# 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 | A syndrome of altered cardiovascular, craniofacial, neurocognitive and skeletal development caused by mutations in TGFBR1 or TGFBR2. Nature Genetics, 2005, 37, 275-281. | 21.4 | 1,543 |
| 2 | Aneurysm Syndromes Caused by Mutations in the TGF-Î² Receptor. New England Journal of Medicine, 2006, 355, 788-798. | 27.0 | 1,490 |
| 3 | Consortium for osteogenesis imperfecta mutations in the helical domain of type I collagen: regions rich in lethal mutations align with collagen binding sites for integrins and proteoglycans. Human Mutation, 2007, 28, 209-221. | 2.5 | 620 |
| 4 | 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 |
| 5 | Loss-of-function mutations in LEMD3 result in osteopoikilosis, Buschke-Ollendorff syndrome and melorheostosis. Nature Genetics, 2004, 36, 1213-1218. | 21.4 | 410 |
| 6 | Nephrocystin-5, a ciliary IQ domain protein, is mutated in Senior-Loken syndrome and interacts with RPGR and calmodulin. Nature Genetics, 2005, 37, 282-288. | 21.4 | 367 |
| 7 | Mutations in the facilitative glucose transporter GLUT10 alter angiogenesis and cause arterial tortuosity syndrome. Nature Genetics, 2006, 38, 452-457. | 21.4 | 354 |
| 8 | Mutations in the human Î±-tectorin gene cause autosomal dominant non-syndromic hearing impairment. Nature Genetics, 1998, 19, 60-62. | 21.4 | 323 |
| 9 | Arterial tortuosity syndrome: clinical and molecular findings in 12 newly identified families. Human Mutation, 2008, 29, 150-158. | 2.5 | 295 |
| 10 | Homozygosity for a missense mutation in fibulin-5 (FBLN5) results in a severe form of cutis laxa. Human Molecular Genetics, 2002, 11, 2113-2118. | 2.9 | 283 |
| 11 | Spectrum of mutations in fucosidosis. European Journal of Human Genetics, 1999, 7, 60-67. | 2.8 | 221 |
| 12 | Phenotypic spectrum of the SMAD3-related aneurysmsâ€“osteoarthritis syndrome. Journal of Medical Genetics, 2012, 49, 47-57. | 3.2 | 221 |
| 13 | Comprehensive molecular screening of theFBN1gene favors locus homogeneity of classical Marfan syndrome. Human Mutation, 2004, 24, 140-146. | 2.5 | 210 |
| 14 | Mutations in Fibrillin-1 Cause Congenital Scleroderma: Stiff Skin Syndrome. Science Translational Medicine, 2010, 2, 23ra20. | 12.4 | 195 |
| 15 | Two Frequent Missense Mutations in Pendred Syndrome. Human Molecular Genetics, 1998, 7, 1099-1104. | 2.9 | 174 |
| 16 | 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 |
| 17 | MASA syndrome is due to mutations in the neural cell adhesion gene L1CAM. Nature Genetics, 1994, 7, 408-413. | 21.4 | 165 |
| 18 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Mutations in the KCNQ4 gene are responsible for autosomal dominant deafness in four DFNA2 families. Human Molecular Genetics, 1999, 8, 1321-1328. | 2.9 | 154 |
| 20 | 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 |
| 21 | Three arginine to cysteine substitutions in the pro-alpha (I)-collagen chain cause Ehlers-Danlos syndrome with a propensity to arterial rupture in early adulthood. Human Mutation, 2007, 28, 387-395. | 2.5 | 139 |
| 22 | Linkage of Autosomal Dominant Hearing Loss to the Short Arm of Chromosome 1 in Two Families. New England Journal of Medicine, 1994, 331, 425-431. | 27.0 | 137 |
| 23 | 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 |
| 24 | 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 |
| 25 | 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 |
| 26 | 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 |
| 27 | 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 |
| 28 | The molecular basis of classic Ehlers-Danlos syndrome: A comprehensive study of biochemical and molecular findings in 48 unrelated patients. Human Mutation, 2005, 25, 28-37. | 2.5 | 117 |
| 29 | New insights into the pathogenesis of autosomalâ€dominant cutis laxa with report of fiveELNmutations. Human Mutation, 2011, 32, 445-455. | 2.5 | 116 |
