

Fetal 2:1 Atrioventricular Block Progressing to Ventricular Arrhythmia and Hydrops: A Case of Congenital Long QT Syndrome

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INTRODUCTION

- Fetal arrhythmias complicate 1–3% of pregnancies and carry a risk of significant fetal/neonatal morbidity and mortality.
- Congenital Long QT Syndrome (LQTS) is a rare cause of fetal bradyarrhythmia that can closely mimic immune-mediated AV block.
- We describe a case of fetal 2:1 AV block that progressed to malignant ventricular arrhythmia and hydrops, later confirmed as congenital LQTS.

GENETIC TESTING — ARRHYTHMIA PANEL

Result: UNCERTAIN — 69-gene panel; no pathogenic variant identified

ACTN2 heterozygous · VUS
c.1839+4G>C (intronic)
Assoc. hypertrophic cardiomyopathy, congenital myopathy

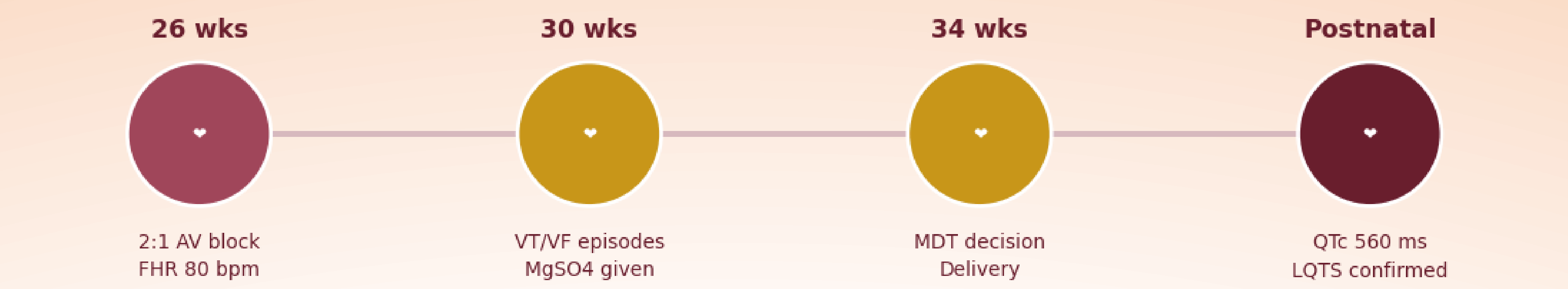
SCN5A heterozygous · VUS
c.2821T>C (p.Ser941Pro)
Assoc. Brugada syndrome, LQTS type 3, dilated cardiomyopathy

SNTA1 heterozygous · VUS
c.272G>T (p.Arg91Leu)
Preliminary evidence — LQTS type 12

