Joris A Veltman

List of Publications by Year
in descending order

258	31,550	4388	5255
papers	citations	86 h-index	165 g-index

281	281	281	40879
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Transcriptome and genome sequencing uncovers functional variation in humans. Nature, 2013, 501, 506-511.	27.8	1,857
2	Diagnostic Exome Sequencing in Persons with Severe Intellectual Disability. New England Journal of Medicine, 2012, 367, 1921-1929.	27.0	1,367
3	Mutations in a new member of the chromodomain gene family cause CHARGE syndrome. Nature Genetics, 2004, 36, 955-957.	21.4	1,098
4	Genome sequencing identifies major causes of severe intellectual disability. Nature, 2014, 511, 344-347.	27.8	996
5	A de novo paradigm for mental retardation. Nature Genetics, 2010, 42, 1109-1112.	21.4	751
6	De novo mutations in human genetic disease. Nature Reviews Genetics, 2012, 13, 565-575.	16.3	715
7	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	27.0	663
8	STAT1Mutations in Autosomal Dominant Chronic Mucocutaneous Candidiasis. New England Journal of Medicine, 2011, 365, 54-61.	27.0	614
9	Genetic studies in intellectual disability and related disorders. Nature Reviews Genetics, 2016, 17, 9-18.	16.3	614
10	Identification of common variants associated with human hippocampal and intracranial volumes. Nature Genetics, 2012, 44, 552-561.	21.4	594
11	Diagnostic Genome Profiling in Mental Retardation. American Journal of Human Genetics, 2005, 77, 606-616.	6.2	514
12	Disruption of the neurexin 1 gene is associated with schizophrenia. Human Molecular Genetics, 2009, 18, 988-996.	2.9	424
13	Array-Based Comparative Genomic Hybridization for the Genomewide Detection of Submicroscopic Chromosomal Abnormalities. American Journal of Human Genetics, 2003, 73, 1261-1270.	6.2	423
14	A new chromosome 17q21.31 microdeletion syndrome associated with a common inversion polymorphism. Nature Genetics, 2006, 38, 999-1001.	21.4	418
15	De novo mutations of SETBP1 cause Schinzel-Giedion syndrome. Nature Genetics, 2010, 42, 483-485.	21.4	417
16	Disease gene identification strategies for exome sequencing. European Journal of Human Genetics, 2012, 20, 490-497.	2.8	412
17	Meta-analysis of 2,104 trios provides support for 10 new genes for intellectual disability. Nature Neuroscience, 2016, 19, 1194-1196.	14.8	407
18	CHARGE syndrome: the phenotypic spectrum of mutations in the CHD7 gene. Journal of Medical Genetics, 2005, 43, 306-314.	3.2	382

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19	A recent bottleneck of Y chromosome diversity coincides with a global change in culture. Genome Research, 2015, 25, 459-466.	5.5	348
20	New insights into the generation and role of de novo mutations in health and disease. Genome Biology, 2016, 17, 241.	8.8	339
21	Exome Sequencing Identifies Truncating Mutations in Human SERPINF1 in Autosomal-Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2011, 88, 362-371.	6.2	316
22	A Post-Hoc Comparison of the Utility of Sanger Sequencing and Exome Sequencing for the Diagnosis of Heterogeneous Diseases. Human Mutation, 2013, 34, 1721-1726.	2.5	303
23	CNTNAP2 gene dosage variation is associated with schizophrenia and epilepsy. Molecular Psychiatry, 2008, 13, 261-266.	7.9	300
24	Whole-genome sequencing reveals important role for TBK1 and OPTN mutations in frontotemporal lobar degeneration without motor neuron disease. Acta Neuropathologica, 2015, 130, 77-92.	7.7	267
25	Parent-of-origin-specific signatures of de novo mutations. Nature Genetics, 2016, 48, 935-939.	21.4	266
26	Exome Sequencing Identifies WDR35 Variants Involved in Sensenbrenner Syndrome. American Journal of Human Genetics, 2010, 87, 418-423.	6.2	260
27	Nextâ€generation genetic testing for retinitis pigmentosa. Human Mutation, 2012, 33, 963-972.	2.5	258
28	Recurrent CNVs Disrupt Three Candidate Genes in Schizophrenia Patients. American Journal of Human Genetics, 2008, 83, 504-510.	6.2	248
