Noriko Miyake

List of Publications by Year
in descending order

151 papers	6,159 citations	81839 39 h-index	88593 70 g-index
154 all docs	154 docs citations	154 times ranked	12848 citing authors

#	Article	IF	Citations
1	Mutations affecting components of the SWI/SNF complex cause Coffin-Siris syndrome. Nature Genetics, 2012, 44, 376-378.	9.4	435
2	De novo mutations in the autophagy gene WDR45 cause static encephalopathy of childhood with neurodegeneration in adulthood. Nature Genetics, 2013, 45, 445-449.	9.4	396
3	Human genetic variation database, a reference database of genetic variations in the Japanese population. Journal of Human Genetics, 2016, 61, 547-553.	1.1	270
4	KDM6A Point Mutations Cause Kabuki Syndrome. Human Mutation, 2013, 34, 108-110.	1.1	168
5	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
6	MLL2 and KDM6A mutations in patients with Kabuki syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 2234-2243.	0.7	148
7	Kabuki syndrome: international consensus diagnostic criteria. Journal of Medical Genetics, 2019, 56, 89-95.	1.5	146
8	Loss-of-function mutations of CHST14 in a new type of Ehlers-Danlos syndrome. Human Mutation, 2010, 31, 966-974.	1.1	137
9	Mutations in POLR3A and POLR3B Encoding RNA Polymerase III Subunits Cause an Autosomal-Recessive Hypomyelinating Leukoencephalopathy. American Journal of Human Genetics, 2011, 89, 644-651.	2.6	137
10	STXBP1 mutations in early infantile epileptic encephalopathy with suppressionâ€burst pattern. Epilepsia, 2010, 51, 2397-2405.	2.6	133
11	Coffinâ€“Siris syndrome is a <scp>SWI</scp>/<scp>SNF</scp> complex disorder. Clinical Genetics, 2014, 85, 548-554.	1.0	118
12	De novo SOX11 mutations cause Coffinâ€“Siris syndrome. Nature Communications, 2014, 5, 4011.	5.8	118
13	Whole genome sequencing in patients with retinitis pigmentosa reveals pathogenic DNA structural changes and NEK2 as a new disease gene. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 16139-16144.	3.3	115
14	Mutations in B3GALT6, which Encodes a Glycosaminoglycan Linker Region Enzyme, Cause a Spectrum of Skeletal and Connective Tissue Disorders. American Journal of Human Genetics, 2013, 92, 927-934.	2.6	112
15	Diagnostic utility of whole exome sequencing in patients showing cerebellar and/or vermis atrophy in childhood. Neurogenetics, 2013, 14, 225-232.	0.7	104
16	Impaired neuronal KCC2 function by biallelic SLC12A5 mutations in migrating focal seizures and severe developmental delay. Scientific Reports, 2016, 6, 30072.	1.6	102
17	SMOC1 Is Essential for Ocular and Limb Development in Humans and Mice. American Journal of Human Genetics, 2011, 88, 30-41.	2.6	100
18	GGC Repeat Expansion of NOTCH2NLC in Adult Patients with Leukoencephalopathy. Annals of Neurology, 2019, 86, 962-968.	2.8	98

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19	De novo <scp>KCNT</scp>1 mutations in earlyâ€onset epileptic encephalopathy. Epilepsia, 2015, 56, e121-8.	2.6	95
20	Biallelic Mutations in Nuclear Pore Complex Subunit NUP107 Cause Early-Childhood-Onset Steroid-Resistant Nephrotic Syndrome. American Journal of Human Genetics, 2015, 97, 555-566.	2.6	91
21	Mutations in genes encoding polycomb repressive complex 2 subunits cause Weaver syndrome. Human Mutation, 2017, 38, 637-648.	1.1	80
22	Exome Sequencing Reveals a Homozygous SYT14 Mutation in Adult-Onset, Autosomal-Recessive Spinocerebellar Ataxia with Psychomotor Retardation. American Journal of Human Genetics, 2011, 89, 320-327.	2.6	79
