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	Stepwise ABC system for classification of any type of genetic variant. European Journal of Human Genetics, 2022, 30, 150-159.	2.8	37
2	New locus underlying auriculocondylar syndrome (ARCND): 430 kb duplication involvingTWIST1regulatory elements. Journal of Medical Genetics, 2022, 59, 895-905.	3.2	4
3	NR2F1database: 112 variants and 84 patients support refining the clinical synopsis of Boschâ€“Boonstraâ€“Schaaf optic atrophy syndrome. Human Mutation, 2022, 43, 128-142.	2.5	12
4	Integrating Whole-Genome Sequencing in Clinical Genetics: A Novel Disruptive Structural Rearrangement Identified in the Dystrophin Gene (DMD). International Journal of Molecular Sciences, 2022, 23, 59.	4.1	3
5	Verifying nomenclature of DNA variants in submitted manuscripts: Guidance for journals. Human Mutation, 2021, 42, 3-7.	2.5	10
6	Data sharing and gene variant databases. , 2021, , 221-236.		0
7	Newborn screening of duchenne muscular dystrophy specifically targeting deletions amenable to exon-skipping therapy. Scientific Reports, 2021, 11, 3011.	3.3	14
8	Mutalyzer 2: next generation HGVS nomenclature checker. Bioinformatics, 2021, 37, 2811-2817.	4.1	44
9	A Case Series of Familial ARID1B Variants Illustrating Variable Expression and Suggestions to Update the ACMG Criteria. Genes, 2021, 12, 1275.	2.4	5
10	ACO2 clinicobiological dataset with extensive phenotype ontology annotation. Scientific Data, 2021, 8, 205.	5.3	2
11	The Role of the European Society of Human Genetics in Delivering Genomic Education. Frontiers in Genetics, 2021, 12, 693952.	2.3	6
12	The LOVD3 platform: efficient genome-wide sharing of genetic variants. European Journal of Human Genetics, 2021, 29, 1796-1803.	2.8	52
13	Using Personal Genomic Data within Primary Care: A Bioinformatics Approach to Pharmacogenomics. Genes, 2020, 11, 1443.	2.4	8
14	Efficient variant data preparation for Human Mutation manuscripts: Variants and phenotypes. Human Mutation, 2019, 40, 1009-1009.	2.5	6
15	Dutch genome diagnostic laboratories accelerated and improved variant interpretation and increased accuracy by sharing data. Human Mutation, 2019, 40, 2230-2238.	2.5	32
16	Templated Insertions: A Smoking Gun for Polymerase Theta-Mediated End Joining. Trends in Genetics, 2019, 35, 632-644.	6.7	103
17	Phenotype predictions for exon deletions/duplications: A user guide for professionals and clinicians using Becker and Duchenne muscular dystrophy as examples. Human Mutation, 2019, 40, 1630-1633.	2.5	8
18	OPA1: 516 unique variants and 831 patients registered in an updated centralized Variome database. Orphanet Journal of Rare Diseases, 2019, 14, 214.	2.7	39

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19	WGS-based telomere length analysis in Dutch family trios implicates stronger maternal inheritance and a role for RRM1 gene. Scientific Reports, 2019, 9, 18758.	3.3	8
20	Skewed X-inactivation is common in the general female population. European Journal of Human Genetics, 2019, 27, 455-465.	2.8	119
21	Integrated clinical and omics approach to rare diseases: novel genes and oligogenic inheritance in holoprosencephaly. Brain, 2019, 142, 35-49.	7.6	44
22	Yet another database?. Human Mutation, 2018, 39, 755-755.	2.5	3
23	Structural and sequence variants in patients with Silver-Russell syndrome or similar features-Curation of a disease database. Human Mutation, 2018, 39, 345-364.	2.5	23
24	Full-length mRNA sequencing uncovers a widespread coupling between transcription initiation and mRNA processing. Genome Biology, 2018, 19, 46.	8.8	106
25	Recommendations for a nomenclature system for reporting methylation aberrations in imprinted domains. Epigenetics, 2018, 13, 117-121.	2.7	70
26	BRCA Challenge: BRCA Exchange as a global resource for variants in BRCA1 and BRCA2. PLoS Genetics, 2018, 14, e1007752.	3.5	148
27	Properties and units in the clinical laboratory sciences part XXIV. Properties and units in clinical molecular genetics (IUPAC Technical Report). Pure and Applied Chemistry, 2018, 90, 1199-1220.	1.9	1