29	De novo mutations in the actin genes ACTB and ACTG1 cause Baraitser-Winter syndrome. Nature Genetics, 2012, 44, 440-444.	21.4	237
30	De novo nonsense mutations in ASXL1 cause Bohring-Opitz syndrome. Nature Genetics, 2011, 43, 729-731.	21.4	236
31	Multiple Phenotypes in Phosphoglucomutase 1 Deficiency. New England Journal of Medicine, 2014, 370, 533-542.	27.0	236
32	Unlocking Mendelian disease using exome sequencing. Genome Biology, 2011, 12, 228.	9.6	228
33	A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology. Genetics in Medicine, 2017, 19, 1055-1063.	2.4	220
34	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. American Journal of Human Genetics, 2014, 95, 173-182.	6.2	219
35	Post-zygotic Point Mutations Are an Underrecognized Source of De Novo Genomic Variation. American Journal of Human Genetics, 2015, 97, 67-74.	6.2	215
36	Disruption of an EHMT1-Associated Chromatin-Modification Module Causes Intellectual Disability. American Journal of Human Genetics, 2012, 91, 73-82.	6.2	214

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37	Common variants at 12q14 and 12q24 are associated with hippocampal volume. Nature Genetics, 2012, 44, 545-551.	21.4	212
38	Ciliopathies with Skeletal Anomalies and Renal Insufficiency due to Mutations in the IFT-A Gene WDR19. American Journal of Human Genetics, 2011, 89, 634-643.	6.2	210
39	Ultra-sensitive Sequencing Identifies High Prevalence of Clonal Hematopoiesis-Associated Mutations throughout Adult Life. American Journal of Human Genetics, 2017, 101, 50-64.	6.2	210
40	Genomic and Expression Profiling of Human Spermatocytic Seminomas: Primary Spermatocyte as Tumorigenic Precursor and DMRT1 as Candidate Chromosome 9 Gene. Cancer Research, 2006, 66, 290-302.	0.9	208
41	Array-based comparative genomic hybridization for genome-wide screening of DNA copy number in bladder tumors. Cancer Research, 2003, 63, 2872-80.	0.9	208
42	Next-Generation Sequencing of a 40 Mb Linkage Interval Reveals TSPAN12 Mutations in Patients with Familial Exudative Vitreoretinopathy. American Journal of Human Genetics, 2010, 86, 240-247.	6.2	202
43	High-Throughput Analysis of Subtelomeric Chromosome Rearrangements by Use of Array-Based Comparative Genomic Hybridization. American Journal of Human Genetics, 2002, 70, 1269-1276.	6.2	196
44	Identification of Tumor-Specific Molecular Signatures in Intracranial Ependymoma and Association With Clinical Characteristics. Journal of Clinical Oncology, 2006, 24, 5223-5233.	1.6	194
45	Mutations in the chromatin modifier gene KANSL1 cause the 17q21.31 microdeletion syndrome. Nature Genetics, 2012, 44, 639-641.	21.4	194
46	Clinical and molecular delineation of the 17q21.31 microdeletion syndrome. Journal of Medical Genetics, 2008, 45, 710-720.	3.2	191
47	Mutations in ISPD cause Walker-Warburg syndrome and defective glycosylation of Î±-dystroglycan. Nature Genetics, 2012, 44, 581-585.	21.4	191
48	Mutations in LCA5, encoding the ciliary protein lebercilin, cause Leber congenital amaurosis. Nature Genetics, 2007, 39, 889-895.	21.4	186
49	Pyrosequencing of 16S rRNA gene amplicons to study the microbiota in the gastrointestinal tract of carp (Cyprinus carpio L.). AMB Express, 2011, 1, 41.	3.0	186
50	OFD1 Is Mutated in X-Linked Joubert Syndrome and Interacts with LCA5-Encoded Lebercilin. American Journal of Human Genetics, 2009, 85, 465-481.	6.2	180
51	Mutations in the phospholipid remodeling gene SERAC1 impair mitochondrial function and intracellular cholesterol trafficking and cause dystonia and deafness. Nature Genetics, 2012, 44, 797-802.	21.4	175
52	Characterization of a recurrent 15q24 microdeletion syndrome. Human Molecular Genetics, 2007, 16, 567-572.	2.9	173
53	Genome-Wide Profiling of p63 DNAâ€“Binding Sites Identifies an Element that Regulates Gene Expression during Limb Development in the 7q21 SHFM1 Locus. PLoS Genetics, 2010, 6, e1001065.	3.5	169
