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Case Report

Paediatrics Section

Unique Presentation of Colpocephaly in a Preterm Triplet: A Case Report

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ABSTRACT

Colpocephaly is a rare congenital cerebral malformation characterised by disproportionate dilatation of the occipital horns of the lateral ventricles, most commonly associated with partial or complete agenesis of the corpus callosum. It results from defective neuronal migration and abnormal white-matter development during embryogenesis. The condition may manifest during infancy with developmental delay, seizures, and varying degrees of intellectual or visual impairment. Present case report a 10-month-old male infant, the first-born of triplets conceived through in-vitro fertilisation, who presented with global developmental delay and recurrent seizures. The perinatal period was complicated by respiratory distress and neonatal sepsis, necessitating ventilation and intensive care. MRI brain revealed thinning of the corpus callosum, bilateral cerebral volume loss, and disproportionate enlargement of the occipital horns-findings consistent with colpocephaly. The infant was managed with physiotherapy, occupational therapy, and speech therapy, leading to gradual improvement in tone, posture, and social interaction on follow-up. Antiepileptic therapy was continued with good seizure control. This case is unique due to the coexistence of colpocephaly, prematurity, perinatal depression, and neonatal sepsis in a triplet born via assisted reproduction- an extremely uncommon combination. The report underscores the multifactorial aetiopathogenesis of neurodevelopmental impairment in such cases. Early recognition, timely neuroimaging, and multidisciplinary intervention play a vital role in optimising outcomes and providing appropriate parental counselling.

Keywords: Corpus callosum dysgenesis, Developmental delay, Neonatal sepsis, Neurodevelopmental disorder, Perinatal depression, Ventriculomegaly

CASE REPORT

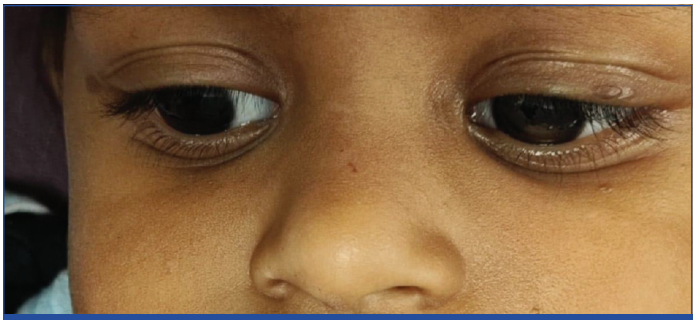
A 10-month-old male infant was brought to the Department of Paediatrics with complaints of delayed attainment of developmental milestones and recurrent seizures. The mother reported poor head control, inability to roll over, and lack of visual tracking since seven months of age, noted especially compared to his two triplet sisters developing normally. The infant produced only monosyllabic sounds and developmentally corresponded to a four to five-month-old child. Seizures began at seven months of age, described as generalised tonic spasms lasting 30-60 seconds, occurring two to three times per week without postictal drowsiness. The child was started on levetiracetam (10 mg/kg /dose twice daily) at a local hospital with partial control.

The infant was the first-born male among triplets conceived via in-vitro fertilisation to a 40-year-old primigravida in a non-consanguineous marriage. The mother had gestational hypertension managed with labetalol (100 mg twice daily) and no diabetes, hypothyroidism, or infections. Antenatal scans were normal. The infant was delivered in an outside hospital, preterm at 30 weeks of gestation via cesarean section due to maternal hypertension, with a birth weight of 978 grams. He required resuscitation for poor cry and respiratory distress at birth and was admitted to the Neonatal Intensive Care Unit (NICU). He was ventilated for eight days for respiratory distress syndrome, received intratracheal surfactant (100 mg/kg), and was managed on Continuous Positive Airway Pressure (CPAP) support. Sepsis screening was positive, and the neonate was treated with intravenous ampicillin (100 mg/kg/day) and gentamycin (5 mg/kg/day) for 14 days. He was discharged on day 57 in stable condition after attaining a weight of two kilograms. There was no family history of neurological disorders or congenital anomalies.

