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Complex cerebral malformation in prenatal diagnosis: consider Joubert Syndrome

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Introduction

Joubert syndrome JS is a rare autosomal recessive neurodevelopmental disorder characterized by midbrain and hindbrain malformations

Case

We report the case of a 27 Years old ,primigravid primiparous, patient in whom a complex cerebral malformation was detected at 23 weeks of amenorrhea during prenatal assessment . Fetal ultrasound revealed vermian hypoplasia associated with brainstem anomalies and thickened superior cerebellar peduncles on ultrasound **Molar tooth sign** (Figure 1) , raising suspicion of a ciliopathy. Following termination of pregnancy, a detailed fetopathological examination confirmed the diagnosis of JS (Figure 2) .

Histological analysis demonstrated characteristic cerebellar and brainstem malformations, supporting the prenatal imaging findings particularly the "molar tooth sign" observed on imaging. This case highlights the importance of prenatal imaging in detecting complex brain malformations and the role of fetal pathology in confirming the diagnosis although prenatal diagnosis of JS remains challenging due to overlapping features with other mid-hindbrain malformations.

Conclusion

Early recognition of JS is crucial for genetic counseling and guiding pregnancy management. A multidisciplinary approach involving obstetricians, neuropathologists, and geneticists is essential to optimize diagnostic accuracy and parental guidance.

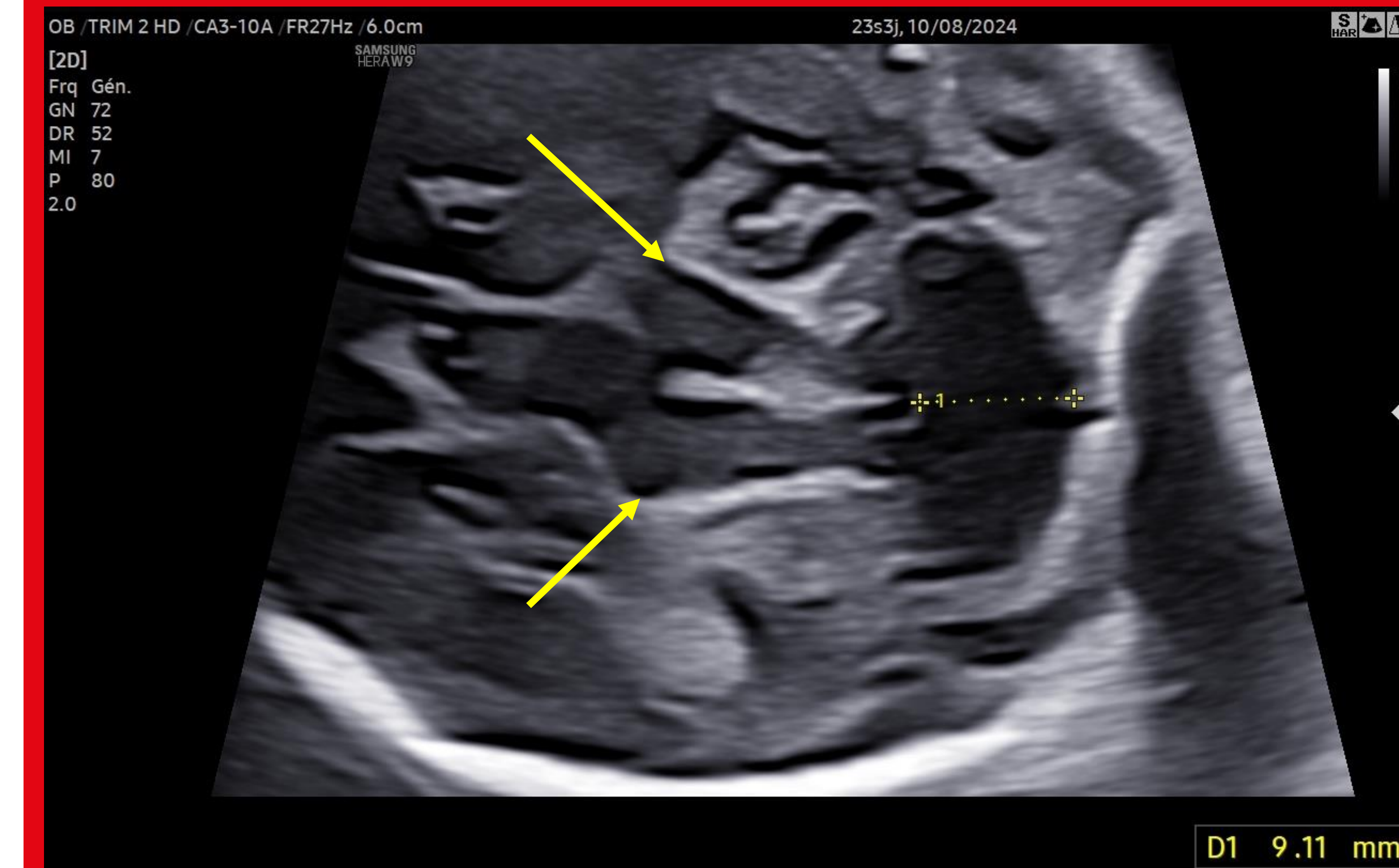


Figure 1 thickened superior cerebellar peduncles on ultrasound Molar tooth sign



Figure 2: cerebellar peduncles aspect(molar tooth)