54	Mutations in DDHD2, Encoding an Intracellular Phospholipase A1, Cause a Recessive Form of Complex Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2012, 91, 1073-1081.	6.2	159

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55	Comparison of Exome and Genome Sequencing Technologies for the Complete Capture of Proteinâ€Coding Regions. Human Mutation, 2015, 36, 815-822.	2.5	156
56	Mutations inDYNC1H1cause severe intellectual disability with neuronal migration defects. Journal of Medical Genetics, 2012, 49, 179-183.	3.2	151
57	Mutations in BICD2, which Encodes a Golgin and Important Motor Adaptor, Cause Congenital Autosomal-Dominant Spinal Muscular Atrophy. American Journal of Human Genetics, 2013, 92, 946-954.	6.2	150
58	Gene identification in the congenital disorders of glycosylation type I by whole-exome sequencing. Human Molecular Genetics, 2012, 21, 4151-4161.	2.9	147
59	ZNF408 is mutated in familial exudative vitreoretinopathy and is crucial for the development of zebrafish retinal vasculature. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 9856-9861.	7.1	144
60	A systematic review and standardized clinical validity assessment of male infertility genes. Human Reproduction, 2019, 34, 932-941.	0.9	144
61	Rare pathogenic microdeletions and tandem duplications are microhomology-mediated and stimulated by local genomic architecture. Human Molecular Genetics, 2009, 18, 3579-3593.	2.9	143
62	Detection of clinically relevant copy-number variants by exome sequencing in a large cohort of genetic disorders. Genetics in Medicine, 2017, 19, 667-675.	2.4	143
63	CantÃº Syndrome Is Caused by Mutations in ABCC9. American Journal of Human Genetics, 2012, 90, 1094-1101.	6.2	141
64	Identification of disease genes by whole genome CGH arrays. Human Molecular Genetics, 2005, 14, R215-R223.	2.9	140
65	Homozygous and heterozygous disruptions of ANK3: at the crossroads of neurodevelopmental and psychiatric disorders. Human Molecular Genetics, 2013, 22, 1960-1970.	2.9	137
66	Genomic microarrays in mental retardation: A practical workflow for diagnostic applications. Human Mutation, 2009, 30, 283-292.	2.5	136
67	Genomic microarrays in mental retardation: from copy number variation to gene, from research to diagnosis. Journal of Medical Genetics, 2010, 47, 289-297.	3.2	135
68	MetaDome: Pathogenicity analysis of genetic variants through aggregation of homologous human protein domains. Human Mutation, 2019, 40, 1030-1038.	2.5	133
69	Array-based comparative genomic hybridization for the differential diagnosis of renal cell cancer. Cancer Research, 2002, 62, 957-60.	0.9	132
70	Exome sequencing identifiesDYNC2H1mutations as a common cause of asphyxiating thoracic dystrophy (Jeune syndrome) without major polydactyly, renal or retinal involvement. Journal of Medical Genetics, 2013, 50, 309-323.	3.2	127
71	Role of gain of 12p in germ cell tumour development. Apmis, 2003, 111, 161-170.	2.0	126
72	Is the $1000 Genome as Near as We Think? A Cost Analysis of Next-Generation Sequencing. Clinical Chemistry, 2016, 62, 1458-1464.	3.2	126

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73	Targeted Next-Generation Sequencing of a 12.5 Mb Homozygous Region Reveals ANO10 Mutations in Patients with Autosomal-Recessive Cerebellar Ataxia. American Journal of Human Genetics, 2010, 87, 813-819.	6.2	125
74	Functional Differences Between Mesenchymal Stem Cell Populations Are Reflected by Their Transcriptome. Stem Cells and Development, 2010, 19, 481-490.	2.1	124
75	A systematic review of the validated monogenic causes of human male infertility: 2020 update and a discussion of emerging geneâ€“disease relationships. Human Reproduction Update, 2021, 28, 15-29.	10.8	121
76	Exome sequencing of Pakistani consanguineous families identifies 30 novel candidate genes for recessive intellectual disability. Molecular Psychiatry, 2017, 22, 1604-1614.	7.9	118