28	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
29	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
30	The InSiGHT Database: An Example LOVD System. , 2018, , 469-478.		0
31	Properties and units in the clinical laboratory sciences part XXIV. Properties and units in clinical molecular genetics (technical report 2017). Clinica Chimica Acta, 2018, 484, 122-131.	1.1	2
32	A dystrophic Duchenne mouse model for testing human antisense oligonucleotides. PLoS ONE, 2018, 13, e0193289.	2.5	44
33	In SilicoFunctional Meta-Analysis of 5,962ABCA4Variants in 3,928 Retinal Dystrophy Cases. Human Mutation, 2017, 38, 400-408.	2.5	118
34	Critical points for an accurate human genome analysis. Human Mutation, 2017, 38, 912-921.	2.5	5
35	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	6.2	305
36	DetectingPKD1variants in polycystic kidney disease patients by single-molecule long-read sequencing. Human Mutation, 2017, 38, 870-879.	2.5	44

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37	Flexible and Scalable Full-Length CYP2D6 Long Amplicon PacBio Sequencing. Human Mutation, 2017, 38, 310-316.	2.5	69
38	The UCL low-density lipoprotein receptor gene variant database: pathogenicity update. Journal of Medical Genetics, 2017, 54, 217-223.	3.2	75
39	Describing Sequence Variants Using HGVS Nomenclature. Methods in Molecular Biology, 2017, 1492, 243-251.	0.9	30
40	Effect of post-mortem delay on N-terminal huntingtin protein fragments in human control and Huntington disease brain lysates. PLoS ONE, 2017, 12, e0178556.	2.5	2
41	Decay of sexual trait genes in an asexual parasitoid wasp. Genome Biology and Evolution, 2016, 8, evw273.	2.5	33
42	The Complete Genome Sequence of the Murine Pathobiont Helicobacter typhlonius. Frontiers in Microbiology, 2016, 6, 1549.	3.5	8
43	The Implicitome: A Resource for Rationalizing Gene-Disease Associations. PLoS ONE, 2016, 11, e0149621.	2.5	22
44	Pharmacogenetic allele nomenclature: International workgroup recommendations for test result reporting. Clinical Pharmacology and Therapeutics, 2016, 99, 172-185.	4.7	146
45	HGVS Nomenclature in Practice: An Example from the United Kingdom National External Quality Assessment Scheme. Human Mutation, 2016, 37, 576-578.	2.5	22
46	Hypomorphic MKS1 mutation in a Pakistani family with mild Joubert syndrome and atypical features: Expanding the phenotypic spectrum of MKS1â€related ciliopathies. American Journal of Medical Genetics, Part A, 2016, 170, 3289-3293.	1.2	9
47	A full-body transcriptome and proteome resource for the European common carp. BMC Genomics, 2016, 17, 701.	2.8	55
48	Sequence Variant Descriptions: HGVS Nomenclature and Mutalyzer. Current Protocols in Human Genetics, 2016, 90, 7.13.1-7.13.19.	3.5	37
49	Deciphering Variability of PKD1 and PKD2 in an Italian Cohort of 643 Patients with Autosomal Dominant Polycystic Kidney Disease (ADPKD). Scientific Reports, 2016, 6, 30850.	3.3	28
50	HGVS Recommendations for the Description of Sequence Variants: 2016 Update. Human Mutation, 2016, 37, 564-569.	2.5	1,194
51	The DNA Bank: High-Security Bank Accounts to Protect and Share Your Genetic Identity. Human Mutation, 2015, 36, 657-659.	2.5	12
52	Collembolan Transcriptomes Highlight Molecular Evolution of Hexapods and Provide Clues on the Adaptation to Terrestrial Life. PLoS ONE, 2015, 10, e0130600.	2.5	25
53	Novel variants in GNAI3 associated with auriculocondylar syndrome strengthen a common dominant negative effect. European Journal of Human Genetics, 2015, 23, 481-485.	2.8	21
54	A novel Fanconi anaemia subtype associated with a dominant-negative mutation in RAD51. Nature Communications, 2015, 6, 8829.	12.8	130

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55	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
56	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
57	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
