Joris A Veltman

List of Publications by Year
in descending order

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Version: 2024-02-01

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	Screening by single-molecule molecular inversion probes targeted sequencing panel of candidate genes of infertility in azoospermic infertile Jordanian males. Human Fertility, 2022, 25, 939-946.	1.7	2
2	A de novo paradigm for male infertility. Nature Communications, 2022, 13, 154.	12.8	38
3	De novo mutations in children born after medical assisted reproduction. Human Reproduction, 2022, 37, 1360-1369.	0.9	12
4	Large-scale analyses of the X chromosome in 2,354 infertile men discover recurrently affected genes associated with spermatogenic failure. American Journal of Human Genetics, 2022, 109, 1458-1471.	6.2	10
5	Disease gene discovery in male infertility: past, present and future. Human Genetics, 2021, 140, 7-19.	3.8	50
6	A global approach to addressing the policy, research and social challenges of male reproductive health. Human Reproduction Open, 2021, 2021, hoab009.	5.4	19
7	Deleterious variants in X-linked CFAP47 induce asthenoteratozoospermia and primary male infertility. American Journal of Human Genetics, 2021, 108, 309-323.	6.2	74
8	Lack of evidence for a role of PIWIL1 variants in human male infertility. Cell, 2021, 184, 1941-1942.	28.9	11
9	Variants in GCNA, X-linked germ-cell genome integrity gene, identified in men with primary spermatogenic failure. Human Genetics, 2021, 140, 1169-1182.	3.8	27
10	Exome sequencing reveals variants in known and novel candidate genes for severe sperm motility disorders. Human Reproduction, 2021, 36, 2597-2611.	0.9	32
11	Differences in the number of de novo mutations between individuals are due to small family-specific effects and stochasticity. Genome Research, 2021, 31, 1513-1518.	5.5	6
12	Variant PNLDC1, Defective piRNA Processing, and Azoospermia. New England Journal of Medicine, 2021, 385, 707-719.	27.0	54
13	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
14	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
15	Programmed Cell Death 2-Like (Pdcd2l) Is Required for Mouse Embryonic Development. G3: Genes, Genomes, Genetics, 2020, 10, 4449-4457.	1.8	2
16	Mutations in the Vâ€ATPase Assembly Factor VMA21 Cause a Congenital Disorder of Glycosylation With Autophagic Liver Disease. Hepatology, 2020, 72, 1968-1986.	7.3	32
17	Exome sequencing reveals novel causes as well as new candidate genes for human globozoospermia. Human Reproduction, 2020, 35, 240-252.	0.9	37
18	Aberrant Expressions and Variant Screening of SEMA3D in Indonesian Hirschsprung Patients. Frontiers in Pediatrics, 2020, 8, 60.	1.9	7

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19	De Novo Mutations Reflect Development and Aging of the Human Germline. Trends in Genetics, 2019, 35, 828-839.	6.7	80
20	Front Cover, Volume 40, Issue 8. Human Mutation, 2019, 40, i-i.	2.5	51
21	De novo variants in FBXO11 cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms. European Journal of Human Genetics, 2019, 27, 738-746.	2.8	32
22	Missense variants in NOX1 and p22phox in a case of very-early-onset inflammatory bowel disease are functionally linked to NOD2. Journal of Physical Education and Sports Management, 2019, 5, a002428.	1.2	13
23	Exome sequencing in routine diagnostics: a generic test for 254 patients with primary immunodeficiencies. Genome Medicine, 2019, 11, 38.	8.2	49
24	A systems genomics approach identifies SIGLEC15 as a susceptibility factor in recurrent vulvovaginal candidiasis. Science Translational Medicine, 2019, 11, .	12.4	38
25	MetaDome: Pathogenicity analysis of genetic variants through aggregation of homologous human protein domains. Human Mutation, 2019, 40, 1030-1038.	2.5	133
26	A systematic review and standardized clinical validity assessment of male infertility genes. Human Reproduction, 2019, 34, 932-941.	0.9	144
27	The role of de novo mutations in adult-onset neurodegenerative disorders. Acta Neuropathologica, 2019, 137, 183-207.	7.7	39
28	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
29	Estimation of minimal disease prevalence from population genomic data: Application to primary familial brain calcification. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 68-74.	1.7	33
30	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. European Journal of Human Genetics, 2018, 26, 54-63.	2.8	32
31	Pathogenic variants in glutamyl-tRNAGln amidotransferase subunits cause a lethal mitochondrial cardiomyopathy disorder. Nature Communications, 2018, 9, 4065.	12.8	44
32	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
33	Upstream SLC2A1 translation initiation causes GLUT1 deficiency syndrome. European Journal of Human Genetics, 2017, 25, 771-774.	2.8	15
34	Rare NOX3 Variants Confer Susceptibility to Agranulocytosis During Thyrostatic Treatment of Graves' Disease. Clinical Pharmacology and Therapeutics, 2017, 102, 1017-1024.	4.7	12
35	Next-generation DNA sequencing identifies novel gene variants and pathways involved in specific language impairment. Scientific Reports, 2017, 7, 46105.	3.3	79
36	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

