Paul J Coucke

List of Publications by Year
in descending order

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223 papers	15,867 citations	18482 62 h-index	19190 118 g-index
235 all docs	235 docs citations	235 times ranked	14666 citing authors

#	Article	IF	Citations
1	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
2	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
3	Human germline nuclear transfer to overcome mitochondrial disease and failed fertilization after ICSI. Journal of Assisted Reproduction and Genetics, 2022, 39, 609-618.	2.5	11
4	High myopia and vitreal veils in a patient with Porettiâ€“ Boltshauser syndrome due to a novel homozygous LAMA1 mutation. Ophthalmic Genetics, 2022, , 1-5.	1.2	0
5	G Proteinâ€Coupled Receptor Kinase 6 (GRK6) Regulation of Insulin Processing and Secretion. FASEB Journal, 2022, 36, .	0.5	1
6	Shortcutting the diagnostic odyssey: the multidisciplinary Program for Undiagnosed Rare Diseases in adults (UD-PrOZA). Orphanet Journal of Rare Diseases, 2022, 17, .	2.7	12
7	Serum Calcification Propensity T50 Associates with Disease Severity in Patients with Pseudoxanthoma Elasticum. Journal of Clinical Medicine, 2022, 11, 3727.	2.4	7
8	The corneoscleral shape in Marfan syndrome. Acta Ophthalmologica, 2021, 99, 405-410.	1.1	8
9	Photoconvertible fluorescent proteins: a versatile tool in zebrafish skeletal imaging. Journal of Fish Biology, 2021, 98, 1007-1017.	1.6	6
10	Reassessment of causality of ABCC6 missense variants associated with pseudoxanthoma elasticum based on Sherloc. Genetics in Medicine, 2021, 23, 131-139.	2.4	17
11	Arterial Tortuosity Syndrome: An Ascorbate Compartmentalization Disorder?. Antioxidants and Redox Signaling, 2021, 34, 875-889.	5.4	11
12	The ZE-Tunnel: An Affordable, Easy-to-Assemble, and User-Friendly Benchtop Zebrafish Swim Tunnel. Zebrafish, 2021, 18, 29-41.	1.1	3
13	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
14	Digital Polymerase Chain Reaction for Assessment of Mutant Mitochondrial Carry-over after Nuclear Transfer for In Vitro Fertilization. Clinical Chemistry, 2021, 67, 968-976.	3.2	4
15	G Proteinâ€Coupled Receptor Kinase 6 (GRK6) Regulation of Insulin Processing and Secretion. FASEB Journal, 2021, 35, .	0.5	0
16	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
17	Loss of zebrafish atp6v1e1b, encoding a subunit of vacuolar ATPase, recapitulates human ARCL type 2C syndrome and identifies multiple pathobiological signatures. PLoS Genetics, 2021, 17, e1009603.	3.5	3
18	A Reassessment of Copy Number Variations in Congenital Heart Defects: Picturing the Whole Genome. Genes, 2021, 12, 1048.	2.4	6

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19	Biallelic variants in MESD, which encodes a WNT-signaling-related protein, in four new families with recessively inherited osteogenesis imperfecta. Human Genetics and Genomics Advances, 2021, 2, 100051.	1.7	3
20	CRISPR-SID: Identifying EZH2 as a druggable target for desmoid tumors via inÂvivo dependency mapping. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	6
21	A clinical scoring system for congenital contractural arachnodactyly. Genetics in Medicine, 2020, 22, 124-131.	2.4	17
22	Hypomorphic zebrafish models mimic the musculoskeletal phenotype of Î²4GalT7-deficient Ehlers-Danlos syndrome. Matrix Biology, 2020, 89, 59-75.	3.6	19
23	Zebrafish: A Resourceful Vertebrate Model to Investigate Skeletal Disorders. Frontiers in Endocrinology, 2020, 11, 489.	3.5	74
24	Maximizing CRISPR/Cas9 phenotype penetrance applying predictive modeling of editing outcomes in Xenopus and zebrafish embryos. Scientific Reports, 2020, 10, 14662.	3.3	28
