Prenatal Diagnosis of Rhombencephalosynapsis Associated With Multiple Fetal Anomalies and a Maternally Inherited EP300 Gene Variant: A Case Report

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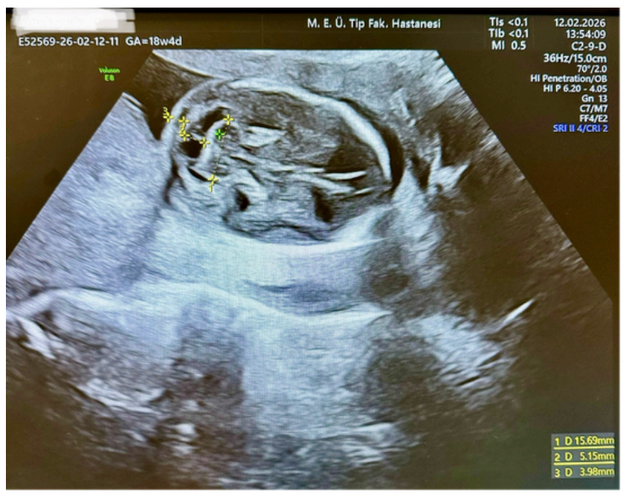
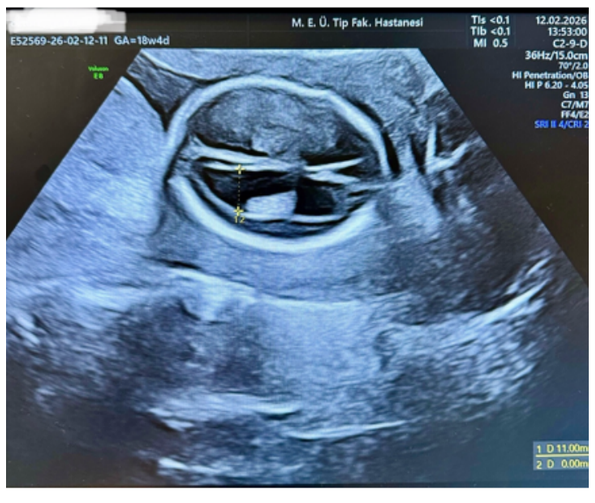
INTRODUCTION

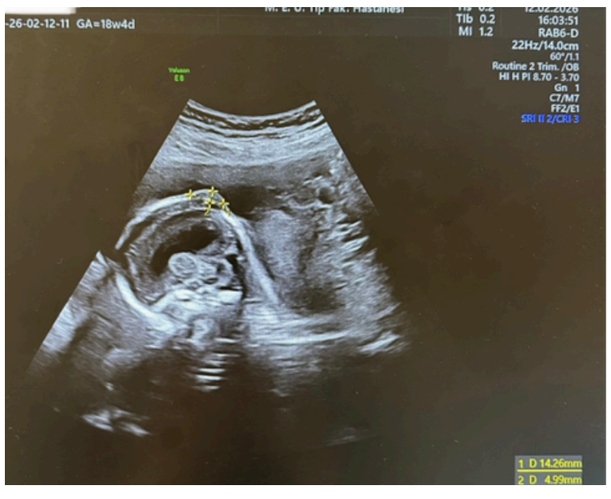
Rhombencephalosynapsis (RES) is a rare cerebellar malformation characterized by midline fusion of the cerebellar hemispheres with vermian agenesis, arising from disrupted dorsal–ventral hindbrain patterning [1].
While its etiology is often linked to ciliopathies or the SHH signaling pathway, many cases remain genetically unexplained. [2].
We report a unique case of RES associated with an EP300 variant, expanding the known phenotypic spectrum of this gene.

