Geert R Mortier

List of Publications by Year in descending order

243 papers	19,536 citations	17776 65 h-index	14386 132 g-index
249 all docs	249 docs citations	249 times ranked	27470 citing authors

#	Article	IF	Citations
1	International Consensus Statement on the diagnosis, multidisciplinary management and lifelong care of individuals with achondroplasia. Nature Reviews Endocrinology, 2022, 18, 173-189.	4.3	44
2	Preexisting memory CD4 T cells in naÃ¯ve individuals confer robust immunity upon hepatitis B vaccination. ELife, 2022, 11, .	2.8	11
3	Broadening the spectrum of loss-of-function variants in NPR-C-related extreme tall stature. Journal of the Endocrine Society, 2022, 6, bvac019.	0.1	2
4	A homozygous hypomorphicBNIP1variant causes an increase in autophagosomes and reduced autophagic flux and results in a spondyloâ€epiphyseal dysplasia. Human Mutation, 2022, 43, 625-642.	1.1	3
5	Identification of Compound Heterozygous Variants in LRP4 Demonstrates That a Pathogenic Variant outside the Third Î²-Propeller Domain Can Cause Sclerosteosis. Genes, 2022, 13, 80.	1.0	3
6	Steel syndrome: Report of three patients, including monozygotic twins and review of clinical and mutation profiles. European Journal of Medical Genetics, 2022, 65, 104521.	0.7	1
7	The European registry for rare bone and mineral conditions (EuRR-Bone): results of a survey on osteogenesis imperfecta and fibrous dysplasia McCune-Albright syndrome. Bone Reports, 2022, 16, 101297.	0.2	0
8	New cases of rare bone and mineral conditions reported within the first 18 months of the European registry for rare bone and mineral conditions. Bone Reports, 2022, 16, 101510.	0.2	0
9	The sqstm1tmÎ”UBA zebrafish model, a proof-of-concept in vivo model for Pagetâ€™s disease of bone?. Bone Reports, 2022, 16, 101483.	0.2	1
10	Clinical characterization of the first Belgian SCN5A founder mutation cohort. Europace, 2021, 23, 918-927.	0.7	3
11	Consensus Recommendations for the Diagnosis and Management of X-Linked Hypophosphatemia in Belgium. Frontiers in Endocrinology, 2021, 12, 641543.	1.5	26
12	Resequencing of candidate genes for Keratoconus reveals a role for Ehlersâ€“Danlos Syndrome genes. European Journal of Human Genetics, 2021, 29, 1745-1755.	1.4	8
13	Destructive juvenile idiopathic arthritis: do not overlook rare genetic skeletal disorders. Lancet Rheumatology, The, 2021, 3, e404.	2.2	0
14	A Panel-Based Sequencing Analysis of Patients with Pagetâ€™s Disease of Bone Suggests Enrichment of Rare Genetic Variation in regulators of NF-Î²B Signaling and Supports the Importance of the 7q33 Locus. Calcified Tissue International, 2021, 109, 656-665.	1.5	1
15	Caffey disease is associated with distinct arginine to cysteine substitutions in the proÎ±1(I) chain of type I procollagen. Genetics in Medicine, 2021, 23, 2378-2385.	1.1	1
16	Phenotypes and genotypes in nonâ€consanguineous and consanguineous primary microcephaly: High incidence of epilepsy. Molecular Genetics & Genomic Medicine, 2021, 9, e1768.	0.6	6
17	Biallelic variants p.Arg1133Cys and p.Arg1379Cys in COL2A1: Further delineation of phenotypic spectrum of recessive Type 2 collagenopathies. American Journal of Medical Genetics, Part A, 2020, 182, 338-347.	0.7	6
18	WNT16 Requires GÎ± Subunits as Intracellular Partners for Both Its Canonical and Non-Canonical WNT Signalling Activity in Osteoblasts. Calcified Tissue International, 2020, 106, 294-302.	1.5	9

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19	Germline and Mosaic Variants in PRKACA and PRKACB Cause a Multiple Congenital Malformation Syndrome. American Journal of Human Genetics, 2020, 107, 977-988.	2.6	33