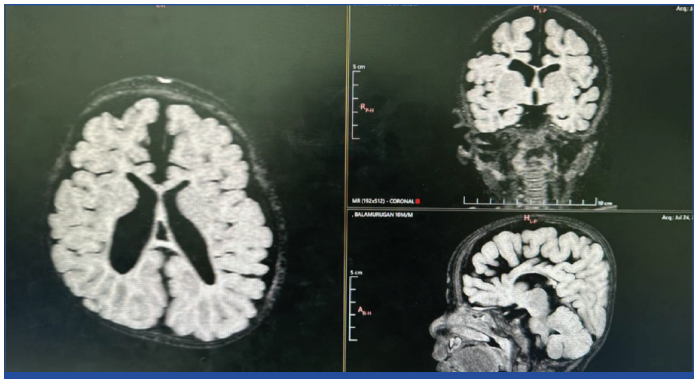
At 10 months, the infant's vital signs were stable (heart rate 120/min, respiratory rate 30/min, temperature 98.4°F, SpO_2 99%). He exhibited poor eye contact, strabismus [Table/Fig-1], and absent head control. Neurological examination revealed generalised spasticity, brisk reflexes, and withdrawal-type plantar responses. Developmental quotient was <70%. Hearing assessment was normal. There were no dysmorphic features, neurocutaneous stigmata, or fundus abnormalities. Laboratory evaluations, including thyroid function, metabolic profile, and TORCH screen, were normal. MRI showed thinning of the corpus callosum, bilateral cerebral volume loss, and disproportionate enlargement of the occipital horns- findings consistent with colpocephaly [Table/Fig-2-4]. Neonatal MRI had earlier shown periventricular leukomalacia.

Differential diagnoses included hydrocephalus, schizencephaly, and periventricular leukomalacia. Hydrocephalus was ruled out as there was no uniform ventricular dilatation or raised intracranial pressure. Schizencephaly was excluded due to the absence of gray matter-lined clefts, and periventricular leukomalacia was not the primary finding given the occipital horn predominance. Hence, based on MRI and clinical correlation, a final diagnosis of colpocephaly with corpus callosum thinning was made.

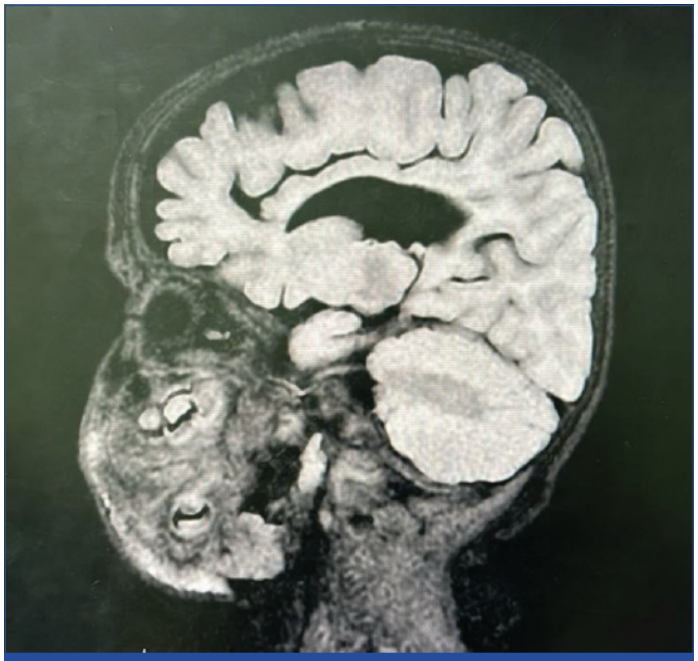
The infant was managed with daily physiotherapy (30 minutes), occupational therapy twice weekly (45 minutes), and speech therapy for stimulation. Levetiracetam (15 mg/ kg/ dose twice daily) was continued for seizure control. Parents received counselling about developmental prognosis, therapy adherence, and follow-up.



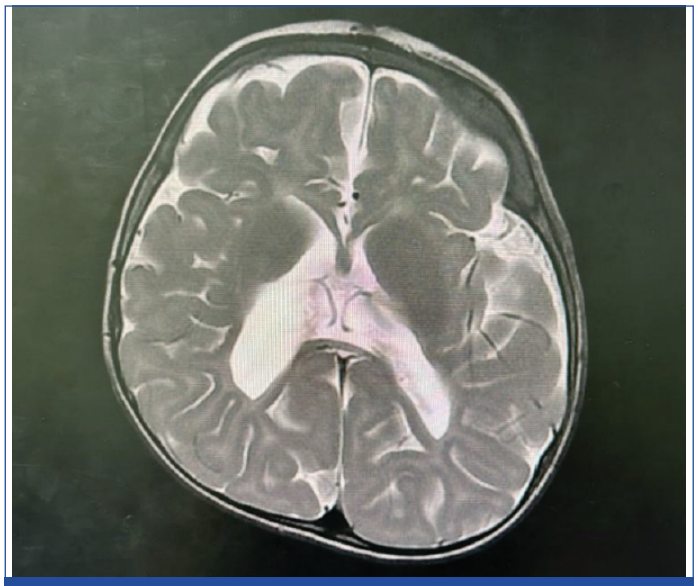
[Table/Fig-1]: Patient with strabismus.



