Donald Love

List of Publications by Year in descending order

159 papers	9,124 citations	61945 43 h-index	40954 93 g-index
159 all docs	159 docs citations	159 times ranked	9455 citing authors

#	Article	IF	Citations
1	Impact and predictors of quality of life in adults diagnosed with a genetic muscle disorder: a nationwide population-based study. Quality of Life Research, 2022, 31, 1657-1666.	1.5	2
2	A Novel STK4 Mutation Impairs T Cell Immunity Through Dysregulation of Cytokine-Induced Adhesion and Chemotaxis Genes. Journal of Clinical Immunology, 2021, 41, 1839-1852.	2.0	3
3	The yield of postmortem genetic testing in sudden death cases with structural findings at autopsy. European Journal of Human Genetics, 2020, 28, 17-22.	1.4	38
4	Screening for anaplastic lymphoma kinase (ALK) gene rearrangements in nonâ€smallâ€cell lung cancer in New Zealand. Internal Medicine Journal, 2020, 50, 716-725.	0.5	9
5	Genetic testing in Polynesian long QT syndrome probands reveals a lower diagnostic yield and an increased prevalence of rare variants. Heart Rhythm, 2020, 17, 1304-1311.	0.3	3
6	Next-generation sequencing using microfluidic PCR enrichment for molecular autopsy. BMC Cardiovascular Disorders, 2019, 19, 174.	0.7	7
7	Observations on the Natural History of Camurati-Engelmann Disease. Journal of Bone and Mineral Research, 2019, 34, 875-882.	3.1	11
8	A Nationwide, Population-Based Prevalence Study of Genetic Muscle Disorders. Neuroepidemiology, 2019, 52, 128-135.	1.1	27
9	Development of a cardiac inherited disease service and clinical registry: A 15-year perspective. American Heart Journal, 2019, 209, 126-130.	1.2	10
10	Penetrance and expressivity of the R858H CACNA1C variant in a fiveâ€generation pedigree segregating an arrhythmogenic channelopathy. Molecular Genetics & Genomic Medicine, 2019, 7, e00476.	0.6	11
11	Determination of Pathogenicity of Breast Cancer 1 Gene Variants using the American College of Medical Genetics and Genomics and the Association for Molecular Pathology Guidelines. Sultan Qaboos University Medical Journal, 2019, 19, 324.	0.3	0
12	Long QT molecular autopsy in sudden unexplained death in the young (1-40 years old): Lessons learnt from an eight year experience in New Zealand. PLoS ONE, 2018, 13, e0196078.	1.1	24
13	Impacts for Children Living with Genetic Muscle Disorders and their Parents â€“ Findings from a Population-Based Study. Journal of Neuromuscular Diseases, 2018, 5, 341-352.	1.1	4
14	Utility of Post-Mortem Genetic Testing in Cases of Sudden Arrhythmic Death Syndrome. Journal of the American College of Cardiology, 2017, 69, 2134-2145.	1.2	219
15	Compound heterozygous SLC19A3 mutations further refine the critical promoter region for biotin-thiamine-responsive basal ganglia disease. Journal of Physical Education and Sports Management, 2017, 3, a001909.	0.5	20
16	Compound Heterozygous Inheritance of Mutations in Coenzyme Q8A Results in Autosomal Recessive Cerebellar Ataxia and Coenzyme Q10 Deficiency in a Female Sib-Pair. JIMD Reports, 2017, 42, 31-36.	0.7	21
17	EGFR Mutation Testing of non-squamous NSCLC: Impact and Uptake during Implementation of Testing Guidelines in a Population-Based Registry Cohort from Northern New Zealand. Targeted Oncology, 2017, 12, 663-675.	1.7	12
18	Massively Parallel Sequencing of Genes Implicated in Heritable Cardiac Disorders: A Strategy for a Small Diagnostic Laboratory. Medical Sciences (Basel, Switzerland), 2017, 5, 22.	1.3	2

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19	Splice Site Variants in the KCNQ1 and SCN5A Genes: Transcript Analysis as a Tool in Supporting Pathogenicity. Journal of Clinical Medicine Research, 2017, 9, 709-718.	0.6	4
