Olivier M Vanakker

List of Publications by Year
in descending order

87 papers	2,862 citations	201674 27 h-index	197818 49 g-index
89 all docs	89 docs citations	89 times ranked	4335 citing authors

#	Article	IF	Citations
1	Mutations in XPR1 cause primary familial brain calcification associated with altered phosphate export. Nature Genetics, 2015, 47, 579-581.	21.4	237
2	Pseudoxanthoma Elasticum-Like Phenotype with Cutis Laxa and Multiple Coagulation Factor Deficiency Represents a Separate Genetic Entity. Journal of Investigative Dermatology, 2007, 127, 581-587.	0.7	168
3	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
4	Exonic Deletions in AUTS2 Cause a Syndromic Form of Intellectual Disability and Suggest a Critical Role for the C Terminus. American Journal of Human Genetics, 2013, 92, 210-220.	6.2	135
5	Comprehensive molecular analysis demonstrates type V collagen mutations in over 90% of patients with classic EDS and allows to refine diagnostic criteria. Human Mutation, 2012, 33, 1485-1493.	2.5	133
6	Mutations in CDC45 , Encoding an Essential Component of the Pre-initiation Complex, Cause Meier-Gorlin Syndrome and Craniosynostosis. American Journal of Human Genetics, 2016, 99, 125-138.	6.2	92
7	Implementation of genomic arrays in prenatal diagnosis: The Belgian approach to meet the challenges. European Journal of Medical Genetics, 2014, 57, 151-156.	1.3	91
8	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
9	Endogenous Calcification Inhibitors in the Prevention of Vascular Calcification: A Consensus Statement From the COST Action EuroSoftCalcNet. Frontiers in Cardiovascular Medicine, 2018, 5, 196.	2.4	82
10	Low serum vitamin K in PXE results in defective carboxylation of mineralization inhibitors similar to the GGCX mutations in the PXE-like syndrome. Laboratory Investigation, 2010, 90, 895-905.	3.7	72
11	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. JAMA Neurology, 2018, 75, 557.	9.0	69
12	Comprehensive Clinical and Molecular Analysis of 12 Families with Type 1 Recessive Cutis Laxa. Human Mutation, 2013, 34, 111-121.	2.5	67
13	Left ventricular outflow obstruction in rhabdomyoma of infancy: meta-analysis of the literature. Journal of Pediatrics, 2003, 143, 258-263.	1.8	64
14	Characterization of Cardiovascular Involvement in Pseudoxanthoma Elasticum Families. Arteriosclerosis, Thrombosis, and Vascular Biology, 2013, 33, 2646-2652.	2.4	62
15	SNX10 mutations define a subgroup of human autosomal recessive osteopetrosis with variable clinical severity. Journal of Bone and Mineral Research, 2013, 28, 1041-1049.	2.8	59
16	Redefining the MED13L syndrome. European Journal of Human Genetics, 2015, 23, 1308-1317.	2.8	53
17	Vitamin K does not prevent soft tissue mineralization in a mouse model of pseudoxanthoma elasticum. Cell Cycle, 2011, 10, 1810-1820.	2.6	50
18	The Genetics of Soft Connective Tissue Disorders. Annual Review of Genomics and Human Genetics, 2015, 16, 229-255.	6.2	50

