Case 14351

Prenatal diagnosis of occipital encephalocele
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Section: Genital (female) imaging
Area of Interest: Foetal imaging
Procedure: Diagnostic procedure
Imaging Technique: MR
Imaging Technique: Ultrasound
Special Focus: Congenital Case Type: Clinical Cases
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Patient: 25 years, female

Clinical History:

A 23-year-old female patient with 5 months of amenorrhoea, came for a routine antenatal scan. No complaints of pain in the abdomen, or bleeding per vaginum. No symptoms of pregnancy induced hypertension.

Imaging Findings:

On USG, a well defined anechoic lesion with internal iso-hyperechoic area was seen arising postero-inferiorly from the occipital region. There was bone defect through which intracranial contents (brain parenchyma and meninges) were seen protruding.

On MRI, T2W images show a well defined hyperintense lesion with internal iso-hypointense brain parenchyma protruding through a bony defect in the occipital region. However, the fetal spine was normal. The fetal movements, cardiac activity and rest of the fetal organs including kidneys were normal. The above imaging findings suggest occipital meningoencephalocele.

The pregnancy was terminated immediately. The fetus showed findings described on the antenatal study and confirmed our diagnosis.

Discussion:

An encephalocele is a bony defect in the skull table, which allows herniation of the meninges or of the brain tissue. It results from failure of the surface ectoderm to separate from the neuroectoderm [2]. These occur 1-4 times per 10,000 live births [1].

Encephaloceles are divided into three major types: sincipital (frontoethmoidal), basal (trans-sphenoidal, sphenoethmoidal, transethmoidal, and spheno-orbital), and occipital [4]. Approximately 75% are occipital, 13% frontal and 12% occur in parietal region [1].

Occipital encephaloceles have been diagnosed on ultrasound from about 9 weeks. After cranial ossification at 10 weeks, the skull defect and occipital sac may be demonstrated. In 30% cases, there is associated spina bifida [1]. Maternal serum alpha-fetoprotein levels are elevated in only 3% of patients, because most encephaloceles are
covered with skin [2].

With ultrasound, encephalocele appear as a defect in the calvarium containing a cystic or solid mass with a gyral pattern that is contiguous with the brain [4]. If the mass appears cystic, the meningocele component predominates, while a solid mass indicates predominantly encephalocele [1]. Associated findings include flattening of the basiocciput (an acute angle between the mass and skin line of the neck and occiput), ventriculomegaly, and lemon sign (inward scalloping of the frontal bones). A cyst-within-a-cyst appearance is occasionally seen. This results from herniation of the fourth ventricle in the encephalocele surrounded by CSF [2].

MRI can provide exquisite detail of the cranial defect and the herniated contents. Contents of the herniated sac may include CSF, disorganized brain tissue, and even ventricles [2]. Corpus callosum dysgenesis, Arnold-Chiari malformation, Dandy-Walker malformation, Meckel-Gruber Syndrome, Amniotic band syndrome, migration abnormalities and chromosomal anomalies are associated features [3]. The fetal kidneys should be examined to exclude Meckel-Gruber syndrome (encephalocele, polycystic kidneys, polydactyly) [1]. Walker-Warburg syndrome, which is also associated with encephaloceles, is a lethal complex of the CNS and eyes. The diagnosis is established by the detection of lissencephaly, hydrocephalus, and a cerebellar malformation. Recognizing these syndromes is important because they are autosomal recessive conditions. Since they can be recognized on prenatal US scans, targeted screening may be possible in the mother's subsequent pregnancies [2].

Differential considerations with an anterior mass include teratoma and cystic hygroma. With a posterior mass, the possibilities include cystic hygroma (if it is predominantly cystic with internal septations), teratoma (if the lesion is predominantly solid), iniencephaly, scalp edema, branchial cleft cyst, hemangioma [1].

The prognosis of encephalocele depends on location of lesion, amount of brain herniation and associated anomalies. Mortality can be up to 44%, and in survivors, intellectual impairment ranges from 40-91% [1].

**Differential Diagnosis List:** Occipital meningoencephalocele, Cystic hygroma, Branchial Cleft cyst, Meckel Gruber syndrome, Scalp hemangioma

**Final Diagnosis:** Occipital meningoencephalocele

**References:**


Description: USG shows a well defined anechoic lesion (arrowheads) protruding through a bone defect in the occipital region with herniation of brain parenchyma within the lesion. Origin:
Department of Radiology, C.U.SHAH Medical College, Surendranagar, Gujarat, India
**Description:** USG shows a well defined anechoic lesion (arrowheads) protruding through a bone defect (arrow) with herniation of brain parenchyma within the lesion. **Origin:** Department of Radiology, C.U.SHAH Medical College, Surendranagar, Gujarat, India
Description: MRI T2W axial image shows a well defined hyperintense lesion (arrows) with internal area (arrowhead) which is isointense to the brain parenchyma. Origin: Department of Radiology, C.U.SHAH Medical College, Surendranagar, Gujarat, India
Description: MRI T2W image shows a well defined hyperintense lesion (arrow) protruding through a bone defect in the occipital region with herniation of the brain parenchyma within the lesion (arrowhead).
Origin: Department of Radiology, C.U.SHAH Medical College, Surendranagar, Gujarat, India
Figure 3

Description: Delivered fetus shows outpouching from the occipital region and confirmed our diagnosis.
Origin: Department of Radiology, C.U.SHAH Medical College, Surendranagar, Gujarat, India
**Description:** Delivered fetus shows outpouching from the occipital region and confirmed our diagnosis.

**Origin:** Department of Radiology, C.U.SHAH Medical College, Surendranagar, Gujarat, India
Description: Delivered fetus shows an outpouching from the occipital region. The spine appears normal. Origin: Department of Radiology, C.U.SHAH Medical College, Surendranagar, Gujarat, India