Kenji Kurosawa

List of Publications by Year
in descending order

214 papers	5,658 citations	117453 34 h-index	102304 66 g-index
219 all docs	219 docs citations	219 times ranked	7366 citing authors

#	Article	IF	Citations
1	Germline mutations in HRAS proto-oncogene cause Costello syndrome. Nature Genetics, 2005, 37, 1038-1040.	9.4	597
2	Germline KRAS and BRAF mutations in cardio-facio-cutaneous syndrome. Nature Genetics, 2006, 38, 294-296.	9.4	517
3	Gain-of-Function Mutations in RIT1 Cause Noonan Syndrome, a RAS/MAPK Pathway Syndrome. American Journal of Human Genetics, 2013, 93, 173-180.	2.6	279
4	Deletions and epimutations affecting the human 14q32.2 imprinted region in individuals with paternal and maternal upd(14)-like phenotypes. Nature Genetics, 2008, 40, 237-242.	9.4	266
5	Mutations in transcriptional regulator ATRX establish the functional significance of a PHD-like domain. Nature Genetics, 1997, 17, 146-148.	9.4	196
6	Spectrum of MLL2 (ALR) mutations in 110 cases of Kabuki syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 1511-1516.	0.7	160
7	Fifty microdeletions among 112 cases of Sotos syndrome: Low copy repeats possibly mediate the common deletion. Human Mutation, 2003, 22, 378-387.	1.1	121
8	Functional analysis of PTPN11/SHP-2 mutants identified in Noonan syndrome and childhood leukemia. Journal of Human Genetics, 2005, 50, 192-202.	1.1	113
9	Molecular and clinical characterization of cardio-facio-cutaneous (CFC) syndrome: Overlapping clinical manifestations with Costello syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 799-807.	0.7	96
10	Molecular and clinical analysis ofRAF1in Noonan syndrome and related disorders: dephosphorylation of serine 259 as the essential mechanism for mutant activation. Human Mutation, 2010, 31, 284-294.	1.1	96
11	Comprehensive clinical studies in 34 patients with molecularly defined UPD(14)pat and related conditions (Kagamiâ€“Ogata syndrome). European Journal of Human Genetics, 2015, 23, 1488-1498.	1.4	85
12	Spectrum of mutations and genotypeâ€“phenotype analysis in Noonan syndrome patients with RIT1 mutations. Human Genetics, 2016, 135, 209-222.	1.8	75
13	Prevalence and clinical features of Costello syndrome and cardioâ€facioâ€cutaneous syndrome in Japan: Findings from a nationwide epidemiological survey. American Journal of Medical Genetics, Part A, 2012, 158A, 1083-1094.	0.7	74
14	Targeting G-quadruplex DNA as cognitive function therapy for ATR-X syndrome. Nature Medicine, 2018, 24, 802-813.	15.2	69
15	Segmental and full paternal isodisomy for chromosome 14 in three patients: Narrowing the critical region and implication for the clinical features. American Journal of Medical Genetics, Part A, 2005, 138A, 127-132.	0.7	64
16	Paternal UPD14 is responsible for a distinctive malformation complex. American Journal of Medical Genetics Part A, 2002, 110, 268-272.	2.4	63
17	Molecular characterization of inv dup del(8p): Analysis of five cases. American Journal of Medical Genetics Part A, 2004, 128A, 133-137.	2.4	57
18	BAC array CGH reveals genomic aberrations in idiopathic mental retardation. American Journal of Medical Genetics, Part A, 2006, 140A, 205-211.	0.7	57

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19	A new case of GABA transaminase deficiency facilitated by proton MR spectroscopy. Journal of Inherited Metabolic Disease, 2010, 33, 85-90.	1.7	57
20	Graves' disease in patients with 22q11.2 deletion. Journal of Pediatrics, 2001, 139, 892-895.	0.9	54
21	Sotos syndrome associated with a de novo balanced reciprocal translocation t(5;8)(q35;q24.1). American Journal of Medical Genetics Part A, 2002, 107, 58-60.	2.4	52
22	CHARGE syndrome modeling using patient-iPSCs reveals defective migration of neural crest cells harboring CHD7 mutations. ELife, 2017, 6, .	2.8	52
23	Further delineation of the behavioral and neurologic features in Costello syndrome. American Journal of Medical Genetics Part A, 2003, 118A, 8-14.	2.4	51