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19	Histopathology of Pseudoxanthoma Elasticum and Related Disorders: Histological Hallmarks and Diagnostic Clues. Scientifica, 2012, 2012, 1-15.	1.7	48
20	Novel pathogenic COL11A1/COL11A2 variants in Stickler syndrome detected by targeted NGS and exome sequencing. Molecular Genetics and Metabolism, 2014, 113, 230-235.	1.1	48
21	FOXP1-related intellectual disability syndrome: a recognisable entity. Journal of Medical Genetics, 2017, 54, 613-623.	3.2	48
22	Primary brain calcification: an international study reporting novel variants and associated phenotypes. European Journal of Human Genetics, 2018, 26, 1462-1477.	2.8	48
23	Perturbation of specific pro-mineralizing signalling pathways in human and murine pseudoxanthoma elasticum. Orphanet Journal of Rare Diseases, 2014, 9, 66.	2.7	35
24	Twenty patients including 7 probands with autosomal dominant cutis laxa confirm clinical and molecular homogeneity. Orphanet Journal of Rare Diseases, 2013, 8, 36.	2.7	33
25	Novel defects in collagen XII and VI expand the mixed myopathy/Ehlersâ€“Danlos syndrome spectrum and lead to variant-specific alterations in the extracellular matrix. Genetics in Medicine, 2020, 22, 112-123.	2.4	33
26	Added value of infrared, red-free and autofluorescence fundus imaging in pseudoxanthoma elasticum. British Journal of Ophthalmology, 2010, 94, 479-486.	3.9	32
27	Mate pair sequencing for the detection of chromosomal aberrations in patients with intellectual disability and congenital malformations. European Journal of Human Genetics, 2014, 22, 652-659.	2.8	32
28	From variome to phenome: Pathogenesis, diagnosis and management of ectopic mineralization disorders. World Journal of Clinical Cases, 2015, 3, 556.	0.8	32
29	GGCX-Associated Phenotypes: An Overview in Search of Genotype-Phenotype Correlations. International Journal of Molecular Sciences, 2017, 18, 240.	4.1	30
30	Pseudoxanthoma Elasticum with Generalized Retinal Dysfunction, a Common Finding?. , 2007, 48, 4250.		28
31	Pathogenic variants in the ABCC6 gene are associated with an increased risk for ischemic stroke. Brain Pathology, 2018, 28, 822-831.	4.1	28
32	De novo mutations in MSL3 cause an X-linked syndrome marked by impaired histone H4 lysine 16 acetylation. Nature Genetics, 2018, 50, 1442-1451.	21.4	28
33	Plasma inorganic pyrophosphate and alkaline phosphatase in patients with pseudoxanthoma elasticum. Annals of Translational Medicine, 2019, 7, 798-798.	1.7	26
34	Efficiency of Exome Sequencing for the Molecular Diagnosis of Pseudoxanthoma Elasticum. Journal of Investigative Dermatology, 2015, 135, 992-998.	0.7	25
35	Severe earlyâ€onset manifestations of pseudoxanthoma elasticum resulting from the cumulative effects of several deleterious mutations inENPP1,ABCC6andHBB: transient improvement in ectopic calcification with sodium thiosulfate. British Journal of Dermatology, 2020, 183, 367-372.	1.5	25
36	Molecular Docking Simulations Provide Insights in the Substrate Binding Sites and Possible Substrates of the ABCC6 Transporter. PLoS ONE, 2014, 9, e102779.	2.5	25