23	Mitochondrial Complex III Deficiency Caused by a HomozygousUQCRC2Mutation Presenting with Neonatal-Onset Recurrent Metabolic Decompensation. Human Mutation, 2013, 34, 446-452.	1.1	79
24	Targeted capture and sequencing for detection of mutations causing early onset epileptic encephalopathy. Epilepsia, 2013, 54, 1262-1269.	2.6	76
25	<scp>GRIN</scp>1 mutations cause encephalopathy with infantileâ€onset epilepsy, and hyperkinetic and stereotyped movement disorders. Epilepsia, 2015, 56, 841-848.	2.6	76
26	Delineation of dermatan 4â€Oâ€sulfotransferase 1 deficient Ehlersâ€“Danlos syndrome: Observation of two additional patients and comprehensive review of 20 reported patients. American Journal of Medical Genetics, Part A, 2011, 155, 1949-1958.	0.7	60
27	De novo KIF1A mutations cause intellectual deficit, cerebellar atrophy, lower limb spasticity and visual disturbance. Journal of Human Genetics, 2015, 60, 739-742.	1.1	58
28	PIGN mutations cause congenital anomalies, developmental delay, hypotonia, epilepsy, and progressive cerebellar atrophy. Neurogenetics, 2014, 15, 85-92.	0.7	57
29	Delineation of clinical features in Wiedemannâ€“Steiner syndrome caused by <scp>KMT2A</scp> mutations. Clinical Genetics, 2016, 89, 115-119.	1.0	56
30	PARS2 and NARS2 mutations in infantile-onset neurodegenerative disorder. Journal of Human Genetics, 2017, 62, 525-529.	1.1	55
31	Detecting copy-number variations in whole-exome sequencing data using the eXome Hidden Markov Model: an â€˜exome-firstâ€™ approach. Journal of Human Genetics, 2015, 60, 175-182.	1.1	54
32	A new Ehlersâ€“Danlos syndrome with craniofacial characteristics, multiple congenital contractures, progressive joint and skin laxity, and multisystem fragilityâ€related manifestations. American Journal of Medical Genetics, Part A, 2010, 152A, 1333-1346.	0.7	53
33	Biallelic TBCD Mutations Cause Early-Onset Neurodegenerative Encephalopathy. American Journal of Human Genetics, 2016, 99, 950-961.	2.6	51
34	De novo WDR45 mutation in a patient showing clinically Rett syndrome with childhood iron deposition in brain. Journal of Human Genetics, 2014, 59, 292-295.	1.1	49
35	Molecular genetic analysis of 30 families with Joubert syndrome. Clinical Genetics, 2016, 90, 526-535.	1.0	45
36	PAPSS2mutations cause autosomal recessive brachyolmia. Journal of Medical Genetics, 2012, 49, 533-538.	1.5	44

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37	ANKRD11 variants cause variable clinical features associated with KBG syndrome and Coffinâ€“Siris-like syndrome. Journal of Human Genetics, 2017, 62, 741-746.	1.1	43
38	Comprehensive genetic analysis of 57 families with clinically suspected Cornelia de Lange syndrome. Journal of Human Genetics, 2019, 64, 967-978.	1.1	43
39	A case of autism spectrum disorder arising from a de novo missense mutation in POGZ. Journal of Human Genetics, 2015, 60, 277-279.	1.1	42
40	A novel mutation in SLC1A3 causes episodic ataxia. Journal of Human Genetics, 2018, 63, 207-211.	1.1	42
41	Compound Heterozygosity for Null Mutations and a Common Hypomorphic Risk Haplotype inTBX6Causes Congenital Scoliosis. Human Mutation, 2017, 38, 317-323.	1.1	41
42	WDR45 mutations in three male patients with West syndrome. Journal of Human Genetics, 2016, 61, 653-661.	1.1	39