20	Fibrous Dysplasia, Paget's Disease of Bone, and Other Uncommon Sclerotic Bone Lesions of the Craniofacial Bones. Seminars in Musculoskeletal Radiology, 2020, 24, 570-578.	0.4	3
21	DNA Methylation Profiling and Genomic Analysis in 20 Children with Short Stature Who Were Born Small for Gestational Age. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e4730-e4741.	1.8	12
22	Chondrodysplasias and Aneurysmal Thoracic Aortopathy: An Emerging Tale of Molecular Intersection. Trends in Molecular Medicine, 2020, 26, 783-795.	3.5	2
23	Delineation of a new fibrillino-2-pathy with evidence for a role of FBN2 in the pathogenesis of carpal tunnel syndrome. Journal of Medical Genetics, 2020, 58, jmedgenet-2020-107085.	1.5	4
24	Transcriptomic profiling of different responder types in adults after a PriorixÂ® vaccination. Vaccine, 2020, 38, 3218-3226.	1.7	1
25	Spondylo-epi-metaphyseal dysplasia due to a homozygous missense mutation in the gene encoding Matrilin-3 (T120M). Bone Reports, 2020, 12, 100245.	0.2	2
26	WNT Signaling and Bone: Lessons From Skeletal Dysplasias and Disorders. Frontiers in Endocrinology, 2020, 11, 165.	1.5	61
27	A multi-omics approach expands the mutational spectrum of MAP2K1-related melorheostosis. Bone, 2020, 137, 115406.	1.4	6
28	Memory CD4+ T cell receptor repertoire data mining as a tool for identifying cytomegalovirus serostatus. Genes and Immunity, 2019, 20, 255-260.	2.2	19
29	The third family with Eiken syndrome. Clinical Genetics, 2019, 96, 378-379.	1.0	7
30	An emerging ribosomopathy affecting the skeleton due to biallelic variations in NEPRO. American Journal of Medical Genetics, Part A, 2019, 179, 1709-1717.	0.7	3
31	Nosology and classification of genetic skeletal disorders: 2019 revision. American Journal of Medical Genetics, Part A, 2019, 179, 2393-2419.	0.7	431
32	Diagnosing enterovirus meningitis via blood transcriptomics: an alternative for lumbar puncture?. Journal of Translational Medicine, 2019, 17, 282.	1.8	10
33	Aortic aneurysm/dissection and osteogenesis imperfecta: Four new families and review of the literature. Bone, 2019, 121, 191-195.	1.4	18
34	Pathogenic Variants in GPC4 Cause Keipert Syndrome. American Journal of Human Genetics, 2019, 104, 914-924.	2.6	23
35	Genetic Variation in RIN3 in the Belgian Population Supports Its Involvement in the Pathogenesis of Pagetâ€™s Disease of Bone and Modifies the Age of Onset. Calcified Tissue International, 2019, 104, 613-621.	1.5	5
36	Camuratiâ€“Engelmann Disease. Calcified Tissue International, 2019, 104, 554-560.	1.5	25

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37	The homozygous variant c.797G>A/p.(Cys266Tyr) inPISDis associated with a Spondyloepimetaphyseal dysplasia with large epiphyses and disturbed mitochondrial function. Human Mutation, 2019, 40, 299-309.	1.1	54
38	Human Genetics of Sclerosing Bone Disorders. Current Osteoporosis Reports, 2018, 16, 256-268.	1.5	13
39	Genotype-Phenotype Correlation in NF1: Evidence for a More Severe Phenotype Associated with Missense Mutations Affecting NF1 Codons 844â€“848. American Journal of Human Genetics, 2018, 102, 69-87.	2.6	144
40	Increased herpes zoster risk associated with poor HLA-A immediate early 62 protein (IE62) affinity. Immunogenetics, 2018, 70, 363-372.	1.2	8
41	Transcriptome profiling in blood before and after hepatitis B vaccination shows significant differences in gene expression between responders and non-responders. Vaccine, 2018, 36, 6282-6289.	1.7	47
42	Conditional mouse models support the role of SLC39A14 (ZIP14) in Hyperostosis Cranialis Interna and in bone homeostasis. PLoS Genetics, 2018, 14, e1007321.	1.5	13
43	Bi-allelic Loss-of-Function Mutations in the NPR-C Receptor Result in Enhanced Growth and Connective Tissue Abnormalities. American Journal of Human Genetics, 2018, 103, 288-295.	2.6	25
