

# The Clinical Spectrum of ENPP1 and ABCC6 Deficiencies

Rare mineralization disorders with overlapping cardiovascular phenotypes

Thursday, February 8, 2024 | 3:00-4:00 pm  
Manchester Grand Hyatt San Diego  
San Diego, CA, USA

## Speaker



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## Objectives

- Review the critical role of ENPP1 and ABCC6 proteins in the generation of pyrophosphate and prevention of ectopic calcification
- Discuss the clinical phenotypes induced by ENPP1 or ABCC6 Deficiency: Generalized Arterial Calcification of Infancy (GACI), Autosomal Recessive Hypophosphatemic Rickets (ARHR2), and Pseudoxanthoma Elasticum (PXE)
- Appreciate the role of the geneticist in the diagnosis and management of patients with ENPP1 or ABCC6 Deficiency

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