

# Arthur H M Burghes

## List of Publications by Year in descending order

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149  
papers

18,078  
citations

20036

63  
h-index

14386

132  
g-index

154  
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154  
docs citations

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times ranked

11458  
citing authors

#	ARTICLE	IF	CITATIONS
1	Conditional deletion of SMN in cell culture identifies functional SMN alleles. <i>Human Molecular Genetics</i> , 2021, 29, 3477-3492.	1.4	9
2	Intragenic complementation of amino and carboxy terminal SMN missense mutations can rescue Smn null mice. <i>Human Molecular Genetics</i> , 2021, 29, 3493-3503.	1.4	4
3	Early Inflammation in Muscular Dystrophy Differs between Limb and Respiratory Muscles and Increases with Dystrophic Severity. <i>American Journal of Pathology</i> , 2021, 191, 730-747.	1.9	13
4	Voluntary wheel running with and without follistatin overexpression improves NMJ transmission but not motor unit loss in late life of C57BL/6J mice. <i>Neurobiology of Aging</i> , 2021, 101, 285-296.	1.5	5
5	Follistatin-induced muscle hypertrophy in aged mice improves neuromuscular junction innervation and function. <i>Neurobiology of Aging</i> , 2021, 104, 32-41.	1.5	11
6	What Genetics Has Told Us and How It Can Inform Future Experiments for Spinal Muscular Atrophy, a Perspective. <i>International Journal of Molecular Sciences</i> , 2021, 22, 8494.	1.8	6
7	Persistent neuromuscular junction transmission defects in adults with spinal muscular atrophy treated with nusinersen. <i>BMJ Neurology Open</i> , 2021, 3, e000164.	0.7	16
8	Dual SMN inducing therapies can rescue survival and motor unit function in symptomatic $\alpha^1$ 7SMA mice. <i>Neurobiology of Disease</i> , 2021, 159, 105488.	2.1	14
9	Biodistribution of onasemnogene abeparvovec DNA, mRNA and SMN protein in human tissue. <i>Nature Medicine</i> , 2021, 27, 1701-1711.	15.2	49
10	244th ENMC international workshop: Newborn screening in spinal muscular atrophy May 10-12, 2019, Hoofddorp, The Netherlands. <i>Neuromuscular Disorders</i> , 2020, 30, 93-103.	0.3	55
11	Complete sequencing of the SMN2 gene in SMA patients detects SMN gene deletion junctions and variants in SMN2 that modify the SMA phenotype. <i>Human Genetics</i> , 2019, 138, 241-256.	1.8	57
12	Muscle strength and size are associated with motor unit connectivity in aged mice. <i>Neurobiology of Aging</i> , 2018, 67, 128-136.	1.5	74
13	Mild SMN missense alleles are only functional in the presence of SMN2 in mammals. <i>Human Molecular Genetics</i> , 2018, 27, 3404-3416.	1.4	15
14	IP 853. AVXS-101 Phase-1-Gene Therapy Clinical Trial in SMA Type 1: Event-Free Survival and Achievement of Developmental Milestones. <i>Neuropediatrics</i> , 2018, 49, .	0.3	0
15	SMN Blood Levels in a Porcine Model of Spinal Muscular Atrophy. <i>Journal of Neuromuscular Diseases</i> , 2017, 4, 59-66.	1.1	9
16	Facilitating orphan drug development: Proceedings of the TREAT-NMD International Conference, December 2015, Washington, DC, USA. <i>Neuromuscular Disorders</i> , 2017, 27, 693-701.	0.3	1
17	Single-Dose Gene-Replacement Therapy for Spinal Muscular Atrophy. <i>New England Journal of Medicine</i> , 2017, 377, 1713-1722.	13.9	1,642
18	AVXS-101 Phase 1 gene therapy clinical trial in SMA Type 1: Interim data demonstrates improvements in supportive care use. <i>European Journal of Paediatric Neurology</i> , 2017, 21, e14.	0.7	4