| 30 | A mutation update on the LDS-associated genesTGFB2/3andSMAD2/3. Human Mutation, 2018, 39, 621-634. | 2.5 | 116 |
| 31 | 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 |
| 32 | Homozygous Mutations in IHH Cause Acrocapitofemoral Dysplasia, an Autosomal Recessive Disorder with Cone-Shaped Epiphyses in Hands and Hips. American Journal of Human Genetics, 2003, 72, 1040-1046. | 6.2 | 113 |
| 33 | Localization of a gene for otosclerosis to chromosome 15q25-q26. Human Molecular Genetics, 1998, 7, 285-290. | 2.9 | 112 |
| 34 | 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 |
| 35 | 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 |
| 36 | Hearing loss in Waardenburg syndrome: a systematic review. Clinical Genetics, 2016, 89, 416-425. | 2.0 | 98 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | 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 |
| 38 | 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 |
| 39 | Total absence of the Â2(I) chain of collagen type I causes a rare form of Ehlers-Danlos syndrome with hypermobility and propensity to cardiac valvular problems. Journal of Medical Genetics, 2005, 43, e36-e36. | 3.2 | 95 |
| 40 | A duplication in the L1CAM gene associated with Xâ€“linked hydrocephalus. Nature Genetics, 1993, 4, 421-425. | 21.4 | 91 |
| 41 | A Second Gene for Otosclerosis, OTSC2, Maps to Chromosome 7q34-36. American Journal of Human Genetics, 2001, 68, 495-500. | 6.2 | 91 |
| 42 | The new Ghent criteria for Marfan syndrome: what do they change?. Clinical Genetics, 2012, 81, 433-442. | 2.0 | 90 |
| 43 | Localization of a gene for non-syndromic hearing loss (DFNA5) to chromosome 7p15. Human Molecular Genetics, 1995, 4, 2159-2163. | 2.9 | 89 |
| 44 | Linkage of a gene for dominant non-syndromic deafness to chromosome 19. Human Molecular Genetics, 1995, 4, 1073-1076. | 2.9 | 86 |
| 45 | Three new families with arterial tortuosity syndrome. American Journal of Medical Genetics Part A, 2004, 131A, 134-143. | 2.4 | 85 |
| 46 | Fibulin-5 mutations: mechanisms of impaired elastic fiber formation in recessive cutis laxa. Human Molecular Genetics, 2006, 15, 3379-3386. | 2.9 | 84 |
| 47 | 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 |
| 48 | 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 |
| 49 | BATCH-GE: Batch analysis of Next-Generation Sequencing data for genome editing assessment. Scientific Reports, 2016, 6, 30330. | 3.3 | 82 |
| 50 | Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa. American Journal of Human Genetics, 2017, 100, 216-227. | 6.2 | 82 |
| 51 | 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 |
| 52 | 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 |
| 53 | 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 |
| 54 | 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 |

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|---|---|---|---|
| 55 | 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 |
| 56 | GermlineLEMD3 mutations are rare in sporadic patients with isolated melorheostosis. Human Mutation, 2006, 27, 290-290. | 2.5 | 75 |
| 57 | Defective protein glycosylation in patients with cutis laxa syndrome. European Journal of Human Genetics, 2005, 13, 414-421. | 2.8 | 74 |
| 58 | Zebrafish: A Resourceful Vertebrate Model to Investigate Skeletal Disorders. Frontiers in Endocrinology, 2020, 11, 489. | 3.5 | 74 |
| 59 | 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 |
| 60 | 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 |
| 61 | The phenotypic spectrum in patients with arginine to cysteine mutations in the COL2A1 gene. Journal of Medical Genetics, 2005, 43, 406-413. | 3.2 | 71 |
| 62 | Applying massive parallel sequencing to molecular diagnosis of Marfan and Loeys-Dietz syndromes. Human Mutation, 2011, 32, 1053-1062. | 2.5 | 71 |