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37	A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology. Genetics in Medicine, 2017, 19, 1055-1063.	2.4	220
38	MST1R mutation as a genetic cause of Lady Windermere syndrome. European Respiratory Journal, 2017, 49, 1601478.	6.7	18
39	Copy Number Variation in Syndromic Forms of Psychiatric Illness: The Emerging Value of Clinical Genetic Testing in Psychiatry. American Journal of Psychiatry, 2017, 174, 1036-1050.	7.2	16
40	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
41	Biallelic variants inWARS2encoding mitochondrial tryptophanyl-tRNA synthase in six individuals with mitochondrial encephalopathy. Human Mutation, 2017, 38, 1786-1795.	2.5	24
42	Aggregation of populationâ€based genetic variation over protein domain homologues and its potential use in genetic diagnostics. Human Mutation, 2017, 38, 1454-1463.	2.5	36
43	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
44	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
45	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
46	Exome sequencing of Pakistani consanguineous families identifies 30 novel candidate genes for recessive intellectual disability. Molecular Psychiatry, 2017, 22, 1604-1614.	7.9	118
47	Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. PLoS Genetics, 2017, 13, e1006683.	3.5	35
48	Different Balance of Wnt Signaling in Adult and Fetal Bone Marrow-Derived Mesenchymal Stromal Cells. Stem Cells and Development, 2016, 25, 934-947.	2.1	14
49	De novo lossâ€ofâ€function mutations in Xâ€linked <scp>SMC1A</scp> cause severe <scp>ID</scp> and therapyâ€resistant epilepsy in females: expanding the phenotypic spectrum. Clinical Genetics, 2016, 90, 413-419.	2.0	32
50	New insights into the generation and role of de novo mutations in health and disease. Genome Biology, 2016, 17, 241.	8.8	339
51	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
52	The Cost-Effectiveness of whole-Exome Sequencing in Complex Paediatric Neurology. Value in Health, 2016, 19, A695.	0.3	1
53	Novel bioinformatic developments for exome sequencing. Human Genetics, 2016, 135, 603-614.	3.8	37
54	Immunologic defects in severe mucocutaneous HSV-2 infections: Response to IFN-Î³ therapy. Journal of Allergy and Clinical Immunology, 2016, 138, 895-898.	2.9	6

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55	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
56	Meta-analysis of 2,104 trios provides support for 10 new genes for intellectual disability. Nature Neuroscience, 2016, 19, 1194-1196.	14.8	407
57	Chromosomal abnormalities in hepatic cysts point to novel polycystic liver disease genes. European Journal of Human Genetics, 2016, 24, 1707-1714.	2.8	14
58	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
59	ATP6AP1 deficiency causes an immunodeficiency with hepatopathy, cognitive impairment and abnormal protein glycosylation. Nature Communications, 2016, 7, 11600.	12.8	110
60	Sequence variation between 462 human individuals fine-tunes functional sites of RNA processing. Scientific Reports, 2016, 6, 32406.	3.3	28
61	Parent-of-origin-specific signatures of de novo mutations. Nature Genetics, 2016, 48, 935-939.	21.4	266
62	De novo loss-of-function mutations in WAC cause a recognizable intellectual disability syndrome and learning deficits in Drosophila. European Journal of Human Genetics, 2016, 24, 1145-1153.	2.8	34
63	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
64	Influence of paternal age on ongoing pregnancy rate at eight weeks' gestation in assisted reproduction. Reproductive BioMedicine Online, 2016, 32, 96-103.	2.4	21
65	CCDC115 Deficiency Causes a Disorder of Golgi Homeostasis with Abnormal Protein Glycosylation. American Journal of Human Genetics, 2016, 98, 310-321.	6.2	88
66	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
67	Genetic studies in intellectual disability and related disorders. Nature Reviews Genetics, 2016, 17, 9-18.	16.3	614
68	Missense variants in AIMP1 gene are implicated in autosomal recessive intellectual disability without neurodegeneration. European Journal of Human Genetics, 2016, 24, 392-399.	2.8	17
69	LRP5 variants may contribute to ADPKD. European Journal of Human Genetics, 2016, 24, 237-242.	2.8	28
70	Evaluating a counselling strategy for diagnostic WES in paediatric neurology: an exploration of parents' information and communication needs. Clinical Genetics, 2016, 89, 244-250.	2.0	22
71	A Next-Generation Framework: Deciding On The Role Of Costs In The Clinical Use Of Targeted Gene Panels, Exome And Genome Sequencing. Value in Health, 2015, 18, A352.	0.3	2
72	Comparison of Exome and Genome Sequencing Technologies for the Complete Capture of Proteinâ€Coding Regions. Human Mutation, 2015, 36, 815-822.	2.5	156