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19	Natural history of infantile-onset spinal muscular atrophy. <i>Annals of Neurology</i> , 2017, 82, 883-891.	2.8	276
20	Variable phenotypic expression and onset in MYH14 distal hereditary motor neuropathy phenotype in a large, multigenerational North American family. <i>Muscle and Nerve</i> , 2017, 56, 341-345.	1.0	11
21	Spinal Muscular Atrophy. , 2017, , .		1
22	The neuromuscular impact of symptomatic SMN restoration in a mouse model of spinal muscular atrophy. <i>Neurobiology of Disease</i> , 2016, 87, 116-123.	2.1	42
23	Baseline results of the NeuroNEXT spinal muscular atrophy infant biomarker study. <i>Annals of Clinical and Translational Neurology</i> , 2016, 3, 132-145.	1.7	106
24	Protective effects of butyrate-based compounds on a mouse model for spinal muscular atrophy. <i>Experimental Neurology</i> , 2016, 279, 13-26.	2.0	25
25	Normalization of Patient-Identified Plasma Biomarkers in SMN <sup>0/7</sup> Mice following Postnatal SMN Restoration. <i>PLoS ONE</i> , 2016, 11, e0167077.	1.1	11
26	A large animal model of spinal muscular atrophy and correction of phenotype. <i>Annals of Neurology</i> , 2015, 77, 399-414.	2.8	114
27	SMN expression is required in motor neurons to rescue electrophysiological deficits in the SMN <sup>0/7</sup> mouse model of SMA. <i>Human Molecular Genetics</i> , 2015, 24, 5524-5541.	1.4	53
28	SMN deficiency disrupts gastrointestinal and enteric nervous system function in mice. <i>Human Molecular Genetics</i> , 2015, 24, 3847-3860.	1.4	44
29	Low levels of Survival Motor Neuron protein are sufficient for normal muscle function in the SMN <sup>0/7</sup> mouse model of SMA. <i>Human Molecular Genetics</i> , 2015, 24, 6160-6173.	1.4	39
30	Improving Single Injection CSF Delivery of AAV9-mediated Gene Therapy for SMA: A Dose-response Study in Mice and Nonhuman Primates. <i>Molecular Therapy</i> , 2015, 23, 477-487.	3.7	217
31	Plastin 3 Expression Does Not Modify Spinal Muscular Atrophy Severity in the <sup>0/7</sup> SMA Mouse. <i>PLoS ONE</i> , 2015, 10, e0132364.	1.1	41
32	A Novel Approach to Identify Genes Related to FGID. <i>FASEB Journal</i> , 2015, 29, 1002.16.	0.2	0
33	Direct conversion of patient fibroblasts demonstrates non-cell autonomous toxicity of astrocytes to motor neurons in familial and sporadic ALS. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 829-832.	3.3	296
34	Electrophysiological biomarkers in spinal muscular atrophy: proof of concept. <i>Annals of Clinical and Translational Neurology</i> , 2014, 1, 34-44.	1.7	55
35	Deletion of atrophy enhancing genes fails to ameliorate the phenotype in a mouse model of spinal muscular atrophy. <i>Neuromuscular Disorders</i> , 2014, 24, 436-444.	0.3	11
36	The effect of diet on the protective action of D156844 observed in spinal muscular atrophy mice. <i>Experimental Neurology</i> , 2014, 256, 1-6.	2.0	15

