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	Glutamate-Induced Deregulation of Krebs Cycle in Mitochondrial Encephalopathy Lactic Acidosis Syndrome Stroke-Like Episodes (MELAS) Syndrome Is Alleviated by Ketone Body Exposure. Biomedicines, 2022, 10, 1665.	3.2	4
2	Next-Generation Sequencing Identifies Novel PMPCA Variants in Patients with Late-Onset Dominant Optic Atrophy. Genes, 2022, 13, 1202.	2.4	0
3	Prenatal diagnosis of Desbuquois dysplasia type 1 by whole exome sequencing before the occurrence of specific ultrasound signs. Journal of Maternal-Fetal and Neonatal Medicine, 2021, 34, 2217-2220.	1.5	5
4	A plasma metabolomic signature of Leber hereditary optic neuropathy showing taurine and nicotinamide deficiencies. Human Molecular Genetics, 2021, 30, 21-29.	2.9	14
5	Dominant mutations in MIEF1 affect mitochondrial dynamics and cause a singular late onset optic neuropathy. Molecular Neurodegeneration, 2021, 16, 12.	10.8	13
6	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
7	Metabolomic Sexual Dimorphism of the Mouse Brain is Predominantly Abolished by Gonadectomy with a Higher Impact on Females. Journal of Proteome Research, 2021, 20, 2772-2779.	3.7	1
8	Are Your Mitochondria Ready for a Space Odyssey?. Trends in Endocrinology and Metabolism, 2021, 32, 193-195.	7.1	1
9	DominantACO2mutations are a frequent cause of isolated optic atrophy. Brain Communications, 2021, 3, fcab063.	3.3	16
10	Improved detection of mitochondrial DNA instability in mitochondrial genome maintenance disorders. Genetics in Medicine, 2021, 23, 1769-1778.	2.4	4
11	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
12	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
13	Heterozygous <scp>HMGB1</scp> lossâ€ofâ€function variants are associated with developmental delay and microcephaly. Clinical Genetics, 2021, 100, 386-395.	2.0	3
14	ZNF668 deficiency causes a recognizable disorder of DNA damage repair. Human Genetics, 2021, 140, 1395-1401.	3.8	1
15	Psychiatric Symptoms of Children and Adolescents With Mitochondrial Disorders: A Descriptive Case Series. Frontiers in Psychiatry, 2021, 12, 685532.	2.6	1
16	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
17	Congenital hypothyroidism and hearing loss without inner ear malformation: Think <scp>TPO</scp>. Clinical Genetics, 2021, 99, 604-606.	2.0	1
18	Metabolomics reveals highly regional specificity of cerebral sexual dimorphism in mice. Progress in Neurobiology, 2020, 184, 101698.	5.7	11

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19	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
20	Oxidative stress contributes differentially to the pathophysiology of Charcot-Marie-Tooth disease type 2K. Experimental Neurology, 2020, 323, 113069.	4.1	22
21	Specific cognitive theory of mind and behavioral dysfunctions in early manifest Huntington disease: a case report. Neurocase, 2020, 26, 36-41.	0.6	1
22	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
23	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
24	Dysfunctional T Cell Mitochondria Lead to Premature Aging. Trends in Molecular Medicine, 2020, 26, 799-800.	6.7	5
25	Clinical and molecular description of 19 patients with GATAD2B-Associated Neurodevelopmental Disorder (GAND). European Journal of Medical Genetics, 2020, 63, 104004.	1.3	7
26	Neuropsychological and Psychiatric Features of Children and Adolescents Affected With Mitochondrial Diseases: A Systematic Review. Frontiers in Psychiatry, 2020, 11, 747.	2.6	12
27	De novo missense variants in LMBRD2 are associated with developmental and motor delays, brain structure abnormalities and dysmorphic features. Journal of Medical Genetics, 2020, 58, jmedgenet-2020-107137.	3.2	3
28	Hypopituitarism in Patients with Blepharophimosis and <i>FOXL2</i> Mutations. Hormone Research in Paediatrics, 2020, 93, 30-39.	1.8	8
29	Metabolomics hallmarks OPA1 variants correlating with their in vitro phenotype and predicting clinical severity. Human Molecular Genetics, 2020, 29, 1319-1329.	2.9	17
30	A Data Mining Metabolomics Exploration of Glaucoma. Metabolites, 2020, 10, 49.	2.9	25
31	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
32	Mutations in aARS genes revealed by targeted next-generation sequencing in patients with mitochondrial diseases. Molecular Biology Reports, 2020, 47, 3779-3787.	2.3	5
33	Mutations in the m-AAA proteases AFG3L2 and SPG7 are causing isolated dominant optic atrophy. Neurology: Genetics, 2020, 6, e428.	1.9	31
34	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
35	Confirmation that variants in TTI2 are responsible for autosomal recessive intellectual disability. Clinical Genetics, 2019, 96, 354-358.	2.0	14
36	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

