

Wim Wuyts

List of Publications by Year in descending order

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Version: 2024-02-01

82
papers

4,160
citations

136950

32
h-index

118850

62
g-index

84
all docs

84
docs citations

84
times ranked

7423
citing authors

#	ARTICLE	IF	CITATIONS
1	Domain-specific mutations in sequestosome 1 (SQSTM1) cause familial and sporadic Paget's disease. <i>Human Molecular Genetics</i> , 2002, 11, 2735-2739.	2.9	307
2	Prevalence and Evolutionary Origins of the del(GJB6-D13S1830) Mutation in the DFNB1 Locus in Hearing-Impaired Subjects: a Multicenter Study. <i>American Journal of Human Genetics</i> , 2003, 73, 1452-1458.	6.2	269
3	Molecular basis of multiple exostoses: mutations in the EXT1 and EXT2 genes. <i>Human Mutation</i> , 2000, 15, 220-227.	2.5	189
4	Multiple osteochondromas: mutation update and description of the multiple osteochondromas mutation database (MOdb). <i>Human Mutation</i> , 2009, 30, 1620-1627.	2.5	176
5	Mutations in the EXT1 and EXT2 Genes in Hereditary Multiple Exostoses. <i>American Journal of Human Genetics</i> , 1998, 62, 346-354.	6.2	174
6	EXT-Mutation Analysis and Loss of Heterozygosity in Sporadic and Hereditary Osteochondromas and Secondary Chondrosarcomas. <i>American Journal of Human Genetics</i> , 1999, 65, 689-698.	6.2	174
7	Two-color multiplex ligation-dependent probe amplification: Detecting genomic rearrangements in hereditary multiple exostoses. <i>Human Mutation</i> , 2004, 24, 86-92.	2.5	142
8	Identification of a Third EXT-like Gene (EXTL3) Belonging to the EXT Gene Family. <i>Genomics</i> , 1998, 47, 230-237.	2.9	124
9	Genomewide Search in Familial Paget Disease of Bone Shows Evidence of Genetic Heterogeneity with Candidate Loci on Chromosomes 2q36, 10p13, and 5q35. <i>American Journal of Human Genetics</i> , 2001, 69, 1055-1061.	6.2	113
10	Recurrent Mutation in the First Zinc Finger of the Orphan Nuclear Receptor NR2E3 Causes Autosomal Dominant Retinitis Pigmentosa. <i>American Journal of Human Genetics</i> , 2007, 81, 147-157.	6.2	110
11	Genotype-Phenotype Correlation Study in 529 Patients with Multiple Hereditary Exostoses: Identification of "Protective" and "Risk" Factors. <i>Journal of Bone and Joint Surgery - Series A</i> , 2011, 93, 2294-2302.	3.0	109
12	Loss-of-Function Mutations in PTPN11 Cause Metachondromatosis, but Not Ollier Disease or Maffucci Syndrome. <i>PLoS Genetics</i> , 2011, 7, e1002050.	3.5	104
13	tRNA Methyltransferase Homolog Gene TRMT10A Mutation in Young Onset Diabetes and Primary Microcephaly in Humans. <i>PLoS Genetics</i> , 2013, 9, e1003888.	3.5	103
14	Gain-of-Function Mutations of ARHGAP31, a Cdc42/Rac1 GTPase Regulator, Cause Syndromic Cutis Aplasia and Limb Anomalies. <i>American Journal of Human Genetics</i> , 2011, 88, 574-585.	6.2	100
15	Performant Mutation Identification Using Targeted Next-Generation Sequencing of 14 Thoracic Aortic Aneurysm Genes. <i>Human Mutation</i> , 2015, 36, 808-814.	2.5	97
16	Loss-of-function mutations in the X-linked biglycan gene cause a severe syndromic form of thoracic aortic aneurysms and dissections. <i>Genetics in Medicine</i> , 2017, 19, 386-395.	2.4	94
17	The spectra of clinical phenotypes in aplasia cutis congenita and terminal transverse limb defects. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1860-1881.	1.2	92
18	Hereditary multiple exostoses: from genetics to clinical syndrome and complications. <i>European Journal of Radiology</i> , 2001, 40, 208-217.	2.6	88