43	Haploinsufficiency of A20 caused by a novel nonsense variant or entire deletion of TNFAIP3 is clinically distinct from BehÃ§etâ€™s disease. Arthritis Research and Therapy, 2019, 21, 137.	1.6	39
44	De novo KCNH1 mutations in four patients with syndromic developmental delay, hypotonia and seizures. Journal of Human Genetics, 2016, 61, 381-387.	1.1	38
45	X-linked hypomyelination with spondylometaphyseal dysplasia (H-SMD) associated with mutations in AIFM1. Neurogenetics, 2017, 18, 185-194.	0.7	38
46	Rapid detection of a mutation causing X-linked leucoencephalopathy by exome sequencing. Journal of Medical Genetics, 2011, 48, 606-609.	1.5	36
47	Loss-of-function and gain-of-function mutations in PPP3CA cause two distinct disorders. Human Molecular Genetics, 2018, 27, 1421-1433.	1.4	36
48	Novel SUZ12 mutations in Weaverâ€like syndrome. Clinical Genetics, 2018, 94, 461-466.	1.0	36
49	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	1.1	36
50	Neuropathology of leukoencephalopathy with brainstem and spinal cord involvement and high lactate caused by a homozygous mutation of DARS2. Brain and Development, 2013, 35, 312-316.	0.6	35
51	Detection of copy number variations in epilepsy using exome data. Clinical Genetics, 2018, 93, 577-587.	1.0	35
52	The Liberfarb syndrome, a multisystem disorder affecting eye, ear, bone, and brain development, is caused by a founder pathogenic variant in the PISD gene. Genetics in Medicine, 2019, 21, 2734-2743.	1.1	33
53	A novel homozygous mutation of DARS2 may cause a severe LBSL variant. Clinical Genetics, 2011, 80, 293-296.	1.0	31
54	Milder progressive cerebellar atrophy caused by biallelic SEPSECS mutations. Journal of Human Genetics, 2016, 61, 527-531.	1.1	30

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55	Genetic landscape of Rett syndrome-like phenotypes revealed by whole exome sequencing. Journal of Medical Genetics, 2019, 56, 396-407.	1.5	30
56	Clinical delineation, sex differences, and genotypeâ€“phenotype correlation in pathogenic KDM6A variants causing X-linked Kabuki syndrome type 2. Genetics in Medicine, 2021, 23, 1202-1210.	1.1	30
57	Numerous BAF complex genes are mutated in Coffinâ€“Siris syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 257-261.	0.7	29
58	Late-onset spastic ataxia phenotype in a patient with a homozygous DDHD2 mutation. Scientific Reports, 2015, 4, 7132.	1.6	29
59	The diagnostic utility of exome sequencing in Joubert syndrome and related disorders. Journal of Human Genetics, 2013, 58, 113-115.	1.1	28
60	Identification of novel <scp>SNORD118</scp> mutations in seven patients with leukoencephalopathy with brain calcifications and cysts. Clinical Genetics, 2017, 92, 180-187.	1.0	28
61	Novel KCNB1 mutation associated with non-syndromic intellectual disability. Journal of Human Genetics, 2017, 62, 569-573.	1.1	28
62	Equivalent missense variant in the FOXP2 and FOXP1 transcription factors causes distinct neurodevelopmental disorders. Human Mutation, 2017, 38, 1542-1554.	1.1	28
63	A novel SLC9A1 mutation causes cerebellar ataxia. Journal of Human Genetics, 2018, 63, 1049-1054.	1.1	28
64	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.	1.1	28
65	Heterozygous Mutations in OAS1 Cause Infantile-Onset Pulmonary Alveolar Proteinosis with Hypogammaglobulinemia. American Journal of Human Genetics, 2018, 102, 480-486.	2.6	26
66	Structural alteration of glycosaminoglycan side chains and spatial disorganization of collagen networks in the skin of patients with mcEDS-CHST14. Biochimica Et Biophysica Acta - General Subjects, 2019, 1863, 623-631.	1.1	26
