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Prenatal Diagnosis of Partial Trisomy 9p and 15q11.1-q13.2 Deletion

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Introduction

A rare prenatal chromosomal imbalance presenting with multisystem fetal abnormalities, highlighting the complementary diagnostic roles of detailed ultrasound, fetal MRI, karyotyping, and chromosomal microarray analysis (CMA).

Case & Diagnostic

28-year-old primigravida 28+2weeks

Multiple fetal anomalies

- Targeted US+ neurosonography
- Fetal MRI
- Amniocentesis
- Karyotype + CMA

Partial trisomy 9p + 15q11.1-13.2 deletion

Genomic findings

CHROMOSOMAL MICROARRAY

9p24.3–q21.1 duplication → Partial trisomy 9p
15q11.1–q13.2 heterozygous → Prader- Willi / Angelman
critical region spared

Integrated Diagnosis

Partial Trisomy 46,XY + 9p + 15q11.1-q13.2

Rare complex chromosomal imbalance associated with a severe multisystem fetal phenotype.

Conclusion

Integrated fetal imaging and genomic testing enabled precise prenatal diagnosis of an exceptionally rare chromosomal imbalance. Multimodal assessment refined genotype-phenotype correlation, supported prognostic counseling, and informed individualized perinatal decision-making.



Figure 1. Bilateral cleft lip, jaw and palate

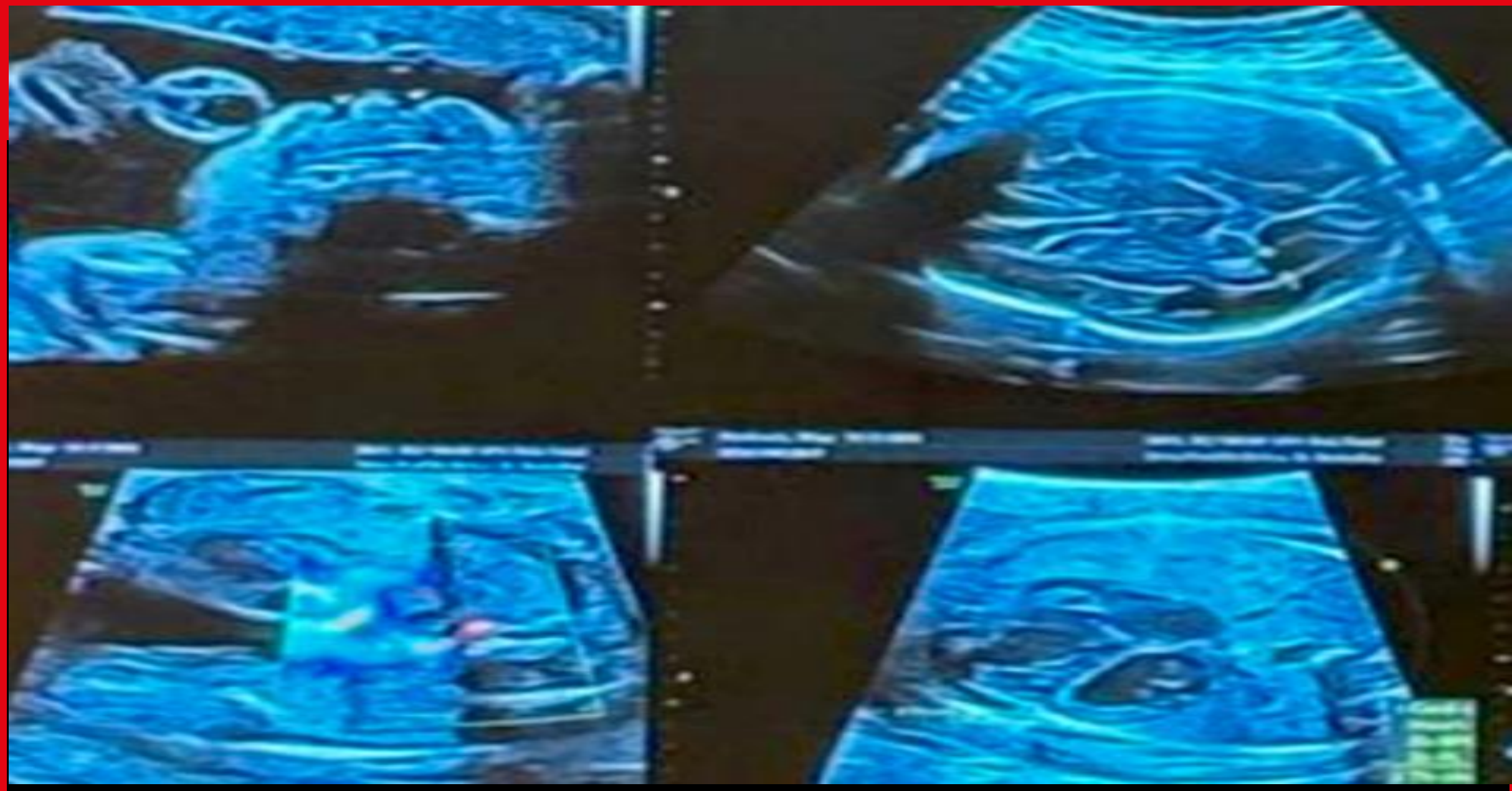


Figure 2. Bilateral ventriculomegaly, cardiomegaly

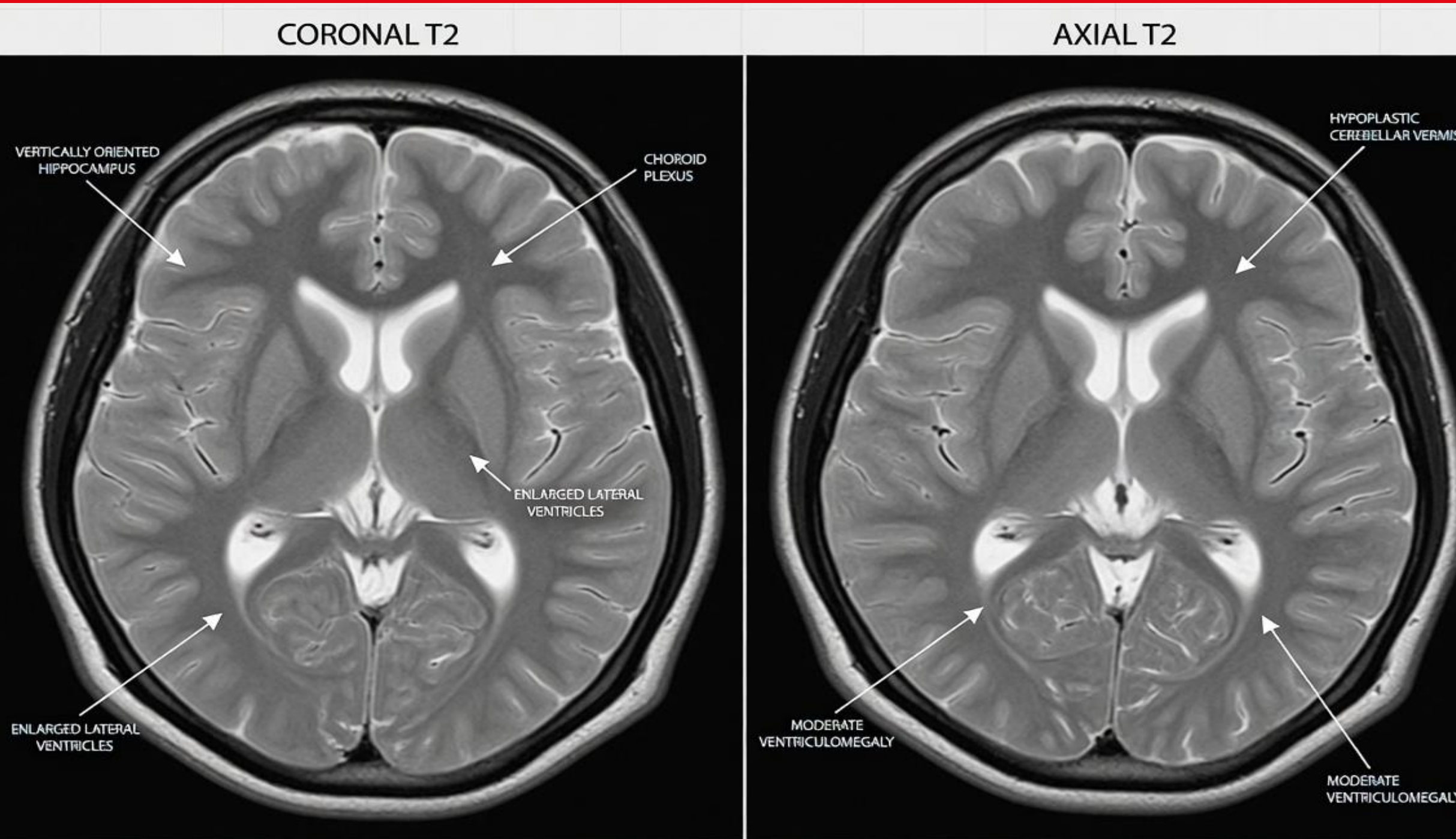


Figure 3. Moderate ventriculomegaly, vertically oriented hippocampi, and hypoplastic cerebellar vermis