44	The Lrp4R1170Q Homozygous Knock-In Mouse Recapitulates the Bone Phenotype of Sclerosteosis in Humans. Journal of Bone and Mineral Research, 2017, 32, 1739-1749.	3.1	27
45	Targeted Next-Generation Sequencing of 51 Genes Involved in Primary Electrical Disease. Journal of Molecular Diagnostics, 2017, 19, 445-459.	1.2	15
46	Metatarsal bony syndactyly in 2 fetuses with Smithâ€Lemliâ€Opitz syndrome: An underâ€recognized part of the clinical spectrum. Clinical Genetics, 2017, 92, 342-343.	1.0	1
47	Mutations in two large pedigrees highlight the role of ZNF711 in X-linked intellectual disability. Gene, 2017, 605, 92-98.	1.0	26
48	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.	1.1	94
49	Inborn errors in RNA polymerase III underlie severe varicella zoster virus infections. Journal of Clinical Investigation, 2017, 127, 3543-3556.	3.9	125
50	Heterozygous Loss-of-Function SEC61A1 Mutations Cause Autosomal-Dominant Tubulo-Interstitial and Glomerulocystic Kidney Disease with Anemia. American Journal of Human Genetics, 2016, 99, 174-187.	2.6	124
51	A Novel Domain-Specific Mutation in a Sclerosteosis Patient Suggests a Role of LRP4 as an Anchor for Sclerostin in Human Bone. Journal of Bone and Mineral Research, 2016, 31, 874-881.	3.1	65
52	DNA Diagnostics of Hereditary Hearing Loss: A Targeted Resequencing Approach Combined with a Mutation Classification System. Human Mutation, 2016, 37, 812-819.	1.1	76
53	Detection of a case of chronic myeloid leukaemia with deletions at the t(9;22) translocation breakpoints by a genome-wide non-invasive prenatal test. Prenatal Diagnosis, 2016, 36, 760-765.	1.1	9
54	Identification of biallelicLRRK1mutations in osteosclerotic metaphyseal dysplasia and evidence for locus heterogeneity. Journal of Medical Genetics, 2016, 53, 568-574.	1.5	43

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55	Whole genome sequencing of a dizygotic twin suggests a role for the serotonin receptor HTR7 in autism spectrum disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2016, 171, 1049-1056.	1.1	8
56	Hyperkalemia in young children: blood pressure checked?. European Journal of Pediatrics, 2016, 175, 2011-2013.	1.3	6
57	Novel microdeletions on chromosome 14q32.2 suggest a potential role for non-coding RNAs in Kagami-Ogata syndrome. European Journal of Human Genetics, 2016, 24, 1724-1729.	1.4	27
58	Long-term acquired everolimus resistance in pancreatic neuroendocrine tumours can be overcome with novel PI3K-AKT-mTOR inhibitors. British Journal of Cancer, 2016, 114, 650-658.	2.9	69
59	Eight mutations including 5 novel ones in the COL1A1 gene in Czech patients with osteogenesis imperfecta. Biomedical Papers of the Medical Faculty of the University Palacký, Olomouc, Czechoslovakia, 2016, 160, 442-447.	0.2	9
60	Regulation of body and brain size: role of MAP4 and other centrosomal proteins. Human Mutation, 2015, 36, v-v.	1.1	0
61	Nosology and classification of genetic skeletal disorders: 2015 revision. American Journal of Medical Genetics, Part A, 2015, 167, 2869-2892.	0.7	453
62	Performant Mutation Identification Using Targeted Next-Generation Sequencing of 14 Thoracic Aortic Aneurysm Genes. Human Mutation, 2015, 36, 808-814.	1.1	97
63	Mutations in the latent TGF-beta binding protein 3 (LTBP3) gene cause brachyolmia with amelogenesis imperfecta. Human Molecular Genetics, 2015, 24, 3038-3049.	1.4	40
64	Whole-exome characterization of pancreatic neuroendocrine tumor cell lines BON-1 and QGP-1. Journal of Molecular Endocrinology, 2015, 54, 137-147.	1.1	83
65	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.	0.7	73