20	Lung cancer mutation testing: a clinical retesting study of agreement between a real-time PCR and a mass spectrometry test. Oncotarget, 2017, 8, 101437-101451.	0.8	12
21	Evaluation of Bioinformatic Programmes for the Analysis of Variants within Splice Site Consensus Regions. Advances in Bioinformatics, 2016, 2016, 1-10.	5.7	43
22	The natural history of elevated tetradecenoylâ€Lâ€carnitine detected by newborn screening in New Zealand: implications for very long chain acylâ€CoA dehydrogenase deficiency screening and treatment. Journal of Inherited Metabolic Disease, 2016, 39, 409-414.	1.7	11
23	Two Novel GLDC Mutations in a Neonate with Nonketotic Hyperglycinemia. Journal of Pediatric Genetics, 2016, 05, 174-180.	0.3	5
24	A Prospective Study of Sudden Cardiac Death among Children and Young Adults. New England Journal of Medicine, 2016, 374, 2441-2452.	13.9	619
25	Microarray testing in clinical diagnosis: an analysis of 5,300 New Zealand patients. Molecular Cytogenetics, 2016, 9, 29.	0.4	6
26	Brain dopamineâ€serotonin vesicular transport disease presenting as a severe infantile hypotonic parkinsonian disorder. Journal of Inherited Metabolic Disease, 2016, 39, 305-308.	1.7	41
27	Detection of sudden death syndromes in New Zealand. New Zealand Medical Journal, 2016, 129, 67-74.	0.5	16
28	NOS1AP Polymorphisms Modify QTc Interval Duration But Not Cardiac Arrest Risk in Hypertrophic Cardiomyopathy. Journal of Cardiovascular Electrophysiology, 2015, 26, 1346-1351.	0.8	4
29	SNP Analysis and Whole Exome Sequencing: Their Application in the Analysis of a Consanguineous Pedigree Segregating Ataxia. Microarrays (Basel, Switzerland), 2015, 4, 490-502.	1.4	5
30	Whole Exome Sequencing Reveals Compound Heterozygosity for Ethnically Distinct PEX7 Mutations Responsible for Rhizomelic Chondrodysplasia Punctata, Type 1. Case Reports in Genetics, 2015, 2015, 1-4.	0.1	3
31	12q14 Microdeletions: Additional Case Series with Confirmation of a Macrocephaly Region. Case Reports in Genetics, 2015, 2015, 1-7.	0.1	10
32	Congestive myeloradiculopathy in a patient with Cowden syndrome. Journal of Clinical Neuroscience, 2015, 22, 431-433.	0.8	6
33	The New Zealand Neuromuscular Disease Registry: Rate of diagnoses confirmed by molecular testing. Journal of Clinical Neuroscience, 2015, 22, 434-436.	0.8	2
34	Assessment of the predictive accuracy of five in silico prediction tools, alone or in combination, and two metaservers to classify long QT syndrome gene mutations. BMC Medical Genetics, 2015, 16, 34.	2.1	73
35	Merosin-deficient congenital muscular dystrophy: A novel homozygous mutation in the laminin-2 gene. Journal of Clinical Neuroscience, 2015, 22, 1983-1985.	0.8	6
36	Array comparative genomic hybridization identifies a heterozygous deletion of exon 3 of theRYR2gene. Upsala Journal of Medical Sciences, 2015, 120, 190-197.	0.4	5

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37	The Diagnosis of Choriocarcinoma in Molar Pregnancies: A Revised Approach in Clinical Testing. Journal of Clinical Medicine Research, 2015, 7, 961-966.	0.6	7
38	Diagnostic Screening Workflow for Mutations in the BRCA1 and BRCA2 Genes. Sultan Qaboos University Medical Journal, 2015, 15, e58-70.	0.3	4
39	Predicting the Pathogenic Potential of BRCA1 and BRCA2 Gene Variants Identified in Clinical Genetic Testing. Sultan Qaboos University Medical Journal, 2015, 15, e218-25.	0.3	2
40	A Novel Glycine Decarboxylase Gene Mutation in an Indian Family With Nonketotic Hyperglycinemia. Journal of Child Neurology, 2014, 29, 122-127.	0.7	10
41	Long QT molecular autopsy in sudden infant death syndrome. Archives of Disease in Childhood, 2014, 99, 635-640.	1.0	36
42	Diabetic Dead-in-Bed Syndrome: A Possible Link to a Cardiac Ion Channelopathy. Case Reports in Medicine, 2014, 2014, 1-5.	0.3	14
