

CITATION REPORT

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Generalized arterial calcification of infancy and pseudoxanthoma elasticum can be caused by mutations in either ENPP1 or ABCC6

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#	Paper	IF	Citations
254	Heritable ectopic mineralization disorders: the paradigm of pseudoxanthoma elasticum. 2012 , 132, E15-9		9
253	Histopathology of pseudoxanthoma elasticum and related disorders: histological hallmarks and diagnostic clues. 2012 , 2012, 598262		32
252	P2Y purinoceptors as potential emerging therapeutical target in vascular disease. 2012 , 18, 6169-80		14
251	Hypertension and vascular calcification: a vicious cycle?. 2012 , 30, 1885-93		39
250	Structure of NPP1, an ectonucleotide pyrophosphatase/phosphodiesterase involved in tissue calcification. 2012 , 20, 1948-59		53
249	Two new cases of idiopathic arterial calcification in the newborn: watch out for lineal calcifications in plain radiographs. 2012 , 161, 767-767.e1		2
248	New insights into NPP1 function: lessons from clinical and animal studies. 2012 , 51, 961-8		65
247	Genetic pathways of vascular calcification. 2012 , 22, 93-8		59
246	Genetics in arterial calcification: lessons learned from rare diseases. 2012 , 22, 145-9		44
245	A novel animal model for pseudoxanthoma elasticum: the KK/HIJ mouse. 2012 , 181, 1190-6		19
244	The molecular and physiological roles of ABCC6: more than meets the eye. <i>Frontiers in Genetics</i> , 2012 , 3, 289	4.5	47
243	Generalized arterial calcification of infancy and pseudoxanthoma elasticum: two sides of the same coin. <i>Frontiers in Genetics</i> , 2012 , 3, 302	4.5	49
242	Hypophosphatemia and growth. 2013 , 28, 595-603		42
241	Cole Disease Results from Mutations in ENPP1. <i>American Journal of Human Genetics</i> , 2013 , 93, 752-7	11	28
240	Genetic mapping and exome sequencing identify 2 mutations associated with stroke protection in pediatric patients with sickle cell anemia. 2013 , 121, 3237-45		44
239	The mystery of persistent pulmonary hypertension: an idiopathic infantile arterial calcification. 2013 , 13, 107		7
238	Cardiovascular Calcifications in Old Age: Mechanisms and Clinical Implications. 2013 , 2, 255-267		7

237	Pseudoxanthoma elasticum: progress in research toward treatment: summary of the 2012 PXE international research meeting. 2013 , 133, 1444-9		40
236	Pseudoxanthoma elasticum: the paradigm of ectopic mineralization disorders [diagnosis and treatment. 2013 , 8, 257-266		
235	Is classical pseudoxanthoma elasticum a consequence of hepatic intoxication due to ABCC6 substrate accumulation in the liver?. 2013 , 8, 37-46		2
234	Severe skeletal toxicity from protracted etidronate therapy for generalized arterial calcification of infancy. <i>Journal of Bone and Mineral Research</i> , 2013 , 28, 419-30	6.3	66
233	Warfarin accelerates ectopic mineralization in Abcc6(-/-) mice: clinical relevance to pseudoxanthoma elasticum. 2013 , 182, 1139-50		16
232	Mineralization/anti-mineralization networks in the skin and vascular connective tissues. 2013 , 183, 10-8		48
231	New findings in genodermatoses. 2013 , 31, 303-15		0
230	Insights from genetic disorders of phosphate homeostasis. 2013 , 33, 143-57		29
229	Clinical phenotypes and ABCC6 gene mutations in Brazilian families with pseudoxanthoma elasticum. 2013 , 93, 739-40		2
228	Treatment of choroidal neovascularization due to angioid streaks: a comprehensive review. 2013 , 33, 1300-14		62
227	Paediatric pseudoxanthoma elasticum with cardiovascular involvement. 2013 , 169, 1148-51		7
226	Progressive extreme heterotopic calcification. 2013 , 161A, 1706-13		2
225	ABCC6 prevents ectopic mineralization seen in pseudoxanthoma elasticum by inducing cellular nucleotide release. 2013 , 110, 20206-11		171
224	Pseudoxanthoma elasticum: cardiac findings in patients and Abcc6-deficient mouse model. <i>PLoS ONE</i> , 2013 , 8, e68700	3.7	21
223	The vascular phenotype in Pseudoxanthoma elasticum and related disorders: contribution of a genetic disease to the understanding of vascular calcification. <i>Frontiers in Genetics</i> , 2013 , 4, 4	4.5	63
222	Fibroblast involvement in soft connective tissue calcification. <i>Frontiers in Genetics</i> , 2013 , 4, 22	4.5	38
221	Transcriptional regulation of the ABCC6 gene and the background of impaired function of missense disease-causing mutations. <i>Frontiers in Genetics</i> , 2013 , 4, 27	4.5	12
220	New insights into the pathogenesis of pseudoxanthoma elasticum and related soft tissue calcification disorders by identifying genetic interactions and modifiers. <i>Frontiers in Genetics</i> , 2013 , 4, 114	4.5	15