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37	The motor neuron response to <i>SMN1</i> deficiency in spinal muscular atrophy. <i>Muscle and Nerve</i> , 2014, 50, 457-458.	1.0	2
38	SMA-EUROPE workshop report: opportunities and challenges in developing clinical trials for spinal muscular atrophy in Europe. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 44.	1.2	17
39	Spinal muscular atrophy: Development and implementation of potential treatments. <i>Annals of Neurology</i> , 2013, 74, 348-362.	2.8	76
40	Antisense Oligonucleotides for the Treatment of Spinal Muscular Atrophy. <i>Human Gene Therapy</i> , 2013, 24, 489-498.	1.4	74
41	Improved Antisense Oligonucleotide Design to Suppress Aberrant SMN2 Gene Transcript Processing: Towards a Treatment for Spinal Muscular Atrophy. <i>PLoS ONE</i> , 2013, 8, e62114.	1.1	63
42	A Role for SMN Exon 7 Splicing in the Selective Vulnerability of Motor Neurons in Spinal Muscular Atrophy. <i>Molecular and Cellular Biology</i> , 2012, 32, 126-138.	1.1	98
43	A single administration of morpholino antisense oligomer rescues spinal muscular atrophy in mouse. <i>Human Molecular Genetics</i> , 2012, 21, 1625-1638.	1.4	226
44	The zinc finger protein ZPR1 is a potential modifier of spinal muscular atrophy. <i>Human Molecular Genetics</i> , 2012, 21, 2745-2758.	1.4	57
45	Astrocytes from familial and sporadic ALS patients are toxic to motor neurons. <i>Nature Biotechnology</i> , 2011, 29, 824-828.	9.4	696
46	Generation and Characterization of a genetic zebrafish model of SMA carrying the human SMN2 gene. <i>Molecular Neurodegeneration</i> , 2011, 6, 24.	4.4	41
47	Temporal requirement for high SMN expression in SMA mice. <i>Human Molecular Genetics</i> , 2011, 20, 3578-3591.	1.4	118
48	Systemic Gene Delivery in Large Species for Targeting Spinal Cord, Brain, and Peripheral Tissues for Pediatric Disorders. <i>Molecular Therapy</i> , 2011, 19, 1971-1980.	3.7	290
49	A genetic model of amyotrophic lateral sclerosis in zebrafish displays phenotypic hallmarks of motoneuron disease. <i>DMM Disease Models and Mechanisms</i> , 2010, 3, 652-662.	1.2	130
50	Antisense oligonucleotides and spinal muscular atrophy: skipping along: Figure 1.. <i>Genes and Development</i> , 2010, 24, 1574-1579.	2.7	33
51	Early heart failure in the SMN <sup>Δ7</sup> model of spinal muscular atrophy and correction by postnatal scAAV9-SMN delivery. <i>Human Molecular Genetics</i> , 2010, 19, 3895-3905.	1.4	192
52	Effects of 2,4-diaminoquinazoline derivatives on SMN expression and phenotype in a mouse model for spinal muscular atrophy. <i>Human Molecular Genetics</i> , 2010, 19, 454-467.	1.4	110
53	A SMN missense mutation complements SMN2 restoring snRNPs and rescuing SMA mice. <i>Human Molecular Genetics</i> , 2009, 18, 2215-2229.	1.4	97
54	Spinal muscular atrophy: why do low levels of survival motor neuron protein make motor neurons sick?. <i>Nature Reviews Neuroscience</i> , 2009, 10, 597-609.	4.9	632

