Johan T Den Dunnen

List of Publications by Year
in descending order

339 papers	30,130 citations	4960 84 h-index	6300 158 g-index
355 all docs	355 docs citations	355 times ranked	39914 citing authors

#	Article	IF	Citations
1	Mutation nomenclature extensions and suggestions to describe complex mutations: A discussion. Human Mutation, 2000, 15, 7-12.	2.5	1,688
2	HGVS Recommendations for the Description of Sequence Variants: 2016 Update. Human Mutation, 2016, 37, 564-569.	2.5	1,194
3	LOVD v.2.0: the next generation in gene variant databases. Human Mutation, 2011, 32, 557-563.	2.5	854
4	Local Dystrophin Restoration with Antisense Oligonucleotide PRO051. New England Journal of Medicine, 2007, 357, 2677-2686.	27.0	735
5	Deep sequencing-based expression analysis shows major advances in robustness, resolution and inter-lab portability over five microarray platforms. Nucleic Acids Research, 2008, 36, e141-e141.	14.5	653
6	Whole-genome sequence variation, population structure and demographic history of the Dutch population. Nature Genetics, 2014, 46, 818-825.	21.4	641
7	Digenic inheritance of an SMCHD1 mutation and an FSHD-permissive D4Z4 allele causes facioscapulohumeral muscular dystrophy type 2. Nature Genetics, 2012, 44, 1370-1374.	21.4	582
8	Entries in the Leiden Duchenne muscular dystrophy mutation database: An overview of mutation types and paradoxical cases that confirm the reading-frame rule. Muscle and Nerve, 2006, 34, 135-144.	2.2	569
9	Next generation sequencing technology: Advances and applications. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 1932-1941.	3.8	557
10	Phenotypically Concordant and Discordant Monozygotic Twins Display Different DNA Copy-Number-Variation Profiles. American Journal of Human Genetics, 2008, 82, 763-771.	6.2	533
11	Theoretic applicability of antisense-mediated exon skipping for Duchenne muscular dystrophy mutations. Human Mutation, 2009, 30, 293-299.	2.5	485
12	Genetic Heterogeneity in Rubinstein-Taybi Syndrome: Mutations in Both the CBP and EP300 Genes Cause Disease. American Journal of Human Genetics, 2005, 76, 572-580.	6.2	416
13	High-Resolution Melting Analysis (HRMA)-More than just sequence variant screening. Human Mutation, 2009, 30, 860-866.	2.5	414
14	Application of a 5-tiered scheme for standardized classification of 2,360 unique mismatch repair gene variants in the InSiGHT locus-specific database. Nature Genetics, 2014, 46, 107-115.	21.4	410
15	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. Human Mutation, 2015, 36, 915-921.	2.5	390
16	Improving sequence variant descriptions in mutation databases and literature using the Mutalyzer sequence variation nomenclature checker. Human Mutation, 2008, 29, 6-13.	2.5	383
17	Mutations in SWI/SNF chromatin remodeling complex gene ARID1B cause Coffin-Siris syndrome. Nature Genetics, 2012, 44, 379-380.	21.4	312
18	Loss-of-function mutations in MICU1 cause a brain and muscle disorder linked to primary alterations in mitochondrial calcium signaling. Nature Genetics, 2014, 46, 188-193.	21.4	311

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19	Protein truncation test (PTT) for rapid detection of translation-terminating mutations. Human Molecular Genetics, 1993, 2, 1719-1721.	2.9	307
20	Rapid detection of BRCA1 mutations by the protein truncation test. Nature Genetics, 1995, 10, 208-212.	21.4	307
21	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	6.2	305
22	WHSC1, a 90 kb SET Domain-Containing Gene, Expressed in Early Development and Homologous to a Drosophila Dysmorphy Gene Maps in the Wolf-Hirschhorn Syndrome Critical Region and is Fused to IgH in t(1;14) Multiple Myeloma. Human Molecular Genetics, 1998, 7, 1071-1082.	2.9	296
23	High-Resolution Whole-Genome Sequencing Reveals That Specific Chromatin Domains from Most Human Chromosomes Associate with Nucleoli. Molecular Biology of the Cell, 2010, 21, 3735-3748.	2.1	274
