Katherine Mathews

List of Publications by Year
in descending order

89 papers	5,972 citations	81900 39 h-index	74163 75 g-index
90 all docs	90 docs citations	90 times ranked	7452 citing authors

#	Article	IF	Citations
1	Post-translational disruption of dystroglycanâ€“ligand interactions in congenital muscular dystrophies. Nature, 2002, 418, 417-421.	27.8	747
2	Ataluren in patients with nonsense mutation Duchenne muscular dystrophy (ACT DMD): a multicentre, randomised, double-blind, placebo-controlled, phase 3 trial. Lancet, The, 2017, 390, 1489-1498.	13.7	365
3	Mutational spectrum of DMD mutations in dystrophinopathy patients: application of modern diagnostic techniques to a large cohort. Human Mutation, 2009, 30, 1657-1666.	2.5	279
4	Diagnostic approach to the congenital muscular dystrophies. Neuromuscular Disorders, 2014, 24, 289-311.	0.6	275
5	Mortality in Friedreich Ataxia. Journal of the Neurological Sciences, 2011, 307, 46-49.	0.6	236
6	Prevalence of Duchenne and Becker Muscular Dystrophies in the United States. Pediatrics, 2015, 135, 513-521.	2.1	233
7	ISPD loss-of-function mutations disrupt dystroglycan O-mannosylation and cause Walker-Warburg syndrome. Nature Genetics, 2012, 44, 575-580.	21.4	212
8	<scp>LTBP4</scp> genotype predicts age of ambulatory loss in duchenne muscular dystrophy. Annals of Neurology, 2013, 73, 481-488.	5.3	202
9	Mutations in PHF6 are associated with BÃ¶rjesonâ€“ForssmanÂâ€“Lehmann syndrome. Nature Genetics, 2002, 32, 661-665.	21.4	192
10	Delayed Diagnosis in Duchenne Muscular Dystrophy: Data from the Muscular Dystrophy Surveillance, Tracking, and Research Network (MD STARnet). Journal of Pediatrics, 2009, 155, 380-385.	1.8	187
11	Limb-Girdle Muscular Dystrophy in the United States. Journal of Neuropathology and Experimental Neurology, 2006, 65, 995-1003.	1.7	144
12	Clinical and genetic characterization of manifesting carriers of DMD mutations. Neuromuscular Disorders, 2010, 20, 499-504.	0.6	136
13	Mutations in SPATA5 Are Associated with Microcephaly, Intellectual Disability, Seizures, and Hearing Loss. American Journal of Human Genetics, 2015, 97, 457-464.	6.2	134
14	Progression of Friedreich ataxia: quantitative characterization over 5 years. Annals of Clinical and Translational Neurology, 2016, 3, 684-694.	3.7	117
15	Mutation spectrum in the large GTPase dynamin 2, and genotype-phenotype correlation in autosomal dominant centronuclear myopathy. Human Mutation, 2012, 33, 949-959.	2.5	115
16	Steroid Therapy and Cardiac Function in Duchenne Muscular Dystrophy. Pediatric Cardiology, 2005, 26, 768-771.	1.3	104
17	Putamen Lesions and the Development of Attention-Deficit/Hyperactivity Symptomatology. Journal of the American Academy of Child and Adolescent Psychiatry, 2002, 41, 563-571.	0.5	103
18	Nonsense mutation-associated Becker muscular dystrophy: interplay between exon definition and splicing regulatory elements within the DMD gene. Human Mutation, 2011, 32, 299-308.	2.5	103

