

A Case of Recurrent Exencephaly

Divya Gautam, Ciaran McKeown, Mark Dempsey, Department of Obstetrics and Gynaecology, University College Hospital Galway

Objective: The majority of exencephaly cases are sporadic, however, in those who have had one prior pregnancy affected by exencephaly, the recurrence risk is estimated to be 2-5%. This case describes a rare case of recurrent exencephaly in a woman with no known medical conditions or genetic abnormalities, who had been taking 5mg of folic acid preconception and highlights the usefulness of ultrasound in identifying such structural abnormalities.

Case Report: A 35-year-old lady (G3P2) was referred to the high-risk clinic in a tertiary care centre at 14+2 weeks gestation due to a history of exencephaly, in her last pregnancy, which resulted in an early neonatal death. In the current pregnancy, foetal ultrasound performed at 14⁺³ weeks gestation, demonstrated features of exencephaly. The option of amniocentesis was offered, as well as the option to terminate the pregnancy. Amniocentesis was performed at 15⁺², revealing a normal karyotype. The patient decided to proceed with medical termination of pregnancy at 17 weeks gestation, using a combination of mifepristone and misoprostol. During her care, the patient met with bereavement support services within the hospital, who outlined the care pathway from diagnosis to delivery, and the investigations that may be undertaken in such cases. In this case, cytogenetics and parental chromosomes did not reveal any cause of the recurrence of exencephaly.

Conclusion: This case illustrates the sensitivity of obstetric ultrasound in the early diagnosis of exencephaly. Couples should be counselled in relation to recurrence risk and close monitoring in subsequent pregnancies, despite recurrence being uncommon.

Discussion: In cases such as these, further investigations such as whole exome sequencing may be undertaken to identify genes that may be associated with the condition. Recognised examples are genes involved in folate metabolism. Further investigation is, however, considered academic as the risk of anencephaly after two pregnancies with exencephaly is 10% and it can be diagnosed on foetal ultrasound scan. Other than an absent/poorly formed cranial vault, radiographic features may include an absent nasal bone, brain tissue seen herniated into amniotic fluid or attached to the amniotic membrane. Recognition of this syndrome is important, so guidance can be offered, in terms of obstetric care and psychological support. Conversations and the procedures surrounding termination can be complicated by psychological barriers and obstetric circumstances like non cephalic presentation.