| 63 | Radiological sacroiliitis, a hallmark of spondylitis, is linked with CARD15 gene polymorphisms in patients with Crohn's disease. Annals of the Rheumatic Diseases, 2004, 63, 1131-1134. | 0.9 | 70 |
| 64 | IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260. | 6.2 | 69 |
| 65 | Comprehensive Clinical and Molecular Analysis of 12 Families with Type 1 Recessive Cutis Laxa. Human Mutation, 2013, 34, 111-121. | 2.5 | 67 |
| 66 | 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 |
| 67 | Arterial tortuosity syndrome: 40 new families and literature review. Genetics in Medicine, 2018, 20, 1236-1245. | 2.4 | 66 |
| 68 | 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 |
| 69 | 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 |
| 70 | 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 |
| 71 | MicroCT-based phenomics in the zebrafish skeleton reveals virtues of deep phenotyping in a distributed organ system. ELife, 2017, 6, . | 6.0 | 60 |
| 72 | Mutations in the KCNQ4 K+ channel gene, responsible for autosomal dominant hearing loss, cluster in the channel pore region. American Journal of Medical Genetics Part A, 2000, 93, 184-187. | 2.4 | 59 |

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|---|---|---|---|
| 73 | 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 |
| 74 | RNF216 mutations as a novel cause of autosomal recessive Huntington-like disorder. Neurology, 2015, 84, 1760-1766. | 1.1 | 59 |
| 75 | 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 |
| 76 | 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 |
| 77 | COL5A1 signal peptide mutations interfere with protein secretion and cause classic Ehlers-Danlos syndrome. Human Mutation, 2009, 30, E395-E403. | 2.5 | 57 |
| 78 | Linkage Analysis of Progressive Hearing Loss in Five Extended Families Maps the DFNA2 Gene to a 1.25-Mb Region on Chromosome 1p. Genomics, 1997, 41, 70-74. | 2.9 | 52 |
| 79 | 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 |
| 80 | Consanguineous nuclear families used to identify a new locus for recessive non-syndromic hearing loss on 14q. Human Molecular Genetics, 1995, 4, 1643-1648. | 2.9 | 51 |
| 81 | The Genetics of Soft Connective Tissue Disorders. Annual Review of Genomics and Human Genetics, 2015, 16, 229-255. | 6.2 | 50 |
| 82 | Identification of binding partners interacting with the Î±1-N-propeptide of typeÂV collagen. Biochemical Journal, 2011, 433, 371-381. | 3.7 | 49 |
| 83 | 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 |
| 84 | A mutational hot spot in theKCNQ4 gene responsible for autosomal dominant hearing impairment. Human Mutation, 2002, 20, 15-19. | 2.5 | 48 |
| 85 | Osteogenesis imperfecta: the audiological phenotype lacks correlation with the genotype. Orphanet Journal of Rare Diseases, 2011, 6, 88. | 2.7 | 48 |
| 86 | 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 |
| 87 | Longitudinal and Cross-Sectional Phenotype Analysis in a New, Large Dutch DFNA2/KCNQ4 Family. Annals of Otology, Rhinology and Laryngology, 2002, 111, 267-274. | 1.1 | 47 |
| 88 | Utility of molecular analyses in the exploration of extreme intrafamilial variability in the Marfan syndrome. Clinical Genetics, 2007, 72, 188-198. | 2.0 | 47 |
| 89 | Localization of a novel gene for nonsyndromic hearing loss (DFNB17) to chromosome region 7q31. American Journal of Medical Genetics Part A, 1998, 78, 107-113. | 2.4 | 45 |
| 90 | 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 |

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|---|---|---|---|
| 91 | Inherited Nonsyndromic Hearing Loss: An Audiovestibular Study in a Large Family With Autosomal Dominant Progressive Hearing Loss Related to DFNA2. JAMA Otolaryngology, 1997, 123, 573-577. | 1.2 | 43 |
| 92 | 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 |
| 93 | 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 |
| 94 | Czech dysplasia metatarsal type: another type II collagen disorder. European Journal of Human Genetics, 2007, 15, 1269-1275. | 2.8 | 41 |
