Chong Ae Kim

List of Publications by Year
in descending order

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203	9,103	76326	46799
papers	citations	40 h-index	89 g-index

211	211	211	11709
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	LDL Receptor-Related Protein 5 (LRP5) Affects Bone Accrual and Eye Development. Cell, 2001, 107, 513-523.	28.9	2,055
2	Localized mutations in the gene encoding the cytoskeletal protein filamin A cause diverse malformations in humans. Nature Genetics, 2003, 33, 487-491.	21.4	375
3	Association of a Homozygous Nonsense Caveolin-1 Mutation with Berardinelli-Seip Congenital Lipodystrophy. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 1129-1134.	3.6	343
4	Mutations in the Transmembrane Natriuretic Peptide Receptor NPR-B Impair Skeletal Growth and Cause Acromesomelic Dysplasia, Type Maroteaux. American Journal of Human Genetics, 2004, 75, 27-34.	6.2	325
5	Mutations in NOTCH2 cause Hajdu-Cheney syndrome, a disorder of severe and progressive bone loss. Nature Genetics, 2011, 43, 303-305.	21.4	291
6	CACP, encoding a secreted proteoglycan, is mutated in camptodactyly-arthropathy-coxa vara-pericarditis syndrome. Nature Genetics, 1999, 23, 319-322.	21.4	286
7	Mutations in the gene encoding filamin B disrupt vertebral segmentation, joint formation and skeletogenesis. Nature Genetics, 2004, 36, 405-410.	21.4	252
8	Congenital abnormalities in Brazilian children associated with misoprostol misuse in first trimester of pregnancy. Lancet, The, 1998, 351, 1624-1627.	13.7	225
9	Rare variants inSOS2andLZTR1are associated with Noonan syndrome. Journal of Medical Genetics, 2015, 52, 413-421.	3.2	187
10	Limb deficiency with or without MÃ¶bius sequence in seven Brazilian children associated with misoprostol use in the first trimester of pregnancy. American Journal of Medical Genetics Part A, 1993, 47, 59-64.	2.4	167
11	Mutations in SRCAP, Encoding SNF2-Related CREBBP Activator Protein, Cause Floating-Harbor Syndrome. American Journal of Human Genetics, 2012, 90, 308-313.	6.2	157
12	Mucopolysaccharidosis I, II, and VI: brief review and guidelines for treatment. Genetics and Molecular Biology, 2010, 33, 589-604.	1.3	150
13	Haploinsufficiency of the human homeobox gene ALX4 causes skull ossification defects. Nature Genetics, 2001, 27, 17-18.	21.4	142
14	A clinical study of 77 patients with mucopolysaccharidosis type II. Acta Paediatrica, International Journal of Paediatrics, 2007, 96, 63-70.	1.5	112
15	Mechanisms of ring chromosome formation, ring instability and clinical consequences. BMC Medical Genetics, 2011, 12, 171.	2.1	106
16	Whole-genome array-CGH screening in undiagnosed syndromic patients: old syndromes revisited and new alterations. Cytogenetic and Genome Research, 2006, 115, 254-261.	1.1	103
17	Williams syndrome. Nature Reviews Disease Primers, 2021, 7, 42.	30.5	103
18	A molecular and clinical study of Larsen syndrome caused by mutations in FLNB. Journal of Medical Genetics, 2006, 44, 89-98.	3.2	102

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19	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.	6.2	97
20	Bi-allelic CSF1R Mutations Cause Skeletal Dysplasia of Dysosteosclerosis-Pyle Disease Spectrum and Degenerative Encephalopathy with Brain Malformation. American Journal of Human Genetics, 2019, 104, 925-935.	6.2	92
21	Prevalence of Mutations in AGPAT2 Among Human Lipodystrophies. Diabetes, 2003, 52, 1573-1578.	0.6	87
22	Echocardiographic study of paediatric patients with mucopolysaccharidosis. Cardiology in the Young, 2010, 20, 254-261.	0.8	77
23	SHOX mutations in idiopathic short stature and Leri-Weill dyschondrosteosis: frequency and phenotypic variability. Clinical Endocrinology, 2006, 66, 061031010617004-???.	2.4	75
24	Neurofibromatosis-Noonan syndrome: Molecular evidence of the concurrence of both disorders in a patient. American Journal of Medical Genetics, Part A, 2005, 136A, 242-245.	1.2	74