66	A STAT3 mutation in hyper-immunoglobulin E syndrome: A case report. Journal of Pediatric Genetics, 2015, 02, 091-096.	0.3	3
67	Five patients with a chromosome 1q21.1 triplication show macrocephaly, increased weight and facial similarities. European Journal of Medical Genetics, 2015, 58, 503-508.	0.7	15
68	The SMAD-binding domain of SKI: a hotspot for de novo mutations causing Shprintzenâ€“Goldberg syndrome. European Journal of Human Genetics, 2015, 23, 224-228.	1.4	48
69	Early presentation of cystic kidneys in a family with a homozygous INVS mutation. American Journal of Medical Genetics, Part A, 2014, 164, 1627-1634.	0.7	7
70	Familial hypertryptasemia with associated mast cell activation syndrome. Journal of Allergy and Clinical Immunology, 2014, 134, 1448-1450.e3.	1.5	44
71	Disfluency: it is not always stuttering. Clinical Genetics, 2014, 85, 298-299.	1.0	2
72	Mutation in the type II collagen gene (COL2AI) as a cause of primary osteoarthritis associated with mild spondyloepiphyseal involvement. Seminars in Arthritis and Rheumatism, 2014, 44, 101-104.	1.6	13

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73	Dysmorphology at a distance: results of a web-based diagnostic service. European Journal of Human Genetics, 2014, 22, 327-332.	1.4	14
74	XYLT1 Mutations in Desbuquois Dysplasia Type 2. American Journal of Human Genetics, 2014, 94, 405-414.	2.6	92
75	Novel pathogenic COL11A1/COL11A2 variants in Stickler syndrome detected by targeted NGS and exome sequencing. Molecular Genetics and Metabolism, 2014, 113, 230-235.	0.5	48
76	A novel mutation (g.106737G>T) in zone of polarizing activity regulatory sequence (ZRS) causes variable limb phenotypes in Werner mesomelia. American Journal of Medical Genetics, Part A, 2014, 164, 898-906.	0.7	15
77	Camuratiâ€“Engelmann Disease (Progressive Diaphyseal Dysplasia): Reports of an Indian Kindred. Calcified Tissue International, 2014, 94, 240-247.	1.5	10
78	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.	1.4	32
79	Sclerosing Bone Dysplasias: Leads Toward Novel Osteoporosis Treatments. Current Osteoporosis Reports, 2014, 12, 243-251.	1.5	12
80	Loss-of-function HDAC8 mutations cause a phenotypic spectrum of Cornelia de Lange syndrome-like features, ocular hypertelorism, large fontanelle and X-linked inheritance. Human Molecular Genetics, 2014, 23, 2888-2900.	1.4	120
81	Multicentric Carpotarsal Osteolysis Is Caused by Mutations Clustering in the Amino-Terminal Transcriptional Activation Domain of MAFB. American Journal of Human Genetics, 2014, 94, 643.	2.6	2
82	Helical mutations in type I collagen that affect the processing of the amino-propeptide result in an Osteogenesis Imperfecta/Ehlers-Danlos Syndrome overlap syndrome. Orphanet Journal of Rare Diseases, 2013, 8, 78.	1.2	73
83	FGFR1mutations cause Hartsfield syndrome, the unique association of holoprosencephaly and ectrodactyly. Journal of Medical Genetics, 2013, 50, 585-592.	1.5	75
84	No mutations in the serotonin related TPH1 and HTR1B genes in patients with monogenic sclerosing bone disorders. Bone, 2013, 55, 52-56.	1.4	7
85	Mutations in sFRP1 or sFRP4 are not a common cause of craniotubular hyperostosis. Bone, 2013, 52, 292-295.	1.4	4
86	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.	2.6	135
87	Complex genetics of radial ray deficiencies: screening of a cohort of 54 patients. Genetics in Medicine, 2013, 15, 195-202.	1.1	15
88	A new form of severe spondyloepimetaphyseal dysplasia: Clinical and radiological characterization. , 2013, 161A, n/a-n/a.		4
89	Nasal speech in patients with 12q15 microdeletions. European Journal of Human Genetics, 2012, 20, 367-367.	1.4	0
