University College London Hospitals NHS
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First trimester diagnosis of Joubert syndrome - a case report

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Joubert syndrome is a rare autosomal recessive disease that includes cerebellar vermian hypoplasia as a distinct feature along with other structural abnormalities. It is associated with severe neurodevelopmental delay. **Reports on first trimester diagnosis are scarce** and early detection usually only occurs in those cases with other brain abnormalities, or in cases where the patient's obstetric history (eg. a previously affected pregnancy) prompts early fetal medicine specialist review.

Our case was in a primigravida. A brain anomaly in keeping with Joubert syndrome was detected at **12 weeks** during her **routine first trimester scan** and was initially suspected owing to an enlarged 4th ventricle and abnormal cerebellum and brain stem on axial views of the brain posterior fossa (Image A), with normal first trimester standard brain images (choroid plexuses, midline echo) (Image B). In addition, there was left hand postaxial polydactyly. Subsequent review in the fetal medicine unit confirmed the findings. Following counselling, the parents opted to continue the pregnancy. An amniocentesis demonstrated normal fetal karyotype and microarrays. Postnatal genetic investigations confirmed diagnosis of Joubert syndrome.

Due to severe ventriculomegaly and macrocephaly (Image C), an elective caesarean section was planned for 36 weeks' gestation. The foetus was born alive and was transferred to the neonatology intensive care unit in the local paediatric hospital.

In our ultrasound unit, the first trimester scan protocol includes assessment of the cerebral posterior fossa in the axial plane, and in this instance facilitated early diagnosis of a rare and severe condition.

Conclusions: 11-13 weeks' routine scan detection rates of cerebellar-vermian abnormalities are low. **The addition of posterior fossa axial views to the national first trimester screening programme recommendations may improve nationwide screening performance in detecting severe brain defects.**

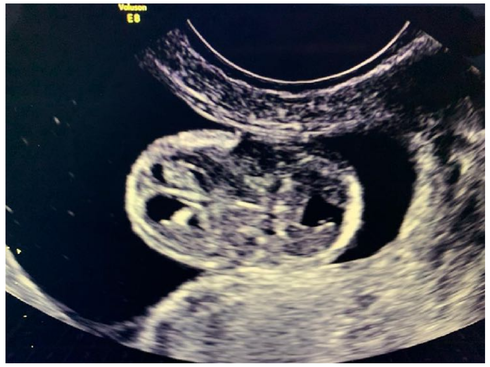

Image A

Image B

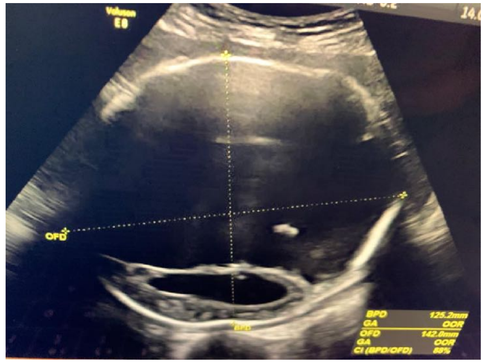

Image C

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