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	Paving the Way Toward Meaningful Trials in Ataxias: An Ataxia Global Initiative Perspective. Movement Disorders, 2022, 37, 1125-1130.	3.9	21
2	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
3	Family genetic result communication in rare and undiagnosed disease communities: Understanding the practice. Journal of Genetic Counseling, 2021, 30, 439-447.	1.6	4
4	miR-142-3p regulates cortical oligodendrocyte gene co-expression networks associated with tauopathy. Human Molecular Genetics, 2021, 30, 103-118.	2.9	5
5	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
6	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
7	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
8	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
9	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
10	Diagnostic utility of transcriptome sequencing for rare Mendelian diseases. Genetics in Medicine, 2020, 22, 490-499.	2.4	136
11	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
12	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. Human Mutation, 2020, 41, 487-501.	2.5	58
13	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
14	The Neurodevelopmental and Motor Phenotype of SCA21 (ATX-TMEM240). Journal of Child Neurology, 2020, 35, 953-962.	1.4	4
15	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	7.6	21
16	Emotional detachment, gait ataxia, and cerebellar dysconnectivity associated with compound heterozygous mutations in the SPG7 gene. Neurocase, 2020, 26, 299-304.	0.6	2
17	Novel NUDT2 variant causes intellectual disability and polyneuropathy. Annals of Clinical and Translational Neurology, 2020, 7, 2320-2325.	3.7	5
18	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

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19	KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. Brain, 2020, 143, 3242-3261.	7.6	57
20	Prevalence of RFC1 -mediated spinocerebellar ataxia in a North American ataxia cohort. Neurology: Genetics, 2020, 6, e440.	1.9	40
21	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
22	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
23	Chimeric Peptide Species Contribute to Divergent Dipeptide Repeat Pathology in c9ALS/FTD and SCA36. Neuron, 2020, 107, 292-305.e6.	8.1	51
24	Genotypeâ€“phenotype considerations in neurogenetic disease. , 2020, , 59-69.		1
25	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	2.4	60
26	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
27	Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy. Clinical Imaging, 2019, 58, 108-113.	1.5	6
28	Spinocerebellar Ataxia type 29 in a family of MÄori descent. Cerebellum and Ataxias, 2019, 6, 14.	1.9	2
29	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
30	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
31	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
32	Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. Human Mutation, 2019, 40, 1115-1126.	2.5	19
33	IgG4â€related disease: Association with a rare gene variant expressed in cytotoxic T cells. Molecular Genetics & Genomic Medicine, 2019, 7, e686.	1.2	8
34	Clinical application of next-generation sequencing to the practice of neurology. Lancet Neurology, The, 2019, 18, 492-503.	10.2	76
35	Next generation sequencing in clinical diagnosis. Lancet Neurology, The, 2019, 18, 426.	10.2	11
36	Disruption of Spermatogenesis and Infertility in Ataxia with Oculomotor Apraxia Type 2 (AOA2). Cerebellum, 2019, 18, 448-456.	2.5	19

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37	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
38	A family with spinocerebellar ataxia and retinitis pigmentosa attributed to an ELOVL4 mutation. Neurology: Genetics, 2019, 5, e357.	1.9	25
39	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
40	Progressive Ataxia with Elevated Alpha-Fetoprotein: Diagnostic Issues and Review of the Literature. Tremor and Other Hyperkinetic Movements, 2019, 9, .	2.0	6
41	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
42	Expanding the global prevalence of spinocerebellar ataxia type 42. Neurology: Genetics, 2018, 4, e232.	1.9	14
43	Successful treatment of a genetic childhood ataxia due to riboflavin transporter deficiency. Cerebellum and Ataxias, 2018, 5, 12.	1.9	9
44	Primary brain calcification: an international study reporting novel variants and associated phenotypes. European Journal of Human Genetics, 2018, 26, 1462-1477.	2.8	48
45	Collaborative science unites researchers and a novel spastic ataxia gene. Annals of Neurology, 2018, 83, 1072-1074.	5.3	4
46	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
47	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
48	Autosomal-recessive cerebellar ataxias. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 147, 187-209.	1.8	25
49	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
50	Prevalence of spinocerebellar ataxia 36 in a US population. Neurology: Genetics, 2017, 3, e174.	1.9	15
51	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
52	Clinical exome sequencing in neurogenetic and neuropsychiatric disorders. Annals of the New York Academy of Sciences, 2016, 1366, 49-60.	3.8	23
53	Whole exome sequencing in patients with white matter abnormalities. Annals of Neurology, 2016, 79, 1031-1037.	5.3	116
54	ELAVL2-regulated transcriptional and splicing networks in human neurons link neurodevelopment and autism. Human Molecular Genetics, 2016, 25, ddw110.	2.9	63