24	Reproducibility of high-throughput mRNA and small RNA sequencing across laboratories. Nature Biotechnology, 2013, 31, 1015-1022.	17.5	251
25	The Genome of the Netherlands: design, and project goals. European Journal of Human Genetics, 2014, 22, 221-227.	2.8	246
26	LOVD: Easy creation of a locus-specific sequence variation database using an â€œLSDB-in-a-boxâ€•approach. Human Mutation, 2005, 26, 63-68.	2.5	235
27	Targeted sequencing by proximity ligation for comprehensive variant detection and local haplotyping. Nature Biotechnology, 2014, 32, 1019-1025.	17.5	231
28	Prorenin Induces Intracellular Signaling in Cardiomyocytes Independently of Angiotensin II. Hypertension, 2006, 48, 564-571.	2.7	228
29	Therapeutic antisense-induced exon skipping in cultured muscle cells from six different DMD patients. Human Molecular Genetics, 2003, 12, 907-914.	2.9	226
30	Complex SNP-related sequence variation in segmental genome duplications. Nature Genetics, 2004, 36, 861-866.	21.4	220
31	Three Genome-wide Association Studies and a Linkage Analysis Identify HERC2 as a Human Iris Color Gene. American Journal of Human Genetics, 2008, 82, 411-423.	6.2	220
32	Rapid Detection of Translation-Terminating Mutations at the Adenomatous Polyposis Coli (APC) Gene by Direct Protein Truncation Test. Genomics, 1994, 20, 1-4.	2.9	218
33	Nine unknown rearrangements in 16p13.3 and 11p15.4 causing Â- and Â-thalassaemia characterised by high resolution multiplex ligation-dependent probe amplification. Journal of Medical Genetics, 2005, 42, 922-931.	3.2	213
34	Direct detection of more than 50% of the Duchenne muscular dystrophy mutations by field inversion gels. Nature, 1987, 329, 640-642.	27.8	202
35	The PWWP domain: a potential proteinâ€“protein interaction domain in nuclear proteins influencing differentiation?. FEBS Letters, 2000, 473, 1-5.	2.8	196
36	Efficient and sensitive identification and quantification of airborne pollen using nextâ€generation <scp>DNA</scp> sequencing. Molecular Ecology Resources, 2015, 15, 8-16.	4.8	192

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37	Germinal mosaicism increases the recurrence risk for 'new' Duchenne muscular dystrophy mutations.. Journal of Medical Genetics, 1989, 26, 553-559.	3.2	187
38	Antisense-Induced Multiexon Skipping for Duchenne Muscular Dystrophy Makes More Sense. American Journal of Human Genetics, 2004, 74, 83-92.	6.2	180
39	Peters Plus Syndrome Is Caused by Mutations in B3GALTL, a Putative Glycosyltransferase. American Journal of Human Genetics, 2006, 79, 562-566.	6.2	178
40	Coffin-Siris Syndrome and the BAF Complex: Genotype-Phenotype Study in 63 Patients. Human Mutation, 2013, 34, 1519-1528.	2.5	178
41	Standardizing mutation nomenclature: Why bother?. Human Mutation, 2003, 22, 181-182.	2.5	176
42	Molecular genetic analysis of familial early-onset Alzheimer's disease linked to chromosome 14q24.3. Human Molecular Genetics, 1995, 4, 2363-2371.	2.9	171
43	Deletion and duplication screening in the DMD gene using MLPA. European Journal of Human Genetics, 2005, 13, 1231-1234.	2.8	171
44	Loss-of-function mutations in IGSF1 cause an X-linked syndrome of central hypothyroidism and testicular enlargement. Nature Genetics, 2012, 44, 1375-1381.	21.4	169
45	Comprehensive Detection of Genomic Duplications and Deletions in the DMD Gene, by Use of Multiplex Amplifiable Probe Hybridization. American Journal of Human Genetics, 2002, 71, 365-374.	6.2	163
46	High-resolution DNA Fiber-FISH for genomic DNA mapping and colour bar-coding of large genes. Human Molecular Genetics, 1995, 4, 831-836.	2.9	162
47	Mutations in ZBTB24 Are Associated with Immunodeficiency, Centromeric Instability, and Facial Anomalies Syndrome Type 2. American Journal of Human Genetics, 2011, 88, 796-804.	6.2	158
48	Targeted exon skipping as a potential gene correction therapy for Duchenne muscular dystrophy. Neuromuscular Disorders, 2002, 12, S71-S77.	0.6	157