25	b3galt6 Knock-Out Zebrafish Recapitulate Î²3GalT6-Deficiency Disorders in Human and Reveal a Trisaccharide Proteoglycan Linkage Region. Frontiers in Cell and Developmental Biology, 2020, 8, 597857.	3.7	11
26	New insights on the clinical variability of FKBP10 mutations. European Journal of Medical Genetics, 2020, 63, 103980.	1.3	2
27	Phenomics-Based Quantification of CRISPR-Induced Mosaicism in Zebrafish. Cell Systems, 2020, 10, 275-286.e5.	6.2	38
28	<scp>VEGFA</scp> variants as prognostic markers for the retinopathy in pseudoxanthoma elasticum. Clinical Genetics, 2020, 98, 74-79.	2.0	8
29	Slc2a10 knock-out mice deficient in ascorbic acid synthesis recapitulate aspects of arterial tortuosity syndrome and display mitochondrial respiration defects. Human Molecular Genetics, 2020, 29, 1476-1488.	2.9	5
30	Lrp5 Mutant and Crispant Zebrafish Faithfully Model Human Osteoporosis, Establishing the Zebrafish as a Platform for CRISPR-Based Functional Screening of Osteoporosis Candidate Genes. Journal of Bone and Mineral Research, 2020, 36, 1749-1764.	2.8	19
31	Comprehensive in silico Study of GLUT10: Prediction of Possible Substrate Binding Sites and Interacting Molecules. Current Pharmaceutical Biotechnology, 2020, 21, 117-130.	1.6	2
32	A homozygous pathogenic missense variant broadens the phenotypic and mutational spectrum of CREB3L1-related osteogenesis imperfecta. Human Molecular Genetics, 2019, 28, 1801-1809.	2.9	21
33	Decreased Nuclear Ascorbate Accumulation Accompanied with Altered Genomic Methylation Pattern in Fibroblasts from Arterial Tortuosity Syndrome Patients. Oxidative Medicine and Cellular Longevity, 2019, 2019, 1-11.	4.0	4
34	Nucleic acids enrichment of fungal pathogens to study host-pathogen interactions. Scientific Reports, 2019, 9, 18037.	3.3	2
35	Vascular Ehlersâ€“Danlos syndrome in 2 Polish patients: identification of 2 novel COL3A1 gene mutations. Kardiologia Polska, 2019, 77, 1070-1073.	0.6	2
36	Future perspectives of genome-scale sequencing. Acta Clinica Belgica, 2018, 73, 7-10.	1.2	11

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37	A mutation update on the LDS-associated genesTGFB2/3andSMAD2/3. Human Mutation, 2018, 39, 621-634.	2.5	116
38	Arterial tortuosity syndrome: 40 new families and literature review. Genetics in Medicine, 2018, 20, 1236-1245.	2.4	66
39	MicroCT-Based Phenomics in the Zebrafish Skeleton Reveals Virtues of Deep Phenotyping in a Distributed Organ System. Zebrafish, 2018, 15, 77-78.	1.1	13
40	Genetic study of nonâ€syndromic tooth agenesis through the screening of paired box 9, msh homeobox 1, axin 2, and Wnt family member 10A genes: a caseâ€series. European Journal of Oral Sciences, 2018, 126, 24-32.	1.5	8
41	Genetic analysis of osteogenesis imperfecta in the <scp>P</scp>alestinian population: molecular screening of 49 affected families. Molecular Genetics & Genomic Medicine, 2018, 6, 15-26.	1.2	26
42	Pathogenic variants in the ABCC6 gene are associated with an increased risk for ischemic stroke. Brain Pathology, 2018, 28, 822-831.	4.1	28
43	CRISPR/Cas9-mediated homology-directed repair by ssODNs in zebrafish induces complex mutational patterns resulting from genomic integration of repair-template fragments. DMM Disease Models and Mechanisms, 2018, 11, .	2.4	77
44	BATCH-GE: Analysis of NGS Data for Genome Editing Assessment. Methods in Molecular Biology, 2018, 1865, 83-90.	0.9	3
45	Homozygosity for CREB3L1 premature stop codon in first case of recessive osteogenesis imperfecta associated with OASIS-deficiency to survive infancy. Bone, 2018, 114, 268-277.	2.9	23
46	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
47	Generation and Validation of a Complete Knockout Model of abcc6a in Zebrafish. Journal of Investigative Dermatology, 2018, 138, 2333-2342.	0.7	18
