Alfredo Brusco

List of Publications by Year
in descending order

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184 papers	7,746 citations	100601 38 h-index	81351 76 g-index
194 all docs	194 docs citations	194 times ranked	13944 citing authors

#	Article	IF	Citations
1	Functional analysis of TLK2 variants and their proximal interactomes implicates impaired kinase activity and chromatin maintenance defects in their pathogenesis. Journal of Medical Genetics, 2022, 59, 170-179.	1.5	9
2	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. Brain, 2022, 145, 208-223.	3.7	15
3	Digenic inheritance of STUB1 variants and TBP polyglutamine expansions explains the incomplete penetrance of SCA17 and SCA48. Genetics in Medicine, 2022, 24, 29-40.	1.1	24
4	Analysis of the DNA methylation pattern of the promoter region of calcitonin gene-related peptide 1 gene in patients with episodic migraine: An exploratory case-control study. Neurobiology of Pain (Cambridge, Mass), 2022, 11, 100089.	1.0	10
5	Drosophila functional screening of de novo variants in autism uncovers damaging variants and facilitates discovery of rare neurodevelopmental diseases. Cell Reports, 2022, 38, 110517.	2.9	24
6	MEK Inhibition in a Newborn with RAF1-Associated Noonan Syndrome Ameliorates Hypertrophic Cardiomyopathy but Is Insufficient to Revert Pulmonary Vascular Disease. Genes, 2022, 13, 6.	1.0	20
7	Heterozygous <scp>PNPT1</scp> Variants Cause Spinocerebellar Ataxia Type 25. Annals of Neurology, 2022, 92, 122-137.	2.8	8
8	The Emerging Roles of Long Non-Coding RNAs in Intellectual Disability and Related Neurodevelopmental Disorders. International Journal of Molecular Sciences, 2022, 23, 6118.	1.8	1
9	De novo truncating NOVA2 variants affect alternative splicing and lead to heterogeneous neurodevelopmental phenotypes. Human Mutation, 2022, 43, 1299-1313.	1.1	6
10	Next-Generation Sequencing Advances the Genetic Diagnosis of Cerebral Cavernous Malformation (CCM). Antioxidants, 2022, 11, 1294.	2.2	7
11	Clinical spectrum and followâ€up in six individuals with Lambâ€“Shaffer syndrome (<scp>SOX5</scp>). American Journal of Medical Genetics, Part A, 2021, 185, 608-613.	0.7	6
12	A highâ€content drug screening strategy to identify protein level modulators for genetic diseases: A proofâ€ofâ€principle in autosomal dominant leukodystrophy. Human Mutation, 2021, 42, 102-116.	1.1	1
13	Evidence that <scp>FGFRL1</scp> contributes to congenital diaphragmatic hernia development in humans. American Journal of Medical Genetics, Part A, 2021, 185, 836-840.	0.7	8
14	Electroclinical features and outcome of ANKRD11-related KBG syndrome: A novel report and literature review. Seizure: the Journal of the British Epilepsy Association, 2021, 85, 151-154.	0.9	7
15	Expanding the clinical phenotype of the ultraâ€rare <scp>Skrabanâ€Deardorff</scp> syndrome: Two novel individuals with <scp>WDR26</scp> lossâ€ofâ€function variants and a literature review. American Journal of Medical Genetics, Part A, 2021, 185, 1712-1720.	0.7	6
16	Haploinsufficiency of PRR12 causes a spectrum of neurodevelopmental, eye, and multisystem abnormalities. Genetics in Medicine, 2021, 23, 1234-1245.	1.1	6
17	Wilms tumour occurring in a patient with osteopathia striata with cranial sclerosis: A still unsolved biological question. Pediatric Blood and Cancer, 2021, 68, e29132.	0.8	1
18	Biallelic mutations in RNF220 cause laminopathies featuring leukodystrophy, ataxia and deafness. Brain, 2021, 144, 3020-3035.	3.7	11

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19	Motor and cognitive outcomes of cerebello-spinal stimulation in neurodegenerative ataxia. Brain, 2021, 144, 2310-2321.	3.7	38
20	Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. American Journal of Human Genetics, 2021, 108, 857-873.	2.6	19
21	Elovl5 is required for proper action potential conduction along peripheral myelinated fibers. Glia, 2021, 69, 2419-2428.	2.5	8