90	17q24.2 microdeletions: a new syndromal entity with intellectual disability, truncal obesity, mood swings and hallucinations. European Journal of Human Genetics, 2012, 20, 534-539.	1.4	28

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91	Loss-of-function mutations in TGFB2 cause a syndromic presentation of thoracic aortic aneurysm. Nature Genetics, 2012, 44, 922-927.	9.4	391
92	NEK1andDYNC2H1are both involved in short rib polydactyly Majewski type but not in Beemer Langer cases. Journal of Medical Genetics, 2012, 49, 227-233.	1.5	57
93	HDAC8 mutations in Cornelia de Lange syndrome affect the cohesin acetylation cycle. Nature, 2012, 489, 313-317.	13.7	488
94	Mutations in the TGF-Î² repressor SKI cause Shprintzen-Goldberg syndrome with aortic aneurysm. Nature Genetics, 2012, 44, 1249-1254.	9.4	237
95	Mutations in PIGO, a Member of the GPI-Anchor-Synthesis Pathway, Cause Hyperphosphatasia with Mental Retardation. American Journal of Human Genetics, 2012, 91, 146-151.	2.6	135
96	Voice-Related Quality of Life in Adults With Neurofibromatosis Type 1. Journal of Voice, 2012, 26, e57-e62.	0.6	7
97	X-linked sideroblastic anemia and ataxia: A new family with identification of a fourth ABCB7 gene mutation. European Journal of Paediatric Neurology, 2012, 16, 730-735.	0.7	39
98	Genomics, Epigenetics and Growth. , 2012, , 153-171.		2
99	Sclerosing Bone Dysplasias. , 2012, , 541-556.		0
100	Further delineation of CANT1 phenotypic spectrum and demonstration of its role in proteoglycan synthesis. Human Mutation, 2012, 33, 1261-1266.	1.1	47
101	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.	0.7	14
102	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.	0.7	26
103	The diagnostic challenge of progressive pseudorheumatoid dysplasia (PPRD): A review of clinical features, radiographic features, and WISP3 mutations in 63 affected individuals. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 217-229.	0.7	74
104	Severe neurologic manifestations from cervical spine instability in spondylo-megaepiphyseal-metaphyseal dysplasia. , 2012, 160C, 230-237.		10
105	Haploinsufficiency of <scp>CMIP</scp> in a Girl With Autism Spectrum Disorder and Developmental Delay due to a De Novo Deletion on Chromosome 16q23.2. Autism Research, 2012, 5, 277-281.	2.1	19
106	Recurrent Dominant Mutations Affecting Two Adjacent Residues in the Motor Domain of the Monomeric Kinesin KIF22 Result in Skeletal Dysplasia and Joint Laxity. American Journal of Human Genetics, 2012, 90, 170.	2.6	0
107	Multicentric Carpotarsal Osteolysis Is Caused by Mutations Clustering in the Amino-Terminal Transcriptional Activation Domain of MAFB. American Journal of Human Genetics, 2012, 90, 494-501.	2.6	97
108	Articulation in schoolchildren and adults with neurofibromatosis type 1. Journal of Communication Disorders, 2012, 45, 111-120.	0.8	11

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109	Pseudoachondroplasia and multiple epiphyseal dysplasia: A 7â€year comprehensive analysis of the known disease genes identify novel and recurrent mutations and provides an accurate assessment of their relative contribution. Human Mutation, 2012, 33, 144-157.	1.1	104
110	Overall intelligibility, articulation, resonance, voice and language in a child with Nager syndrome. International Journal of Pediatric Otorhinolaryngology, 2011, 75, 270-276.	0.4	5
111	Voice Characteristics in Adults With Neurofibromatosis Type 1. Journal of Voice, 2011, 25, 759-764.	0.6	4
112	Two novel WTX mutations underscore the unpredictability of male survival in osteopathia striata with cranial sclerosis. Clinical Genetics, 2011, 80, 383-388.	1.0	25
113	Nasal speech and hypothyroidism are common hallmarks of 12q15 microdeletions. European Journal of Human Genetics, 2011, 19, 1032-1037.	1.4	11