219	The ABCC6 transporter: what lessons can be learnt from other ATP-binding cassette transporters?. <i>Frontiers in Genetics</i> , 2013 , 4, 203	4.5	8
218	Coordinated orphan disease research: yes, we can!. <i>Frontiers in Genetics</i> , 2013 , 4, 207	4.5	
217	Mouse models for pseudoxanthoma elasticum: genetic and dietary modulation of the ectopic mineralization phenotypes. <i>PLoS ONE</i> , 2014 , 9, e89268	3.7	15
216	The contribution of arterial calcification to peripheral arterial disease in pseudoxanthoma elasticum. <i>PLoS ONE</i> , 2014 , 9, e96003	3.7	16
215	Heart transplant and 2-year follow up in a child with generalized arterial calcification of infancy. 2014 , 173, 1735-40		7
214	Physiology of the Developing Kidney: Disorders and Therapy of Calcium and Phosphorous Homeostasis. 2014 , 1-59		
213	Analysis of pseudoxanthoma elasticum-causing missense mutants of ABCC6 in vivo; pharmacological correction of the mislocalized proteins. 2014 , 134, 946-953		39
212	Disseminated arterial calcification and enhanced myogenic response are associated with abcc6 deficiency in a mouse model of pseudoxanthoma elasticum. 2014 , 34, 1045-56		20
211	Mineralisation of collagen rich soft tissues and osteocyte lacunae in Enpp1(-/-) mice. 2014 , 69, 139-47		40
210	Zebrafish enpp1 mutants exhibit pathological mineralization, mimicking features of generalized arterial calcification of infancy (GACI) and pseudoxanthoma elasticum (PXE). 2014 , 7, 811-22		40
209	Mono-allelic and bi-allelic ENPP1 deficiency promote post-injury neointimal hyperplasia associated with increased C/EBP homologous protein expression. 2014 , 233, 493-502		8
208	Mutations in the ABCC6 gene as a cause of generalized arterial calcification of infancy: genotypic overlap with pseudoxanthoma elasticum. 2014 , 134, 658-665		58
207	Molecular Biology of Valvular Heart Disease. 2014 ,		0
206	Hearing loss is part of the clinical picture of ENPP1 loss of function mutation. 2014 , 81, 63-6		17
205	Dysregulation of gene expression in ABCC6 knockdown HepG2 cells. 2014 , 19, 517-26		25
204	Genetic modulation of nephrocalcinosis in mouse models of ectopic mineralization: the Abcc6(tm1Jfk) and Enpp1(asj) mutant mice. 2014 , 94, 623-32		16
203	Inflammatory, metabolic, and genetic mechanisms of vascular calcification. 2014 , 34, 715-23		220
202	Bleeding and non-bleeding phenotypes in patients with GGCX gene mutations. 2014 , 134, 856-65		24

201	Polymicrogyria: pathology, fetal origins and mechanisms. 2014 , 2, 80		68
200	Perturbation of specific pro-mineralizing signalling pathways in human and murine pseudoxanthoma elasticum. <i>Orphanet Journal of Rare Diseases</i> , 2014 , 9, 66	4.2	31
199	Variants in genes encoding pyrophosphate metabolizing enzymes are associated with Pseudoxanthoma elasticum. 2014 , 47, 60-7		11
198	Ectopic mineralization disorders of the extracellular matrix of connective tissue: molecular genetics and pathomechanisms of aberrant calcification. 2014 , 33, 23-8		46
197	[GACI syndrome: a case report with a neonatal beginning]. 2014 , 21, 632-6		8
196	The level of hepatic ABCC6 expression determines the severity of calcification after cardiac injury. 2014 , 184, 159-70		21
195	PSEUDOXANTHOMA ELASTICUM: DIAGNOSTIC FEATURES, CLASSIFICATION, AND TREATMENT OPTIONS. 2014 , 2, 567-577		49
194	Pyrophosphates as a major inhibitor of matrix calcification in Pseudoxanthoma elasticum. 2014 , 75, 109-20		27
193	Prenatal ultrasonographic diagnosis of generalized arterial calcification of infancy. 2015 , 43, 50-4		7
192	Pseudoxanthoma elasticum. 2015 , 132, 215-21		13
191	Calcinosis: pathophysiology and management. 2015 , 27, 542-8		60
190	Cerebral Vasculopathies. 2015 , 1211-1222		1
189	Pseudoxanthoma elasticum and skin: Clinical manifestations, histopathology, pathomechanism, perspectives of treatment. <i>Intractable and Rare Diseases Research</i> , 2015 , 4, 113-22	1.4	38
188	The Genetics of Soft Connective Tissue Disorders. 2015 , 16, 229-55		41
187	Pathological calcification and the mystery of Lot's wife. 2015 , 14, 3354-5		
186	ENPP1-Fc prevents mortality and vascular calcifications in rodent model of generalized arterial calcification of infancy. 2015 , 6, 10006		71
185	Vitamin K reduces hypermineralisation in zebrafish models of PXE and GACI. 2015 , 142, 1095-101		33
184	Efficiency of exome sequencing for the molecular diagnosis of pseudoxanthoma elasticum. 2015 , 135, 992-998		20