67	Genome sequencing in persistently unsolved white matter disorders. Annals of Clinical and Translational Neurology, 2020, 7, 144-152.	1.7	26
68	Whole exome sequencing revealed biallelic <scp>IFT122</scp> mutations in a family with <scp>CED1</scp> and recurrent pregnancy loss. Clinical Genetics, 2014, 85, 592-594.	1.0	25
69	A hemizygous GYG2 mutation and Leigh syndrome: a possible link?. Human Genetics, 2014, 133, 225-234.	1.8	25
70	Identification andIn VivoFunctional Characterization of Novel Compound HeterozygousBMP1Variants in Osteogenesis Imperfecta. Human Mutation, 2015, 36, 191-195.	1.1	25
71	Defect in dermatan sulfate in urine of patients with Ehlers-Danlos syndrome caused by a CHST14/D4ST1 deficiency. Clinical Biochemistry, 2017, 50, 670-677.	0.8	25
72	Novel biallelic <scp>SZT2</scp> mutations in 3 cases of earlyâ€onset epileptic encephalopathy. Clinical Genetics, 2018, 93, 266-274.	1.0	25

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73	De novo variants in RHOBTB2 , an atypical Rho GTPase gene, cause epileptic encephalopathy. Human Mutation, 2018, 39, 1070-1075.	1.1	25
74	Gain-of-Function MN1 Truncation Variants Cause a Recognizable Syndrome with Craniofacial and Brain Abnormalities. American Journal of Human Genetics, 2020, 106, 13-25.	2.6	25
75	Pathogenic variants of DYNC2H1 , KIAA0556 , and PTPN11 associated with hypothalamic hamartoma. Neurology, 2019, 93, e237-e251.	1.5	24
76	Recent Advances in the Pathophysiology of Musculocontractural Ehlers-Danlos Syndrome. Genes, 2020, 11, 43.	1.0	24
77	Biotin-responsive basal ganglia disease: a case diagnosed by whole exome sequencing. Journal of Human Genetics, 2015, 60, 381-385.	1.1	22
78	Genetic analysis of adult leukoencephalopathy patients using a customâ€designed gene panel. Clinical Genetics, 2018, 94, 232-238.	1.0	22
79	A response to: Loss of dermatan-4-sulfotransferase 1 (D4ST1/CHST14) function represents the first dermatan sulfate biosynthesis defect, â€œdermatan sulfate-deficient Adducted Thumb-Clubfoot Syndromeâ€• Which name is appropriate, â€œAdducted Thumb-Clubfoot Synd. Human Mutation, 2011, 32, 1507-1509.	1.1	21
80	A novel missense mutation affecting the same amino acid as the recurrent <scp>PACS1</scp> mutation in Schuursâ€Hoeijmakers syndrome. Clinical Genetics, 2018, 93, 929-930.	1.0	21
81	A novel CYCS mutation in the Î±â€helix of the CYCS Câ€terminal domain causes nonâ€syndromic thrombocytopenia. Clinical Genetics, 2018, 94, 548-553.	1.0	20
82	Clinical and molecular spectrum of CHOPS syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 1126-1138.	0.7	20
83	Novel recessive mutations in MSTO1 cause cerebellar atrophy with pigmentary retinopathy. Journal of Human Genetics, 2018, 63, 263-270.	1.1	19
84	Characteristics of epilepsy in patients with Kabuki syndrome with KMT2D mutations. Brain and Development, 2017, 39, 672-677.	0.6	18
85	Delineation of musculocontractural Ehlersâ€“Danlos Syndrome caused by dermatan sulfate epimerase deficiency. Molecular Genetics & Genomic Medicine, 2020, 8, e1197.	0.6	18
86	Efficient detection of copyâ€number variations using exome data: Batchâ€and sexâ€based analyses. Human Mutation, 2021, 42, 50-65.	1.1	18
87	Clinical and molecular features of 66 patients with musculocontractural Ehlersâ€Danlos syndrome caused by pathogenic variants in CHST14 (mcEDS-CHST14). Journal of Medical Genetics, 2021, , jmedgenet-2020-107623.	1.5	18