22	KCNK18 Biallelic Variants Associated with Intellectual Disability and Neurodevelopmental Disorders Alter TRESK Channel Activity. International Journal of Molecular Sciences, 2021, 22, 6064.	1.8	3
23	Biallelic PI4KA variants cause a novel neurodevelopmental syndrome with hypomyelinating leukodystrophy. Brain, 2021, 144, 2659-2669.	3.7	19
24	CCGâ€¢CGG interruptions in highâ€penetrance SCA8 families increase RAN translation and protein toxicity. EMBO Molecular Medicine, 2021, 13, e14095.	3.3	12
25	Trace elements profile in the blood of Huntingtonâ€™ disease patients. Journal of Trace Elements in Medicine and Biology, 2020, 57, 18-20.	1.5	20
26	Novel LRPPRC compound heterozygous mutation in a child with early-onset Leigh syndrome French-Canadian type: case report of an Italian patient. Italian Journal of Pediatrics, 2020, 46, 140.	1.0	18
27	A Novel CCT5 Missense Variant Associated with Early Onset Motor Neuropathy. International Journal of Molecular Sciences, 2020, 21, 7631.	1.8	8
28	ACE2 gene variants may underlie interindividual variability and susceptibility to COVID-19 in the Italian population. European Journal of Human Genetics, 2020, 28, 1602-1614.	1.4	208
29	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	4.7	43
30	SLC20A1 Is Involved in Urinary Tract and Urorectal Development. Frontiers in Cell and Developmental Biology, 2020, 8, 567.	1.8	22
31	Clinical, neuropathological, and genetic characterization of STUB1 variants in cerebellar ataxias: a frequent cause of predominant cognitive impairment. Genetics in Medicine, 2020, 22, 1851-1862.	1.1	30
32	New Insights into Potocki-Shaffer Syndrome: Report of Two Novel Cases and Literature Review. Brain Sciences, 2020, 10, 788.	1.1	7
33	A novel case of congenital spinocerebellar ataxia 5: further support for a specific phenotype associated with the p.(Arg480Trp) variant in SPTBN2. BMJ Case Reports, 2020, 13, e238108.	0.2	4
34	In vitro dexamethasone treatment does not induce alternative ATM transcripts in cells from Ataxiaâ€“Telangiectasia patients. Scientific Reports, 2020, 10, 20182.	1.6	3
35	Missense variant contribution to USP9X-female syndrome. Npj Genomic Medicine, 2020, 5, 53.	1.7	17
36	Challenging arterial calcification disease associated with rare NT5E gene mutation. BMJ Case Reports, 2020, 13, e235365.	0.2	2

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37	Autozygosity-driven genetic diagnosis in consanguineous families from Italy and the Greater Middle East. Human Genetics, 2020, 139, 1429-1441.	1.8	8
38	A 20â€year long term experience of the Italian Diamondâ€Blackfan Anaemia Registry: RPS and RPL genes, different faces of the same disease?. British Journal of Haematology, 2020, 190, 93-104.	1.2	35
39	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell, 2020, 180, 568-584.e23.	13.5	1,422
40	Next Generation Sequencing (NGS) Strategies for Genetic Testing of Cerebral Cavernous Malformation (CCM) Disease. Methods in Molecular Biology, 2020, 2152, 59-75.	0.4	2
41	Prevalence and phenotype of the c.1529C>T <scp>SPG</scp>7 variant in adultâ€onset cerebellar ataxia in Italy. European Journal of Neurology, 2019, 26, 80-86.	1.7	12
42	Congenital Sensorineural Hearing Loss and Inborn Pigmentary Disorders: First Report of Multilocus Syndrome in Piebaldism. Medicina (Lithuania), 2019, 55, 345.	0.8	2
43	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	5.8	150
44	Design of a multiplex ligation-dependent probe amplification assay for SLC20A2: identification of two novel deletions in primary familial brain calcification. Journal of Human Genetics, 2019, 64, 1083-1090.	1.1	2
45	Front Cover, Volume 40, Issue 6. Human Mutation, 2019, 40, i.	1.1	0
46	Allele-specific silencing as treatment for gene duplication disorders: proof-of-principle in autosomal dominant leukodystrophy. Brain, 2019, 142, 1905-1920.	3.7	15
