Brent L Fogel

List of Publications by Year
in descending order

91	4,679	109321	110387
papers	citations	35 h-index	64 g-index

93	93	93	8959
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinical Exome Sequencing for Genetic Identification of Rare Mendelian Disorders. JAMA - Journal of the American Medical Association, 2014, 312, 1880.	7.4	842
2	Clinical features and molecular genetics of autosomal recessive cerebellar ataxias. Lancet Neurology, The, 2007, 6, 245-257.	10.2	264
3	Mutations in XPR1 cause primary familial brain calcification associated with altered phosphate export. Nature Genetics, 2015, 47, 579-581.	21.4	237
4	Exome Sequencing in the Clinical Diagnosis of Sporadic or Familial Cerebellar Ataxia. JAMA Neurology, 2014, 71, 1237.	9.0	211
5	RBFOX1 regulates both splicing and transcriptional networks in human neuronal development. Human Molecular Genetics, 2012, 21, 4171-4186.	2.9	192
6	Bioinformatics-Based Identification of Expanded Repeats: A Non-reference Intronic Pentamer Expansion in RFC1 Causes CANVAS. American Journal of Human Genetics, 2019, 105, 151-165.	6.2	170
7	Diagnostic utility of transcriptome sequencing for rare Mendelian diseases. Genetics in Medicine, 2020, 22, 490-499.	2.4	136
8	Mutations in SLC20A2 are a major cause of familial idiopathic basal ganglia calcification. Neurogenetics, 2013, 14, 11-22.	1.4	131
9	Adult polyglucosan body disease: Natural History and Key Magnetic Resonance Imaging Findings. Annals of Neurology, 2012, 72, 433-441.	5.3	125
10	Whole exome sequencing in patients with white matter abnormalities. Annals of Neurology, 2016, 79, 1031-1037.	5.3	116
11	Î±-Synuclein in blood exosomes immunoprecipitated using neuronal and oligodendroglial markers distinguishes Parkinsonâ€™s disease from multiple system atrophy. Acta Neuropathologica, 2021, 142, 495-511.	7.7	80
12	Clinical Neurogenetics. Neurologic Clinics, 2013, 31, 987-1007.	1.8	78
13	Do mutations in the murine ataxia gene TRPC3 cause cerebellar ataxia in humans?. Movement Disorders, 2015, 30, 284-286.	3.9	78
14	Clinical application of next-generation sequencing to the practice of neurology. Lancet Neurology, The, 2019, 18, 492-503.	10.2	76
15	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
16	Orchestration of Neurodevelopmental Programs by RBFOX1. International Review of Neurobiology, 2013, 113, 251-267.	2.0	64
17	ELAVL2-regulated transcriptional and splicing networks in human neurons link neurodevelopment and autism. Human Molecular Genetics, 2016, 25, ddw110.	2.9	63
18	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	2.4	60

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19	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. American Journal of Human Genetics, 2018, 102, 494-504.	6.2	59
20	Lysosomal Storage and Albinism Due to Effects of a De Novo CLCN7 Variant on Lysosomal Acidification. American Journal of Human Genetics, 2019, 104, 1127-1138.	6.2	59
21	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. American Journal of Human Genetics, 2019, 104, 164-178.	6.2	59
22	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. Human Mutation, 2020, 41, 487-501.	2.5	58
23	KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. Brain, 2020, 143, 3242-3261.	7.6	57
24	Clinical exome sequencing in neurologic disease. Neurology: Clinical Practice, 2016, 6, 164-176.	1.6	56
25	An approach to the patient with late-onset cerebellar ataxia. Nature Clinical Practice Neurology, 2006, 2, 629-635.	2.5	55
26	Emerging therapies in Friedreich's ataxia. Neurodegenerative Disease Management, 2016, 6, 49-65.	2.2	55
27	A Cellular Protein, hnRNP H, Binds to the Negative Regulator of Splicing Element from Rous Sarcoma Virus. Journal of Biological Chemistry, 2000, 275, 32371-32378.	3.4	54
28	Chimeric Peptide Species Contribute to Divergent Dipeptide Repeat Pathology in c9ALS/FTD and SCA36. Neuron, 2020, 107, 292-305.e6.	8.1	51
29	Childhood Cerebellar Ataxia. Journal of Child Neurology, 2012, 27, 1138-1145.	1.4	50
30	Primary brain calcification: an international study reporting novel variants and associated phenotypes. European Journal of Human Genetics, 2018, 26, 1462-1477.	2.8	48
31	Novel mutations in the senataxin DNA/RNA helicase domain in ataxia with oculomotor apraxia 2. Neurology, 2006, 67, 2083-2084.	1.1	47
32	Mutation of senataxin alters disease-specific transcriptional networks in patients with ataxia with oculomotor apraxia type 2. Human Molecular Genetics, 2014, 23, 4758-4769.	2.9	43
33	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. American Journal of Human Genetics, 2019, 105, 413-424.	6.2	43
34	Spinocerebellar ataxia type 29 due to mutations in ITPR1: a case series and review of this emerging congenital ataxia. Orphanet Journal of Rare Diseases, 2017, 12, 121.	2.7	42
35	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
36	Prevalence of RFC1 -mediated spinocerebellar ataxia in a North American ataxia cohort. Neurology: Genetics, 2020, 6, e440.	1.9	40