48	Zebrafish type I collagen mutants faithfully recapitulate human type I collagenopathies. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E8037-E8046.	7.1	77
49	Tailoring the American College of Medical Genetics and Genomics and the Association for Molecular Pathology Guidelines for the Interpretation of Sequenced Variants in the FBN1 Gene for Marfan Syndrome. Circulation Genomic and Precision Medicine, 2018, 11, e002039.	3.6	20
50	Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa. American Journal of Human Genetics, 2017, 100, 216-227.	6.2	82
51	Tissueâ€specific mosaicism for a lethal osteogenesis imperfecta COL1A1 mutation causes mild OI/EDS overlap syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1047-1050.	1.2	13
52	A novel case of autosomal dominant cutis laxa in a consanguineous family: report and literature review. Clinical Dysmorphology, 2017, 26, 142-147.	0.3	7
53	A mild form of Stickler syndrome type II caused by mosaicism of COL11A1. European Journal of Medical Genetics, 2017, 60, 275-278.	1.3	14
54	Accurate quantification of homologous recombination in zebrafish: brca2 deficiency as a paradigm. Scientific Reports, 2017, 7, 16518.	3.3	9

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55	Sequence variants in nine different genes underlying rare skin disorders in 10 consanguineous families. International Journal of Dermatology, 2017, 56, 1406-1413.	1.0	6
56	GLUT10â€"Lacking in Arterial Tortuosity Syndromeâ€"Is Localized to the Endoplasmic Reticulum of Human Fibroblasts. International Journal of Molecular Sciences, 2017, 18, 1820.	4.1	15
57	A novel fibrillin-1 mutation in an egyptian marfan family: A proband showing nephrotic syndrome due to focal segmental glomerulosclerosis. Saudi Journal of Kidney Diseases and Transplantation: an Official Publication of the Saudi Center for Organ Transplantation, Saudi Arabia, 2017, 28, 141.	0.3	2
58	MicroCT-based phenomics in the zebrafish skeleton reveals virtues of deep phenotyping in a distributed organ system. ELife, 2017, 6, .	6.0	60
59	Hearing loss in Waardenburg syndrome: a systematic review. Clinical Genetics, 2016, 89, 416-425.	2.0	98
60	DNA Diagnostics of Hereditary Hearing Loss: A Targeted Resequencing Approach Combined with a Mutation Classification System. Human Mutation, 2016, 37, 812-819.	2.5	76
61	Identification of von Willebrand disease type 1 in a patient with Ehlersâ€"Danlos syndrome classic type. Haemophilia, 2016, 22, e309-11.	2.1	4
62	Glucose transporter type 10â€"lacking in arterial tortuosity syndromeâ€"facilitates dehydroascorbic acid transport. FEBS Letters, 2016, 590, 1630-1640.	2.8	25
63	Exome sequencing revealed a novel biallelic deletion in theDCAF17gene underlying Woodhouse Sakati syndrome. Clinical Genetics, 2016, 90, 263-269.	2.0	23
64	BATCH-GE: Batch analysis of Next-Generation Sequencing data for genome editing assessment. Scientific Reports, 2016, 6, 30330.	3.3	82
65	Characterization of a novel mutation in PAX9 gene in a family with non-syndromic dental agenesis. Archives of Oral Biology, 2016, 71, 110-116.	1.8	13
66	Loss of Type I Collagen Telopeptide Lysyl Hydroxylation Causes Musculoskeletal Abnormalities in a Zebrafish Model of Bruck Syndrome. Journal of Bone and Mineral Research, 2016, 31, 1930-1942.	2.8	65
67	Zebrafish Collagen Type I: Molecular and Biochemical Characterization of the Major Structural Protein in Bone and Skin. Scientific Reports, 2016, 6, 21540.	3.3	97
68	Mitral regurgitation as a phenotypic manifestation of nonphotosensitive trichothiodystrophy due to a splice variant in MPLKIP. BMC Medical Genetics, 2016, 17, 13.	2.1	5