25	Associations among genotype, clinical phenotype, and intracellular localization of trafficking proteins in ARC syndrome. Human Mutation, 2012, 33, 1656-1664.	2.5	74
26	Autoimmune disease and multiple autoantibodies in 42 patients with RASopathies. American Journal of Medical Genetics, Part A, 2012, 158A, 1077-1082.	1.2	73
27	Report of a del22q11 in a patient with Mayer-Rokitansky-KÃ¼ster-Hauser (MRKH) anomaly and exclusion ofWNT-4,RAR-gamma, andRXR-alpha as major genes determining MRKH anomaly in a study of 25 affected women. American Journal of Medical Genetics, Part A, 2006, 140A, 1339-1342.	1.2	64
28	Mucopolysaccharidoses in Brazil: What happens from birth to biochemical diagnosis?. American Journal of Medical Genetics, Part A, 2008, 146A, 1741-1747.	1.2	63
29	Identification of 2 novel ANTXR2 mutations in patients with hyaline fibromatosis syndrome and proposal of a modified grading system. American Journal of Medical Genetics, Part A, 2012, 158A, 732-742.	1.2	61
30	Serpentine fibula polycystic kidney syndrome is part of the phenotypic spectrum of Hajduâ€“Cheney syndrome. European Journal of Human Genetics, 2012, 20, 122-124.	2.8	60
31	The phenotype of Floating-Harbor syndrome: clinical characterization of 52 individuals with mutations in exon 34 of SRCAP. Orphanet Journal of Rare Diseases, 2013, 8, 63.	2.7	60
32	Are Noonan syndrome and Noonan-like/multiple giant cell lesion syndrome distinct entities?. American Journal of Medical Genetics Part A, 2001, 98, 230-234.	2.4	54
33	Further evidence of the importance of RIT1 in Noonan syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 2952-2957.	1.2	53
34	Molecular evidence that AEC syndrome and Rapp-Hodgkin syndrome are variable expression of a single genetic disorder. Clinical Genetics, 2004, 66, 79-80.	2.0	52
35	Mutations in MAP3K7 that Alter the Activity of the TAK1 Signaling Complex Cause Frontometaphyseal Dysplasia. American Journal of Human Genetics, 2016, 99, 392-406.	6.2	52
36	Patients with a Kabuki syndrome phenotype demonstrate DNA methylation abnormalities. European Journal of Human Genetics, 2017, 25, 1335-1344.	2.8	52

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37	A new case ofÂinterstitial 6q16.2Âdeletion inÂaÂpatient with Praderâ€“Willi-like phenotype andÂinvestigation ofÂSIM1 gene deletion inÂ87Âpatients with syndromic obesity. European Journal of Medical Genetics, 2006, 49, 298-305.	1.3	50
38	Prader-Willi-like phenotype: investigation ofÂ1p36 deletion inÂ41Âpatients with delayed psychomotor development, hypotonia, obesity and/orÂhyperphagia, learning disabilities andÂbehavioral problems. European Journal of Medical Genetics, 2006, 49, 451-460.	1.3	46
39	Using a combination of MLPA kits to detect chromosomal imbalances in patients with multiple congenital anomalies and mental retardation is a valuable choice for developing countries. European Journal of Medical Genetics, 2011, 54, e425-e432.	1.3	44
40	PTPN11 Gene Analysis in 74 Brazilian Patients with Noonan Syndrome or Noonan-like Phenotype. Genetic Testing and Molecular Biomarkers, 2006, 10, 186-191.	1.7	43
41	Two distinct regions in 2q24.2â€q24.3 associated with idiopathic epilepsy. Epilepsia, 2010, 51, 2457-2460.	5.1	43
42	Comprehensive genetic analysis of 57 families with clinically suspected Cornelia de Lange syndrome. Journal of Human Genetics, 2019, 64, 967-978.	2.3	43
43	Metabolic effects of C677T and A1298C mutations at the MTHFR gene in Brazilian children with neural tube defects. Clinica Chimica Acta, 2002, 318, 139-143.	1.1	40
44	Clinical variability in a Noonan syndrome family with a new PTPN11 gene mutation. American Journal of Medical Genetics Part A, 2004, 130A, 378-383.	2.4	40
45	DiGeorge Syndrome: a not so rare disease. Clinics, 2010, 65, 865-869.	1.5	40
