Tobias B Haack

List of Publications by Year
in descending order

174	7,033	53751 45	79644 73
papers	citations	h-index	g-index

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

#	Article	IF	Citations
1	Genetic diagnosis of Mendelian disorders via RNA sequencing. Nature Communications, 2017, 8, 15824.	5.8	432
2	Exome Sequencing Reveals De Novo WDR45 Mutations Causing a Phenotypically Distinct, X-Linked Dominant Form of NBIA. American Journal of Human Genetics, 2012, 91, 1144-1149.	2.6	309
3	Exome sequencing identifies ACAD9 mutations as a cause of complex I deficiency. Nature Genetics, 2010, 42, 1131-1134.	9.4	234
4	Molecular diagnosis in mitochondrial complex I deficiency using exome sequencing. Journal of Medical Genetics, 2012, 49, 277-283.	1.5	182
5	Exome Sequence Reveals Mutations in CoA Synthase as a Cause of Neurodegeneration with Brain Iron Accumulation. American Journal of Human Genetics, 2014, 94, 11-22.	2.6	176
6	ELAC2 Mutations Cause a Mitochondrial RNA Processing Defect Associated with Hypertrophic Cardiomyopathy. American Journal of Human Genetics, 2013, 93, 211-223.	2.6	127
7	Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. American Journal of Human Genetics, 2017, 100, 257-266.	2.6	127
8	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
9	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
10	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
11	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
12	CADmutations and uridine-responsive epileptic encephalopathy. Brain, 2017, 140, 279-286.	3.7	106
13	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
14	Absence of BiP Co-chaperone DNAJC3 Causes Diabetes Mellitus and Multisystemic Neurodegeneration. American Journal of Human Genetics, 2014, 95, 689-697.	2.6	100
15	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
16	SYT1-associated neurodevelopmental disorder: a case series. Brain, 2018, 141, 2576-2591.	3.7	98
17	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
18	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

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19	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
20	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
21	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
22	Human thioredoxin 2 deficiency impairs mitochondrial redox homeostasis and causes early-onset neurodegeneration. Brain, 2016, 139, 346-354.	3.7	86
23	Homozygous missense mutation in BOLA3 causes multiple mitochondrial dysfunctions syndrome in two siblings. Journal of Inherited Metabolic Disease, 2013, 36, 55-62.	1.7	83
24	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
25	Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa. American Journal of Human Genetics, 2017, 100, 216-227.	2.6	82
26	Clinical, biochemical, and genetic spectrum of seven patients with NFU1 deficiency. Frontiers in Genetics, 2015, 06, 123.	1.1	81
27	LRPPRCmutations cause early-onset multisystem mitochondrial disease outside of the French-Canadian population. Brain, 2015, 138, 3503-3519.	3.7	81
28	BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. Brain, 2018, 141, 2299-2311.	3.7	81
29	Mutation screening of 75 candidate genes in 152 complex I deficiency cases identifies pathogenic variants in 16 genes includingNDUFB9. Journal of Medical Genetics, 2012, 49, 83-89.	1.5	78
30	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
31	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
32	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
33	Delineating MT-ATP6 -associated disease. Neurology: Genetics, 2020, 6, e393.	0.9	73
34	GestaltMatcher facilitates rare disease matching using facial phenotype descriptors. Nature Genetics, 2022, 54, 349-357.	9.4	73
35	Impaired riboflavin transport due to missense mutations in SLC52A2 causes Brownâ€Vialettoâ€Van Laere syndrome. Journal of Inherited Metabolic Disease, 2012, 35, 943-948.	1.7	72
36	De Novo Mutations in CHAMP1 Cause Intellectual Disability with Severe Speech Impairment. American Journal of Human Genetics, 2015, 97, 493-500.	2.6	71

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37	Phenotypic spectrum of eleven patients and five novel MTFMT mutations identified by exome sequencing and candidate gene screening. Molecular Genetics and Metabolism, 2014, 111, 342-352.	0.5	65
38	Mutations in APOPT1, Encoding a Mitochondrial Protein, Cause Cavitating Leukoencephalopathy with Cytochrome c Oxidase Deficiency. American Journal of Human Genetics, 2014, 95, 315-325.	2.6	64
39	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
40	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
41	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
42	Treatable mitochondrial diseases: cofactor metabolism and beyond. Brain, 2017, 140, e11-e11.	3.7	57
43	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	2.6	56
44	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
45	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
46	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
47	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
48	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
49	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
50	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. American Journal of Human Genetics, 2016, 99, 674-682.	2.6	48
51	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
52	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
53	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	2.6	45
54	MRPL44 mutations cause a slowly progressive multisystem disease with childhood-onset hypertrophic cardiomyopathy. Neurogenetics, 2015, 16, 319-323.	0.7	44

