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	Recurrent de novo missense variants in GNB2 can cause syndromic intellectual disability. Journal of Medical Genetics, 2022, 59, 511-516.	1.5	4
2	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
3	Polymicrogyria in a child with KCNMA1-related channelopathy. Brain and Development, 2022, 44, 173-177.	0.6	7
4	De novo heterozygous variants in KIF5B cause kyphomelic dysplasia. Clinical Genetics, 2022, 102, 3-11.	1.0	5
5	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
6	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
7	Monogenic causes of pigmentary mosaicism. Human Genetics, 2022, , .	1.8	2
8	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
9	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
10	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
11	Efficient detection of copyâ€number variations using exome data: Batchâ€and sexâ€based analyses. Human Mutation, 2021, 42, 50-65.	1.1	18
12	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
13	OTUD5 Variants Associated With X-Linked Intellectual Disability and Congenital Malformation. Frontiers in Cell and Developmental Biology, 2021, 9, 631428.	1.8	4
14	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
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.	0.7	4
16	<scp>COG1â€</scp>congenital disorders of glycosylation: Milder presentation and review. Clinical Genetics, 2021, 100, 318-323.	1.0	5
17	De novo pathogenic <scp>DHX30</scp> variants in two cases. Clinical Genetics, 2021, 100, 350-351.	1.0	1
18	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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19	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
20	Ehlers Danlos Syndrome with Glycosaminoglycan Abnormalities. Advances in Experimental Medicine and Biology, 2021, 1348, 235-249.	0.8	1
21	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
22	Recent Advances in the Pathophysiology of Musculocontractural Ehlers-Danlos Syndrome. Genes, 2020, 11, 43.	1.0	24
23	Genome sequencing in persistently unsolved white matter disorders. Annals of Clinical and Translational Neurology, 2020, 7, 144-152.	1.7	26
24	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
25	The recurrent postzygotic pathogenic variant p.Glu47Lys in RHOA causes a novel recognizable neuroectodermal phenotype. Human Mutation, 2020, 41, 591-599.	1.1	6
26	De novo CACNA1G variants in developmental delay and early-onset epileptic encephalopathies. Journal of the Neurological Sciences, 2020, 416, 117047.	0.3	8
27	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
28	Hemizygous FLNA variant in West syndrome without periventricular nodular heterotopia. Human Genome Variation, 2020, 7, 43.	0.4	3
29	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
30	A novel ITPA variant causes epileptic encephalopathy with multiple-organ dysfunction. Journal of Human Genetics, 2020, 65, 751-757.	1.1	13
31	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
32	Delineation of musculocontractural Ehlersâ€“Danlos Syndrome caused by dermatan sulfate epimerase deficiency. Molecular Genetics & Genomic Medicine, 2020, 8, e1197.	0.6	18
33	GGC Repeat Expansion of NOTCH2NLC in Adult Patients with Leukoencephalopathy. Annals of Neurology, 2019, 86, 962-968.	2.8	98
34	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
35	Comprehensive genetic analysis of 57 families with clinically suspected Cornelia de Lange syndrome. Journal of Human Genetics, 2019, 64, 967-978.	1.1	43
36	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

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37	Pathogenic variants of DYNC2H1 , KIAA0556 , and PTPN11 associated with hypothalamic hamartoma. Neurology, 2019, 93, e237-e251.	1.5	24
38	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	1.1	36
39	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
40	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
41	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
42	Clinical and molecular spectrum of CHOPS syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 1126-1138.	0.7	20
43	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
44	Genetic landscape of Rett syndrome-like phenotypes revealed by whole exome sequencing. Journal of Medical Genetics, 2019, 56, 396-407.	1.5	30
45	SOFT syndrome in a patient from Chile. American Journal of Medical Genetics, Part A, 2019, 179, 338-340.	0.7	10
46	Kabuki syndrome: international consensus diagnostic criteria. Journal of Medical Genetics, 2019, 56, 89-95.	1.5	146
47	The second point mutation in PREPL: a case report and literature review. Journal of Human Genetics, 2018, 63, 677-681.	1.1	10
48	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
49	Heterozygous Mutations in OAS1 Cause Infantile-Onset Pulmonary Alveolar Proteinosis with Hypogammaglobulinemia. American Journal of Human Genetics, 2018, 102, 480-486.	2.6	26
50	A homozygous NOP14 variant is likely to cause recurrent pregnancy loss. Journal of Human Genetics, 2018, 63, 425-430.	1.1	14
51	Loss-of-function and gain-of-function mutations in PPP3CA cause two distinct disorders. Human Molecular Genetics, 2018, 27, 1421-1433.	1.4	36
52	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
53	Novel recessive mutations in MSTO1 cause cerebellar atrophy with pigmentary retinopathy. Journal of Human Genetics, 2018, 63, 263-270.	1.1	19
54	A novel homozygous DPH1 mutation causes intellectual disability and unique craniofacial features. Journal of Human Genetics, 2018, 63, 487-491.	1.1	14