49	Phage display screening without repetitious selection rounds. Analytical Biochemistry, 2012, 421, 622-631.	2.4	149
50	Poly(A) binding protein nuclear 1 levels affect alternative polyadenylation. Nucleic Acids Research, 2012, 40, 9089-9101.	14.5	148
51	BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. PLoS Genetics, 2018, 14, e1007752.	3.5	148
52	Standard Mutation Nomenclature in Molecular Diagnostics. Journal of Molecular Diagnostics, 2007, 9, 1-6.	2.8	146
53	Pharmacogenetic allele nomenclature: International workgroup recommendations for test result reporting. Clinical Pharmacology and Therapeutics, 2016, 99, 172-185.	4.7	146
54	Duplications in theDMD gene. Human Mutation, 2006, 27, 938-945.	2.5	145

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55	Two-color multiplex ligation-dependent probe amplification: Detecting genomic rearrangements in hereditary multiple exostoses. Human Mutation, 2004, 24, 86-92.	2.5	142
56	Long-term persistence of donor nuclei in a Duchenne muscular dystrophy patient receiving bone marrow transplantation. Journal of Clinical Investigation, 2002, 110, 807-814.	8.2	140
57	Identification of a hot spot for microdeletions in patients with X- linked deafness type 3 (DFN3) 900 kb proximal to the DFN3 gene POU3F4. Human Molecular Genetics, 1996, 5, 1229-1235.	2.9	138
58	Improved molecular diagnosis of dystrophinopathies in an unselected clinical cohort. American Journal of Medical Genetics, Part A, 2005, 134A, 295-298.	1.2	135
59	AHNAK a novel component of the dysferlin protein complex, redistributes to the cytoplasm with dysferlin during skeletal muscle regeneration. FASEB Journal, 2007, 21, 732-742.	0.5	133
60	Genome-wide assessment of differential roles for p300 and CBP in transcription regulation. Nucleic Acids Research, 2010, 38, 5396-5408.	14.5	133
61	Serum matrix metalloproteinase-9 (MMP-9) as a biomarker for monitoring disease progression in Duchenne muscular dystrophy (DMD). Neuromuscular Disorders, 2011, 21, 569-578.	0.6	132
62	A novel Fanconi anaemia subtype associated with a dominant-negative mutation in RAD51. Nature Communications, 2015, 6, 8829.	12.8	130
63	Comparative analysis of antisense oligonucleotide analogs for targeted DMD exon 46 skipping in muscle cells. Gene Therapy, 2004, 11, 1391-1398.	4.5	126
64	The value of data. Nature Genetics, 2011, 43, 281-283.	21.4	126
65	Next-Generation Diagnostics: Gene Panel, Exome, or Whole Genome?. Human Mutation, 2015, 36, 648-655.	2.5	124
66	Skewed X-inactivation is common in the general female population. European Journal of Human Genetics, 2019, 27, 455-465.	2.8	119
67	In Silico Functional Meta-Analysis of 5,962 ABCA4 Variants in 3,928 Retinal Dystrophy Cases. Human Mutation, 2017, 38, 400-408.	2.5	118
68	Calling on a million minds for community annotation in WikiProteins. Genome Biology, 2008, 9, R89.	9.6	117
69	Targeted Exon Skipping in Transgenic hDMD Mice: A Model for Direct Preclinical Screening of Human-Specific Antisense Oligonucleotides. Molecular Therapy, 2004, 10, 232-240.	8.2	111
70	Functional Analysis of 114 Exon-Internal AONs for Targeted DMD Exon Skipping: Indication for Steric Hindrance of SR Protein Binding Sites. Oligonucleotides, 2005, 15, 284-197.	2.7	108
71	An overview of L-2-hydroxyglutarate dehydrogenase gene (L2HGDH) variants: a genotype-phenotype study. Human Mutation, 2010, 31, 380-390.	2.5	108
72	Mutation Update: The Spectra of Nebulin Variants and Associated Myopathies. Human Mutation, 2014, 35, 1418-1426.	2.5	107

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73	Full-length mRNA sequencing uncovers a widespread coupling between transcription initiation and mRNA processing. Genome Biology, 2018, 19, 46.	8.8	106
74	Subcellular localization of the Huntington's disease gene product in cell lines by immunofluorescence and biochemical subcellular fractionation. Human Molecular Genetics, 1996, 5, 1093-1099.	2.9	103
75	Templated Insertions: A Smoking Gun for Polymerase Theta-Mediated End Joining. Trends in Genetics, 2019, 35, 632-644.	6.7	103