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37	The Belgian MicroArray Prenatal (BEMAPRE) database: A systematic nationwide repository of fetal genomic aberrations. Prenatal Diagnosis, 2018, 38, 1120-1128.	2.3	24
38	Cellular signaling in pseudoxanthoma elasticum: an update. Cellular Signalling, 2019, 55, 119-129.	3.6	22
39	Nonlinear optical microscopy is a novel tool for the analysis of cutaneous alterations in pseudoxanthoma elasticum. Lasers in Medical Science, 2020, 35, 1821-1830.	2.1	21
40	Disruption of NEUROD2 causes a neurodevelopmental syndrome with autistic features via cell-autonomous defects in forebrain glutamatergic neurons. Molecular Psychiatry, 2021, 26, 6125-6148.	7.9	21
41	Hereditary Connective Tissue Diseases in Young Adult Stroke: A Comprehensive Synthesis. Stroke Research and Treatment, 2011, 2011, 1-18.	0.8	20
42	Retinitis Pigmentosa, Cutis Laxa, and Pseudoxanthoma Elasticumâ€“Like Skin Manifestations Associated with GGCX Mutations. Journal of Investigative Dermatology, 2014, 134, 2331-2338.	0.7	20
43	Chikungunya outbreak in Bangladesh (2017): Clinical and hematological findings. PLoS Neglected Tropical Diseases, 2020, 14, e0007466.	3.0	19
44	Generation and Validation of a Complete Knockout Model of abcc6a in Zebrafish. Journal of Investigative Dermatology, 2018, 138, 2333-2342.	0.7	18
45	Acquired pseudoxanthoma elasticum presenting after liver transplantation. Journal of the American Academy of Dermatology, 2011, 64, 873-878.	1.2	17
46	Reassessment of causality of ABCC6 missense variants associated with pseudoxanthoma elasticum based on Sherloc. Genetics in Medicine, 2021, 23, 131-139.	2.4	17
47	Atypical presentation of pseudoxanthoma elasticum with abdominal cutis laxa: Evidence for a spectrum of ectopic calcification disorders?. American Journal of Medical Genetics, Part A, 2011, 155, 2855-2859.	1.2	16
48	Second family with the bostonâ€type craniosynostosis syndrome: Novel mutation and expansion of the clinical spectrum. American Journal of Medical Genetics, Part A, 2013, 161, 2352-2357.	1.2	16
49	Deletions in 14q24.1q24.3 are associated with congenital heart defects, brachydactyly, and mild intellectual disability. American Journal of Medical Genetics, Part A, 2014, 164, 620-626.	1.2	16
50	Delineating the psychiatric and behavioral phenotype of recurrent 2q13 deletions and duplications. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 397-405.	1.7	16
51	Can APOE and MTHFR polymorphisms have an influence on the severity of cardiovascular manifestations in Italian Pseudoxanthoma elasticum affected patients?. Molecular Genetics and Metabolism Reports, 2014, 1, 477-482.	1.1	15
52	Genotype-phenotype correlation in pseudoxanthoma elasticum. Atherosclerosis, 2021, 324, 18-26.	0.8	15
53	Zebrafish models for ectopic mineralization disorders: practical issues from morpholino design to post-injection observations. Frontiers in Genetics, 2013, 4, 74.	2.3	13
54	Internal Carotid Artery Hypoplasia: A New Clinical Feature in Pseudoxanthoma Elasticum. Journal of Stroke, 2019, 21, 108-111.	3.2	13

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55	Cellular and Molecular Biomarkers Indicate Premature Aging in Pseudoxanthoma Elasticum Patients. , 2020, 11, 536.		12
56	Minocycline Attenuates Excessive DNA Damage Response and Reduces Ectopic Calcification in Pseudoxanthoma Elasticum. Journal of Investigative Dermatology, 2022, 142, 1629-1638.e6.	0.7	12
57	The ABCC6 transporter: what lessons can be learnt from other ATP-binding cassette transporters?. Frontiers in Genetics, 2013, 4, 203.	2.3	11
58	The Role of Vitamin K and Its Related Compounds in Mendelian and Acquired Ectopic Mineralization Disorders. International Journal of Molecular Sciences, 2019, 20, 2142.	4.1	11
59	Genetic deletion of Abcc6 disturbs cholesterol homeostasis in mice. Scientific Reports, 2021, 11, 2137.	3.3	11
60	Pharmacogenomics in Children: Advantages and Challenges of Next Generation Sequencing Applications. International Journal of Pediatrics (United Kingdom), 2013, 2013, 1-8.	0.8	10
61	Clinical Zinc Deficiency as Early Presentation of Wilson Disease. Journal of Pediatric Gastroenterology and Nutrition, 2015, 60, 457-459.	1.8	10
62	Phenotype of a Belgian Family With 6p25 Deletion Syndrome. Annals of Otology, Rhinology and Laryngology, 2016, 125, 734-745.	1.1	10
63	7p22.1 microdeletions involving ACTB associated with developmental delay, short stature, and microcephaly. European Journal of Medical Genetics, 2016, 59, 502-506.	1.3	10
64	Phenotypic spectrum of the <scp>RBM10</scp>â€mediated intellectual disability and congenital malformation syndrome beyond classic <scp>TARP</scp> syndrome features. Clinical Genetics, 2021, 99, 449-456.	2.0	10
65	The ABCC6 Transporter as a Paradigm for Networking from an Orphan Disease to Complex Disorders. BioMed Research International, 2015, 2015, 1-18.	1.9	9
66	A Belgian consensus strategy to identify familial hypercholesterolaemia in the coronary care unit and its subsequent cascade screening and treatment: BEL-FaHST (The BELgium Familial) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 297 Td (Hyper	0.8	9
67	Acral Acquired Cutis Laxa Associated with IgA Multiple Myeloma, Joint Hyperlaxity and Urticarial Neutrophilic Dermatosis. Acta Dermato-Venereologica, 2014, 94, 743-744.	1.3	8
68	From membrane to mineralization: the curious case of the ABCC6 transporter. FEBS Letters, 2020, 594, 4109-4133.	2.8	8
69	<scp>VEGFA</scp> variants as prognostic markers for the retinopathy in pseudoxanthoma elasticum. Clinical Genetics, 2020, 98, 74-79.	2.0	8
70	Serum Calcification Propensity T50 Associates with Disease Severity in Patients with Pseudoxanthoma Elasticum. Journal of Clinical Medicine, 2022, 11, 3727.	2.4	7
71	Lambertâ€“Eaton myasthenic syndrome in a 13-year-old girl with Xp11.22-p11.23 duplication. European Journal of Paediatric Neurology, 2014, 18, 439-443.	1.6	6
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. Frontiers in Cell and Developmental Biology, 2021, 9, 612581.	3.7	6