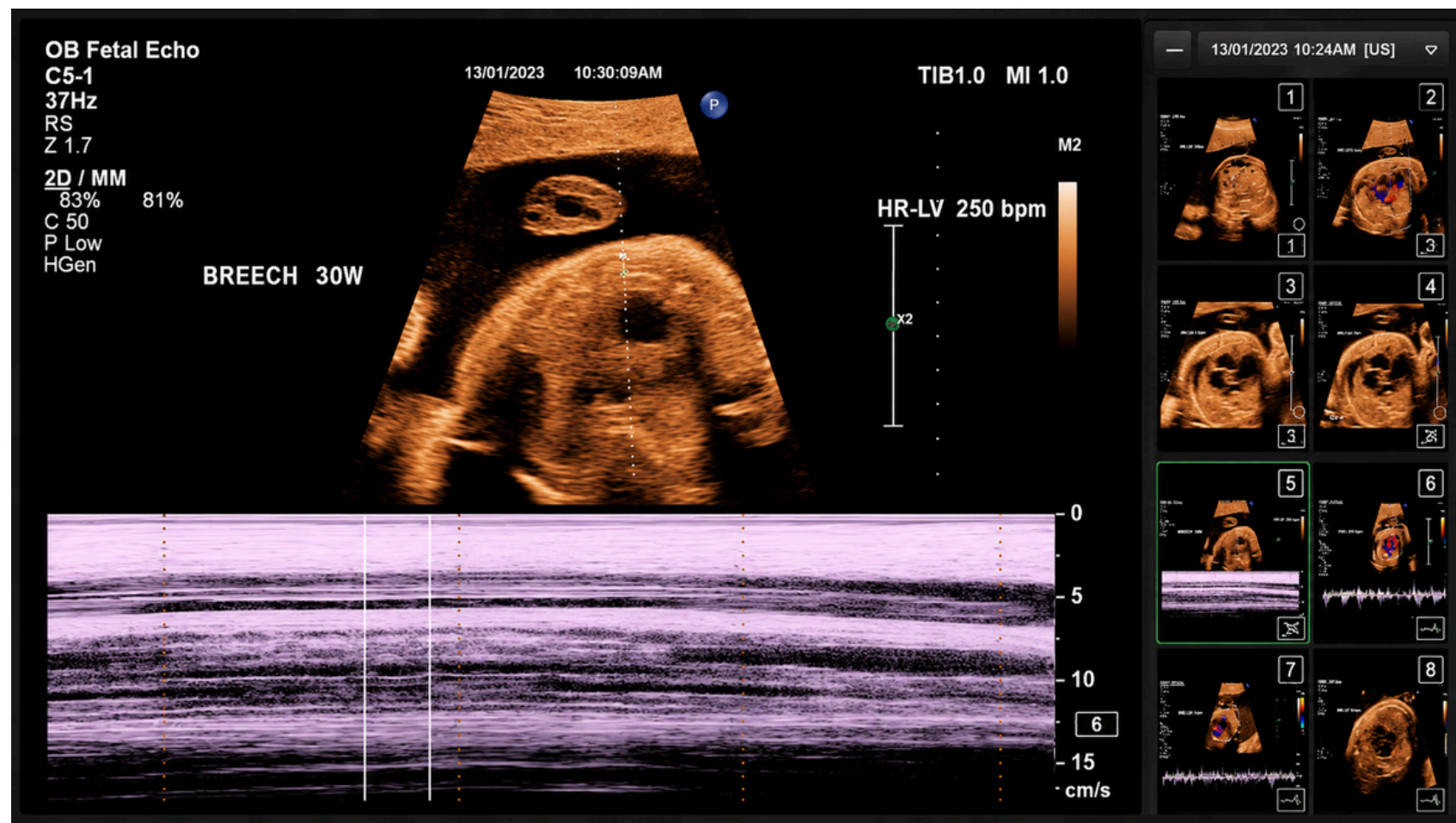
The deceased infant's DNA was tested for a cardiac arrhythmia gene panel consisting of 69 genes. It yielded *SCN5A* heterozygous mutation c.2821T>C (p.Ser941Pro), which was classified pathogenic according to ACMG guideline. This confirmed the diagnosis of autosomal dominant *SCN5A*-related disorder. *SCN5A*-related disorder is associated with variable phenotypes namely long QT syndrome type 3 (LQT3), Brugada syndrome type 1, progressive familial heart block type 1, sick sinus syndrome type 1, isolated atrial fibrillation or ventricular fibrillation, dilated cardiomyopathy type 1E and arrhythmogenic right ventricular cardiomyopathy. For accurate genetic counselling, both parents proceeded to have ECG and *SCN5A* testing, and both were normal.