77	ATP6AP1 deficiency causes an immunodeficiency with hepatopathy, cognitive impairment and abnormal protein glycosylation. Nature Communications, 2016, 7, 11600.	12.8	110
78	Genotypeâ€“phenotype mapping of chromosome 18q deletions by highâ€resolution array CGH: An update of the phenotypic map. American Journal of Medical Genetics, Part A, 2007, 143A, 1858-1867.	1.2	106
79	Detection of Clinically Relevant Copy Number Variants with Whole-Exome Sequencing. Human Mutation, 2013, 34, 1439-1448.	2.5	105
80	Definition of a Critical Region on Chromosome 18 for Congenital Aural Atresia by ArrayCGH. American Journal of Human Genetics, 2003, 72, 1578-1584.	6.2	102
81	STAT1 Hyperphosphorylation and Defective IL12R/IL23R Signaling Underlie Defective Immunity in Autosomal Dominant Chronic Mucocutaneous Candidiasis. PLoS ONE, 2011, 6, e29248.	2.5	101
82	Common variants in DGKK are strongly associated with risk of hypospadias. Nature Genetics, 2011, 43, 48-50.	21.4	99
83	Recurrent De Novo Mutations in PACS1 Cause Defective Cranial-Neural-Crest Migration and Define a Recognizable Intellectual-Disability Syndrome. American Journal of Human Genetics, 2012, 91, 1122-1127.	6.2	96
84	Microarray analyses reveal strong influence of DNA copy number alterations on the transcriptional patterns in pancreatic cancer: implications for the interpretation of genomic amplifications. Oncogene, 2005, 24, 1794-1801.	5.9	95
85	Genetic Variation in CACNA1C, a Gene Associated with Bipolar Disorder, Influences Brainstem Rather than Gray Matter Volume in Healthy Individuals. Biological Psychiatry, 2010, 68, 586-588.	1.3	95
86	De novo copy number variants associated with intellectual disability have a paternal origin and age bias. Journal of Medical Genetics, 2011, 48, 776-778.	3.2	95
87	Intragenic deletion in DYRK1A leads to mental retardation and primary microcephaly. Clinical Genetics, 2011, 79, 296-299.	2.0	94
88	TRIOloss of function is associated with mild intellectual disability and affects dendritic branching and synapse function. Human Molecular Genetics, 2016, 25, 892-902.	2.9	94
89	Identification of pathogenic gene variants in small families with intellectually disabled siblings by exome sequencing. Journal of Medical Genetics, 2013, 50, 802-811.	3.2	93
90	De novo mutations in beta-catenin (CTNNB1) appear to be a frequent cause of intellectual disability: expanding the mutational and clinical spectrum. Human Genetics, 2015, 134, 97-109.	3.8	93

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91	Homozygosity Mapping in Patients with Coneâ€“Rod Dystrophy: Novel Mutations and Clinical Characterizations. , 2010, 51, 5943.		92
92	Trisomy for Synaptojanin1 in Down syndrome is functionally linked to the enlargement of early endosomes. Human Molecular Genetics, 2012, 21, 3156-3172.	2.9	92
93	Genome-wide Copy Number Profiling on High-density Bacterial Artificial Chromosomes, Single-nucleotide Polymorphisms, and Oligonucleotide Microarrays: A Platform Comparison based on Statistical Power Analysis. DNA Research, 2007, 14, 1-11.	3.4	91
94	Identification of Novel Mutations in Patients with Leber Congenital Amaurosis and Juvenile RP by Genome-wide Homozygosity Mapping with SNP Microarrays. , 2007, 48, 5690.		90
95	Chondrodysplasia and Abnormal Joint Development Associated with Mutations in IMPAD1, Encoding the Golgi-Resident Nucleotide Phosphatase, gPAPP. American Journal of Human Genetics, 2011, 88, 608-615.	6.2	88
96	CCDC115 Deficiency Causes a Disorder of Golgi Homeostasis with Abnormal Protein Glycosylation. American Journal of Human Genetics, 2016, 98, 310-321.	6.2	88
97	Massively parallel sequencing of ataxia genes after array-based enrichment. Human Mutation, 2010, 31, 494-499.	2.5	86
98	Mobster: accurate detection of mobile element insertions in next generation sequencing data. Genome Biology, 2014, 15, 488.	8.8	86
99	Chromosomal copy number changes in patients with non-syndromic X linked mental retardation detected by array CGH. Journal of Medical Genetics, 2005, 43, 362-370.	3.2	85
