Tobias B Haack

List of Publications by Year
in descending order

174	7,033	53660 45	79541 73
papers	citations	h-index	g-index

177	177	177	12911
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Bi-allelic loss-of-function variants inKIF21Acause severe fetal akinesia with arthrogryposis multiplex. Journal of Medical Genetics, 2023, 60, 48-56.	1.5	26
2	Mutations at a split codon in the GTPase-encoding domain of OPA1 cause dominant optic atrophy through different molecular mechanisms. Human Molecular Genetics, 2022, 31, 761-774.	1.4	6
3	Human COQ4 deficiency: delineating the clinical, metabolic and neuroimaging phenotypes. Journal of Medical Genetics, 2022, 59, 878-887.	1.5	9
4	NPTX1 mutations trigger endoplasmic reticulum stress and cause autosomal dominant cerebellar ataxia. Brain, 2022, 145, 1519-1534.	3.7	10
5	Sensory axonal neuropathy in RFC1-disease: tip of the iceberg of broad subclinical multisystemic neurodegeneration. Brain, 2022, 145, e6-e9.	3.7	6
6	Multisystemic neurodegeneration caused by biallelic pentanucleotide expansions in RFC1. Parkinsonism and Related Disorders, 2022, 95, 54-56.	1.1	8
7	DNAJC30 disease-causing gene variants in a large Central European cohort of patients with suspected Leberâ€™s hereditary optic neuropathy and optic atrophy. Journal of Medical Genetics, 2022, 59, 1027-1034.	1.5	15
8	Characterization of cognitive impairment in adult polyglucosan body disease. Journal of Neurology, 2022, 269, 2854-2861.	1.8	6
9	Disruption of MeCP2â€“TCF20 complex underlies distinct neurodevelopmental disorders. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	3.3	15
10	Clinicoâ€radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. Human Mutation, 2022, 43, 403-419.	1.1	9
11	GestaltMatcher facilitates rare disease matching using facial phenotype descriptors. Nature Genetics, 2022, 54, 349-357.	9.4	73
12	De Novo and Dominantly Inherited <scp>SPTAN1</scp> Mutations Cause Spastic Paraplegia and Cerebellar Ataxia. Movement Disorders, 2022, 37, 1175-1186.	2.2	9
13	A Novel, Apparently Silent Variant in MFSD8 Causes Neuronal Ceroid Lipofuscinosis with Marked Intrafamilial Variability. International Journal of Molecular Sciences, 2022, 23, 2271.	1.8	3
14	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. Genetics in Medicine, 2022, 24, 1283-1296.	1.1	9
15	A Novel <scp>NPTX1 de novo</scp> Variant in a Lateâ€Onset Ataxia Patient. Movement Disorders, 2022, 37, 1319-1321.	2.2	2
16	Molecular Properties of Human Guanylate Cyclase-Activating Protein 3 (GCAP3) and Its Possible Association with Retinitis Pigmentosa. International Journal of Molecular Sciences, 2022, 23, 3240.	1.8	3
17	Biallelic ADAM22 pathogenic variants cause progressive encephalopathy and infantile-onset refractory epilepsy. Brain, 2022, 145, 2301-2312.	3.7	8
18	<scp>CCDC82</scp> frameshift mutation associated with intellectual disability, spastic paraparesis, and dysmorphic features. Clinical Genetics, 2022, 102, 80-81.	1.0	4

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19	The TLRâ€chaperone CNPY3 is a critical regulator of NLRP3â€inflammasome activation. European Journal of Immunology, 2022, 52, 907-923.	1.6	6
20	Successful treatment with azacitidine in VEXAS syndrome with prominent myofasciitis. Rheumatology, 2022, 61, e117-e119.	0.9	16
21	Transcript-Specific Loss-of-Function Variants in VPS16 Are Enriched in Patients With Dystonia. Neurology: Genetics, 2022, 8, e644.	0.9	9
22	Heterozygous frameshift variants in HNRNPA2B1 cause early-onset oculopharyngeal muscular dystrophy. Nature Communications, 2022, 13, 2306.	5.8	20
