Chong Ae Kim

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3235421/publications.pdf

Version: 2024-02-01

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	<scp>Cardiovascular findings in Williamsâ€“Beuren Syndrome</scp>: Experience of a single center with 127 cases. American Journal of Medical Genetics, Part A, 2022, 188, 676-682.	1.2	4
2	Sanfilippo syndrome type B: Analysis of patients diagnosed by the <scp>MPS</scp> Brazil Network. American Journal of Medical Genetics, Part A, 2022, 188, 760-767.	1.2	3
3	The recurrent homozygous translation start site variant in CCDC134 in an individual with severe osteogenesis imperfecta of nonâ€Morrocan ancestry. American Journal of Medical Genetics, Part A, 2022, , .	1.2	3
4	Vertebral segmentation defects in a Brazilian cohort: Clinical and molecular analysis focused on spondylocostal dysostosis. Clinical Genetics, 2022, 101, 476-478.	2.0	0
5	Frequency of carriers for rare metabolic diseases in a Brazilian cohort of 320 patients. Molecular Biology Reports, 2022, 49, 3911-3918.	2.3	2
6	Phenotypic and mutational spectrum of ROR2 â€related Robinow syndrome. Human Mutation, 2022, 43, 900-918.	2.5	8
7	Parental segregation study reveals rare benign and likely benign variants in a Brazilian cohort of rare diseases. Scientific Reports, 2022, 12, 7764.	3.3	1
8	Cri-du-Chat Syndrome: Revealing a Familial Atypical Deletion in 5p. Molecular Syndromology, 2022, 13, 527-536.	0.8	0
9	Novel rearrangements between different chromosomes with direct impact on the diagnosis of 5p- syndrome. Clinics, 2022, 77, 100045.	1.5	2
10	Back Cover, Volume 43, Issue 7. Human Mutation, 2022, 43, .	2.5	0
11	Insights from the genetic characterization of central precocious puberty associated with multiple anomalies. Human Reproduction, 2021, 36, 506-518.	0.9	16
12	Efficient detection of copyâ€number variations using exome data: Batchâ€and sexâ€based analyses. Human Mutation, 2021, 42, 50-65.	2.5	18
13	Exome sequencing and targeted gene panels: a simulated comparison of diagnostic yield using data from 158 patients with rare diseases. Genetics and Molecular Biology, 2021, 44, 20210061.	1.3	4
14	Abnormal auditory event-related potentials in Williams syndrome. European Journal of Medical Genetics, 2021, 64, 104163.	1.3	2
15	Cerebellofaciodental syndrome in an adult patient: Expanding the phenotypic and natural history characteristics. American Journal of Medical Genetics, Part A, 2021, 185, 1561-1568.	1.2	4
16	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
17	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
18	Mucopolysaccharidosis VII in Brazil: natural history and clinical findings. Orphanet Journal of Rare Diseases, 2021, 16, 238.	2.7	11

#	Article	IF	Citations
19	Atypical, severe hypertrophic cardiomyopathy in a newborn presenting Noonan syndrome harboring a recurrent heterozygous <scp>MRAS</scp> variant. American Journal of Medical Genetics, Part A, 2021, 185, 3099-3103.	1.2	4
20	Williams syndrome. Nature Reviews Disease Primers, 2021, 7, 42.	30.5	103
21	Truncating variants in the SHANK1 gene are associated with a spectrum of neurodevelopmental disorders. Genetics in Medicine, 2021, 23, 1912-1921.	2.4	5
22	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
23	Novel CLTC variants cause new brain and kidney phenotypes. Journal of Human Genetics, 2021, , .	2.3	4
24	Auditory hypersensitivity in Williams syndrome. International Journal of Pediatric Otorhinolaryngology, 2021, 146, 110740.	1.0	3
25	Twentyâ€year followâ€up of the facial phenotype of Brazilian patients with Sotos syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3916-3923.	1.2	0
26	Congenital limb deficiency: Genetic investigation of 44 individuals presenting mainly longitudinal defects in isolated or syndromic forms. Clinical Genetics, 2021, 100, 615-623.	2.0	4
27	A Brazilian case arising from a homozygous canonical splice site <scp>SLC35A3</scp> variant leading to an inâ€frame deletion. Clinical Genetics, 2021, 99, 607-608.	2.0	2
28	Nationwide questionnaire data of 229 Williams-Beuren syndrome patients using WhatsApp tool. Arquivos De Neuro-Psiquiatria, 2021, 79, 950-956.	0.8	1
29	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
30	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
31	Expanding the role of SETD5 haploinsufficiency in neurodevelopment and neuroblastoma. Pediatric Blood and Cancer, 2020, 67, e28376.	1.5	3
32	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
33	Lymphoproliferative disorder with polyautoimmunity and hypogammaglobulinemia: An unusual presentation of 22q11.2 deletion syndrome. Clinical Immunology, 2020, 220, 108590.	3.2	1
34	Gene expression profile suggesting immunological dysregulation in two Brazilian Bloom's syndrome cases. Molecular Genetics & Genomic Medicine, 2020, 8, e1133.	1.2	7
35	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
36	Genetic investigation of patients with tall stature. European Journal of Endocrinology, 2020, 182, 139-147.	3.7	3

