

Encephalocele: A rare clinical image

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Clinical Image Description

Encephalocele is a rare birth defect of the neural tube in which there is a sac like protrusion or projection of brain and the meninges. It occurs when the neural tube, a narrow channel that allows the brain and spinal cord to develop, does not close completely during pregnancy. The portion of the brain that sticks outside the skull is usually covered by skin or thin membrane [1]. It can be either frontal, parietal, occipital or sphenoidal. The exact cause remains unknown but it often occurs in families whose members have had spina bifida or anencephaly. An infant with encephalocele suffers from developmental delay, seizures, hydrocephalus and vision problems. Affected individuals may have progressive weakness in arms and legs due to increased muscle tone. The location of the encephalocele is crucial as there are distinct treatments for anterior and posterior encephalocele. Anterior ones generally have better prognosis as they do not contain brain tissue but posterior ones are associated with neurological problems. Diagnosis is based on prenatal ultrasounds but a small nasal or forehead encephalocele may go undetected [2]. It can be treated with surgery which is done between birth to 4 months of age, but neurological problems may still persist. Studies showed that addition of folic acid to diet of pregnant women reduced the risk of neural tube defects. This infant born with posterior encephalocele could not get proper treatment on time. Thus, the mother and community needs to be educated regarding folic acid supplements and timely intervention for such defects.

Final Diagnosis: Encephalocele

Differential Diagnosis: Meningoencephalocele, cranial encephalocele, meningocele.



Figure 1: Drawings in 6 to 9 months after UCBC therapy.

References

1. <https://www.cdc.gov/ncbddd/birthdefects/encephalocele.html>
2. Ong AGJ, Rolnik DL, Menezes M, Meagher S. Early Diagnosis and Differences in Progression of Fetal Encephalocele. *J Ultrasound Med.* 2020; 39: 1435-1440.

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