Dominique Bonneau

List of Publications by Year
in descending order

186	13,372	31976 53	25787 108
papers	citations	h-index	g-index

196	196	196	17932
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The putative forkhead transcription factor FOXL2 is mutated in blepharophimosis/ptosis/epicanthus inversus syndrome. Nature Genetics, 2001, 27, 159-166.	21.4	886
2	Germline KRAS and BRAF mutations in cardio-facio-cutaneous syndrome. Nature Genetics, 2006, 38, 294-296.	21.4	517
3	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: A Gradient of Severity in Cognitive Impairments. PLoS Genetics, 2014, 10, e1004580.	3.5	501
4	OPA1 mutations induce mitochondrial DNA instability and optic atrophy 'plus' phenotypes. Brain, 2008, 131, 338-351.	7.6	454
5	Mutations in TUBG1, DYNC1H1, KIF5C and KIF2A cause malformations of cortical development and microcephaly. Nature Genetics, 2013, 45, 639-647.	21.4	399
6	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	27.8	394
7	Genetic and Functional Analyses of SHANK2 Mutations Suggest a Multiple Hit Model of Autism Spectrum Disorders. PLoS Genetics, 2012, 8, e1002521.	3.5	358
8	Xâ€“linked spastic paraplegia and Pelizaeusâ€“Merzbacher disease are allelic disorders at the proteolipid protein locus. Nature Genetics, 1994, 6, 257-262.	21.4	353
9	Mutations of the human PTEN gene. Human Mutation, 2000, 16, 109-122.	2.5	304
10	Mutations ofARX are associated with striking pleiotropy and consistent genotype-phenotype correlation. Human Mutation, 2004, 23, 147-159.	2.5	293
11	Mutation of ALMS1, a large gene with a tandem repeat encoding 47 amino acids, causes AlstrÃ¶m syndrome. Nature Genetics, 2002, 31, 79-83.	21.4	291
12	High cumulative risks of cancer in patients withPTENhamartoma tumour syndrome. Journal of Medical Genetics, 2013, 50, 255-263.	3.2	290
13	Mutation in the iron responsive element of the L ferritin mRNA in a family with dominant hyperferritinaemia and cataract. Nature Genetics, 1995, 11, 444-446.	21.4	266
14	BBS10 encodes a vertebrate-specific chaperonin-like protein and is a major BBS locus. Nature Genetics, 2006, 38, 521-524.	21.4	259
15	Identification of genetic variants associated with Huntington's disease progression: a genome-wide association study. Lancet Neurology, The, 2017, 16, 701-711.	10.2	248
16	Efficient strategy for the molecular diagnosis of intellectual disability using targeted high-throughput sequencing. Journal of Medical Genetics, 2014, 51, 724-736.	3.2	229
17	A Gene Predisposing to Familial Thyroid Tumors with Cell Oxyphilia Maps to Chromosome 19p13.2. American Journal of Human Genetics, 1998, 63, 1743-1748.	6.2	221
18	Identification of a Novel BBS Gene (BBS12) Highlights the Major Role of a Vertebrate-Specific Branch of Chaperonin-Related Proteins in Bardet-Biedl Syndrome. American Journal of Human Genetics, 2007, 80, 1-11.	6.2	219

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19	Dominant optic atrophy. Orphanet Journal of Rare Diseases, 2012, 7, 46.	2.7	213
20	Mitochondrial dynamics and disease, OPA1. Biochimica Et Biophysica Acta - Molecular Cell Research, 2006, 1763, 500-509.	4.1	195
21	OPA1 R445H mutation in optic atrophy associated with sensorineural deafness. Annals of Neurology, 2005, 58, 958-963.	5.3	155
22	Cobblestone lissencephaly: neuropathological subtypes and correlations with genes of dystroglycanopathies. Brain, 2012, 135, 469-482.	7.6	151
23	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
24	Molecular screening of 980 cases of suspected hereditary optic neuropathy with a report on 77 novelOPA1mutations. Human Mutation, 2009, 30, E692-E705.	2.5	140
25	Clinical and genetic heterogeneity in retinitis pigmentosa. Human Genetics, 1990, 85, 635-42.	3.8	136
26	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
