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A Rare Case of Generalized Arterial Calcification of Infancy (GACI) in an Infant Presenting with Respiratory Failure and Arterial Calcification

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Background

- Generalized Arterial Calcification of Infancy (GACI) is a **rare, life-threatening condition** characterized by arterial calcification and narrowing of medium to large-sized vessels¹
- Patients can present in utero with hydrops fetalis, at birth with **respiratory distress** and **pulmonary hypertension**, or within 3 months with sequela of cardiovascular disease including **systemic hypertension** and **heart failure**
- Deposition of hydroxyapatite in the internal elastic lamina with proliferation of fibroblasts in the tunica intima results in **severely reduced arterial elasticity**
- Deficiencies in ENPP1 and ABCC6** comprise 75-85% of cases, demonstrating autosomal recessive pattern. No known genetic cause is found in 15-25% of cases²

Case Report

- A male infant born at 31-weeks secondary to maternal pre-eclampsia presents with **respiratory distress out of proportion to gestational age**.

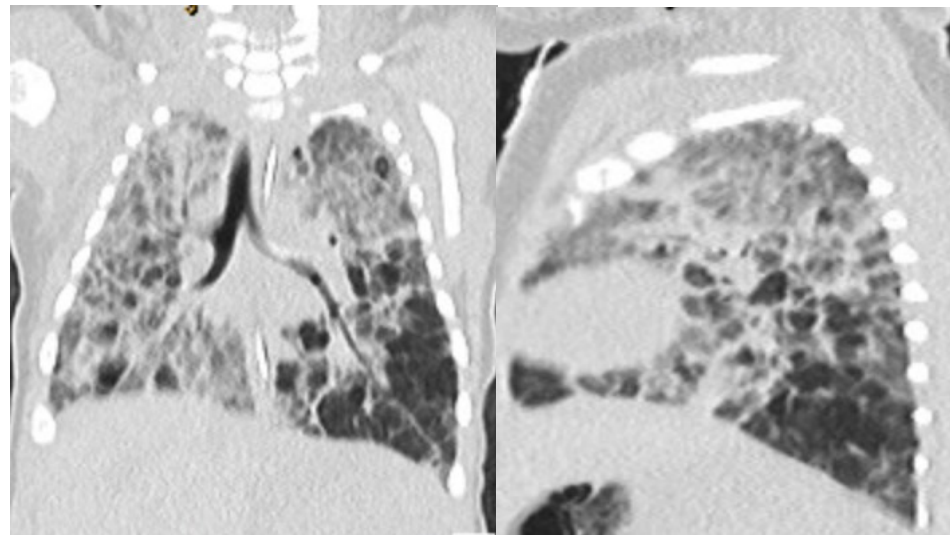


Figure 1. Chest CT with calcific arterial densities, air trapping.

Discussion

- GACI should be considered in infants with signs of **myocardial ischemia** with normal coronary arteries on echocardiogram
- Bisphosphonate therapy has **debatable benefit**
- Mortality is approximately 50%** due to complications of cardiovascular disease; most do not live past 6 months
- Given the inheritance pattern, prenatal **genetic counseling** is recommended for appropriate families

References

- Boyce AM et al. Generalized Arterial Calcification of Infancy: New Insights, Controversies, and Approach to Management. Curr Osteoporos Rep. 2020 Jun;18(3):232-241.
- Ziegler SG et al. Generalized Arterial Calcification of Infancy. GeneReviews; 2020 Dec:1993-2022.