CASE PRESENTATION

A 26-year-old (G3P1) woman presented at 18 weeks of gestation



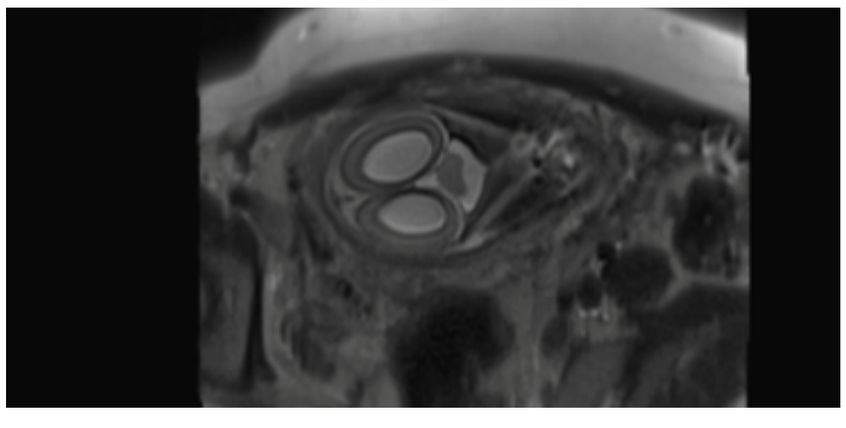


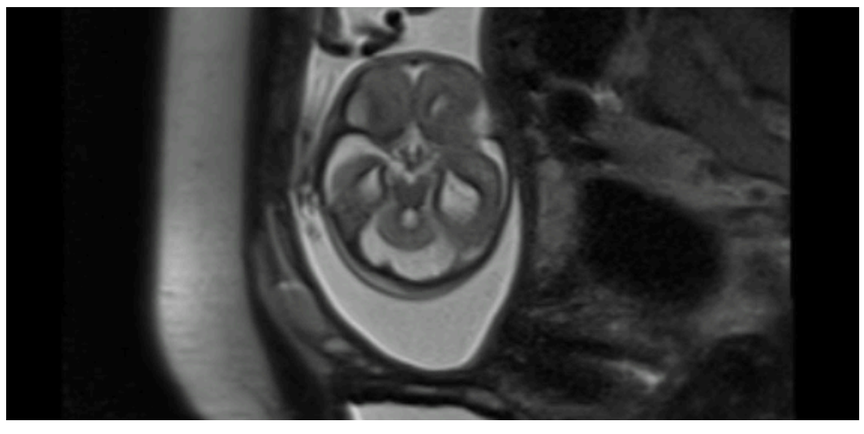


18 Week USG Findings :

- Hypoplastic cerebellum (TCD: 15.69 mm)
- Mild ventriculomegaly (LV:11 mm)
- Protruding 14.6 × 4.99 mm parietooccipital myeloencephalocele
- Grade 2 hyperechogenic bowel and a small mid-muscular VSD.

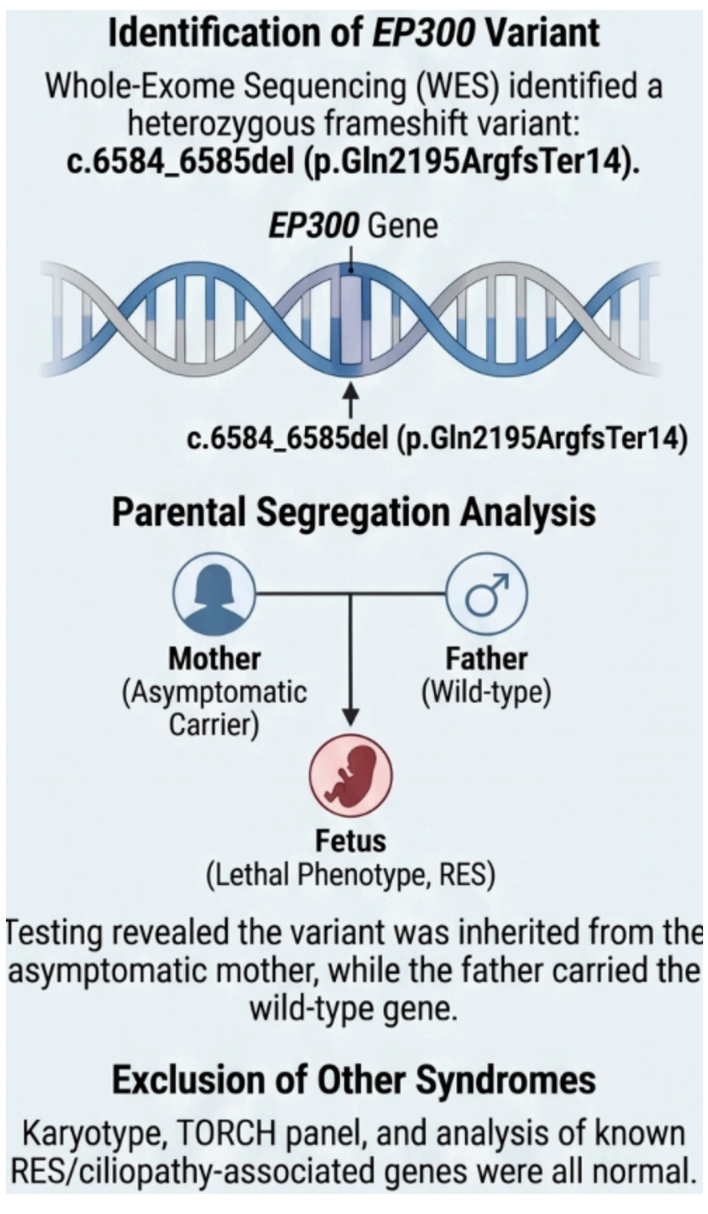
19- Week Fetal MRI
confirmed dorsal fusion of the cerebellar hemispheres with a continuous folial pattern, diagnosing RES.