27	Abnormal methylation pattern in constitutive and facultative (X inactive chromosome) heterochromatin of ICF patients. Human Molecular Genetics, 1994, 3, 2093-2102.	2.9	135
28	Thyroid pathologic findings in patients with Cowden disease. Annals of Diagnostic Pathology, 1999, 3, 331-340.	1.3	130
29	Effects of OPA1 mutations on mitochondrial morphology and apoptosis: Relevance to ADOA pathogenesis. Journal of Cellular Physiology, 2007, 211, 423-430.	4.1	128
30	Mitochondrial coupling defect in Charcot-Marie-Tooth type 2A disease. Annals of Neurology, 2007, 61, 315-323.	5.3	127
31	OPA1-associated disorders: Phenotypes and pathophysiology. International Journal of Biochemistry and Cell Biology, 2009, 41, 1855-1865.	2.8	122
32	Whole-Exome Sequencing Identifies Mutations in GPR179 Leading to Autosomal-Recessive Complete Congenital Stationary Night Blindness. American Journal of Human Genetics, 2012, 90, 321-330.	6.2	121
33	Hereditary optic neuropathies share a common mitochondrial coupling defect. Annals of Neurology, 2008, 63, 794-798.	5.3	112
34	Resveratrol Induces a Mitochondrial Complex I-dependent Increase in NADH Oxidation Responsible for Sirtuin Activation in Liver Cells. Journal of Biological Chemistry, 2013, 288, 36662-36675.	3.4	110
35	Loss-of-Function Mutations in WDR73 Are Responsible for Microcephaly and Steroid-Resistant Nephrotic Syndrome: Galloway-Mowat Syndrome. American Journal of Human Genetics, 2014, 95, 637-648.	6.2	108
36	Mutations in DNM1L, as in OPA1, result in dominant optic atrophy despite opposite effects on mitochondrial fusion and fission. Brain, 2017, 140, 2586-2596.	7.6	100

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37	Molecular and in silico analyses of the full-length isoform of usherin identify new pathogenic alleles in Usher type II patients. Human Mutation, 2007, 28, 781-789.	2.5	98
38	Mutations in CNTNAP1 and ADCY6 are responsible for severe arthrogryposis multiplex congenita with axoglial defects. Human Molecular Genetics, 2014, 23, 2279-2289.	2.9	98
39	Mutation screening of the EYA1, SIX1, and SIX5 genes in a large cohort of patients harboring branchio-oto-renal syndrome calls into question the pathogenic role of SIX5 mutations. Human Mutation, 2011, 32, 183-190.	2.5	93
40	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.	6.2	87
41	Spectrum of mutations in the renin-angiotensin system genes in autosomal recessive renal tubular dysgenesis. Human Mutation, 2012, 33, 316-326.	2.5	86
42	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	6.2	86
43	Hereditary spastic paraplegia-like disorder due to a mitochondrial ATP6 gene point mutation. Mitochondrion, 2011, 11, 70-75.	3.4	74
44	Mitochondrial complex I deficiency in GDAP1-related autosomal dominant Charcot-Marie-Tooth disease (CMT2K). Neurogenetics, 2009, 10, 145-150.	1.4	72
45	Spastic paraplegia due to SPAST mutations is modified by the underlying mutation and sex. Brain, 2018, 141, 3331-3342.	7.6	72
46	Testing for triallelism: analysis of six BBS genes in a Bardetâ€“Biedl syndrome family cohort. European Journal of Human Genetics, 2005, 13, 607-616.	2.8	69
47	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 101, 716-724.	6.2	66
48	Heterogeneity ofNSD1alterations in 116 patients with Sotos syndrome. Human Mutation, 2007, 28, 1098-1107.	2.5	62
49	Early-onset Behr syndrome due to compound heterozygous mutations in OPA1. Brain, 2014, 137, e301-e301.	7.6	62
50	New practical definitions for the diagnosis of autosomal recessive spastic ataxia of <scp>C</scp>harlevoixâ€“<scp>S</scp>aguenay. Annals of Neurology, 2015, 78, 871-886.	5.3	62
51	Nicotinamide Deficiency in Primary Open-Angle Glaucoma. , 2019, 60, 2509.		61