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19	Haploinsufficiency of the NOTCH1 Receptor as a Cause of Adams-Oliver Syndrome With Variable Cardiac Anomalies. <i>Circulation: Cardiovascular Genetics</i> , 2015, 8, 572-581.	5.1	84
20	Identification and Characterization of a Novel Member of the EXT Gene Family, EXTL2. <i>European Journal of Human Genetics</i> , 1997, 5, 382-389.	2.8	77
21	Localization of a Gene for Autosomal Dominant Osteopetrosis (Albers-Schönberg Disease) to Chromosome 1p21. <i>American Journal of Human Genetics</i> , 1997, 61, 363-369.	6.2	76
22	DNA Diagnostics of Hereditary Hearing Loss: A Targeted Resequencing Approach Combined with a Mutation Classification System. <i>Human Mutation</i> , 2016, 37, 812-819.	2.5	76
23	Expanding the Spectrum of <i>FOXC1</i> and <i>PITX2</i> Mutations and Copy Number Changes in Patients with Anterior Segment Malformations. , 2011, 52, 324.		73
24	Heterozygous Loss-of-Function Mutations in <i>DLL4</i> Cause Adams-Oliver Syndrome. <i>American Journal of Human Genetics</i> , 2015, 97, 475-482.	6.2	73
25	CNV-WebStore: Online CNV Analysis, Storage and Interpretation. <i>BMC Bioinformatics</i> , 2011, 12, 4.	2.6	54
26	Tiling resolution array-CGH shows that somatic mosaic deletion of the EXT gene is causative in EXT gene mutation negative multiple osteochondromas patients. <i>Human Mutation</i> , 2011, 32, E2036-E2049.	2.5	50
27	Mutation Screening of <i>EXT1</i> and <i>EXT2</i> by Denaturing High-Performance Liquid Chromatography, Direct Sequencing Analysis, Fluorescence in Situ Hybridization, and a New Multiplex Ligation-Dependent Probe Amplification Probe Set in Patients with Multiple Osteochondromas. <i>Journal of Molecular Diagnostics</i> , 2008, 10, 85-92.	2.8	46
28	A subterminal deletion of the long arm of chromosome 10: A clinical report and review. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 402-409.	1.2	45
29	Clinical and molecular analysis of nine families with Adams-Oliver syndrome. <i>European Journal of Human Genetics</i> , 2003, 11, 457-463.	2.8	44
30	A combined analytical approach reveals novel <i>EXT1/2</i> gene mutations in a large cohort of Italian multiple osteochondromas patients. <i>Genes Chromosomes and Cancer</i> , 2007, 46, 470-477.	2.8	43
31	Multiple exostoses, mental retardation, hypertrichosis, and brain abnormalities in a boy with a de novo 8q24 submicroscopic interstitial deletion. <i>American Journal of Medical Genetics Part A</i> , 2002, 113, 326-332.	2.4	42
32	Denaturing HPLC-Based Approach for Detecting <i>RYR2</i> Mutations Involved in Malignant Arrhythmias. <i>Clinical Chemistry</i> , 2004, 50, 1148-1155.	3.2	41
33	Detection of Microsatellite Instability in Colorectal Cancer Using an Alternative Multiplex Assay of Quasi-Monomorphic Mononucleotide Markers. <i>Journal of Molecular Diagnostics</i> , 2008, 10, 154-159.	2.8	40
34	Somatic and gonadal mosaicism in Hutchinson-Gilford progeria. <i>American Journal of Medical Genetics, Part A</i> , 2005, 135A, 66-68.	1.2	38
35	<i>DOCK6</i> Mutations Are Responsible for a Distinct Autosomal-Recessive Variant of Adams-Oliver Syndrome Associated with Brain and Eye Anomalies. <i>Human Mutation</i> , 2015, 36, 593-598.	2.5	32
36	Mutation analysis of mitochondrial DNA 12SrRNA and tRNASer(UCN) genes in non-syndromic hearing loss patients. <i>Mitochondrion</i> , 2008, 8, 377-382.	3.4	31