24	Delineation of LZTR1 mutation-positive patients with Noonan syndrome and identification of LZTR1 binding to RAF1â€“PPP1CB complexes. Human Genetics, 2019, 138, 21-35.	1.8	50
25	Preferential Paternal Origin of Microdeletions Caused by Prezygotic Chromosome or Chromatid Rearrangements in Sotos Syndrome. American Journal of Human Genetics, 2003, 72, 1331-1337.	2.6	48
26	Mutations in CD96, a Member of the Immunoglobulin Superfamily, Cause a Form of the C (Opitz) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	2.6	48
27	Maternal Uniparental Disomy 14 Syndrome Demonstrates Prader-Willi Syndrome-Like Phenotype. Journal of Pediatrics, 2009, 155, 900-903.e1.	0.9	46
28	Epilepsy and neurological findings in 11 individuals with 1p36 deletion syndrome. Brain and Development, 2005, 27, 378-382.	0.6	45
29	Endocrine and Radiological Studies in Patients with Molecularly Confirmed CHARGE Syndrome. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 920-924.	1.8	44
30	Origin and mechanisms of formation of fetus-in-fetu: Two cases with genotype and methylation analyses. American Journal of Medical Genetics, Part A, 2006, 140A, 1737-1743.	0.7	39
31	Two patients with atypical interstitial deletions of 8p23.1: Mapping of phenotypical traits. American Journal of Medical Genetics, Part A, 2008, 146A, 1158-1165.	0.7	39
32	Mutation analysis of the SHOC2 gene in Noonan-like syndrome and in hematologic malignancies. Journal of Human Genetics, 2010, 55, 801-809.	1.1	38
33	Molecular karyotyping in 17 patients and mutation screening in 41 patients with Kabuki syndrome. Journal of Human Genetics, 2009, 54, 304-309.	1.1	37
34	Patients with a sodium channel alpha 1 gene mutation show wide phenotypic variation. Epilepsy Research, 2007, 75, 46-51.	0.8	36
35	Epidemiological, clinical, and genetic landscapes of hypomyelinating leukodystrophies. Journal of Neurology, 2014, 261, 752-758.	1.8	36
36	DisruptedSOX10regulation ofGJC2transcription causes Pelizaeus-Merzbacher-Like Disease. Annals of Neurology, 2010, 68, NA-NA.	2.8	34

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37	Paternal uniparental disomy 14 and related disorders. Epigenetics, 2012, 7, 1142-1150.	1.3	34
38	The spectrum of ZEB2 mutations causing the Mowatâ€“Wilson syndrome in Japanese populations. American Journal of Medical Genetics, Part A, 2014, 164, 1899-1908.	0.7	34
39	Recurrent de novo MAPK8IP3 variants cause neurological phenotypes. Annals of Neurology, 2019, 85, 927-933.	2.8	34
40	On the reported 8p22-p23.1 duplication in Kabuki make-up syndrome (KMS) and its absence in patients with typical KMS. American Journal of Medical Genetics Part A, 2004, 128A, 170-172.	2.4	32
41	Clinical manifestations in patients with SOS1 mutations range from Noonan syndrome to CFC syndrome. Journal of Human Genetics, 2008, 53, 834-841.	1.1	31
42	Early infantile epileptic encephalopathy associated with the disrupted gene encoding Slitâ€Robo Rho GTPase activating protein 2 (SRGAP2). American Journal of Medical Genetics, Part A, 2012, 158A, 199-205.	0.7	31
43	Four novelNIPBL mutations in Japanese patients with Cornelia de Lange syndrome. American Journal of Medical Genetics, Part A, 2005, 135A, 103-105.	0.7	30
44	Further delineation of 9q22 deletion syndrome associated with basal cell nevus (Gorlin) syndrome: Report of two cases and review of the literature. Congenital Anomalies (discontinued), 2009, 49, 8-14.	0.3	30
45	Increased oxidative stress biomarkers in the saliva of Down syndrome patients. Archives of Oral Biology, 2013, 58, 1246-1250.	0.8	30
46	Submicroscopic deletion of chromosome region 16p13.3 in a Japanese patient with Rubinstein-Taybi syndrome. American Journal of Medical Genetics Part A, 1994, 53, 352-354.	2.4	29
47	Detection of cryptic chromosome aberrations in a patient with a balanced t(1;9)(p34.2;p24) by array-based comparative genomic hybridization. American Journal of Medical Genetics, Part A, 2005, 139A, 32-36.	0.7	29