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37	Developmental trajectories of neuroanatomical alterations associated with the 16p11.2 Copy Number Variations. NeuroImage, 2019, 203, 116155.	4.2	9
38	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
39	Nicotinamide Deficiency in Primary Open-Angle Glaucoma. , 2019, 60, 2509.		61
40	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
41	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
42	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
43	A snapshot of some pLI score pitfalls. Human Mutation, 2019, 40, 839-841.	2.5	29
44	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
45	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
46	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
47	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
48	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
49	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
50	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
51	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
52	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
53	Reply: The expanding neurological phenotype of DNM1L-related disorders. Brain, 2018, 141, e29-e29.	7.6	5
54	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. JAMA Neurology, 2018, 75, 105.	9.0	26

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55	Spastic paraplegia due to SPAST mutations is modified by the underlying mutation and sex. Brain, 2018, 141, 3331-3342.	7.6	72
56	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
57	A Plasma Metabolomic Signature Involving Purine Metabolism in Human Optic Atrophy 1 (OPA1)-Related Disorders. , 2018, 59, 185.		21
58	A Metabolomics Profiling of Glaucoma Points to Mitochondrial Dysfunction, Senescence, and Polyamines Deficiency. , 2018, 59, 4355.		51
59	A Plasma Metabolomic Signature of the Exfoliation Syndrome Involves Amino Acids, Acylcarnitines, and Polyamines. , 2018, 59, 1025.		18
60	The Metabolomic Bioenergetic Signature of Opa1-Disrupted Mouse Embryonic Fibroblasts Highlights Aspartate Deficiency. Scientific Reports, 2018, 8, 11528.	3.3	20
61	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
62	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
63	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
64	CLUH couples mitochondrial distribution to the energetic and metabolic status. Journal of Cell Science, 2017, 130, 1940-1951.	2.0	38
65	A randomized, double-blind, placebo-controlled trial evaluating cysteamine in Huntington's disease. Movement Disorders, 2017, 32, 932-936.	3.9	31
66	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
67	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
68	A Nontargeted UHPLC-HRMS Metabolomics Pipeline for Metabolite Identification: Application to Cardiac Remote Ischemic Preconditioning. Analytical Chemistry, 2017, 89, 2138-2146.	6.5	43
69	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
70	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
71	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
72	A recessive ataxia diagnosis algorithm for the next generation sequencing era. Annals of Neurology, 2017, 82, 892-899.	5.3	27

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73	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
74	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
75	Novel NDUFS4 gene mutation in an atypical late-onset mitochondrial form of multifocal dystonia. Neurology: Genetics, 2017, 3, e205.	1.9	7
76	Targeted Metabolomics Reveals Early Dominant Optic Atrophy Signature in Optic Nerves of Opa1^{delTTAG/+} Mice. , 2017, 58, 812.		22
77	Rationale and protocol for using a smartphone application to study autism spectrum disorders: SMARTAUTISM. BMJ Open, 2016, 6, e012135.	1.9	5
78	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
79	The metabolomic signature of Leberâ€™s hereditary optic neuropathy reveals endoplasmic reticulum stress. Brain, 2016, 139, 2864-2876.	7.6	45
80	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
81	De Novo Truncating Mutations in the Kinetochore-Microtubules Attachment GeneCHAMP1Cause Syndromic Intellectual Disability. Human Mutation, 2016, 37, 354-358.	2.5	40
82	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
83	OPA1-related disorders: Diversity of clinical expression, modes of inheritance and pathophysiology. Neurobiology of Disease, 2016, 90, 20-26.	4.4	45
84	Theory of mind and empathy in preclinical and clinical Huntingtonâ€™s disease. Social Cognitive and Affective Neuroscience, 2016, 11, 89-99.	3.0	27
85	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
86	Improved Locus-Specific Database forOPA1Mutations Allows Inclusion of Advanced Clinical Data. Human Mutation, 2015, 36, 20-25.	2.5	39
87	A phase II, openâ€label evaluation of cysteamine tolerability in patients with Huntington's disease. Movement Disorders, 2015, 30, 288-289.	3.9	16
88	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
89	Adult-onset genetic leukoencephalopathies: A MRI pattern-based approach in a comprehensive study of 154 patients. Brain, 2015, 138, 284-292.	7.6	58
90	Recessive Mutations in RTN4IP1 Cause Isolated and Syndromic Optic Neuropathies. American Journal of Human Genetics, 2015, 97, 754-760.	6.2	54

