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Case report

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Craniorachischisis: a case report

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Abstract

Neural tube defects (NTDs) are congenital disorders. They involve incomplete spinal column development or even the absence of the cranial vault. The presence of both anencephaly and spina bifida is known as craniorachischisis. It can detect fetal scans at 18 to 22 weeks' gestation. We report a case of Mrs J.R, 24 years old. She is primigravida; the result of a nonconsanguineous marriage; free of dysgravidia. Pregnant at 18 weeks of amenorrhea. Second-trimester prenatal screening test revealed a neural tube defect (NTD). The diagnosis was confirmed with ultrasonography. After a multidisciplinary prenatal consultation and with their informed consent, the parents agree to terminate the pregnancy. The findings suggest that prenatal counseling, and regular folic acid intake can help prevent additional complications.

Therefore, reporting our clinical case and explaining some ultrasound characteristics of these defects is important, as well as clarifying prenatal diagnostics, and prevention methods.

Introduction

Neural tube defects arise due to a neurulation failure approximately 28 days post-conception, usually before most women are aware of their pregnancy. The three primary forms are anencephaly, encephalocele, and spina bifida cystica (open spina bifida) [1]. These anomalies are the result of a complex interaction between genetic factors and environmental influences, with numerous studies suggesting a genetic component [1]. The typical occurrence rate of neural tube defects (NTDs) is 1 in every 1,000 births, while the frequency of craniorachischisis is less than 0.1% [2]. Craniorachischisis is a very rare form of neural tube defect, characterized by the simultaneous presence of spina bifida and anencephaly in a newborn baby, and is considered the most severe and fatal type [3]. Despite this, it has been mostly ignored and has only recently become the subject of more extensive research. Despite the decline in NTDs documented in low- and middle-income countries due to folic acid fortification, prenatal counseling, fetal scans, and second-trimester screenings, surveillance for NTDs remains underdeveloped in less developed countries [4]. We report a case of craniorachischisis.

Patient and observation

Patient information: Mrs. J.R a 24-year-old, has no medical or surgical antecedents, no alcohol or tobacco consumption, primigravida, nonconsanguineous marriage, free of dysgravidia, pregnant at 18 weeks of amenorrhea, on martial therapy with no folic acid. The patient missed the first-trimester screening.

Clinical findings: the patient was conscious, well-oriented, and in good haemodynamic condition with no functional complaints.

Timeline of the current episode: the patient conveyed worry regarding her initial pregnancy, so, second-trimester prenatal screening at 16 weeks of gestation and morphological ultrasound scan at 18 weeks of gestation were suggested.

Diagnostic assessment: the second-trimester prenatal screening at 16 weeks of gestation revealed a neural tube defect by the high level of alpha-fetoprotein in the mothers' blood. The diagnosis was confirmed with a morphological ultrasound scan at 18 weeks of gestation that shows thoracic-cervical myelomeningocele measuring 32.9 millimeters and anencephaly (Figure 1, Figure 2). The remainder of the morphological examination did not show any abnormalities.

Diagnosis: the craniorachischisis was confirmed. All newborns with craniorachischisis, like ours, either die soon after birth or are stillborn.

Therapeutic interventions: following antenatal diagnostic counseling and giving their informed consent, the parents chose to terminate the pregnancy. The patient underwent cervical ripening using a Foley catheter and misoprostol (400 micrograms sublingually every 3 hours).

Follow-up and outcome of interventions: subsequently, the patient expelled a dead male fetus, weighing 100 grams. There was a defect in the formation of the scalp and vault (acrania) which was extending up to the thoracic part of the vertebral column brain tissue and spinal cord were covered only by a membranous tissue (Figure 3). Retroflexion of the spine was observed. The neck was short, the nose was broad, and the eyes were bulging (Figure 4). No other abnormalities were seen. Follow-up proceeded without any clinical or biological complications, and the patient was discharged after being administered cabergoline to inhibit lactation. It is recommended to provide

genetic counseling and folic acid supplementation during the periconceptional period for future pregnancies as preventive measures.

Patient perspective: the patient may experience intense grief and a sense of loss. This is not just about the termination, but also the loss of hopes and dreams associated with pregnancy. The patient has gone beyond this acute stage.

