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Unusual Early Prenatal Presentation of Aicardi-Goutieres Syndrome

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Aicardi–Goutieres syndrome (AGS) is a rare genetic disease arising during the first year of life mainly affecting the central nervous system. Fetal AGS mimics in-utero infection and presents with microcephaly and cerebral calcifications.
We report on prenatal myocardial calcifications as the first manifestation of AGS. The parents are consanguineous with two healthy children and one who suffered from severe intellectual disability, epilepsy and diffuse brain calcifications on CT scan. He died at 4 years of age without a definitive diagnosis.
At 21 weeks of the current pregnancy, myocardial calcification with premature atrial beats and pericardial effusion were observed. No calcifications were detected in the brain, liver, spleen, or placenta. Maternal serology for intrauterine infection, ANA, anti-SSA and anti-SSB were negative. Ultrasound at 27 weeks revealed calcifications of the myocardium, caudothalamic groove and liver. At 32 weeks, reduced cardiac contractility, multiple cardiac, thalamic, brainstem and striatal calcifications and a parenchymal temporal lobe cyst were detected.
The couple chose to terminate the pregnancy. Genetic analysis of the amniotic fluid revealed biallelic mutations in TREX1 gene compatible with AGS. Notably, this same mutation was detected in the preserved DNA of the deceased sibling.
AGS is associated with a high risk of recurrence. Since AGS calcifications may mimic in-utero infection, AGS should be considered when TORCH is negative. Cardiac involvement with abnormal contractility, calcifications, effusion and arrhythmia are unusual in AGS but may be the earliest manifestation of AGS due to TREX1 mutation.