69	An Exploratory Caseâ€Control Study on the Impact of <scp>IL</scp>â€1 Gene Polymorphisms on Early Implant Failure. Clinical Implant Dentistry and Related Research, 2016, 18, 234-240.	3.7	19
70	Association between Kniest dysplasia and chondrosarcoma in a child. American Journal of Medical Genetics, Part A, 2015, 167, 3204-3208.	1.2	3
71	Comparison of Methods for In-House Screening of HLA-B*57:01 to Prevent Abacavir Hypersensitivity in HIV-1 Care. PLoS ONE, 2015, 10, e0123525.	2.5	11
72	Ehlers-Danlos Syndrome, Hypermobility Type, Is Linked to Chromosome 8p22-8p21.1 in an Extended Belgian Family. Disease Markers, 2015, 2015, 1-9.	1.3	28

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73	Next Generation Sequencing to Determine the Cystic Fibrosis Mutation Spectrum in Palestinian Population. Disease Markers, 2015, 2015, 1-6.	1.3	11
74	The Genetics of Soft Connective Tissue Disorders. Annual Review of Genomics and Human Genetics, 2015, 16, 229-255.	6.2	50
75	Flexible, Scalable, and Efficient Targeted Resequencing on a Benchtop Sequencer for Variant Detection in Clinical Practice. Human Mutation, 2015, 36, 379-387.	2.5	43
76	Defective Proteolytic Processing of Fibrillar Procollagens and Prodecorin Due to Biallelic BMP1 Mutations Results in a Severe, Progressive Form of Osteogenesis Imperfecta. Journal of Bone and Mineral Research, 2015, 30, 1445-1456.	2.8	42
77	A study of the clinical and radiological features in a cohort of 93 patients with a COL2A1 mutation causing spondyloepiphyseal dysplasia congenita or a related phenotype. American Journal of Medical Genetics, Part A, 2015, 167, 461-475.	1.2	73
78	Gene panel sequencing in heritable thoracic aortic disorders and related entities â€“ results of comprehensive testing in a cohort of 264 patients. Orphanet Journal of Rare Diseases, 2015, 10, 9.	2.7	62
79	Genetics of the Ehlersâ€“Danlos syndrome: more than collagen disorders. Expert Opinion on Orphan Drugs, 2015, 3, 379-392.	0.8	3
80	Efficiency of Exome Sequencing for the Molecular Diagnosis of Pseudoxanthoma Elasticum. Journal of Investigative Dermatology, 2015, 135, 992-998.	0.7	25
81	RNF216 mutations as a novel cause of autosomal recessive Huntington-like disorder. Neurology, 2015, 84, 1760-1766.	1.1	59
82	Genetic Defects in TAPT1 Disrupt Ciliogenesis and Cause a Complex Lethal Osteochondrodysplasia. American Journal of Human Genetics, 2015, 97, 521-534.	6.2	39
83	Congenital contractural arachnodactyly due to a novel splice site mutation in the FBN2 gene. Journal of Pediatric Genetics, 2015, 03, 163-166.	0.7	3
84	The Soft Tissue Immunologic Response to Hydroxyapatiteâ€Coated Transmucosal Implant Surfaces: A Study in Humans. Clinical Implant Dentistry and Related Research, 2015, 17, e65-74.	3.7	19
85	Altered cytoskeletal organization characterized lethal but not surviving Brtl^{+/â^'}mice: insight on phenotypic variability in osteogenesis imperfecta. Human Molecular Genetics, 2015, 24, 6118-6133.	2.9	29
86	Refinement of the critical 2p25.3 deletion region: the role of MYT1L in intellectual disability and obesity. Genetics in Medicine, 2015, 17, 460-466.	2.4	45
87	Marfan Syndrome and Related Heritable Thoracic Aortic Aneurysms and Dissections. Current Pharmaceutical Design, 2015, 21, 4061-4075.	1.9	13
88	Absence of Cardiovascular Manifestations in a Haploinsufficient Tgfbr1 Mouse Model. PLoS ONE, 2014, 9, e89749.	2.5	9
89	Expressed Repeat Elements Improve RT-qPCR Normalization across a Wide Range of Zebrafish Gene Expression Studies. PLoS ONE, 2014, 9, e109091.	2.5	38
90	Type I Procollagen C-Propeptide Defects: Study of Genotype-Phenotype Correlation and Predictive Role of Crystal Structure. Human Mutation, 2014, 35, n/a-n/a.	2.5	38