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19	Measuring the rate of progression in Friedreich ataxia: Implications for clinical trial design. Movement Disorders, 2010, 25, 426-432.	3.9	102
20	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome Biology, 2014, 15, R53.	9.6	101
21	Psychiatric Disorders After Childhood Stroke. Journal of the American Academy of Child and Adolescent Psychiatry, 2002, 41, 555-562.	0.5	86
22	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	7.6	85
23	Correspondence. Journal of Child Neurology, 1991, 6, 90-90.	1.4	74
24	Association Study of Exon Variants in the NF-Î¹B and TGFÎ² Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. American Journal of Human Genetics, 2016, 99, 1163-1171.	6.2	71
25	Attention deficit hyperactivity disorder and neurocognitive correlates after childhood stroke. Journal of the International Neuropsychological Society, 2003, 9, 815-829.	1.8	67
26	Muscular Dystrophy Surveillance Tracking and Research Network (MD STARnet): Case Definition in Surveillance for Childhood-Onset Duchenne/Becker Muscular Dystrophy. Journal of Child Neurology, 2010, 25, 1098-1102.	1.4	63
27	Analysis of the visual system in Friedreich ataxia. Journal of Neurology, 2013, 260, 2362-2369.	3.6	55
28	Frataxin levels in peripheral tissue in Friedreich ataxia. Annals of Clinical and Translational Neurology, 2015, 2, 831-842.	3.7	55
29	Corticosteroid Treatment and Growth Patterns in Ambulatory Males with Duchenne Muscular Dystrophy. Journal of Pediatrics, 2016, 173, 207-213.e3.	1.8	51
30	Analysis of Echocardiograms in a Large Heterogeneous Cohort of Patients With Friedreich Ataxia. American Journal of Cardiology, 2012, 109, 401-405.	1.6	50
31	Friedreich Ataxia Clinical Outcome Measures. Journal of Child Neurology, 2012, 27, 1152-1158.	1.4	48
32	Cerebral infarction complicating Fontan surgery for cyanotic congenital heart disease. Pediatric Cardiology, 1986, 7, 161-166.	1.3	46
33	Health related quality of life measures in Friedreich Ataxia. Journal of the Neurological Sciences, 2008, 272, 123-128.	0.6	46
34	Brain structure in juvenile-onset Huntington disease. Neurology, 2019, 92, e1939-e1947.	1.1	45
35	Hypertensive Encephalopathy in Childhood. Journal of Child Neurology, 1996, 11, 193-196.	1.4	44
36	Prefrontal and Executive Attention Network Lesions and the Development of Attention-Deficit/Hyperactivity Symptomatology. Journal of the American Academy of Child and Adolescent Psychiatry, 2005, 44, 443-450.	0.5	44