23	A single center experience of prenatal parentâ€fetus trio exome sequencing for pregnancies with congenital anomalies. Prenatal Diagnosis, 2022, 42, 901-910.	1.1	4
24	Expanded Genetic Spectrum and Variable Disease Onset in <scp>AOPEP</scp>â€Associated Dystonia. Movement Disorders, 2022, 37, 1113-1115.	2.2	3
25	Biallelic variants in <scp>ZNF142</scp> lead to a syndromic neurodevelopmental disorder. Clinical Genetics, 2022, 102, 98-109.	1.0	6
26	Adultâ€Onset Neurodegeneration in Nucleotide Excision Repair Disorders (<scp>NERD_{ND}</scp>): Time to Move Beyond the Skin. Movement Disorders, 2022, 37, 1707-1718.	2.2	7
27	GFPT1-Associated Congenital Myasthenic Syndrome Mimicking a Glycogen Storage Disease â€“ Diagnostic Pitfalls in Myopathology Solved by Next-Generation-Sequencing. Journal of Neuromuscular Diseases, 2022, , 1-9.	1.1	1
28	Ophthalmic and Genetic Features of Bardet Biedl Syndrome in a German Cohort. Genes, 2022, 13, 1218.	1.0	5
29	Expanding PRDX3 disease: broad range of onset age and infratentorial MRI signal changes. Brain, 2022, 145, e95-e98.	3.7	3
30	Tetraparesis and sensorimotor axonal polyneuropathy due to co-occurrence of Pompe disease and hereditary ATTR amyloidosis. Neurological Sciences, 2021, 42, 1523-1525.	0.9	2
31	De novo stop-loss variants in CLDN11 cause hypomyelinating leukodystrophy. Brain, 2021, 144, 411-419.	3.7	12
32	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	1.1	16
33	Correspondence on â€œClinical, neuropathological, and genetic characterization of STUB1 variants in cerebellar ataxias: a frequent cause of predominant cognitive impairmentâ€by Roux et al.. Genetics in Medicine, 2021, 23, 1171-1172.	1.1	2
34	Clinical Phenotype of PDE6B-Associated Retinitis Pigmentosa. International Journal of Molecular Sciences, 2021, 22, 2374.	1.8	12
35	Identification and Characterization of a Novel Splice Site Mutation Associated with Glycogen Storage Disease Type VI in Two Unrelated Turkish Families. Diagnostics, 2021, 11, 500.	1.3	3
36	Pitfalls in Genetic Diagnostics: Why Phenotyping is Essential. Neuropediatrics, 2021, 52, 274-283.	0.3	1

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37	A de novo STUB1 variant associated with an early adult-onset multisystemic ataxia phenotype. Journal of Neurology, 2021, 268, 3845-3851.	1.8	7
38	Clinical Characteristics of POC1B-Associated Retinopathy and Assignment of Pathogenicity to Novel Deep Intronic and Non-Canonical Splice Site Variants. International Journal of Molecular Sciences, 2021, 22, 5396.	1.8	13
39	Characterization of PARP6 Function in Knockout Mice and Patients with Developmental Delay. Cells, 2021, 10, 1289.	1.8	7
40	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	3.7	22
41	Teaching Video NeuroImages: New STUB1 Variant Causes Chorea, Tremor, Dystonia, Myoclonus, Ataxia, Depression, Cognitive Impairment, Epilepsy, and Superficial Siderosis. Neurology, 2021, 97, 10.1212/WNL.0000000000012264.	1.5	4
42	Bi-allelic loss-of-function variants in BCAS3 cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2021, 108, 1069-1082.	2.6	8
43	Impaired glucose-1,6-biphosphate production due to bi-allelic PGM2L1 mutations is associated with a neurodevelopmental disorder. American Journal of Human Genetics, 2021, 108, 1151-1160.	2.6	9
44	Zonisamideâ€responsive myoclonus in SEMA6Bâ€associated progressive myoclonic epilepsy. Annals of Clinical and Translational Neurology, 2021, 8, 1524-1527.	1.7	10
45	Further evidence for de novo variants in SYNCRIP as the cause of a neurodevelopmental disorder. Human Mutation, 2021, 42, 1094-1100.	1.1	9
46	Expansion of the mutational spectrum of BMPER leading to diaphanospondylodysostosis and description of the associated disease process. Molecular Genetics & Genomic Medicine, 2021, 9, e1767.	0.6	3