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55	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
56	KCNC1â€related disorders: new de novo variants expand the phenotypic spectrum. Annals of Clinical and Translational Neurology, 2019, 6, 1319-1326.	1.7	43
57	Mutations in PPCS, Encoding Phosphopantothenoylcysteine Synthetase, Cause Autosomal-Recessive Dilated Cardiomyopathy. American Journal of Human Genetics, 2018, 102, 1018-1030.	2.6	42
58	BPAN. International Review of Neurobiology, 2013, 110, 85-90.	0.9	41
59	Clinical Features, Molecular Heterogeneity, and Prognostic Implications in YARS2-Related Mitochondrial Myopathy. JAMA Neurology, 2017, 74, 686.	4.5	41
60	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
61	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
62	Truncating Mutations in UBAP1 Cause Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2019, 104, 767-773.	2.6	39
63	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
64	Infantile Leigh-like syndrome caused by SLC19A3 mutations is a treatable disease. Brain, 2014, 137, e295-e295.	3.7	36
65	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
66	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
67	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
68	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
69	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
70	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
71	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
72	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

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73	De novo FBXO11 mutations are associated with intellectual disability and behavioural anomalies. Human Genetics, 2018, 137, 401-411.	1.8	29
74	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
75	Neonatal encephalocardiomyopathy caused by mutations in VARS2. Metabolic Brain Disease, 2017, 32, 267-270.	1.4	26
76	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. JAMA Neurology, 2018, 75, 105.	4.5	26
77	Bi-allelic loss-of-function variants inKIF21Acause severe fetal akinesia with arthrogryposis multiplex. Journal of Medical Genetics, 2023, 60, 48-56.	1.5	26
78	Mutations in TTC19: expanding the molecular, clinical and biochemical phenotype. Orphanet Journal of Rare Diseases, 2015, 10, 40.	1.2	25
79	Severe respiratory complex III defect prevents liver adaptation to prolonged fasting. Journal of Hepatology, 2016, 65, 377-385.	1.8	25
80	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. Genetics in Medicine, 2019, 21, 2521-2531.	1.1	25
81	The genotypic and phenotypic spectrum of MTO1 deficiency. Molecular Genetics and Metabolism, 2018, 123, 28-42.	0.5	24
82	Rare causes of early-onset dystonia-parkinsonism with cognitive impairment: a de novo PSEN-1 mutation. Neurogenetics, 2017, 18, 175-178.	0.7	23
83	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
84	Clinical and molecular delineation of <scp>PUS3</scp>â€associated neurodevelopmental disorders. Clinical Genetics, 2021, 100, 628-633.	1.0	23
85	Clinical, biochemical, and genetic features associated with VARS2 -related mitochondrial disease. Human Mutation, 2018, 39, 563-578.	1.1	22
86	Diverse phenotype in patients with complex I deficiency due to mutations in NDUFB11. European Journal of Medical Genetics, 2019, 62, 103572.	0.7	22
87	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	3.7	22
88	Bain type of Xâ€linked syndromic mental retardation in boys. Clinical Genetics, 2019, 95, 734-735.	1.0	21
89	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
90	The many faces of paediatric mitochondrial disease on neuroimaging. Child's Nervous System, 2016, 32, 2077-2083.	0.6	20

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91	LYRM7 - associated complex III deficiency: A clinical, molecular genetic, MR tomographic, and biochemical study. Mitochondrion, 2017, 37, 55-61.	1.6	20
92	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
93	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
94	Heterozygous frameshift variants in HNRNPA2B1 cause early-onset oculopharyngeal muscular dystrophy. Nature Communications, 2022, 13, 2306.	5.8	20
95	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
96	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
97	Genetic basis of neurodevelopmental disorders in 103 Jordanian families. Clinical Genetics, 2020, 97, 621-627.	1.0	19
98	Sequence variants in four genes underlying Bardet-Biedl syndrome in consanguineous families. Molecular Vision, 2017, 23, 482-494.	1.1	19
99	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
100	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
101	Genetic cause and prevalence of hydroxyprolinemia. Journal of Inherited Metabolic Disease, 2016, 39, 625-632.	1.7	17
102	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
103	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	1.1	16
104	Bi-allelic truncating mutations in VWA1 cause neuromyopathy. Brain, 2021, 144, 574-583.	3.7	16
105	Successful treatment with azacitidine in VEXAS syndrome with prominent myofasciitis. Rheumatology, 2022, 61, e117-e119.	0.9	16
106	EARS2 mutations cause fatal neonatal lactic acidosis, recurrent hypoglycemia and agenesis of corpus callosum. Metabolic Brain Disease, 2016, 31, 717-721.	1.4	15
107	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
108	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