43	Expression of a Mutant kcnj2 Gene Transcript in Zebrafish. , 2014, 2014, 1-14.		7
44	Microduplication of 3p26.3 Implicated in Cognitive Development. Case Reports in Genetics, 2014, 2014, 1-6.	0.1	17
45	A Turner Syndrome Patient Carrying a Mosaic Distal X Chromosome Marker. Case Reports in Genetics, 2014, 2014, 1-5.	0.1	10
46	A Case of 17q21.31 Microduplication and 7q31.33 Microdeletion, Associated with Developmental Delay, Microcephaly, and Mild Dysmorphic Features. Case Reports in Genetics, 2014, 2014, 1-6.	0.1	8
47	Single nucleotide polymorphisms in arrhythmia genes modify the risk of cardiac events and sudden death in long QT syndrome. Heart Rhythm, 2014, 11, 76-82.	0.3	53
48	Array Comparative Genomic Hybridization Identifies a Heterozygous Deletion of the Entire KCNJ2 Gene as a Cause of Sudden Cardiac Death. Circulation: Cardiovascular Genetics, 2014, 7, 17-22.	5.1	7
49	The left and right atria finally express themselves. Heart Rhythm, 2014, 11, 272-273.	0.3	0
50	Application of Massively Parallel Sequencing in the Clinical Diagnostic Testing of Inherited Cardiac Conditions. Medical Sciences (Basel, Switzerland), 2014, 2, 98-126.	1.3	2
51	Tumour Mutation Profiling with High-throughput Multiplexed Genotyping: A Review of its Use for Guiding Targeted Cancer Therapy. Current Cancer Therapy Reviews, 2014, 9, 236-244.	0.2	0
52	The p.Ala510Val mutation in the SPG7 (paraplegin) gene is the most common mutation causing adult onset neurogenetic disease in patients of British ancestry. Journal of Neurology, 2013, 260, 1286-1294.	1.8	45
53	A novel microdeletion syndrome at 9q21.13 characterised by mental retardation, speech delay, epilepsy and characteristic facial features. European Journal of Medical Genetics, 2013, 56, 163-170.	0.7	35
54	Community detection of long QT syndrome with a clinical registry: An alternative to ECG screening programs?. Heart Rhythm, 2013, 10, 233-238.	0.3	56

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55	Bacterial artificial chromosomes (BACs)-on-Beadsâ„¢ as a diagnostic platform for the rapid aneuploidy screening of products of conception. Molecular Medicine Reports, 2013, 8, 650-654.	1.1	6
56	Gene Dosage Analysis in a Clinical Environment: Gene-Targeted Microarrays as the Platform-of-Choice. Microarrays (Basel, Switzerland), 2013, 2, 51-62.	1.4	2
57	A Streamlined Protocol for Molecular Testing of the DMD Gene within a Diagnostic Laboratory: A Combination of Array Comparative Genomic Hybridization and Bidirectional Sequence Analysis. ISRN Neurology, 2013, 2013, 1-7.	1.5	6
58	Delineation of 2q32q35 Deletion Phenotypes: Two Apparent â€œProximalâ€•and â€œDistalâ€•Syndromes. Case Reports in Genetics, 2013, 2013, 1-8.	0.1	8
59	Novel mutation in the <scp>TMEM</scp>127 gene associated with phaeochromocytoma. Internal Medicine Journal, 2013, 43, 449-451.	0.5	14
60	Frequency and genetic spectrum of maturity-onset diabetes of the young (MODY) in southern New Zealand. Journal of Diabetes and Metabolic Disorders, 2013, 12, 46.	0.8	10
61	Developmental delay referrals and the roles of Fragile X testing and molecular karyotyping: A New Zealand perspective. Molecular Medicine Reports, 2013, 7, 1710-1714.	1.1	4
62	Simple Repeat-Primed PCR Analysis of the Myotonic Dystrophy Type 1 Gene in a Clinical Diagnostics Environment. Journal of Neurodegenerative Diseases, 2013, 2013, 1-8.	1.1	8
63	Array-Based Identification of Copy Number Changes in a Diagnostic Setting : Simultaneous Gene - Focused and Low Resolution Whole Human Genome Analysis. Sultan Qaboos University Medical Journal, 2013, 13, 69-79.	0.3	5
64	Implications of a Chr7q21.11 Microdeletion and the Role of the PCLO Gene in Developmental Delay = Ø§Ù„Ø£Ø«Ø§Ø± Ø§Ù„Ù…Ø±ØªØ¨Ø·Ø© Ø¨ØØ°Ù Chr7q21.11 ÙˆØ¯ÙˆØ± Ø¬ÙŠÙ† PCLO ÙŠÙŠØ ØµØ¹ÙˆØ¨Ø§Øª Ø§Ù„ØªØ¹Ù„Ù….... Sultan Qaboos University Medical Journal, 2013, 13, 306-310.	0.3	3
