Alan H Beggs

List of Publications by Year
in descending order

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267	24,222	7096 78	9103 144
papers	citations	h-index	g-index

281	281	281	23739
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Genotype-Phenotype Correlation in the Long-QT Syndrome. Circulation, 2001, 103, 89-95.	1.6	1,641
2	Mutations in ACTN4, encoding Î±-actinin-4, cause familial focal segmental glomerulosclerosis. Nature Genetics, 2000, 24, 251-256.	21.4	1,124
3	ACTN3 Genotype Is Associated with Human Elite Athletic Performance. American Journal of Human Genetics, 2003, 73, 627-631.	6.2	708
4	Competitive binding of Î±-actinin and calmodulin to the NMDA receptor. Nature, 1997, 385, 439-442.	27.8	567
5	Severe arrhythmia disorder caused by cardiac L-type calcium channel mutations. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 8089-8096.	7.1	558
6	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. Science Translational Medicine, 2017, 9, .	12.4	516
7	Variant of SCN5A Sodium Channel Implicated in Risk of Cardiac Arrhythmia. Science, 2002, 297, 1333-1336.	12.6	506
8	Patient-Customized Oligonucleotide Therapy for a Rare Genetic Disease. New England Journal of Medicine, 2019, 381, 1644-1652.	27.0	481
9	Distinctive patterns of microRNA expression in primary muscular disorders. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 17016-17021.	7.1	458
10	Multiple Serotonergic Brainstem Abnormalities in Sudden Infant Death Syndrome. JAMA - Journal of the American Medical Association, 2006, 296, 2124.	7.4	443
11	Mutations in dynamin 2 cause dominant centronuclear myopathy. Nature Genetics, 2005, 37, 1207-1209.	21.4	390
12	Mutations in the skeletal muscle Î±-actin gene in patients with actin myopathy and nemaline myopathy. Nature Genetics, 1999, 23, 208-212.	21.4	389
13	A common nonsense mutation results in Î±-actinin-3 deficiency in the general population. Nature Genetics, 1999, 21, 353-354.	21.4	378
14	Ribosomal Protein L5 and L11 Mutations Are Associated with Cleft Palate and Abnormal Thumbs in Diamond-Blackfan Anemia Patients. American Journal of Human Genetics, 2008, 83, 769-780.	6.2	363
15	Gene expression comparison of biopsies from Duchenne muscular dystrophy (DMD) and normal skeletal muscle. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 15000-15005.	7.1	312
16	Mutations in the nebulin gene associated with autosomal recessive nemaline myopathy. Proceedings of the National Academy of Sciences of the United States of America, 1999, 96, 2305-2310.	7.1	304
17	Exome sequencing identifies GATA1 mutations resulting in Diamond-Blackfan anemia. Journal of Clinical Investigation, 2012, 122, 2439-2443.	8.2	292
18	Type I interferonâ€“inducible gene expression in blood is present and reflects disease activity in dermatomyositis and polymyositis. Arthritis and Rheumatism, 2007, 56, 3784-3792.	6.7	264

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19	Ribosomal Protein S24 Gene Is Mutated in Diamond-Blackfan Anemia. American Journal of Human Genetics, 2006, 79, 1110-1118.	6.2	257
20	Altered translation of GATA1 in Diamond-Blackfan anemia. Nature Medicine, 2014, 20, 748-753.	30.7	243
21	Approach to the diagnosis of congenital myopathies. Neuromuscular Disorders, 2014, 24, 97-116.	0.6	239
22	Nemaline myopathy: A clinical study of 143 cases. Annals of Neurology, 2001, 50, 312-320.	5.3	236
23	Nemaline Myopathy with Minicores Caused by Mutation of the CFL2 Gene Encoding the Skeletal Muscle Actinâ€“Binding Protein, Cofilin-2. American Journal of Human Genetics, 2007, 80, 162-167.	6.2	213