183	Genetic heterogeneity of pseudoxanthoma elasticum: the Chinese signature profile of ABCC6 and ENPP1 mutations. 2015 , 135, 1294-1302	17
182	Antenatal diagnosis of idiopathic arterial calcification: a systematic review with a report of two cases. 2015 , 291, 977-86	5
181	A genetic component of stroke in sickle-cell disease. 2015 , 27, 176-177	
180	Mineral content of the maternal diet influences ectopic mineralization in offspring of Abcc6(-/-) mice. 2015 , 14, 3184-9	12
179	Calcification of joints and arteries: second report with novel NT5E mutations and expansion of the phenotype. 2015 , 60, 561-4	21
178	Early onset hearing loss in autosomal recessive hypophosphatemic rickets caused by loss of function mutation in ENPP1. 2015 , 28, 967-70	8
177	The effects of bisphosphonates on ectopic soft tissue mineralization caused by mutations in the ABCC6 gene. 2015 , 14, 1082-9	44
176	Basic data underlying decision making in nonatherosclerotic causes of intermittent claudication. 2015 , 29, 138-53	13
175	Vascular calcification: Mechanisms of vascular smooth muscle cell calcification. 2015 , 25, 267-74	237
174	Treatment of hypophosphatemic rickets in generalized arterial calcification of infancy (GACI) without worsening of vascular calcification. 2016 , 170A, 1308-11	16
173	Bidirectional Translation in Cardiovascular Calcification. 2016 , 36, e19-24	5
172	Auricular ossification: A newly recognized feature of osteoprotegerin-deficiency juvenile Paget disease. 2016 , 170A, 978-85	10
171	Increased activity of TNAP compensates for reduced adenosine production and promotes ectopic calcification in the genetic disease ACDC. 2016 , 9, ra121	47
170	Diseases of Small and Medium-sized Blood Vessels. 2016 , 125-168	1
169	Pyrophosphate: a key inhibitor of mineralisation. 2016 , 28, 57-68	81
168	Antenatal manifestations of inborn errors of metabolism: autopsy findings suggestive of a metabolic disorder. 2016 , 39, 597-610	9
167	Arterial calcification due to CD73 deficiency (ACDC): imaging manifestations of ectopic mineralization. 2016 , 45, 1583-7	7
166	Effects of Different Variants in the ENPP1 Gene on the Functional Properties of Ectonucleotide Pyrophosphatase/Phosphodiesterase Family Member 1. <i>Human Mutation</i> , 2016 , 37, 1190-1201	4-7 24

165	Large animal models of cardiovascular disease. 2016 , 34, 113-32	67
164	Prenatal Diagnosis of Idiopathic Infantile Arterial Calcification. 2016 , 3, 143-146	0
163	New perspectives on rare connective tissue calcifying diseases. 2016 , 28, 14-23	17
162	Association of Cole disease with novel heterozygous mutations in the somatomedin-B domains of the ENPP1 gene: necessary, but not always sufficient. 2016 , 174, 1152-6	6
161	ABC Transporters - 40 Years on. 2016 ,	10
160	[Pseudoxanthoma elasticum-like disease with deficiency of vitamin K-dependent clotting factors and cutis laxa features]. 2016 , 143, 279-83	3
159	Research Progress in Pseudoxanthoma Elasticum and Related Ectopic Mineralization Disorders. 2016 , 136, 550-556	26
158	Two Liver Transporters, ABCB11 and ABCC6; Novel Therapeutic Approaches in the Related Disorders. 2016 , 357-376	
157	Dual Effects of Bisphosphonates on Ectopic Skin and Vascular Soft Tissue Mineralization versus Bone Microarchitecture in a Mouse Model of Generalized Arterial Calcification of Infancy. 2016 , 136, 275-283	31
156	Generalized Arterial Calcification in a Recipient Twin: Discordant Fetal Hemodynamics Result in Differing Phenotypes in Monozygotic Twins with an ABCC6 Mutation. 2017 , 41, 234-236	7
155	Cerebral disease in a nationwide Dutch pseudoxanthoma elasticum cohort with a systematic review of the literature. 2017 , 373, 167-172	14
154	Condylar geometry variation is associated with ENPP1 variant in a population of patients with dento-facial deformities. 2017 , 45, 826-830	7
153	Pyrophosphate Supplementation Prevents Chronic and Acute Calcification in ABCC6-Deficient Mice. 2017 , 187, 1258-1272	40
152	Inherited Arterial Calcification Syndromes: Etiologies and Treatment Concepts. 2017 , 15, 255-270	37
151	Ectopic calcification in pseudoxanthoma elasticum responds to inhibition of tissue-nonspecific alkaline phosphatase. 2017 , 9,	63
150	Insights into Pathomechanisms and Treatment Development in Heritable Ectopic Mineralization Disorders: Summary of the PXE International Biennial Research Symposium-2016. 2017 , 137, 790-795	25
149	Prevalence and severity of arterial calcifications in pseudoxanthoma elasticum (PXE) compared to hospital controls. Novel insights into the vascular phenotype of PXE. 2017 , 256, 7-14	21
148	Gene2DisCo: Gene to disease using disease commonalities. 2017 , 82, 34-46	6