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55	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
56	Genetic analysis of adult leukoencephalopathy patients using a customâ€designed gene panel. Clinical Genetics, 2018, 94, 232-238.	1.0	22
57	Novel biallelic <scp>SZT2</scp> mutations in 3 cases of earlyâ€onset epileptic encephalopathy. Clinical Genetics, 2018, 93, 266-274.	1.0	25
58	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
59	Detection of copy number variations in epilepsy using exome data. Clinical Genetics, 2018, 93, 577-587.	1.0	35
60	A novel mutation in SLC1A3 causes episodic ataxia. Journal of Human Genetics, 2018, 63, 207-211.	1.1	42
61	Phenotypic and molecular insights into PQBP1â€related intellectual disability. American Journal of Medical Genetics, Part A, 2018, 176, 2446-2450.	0.7	5
62	GRIN2D variants in three cases of developmental and epileptic encephalopathy. Clinical Genetics, 2018, 94, 538-547.	1.0	17
63	New SMARCE1 variant in a patient with features overlapping with oculoauriculofrontonasal syndrome. Clinical Genetics, 2018, 94, 487-488.	1.0	2
64	De novo variants in RHOBTB2 , an atypical Rho GTPase gene, cause epileptic encephalopathy. Human Mutation, 2018, 39, 1070-1075.	1.1	25
65	Confirmation of SLC5A7â€related distal hereditary motor neuropathy 7 in a family outside Wales. Clinical Genetics, 2018, 94, 274-275.	1.0	9
66	PRUNE1â€related disorder: Expanding the clinical spectrum. Clinical Genetics, 2018, 94, 362-367.	1.0	11
67	A novel SLC9A1 mutation causes cerebellar ataxia. Journal of Human Genetics, 2018, 63, 1049-1054.	1.1	28
68	Novel SUZ12 mutations in Weaverâ€like syndrome. Clinical Genetics, 2018, 94, 461-466.	1.0	36
69	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
70	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
71	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
72	PARS2 and NARS2 mutations in infantile-onset neurodegenerative disorder. Journal of Human Genetics, 2017, 62, 525-529.	1.1	55

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73	Mutations in genes encoding polycomb repressive complex 2 subunits cause Weaver syndrome. Human Mutation, 2017, 38, 637-648.	1.1	80
74	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
75	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
76	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
77	Characteristics of epilepsy in patients with Kabuki syndrome with KMT2D mutations. Brain and Development, 2017, 39, 672-677.	0.6	18
78	Novel KCNB1 mutation associated with non-syndromic intellectual disability. Journal of Human Genetics, 2017, 62, 569-573.	1.1	28
79	Compound Heterozygosity for Null Mutations and a Common Hypomorphic Risk Haplotype inTBX6Causes Congenital Scoliosis. Human Mutation, 2017, 38, 317-323.	1.1	41
80	Response to Lefebvre et al. Clinical Genetics, 2017, 92, 563-564.	1.0	2
81	X-linked hypomyelination with spondylometaphyseal dysplasia (H-SMD) associated with mutations in AIFM1. Neurogenetics, 2017, 18, 185-194.	0.7	38
82	An atypical case of SPG56/CYP2U1-related spastic paraplegia presenting with delayed myelination. Journal of Human Genetics, 2017, 62, 997-1000.	1.1	9
83	Equivalent missense variant in the FOXP2 and FOXP1 transcription factors causes distinct neurodevelopmental disorders. Human Mutation, 2017, 38, 1542-1554.	1.1	28
84	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
85	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
86	Clinical features of SMARCA2 duplication overlap with Coffinâ€“Siris syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2662-2670.	0.7	15
87	Different X-linkedKDM5Cmutations in affected male siblings: is maternal reversion error involved?. Clinical Genetics, 2016, 90, 276-281.	1.0	10
88	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
89	Impaired neuronal KCC2 function by biallelic SLC12A5 mutations in migrating focal seizures and severe developmental delay. Scientific Reports, 2016, 6, 30072.	1.6	102
90	WDR45 mutations in three male patients with West syndrome. Journal of Human Genetics, 2016, 61, 653-661.	1.1	39

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91	Biallelic TBCD Mutations Cause Early-Onset Neurodegenerative Encephalopathy. American Journal of Human Genetics, 2016, 99, 950-961.	2.6	51
92	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
93	Molecular genetic analysis of 30 families with Joubert syndrome. Clinical Genetics, 2016, 90, 526-535.	1.0	45
94	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
95	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
96	Milder progressive cerebellar atrophy caused by biallelic SEPSECS mutations. Journal of Human Genetics, 2016, 61, 527-531.	1.1	30
97	Detection of low-prevalence somatic TSC2 mutations in sporadic pulmonary lymphangioleiomyomatosis tissues by deep sequencing. Human Genetics, 2016, 135, 61-68.	1.8	16
98	Delineation of clinical features in Wiedemannâ€“Steiner syndrome caused by <scp>KMT2A</scp> mutations. Clinical Genetics, 2016, 89, 115-119.	1.0	56
99	<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
100	Late-onset spastic ataxia phenotype in a patient with a homozygous DDHD2 mutation. Scientific Reports, 2015, 4, 7132.	1.6	29
101	Two novel homozygous RAB3GAP1 mutations cause Warburg micro syndrome. Human Genome Variation, 2015, 2, 15034.	0.4	12
102	De novo <scp>KCNT</scp>1 mutations in earlyâ€onset epileptic encephalopathy. Epilepsia, 2015, 56, e121-8.	2.6	95
103	<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
104	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
105	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
106	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
107	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
108	Biotin-responsive basal ganglia disease: a case diagnosed by whole exome sequencing. Journal of Human Genetics, 2015, 60, 381-385.	1.1	22

