

Identification of Two Novel Missense Mutations (p.R122 (MRP6) Gene in a Japanese Patient with Pseudoxanthom

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Citation Report

#	ARTICLE	IF	CITATIONS
1	Identification of a DNA Methylation-dependent Activator Sequence in the Pseudoxanthoma Elasticum Gene, ABCC6. Journal of Biological Chemistry, 2005, 280, 18643-18650.	3.4	35
2	Identification of a novel deletion in the ABCC6 gene leading to Pseudoxanthoma elasticum. Journal of Dermatological Science, 2005, 40, 115-121.	1.9	17
4	HNF4 β and NF-E2 are key transcriptional regulators of the murine Abcc6 gene expression. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 2006, 1759, 426-436.	2.4	21
5	Mutational analysis of the ABCC6 gene and the proximal ABCC6 gene promoter in German patients with pseudoxanthoma elasticum (PXE). Human Mutation, 2006, 27, 831-831.	2.5	55
6	Mutation detection in the ABCC6 gene and genotype phenotype analysis in a large international case series affected by pseudoxanthoma elasticum. Journal of Medical Genetics, 2007, 44, 621-628.	3.2	161
7	Acute myocardial infarction and a new ABCC6 mutation in a 16-year-old boy with pseudoxanthoma elasticum. International Journal of Cardiology, 2007, 116, 261-262.	1.7	18
8	Development of a Rapid, Reliable Genetic Test for Pseudoxanthoma Elasticum. Journal of Molecular Diagnostics, 2007, 9, 105-112.	2.8	16
9	Novel clinico-molecular insights in pseudoxanthoma elasticum provide an efficient molecular screening method and a comprehensive diagnostic flowchart. Human Mutation, 2008, 29, 205-205.	2.5	82
10	Pseudoxanthoma elasticum: genetic diagnostic markers. Expert Opinion on Medical Diagnostics, 2008, 2, 63-79.	1.6	24
11	Pseudoxanthoma Elasticum: A Streamlined, Ethnicity-based Mutation Detection Strategy. Clinical and Translational Science, 2010, 3, 295-298.	3.1	9
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13	The Multifaceted Complexity of Genetic Diseases: A Lesson from Pseudoxanthoma Elasticum. , 2011, , .		4
14	Pseudoxanthoma Elasticum and Sudden Death. Journal of Forensic Sciences, 2011, 56, 418-422.	1.6	17
15	Genetic Heterogeneity of Pseudoxanthoma Elasticum: The Chinese Signature Profile of ABCC6 and ENPP1 Mutations. Journal of Investigative Dermatology, 2015, 135, 1294-1302.	0.7	22
16	Resistance to Targeted Therapies Against Adult Brain Cancers. Resistance To Targeted Anti-cancer Therapeutics, 2016, , .	0.1	4
17	Targeting Chemotherapy Resistance in Glioblastoma Through Modulation of ABC Transporters. Resistance To Targeted Anti-cancer Therapeutics, 2016, , 25-54.	0.1	1
18	Analysis of clinical symptoms and <i>ABCC6</i> mutations in 76 Japanese patients with pseudoxanthoma elasticum. Journal of Dermatology, 2017, 44, 644-650.	1.2	20
19	<i>ABCC6</i> Gene Analysis in 20 Japanese Patients with Angioid Streaks Revealing Four Frequent and Two Novel Variants and Pseudodominant Inheritance. Journal of Ophthalmology, 2017, 2017, 1-7.	1.3	4

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20	Coquille d'oeuf in young patients affected with Pseudoxantoma elasticum. <i>Ophthalmic Genetics</i> , 2019, 40, 242-246.	1.2	11
21	Adaptive Optics Imaging in Patients Affected by Pseudoxanthoma Elasticum. <i>American Journal of Ophthalmology</i> , 2021, 224, 84-95.	3.3	4
22	Mutagenic Analysis of the Putative ABCC6 Substrate-Binding Cavity Using a New Homology Model. <i>International Journal of Molecular Sciences</i> , 2021, 22, 6910.	4.1	8
23	Phenotypic Features and Genetic Findings in a Cohort of Italian Pseudoxanthoma Elasticum Patients and Update of the Ophthalmologic Evaluation Score. <i>Journal of Clinical Medicine</i> , 2021, 10, 2710.	2.4	8