65	Expression of a Mutant kcnj2 Gene Transcript in Zebrafish. , 2013, 2013, 324839.		5
66	In VivoTesting of MicroRNA-Mediated Gene Knockdown in Zebrafish. Journal of Biomedicine and Biotechnology, 2012, 2012, 1-7.	3.0	8
67	Effects of kiwifruit extracts on colonic gene and protein expression levels in IL-10 gene-deficient mice. British Journal of Nutrition, 2012, 108, 113-129.	1.2	24
68	A Novel 2.3 Mb Microduplication of 9q34.3 Inserted into 19q13.4 in a Patient with Learning Disabilities. Case Reports in Pediatrics, 2012, 2012, 1-7.	0.2	3
69	The New Zealand Neuromuscular Disease Registry. Journal of Clinical Neuroscience, 2012, 19, 1749-1750.	0.8	20
70	Amino-Terminal Microdeletion within theCNTNAP2Gene Associated with Variable Expressivity of Speech Delay. Case Reports in Genetics, 2012, 2012, 1-4.	0.1	17
71	Fluorescent Function-Spacer-Lipid Construct Labelling Allows for Real-Time in Vivo Imaging of Cell Migration and Behaviour in Zebrafish (Danio Rerio). Journal of Fluorescence, 2012, 22, 1055-1063.	1.3	10
72	Application of complementary luminescent and fluorescent imaging techniques to visualize nuclear and cytoplasmic Ca²⁺ signalling during the in vivo differentiation of slow muscle cells in zebrafish embryos under normal and dystrophic conditions. Clinical and Experimental Pharmacology and Physiology, 2012, 39, 78-86.	0.9	12

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73	Association of vitamin D receptor gene polymorphisms with insulin resistance and response to vitamin D. Metabolism: Clinical and Experimental, 2012, 61, 293-301.	1.5	51
74	Automation of a primer design and evaluation pipeline for subsequent sequencing of the coding regions of all human Refseq genes. Bioinformation, 2012, 8, 365-368.	0.2	6
75	Indolent Medullary Thyroid Cancer with a RET Proto-Oncogene Cys618Phe Mutation Presenting As Sporadic Unilateral Pheochromocytoma in a 55-Year-Old Korean Woman. Thyroid, 2011, 21, 325-326.	2.4	2
76	Elevated serum gastrin levels in Jervell and Lange-Nielsen syndrome: A marker of severe KCNQ1 dysfunction?. Heart Rhythm, 2011, 8, 551-554.	0.3	26
77	Pseudotrisomy 13 syndrome: Use of homozygosity mapping to target candidate genes. Gene, 2011, 486, 37-40.	1.0	6
78	Prospective, population-based long QT molecular autopsy study of postmortem negative sudden death in 1 to 40 year olds. Heart Rhythm, 2011, 8, 412-419.	0.3	148
79	Pure Duplication of the Distal Long Arm of Chromosome 15 with Ebstein Anomaly and Clavicular Anomaly. Case Reports in Genetics, 2011, 2011, 1-5.	0.1	7
80	Inheritance of a Ring Chromosome 21 in a Couple UndergoingIn VitroFertilization (IVF): A Case Report. Case Reports in Genetics, 2011, 2011, 1-5.	0.1	4
81	Zebrafish dystrophin and utrophin genes: Dissecting transcriptional expression during embryonic development. International Journal of Molecular Medicine, 2011, 29, 338-48.	1.8	2
82	Citrullinaemia type I: A common mutation in the Pacific Island population. Journal of Paediatrics and Child Health, 2011, 47, 262-265.	0.4	10
83	Kiwifruit extracts inhibit cytokine production by lipopolysaccharide-activated macrophages, and intestinal epithelial cells isolated from IL10 gene deficient mice. Cellular Immunology, 2011, 270, 70-79.	1.4	29
84	Targeted mutagenesis of zebrafish: Use of zinc finger nucleases. Birth Defects Research Part C: Embryo Today Reviews, 2011, 93, 249-255.	3.6	11
85	Disease Modeling by Gene Targeting Using MicroRNAs. Methods in Cell Biology, 2011, 105, 419-436.	0.5	6
86	Visualization, characterization and modulation of calcium signaling during the development of slow muscle cells in intact zebrafish embryos. International Journal of Developmental Biology, 2011, 55, 153-174.	0.3	46