48	GPC3 mutations in seven patients with Simpsonâ€“Golabiâ€“Behmel syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1703-1707.	0.7	29
49	HRAS mutants identified in Costello syndrome patients can induce cellular senescence: possible implications for the pathogenesis of Costello syndrome. Journal of Human Genetics, 2011, 56, 707-715.	1.1	29
50	1p36 deletion syndrome associated with Praderâ€“Williâ€like phenotype. Pediatrics International, 2010, 52, 547-550.	0.2	28
51	Pregnancy outcome of fetuses with trisomy 18 identified by prenatal sonography and chromosomal analysis in a perinatal center. American Journal of Medical Genetics, Part A, 2006, 140A, 1177-1182.	0.7	27
52	Expansion of the phenotype of Kosaki overgrowth syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 2422-2427.	0.7	27
53	A novel UBE2A mutation causes X-linked intellectual disability type Nascimento. Human Genome Variation, 2017, 4, 17019.	0.4	26
54	West Syndrome in a Patient With Schinzel-Giedion Syndrome. Journal of Child Neurology, 2015, 30, 932-936.	0.7	25

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55	Epidemiology of limb-body wall complex in Japan. American Journal of Medical Genetics Part A, 1994, 51, 143-146.	2.4	24
56	Ophthalmic features of CHARGE syndrome with CHD7 mutations. American Journal of Medical Genetics, Part A, 2012, 158A, 514-518.	0.7	24
57	A postzygotic NRAS mutation in a patient with Schimmelpenning syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 2223-2225.	0.7	24
58	Congenital anomaly of cervical vertebrae is a major complication of Rubinstein-Taybi syndrome. American Journal of Medical Genetics, Part A, 2005, 135A, 130-133.	0.7	23
59	Delineation of the KIAA2022 mutation phenotype: Two patients with Xâ€linked intellectual disability and distinctive features. American Journal of Medical Genetics, Part A, 2015, 167, 1349-1353.	0.7	23
60	CTCFdeletion syndrome: clinical features and epigenetic delineation. Journal of Medical Genetics, 2017, 54, 836-842.	1.5	23
61	Comprehensive screening of CREB-binding protein gene mutations among patients with Rubinstein-Taybi syndrome using denaturing high-performance liquid chromatography. Congenital Anomalies (discontinued), 2005, 45, 125-131.	0.3	22
62	Clinical application of array-based comparative genomic hybridization by two-stage screening for 536 patients with mental retardation and multiple congenital anomalies. Journal of Human Genetics, 2011, 56, 110-124.	1.1	22
63	Patellar dislocation in Kabuki syndrome. American Journal of Medical Genetics Part A, 2002, 108, 160-163.	2.4	21
64	Genomic rearrangement at 10q24 in non-syndromic split-hand/split-foot malformation. Human Genetics, 2005, 118, 477-483.	1.8	21
65	Refining the clinical phenotype of Okurâ€“Chung neurodevelopmental syndrome. Human Genome Variation, 2018, 5, 18011.	0.4	21
66	A novel gene (FAM20B encoding glycosaminoglycan xylosylkinase) for neonatal short limb dysplasia resembling Desbuquois dysplasia. Clinical Genetics, 2019, 95, 713-717.	1.0	21
67	Update of the genotype and phenotype of <scp>KMT2D</scp> and <scp>KDM6A</scp> by genetic screening of 100 patients with clinically suspected Kabuki syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2333-2344.	0.7	21
68	A female patient with Xâ€linked Ohdo syndrome of the Maatâ€Kievitâ€Brunner phenotype caused by a novel variant of MED12. Congenital Anomalies (discontinued), 2020, 60, 91-93.	0.3	20
69	No detectable genomic aberrations by BAC array CGH in Kabuki make-up syndrome patients. American Journal of Medical Genetics, Part A, 2006, 140A, 291-293.	0.7	19
70	Male with type II autosomal recessive cutis laxa. Clinical Genetics, 1994, 45, 40-43.	1.0	19
71	Sirenomelia with a de novo balanced translocation 46,X,t(X;16)(p11.23;p12.3). Congenital Anomalies (discontinued), 2012, 52, 106-110.	0.3	19