58	Selection and characterization of llama single domain antibodies against N-terminal huntingtin. Neurological Sciences, 2015, 36, 429-434.	1.9	16
59	SplicePie: a novel analytical approach for the detection of alternative, non-sequential and recursive splicing. Nucleic Acids Research, 2015, 43, e80-e80.	14.5	17
60	Next-Generation Diagnostics: Gene Panel, Exome, or Whole Genome?. Human Mutation, 2015, 36, 648-655.	2.5	124
61	Assessing the translational landscape of myogenic differentiation by ribosome profiling. Nucleic Acids Research, 2015, 43, 4408-4428.	14.5	43
62	Akkermansia muciniphilaandHelicobacter typhloniusmodulate intestinal tumor development in mice. Carcinogenesis, 2015, 36, 1388-1396.	2.8	87
63	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. Human Mutation, 2015, 36, 915-921.	2.5	390
64	The Human Variome Project: ensuring the quality of DNA variant databases in inherited renal disease. Pediatric Nephrology, 2015, 30, 1893-1901.	1.7	2
65	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
66	The genomic landscape of the verrucomicrobial methanotroph Methylacidiphilum fumariolicum SolV. BMC Genomics, 2014, 15, 914.	2.8	39
67	Determining the quality and complexity of next-generation sequencing data without a reference genome. Genome Biology, 2014, 15, 555.	8.8	30
68	Mutation Update: The Spectra of Nebulin Variants and Associated Myopathies. Human Mutation, 2014, 35, 1418-1426.	2.5	107
69	Pathogenic: Light or Dark Skin?. Human Mutation, 2014, 35, 520-520.	2.5	1
70	Comprehensive Registration of DNA Sequence Variants Associated with Inherited Retinal Diseases in Leiden Open Variation Databases. Human Mutation, 2014, 35, 147-148.	2.5	5
71	A 3-base pair deletion, c.9711_9713del, in DMD results in intellectual disability without muscular dystrophy. European Journal of Human Genetics, 2014, 22, 480-485.	2.8	30
72	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

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73	TSSV: a tool for characterization of complex allelic variants in pure and mixed genomes. Bioinformatics, 2014, 30, 1651-1659.	4.1	39
74	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
75	The Genome of the Netherlands: design, and project goals. European Journal of Human Genetics, 2014, 22, 221-227.	2.8	246
76	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
77	Redefining Mutational Spectra via Updated Locus-specific Databases. Human Mutation, 2014, 35, v-v.	2.5	0
78	Preventing Formation of Toxic N-Terminal Huntingtin Fragments Through Antisense Oligonucleotide-Mediated Protein Modification. Nucleic Acid Therapeutics, 2014, 24, 4-12.	3.6	47
79	Targeted sequencing by proximity ligation for comprehensive variant detection and local haplotyping. Nature Biotechnology, 2014, 32, 1019-1025.	17.5	231
80	Next generation sequencing technology: Advances and applications. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 1932-1941.	3.8	557
81	RNA sequencing: from tag-based profiling to resolving complete transcript structure. Cellular and Molecular Life Sciences, 2014, 71, 3537-3551.	5.4	33
82	Whole-genome sequence variation, population structure and demographic history of the Dutch population. Nature Genetics, 2014, 46, 818-825.	21.4	641
83	Molecular diagnostics of the HBB gene in an Omani cohort using bench-top DNA Ion Torrent PGM technology. Blood Cells, Molecules, and Diseases, 2014, 53, 133-137.	1.4	4
84	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
85	The InSiGHT database: utilizing 100Âyears of insights into Lynch Syndrome. Familial Cancer, 2013, 12, 175-180.	1.9	100
86	Reproducibility of high-throughput mRNA and small RNA sequencing across laboratories. Nature Biotechnology, 2013, 31, 1015-1022.	17.5	251
87	Coffin-Siris Syndrome and the BAF Complex: Genotype-Phenotype Study in 63 Patients. Human Mutation, 2013, 34, 1519-1528.	2.5	178
88	Transcriptional responses indicate attenuated oxidative stress in the springtail Folsomia candida exposed to mixtures of cadmium and phenanthrene. Ecotoxicology, 2013, 22, 619-631.	2.4	19