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91	Congenital Fixed Dilated Pupils Due to ACTA2â€“ Multisystemic Smooth Muscle Dysfunction Syndrome. Journal of Neuro-Ophthalmology, 2014, 34, 137-143.	0.8	29
92	Neonatal progeroid variant of Marfan syndrome with congenital lipodystrophy results from mutations at the 3â€² end of FBN1 gene. European Journal of Medical Genetics, 2014, 57, 230-234.	1.3	41
93	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
94	Illumina sequencing of 15 deafness genes using fragmented amplicons. BMC Research Notes, 2014, 7, 509.	1.4	0
95	Perturbation of specific pro-mineralizing signalling pathways in human and murine pseudoxanthoma elasticum. Orphanet Journal of Rare Diseases, 2014, 9, 66.	2.7	35
96	Severe congenital cutis laxa with cardiovascular manifestations due to homozygous deletions in ALDH18A1. Molecular Genetics and Metabolism, 2014, 112, 310-316.	1.1	41
97	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
98	Deficiency for the ER-stress transducer OASIS causes severe recessive osteogenesis imperfecta in humans. Orphanet Journal of Rare Diseases, 2013, 8, 154.	2.7	98
99	A sensitive and specific diagnostic test for hearing loss using a microdroplet PCRâ€based approach and next generation sequencing. American Journal of Medical Genetics, Part A, 2013, 161, 145-152.	1.2	61
100	Novel MYH11 and ACTA2 mutations reveal a role for enhanced TGFÎ² signaling in FTAAD. International Journal of Cardiology, 2013, 165, 314-321.	1.7	134
101	Thoracic aortic-aneurysm and dissection in association with significant mitral valve disease caused by mutations in TGFB2. International Journal of Cardiology, 2013, 165, 584-587.	1.7	58
102	Comprehensive Clinical and Molecular Analysis of 12 Families with Type 1 Recessive Cutis Laxa. Human Mutation, 2013, 34, 111-121.	2.5	67
103	Whole exome sequencing is an efficient, sensitive and specific method of mutation detection in osteogenesis imperfecta and Marfan syndrome. BoneKEy Reports, 2013, 2, 456.	2.7	24
104	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
105	Dermatosparaxis (<scp>E</scp>hlersâ€“<scp>D</scp>anlos Type <scp>VIIC</scp>): Prenatal Diagnosis Following a Previous Pregnancy With Unexpected Skull Fractures at Delivery. American Journal of Medical Genetics, Part A, 2013, 161, 1122-1125.	1.2	19
106	Zebrafish models for ectopic mineralization disorders: practical issues from morpholino design to post-injection observations. Frontiers in Genetics, 2013, 4, 74.	2.3	13
107	Genes in Thoracic Aortic Aneurysms and Dissections - Do they Matter?: Translation and Integration of Research and Modern Genetic Techniques into Daily Clinical Practice. Aorta, 2013, 1, 135-145.	0.5	3
108	New insights into the molecular diagnosis and management of heritable thoracic aortic aneurysms and dissections. Polish Archives of Internal Medicine, 2013, 123, 693-700.	0.4	5

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109	First report of the genetic background of Marfan syndrome in Polish patients. Polish Archives of Internal Medicine, 2013, 123, 646-647.	0.4	2
110	GLUT10 is required for the development of the cardiovascular system and the notochord and connects mitochondrial function to TGFÎ² signaling. Human Molecular Genetics, 2012, 21, 1248-1259.	2.9	52
111	Stapes Surgery in Osteogenesis Imperfecta: Retrospective Analysis of 34 Operated Ears. Audiology and Neuro-Otology, 2012, 17, 198-206.	1.3	19
112	Phenotypic spectrum of the SMAD3-related aneurysmsâ€“osteoarthritis syndrome. Journal of Medical Genetics, 2012, 49, 47-57.	3.2	221
113	Audiologic Phenotype of Osteogenesis Imperfecta. Otology and Neurotology, 2012, 33, 115-122.	1.3	25
114	The Ghent Marfan Trial â€" A randomized, double-blind placebo controlled trial with losartan in Marfan patients treated with Î²-blockers. International Journal of Cardiology, 2012, 157, 354-358.	1.7	59