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37	LMNA variants cause cytoplasmic distribution of nuclear pore proteins in Drosophila and human muscle. Human Molecular Genetics, 2012, 21, 1544-1556.	2.9	44
38	Trafficking-competent and trafficking-defectiveKCNJ2 mutations in Andersen syndrome. Human Mutation, 2006, 27, 388-388.	2.5	42
39	Use of Corticosteroids in a Population-Based Cohort of Boys With Duchenne and Becker Muscular Dystrophy. Journal of Child Neurology, 2010, 25, 1319-1324.	1.4	42
40	Multiminicore Myopathy, Central Core Disease, Malignant Hyperthermia Susceptibility, and RYR1 Mutations. Archives of Neurology, 2004, 61, 27.	4.5	40
41	Bone density and alendronate effects in Duchenne Muscular Dystrophy patients. Muscle and Nerve, 2014, 49, 506-511.	2.2	39
42	GMPPB-Associated Dystroglycanopathy: Emerging Common Variants with Phenotype Correlation. Human Mutation, 2015, 36, 1159-1163.	2.5	39
43	Clinical phenotypes as predictors of the outcome of skipping around <scp>DMD</scp> exon 45. Annals of Neurology, 2015, 77, 668-674.	5.3	38
44	Limb-girdle muscular dystrophy. Current Neurology and Neuroscience Reports, 2003, 3, 78-85.	4.2	33
45	Antioxidant use in Friedreich ataxia. Journal of the Neurological Sciences, 2008, 267, 174-176.	0.6	33
46	Autosomal Recessive Cerebellar Hypoplasia. Journal of Child Neurology, 1989, 4, 189-194.	1.4	30
47	Age at onset of first signs or symptoms predicts age at loss of ambulation in Duchenne and Becker Muscular Dystrophy: Data from the MD STARnet. Journal of Pediatric Rehabilitation Medicine, 2016, 9, 5-11.	0.5	29
48	Infantile onset CMT2D/dSMA V in monozygotic twins due to a mutation in the anticodonâ€binding domain of GARS. Journal of the Peripheral Nervous System, 2012, 17, 132-134.	3.1	28
49	Placeboâ€controlled Phase 2 Trial of Drisapersen for Duchenne Muscular Dystrophy. Annals of Clinical and Translational Neurology, 2018, 5, 913-926.	3.7	28
50	Phenotypic and Pathologic Evaluation of the myd Mouse. A Candidate Model for Facioscapulohumeral Dystrophy. Journal of Neuropathology and Experimental Neurology, 1995, 54, 601-606.	1.7	27
51	Attention function after childhood stroke. Journal of the International Neuropsychological Society, 2004, 10, 976-986.	1.8	27
52	Neurobehavioral Concerns Among Males with Dystrophinopathy Using Population-Based Surveillance Data from the Muscular Dystrophy Surveillance, Tracking, and Research Network. Journal of Developmental and Behavioral Pediatrics, 2015, 36, 455-463.	1.1	26
53	A survey-based study identifies common but unrecognized symptoms in a large series of juvenile Huntington's disease. Neurodegenerative Disease Management, 2017, 7, 307-315.	2.2	25
54	Editorial by concerned physicians: Unintended effect of the orphan drug act on the potential cost of 3,4-diaminopyridine. Muscle and Nerve, 2016, 53, 165-168.	2.2	24

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55	Presentation, Management and Follow-Up of Schilderâ€™s Disease. Pediatric Neurosurgery, 1998, 29, 86-91.	0.7	23
56	Risk Factors for First Fractures Among Males With Duchenne or Becker Muscular Dystrophy. Journal of Pediatric Orthopaedics, 2015, 35, 640-644.	1.2	23
57	Mutation Analysis in a Population-Based Cohort of Boys With Duchenne or Becker Muscular Dystrophy. Journal of Child Neurology, 2009, 24, 425-430.	1.4	22
58	Natural History of Friedreich Ataxia. Neurology, 2022, 99, .	1.1	21
59	Use of Complementary and Alternative Medicine by Males With Duchenne or Becker Muscular Dystrophy. Journal of Child Neurology, 2012, 27, 734-740.	1.4	20
60	Uniparental disomy unveils a novel recessive mutation in POMT2. Neuromuscular Disorders, 2018, 28, 592-596.	0.6	20
61	Global FKRP Registry: observations in more than 300 patients with Limb Girdle Muscular Dystrophy R9. Annals of Clinical and Translational Neurology, 2020, 7, 757-766.	3.7	20
62	Perceived quality of life among caregivers of children with a childhood-onset dystrophinopathy: a double ABCX model of caregiver stressors and perceived resources. Health and Quality of Life Outcomes, 2017, 15, 33.	2.4	19
63	Descriptive Phenotype of Obsessive Compulsive Symptoms in Males With Duchenne Muscular Dystrophy. Journal of Child Neurology, 2018, 33, 572-579.	1.4	18
64	Genetic mapping near the myd locus on mouse Chromosome 8. Mammalian Genome, 1995, 6, 278-280.	2.2	17
65	Novel pathogenic <scp>COX</scp>20 variants causing dysarthria, ataxia, and sensory neuropathy. Annals of Clinical and Translational Neurology, 2019, 6, 154-160.	3.7	17
66	Muscular dystrophy overview: genetics and diagnosis. Neurologic Clinics, 2003, 21, 795-816.	1.8	15
67	Longitudinal analysis of contrast acuity in Friedreich ataxia. Neurology: Genetics, 2018, 4, e250.	1.9	15
68	Exercise-Induced Left Ventricular Systolic Dysfunction in Women Heterozygous for Dystrophinopathy. Journal of the American Society of Echocardiography, 2010, 23, 848-853.	2.8	13
69	Implementation of Duchenne Muscular Dystrophy Care Considerations. Pediatrics, 2018, 142, .	2.1	11
70	Reâ€analysis of an original <scp>CMTX3</scp> family using exome sequencing identifies a known <scp>BSCL2</scp> mutation. Muscle and Nerve, 2013, 47, 922-924.	2.2	10
71	Mouse myodystrophy (myd) mutation: Refined mapping in an interval flanked by homology with distal human 4q. Muscle and Nerve, 1995, 18, S98-S102.	2.2	9
72	Genetic and Physical Mapping of a Voltage-Dependent Chloride Channel Gene to Human 4q32 and to Mouse 8. Genomics, 1996, 36, 374-376.	2.9	8

