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List of Publications by Year
in descending order

190 papers	11,522 citations	27035 58 h-index	42259 96 g-index
196 all docs	196 docs citations	196 times ranked	17937 citing authors

#	Article	IF	Citations
1	Biallelic <scp>TMEM260</scp> variants cause truncus arteriosus, with or without renal defects. Clinical Genetics, 2022, 101, 127-133.	1.0	10
2	Clinical presentation and analysis of genotype-phenotype correlations in patients with malignant infantile osteopetrosis. Bone, 2022, 154, 116229.	1.4	9
3	Bi-allelic variants in neuronal cell adhesion molecule cause a neurodevelopmental disorder characterized by developmental delay, hypotonia, neuropathy/spasticity. American Journal of Human Genetics, 2022, 109, 518-532.	2.6	8
4	A Zebrafish Model for a Rare Genetic Disease Reveals a Conserved Role for FBXL3 in the Circadian Clock System. International Journal of Molecular Sciences, 2022, 23, 2373.	1.8	3
5	Orbital nodular fasciitis in child with biallelic germline RBL2 variant. European Journal of Medical Genetics, 2022, 65, 104513.	0.7	0
6	Parental exome analysis identifies shared carrier status for a second recessive disorder in couples with an affected child. European Journal of Human Genetics, 2021, 29, 455-462.	1.4	8
7	Bacillus Calmetteâ€“Guerin (BCG) Vaccine-associated Complications in Immunodeficient Patients Following Stem Cell Transplantation. Journal of Clinical Immunology, 2021, 41, 147-162.	2.0	11
8	A novel de novo heterozygous pathogenic variant in the SDHA gene results in childhood onset bilateral optic atrophy and cognitive impairment. Metabolic Brain Disease, 2021, 36, 581-588.	1.4	4
9	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	2.6	30
10	A novel homozygous MSTO1 mutation in Ashkenazi Jewish siblings with ataxia and myopathy. Journal of Human Genetics, 2021, 66, 835-840.	1.1	3
11	Homozygous variant in MADD, encoding a Rab guanine nucleotide exchange factor, results in pleiotropic effects and a multisystemic disorder. European Journal of Human Genetics, 2021, 29, 977-987.	1.4	6
12	Biallelic loss-of-function variants in PLD1 cause congenital right-sided cardiac valve defects and neonatal cardiomyopathy. Journal of Clinical Investigation, 2021, 131, .	3.9	16
13	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	3.6	50
14	Characterization of a L136P mutation in Formin-like 2 (FMNL2) from a patient with chronic inflammatory bowel disease. PLoS ONE, 2021, 16, e0252428.	1.1	5
15	Biallelic deletion in a minimal <scp>CAPN15</scp> intron in siblings with a recognizable syndrome of congenital malformations and developmental delay. Clinical Genetics, 2021, 99, 577-582.	1.0	9
16	A human case of GIMAP6 deficiency: a novel primary immune deficiency. European Journal of Human Genetics, 2021, 29, 657-662.	1.4	10
17	PNC2 (SLC25A36) Deficiency Associated With the Hyperinsulinism/Hyperammonemia Syndrome. Journal of Clinical Endocrinology and Metabolism, 2021, , .	1.8	5
18	Loss of ADAMTS19 causes progressive non-syndromic heart valve disease. Nature Genetics, 2020, 52, 40-47.	9.4	46

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19	A mutation in POLR3E impairs antiviral immune response and RNA polymerase III. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 22113-22121.	3.3	17
20	CFAP45 deficiency causes situs abnormalities and asthenospermia by disrupting an axonemal adenine nucleotide homeostasis module. Nature Communications, 2020, 11, 5520.	5.8	36
21	De novo mutations in the X-linked TFE3 gene cause intellectual disability with pigmentary mosaicism and storage disorder-like features. Journal of Medical Genetics, 2020, 57, 808-819.	1.5	11
22	Cell-based analysis of CAD variants identifies individuals likely to benefit from uridine therapy. Genetics in Medicine, 2020, 22, 1598-1605.	1.1	18
23	Grandparental genotyping enhances exome variant interpretation. American Journal of Medical Genetics, Part A, 2020, 182, 689-696.	0.7	4