47	Expanded phenotype of AARS1-related white matter disease. Genetics in Medicine, 2021, 23, 2352-2359.	1.1	8
48	Clinical and molecular delineation of <scp>PUS3</scp>â€associated neurodevelopmental disorders. Clinical Genetics, 2021, 100, 628-633.	1.0	23
49	Detection of mobile elements insertions for routine clinical diagnostics in targeted sequencing data. Molecular Genetics & Genomic Medicine, 2021, 9, e1807.	0.6	6
50	Bi-allelic truncating mutations in VWA1 cause neuromyopathy. Brain, 2021, 144, 574-583.	3.7	16
51	Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. American Journal of Human Genetics, 2021, 108, 2006-2016.	2.6	11
52	Angiokeratoma corporis diffusum with severe acroparesthesia, an endothelial abnormality, and inconspicuous genetic findings. Journal of Cutaneous Pathology, 2021, , .	0.7	1
53	De novo variants in SLC12A6 cause sporadic early-onset progressive sensorimotor neuropathy. Journal of Medical Genetics, 2020, 57, 283-288.	1.5	14
54	First submicroscopic inversion of the OPA1 gene identified in dominant optic atrophy â€“ a case report. BMC Medical Genetics, 2020, 21, 236.	2.1	17

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55	Biallelic loss of function variants in <scp>SYT2</scp> cause a treatable congenital onset presynaptic myasthenic syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2272-2283.	0.7	20
56	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. Genetics in Medicine, 2020, 22, 1863-1873.	1.1	19
57	De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. American Journal of Human Genetics, 2020, 107, 311-324.	2.6	32
58	Clinical and molecular description of 19 patients with GATAD2B-Associated Neurodevelopmental Disorder (GAND). European Journal of Medical Genetics, 2020, 63, 104004.	0.7	7
59	Paroxysmal and non-paroxysmal dystonia in 3 patients with biallelic ECHS1 variants: Expanding the neurological spectrum and therapeutic approaches. European Journal of Medical Genetics, 2020, 63, 104046.	0.7	12
60	Novel mutation points to a hot spot in CDKN1C causing Silverâ€“Russell syndrome. Clinical Epigenetics, 2020, 12, 152.	1.8	12
61	Bi-allelic HPDL Variants Cause a Neurodegenerative Disease Ranging from Neonatal Encephalopathy to Adolescent-Onset Spastic Paraplegia. American Journal of Human Genetics, 2020, 107, 364-373.	2.6	30
62	Defining diagnostic cutoffs in neurological patients for serum very long chain fatty acids (VLCFA) in genetically confirmed X-Adrenoleukodystrophy. Scientific Reports, 2020, 10, 15093.	1.6	12
63	LINS1-associated neurodevelopmental disorder. Neurology: Genetics, 2020, 6, e500.	0.9	3
64	Novel Biallelic <scp>CTSD</scp> Gene Variants Cause Lateâ€Onset Ataxia and Retinitis Pigmentosa. Movement Disorders, 2020, 35, 1280-1282.	2.2	3
65	Delineating MT-ATP6 -associated disease. Neurology: Genetics, 2020, 6, e393.	0.9	73
66	First-line exome sequencing in Palestinian and Israeli Arabs with neurological disorders is efficient and facilitates disease gene discovery. European Journal of Human Genetics, 2020, 28, 1034-1043.	1.4	20
67	Genetic basis of neurodevelopmental disorders in 103 Jordanian families. Clinical Genetics, 2020, 97, 621-627.	1.0	19
68	Pontocerebellar hypoplasia type 11: Does the genetic defect determine timing of cerebellar pathology?. European Journal of Medical Genetics, 2020, 63, 103938.	0.7	4
69	Clinicoâ€Genetic, Imaging and Molecular Delineation of <scp>COQ8A</scp>â€Ataxia: A Multicenter Study of 59 Patients. Annals of Neurology, 2020, 88, 251-263.	2.8	52