47	Long-term efficacy of docosahexaenoic acid (DHA) for Spinocerebellar Ataxia 38 (SCA38) treatment: An open label extension study. Parkinsonism and Related Disorders, 2019, 63, 191-194.	1.1	19
48	NBAS pathogenic variants: Defining the associated clinical and facial phenotype and genotypeâ€“phenotype correlations. Human Mutation, 2019, 40, 721-728.	1.1	26
49	Genomic deletions upstream of lamin B1 lead to atypical autosomal dominant leukodystrophy. Neurology: Genetics, 2019, 5, e305.	0.9	16
50	A novel case of Greenberg dysplasia and genotypeâ€“phenotype correlation analysis for LBR pathogenic variants: An instructive example of one geneâ€multiple phenotypes. American Journal of Medical Genetics, Part A, 2019, 179, 306-311.	0.7	8
51	Mice harbouring a SCA28 patient mutation in AFG3L2 develop late-onset ataxia associated with enhanced mitochondrial proteotoxicity. Neurobiology of Disease, 2019, 124, 14-28.	2.1	23
52	Spontaneous remission in a Diamondâ€Blackfan anaemia patient due to a revertant uniparental disomy ablating a de novo RPS19 mutation. British Journal of Haematology, 2019, 185, 994-998.	1.2	24
53	Small supernumerary marker chromosomes: A legacy of trisomy rescue?. Human Mutation, 2019, 40, 193-200.	1.1	33
54	A fetal case of microphthalmia and limb anomalies with abnormal neuronal migration associated with SMOC1 biallelic variants. European Journal of Medical Genetics, 2019, 62, 103578.	0.7	4

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55	ATXN2 intermediate repeat expansions influence the clinical phenotype in frontotemporal dementia. Neurobiology of Aging, 2019, 73, 231.e7-231.e9.	1.5	21
56	Mitochondrial stress response triggered by defects in protein synthesis quality control. Life Science Alliance, 2019, 2, e201800219.	1.3	26
57	Genomic Architecture of ASD. , 2019, , 23-34.		0
58	Spinocerebellar Ataxia Tethering PCR. Journal of Molecular Diagnostics, 2018, 20, 289-297.	1.2	16
59	Quantifying the Effects of 16p11.2 Copy Number Variants on Brain Structure: A Multisite Genetic-First Study. Biological Psychiatry, 2018, 84, 253-264.	0.7	56
60	Functional evaluation of natural killer cell cytotoxic activity in NFKB2-mutated patients. Immunology Letters, 2018, 194, 40-43.	1.1	12
61	Altered homeostasis of trace elements in the blood of SCA2 patients. Journal of Trace Elements in Medicine and Biology, 2018, 47, 111-114.	1.5	7
62	Complexity of the Genetics and Clinical Presentation of Spinocerebellar Ataxia 17. Frontiers in Cellular Neuroscience, 2018, 12, 429.	1.8	21
63	X chromosome dosage and presence of SRY shape sex-specific differences in DNA methylation at an autosomal region in human cells. Biology of Sex Differences, 2018, 9, 10.	1.8	20
64	ISL1 is a major susceptibility gene for classic bladder exstrophy and a regulator of urinary tract development. Scientific Reports, 2017, 7, 42170.	1.6	41
65	A case of Feingold type 2 syndrome associated with keratoconus refines keratoconus type 7 locus on chromosome 13q. European Journal of Medical Genetics, 2017, 60, 224-227.	0.7	10
66	A novel homozygous change ofCLCN2(p.His590Pro) is associated with a subclinical form of leukoencephalopathy with ataxia (LKPAT). Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 894-896.	0.9	20
67	Three novel missense mutations in SLC20A2 associated with idiopathic basal ganglia calcification. Journal of the Neurological Sciences, 2017, 377, 62-64.	0.3	7
68	Copy number variants analysis in a cohort of isolated and syndromic developmental delay/intellectual disability reveals novel genomic disorders, position effects and candidate disease genes. Clinical Genetics, 2017, 92, 415-422.	1.0	43
69	Exome sequencing in children of women with skewed X-inactivation identifies atypical cases and complex phenotypes. European Journal of Paediatric Neurology, 2017, 21, 475-484.	0.7	14
