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Case Report

Agenesis of corpus callosum in a preterm neonate

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Abstract

A preterm female neonate was born to a 20-year-old primiparous woman with a history of second degree consanguineous marriage, septic abortion, PCOS and hypothyroidism at 34 weeks of gestation. Prenatal screenings were negative for trisomies and major genetic abnormalities. Postnatal neuro imaging revealed agenesis of the corpus callosum (ACC), a rare congenital malformation. Despite the absence of advanced maternal age. The neonate required neonatal intensive care unit (NICU) support and was placed on continuous positive airway pressure (CPAP) for respiratory assistance. During NICU management, the neonate displayed poor heart and respiratory function and a multidisciplinary care approach was implemented. This case highlights the significance of thorough prenatal and postnatal monitoring in consanguineous pregnancies, even in young mothers with atypical maternal histories, and emphasises the need for specialised care in such cases.

Keywords: Preterm Infant, Agenesis of corpus callosum, Consanguinity, Congenital malformations, Prenatal screening, Postnatal neuroimaging, Maternal hypothyroidism

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1. Introduction

Agenesis of corpus callosum is a congenital brain deformity which may present as an independent condition or with associations to congenital syndromes. The incidence of ACC is 0.5-70 per 10,000 births. It is caused due to improper migration of glial cell populations and migratory neurons during developmental period. Usually, it is diagnosed prenatally via ultrasonography at 18-20 weeks of gestation and the diagnosis is confirmed by MRI brain. In isolated ACC, 65% cases show normal neurological development while 35% show impairment. Hence, it must be noted that ACC is not an absolute indication for MTP.

Table 1: Timeline

2024-01-03	LMP
2024-01-19	USG scan done
2024-03-17	USG: B/L PCOS
2024-05-21	Anomaly Scan
2024-05-21	USG (growth scan) frontal bossing with hypotelorism and hypognathism, small posterior fossa and b/l
Dilated lateral ventricles	
2024-06-19	USG
2024-07-24	USG: Mild Oligohydromnios
2024-07-25	CBC
2024-08-03	District Hospital visit
2024-08-08	Fetal growth scan
2024-08-28	Date of Admission
2024-08-29	USG before delivery
2024-08-31	Baby Admitted in NICU on cpap
2024-08-31	Delivery by LSCS

2. Case Report

A 20-year-old G2A1 mother delivered a female baby at 34 weeks of gestation secondary to threatened preterm by LSCS secondary to threatened.

Preterm at our institute. The neonate was born to a secondary consanguineous marriage with a history of septic abortion, PCOS and hypothyroidism. The mother's first pregnancy was terminated by dilation and curettage due to septic abortion at 13 weeks of gestation.

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The current pregnancy was booked, and, on an anomaly, scan done at 19 weeks of gestation the fetus was found to have gross ventriculomegaly. A second Sonography performed at 31 weeks.

1. Absence of Cavum septum pellucidum.
2. Parallel oriented lateral ventricles showing dilatation of occipital horns (colpocephaly) (**Figure 5**).
3. upward cystic dilatation of roof of third ventricle is noted, forming a tubular interhemispheric cyst just caudal to inferior edge of falx cerebri, measuring 28 x 11 (**Figure 1**, **Figure 2**).
4. Deviation of pericallosal artery.
5. Radially oriented parasagittal sulci (sunburst appearance),
6. Posterior most segment of body and splenium of corpus callosum however are visualized, suggesting it be a partial agenesis of corpus callosum.
7. Dilatation of atria of bilateral lateral ventricles noted, measuring 12.6 mm and 13.6 mm- ventriculomegaly. (**Figure 4**).
8. Third ventricle is also dilated (**Figure 2**).
9. The above-mentioned interhemispheric cyst representing upward cystic dilatation of roof the third ventricle in the region of corpus callosal agenesis. (**Figure 3**)
10. Aqueduct of sylvius and fourth ventricles appeared normal to the visualized extent.
11. Posterior fossa appeared normal with normal Cisterna magna.

A provisional diagnosis of agenesis of corpus callosum was made. She did not have any history of inter uterine infection during her current pregnancy. She had no history of exposure to teratogenic drugs or radiation. She was diagnosed with PCOS in the 1st trimester and hypothyroidism during 2nd trimester. The rest of the antenatal period was uneventful.

