

# WHEN YOU SEE...

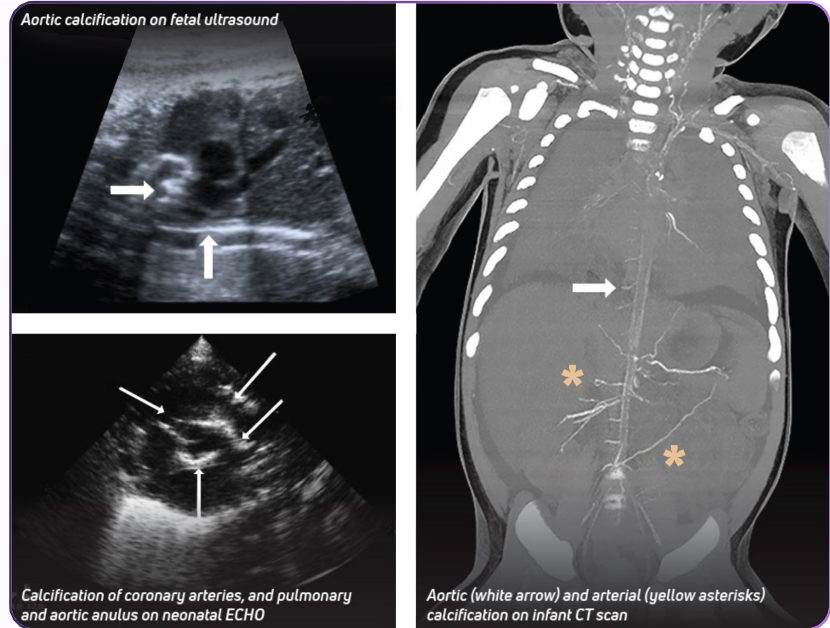
## Abnormal Brightness of the Arteries on Prenatal or Postnatal Ultrasound, ECHO, or CT Scan

### THINK...

**G**eneralized  
**A**rterial  
**C**alcification of  
**I**nfancy



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



CT, computerized tomography; ECHO, echocardiogram.

Neonatal ECHO: Reprinted by permission from the Copyright Clearance Center: Springer Nature, Galletti S, et al. *JIMD Rep.* 2011;1:23-27, © 2011, SSIEM and Springer-Verlag Berlin Heidelberg

**References:** **1.** Mulcahy, C.H., et al. *J Congenit Heart Dis.* 2019; 3(1):1-6. **2.** Galletti S, et al. *JIMD Rep.* 2011;1:23-27. **3.** Boyce AM, et al. *Curr Osteoporos Rep.* 2020;18(3):232-241

**4.** Ferreira CR, et al. *J Bone Miner Res.* 2021;36(11):2193-2202. **5.** Rutsch F, et al. *Circ Cardiovasc Genet.* 2008;1(2):133-140.

# Prenatal/Infant Diagnostic Pathway



## Common presenting signs/symptoms

### Fetal ultrasound findings

- Pericardial effusion
- Cardiomyopathy (cardiac hypertrophy)
- Non-immune hydrops
- Polyhydramnios

### Neonatal/infant

- Cardiomyopathy (cardiac hypertrophy)
- Pulmonary hypertension
- Diminished pulses
- Hypertension
- Respiratory distress
- Heart failure

*Other complications may include seizure, stroke, encephalopathy, pleural effusion, ascites, and gastrointestinal complications.*

## Indications to test

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

OR

**Vascular stenosis**  
(CTA, MRA, ECHO)

OR

**Family history**  
of fetal/neonatal death or hypophosphatemic rickets



**Genetic testing for *ENPP1* and *ABCC6***

ABCC6, adenosine triphosphate binding cassette subfamily C member 6; CT, computed tomography; CTA, computed tomography angiography; ECHO, echocardiogram; ENPP1, ectonucleotide pyrophosphatase/phosphodiesterase 1; MRA, magnetic resonance angiography; MRI, magnetic resonance imaging.

**References:** **1.** Mulcahy CH, et al. *J Congenit Cardiol.* 2019;3(1). doi:10.1186/s40949-018-0022-1 **2.** Nasrallah FK, et al. *Ultrasound Obstet Gynecol.* 2009;34(5):601-604.

**3.** Rutsch F, et al. *Circ Cardiovasc Genet.* 2008;1(2):133-140. **4.** Ferreira CR, et al. In: Adam MP, et al, eds. *GeneReviews® | Internet.* University of Washington, Seattle; 1993-2020.

**5.** Ferreira CR, et al. *J Bone Miner Res.* 2021;36(11):2193-2202. **6.** Ferreira CR, et al. *Genet Med.* 2021;23(2):396-407. **7.** Chong CR, Hutchins GM. *Pediatr Dev Pathol.* 2008;11(5):405-415.

**8.** Ramirez-Suarez Kl, et al. *Pediatr Radiol.* Published online April 19, 2022. doi:10.1007/s00247-022-05364-0