114	Mutations in the TGFÎ² Binding-Protein-Like Domain 5 of FBN1 Are Responsible for Acromicric and Geleophysic Dysplasias. American Journal of Human Genetics, 2011, 89, 7-14.	2.6	199
115	Recurrent Dominant Mutations Affecting Two Adjacent Residues in the Motor Domain of the Monomeric Kinesin KIF22 Result in Skeletal Dysplasia and Joint Laxity. American Journal of Human Genetics, 2011, 89, 767-772.	2.6	31
116	Tetrasomy and pentasomy of the X chromosome. European Journal of Pediatrics, 2011, 170, 1325-1327.	1.3	15
117	Genotypeâ€“phenotype analysis of the branchioâ€oculoâ€facial syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 22-32.	0.7	61
118	Nosology and classification of genetic skeletal disorders: 2010 revision. American Journal of Medical Genetics, Part A, 2011, 155, 943-968.	0.7	573
119	Five patients with novel overlapping interstitial deletions in 8q22.2q22.3. American Journal of Medical Genetics, Part A, 2011, 155, 1857-1864.	0.7	23
120	Objective assessment of nasality in flemish adults with neurofibromatosis type 1. American Journal of Medical Genetics, Part A, 2011, 155, 2974-2981.	0.7	2
121	Haploinsufficiency of TAB2 Causes Congenital Heart Defects in Humans. American Journal of Human Genetics, 2010, 86, 839-849.	2.6	97
122	Mesomelia-Synostoses Syndrome Results from Deletion of SULF1 and SLCO5A1 Genes at 8q13. American Journal of Human Genetics, 2010, 87, 95-100.	2.6	42
123	Distinct Effects of Allelic NFIX Mutations on Nonsense-Mediated mRNA Decay Engender Either a Sotos-like or a Marshall-Smith Syndrome. American Journal of Human Genetics, 2010, 87, 189-198.	2.6	131
124	Speech disorders in neurofibromatosis type 1: a sample survey. International Journal of Language and Communication Disorders, 2010, 45, 600-607.	0.7	11
125	Speech fluency in neurofibromatosis type 1. Journal of Fluency Disorders, 2010, 35, 59-69.	0.7	7
126	Word-final prolongations in an adult male with neurofibromatosis type 1. Journal of Fluency Disorders, 2010, 35, 235-245.	0.7	10

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127	Genotypeâ€“phenotype correlation in eight new patients with a deletion encompassing 2q31.1. American Journal of Medical Genetics, Part A, 2010, 152A, 1213-1224.	0.7	29
128	Templeâ€“Baraitser syndrome: A rare and possibly unrecognized condition. American Journal of Medical Genetics, Part A, 2010, 152A, 2322-2326.	0.7	12
129	Phenotype and natural history in Marshallâ€“Smith syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 2714-2726.	0.7	44
130	Stickler syndrome caused by COL2A1 mutations: genotypeâ€“phenotype correlation in a series of 100 patients. European Journal of Human Genetics, 2010, 18, 872-880.	1.4	114
131	De novo mutations of SETBP1 cause Schinzel-Giedion syndrome. Nature Genetics, 2010, 42, 483-485.	9.4	417
132	Duplications of the critical Rubinstein-Taybi deletion region on chromosome 16p13.3 cause a novel recognisable syndrome. Journal of Medical Genetics, 2010, 47, 155-161.	1.5	47
133	Improved Molecular Diagnostics of Idiopathic Short Stature and Allied Disorders: Quantitative Polymerase Chain Reaction-Based Copy Number Profiling of SHOX and Pseudoautosomal Region 1. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 3010-3018.	1.8	15
134	Long-term impact of tongue reduction on speech intelligibility, articulation and oromyofunctional behaviour in a child with Beckwithâ€“Wiedemann syndrome. International Journal of Pediatric Otorhinolaryngology, 2010, 74, 309-318.	0.4	26
135	MHC Class II Deficiency. , 2009, , 1306-1308.		0
136	Hartsfield holoprosencephalyâ€“ectrodactyly syndrome in five male patients: Further delineation and review. American Journal of Medical Genetics, Part A, 2009, 149A, 1476-1481.	0.7	17