88	GRIN2D variants in three cases of developmental and epileptic encephalopathy. Clinical Genetics, 2018, 94, 538-547.	1.0	17
89	Exome sequencing identifies a novel INPPL1 mutation in opsismodysplasia. Journal of Human Genetics, 2013, 58, 391-394.	1.1	16
90	Detection of low-prevalence somatic TSC2 mutations in sporadic pulmonary lymphangioleiomyomatosis tissues by deep sequencing. Human Genetics, 2016, 135, 61-68.	1.8	16

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91	De novo variants in CELF2 that disrupt the nuclear localization signal cause developmental and epileptic encephalopathy. Human Mutation, 2021, 42, 66-76.	1.1	16
92	Severe manifestations of handâ€footâ€genital syndrome associated with a novel HOXA13 mutation. American Journal of Medical Genetics, Part A, 2014, 164, 2398-2402.	0.7	15
93	Clinical features of SMARCA2 duplication overlap with Coffinâ€“Siris syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2662-2670.	0.7	15
94	Dermatan 4â€Oâ€sulfotransferase 1â€deficient Ehlersâ€“Danlos syndrome complicated by a large subcutaneous hematoma on the back. Journal of Dermatology, 2016, 43, 832-833.	0.6	15
95	Three patients with Schaafâ€“Yang syndrome exhibiting arthrogryposis and endocrinological abnormalities. American Journal of Medical Genetics, Part A, 2018, 176, 707-711.	0.7	15
96	Novel EXOSC9 variants cause pontocerebellar hypoplasia type 1D with spinal motor neuronopathy and cerebellar atrophy. Journal of Human Genetics, 2021, 66, 401-407.	1.1	15
97	Ehlersâ€“Danlos Syndrome Associated with Glycosaminoglycan Abnormalities. Advances in Experimental Medicine and Biology, 2014, 802, 145-159.	0.8	14
98	A Japanese case of cerebellar ataxia, spastic paraparesis and deep sensory impairment associated with a novel homozygous TTC19 mutation. Journal of Human Genetics, 2015, 60, 187-191.	1.1	14
99	A homozygous NOP14 variant is likely to cause recurrent pregnancy loss. Journal of Human Genetics, 2018, 63, 425-430.	1.1	14
100	A novel homozygous DPH1 mutation causes intellectual disability and unique craniofacial features. Journal of Human Genetics, 2018, 63, 487-491.	1.1	14
101	Hypothalamic pituitary complications in Kabuki syndrome. Pituitary, 2013, 16, 133-138.	1.6	13
102	A unique case of de novo 5q33.3â€“q34 triplication with uniparental isodisomy of 5q34â€“qter. American Journal of Medical Genetics, Part A, 2013, 161, 1904-1909.	0.7	13
103	Cancer Management in Kabuki Syndrome: The First Case of Wilms Tumor and a Literature Review. Journal of Pediatric Hematology/Oncology, 2018, 40, 391-394.	0.3	13
104	Bi-allelic loss of function variants ofTBX6causes a spectrum of malformation of spine and rib including congenital scoliosis and spondylocostal dysostosis. Journal of Medical Genetics, 2019, 56, 622-628.	1.5	13
105	A novel ITPA variant causes epileptic encephalopathy with multiple-organ dysfunction. Journal of Human Genetics, 2020, 65, 751-757.	1.1	13
106	Large-scale discovery of novel neurodevelopmental disorder-related genes through a unified analysis of single-nucleotide and copy number variants. Genome Medicine, 2022, 14, 40.	3.6	13
107	Two novel homozygous RAB3GAP1 mutations cause Warburg micro syndrome. Human Genome Variation, 2015, 2, 15034.	0.4	12
108	Dual genetic diagnoses: Atypical handâ€footâ€genital syndrome and developmental delay due to de novo mutations in HOXA13 and NRXN1. American Journal of Medical Genetics, Part A, 2016, 170, 717-724.	0.7	11