24	Loss of function mutations in CCDC32 cause a congenital syndrome characterized by craniofacial, cardiac and neurodevelopmental anomalies. Human Molecular Genetics, 2020, 29, 1489-1497.	1.4	6
25	Homozygous frameshift variant in NTNG2, encoding a synaptic cell adhesion molecule, in individuals with developmental delay, hypotonia, and autistic features. Neurogenetics, 2019, 20, 209-213.	0.7	7
26	Pathogenic Variants in NUP214 Cause â€œPluggedâ€•Nuclear Pore Channels and Acute Febrile Encephalopathy. American Journal of Human Genetics, 2019, 105, 48-64.	2.6	29
27	Homozygous stop-gain variant in LRRC32, encoding a TGFÎ² receptor, associated with cleft palate, proliferative retinopathy, and developmental delay. European Journal of Human Genetics, 2019, 27, 1315-1319.	1.4	7
28	De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smithâ€“Magenis syndrome. Genome Medicine, 2019, 11, 12.	3.6	23
29	Biallelic variants in AGTPBP1, involved in tubulin deglutamylation, are associated with cerebellar degeneration and motor neuropathy. European Journal of Human Genetics, 2019, 27, 1419-1426.	1.4	25
30	Combined loss of LAP1B and LAP1C results in an early onset multisystemic nuclear envelopathy. Nature Communications, 2019, 10, 605.	5.8	40
31	A novel variant of the human mitochondrial DnaJ protein, Tid1, associates with a human disease exhibiting developmental delay and polyneuropathy. European Journal of Human Genetics, 2019, 27, 1072-1080.	1.4	9
32	Biallelic VARS variants cause developmental encephalopathy with microcephaly that is recapitulated in vars knockout zebrafish. Nature Communications, 2019, 10, 708.	5.8	40
33	Stem cell transplantation for osteopetrosis in patients beyond the age of 5 years. Blood Advances, 2019, 3, 862-868.	2.5	34
34	Two separate functions of NME3 critical for cell survival underlie a neurodegenerative disorder. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 566-574.	3.3	36
35	Heterozygous RNF13 Gain-of-Function Variants Are Associated with Congenital Microcephaly, Epileptic Encephalopathy, Blindness, and Failure to Thrive. American Journal of Human Genetics, 2019, 104, 179-185.	2.6	10
36	Severe infantile epileptic encephalopathy associated with D-glyceric aciduria: report of a novel case and review. Metabolic Brain Disease, 2019, 34, 557-563.	1.4	4

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37	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	0.7	108
38	A homozygous TTN gene variant associated with lethal congenital contracture syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1001-1005.	0.7	15
39	MARS variant associated with both recessive interstitial lung and liver disease and dominant Charcot-Marie-Tooth disease. European Journal of Medical Genetics, 2018, 61, 616-620.	0.7	14
40	Clinical and experimental evidence suggest a link between KIF7 and C5orf42-related ciliopathies through Sonic Hedgehog signaling. European Journal of Human Genetics, 2018, 26, 197-209.	1.4	23
41	EPT1 (selenoprotein I) is critical for the neural development and maintenance of plasmalogen in humans. Journal of Lipid Research, 2018, 59, 1015-1026.	2.0	79
42	Organic solute transporterâ€Î² (SLC51B) deficiency in two brothers with congenital diarrhea and features of cholestasis. Hepatology, 2018, 68, 590-598.	3.6	41
43	A homozygous deleterious CDK10 mutation in a patient with agenesis of corpus callosum, retinopathy, and deafness. American Journal of Medical Genetics, Part A, 2018, 176, 92-98.	0.7	21
44	Further delineation of the clinical spectrum of de novo TRIM8 truncating mutations. American Journal of Medical Genetics, Part A, 2018, 176, 2470-2478.	0.7	19
45	Homozygous loss-of-function mutations in MNS1 cause laterality defects and likely male infertility. PLoS Genetics, 2018, 14, e1007602.	1.5	49
46	Respiratory manifestations in LPS-responsive beige-like anchor (LRBA) protein-deficient patients. European Journal of Pediatrics, 2018, 177, 1163-1172.	1.3	20