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55	A Positive Modifier of Spinal Muscular Atrophy in the SMN2 Gene. American Journal of Human Genetics, 2009, 85, 408-413.	2.6	253
56	The SMN binding protein gemin2 is not involved in motor axon outgrowth. Developmental Neurobiology, 2008, 68, 182-194.	1.5	37
57	Other forms of survival motor neuron protein and spinal muscular atrophy: An opinion. Neuromuscular Disorders, 2008, 18, 82-83.	0.3	5
58	Synthesis and Biological Evaluation of Novel 2,4-Diaminoquinazoline Derivatives as SMN2 Promoter Activators for the Potential Treatment of Spinal Muscular Atrophy. Journal of Medicinal Chemistry, 2008, 51, 449-469.	2.9	88
59	Neuronal SMN expression corrects spinal muscular atrophy in severe SMA mice while muscle-specific SMN expression has no phenotypic effect. Human Molecular Genetics, 2008, 17, 1063-1075.	1.4	199
60	Embryonic motor axon development in the severe SMA mouse. Human Molecular Genetics, 2008, 17, 2900-2909.	1.4	136
61	Protein phosphatase 1 binds to the RNA recognition motif of several splicing factors and regulates alternative pre-mRNA processing. Human Molecular Genetics, 2008, 17, 52-70.	1.4	76
62	Let all DNA vote. Neurology, 2008, 70, 662-663.	1.5	3
63	The human centromeric survival motor neuron gene (SMN2) rescues embryonic lethality in Smn <sup>-/-</sup> mice and results in a mouse with spinal muscular atrophy. Human Molecular Genetics, 2007, 16, 2648-2648.	1.4	1
64	Ribonucleoprotein Assembly Defects Correlate with Spinal Muscular Atrophy Severity and Preferentially Affect a Subset of Spliceosomal snRNPs. PLoS ONE, 2007, 2, e921.	1.1	266
65	Protein- and mRNA-based phenotype-genotype correlations in DMD/BMD with point mutations and molecular basis for BMD with nonsense and frameshift mutations in the DMD gene. Human Mutation, 2007, 28, 183-195.	1.1	107
66	A novel method for oral delivery of drug compounds to the neonatal SMN <sup>Δ7</sup> mouse model of spinal muscular atrophy. Journal of Neuroscience Methods, 2007, 161, 285-290.	1.3	37
67	Absence of gemin5 from SMN complexes in nuclear Cajal bodies. BMC Cell Biology, 2007, 8, 28.	3.0	44
68	Abnormal motor phenotype in the SMN <sup>Δ7</sup> mouse model of spinal muscular atrophy. Neurobiology of Disease, 2007, 27, 207-219.	2.1	96
69	Survival Motor Neuron Function in Motor Axons Is Independent of Functions Required for Small Nuclear Ribonucleoprotein Biogenesis. Journal of Neuroscience, 2006, 26, 11014-11022.	1.7	156
70	Dystrophin glycoprotein complex dysfunction: A regulatory link between muscular dystrophy and cancer cachexia. Cancer Cell, 2005, 8, 421-432.	7.7	260
71	SMN <sup>Δ7</sup> , the major product of the centromeric survival motor neuron (SMN2) gene, extends survival in mice with spinal muscular atrophy and associates with full-length SMN. Human Molecular Genetics, 2005, 14, 845-857.	1.4	550
72	Diverse small-molecule modulators of SMN expression found by high-throughput compound screening: early leads towards a therapeutic for spinal muscular atrophy. Human Molecular Genetics, 2005, 14, 2003-2018.	1.4	143

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73	A role for complexes of survival of motor neurons (SMN) protein with gemins and profilin in neurite-like cytoplasmic extensions of cultured nerve cells. <i>Experimental Cell Research</i> , 2005, 309, 185-197.	1.2	118
74	Identification of a Novel Cyclic AMP-response Element (CRE-II) and the Role of CREB-1 in the cAMP-induced Expression of the Survival Motor Neuron (SMN) Gene. <i>Journal of Biological Chemistry</i> , 2004, 279, 14803-14811.	1.6	52
75	Indoprofen Upregulates the Survival Motor Neuron Protein through a Cyclooxygenase-Independent Mechanism. <i>Chemistry and Biology</i> , 2004, 11, 1489-1493.	6.2	135
76	Perspectives on models of spinal muscular atrophy for drug discovery. <i>Drug Discovery Today: Disease Models</i> , 2004, 1, 151-156.	1.2	13
77	Lentivector-mediated SMN replacement in a mouse model of spinal muscular atrophy. <i>Journal of Clinical Investigation</i> , 2004, 114, 1726-1731.	3.9	183
78	Valproic acid increases SMN levels in spinal muscular atrophy patient cells. <i>Annals of Neurology</i> , 2003, 54, 647-654.	2.8	269
79	A transgene carrying an A2G missense mutation in the SMN gene modulates phenotypic severity in mice with severe (type I) spinal muscular atrophy. <i>Journal of Cell Biology</i> , 2003, 160, 41-52.	2.3	140
80	Knockdown of the survival motor neuron (Smn) protein in zebrafish causes defects in motor axon outgrowth and pathfinding. <i>Journal of Cell Biology</i> , 2003, 162, 919-932.	2.3	387
81	Molecular analysis of spinal muscular atrophy and modification of the phenotype by SMN2. <i>Genetics in Medicine</i> , 2002, 4, 20-26.	1.1	296
82	Expression of the survival of motor neuron (SMN) gene in primary neurons and increase in SMN levels by activation of the N-methyl-D-aspartate glutamate receptor. <i>Neurogenetics</i> , 2002, 4, 29-36.	0.7	16
83	Spinal muscular atrophies. <i>Advances in Neurology</i> , 2002, 88, 83-98.	0.8	4
84	Nuclear Gems and Cajal (Coiled) Bodies in Fetal Tissues: Nucleolar Distribution of the Spinal Muscular Atrophy Protein, SMN. <i>Experimental Cell Research</i> , 2001, 265, 252-261.	1.2	126
85	Hybrids monosomal for human chromosome 5 reveal the presence of a spinal muscular atrophy (SMA) carrier with two SMN1 copies on one chromosome. <i>Human Genetics</i> , 2001, 108, 109-115.	1.8	38
86	GENETICS: The Land Between Mendelian and Multifactorial Inheritance. <i>Science</i> , 2001, 293, 2213-2214.	6.0	72
87	Genomic organization and alternative splicing of the human and mouse RPTP $\beta$ -genes. <i>BMC Genomics</i> , 2001, 2, 1.	1.2	21
88	Use of western immunoblot for evaluation of myocardial dystrophin, -sarcoglycan, and -dystroglycan in dogs with idiopathic dilated cardiomyopathy. <i>American Journal of Veterinary Research</i> , 2001, 62, 67-71.	0.3	8
89	The survival motor neuron (SMN) protein: effect of exon loss and mutation on protein localization. <i>Neurogenetics</i> , 2000, 3, 7-16.	0.7	35
90	The Relationship between SMN, the Spinal Muscular Atrophy Protein, and Nuclear Coiled Bodies in Differentiated Tissues and Cultured Cells. <i>Experimental Cell Research</i> , 2000, 256, 365-374.	1.2	183

