

14a04ed Prenatal suspicion of Joubert syndrome by targeted neurosonography: diagnostic value and clinical implications

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BACKGROUND

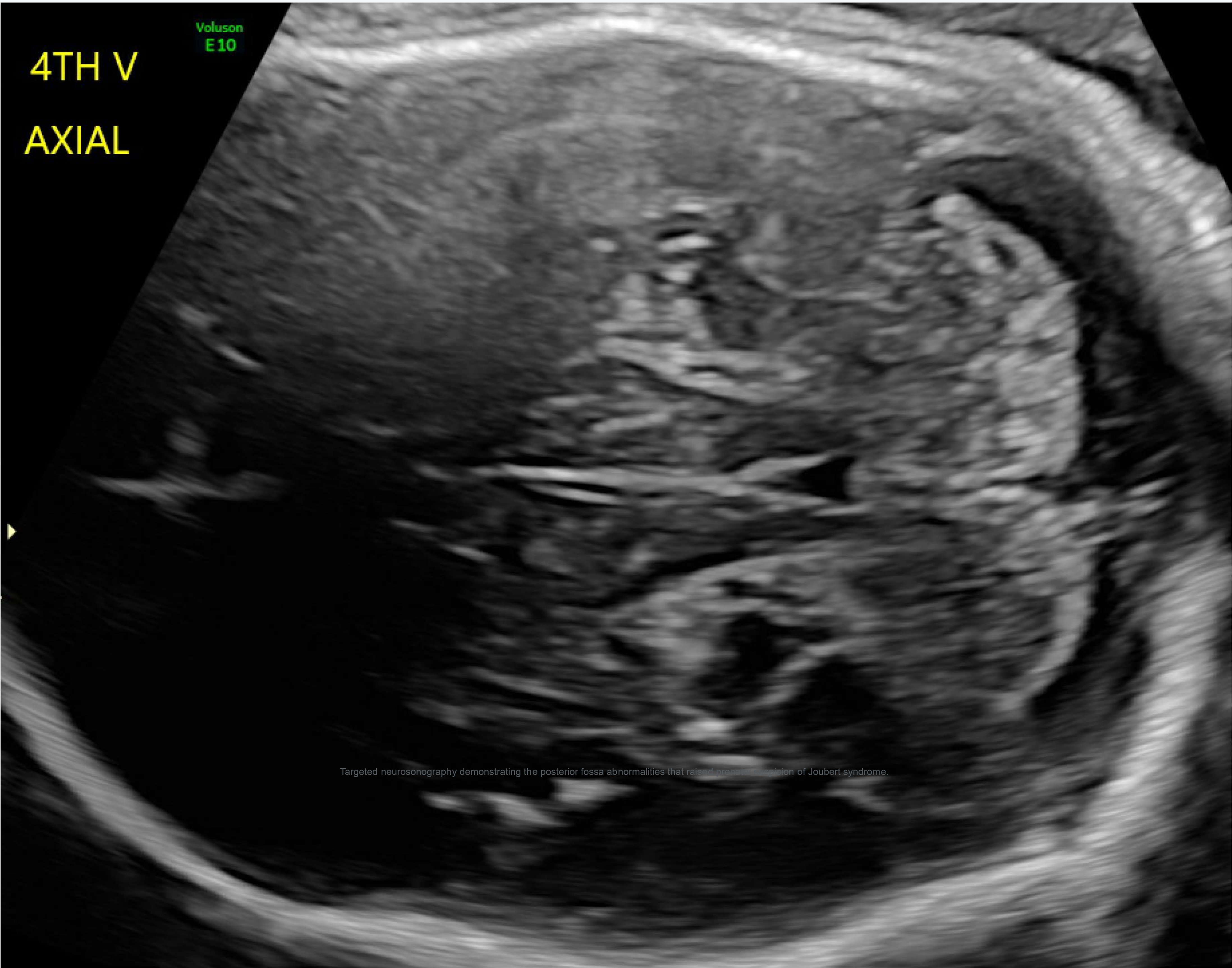
Joubert syndrome is a rare ciliopathy characterized by cerebellar vermian hypoplasia and brainstem malformation. Prenatal diagnosis remains challenging and is often made postnatally.

CASE

- 39-year-old G4P0030 referred at 23 weeks for suspected vermian agenesis
- Transvaginal neurosonography raised suspicion for Joubert syndrome
- Fetal MRI was limited by motion artifact
- Repeat neurosonography at 32 weeks confirmed persistent findings
- Postnatal MRI established the diagnosis

CLINICAL IMPLICATION

Serial targeted neurosonography can provide decisive prenatal information when fetal MRI is technically limited, allowing multidisciplinary counseling and postnatal care planning.



KEY PRENATAL FEATURES

- 1 Vermian hypoplasia
- 2 Abnormal 4th ventricle morphology
- 3 Laterolateral/AP diameter ratio <1
- 4 Deepened interpeduncular fossa

DIAGNOSTIC PATHWAY

- | | |
|---------------|---|
| 23 w | Targeted NSG → suspicion |
| Fetal MRI | Vermian hypoplasia; brainstem limited by motion |
| 32 w | Serial NSG → persistent findings |
| Postnatal MRI | Thickened superior cerebellar peduncles + dysplastic 4th ventricle → JS confirmed |

TAKE-HOME MESSAGE

For suspected Joubert syndrome, targeted neurosonography should actively assess the vermis, fourth ventricle and interpeduncular fossa.