Wim Wuyts

List of Publications by Year in descending order

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	A New Pathogenic Variant in POU3F4 Causing Deafness Due to an Incomplete Partition of the Cochlea Paved the Way for Innovative Surgery. Genes, 2021, 12, 613.	2.4	13
2	ABCC8 variants in MODY12: Review of the literature and report of a case with severe complications. Diabetes/Metabolism Research and Reviews, 2021, 37, e3459.	4.0	10
3	Frequency of Participation in External Quality Assessment Programs Focused on Rare Diseases: Belgian Guidelines for Human Genetics Centers. JMIR Medical Informatics, 2021, 9, e27980.	2.6	0
4	Etiological Work-up in Referrals From Neonatal Hearing Screening: 20 Years of Experience. Otology and Neurotology, 2020, 41, 1240-1248.	1.3	8
5	A New Pathogenic Variant in the TRIOBP Associated with Profound Deafness Is Remediable with Cochlear Implantation. Audiology and Neuro-Otology, 2020, 26, 1-9.	1.3	4
6	Adamsâ€“Oliver syndrome caused by mutations of the EOGT gene. American Journal of Medical Genetics, Part A, 2019, 179, 2246-2251.	1.2	17
7	RNAâ€Seq detects a SAMD12â€EXT1 fusion transcript and leads to the discovery of an EXT1 deletion in a child with multiple osteochondromas. Molecular Genetics & Genomic Medicine, 2019, 7, e00560.	1.2	17
8	Bi-allelic inactivating variants in the COCH gene cause autosomal recessive prelingual hearing impairment. European Journal of Human Genetics, 2018, 26, 587-591.	2.8	22
9	Fluxomic assay-assisted diagnosis orientation in a cohort of 11 patients with myopathic form of CPT2 deficiency. Molecular Genetics and Metabolism, 2018, 123, 441-448.	1.1	13
10	The SLC40A1 R178Q mutation is a recurrent cause of hemochromatosis and is associated with a novel pathogenic mechanism. Haematologica, 2018, 103, 1796-1805.	3.5	19
11	Role of Targeted Next Generation Sequencing in the Etiological Work-Up of Congenitally Deaf Children. Otology and Neurotology, 2018, 39, 732-738.	1.3	10
12	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. Human Mutation, 2018, 39, 1246-1261.	2.5	31
13	Molecular diagnostics for hereditary hearing loss in children. Expert Review of Molecular Diagnostics, 2017, 17, 751-760.	3.1	21
14	Targeted Next-Generation Sequencing of 51 Genes Involved in Primary Electrical Disease. Journal of Molecular Diagnostics, 2017, 19, 445-459.	2.8	15
15	Loss-of-function mutations in the X-linked biglycan gene cause a severe syndromic form of thoracic aortic aneurysms and dissections. Genetics in Medicine, 2017, 19, 386-395.	2.4	94
16	Trichorhinophalangeal syndrome type II presenting with short stature in a child. Archivos Argentinos De Pediatria, 2016, 114, e403-e407.	0.2	2
17	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
18	NovelLMNAmutations cause an aggressive atypical neonatal progeria without progerin accumulation. Journal of Medical Genetics, 2016, 53, 776-785.	3.2	17

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19	Performant Mutation Identification Using Targeted Next-Generation Sequencing of 14 Thoracic Aortic Aneurysm Genes. Human Mutation, 2015, 36, 808-814.	2.5	97
20	Haploinsufficiency of the NOTCH1 Receptor as a Cause of Adamsâ€“Oliver Syndrome With Variable Cardiac Anomalies. Circulation: Cardiovascular Genetics, 2015, 8, 572-581.	5.1	84
21	Intraosseous Atypical Chondroid Tumor or Chondrosarcoma Grade 1 in Patients with Multiple Osteochondromas. Journal of Bone and Joint Surgery - Series A, 2015, 97, 24-31.	3.0	18