52	The association of autosomal dominant optic atrophy and moderate deafness may be due to the R445H mutation in the OPA1 gene. American Journal of Ophthalmology, 2003, 136, 1170-1171.	3.3	58
53	Adult-onset genetic leukoencephalopathies: A MRI pattern-based approach in a comprehensive study of 154 patients. Brain, 2015, 138, 284-292.	7.6	58
54	A gene for blepharophimosis-ptosis-epicanthus inversus syndrome maps to chromosome 3q23. Human Genetics, 1995, 96, 213-215.	3.8	57

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55	Fourteen novel OPA1 mutations in autosomal dominant optic atrophy including two de novo mutations in sporadic optic atrophy. Human Mutation, 2003, 21, 656-656.	2.5	57
56	A gene for Leber's congenital amaurosis maps to chromosome 17p. Human Molecular Genetics, 1995, 4, 1447-1452.	2.9	56
57	Pitfalls of homozygosity mapping: an extended consanguineous Bardetâ€“Biedl syndrome family with two mutant genes (BBS2, BBS10), three mutations, but no triallelism. European Journal of Human Genetics, 2006, 14, 1195-1203.	2.8	56
58	Quantifying the Effects of 16p11.2 Copy Number Variants on Brain Structure: A Multisite Genetic-First Study. Biological Psychiatry, 2018, 84, 253-264.	1.3	56
59	Enrichment of LOVD-USHbases with 152USH2AGenotypes Defines an Extensive Mutational Spectrum and Highlights Missense Hotspots. Human Mutation, 2014, 35, 1179-1186.	2.5	55
60	Recessive Mutations in RTN4IP1 Cause Isolated and Syndromic Optic Neuropathies. American Journal of Human Genetics, 2015, 97, 754-760.	6.2	54
61	Effect of Single and Multiple Courses of Prenatal Corticosteroids on 17-Hydroxyprogesterone Levels: Implication for Neonatal Screening of Congenital Adrenal Hyperplasia. Pediatric Research, 2004, 56, 701-705.	2.3	53
62	A Metabolomics Profiling of Glaucoma Points to Mitochondrial Dysfunction, Senescence, and Polyamines Deficiency. , 2018, 59, 4355.		51
63	Idebenone increases mitochondrial complex I activity in fibroblasts from LHON patients while producing contradictory effects on respiration. BMC Research Notes, 2011, 4, 557.	1.4	50
64	Schimke immunoosseous dysplasia: suggestions of genetic diversity. Human Mutation, 2007, 28, 273-283.	2.5	49
65	Ethambutol-induced optic neuropathy linked to OPA1 mutation and mitochondrial toxicity. Mitochondrion, 2010, 10, 115-124.	3.4	49
66	Metabolomic Profiling of Aqueous Humor in Glaucoma Points to Taurine and Spermine Deficiency: Findings from the Eye-D Study. Journal of Proteome Research, 2019, 18, 1307-1315.	3.7	49
67	A large-scale mutation search reveals genetic heterogeneity in 3M syndrome. European Journal of Human Genetics, 2009, 17, 395-400.	2.8	48
68	Bioinformatics Tools and Databases to Assess the Pathogenicity of Mitochondrial DNA Variants in the Field of Next Generation Sequencing. Frontiers in Genetics, 2018, 9, 632.	2.3	48
69	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. Genetics in Medicine, 2019, 21, 2723-2733.	2.4	48
70	The metabolomic signature of Leberâ€™s hereditary optic neuropathy reveals endoplasmic reticulum stress. Brain, 2016, 139, 2864-2876.	7.6	45
71	OPA1-related disorders: Diversity of clinical expression, modes of inheritance and pathophysiology. Neurobiology of Disease, 2016, 90, 20-26.	4.4	45
72	Relative Frequencies of Inherited Retinal Dystrophies and Optic Neuropathies in Southern France: Assessment of 21-year Data Management. Ophthalmic Epidemiology, 2013, 20, 13-25.	1.7	44

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73	A Nontargeted UHPLC-HRMS Metabolomics Pipeline for Metabolite Identification: Application to Cardiac Remote Ischemic Preconditioning. Analytical Chemistry, 2017, 89, 2138-2146.	6.5	43
74	Genetic Heterogeneity of Usher Syndrome Type 1 in French Families. Genomics, 1994, 21, 138-143.	2.9	42
75	USH1A: Chronicle of a Slow Death. American Journal of Human Genetics, 2006, 78, 357-359.	6.2	41