CASE PRESENTATION

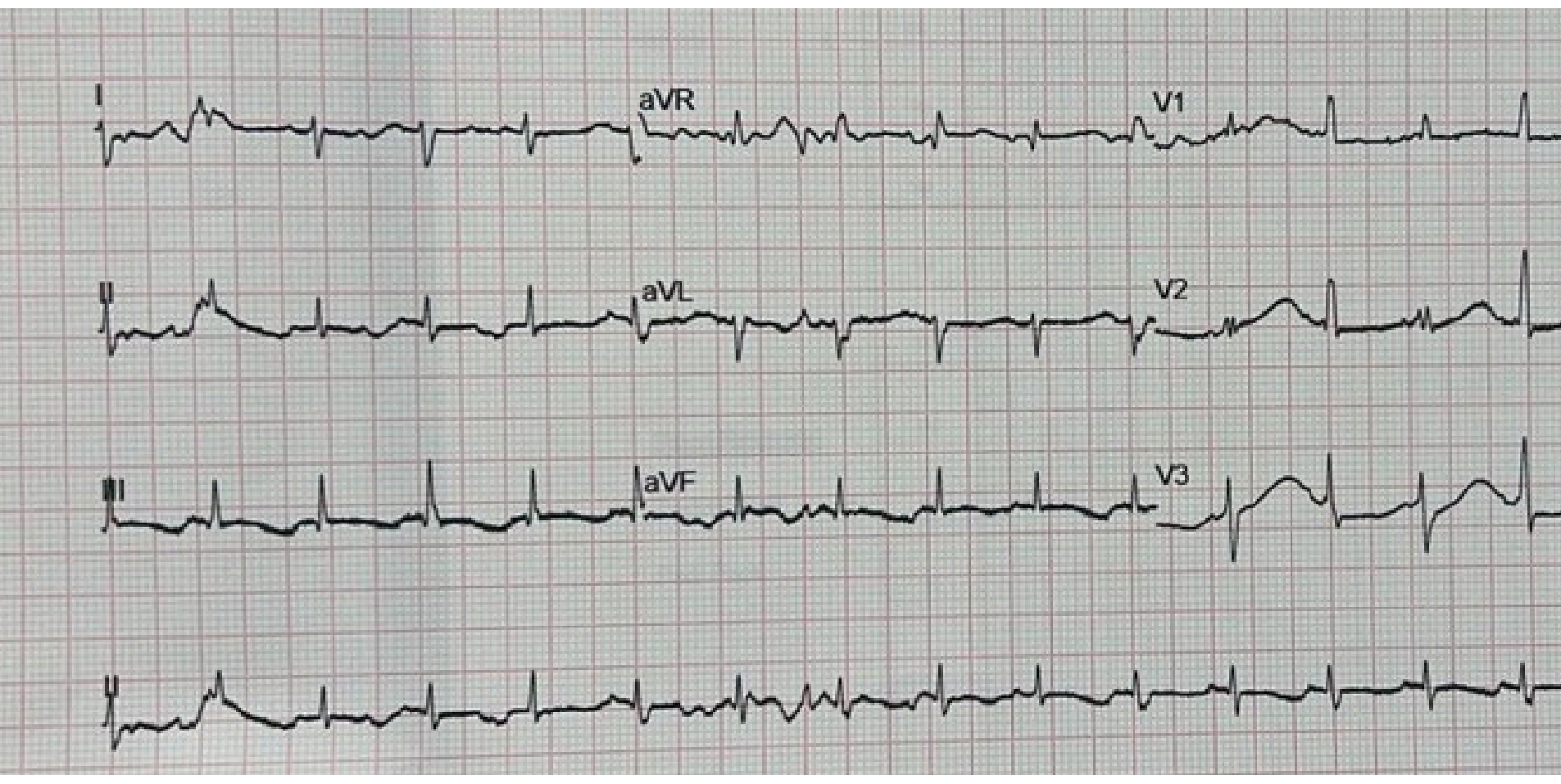
39-year-old G2P1, IVF pregnancy, referred for fetal bradycardia and polyhydramnios. Maternal serology: anti-Ro positive, anti-La negative. Echocardiography revealed a structurally normal heart with persistent 2:1 AV block, cardiomegaly and mild pericardial effusion — immune-mediated block was suspected; IVIG and corticosteroids were started.



Ventricular arrhythmia remained refractory to immune-directed and antiarrhythmic therapy, with progressive deterioration to hydrops fetalis. Postnatal management comprised mechanical ventilation, thoracic drainage, serial cardioversion and antiarrhythmic agents; the neonate nonetheless demised. Dysmorphic features (low-set ears, overlapping fingers, clinodactyly) were recorded, and molecular genetic testing confirmed congenital long QT syndrome.



Fetal echocardiogram, breech, 30 wks (HR-LV 250 bpm)



Postnatal ECG: Calculated QTc : 560 ms

DISCUSSION

- Prolonged ventricular repolarisation in fetal LQTS can produce a functional 2:1 AV block — indistinguishable in utero from immune-mediated block.
- Maternal anti-Ro positivity is a diagnostic confounder that can delay the correct diagnosis and management pathway.
- The key clinical clue is progression from bradycardia to refractory ventricular tachyarrhythmia (VT/VF) despite immune-directed therapy.
- Outcomes for fetal LQTS complicated by malignant arrhythmia and hydrops remain poor despite aggressive antiarrhythmic therapy.

CONCLUSION

- A 2nd-trimester fetal 2:1 AV block — even with positive maternal anti-Ro — is not always immune-mediated.
- Congenital LQTS should be considered when rhythm progresses to ventricular tachyarrhythmia.
- Early suspicion can guide counselling and care planning, though prognosis may remain poor.

1–3%

Pregnancies complicated by fetal arrhythmia

560 ms

Postnatal QTc (markedly prolonged)

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- Samples S, Cherry S, Madan N, Hong J, Mansukhani SA, Strasburger JF, et al. The prenatal diagnosis and perinatal management of congenital long QT syndrome: a comprehensive literature review and recent updates. *J Cardiovasc Dev Dis.* 2025;12(4):156. doi:10.3390/jcdd12040156.