72	A girl with 1p36 deletion syndrome and congenital fiber type disproportion myopathy. Journal of Human Genetics, 2002, 47, 0556-0559.	1.1	18

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73	Successful Endovascular Treatment of a Ruptured Superior Mesenteric Artery in a Patient with Ehlersâ€'Danlos Syndrome. Annals of Vascular Surgery, 2013, 27, 975.e1-975.e5.	0.4	18
74	The magnetic resonance imaging spectrum of Pelizaeusâ€"Merzbacher disease: A multicenter study of 19 patients. Brain and Development, 2016, 38, 571-580.	0.6	18
75	A large interstitial deletion of 17p13.1p11.2 involving the Smith-Magenis chromosome region in a girl with multiple congenital anomalies. American Journal of Medical Genetics, Part A, 2006, 140A, 88-91.	0.7	17
76	Characterization of the complex 7q21.3 rearrangement in a patient with bilateral splitâ€foot malformation and hearing loss. American Journal of Medical Genetics, Part A, 2009, 149A, 1224-1230.	0.7	17
77	5,10-Methylenetetrahydrofolate reductase deficiency with progressive polyneuropathy in an infant. Brain and Development, 2011, 33, 521-524.	0.6	17
78	Ageâ€dependent change in behavioral feature in Rubinsteinâ€Taybi syndrome. Congenital Anomalies (discontinued), 2012, 52, 82-86.	0.3	17
79	Neuroimaging findings in Joubert syndrome with C5orf42 gene mutations: A milder form of molar tooth sign and vermian hypoplasia. Journal of the Neurological Sciences, 2017, 376, 7-12.	0.3	17
80	Tetralogy of Fallot associated with pulmonary atresia and major aortopulmonary collateral arteries in a patient with interstitial deletion of 16q21â€"q22.1. American Journal of Medical Genetics, Part A, 2008, 146A, 1575-1580.	0.7	16
81	Proximal interstitial 1p36 deletion syndrome: The most proximal 3.5-Mb microdeletion identified on a dysmorphic and mentally retarded patient with inv(3)(p14.1q26.2). Brain and Development, 2009, 31, 629-633.	0.6	16
82	Brain MRI findings of older patients with Pallisterâ€"Killian syndrome. Brain and Development, 2006, 28, 34-38.	0.6	15
83	Prenatal findings of paternal uniparental disomy 14: Report of four patients. American Journal of Medical Genetics, Part A, 2010, 152A, 789-791.	0.7	15
84	Clinical Characteristics and Outcomes of MÃ¶bius Syndrome in a Children's Hospital. Pediatric Neurology, 2014, 51, 781-789.	1.0	15
85	Polymicrogyria and infantile spasms in a patient with 1p36 deletion syndrome. Brain and Development, 2011, 33, 437-441.	0.6	14
86	The incidence of hypoplasia of the corpus callosum in patients with dup (X)(q28) involving MECP2 is associated with the location of distal breakpoints. American Journal of Medical Genetics, Part A, 2012, 158A, 1292-1303.	0.7	14
87	De novo duplication of 17p13.1â€"p13.2 in a patient with intellectual disability and obesity. American Journal of Medical Genetics, Part A, 2014, 164, 1550-1554.	0.7	14
88	Fetal outcome of trisomy 18 diagnosed after 22 weeks of gestation: Experience of 123 cases at a single perinatal center. Congenital Anomalies (discontinued), 2016, 56, 35-40.	0.3	14
89	Redefining the phenotypic spectrum of de novo heterozygous CDK13 variants: Three patients without cardiac defects. European Journal of Medical Genetics, 2018, 61, 243-247.	0.7	14
90	Novel COL4A1 mutation in a fetus with early prenatal onset of schizencephaly. Human Genome Variation, 2018, 5, 4.	0.4	14

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91	A case of de novo splice site variant in SLC35A2 showing developmental delays, spastic paraplegia, and delayed myelination. Molecular Genetics & Genomic Medicine, 2019, 7, e814.	0.6	14
92	CNV analysis using whole exome sequencing identified biallelic CNVs of VPS13B in siblings with intellectual disability. European Journal of Medical Genetics, 2020, 63, 103610.	0.7	14
93	Congenital Neuroblastoma in a Patient With Partial Trisomy of 2p. Journal of Pediatric Hematology/Oncology, 2006, 28, 379-382.	0.3	13