76	The addition of ketone bodies alleviates mitochondrial dysfunction by restoring complex I assembly in a MELAS cellular model. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 284-291.	3.8	41
77	De Novo Truncating Mutations in the Kinetochore-Microtubules Attachment GeneCHAMP1Cause Syndromic Intellectual Disability. Human Mutation, 2016, 37, 354-358.	2.5	40
78	Molecular diagnosis reveals genetic heterogeneity for the overlapping MKKS and BBS phenotypes. European Journal of Medical Genetics, 2011, 54, 157-160.	1.3	39
79	Improved Locus-Specific Database forOPA1Mutations Allows Inclusion of Advanced Clinical Data. Human Mutation, 2015, 36, 20-25.	2.5	39
80	A New Case of PCSK1 Pathogenic Variant With Congenital Proprotein Convertase 1/3 Deficiency and Literature Review. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 985-993.	3.6	39
81	Metabolically induced heteroplasmy shifting and l-arginine treatment reduce the energetic defect in a neuronal-like model of MELAS. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1019-1029.	3.8	38
82	CLUH couples mitochondrial distribution to the energetic and metabolic status. Journal of Cell Science, 2017, 130, 1940-1951.	2.0	38
83	eKLIPse: a sensitive tool for the detection and quantification of mitochondrial DNA deletions from next-generation sequencing data. Genetics in Medicine, 2019, 21, 1407-1416.	2.4	38
84	Acute and late-onset optic atrophy due to a novel OPA1 mutation leading to a mitochondrial coupling defect. Molecular Vision, 2009, 15, 598-608.	1.1	37
85	Reversible optic neuropathy with OPA1 exon 5b mutation. Annals of Neurology, 2008, 63, 667-671.	5.3	36
86	Heterozygosity for a Single Mutation in the ABCC6 Gene May Closely Mimic PXE. Archives of Dermatology, 2008, 144, 301-6.	1.4	36
87	Bioenergetic defect associated with mK _{ATP} channel opening in a mouse model carrying a mitofusin 2 mutation. FASEB Journal, 2011, 25, 1618-1627.	0.5	36
88	Sensorineural hearing loss in OPA1-linked disorders. Brain, 2013, 136, e236-e236.	7.6	36
89	SLC24A5 Mutations Are Associated with Non-Syndromic Oculocutaneous Albinism. Journal of Investigative Dermatology, 2014, 134, 568-571.	0.7	36
90	Haploinsufficiency of the E3 ubiquitin-protein ligase gene TRIP12 causes intellectual disability with or without autism spectrum disorders, speech delay, and dysmorphic features. Human Genetics, 2017, 136, 377-386.	3.8	36

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91	Combination of WAGR and Potockiâ€“Shaffer contiguous deletion syndromes in a patient with an 11p11.2â€“p14 deletion. European Journal of Human Genetics, 2005, 13, 409-413.	2.8	35
92	Adenine nucleotide translocase is involved in a mitochondrial coupling defect in MFN2-related Charcotâ€“Marieâ€“Tooth type 2A disease. Neurogenetics, 2010, 11, 127-133.	1.4	34
93	Standardized mitochondrial analysis gives new insights into mitochondrial dynamics and OPA1 function. International Journal of Biochemistry and Cell Biology, 2012, 44, 980-988.	2.8	34
94	Germline and Mosaic Variants in PRKACA and PRKACB Cause a Multiple Congenital Malformation Syndrome. American Journal of Human Genetics, 2020, 107, 977-988.	6.2	33
95	Allelic loss on chromosomes 2q21 and 19p 13.2 in oxyphilic thyroid tumors. International Journal of Cancer, 2004, 111, 463-467.	5.1	32
96	Genetically determined optic neuropathies. Current Opinion in Neurology, 2010, 23, 24-28.	3.6	31
97	Search for the best indicators for the presence of a VPS13B gene mutation and confirmation of diagnostic criteria in a series of 34 patients genotyped for suspected Cohen syndrome. Journal of Medical Genetics, 2010, 47, 549-553.	3.2	31
98	A randomized, double-blind, placebo-controlled trial evaluating cysteamine in Huntington's disease. Movement Disorders, 2017, 32, 932-936.	3.9	31