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109	PRUNE1â€related disorder: Expanding the clinical spectrum. Clinical Genetics, 2018, 94, 362-367.	1.0	11
110	Hemorrhagic stroke and renovascular hypertension with Grange syndrome arising from a novel pathogenic variant in YY1AP1. Journal of Human Genetics, 2019, 64, 885-890.	1.1	11
111	Pathogenic 12-kb copy-neutral inversion in syndromic intellectual disability identified by high-fidelity long-read sequencing. Genomics, 2021, 113, 1044-1053.	1.3	11
112	Different X-linkedKDM5Cmutations in affected male siblings: is maternal reversion error involved?. Clinical Genetics, 2016, 90, 276-281.	1.0	10
113	A case of atypical Kabuki syndrome arising from a novel missense variant in <scp>HNRNPK</scp>. Clinical Genetics, 2017, 92, 554-555.	1.0	10
114	The second point mutation in PREPL: a case report and literature review. Journal of Human Genetics, 2018, 63, 677-681.	1.1	10
115	Novel compound heterozygous DPH1 mutations in a patient with the unique clinical features of airway obstruction and external genital abnormalities. Journal of Human Genetics, 2018, 63, 529-532.	1.1	10
116	SOFT syndrome in a patient from Chile. American Journal of Medical Genetics, Part A, 2019, 179, 338-340.	0.7	10
117	Exome sequencing of two patients in a family with atypical Xâ€linked leukodystrophy. Clinical Genetics, 2011, 80, 161-166.	1.0	9
118	An atypical case of SPG56/CYP2U1-related spastic paraplegia presenting with delayed myelination. Journal of Human Genetics, 2017, 62, 997-1000.	1.1	9
119	Confirmation of SLC5A7â€related distal hereditary motor neuropathy 7 in a family outside Wales. Clinical Genetics, 2018, 94, 274-275.	1.0	9
120	A 45-year-old Woman with Ehlers-Danlos Syndrome Caused by Dermatan 4-O-sulfotransferase-1 Deficiency: Implications for Early Ageing. Acta Dermato-Venereologica, 2014, 96, 830-1.	0.6	8
121	â€˜Cortical cerebellar atrophyâ€™ dwindles away in the era of next-generation sequencing. Journal of Human Genetics, 2014, 59, 589-590.	1.1	8
122	De novo CACNA1G variants in developmental delay and early-onset epileptic encephalopathies. Journal of the Neurological Sciences, 2020, 416, 117047.	0.3	8
123	Polymicrogyria in a child with KCNMA1-related channelopathy. Brain and Development, 2022, 44, 173-177.	0.6	7
124	A novel DARS2 mutation in a Japanese patient with leukoencephalopathy with brainstem and spinal cord involvement but no lactate elevation. Human Genome Variation, 2017, 4, 17051.	0.4	6
125	Independent occurrence of de novo HSPD1 and HIP1 variants in brothers with different neurological disorders â€“ leukodystrophy and autism. Human Genome Variation, 2018, 5, 18.	0.4	6
126	Bilateral cerebellar cysts and cerebral white matter lesions with cortical dysgenesis: Expanding the phenotype of LAMB1 gene mutations. Clinical Genetics, 2018, 94, 391-392.	1.0	6

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127	The recurrent postzygotic pathogenic variant p.Glu47Lys in RHOA causes a novel recognizable neuroectodermal phenotype. Human Mutation, 2020, 41, 591-599.	1.1	6
128	A case of cerebral hypomyelination with spondyloâ€epiâ€metaphyseal dysplasia. American Journal of Medical Genetics, Part A, 2013, 161, 203-207.	0.7	5
129	Phenotypic and molecular insights into PQBP1â€related intellectual disability. American Journal of Medical Genetics, Part A, 2018, 176, 2446-2450.	0.7	5