87	A transient assay for recombination demonstrates that Arabidopsis SNM1 and XRCC3 enhance non-homologous recombination. Genetics and Molecular Research, 2011, 10, 2104-32.	0.3	4
88	Distal 5q deletion with associated parietal foramina. Clinical Dysmorphology, 2010, 19, 43-47.	0.1	4
89	The clinical utility of molecular diagnostic testing for primary immune deficiency disorders: a case based review. Allergy, Asthma and Clinical Immunology, 2010, 6, 12.	0.9	25
90	Zebrafish as a model for long QT syndrome: the evidence and the means of manipulating zebrafish gene expression. Acta Physiologica, 2010, 199, 257-276.	1.8	47

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91	Application of Nutrigenomics in Gastrointestinal Health. , 2010, , 83-94.		0
92	The SCN5A gene in Brugada syndrome: mutations, variants, missense and nonsense. What's a clinician to do?. Heart Rhythm, 2010, 7, 50-51.	0.3	2
93	Identification and expression analysis of kcnh2 genes in the zebrafish. Biochemical and Biophysical Research Communications, 2010, 396, 817-824.	1.0	30
94	Posthumous diagnosis of long QT syndrome from neonatal screening cards. Heart Rhythm, 2010, 7, 481-486.	0.3	56
95	Array comparative genomic hybridisation: a new tool in the diagnostic genetic armoury. New Zealand Medical Journal, 2010, 123, 50-61.	0.5	1
96	Activin is a potent growth suppressor of epithelial ovarian cancer cells. Cancer Letters, 2009, 285, 157-165.	3.2	24
97	Role of gut microbiota in Crohnâ€™s disease. Expert Review of Gastroenterology and Hepatology, 2009, 3, 535-546.	1.4	57
98	Quantitative Real-Time RT-PCR (qRT-PCR) of Zebrafish Transcripts: Optimization of RNA Extraction, Quality Control Considerations, and Data Analysis. Cold Spring Harbor Protocols, 2009, 2009, pdb.prot5314.	0.2	29
99	Phenotypic expansion and further characterisation of the 17q21.31 microdeletion syndrome. Journal of Medical Genetics, 2009, 46, 480-489.	1.5	66
100	Chimerism detected in fraternal twins using ABI AmpFlSTRÂ® Identifiler. Forensic Science International: Genetics Supplement Series, 2009, 2, 226-227.	0.1	0
101	Spectrum of mutations in sarcoglycan genes in the Mumbai region of western India: High prevalence of 525del T. Neurology India, 2009, 57, 406.	0.2	25
102	Identification of large gene deletions and duplications in KCNQ1 and KCNH2 in patients with long QT syndrome. Heart Rhythm, 2008, 5, 1275-1281.	0.3	79
103	Discovery of three related females who type XY at the amelogenin locus. Forensic Science International: Genetics Supplement Series, 2008, 1, 577-579.	0.1	2
104	Whole organism approaches to chemical genomics: the promising role of zebrafish (Danio rerio). Expert Opinion on Drug Discovery, 2007, 2, 1389-1401.	2.5	3
105	Modeling inflammatory bowel disease: the zebrafish as a way forward. Expert Review of Molecular Diagnostics, 2007, 7, 177-193.	1.5	11
106	Validation of Zebrafish (Danio rerio) Reference Genes for Quantitative Real-time RT-PCR Normalization. Acta Biochimica Et Biophysica Sinica, 2007, 39, 384-390.	0.9	540
107	Zebrafish: At the Nexus of Functional and Chemical Genomics. Biotechnology and Genetic Engineering Reviews, 2006, 22, 77-100.	2.4	2
108	Technology for high-throughput screens: the present and future using zebrafish. Current Opinion in Biotechnology, 2004, 15, 564-571.	3.3	102

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109	Global gene expression analysis in the zebrafish: the challenge and the promise. Drug Discovery Today: Technologies, 2004, 1, 79-84.	4.0	5
110	Short interfering RNA-mediated gene targeting in the zebrafish. FEBS Letters, 2004, 561, 89-93.	1.3	74