[Table/Fig-2]: a) Axial T1 weighted shows parallel orientation with separation of frontal horns from the enlarged occipital horns known as the " race car sign" due to the partial agenesis of the corpus callosum (colpocephaly). b) Coronal T1 MRI shows the eversion of the cingulate gyri, giving a "Moose head" appearance, which is seen due to lateral loss of corpus callosum. Bilateral reduction of the cerebral white matter volume can also be visualised. c) Sagittal T1 MRI shows thinned out genu and trunk of corpus callosum with a normally sized fourth ventricle.



[Table/Fig-3]: Mid-Sagittal T1-weighted shows distorted genu with prominent anterior commissure and partial agenesis of corpus callosum and the reduced column of cerebral matter.



[Table/Fig-4]: Axial, T2-weighted sequence MRI shows asymmetrical dilatation of the occipital horns in comparison to the frontal horns of the lateral ventricles.

After three months, there was improvement in tone, posture, and initiation of social smile. Continued neurological follow-up and early intervention were advised.

DISCUSSION

In the present case, a 10-month-old male infant presented with global developmental delay, seizures, perinatal depression, neonatal sepsis, and MRI-confirmed colpocephaly.

Colpocephaly is a rare congenital malformation characterised by disproportionate dilatation of the occipital horns of the lateral ventricles. It is commonly associated with dysgenesis or agenesis of the corpus callosum, lissencephaly, and other neuronal migration disorders [1,2]. The pathogenesis involves impaired neuronal migration, persistence of foetal ventricular configuration, or perinatal hypoxic insults [3,4]. Infections such as cytomegalovirus and toxoplasmosis have also been implicated, but our patient tested negative for TORCH infections [5].

Colpocephaly may present with seizures, intellectual disability, visual impairment, or motor deficits. Our patient had developmental delay, strabismus, and spasticity, but no hearing loss. Genetic associations, including syndromes such as Aicardi, trisomy 8 and 9, and familial cases, have been reported [6-9]. Cerullo A et al., described siblings with colpocephaly and different fathers, suggesting maternal transmission [8].

Diagnosis relies on neuroimaging. CT may reveal ventricular dilatation, but MRI is superior for identifying associated anomalies. A posterior-to-anterior horn width ratio ≥3 is considered diagnostic [10]. In our case, MRI at seven months showed thinning of the corpus callosum and occipital horn dilatation consistent with colpocephaly. Differentials include hydrocephalus, schizencephaly, and periventricular leukomalacia. Unlike hydrocephalus, colpocephaly does not show uniform ventricular dilatation or raised intracranial pressure [10].

In this case, colpocephaly coexisted with prematurity, neonatal sepsis, and perinatal depression- factors that may have compounded the neurodevelopmental delay. MRI findings confirmed corpus callosum thinning with occipital horn enlargement. Genetic, metabolic, and infectious causes were excluded. Few paediatric cases similar to this have been described, such as Patnaik A et al., (2012), and Saldanha RP et al., (2017), further illustrating the variability of clinical presentation in colpocephaly [2,4]. Saldanha RP et al., documented neonatal colpocephaly associated with prematurity and early neurological deficits, emphasising the role of perinatal insults and the characteristic MRI pattern of disproportionate occipital horn dilatation- findings similar to our patient [4]. In contrast, Patnaik A et al., reported colpocephaly with macrocephaly and raised intracranial pressure that required ventriculo-peritoneal shunting, demonstrating that some cases may present with obstructive-type symptoms requiring surgical intervention [2]. Together, these studies highlight the broad phenotypic spectrum of colpocephaly, ranging from cases requiring neurosurgical management to those like our patient, who benefit from multidisciplinary developmental rehabilitation. The combination of prematurity, perinatal insult and callosal dysgenesis makes this presentation uniquely illustrative of multifactorial brain injury. Diagnosis relies on MRI with characteristic "race car" and "moose head" signs. Management is supportive and multidisciplinary. Antiepileptics are prescribed for seizures, physiotherapy and occupational therapy help with tone and posture, and speech therapy supports communication [11]. In our patient, physiotherapy and occupational therapy sessions led to improvements in tone and social smile during follow-up. Surgical intervention (ventriculo-peritoneal shunt) may be considered in cases with raised intracranial pressure, though rarely required [12]. Outcomes vary widely. While some cases are asymptomatic and diagnosed incidentally in adults [12,13], children with associated anomalies often have significant neurodevelopmental impairment.

CONCLUSION(S)

Colpocephaly is a rare neuronal migration disorder that should be considered in infants presenting with global developmental

delay, seizures, or atypical neuroimaging findings. In our case, the presence of perinatal depression, neonatal sepsis, and colpocephaly with corpus callosum thinning emphasises the multifactorial etiologies contributing to developmental delay. Early recognition and appropriate supportive interventions-including physiotherapy, occupational therapy, and parental counselling- play a critical role in improving developmental outcomes. A multidisciplinary approach with regular follow-up is essential.

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