76	Rapid genotyping of blood group antigens by multiplex polymerase chain reaction and DNA microarray hybridization. Transfusion, 2005, 45, 667-679.	1.6	100
77	Locus Reference Genomic sequences: an improved basis for describing human DNA variants. Genome Medicine, 2010, 2, 24.	8.2	100
78	The InSiGHT database: utilizing 100Âyears of insights into Lynch Syndrome. Familial Cancer, 2013, 12, 175-180.	1.9	100
79	Nutrigenomics: The Impact of Biomics Technology on Nutrition Research. Annals of Nutrition and Metabolism, 2005, 49, 355-365.	1.9	98
80	The Effects of Low Levels of Dystrophin on Mouse Muscle Function and Pathology. PLoS ONE, 2012, 7, e31937.	2.5	96
81	Reconstruction of the 204 Mb human DMD-gene bhy homologous YAC recombination. Human Molecular Genetics, 1992, 1, 19-28.	2.9	95
82	Muscle regeneration in dystrophin-deficient mdx mice studied by gene expression profiling. BMC Genomics, 2005, 6, 98.	2.8	95
83	mRNA degradation controls differentiation state-dependent differences in transcript and splice variant abundance. Nucleic Acids Research, 2011, 39, 556-566.	14.5	95
84	Fluorescent labelling of cRNA for microarray applications. Nucleic Acids Research, 2003, 31, 20e-20.	14.5	92
85	Calpain 3 is a modulator of the dysferlin protein complex in skeletal muscle. Human Molecular Genetics, 2008, 17, 1855-1866.	2.9	89
86	Long-term persistence of donor nuclei in a Duchenne muscular dystrophy patient receiving bone marrow transplantation. Journal of Clinical Investigation, 2002, 110, 807-814.	8.2	89
87	Concerted and divergent evolution within the rat Î³-crystallin gene family. Journal of Molecular Biology, 1986, 189, 37-46.	4.2	88
88	Detection of mutations in the dystrophin gene via automated DHPLC screening and direct sequencing. BMC Genetics, 2001, 2, 17.	2.7	88
89	Akkermansia muciniphilaandHelicobacter typhloniusmodulate intestinal tumor development in mice. Carcinogenesis, 2015, 36, 1388-1396.	2.8	87
90	Distribution of Inclusions in Neuronal Nuclei and Dystrophic Neurites in Huntington Disease Brain. Journal of Neuropathology and Experimental Neurology, 1999, 58, 129-137.	1.7	85

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91	Dystrophin nonsense mutation induces different levels of exon 29 skipping and leads to variable phenotypes within one BMD family. European Journal of Human Genetics, 2000, 8, 793-796.	2.8	85
92	New methods for next generation sequencing based microRNA expression profiling. BMC Genomics, 2010, 11, 716.	2.8	85
93	Targeting Several CAG Expansion Diseases by a Single Antisense Oligonucleotide. PLoS ONE, 2011, 6, e24308.	2.5	85
94	Copy number variation in the genome; the human <i>DMD</i> gene as an example. Cytogenetic and Genome Research, 2006, 115, 240-246.	1.1	82
95	Recurrence risk due to germ line mosaicism: Duchenne and Becker muscular dystrophy. Clinical Genetics, 2009, 75, 465-472.	2.0	81
96	Severe Myocardial Fibrosis Caused by a Deletion of the 5â€™ End of the Lamin A/C Gene. Journal of the American College of Cardiology, 2007, 49, 2430-2439.	2.8	79
97	Huntingtonâ€™s disease biomarker progression profile identified by transcriptome sequencing in peripheral blood. European Journal of Human Genetics, 2015, 23, 1349-1356.	2.8	79
98	Exploring the Frontiers of Therapeutic Exon Skipping for Duchenne Muscular Dystrophy by Double Targeting within One or Multiple Exons. Molecular Therapy, 2006, 14, 401-407.	8.2	76
99	Mitochondrial Disease Sequence Data Resource (MSeqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. Molecular Genetics and Metabolism, 2015, 114, 388-396.	1.1	76
100	The UCL low-density lipoprotein receptor gene variant database: pathogenicity update. Journal of Medical Genetics, 2017, 54, 217-223.	3.2	75
101	Large scale single nucleotide polymorphism discovery in unsequenced genomes using second generation high throughput sequencing technology: applied to turkey. BMC Genomics, 2009, 10, 479.	2.8	73