70	Docosahexaenoic acid is a beneficial replacement treatment for spinocerebellar ataxia 38. Annals of Neurology, 2017, 82, 615-621.	2.8	30
71	A syndromic extreme insulin resistance caused by biallelic POC1A mutations in exon 10. European Journal of Endocrinology, 2017, 177, K21-K27.	1.9	8
72	Human canonical CD157/Bst1 is an alternatively spliced isoform masking a previously unidentified primate-specific exon included in a novel transcript. Scientific Reports, 2017, 7, 15923.	1.6	10

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73	Motor Deficits and Cerebellar Atrophy in Elovl5 Knock Out Mice. Frontiers in Cellular Neuroscience, 2017, 11, 343.	1.8	29
74	Synaptic Interactome Mining Reveals p140Cap as a New Hub for PSD Proteins Involved in Psychiatric and Neurological Disorders. Frontiers in Molecular Neuroscience, 2017, 10, 212.	1.4	30
75	Cover Image, Volume 170A, Number 7, July 2016. , 2016, 170, i-i.		0
76	Clinical and neuroradiological features of spinocerebellar ataxia 38 (SCA38). Parkinsonism and Related Disorders, 2016, 28, 80-86.	1.1	27
77	Long-term treatment with thiamine as possible medical therapy for Friedreich ataxia. Journal of Neurology, 2016, 263, 2170-2178.	1.8	18
78	Whole exome sequencing is necessary to clarify ID/DD cases with de novo copy number variants of uncertain significance: Two proofâ€ofâ€concept examples. American Journal of Medical Genetics, Part A, 2016, 170, 1772-1779.	0.7	26
79	A novel 3q29 deletion associated with autism, intellectual disability, psychiatric disorders, and obesity. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2016, 171, 290-299.	1.1	34
80	CNV analysis in 169 patients with bladder exstrophy-epispadias complex. BMC Medical Genetics, 2016, 17, 35.	2.1	15
81	Heterozygous Deletion of KLHL1/ATX8OS at the SCA8 Locus Is Unlikely Associated With Cerebellar Impairment in Humans. Cerebellum, 2016, 15, 208-212.	1.4	3
82	A Novel CSF1R Mutation in a Patient with Clinical and Neuroradiological Features of Hereditary Diffuse Leukoencephalopathy with Axonal Spheroids. Journal of Alzheimer's Disease, 2015, 47, 319-322.	1.2	14
83	O056. Migraine as presenting symptom of SLC20A2gene mutations. Journal of Headache and Pain, 2015, 16, A121.	2.5	0
84	Array-Comparative Genomic Hybridization Analysis in Fetuses with Major Congenital Malformations Reveals that 24% of Cases Have Pathogenic Deletions/Duplications. Cytogenetic and Genome Research, 2015, 147, 10-16.	0.6	4
85	The 16p11.2 locus modulates brain structures common to autism, schizophrenia and obesity. Molecular Psychiatry, 2015, 20, 140-147.	4.1	160
86	Messenger RNA processing is altered in autosomal dominant leukodystrophy. Human Molecular Genetics, 2015, 24, 2746-2756.	1.4	27
87	Adult-onset autosomal recessive ataxia associated with neuronal ceroid lipofuscinosis type 5 gene (CLN5) mutations. Journal of Neurology, 2015, 262, 173-178.	1.8	29
88	An atypical form of AOA2 with myoclonus associated with mutations in SETX and AFG3L2. BMC Medical Genetics, 2015, 16, 16.	2.1	12
89	Two families with novel missense mutations in COL4A1: When diagnosis can be missed. Journal of the Neurological Sciences, 2015, 352, 99-104.	0.3	21
90	A Recurrent Mutation in CACNA1G Alters Cav3.1 T-Type Calcium-Channel Conduction and Causes Autosomal-Dominant Cerebellar Ataxia. American Journal of Human Genetics, 2015, 97, 726-737.	2.6	87

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91	A large genomic deletion leads to enhancer adoption by the lamin B1 gene: a second path to autosomal dominant adult-onset demyelinating leukodystrophy (ADLD). Human Molecular Genetics, 2015, 24, 3143-3154.	1.4	117
92	Intrafamilial phenotypic variability in Spinocerebellar ataxia type 8. Journal of the Neurological Sciences, 2015, 357, e255.	0.3	2
93	Blood metal levels and related antioxidant enzyme activities in patients with ataxia telangiectasia. Neurobiology of Disease, 2015, 81, 162-167.	2.1	13