24	Abnormalities of the large ribosomal subunit protein, Rpl35a, in Diamond-Blackfan anemia. Blood, 2008, 112, 1582-1592.	1.4	208
25	Mutation of the Gene for I sK Associated With Both Jervell and Lange-Nielsen and Romano-Ward Forms of Long-QT Syndrome. Circulation, 1998, 97, 142-146.	1.6	205
26	The ribosomal basis of diamond-blackfan anemia: mutation and database update. Human Mutation, 2010, 31, 1269-1279.	2.5	202
27	Mutations and polymorphisms of the skeletal muscle ÃŽÂ±-actin gene (ACTA1). Human Mutation, 2009, 30, 1267-1277.	2.5	198
28	Directed evolution of a family of AAV capsid variants enabling potent muscle-directed gene delivery across species. Cell, 2021, 184, 4919-4938.e22.	28.9	193
29	Muscle disease caused by mutations in the skeletal muscle alpha-actin gene (ACTA1). Neuromuscular Disorders, 2003, 13, 519-531.	0.6	192
30	Mutations in KLHL40 Are a Frequent Cause of Severe Autosomal-Recessive Nemaline Myopathy. American Journal of Human Genetics, 2013, 93, 6-18.	6.2	186
31	The Genetic Landscape of Diamond-Blackfan Anemia. American Journal of Human Genetics, 2018, 103, 930-947.	6.2	184
32	MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. American Journal of Human Genetics, 2017, 100, 843-853.	6.2	181
33	Recessive truncating titin gene, TTN , mutations presenting as centronuclear myopathy. Neurology, 2013, 81, 1205-1214.	1.1	177
34	Interpretation of Genomic Sequencing Results in Healthy and Ill Newborns: Results from the BabySeq Project. American Journal of Human Genetics, 2019, 104, 76-93.	6.2	176
35	Newborn Sequencing in Genomic Medicine and Public Health. Pediatrics, 2017, 139, .	2.1	174
36	Evidence by molecular profiling for a placental origin of infantile hemangioma. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 19097-19102.	7.1	170

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37	T-tubule disorganization and defective excitation-contraction coupling in muscle fibers lacking myotubularin lipid phosphatase. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 18763-18768.	7.1	167
38	Exome Sequencing and Functional Validation in Zebrafish Identify GTDC2 Mutations as a Cause of Walker-Warburg Syndrome. American Journal of Human Genetics, 2012, 91, 541-547.	6.2	167
39	Oculomotor nerve and muscle abnormalities in congenital fibrosis of the extraocular muscles. Annals of Neurology, 1997, 41, 314-325.	5.3	165
40	Differential Regional Expression and Ultrastructural Localization of Î±-Actinin-2, a Putative NMDA Receptor-Anchoring Protein, in Rat Brain. Journal of Neuroscience, 1998, 18, 1383-1392.	3.6	164
41	Expression profiling and identification of novel genes involved in myogenic differentiation. FASEB Journal, 2004, 18, 1-23.	0.5	157
42	Leiomodin-3 dysfunction results in thin filament disorganization and nemaline myopathy. Journal of Clinical Investigation, 2014, 124, 4693-4708.	8.2	153
43	Identification of KLHL41 Mutations Implicates BTB-Kelch-Mediated Ubiquitination as an Alternate Pathway to Myofibrillar Disruption in Nemaline Myopathy. American Journal of Human Genetics, 2013, 93, 1108-1117.	6.2	147
44	Clinical and genetic heterogeneity in nemaline myopathy â€“ a disease of skeletal muscle thin filaments. Trends in Molecular Medicine, 2001, 7, 362-368.	6.7	145
45	SPEG Interacts with Myotubularin, and Its Deficiency Causes Centronuclear Myopathy with Dilated Cardiomyopathy. American Journal of Human Genetics, 2014, 95, 218-226.	6.2	143
46	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	6.2	142