The female neonate cried immediately after birth and had an APGAR score 8 and 9 at 1 min and 5 min respectively. The birthweight was 1.73kg which was appropriate for gestational age due to preterm delivery 17 inches length and head circumference were 33cm and 37cm at birth. (Within normal range).

The neonate received routine care at birth, general physical examination and neurological examination was normal. The neonate was admitted in NICU and was placed on continuous positive airway pressure which followed with eventual weaning to room air.

Lab tests conducted soon after birth consisted of CBC, CRP levels and CSF culture were done which showed no significant findings. (**Table 2**)

2D Echo revealed the following:

1. Small PDA-20.2mm left to right
2. Patent foramen ovale
3. Trivial TR-27mmhg
4. Mild Pulmonary artery hypertension.

MRI was done on 9th day which revealed the following impressions:

1. Near total agenesis of corpus callosum (thin strip is seen in the anterior body) (**Figure 6**, **Figure 7**) with dilated and cranial migrated 3rd ventricle. associated with parallel running lateral ventricles giving "Race- car apperance"
2. Abnormal contour of right hippocampi with rounded morphology and vertical orientation on the right side is noted.

Post the period of 10-day observation Retinopathy of prematurity (ROP) test was carried out and the neonate was discharged.

Table 2: Diagnostics

Type	Value	Unit
2024-08-31		
Calcium	8.1	mg/dL
CRP (C-reactive protein)	6.8	mg/L
CSF culture	Negative	
Hb (hemoglobin)	21.4	g/dL
Platelet count	57	cells/µL
Potassium	5	mEq/L
RBC (total red blood cells)	5.26	cells/mm^3
Sodium	139	mEq/L
WBC (total white blood cells)	7890	cells/mm^3

Figure 1: Interhemispheric cyst IHC communicating with 3rd ventricle

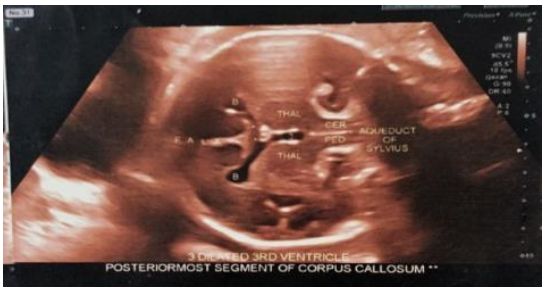

Figure 2: Interhemispheric cyst contiguous with 3rd ventricular cavity

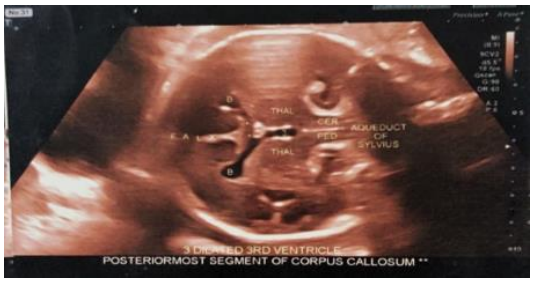

Figure 3: Posterior most segment of Corpus Callosum

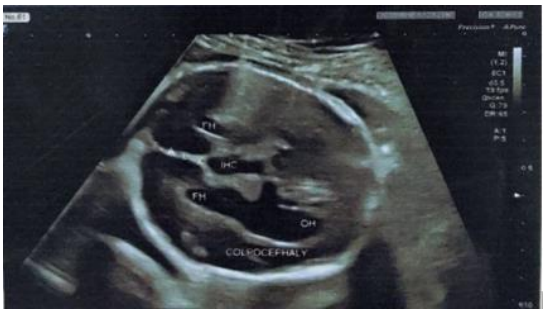

Figure 4: Dilatation of atria of bilateral lateral ventricles noted, measuring 12.6 mm and 13.6 mm- ventriculomegaly. Absence of Cavum septum pellucidum along with wide intrahemispheric fissure is noted.