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73	Functional Polymorphism in Gamma-Glutamylcarboxylase is a Risk Factor for Severe Neonatal Hemorrhage. Journal of Pediatrics, 2011, 159, 347-349.	1.8	5
74	Morpholino-Mediated Gene Knockdown in Zebrafish: It Is All About Dosage and Validation. Journal of Investigative Dermatology, 2019, 139, 1599-1600.	0.7	5
75	Clinical and subclinical findings in heterozygous ABCC6 carriers: results from a Belgian cohort and clinical practice guidelines. Journal of Medical Genetics, 2022, 59, 496-504.	3.2	5
76	Minocycline Counteracts Ectopic Calcification in a Murine Model of Pseudoxanthoma Elasticum: A Proof-of-Concept Study. International Journal of Molecular Sciences, 2022, 23, 1838.	4.1	5
77	Reply to the article of C. Markello et al. entitled â€œVascular pathology of medial arterial calcifications in NT5E deficiency: Implications for the role of adenosine in pseudoxanthoma elasticumâ€• Molecular Genetics and Metabolism, 2011, 103, 199-200.	1.1	4
78	Hypotonia and delayed motor development as an early presentation of Lowe syndrome: case report and literature review. Acta Clinica Belgica, 2019, 74, 460-464.	1.2	4
79	Genetic counseling in the context of Bangladesh: current scenario, challenges, and a framework for genetic service implementation. Orphanet Journal of Rare Diseases, 2021, 16, 168.	2.7	4
80	A likely pathogenic variant in theSLC20A2gene presenting with progressive myoclonus. Annals of Clinical and Translational Neurology, 2019, 6, 605-609.	3.7	3
81	Comprehensive validation of a diagnostic strategy for sequencing genes with one or multiple pseudogenes using pseudoxanthoma elasticum as a model. Journal of Genetics and Genomics, 2021, 48, 289-299.	3.9	2
82	â€˜De novoâ€™ Col4A2 mutation in a patient with migraine, leukoencephalopathy, and small carotid aneurysms. Journal of Neurology, 2016, 263, 2327-2329.	3.6	1
83	Various vascular malformations are prevalent in Finnish pseudoxanthoma elasticum (PXE) patients: a national registry study. Orphanet Journal of Rare Diseases, 2022, 17, 185.	2.7	1
84	Use of an event recorder in the decision for pacemaker implantation in a child with syncope. European Journal of Pediatrics, 2002, 161, 267-269.	2.7	0
85	P1.14 Overt myopathy in Ehlersâ€“Danlos syndrome caused by tenascin-X deficiency: extending the clinical spectrum and refining the muscle pathology. Neuromuscular Disorders, 2010, 20, 604.	0.6	0
86	Coordinated orphan disease research: yes, we can!. Frontiers in Genetics, 2013, 4, 207.	2.3	0
87	Angioid streaks beyond pseudoxanthoma elasticum. Acta Ophthalmologica, 2010, 88, 0-0.	1.1	0