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55	Emerging therapies in Friedreich's ataxia. Neurodegenerative Disease Management, 2016, 6, 49-65.	2.2	55
56	Clinical exome sequencing in neurologic disease. Neurology: Clinical Practice, 2016, 6, 164-176.	1.6	56
57	Do mutations in the murine ataxia gene TRPC3 cause cerebellar ataxia in humans?. Movement Disorders, 2015, 30, 284-286.	3.9	78
58	A new model to study neurodegeneration in ataxia oculomotor apraxia type 2. Human Molecular Genetics, 2015, 24, 5759-5774.	2.9	34
59	Mutations in XPR1 cause primary familial brain calcification associated with altered phosphate export. Nature Genetics, 2015, 47, 579-581.	21.4	237
60	The Neurogenetics of Atypical Parkinsonian Disorders. Seminars in Neurology, 2014, 34, 217-224.	1.4	14
61	Exome Sequencing in the Clinical Diagnosis of Sporadic or Familial Cerebellar Ataxia. JAMA Neurology, 2014, 71, 1237.	9.0	211
62	Clinical Exome Sequencing for Genetic Identification of Rare Mendelian Disorders. JAMA - Journal of the American Medical Association, 2014, 312, 1880.	7.4	842
63	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
64	Mutations in PDYN are not responsible for multiple system atrophy. Journal of Neurology, 2013, 260, 927-928.	3.6	4
65	Clinical Neurogenetics. Neurologic Clinics, 2013, 31, 987-1007.	1.8	78
66	Mutations in SLC20A2 are a major cause of familial idiopathic basal ganglia calcification. Neurogenetics, 2013, 14, 11-22.	1.4	131
67	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
68	Utilization of Genetic Testing Prior to Subspecialist Referral for Cerebellar Ataxia. Genetic Testing and Molecular Biomarkers, 2013, 17, 588-594.	0.7	9
69	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
70	Orchestration of Neurodevelopmental Programs by RBFOX1. International Review of Neurobiology, 2013, 113, 251-267.	2.0	64
71	Childhood Cerebellar Ataxia. Journal of Child Neurology, 2012, 27, 1138-1145.	1.4	50
72	C9ORF72 expansion is not a significant cause of sporadic spinocerebellar ataxia. Movement Disorders, 2012, 27, 1835-1836.	3.9	21

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73	RBFOX1 regulates both splicing and transcriptional networks in human neuronal development. Human Molecular Genetics, 2012, 21, 4171-4186.	2.9	192
74	Adult polyglucosan body disease: Natural History and Key Magnetic Resonance Imaging Findings. Annals of Neurology, 2012, 72, 433-441.	5.3	125
75	Mutations in rare ataxia genes are uncommon causes of sporadic cerebellar ataxia. Movement Disorders, 2012, 27, 442-446.	3.9	27
76	Clinical Neurogenetics. , 2012, , 704-734.		3
77	Cerebellar disorders. , 2011, , 198-216.		6
78	Interpretation of Genetic Testing. CONTINUUM Lifelong Learning in Neurology, 2011, 17, 347-352.	0.8	11
79	Candidate Screening of the TRPC3 Gene in Cerebellar Ataxia. Cerebellum, 2011, 10, 296-299.	2.5	27
80	New-Onset Psychosis in a Patient With Spinocerebellar Ataxia Type 10. American Journal of Psychiatry, 2011, 168, 1339-1340.	7.2	10
81	Aberrant Splicing of the Senataxin Gene in a Patient with Ataxia with Oculomotor Apraxia Type 2. Cerebellum, 2009, 8, 448-453.	2.5	20
82	Progressive spinocerebellar ataxia mimicked by a presumptive cerebellar arteriovenous malformation. European Journal of Radiology Extra, 2009, 71, e1-e2.	0.1	4
83	A family with combined mutations of the hemophilia A and X-linked adrenoleukodystrophy genes. Neurogenetics, 2008, 9, 215-218.	1.4	2
84	Clinical features and molecular genetics of autosomal recessive cerebellar ataxias. Lancet Neurology, The, 2007, 6, 245-257.	10.2	264
85	An approach to the patient with late-onset cerebellar ataxia. Nature Clinical Practice Neurology, 2006, 2, 629-635.	2.5	55
86	Magnetic Resonance Imaging Abnormalities in the Corpus Callosum of a Patient With Neuropsychiatric Lupus. Neurologist, 2006, 12, 271-273.	0.7	8
87	Creutzfeldtâ€"Jakob disease presenting with alien limb sign. Movement Disorders, 2006, 21, 1040-1042.	3.9	15
88	Novel mutations in the senataxin DNA/RNA helicase domain in ataxia with oculomotor apraxia 2. Neurology, 2006, 67, 2083-2084.	1.1	47
89	Efficient polyadenylation of Rous sarcoma virus RNA requires the negative regulator of splicing element. Nucleic Acids Research, 2002, 30, 810-817.	14.5	27
90	Trace Contamination Following Reuse of Anion-Exchange DNA Purification Resins. BioTechniques, 2000, 28, 299-302.	1.8	6

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91	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