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37	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. American Journal of Human Genetics, 2020, 106, 570-583.	6.2	37
38	A new model to study neurodegeneration in ataxia oculomotor apraxia type 2. Human Molecular Genetics, 2015, 24, 5759-5774.	2.9	34
39	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.	2.5	33
40	De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Callosum, Axon, Cardiac, Ocular, and Genital Defects. American Journal of Human Genetics, 2019, 105, 854-868.	6.2	29
41	Efficient polyadenylation of Rous sarcoma virus RNA requires the negative regulator of splicing element. Nucleic Acids Research, 2002, 30, 810-817.	14.5	27
42	Candidate Screening of the TRPC3 Gene in Cerebellar Ataxia. Cerebellum, 2011, 10, 296-299.	2.5	27
43	Mutations in rare ataxia genes are uncommon causes of sporadic cerebellar ataxia. Movement Disorders, 2012, 27, 442-446.	3.9	27
44	A Family with Spinocerebellar Ataxia Type 5 Found to Have a Novel Missense Mutation within a SPTBN2 Spectrin Repeat. Cerebellum, 2013, 12, 162-164.	2.5	27
45	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	6.2	27
46	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in POLR3A, POLR3B, and POLR1C. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e660-e674.	3.6	26
47	Autosomal-recessive cerebellar ataxias. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 147, 187-209.	1.8	25
48	A family with spinocerebellar ataxia and retinitis pigmentosa attributed to an ELOVL4 mutation. Neurology: Genetics, 2019, 5, e357.	1.9	25
49	Clinical exome sequencing in neurogenetic and neuropsychiatric disorders. Annals of the New York Academy of Sciences, 2016, 1366, 49-60.	3.8	23
50	C9ORF72 expansion is not a significant cause of sporadic spinocerebellar ataxia. Movement Disorders, 2012, 27, 1835-1836.	3.9	21
51	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	7.6	21
52	Paving the Way Toward Meaningful Trials in Ataxias: An Ataxia Global Initiative Perspective. Movement Disorders, 2022, 37, 1125-1130.	3.9	21
53	Aberrant Splicing of the Senataxin Gene in a Patient with Ataxia with Oculomotor Apraxia Type 2. Cerebellum, 2009, 8, 448-453.	2.5	20
54	Genetic and genomic testing for neurologic disease in clinical practice. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 147, 11-22.	1.8	20