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109	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
110	Identification andIn VivoFunctional Characterization of Novel Compound HeterozygousBMP1Variants in Osteogenesis Imperfecta. Human Mutation, 2015, 36, 191-195.	1.1	25
111	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
112	â€˜Cortical cerebellar atrophyâ€™ dwindles away in the era of next-generation sequencing. Journal of Human Genetics, 2014, 59, 589-590.	1.1	8
113	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
114	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
115	Cono-spondylar dysplasia: Clinical, radiographic, and molecular findings of a previously unreported disorder. , 2014, 164, 2147-2152.		0
116	Ehlersâ€“Danlos Syndrome Associated with Glycosaminoglycan Abnormalities. Advances in Experimental Medicine and Biology, 2014, 802, 145-159.	0.8	14
117	A hemizygous GYG2 mutation and Leigh syndrome: a possible link?. Human Genetics, 2014, 133, 225-234.	1.8	25
118	PIGN mutations cause congenital anomalies, developmental delay, hypotonia, epilepsy, and progressive cerebellar atrophy. Neurogenetics, 2014, 15, 85-92.	0.7	57
119	Coffinâ€“Siris syndrome is a <scp>SWI</scp>/<scp>SNF</scp> complex disorder. Clinical Genetics, 2014, 85, 548-554.	1.0	118
120	De novo SOX11 mutations cause Coffinâ€“Siris syndrome. Nature Communications, 2014, 5, 4011.	5.8	118
121	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
122	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
123	Hypothalamic pituitary complications in Kabuki syndrome. Pituitary, 2013, 16, 133-138.	1.6	13
124	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
125	Diagnostic utility of whole exome sequencing in patients showing cerebellar and/or vermis atrophy in childhood. Neurogenetics, 2013, 14, 225-232.	0.7	104
126	Targeted capture and sequencing for detection of mutations causing early onset epileptic encephalopathy. Epilepsia, 2013, 54, 1262-1269.	2.6	76

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127	A case of cerebral hypomyelination with spondyloâ€epiâ€metaphyseal dysplasia. American Journal of Medical Genetics, Part A, 2013, 161, 203-207.	0.7	5
128	KDM6A Point Mutations Cause Kabuki Syndrome. Human Mutation, 2013, 34, 108-110.	1.1	168
129	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
130	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
131	Mitochondrial Complex III Deficiency Caused by a HomozygousUQCRC2Mutation Presenting with Neonatal-Onset Recurrent Metabolic Decompensation. Human Mutation, 2013, 34, 446-452.	1.1	79
132	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
133	The diagnostic utility of exome sequencing in Joubert syndrome and related disorders. Journal of Human Genetics, 2013, 58, 113-115.	1.1	28
134	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
135	MLL2 and KDM6A mutations in patients with Kabuki syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 2234-2243.	0.7	148
136	Exome sequencing identifies a novel INPPL1 mutation in opsismodysplasia. Journal of Human Genetics, 2013, 58, 391-394.	1.1	16
137	PAPSS2mutations cause autosomal recessive brachyolmia. Journal of Medical Genetics, 2012, 49, 533-538.	1.5	44
138	Mutations affecting components of the SWI/SNF complex cause Coffin-Siris syndrome. Nature Genetics, 2012, 44, 376-378.	9.4	435
139	A novel homozygous mutation of DARS2 may cause a severe LBSL variant. Clinical Genetics, 2011, 80, 293-296.	1.0	31
140	Exome sequencing of two patients in a family with atypical Xâ€linked leukodystrophy. Clinical Genetics, 2011, 80, 161-166.	1.0	9
141	SMOC1 Is Essential for Ocular and Limb Development in Humans and Mice. American Journal of Human Genetics, 2011, 88, 30-41.	2.6	100
142	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
143	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
144	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

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145	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
146	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
147	Rapid detection of a mutation causing X-linked leucoencephalopathy by exome sequencing. Journal of Medical Genetics, 2011, 48, 606-609.	1.5	36
148	Loss-of-function mutations of CHST14 in a new type of Ehlers-Danlos syndrome. Human Mutation, 2010, 31, 966-974.	1.1	137
149	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
150	STXBP1 mutations in early infantile epileptic encephalopathy with suppressionâ€burst pattern. Epilepsia, 2010, 51, 2397-2405.	2.6	133
151	Cockayne syndrome without UV-sensitivity in Vietnamese siblings with novel ERCC8 variants. Aging, 0, , .	1.4	3