115	Molecular diagnostics for congenital hearing loss including 15 deafness genes using a next generation sequencing platform. BMC Medical Genomics, 2012, 5, 17.	1.5	49
116	Aggressive Cardiovascular Phenotype of Aneurysms-Osteoarthritis Syndrome Caused by Pathogenic SMAD3 Variants. Journal of the American College of Cardiology, 2012, 60, 397-403.	2.8	135
117	A Mutation in CABP2 , Expressed in Cochlear Hair Cells, Causes Autosomal-Recessive Hearing Impairment. American Journal of Human Genetics, 2012, 91, 636-645.	6.2	96
118	Characterization of a distinct lethal arteriopathy syndrome in twenty-two infants associated with an identical, novel mutation in FBLN4 gene, confirms fibulin-4 as a critical determinant of human vascular elastogenesis. Orphanet Journal of Rare Diseases, 2012, 7, 61.	2.7	38
119	Association between bone mineral density and hearing loss in osteogenesis imperfecta. Laryngoscope, 2012, 122, 401-408.	2.0	33
120	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
121	Spondyloperipheral dysplasia as the mosaic form of platyspondylic lethal skeletal dyplasia torrance type in mother and fetus with the same COL2A1 mutation. American Journal of Medical Genetics, Part A, 2012, 158A, 1948-1952.	1.2	14
122	Mutationâ€based growth charts for SEDC and other COL2A1 related dysplasias. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 205-216.	1.6	26
123	The new Ghent criteria for Marfan syndrome: what do they change?. Clinical Genetics, 2012, 81, 433-442.	2.0	90
124	Identification of binding partners interacting with the Î±1-N-propeptide of typeÂV collagen. Biochemical Journal, 2011, 433, 371-381.	3.7	49
125	Functional Polymorphism in Gamma-Glutamylcarboxylase is a Risk Factor for Severe Neonatal Hemorrhage. Journal of Pediatrics, 2011, 159, 347-349.	1.8	5
126	Osteogenesis imperfecta: the audiological phenotype lacks correlation with the genotype. Orphanet Journal of Rare Diseases, 2011, 6, 88.	2.7	48

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127	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
128	Massive parallel amplicon sequencing of the breast cancer genes BRCA1 and BRCA2: opportunities, challenges, and limitations. Human Mutation, 2011, 32, 335-344.	2.5	58
129	New insights into the pathogenesis of autosomalâ€dominant cutis laxa with report of fiveELNmutations. Human Mutation, 2011, 32, 445-455.	2.5	116
130	Applying massive parallel sequencing to molecular diagnosis of Marfan and Loeys-Dietz syndromes. Human Mutation, 2011, 32, 1053-1062.	2.5	71
131	Practical Tools to Implement Massive Parallel Pyrosequencing of PCR Products in Next Generation Molecular Diagnostics. PLoS ONE, 2011, 6, e25531.	2.5	40
132	Analysing 454 amplicon resequencing experiments using the modular and database oriented Variant Identification Pipeline. BMC Bioinformatics, 2010, 11, 269.	2.6	15
133	Genetic screening of LCA in Belgium: predominance of CEP290 and identification of potential modifier alleles in AHI1 of CEP290-related phenotypes. Human Mutation, 2010, 31, E1709-E1766.	2.5	127
134	Cytogenetic and arrayâ€CGH characterization of a 6q27 deletion in a patient with developmental delay and features of Ehlersâ€“Danlos syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 1314-1317.	1.2	10
135	Stickler syndrome caused by COL2A1 mutations: genotypeâ€“phenotype correlation in a series of 100 patients. European Journal of Human Genetics, 2010, 18, 872-880.	2.8	114
136	Altered TGFÎ² signaling and cardiovascular manifestations in patients with autosomal recessive cutis laxa type I caused by fibulin-4 deficiency. European Journal of Human Genetics, 2010, 18, 895-901.	2.8	132