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91	Identification of survival motor neuron as a transcriptional activator-binding protein. Human Molecular Genetics, 1999, 8, 1219-1226.	1.4	98
92	Analysis of mutations in the tudor domain of the survival motor neuron protein SMN. European Journal of Human Genetics, 1999, 7, 519-525.	1.4	24
93	Promoter analysis of the human centromeric and telomeric survival motor neuron genes (SMNC and Tj ETQq1 1 0.784314 rgBT /Ove	2.4	77
94	A single nucleotide difference that alters splicing patterns distinguishes the SMA gene SMN1 from the copy gene SMN2. Human Molecular Genetics, 1999, 8, 1177-1183.	1.4	806
95	Genomic Organization and Biological Characterization of the Novel Human CC Chemokine DC-CK-1/PARC/MIP-4/SCYA18. Genomics, 1999, 56, 296-302.	1.3	45
96	SMN oligomerization defect correlates with spinal muscular atrophy severity. Nature Genetics, 1998, 19, 63-66.	9.4	470
97	Novel receptor protein tyrosine phosphatase (RPTP?) and acidic fibroblast growth factor (FGF-1) transcripts delineate a rostrocaudal boundary in the granule cell layer of the murine cerebellar cortex. , 1998, 391, 444-455.		33
98	Identification and characterization of RPTP $\beta$ , a novel RPTP $\beta$ -like receptor protein tyrosine phosphatase whose expression is restricted to the central nervous system. Molecular Brain Research, 1998, 56, 9-21.	2.5	26
99	Reply to Mackenzie. American Journal of Human Genetics, 1998, 62, 486-488.	2.6	0
100	DAX1 Mutations Map to Putative Structural Domains in a Deduced Three-Dimensional Model. American Journal of Human Genetics, 1998, 62, 855-864.	2.6	91
101	Intragenic telSMN Mutations: Frequency, Distribution, Evidence of a Founder Effect, and Modification of the Spinal Muscular Atrophy Phenotype by cenSMN Copy Number. American Journal of Human Genetics, 1998, 63, 1712-1723.	2.6	168
102	The survival motor neuron protein in spinal muscular atrophy. Human Molecular Genetics, 1997, 6, 1205-1214.	1.4	604
103	When Is a Deletion Not a Deletion? When It Is Converted. American Journal of Human Genetics, 1997, 61, 9-15.	2.6	254
104	Identification of Proximal Spinal Muscular Atrophy Carriers and Patients by Analysis of SMNT and SMNC Gene Copy Number. American Journal of Human Genetics, 1997, 60, 1411-1422.	2.6	494
105	Molecular diagnosis of non-deletion SMA patients using quantitative PCR of SMN exon 7. Neurogenetics, 1997, 1, 141-147.	0.7	54
106	Deletion and conversion in spinal muscular atrophy patients: Is there a relationship to severity?. Annals of Neurology, 1997, 41, 230-237.	2.8	98
107	Nonsense mutations in a Becker muscular dystrophy and an intermediate patient. , 1996, 7, 72-75.		10
108	An 11 base pair duplication in exon 6 of the SMN gene produces a type I spinal muscular atrophy (SMA) phenotype: further evidence for SMN as the primary SMA-determining gene. Human Molecular Genetics, 1996, 5, 1727-1732.	1.4	123

