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Case Report

Developmental brain anomaly due to fetal hydrocephalus

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Abstract. In this article, the authors intend to illustrate a striking brain malformation due to early onset severe fetal hydrocephalus that superficially suggests the presence of semilobar holoprosencephaly.

Keywords: Developmental brain anomaly, fetal hydrocephalus, semilobar holoprosencephaly

1. Case report

A 22-year-old primigravida with no antenatal care was admitted to the Obstetrics Emergency Department with labour pain where she delivered a male infant at 38 wk gestation by caesarean section. Maternal history was unremarkable for any prenatal infections, trauma, drug abuse or other chronic diseases. Family history was negative for early neonatal death. Apgar scores at 1, 5, 10 and 15 minutes were 6, 6, 7, and 7 respectively. The infant's anthropometric parameters were as follows: weight - 2.4 kg, length - 46 cm, head circumference - 50 cm (> 90th percentile of Lubchenco's intrauterine growth chart). His anterior frontanelle was bulging (6 × 6 cm), and the sunset sign was present. Ill defined soft swellings could be seen over bilateral parietal and occipital regions. Canthal index was 0.34, and facial abnormality was characterized by a depressed nasal bridge (Fig. 1A). A meningomyelocoele of dimensions 8 cm in vertical and 5 cm in the horizontal axes was present in the thoracolumbar region, which had ruptured during delivery (Fig. 1B). He was grossly hypotonic, and primitive reflexes were absent. Doll's eye phenomenon was positive and gag reflex was present. Power in lower limb muscles was grade 2/5. Anal sphincter was hypotonic, and ankle and knee jerks were absent. Heart sounds were normal; there was no hepatosplenomegaly and no abnormalities of the external genitalia. He had tonic seizures from 2 h of age, recurrent apnoeic spells and hypothermia despite using standard servo controlled warmer. Intravenous fluid (10% dextrose) was given as appropriate for his age. For seizure control, phenobarbitone injection and subsequently phenytoin injection was used (20 mg/kg loading dose and 5 mg/kg maintenance dose for both anticonvulsant drugs). The baby received positive pressure ventilation using artificial manual breathing unit bags during episodes of recurrent apnea. The ruptured meningomyelocoele was kept covered with sterile gauze soaked in saline. Ultrasound of brain revealed a large single fluid filled structure (monoventricle) with echogenic material within it that

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Fig. 1. (A) The image shows absence of any significant midline facial defect except depressed nasal bridge. (B) The image shows the ruptured meningomyelocoele in the thoracolumbar region with dimensions 8 cm in vertical and 5 cm in horizontal axes.

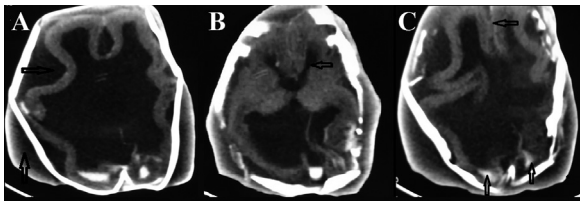

Fig. 2. (A) Axial computed tomography image showing dysmorphic monoventricle, extra-axial and extracranial cerebrospinal fluid collection (marked with arrows) and CSF collection within the frontal interhemispheric fissure. (B) Axial computed tomography image from lateral ventricle level demonstrates well differentiated frontal horns of the lateral ventricle (marked with arrow), dilated occipital horns (especially the right horn) and fragmented skull bones. (C) Axial computed tomography image showing frontal interhemispheric fissure (marked by horizontal arrow) and intraventricular hemorrhage in the posterior aspect of the monoventricle (marked by two vertical arrows).

could represent haemorrhage. The brain mantle appeared thin and without sulcal pattern. A computed tomography showed a dysmorphic monoventricle without septum pellucidum (Fig. 2A). The frontal lobes were divided by an interhemispheric fissure within which there was a collection of cerebrospinal fluid (CSF) (Fig. 2A). There was extra axial CSF with extracranial seepage through fragmented skull bones resulting in the scalp swellings (Fig. 2A). The frontal horns of lateral ventricle were well differentiated (Fig. 2B). The occipital horns were dilated (Fig. 2B) and contained intraventricular blood (Fig. 2C). The third ventricle and basal cisterns were normally differentiated.