100	Clinical and cytogenetic characterization of 13 Dutch patients with deletion 9p syndrome: Delineation of the critical region for a consensus phenotype. American Journal of Medical Genetics, Part A, 2008, 146A, 1430-1438.	1.2	85
101	<scp>MLL2</scp> mutation detection in 86 patients with Kabuki syndrome: a genotypeâ€“phenotype study. Clinical Genetics, 2013, 84, 539-545.	2.0	85
102	Spatial Clustering of de Novo Missense Mutations Identifies Candidate Neurodevelopmental Disorder-Associated Genes. American Journal of Human Genetics, 2017, 101, 478-484.	6.2	84
103	Disruption of the Podosome Adaptor Protein TKS4 (SH3PXD2B) Causes the Skeletal Dysplasia, Eye, and Cardiac Abnormalities of Frank-Ter Haar Syndrome. American Journal of Human Genetics, 2010, 86, 254-261.	6.2	83
104	Genome-Wide Array-Based Comparative Genomic Hybridization Reveals Multiple Amplification Targets and Novel Homozygous Deletions in Pancreatic Carcinoma Cell Lines. Cancer Research, 2004, 64, 3052-3059.	0.9	82
105	Mutations in C8orf37, Encoding a Ciliary Protein, are Associated with Autosomal-Recessive Retinal Dystrophies with Early Macular Involvement. American Journal of Human Genetics, 2012, 90, 102-109.	6.2	82
106	De Novo Mutations in SON Disrupt RNA Splicing of Genes Essential for Brain Development and Metabolism, Causing an Intellectual-Disability Syndrome. American Journal of Human Genetics, 2016, 99, 711-719.	6.2	81
107	Heterozygous Mutations of FREM1 Are Associated with an Increased Risk of Isolated Metopic Craniosynostosis in Humans and Mice. PLoS Genetics, 2011, 7, e1002278.	3.5	80
108	De Novo Mutations Reflect Development and Aging of the Human Germline. Trends in Genetics, 2019, 35, 828-839.	6.7	80

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109	Whole-exome sequencing reveals LRP5 mutations and canonical Wnt signaling associated with hepatic cystogenesis. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 5343-5348.	7.1	79
110	Next-generation DNA sequencing identifies novel gene variants and pathways involved in specific language impairment. Scientific Reports, 2017, 7, 46105.	3.3	79
111	A mutation in the FAM36A gene, the human ortholog of COX20, impairs cytochrome c oxidase assembly and is associated with ataxia and muscle hypotonia. Human Molecular Genetics, 2013, 22, 656-667.	2.9	75
112	Deleterious variants in X-linked CFAP47 induce asthenoteratozoospermia and primary male infertility. American Journal of Human Genetics, 2021, 108, 309-323.	6.2	74
113	Reduced purifying selection prevails over positive selection in human copy number variant evolution. Genome Research, 2008, 18, 1711-1723.	5.5	73
114	Exome sequencing and whole genome sequencing for the detection of copy number variation. Expert Review of Molecular Diagnostics, 2015, 15, 1023-1032.	3.1	73
115	Understanding the Psychosocial Effects of WES Test Results on Parents of Children with Rare Diseases. Journal of Genetic Counseling, 2016, 25, 1207-1214.	1.6	73
116	TMEM199 Deficiency Is a Disorder of Golgi Homeostasis Characterized by Elevated Aminotransferases, Alkaline Phosphatase, and Cholesterol and Abnormal Glycosylation. American Journal of Human Genetics, 2016, 98, 322-330.	6.2	73
117	12p-Amplicon structure analysis in testicular germ cell tumors of adolescents and adults by array CGH. Oncogene, 2003, 22, 7695-7701.	5.9	72
118	Exome sequencing identifies putative drivers of progression of transient myeloproliferative disorder to AMKL in infants with Down syndrome. Blood, 2013, 122, 554-561.	1.4	72
119	The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype. European Journal of Human Genetics, 2010, 18, 163-170.	2.8	71
120	Mutations in the interleukin receptor <scp>IL</scp>11<scp>RA</scp> cause autosomal recessive Crouzonâ€like craniosynostosis. Molecular Genetics & Genomic Medicine, 2013, 1, 223-237.	1.2	70