94	Testis development in the absence of SRY: chromosomal rearrangements at SOX9 and SOX3. European Journal of Human Genetics, 2015, 23, 1025-1032.	1.4	59
95	Novel mutation of SLC20A2 in an Italian patient presenting with migraine. Journal of Neurology, 2014, 261, 2019-2021.	1.8	12
96	A New Case of 13q12.2q13.1 Microdeletion Syndrome Contributes to Phenotype Delineation. Case Reports in Genetics, 2014, 2014, 1-5.	0.1	3
97	Large cryptic genomic rearrangements with apparently normal karyotypes detected by array-CGH. Molecular Cytogenetics, 2014, 7, 82.	0.4	25
98	Defining the phenotype associated with microduplication reciprocal to Sotos syndrome microdeletion. American Journal of Medical Genetics, Part A, 2014, 164, 2084-2090.	0.7	20
99	Modulation of the age at onset in spinocerebellar ataxia by CAG tracts in various genes. Brain, 2014, 137, 2444-2455.	3.7	144
100	ELOVL5 Mutations Cause Spinocerebellar Ataxia 38. American Journal of Human Genetics, 2014, 95, 209-217.	2.6	107
101	Genome-wide expression profiling and functional characterization of SCA28 lymphoblastoid cell lines reveal impairment in cell growth and activation of apoptotic pathways. BMC Medical Genomics, 2013, 6, 22.	0.7	14
102	SETX mutations are a frequent genetic cause of juvenile and adult onset cerebellar ataxia with neuropathy and elevated serum alpha-fetoprotein. Orphanet Journal of Rare Diseases, 2013, 8, 123.	1.2	31
103	Analysis of <scp> LMNB </scp> 1 Duplications in Autosomal Dominant Leukodystrophy Provides Insights into Duplication Mechanisms and Alleleâ€Specific Expression. Human Mutation, 2013, 34, 1160-1171.	1.1	33
104	Deep-intronic ATM mutation detected by genomic resequencing and corrected in vitro by antisense morpholino oligonucleotide (AMO). European Journal of Human Genetics, 2013, 21, 774-778.	1.4	31
105	Ade novoX;8 translocation creates aPTK2-THOC2gene fusion withTHOC2expression knockdown in a patient with psychomotor retardation and congenital cerebellar hypoplasia. Journal of Medical Genetics, 2013, 50, 543-551.	1.5	42
106	Different electroclinical picture of generalized epilepsy in two families with 15q13.3 microdeletion. Epilepsia, 2013, 54, e69-73.	2.6	14
107	Bilaterally cleft lip and bilateral thumb polydactyly with triphalangeal component in a patient with two De novo deletions of HSA 4q32 and 4q34 involving PDGFC, GRIA2, and FBXO8 genes. American Journal of Medical Genetics, Part A, 2013, 161, 2656-2662.	0.7	6
108	Progressive extreme heterotopic calcification. American Journal of Medical Genetics, Part A, 2013, 161, 1706-1713.	0.7	3

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109	Spastic paraplegia gene 7 in patients with spasticity and/or optic neuropathy. Brain, 2012, 135, 2980-2993.	3.7	148
110	High frequency of ribosomal protein gene deletions in Italian Diamond-Blackfan anemia patients detected by multiplex ligation-dependent probe amplification assay. Haematologica, 2012, 97, 1813-1817.	1.7	49
111	Megalencephalic leukoencephalopathy with subcortical cysts type 1 (MLC1) due to a homozygous deep intronic splicing mutation (c.895-226T>G) abrogated in vitro using an antisense morpholino oligonucleotide. Neurogenetics, 2012, 13, 205-214.	0.7	21
112	790ÂKb microduplication in chromosome band 17p13.1 associated with intellectual disability, afebrile seizures, dysmorphic features, diabetes, and hypothyroidism. European Journal of Medical Genetics, 2012, 55, 222-224.	0.7	23
113	De novo 13q12.3â€“q14.11 deletion involving BRCA2 gene in a patient with developmental delay, elevated IgM levels, transient ataxia, and cerebellar hypoplasia, mimicking an Aâ€T like phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 2571-2576.	0.7	6
114	A randomized trial of oral betamethasone to reduce ataxia symptoms in ataxia telangiectasia. Movement Disorders, 2012, 27, 1312-1316.	2.2	73
