Unusual presentation of more common disease/injury

Meningocele following aplasia of the occipital bone

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Summary

We describe a first trimester female foetus with aplasia of the occipital bone allowing a meningocele without skin coverage to be formed. The pregnancy was terminated, and on later autopsy the brain appeared to be intact. The foetus carried an apparently balanced translocation 46,XX,t(3;9)(p21.3;q22.3) inherited from a normal father and grandfather.

BACKGROUND

We describe an uncovered meningocele following aplasia of the occipital bone, and to the best of our knowledge aplasia of the occipital bone has not been described before.

CASE PRESENTATION

A 32-year-old woman in her first pregnancy asked for a first trimester ultrasound examination. A midwife with ultrasound experience recognised an irregular skull contour, and the patient was referred to Stavanger University Hospital. A two- and three-dimensional transabdominal ultrasound examination was performed with Voluson E8 ultrasound scanner (Zipf, Austria) with 3.5–7.5 MHz multifrequency transducer. Crown-rump length was measured to 58 mm, corresponding to 12–13 weeks gestational age. An irregular occipital meningocele was diagnosed. The frontal bones and the parietal bones were visualised, but the occipital bone could not be seen (figure 1A,B and videos 1 and 2).

Video 1 3D ultrasound rotational video (around a sagittal axis) of a first trimester fetus with aplasia of the occipital bone. The frontal bones, the parietal bones, the great fontanelle and a meningocele are visualised.10.1136/bcr.10.2010.3437v1

Video 2 3D ultrasound rotational video (around a transverse axis) of a first trimester fetus with aplasia of the occipital bone. The frontal bones, the parietal bones, the great fontanelle and a meningocele are visualised.10.1136/bcr.10.2010.3437v2

INVESTIGATIONS

An amniocentesis was performed and the fetal karyotype was 46,XX,t(3;9)(p21.3;q22.3). A high-resolution array-based genomic copy number analysis on an Affymetrix Genome-Wide Human SNP Array 6.0 (Affymetrix, Santa Clara, California, USA) gave normal results. This balanced translocation was inherited from a normal father and grandfather. There was no history of fetuses or children with similar skull defects in the father's family.

DIFFERENTIAL DIAGNOSIS

Skull defects may also occur in association with aplasia cutis congenita, but these defects are usually restricted to the skull vertex.¹

OUTCOME AND FOLLOW-UP

The parents were informed about a severe malformation and the pregnancy was terminated. Upon foetal autopsy the brain appeared to be intact. The skull defect, which looked like aplasia of the occipital bone, had no skin coverage (figure 1C). It was a female foetus, and no other abnormalities were detected.

DISCUSSION

Detection of neural tube defects is possible already in the first trimester,² and three-dimensional ultrasound is helpful in visualising the development of the skull bones.³ Acrania is the most common rostral neural tube defect associated with anencephaly, and results from failure of the rostral part of the neural tube to close around week 6 weeks of pregnancy, that is, week 4 of foetal life.⁴ Acrania is usually an isolated birth defect with complete absence of the calvarium, exposing a skin-uncovered and malformed brain to the amniotic fluid.² In encephaloceles and the more unusual condition acalvaria, the skull defect is covered by skin.⁵ Encephaloceles are often associated with other malformations.^{2–6}

In the described case, aplasia of the occipital bone with secondary meningocele was the only finding. An intact foetal brain was localised between the parietal bones and not outside the skull contour, and only the meninges were protruding. Probably the partial skull coverage had prevented the brain from being damaged by the shearing forces of the amniotic fluid. The ultrasound appearance was not like an encephalocele, but like acrania only affecting the occipital bone. Agenesis of frontal, parietal and temporal bones have been described earlier.⁷ The occipital part of the calvarium is formed by direct ossification of the cranial

Figure 1 Images of a first trimester fetus with aplasia of the occipital bone. (A) 3D ultrasound image of the foetal skull showing the frontal bones, the parietal bones, the great fontanelle and a meningocele. (B) 2D ultrasound image illustrating a defect in the occipital region. (C) Image after autopsy showing an open skull defect with apparently normal brain surface.

mesenchyme (the interparietal bone) as well as endochondral ossification of three occipital somites that constitute parts of the skull base.⁸

Neural tube defects like acrania and myelomeningoceles are birth defects of multifactorial etiology, where both genetic and environmental factors, like folate deficiency,⁹ may play a role. The genetic causes are with few exceptions unknown.¹⁰ In this case a balanced and familiar translocation between chromosomes 3 and 9 was discovered. None of the breakpoint regions (3p21.3 and 9q22.3, both gene-rich R-bands) are known to harbour genes with a known or potential role in neural tube development. Most likely, the translocation is a random finding unrelated to this rare phenotype. Alternatively, the disruption of a gene or more long-range regulatory or epigenetic changes affecting genes in the vicinity of translocation breakpoints may have contributed to the phenotype.^{11 12}

Learning points

- ▶ Foetal skull defects can be accurately described by 3D ultrasound in early pregnancy.
- ▶ An exact diagnosis of foetal malformation is important counselling the parents.
- ▶ The diagnosis should be confirmed with autopsy.

Competing interests None.

Patient consent Obtained.

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Eggebo TM, Brathetland J, Dirdal HU, Houge G. Meningocele following aplasia of the occipital bone. *BMJ Case Reports* 2011;**10**:1136/bcr.10.2010.3437, date of publication

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