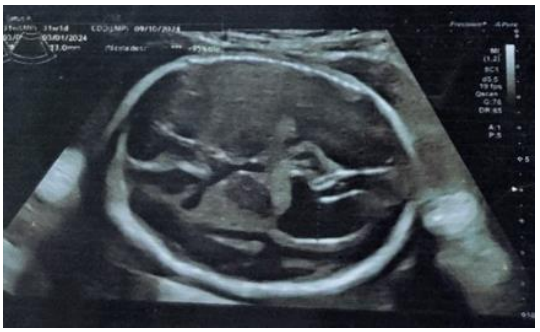

Figure 5: Dilatation of occipital horns (colpocephaly) along with intrahemispheric cyst

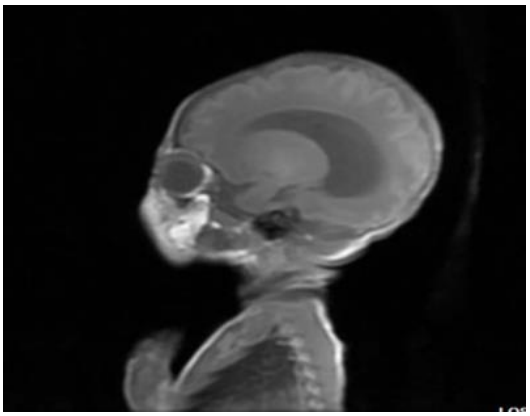

Figure 6: T1WI sagittal image shows near total agenesis of corpus callosum (thin strip is seen in the anterior body)

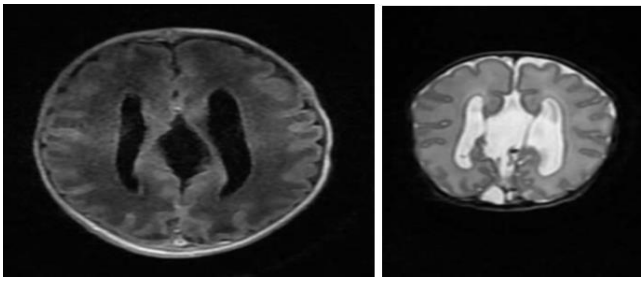

Figure 7: T1WI sagittal image shows near total agenesis of corpus callosum (thin strip is seen in the anterior body)

3. Discussion

Corpus callosum is the largest white matter structure of the brain which helps in communication between the two hemispheres. Its development starts by migration of multiple glial cell populations along with migratory neurons in the brain at 6 weeks of gestation and it is completely formed between 18 to 29 weeks.

Disruption of this process during developmental period can result in dysgenesis or hypogenesis of Corpus Callosum.

Agenesis of Corpus Callosum is a congenital cerebral malformation which may be classified as Complete (Total) or Partial based on the morphology, it depends on the timing of insult during the antenatal period

ACC can present as an isolated condition or in association with chromosomal abnormalities such as Trisomy18 (Edwards's syndrome), Trisomy 13(Patau syndrome), Trisomy 21 (Down syndrome), Dandy- Walker malformation, Aicardi syndrome, Andermann syndrome, X-linked hydrocephalus etc.

Risk factors include exposure to teratogenic drugs, maternal alcoholism, metabolic disorders and CMV infection. While a history of consanguinity may be considered significant risk factors there is no documented evidence about the same.

Fetal Ultrasonography can usually detect ACC prenatally around 18-20 weeks of gestation which can be confirmed on MRI which is the investigation of choice in such cases. MRI findings like colpocephaly, High riding third ventricle and dilated occipital horns are usually characteristic and are consistent with our case.

In most cases, a diagnosis of ACC is made after a few months of birth as they show delayed milestones or development. Although in our case a diagnosis suggestive of ACC was made well in advance which gave the parents a choice of MTP. However, it must be noted that ACC is not an absolute indication for MTP as in isolated ACC, 65% cases show normal neurological development while 35% show impairment. Patients are usually asymptomatic, although if symptomatic they usual present with mental retardation, vision problems, speech delay, seizures and feeding problems. Hence regular follow ups are indicated to

detect potential developmental issues Due to gross ventriculomegaly there is increase in head circumference.

Patients are usually asymptomatic although if symptomatic they usually present with mental retardation, vision problems, speech delay, seizures and feeding problems in the first two years of life. Hence the approach of management is usually symptomatic and supportive.

This case presents us with the classical signs of ACC on the MRI such as race car sign as seen on **Figure 6**, **Figure 7**. There are also a few foci of blooming noted in the bilateral basal ganglia and right cerebellar hemisphere which could indicate micro-bleeds or calcifications. This could indicate further neuro cognitive defects. The case is unique because ACC is usually seen in advancing maternal age and our patient was only 20Y old without any genetic abnormalities. She was also born pre-term to a mother with a complicated maternal history of secondary consanguineous marriage, hypothyroidism, PCOS and septic abortion.

4. Source of Funding

None.

5. Conflict of Interest

None.

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