137	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
138	Mutations in Fibrillin-1 Cause Congenital Scleroderma: Stiff Skin Syndrome. Science Translational Medicine, 2010, 2, 23ra20.	12.4	195
139	Short stature, severe aortic root dilation, skin hyperextensibility, extreme joint laxity and craniofacial dysmorphic features: a probable new syndrome. Clinical Dysmorphology, 2010, 19, 119-122.	0.3	0
140	Added value of infrared, red-free and autofluorescence fundus imaging in pseudoxanthoma elasticum. British Journal of Ophthalmology, 2010, 94, 479-486.	3.9	32
141	Novel deletions causing pseudoxanthoma elasticum underscore the genomic instability of the ABCC6 region. Journal of Human Genetics, 2010, 55, 112-117.	2.3	29
142	Clinical and Molecular Study of 320 Children With Marfan Syndrome and Related Type I Fibrillinopathies in a Series of 1009 Probands With Pathogenic FBN1 Mutations. Pediatrics, 2009, 123, 391-398.	2.1	146
143	Pathogenic FBN1 mutations in 146 adults not meeting clinical diagnostic criteria for Marfan syndrome: Further delineation of type 1 fibrillinopathies and focus on patients with an isolated major criterion. American Journal of Medical Genetics, Part A, 2009, 149A, 854-860.	1.2	40
144	Comprehensive clinical and molecular assessment of 32 probands with congenital contractural arachnodactyly: Report of 14 novel mutations and review of the literature. Human Mutation, 2009, 30, 334-341.	2.5	81

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145	COL5A1 signal peptide mutations interfere with protein secretion and cause classic Ehlers-Danlos syndrome. Human Mutation, 2009, 30, E395-E403.	2.5	57
146	Audiometric, surgical, and genetic findings in 15 ears of patients with osteogenesis imperfecta. Laryngoscope, 2009, 119, 1171-1179.	2.0	40
147	Clinical and mutation-type analysis from an international series of 198 probands with a pathogenic FBN1 exons 24â€“32 mutation. European Journal of Human Genetics, 2009, 17, 491-501.	2.8	66
148	Recessive osteogenesis imperfecta caused by LEPRE1 mutations: clinical documentation and identification of the splice form responsible for prolyl 3-hydroxylation. Journal of Medical Genetics, 2009, 46, 233-241.	3.2	77
149	Absence of arterial phenotype in mice with homozygous slc2A10 missense substitutions. Genesis, 2008, 46, 385-389.	1.6	30
150	Arterial tortuosity syndrome: clinical and molecular findings in 12 newly identified families. Human Mutation, 2008, 29, 150-158.	2.5	295
151	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
152	A genome-wide linkage scan for low spinal bone mineral density in a single extended family confirms linkage to 1p36.3. European Journal of Human Genetics, 2008, 16, 970-976.	2.8	7
153	Contribution of molecular analyses in diagnosing Marfan syndrome and type I fibrillinopathies: an international study of 1009 probands. Journal of Medical Genetics, 2008, 45, 384-390.	3.2	83
154	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
155	Pseudoxanthoma Elasticum with Generalized Retinal Dysfunction, a Common Finding?. , 2007, 48, 4250.		28
156	Recurrent Mutation in the First Zinc Finger of the Orphan Nuclear Receptor NR2E3 Causes Autosomal Dominant Retinitis Pigmentosa. American Journal of Human Genetics, 2007, 81, 147-157.	6.2	110
157	Effect of Mutation Type and Location on Clinical Outcome in 1,013 Probands with Marfan Syndrome or Related Phenotypes and FBN1 Mutations: An International Study. American Journal of Human Genetics, 2007, 81, 454-466.	6.2	485
158	COL2A1â€“related skeletal dysplasias with predominant metaphyseal involvement. American Journal of Medical Genetics, Part A, 2007, 143A, 161-167.	1.2	32
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