94	First case of a Japanese girl with Myhre syndrome due to a heterozygous SMAD4 mutation. American Journal of Medical Genetics, Part A, 2012, 158A, 1982-1986.	0.7	13
95	A case of paternal uniparental isodisomy for chromosome 7 associated with overgrowth. Journal of Medical Genetics, 2018, 55, 567-570.	1.5	13
96	Tatton-Brown-Rahman syndrome with a novel DNMT3A mutation presented severe intellectual disability and autism spectrum disorder. Human Genome Variation, 2020, 7, 15.	0.4	13
97	ATP1A3 variants and slowly progressive cerebellar ataxia without paroxysmal or episodic symptoms in children. Developmental Medicine and Child Neurology, 2021, 63, 111-115.	1.1	13
98	Self-induced vomiting in X-linked Î±-thalassemia/mental retardation syndrome. American Journal of Medical Genetics Part A, 1996, 63, 505-506.	2.4	12
99	Trigonocephaly in a boy with paternally inherited deletion 22q11.2 syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 1302-1304.	0.7	12
100	Neuroradiologic Findings in Sotos Syndrome. Journal of Child Neurology, 2006, 21, 614-618.	0.7	12
101	Screening for Partial Deletions in the CREBBP Gene in Rubinsteinâ€“Taybi Syndrome Patients Using Multiplex PCR/Liquid Chromatography. Genetic Testing and Molecular Biomarkers, 2006, 10, 265-271.	1.7	12
102	Mild phenotype in Pelizaeus-Merzbacher disease caused by a PLP1-specific mutation. Brain and Development, 2010, 32, 703-707.	0.6	12
103	Heterozygous Câ€propeptide mutations in COL1A1: Osteogenesis imperfecta type IIC and dense bone variant. American Journal of Medical Genetics, Part A, 2011, 155, 2269-2273.	0.7	12
104	Congenital Omphalocele and Polyhydramnios: A Study of 52 Cases. Fetal Diagnosis and Therapy, 2011, 30, 184-188.	0.6	12
105	Novel AMER1 frameshift mutation in a girl with osteopathia striata with cranial sclerosis. Congenital Anomalies (discontinued), 2018, 58, 145-146.	0.3	12
106	Analysis of GBE1 mutations via protein expression studies in glycogen storage disease type IV: A report on a non-progressive form with a literature review. Molecular Genetics and Metabolism Reports, 2018, 17, 31-37.	0.4	12
107	Intellectual disability-associated gain-of-function mutations in CERT1 that encodes the ceramide transport protein CERT. PLoS ONE, 2020, 15, e0243980.	1.1	12
108	Prenatal diagnosis of GM2-gangliosidosis. Brain and Development, 1993, 15, 278-282.	0.6	11

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109	1p36 deletion syndrome with intestinal malrotation and annular pancreas. European Journal of Pediatrics, 2005, 164, 193-194.	1.3	11
110	Proportion of malformations and genetic disorders among cases encountered at a high-care unit in a childrenâ€™s hospital. European Journal of Pediatrics, 2012, 171, 301-305.	1.3	11
111	Microdeletion of 19p13.3 in a girl with Peutzâ€“Jeghers syndrome, intellectual disability, hypotonia, and distinctive features. American Journal of Medical Genetics, Part A, 2015, 167, 389-393.	0.7	11
112	SSADH deficiency possibly associated with enzyme activity-reducing SNPs. Brain and Development, 2016, 38, 871-874.	0.6	11
113	The severe clinical phenotype for a heterozygous Fabry female patient correlates to the methylation of non-mutated allele associated with chromosome 10q26 deletion syndrome. Molecular Genetics and Metabolism, 2017, 120, 173-179.	0.5	11
114	Chromosome aberrations in Rubinsteinâ€Taybi syndrome. Clinical Genetics, 1993, 43, 215-216.	1.0	10
115	A case of SjÃ¶gren-Larsson syndrome with minimal MR imaging findings facilitated by proton spectroscopy. Pediatric Radiology, 2012, 42, 380-382.	1.1	10
116	Trends in occurrence of twin births in Japan. American Journal of Medical Genetics, Part A, 2012, 158A, 75-77.	0.7	10
117	Pure duplication of 19p13.3. American Journal of Medical Genetics, Part A, 2013, 161, 2300-2304.	0.7	10