47	Homozygous mutation in MFSD2A, encoding a lysolipid transporter for docosahexanoic acid, is associated with microcephaly and hypomyelination. Neurogenetics, 2018, 19, 227-235.	0.7	45
48	SYT1-associated neurodevelopmental disorder: a case series. Brain, 2018, 141, 2576-2591.	3.7	98
49	A patient-specific induced pluripotent stem cell model for West syndrome caused by ST3GAL3 deficiency. European Journal of Human Genetics, 2018, 26, 1773-1783.	1.4	15
50	T+ NK+ IL-2 Receptor Î³ Chain Mutation: a Challenging Diagnosis of Atypical Severe Combined Immunodeficiency. Journal of Clinical Immunology, 2018, 38, 527-536.	2.0	16
51	Homozygous mutation, p.Pro304His, in IDH3A, encoding isocitrate dehydrogenase subunit is associated with severe encephalopathy in infancy. Neurogenetics, 2017, 18, 57-61.	0.7	23
52	Compound heterozygosity for severe and hypomorphicNDUFS2mutations cause non-syndromic LHON-like optic neuropathy. Journal of Medical Genetics, 2017, 54, 346-356.	1.5	43
53	tRNA N6-adenosine threonylcarbamoyltransferase defect due to KAE1/TCS3 (OSGEP) mutation manifest by neurodegeneration and renal tubulopathy. European Journal of Human Genetics, 2017, 25, 545-551.	1.4	67
54	Tocilizumab Promotes Regulatory T-cell Alleviation in STAT3 Gain-of-functionâ”associated Multi-organ Autoimmune Syndrome. Clinical Therapeutics, 2017, 39, 444-449.	1.1	29

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55	Hypomyelinating leukodystrophy associated with a deleterious mutation in the ATRN gene. Neurogenetics, 2017, 18, 135-139.	0.7	8
56	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. American Journal of Human Genetics, 2017, 100, 676-688.	2.6	54
57	Mutations in the phosphatidylinositol glycan C (PIGC) gene are associated with epilepsy and intellectual disability. Journal of Medical Genetics, 2017, 54, 196-201.	1.5	44
58	De novo GRIN1 mutations: An emerging cause of severe early infantile encephalopathy. European Journal of Medical Genetics, 2017, 60, 317-320.	0.7	32
59	Mutations inEFL1, anSBDSpartner, are associated with infantile pancytopenia, exocrine pancreatic insufficiency and skeletal anomalies in aShwachman-Diamond like syndrome. Journal of Medical Genetics, 2017, 54, 558-566.	1.5	101
60	Mutations in TMEM260 Cause a Pediatric Neurodevelopmental, Cardiac, and Renal Syndrome. American Journal of Human Genetics, 2017, 100, 666-675.	2.6	22
61	Congenital valvular defects associated with deleterious mutations in thePLD1gene. Journal of Medical Genetics, 2017, 54, 278-286.	1.5	36
62	Homozygous mutation in PTRH2 gene causes progressive sensorineural deafness and peripheral neuropathy. American Journal of Medical Genetics, Part A, 2017, 173, 1051-1055.	0.7	9
63	TBCKâ€related intellectual disability syndrome: Case study of two patients. American Journal of Medical Genetics, Part A, 2017, 173, 491-494.	0.7	9
64	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 101, 716-724.	2.6	66
65	Homozygous null variant in CRADD, encoding an adaptor protein that mediates apoptosis, is associated with lissencephaly. American Journal of Medical Genetics, Part A, 2017, 173, 2539-2544.	0.7	18
66	Mutation in the COX4I1 gene is associated with short stature, poor weight gain and increased chromosomal breaks, simulating Fanconi anemia. European Journal of Human Genetics, 2017, 25, 1142-1146.	1.4	38
67	Heterozygous De Novo UBTF Gain-of-Function Variant Is Associated with Neurodegeneration in Childhood. American Journal of Human Genetics, 2017, 101, 267-273.	2.6	41
68	Mutations in TRAPPC12 Manifest in Progressive Childhood Encephalopathy and Golgi Dysfunction. American Journal of Human Genetics, 2017, 101, 291-299.	2.6	37
69	Compound heterozygous variants in PGAP1 causing severe psychomotor retardation, brain atrophy, recurrent apneas and delayed myelination: a case report and literature review. BMC Neurology, 2016, 16, 74.	0.8	19
