Varied presentation of lobar holoprosencephaly as a cause of macrocephaly in a neonate

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DESCRIPTION
A 37 weeks primigravida presented in obstetric department with the report of labour pain. She had undergone antenatal scans two times; first scan in first trimester and second in third trimester. In the antenatal sonography at around 30 weeks, the fetus was diagnosed with hydrocephalus and macrocephaly, as the head circumference was 31.5 cm, which is >97 percentile for gestational age.1 On examination, the cervix was 4 cm dilated and the fetal head was not engaged. Considering the risk of fetal distress and the presence of macrocephaly, the patient was taken for caesarean section and the baby was delivered at 37 weeks. The baby was admitted in Neonatal Intensive Care Unit (NICU) care in view of hyperbilirubinemia, serum bilirubin was 26.1 mg/dL, for which the phototherapy was given. On clinical examination, the newborn had occipitofrontal circumference of 41 cm, >97 percentile for the age2 and suggestive of macrocephaly. The baby also had downward gaze, frontal bossing and was lethargic (figure 1). After 2 weeks, the bilirubin levels were within normal limits and the patient was advised MRI to rule out the cause of hydrocephalus. On MRI, the findings of lobar holoprosencephaly with enlargement of arachnoid space were present: cavum septum was absent and there was a decrease in head circumference, occipitofrontal circumference was 38 cm. The parents were advised genetic testing for the baby as well as them-selves, however, due to lack of financial support, it was not done. The parents were advised regarding the importance of timely antenatal scan as well, in case of future pregnancy.

Figure 1 Neonate presenting with gross macrocephaly and frontal bossing. No other external anomalies were present.

Figure 2 (A) T2-weighted image on MRI showing the presence of falx cerebri anteriorly (red arrow), absent cavum septum and box-shaped fused frontal horns of lateral ventricles (blue arrow) and traversing vessel across the subarachnoid fluid collection giving cortical vein sign(yellow arrow). (B) Post ventriculo-peritoneal shunting CT image, showing the tip (arrow) of the ventricular end in the enlarged subarachnoid space.

Patient’s perspective
Translated from patient’s mother’s language: As soon as the labour pain started, I was referred to the hospital where I delivered the baby. Post the birth of my baby, we noticed her head was enlarged, for which the doctor advised MRI. The doctor informed us that the baby had an enlarged head due to the collection of fluid and for which shunting was done. I know that this is not common and that the baby will have to be dependent on others and have difficulty in performing routine activities as she grows. I feel privileged hoping that my baby’s story can help some other baby and the parents who are suffering from the same.
Learning points

- In case of holoprosencephaly, the cause of macrocephaly is usually internal hydrocephalus; however, in this case, it was the external hydrocephalus associated with holoprosencephaly as an underlying cause of macrocephaly.
- Surgical shunting proved to be a life-saving procedure in the present case where macrocephaly was unconventionally caused due to external hydrocephalus. The proximal and distal tip of the shunt were placed in subarachnoid space and peritoneal cavity, respectively.

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Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

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