147	Plasma PPI Deficiency Is the Major, but Not the Exclusive, Cause of Ectopic Mineralization in an Abcc6 Mouse Model of PXE. 2017 , 137, 2336-2343	33
146	knockdown in HepG2 cells induces a senescent-like cell phenotype. 2017 , 22, 7	21
145	Pseudoxanthoma elasticum. <i>Orphanet Journal of Rare Diseases</i> , 2017 , 12, 85	4.2 72
144	Functional Rescue of ABCC6 Deficiency by 4-Phenylbutyrate Therapy Reduces Dystrophic Calcification in Abcc6 Mice. 2017 , 137, 595-602	29
143	ABC Transport Proteins in Cardiovascular Disease-A Brief Summary. 2017 , 22,	49
142	Finding the culprit: who is turning hearts to stone?. 2017 , 4, 33	1
141	Clinical utility gene card: for pseudoxanthoma elasticum. 2018 , 26, 919-924	1
140	Etidronate for Prevention of Ectopic Mineralization in Patients With Pseudoxanthoma Elasticum. 2018 , 71, 1117-1126	63
139	Persistence of the ABCC6 genes and the emergence of the bony skeleton in vertebrates. 2018 , 8, 6027	7
138	Alteration of Extracellular Nucleotide Metabolism in Pseudoxanthoma Elasticum. 2018 , 138, 1862-1870	19
137	Variant discovery in patients with Mendelian vascular anomalies by next-generation sequencing and their use in patient clinical management. 2018 , 67, 922-932.e11	10
136	Association of arterial calcification with chronic limb ischemia in patients with peripheral artery disease. 2018 , 67, 507-513	23
135	A targeted sequencing approach to find novel pathogenic genes associated with sporadic aortic dissection. 2018 , 61, 1545-1553	10
134	Bisphosphonate therapy in an infant with generalized arterial calcification with an ABCC6 mutation. 2018 , 29, 2575-2579	5
133	Coronary pathology of inherited generalized arterial calcification of infancy: a case report. 2018 , 36, 15-19	3
132	Disorders and Mechanisms of Ectopic Calcification. 2018 , 571-595	1
131	Generalized arterial calcification of infancy with a novel ENPP1 mutation: a case report. 2018 , 18, 217	5
130	inhibits ectopic joint calcification and maintains articular chondrocytes by repressing hedgehog signaling. 2018 , 145,	10

