BIOMARIN

ENPP1 Deficiency Selected Publications

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Disease and Diagnosis	Bruno I, De Sanctis L, Foligno NE, et al. Optimizing diagnosis and management of patients with ENPP1 deficiency: An expert opinion. *J Endocrinol Invest.* 2026; in press. DOI: 10.1007/s40618-026-02843-1[a]
	Ferreira CR, Hackbarth ME, Nitschke Y, et al. Phenotypic characterization of ENPP1 deficiency: Generalized arterial calcification of infancy and autosomal recessive hypophosphatemic rickets type 2. *JBMR Plus.* 2025;9(5):ziaf019. bDOI: 10.1093/jbmrpl/ziaf019
	Collins L, Sandy J, Ly S, et al. Six cases of *ENPP1* pathogenic variants causing autosomal recessive hypophosphatemic rickets type 2 and generalized arterial calcification of infancy. *JBMR Plus.* 2025;9(S5):v47□v57. DOI: 10.1093/jbmrpl/ziae174
	Dursun F, Turan İ, Bitkin EÇ, et al. Natural history of ENPP1 deficiency: Nationwide Turkish cohort study of autosomal-recessive hypophosphataemic rickets type 2. *Clin Endocrinol.* 2024;101(5):475□484. DOI: 10.1111/cen.15028[a]
	Ferreira CR, Ansh AJ, Nester C, et al. Musculoskeletal comorbidities and quality of life in ENPP1-deficient adults and the response of enthesopathy to enzyme replacement therapy in murine models. *JBMR.* 2022;37(3):494□504. DOI: 10.1002/jbmr.4487
	Ferreira CR, Hackbarth ME, Ziegler SG, et al. Prospective phenotyping of long-term survivors of generalized arterial calcification of infancy (GACI). *Genet Med.* 2021;23:396–407. DOI: 10.1038/s41436-020-00983-0
	Höppner J, Kowak U, Sinningen K, et al. Autosomal recessive hypophosphatemic rickets type 2 (ARHR2) due to ENPP1-deficiency. *Bone.* 2021;153:116111. DOI: 10.1016/j.bone.2021.116111
	Ferreira CR, Kintzinger K, Hackbarth ME, et al. Ectopic calcification and hypophosphatemic rickets: Natural history of ENPP1 and ABCC6 deficiencies. *JBMR.* 2021;36(11):2193–2202. DOI: 10.1002/jbmr.4418
Preclinical	Cheng Z, O'Brien K, Howe J, et al. INZ-701 prevents ectopic tissue calcification and restores bone architecture and growth in ENPP1-deficient mice. *JBMR Plus.* 2021;36:1594–1604. DOI: 10.1002/jbmr.4315[a]
	Ferreira CR, Kavanagh D, Oheim R, et al. Response of the ENPP1-deficient skeletal phenotype to oral phosphate supplementation and/or enzyme replacement therapy: Comparative studies in humans and mice. *JBMR.* 2021;36(5):942–955. DOI: 10.1002/jbmr.4254
	Oheim R, Zimmerman K, Maulding ND, et al. Human heterozygous ENPP1 deficiency is associated with early onset osteoporosis, a phenotype recapitulated in a mouse model of Enpp1 deficiency. *JBMR.* 2020;35(3):528–539. DOI: 10.1002/jbmr.3911
Management	Munteanu M, Rutsch F, Nitschke Y, et al. Autosomal recessive hypophosphatemic rickets type 2 (ARHR2): Is phosphate supplementation safe? *Bone.* 2026;202:117698. DOI: 10.1016/j.bone.2025.117698
	Rutsch F, Boÿer P, Nitschke Y, et al. Hypophosphatemia, hyperphosphaturia, and bisphosphonate treatment are associated with survival beyond infancy in generalized arterial calcification of infancy. *Circ Cardiovasc Genet.* 2008;1(2):133–140. DOI: 10.1161/circgenetics.108.797704
Other	Ferreira CR, Carpenter TO, Braddock DT. ENPP1 in blood and bone: Skeletal and soft tissue diseases induced by ENPP1 deficiency. *Annu Rev Path Mech Dis.* 2024;19:507□540. DOI: 10.1146/annurev-pathmechdis-051222-121126
	Chunn LM, Bissonnette J, Heinrich SV, et al. Estimation of ENPP1 deficiency genetic prevalence using a comprehensive literature review and population databases. *Orphanet J Rare Dis.* 2022;17:421. DOI: 10.1186/s13023-022-02577-2
	O'Brien C, Khursigara G, Huertas P, et al. Lifelong impact of ENPP1 deficiency and the early onset form of ABCC6 deficiency from patient or caregiver perspective. *PLoS One.* 2022;17(7):e0270632. DOI: 10.1371/journal.pone.0270632

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