#	Article	IF	Citations
37	Targeted massively parallel sequencing for congenital generalized lipodystrophy. Archives of Endocrinology and Metabolism, 2020, 64, 559-566.	0.6	0
38	SUN-081 High Throughput Genetic Analysis Revealed Novel Genomic Loci and Candidate Genes Involved in Central Precocious Puberty Associated with Complex Phenotypes. Journal of the Endocrine Society, 2020, 4, .	0.2	0
39	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
40	Comprehensive genetic analysis of 57 families with clinically suspected Cornelia de Lange syndrome. Journal of Human Genetics, 2019, 64, 967-978.	2.3	43
41	Autosomal-Recessive Mutations in MESD Cause Osteogenesis Imperfecta. American Journal of Human Genetics, 2019, 105, 836-843.	6.2	36
42	Genetic Disorders in Prenatal Onset Syndromic Short Stature Identified by Exome Sequencing. Journal of Pediatrics, 2019, 215, 192-198.	1.8	36
43	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
44	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
45	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
46	Clinical Characterization of Mucolipidoses II and III: A Multicenter Study. Journal of Pediatric Genetics, 2019, 08, 198-204.	0.7	8
47	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
48	Downregulation of genes outside the deleted region in individuals with 22q11.2 deletion syndrome. Human Genetics, 2019, 138, 93-103.	3.8	8
49	Novel fibronectin mutations and expansion of the phenotype in spondylometaphyseal dysplasia with â€œcorner fracturesâ€• Bone, 2019, 121, 163-171.	2.9	13
50	Audiological characteristics in mucopolysaccharidosis: a systematic literature review. Revista CEFAC: ActualizaÃ§Ã£o CientÃfica Em Fonoaudiologia, 2019, 21, .	0.1	3
51	Chromosomal microarray analysis in the genetic evaluation of 279 patients with syndromic obesity. Molecular Cytogenetics, 2018, 11, 14.	0.9	35
52	Multimodal image analysis of the retina in Hunter syndrome (mucopolysaccharidosis type II): Case report. Ophthalmic Genetics, 2018, 39, 103-107.	1.2	5
53	Recurrent Copy Number Variants Associated with Syndromic Short Stature of Unknown Cause. Hormone Research in Paediatrics, 2018, 89, 13-21.	1.8	29
54	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

#	Article	IF	Citations
55	Large deletion in PIGL: a common mutational mechanism in CHIME syndrome?. Genetics and Molecular Biology, 2018, 41, 85-91.	1.3	6
56	Using a deep learning network to recognise low back pain in static standing. Ergonomics, 2018, 61, 1374-1381.	2.1	36
57	Cognitive and behavioral profile of Williams Syndrome toddlers. CoDAS, 2018, 30, e20170188.	0.7	5
58	Natural history of 39 patients with Achondroplasia. Clinics, 2018, 73, e324.	1.5	20
59	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
60	Autosomal dominant frontometaphyseal dysplasia: Delineation of the clinical phenotype. American Journal of Medical Genetics, Part A, 2017, 173, 1739-1746.	1.2	24
61	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
62	Patients with a Kabuki syndrome phenotype demonstrate DNA methylation abnormalities. European Journal of Human Genetics, 2017, 25, 1335-1344.	2.8	52
63	Herpetiform keratitis and palmoplantar hyperkeratosis: warning signs for Richner-Hanhart syndrome. Journal of Inherited Metabolic Disease, 2017, 40, 461-462.	3.6	6
64	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
65	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
66	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
67	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
68	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
69	Nutritional aspects of Noonan syndrome and Noonanâ€related disorders. American Journal of Medical Genetics, Part A, 2016, 170, 1525-1531.	1.2	18
70	Post-mortem cytogenomic investigations in patients with congenital malformations. Experimental and Molecular Pathology, 2016, 101, 116-123.	2.1	5
71	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
72	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