102	Functional assessment of variants in the TSC1 and TSC2 genes identified in individuals with Tuberous Sclerosis Complex. Human Mutation, 2011, 32, 424-435.	2.5	73
103	Integrated study of 100 patients with Xp21 linked muscular dystrophy using clinical, genetic, immunochemical, and histopathological data. Part 1. Trends across the clinical groups.. Journal of Medical Genetics, 1993, 30, 728-736.	3.2	72
104	Leiden open variation database of the MUTYH gene. Human Mutation, 2010, 31, 1205-1215.	2.5	72
105	Deep sequencing to reveal new variants in pooled DNA samples. Human Mutation, 2009, 30, 1703-1712.	2.5	71
106	LAMA2gene mutation update: Toward a more comprehensive picture of the laminin-Î±2 variome and its related phenotypes. Human Mutation, 2018, 39, 1314-1337.	2.5	71
107	Successful Long-Term Growth Hormone Therapy in a Girl with Haploinsufficiency of the Insulin-Like Growth Factor-I Receptor due to a Terminal 15q26.2->qter Deletion Detected by Multiplex Ligation Probe Amplification. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 2421-2425.	3.6	70
108	Autosomal Recessive Spinocerebellar Ataxia 7 (SCAR7) is Caused by Variants inTPP1, The Gene Involved in Classic Late-Infantile Neuronal Ceroid Lipofuscinosis 2 Disease (CLN2 Disease). Human Mutation, 2013, 34, 706-713.	2.5	70

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109	Recommendations for a nomenclature system for reporting methylation aberrations in imprinted domains. Epigenetics, 2018, 13, 117-121.	2.7	70
110	Characterization and cell type distribution of a novel, major transcript of the Duchenne Muscular Dystrophy gene. Differentiation, 1992, 49, 187-193.	1.9	69
111	Generation and Characterization of Transgenic Mice with the Full-length Human DMD Gene. Journal of Biological Chemistry, 2008, 283, 5899-5907.	3.4	69
112	Flexible and Scalable Full-Length CYP2D6 Long Amplicon PacBio Sequencing. Human Mutation, 2017, 38, 310-316.	2.5	69
113	Common pathological mechanisms in mouse models for muscular dystrophies. FASEB Journal, 2006, 20, 127-129.	0.5	67
114	Keratosis Follicularis Spinulosa Decalvans is caused by mutations in MBTPS2. Human Mutation, 2010, 31, 1125-1133.	2.5	67
115	Mutant huntingtin activates Nrf2-responsive genes and impairs dopamine synthesis in a PC12 model of Huntington's disease. BMC Molecular Biology, 2008, 9, 84.	3.0	66
116	Ataxin-3 protein modification as a treatment strategy for spinocerebellar ataxia type 3: Removal of the CAG containing exon. Neurobiology of Disease, 2013, 58, 49-56.	4.4	66
117	Detecting authorized and unauthorized genetically modified organisms containing vip3A by real-time PCR and next-generation sequencing. Analytical and Bioanalytical Chemistry, 2014, 406, 2603-2611.	3.7	64
118	A common reference for cDNA microarray hybridizations. Nucleic Acids Research, 2002, 30, 116e-116.	14.5	63
119	The Human Variome Project. Science, 2008, 322, 861-862.	12.6	63
120	Reliable and controllable antibody fragment selections from Camelid non-immune libraries for target validation. Biochimica Et Biophysica Acta - Proteins and Proteomics, 2006, 1764, 1307-1319.	2.3	62
121	Relative power and sample size analysis on gene expression profiling data. BMC Genomics, 2009, 10, 439.	2.8	62
122	Increased sensitivity of next generation sequencing-based expression profiling after globin reduction in human blood RNA. BMC Genomics, 2012, 13, 28.	2.8	62
123	Somatic expansion of the (CAG) n repeat in Huntington disease brains. Human Genetics, 1995, 95, 270-274.	3.8	61
124	High resoluation deletion breakpoint mapping in the DMD gene by whole cosmid hybridization. Nucleic Acids Research, 1989, 17, 5611-5621.	14.5	60
125	242 Breakpoints in the 200-kb deletion-prone P20 region of the DMD gene are widely spread. Genomics, 1991, 10, 631-639.	2.9	60
126	Protein studies in dysferlinopathy patients using llama-derived antibody fragments selected by phage display. European Journal of Human Genetics, 2005, 13, 721-730.	2.8	60