Informed consent: an informed consent was obtained from the patient.

Discussion

Neural tube defects (NTDs) are common congenital abnormalities resulting from incomplete closure of the neural tube in early embryonic development. Anencephaly and spina bifida are major factors contributing to mortality rates among fetuses and infants [5]. Craniorachischisis Totalis represents an uncommon and extremely severe type of neural tube defect (NTD). It involves anencephaly and spina bifida along the entire length of the vertebral column, accompanied by the herniation of neural tissue and meninges [5]. Based on 2015 estimates, approximately 260,100 newborns worldwide are born annually with neural tube defects (NTDs), with a global prevalence ranging from 213,800 to 322,000 (95% CI) (4). In Tunisia, the overall prevalence of NTDs from 1991 to 2011 was 2.02 per 10,000 live births. Among NTD cases, 20.5% (n = 158) were diagnosed with craniorachischisis [6].

When the vertebral column and cranial vault do not develop properly, the neural tissue is at risk of substantial damage. This exposure makes the neural tissue vulnerable to injury from amniotic fluid and mechanical trauma as the brain tissue grows. Consequently, the exposed neural tissue undergoes secondary degenerative changes, resulting in the formation of a mass of primitive vascular connective tissue [7]. Based on our observation, we could only observe rudimentary neural tissue, which made it difficult to identify specific neural structures. In addition to defects in the vertebral column and cranial vault, we also noted bulging eyes, a broad nose, and a small neck. The occurrence of NTDs varies based on geographic location, fetal gender, ethnicity, and the socioeconomic status of the parents.

In our case, the couple has a low socioeconomic status, along with advanced maternal age. Inadequate folate intake during early pregnancy represents a significant risk factor for neural tube defects [8]; we find this in our case. Factors such as maternal diabetes, maternal obesity, maternal fever in early pregnancy, and amniotic band disruption during gestation are believed to increase the risk of neural tube defects (NTDs) [3]. Studies have also shown that open neural tube defects (NTDs) such as craniorachischisis, anencephaly, and spina bifida are more prevalent in females than in males [3]. Genetic studies of humans with craniorachischisis, conclude that Missense variants in CELSR1 and SCRIB may contribute to this condition, potentially mirroring findings in mice where defective trafficking of planar cell polarity proteins to the plasma membrane is a plausible pathogenic mechanism [9].

Regular ultrasound exams are the first step towards an early diagnosis. Additionally, elevated alpha-fetoprotein levels in maternal blood and amniotic fluid suggest the existence of a neural tube defect. In our case, the second-trimester prenatal screening test revealed neural tube defects confirmed with ultrasonography that show thoracic-cervical myelomeningocele and anencephaly. All newborns with craniorachischisis, like ours, either die soon after birth or are stillborn. These illnesses consistently lead to death and currently have no approved cure [10]. It is recommended to end the pregnancy if the pathology is discovered early. It is advised that parents whose child is impacted should use preventive measures like folic acid supplements and genetic counseling when they are trying to conceive in the future.

Conclusion

Craniorachischisis is a birth defect caused by the incomplete closure of the neural tube in early embryonic development. If diagnosed early with an ultrasound scan, medical termination of the pregnancy is recommended, as survival is not feasible. Substantial evidence indicates that a significant number of neural tube defects are due to folate deficiency and can be prevented. Parental counseling and the use of folic acid supplements, beginning before conception and continuing during pregnancy, are advised preventive strategies. It is recommended that fertile women take 400 micrograms of folic acid per day.

Competing interests

The authors declare no competing interests.

Authors' contributions

Patient management: Aymen Khalfaoui and Achraf Ouadday. Data collection: Aymen Khalfaoui and Nairouz Mtir. Manuscript drafting: Aymen Khalfaoui. Manuscript revision: Aymen Khalfaoui and Houssem Ragmoun. All the authors have read and agreed to the final manuscript.