70	IRF2BPL mutation causes nigrostriatal degeneration presenting with dystonia, spasticity and keratoconus. Parkinsonism and Related Disorders, 2020, 79, 141-143.	1.1	14
71	Clinico-Genetic, Imaging and Molecular Delineation of COQ8A-Ataxia: A Multicenter Study of 59 Patients. , 2020, 88, 251.		1
72	KCNC1â€related disorders: new de novo variants expand the phenotypic spectrum. Annals of Clinical and Translational Neurology, 2019, 6, 1319-1326.	1.7	43

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73	Mutations in PIGB Cause an Inherited GPI Biosynthesis Defect with an Axonal Neuropathy and Metabolic Abnormality in Severe Cases. American Journal of Human Genetics, 2019, 105, 384-394.	2.6	37
74	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. Genetics in Medicine, 2019, 21, 2521-2531.	1.1	25
75	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	2.6	56
76	SOPH syndrome in three affected individuals showing similarities with progeroid cutis laxa conditions in early infancy. Journal of Human Genetics, 2019, 64, 609-616.	1.1	14
77	Bain type of Xâ€linked syndromic mental retardation in boys. Clinical Genetics, 2019, 95, 734-735.	1.0	21
78	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	2.6	45
79	Wholeâ€exome sequencing revealed a nonsense mutation in STKLD1 causing nonâ€syndromic preâ€axial polydactyly type A affecting only upper limb. Clinical Genetics, 2019, 96, 134-139.	1.0	7
80	Truncating Mutations in UBAP1 Cause Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2019, 104, 767-773.	2.6	39
81	Diverse phenotype in patients with complex I deficiency due to mutations in NDUFB11. European Journal of Medical Genetics, 2019, 62, 103572.	0.7	22
82	The movement disorder spectrum of SCA21 (ATX-TMEM240): 3 novel families and systematic review of the literature. Parkinsonism and Related Disorders, 2019, 62, 215-220.	1.1	18
83	Blue Diaper Syndrome and PCSK1 Mutations. Pediatrics, 2018, 141, S501-S505.	1.0	14
84	Compound heterozygous SPATA5 variants in four families and functional studies of SPATA5 deficiency. European Journal of Human Genetics, 2018, 26, 407-419.	1.4	29
85	SCYL1 variants cause a syndrome with lowÎ³-glutamyl-transferase cholestasis, acute liver failure, and neurodegeneration (CALFAN). Genetics in Medicine, 2018, 20, 1255-1265.	1.1	50
86	The role of the clinician in the multiâ€omics era: are you ready?. Journal of Inherited Metabolic Disease, 2018, 41, 571-582.	1.7	55
87	Mutations outside the N-terminal part of RBCK1 may cause polyglucosan body myopathy with immunological dysfunction: expanding the genotypeâ€“phenotype spectrum. Journal of Neurology, 2018, 265, 394-401.	1.8	36
88	Clinical, biochemical, and genetic features associated with VARS2 -related mitochondrial disease. Human Mutation, 2018, 39, 563-578.	1.1	22
89	SERAC1 deficiency causes complicated HSP: evidence from a novel splice mutation in a large family. Journal of Medical Genetics, 2018, 55, 39-47.	1.5	28
90	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. JAMA Neurology, 2018, 75, 105.	4.5	26

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91	The genotypic and phenotypic spectrum of MTO1 deficiency. Molecular Genetics and Metabolism, 2018, 123, 28-42.	0.5	24
92	Bi-allelic ADPRHL2 Mutations Cause Neurodegeneration with Developmental Delay, Ataxia, and Axonal Neuropathy. American Journal of Human Genetics, 2018, 103, 817-825.	2.6	40
93	De novo FBXO11 mutations are associated with intellectual disability and behavioural anomalies. Human Genetics, 2018, 137, 401-411.	1.8	29
94	PRUNE1 Deficiency: Expanding the Clinical and Genetic Spectrum. Neuropediatrics, 2018, 49, 330-338.	0.3	11
