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CLINICAL IMAGE

Rare and severe neural tube defect: Craniorachischisis totalis

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Abstract

Craniorachischisis totalis is an uncommon and severe form of neural tube defect. It is characterized by anencephaly and spina bifida throughout the vertebral column accompanied by herniation of neural tissue and meninges. Hepatic calcification in the fetus is rare and its clinical significance is not fully established.

KEYWORDS

anencephaly, antenatal diagnosis, craniorachischisis, liver calcifications, neural tube defects

A 24-year-old gravida 2 para 0 woman, was referred to the gynecology department because of suspected cranial abnormality of the fetus.[1,2] The patient's obstetrical history was significant for a consanguineous marriage and a medical abortion of a female fetus with meningoencephalocele. The patient had not taken any folic acid supplementation. Ultrasound examination revealed a singleton pregnancy at 14 weeks and 3 days of gestation with acrania, anencephaly, and complete division in the fetal spine (Figure 1A). After informed consent of the patient, the pregnancy was terminated and the fetus was sent for an autopsy examination. We received a male fetus, weighing 16 g for autopsy. On external examination, the cranial vault was absent (acrania) and the brain was not fully formed (Figure 1B). The orbit was partially formed, with bulging eyes. The spinal cord showed bifid vertebrae exposing the neural tissue up to sacral vertebrae (Figure 1C). There was complete failure of neural tube closure, exposing the spinal cord (Figure 1C). Histological examination of the different organs of the fetus revealed calcifications in the liver (Figure 1D). Karyotyping of the fetus did not disclose chromosomal anomalies. The final diagnosis was craniorachischisis totalis.

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CONFLICT OF INTEREST

None declared.

AUTHOR CONTRIBUTIONS

Dr Faten Limaiem and **Pr Kaouther Dimassi** prepared, organized, wrote, and edited all aspects of the manuscript. **Dr Faten Limaiem** prepared all of the histology figures in the manuscript. **Pr Kaouther Dimassi** participated in the conception and design of the study, the acquisition of data, analysis, and interpretation of the data. All authors contributed equally to preparing the manuscript and participated in the final approval of the manuscript before its submission.

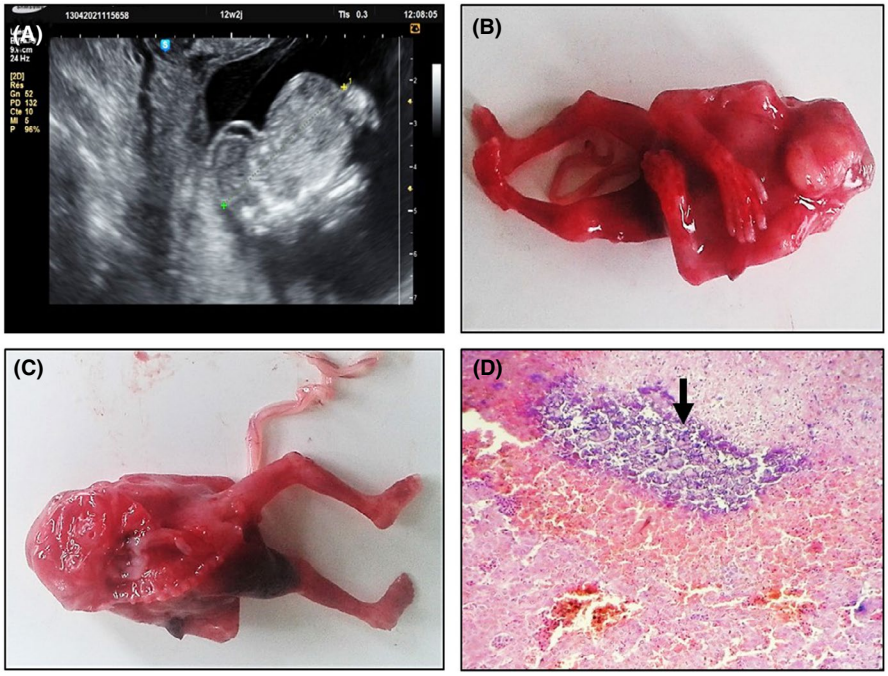

FIGURE 1 (A) Ultrasonographic view of the fesstus with total craniorachischisis. (B) Frontal view of the fetus showing anencephaly. The orbit is partially formed, with bulging eyes. The cranial vault is absent (acrania) and the skull base flattened. (C) Posterior view of the fetus showing complete division of the spine: total spina bifida in the fetus of 14 weeks +3 days of gestational age. There is complete failure of neural tube closure, exposing the spinal cord. There has been no fusion of the rostral tube, resulting in abnormal development and damage of the forebrain structures which are replaced by hemorrhagic tissue. (D), Foci of hepatic calcifications were noted in the liver parenchyma (black arrow) (Hematoxylin and eosin, magnification ×200)

ETHICAL APPROVAL

All procedures performed were in accordance with the ethical standards. The examination was made in accordance with the approved principles.

CONSENT

Published with written consent of the patient.

DATA AVAILABILITY STATEMENT

In accordance with the DFG Guidelines on the Handling of Research Data, we will make all data available upon request.

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