118	Refinement of the deletion in 8q22.2â€“q22.3: The minimum deletion size at 8q22.3 related to intellectual disability and epilepsy. American Journal of Medical Genetics, Part A, 2014, 164, 2104-2108.	0.7	10
119	Novel SYNGAP1 variant in a patient with intellectual disability and distinctive dysmorphisms. Congenital Anomalies (discontinued), 2018, 58, 188-190.	0.3	9
120	Nonsyndromic intellectual disability with novel heterozygous SCN2A mutation and epilepsy. Human Genome Variation, 2018, 5, 20.	0.4	9
121	Attitudes of clinical geneticists and certified genetic counselors to genome editing and its clinical applications: A nation-wide questionnaire survey in Japan. Journal of Human Genetics, 2019, 64, 945-954.	1.1	9
122	Two unrelated girls with intellectual disability associated with a truncating mutation in the PPM1D penultimate exon. Brain and Development, 2019, 41, 538-541.	0.6	9
123	A novel intragenic deletion in OPHN1 in a Japanese patient with Dandy-Walker malformation. Human Genome Variation, 2019, 6, 1.	0.4	9
124	Deletion of UBE3A in brothers with Angelman syndrome at the breakpoint with an inversion at 15q11.2. American Journal of Medical Genetics, Part A, 2014, 164, 2873-2878.	0.7	8
125	Genome Analysis in Sick Neonates and Infants: High-yield Phenotypes and Contribution of Small Copy Number Variations. Journal of Pediatrics, 2022, 244, 38-48.e1.	0.9	8
126	Unmasking 15q12 deletion using microarray-based comparative genomic hybridization in a mentally retarded boy with r(Y). American Journal of Medical Genetics Part A, 2004, 130A, 322-324.	2.4	7

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127	Girl with monosomy 1p36 and Angelman syndrome due to unbalanced der(1) transmission of a maternal translocation t(1;15)(p36.3;q13.1). American Journal of Medical Genetics Part A, 2004, 131A, 94-98.	2.4	7
128	Screening for Alagille Syndrome Mutations in the JAG1 and NOTCH2 Genes Using Denaturing High-Performance Liquid Chromatography. Genetic Testing and Molecular Biomarkers, 2007, 11, 216-227.	1.7	7
129	SNP array screening of cryptic genomic imbalances in 450 Japanese subjects with intellectual disability and multiple congenital anomalies previously negative for large rearrangements. Journal of Human Genetics, 2016, 61, 335-343.	1.1	7
130	Biallelic mutations of EGFR in a compound heterozygous state cause ectodermal dysplasia with severe skin defects and gastrointestinal dysfunction. Human Genome Variation, 2018, 5, 11.	0.4	7
131	Familial total anomalous pulmonary venous return with 15q11.2 (BP1-BP2) microdeletion. Journal of Human Genetics, 2018, 63, 1185-1188.	1.1	7
132	Exploring the unique function of imprinting control centers in the PWS/AS-responsible region: finding from array-based methylation analysis in cases with variously sized microdeletions. Clinical Epigenetics, 2019, 11, 36.	1.8	7
133	Novel CUL7 biallelic mutations alter the skeletal phenotype of 3M syndrome. Human Genome Variation, 2020, 7, 1.	0.4	7
134	A severe form of Ellis-van Creveld syndrome caused by novel mutations in EVC2. Human Genome Variation, 2019, 6, 40.	0.4	6
135	Discordant phenotype caused by CASK mutation in siblings with NF1. Human Genome Variation, 2019, 6, 20.	0.4	6
136	Novel USP9X variants in two patients with X-linked intellectual disability. Human Genome Variation, 2019, 6, 49.	0.4	6
137	Diagnostic utility of integrated analysis of exome and transcriptome: Successful diagnosis of Auâ€Kline syndrome in a patient with submucous cleft palate, scaphocephaly, and intellectual disabilities. Molecular Genetics & Genomic Medicine, 2020, 8, e1364.	0.6	6
138	Blended phenotype of AP4E1 deficiency and Angelman syndrome caused by paternal isodisomy of chromosome 15. Brain and Development, 2020, 42, 289-292.	0.6	6
139	Multiple craniosynostosis and facial dysmorphisms with homozygous <scp>IL11RA</scp> variant caused by maternal uniparental isodisomy of chromosome 9. Congenital Anomalies (discontinued), 2020, 60, 153-155.	0.3	6