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127	Copy number variation in regions flanked (or unflanked) by duplicons among patients with developmental delay and/or congenital malformations; detection of reciprocal and partial Williams-Beuren duplications. European Journal of Human Genetics, 2006, 14, 180-189.	2.8	60
128	The protein truncation test: A review. Human Mutation, 1999, 14, 95-102.	2.5	59
129	Recommendations for locus-specific databases and their curation. Human Mutation, 2008, 29, 2-5.	2.5	59
130	Array-MLPA: comprehensive detection of deletions and duplications and its application to DMD patients. Human Mutation, 2008, 29, 190-197.	2.5	58
131	Strict co-linearity of genetic and protein folding domains in an intragenically duplicated rat lens Î³-crystallin gene. Journal of Molecular Biology, 1983, 171, 353-368.	4.2	57
132	Describing structural changes by extending HGVS sequence variation nomenclature. Human Mutation, 2011, 32, 507-511.	2.5	57
133	Next-generation sequencing-based genome diagnostics across clinical genetics centers: implementation choices and their effects. European Journal of Human Genetics, 2015, 23, 1142-1150.	2.8	56
134	Protein truncation test (PTT) to rapidly screen the DMD gene for translation terminating mutations. Neuromuscular Disorders, 1993, 3, 391-394.	0.6	55
135	A full-body transcriptome and proteome resource for the European common carp. BMC Genomics, 2016, 17, 701.	2.8	55
136	Integrated study of 100 patients with Xp21 linked muscular dystrophy using clinical, genetic, immunochemical, and histopathological data. Part 2. Correlations within individual patients.. Journal of Medical Genetics, 1993, 30, 737-744.	3.2	54
137	Dynamic mutation in Dutch Huntington's disease patients: increased paternal repeat instability extending to within the normal size range.. Journal of Medical Genetics, 1993, 30, 996-1002.	3.2	53
138	Becker muscular dystrophy patients with deletions around exon 51; a promising outlook for exon skipping therapy in Duchenne patients. Neuromuscular Disorders, 2010, 20, 251-254.	0.6	53
139	Two human Î³-crystallin genes are linked and riddled with Alu-repeats. Gene, 1985, 38, 197-204.	2.2	52
140	Mutation rates in the dystrophin gene: A hotspot of mutation at a CpG dinucleotide. Human Mutation, 2005, 25, 177-188.	2.5	52
141	Serum protein profiling in mice: Identification of Factor XIIIa as a potential biomarker for muscular dystrophy. Proteomics, 2008, 8, 1552-1563.	2.2	52
142	The LOVD3 platform: efficient genome-wide sharing of genetic variants. European Journal of Human Genetics, 2021, 29, 1796-1803.	2.8	52
143	Short hypervariable microhaplotypes: A novel set of very short high discriminating power loci without stutter artefacts. Forensic Science International: Genetics, 2018, 35, 169-175.	3.1	51
144	A novel human serine-threonine phosphatase related to the Drosophila retinal degeneration C (rdgC) gene is selectively expressed in sensory neurons of neural crest origin. Human Molecular Genetics, 1997, 6, 1137-1145.	2.9	50

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145	Strong aggregation and increased toxicity of polyleucine over polyglutamine stretches in mammalian cells. Human Molecular Genetics, 2002, 11, 1487-1496.	2.9	50
146	DGGE-based whole-gene mutation scanning of the dystrophin gene in Duchenne and Becker muscular dystrophy patients. Human Mutation, 2004, 23, 57-66.	2.5	50
147	Terminal Osseous Dysplasia Is Caused by a Single Recurrent Mutation in the FLNA Gene. American Journal of Human Genetics, 2010, 87, 146-153.	6.2	50
148	Guidelines for establishing locus specific databases. Human Mutation, 2012, 33, 298-305.	2.5	48
149	WHSC1L1, on Human Chromosome 8p11.2, Closely Resembles WHSC1 and Maps to a Duplicated Region Shared with 4p16.3. Genomics, 2001, 76, 5-8.	2.9	47