47	Gene Therapy Prolongs Survival and Restores Function in Murine and Canine Models of Myotubular Myopathy. Science Translational Medicine, 2014, 6, 220ra10.	12.4	141
48	Selenoproteins and Their Impact on Human Health Through Diverse Physiological Pathways. Physiology, 2006, 21, 307-315.	3.1	136
49	Heterogeneity of nemaline myopathy cases with skeletal muscle Î±â€actin gene mutations. Annals of Neurology, 2004, 56, 86-96.	5.3	135
50	Frameshift mutation in p53 regulatorRPL26is associated with multiple physical abnormalities and a specific pre-ribosomal RNA processing defect in diamond-blackfan anemia. Human Mutation, 2012, 33, 1037-1044.	2.5	135
51	Myotubularin controls desmin intermediate filament architecture and mitochondrial dynamics in human and mouse skeletal muscle. Journal of Clinical Investigation, 2011, 121, 70-85.	8.2	132
52	Deficiency of a skeletal muscle isoform of Î±-actinin (Î±-actinin-3) in merosin-positive congenital muscular dystrophy. Neuromuscular Disorders, 1996, 6, 229-235.	0.6	127
53	Thin filament length dysregulation contributes to muscle weakness in nemaline myopathy patients with nebulin deficiency. Human Molecular Genetics, 2009, 18, 2359-2369.	2.9	124
54	Kelch proteins: emerging roles in skeletal muscle development and diseases. Skeletal Muscle, 2014, 4, 11.	4.2	119

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55	AAV-mediated intramuscular delivery of myotubularin corrects the myotubular myopathy phenotype in targeted murine muscle and suggests a function in plasma membrane homeostasis. Human Molecular Genetics, 2008, 17, 2132-2143.	2.9	115
56	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
57	The BabySeq project: implementing genomic sequencing in newborns. BMC Pediatrics, 2018, 18, 225.	1.7	115
58	MTM1 mutation associated with X-linked myotubular myopathy in Labrador Retrievers. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 14697-14702.	7.1	114
59	Genotype?phenotype correlations in nemaline myopathy caused by mutations in the genes for nebulin and skeletal muscle ?-actin. Neuromuscular Disorders, 2004, 14, 461-470.	0.6	107
60	Mutation Update: The Spectra of Nebulin Variants and Associated Myopathies. Human Mutation, 2014, 35, 1418-1426.	2.5	107
61	Mapping a gene for congenital fibrosis of the extraocular muscles to the centromeric region of chromosome 12. Nature Genetics, 1994, 7, 69-73.	21.4	104
62	The zebrafish dag1 mutant: a novel genetic model for dystroglycanopathies. Human Molecular Genetics, 2011, 20, 1712-1725.	2.9	101
63	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
64	Novel deletion of RPL15 identified by array-comparative genomic hybridization in Diamondâ€“Blackfan anemia. Human Genetics, 2013, 132, 1265-1274.	3.8	97
65	RNA and protein evidence for haplo-insufficiency in Diamond-Blackfan anaemia patients with RPS19 mutations. British Journal of Haematology, 2004, 127, 105-113.	2.5	96
66	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137.	6.2	96
67	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	5.3	93
68	Mutation Update and Genotype-Phenotype Correlations of Novel and Previously Described Mutations inTPM2andTPM3Causing Congenital Myopathies. Human Mutation, 2014, 35, 779-790.	2.5	92
69	Preservation of the C-terminus of dystrophin molecule in the skeletal muscle from Becker muscular dystrophy. Journal of the Neurological Sciences, 1991, 101, 148-156.	0.6	91
70	X-Linked Myotubular and Centronuclear Myopathies. Journal of Neuropathology and Experimental Neurology, 2005, 64, 555-564.	1.7	90
71	Congenital Fibrosis of the Extraocular Muscles Type 2, an Inherited Exotropic Strabismus Fixus, Maps to Distal 11q13. American Journal of Human Genetics, 1998, 63, 517-525.	6.2	89