89	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
90	Exome Sequencing Identifies A Branch Point Variant in Aarskog-Scott Syndrome. Human Mutation, 2013, 34, 430-434.	2.5	21

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91	DeepSAGE Reveals Genetic Variants Associated with Alternative Polyadenylation and Expression of Coding and Non-coding Transcripts. PLoS Genetics, 2013, 9, e1003594.	3.5	45
92	Expanding the MTM1 mutational spectrum: novel variants including the first multi-exonic duplication and development of a locus-specific database. European Journal of Human Genetics, 2013, 21, 540-549.	2.8	29
93	DMD transcript imbalance determines dystrophin levels. FASEB Journal, 2013, 27, 4909-4916.	0.5	30
94	<scp>GPSM</scp>2 and Chudleyâ€“<scp>M</scp>c<scp>C</scp>ullough Syndrome: A Dutch Founder Variant Brought to North America. American Journal of Medical Genetics, Part A, 2013, 161, 973-976.	1.2	13
95	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
96	Poly(A) binding protein nuclear 1 levels affect alternative polyadenylation. Nucleic Acids Research, 2012, 40, 9089-9101.	14.5	148
97	Mutations in SWI/SNF chromatin remodeling complex gene ARID1B cause Coffin-Siris syndrome. Nature Genetics, 2012, 44, 379-380.	21.4	312
98	Single Molecule Sequencing of Free DNA from Maternal Plasma for Noninvasive Trisomy 21 Detection. Clinical Chemistry, 2012, 58, 699-706.	3.2	39
99	CLI-mate. , 2012, , .		2
100	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
101	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
102	Increased sensitivity of next generation sequencing-based expression profiling after globin reduction in human blood RNA. BMC Genomics, 2012, 13, 28.	2.8	62
103	The Effects of Low Levels of Dystrophin on Mouse Muscle Function and Pathology. PLoS ONE, 2012, 7, e31937.	2.5	96
104	Phage display screening without repetitious selection rounds. Analytical Biochemistry, 2012, 421, 622-631.	2.4	149
105	Transposon proliferation in an asexual parasitoid. Molecular Ecology, 2012, 21, 3898-3906.	3.9	33
106	Fine-tiling array CGH to improve diagnostics for Î±- and Î²-thalassemia rearrangements. Human Mutation, 2012, 33, 272-280.	2.5	37
107	Curating gene variant databases (LSDBs): Toward a universal standard. Human Mutation, 2012, 33, 291-297.	2.5	41
108	Guidelines for establishing locus specific databases. Human Mutation, 2012, 33, 298-305.	2.5	48

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109	Mandatory variant submission-Our experiences. Human Mutation, 2012, 33, 1-1.	2.5	2
110	Functional assessment of TSC1 missense variants identified in individuals with tuberous sclerosis complex. Human Mutation, 2012, 33, 476-479.	2.5	45
111	Reporting of Genetic Variants by Diagnostic Laboratories and other Centres. Clinical Biochemist Reviews, 2012, 33, 21-4.	3.3	0
112	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
113	The value of data. Nature Genetics, 2011, 43, 281-283.	21.4	126
114	Mutation (variation) databases and registries: a rationale for coordination of efforts. Nature Reviews Genetics, 2011, 12, 881-881.	16.3	11
115	Characterization of novel SLC6A8 variants with the use of splice-site analysis tools and implementation of a newly developed LOVD database. European Journal of Human Genetics, 2011, 19, 56-63.	2.8	18
116	Experiences with array-based sequence capture; toward clinical applications. European Journal of Human Genetics, 2011, 19, 50-55.	2.8	13
117	MutaDATABASE: a centralized and standardized DNA variation database. Nature Biotechnology, 2011, 29, 117-118.	17.5	36
118	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
119	Call for participation in the neurogenetics consortium within the Human Variome Project. Neurogenetics, 2011, 12, 169-173.	1.4	5
120	High throughput nano-liter RT-qPCR to classify soil contamination using a soil arthropod. BMC Molecular Biology, 2011, 12, 11.	3.0	20