129	Genetic analysis of three families with X-linked dominant hypophosphatemic rickets. 2018 , 31, 789-797		6
128	The prevalence of pseudoxanthoma elasticum: Revised estimations based on genotyping in a high vascular risk cohort. 2019 , 62, 90-92		14
127	Molecular characterization of known and novel ACVR1 variants in phenotypes of aberrant ossification. 2019 , 179, 1764-1777		8
126	Quantitative Trait Locus and Integrative Genomics Revealed Candidate Modifier Genes for Ectopic Mineralization in Mouse Models of Pseudoxanthoma Elasticum. 2019 , 139, 2447-2457.e7		12
125	Basic molecular mechanism of vascular calcification. 2019 , 47-82		
124	Novel homozygous ENPP1 mutation causes generalized arterial calcifications of infancy, thrombocytopenia, and cardiovascular and central nervous system syndrome. 2019 , 179, 2112-2118		10
123	Dietary Pyrophosphate Modulates Calcification in a Mouse Model of Pseudoxanthoma Elasticum: Implication for Treatment of Patients. 2019 , 139, 1082-1088		17
122	Potential Role of H-Ferritin in Mitigating Valvular Mineralization. 2019 , 39, 413-431		13
121	Pseudoxanthoma elasticum with prominent arterial calcifications evoking CD73 deficiency. 2019 , 24, 461-464		6
120	The Role of Vitamin K and Its Related Compounds in Mendelian and Acquired Ectopic Mineralization Disorders. <i>International Journal of Molecular Sciences</i> , 2019 , 20,	6.3	4
119	Understanding the basis of Ehlers-Danlos syndrome in the era of the next-generation sequencing. 2019 , 311, 265-275		6
118	Adenovirus-Mediated ABCC6 Gene Therapy for Heritable Ectopic Mineralization Disorders. 2019 , 139, 1254-1263		11
117	Pseudoxanthoma Elasticum and Cutis Laxa. 2019 , 1125-1138		
116	The Parathyroid and Disorders of Calcium and Bone Metabolism. 2019 , 409-479		0
115	Pseudoxanthoma Elasticum, Kidney Stones and Pyrophosphate: From a Rare Disease to Urolithiasis and Vascular Calcifications. <i>International Journal of Molecular Sciences</i> , 2019 , 20,	6.3	10
114	Pseudoxanthoma Elasticum as a Paradigm of Heritable Ectopic Mineralization Disorders: Pathomechanisms and Treatment Development. 2019 , 189, 216-225		37
113	Inhibition of Tissue-Nonspecific Alkaline Phosphatase Attenuates Ectopic Mineralization in the Abcc6 Mouse Model of PXE but Not in the Enpp1 Mutant Mouse Models of GACI. 2019 , 139, 360-368		31
112	Cellular signaling in pseudoxanthoma elasticum: an update. 2019 , 55, 119-129		18

111	Investigational Pharmacological Treatments for Vascular Calcification. 2019 , 2, 1800094		20
110	Zebrafish and medaka as models for biomedical research of bone diseases. 2020 , 457, 191-205		25
109	Human Heterozygous ENPP1 Deficiency Is Associated With Early Onset Osteoporosis, a Phenotype Recapitulated in a Mouse Model of Enpp1 Deficiency. <i>Journal of Bone and Mineral Research</i> , 2020 , 35, 528-539	6.3	18
108	Diseases of the circulatory system. 2020 , 327-390		0
107	Clinical and Biochemical Phenotypes in a Family With ENPP1 Mutations. <i>Journal of Bone and Mineral Research</i> , 2020 , 35, 662-670	6.3	21
106	Phylogeny and chemistry of biological mineral transport. 2020 , 141, 115621		3
105	Osteoarthritis in Pseudoxanthoma Elasticum Patients: An Explorative Imaging Study. <i>Journal of Clinical Medicine</i> , 2020 , 9,	5.1	1
104	Analyses of key genes involved in Arctic adaptation in polar bears suggest selection on both standing variation and de novo mutations played an important role. 2020 , 21, 543		
103	Evaluation of sulfonate and sulfamate derivatives possessing benzofuran or benzothiophene nucleus as inhibitors of nucleotide pyrophosphatases/phosphodiesterases and anticancer agents. 2020 , 104, 104305		2
102	Hereditary Disorders of Cardiovascular Calcification. 2021 , 41, 35-47		5
101	From membrane to mineralization: the curious case of the ABCC6 transporter. 2020 , 594, 4109-4133		2
100	The Role of Sclerostin in Bone and Ectopic Calcification. <i>International Journal of Molecular Sciences</i> , 2020 , 21,	6.3	14
99	Generalized Arterial Calcification of Infancy: New Insights, Controversies, and Approach to Management. 2020 , 18, 232-241		11
98	Arterial Stiffness: A Focus on Vascular Calcification and Its Link to Bone Mineralization. 2020 , 40, 1078-1093		40
97	The biology of vascular calcification. 2020 , 354, 261-353		6
96	Skeletal abnormalities secondary to antenatal etidronate treatment for suspected generalised arterial calcification of infancy. 2020 , 12, 100280		2
95	Rare Co-occurrence of Beta-Thalassemia and : Novel Biomolecular Findings. 2019 , 6, 322		17
94	Extracellular pyrophosphate: The body's "water softener". 2020 , 134, 115243		12