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73	Genitourinary health in a population-based cohort of males with Duchenne and Becker Muscular dystrophies. Muscle and Nerve, 2015, 52, 22-27.	2.2	7
74	Subacute sclerosing panencephalitis with remission in a Bosnian refugee child. Pediatric Infectious Disease Journal, 2003, 22, 757-758.	2.0	6
75	Comorbid Medical Conditions in Friedreich Ataxia. Journal of Child Neurology, 2016, 31, 1161-1165.	1.4	6
76	Childhood Activity on Progression in Limb Girdle Muscular Dystrophy 2I. Journal of Child Neurology, 2017, 32, 204-209.	1.4	6
77	Impact of Mobility Device Use on Quality of Life in Children With Friedreich Ataxia. Journal of Child Neurology, 2018, 33, 397-404.	1.4	5
78	Hereditary causes of chorea in childhood. Seminars in Pediatric Neurology, 2003, 10, 20-25.	2.0	3
79	Dystrophinopathy muscle biopsies in the genetic testing ERA: One center's data. Muscle and Nerve, 2018, 58, 149-153.	2.2	3
80	A syntrophin gene maps to mouse Chromosome 8 and is not the myodystrophy gene. Mammalian Genome, 1995, 6, 664-665.	2.2	2
81	214th ENMC International Workshop: Establishing an international consortium for gene discovery and clinical research for Congenital Muscle Disease, Heemskerk, the Netherlands, 6â€“18 October 2015. Neuromuscular Disorders, 2019, 29, 644-650.	0.6	2
82	The Boy Who Lost His Smile. Annals of Otology, Rhinology and Laryngology, 2015, 124, 148-152.	1.1	1
83	Facial Weakness and Ophthalmoplegia in a 4-Day-Old Infant. Seminars in Pediatric Neurology, 2018, 26, 63-66.	2.0	1
84	NINDS Common Data Elements for Congenital Muscular Dystrophy Clinical Research: A National Institute for Neurological Disorders and Stroke Project. Journal of Neuromuscular Diseases, 2018, 5, 75-84.	2.6	1
85	Diagnostic delay in patients with FKRP-related muscular dystrophy. Neuromuscular Disorders, 2021, , .	0.6	1
86	eP213: Phase 2 multiple ascending-dose study of SRP-5051 PPMO in patients with DMD amenable to exon 51 skipping: Part A results. Genetics in Medicine, 2022, 24, S133.	2.4	1
87	GENETICS of MUSCLE DISEASE. CONTINUUM Lifelong Learning in Neurology, 2005, 11, 95-114.	0.8	0
88	A 16-Week-Old Infant With Failure to Thrive and Hypotonia. Clinical Pediatrics, 2013, 52, 1075-1078.	0.8	0
89	Neuromuscular disease - Gene transfer for children. Journal of International Child Neurology Association, 2021, 1, .	0.0	0