95	Isolated PREPL deficiency associated with congenital myasthenic syndrome-22. Klinische Padiatrie, 2018, 230, 281-283.	0.2	9
96	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. Orphanet Journal of Rare Diseases, 2018, 13, 120.	1.2	61
97	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 103, 305-316.	2.6	48
98	BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. Brain, 2018, 141, 2299-2311.	3.7	81
99	Mutations in PPCS, Encoding Phosphopantothenoylcysteine Synthetase, Cause Autosomal-Recessive Dilated Cardiomyopathy. American Journal of Human Genetics, 2018, 102, 1018-1030.	2.6	42
100	SYT1-associated neurodevelopmental disorder: a case series. Brain, 2018, 141, 2576-2591.	3.7	98
101	Clinical presentation and natural history of infantile-onset ascending spastic paralysis from three families with an ALS2 founder variant. Neurological Sciences, 2018, 39, 1917-1925.	0.9	18
102	Bi-allelic mutations in TRAPPC2L result in a neurodevelopmental disorder and have an impact on RAB11 in fibroblasts. Journal of Medical Genetics, 2018, 55, 753-764.	1.5	39
103	A Homozygous Splice Site Mutation in SLC25A42, Encoding the Mitochondrial Transporter of Coenzyme A, Causes Metabolic Crises and Epileptic Encephalopathy. JIMD Reports, 2018, 44, 1-7.	0.7	15
104	P 968. Vitamine B6-Dependent Epilepsy in a 14-Year-Old Girl with Drug-Resistant Seizures and Recurring Status Epilepticus. Neuropediatrics, 2018, 49, .	0.3	0
105	Exome sequencing is a valuable approach in critically ill patients with suspected monogenic disease: Diagnosis of X-linked centronuclear myopathy in preterm twins. Pediatrics and Neonatology, 2017, 58, 458-459.	0.3	2
106	Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa. American Journal of Human Genetics, 2017, 100, 216-227.	2.6	82
107	A homozygous splice variant in AP4S1 mimicking neurodegeneration with brain iron accumulation. Movement Disorders, 2017, 32, 797-799.	2.2	14
108	Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. American Journal of Human Genetics, 2017, 100, 257-266.	2.6	127

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109	Clinical Features, Molecular Heterogeneity, and Prognostic Implications in YARS2-Related Mitochondrial Myopathy. JAMA Neurology, 2017, 74, 686.	4.5	41
110	Exome sequencing revealed a splice site variant in the IQCE gene underlying post-axial polydactyly type A restricted to lower limb. European Journal of Human Genetics, 2017, 25, 960-965.	1.4	53
111	3-Methylglutaconic aciduria, a frequent but underrecognized finding in carbamoyl phosphate synthetase I deficiency. Clinica Chimica Acta, 2017, 471, 95-100.	0.5	14
112	Coexisting variants in OSTM1 and MANEAL cause a complex neurodegenerative disorder with NBIA-like brain abnormalities. European Journal of Human Genetics, 2017, 25, 1092-1095.	1.4	13
113	Genetic diagnosis of Mendelian disorders via RNA sequencing. Nature Communications, 2017, 8, 15824.	5.8	432
114	CADmutations and uridine-responsive epileptic encephalopathy. Brain, 2017, 140, 279-286.	3.7	106
115	Bainbridgeâ€“Ropers syndrome caused by loss-of-function variants in ASXL3: a recognizable condition. European Journal of Human Genetics, 2017, 25, 183-191.	1.4	35
116	Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy. American Journal of Human Genetics, 2017, 100, 151-159.	2.6	63
117	Treatable mitochondrial diseases: cofactor metabolism and beyond. Brain, 2017, 140, e11-e11.	3.7	57
118	Novel GFM2 variants associated with early-onset neurological presentations of mitochondrial disease and impaired expression of OXPHOS subunits. Neurogenetics, 2017, 18, 227-235.	0.7	10