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37	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. <i>Human Mutation</i> , 2018, 39, 1246-1261.	2.5	31
38	Hypoparathyroidism-retardation-dysmorphism syndrome in a girl: A new variant not caused by a TBCE mutation-clinical report and review. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 611-617.	1.2	30
39	Myopathy and phosphorylase kinase deficiency caused by a mutation in the PHKA1 gene. <i>American Journal of Medical Genetics, Part A</i> , 2005, 133A, 82-84.	1.2	29
40	Molecular and clinical examination of an Italian DEFECT 11 family. <i>European Journal of Human Genetics</i> , 1999, 7, 579-584.	2.8	27
41	Prenatal diagnosis of Pfeiffer syndrome type II. <i>Prenatal Diagnosis</i> , 2004, 24, 644-646.	2.3	27
42	GM1 gangliosidosis: molecular analysis of nine patients and development of an RT-PCR assay for GLB1 gene expression profiling. <i>Human Mutation</i> , 2007, 28, 204-204.	2.5	27
43	Breakpoint characterization of large deletions in EXT1 or EXT2 in 10 Multiple Osteochondromas families. <i>BMC Medical Genetics</i> , 2011, 12, 85.	2.1	26
44	An interstitial deletion of chromosome 7 at band q21: A case report and review. , 2005, 134A, 12-23.		23
45	Bi-allelic inactivating variants in the COCH gene cause autosomal recessive prelingual hearing impairment. <i>European Journal of Human Genetics</i> , 2018, 26, 587-591.	2.8	22
46	Molecular diagnostics for hereditary hearing loss in children. <i>Expert Review of Molecular Diagnostics</i> , 2017, 17, 751-760.	3.1	21
47	Proximal 11p deletion syndrome (P11pDS): additional evaluation of the clinical and molecular aspects. <i>European Journal of Human Genetics</i> , 2004, 12, 400-406.	2.8	19
48	KID Syndrome: Report of a Scandinavian Patient with Connexin 26 Gene Mutation. <i>Acta Dermato-Venereologica</i> , 2005, 85, 152-155.	1.3	19
49	Microsatellite instability in sporadic colon carcinomas has no independent prognostic value in a Belgian study population. <i>European Journal of Cancer</i> , 2008, 44, 2288-2295.	2.8	19
50	The <i>SLC40A1</i> R178Q mutation is a recurrent cause of hemochromatosis and is associated with a novel pathogenic mechanism. <i>Haematologica</i> , 2018, 103, 1796-1805.	3.5	19
51	Identification and functional characterization of the human EXT1 promoter region. <i>Gene</i> , 2012, 492, 148-159.	2.2	18
52	Intraosseous Atypical Chondroid Tumor or Chondrosarcoma Grade 1 in Patients with Multiple Osteochondromas. <i>Journal of Bone and Joint Surgery - Series A</i> , 2015, 97, 24-31.	3.0	18
53	Isolated terminal limb reduction defects: Extending the clinical spectrum of Adams-Oliver syndrome and <i>ARHGAP31</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1576-1579.	1.2	17
54	Novel <i>LMNA</i> mutations cause an aggressive atypical neonatal progeria without progerin accumulation. <i>Journal of Medical Genetics</i> , 2016, 53, 776-785.	3.2	17