115	Overexpression of CD157 Contributes to Epithelial Ovarian Cancer Progression by Promoting Mesenchymal Differentiation. PLoS ONE, 2012, 7, e43649.	1.1	22
116	Functional characterization and targeted correction of ATM mutations identified in Japanese patients with ataxia-telangiectasia. Human Mutation, 2012, 33, 198-208.	1.1	39
117	NT5EMutations and Arterial Calcifications. New England Journal of Medicine, 2011, 364, 432-442.	13.9	403
118	A novel spinocerebellar ataxia type 15 family with involuntary movements and cognitive decline. European Journal of Neurology, 2011, 18, 1263-1265.	1.7	24
119	ATXN-2 CAG repeat expansions are interrupted in ALS patients. Human Genetics, 2011, 130, 575-580.	1.8	52
120	Gene-targeted embryonic stem cells: real-time PCR assay for estimation of the number of neomycin selection cassettes. Biological Procedures Online, 2011, 13, 10.	1.4	12
121	Molecular Mechanisms Generating and Stabilizing Terminal 22q13 Deletions in 44 Subjects with Phelan/McDermid Syndrome. PLoS Genetics, 2011, 7, e1002173.	1.5	172
122	High Frequency of Large Gene Deletions Detected by Multiplex Ligation-Dependent Probe Amplification in Diamond Blackfan Anemia,. Blood, 2011, 118, 3428-3428.	0.6	0
123	Diamond-Blackfan anemia: genotype-phenotype correlations in Italian patients with RPL5 and RPL11 mutations. Haematologica, 2010, 95, 206-213.	1.7	78
124	Two Italian Families with ITPR1 Gene Deletion Presenting a Broader Phenotype of SCA15. Cerebellum, 2010, 9, 115-123.	1.4	39
125	Mouse brain expression patterns of Spg7, Afg3l1, and Afg3l2 transcripts, encoding for the mitochondrial m-AAA protease. BMC Neuroscience, 2010, 11, 55.	0.8	26
126	Missense mutations in the AFG3L2 proteolytic domain account for â¼1.5% of European autosomal dominant cerebellar ataxias. Human Mutation, 2010, 31, 1117-1124.	1.1	81

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127	Spinocerebellar ataxia type 12 identified in two Italian families may mimic sporadic ataxia. Movement Disorders, 2010, 25, 1269-1273.	2.2	25
128	A family with autosomal dominant leukodystrophy linked to 5q23.2â€“q23.3 without lamin B1 mutations. European Journal of Neurology, 2010, 17, 541-549.	1.7	36
129	Mutations in the mitochondrial protease gene AFG3L2 cause dominant hereditary ataxia SCA28. Nature Genetics, 2010, 42, 313-321.	9.4	291
130	A novel family with Lamin B1 duplication associated with adult-onset leucoencephalopathy. Journal of Neurology, Neurosurgery and Psychiatry, 2009, 80, 237-240.	0.9	35
131	Mutations in the lamin B1 gene are not present in multiple sclerosis. European Journal of Neurology, 2009, 16, 544-546.	1.7	4
132	Two-tier analysis of histone H2AX phosphorylation allows the identification of Ataxia Telangiectasia heterozygotes. Radiotherapy and Oncology, 2009, 92, 133-137.	0.3	17
133	Spinocerebellar ataxia type 28: A novel autosomal dominant cerebellar ataxia characterized by slow progression and ophthalmoparesis. Cerebellum, 2008, 7, 184-188.	1.4	57
134	Mutations in the POLG1 gene are not a relevant cause of cerebellar ataxia in Italy. Journal of Neurology, 2008, 255, 1079-1080.	1.8	8
135	A previously undiagnosed case of Gerstmannâ€šStrÃusslerâ€šScheinker disease revealed by PRNP gene analysis in patients with adultâ€onset ataxia. Movement Disorders, 2008, 23, 1468-1471.	2.2	10
136	A rapid flow cytometry test based on histone H2AX phosphorylation for the sensitive and specific diagnosis of ataxia telangiectasia. Cytometry Part A: the Journal of the International Society for Analytical Cytology, 2008, 73A, 508-516.	1.1	73
137	Multiplex ligation-dependent probe amplification enhances molecular diagnosis of Diamond-Blackfan anemia due to RPS19 deficiency. Haematologica, 2008, 93, 1748-1750.	1.7	16