119	Survival among children with â€œLethalâ€•congenital contracture syndrome 11 caused by novel mutations in the gliomedin gene (GLDN). Human Mutation, 2017, 38, 1477-1484.	1.1	19
120	Biallelic Mutations in LIPT2 Cause a Mitochondrial Lipoylation Defect Associated with Severe Neonatal Encephalopathy. American Journal of Human Genetics, 2017, 101, 283-290.	2.6	55
121	Monoallelic BMP2 Variants Predicted to Result in Haploinsufficiency Cause Craniofacial, Skeletal, and Cardiac Features Overlapping Those of 20p12 Deletions. American Journal of Human Genetics, 2017, 101, 985-994.	2.6	44
122	LYRM7 - associated complex III deficiency: A clinical, molecular genetic, MR tomographic, and biochemical study. Mitochondrion, 2017, 37, 55-61.	1.6	20
123	Exome sequencing reveals a novel homozygous splice site variant in the WNT1 gene underlying osteogenesis imperfecta type 3. Pediatric Research, 2017, 82, 753-758.	1.1	34
124	Rare causes of early-onset dystonia-parkinsonism with cognitive impairment: a de novo PSEN-1 mutation. Neurogenetics, 2017, 18, 175-178.	0.7	23
125	Neonatal encephalocardiomyopathy caused by mutations in VARS2. Metabolic Brain Disease, 2017, 32, 267-270.	1.4	26
126	Combined Respiratory Chain Deficiency and UQCC2 Mutations in Neonatal Encephalomyopathy: Defective Supercomplex Assembly in Complex III Deficiencies. Oxidative Medicine and Cellular Longevity, 2017, 2017, 1-11.	1.9	33

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127	Hemodialysis in MNGIE transiently reduces serum and urine levels of thymidine and deoxyuridine, but not CSF levels and neurological function. Orphanet Journal of Rare Diseases, 2017, 12, 135.	1.2	23
128	A Case of Beta-propeller Protein-associated Neurodegeneration due to a Heterozygous Deletion of. Tremor and Other Hyperkinetic Movements, 2017, 7, 465.	1.1	3
129	Sequence variants in four genes underlying Bardet-Biedl syndrome in consanguineous families. Molecular Vision, 2017, 23, 482-494.	1.1	19
130	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 217-227.	2.6	57
131	Genetic cause and prevalence of hydroxyprolinemia. Journal of Inherited Metabolic Disease, 2016, 39, 625-632.	1.7	17
132	SYNE1 ataxia is a common recessive ataxia with major non-cerebellar features: a large multi-centre study. Brain, 2016, 139, 1378-1393.	3.7	87
133	Severe respiratory complex III defect prevents liver adaptation to prolonged fasting. Journal of Hepatology, 2016, 65, 377-385.	1.8	25
134	The many faces of paediatric mitochondrial disease on neuroimaging. Child's Nervous System, 2016, 32, 2077-2083.	0.6	20
135	Absence of the Autophagy Adaptor SQSTM1/p62 Causes Childhood-Onset Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy. American Journal of Human Genetics, 2016, 99, 735-743.	2.6	99
136	Biallelic Variants in UBA5 Reveal that Disruption of the UFM1 Cascade Can Result in Early-Onset Encephalopathy. American Journal of Human Genetics, 2016, 99, 695-703.	2.6	87
137	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. American Journal of Human Genetics, 2016, 99, 674-682.	2.6	48
138	NAXE Mutations Disrupt the Cellular NAD(P)HX Repair System and Cause a Lethal Neurometabolic Disorder of Early Childhood. American Journal of Human Genetics, 2016, 99, 894-902.	2.6	75
139	Biallelic IARS Mutations Cause Growth Retardation with Prenatal Onset, Intellectual Disability, Muscular Hypotonia, and Infantile Hepatopathy. American Journal of Human Genetics, 2016, 99, 414-422.	2.6	73
140	Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency. American Journal of Human Genetics, 2016, 98, 1130-1145.	2.6	118
141	Disturbed mitochondrial and peroxisomal dynamics due to loss of MFF causes Leigh-like encephalopathy, optic atrophy and peripheral neuropathy. Journal of Medical Genetics, 2016, 53, 270-278.	1.5	105