150	Developing a set of ancestry-sensitive DNA markers reflecting continental origins of humans. BMC Genetics, 2009, 10, 69.	2.7	47
151	Therapeutic exon skipping for dysferlinopathies?. European Journal of Human Genetics, 2010, 18, 889-894.	2.8	47
152	Preventing Formation of Toxic N-Terminal Huntingtin Fragments Through Antisense Oligonucleotide-Mediated Protein Modification. Nucleic Acid Therapeutics, 2014, 24, 4-12.	3.6	47
153	Embryonic expression patterns of the Drosophila dystrophin-associated glycoprotein complex orthologs. Gene Expression Patterns, 2004, 4, 153-159.	0.8	46
154	Variation of CNV distribution in five different ethnic populations. Cytogenetic and Genome Research, 2007, 118, 19-30.	1.1	46
155	Extensive intragenic sequence homology in two distinct rat lens gamma-crystallin cDNAs suggests duplications of a primordial gene.. Proceedings of the National Academy of Sciences of the United States of America, 1982, 79, 6876-6880.	7.1	45
156	Can subtle changes in gene expression be consistently detected with different microarray platforms?. BMC Genomics, 2008, 9, 124.	2.8	45
157	Functional assessment of TSC1 missense variants identified in individuals with tuberous sclerosis complex. Human Mutation, 2012, 33, 476-479.	2.5	45
158	DeepSAGE Reveals Genetic Variants Associated with Alternative Polyadenylation and Expression of Coding and Non-coding Transcripts. PLoS Genetics, 2013, 9, e1003594.	3.5	45
159	Application of massive parallel sequencing to whole genome SNP discovery in the porcine genome. BMC Genomics, 2009, 10, 374.	2.8	44
160	DetectingPKD1variants in polycystic kidney disease patients by single-molecule long-read sequencing. Human Mutation, 2017, 38, 870-879.	2.5	44
161	A dystrophic Duchenne mouse model for testing human antisense oligonucleotides. PLoS ONE, 2018, 13, e0193289.	2.5	44
162	Integrated clinical and omics approach to rare diseases: novel genes and oligogenic inheritance in holoprosencephaly. Brain, 2019, 142, 35-49.	7.6	44

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163	Mutalyzer 2: next generation HGVS nomenclature checker. Bioinformatics, 2021, 37, 2811-2817.	4.1	44
164	Assessing the translational landscape of myogenic differentiation by ribosome profiling. Nucleic Acids Research, 2015, 43, 4408-4428.	14.5	43
165	Characterization of the rat Î³-crystallin gene family and its expression in the eye lens. Journal of Molecular Biology, 1985, 182, 419-430.	4.2	41
166	Curating gene variant databases (LSDBs): Toward a universal standard. Human Mutation, 2012, 33, 291-297.	2.5	41
167	Novel Protein-Protein Interactions Inferred from Literature Context. PLoS ONE, 2009, 4, e7894.	2.5	41
168	Structural variation in mammalian Î³-crystallins based on computer graphics analyses of human, rat and calf sequences 1. Core packing and surface properties. Experimental Eye Research, 1986, 43, 77-92.	2.6	40
169	Dual exon skipping in myostatin and dystrophin for Duchenne muscular dystrophy. BMC Medical Genomics, 2011, 4, 36.	1.5	40
170	Aging as Accelerated Accumulation of Somatic Variants: Whole-Genome Sequencing of Centenarian and Middle-Aged Monozygotic Twin Pairs. Twin Research and Human Genetics, 2013, 16, 1026-1032.	0.6	40
171	Identification of SCML2, a Second Human Gene Homologous to theDrosophila Sex comb on midleg(Scm): A New Gene Cluster on Xp22. Genomics, 1999, 58, 65-72.	2.9	39
172	Three-tiered noninvasive diagnosis in 96% of patients with Duchenne muscular dystrophy (DMD). Human Mutation, 2004, 23, 203-204.	2.5	39
173	Single Molecule Sequencing of Free DNA from Maternal Plasma for Noninvasive Trisomy 21 Detection. Clinical Chemistry, 2012, 58, 699-706.	3.2	39
174	The genomic landscape of the verrucomicrobial methanotroph Methylacidiphilum fumariolicum SolV. BMC Genomics, 2014, 15, 914.	2.8	39
175	TSSV: a tool for characterization of complex allelic variants in pure and mixed genomes. Bioinformatics, 2014, 30, 1651-1659.	4.1	39
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