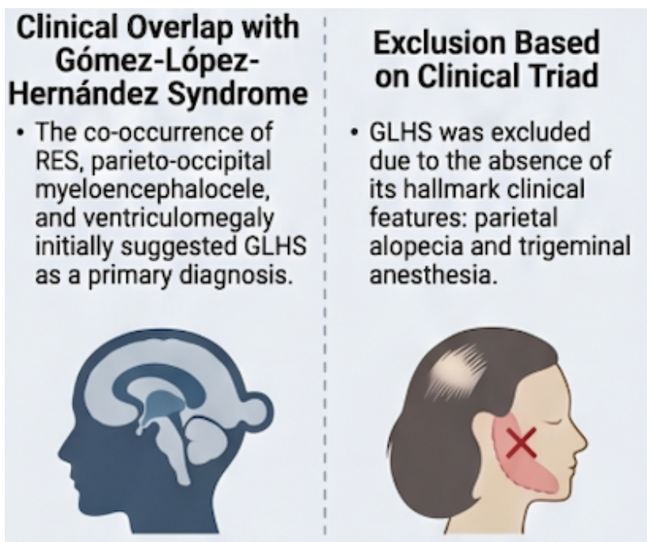


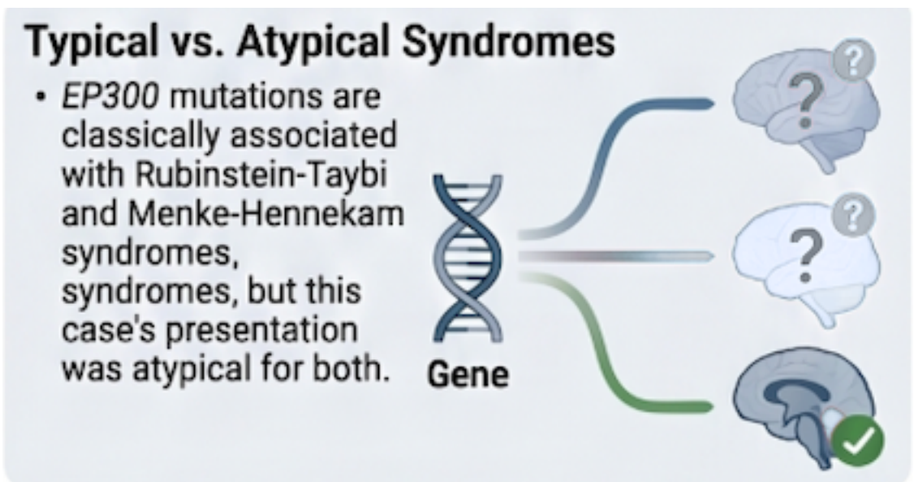
There was no consanguinity between the parents. The mother had no known systemic illnesses and there were no pathological findings on the first-trimester screening test. Based on the findings, **fetal karyotype** and **WES** (whole-exome sequencing), **TORCH panel** was performed .

Genetic Findings (WES Result)



DISCUSSION





While the progression from WES finding to MRI confirmation underscores the strength of genotype-phenotype correlation in this instance, suggests EP300 as a candidate in the complex molecular architecture of RES.

CONCLUSION

- **This case highlights EP300's potential role as a transcriptional regulator in posterior fossa development; however additional case studies are needed to establish a definitive causal relationship. [3].**
- **Furthermore highlights the importance of comprehensive genetic evaluation in patients with rhombencephalosynapsis.**

REFERENCES

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