72	Altered myofilament function depresses force generation in patients with nebulin-based nemaline myopathy (NEM2). Journal of Structural Biology, 2010, 170, 334-343.	2.8	87

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73	Drug discovery for Diamond-Blackfan anemia using reprogrammed hematopoietic progenitors. Science Translational Medicine, 2017, 9, .	12.4	87
74	A polymorphic CACA repeat in the 3â€² untranslated region of dystrophin. Nucleic Acids Research, 1990, 18, 1931-1931.	14.5	85
75	Clinical and genetic heterogeneity in autosomal recessive nemaline myopathy. Neuromuscular Disorders, 1999, 9, 564-572.	0.6	84
76	Evidence for linkage of familial Diamond-Blackfan anemia to chromosome 8p23.3-p22 and for non-19q non-8p disease. Blood, 2001, 97, 2145-2150.	1.4	84
77	Dominant Mutation of CCDC78 in a Unique Congenital Myopathy with Prominent Internal Nuclei and Atypical Cores. American Journal of Human Genetics, 2012, 91, 365-371.	6.2	84
78	Prospective, phenotype-driven selection of critically ill neonates for rapid exome sequencing is associated with high diagnostic yield. Genetics in Medicine, 2020, 22, 736-744.	2.4	83
79	Gene expression profiling of Duchenne muscular dystrophy skeletal muscle. Neurogenetics, 2003, 4, 163-171.	1.4	82
80	Effect of ageing on reactivation of the human X-linked HPRT locus. Nature, 1988, 335, 93-96.	27.8	81
81	Systemic AAV8-Mediated Gene Therapy Drives Whole-Body Correction of Myotubular Myopathy in Dogs. Molecular Therapy, 2017, 25, 839-854.	8.2	81
82	Normal myofibrillar development followed by progressive sarcomeric disruption with actin accumulations in a mouse Cfl2 knockout demonstrates requirement of cofilin-2 for muscle maintenance. Human Molecular Genetics, 2012, 21, 2341-2356.	2.9	80
83	Mutation of KCNJ8 in a patient with CantÃº syndrome with unique vascular abnormalities â€“ Support for the role of K(ATP) channels in this condition. European Journal of Medical Genetics, 2013, 56, 678-682.	1.3	79
84	Novel Mutations Widen the Phenotypic Spectrum of Slow Skeletal/Î²-Cardiac Myosin (MYH7) Distal Myopathy. Human Mutation, 2014, 35, 868-879.	2.5	79
85	A curated gene list for reporting results of newborn genomic sequencing. Genetics in Medicine, 2017, 19, 809-818.	2.4	79
86	Defective Ribosomal Protein Gene Expression Alters Transcription, Translation, Apoptosis, and Oncogenic Pathways in Diamond-Blackfan Anemia. Stem Cells, 2006, 24, 2034-2044.	3.2	75
87	A natural history study of X-linked myotubular myopathy. Neurology, 2017, 89, 1355-1364.	1.1	75
88	Enzyme replacement therapy rescues weakness and improves muscle pathology in mice with X-linked myotubular myopathy. Human Molecular Genetics, 2013, 22, 1525-1538.	2.9	71
89	Mutations of tropomyosin 3 (TPM3) are common and associated with type 1 myofiber hypotrophy in congenital fiber type disproportion. Human Mutation, 2010, 31, 176-183.	2.5	70
90	Human Skeletal Muscle-Specific Î±-Actinin-2 and -3 Isoforms Form Homodimers and Heterodimersin Vitroandin Vivo. Biochemical and Biophysical Research Communications, 1998, 248, 134-139.	2.1	69

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91	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
92	Expression profiling reveals altered satellite cell numbers and glycolytic enzyme transcription in nemaline myopathy muscle. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 4666-4671.	7.1	68
93	Mutations in the satellite cell gene MEGF10 cause a recessive congenital myopathy with minicores. Neurogenetics, 2012, 13, 115-124.	1.4	68