137	Mesomelic dysplasia with acral synostoses Verloesâ€“Davidâ€“Pfeiffer type: Followâ€up study documents progressive clinical course. American Journal of Medical Genetics, Part A, 2009, 149A, 2220-2225.	0.7	3
138	The Heterozygous Lemd3 +/GT Mouse Is Not a Murine Model for Osteopoikilosis in Humans. Calcified Tissue International, 2009, 85, 546-551.	1.5	3
139	Identification of CANT1 Mutations in Desbuquois Dysplasia. American Journal of Human Genetics, 2009, 85, 706-710.	2.6	81
140	Homozygous Inactivating Mutations in the NKX3-2 Gene Result in Spondylo-Megaepiphyseal-Metaphyseal Dysplasia. American Journal of Human Genetics, 2009, 85, 916-922.	2.6	30
141	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.	1.5	77
142	Unusual 8p inverted duplication deletion with telomere capture from 8q. European Journal of Medical Genetics, 2009, 52, 31-36.	0.7	26
143	Fourteen new cases contribute to the characterization of the 7q11.23 microduplication syndrome. European Journal of Medical Genetics, 2009, 52, 94-100.	0.7	157
144	The 12q14 microdeletion syndrome: Additional patients and further evidence that HMGA2 is an important genetic determinant for human height. European Journal of Medical Genetics, 2009, 52, 101-107.	0.7	46

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145	Whole body MR imaging in neurofibromatosis type 1. European Journal of Radiology, 2009, 69, 236-242.	1.2	32
146	Challenges for CNV interpretation in clinical molecular karyotyping: Lessons learned from a 1001 sample experience. European Journal of Medical Genetics, 2009, 52, 398-403.	0.7	90
147	Further delineation of the 15q13 microdeletion and duplication syndromes: a clinical spectrum varying from non-pathogenic to a severe outcome. Journal of Medical Genetics, 2009, 46, 511-523.	1.5	250
148	Delineation of a critical region on chromosome 18 for the del(18)(q12.2q21.1) syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 1330-1334.	0.7	28
149	Mutations in the cyclin family member FAM58A cause an X-linked dominant disorder characterized by syndactyly, telecanthus and anogenital and renal malformations. Nature Genetics, 2008, 40, 287-289.	9.4	45
150	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	13.9	663
151	Buschke-Ollendorff syndrome: A manifestation of a heterozygous nonsense mutation in the LEMD3 gene. Journal of the American Academy of Dermatology, 2008, 58, S103-S104.	0.6	24
152	Mapping of 5q35 chromosomal rearrangements within a genomically unstable region. Journal of Medical Genetics, 2008, 45, 672-678.	1.5	7
153	High prevalence of SDHB mutations in head and neck paraganglioma in Belgium. Journal of Hypertension, 2008, 26, 1395-1401.	0.3	16
154	Osteopoikilosis, short stature and mental retardation as key features of a new microdeletion syndrome on 12q14. Journal of Medical Genetics, 2007, 44, 264-268.	1.5	58
155	Report of a female patient with mental retardation and tall stature due to a chromosomal rearrangement disrupting the OPHN1 gene on Xq12. European Journal of Medical Genetics, 2007, 50, 446-454.	0.7	16
156	Overall intelligibility, language, articulation, voice and resonance characteristics in a child with Shprintzenâ€“Goldberg syndrome. International Journal of Pediatric Otorhinolaryngology, 2007, 71, 721-728.	0.4	9
157	Mutations in STRA6 Cause a Broad Spectrum of Malformations Including Anophthalmia, Congenital Heart Defects, Diaphragmatic Hernia, Alveolar Capillary Dysplasia, Lung Hypoplasia, and Mental Retardation. American Journal of Human Genetics, 2007, 80, 550-560.	2.6	316
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