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55	Adams's Oliver syndrome caused by mutations of the <i>EOGT</i> gene. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2246-2251.	1.2	17
56	RNA-seq detects a <i>SAMD12-EXT1</i> fusion transcript and leads to the discovery of an <i>EXT1</i> deletion in a child with multiple osteochondromas. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00560.	1.2	17
57	Carnitine-palmitoyltransferase 2 deficiency: Novel mutations and relevance of newborn screening. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 2925-2928.	1.2	15
58	Targeted Next-Generation Sequencing of 51 Genes Involved in Primary Electrical Disease. <i>Journal of Molecular Diagnostics</i> , 2017, 19, 445-459.	2.8	15
59	Fluomic assay-assisted diagnosis orientation in a cohort of 11 patients with myopathic form of CPT2 deficiency. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 441-448.	1.1	13
60	A New Pathogenic Variant in <i>POU3F4</i> Causing Deafness Due to an Incomplete Partition of the Cochlea Paved the Way for Innovative Surgery. <i>Genes</i> , 2021, 12, 613.	2.4	13
61	Comparison of three commonly used PCR-based techniques to analyze MSI status in sporadic colorectal cancer. <i>Journal of Clinical Laboratory Analysis</i> , 2006, 20, 52-61.	2.1	12
62	A novel mutation in the <i>MSX2</i> gene in a family with foramina parietalia permagna (FPP). <i>American Journal of Medical Genetics, Part A</i> , 2005, 139A, 45-47.	1.2	11
63	Burning down <i>DEFECT11</i> . <i>American Journal of Medical Genetics Part A</i> , 2001, 100, 331-332.	2.4	10
64	Role of Targeted Next Generation Sequencing in the Etiological Work-Up of Congenitally Deaf Children. <i>Otology and Neurotology</i> , 2018, 39, 732-738.	1.3	10
65	<i>ABCC8</i> variants in <i>MODY12</i> : Review of the literature and report of a case with severe complications. <i>Diabetes/Metabolism Research and Reviews</i> , 2021, 37, e3459.	4.0	10
66	Solitary thyroid nodule as presenting symptom of Pendred syndrome caused by a novel splice-site mutation in intron 8 of the <i>SLC26A4</i> gene. <i>European Journal of Pediatrics</i> , 2003, 162, 674-677.	2.7	9
67	A de novo subterminal trisomy 10p and monosomy 18q in a girl with MCA/MR: case report and review. <i>European Journal of Medical Genetics</i> , 2006, 49, 402-413.	1.3	8
68	Clinical and molecular studies of <i>EXT1/EXT2</i> in Bulgaria. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 917-921.	3.6	8
69	Etiological Work-up in Referrals From Neonatal Hearing Screening: 20 Years of Experience. <i>Otology and Neurotology</i> , 2020, 41, 1240-1248.	1.3	8
70	Genetic Defects in the Development of the Skull Vault in Humans and Mice. <i>Critical Reviews in Eukaryotic Gene Expression</i> , 2006, 16, 119-142.	0.9	8
71	A de novo subtelomeric monosomy 11q (11q24.2-qter) and trisomy 20q (20q13.3-qter) in a girl with findings compatible with Jacobsen syndrome: case report and review. <i>Clinical Dysmorphology</i> , 2007, 16, 231-239.	0.3	7
72	High incidence of the <i>CFTR</i> mutations 3272-26A>G and L927P in Belgian cystic fibrosis patients, and identification of three new <i>CFTR</i> mutations (186-2A>G, E588V, and 1671insTATCA). <i>Journal of Cystic Fibrosis</i> , 2007, 6, 371-375.	0.7	5

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73	A new double substitution mutation in the MEN1 gene: a limited penetrance and a specific phenotype. <i>European Journal of Human Genetics</i> , 2013, 21, 695-697.	2.8	5
74	A New Pathogenic Variant in the TRIOBP Associated with Profound Deafness Is Remediable with Cochlear Implantation. <i>Audiology and Neuro-Otology</i> , 2020, 26, 1-9.	1.3	4
75	Phenotypic Differences in Multiple Osteochondromas in Monozygotic Twins. <i>JBJS Case Connector</i> , 2012, 2, e60.	0.3	2
76	Trichorhinophalangeal syndrome type II presenting with short stature in a child. <i>Archivos Argentinos De Pediatria</i> , 2016, 114, e403-e407.	0.2	2
77	Organization of the Ribosomal Operon 16S-23S Gene Spacer Region in Representatives of <i>Neisseria gonorrhoeae</i> . <i>Systematic and Applied Microbiology</i> , 2000, 23, 9-14.	2.8	1
78	Recurrent post-infectious rhabdomyolysis in muscle CPT-II deficiency caused by a novel missense mutation. <i>Acta Neurologica Belgica</i> , 2008, 108, 155-60.	1.1	1
79	Response to letter to the editor: "Hypoparathyroidism-Retardation-Dysmorphism Syndrome in a Female Child: A New Variant Not Caused by a TBCE Mutation" Clinical Report and Review. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 303-304.	1.2	0
80	Erratum to "High incidence of the CFTR mutations 3272-26A → G and L927P in Belgian cystic fibrosis patients, and identification of three new CFTR mutations (186-2A → G, E588V, and 1671insTATCA)". <i>Journal of Cystic Fibrosis</i> 6(2007)371-375]. <i>Journal of Cystic Fibrosis</i> , 2008, 7, 461.	0.7	0
81	Frequency of Participation in External Quality Assessment Programs Focused on Rare Diseases: Belgian Guidelines for Human Genetics Centers. <i>JMIR Medical Informatics</i> , 2021, 9, e27980.	2.6	0
82	Abstract 2122:KRAS mutation detection using high resolution melting analysis and its prognostic value in archival colorectal cancer tissues. , 2010, , .		0