93	Creation of the first monoclonal antibody recognizing an extracellular epitope of hABCC6. 2021 , 595, 789-798		1
92	Prospective phenotyping of long-term survivors of generalized arterial calcification of infancy (GACI). 2021 , 23, 396-407		14
91	Genetic pathways disrupted by ENPP1 deficiency provide insight into mechanisms of osteoporosis, osteomalacia, and paradoxical mineralization. 2021 , 142, 115656		4
90	A systematic review of monogenic etiologies of nonimmune hydrops fetalis. 2021 , 23, 3-12		11
89	Adaptive Optics Imaging in Patients Affected by Pseudoxanthoma Elasticum. 2021 , 224, 84-95		4
88	Neonatal myocardial ischemia and calcifications. Report of a case of generalized arterial calcification of infancy. 2021 , 74, 187-189		0
87	Molecular Genetics and Modifier Genes in Pseudoxanthoma Elasticum, a Heritable Multisystem Ectopic Mineralization Disorder. 2021 , 141, 1148-1156		10
86	Pseudoxanthoma elasticum overlaps hereditary spastic paraplegia type 56. 2021 , 289, 709-725		1
85	Reassessment of causality of ABCC6 missense variants associated with pseudoxanthoma elasticum based on Sherlock. 2021 , 23, 131-139		8
84	An update on vascular calcification and potential therapeutics. 2021 , 48, 887-896		11
83	A phytic acid analogue INS-3001 prevents ectopic calcification in an Abcc6 mouse model of pseudoxanthoma elasticum. <i>Experimental Dermatology</i> , 2021 , 30, 853-858	4	1
82	Isquemia miocárdica neonatal y calcificaciones, presentación de un caso de calcificación arterial generalizada de la infancia. 2021 , 74, 187-189		0
81	Dermal Alterations in Clinically Unaffected Skin of Patients. <i>Journal of Clinical Medicine</i> , 2021 , 10,	5.1	2
80	Response of the ENPP1-Deficient Skeletal Phenotype to Oral Phosphate Supplementation and/or Enzyme Replacement Therapy: Comparative Studies in Humans and Mice. <i>Journal of Bone and Mineral Research</i> , 2021 , 36, 942-955	6.3	2
79	ABCC6 deficiency promotes dyslipidemia and atherosclerosis. 2021 , 11, 3881		8
78	A New Zebrafish Model for Pseudoxanthoma Elasticum. 2021 , 9, 628699		1
77	Experience with the targeted next-generation sequencing in the diagnosis of hereditary hypophosphatemic rickets. 2021 , 34, 639-648		0
76	Comprehensive validation of a diagnostic strategy for sequencing genes with one or multiple pseudogenes using pseudoxanthoma elasticum as a model. 2021 , 48, 289-299		0

75	ABCC6, Pyrophosphate and Ectopic Calcification: Therapeutic Solutions. <i>International Journal of Molecular Sciences</i> , 2021 , 22,	6.3	4
74	Genotype-phenotype correlation in pseudoxanthoma elasticum. 2021 , 324, 18-26		7
73	INZ-701 Prevents Ectopic Tissue Calcification and Restores Bone Architecture and Growth in ENPP1-Deficient Mice. <i>Journal of Bone and Mineral Research</i> , 2021 , 36, 1594-1604	6.3	3
72	Rare Modifier Variants Alter the Severity of Cardiovascular Disease in Pseudoxanthoma Elasticum: Identification of Novel Candidate Modifier Genes and Disease Pathways Through Mixture of Effects Analysis. 2021 , 9, 612581		2
71	Generalized Arterial Calcification of Infancy Type 1 (GACI1): Identification of a Novel Pathogenic Variant (c.1715T>C (p.Leu572Ser)). 2021 , 11,		0
70	Mutagenic Analysis of the Putative ABCC6 Substrate-Binding Cavity Using a New Homology Model. <i>International Journal of Molecular Sciences</i> , 2021 , 22,	6.3	3
69	Phenotypic Features and Genetic Findings in a Cohort of Italian Patients and Update of the Ophthalmologic Evaluation Score. <i>Journal of Clinical Medicine</i> , 2021 , 10,	5.1	3
68	From Clinical Diagnosis to the Discovery of Multigene Rare Sequence Variants in : A Case Report. 2021 , 8, 726856		
67	Ectopic Calcification and Hypophosphatemic Rickets: Natural History of ENPP1 and ABCC6 Deficiencies. <i>Journal of Bone and Mineral Research</i> , 2021 , 36, 2193-2202	6.3	12
66	A reference range for plasma levels of inorganic pyrophosphate in children using the ATP sulfurylase method. 2021 ,		0
65	A Case Report of with Rare Sequence Variants in Genes Related to Inherited Retinal Diseases. 2021 , 11,		1
64	Functional Assessment of Missense Variants in the ABCC6 Gene Implicated in Pseudoxanthoma Elasticum, a Heritable Ectopic Mineralization Disorder. 2021 ,		2
63	Autosomal recessive hypophosphatemic rickets type 2 (ARHR2) due to ENPP1-deficiency. 2021 , 153, 116111		1
62	The 3-step Greek protocol of neck skin rejuvenation inspired by a case of pseudoxanthoma elasticum: combination of laser skin resurfacing, collagen booster and PRP. 2021 , 5, 66-73		
61	Genetische Bindegewebskrankheiten. 2014 , 1912-1925		1
60	Hereditäre Bindegewebskrankheiten bei Kindern und Jugendlichen. <i>Springer Reference Medizin</i> , 2019 , 1-25	0	1
59	Pattern dystrophy-like changes and coquille d'oeuf atrophy in elderly patients affected by pseudoxanthoma elasticum. 2020 , 258, 1881-1892		6
58	Ectonucleotidases in Immunobiology. 2016 , 424-431		9