70	Deleterious mutation in GPR88 is associated with chorea, speech delay, and learning disabilities. Neurology: Genetics, 2016, 2, e64.	0.9	36
71	Nemaline body myopathy caused by a novel mutation in troponin T1 (TNNT1). Muscle and Nerve, 2016, 53, 564-569.	1.0	39
72	Deficiency of HTRA2/Omi is associated with infantile neurodegeneration and 3-methylglutaconic aciduria. Journal of Medical Genetics, 2016, 53, 690-696.	1.5	30

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73	Fatal infantile mitochondrial encephalomyopathy, hypertrophic cardiomyopathy and optic atrophy associated with a homozygousOPA1mutation. Journal of Medical Genetics, 2016, 53, 127-131.	1.5	91
74	Altered RNA metabolism due to a homozygousRBM7mutation in a patient with spinal motor neuropathy. Human Molecular Genetics, 2016, 25, ddw149.	1.4	35
75	Mutations in TBCK, Encoding TBC1-Domain-Containing Kinase, Lead to a Recognizable Syndrome of Intellectual Disability and Hypotonia. American Journal of Human Genetics, 2016, 98, 782-788.	2.6	50
76	Homozygous mutation in the APOA1BP is associated with a lethal infantile leukoencephalopathy. Neurogenetics, 2016, 17, 187-190.	0.7	32
77	Recurrent De Novo Dominant Mutations in SLC25A4 Cause Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number. American Journal of Human Genetics, 2016, 99, 860-876.	2.6	93
78	Infantile Neurodegenerative Disorder Associated with Mutations inTBCD, an Essential Gene in the Tubulin Heterodimer Assembly Pathway. Human Molecular Genetics, 2016, 25, ddw292.	1.4	25
79	Mitochondrial hepato-encephalopathy due to deficiency of QIL1/MIC13 (C19orf70), a MICOS complex subunit. European Journal of Human Genetics, 2016, 24, 1778-1782.	1.4	44
80	Therapy with eculizumab for patients with CD59 p.Cys89Tyr mutation. Annals of Neurology, 2016, 80, 708-717.	2.8	41
81	Novel Homozygous Missense Mutation in SPG20 Gene Results in Troyer Syndrome Associated with Mitochondrial Cytochrome c Oxidase Deficiency. JIMD Reports, 2016, 33, 55-60.	0.7	15
82	Variants in HNRNPH2 on the X Chromosome Are Associated with a Neurodevelopmental Disorder in Females. American Journal of Human Genetics, 2016, 99, 728-734.	2.6	75
83	Extending the Clinical Phenotype of Adenosine Deaminase 2 Deficiency. Journal of Pediatrics, 2016, 177, 316-320.	0.9	87
84	PARP10 deficiency manifests by severe developmental delay and DNA repair defect. Neurogenetics, 2016, 17, 227-232.	0.7	17
85	A mutation in the THG1L gene in a family with cerebellar ataxia and developmental delay. Neurogenetics, 2016, 17, 219-225.	0.7	17
86	Early onset combined immunodeficiency and autoimmunity in patients with loss-of-function mutation in LAT. Journal of Experimental Medicine, 2016, 213, 1185-1199.	4.2	57
87	An Ashkenazi founder mutation in the PKHD1 gene. European Journal of Medical Genetics, 2016, 59, 86-90.	0.7	6
88	Deep intronic mis-splicing mutation in JAK3 gene underlies T â˚' B + NK â˚' severe combined immunodeficiency phenotype. Clinical Immunology, 2016, 163, 91-95.	1.4	13
89	Microcephaly-dystonia due to mutated PLEKHG2 with impaired actin polymerization. Neurogenetics, 2016, 17, 25-30.	0.7	8
90	TECPR2 mutations cause a new subtype of familial dysautonomia like hereditary sensory autonomic neuropathy with intellectual disability. European Journal of Paediatric Neurology, 2016, 20, 69-79.	0.7	45

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91	Deficiency of the alkaline ceramidase ACER3 manifests in early childhood by progressive leukodystrophy. Journal of Medical Genetics, 2016, 53, 389-396.	1.5	49
92	Leukoencephalopathy and early death associated with an Ashkenazi-Jewish founder mutation in the Hikeshi gene. Journal of Medical Genetics, 2016, 53, 132-137.	1.5	21
93	Early-onset Evans syndrome, immunodeficiency, and premature immunosenescence associated with tripeptidyl-peptidase II deficiency. Blood, 2015, 125, 753-761.	0.6	66
