

EP15.55: Prenatal diagnosis of spastic paraplegia type 47 (SPG47) presenting with ventriculomegaly and clubfoot: a case report

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Background

Spastic paraplegia type 47 (SPG47) is a rare autosomal-recessive hereditary spastic paraplegia caused by biallelic AP4B1 variants. Prenatal sonographic descriptions remain sparse. We report a case identified through detailed neurosonography combined with trio whole-exome sequencing (WES).

Case Report

A second-trimester scan at 23+2 weeks (GE Voluson E22, ISUOG protocol) with Omniview neurosonography prompted amniocentesis with trio WES and karyotyping. Findings: EFW <10th centile, shortened long bones, bilateral clubfoot, and asymmetric ventriculomegaly (anterior horn 5 mm, posterior horn 13 mm) with floating choroid plexus; corpus callosum, cerebellum, and posterior fossa were normal.

Genetic Findings

Trio WES identified a homozygous frameshift variant c.1160_1161del (p.Thr387ArgfsTer30) in AP4B1 (1p13.2), confirming SPG47. Both parents were heterozygous carriers. After counseling, the pregnancy was terminated.

Key Sonographic Clues

No single marker was specific, but posterior-dominant ventriculomegaly, floating choroid plexus, short long bones, growth restriction, and clubfoot prompted genetic workup.

Clinical Significance

Such combinations should raise suspicion for a monogenic etiology and prompt comprehensive genomic testing, even when no isolated finding is diagnostic.

With both parents confirmed heterozygous carriers and autosomal-recessive inheritance, recurrence risk in future pregnancies is 25%, supporting early targeted prenatal diagnosis.

Conclusion

This case adds to the sparse literature on prenatal SPG47. Combining neurosonography with trio WES enabled a precise diagnosis, supporting counseling and risk assessment.



Figure 1. Bilateral clubfoot.