22	DOCK6Mutations Are Responsible for a Distinct Autosomal-Recessive Variant of Adams-Oliver Syndrome Associated with Brain and Eye Anomalies. Human Mutation, 2015, 36, 593-598.	2.5	32
23	Heterozygous Loss-of-Function Mutations in DLL4 Cause Adams-Oliver Syndrome. American Journal of Human Genetics, 2015, 97, 475-482.	6.2	73
24	Isolated terminal limb reduction defects: Extending the clinical spectrum of Adamsâ€“Oliver syndrome and ARHGAP31 mutations. American Journal of Medical Genetics, Part A, 2014, 164, 1576-1579.	1.2	17
25	A new double substitution mutation in the MEN1 gene: a limited penetrance and a specific phenotype. European Journal of Human Genetics, 2013, 21, 695-697.	2.8	5
26	tRNA Methyltransferase Homolog Gene TRMT10A Mutation in Young Onset Diabetes and Primary Microcephaly in Humans. PLoS Genetics, 2013, 9, e1003888.	3.5	103
27	Identification and functional characterization of the human EXT1 promoter region. Gene, 2012, 492, 148-159.	2.2	18
28	Phenotypic Differences in Multiple Osteochondromas in Monozygotic Twins. JBJS Case Connector, 2012, 2, e60.	0.3	2
29	Gain-of-Function Mutations of ARHGAP31, a Cdc42/Rac1 GTPase Regulator, Cause Syndromic Cutis Aplasia and Limb Anomalies. American Journal of Human Genetics, 2011, 88, 574-585.	6.2	100
30	Clinical and molecular studies of EXT1/EXT2 in Bulgaria. Journal of Inherited Metabolic Disease, 2011, 34, 917-921.	3.6	8
31	Breakpoint characterization of large deletions in EXT1 or EXT2 in 10 Multiple Osteochondromas families. BMC Medical Genetics, 2011, 12, 85.	2.1	26
32	CNV-WebStore: Online CNV Analysis, Storage and Interpretation. BMC Bioinformatics, 2011, 12, 4.	2.6	54
33	Tiling resolution array-CGH shows that somatic mosaic deletion of the EXT gene is causative in EXT gene mutation negative multiple osteochondromas patients. Human Mutation, 2011, 32, E2036-E2049.	2.5	50
34	Expanding the Spectrum ofFOXC1andPITX2Mutations and Copy Number Changes in Patients with Anterior Segment Malformations. , 2011, 52, 324.		73
35	Genotype-Phenotype Correlation Study in 529 Patients with Multiple Hereditary Exostoses: Identification of â€œProtectiveâ€•and â€œRiskâ€•Factors. Journal of Bone and Joint Surgery - Series A, 2011, 93, 2294-2302.	3.0	109
36	Loss-of-Function Mutations in PTPN11 Cause Metachondromatosis, but Not Ollier Disease or Maffucci Syndrome. PLoS Genetics, 2011, 7, e1002050.	3.5	104

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37	Abstract 2122:KRASmutation detection using high resolution melting analysis and its prognostic value in archival colorectal cancer tissues. , 2010, , .		0
38	The spectra of clinical phenotypes in aplasia cutis congenita and terminal transverse limb defects. American Journal of Medical Genetics, Part A, 2009, 149A, 1860-1881.	1.2	92
39	Multiple osteochondromas: mutation update and description of the multiple osteochondromas mutation database (MOdb). Human Mutation, 2009, 30, 1620-1627.	2.5	176
40	Carnitineâ€palmitoyltransferase 2 deficiency: Novel mutations and relevance of newborn screening. American Journal of Medical Genetics, Part A, 2008, 146A, 2925-2928.	1.2	15
41	Mutation Screening of EXT1 and EXT2 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. Journal of Molecular Diagnostics, 2008, 10, 85-92.	2.8	46