94	Extending the clinical and immunological phenotype of human interleukin-21 receptor deficiency. Haematologica, 2015, 100, e72-e76.	1.7	41
95	Clinical and biochemical characterization of four patients with mutations in ECHS1. Orphanet Journal of Rare Diseases, 2015, 10, 79.	1.2	68
96	Devastating recurrent brain ischemic infarctions and retinal disease in pediatric patients with CD59 deficiency. European Journal of Paediatric Neurology, 2015, 19, 688-693.	0.7	18
97	Mitochondrial complex IV deficiency, caused by mutated COX6B1, is associated with encephalomyopathy, hydrocephalus and cardiomyopathy. European Journal of Human Genetics, 2015, 23, 159-164.	1.4	82
98	Truncating Mutation in the Nitric Oxide Synthase 1 Gene Is Associated With Infantile Achalasia. Gastroenterology, 2015, 148, 533-536.e4.	0.6	37
99	Hindbrain malformation and myoclonic seizures associated with a deleterious mutation in the INPP4A gene. Neurogenetics, 2015, 16, 23-26.	0.7	10
100	A defect in the retromer accessory protein, SNX27, manifests by infantile myoclonic epilepsy and neurodegeneration. Neurogenetics, 2015, 16, 215-221.	0.7	44
101	Enteroviral Infection in a Patient with BLNK Adaptor Protein Deficiency. Journal of Clinical Immunology, 2015, 35, 356-360.	2.0	22
102	The 3â€² addition of CCA to mitochondrial tRNASer(AGY) is specifically impaired in patients with mutations in the tRNA nucleotidyl transferase TRNT1. Human Molecular Genetics, 2015, 24, 2841-2847.	1.4	65
103	Autoimmune lymphoproliferative syndrome-like disease in patients with LRBA mutation. Clinical Immunology, 2015, 159, 84-92.	1.4	96
104	A human laterality disorder caused by a homozygous deleterious mutation inMMP21. Journal of Medical Genetics, 2015, 52, 840-847.	1.5	46
105	Joubert Syndrome in French Canadians and Identification of Mutations in CEP104. American Journal of Human Genetics, 2015, 97, 744-753.	2.6	56
106	Mutations inSLC1A4, encoding the brain serine transporter, are associated with developmental delay, microcephaly and hypomyelination. Journal of Medical Genetics, 2015, 52, 541-547.	1.5	68
107	Hypomyelination and developmental delay associated withVPS11mutation in Ashkenazi-Jewish patients. Journal of Medical Genetics, 2015, 52, 749-753.	1.5	41
108	A human laterality disorder associated with a homozygous WDR16 deletion. European Journal of Human Genetics, 2015, 23, 1262-1265.	1.4	43

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109	Cytokine secretion and NK cell activity in human ADAM17 deficiency. Oncotarget, 2015, 6, 44151-44160.	0.8	33
110	Delineation of C12orf65-related phenotypes: a genotypeâ€“phenotype relationship. European Journal of Human Genetics, 2014, 22, 1019-1025.	1.4	48
111	Deleterious mutation in FDX1L gene is associated with a novel mitochondrial muscle myopathy. European Journal of Human Genetics, 2014, 22, 902-906.	1.4	65
112	A novel mutation in TTC19 associated with isolated complex III deficiency, cerebellar hypoplasia, and bilateral basal ganglia lesions. Frontiers in Genetics, 2014, 5, 397.	1.1	17
113	Conotruncal malformations and absent thymus due to a deleterious NKX2-6 mutation. Journal of Medical Genetics, 2014, 51, 268-270.	1.5	28
114	TRMT10A dysfunction is associated with abnormalities in glucose homeostasis, short stature and microcephaly. Journal of Medical Genetics, 2014, 51, 581-586.	1.5	83
115	Mutations in the Mitochondrial Citrate Carrier SLC25A1 are Associated with Impaired Neuromuscular Transmission. Journal of Neuromuscular Diseases, 2014, 1, 75-90.	1.1	69
116	A Novel Familial Mutation in the PCSK1 Gene That Alters the Oxyanion Hole Residue of Proprotein Convertase 1/3 and Impairs Its Enzymatic Activity. PLoS ONE, 2014, 9, e108878.	1.1	19
117	New Mutation in Chromosome 9 Associated with Familial Autosomal Recessive Macrothrombocytopenia. Blood, 2014, 124, 2776-2776.	0.6	0
118	West syndrome caused by ST3Galâ€II deficiency. Epilepsia, 2013, 54, e24-7.	2.6	58