99	Mutations in the m-AAA proteases AFG3L2 and SPG7 are causing isolated dominant optic atrophy. Neurology: Genetics, 2020, 6, e428.	1.9	31
100	Autophagy controls the pathogenicity of <scp>OPA</scp>1 mutations in dominant optic atrophy. Journal of Cellular and Molecular Medicine, 2017, 21, 2284-2297.	3.6	30
101	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	6.2	30
102	Colorectal Adenomatous Polyposis Associated with MYH Mutations: Genotype and Phenotype Characteristics. Diseases of the Colon and Rectum, 2007, 50, 1612-1617.	1.3	29
103	Identification of gene copy number variations in patients with mental retardation using array-CGH: Novel syndromes in a large French series. European Journal of Medical Genetics, 2010, 53, 66-75.	1.3	29
104	Thrombocytopenia-absent radius (TAR) syndrome: A clinical genetic series of 14 further cases. Impact of the associated 1q21.1 deletion on the genetic counselling. European Journal of Medical Genetics, 2011, 54, e471-e477.	1.3	29
105	Assembly defects induce oxidative stress in inherited mitochondrial complex I deficiency. International Journal of Biochemistry and Cell Biology, 2015, 65, 91-103.	2.8	29
106	A snapshot of some pLI score pitfalls. Human Mutation, 2019, 40, 839-841.	2.5	29
107	Gain-of-Function Mutation in Filamin A Potentiates Platelet Integrin Î± _{IIb} Î² ₃ Activation. Arteriosclerosis, Thrombosis, and Vascular Biology, 2017, 37, 1087-1097.	2.4	28
108	BBS8 is rarely mutated in a cohort of 128 Bardetâ€“Biedl syndrome families. Journal of Human Genetics, 2006, 51, 81-84.	2.3	27

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109	Theory of mind and empathy in preclinical and clinical Huntingtonâ€™s disease. Social Cognitive and Affective Neuroscience, 2016, 11, 89-99.	3.0	27
110	A recessive ataxia diagnosis algorithm for the next generation sequencing era. Annals of Neurology, 2017, 82, 892-899.	5.3	27
111	Reduced elastogenesis: a clue to the arteriosclerosis and emphysematous changes in Schimke immuno-osseous dysplasia?. Orphanet Journal of Rare Diseases, 2012, 7, 70.	2.7	26
112	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. JAMA Neurology, 2018, 75, 105.	9.0	26
113	Missense variants in DPYSL5 cause a neurodevelopmental disorder with corpus callosum agenesis and cerebellar abnormalities. American Journal of Human Genetics, 2021, 108, 951-961.	6.2	26
114	A Data Mining Metabolomics Exploration of Glaucoma. Metabolites, 2020, 10, 49.	2.9	25
115	Where are the missing gene defects in inherited retinal disorders? Intronic and synonymous variants contribute at least to 4% of CACNA1F â€mediated inherited retinal disorders. Human Mutation, 2019, 40, 765-787.	2.5	24
116	Mild form of oculocutaneous albinism type 1: phenotypic analysis of compound heterozygous patients with the R402Q variant of the TYR gene. British Journal of Ophthalmology, 2019, 103, 1239-1247.	3.9	24
117	Phenotypic spectrum associated with SPECC1L pathogenic variants: new families and critical review of the nosology of Teebi, Opitz GBBB, and Baraitser-Winter syndromes. European Journal of Medical Genetics, 2019, 62, 103588.	1.3	24
118	De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. Genetics in Medicine, 2020, 22, 538-546.	2.4	24
119	Molecular cytogenetic characterization of terminal 14q32 deletions in two children with an abnormal phenotype and corpus callosum hypoplasia. European Journal of Human Genetics, 2008, 16, 680-687.	2.8	23
120	Microchimerism from a dizygotic twin in juvenile ulcerative lichen planus. Lancet, The, 2002, 359, 1861-1862.	13.7	22
121	Schimke immunoosseous dysplasia: defining skeletal features. European Journal of Pediatrics, 2010, 169, 801-811.	2.7	22