57	Late-onset Pseudoxanthoma Elasticum Associated with a Hypomorphic ABCC6 Variant. 2020 , 218, 255-260		4
56	Inhibition of vascular calcification by inositol phosphates derivatized with ethylene glycol oligomers. 2020 , 11, 721		23
55	The pathogenic p.(R391G) ABCC6 displays incomplete penetrance implying the necessity of an interacting partner for the development of pseudoxanthoma elasticum.		2
54	Pathology of the Elastic Matrix. 2016 , 31-80		1
53	From variome to phenome: Pathogenesis, diagnosis and management of ectopic mineralization disorders. <i>World Journal of Clinical Cases</i> , 2015 , 3, 556-74	1.6	21
52	Ectopic Mineralization and Conductive Hearing Loss in Enpp1asj Mutant Mice, a New Model for Otitis Media and Tympanosclerosis. <i>PLoS ONE</i> , 2016 , 11, e0168159	3-7	13
51	Calcification in dermal fibroblasts from a patient with GGCX syndrome accompanied by upregulation of osteogenic molecules. <i>PLoS ONE</i> , 2017 , 12, e0177375	3-7	5
50	Etidronate prevents, but does not reverse, ectopic mineralization in a mouse model of pseudoxanthoma elasticum (. <i>Oncotarget</i> , 2018 , 9, 30721-30730	3-3	20
49	Ectopic mineralization of cartilage and collagen-rich tendons and ligaments in Enpp1asj-2J mice. <i>Oncotarget</i> , 2016 , 7, 12000-9	3-3	9
48	Homology Modeling and Virtual Screening of Proteins Related to PXE and PXE-like Diseases: Insights for Overlapping Metabolites. <i>Current Pharmaceutical Biotechnology</i> , 2020 , 21, 1470-1478	2.6	1
47	Therapeutics Development for Pseudoxanthoma Elasticum and Related Ectopic Mineralization Disorders: Update 2020. <i>Journal of Clinical Medicine</i> , 2020 , 10,	5-1	10
46	Vascular calcification; Stony bridge between kidney and heart. <i>Journal of Cardiovascular and Thoracic Research</i> , 2020 , 12, 165-171	1-3	3
45	Magnesium and Anti-phosphate Treatment with Bisphosphonates for Generalised Arterial Calcification of Infancy: A Case Report. <i>JCRPE Journal of Clinical Research in Pediatric Endocrinology</i> , 2019 , 11, 311-318	1-9	2
44	IL-1 β in atherosclerotic vascular calcification: From bench to bedside. <i>International Journal of Biological Sciences</i> , 2021 , 17, 4353-4364	11.2	2
43	A meta-analysis of prognostic biomarkers in neonatal retinal hemorrhage. <i>International Ophthalmology</i> , 2021 , 1	2.2	1
42	Role of Ectonucleotidases and Purinergic Receptors in Calcific Aortic Valve Disease. 2014 , 117-126		
41	Pseudoxanthoma Elasticum. 2014 , 441-447		
40	Vascular Genetics. 2014 , 1-41		