121	Dual exon skipping in myostatin and dystrophin for Duchenne muscular dystrophy. BMC Medical Genomics, 2011, 4, 36.	1.5	40
122	A formalized description of the standard human variant nomenclature in Extended Backus-Naur Form. BMC Bioinformatics, 2011, 12, S5.	2.6	18
123	Describing structural changes by extending HGVS sequence variation nomenclature. Human Mutation, 2011, 32, 507-511.	2.5	57
124	LOVD v.2.0: the next generation in gene variant databases. Human Mutation, 2011, 32, 557-563.	2.5	854
125	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
126	Using systematic nomenclature for CFTR variants: Improvement needed. Human Mutation, 2011, 32, v-v.	2.5	0

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127	Literature-aided interpretation of gene expression data with the weighted global test. Briefings in Bioinformatics, 2011, 12, 518-529.	6.5	19
128	Reply to Clarity and claims in variation/mutation databasing. Nature Biotechnology, 2011, 29, 792-794.	17.5	0
129	Genetic Tests Need the Human Variome Project. Genetic Testing and Molecular Biomarkers, 2011, 15, 3-3.	0.7	4
130	mRNA degradation controls differentiation state-dependent differences in transcript and splice variant abundance. Nucleic Acids Research, 2011, 39, 556-566.	14.5	95
131	Targeting Several CAG Expansion Diseases by a Single Antisense Oligonucleotide. PLoS ONE, 2011, 6, e24308.	2.5	85
132	Development of NIPBL Locus-Specific Database Using LOVD: From Novel Mutations to Further Genotype-Phenotype Correlations in Cornelia de Lange Syndrome. Human Mutation, 2010, 31, 1216-1222.	2.5	19
133	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
134	New methods for next generation sequencing based microRNA expression profiling. BMC Genomics, 2010, 11, 716.	2.8	85
135	An overview of L-2-hydroxyglutarate dehydrogenase gene (L2HGDH) variants: a genotype-phenotype study. Human Mutation, 2010, 31, 380-390.	2.5	108
136	High-throughput genotyping of mannose-binding lectin variants using high-resolution DNA-melting analysis. Human Mutation, 2010, 31, E1286-E1293.	2.5	17
137	Keratosis Follicularis Spinulosa Decalvans is caused by mutations in MBTPS2. Human Mutation, 2010, 31, 1125-1133.	2.5	67
138	Practical guidelines addressing ethical issues pertaining to the curation of human locus-specific variation databases (LSDBs). Human Mutation, 2010, 31, 1179-1184.	2.5	36
139	Leiden open variation database of the MUTYH gene. Human Mutation, 2010, 31, 1205-1215.	2.5	72
140	Therapeutic exon skipping for dysferlinopathies?. European Journal of Human Genetics, 2010, 18, 889-894.	2.8	47
141	Accurate quantification of dystrophin mRNA and exon skipping levels in Duchenne Muscular Dystrophy. Laboratory Investigation, 2010, 90, 1396-1402.	3.7	37
142	New insights in geneâ€derived therapy: the example of Duchenne muscular dystrophy. Annals of the New York Academy of Sciences, 2010, 1214, 199-212.	3.8	21
143	Genome-wide assessment of differential roles for p300 and CBP in transcription regulation. Nucleic Acids Research, 2010, 38, 5396-5408.	14.5	133
144	Tissue-specific transcript annotation and expression profiling with complementary next-generation sequencing technologies. Nucleic Acids Research, 2010, 38, e165-e165.	14.5	32

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145	RNA-Based Variant Detection. , 2010, , 293-298.		0
146	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
147	Databases in Human and Medical Genetics. , 2010, , 941-960.		0
148	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
149	Locus Reference Genomic sequences: an improved basis for describing human DNA variants. Genome Medicine, 2010, 2, 24.	8.2	100
150	Comprehensive Gene-Expression Survey Identifies Wif1 as a Modulator of Cardiomyocyte Differentiation. PLoS ONE, 2010, 5, e15504.	2.5	18
151	Application of massive parallel sequencing to whole genome SNP discovery in the porcine genome. BMC Genomics, 2009, 10, 374.	2.8	44
152	Relative power and sample size analysis on gene expression profiling data. BMC Genomics, 2009, 10, 439.	2.8	62