46	Mutations in PCYT1A Cause Spondylometaphyseal Dysplasia with Cone-Rod Dystrophy. American Journal of Human Genetics, 2014, 94, 113-119.	6.2	39
47	Microduplication of the ICR2 domain at chromosome 11p15 and familial Silverâ€“Russell syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 2479-2483.	1.2	38
48	Further evidence of genetic heterogeneity in Costello syndrome: involvement of the KRAS gene. Journal of Human Genetics, 2007, 52, 521-526.	2.3	36
49	High frequency of submicroscopic chromosomal imbalances in patients with syndromic craniosynostosis detected by a combined approach of microsatellite segregation analysis, multiplex ligation-dependent probe amplification and array-based comparative genome hybridisation. Journal of Medical Genetics, 2008, 45, 447-450.	3.2	36
50	Using a deep learning network to recognise low back pain in static standing. Ergonomics, 2018, 61, 1374-1381.	2.1	36
51	Autosomal-Recessive Mutations in MESD Cause Osteogenesis Imperfecta. American Journal of Human Genetics, 2019, 105, 836-843.	6.2	36
52	Genetic Disorders in Prenatal Onset Syndromic Short Stature Identified by Exome Sequencing. Journal of Pediatrics, 2019, 215, 192-198.	1.8	36
53	Chromosomal microarray analysis in the genetic evaluation of 279 patients with syndromic obesity. Molecular Cytogenetics, 2018, 11, 14.	0.9	35
54	Enzyme replacement therapy with galsulfase in 34 children younger than five years of age with MPS VI. Molecular Genetics and Metabolism, 2013, 109, 62-69.	1.1	34

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55	The clinical impact of chromosomal rearrangements with breakpoints upstream of the SOX9gene: two novel de novo balanced translocations associated with acampomelic campomelic dysplasia. BMC Medical Genetics, 2013, 14, 50.	2.1	33
56	Extending the phenotype of monosomy 1p36 syndrome and mapping of a critical region for obesity and hyperphagia. American Journal of Medical Genetics, Part A, 2010, 152A, 102-110.	1.2	32
57	Recurrent Copy Number Variants Associated with Syndromic Short Stature of Unknown Cause. Hormone Research in Paediatrics, 2018, 89, 13-21.	1.8	29
58	Neuroblastoma in a boy with MCA/MR syndrome, deletion 11q, and duplication 12q. American Journal of Medical Genetics Part A, 1995, 58, 46-49.	2.4	28
59	Exuberant Juvenile Hyaline Fibromatosis in Two Patients. Pediatric Dermatology, 2006, 23, 458-464.	0.9	28
60	Cockayne syndrome type A: novel mutations in eight typical patients. Journal of Human Genetics, 2006, 51, 701-705.	2.3	28
61	A Known SOST Gene Mutation Causes Sclerosteosis in a Familial and an Isolated Case from Brazilian Origin. Genetic Testing and Molecular Biomarkers, 2008, 12, 475-479.	1.7	28
62	Primary immunodeficiency with chronic enteropathy and developmental delay in a boy arising from a novel homozygous RIPK1 variant. Journal of Human Genetics, 2019, 64, 955-960.	2.3	28
63	PTPN11 and KRAS Gene Analysis in Patients with Noonan and Noonan-Like Syndromes. Genetic Testing and Molecular Biomarkers, 2010, 14, 425-432.	0.7	27
64	CHILD Syndrome Caused by a Deletion of Exons 6â€“8 of the <i>NSDHL</i> Gene. Dermatology, 2005, 211, 155-158.	2.1	25
65	An 11q11â€“q13.3 duplication, including FGF3 and FGF4 genes, in a patient with syndromic multiple craniosynostoses. American Journal of Medical Genetics, Part A, 2007, 143A, 1912-1918.	1.2	25
66	Further delineation of nonhomologous-based recombination and evidence for subtelomeric segmental duplications in 1p36 rearrangements. Human Genetics, 2009, 125, 551-563.	3.8	25
67	Obesity with associated developmental delay and/or learning disability in patients exhibiting additional features: Report of novel pathogenic copy number variants. American Journal of Medical Genetics, Part A, 2013, 161, 479-486.	1.2	25
68	Schinzelâ€“Giedion syndrome in two Brazilian patients: Report of a novel mutation in SETBP1 and literature review of the clinical features. American Journal of Medical Genetics, Part A, 2015, 167, 1039-1046.	1.2	25