130	<scp>COG1â€</scp>congenital disorders of glycosylation: Milder presentation and review. Clinical Genetics, 2021, 100, 318-323.	1.0	5
131	De novo heterozygous variants in KIF5B cause kyphomelic dysplasia. Clinical Genetics, 2022, 102, 3-11.	1.0	5
132	OTUD5 Variants Associated With X-Linked Intellectual Disability and Congenital Malformation. Frontiers in Cell and Developmental Biology, 2021, 9, 631428.	1.8	4
133	Cerebellofaciodental syndrome in an adult patient: Expanding the phenotypic and natural history characteristics. American Journal of Medical Genetics, Part A, 2021, 185, 1561-1568.	0.7	4
134	Recurrent de novo missense variants in GNB2 can cause syndromic intellectual disability. Journal of Medical Genetics, 2022, 59, 511-516.	1.5	4
135	De novo missense variants in LMBRD2 are associated with developmental and motor delays, brain structure abnormalities and dysmorphic features. Journal of Medical Genetics, 2020, 58, jmedgenet-2020-107137.	1.5	3
136	Hemizygous FLNA variant in West syndrome without periventricular nodular heterotopia. Human Genome Variation, 2020, 7, 43.	0.4	3
137	Expanding the phenotypic spectrum of cardiospondylocarpofacial syndrome: From a detailed clinical and radiological observation of a boy with a novel missense variant in <scp>MAP3K7</scp>. American Journal of Medical Genetics, Part A, 2022, 188, 350-356.	0.7	3
138	Cockayne syndrome without UV-sensitivity in Vietnamese siblings with novel ERCC8 variants. Aging, 0, , .	1.4	3
139	<scp>J</scp>apanese familial case of myoclonusâ€“dystonia syndrome with a splicing mutation in <scp>SGCE</scp>. Pediatrics International, 2015, 57, 324-326.	0.2	2
140	Response to Lefebvre et al. Clinical Genetics, 2017, 92, 563-564.	1.0	2
141	New SMARCE1 variant in a patient with features overlapping with oculoauriculofrontonasal syndrome. Clinical Genetics, 2018, 94, 487-488.	1.0	2
142	Reply to â€œ<scp>GGC</scp> Repeat Expansion of <scp>NOTCH2NLC</scp> is Rare in European Leukoencephalopathyâ€. Annals of Neurology, 2020, 88, 642-643.	2.8	2
143	A patient with a 6q22.1 deletion and a phenotype of non-progressive early-onset generalized epilepsy with tremor. Epilepsy and Behavior Reports, 2021, 15, 100405.	0.5	2
144	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.	1.0	2

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145	A homozygous <scp>ABHD16A</scp> variant causes a complex hereditary spastic paraplegia with developmental delay, absent speech, and characteristic face. Clinical Genetics, 2022, 101, 359-363.	1.0	2
146	Monogenic causes of pigmentary mosaicism. Human Genetics, 2022, , .	1.8	2
147	Leggâ€CalvÃ©â€Perthes disease in a patient with Bardetâ€Biedl syndrome: A case report of a novelMKKS/BBS6mutation. Clinical Case Reports (discontinued), 2020, 8, 3110-3115.	0.2	1
148	De novo pathogenic <scp>DHX30</scp> variants in two cases. Clinical Genetics, 2021, 100, 350-351.	1.0	1
149	Ehlers Danlos Syndrome with Glycosaminoglycan Abnormalities. Advances in Experimental Medicine and Biology, 2021, 1348, 235-249.	0.8	1
150	Cono-spondylar dysplasia: Clinical, radiographic, and molecular findings of a previously unreported disorder. , 2014, 164, 2147-2152.		0
151	Autosomal dominant Alport syndrome due to a COL4A4 mutation with an additional ESPN variant detected by whole-exome analysis. CEN Case Reports, 2020, 9, 59-64.	0.5	0