42	Erratum to â€œHigh incidence of the CFTR mutations 3272-26A â†' G and L927P in Belgian cystic fibrosis patients, and identification of three new CFTR mutations (186-2A â†' G, E588V, and 1671insTATCA)â€•[Journal of Cystic Fibrosis 6(2007)371â€“375]. Journal of Cystic Fibrosis, 2008, 7, 461.	0.7	0
43	Detection of Microsatellite Instability in Colorectal Cancer Using an Alternative Multiplex Assay of Quasi-Monomorphic Mononucleotide Markers. Journal of Molecular Diagnostics, 2008, 10, 154-159.	2.8	40
44	Mutation analysis of mitochondrial DNA 12SrRNA and tRNASer(UCN) genes in non-syndromic hearing loss patients. Mitochondrion, 2008, 8, 377-382.	3.4	31
45	Microsatellite instability in sporadic colon carcinomas has no independent prognostic value in a Belgian study population. European Journal of Cancer, 2008, 44, 2288-2295.	2.8	19
46	Recurrent post-infectious rhabdomyolysis in muscle CPT-II deficiency caused by a novel missense mutation. Acta Neurologica Belgica, 2008, 108, 155-60.	1.1	1
47	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. Clinical Dysmorphology, 2007, 16, 231-239.	0.3	7
48	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
49	High incidence of the CFTR mutations 3272-26Aâ†'G and L927P in Belgian cystic fibrosis patients, and identification of three new CFTR mutations (186-2Aâ†'G, E588V, and 1671insTATCA). Journal of Cystic Fibrosis, 2007, 6, 371-375.	0.7	5
50	Response to letter to the editor: â€œHypoparathyroidism-Retardation-Dysmorphism Syndrome in a Female Child: A New Variant Not Caused by aTBCE Mutationâ€"Clinical Report and Reviewâ€• American Journal of Medical Genetics, Part A, 2007, 143A, 303-304.	1.2	0
51	GM1 gangliosidosis: molecular analysis of nine patients and development of an RT-PCR assay for GLB1 gene expression profiling. Human Mutation, 2007, 28, 204-204.	2.5	27
52	A combined analytical approach reveals novelEXT1/2 gene mutations in a large cohort of Italian multiple osteochondromas patients. Genes Chromosomes and Cancer, 2007, 46, 470-477.	2.8	43
53	A de novo subterminal trisomy 10p andÂmonosomy 18q inÂaÂgirl with MCA/MR: case report andÂreview. European Journal of Medical Genetics, 2006, 49, 402-413.	1.3	8
54	Comparison of three commonly used PCR-based techniques to analyze MSI status in sporadic colorectal cancer. Journal of Clinical Laboratory Analysis, 2006, 20, 52-61.	2.1	12

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55	A subterminal deletion of the long arm of chromosome 10: A clinical report and review. American Journal of Medical Genetics, Part A, 2006, 140A, 402-409.	1.2	45
56	Hypoparathyroidism-retardation-dysmorphism syndrome in a girl: A new variant not caused by aTBCEmutation-clinical report and review. American Journal of Medical Genetics, Part A, 2006, 140A, 611-617.	1.2	30
57	Genetic Defects in the Development of the Skull Vault in Humans and Mice. Critical Reviews in Eukaryotic Gene Expression, 2006, 16, 119-142.	0.9	8
58	An interstitial deletion of chromosome 7 at band q21: A case report and review. , 2005, 134A, 12-23.		23
59	Myopathy and phosphorylase kinase deficiency caused by a mutation in thePHKA1 gene. American Journal of Medical Genetics, Part A, 2005, 133A, 82-84.	1.2	29
60	Somatic and gonadal mosaicism in Hutchinson-Gilford progeria. American Journal of Medical Genetics, Part A, 2005, 135A, 66-68.	1.2	38
61	A novel mutation in the MSX2 gene in a family with foramina parietalia permagna (FPP). American Journal of Medical Genetics, Part A, 2005, 139A, 45-47.	1.2	11