119	Isolated truncus arteriosus associated with a mutation in the plexinâ€Đ1 gene. American Journal of Medical Genetics, Part A, 2013, 161, 3115-3120.	0.7	33
120	Deficiency of caspase recruitment domain family, memberÂ11 (CARD11), causes profound combined immunodeficiency in human subjects. Journal of Allergy and Clinical Immunology, 2013, 131, 477-485.e1.	1.5	166
121	Exome sequencing identifies a new mutation in SERAC1 in a patient with 3-methylglutaconic aciduria. Molecular Genetics and Metabolism, 2013, 110, 73-77.	0.5	33
122	Agenesis of corpus callosum and optic nerve hypoplasia due to mutations inSLC25A1encoding the mitochondrial citrate transporter. Journal of Medical Genetics, 2013, 50, 240-245.	1.5	60
123	West syndrome, microcephaly, grey matter heterotopia and hypoplasia of corpus callosum due to a novel ARFGEF2 mutation. Journal of Medical Genetics, 2013, 50, 772-775.	1.5	24
124	Mutations in SLC35A3 cause autism spectrum disorder, epilepsy and arthrogryposis. Journal of Medical Genetics, 2013, 50, 733-739.	1.5	55
125	<scp>VPS</scp> 45â€associated primary infantile myelofibrosis â€“ Successful treatment with hematopoietic stem cell transplantation. Pediatric Transplantation, 2013, 17, 820-825.	0.5	13
126	Early infantile epileptic encephalopathy associated with a high voltage gated calcium channelopathy. Journal of Medical Genetics, 2013, 50, 118-123.	1.5	60

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127	CD59 deficiency is associated with chronic hemolysis and childhood relapsing immune-mediated polyneuropathy. Blood, 2013, 121, 129-135.	0.6	142
128	The Thr224Asn mutation in the VPS45 gene is associated with the congenital neutropenia and primary myelofibrosis of infancy. Blood, 2013, 121, 5078-5087.	0.6	70
129	LRRC6 Mutation Causes Primary Ciliary Dyskinesia with Dynein Arm Defects. PLoS ONE, 2013, 8, e59436.	1.1	87
130	CCDC65 Mutation Causes Primary Ciliary Dyskinesia with Normal Ultrastructure and Hyperkinetic Cilia. PLoS ONE, 2013, 8, e72299.	1.1	108
131	AnSNX10mutation causes malignant osteopetrosis of infancy. Journal of Medical Genetics, 2012, 49, 221-226.	1.5	102
132	Two novel CCDC88C mutations confirm the role of DAPLE in autosomal recessive congenital hydrocephalus. Journal of Medical Genetics, 2012, 49, 708-712.	1.5	67
133	Intractable epilepsy of infancy due to homozygous mutation in the EFHC1 gene. Epilepsia, 2012, 53, 1436-1440.	2.6	16
134	A human laterality disorder associated with recessiveCCDC11mutation. Journal of Medical Genetics, 2012, 49, 386-390.	1.5	49
135	A Deleterious Mutation in DNAJC6 Encoding the Neuronal-Specific Clathrin-Uncoating Co-Chaperone Auxilin, Is Associated with Juvenile Parkinsonism. PLoS ONE, 2012, 7, e36458.	1.1	256
136	Molecular and biochemical characterization of a unique mutation in CCS, the human copper chaperone to superoxide dismutase. Human Mutation, 2012, 33, 1207-1215.	1.1	34
137	Hereditary sensory autonomic neuropathy caused by a mutation in dystonin. Annals of Neurology, 2012, 71, 569-572.	2.8	128
138	Mutations in SLC33A1 Cause a Lethal Autosomal-Recessive Disorder with Congenital Cataracts, Hearing Loss, and Low Serum Copper and Ceruloplasmin. American Journal of Human Genetics, 2012, 90, 61-68.	2.6	85
139	Infantile Cerebellar-Retinal Degeneration Associated with a Mutation in Mitochondrial Aconitase, ACO2. American Journal of Human Genetics, 2012, 90, 518-523.	2.6	93
140	Combined OXPHOS complex I and IV defect, due to mutated complex I assembly factor C20ORF7. Journal of Inherited Metabolic Disease, 2012, 35, 125-131.	1.7	52
141	CD59 Deficiency Is Associated with Chronic Hemolysis and Childhood Chronic Immune Mediated Neuropathy. Blood, 2012, 120, 3187-3187.	0.6	14
142	Early prenatal ventriculomegaly due to an AIFM1 mutation identified by linkage analysis and whole exome sequencing. Molecular Genetics and Metabolism, 2011, 104, 517-520.	0.5	89
143	TMEM70 mutations are a common cause of nuclear encoded ATP synthase assembly defect: further delineation of a new syndrome. Journal of Medical Genetics, 2011, 48, 177-182.	1.5	61