69	AEC Syndrome and CHAND Syndrome: Further Evidence of Clinical Overlapping in the Ectodermal Dysplasias. Pediatric Dermatology, 2000, 17, 218-221.	0.9	24
70	Craniosynostosis in pycnodysostosis: Broadening the spectrum of the cranial flat bone abnormalities. American Journal of Medical Genetics, Part A, 2010, 152A, 2599-2603.	1.2	24
71	Vertical transmission of a frontonasal phenotype caused by a novel ALX4 mutation. American Journal of Medical Genetics, Part A, 2013, 161, 600-604.	1.2	24
72	The recurrent PPP1CB mutation p.Pro49Arg in an additional Noonanâ€like syndrome individual: Broadening the clinical phenotype. American Journal of Medical Genetics, Part A, 2017, 173, 824-828.	1.2	24

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73	Autosomal dominant frontometaphyseal dysplasia: Delineation of the clinical phenotype. American Journal of Medical Genetics, Part A, 2017, 173, 1739-1746.	1.2	24
74	Mutations, Clinical Findings and Survival Estimates in South American Patients with X-Linked Adrenoleukodystrophy. PLoS ONE, 2012, 7, e34195.	2.5	24
75	Williams-Beuren syndrome: cardiovascular abnormalities in 20 patients diagnosed with fluorescence in situ hybridization. Arquivos Brasileiros De Cardiologia, 2003, 81, 462-73.	0.8	23
76	Williams Syndrome: development of a new scoring system for clinical diagnosis. Clinics, 2007, 62, 159-166.	1.5	23
77	Congenital genitourinary abnormalities in children with Williamsâ€“Beuren syndrome. Journal of Pediatric Urology, 2014, 10, 804-809.	1.1	22
78	Diagnostic power and clinical impact of exome sequencing in a cohort of 500 patients with rare diseases. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 955-964.	1.6	22
79	Phenotypic overlap in Melnickâ€“Needles, serpentine fibulaâ€“polycystic kidney and Hajduâ€“Cheney syndromes: a clinical and molecular study in three patients. Clinical Dysmorphology, 2007, 16, 27-33.	0.3	20
80	New insights in mucopolysaccharidosis type VI: Neurological perspective. Brain and Development, 2014, 36, 585-592.	1.1	20
81	Natural history of 39 patients with Achondroplasia. Clinics, 2018, 73, e324.	1.5	20
82	A study of EEG and epilepsy profile in Wolfâ€“Hirschhorn syndrome and considerations regarding its correlation with other chromosomal disorders. Brain and Development, 2003, 25, 283-287.	1.1	19
83	Copy number variation in Williams-Beuren syndrome: suitable diagnostic strategy for developing countries. BMC Research Notes, 2012, 5, 13.	1.4	19
84	Multiple, diffuse schwannomas in a RASopathy phenotype patient with germline KRAS mutation: a causal relationship?. Clinical Genetics, 2012, 81, 595-597.	2.0	19
85	Enzyme replacement therapy for Mucopolysaccharidosis Type I among patients followed within the MPS Brazil Network. Genetics and Molecular Biology, 2014, 37, 23-29.	1.3	19
86	Mucopolysaccharidosis type IVA: Evidence of primary and secondary central nervous system involvement. American Journal of Medical Genetics, Part A, 2014, 164, 1162-1169.	1.2	19
87	Impact of Growth Hormone Therapy on Adult Height in Patients with <i>PTPN11</i> Mutations Related to Noonan Syndrome. Hormone Research in Paediatrics, 2019, 91, 252-261.	1.8	19
88	Malformation of cortical and vascular development in one family with parietal foramina determined by an ALX4 homeobox gene mutation. American Journal of Neuroradiology, 2004, 25, 1836-9.	2.4	19
89	Is Shwachman syndrome (McKusick 26040) a chromosome breakage syndrome?. Human Genetics, 1991, 87, 106-107.	3.8	18
90	Detection of deletions at 7q11.23 in Williams-Beuren syndrome by polymorphic markers. Clinics, 2011, 66, 959-964.	1.5	18

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91	Nutritional aspects of Noonan syndrome and Noonanâ€related disorders. American Journal of Medical Genetics, Part A, 2016, 170, 1525-1531.	1.2	18