Figures

Figure 1: ultrasonographic view of the fetus with craniorachischisis (anencephaly - myelomeningocele measuring 32.9 millimeters; thoraco-cervical myelomeningocele arrow: anencephaly

Figure 2: 4D ultrasonographic view of the fetus with craniorachischisis; arrow: thoraco-cervical myelomeningocele flash: anencephaly

Figure 3: posterior view of the fetus at 18 weeks gestational age, demonstrating complete division of the spine and total spina bifida; there is complete failure of neural tube closure, leading to exposure of the spinal cord; the rostral tube has not fused, causing abnormal development and damage to forebrain structures, which are replaced by hemorrhagic tissue; arrow: spina bifida flash: forebrain structure is replaced by hemorrhagic tissue

Figure 4: frontal view of the fetus showing anencephaly; the orbit is partially formed, with bulging eyes and broad nose

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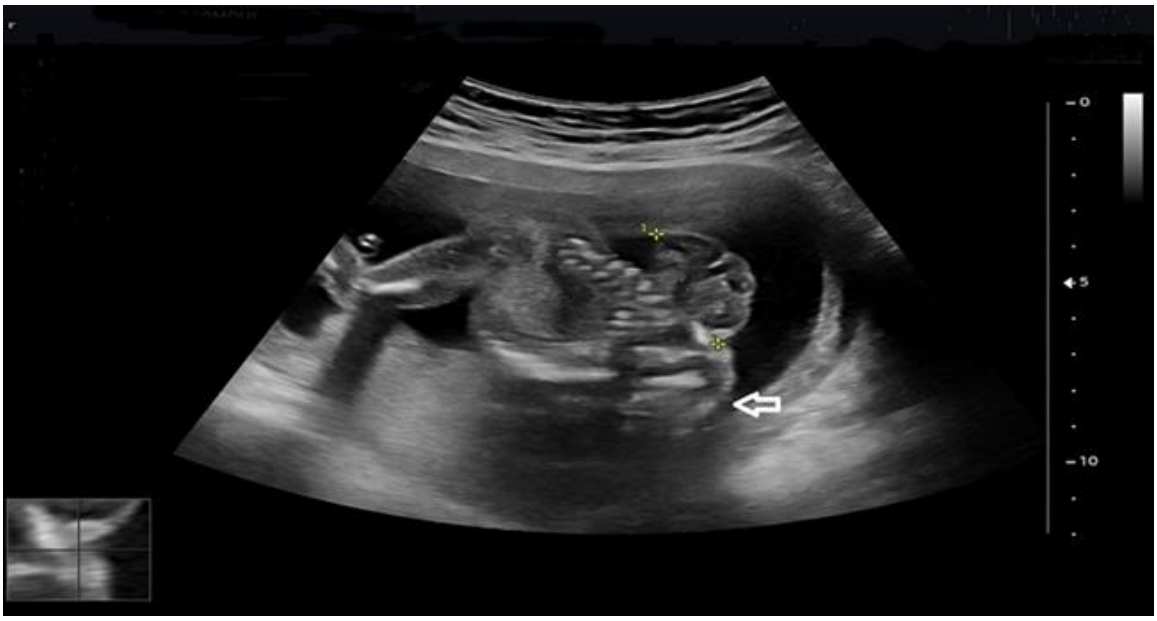