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91	Dermatologic Features of Smithâ€“Magenis Syndrome. Pediatric Dermatology, 2015, 32, 337-341.	0.9	10
92	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
93	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: A Gradient of Severity in Cognitive Impairments. PLoS Genetics, 2014, 10, e1004580.	3.5	501
94	Early-onset Behr syndrome due to compound heterozygous mutations in OPA1. Brain, 2014, 137, e301-e301.	7.6	62
95	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
96	Enrichment of LOVD-USHbases with 152USH2AGenotypes Defines an Extensive Mutational Spectrum and Highlights Missense Hotspots. Human Mutation, 2014, 35, 1179-1186.	2.5	55
97	Extensive Mongolian spots in 4p16.3 deletion (Wolfâ€“Hirschhorn syndrome). Clinical Dysmorphology, 2014, 23, 109-110.	0.3	2
98	SLC24A5 Mutations Are Associated with Non-Syndromic Oculocutaneous Albinism. Journal of Investigative Dermatology, 2014, 134, 568-571.	0.7	36
99	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
100	Evaluation of the colorectal cancer risk conferred by rareUNC5Calleles. World Journal of Gastroenterology, 2014, 20, 204.	3.3	11
101	Mutations in TUBG1, DYNC1H1, KIF5C and KIF2A cause malformations of cortical development and microcephaly. Nature Genetics, 2013, 45, 639-647.	21.4	399
102	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
103	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
104	High cumulative risks of cancer in patients withPTENhamartoma tumour syndrome. Journal of Medical Genetics, 2013, 50, 255-263.	3.2	290
105	Sensorineural hearing loss in OPA1-linked disorders. Brain, 2013, 136, e236-e236.	7.6	36
106	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
107	Genetic and Functional Analyses of SHANK2 Mutations Suggest a Multiple Hit Model of Autism Spectrum Disorders. PLoS Genetics, 2012, 8, e1002521.	3.5	358
108	Cobblestone lissencephaly: neuropathological subtypes and correlations with genes of dystroglycanopathies. Brain, 2012, 135, 469-482.	7.6	151

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109	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
110	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
111	Dominant optic atrophy. Orphanet Journal of Rare Diseases, 2012, 7, 46.	2.7	213
112	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
113	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
114	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
115	Spectrum of mutations in the renin-angiotensin system genes in autosomal recessive renal tubular dysgenesis. Human Mutation, 2012, 33, 316-326.	2.5	86
116	Ade novogermlineMLH1mutation in a Lynch syndrome patient with discordant immunohistochemical and molecular biology test results. World Journal of Gastroenterology, 2012, 18, 5635.	3.3	5
117	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	27.8	394
118	Molecular diagnosis reveals genetic heterogeneity for the overlapping MKKS and BBS phenotypes. European Journal of Medical Genetics, 2011, 54, 157-160.	1.3	39
119	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
120	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
121	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
122	Hereditary spastic paraplegia-like disorder due to a mitochondrial ATP6 gene point mutation. Mitochondrion, 2011, 11, 70-75.	3.4	74
123	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
124	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
125	Genetically determined optic neuropathies. Current Opinion in Neurology, 2010, 23, 24-28.	3.6	31
126	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

