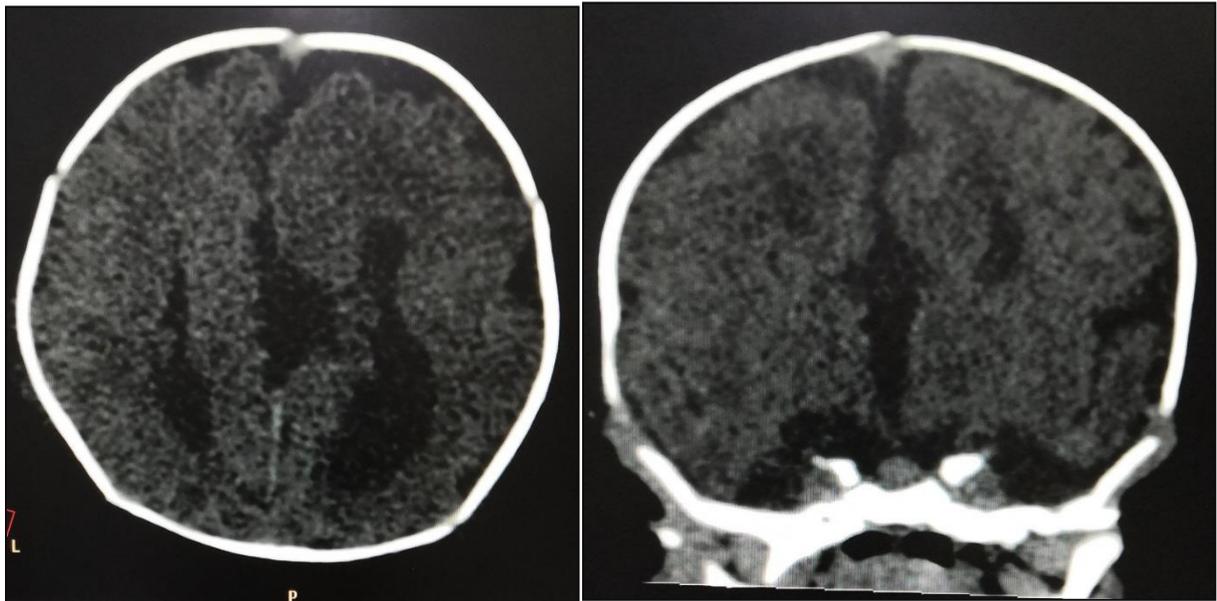
## 2. CASE PRESENTATION

This study involved a full-term neonate baby with ambiguous gender who was born by Normal Spontaneous Vaginal Delivery (NSVD), after a gestational period of 41 weeks and who was the couple's first child. The baby was found to have dysmorphic facial features and ambiguous genitalia. Head Computed tomography scan (CT) performed in King Khaled Hospital in Kharj revealed the indication of Agenesis of the Corpus Callosum ACC, with wide cisterna magna and small cerebellum, regarding the ventricular system, revealed enlarged 3rd and left lateral ventricles and abnormalities in the development of the cerebral gyrus and brain sulcus (fig-1). There is no choanal atresia. The baby was admitted to the neonatal intensive care unit for the ACC examination and then referred for consultation. No past history has been reported of consanguineous marriage, albinism, or any other systemic disease.

### Findings at Birth

The weight of the baby at birth was 3 Kg. HC - 33 cm and an Apgar score of 7/9. The baby was found to have dysmorphic facial features and syndactyly all extremities. Regarding feeding the baby starts with OGT feeding for four days after that sucking became

stable. There was no amniotic fluid turbidity or coiling of the umbilical cord at the time of birth. Echocardiography demonstrated good heart contraction. No significant anomalies of the heart, liver, or immune system were observed among other systemic findings.



**Figure 1-** CT Brain (Cronel and Axial planes), manifest agenesis of the corpus callosum

### 3. DISCUSSION

Agenesis of the corpus callosum considered as rare or uncommon cerebral malformation with unknown clear aetiology (Zamurović & Andjelic. 2014). It may be associated with other nervous system defects such as ventriculomegaly, abnormal development of cerebellum and medulla oblongata, displacement of one or both cerebellar tonsils as in the case of Chiari II malformation. Also, it could be combined with other organ problems like congenital heart defects.

The genetic factors also play an essential role in some cases of agenesis of the corpus callosum, due to chromosome anomalies as occur in a patient with apparently balanced reciprocal translocation which causes disturbance and an anticipated loss in the FOXP1B gene quality (Schell-Apacik et al., 2008) also in case of oro-facial-digital syndrome type 1 (Thauvin-Robinet et al., 2006), and Mowat–Wilson syndrome affect ZFX1B gene (Zweier et al., 2002).

Our recent case describes a case of agenesis of the corpus callosum as appear in head Computed tomography scan with ventriculomegaly due to enlarged 3rd and left lateral ventricles, associated with wide cisterna magna and small cerebellum, atrophic changes in the left hemisphere.

The causes of this case are unclear, but we can suggest the genetic factors which affected in the development of the corpus callosum, embryologically the corpus callosum starts to develop on the seventh week of gestation and is achieved by 18 to 20 weeks (Paul et al., 2011), in case of the presence of an autosomal recessive trait or an X-linked dominant trait the agenesis can occur by causing the absence of callosal fibres.

### 4. CONCLUSION

The importance of this rare case report is to highlight the rarity disorder of corpus callosum agenesis with ambiguous genitalia and multiple congenital anomalies and dysmorphic features. Diagnosis of congenital brain malformation considered to be a challenging issue, further studies on the antenatal sonographic diagnosis of callosal underdevelopment also the precise genetic counselling should be performed.

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**Authors' contributions**

All authors contributed equally to this work and they have read and agreed to the final manuscript.

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**Conflict of interest**

The authors declare that they have no conflict of interest.

**Informed consent**

Written & Oral informed consent was obtained from all individual participants included in the study.

**Data and materials availability**

All data associated with this study are present in the paper.

**Peer-review**

External peer-review was done through double-blind method.

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