140	A Recurrent Variant in <i>POLR1B</i>, c.3007C>T; p.Arg1003Cys, Associated with Atresia of the External Canal and Microtia in Treacher Collins Syndrome Type 4. Molecular Syndromology, 2021, 12, 127-132.	0.3	6
141	Early diagnosis of MECP2 duplication syndrome: Insights from a nationwide survey in Japan. Journal of the Neurological Sciences, 2021, 422, 117321.	0.3	6
142	Analysis of Gene-Environment Interactions Related to Developmental Disorders. Frontiers in Pharmacology, 2022, 13, 863664.	1.6	6
143	Premature thelarche in Rubinstein-Taybi syndrome. American Journal of Medical Genetics Part A, 2002, 109, 72-73.	2.4	5
144	Gonadal sex cord stromal tumor in a patient with Rubinstein-Taybi syndrome. Pediatrics International, 2002, 44, 330-332.	0.2	5

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145	Expression analysis of a 17p terminal deletion, including YWHAE, but not PAFAH1B1, associated with normal brain structure on MRI in a young girl. American Journal of Medical Genetics, Part A, 2012, 158A, 2347-2352.	0.7	5
146	Combination of Millerâ€“Dieker syndrome and VACTERL association causes extremely severe clinical presentation. European Journal of Pediatrics, 2014, 173, 1541-1544.	1.3	5
147	MELAS syndrome with m.4450 Gâ€¯>â€¯A mutation in mitochondrial tRNAMet gene. Brain and Development, 2019, 41, 465-469.	0.6	5
148	Wholeâ€exome sequencing reveals the subclonal expression of NUP214â€ABL1 fusion gene in Tâ€cell acute lymphoblastic leukemia. Pediatric Blood and Cancer, 2020, 67, e28019.	0.8	5
149	Epilepsy in Christianson syndrome: Two cases of Lennoxâ€“Gastaut syndrome and a review of literature. Epilepsy and Behavior Reports, 2020, 13, 100349.	0.5	5
150	Prenatal diagnosis of Fraser syndrome caused by novel variants of FREM2. Human Genome Variation, 2020, 7, 32.	0.4	5
151	13q13.3 microdeletion associated with apparently balanced translocation of 46,XX,t(7;13) suggests NBEA involvement. Brain and Development, 2020, 42, 581-586.	0.6	5
152	A recurrent TMEM106B mutation in hypomyelinating leukodystrophy: A rapid diagnostic assay. Brain and Development, 2020, 42, 603-606.	0.6	5
153	Divergent variant patterns among 19 patients with<scp>Rubinsteinâ€Taybi</scp>syndrome uncovered by comprehensive genetic analysis including whole genome sequencing. Clinical Genetics, 2022, 101, 335-345.	1.0	5
154	De novo heterozygous variants in KIF5B cause kyphomelic dysplasia. Clinical Genetics, 2022, 102, 3-11.	1.0	5
155	No sex differences in 18 trisomy births in the Kanagawa Birth Defects Monitoring Program. Congenital Anomalies (discontinued), 2004, 44, 97-98.	0.3	4
156	Prenatal genetic testing for a microdeletion at chromosome 14q32.2 imprinted region leading to UPD(14)patâ€like phenotype. American Journal of Medical Genetics, Part A, 2014, 164, 264-266.	0.7	4
157	A splicing mutation of proteolipid protein 1 in Pelizaeus-Merzbacher disease. Brain and Development, 2016, 38, 581-584.	0.6	4
158	Hearing impairment in a female infant with interstitial deletion of 2q24.1q24.3. Congenital Anomalies (discontinued), 2017, 57, 118-121.	0.3	4
159	Somatic mosaicism of a heterogeneous mutation of <scp>ACTA</scp>1 in nemaline myopathy. Pediatrics International, 2019, 61, 1169-1171.	0.2	4
160	Tumor predisposition in an individual with chromosomal rearrangements of 1q31.2â€q41 encompassing cell division cycle protein 73. Congenital Anomalies (discontinued), 2020, 60, 128-130.	0.3	4
161	POLR1C variants dysregulate splicing and cause hypomyelinating leukodystrophy. Neurology: Genetics, 2020, 6, e524.	0.9	4
162	Unique skeletal manifestations in patients with Primrose syndrome. European Journal of Medical Genetics, 2020, 63, 103967.	0.7	4

#	Article	IF	Citations
163	An efficient genetic test flow for multiple congenital anomalies and intellectual disability. Pediatrics International, 2020, 62, 556-561.	0.2	4
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