92	Molecular analysis of the CTSK gene in a cohort of 33 Brazilian families with pycnodysostosis from a cluster in a Brazilian Northeast region. European Journal of Medical Research, 2016, 21, 33.	2.2	18
93	Efficient detection of copyâ€number variations using exome data: Batchâ€and sexâ€based analyses. Human Mutation, 2021, 42, 50-65.	2.5	18
94	Ocular manifestations of Noonan syndrome. Ophthalmic Genetics, 2012, 33, 1-5.	1.2	17
95	Dental Evaluation of Kabuki Syndrome Patients. Cleft Palate-Craniofacial Journal, 2009, 46, 668-673.	0.9	16
96	Clinical, cytogenetic, and molecular characterization of six patients with ring chromosomes 22, including one with concomitant 22q11.2 deletion. American Journal of Medical Genetics, Part A, 2014, 164, 1659-1665.	1.2	16
97	Insights from the genetic characterization of central precocious puberty associated with multiple anomalies. Human Reproduction, 2021, 36, 506-518.	0.9	16
98	Mutational Screening of FGFR1, CER1, and CDON in a Large Cohort of Trigonocephalic Patients. Cleft Palate-Craniofacial Journal, 2006, 43, 148-151.	0.9	15
99	KRAS gene mutations in Noonan syndrome familial cases cluster in the vicinity of the switch II region of the Gâ€domain: Report of another family with metopic craniosynostosis. American Journal of Medical Genetics, Part A, 2012, 158A, 1178-1184.	1.2	15
100	Challenges in the Orthodontic Treatment of a Patient with Pycnodysostosis. Cleft Palate-Craniofacial Journal, 2014, 51, 735-739.	0.9	15
101	Long-term follow-up of a female with congenital adrenal hyperplasia due to P450-oxidoreductase deficiency. Archives of Endocrinology and Metabolism, 2016, 60, 500-504.	0.6	15
102	Mowat-Wilson syndrome: neurological and molecular study in seven patients. Arquivos De Neuro-Psiquiatria, 2015, 73, 12-17.	0.8	14
103	Co-occurring PTPN11 and SOS1 gene mutations in Noonan syndrome: does this predict a more severe phenotype?. Arquivos Brasileiros De Endocrinologia E Metabologia, 2010, 54, 717-722.	1.3	14
104	Investigation of copy number variation in children with conotruncal heart defects. Arquivos Brasileiros De Cardiologia, 2014, 104, 24-31.	0.8	14
105	Hydronephrosis in Schinzel-Giedion syndrome: an important clue for the diagnosis. Revista Do Hospital Das Clinicas, 2004, 59, 89-92.	0.5	13
106	Renal and urinary findings in 20 patients with Williams-Beuren syndrome diagnosed by fluorescence in situ hybridization (FISH). Revista Do Hospital Das Clinicas, 2004, 59, 266-272.	0.5	13
107	MLPA analysis in 30 Sotos syndrome patients revealed one total NSD1 deletion and two partial deletions not previously reported. European Journal of Medical Genetics, 2009, 52, 333-336.	1.3	13
108	Complex structural rearrangement features suggesting chromoanagenesis mechanism in a case of 1p36 deletion syndrome. Molecular Genetics and Genomics, 2014, 289, 1037-1043.	2.1	13

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109	Atypical 581-kb 22q11.21 Deletion in a Patient with Oculo-Auriculo-Vertebral Spectrum Phenotype. Cytogenetic and Genome Research, 2015, 147, 130-134.	1.1	13
110	Williams-Beuren Syndrome: A Clinical Study of 55 Brazilian Patients and the Diagnostic Use of MLPA. BioMed Research International, 2015, 2015, 1-6.	1.9	13
111	Novel fibronectin mutations and expansion of the phenotype in spondylometaphyseal dysplasia with â€œcorner fracturesâ€• Bone, 2019, 121, 163-171.	2.9	13
112	Proteus syndrome: report of a case with recurrent abdominal lipomatosis. Journal of Pediatric Surgery, 2009, 44, e1-e3.	1.6	12
113	Copy Number Variations on Chromosome 4q26â€“27 Are Associated with Cantu Syndrome. Dermatology, 2011, 223, 316-320.	2.1	12
114	Tegumentary manifestations of Noonan and Noonan-related syndromes. Clinics, 2013, 68, 1079-1083.	1.5	12
115	Advantages of early replacement therapy for mucopolysaccharidosis type VI: echocardiographic follow-up of siblings. Cardiology in the Young, 2014, 24, 229-235.	0.8	12