122	Targeted Metabolomics Reveals Early Dominant Optic Atrophy Signature in Optic Nerves of Opa1^{delTTAG/+} Mice. , 2017, 58, 812.		22
123	Oxidative stress contributes differentially to the pathophysiology of Charcot-Marie-Tooth disease type 2K. Experimental Neurology, 2020, 323, 113069.	4.1	22
124	A Plasma Metabolomic Signature Involving Purine Metabolism in Human Optic Atrophy 1 (OPA1)-Related Disorders. , 2018, 59, 185.		21
125	Retinal angioma in a patient with Cowden disease. American Journal of Ophthalmology, 2003, 135, 400-402.	3.3	20
126	The Metabolomic Bioenergetic Signature of Opa1-Disrupted Mouse Embryonic Fibroblasts Highlights Aspartate Deficiency. Scientific Reports, 2018, 8, 11528.	3.3	20

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127	Five novel missense mutations of the rhodopsin gene in autosomal dominant retinitis pigmentosa. Human Molecular Genetics, 1994, 3, 1433-1434.	2.9	19
128	A Plasma Metabolomic Signature of the Exfoliation Syndrome Involves Amino Acids, Acylcarnitines, and Polyamines. , 2018, 59, 1025.		18
129	Refined genetic mapping of autosomal recessive chronic distal spinal muscular atrophy to chromosome 11q13.3 and evidence of linkage disequilibrium in European families. European Journal of Human Genetics, 2004, 12, 483-488.	2.8	17
130	Metabolomics hallmarks OPA1 variants correlating with their in vitro phenotype and predicting clinical severity. Human Molecular Genetics, 2020, 29, 1319-1329.	2.9	17
131	A phase II, openâ€label evaluation of cysteamine tolerability in patients with Huntington's disease. Movement Disorders, 2015, 30, 288-289.	3.9	16
132	DominantACO2mutations are a frequent cause of isolated optic atrophy. Brain Communications, 2021, 3, fcab063.	3.3	16
133	Prenatal diagnosis of CHARGE syndrome by identification of a novel CHD7 mutation in a previously unaffected family. Prenatal Diagnosis, 2012, 32, 692-694.	2.3	15
134	The accumulation of assembly intermediates of the mitochondrial complex I matrix arm is reduced by limiting glucose uptake in a neuronal-like model of MELAS syndrome. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 1596-1608.	3.8	15
135	Phenotypic spectrum of TGFB3 diseaseâ€causing variants in a Dutchâ€French cohort and first report of a homozygous patient. Clinical Genetics, 2020, 97, 723-730.	2.0	15
136	Confirmation that variants in TTI2 are responsible for autosomal recessive intellectual disability. Clinical Genetics, 2019, 96, 354-358.	2.0	14
137	A plasma metabolomic signature of Leber hereditary optic neuropathy showing taurine and nicotinamide deficiencies. Human Molecular Genetics, 2021, 30, 21-29.	2.9	14
138	Bi-allelic variants in IPO8 cause a connective tissue disorder associated with cardiovascular defects, skeletal abnormalities, and immune dysregulation. American Journal of Human Genetics, 2021, 108, 1126-1137.	6.2	14
139	Warburg-like effect is a hallmark of complex I assembly defects. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2019, 1865, 2475-2489.	3.8	13
140	Dominant mutations in MIEF1 affect mitochondrial dynamics and cause a singular late onset optic neuropathy. Molecular Neurodegeneration, 2021, 16, 12.	10.8	13
141	Paradoxical effect of inhaled nitric oxide in a newborn with pulmonary hypertension. Lancet, The, 1993, 342, 364-365.	13.7	12
142	Next generation sequencing in family with MNGIE syndrome associated to optic atrophy: Novel homozygous POLG mutation in the C-terminal sub-domain leading to mtDNA depletion. Clinica Chimica Acta, 2019, 488, 104-110.	1.1	12
143	Neuropsychological and Psychiatric Features of Children and Adolescents Affected With Mitochondrial Diseases: A Systematic Review. Frontiers in Psychiatry, 2020, 11, 747.	2.6	12
144	Use of Next-Generation Sequencing for the Molecular Diagnosis of 1,102 Patients With a Autosomal Optic Neuropathy. Frontiers in Neurology, 2021, 12, 602979.	2.4	12