121	Germline de novo mutation clusters arise during oocyte aging in genomic regions with high double-strand-break incidence. Nature Genetics, 2018, 50, 487-492.	21.4	68
122	Bi-allelic Mutations in M1AP Are a Frequent Cause of Meiotic Arrest and Severely Impaired Spermatogenesis Leading to Male Infertility. American Journal of Human Genetics, 2020, 107, 342-351.	6.2	68
123	Understanding variable expressivity in microdeletion syndromes. Nature Genetics, 2010, 42, 192-193.	21.4	67
124	Anomalies of the CD8+ T cell pool in haemochromatosis: HLA-A3-linked expansions of CD8+ CD28â^' T cells. Clinical and Experimental Immunology, 1997, 107, 548-554.	2.6	65
125	Molecular karyotyping of patients with unexplained mental retardation by SNP arrays: A multicenter study. Human Mutation, 2009, 30, 1082-1092.	2.5	65
126	GATAD2Bloss-of-function mutations cause a recognisable syndrome with intellectual disability and are associated with learning deficits and synaptic undergrowth inDrosophila. Journal of Medical Genetics, 2013, 50, 507-514.	3.2	63

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127	Validation Study of Existing Gene Expression Signatures for Anti-TNF Treatment in Patients with Rheumatoid Arthritis. PLoS ONE, 2012, 7, e33199.	2.5	61
128	Heterozygous germline mutations in A2ML1 are associated with a disorder clinically related to Noonan syndrome. European Journal of Human Genetics, 2015, 23, 317-324.	2.8	61
129	Mutations Affecting the SAND Domain of DEAF1 Cause Intellectual Disability with Severe Speech Impairment and Behavioral Problems. American Journal of Human Genetics, 2014, 94, 649-661.	6.2	59
130	Homozygosity Mapping Reveals Mutations of GRXCR1 as a Cause of Autosomal-Recessive Nonsyndromic Hearing Impairment. American Journal of Human Genetics, 2010, 86, 138-147.	6.2	58
131	Association of the Alzheimer's GeneSORL1With Hippocampal Volume in Young, Healthy Adults. American Journal of Psychiatry, 2011, 168, 1083-1089.	7.2	58
132	Exome sequencing identifies a de novo <scp>SCN</scp>2<scp>A</scp> mutation in a patient with intractable seizures, severe intellectual disability, optic atrophy, muscular hypotonia, and brain abnormalities. Epilepsia, 2014, 55, e25-9.	5.1	58
133	Mutations in TPRN Cause a Progressive Form of Autosomal-Recessive Nonsyndromic Hearing Loss. American Journal of Human Genetics, 2010, 86, 479-484.	6.2	56
134	Nuclear ReceptorsNur77andNurr1Modulate Mesenchymal Stromal Cell Migration. Stem Cells and Development, 2012, 21, 228-238.	2.1	56
135	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
136	De Novo Truncating Mutations in the Last and Penultimate Exons of PPM1D Cause an Intellectual Disability Syndrome. American Journal of Human Genetics, 2017, 100, 650-658.	6.2	56
137	Variant PNLDC1, Defective piRNA Processing, and Azoospermia. New England Journal of Medicine, 2021, 385, 707-719.	27.0	54
138	Somatic variants in autosomal dominant genes are a rare cause of sporadic Alzheimer's disease. Alzheimer's and Dementia, 2018, 14, 1632-1639.	0.8	51
139	Front Cover, Volume 40, Issue 8. Human Mutation, 2019, 40, i-i.	2.5	51
140	High resolution profiling of X chromosomal aberrations by array comparative genomic hybridisation. Journal of Medical Genetics, 2004, 41, 425-432.	3.2	50
141	Exome Sequencing in an Admixed Isolated Population Indicates NFXL1 Variants Confer a Risk for Specific Language Impairment. PLoS Genetics, 2015, 11, e1004925.	3.5	50
142	Disease gene discovery in male infertility: past, present and future. Human Genetics, 2021, 140, 7-19.	3.8	50
143	Loss of a small region around thePTEN locus is a major chromosome 10 alteration in prostate cancer xenografts and cell lines. Genes Chromosomes and Cancer, 2004, 39, 171-184.	2.8	49
144	Exome sequencing in routine diagnostics: a generic test for 254 patients with primary immunodeficiencies. Genome Medicine, 2019, 11, 38.	8.2	49