94	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 991-999.	6.2	68
95	Reproducibility of gene expression across generations of Affymetrix microarrays. BMC Bioinformatics, 2003, 4, 27.	2.6	67
96	ClinPhen extracts and prioritizes patient phenotypes directly from medical records to expedite genetic disease diagnosis. Genetics in Medicine, 2019, 21, 1585-1593.	2.4	67
97	Isoform Cloning, Actin Binding, and Chromosomal Localization of Human Erythroid Dematin, a Member of the Villin Superfamily. Journal of Biological Chemistry, 1995, 270, 17407-17413.	3.4	66
98	Inhibition of Activin Receptor Type IIB Increases Strength and Lifespan in Myotubularin-Deficient Mice. American Journal of Pathology, 2011, 178, 784-793.	3.8	63
99	Parental interest in genomic sequencing of newborns: enrollment experience from the BabySeq Project. Genetics in Medicine, 2019, 21, 622-630.	2.4	61
100	Human pituitary adenomas show no loss of heterozygosity at the retinoblastoma gene locus. Journal of Clinical Endocrinology and Metabolism, 1994, 78, 922-927.	3.6	61
101	Changes in cross-bridge cycling underlie muscle weakness in patients with tropomyosin 3-based myopathy. Human Molecular Genetics, 2011, 20, 2015-2025.	2.9	60
102	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	2.4	60
103	Unique bioinformatic approach and comprehensive reanalysis improve diagnostic yield of clinical exomes. European Journal of Human Genetics, 2019, 27, 1398-1405.	2.8	60
104	AMELIE speeds Mendelian diagnosis by matching patient phenotype and genotype to primary literature. Science Translational Medicine, 2020, 12, .	12.4	60
105	Skeletal Muscle Pathology in X-Linked Myotubular Myopathy: Review With Cross-Species Comparisons. Journal of Neuropathology and Experimental Neurology, 2016, 75, 102-110.	1.7	59
106	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
107	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
108	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

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109	Multiple different missense mutations in the pore region of HERG in patients with long QT syndrome. Human Genetics, 1998, 102, 265-272.	3.8	57
110	Cell Membrane Expression of Cardiac Sodium Channel Na_v1.5 Is Modulated by Î±-Actinin-2 Interaction. Biochemistry, 2010, 49, 166-178.	2.5	57
111	The Pediatric Cell Atlas: Defining the Growth Phase of Human Development at Single-Cell Resolution. Developmental Cell, 2019, 49, 10-29.	7.0	57
112	Analysis of Skeletal Muscle Defects in Larval Zebrafish by Birefringence and Touch-evoke Escape Response Assays. Journal of Visualized Experiments, 2013, , e50925.	0.3	56
113	Deleting exon 55 from the nebulin gene induces severe muscle weakness in a mouse model for nemaline myopathy. Brain, 2013, 136, 1718-1731.	7.6	55
114	Filamin C accumulation is a strong but nonspecific immunohistochemical marker of core formation in muscle. Journal of the Neurological Sciences, 2003, 206, 71-78.	0.6	54
115	The exon 55 deletion in the nebulin gene â€“ One single founder mutation with world-wide occurrence. Neuromuscular Disorders, 2009, 19, 179-181.	0.6	54
116	Modeling the human MTM1 p.R69C mutation in murine Mtm1 results in exon 4 skipping and a less severe myotubular myopathy phenotype. Human Molecular Genetics, 2012, 21, 811-825.	2.9	54
117	Mutationâ€specific effects on thin filament length in thin filament myopathy. Annals of Neurology, 2016, 79, 959-969.	5.3	54
118	A multicenter, retrospective medical record review of Xâ€linked myotubular myopathy: The recensus study. Muscle and Nerve, 2018, 57, 550-560.	2.2	54