144	Mutations in the Mitochondrial Seryl-tRNA Synthetase Cause Hyperuricemia, Pulmonary Hypertension, Renal Failure in Infancy and Alkalosis, HUPRA Syndrome. American Journal of Human Genetics, 2011, 88, 193-200.	2.6	161

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145	A Fatal Mitochondrial Disease Is Associated with Defective NFU1 Function in the Maturation of a Subset of Mitochondrial Fe-S Proteins. American Journal of Human Genetics, 2011, 89, 656-667.	2.6	262
146	Exome sequencing and disease-network analysis of a single family implicate a mutation in KIF1A in hereditary spastic paraparesis. Genome Research, 2011, 21, 658-664.	2.4	172
147	IL-2-inducible T-cell kinase deficiency: clinical presentation and therapeutic approach. Haematologica, 2011, 96, 472-476.	1.7	105
148	Joubert Syndrome 2 (JBTS2) in Ashkenazi Jews Is Associated with a TMEM216 Mutation. American Journal of Human Genetics, 2010, 86, 93-97.	2.6	89
149	Acute Infantile Liver Failure Due to Mutations in the TRMU Gene. American Journal of Human Genetics, 2010, 86, 295.	2.6	0
150	Infantile Cerebral and Cerebellar Atrophy Is Associated with a Mutation in the MED17 Subunit of the Transcription Preinitiation Mediator Complex. American Journal of Human Genetics, 2010, 87, 667-670.	2.6	58
151	LPIN1 gene mutations: a major cause of severe rhabdomyolysis in early childhood. Human Mutation, 2010, 31, E1564-E1573.	1.1	112
152	SLC25A19 mutation as a cause of neuropathy and bilateral striatal necrosis. Annals of Neurology, 2009, 66, 419-424.	2.8	74
153	Mutated NDUFS6 is the cause of fatal neonatal lactic acidemia in Caucasus Jews. European Journal of Human Genetics, 2009, 17, 1200-1203.	1.4	41
154	Mutations in LPIN1 Cause Recurrent Acute Myoglobinuria in Childhood. American Journal of Human Genetics, 2009, 84, 95.	2.6	1
155	Exocrine Pancreatic Insufficiency, Dyserythropoeitic Anemia, and Calvarial Hyperostosis Are Caused by a Mutation in the COX4I2 Gene. American Journal of Human Genetics, 2009, 84, 412-417.	2.6	78
156	Acute Infantile Liver Failure Due to Mutations in the TRMU Gene. American Journal of Human Genetics, 2009, 85, 401-407.	2.6	205
157	Cardiac-Targeted Transgenic Mutant Mitochondrial Enzymes: mtDNA Defects, Antiretroviral Toxicity and Cardiomyopathy. Cardiovascular Toxicology, 2008, 8, 57-69.	1.1	17
158	Mitochondrial complex I deficiency caused by a deleterious NDUFA11 mutation. Annals of Neurology, 2008, 63, 405-408.	2.8	103
159	FASTKD2 Nonsense Mutation in an Infantile Mitochondrial Encephalomyopathy Associated with Cytochrome C Oxidase Deficiency. American Journal of Human Genetics, 2008, 83, 415-423.	2.6	107
160	Mutations in LPIN1 Cause Recurrent Acute Myoglobinuria in Childhood. American Journal of Human Genetics, 2008, 83, 489-494.	2.6	189
161	The H Syndrome Is Caused by Mutations in the Nucleoside Transporter hENT3. American Journal of Human Genetics, 2008, 83, 529-534.	2.6	166
162	Mutations in the Fatty Acid 2-Hydroxylase Gene Are Associated with Leukodystrophy with Spastic Paraparesis and Dystonia. American Journal of Human Genetics, 2008, 83, 643-648.	2.6	193

#	Article	IF	Citations
163	The H syndrome: A genodermatosis characterized by indurated, hyperpigmented, and hypertrichotic skin with systemic manifestations. Journal of the American Academy of Dermatology, 2008, 59, 79-85.	0.6	117
164	C6ORF66 Is an Assembly Factor of Mitochondrial Complex I. American Journal of Human Genetics, 2008, 82, 32-38.	2.6	155
165	Short-chain acyl-CoA dehydrogenase gene mutation (c.319C>T) presents with clinical heterogeneity and is candidate founder mutation in individuals of Ashkenazi Jewish origin. Molecular Genetics and Metabolism, 2008, 93, 179-189.	0.5	61
166	The unique neuroradiology of complex I deficiency due to NDUFA12L defect. Molecular Genetics and Metabolism, 2008, 94, 78-82.	0.5	46