#	Article	IF	Citations
73	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
74	CD4+CD25highFoxp3+ Treg deficiency in a Brazilian patient with Gaucher disease and lupus nephritis. Human Immunology, 2016, 77, 196-200.	2.4	8
75	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
76	Rare genomic rearrangement in a boy with Williamsâ€“Beuren syndrome associated to XYY syndrome and intriguing behavior. American Journal of Medical Genetics, Part A, 2015, 167, 3197-3203.	1.2	2
77	Cytogenomic delineation and clinical follow-up of 10 Brazilian patients with Pallister-Killian syndrome. Molecular Cytogenetics, 2015, 8, 43.	0.9	10
78	Recurrence of frontometaphyseal dysplasia in two sisters with a mutation in FLNA and an atypical paternal phenotype: Insights into genotypeâ€“phenotype correlation. American Journal of Medical Genetics, Part A, 2015, 167, 1161-1164.	1.2	2
79	Mowat-Wilson syndrome: neurological and molecular study in seven patients. Arquivos De Neuro-Psiquiatria, 2015, 73, 12-17.	0.8	14
80	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
81	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
82	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
83	Multicentric study on the diagnosis of Fabry's disease using angiokeratoma biopsy registries. International Journal of Dermatology, 2015, 54, e241-4.	1.0	2
84	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
85	Rare variants inSOS2andLZTR1are associated with Noonan syndrome. Journal of Medical Genetics, 2015, 52, 413-421.	3.2	187
86	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
87	Lipoid proteinosis: Rare case confirmed by ECM1 mutation detection. International Journal of Pediatric Otorhinolaryngology, 2014, 78, 2314-2315.	1.0	3
88	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
89	Remote spinal cord injury in mucopolysaccharidosis type IVA after cervical decompression. Neurology, 2014, 82, 1382-1383.	1.1	3
90	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

#	Article	IF	Citations
91	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
92	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
93	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
94	Further evidence of the importance of RIT1 in Noonan syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 2952-2957.	1.2	53
95	NK and B cell deficiency in a MPS type II family with novel mutation in the IDS gene. Clinical Immunology, 2014, 154, 100-104.	3.2	3
96	Congenital genitourinary abnormalities in children with Williamsâ€“Beuren syndrome. Journal of Pediatric Urology, 2014, 10, 804-809.	1.1	22
97	Mutations in PCYT1A Cause Spondylometaphyseal Dysplasia with Cone-Rod Dystrophy. American Journal of Human Genetics, 2014, 94, 113-119.	6.2	39
98	New insights in mucopolysaccharidosis type VI: Neurological perspective. Brain and Development, 2014, 36, 585-592.	1.1	20
99	Challenges in the Orthodontic Treatment of a Patient with Pycnodysostosis. Cleft Palate-Craniofacial Journal, 2014, 51, 735-739.	0.9	15
100	Case Report Johanson-Blizzard syndrome: a report of gender-discordant twins with a novel UBR1 mutation. Genetics and Molecular Research, 2014, 13, 4159-4164.	0.2	1
101	Investigation of copy number variation in children with conotruncal heart defects. Arquivos Brasileiros De Cardiologia, 2014, 104, 24-31.	0.8	14
102	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
103	A Novel Mutation in HPRT1 Gene Causing Variant Form of Lesch-Nyhan Disease. Pediatric Neurology, 2013, 49, e5-e7.	2.1	0
104	Single-Nucleotide Polymorphism Array-Based Characterization of Ring Chromosome 18. Journal of Pediatrics, 2013, 163, 1174-1178.e3.	1.8	8
105	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
106	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
107	Ring chromosome 10: report on two patients and review of the literature. Journal of Applied Genetics, 2013, 54, 35-41.	1.9	10
108	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