119	Artificial intelligence enables comprehensive genome interpretation and nomination of candidate diagnoses for rare genetic diseases. Genome Medicine, 2021, 13, 153.	8.2	53
120	Novel mutations in NEB cause abnormal nebulin expression and markedly impaired muscle force generation in severe nemaline myopathy. Skeletal Muscle, 2011, 1, 23.	4.2	51
121	Longâ€term effects of systemic gene therapy in a canine model of myotubular myopathy. Muscle and Nerve, 2017, 56, 943-953.	2.2	50
122	Telethonin protein expression in neuromuscular disorders. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2002, 1588, 33-40.	3.8	49
123	Congenital myopathy caused by a novel missense mutation in the CFL2 gene. Neuromuscular Disorders, 2012, 22, 632-639.	0.6	49
124	Sodium channel abnormalities are infrequent in patients with long QT Syndrome: Identification of two novelSCN5A mutations. , 1999, 86, 470-476.		48
125	Tissue Triage and Freezing for Models of Skeletal Muscle Disease. Journal of Visualized Experiments, 2014, , .	0.3	48
126	A Splice Site Mutation in Laminin-Î±2 Results in a Severe Muscular Dystrophy and Growth Abnormalities in Zebrafish. PLoS ONE, 2012, 7, e43794.	2.5	48

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127	Perceived Benefits, Risks, and Utility of Newborn Genomic Sequencing in the BabySeq Project. Pediatrics, 2019, 143, S6-S13.	2.1	47
128	Discordant results between conventional newborn screening and genomic sequencing in the BabySeq Project. Genetics in Medicine, 2021, 23, 1372-1375.	2.4	47
129	Dystrophinopathy, The Expanding Phenotype. Circulation, 1997, 95, 2344-2347.	1.6	47
130	Troponin activator augments muscle force in nemaline myopathy patients with nebulin mutations. Journal of Medical Genetics, 2013, 50, 383-392.	3.2	46
131	X-linked myotubular myopathy in Rottweiler dogs is caused by a missense mutation in Exon 11 of the MTM1 gene. Skeletal Muscle, 2015, 5, 1.	4.2	46
132	A novel de novo mutation inATP1A3and childhood-onset schizophrenia. Journal of Physical Education and Sports Management, 2016, 2, a001008.	1.2	46
133	Side Population cells isolated from different tissues share transcriptome signatures and express tissue-specific markers. Experimental Cell Research, 2005, 303, 360-374.	2.6	45
134	Functional muscle analysis of the Tcap knockout mouse. Human Molecular Genetics, 2010, 19, 2268-2283.	2.9	45
135	Returning a Genomic Result for an Adult-Onset Condition to the Parents of a Newborn: Insights From the BabySeq Project. Pediatrics, 2019, 143, S37-S43.	2.1	45
136	Novel actin crosslinker superfamily member identified by a two step degenerate PCR procedure. FEBS Letters, 1995, 368, 500-504.	2.8	44
137	The Mouse Region Syntenic for Human Spinal Muscular Atrophy Lies within theLgn1Critical Interval and Contains Multiple Copies ofNaipExon 5. Genomics, 1996, 38, 405-417.	2.9	44
138	Skeletal muscle repair in a mouse model of nemaline myopathy. Human Molecular Genetics, 2006, 15, 2603-2612.	2.9	44
139	SLC6A1 Mutation and Ketogenic Diet in Epilepsy With Myoclonic-Atonic Seizures. Pediatric Neurology, 2016, 64, 77-79.	2.1	44
140	Ddx18 is essential for cell-cycle progression in zebrafish hematopoietic cells and is mutated in human AML. Blood, 2011, 118, 903-915.	1.4	43
141	Myotubular myopathy and the neuromuscular junction: a novel therapeutic approach from mouse models. DMM Disease Models and Mechanisms, 2012, 5, 852-9.	2.4	43
142	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
143	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