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145	Dissociation between decision-making under risk and decision-making under ambiguity in premanifest and manifest Huntington's disease. Neuropsychologia, 2017, 103, 87-95.	1.6	11
146	Metabolomics reveals highly regional specificity of cerebral sexual dimorphism in mice. Progress in Neurobiology, 2020, 184, 101698.	5.7	11
147	Evaluation of the colorectal cancer risk conferred by rareUNC5Calleles. World Journal of Gastroenterology, 2014, 20, 204.	3.3	11
148	Arithmetic word-problem-solving in Huntingtonâ€™s disease. Brain and Cognition, 2005, 57, 1-3.	1.8	10
149	Dermatologic Features of Smithâ€“Magenis Syndrome. Pediatric Dermatology, 2015, 32, 337-341.	0.9	10
150	Increased mitochondrial fusion in a autosomal recessive CMT2A family with mitochondrial GTPase mitofusin 2 mutations. Journal of the Peripheral Nervous System, 2016, 21, 365-369.	3.1	10
151	Refsumâ€™s disease may mimic familial Guillain Barre syndrome. Neuromuscular Disorders, 2006, 16, 805-808.	0.6	9
152	The neural substrates of script knowledge deficits as revealed by a PET study in Huntington's disease. Neuropsychologia, 2011, 49, 2673-2684.	1.6	9
153	Developmental trajectories of neuroanatomical alterations associated with the 16p11.2 Copy Number Variations. NeuroImage, 2019, 203, 116155.	4.2	9
154	Lipidomics Reveals Triacylglycerol Accumulation Due to Impaired Fatty Acid Flux in Opa1-Disrupted Fibroblasts. Journal of Proteome Research, 2019, 18, 2779-2790.	3.7	9
155	Childhood Dermatosis due to Microchimerism. Dermatology, 2005, 211, 388-389.	2.1	8
156	Expanding the phenotypic spectrum associated with OPHN1 mutations: Report of 17 individuals with intellectual disability but no cerebellar hypoplasia. European Journal of Medical Genetics, 2018, 61, 442-450.	1.3	8
157	Hypopituitarism in Patients with Blepharophimosis and <i>FOXL2</i> Mutations. Hormone Research in Paediatrics, 2020, 93, 30-39.	1.8	8
158	A new mutational hotspot in the SKI gene in the context of MFS/TAA molecular diagnosis. Human Genetics, 2020, 139, 461-472.	3.8	8
159	Run of homozygosity analysis reveals a novel nonsense variant of the CNGB1 gene involved in retinitis pigmentosa 45. Ophthalmic Genetics, 2016, 37, 357-359.	1.2	7
160	Novel NDUFS4 gene mutation in an atypical late-onset mitochondrial form of multifocal dystonia. Neurology: Genetics, 2017, 3, e205.	1.9	7
161	Clinical and molecular description of 19 patients with GATAD2B-Associated Neurodevelopmental Disorder (GAND). European Journal of Medical Genetics, 2020, 63, 104004.	1.3	7
162	Mobile App for Parental Empowerment for Caregivers of Children With Autism Spectrum Disorders: Prospective Open Trial. JMIR Mental Health, 2021, 8, e27803.	3.3	7

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163	Benzathine as a cause for a false-positive test result for amphetamines. Journal of Pediatrics, 1995, 127, 669-670.	1.8	6
164	Pitfalls in Clinical Diagnosis of Female Carriers of X-linked Hypohidrotic Ectodermal Dysplasia. Archives of Dermatology, 2002, 138, 1256-1258.	1.4	6
165	Intellectual disability associated with retinal dystrophy in the Xp11.3 deletion syndrome: ZNF674 on trial. Guilty or innocent?. European Journal of Human Genetics, 2012, 20, 352-356.	2.8	5
166	Rationale and protocol for using a smartphone application to study autism spectrum disorders: SMARTAUTISM. BMJ Open, 2016, 6, e012135.	1.9	5
167	Reply: The expanding neurological phenotype of DNM1L-related disorders. Brain, 2018, 141, e29-e29.	7.6	5
168	Dysfunctional T Cell Mitochondria Lead to Premature Aging. Trends in Molecular Medicine, 2020, 26, 799-800.	6.7	5
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