111	Modeling Human Disease by Gene Targeting. Methods in Cell Biology, 2004, 76, 593-612.	0.5	6
112	Idiopathic Hyperphosphatasia andTNFRSF11BMutations: Relationships Between Phenotype and Genotype. Journal of Bone and Mineral Research, 2003, 18, 2095-2104.	3.1	113
113	Chemical discovery and global gene expression analysis in zebrafish. Nature Biotechnology, 2003, 21, 879-883.	9.4	142
114	Sarcoglycans of the zebrafish: orthology and localization to the sarcolemma and myosepta of muscle. Biochemical and Biophysical Research Communications, 2003, 303, 488-495.	1.0	25
115	A mutation in the gene TNFRSF11B encoding osteoprotegerin causes an idiopathic hyperphosphatasia phenotype. Human Molecular Genetics, 2002, 11, 2119-2127.	1.4	190
116	Melanocortin-3 receptor gene variants in a Maori kindred with obesity and early onset type 2 diabetes. Diabetes Research and Clinical Practice, 2002, 58, 61-71.	1.1	32
117	Detection of Sequence Variations in the Adenomatous Polyposis Coli (APC) Gene Using Denaturing High-Performance Liquid Chromatography. Genetic Testing and Molecular Biomarkers, 2001, 5, 281-290.	1.7	32
118	Dystrophin in Adult Zebrafish Muscle. Biochemical and Biophysical Research Communications, 2001, 286, 478-483.	1.0	38
119	Gene flow on the ice: genetic differentiation among AdÃ©lie penguin colonies around Antarctica. Molecular Ecology, 2001, 10, 1645-1656.	2.0	71
120	Analysis of the TGF Î² functional pathway in epithelial ovarian carcinoma. British Journal of Cancer, 2001, 85, 687-691.	2.9	28
121	Hierarchical mutation screening protocol for theBRCA1 gene. Human Mutation, 2000, 16, 422-430.	1.1	2
122	Null Alleles at the Huntington Disease Locus: Implications for Diagnostics and CAG Repeat Instability. Genetic Testing and Molecular Biomarkers, 2000, 4, 55-60.	1.7	18
123	Zebrafish: bridging the gap between development and disease. Human Molecular Genetics, 2000, 9, 2443-2449.	1.4	137
124	Direct Interaction between Emerin and Lamin A. Biochemical and Biophysical Research Communications, 2000, 267, 709-714.	1.0	226
125	Distribution of emerin and lamins in the heart and implications for Emery-Dreifuss muscular dystrophy. Human Molecular Genetics, 1999, 8, 353-359.	1.4	90
126	Comparative semi-automated analysis of (CAG) repeats in the Huntington disease gene: use of internal standards. Molecular and Cellular Probes, 1999, 13, 283-289.	0.9	24

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127	Trinucleotide (CAG) repeat length is positively correlated with the degree of DNA fragmentation in Huntington's disease striatum. Neuroscience, 1998, 87, 49-53.	1.1	94
128	Reduction in enkephalin and substance P messenger RNA in the striatum of early grade Huntington's disease: A detailed cellularin situ hybridization study. Neuroscience, 1996, 72, 1023-1036.	1.1	141
129	Decreased neuronal nitric oxide synthase messenger RNA and somatostatin messenger RNA in the striatum of Huntington's disease. Neuroscience, 1996, 72, 1037-1047.	1.1	96
130	Structure and Location of the Murine Adrenoleukodystrophy Gene. Genomics, 1996, 32, 395-400.	1.3	9
131	A mutation in the Î± tropomyosin gene TPM3 associated with autosomal dominant nemaline myopathy. Nature Genetics, 1995, 9, 75-79.	9.4	315
132	Molecular characterization of further dystrophin gene microsatellites. Molecular and Cellular Probes, 1995, 9, 361-370.	0.9	9
133	A multiple interval physical map of the pericentromeric region of human chromosome 10. Human Genetics, 1994, 93, 313-318.	1.8	7
134	Germ-line mutations of the RET proto-oncogene in multiple endocrine neoplasia type 2A. Nature, 1993, 363, 458-460.	13.7	1,886
135	Dystrophin and dystrophin-related proteins: A review of protein and RNA studies. Neuromuscular Disorders, 1993, 3, 5-21.	0.3	59