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109	A novel cDNA detects homozygous microdeletions in greater than 50% of type I spinal muscular atrophy patients. <i>Nature Genetics</i> , 1995, 9, 56-62.	9.4	83
110	Myoblast Transfer in the Treatment of Duchenne's Muscular Dystrophy. <i>New England Journal of Medicine</i> , 1995, 333, 832-838.	13.9	489
111	Allelic association and deletions in autosomal recessive proximal spinal muscular atrophy: association of marker genotype with disease severity and candidate cDNAs. <i>Human Molecular Genetics</i> , 1995, 4, 1273-1284.	1.4	101
112	Refined physical map of the spinal muscular atrophy gene (SMA) region at 5q13 based on YAC and cosmid contiguous arrays. <i>Genomics</i> , 1995, 26, 451-460.	1.3	39
113	Mapping of the Spinal Muscular Atrophy (SMA) Gene to a 750-kb Interval Flanked by Two New Microsatellites. <i>European Journal of Human Genetics</i> , 1995, 3, 56-60.	1.4	19
114	Identification of a missense mutation, single base deletion and a polymorphism in the dystrophin exon 16. <i>Human Molecular Genetics</i> , 1994, 3, 1173-1174.	1.4	11
115	Linkage mapping of the spinal muscular atrophy gene. <i>Human Genetics</i> , 1994, 93, 305-312.	1.8	43
116	A Multicopy Dinucleotide Marker That Maps Close to the Spinal Muscular Atrophy Gene. <i>Genomics</i> , 1994, 21, 394-402.	1.3	54
117	A YAC Contig of the Region Containing the Spinal Muscular Atrophy Gene (SMA): Identification of an Unstable Region. <i>Genomics</i> , 1994, 24, 351-356.	1.3	45
118	Two 5q13 simple tandem repeat loci are in linkage disequilibrium with Type 1 spinal muscular atrophy. <i>Human Molecular Genetics</i> , 1994, 3, 1951-1956.	1.4	35
119	Gene therapy for muscle diseases. <i>Current Opinion in Neurology</i> , 1994, 7, 463-470.	1.8	8
120	Immunohistochemical analysis of dystrophin-associated proteins in Becker/Duchenne muscular dystrophy with huge in-frame deletions in the NH2-terminal and rod domains of dystrophin.. <i>Journal of Clinical Investigation</i> , 1994, 93, 99-105.	3.9	46
121	Exon 44 nonsense mutation in two-duchenne muscular dystrophy brothers detected by heteroduplex analysis. <i>Human Mutation</i> , 1993, 2, 192-195.	1.1	20
122	A missense mutation in the dystrophin gene in a Duchenne muscular dystrophy patient. <i>Nature Genetics</i> , 1993, 4, 357-360.	9.4	77
123	Gene delivery to spinal motor neurons. <i>Brain Research</i> , 1993, 606, 126-129.	1.1	25
124	The role of the dystrophin-glycoprotein complex in the molecular pathogenesis of muscular dystrophies. <i>Neuromuscular Disorders</i> , 1993, 3, 533-535.	0.3	51
125	Characterization of translational frame exception patients in Duchenne/Becker muscular dystrophy. <i>Human Molecular Genetics</i> , 1993, 2, 737-744.	1.4	89
126	Identification of two point mutations and a one base deletion in exon 19 of the dystrophin gene by heteroduplex formation. <i>Human Molecular Genetics</i> , 1993, 2, 311-313.	1.4	46