62	KID Syndrome: Report of a Scandinavian Patient with Connexinâ€26 Gene Mutation. Acta Dermato-Venereologica, 2005, 85, 152-155.	1.3	19
63	Denaturing HPLC-Based Approach for Detecting RYR2 Mutations Involved in Malignant Arrhythmias. Clinical Chemistry, 2004, 50, 1148-1155.	3.2	41
64	Proximal 11p deletion syndrome (P11pDS): additional evaluation of the clinical and molecular aspects. European Journal of Human Genetics, 2004, 12, 400-406.	2.8	19
65	Prenatal diagnosis of Pfeiffer syndrome type II. Prenatal Diagnosis, 2004, 24, 644-646.	2.3	27
66	Two-color multiplex ligation-dependent probe amplification: Detecting genomic rearrangements in hereditary multiple exostoses. Human Mutation, 2004, 24, 86-92.	2.5	142
67	Solitary thyroid nodule as presenting symptom of Pendred syndrome caused by a novel splice-site mutation in intron 8 of the SLC26A4 gene. European Journal of Pediatrics, 2003, 162, 674-677.	2.7	9
68	Clinical and molecular analysis of nine families with Adamsâ€“Oliver syndrome. European Journal of Human Genetics, 2003, 11, 457-463.	2.8	44
69	Prevalence and Evolutionary Origins of the del(GJB6-D13S1830) Mutation in the DFNB1 Locus in Hearing-Impaired Subjects: a Multicenter Study. American Journal of Human Genetics, 2003, 73, 1452-1458.	6.2	269
70	Domain-specific mutations in sequestosome 1 (SQSTM1) cause familial and sporadic Paget's disease. Human Molecular Genetics, 2002, 11, 2735-2739.	2.9	307
71	Multiple exostoses, mental retardation, hypertrichosis, and brain abnormalities in a boy with a de novo 8q24 submicroscopic interstitial deletion. American Journal of Medical Genetics Part A, 2002, 113, 326-332.	2.4	42
72	Genomewide Search in Familial Paget Disease of Bone Shows Evidence of Genetic Heterogeneity with Candidate Loci on Chromosomes 2q36, 10p13, and 5q35. American Journal of Human Genetics, 2001, 69, 1055-1061.	6.2	113

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73	Hereditary multiple exostoses: from genetics to clinical syndrome and complications. European Journal of Radiology, 2001, 40, 208-217.	2.6	88
74	Burning down DEFECT11. American Journal of Medical Genetics Part A, 2001, 100, 331-332.	2.4	10
75	Molecular basis of multiple exostoses: mutations in the EXT1 and EXT2 genes. Human Mutation, 2000, 15, 220-227.	2.5	189
76	Organization of the Ribosomal Operon 16S-23S Gene Spacer Region in Representatives of Neisseria gonorrhoeae. Systematic and Applied Microbiology, 2000, 23, 9-14.	2.8	1
77	Molecular and clinical examination of an Italian DEFECT 11 family. European Journal of Human Genetics, 1999, 7, 579-584.	2.8	27
78	EXT-Mutation Analysis and Loss of Heterozygosity in Sporadic and Hereditary Osteochondromas and Secondary Chondrosarcomas. American Journal of Human Genetics, 1999, 65, 689-698.	6.2	174
79	Mutations in the EXT1 and EXT2 Genes in Hereditary Multiple Exostoses. American Journal of Human Genetics, 1998, 62, 346-354.	6.2	174
80	Identification of a Third EXT-like Gene (EXTL3) Belonging to the EXT Gene Family. Genomics, 1998, 47, 230-237.	2.9	124
81	Localization of a Gene for Autosomal Dominant Osteopetrosis (Albers-SchÃ¶nberg Disease) to Chromosome 1p21. American Journal of Human Genetics, 1997, 61, 363-369.	6.2	76
82	Identification and Characterization of a Novel Member of the EXT Gene Family, EXTL2. European Journal of Human Genetics, 1997, 5, 382-389.	2.8	77