144	Fastâ€twitch sarcomeric and glycolytic enzyme protein loss in inclusion body myositis. Muscle and Nerve, 2009, 39, 739-753.	2.2	41

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145	Î±-Actinin-2 Is a New Component of the Dystrophinâ€“Glycoprotein Complex. Archives of Biochemistry and Biophysics, 1999, 365, 216-222.	3.0	40
146	Adult-Onset Nemaline Myopathy and Monoclonal Gammopathy. Archives of Neurology, 2006, 63, 132.	4.5	39
147	Myofiber size correlates with MTM1 mutation type and outcome in X-linked myotubular myopathy. Neuromuscular Disorders, 2007, 17, 562-568.	0.6	39
148	Transcriptional profile of postmortem skeletal muscle. Physiological Genomics, 2004, 16, 222-228.	2.3	38
149	A compound heterozygous mutation in GPD1 causes hepatomegaly, steatohepatitis, and hypertriglyceridemia. European Journal of Human Genetics, 2014, 22, 1229-1232.	2.8	38
150	Muscle weakness inTPM3-myopathy is due to reduced Ca²⁺-sensitivity and impaired acto-myosin cross-bridge cycling in slow fibres. Human Molecular Genetics, 2015, 24, 6278-6292.	2.9	38
151	Childrenâ€™s rare disease cohorts: an integrative research and clinical genomics initiative. Npj Genomic Medicine, 2020, 5, 29.	3.8	38
152	Sarcomeres regulate murine cardiomyocyte maturation through MRTF-SRF signaling. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	38
153	Myotubularin-Deficient Myoblasts Display Increased Apoptosis, Delayed Proliferation, and Poor Cell Engraftment. American Journal of Pathology, 2012, 181, 961-968.	3.8	37
154	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
155	The influence of muscle type and dystrophin deficiency on murine expression profiles. Mammalian Genome, 2005, 16, 739-748.	2.2	35
156	Association of a NovelACTA1Mutation With a Dominant Progressive Scapuloperoneal Myopathy in an Extended Family. JAMA Neurology, 2015, 72, 689.	9.0	35
157	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. American Journal of Human Genetics, 2017, 100, 343-351.	6.2	35
158	Psychosocial Effect of Newborn Genomic Sequencing on Families in the BabySeq Project. JAMA Pediatrics, 2021, 175, 1132.	6.2	35
159	Melanoma cell adhesion molecule is a novel marker for human fetal myogenic cells and affects myoblast fusion. Journal of Cell Science, 2006, 119, 3117-3127.	2.0	34
160	Acute appendicitis is characterized by a uniform and highly selective pattern of inflammatory gene expression. Mucosal Immunology, 2008, 1, 297-308.	6.0	34
161	Î±â€Actininâ€2 deficiency results in sarcomeric defects in zebrafish that cannot be rescued by Î±â€actininâ€3 revealing functional differences between sarcomeric isoforms. FASEB Journal, 2012, 26, 1892-1908.	0.5	34
162	Selenoprotein N deficiency in mice is associated with abnormal lung development. FASEB Journal, 2013, 27, 1585-1599.	0.5	34

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163	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. Genetics in Medicine, 2021, 23, 1028-1040.	2.4	34
164	RNA helicase, DDX27 regulates skeletal muscle growth and regeneration by modulation of translational processes. PLoS Genetics, 2018, 14, e1007226.	3.5	34
165	Whole Exome Sequencing IdentifiesRAI1Mutation in a Morbidly Obese Child Diagnosed With ROHHAD Syndrome. Journal of Clinical Endocrinology and Metabolism, 2015, 100, 1723-1730.	3.6	33
166	Expanding the Phenotype Associated With theNEFLMutation. JAMA Neurology, 2014, 71, 1413.	9.0	30
167	The Genomics Research and Innovation Network: creating an interoperable, federated, genomics learning system. Genetics in Medicine, 2020, 22, 371-380.	2.4	30
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