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127	Identification of a 2 base pair nonsense mutation causing a cryptic splice site in a DMD patient. <i>Human Molecular Genetics</i> , 1992, 1, 645-646.	1.4	32
128	A HindIII/BglII dystrophin gene polymorphism in the African-American population. <i>Human Genetics</i> , 1992, 89, 687-8.	1.8	4
129	Physical mapping at a potential X-linked retinitis pigmentosa locus (RP3) by pulsed-field gel electrophoresis. <i>Genomics</i> , 1991, 11, 263-272.	1.3	21
130	Linkage analysis of a large Latin-American family with X-linked retinitis pigmentosa and metallic sheen in the heterozygote carrier. <i>Genomics</i> , 1989, 4, 601-605.	1.3	15
131	Mapping of four translocation breakpoints within the Duchenne muscular dystrophy gene. <i>Genomics</i> , 1989, 4, 101-104.	1.3	16
132	The Duchenne muscular dystrophy gene product is localized in sarcolemma of human skeletal muscle. <i>Nature</i> , 1988, 333, 466-469.	13.7	650
133	Frame-shift deletions in patients with Duchenne and Becker muscular dystrophy. <i>Science</i> , 1988, 242, 755-759.	6.0	280
134	Duchenne muscular dystrophy gene expression in normal and diseased human muscle. <i>Science</i> , 1988, 239, 1418-1420.	6.0	60
135	Molecular Genetics of Duchenne and Becker Muscular Dystrophy. <i>International Review of Neurobiology</i> , 1988, 29, 1-76.	0.9	17
136	A cDNA clone from the Duchenne/Becker muscular dystrophy gene. <i>Nature</i> , 1987, 328, 434-437.	13.7	280
137	Comparison of focusing in buffers and synthetic carrier ampholytes for use in the first dimension of two dimensional polyacrylamide gel electrophoresis. <i>Electrophoresis</i> , 1985, 6, 453-461.	1.3	11
138	High resolution two-dimensional polyacrylamide gel electrophoresis. I. Methodological procedures. <i>Electrophoresis</i> , 1983, 4, 97-116.	1.3	128
139	High resolution two-dimensional polyacrylamide gel electrophoresis. II. Analysis and applications. <i>Electrophoresis</i> , 1983, 4, 173-189.	1.3	62
140	High resolution two-dimensional polyacrylamide gel electrophoresis. <i>TrAC - Trends in Analytical Chemistry</i> , 1983, 2, 211-214.	5.8	5
141	Isolation and characterization of a mutant liver aldolase in adult hereditary fructose intolerance. Identification of the enzyme variant by radioassay in tissue biopsy specimens. <i>Journal of Clinical Investigation</i> , 1983, 72, 201-213.	3.9	19
142	Analysis of skin fibroblast proteins in Duchenne muscular dystrophy: 1. Sodium dodecyl sulphate polyacrylamide gel electrophoresis. <i>Electrophoresis</i> , 1982, 3, 177-185.	1.3	16
143	Analysis of skin fibroblast proteins in Duchenne muscular dystrophy: 2. Isoelectric focusing under dissociating conditions. <i>Electrophoresis</i> , 1982, 3, 185-196.	1.3	19
144	Improvements of isoelectric focusing in agarose for direct tissue isoelectric focusing. <i>Electrophoresis</i> , 1982, 3, 307-314.	1.3	18

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145	Enhancement of resolution in two-dimensional gel electrophoresis and simultaneous resolution of acidic and basic proteins. <i>Electrophoresis</i> , 1982, 3, 354-363.	1.3	49
146	Erythrocyte membrane (Ca <sup>2+</sup> + Mg <sup>2+</sup> )-activated adenosine triphosphatase in Duchenne muscular dystrophy. <i>Biochemical Society Transactions</i> , 1981, 9, 81-82.	1.6	1
147	Polyacrylamide-gel-electrophoretic analysis of cultured skin fibroblasts from patients with Duchenne muscular dystrophy. <i>Biochemical Society Transactions</i> , 1981, 9, 118-119.	1.6	3
148	The application of direct tissue isoelectric focusing to the study of human skeletal muscle. <i>Electrophoresis</i> , 1981, 2, 251-258.	1.3	17
149	Erythrocyte ghost Na <sup>+</sup> , K <sup>+</sup> -adenosine triphosphatase in Duchenne muscular dystrophy. <i>Journal of the Neurological Sciences</i> , 1980, 46, 209-220.	0.3	9