153	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
154	Split handâ€foot malformation, tetralogy of Fallot, mental retardation and a 1 Mb 19p deletionâ€"evidence for further heterogeneity?. American Journal of Medical Genetics, Part A, 2009, 149A, 975-981.	1.2	19
155	Somatic mutation databases as tools for molecular epidemiology and molecular pathology of cancer: Proposed guidelines for improving data collection, distribution, and integration. Human Mutation, 2009, 30, 275-282.	2.5	14
156	Theoretic applicability of antisense-mediated exon skipping for Duchenne muscular dystrophy mutations. Human Mutation, 2009, 30, 293-299.	2.5	485
157	Sharing data between LSDBs and central repositories. Human Mutation, 2009, 30, 493-495.	2.5	18
158	High-Resolution Melting Analysis (HRMA)-More than just sequence variant screening. Human Mutation, 2009, 30, 860-866.	2.5	414
159	Deep sequencing to reveal new variants in pooled DNA samples. Human Mutation, 2009, 30, 1703-1712.	2.5	71
160	Developing a set of ancestry-sensitive DNA markers reflecting continental origins of humans. BMC Genetics, 2009, 10, 69.	2.7	47
161	Identification of a region required for TSC1 stability by functional analysis of TSC1missense mutations found in individuals with tuberous sclerosis complex. BMC Medical Genetics, 2009, 10, 88.	2.1	29
162	Cost-effective HRMA pre-sequence typing of clone libraries; application to phage display selection. BMC Biotechnology, 2009, 9, 50.	3.3	7

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163	Recurrence risk due to germ line mosaicism: Duchenne and Becker muscular dystrophy. Clinical Genetics, 2009, 75, 465-472.	2.0	81
164	Rapid and cost effective detection of small mutations in the DMD gene by high resolution melting curve analysis. Neuromuscular Disorders, 2009, 19, 383-390.	0.6	33
165	A Study of the SORL1 Gene in Alzheimer's Disease and Cognitive Function. Journal of Alzheimer's Disease, 2009, 18, 51-64.	2.6	36
166	Novel Protein-Protein Interactions Inferred from Literature Context. PLoS ONE, 2009, 4, e7894.	2.5	41
167	Novel synonymous substitution in POMGNT1 promotes exon skipping in a patient with congenital muscular dystrophy. Journal of Human Genetics, 2008, 53, 565-572.	2.3	17
168	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
169	Serum protein profiling in mice: Identification of Factor XIIIa as a potential biomarker for muscular dystrophy. Proteomics, 2008, 8, 1552-1563.	2.2	52
170	Array-MLPA: comprehensive detection of deletions and duplications and its application to DMD patients. Human Mutation, 2008, 29, 190-197.	2.5	58
171	Recommendations for locus-specific databases and their curation. Human Mutation, 2008, 29, 2-5.	2.5	59
172	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
173	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
174	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
175	Three Genome-wide Association Studies and a Linkage Analysis Identify HERC2 as a Human Iris Color Gene. American Journal of Human Genetics, 2008, 82, 801.	6.2	4
176	Literature-aided meta-analysis of microarray data: a compendium study on muscle development and disease. BMC Bioinformatics, 2008, 9, 291.	2.6	21
177	CORE_TF: a user-friendly interface to identify evolutionary conserved transcription factor binding sites in sets of co-regulated genes. BMC Bioinformatics, 2008, 9, 495.	2.6	33
178	Can subtle changes in gene expression be consistently detected with different microarray platforms?. BMC Genomics, 2008, 9, 124.	2.8	45
179	Calling on a million minds for community annotation in WikiProteins. Genome Biology, 2008, 9, R89.	9.6	117
180	Generation and Characterization of Transgenic Mice with the Full-length Human DMD Gene. Journal of Biological Chemistry, 2008, 283, 5899-5907.	3.4	69

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181	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
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