| 95 | 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 |
| 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 | 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 |
| 98 | Audiometric, surgical, and genetic findings in 15 ears of patients with osteogenesis imperfecta. Laryngoscope, 2009, 119, 1171-1179. | 2.0 | 40 |
| 99 | Practical Tools to Implement Massive Parallel Pyrosequencing of PCR Products in Next Generation Molecular Diagnostics. PLoS ONE, 2011, 6, e25531. | 2.5 | 40 |
| 100 | A Gene for Fluctuating, Progressive Autosomal Dominant Nonsyndromic Hearing Loss, DFNA16, Maps to Chromosome 2q23-24.3. American Journal of Human Genetics, 1999, 65, 141-150. | 6.2 | 39 |
| 101 | Genetic Defects in TAPT1 Disrupt Ciliogenesis and Cause a Complex Lethal Osteochondrodysplasia. American Journal of Human Genetics, 2015, 97, 521-534. | 6.2 | 39 |
| 102 | X-linked liver glycogenosis type II (XLG II) is caused by mutations in PHKA2, the gene encoding the liver alpha subunit of phosphorylase kinase. Human Molecular Genetics, 1996, 5, 649-652. | 2.9 | 38 |
| 103 | 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 |
| 104 | Expressed Repeat Elements Improve RT-qPCR Normalization across a Wide Range of Zebrafish Gene Expression Studies. PLoS ONE, 2014, 9, e109091. | 2.5 | 38 |
| 105 | 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 |
| 106 | Phenomics-Based Quantification of CRISPR-Induced Mosaicism in Zebrafish. Cell Systems, 2020, 10, 275-286.e5. | 6.2 | 38 |
| 107 | Deafness linked to DFNA2: one locus but how many genes?. Nature Genetics, 1999, 21, 263-263. | 21.4 | 35 |
| 108 | Perturbation of specific pro-mineralizing signalling pathways in human and murine pseudoxanthoma elasticum. Orphanet Journal of Rare Diseases, 2014, 9, 66. | 2.7 | 35 |

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|---|---|---|---|
| 109 | A combined defect in the biosynthesis of N- and O-glycans in patients with cutis laxa and neurological involvement: the biochemical characteristics. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2005, 1741, 156-164. | 3.8 | 34 |
| 110 | Association between bone mineral density and hearing loss in osteogenesis imperfecta. Laryngoscope, 2012, 122, 401-408. | 2.0 | 33 |
| 111 | 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 |
| 112 | COL2A1â€“related skeletal dysplasias with predominant metaphyseal involvement. American Journal of Medical Genetics, Part A, 2007, 143A, 161-167. | 1.2 | 32 |
| 113 | Added value of infrared, red-free and autofluorescence fundus imaging in pseudoxanthoma elasticum. British Journal of Ophthalmology, 2010, 94, 479-486. | 3.9 | 32 |
| 114 | CARD15 polymorphisms are associated with anti-Saccharomyces cerevisiae antibodies in caucasian Crohn's disease patients. Clinical and Experimental Immunology, 2005, 140, 354-359. | 2.6 | 30 |
| 115 | Absence of arterial phenotype in mice with homozygous slc2A10 missense substitutions. Genesis, 2008, 46, 385-389. | 1.6 | 30 |
| 116 | The Gene for Pendred Syndrome Is Located between D7S501 and D7S692 in a 1.7-cM Region on Chromosome 7q. Genomics, 1997, 40, 48-54. | 2.9 | 29 |
| 117 | Novel deletions causing pseudoxanthoma elasticum underscore the genomic instability of the ABCC6 region. Journal of Human Genetics, 2010, 55, 112-117. | 2.3 | 29 |
| 118 | Congenital Fixed Dilated Pupils Due to ACTA2â€“ Multisystemic Smooth Muscle Dysfunction Syndrome. Journal of Neuro-Ophthalmology, 2014, 34, 137-143. | 0.8 | 29 |
| 119 | Altered cytoskeletal organization characterized lethal but not surviving Brtl<sup>+/â^'</sup>mice: insight on phenotypic variability in osteogenesis imperfecta. Human Molecular Genetics, 2015, 24, 6118-6133. | 2.9 | 29 |
| 120 | Missense Mutations in LRP5 Are Not a Common Cause of Idiopathic Osteoporosis in Adult Men. Journal of Bone and Mineral Research, 2005, 20, 1951-1959. | 2.8 | 28 |
