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	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
2	Natural History of Friedreich Ataxia. Neurology, 2022, 99, .	1.1	21
3	Diagnostic delay in patients with FKRP-related muscular dystrophy. Neuromuscular Disorders, 2021, , .	0.6	1
4	Neuromuscular disease - Gene transfer for children. Journal of International Child Neurology Association, 2021, 1, .	0.0	0
5	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
6	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
7	Brain structure in juvenile-onset Huntington disease. Neurology, 2019, 92, e1939-e1947.	1.1	45
8	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
9	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
10	Uniparental disomy unveils a novel recessive mutation in POMT2. Neuromuscular Disorders, 2018, 28, 592-596.	0.6	20
11	Dystrophinopathy muscle biopsies in the genetic testing ERA: One center's data. Muscle and Nerve, 2018, 58, 149-153.	2.2	3
12	Facial Weakness and Ophthalmoplegia in a 4-Day-Old Infant. Seminars in Pediatric Neurology, 2018, 26, 63-66.	2.0	1
13	Descriptive Phenotype of Obsessive Compulsive Symptoms in Males With Duchenne Muscular Dystrophy. Journal of Child Neurology, 2018, 33, 572-579.	1.4	18
14	Longitudinal analysis of contrast acuity in Friedreich ataxia. Neurology: Genetics, 2018, 4, e250.	1.9	15
15	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
16	Placeboâ€controlled Phase 2 Trial of Drisapersen for Duchenne Muscular Dystrophy. Annals of Clinical and Translational Neurology, 2018, 5, 913-926.	3.7	28
17	Implementation of Duchenne Muscular Dystrophy Care Considerations. Pediatrics, 2018, 142, .	2.1	11
18	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

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19	Childhood Activity on Progression in Limb Girdle Muscular Dystrophy 2I. Journal of Child Neurology, 2017, 32, 204-209.	1.4	6
20	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	7.6	85
21	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
22	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
23	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
24	Comorbid Medical Conditions in Friedreich Ataxia. Journal of Child Neurology, 2016, 31, 1161-1165.	1.4	6
25	Corticosteroid Treatment and Growth Patterns in Ambulatory Males with Duchenne Muscular Dystrophy. Journal of Pediatrics, 2016, 173, 207-213.e3.	1.8	51
26	Progression of Friedreich ataxia: quantitative characterization over 5 years. Annals of Clinical and Translational Neurology, 2016, 3, 684-694.	3.7	117
27	Association Study of Exon Variants in the NF-Î²B and TGFÎ² Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. American Journal of Human Genetics, 2016, 99, 1163-1171.	6.2	71
28	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
29	Frataxin levels in peripheral tissue in Friedreich ataxia. Annals of Clinical and Translational Neurology, 2015, 2, 831-842.	3.7	55
30	GMPPB-Associated Dystroglycanopathy: Emerging Common Variants with Phenotype Correlation. Human Mutation, 2015, 36, 1159-1163.	2.5	39
31	Risk Factors for First Fractures Among Males With Duchenne or Becker Muscular Dystrophy. Journal of Pediatric Orthopaedics, 2015, 35, 640-644.	1.2	23
32	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
33	The Boy Who Lost His Smile. Annals of Otology, Rhinology and Laryngology, 2015, 124, 148-152.	1.1	1
34	Prevalence of Duchenne and Becker Muscular Dystrophies in the United States. Pediatrics, 2015, 135, 513-521.	2.1	233
35	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
36	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

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37	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
38	Bone density and alendronate effects in Duchenne Muscular Dystrophy patients. Muscle and Nerve, 2014, 49, 506-511.	2.2	39
39	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
40	Diagnostic approach to the congenital muscular dystrophies. Neuromuscular Disorders, 2014, 24, 289-311.	0.6	275
41	Analysis of the visual system in Friedreich ataxia. Journal of Neurology, 2013, 260, 2362-2369.	3.6	55
42	<scp>LTBP4</scp> genotype predicts age of ambulatory loss in duchenne muscular dystrophy. Annals of Neurology, 2013, 73, 481-488.	5.3	202
43	A 16-Week-Old Infant With Failure to Thrive and Hypotonia. Clinical Pediatrics, 2013, 52, 1075-1078.	0.8	0
44	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
45	LMNA variants cause cytoplasmic distribution of nuclear pore proteins in Drosophila and human muscle. Human Molecular Genetics, 2012, 21, 1544-1556.	2.9	44
46	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
47	ISPD loss-of-function mutations disrupt dystroglycan O-mannosylation and cause Walker-Warburg syndrome. Nature Genetics, 2012, 44, 575-580.	21.4	212
48	Friedreich Ataxia Clinical Outcome Measures. Journal of Child Neurology, 2012, 27, 1152-1158.	1.4	48
49	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
50	Analysis of Echocardiograms in a Large Heterogeneous Cohort of Patients With Friedreich Ataxia. American Journal of Cardiology, 2012, 109, 401-405.	1.6	50
51	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
52	Mortality in Friedreich Ataxia. Journal of the Neurological Sciences, 2011, 307, 46-49.	0.6	236
53	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
54	Measuring the rate of progression in Friedreich ataxia: Implications for clinical trial design. Movement Disorders, 2010, 25, 426-432.	3.9	102