167	TAT-mediated Delivery of LAD Restores Pyruvate Dehydrogenase Complex Activity in the Mitochondria of Patients with LAD Deficiency. Molecular Therapy, 2008, 16, 691-697.	3.7	38
168	Cryptic proteolytic activity of dihydrolipoamide dehydrogenase. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 6158-6163.	3.3	107
169	Seemingly Neutral Polymorphic Variants May Confer Immunity to Splicing-Inactivating Mutations: A Synonymous SNP in Exon 5 of MCAD Protects from Deleterious Mutations in a Flanking Exonic Splicing Enhancer. American Journal of Human Genetics, 2007, 80, 416-432.	2.6	140
170	Deleterious Mutation in the Mitochondrial Arginylâ€“Transfer RNA Synthetase Gene Is Associated with Pontocerebellar Hypoplasia. American Journal of Human Genetics, 2007, 81, 857-862.	2.6	306
171	Biochemical Assays for Mitochondrial Activity: Assays of TCA Cycle Enzymes and PDHc. Methods in Cell Biology, 2007, 80, 199-222.	0.5	74
172	Distinct Clinical Phenotypes Associated with a Mutation in the Mitochondrial Translation Elongation Factor EFTs. American Journal of Human Genetics, 2006, 79, 869-877.	2.6	169
173	Deficiency of the ADP-Forming Succinyl-CoA Synthase Activity Is Associated with Encephalomyopathy and Mitochondrial DNA Depletion. American Journal of Human Genetics, 2005, 76, 1081-1086.	2.6	284
174	Novel Mutations in the PEX2 Gene of Four Unrelated Patients with a Peroxisome Biogenesis Disorder. Pediatric Research, 2004, 55, 431-436.	1.1	21
175	Evaluation of enzymatic assays and compounds affecting ATP production in mitochondrial respiratory chain complex I deficiency. Analytical Biochemistry, 2004, 335, 66-72.	1.1	56
176	Mitochondrial deoxyribonucleoside triphosphate pools in thymidine kinase 2 deficiency. Biochemical and Biophysical Research Communications, 2003, 310, 963-966.	1.0	41
177	mtDNA depletion myopathy: elucidation of the tissue specificity in the mitochondrial thymidine kinase (TK2) deficiency. Molecular Genetics and Metabolism, 2003, 79, 1-5.	0.5	89
178	Inherited Mitochondrial DNA Depletion. Pediatric Research, 2003, 54, 153-159.	1.1	27
179	Clinical Characteristics and Muscle Pathology in Myopathic Mitochondrial DNA Depletion. Journal of Child Neurology, 2002, 17, 499-504.	0.7	13
180	Hypocarnitinemia in lysinuric protein intolerance. Molecular Genetics and Metabolism, 2002, 76, 81-83.	0.5	9

#	Article	IF	Citations
181	Infantile citrullinemia caused by citrin deficiency with increased dibasic amino acids. Molecular Genetics and Metabolism, 2002, 77, 202-208.	0.5	56
182	N-acetylglutamate synthase deficiency and the treatment of hyperammonemic encephalopathy. Annals of Neurology, 2002, 52, 845-849.	2.8	42
183	Depletion of the other genome-mitochondrial DNA depletion syndromes in humans. Journal of Molecular Medicine, 2002, 80, 389-396.	1.7	54
184	A Recessive Contiguous Gene Deletion of Chromosome 2p16 Associated with Cystinuria and a Mitochondrial Disease. American Journal of Human Genetics, 2001, 69, 869-875.	2.6	69
185	Type III 3-Methylglutaconic Aciduria (Optic Atrophy Plus Syndrome, or Costeff Optic Atrophy) Tj ETQq1 1 0.784314 rgBT /Overlock 10 Tf of Human Genetics, 2001, 69, 1218-1224.	2.6	166
186	The deoxyguanosine kinase gene is mutated in individuals with depleted hepatocerebral mitochondrial DNA. Nature Genetics, 2001, 29, 337-341.	9.4	521
187	Mutant mitochondrial thymidine kinase in mitochondrial DNA depletion myopathy. Nature Genetics, 2001, 29, 342-344.	9.4	551
188	DICHLOROACETATE TREATMENT FOR SEVERE REFRACTORY METABOLIC ACIDOSIS DURING NEONATAL SEPSIS. Pediatric Infectious Disease Journal, 2001, 20, 218-219.	1.1	4
189	Demonstration of a New Pathogenic Mutation in Human Complex I Deficiency: A 5-bp Duplication in the Nuclear Gene Encoding the 18-kD (AQDQ) Subunit. American Journal of Human Genetics, 1998, 62, 262-268.	2.6	268
190	Type IV 3-Methylglutaconic (3-MGC) aciduria: A new case presenting with hepatic dysfunction. Pediatric Neurology, 1997, 17, 353-355.	1.0	10