138	CGG repeat length correlates with age of onset of motor signs of the fragile X-associated tremor/ataxia syndrome (FXTAS). American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2007, 144B, 566-569.	1.1	138
139	The (â^'16C > T) substitution in thePLEKHG4 gene is not present among European ADCA patients. Movement Disorders, 2007, 22, 752-753.	2.2	2
140	Glutathione levels in blood from ataxia telangiectasia patients suggest in vivo adaptive mechanisms to oxidative stress. Clinical Biochemistry, 2007, 40, 666-670.	0.8	15
141	Large Genomic Mutations within the ATM Gene Detected by MLPA, Including a Duplication of 41 kb from Exon 4 to 20. Annals of Human Genetics, 2007, 72, 071003005754001-???.	0.3	27
142	Large Pathogenic Expansions in the SCA2 and SCA7 Genes Can Be Detected by Fluorescent Repeat-Primed Polymerase Chain Reaction Assay. Journal of Molecular Diagnostics, 2006, 8, 128-132.	1.2	31
143	The polymorphic polyglutamine repeat in the mitochondrial DNA polymerase Î³ gene is not associated with oligozoospermia. Journal of Endocrinological Investigation, 2006, 29, 1-4.	1.8	58
144	Prognostic Values of Soluble CD30 and CD30 Gene Polymorphisms in Heart Transplantation. Transplantation, 2006, 81, 1153-1156.	0.5	14

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145	Myelin-Associated Glycoprotein is Altered in a Familial Late-Onset Orthochromatic Leukodystrophy. Brain Pathology, 2006, 15, 116-123.	2.1	9
146	ATM mutations in Italian families with ataxia telangiectasia include two distinct large genomic deletions. Human Mutation, 2006, 27, 1061-1061.	1.1	34
147	SCA28, a novel form of autosomal dominant cerebellar ataxia on chromosome 18p11.22â€“q11.2. Brain, 2006, 129, 235-242.	3.7	122
148	Functional significance of a deep intronic mutation in the ATM gene and evidence for an alternative exon 28a. Human Mutation, 2005, 25, 118-124.	1.1	48
149	FMR1 gene premutation is a frequent genetic cause of late-onset sporadic cerebellar ataxia. Neurology, 2005, 64, 145-147.	1.5	90
150	Association of a new cationic trypsinogen gene mutation (V39A) with chronic pancreatitis in an Italian family. Gut, 2005, 54, 1663-1664.	6.1	10
151	An Enhanced Polymerase Chain Reaction Assay to Detect Pre- and Full Mutation Alleles of the Fragile X Mental Retardation 1 Gene. Journal of Molecular Diagnostics, 2005, 7, 605-612.	1.2	96
152	Somatic Mosaicism and Variable Expressivity in Diamond Blackfan Anemia (DBA): A Large Deletion Involving the 19q13 Locus in a Patient with Transient Anemia.. Blood, 2005, 106, 3550-3550.	0.6	0
153	Molecular Genetics of Hereditary Spinocerebellar Ataxia. Archives of Neurology, 2004, 61, 727.	4.9	130
154	Detection of Large Pathogenic Expansions in FRDA1, SCA10, and SCA12 Genes Using a Simple Fluorescent Repeat-Primed PCR Assay. Journal of Molecular Diagnostics, 2004, 6, 96-100.	1.2	48
155	Six novel ATM mutations in Italian patients with classical ataxia-telangiectasia. Human Mutation, 2003, 21, 450-450.	1.1	6
156	A late onset variant of ataxia-telangiectasia with a compound heterozygous genotype, A8030G/7481insA. Journal of Medical Genetics, 2002, 39, 57-61.	1.5	49
157	Analysis of SCA8 and SCA12 loci in 134 Italian ataxic patients negative for SCA1-3, 6 and 7 CAG expansions. Journal of Neurology, 2002, 249, 923-929.	1.8	35
158	COL4A3/COL4A4 mutations: From familial hematuria to autosomal-dominant or recessive Alport syndrome. Kidney International, 2002, 61, 1947-1956.	2.6	187
159	Germline mutations in CFTR and PSTI genes in chronic pancreatitis patients. Digestive Diseases and Sciences, 2002, 47, 2416-2421.	1.1	41
160	TSC1 and TSC2 deletions differ in size, preference for recombinatorial sequences, and location within the gene. Human Genetics, 2001, 108, 156-166.	1.8	19
161	Defective Function of Fas in Patients With Type 1 Diabetes Associated With Other Autoimmune Diseases. Diabetes, 2001, 50, 483-488.	0.3	45
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