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55	Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. Human Mutation, 2019, 40, 1115-1126.	2.5	19
56	Disruption of Spermatogenesis and Infertility in Ataxia with Oculomotor Apraxia Type 2 (AOA2). Cerebellum, 2019, 18, 448-456.	2.5	19
57	Creutzfeldtâ€"Jakob disease presenting with alien limb sign. Movement Disorders, 2006, 21, 1040-1042.	3.9	15
58	Prevalence of spinocerebellar ataxia 36 in a US population. Neurology: Genetics, 2017, 3, e174.	1.9	15
59	Early infantile epileptic encephalopathy due to biallelic pathogenic variants in <scp>PIGQ</scp>: Report of seven new subjects and review of the literature. Journal of Inherited Metabolic Disease, 2020, 43, 1321-1332.	3.6	15
60	The Neurogenetics of Atypical Parkinsonian Disorders. Seminars in Neurology, 2014, 34, 217-224.	1.4	14
61	Expanding the global prevalence of spinocerebellar ataxia type 42. Neurology: Genetics, 2018, 4, e232.	1.9	14
62	Variants in SCAF4 Cause a Neurodevelopmental Disorder and Are Associated with Impaired mRNA Processing. American Journal of Human Genetics, 2020, 107, 544-554.	6.2	13
63	Interpretation of Genetic Testing. CONTINUUM Lifelong Learning in Neurology, 2011, 17, 347-352.	0.8	11
64	Next generation sequencing in clinical diagnosis. Lancet Neurology, The, 2019, 18, 426.	10.2	11
65	Detection of a mosaic CDKL5 deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. Molecular Genetics & Genomic Medicine, 2021, 9, e1665.	1.2	11
66	New-Onset Psychosis in a Patient With Spinocerebellar Ataxia Type 10. American Journal of Psychiatry, 2011, 168, 1339-1340.	7.2	10
67	Utilization of Genetic Testing Prior to Subspecialist Referral for Cerebellar Ataxia. Genetic Testing and Molecular Biomarkers, 2013, 17, 588-594.	0.7	9
68	Successful treatment of a genetic childhood ataxia due to riboflavin transporter deficiency. Cerebellum and Ataxias, 2018, 5, 12.	1.9	9
69	Magnetic Resonance Imaging Abnormalities in the Corpus Callosum of a Patient With Neuropsychiatric Lupus. Neurologist, 2006, 12, 271-273.	0.7	8
70	IgG4â€related disease: Association with a rare gene variant expressed in cytotoxic T cells. Molecular Genetics & Genomic Medicine, 2019, 7, e686.	1.2	8
71	DYRK1A pathogenic variants in two patients with syndromic intellectual disability and a review of the literature. Molecular Genetics & Genomic Medicine, 2020, 8, e1544.	1.2	8
72	The need to develop a patient-centered precision medicine model for adults with chronic disability. Expert Review of Molecular Diagnostics, 2017, 17, 415-418.	3.1	7

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73	Trace Contamination Following Reuse of Anion-Exchange DNA Purification Resins. BioTechniques, 2000, 28, 299-302.	1.8	6
74	Cerebellar disorders. , 2011, , 198-216.		6
75	Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy. Clinical Imaging, 2019, 58, 108-113.	1.5	6
76	Progressive Ataxia with Elevated Alpha-Fetoprotein: Diagnostic Issues and Review of the Literature. Tremor and Other Hyperkinetic Movements, 2019, 9, .	2.0	6
77	Novel NUDT2 variant causes intellectual disability and polyneuropathy. Annals of Clinical and Translational Neurology, 2020, 7, 2320-2325.	3.7	5
78	miR-142-3p regulates cortical oligodendrocyte gene co-expression networks associated with tauopathy. Human Molecular Genetics, 2021, 30, 103-118.	2.9	5
79	Lack of Association Between GBA Mutations and Motor Complications in European and American Parkinsonâ€™s Disease Cohorts. Journal of Parkinson's Disease, 2021, 11, 1569-1578.	2.8	5
80	De novo pathogenic variant in SETX causes a rapidly progressive neurodegenerative disorder of early childhood-onset with severe axonal polyneuropathy. Acta Neuropathologica Communications, 2021, 9, 194.	5.2	5
81	Progressive spinocerebellar ataxia mimicked by a presumptive cerebellar arteriovenous malformation. European Journal of Radiology Extra, 2009, 71, e1-e2.	0.1	4
82	Mutations in PDYN are not responsible for multiple system atrophy. Journal of Neurology, 2013, 260, 927-928.	3.6	4
83	Collaborative science unites researchers and a novel spastic ataxia gene. Annals of Neurology, 2018, 83, 1072-1074.	5.3	4
84	The Neurodevelopmental and Motor Phenotype of SCA21 (ATX-TMEM240). Journal of Child Neurology, 2020, 35, 953-962.	1.4	4
85	Family genetic result communication in rare and undiagnosed disease communities: Understanding the practice. Journal of Genetic Counseling, 2021, 30, 439-447.	1.6	4
86	Acute pharmacogenetic dystonic reactions in a family with the CYP2D6 *41 allele: a case report. Journal of Medical Case Reports, 2021, 15, 432.	0.8	3
87	Clinical Neurogenetics. , 2012, , 704-734.		3
88	A family with combined mutations of the hemophilia A and X-linked adrenoleukodystrophy genes. Neurogenetics, 2008, 9, 215-218.	1.4	2
89	Spinocerebellar Ataxia type 29 in a family of MÄori descent. Cerebellum and Ataxias, 2019, 6, 14.	1.9	2
90	Emotional detachment, gait ataxia, and cerebellar dysconnectivity associated with compound heterozygous mutations in the SPG7 gene. Neurocase, 2020, 26, 299-304.	0.6	2

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91	Genotypeâ€“phenotype considerations in neurogenetic disease. , 2020, , 59-69.		1