116	Clinical description of 41 Brazilian patients with oculo-auriculo-vertebral dysplasia. Revista Da AssociaÃ§Ã£o MÃ©dica Brasileira, 2016, 62, 202-206.	0.7	12
117	FenÃ³tipo comportamental e cognitivo de crianÃ§as e adolescentes com SÃndrome de Williams-Beuren. PrÃ³-fono: Revista De AtualizaÃ§Ã£o CientÃfica, 2010, 22, 215-220.	0.5	11
118	GNPTAB missense mutations cause loss of GlcNAc-1-phosphotransferase activity in mucolipidosis type II through distinct mechanisms. International Journal of Biochemistry and Cell Biology, 2017, 92, 90-94.	2.8	11
119	Mucopolysaccharidosis type I, II and VI and response to enzyme replacement therapy: Results from a single-center case series study. Intractable and Rare Diseases Research, 2017, 6, 183-190.	0.9	11
120	Hemorrhagic stroke and renovascular hypertension with Grange syndrome arising from a novel pathogenic variant in YY1AP1. Journal of Human Genetics, 2019, 64, 885-890.	2.3	11
121	Mucopolysaccharidosis VII in Brazil: natural history and clinical findings. Orphanet Journal of Rare Diseases, 2021, 16, 238.	2.7	11
122	FenÃ³tipo comportamental de crianÃ§as e adolescentes com sÃndrome de Prader-Willi. Revista Paulista De Pediatria, 2010, 28, 63-69.	1.0	10
123	Ring chromosome 10: report on two patients and review of the literature. Journal of Applied Genetics, 2013, 54, 35-41.	1.9	10
124	Investigation of selected genomic deletions and duplications in a cohort of 338 patients presenting with syndromic obesity by multiplex ligation-dependent probe amplification using synthetic probes. Molecular Cytogenetics, 2014, 7, 75.	0.9	10
125	Cytogenomic delineation and clinical follow-up of 10 Brazilian patients with Pallister-Killian syndrome. Molecular Cytogenetics, 2015, 8, 43.	0.9	10
126	Cytogenomic assessment of the diagnosis of 93 patients with developmental delay and multiple congenital abnormalities: The Brazilian experience. Clinics, 2017, 72, 526-537.	1.5	10

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127	Phenotypeâ€“genotype analysis of 242 individuals with <scp>RASopathies</scp>: 18â€year experience of a tertiary center in Brazil. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 896-911.	1.6	10
128	Atypical Deletion in Williamsâ€“Beuren Syndrome Critical Region Detected by MLPA in a Patient with Supravalvular Aortic Stenosis and Learning Difficulty. Journal of Genetics and Genomics, 2012, 39, 571-574.	3.9	9
129	Discrepant outcomes in two Brazilian patients with Bloom syndrome and Wilmsâ€™ tumor: two case reports. Journal of Medical Case Reports, 2013, 7, 284.	0.8	9
130	Steric Clash in the SET Domain of Histone Methyltransferase NSD1 as a Cause of Sotos Syndrome and Its Genetic Heterogeneity in a Brazilian Cohort. Genes, 2016, 7, 96.	2.4	9
131	Further delineation of Char syndrome. Pediatrics International, 2000, 42, 85-88.	0.5	8
132	Single-Nucleotide Polymorphism Array-Based Characterization of Ring Chromosome 18. Journal of Pediatrics, 2013, 163, 1174-1178.e3.	1.8	8
133	CD4+CD25highFoxp3+ Treg deficiency in a Brazilian patient with Gaucher disease and lupus nephritis. Human Immunology, 2016, 77, 196-200.	2.4	8
134	Diagnosis and management of systemic hypertension due to renovascular and aortic stenosis in patients with Williams-Beuren syndrome. Revista Da AssociaÃ§Ã£o MÃ©dica Brasileira, 2018, 64, 723-728.	0.7	8
135	Clinical Characterization of Mucolipidoses II and III: A Multicenter Study. Journal of Pediatric Genetics, 2019, 08, 198-204.	0.7	8
136	Downregulation of genes outside the deleted region in individuals with 22q11.2 deletion syndrome. Human Genetics, 2019, 138, 93-103.	3.8	8
137	Phenotypic and mutational spectrum of ROR2 â€related Robinow syndrome. Human Mutation, 2022, 43, 900-918.	2.5	8
138	Growth and Clinical Characteristics of Children with Floating-Harbor Syndrome: Analysis of Current Original Data and a Review of the Literature. Hormone Research in Paediatrics, 2019, 92, 115-123.	1.8	7