| 121 | Pseudoxanthoma Elasticum with Generalized Retinal Dysfunction, a Common Finding?. , 2007, 48, 4250. | | 28 |
| 122 | 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 |
| 123 | Pathogenic variants in the ABCC6 gene are associated with an increased risk for ischemic stroke. Brain Pathology, 2018, 28, 822-831. | 4.1 | 28 |
| 124 | Maximizing CRISPR/Cas9 phenotype penetrance applying predictive modeling of editing outcomes in Xenopus and zebrafish embryos. Scientific Reports, 2020, 10, 14662. | 3.3 | 28 |
| 125 | Functional haplotypes of PADI4: relevance for rheumatoid arthritis specific synovial intracellular citrullinated proteins and anticitrullinated protein antibodies. Annals of the Rheumatic Diseases, 2005, 64, 1316-1320. | 0.9 | 26 |
| 126 | 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 |

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|---|---|---|---|
| 127 | 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 |
| 128 | Nonsyndromic Autosomal Dominant Progressive Sensorineural Hearing Loss: Audiologic Analysis of a Pedigree Linked to DFNA2. Laryngoscope, 1998, 108, 74-80. | 2.0 | 25 |
| 129 | Audiologic Phenotype of Osteogenesis Imperfecta. Otology and Neurotology, 2012, 33, 115-122. | 1.3 | 25 |
| 130 | Efficiency of Exome Sequencing for the Molecular Diagnosis of Pseudoxanthoma Elasticum. Journal of Investigative Dermatology, 2015, 135, 992-998. | 0.7 | 25 |
| 131 | Glucose transporter type 10â€"lacking in arterial tortuosity syndromeâ€"facilitates dehydroascorbic acid transport. FEBS Letters, 2016, 590, 1630-1640. | 2.8 | 25 |
| 132 | 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 |
| 133 | Exome sequencing revealed a novel biallelic deletion in theDCAF17gene underlying Woodhouse Sakati syndrome. Clinical Genetics, 2016, 90, 263-269. | 2.0 | 23 |
| 134 | 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 |
| 135 | Visceral and testicular calcifications as part of the phenotype in pseudoxanthoma elasticum: ultrasound findings in Belgian patients and healthy carriers. British Journal of Radiology, 2006, 79, 221-225. | 2.2 | 22 |
| 136 | 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 |
| 137 | Complementary Deoxyribonucleic Acid Cloning and Characterization of a Putative Human Axonemal Dynein Light Chain Gene. Journal of Clinical Endocrinology and Metabolism, 1997, 82, 3047-3053. | 3.6 | 21 |
| 138 | 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 |
| 139 | Molecular study of chromosome 15 in 22 patients with Angelman syndrome. Human Genetics, 1993, 90, 489-495. | 3.8 | 19 |
| 140 | Recurrence of achondrogenesis type II within the same family: Evidence for germline mosaicism. American Journal of Medical Genetics Part A, 2004, 126A, 308-312. | 2.4 | 19 |
| 141 | Stapes Surgery in Osteogenesis Imperfecta: Retrospective Analysis of 34 Operated Ears. Audiology and Neuro-Otology, 2012, 17, 198-206. | 1.3 | 19 |
| 142 | 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 |
| 143 | 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 |
| 144 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 145 | Hypomorphic zebrafish models mimic the musculoskeletal phenotype of Î²4GalT7-deficient Ehlers-Danlos syndrome. Matrix Biology, 2020, 89, 59-75. | 3.6 | 19 |
| 146 | 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 |
| 147 | Generation and Validation of a Complete Knockout Model of abcc6a in Zebrafish. Journal of Investigative Dermatology, 2018, 138, 2333-2342. | 0.7 | 18 |
| 148 | A clinical scoring system for congenital contractural arachnodactyly. Genetics in Medicine, 2020, 22, 124-131. | 2.4 | 17 |
| 149 | Reassessment of causality of ABCC6 missense variants associated with pseudoxanthoma elasticum based on Sherloc. Genetics in Medicine, 2021, 23, 131-139. | 2.4 | 17 |
| 150 | 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 |
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