#	Article	IF	Citations
109	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
110	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
111	Tegumentary manifestations of Noonan and Noonan-related syndromes. Clinics, 2013, 68, 1079-1083.	1.5	12
112	Estresse em crianÃ§as e adolescentes com SÃndrome de Williams-Beuren em idade escolar. Psicologia Escolar E Educacional, 2013, 17, 105-112.	0.3	1
113	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
114	The Effects of Oxybutynin on Urinary Symptoms in Children with Williams-Beuren Syndrome. Journal of Urology, 2012, 188, 253-257.	0.4	5
115	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
116	Ocular manifestations of Noonan syndrome. Ophthalmic Genetics, 2012, 33, 1-5.	1.2	17
117	Associations among genotype, clinical phenotype, and intracellular localization of trafficking proteins in ARC syndrome. Human Mutation, 2012, 33, 1656-1664.	2.5	74
118	The first cardiac transplant experience in a patient with mucopolysaccharidosis. Cardiovascular Pathology, 2012, 21, 358-360.	1.6	6
119	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
120	A clinical follow-up of 35 Brazilian patients with Prader-Willi Syndrome. Clinics, 2012, 67, 917-921.	1.5	4
121	Copy number variation in Williams-Beuren syndrome: suitable diagnostic strategy for developing countries. BMC Research Notes, 2012, 5, 13.	1.4	19
122	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
123	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
124	Autoimmune disease and multiple autoantibodies in 42 patients with RASopathies. American Journal of Medical Genetics, Part A, 2012, 158A, 1077-1082.	1.2	73
125	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
126	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

#	Article	IF	Citations
127	A Possible Role of Different PTPN Genes in Immune Regulation. Scandinavian Journal of Immunology, 2012, 75, 540-541.	2.7	2
128	Multiple, diffuse schwannomas in a RASopathy phenotype patient with germline KRAS mutation: a causal relationship?. Clinical Genetics, 2012, 81, 595-597.	2.0	19
129	Mutations, Clinical Findings and Survival Estimates in South American Patients with X-Linked Adrenoleukodystrophy. PLoS ONE, 2012, 7, e34195.	2.5	24
130	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
131	Detection of deletions at 7q11.23 in Williams-Beuren syndrome by polymorphic markers. Clinics, 2011, 66, 959-964.	1.5	18
132	Mutations in NOTCH2 cause Hajdu-Cheney syndrome, a disorder of severe and progressive bone loss. Nature Genetics, 2011, 43, 303-305.	21.4	291
133	Mechanisms of ring chromosome formation, ring instability and clinical consequences. BMC Medical Genetics, 2011, 12, 171.	2.1	106
134	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
135	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
136	Copy Number Variations on Chromosome 4q26â€“27 Are Associated with Cantu Syndrome. Dermatology, 2011, 223, 316-320.	2.1	12
137	An Illustrative Case of Neurofibromatosis Type 1 and NF1 Microdeletion. Molecular Syndromology, 2010, 1, 133-135.	0.8	3
138	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
139	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
140	Two distinct regions in 2q24.2â€q24.3 associated with idiopathic epilepsy. Epilepsia, 2010, 51, 2457-2460.	5.1	43
141	Mucopolysaccharidosis I, II, and VI: brief review and guidelines for treatment. Genetics and Molecular Biology, 2010, 33, 589-604.	1.3	150
142	DiGeorge Syndrome: a not so rare disease. Clinics, 2010, 65, 865-869.	1.5	40
143	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
144	FenÃ³tipo comportamental de crianÃ§as e adolescentes com sÃndrome de Prader-Willi. Revista Paulista De Pediatria, 2010, 28, 63-69.	1.0	10

#	Article	IF	Citations
145	Echocardiographic study of paediatric patients with mucopolysaccharidosis. Cardiology in the Young, 2010, 20, 254-261.	0.8	77
146	Williams-Beuren Syndrome: Diagnosis by Polymorphic Markers. Genetic Testing and Molecular Biomarkers, 2010, 14, 209-214.	0.7	2
147	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
148	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
149	Nephrogenic Diabetes Insipidus (NDI): Clinical, Laboratory and Genetic Characterization of Five Brazilian Patients. Clinics, 2009, 64, 409-414.	1.5	6
150	Spondylocostal Dysostosis Associated with Methylmalonic Aciduria. Genetic Testing and Molecular Biomarkers, 2009, 13, 181-183.	0.7	1
151	Further delineation of nonhomologous-based recombination and evidence for subtelomeric segmental duplications in 1p36 rearrangements. Human Genetics, 2009, 125, 551-563.	3.8	25
152	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
153	Proteus syndrome: report of a case with recurrent abdominal lipomatosis. Journal of Pediatric Surgery, 2009, 44, e1-e3.	1.6	12
154	A Duplex Allele-Specific Amplification PCR to Detect SMN1 Deletion. Genetic Testing and Molecular Biomarkers, 2009, 13, 205-208.	0.7	2
155	Dental Evaluation of Kabuki Syndrome Patients. Cleft Palate-Craniofacial Journal, 2009, 46, 668-673.	0.9	16
156	Infantile autism and 47,XYY karyotype. Arquivos De Neuro-Psiquiatria, 2009, 67, 717-718.	0.8	7
157	Mucopolysaccharidoses in Brazil: What happens from birth to biochemical diagnosis?. American Journal of Medical Genetics, Part A, 2008, 146A, 1741-1747.	1.2	63
158	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
159	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
160	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
161	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
162	Williams Syndrome: development of a new scoring system for clinical diagnosis. Clinics, 2007, 62, 159-166.	1.5	23