Renal ultrasound, echocardiography, fundoscopy and skeletal survey were normal. Karyotyping revealed 46XY karyotype and no other chromosomal abnormalities. The baby expired at 50 h of life - hence a magnetic resonance imaging of the brain and detailed chromosomal analysis for microdeletions, duplications and rearrangements could not be done. The parents refused a necroptic examination.

Although the computed tomography scan revealed a dysmorphic monoventricle, lack of visualization of complete septum pellucidum was not a reliable sign for diagnosis of holoprosencephaly. Early onset of severe fetal hydrocephalus may result in disruption of the septum pellucidum, creating an illusion of a monoventricle in axial sequences- thereby masquerading milder forms of holoprosencephaly (lobar holoprosencephaly) [1]. Additionally, there may be absence of posterior corpus callosum, periventricular gray matter heterotopias, hippocampal and white matter hypoplasia, cortical polygyration and complete disruption of postero-mesial cerebral mantle formation due to severe lateral ventricular enlargement [1]. In fact, the appearance of the wavy membranous structure extending from the left occipital pole is consistent with the residuum of the left mesial posterior cerebral mantle. This interpretation is supported by the fact that there are two collections of intraventricular blood separated by the membranous structure. However for doubtful cases, a magnetic resonance imaging should be done after CSF shunting to delineate the structures which have been affected [2]. Moreover, the simplified gyral pattern (resembling lissencephaly) adds to the complexity of the case. If hydrocephalus develops in-utero, the cerebral hemispheres may show abnormal gyral patterns, characterized by flattened gyri, multiple complex small gyri and reduced sulci [3]. In the setting of delivery of the baby by caesarean section, the fragmented bones were considered to be immature wormian bones.

2. Discussion

Animal models have demonstrated that accumulation of CSF results in reduction of neuronal proliferation from the germinal epithelium. Cell migration is apparently unaffected although a decrease in the number of migrating cells occur [4].

Severe early onset fetal hydrocephalus can be caused by Arnold – Chiari II syndrome, Dandy – Walker syndrome, Congenital infections (Cytomegalovirus [5], Lymphocytic choriomeningitis virus [6], *Toxoplasmosis*), aqueductal atresia/stenosis, brain tumours, vascular malformations or may be genetically determined - microdeletion of 8q12.2 - q21.2 and abnormalities of Xq28 which encodes for

L1CAM protein (a neuronal surface glycoprotein implicated in neuronal migration and axonal differentiation) [7]. This infant had a meningomyelocoele and greater than 95% of such patients have an Arnold – Chiari II malformation [8].

We describe a rare developmental brain anomaly due to fetal hydrocephalus. In their study on 77 subjects with fetal hydrocephalus, Humphreys et al. [1] reported 12 (15.6%) cases with similar anomaly. Koo et al. [9] reported a 30 wk premature infant with congenital hydrocephalus, absent septum pellucidum, thin cerebral parenchyma, focal agenesis of posterior corpus callosum, and rudimentary left sided hippocampus and fornix. Malinger et al. [10] described 10 infants where early onset severe fetal hydrocephalus resulted in disruption of the septum pellucidum; thereby creating an impression of a monoventricle.

Holoprosencephaly, early onset severe fetal hydrocephalus, septo-optic dysplasia and isolated agenesis of cavum septum pellucidum were considered by Malinger et al. [10] as the differential diagnosis of absent septum pellucidum seen in prenatal ultrasounds. Holoprosencephaly signifies lack of cleavage of embryonic prosencephalon into diencephalon and telencephalon, as well as separation of telencephalon into two cerebral hemispheres (which normally occur by embryonic day 35) [11]. Holoprosencephaly is a rare presentation in newborns. Incidence rate is 1:250 in utero, but progressively declines due to high fetal mortality rate to 1:10,000–20,000 live newborns [12]. According to the degree of division in the prosencephalon, holoprosencephaly was classified by De Meyer et al. [13] into alobar, semilobar and lobar types. Septo-optic dysplasia comprises of agenesis of septum pellucidum, optic nerve and pituitary hypoplasia [10]. Isolated agenesis of septum pellucidum is a rare abnormality (incidence being 0.2–0.3/10,000 live birth), and is often associated with corpus callosal abnormalities, midline defects and schizencephaly [14,15].

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