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55	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
56	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
57	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
58	Clinical and genetic characterization of manifesting carriers of DMD mutations. Neuromuscular Disorders, 2010, 20, 499-504.	0.6	136
59	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
60	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
61	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
62	Antioxidant use in Friedreich ataxia. Journal of the Neurological Sciences, 2008, 267, 174-176.	0.6	33
63	Health related quality of life measures in Friedreich Ataxia. Journal of the Neurological Sciences, 2008, 272, 123-128.	0.6	46
64	Limb-Girdle Muscular Dystrophy in the United States. Journal of Neuropathology and Experimental Neurology, 2006, 65, 995-1003.	1.7	144
65	Trafficking-competent and trafficking-defectiveKCNJ2 mutations in Andersen syndrome. Human Mutation, 2006, 27, 388-388.	2.5	42
66	GENETICS of MUSCLE DISEASE. CONTINUUM Lifelong Learning in Neurology, 2005, 11, 95-114.	0.8	0
67	Steroid Therapy and Cardiac Function in Duchenne Muscular Dystrophy. Pediatric Cardiology, 2005, 26, 768-771.	1.3	104
68	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
69	Multiminicore Myopathy, Central Core Disease, Malignant Hyperthermia Susceptibility, and RYR1 Mutations. Archives of Neurology, 2004, 61, 27.	4.5	40
70	Attention function after childhood stroke. Journal of the International Neuropsychological Society, 2004, 10, 976-986.	1.8	27
71	Limb-girdle muscular dystrophy. Current Neurology and Neuroscience Reports, 2003, 3, 78-85.	4.2	33
72	Hereditary causes of chorea in childhood. Seminars in Pediatric Neurology, 2003, 10, 20-25.	2.0	3

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73	Muscular dystrophy overview: genetics and diagnosis. Neurologic Clinics, 2003, 21, 795-816.	1.8	15
74	Attention deficit hyperactivity disorder and neurocognitive correlates after childhood stroke. Journal of the International Neuropsychological Society, 2003, 9, 815-829.	1.8	67
75	Subacute sclerosing panencephalitis with remission in a Bosnian refugee child. Pediatric Infectious Disease Journal, 2003, 22, 757-758.	2.0	6
76	Psychiatric Disorders After Childhood Stroke. Journal of the American Academy of Child and Adolescent Psychiatry, 2002, 41, 555-562.	0.5	86
77	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
78	Mutations in PHF6 are associated with BÃ¶rjesonâ€“ForssmanÂâ€“Lehmann syndrome. Nature Genetics, 2002, 32, 661-665.	21.4	192
79	Post-translational disruption of dystroglycanâ€“ligand interactions in congenital muscular dystrophies. Nature, 2002, 418, 417-421.	27.8	747
80	Presentation, Management and Follow-Up of Schilderâ€™s Disease. Pediatric Neurosurgery, 1998, 29, 86-91.	0.7	23
81	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
82	Hypertensive Encephalopathy in Childhood. Journal of Child Neurology, 1996, 11, 193-196.	1.4	44
83	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
84	A syntrophin gene maps to mouse Chromosome 8 and is not the myodystrophy gene. Mammalian Genome, 1995, 6, 664-665.	2.2	2
85	Genetic mapping near the myd locus on mouse Chromosome 8. Mammalian Genome, 1995, 6, 278-280.	2.2	17
86	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
87	Correspondence. Journal of Child Neurology, 1991, 6, 90-90.	1.4	74
88	Autosomal Recessive Cerebellar Hypoplasia. Journal of Child Neurology, 1989, 4, 189-194.	1.4	30
89	Cerebral infarction complicating Fontan surgery for cyanotic congenital heart disease. Pediatric Cardiology, 1986, 7, 161-166.	1.3	46