#	Article	IF	Citations
163	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
164	A clinical study of 77 patients with mucopolysaccharidosis type II. Acta Paediatrica, International Journal of Paediatrics, 2007, 96, 63-70.	1.5	112
165	Further evidence of genetic heterogeneity in Costello syndrome: involvement of the KRAS gene. Journal of Human Genetics, 2007, 52, 521-526.	2.3	36
166	A molecular and clinical study of Larsen syndrome caused by mutations in FLNB. Journal of Medical Genetics, 2006, 44, 89-98.	3.2	102
167	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
168	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
169	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
170	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
171	Exuberant Juvenile Hyaline Fibromatosis in Two Patients. Pediatric Dermatology, 2006, 23, 458-464.	0.9	28
172	SHOX mutations in idiopathic short stature and Leri-Weill dyschondrosteosis: frequency and phenotypic variability. Clinical Endocrinology, 2006, 66, 061031010617004-???.	2.4	75
173	Cockayne syndrome type A: novel mutations in eight typical patients. Journal of Human Genetics, 2006, 51, 701-705.	2.3	28
174	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
175	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
176	Dermal melanocytosis associated with GM1-gangliosidosis type 1. Acta Dermato-Venereologica, 2006, 86, 156-158.	1.3	6
177	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
178	CHILD Syndrome Caused by a Deletion of Exons 6â€“8 of the <i>NSDHL</i> Gene. Dermatology, 2005, 211, 155-158.	2.1	25
179	Hydronephrosis in Schinzel-Giedion syndrome: an important clue for the diagnosis. Revista Do Hospital Das Clinicas, 2004, 59, 89-92.	0.5	13
180	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

#	Article	IF	Citations
181	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
182	Mutations in the gene encoding filamin B disrupt vertebral segmentation, joint formation and skeletogenesis. Nature Genetics, 2004, 36, 405-410.	21.4	252
183	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
184	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
185	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
186	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
187	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
188	Prevalence of Mutations in AGPAT2 Among Human Lipodystrophies. Diabetes, 2003, 52, 1573-1578.	0.6	87
189	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
190	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
191	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
192	LDL Receptor-Related Protein 5 (LRP5) Affects Bone Accrual and Eye Development. Cell, 2001, 107, 513-523.	28.9	2,055
193	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
194	Haploinsufficiency of the human homeobox gene ALX4 causes skull ossification defects. Nature Genetics, 2001, 27, 17-18.	21.4	142
195	AEC Syndrome and CHAND Syndrome: Further Evidence of Clinical Overlapping in the Ectodermal Dysplasias. Pediatric Dermatology, 2000, 17, 218-221.	0.9	24
196	Further delineation of Char syndrome. Pediatrics International, 2000, 42, 85-88.	0.5	8
197	CACP, encoding a secreted proteoglycan, is mutated in camptodactyly-arthropathy-coxa vara-pericarditis syndrome. Nature Genetics, 1999, 23, 319-322.	21.4	286
198	Congenital abnormalities in Brazilian children associated with misoprostol misuse in first trimester of pregnancy. Lancet, The, 1998, 351, 1624-1627.	13.7	225

#	Article	IF	Citations
199	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
200	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
201	Is Shwachman syndrome (McKusick 26040) a chromosome breakage syndrome?. Human Genetics, 1991, 87, 106-107.	3.8	18
202	Caudal â€œregressionâ€•anomaly in a boy born to a pancreatectomized mother. American Journal of Medical Genetics Part A, 1985, 21, 205-209.	2.4	3
203	Intellectual performance profi le of a sample of children and adolescents from Brazil with 22q11.2 Deletion Syndrome (22q11.2DS) based on the Wechsler Scale. Estudos De Psicologia (Campinas), 0, 36, .	0.8	0