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127	Schimke immunoosseous dysplasia: defining skeletal features. European Journal of Pediatrics, 2010, 169, 801-811.	2.7	22
128	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
129	Ethambutol-induced optic neuropathy linked to OPA1 mutation and mitochondrial toxicity. Mitochondrion, 2010, 10, 115-124.	3.4	49
130	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
131	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
132	Mitochondrial complex I deficiency in GDAP1-related autosomal dominant Charcot-Marie-Tooth disease (CMT2K). Neurogenetics, 2009, 10, 145-150.	1.4	72
133	A large-scale mutation search reveals genetic heterogeneity in 3M syndrome. European Journal of Human Genetics, 2009, 17, 395-400.	2.8	48
134	OPA1-associated disorders: Phenotypes and pathophysiology. International Journal of Biochemistry and Cell Biology, 2009, 41, 1855-1865.	2.8	122
135	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
136	Reversible optic neuropathy with OPA1 exon 5b mutation. Annals of Neurology, 2008, 63, 667-671.	5.3	36
137	Hereditary optic neuropathies share a common mitochondrial coupling defect. Annals of Neurology, 2008, 63, 794-798.	5.3	112
138	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
139	OPA1 mutations induce mitochondrial DNA instability and optic atrophy 'plus' phenotypes. Brain, 2008, 131, 338-351.	7.6	454
140	Heterozygosity for a Single Mutation in the ABCC6 Gene May Closely Mimic PXE. Archives of Dermatology, 2008, 144, 301-6.	1.4	36
141	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
142	Mitochondrial coupling defect in Charcot-Marie-Tooth type 2A disease. Annals of Neurology, 2007, 61, 315-323.	5.3	127
143	Schimke immunoosseous dysplasia: suggestions of genetic diversity. Human Mutation, 2007, 28, 273-283.	2.5	49
144	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

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145	Heterogeneity ofNSD1alterations in 116 patients with Sotos syndrome. Human Mutation, 2007, 28, 1098-1107.	2.5	62
146	Effects of OPA1 mutations on mitochondrial morphology and apoptosis: Relevance to ADOA pathogenesis. Journal of Cellular Physiology, 2007, 211, 423-430.	4.1	128
147	Colorectal Adenomatous Polyposis Associated with MYH Mutations: Genotype and Phenotype Characteristics. Diseases of the Colon and Rectum, 2007, 50, 1612-1617.	1.3	29
148	USH1A: Chronicle of a Slow Death. American Journal of Human Genetics, 2006, 78, 357-359.	6.2	41
149	Refsumâ€™s disease may mimic familial Guillain Barre syndrome. Neuromuscular Disorders, 2006, 16, 805-808.	0.6	9
150	Germline KRAS and BRAF mutations in cardio-facio-cutaneous syndrome. Nature Genetics, 2006, 38, 294-296.	21.4	517
151	BBS10 encodes a vertebrate-specific chaperonin-like protein and is a major BBS locus. Nature Genetics, 2006, 38, 521-524.	21.4	259
152	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
153	BBS8 is rarely mutated in a cohort of 128 Bardetâ€“Biedl syndrome families. Journal of Human Genetics, 2006, 51, 81-84.	2.3	27
154	Mitochondrial dynamics and disease, OPA1. Biochimica Et Biophysica Acta - Molecular Cell Research, 2006, 1763, 500-509.	4.1	195
155	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
156	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
157	OPA1 R445H mutation in optic atrophy associated with sensorineural deafness. Annals of Neurology, 2005, 58, 958-963.	5.3	155
158	Childhood Dermatosis due to Microchimerism. Dermatology, 2005, 211, 388-389.	2.1	8
159	Arithmetic word-problem-solving in Huntingtonâ€™s disease. Brain and Cognition, 2005, 57, 1-3.	1.8	10
160	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
161	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
162	Mutations ofARX are associated with striking pleiotropy and consistent genotype-phenotype correlation. Human Mutation, 2004, 23, 147-159.	2.5	293

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163	Allelic loss on chromosomes 2q21 and 19p 13.2 in oxyphilic thyroid tumors. International Journal of Cancer, 2004, 111, 463-467.	5.1	32
164	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
165	Retinal angioma in a patient with Cowden disease. American Journal of Ophthalmology, 2003, 135, 400-402.	3.3	20
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167	Microchimerism from a dizygotic twin in juvenile ulcerative lichen planus. Lancet, The, 2002, 359, 1861-1862.	13.7	22
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