142	Bi-allelic Truncating Mutations in TANGO2 Cause Infancy-Onset Recurrent Metabolic Crises with Encephalocardiomyopathy. American Journal of Human Genetics, 2016, 98, 358-362.	2.6	77
143	Recurrent acute liver failure due to NBAS deficiency: phenotypic spectrum, disease mechanisms, and therapeutic concepts. Journal of Inherited Metabolic Disease, 2016, 39, 3-16.	1.7	92
144	EARS2 mutations cause fatal neonatal lactic acidosis, recurrent hypoglycemia and agenesis of corpus callosum. Metabolic Brain Disease, 2016, 31, 717-721.	1.4	15

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145	Human thioredoxin 2 deficiency impairs mitochondrial redox homeostasis and causes early-onset neurodegeneration. Brain, 2016, 139, 346-354.	3.7	86
146	Fatal neonatal encephalopathy and lactic acidosis caused by a homozygous loss-of-function variant in COQ9. European Journal of Human Genetics, 2016, 24, 450-454.	1.4	45
147	Deficiency of <scp>ECHS</scp>1 causes mitochondrial encephalopathy with cardiac involvement. Annals of Clinical and Translational Neurology, 2015, 2, 492-509.	1.7	90
148	Clinical, biochemical, and genetic spectrum of seven patients with NFU1 deficiency. Frontiers in Genetics, 2015, 06, 123.	1.1	81
149	Mutations in TTC19: expanding the molecular, clinical and biochemical phenotype. Orphanet Journal of Rare Diseases, 2015, 10, 40.	1.2	25
150	Ataxia meets chorioretinal dystrophy and hypogonadism: Boucher-NeuhÃuser syndrome due toPNPLA6mutations: FigureÃ1. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 580-581.	0.9	20
151	LRPPRCmutations cause early-onset multisystem mitochondrial disease outside of the French-Canadian population. Brain, 2015, 138, 3503-3519.	3.7	81
152	Atypical Clinical Presentations of TAZ Mutations: An Underdiagnosed Cause of Growth Retardation?. JIMD Reports, 2015, 29, 89-93.	0.7	8
153	COQ4 Mutations Cause a Broad Spectrum of Mitochondrial Disorders Associated with CoQ10 Deficiency. American Journal of Human Genetics, 2015, 96, 309-317.	2.6	86
154	CLPB Mutations Cause 3-Methylglutaconic Aciduria, Progressive Brain Atrophy, Intellectual Disability, Congenital Neutropenia, Cataracts, Movement Disorder. American Journal of Human Genetics, 2015, 96, 245-257.	2.6	111
155	TRMT5 Mutations Cause a Defect in Post-transcriptional Modification of Mitochondrial tRNA Associated with Multiple Respiratory-Chain Deficiencies. American Journal of Human Genetics, 2015, 97, 319-328.	2.6	83
156	Biallelic Mutations in NBAS Cause Recurrent Acute Liver Failure with Onset in Infancy. American Journal of Human Genetics, 2015, 97, 163-169.	2.6	110
157	MRPL44 mutations cause a slowly progressive multisystem disease with childhood-onset hypertrophic cardiomyopathy. Neurogenetics, 2015, 16, 319-323.	0.7	44
158	Complex I assembly function and fatty acid oxidation enzyme activity of ACAD9 both contribute to disease severity in ACAD9 deficiency. Human Molecular Genetics, 2015, 24, 3238-3247.	1.4	53
159	De Novo Mutations in CHAMP1 Cause Intellectual Disability with Severe Speech Impairment. American Journal of Human Genetics, 2015, 97, 493-500.	2.6	71
160	A truncating PET100 variant causing fatal infantile lactic acidosis and isolated cytochrome c oxidase deficiency. European Journal of Human Genetics, 2015, 23, 935-939.	1.4	32
161	Mutations in GTPBP3 Cause a Mitochondrial Translation Defect Associated with Hypertrophic Cardiomyopathy, Lactic Acidosis, and Encephalopathy. American Journal of Human Genetics, 2014, 95, 708-720.	2.6	123
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