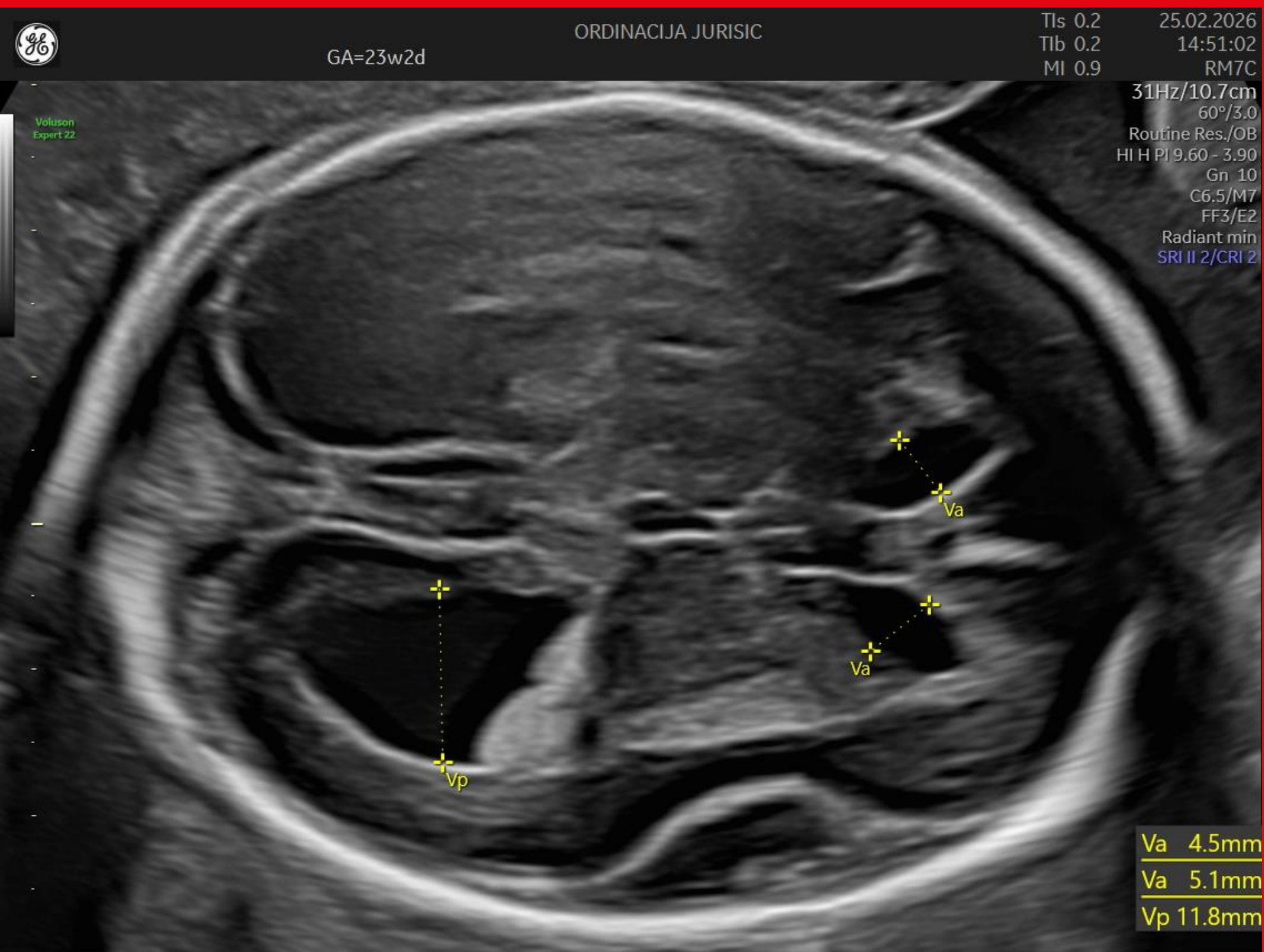


Figure 2. Ventriculomegaly.

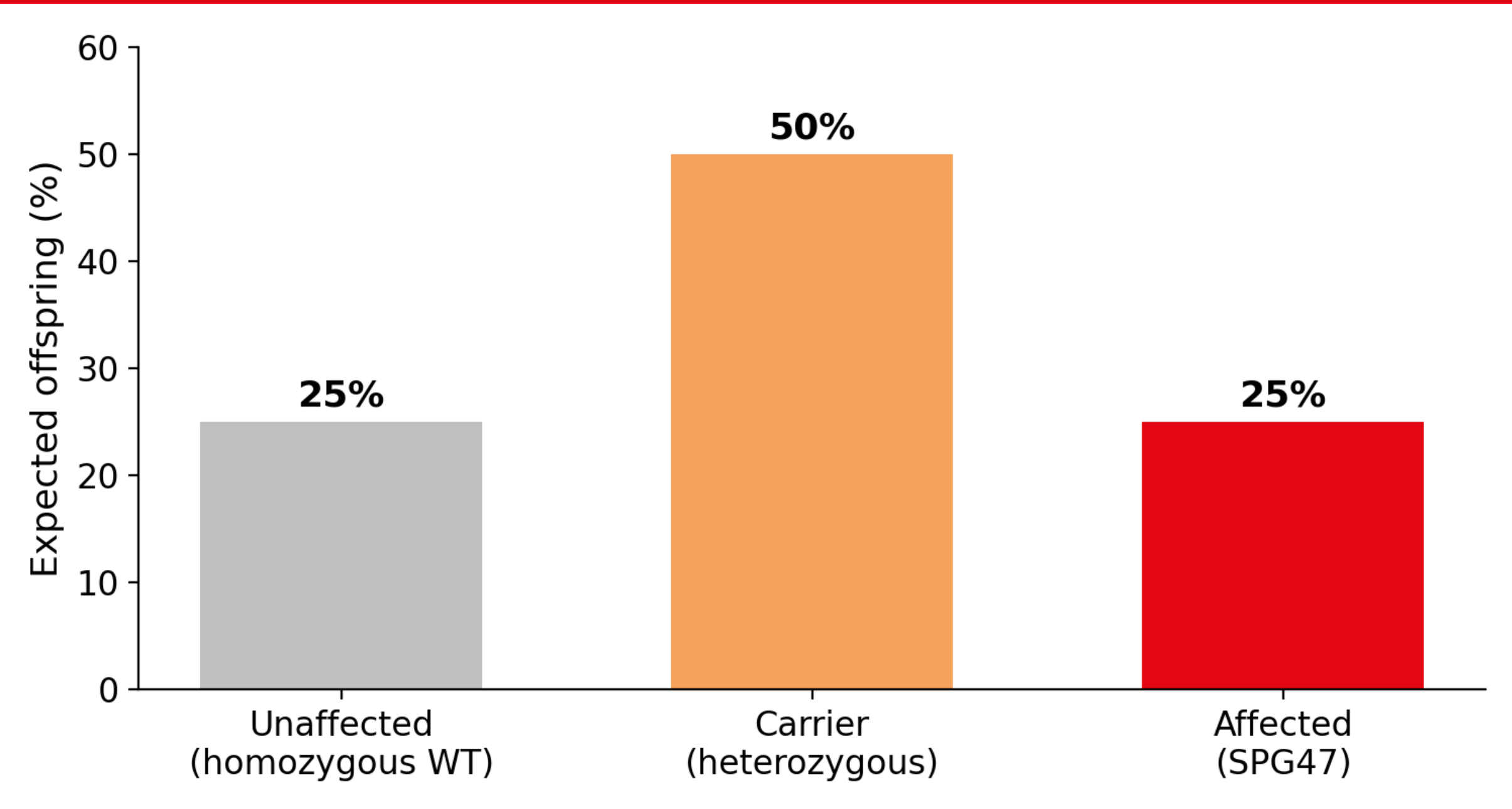


Figure 3. Autosomal-recessive inheritance: 25% recurrence risk.