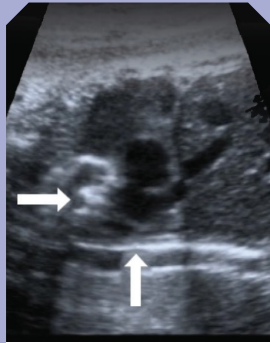
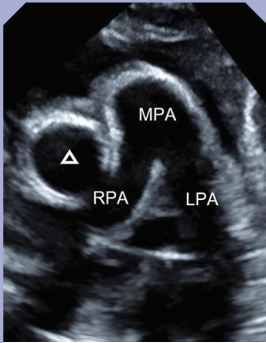


When you see...

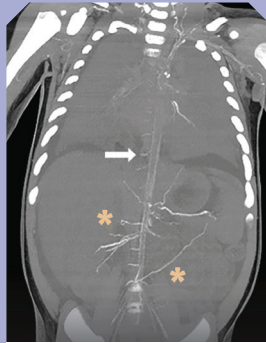
Abnormal Brightness of the Arteries on Prenatal or Postnatal Ultrasound, ECHO, or CT Scan



Aortic calcification on fetal ultrasound¹



Calcification of pulmonary arteries and aortic root on neonatal ECHO²



Aortic (white arrow) and arterial (yellow asterisks) calcification on infant CT scan³

Think...
Generalized
Arterial
Calcification of
Infancy

GACI is a rare mineralization disorder caused by mutations in the *ENPP1* or *ABCC6* genes. It is characterized by pathologic arterial calcification, severe cardiovascular complications, and ~50% mortality within the first 6 months of life.¹⁻¹²

Δ, aortic root; CT, computerized tomography; ECHO, echocardiogram; LPA, proximal left pulmonary artery; MPA, main pulmonary artery; RPA, proximal right pulmonary artery. Neonatal ECHO: Reprinted with permission from the Copyright Clearance Center: Elsevier, Zhang SR, et al. *CASE (Phila)*. 2025;9(8):262-267, ©2025, Elsevier

©2026 BioMarin Pharmaceutical Inc. All rights reserved. MED-ET-0176

GACI Diagnostic Pathway

Common presenting signs/symptoms¹⁻¹²



Fetal ultrasound findings

- Pericardial effusion
- Polyhydramnios
- Hydrops fetalis



Neonatal/infant

- Hypertension
- Heart failure
- Respiratory distress
- Left ventricular hypertrophy
- Heart valve defect

Other complications may include pulmonary, gastrointestinal, and neurologic problems

Indications to test¹⁻¹²

Abnormal brightness

of arteries or heart valves (ultrasound, ECHO, CT, MRI)

OR

Vascular stenosis

(CTA, MRA, ECHO)

OR

Family history

of hypophosphatemic rickets

Genetic testing for *ENPP1* and *ABCC6*



CT, computed tomography; CTA, computed tomography angiography; ECHO, echocardiogram; MRA, magnetic resonance angiography; MRI, magnetic resonance imaging.

References: **1.** Mulcahy CH, et al. *J Congenit Cardiol.* 2019;3(1):1-6. **2.** Zhang SR, et al. *CASE (Phila).* 2025;9(8):262-267. **3.** Boyce AM, et al. *Curr Osteoporos Rep.* 2020;18(3):232-241. **4.** Galletti S, et al. *JIMD Rep.* 2011;1:23-27. **5.** Ferreira CR, et al. *J Bone Miner Res.* 2021;36(11):2193-2202. **6.** Rutsch F, et al. *Circ Cardiovasc Genet.* 2008;1(2):133-140. **7.** Nasrallah FK, et al. *Ultrasound Obstet Gynecol.* 2009;34(5):601-604. **8.** Ziegler SG, et al. In: Adam MP, et al., eds. *GeneReviews*[®] [Internet]. University of Washington, Seattle; 1993-2026. **9.** Ferreira CR, et al. *Genet Med.* 2021;23(2):396-407. **10.** Chong CR, Hutchins GM. *Pediatr Dev Pathol.* 2008;11(5):405-415. **11.** Ramirez-Suarez KI, et al. *Pediatr Radiol.* 2022;52(12):2329-2341. **12.** Ferreira CR, et al. *JBMR Plus.* 2025;9(5):zifa019.