39	Vascular Genetics. 2015 , 53-88		
38	Hereditäre Bindegewebskrankheiten bei Kindern und Jugendlichen. 2015 , 1-24		
37	Physiology of the Developing Kidney: Disorders and Therapy of Calcium and Phosphorous Homeostasis. 2016 , 291-339		
36	VASCULAR CALCIFICATION, ATHEROSCLEROSIS AND BONE LOSS (OSTEOPOROSIS): NEW PATHOPHYSIOLOGICAL MECHANISMS AND FUTURE PERSPECTIVES FOR PHARMACOLOGICAL THERAPY. <i>Almanah Kliničkoj Mediciny</i> , 2016 , 44, 513-534	0.2	2
35	Endocrine Regulation of Phosphate Homeostasis. 2017 , 71-82		
34	Genetic testing for vascular anomalies. <i>The EuroBiotech Journal</i> , 2018 , 2, 26-31	1.5	
33	Hereditäre Bindegewebskrankheiten. <i>Springer Reference Medizin</i> , 2020 , 2835-2859	0	
32	Cerebral vasculopathies. 2020 , 561-578		
31	Profile of genetic variations in severely calcified carotid plaques by whole-exome sequencing. <i>Surgical Neurology International</i> , 2020 , 11, 286	1	0
30	Purinergic signaling in systemic sclerosis. <i>Rheumatology</i> , 2021 ,	3.9	
29	Oral supplementation of inorganic pyrophosphate in pseudoxanthoma elasticum. <i>Experimental Dermatology</i> , 2021 ,	4	4
28	Syndromic retinal disease. 2022 , 267-321		
27	Pulmonary affection of patients with Pseudoxanthoma elasticum: Long-term development and genotype-phenotype-correlation.. <i>Intractable and Rare Diseases Research</i> , 2022 , 11, 7-14	1.4	
26	Disorders of Nucleotide Metabolism. 2022 , 213-233		
25	Vitamin D and Calcium Supplementation Accelerate Vascular Calcification in a Model of Pseudoxanthoma Elasticum.. <i>International Journal of Molecular Sciences</i> , 2022 , 23,	6.3	1
24	The Abcc6a Knockout Zebrafish Model as a Novel Tool for Drug Screening for Pseudoxanthoma Elasticum.. <i>Frontiers in Pharmacology</i> , 2022 , 13, 822143	5.6	1
23	Identification of ENPP1 haploinsufficiency in patients with diffuse idiopathic skeletal hyperostosis and early-onset osteoporosis.. <i>Journal of Bone and Mineral Research</i> , 2022 ,	6.3	2
22	Case Report: A Novel Genetic Mutation Causes Idiopathic Infantile Arterial Calcification in Preterm Infants.. <i>Frontiers in Genetics</i> , 2021 , 12, 763916	4.5	0

21 Data_Sheet_1.PDF. 2020,

20 Mutation update: Variants of the ENPP1 gene in pathologic calcification, hypophosphatemic rickets, and cutaneous hypopigmentation with punctate keratoderma.. *Human Mutation*, 2022, 4-7 ○19 ENPP1 variants in patients with GACI and PXE expand the clinical and genetic heterogeneity of heritable disorders of ectopic calcification.. *PLoS Genetics*, 2022, 18, e1010192 6 318 INZ-701, a recombinant ENPP1 enzyme, prevents ectopic calcification in an Abcc6 mouse model of pseudoxanthoma elasticum.. *Experimental Dermatology*, 2022, 4 317 Generalized Arterial Calcification of Infancy (GACI): Optimizing Care with a Multidisciplinary Approach. *Journal of Multidisciplinary Healthcare*, Volume 15, 1261-1276 2.8 ○16 Inhibition of alkaline phosphatase impairs dyslipidemia and protects mice from atherosclerosis. *Translational Research*, 2022, 11 1

15 Diseases of small and medium-sized blood vessels. 2022, 307-351 1

14 ABCC6 deficiency and bone loss: A double benefit of etidronate for patient presenting with pseudoxanthoma elasticum?. *Experimental Dermatology*, 413 Characterization of hearing-impairment in Generalized Arterial Calcification of Infancy (GACI). *Orphanet Journal of Rare Diseases*, 2022, 17, 4-2 ○

12 Case Report and Review of Literature: Autosomal Recessive Hypophosphatemic Rickets Type 2 Caused by a Pathogenic Variant in ENPP1 Gene. 13, ○

11 Lifelong impact of ENPP1 Deficiency and the early onset form of ABCC6 Deficiency from patient or caregiver perspective. 2022, 17, e0270632 ○

10 PHEXL222P Mutation Increases Phex Expression in a New ENU Mouse Model for XLH Disease. 2022, 13, 1356

9 ENPP1 Deficiency: A Clinical Update on the Relevance of Individual Variants Using a Locus-Specific Patient Database. ○

8 Prenatal diagnosis of generalized arterial calcification of infancy in the second trimester. ○

7 The pathogenic c.1171A>G (p.Arg391Gly) and c.2359G>A (p.Val787Ile) ABCC6 variants display incomplete penetrance causing pseudoxanthoma elasticum in a subset of individuals. ○

6 Severe early-onset manifestations of generalized arterial calcification of infancy (mimicking severe coarctation of the aorta) with ABCC6 gene variant [Case report and literature review. 9, ○

5 Case report: A rare homozygous variation in the ENPP1 gene, presenting with generalized arterial calcification of infancy in a Chinese infant. 10, ○

4 Effects of food, fasting, and exercise on plasma pyrophosphate levels and ENPP1 activity in healthy adults. 2023, 171, 116750 ○

- 3 Review of Basic Research about Ossification of the Spinal Ligaments Focusing on Animal Models. **2023**, 12, 1958 ○
- 2 Inorganic Pyrophosphate Plasma Levels Are Decreased in Pseudoxanthoma Elasticum Patients and Heterozygous Carriers but Do Not Correlate with the Genotype or Phenotype. **2023**, 12, 1893 ○
- 1 Acquired and other retinal diseases (including juvenile X-linked retinoschisis). **2017**, 537-545.e2 ○