136	Genetic linkage studies map the multiple endocrine neoplasia type 2 loci to a small interval on chromosome 10q11.2. Human Molecular Genetics, 1993, 2, 241-246.	1.4	100
137	A YAC contig in Xp21 containing the adrenal hypoplasia congenita and glycerol kinase deficiency genes. Human Molecular Genetics, 1992, 1, 579-585.	1.4	33
138	Human dystrophin expression corrects the myopathic phenotype in transgenic mdx mice. Human Molecular Genetics, 1992, 1, 35-40.	1.4	67
139	Characterization of a 4.8kb transcript from the Duchenne muscular dystrophy locus expressed in Schwannoma cells. Human Molecular Genetics, 1992, 1, 103-109.	1.4	125
140	Retroviral-mediated transfer of a dystrophin minigene intomdxmouse myoblasts in vitro. FEBS Letters, 1992, 296, 128-134.	1.3	72
141	Localization of two new DNA markers on the linkage map of human chromosome 6q. Cytogenetic and Genome Research, 1992, 60, 216-218.	0.6	0
142	Exclusion of the gene responsible for facioscapulohumeral muscular dystrophy (FSH) at 6q23-q27. Journal of the Neurological Sciences, 1991, 102, 206-208.	0.3	1
143	Sequences of junction fragments in the deletion-prone region of the dystrophin gene. Genomics, 1991, 10, 57-67.	1.3	53
144	Human dystrophin expression in mdx mice after intramuscular injection of DNA constructs. Nature, 1991, 352, 815-818.	13.7	501

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145	Linkage analysis in families with autosomal recessive limb-girdle muscular dystrophy (LGMD) and 6q probes flanking the dystrophin-related sequence. American Journal of Medical Genetics Part A, 1991, 38, 140-146.	2.4	10
146	Apparent association of mental retardation and specific patterns of deletions screened with probes cf56a and cf23a in Duchenne muscular dystrophy. American Journal of Medical Genetics Part A, 1991, 39, 437-441.	2.4	66
147	Human dystrophin gene transfer: production and expression of a functional recombinant DNA-based gene. Human Genetics, 1991, 88, 53-58.	1.8	47
148	A new DNA marker, D6S129, identifies aHindlll polymorphismOn ChromoSome 6q. Nucleic Acids Research, 1991, 19, 4310-4310.	6.5	0
149	Localisation of a dystrophin-related autosomal gene to 6q24 in man, and to mouse chromosome 10 in the region of the dystrophia muscularis (dy) locus. Human Genetics, 1990, 85, 324-6.	1.8	64
150	Characterization of deletions in the dystrophin gene giving mild phenotypes. American Journal of Medical Genetics Part A, 1990, 37, 136-142.	2.4	31
151	Physical mapping distal to the DMD locus. Genomics, 1990, 8, 106-112.	1.3	20
152	Monoclonal antibodies against defined regions of the muscular dystrophy protein, dystrophin. FEBS Letters, 1990, 262, 237-240.	1.3	52
153	Nucleotide sequence of a gene fromCaldocellum saccharolyticumencoding for exocellulase and endocellulase activity. Nucleic Acids Research, 1989, 17, 439-439.	6.5	61
154	An autosomal transcript in skeletal muscle with homology to dystrophin. Nature, 1989, 339, 55-58.	13.7	501
155	Sequence structure and expression of a cloned Î²-glucosidase gene from an extreme thermophile. Molecular Genetics and Genomics, 1988, 213, 84-92.	2.4	69
156	Distinct dystrophin mRNA species are expressed in embryonic and adult mouse skeletal muscle. FEBS Letters, 1988, 242, 47-52.	1.3	25
157	Genetics and Potential Biotechnological Applications of Thermophilic and Extremely Thermophilic Microorganisms. Biotechnology and Genetic Engineering Reviews, 1987, 5, 199-244.	2.4	64
158	Molecular Cloning of a Î²-Glucosidase Gene from an Extremely Thermophilic Anaerobe in E. coli and B. subtilis. Nature Biotechnology, 1987, 5, 384-387.	9.4	23
159	Expression of leucine genes from an extremely thermophilic bacterium in Escherichia coli. Molecular Genetics and Genomics, 1987, 210, 490-497.	2.4	35