# Noriko Miyake

List of Publications by Year
in descending order



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|---|---|---|---|
| 151 papers | 6,159 citations | 81743 39 h-index | 88477 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 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 |