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109	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
110	A homozygous splice variant in AP4S1 mimicking neurodegeneration with brain iron accumulation. Movement Disorders, 2017, 32, 797-799.	2.2	14
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	Blue Diaper Syndrome and PCSK1 Mutations. Pediatrics, 2018, 141, S501-S505.	1.0	14
113	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
114	De novo variants in SLC12A6 cause sporadic early-onset progressive sensorimotor neuropathy. Journal of Medical Genetics, 2020, 57, 283-288.	1.5	14
115	IRF2BPL mutation causes nigrostriatal degeneration presenting with dystonia, spasticity and keratoconus. Parkinsonism and Related Disorders, 2020, 79, 141-143.	1.1	14
116	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
117	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
118	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
119	Novel mutation points to a hot spot in CDKN1C causing Silverâ€“Russell syndrome. Clinical Epigenetics, 2020, 12, 152.	1.8	12
120	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
121	De novo stop-loss variants in CLDN11 cause hypomyelinating leukodystrophy. Brain, 2021, 144, 411-419.	3.7	12
122	Clinical Phenotype of PDE6B-Associated Retinitis Pigmentosa. International Journal of Molecular Sciences, 2021, 22, 2374.	1.8	12
123	PRUNE1 Deficiency: Expanding the Clinical and Genetic Spectrum. Neuropediatrics, 2018, 49, 330-338.	0.3	11
124	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
125	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
126	Zonisamideâ€responsive myoclonus in SEMA6Bâ€associated progressive myoclonic epilepsy. Annals of Clinical and Translational Neurology, 2021, 8, 1524-1527.	1.7	10

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127	NPTX1 mutations trigger endoplasmic reticulum stress and cause autosomal dominant cerebellar ataxia. Brain, 2022, 145, 1519-1534.	3.7	10
128	Isolated PREPL deficiency associated with congenital myasthenic syndrome-22. Klinische Padiatrie, 2018, 230, 281-283.	0.2	9
129	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
130	Further evidence for de novo variants in SYNCRIP as the cause of a neurodevelopmental disorder. Human Mutation, 2021, 42, 1094-1100.	1.1	9
131	Human COQ4 deficiency: delineating the clinical, metabolic and neuroimaging phenotypes. Journal of Medical Genetics, 2022, 59, 878-887.	1.5	9
132	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
133	De Novo and Dominantly Inherited <scp>SPTAN1</scp> Mutations Cause Spastic Paraplegia and Cerebellar Ataxia. Movement Disorders, 2022, 37, 1175-1186.	2.2	9
134	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. Genetics in Medicine, 2022, 24, 1283-1296.	1.1	9
135	Transcript-Specific Loss-of-Function Variants in VPS16 Are Enriched in Patients With Dystonia. Neurology: Genetics, 2022, 8, e644.	0.9	9
136	Atypical Clinical Presentations of TAZ Mutations: An Underdiagnosed Cause of Growth Retardation?. JIMD Reports, 2015, 29, 89-93.	0.7	8
137	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
138	Expanded phenotype of AARS1-related white matter disease. Genetics in Medicine, 2021, 23, 2352-2359.	1.1	8
139	Multisystemic neurodegeneration caused by biallelic pentanucleotide expansions in RFC1. Parkinsonism and Related Disorders, 2022, 95, 54-56.	1.1	8
140	Biallelic ADAM22 pathogenic variants cause progressive encephalopathy and infantile-onset refractory epilepsy. Brain, 2022, 145, 2301-2312.	3.7	8
141	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
142	Clinical and molecular description of 19 patients with GATAD2B-Associated Neurodevelopmental Disorder (GAND). European Journal of Medical Genetics, 2020, 63, 104004.	0.7	7
143	A de novo STUB1 variant associated with an early adult-onset multisystemic ataxia phenotype. Journal of Neurology, 2021, 268, 3845-3851.	1.8	7
144	Characterization of PARP6 Function in Knockout Mice and Patients with Developmental Delay. Cells, 2021, 10, 1289.	1.8	7

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145	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
146	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
147	Detection of mobile elements insertions for routine clinical diagnostics in targeted sequencing data. Molecular Genetics & Genomic Medicine, 2021, 9, e1807.	0.6	6
148	Sensory axonal neuropathy in RFC1-disease: tip of the iceberg of broad subclinical multisystemic neurodegeneration. Brain, 2022, 145, e6-e9.	3.7	6
149	Characterization of cognitive impairment in adult polyglucosan body disease. Journal of Neurology, 2022, 269, 2854-2861.	1.8	6
150	The TLRâ€chaperone CNPY3 is a critical regulator of NLRP3â€inflammasome activation. European Journal of Immunology, 2022, 52, 907-923.	1.6	6
151	Biallelic variants in <scp>ZNF142</scp> lead to a syndromic neurodevelopmental disorder. Clinical Genetics, 2022, 102, 98-109.	1.0	6
152	Ophthalmic and Genetic Features of Bardet Biedl Syndrome in a German Cohort. Genes, 2022, 13, 1218.	1.0	5
153	Pontocerebellar hypoplasia type 11: Does the genetic defect determine timing of cerebellar pathology?. European Journal of Medical Genetics, 2020, 63, 103938.	0.7	4
154	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
155	<scp>CCDC82</scp> frameshift mutation associated with intellectual disability, spastic paraparesis, and dysmorphic features. Clinical Genetics, 2022, 102, 80-81.	1.0	4
156	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
157	LINS1-associated neurodevelopmental disorder. Neurology: Genetics, 2020, 6, e500.	0.9	3
158	Novel Biallelic <scp>CTSD</scp> Gene Variants Cause Lateâ€Onset Ataxia and Retinitis Pigmentosa. Movement Disorders, 2020, 35, 1280-1282.	2.2	3
159	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
160	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
161	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
162	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

#	Article	IF	Citations
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