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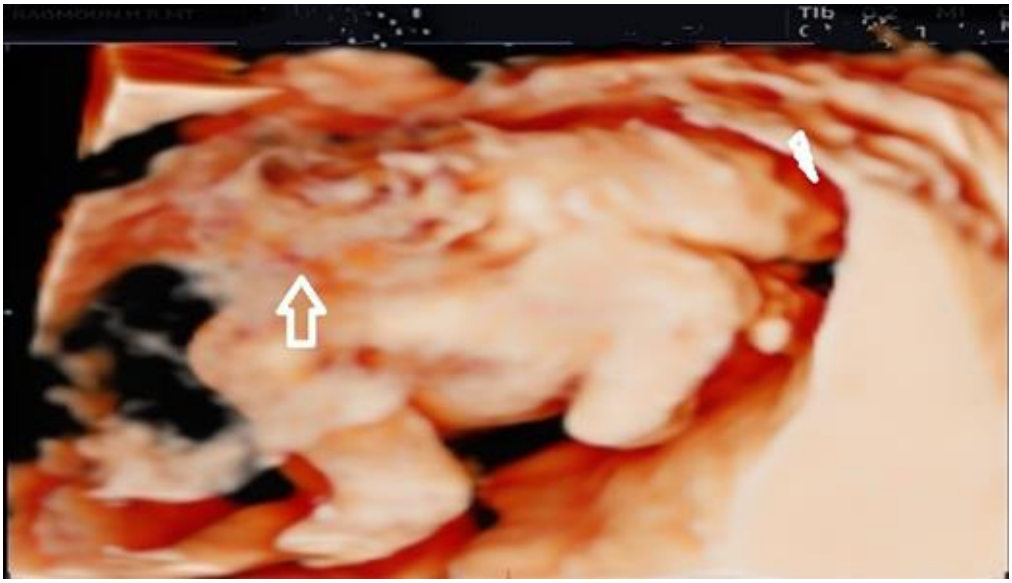
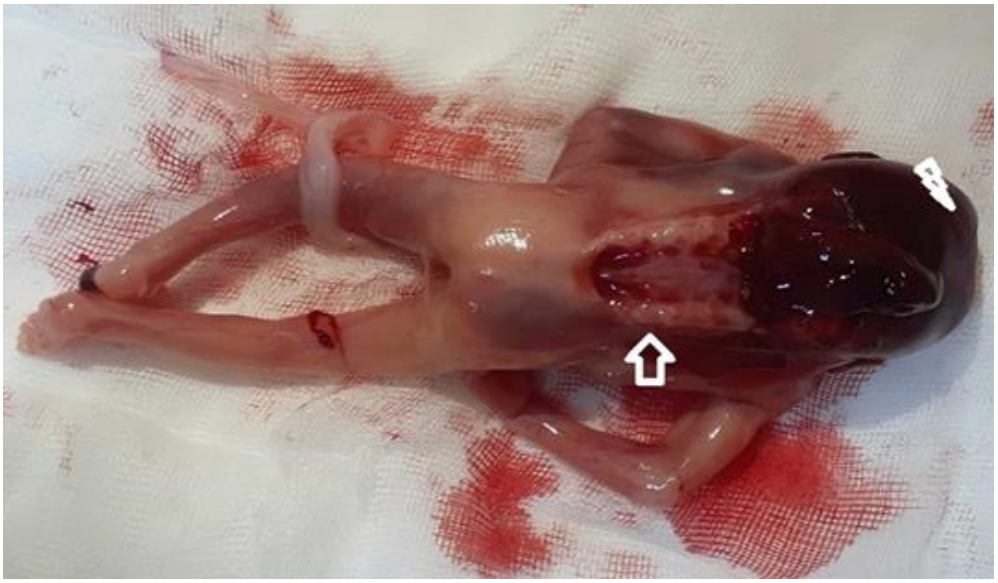


Figure 2: 4D ultrasonographic view of the fetus with craniorachischisis; arrow: thoraco-cervical myelomeningocele flash: anencephaly



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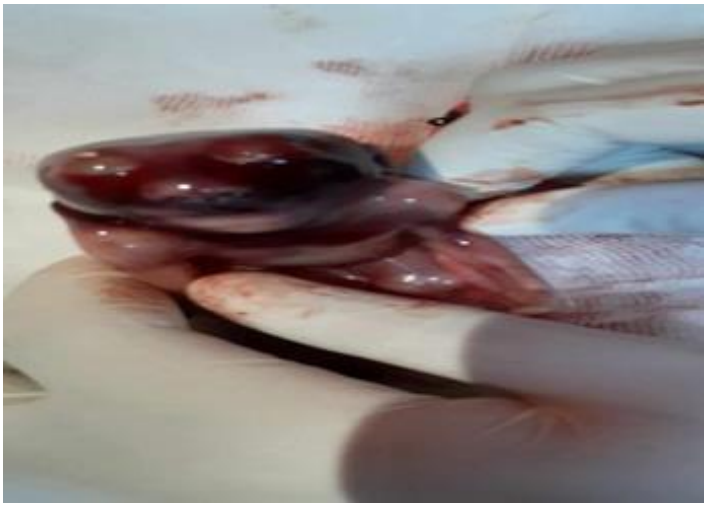

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