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145	Chromosome Instability as an Indicator of Malignant Progression in Laryngeal Mucosa. Journal of Clinical Oncology, 2000, 18, 1644-1651.	1.6	48
146	Wholeâ€exome sequencing detects somatic mutations of IDH1 in metaphyseal chondromatosis with <scp>D</scp>â€2â€hydroxyglutaric aciduria (MCâ€HGA). American Journal of Medical Genetics, Part A, 2011, 155, 2609-2616.	1.2	47
147	Variation of CNV distribution in five different ethnic populations. Cytogenetic and Genome Research, 2007, 118, 19-30.	1.1	46
148	Trisomic dose of several chromosome 21 genes perturbs haematopoietic stem and progenitor cell differentiation in Down's syndrome. Oncogene, 2010, 29, 6102-6114.	5.9	46
149	Accurate Distinction of Pathogenic from Benign CNVs in Mental Retardation. PLoS Computational Biology, 2010, 6, e1000752.	3.2	46
150	Validation and application of a novel integrated genetic screening method to a cohort of 1,112 men with idiopathic azoospermia or severe oligozoospermia. Human Mutation, 2017, 38, 1592-1605.	2.5	45
151	Human Mitochondrial Complex I Deficiency: Investigating Transcriptional Responses by Microarray. Neuropediatrics, 2003, 34, 14-22.	0.6	44
152	Diagnostic Genome Profiling. Journal of Molecular Diagnostics, 2006, 8, 534-537.	2.8	44
153	Pathogenic or not? Assessing the clinical relevance of copy number variants. Clinical Genetics, 2013, 84, 415-421.	2.0	44
154	Point mutations as a source of de novo genetic disease. Current Opinion in Genetics and Development, 2013, 23, 257-263.	3.3	44
155	Pathogenic variants in glutamyl-tRNAGln amidotransferase subunits cause a lethal mitochondrial cardiomyopathy disorder. Nature Communications, 2018, 9, 4065.	12.8	44
156	Ovotestes and XY sex reversal in a female with an interstitial 9q33.3â€q34.1 deletion encompassing NR5A1 and LMX1B causing features of genitopatellar syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1071-1081.	1.2	43
157	Molecular characterisation of patients with subtelomeric 22q abnormalities using chromosome specific array-based comparative genomic hybridisation. European Journal of Human Genetics, 2005, 13, 1019-1024.	2.8	42
158	Identification of recurrent chromosomal aberrations in germ cell tumors of neonates and infants using genomewide array-based comparative genomic hybridization. Genes Chromosomes and Cancer, 2005, 43, 367-376.	2.8	41
159	Diagnostic serum glycosylation profile in patients with intellectual disability as a result of MAN1B1 deficiency. Brain, 2014, 137, 1030-1038.	7.6	41
160	Forging Links between Human Mental Retardationâ€“Associated CNVs and Mouse Gene Knockout Models. PLoS Genetics, 2009, 5, e1000531.	3.5	40
161	The diagnostic pathway in complex paediatric neurology: A cost analysis. European Journal of Paediatric Neurology, 2015, 19, 233-239.	1.6	40
162	The role of de novo mutations in adult-onset neurodegenerative disorders. Acta Neuropathologica, 2019, 137, 183-207.	7.7	39

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163	Genomic microarrays in clinical diagnosis. Current Opinion in Pediatrics, 2006, 18, 598-603.	2.0	38
164	Disruption of Teashirt Zinc Finger Homeobox 1 Is Associated with Congenital Aural Atresia in Humans. American Journal of Human Genetics, 2011, 89, 813-819.	6.2	38
165	A systems genomics approach identifies SIGLEC15 as a susceptibility factor in recurrent vulvovaginal candidiasis. Science Translational Medicine, 2019, 11, .	12.4	38
166	A de novo paradigm for male infertility. Nature Communications, 2022, 13, 154.	12.8	38
167	Novel bioinformatic developments for exome sequencing. Human Genetics, 2016, 135, 603-614.	3.8	37
168	Exome sequencing reveals novel causes as well as new candidate genes for human globozoospermia. Human Reproduction, 2020, 35, 240-252.	0.9	37
169	Chromosomal breakpoint mapping by arrayCGH using flow-sorted chromosomes. BioTechniques, 2003, 35, 1066-1070.	1.8	36
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