139	Gene expression profile suggesting immunological dysregulation in two Brazilian Bloom's syndrome cases. Molecular Genetics & Genomic Medicine, 2020, 8, e1133.	1.2	7
140	Long-term impact of early initiation of enzyme replacement therapy in 34 MPS VI patients: A resurvey study. Molecular Genetics and Metabolism, 2021, 133, 94-99.	1.1	7
141	Infantile autism and 47,XYY karyotype. Arquivos De Neuro-Psiquiatria, 2009, 67, 717-718.	0.8	7
142	Nephrogenic Diabetes Insipidus (NDI): Clinical, Laboratory and Genetic Characterization of Five Brazilian Patients. Clinics, 2009, 64, 409-414.	1.5	6
143	The first cardiac transplant experience in a patient with mucopolysaccharidosis. Cardiovascular Pathology, 2012, 21, 358-360.	1.6	6
144	Role of SNAP29, LZTR1 and P2RXL1 genes on immune regulation in a patient with atypical 0.5Mb deletion in 22q11.2 region. Clinical Immunology, 2012, 145, 55-58.	3.2	6

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145	Intragenic Deletion in the <i>LIFR</i> Gene in a Long-Term Survivor with StÃ¼ve-Wiedemann Syndrome. Molecular Syndromology, 2015, 6, 87-90.	0.8	6
146	Subtelomeric Copy Number Variations: The Importance of 4p/4q Deletions in Patients with Congenital Anomalies and Developmental Disability. Cytogenetic and Genome Research, 2016, 149, 241-246.	1.1	6
147	Herpetiform keratitis and palmoplantar hyperkeratosis: warning signs for Richner-Hanhart syndrome. Journal of Inherited Metabolic Disease, 2017, 40, 461-462.	3.6	6
148	Large deletion in PIGL: a common mutational mechanism in CHIME syndrome?. Genetics and Molecular Biology, 2018, 41, 85-91.	1.3	6
149	Mucopolysaccharidosis type VI: case report with first neonatal presentation with ascites fetalis and rapidly progressive cardiac manifestation. BMC Medical Genetics, 2020, 21, 37.	2.1	6
150	Genotypeâ€“phenotype studies in a large cohort of Brazilian patients with Hunter syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 349-356.	1.6	6
151	Frequency of carriers for rare recessive Mendelian diseases in a Brazilian cohort of 320 patients. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 364-372.	1.6	6
152	Dermal melanocytosis associated with GM1-gangliosidosis type 1. Acta Dermato-Venereologica, 2006, 86, 156-158.	1.3	6
153	Obesity in pycnodysostosis due to UPD1: Possible effect of an imprinted gene on chromosome 1. American Journal of Medical Genetics, Part A, 2011, 155, 1483-1486.	1.2	5
154	The Effects of Oxybutynin on Urinary Symptoms in Children with Williams-Beuren Syndrome. Journal of Urology, 2012, 188, 253-257.	0.4	5
155	Menkes disease: importance of diagnosis with molecular analysis in the neonatal period. Revista Da AssociaÃ§Ã£o MÃ©dica Brasileira, 2015, 61, 407-410.	0.7	5
156	Post-mortem cytogenomic investigations in patients with congenital malformations. Experimental and Molecular Pathology, 2016, 101, 116-123.	2.1	5
157	Multimodal image analysis of the retina in Hunter syndrome (mucopolysaccharidosis type II): Case report. Ophthalmic Genetics, 2018, 39, 103-107.	1.2	5
158	Cognitive and behavioral profile of Williams Syndrome toddlers. CoDAS, 2018, 30, e20170188.	0.7	5
159	Breakpoint delineation in 5pâ€•patients leads to new insights about microcephaly and the typical highâ€pitched cry. Molecular Genetics & Genomic Medicine, 2020, 8, e957.	1.2	5
160	Truncating variants in the SHANK1 gene are associated with a spectrum of neurodevelopmental disorders. Genetics in Medicine, 2021, 23, 1912-1921.	2.4	5
161	Delimitation of duplicated segments and identification of their parental origin in two partial chromosome 3p duplications. American Journal of Medical Genetics Part A, 2002, 113, 144-150.	2.4	4
162	A clinical follow-up of 35 Brazilian patients with Prader-Willi Syndrome. Clinics, 2012, 67, 917-921.	1.5	4

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