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73	From genes to genomes in the clinic. Genome Medicine, 2015, 7, 78.	8.2	13
74	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
75	A missense mutation underlies defective <scp>SOCS</scp>4 function in a family with autoimmunity. Journal of Internal Medicine, 2015, 278, 203-210.	6.0	6
76	The diagnostic pathway in complex paediatric neurology: A cost analysis. European Journal of Paediatric Neurology, 2015, 19, 233-239.	1.6	40
77	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
78	Homozygous mutation of STXBP5L explains an autosomal recessive infantile-onset neurodegenerative disorder. Human Molecular Genetics, 2015, 24, 2000-2010.	2.9	25
79	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
80	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
81	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
82	A recent bottleneck of Y chromosome diversity coincides with a global change in culture. Genome Research, 2015, 25, 459-466.	5.5	348
83	Standardized phenotyping enhances Mendelian disease gene identification. Nature Genetics, 2015, 47, 1222-1224.	21.4	17
84	Cell-Free RNA Is a Reliable Fetoplacental Marker in Noninvasive Fetal Sex Determination. Clinical Chemistry, 2015, 61, 1515-1523.	3.2	11
85	Advantages and Disadvantages of Different Implementation Strategies of Non-Invasive Prenatal Testing in Down Syndrome Screening Programmes. Public Health Genomics, 2015, 18, 260-271.	1.0	5
86	Patient experiences with gene panels based on exome sequencing in clinical diagnostics: high acceptance and low distress. Clinical Genetics, 2015, 87, 319-326.	2.0	23
87	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
88	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
89	Exome Sequencing Identifies Three Novel Candidate Genes Implicated in Intellectual Disability. PLoS ONE, 2014, 9, e112687.	2.5	23
90	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

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91	Mobster: accurate detection of mobile element insertions in next generation sequencing data. Genome Biology, 2014, 15, 488.	8.8	86
92	The effect of enamel matrix derivative (EmdogainÂ®) on gene expression profiles of human primary alveolar bone cells. Journal of Tissue Engineering and Regenerative Medicine, 2014, 8, 463-472.	2.7	17
93	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
94	Diagnostic serum glycosylation profile in patients with intellectual disability as a result of MAN1B1 deficiency. Brain, 2014, 137, 1030-1038.	7.6	41
95	Clinical exome sequencing in daily practice: 1,000 patients and beyond. Genome Medicine, 2014, 6, 2.	8.2	23
96	Platform comparison of detecting copy number variants with microarrays and whole-exome sequencing. Genomics Data, 2014, 2, 144-146.	1.3	13
97	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. American Journal of Human Genetics, 2014, 95, 173-182.	6.2	219
98	Multiple Phenotypes in Phosphoglucomutase 1 Deficiency. New England Journal of Medicine, 2014, 370, 533-542.	27.0	236
99	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
100	Genome sequencing identifies major causes of severe intellectual disability. Nature, 2014, 511, 344-347.	27.8	996
101	Detection of Clinically Relevant Copy Number Variants with Whole-Exome Sequencing. Human Mutation, 2013, 34, 1439-1448.	2.5	105
102	Pathogenic or not? Assessing the clinical relevance of copy number variants. Clinical Genetics, 2013, 84, 415-421.	2.0	44
103	A different balance in wnt-signaling in adult and fetal bone marrow-derived MSC. Experimental Hematology, 2013, 41, S75.	0.4	0
104	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
105	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
106	Homozygous and heterozygous disruptions of ANK3: at the crossroads of neurodevelopmental and psychiatric disorders. Human Molecular Genetics, 2013, 22, 1960-1970.	2.9	137
107	Breast cancer size estimation with MRI in BRCA mutation carriers and other high risk patients. European Journal of Radiology, 2013, 82, 1416-1422.	2.6	18
108	<scp>MLL2</scp> mutation detection in 86 patients with Kabuki syndrome: a genotypeâ€“phenotype study. Clinical Genetics, 2013, 84, 539-545.	2.0	85

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109	Transcriptome and genome sequencing uncovers functional variation in humans. Nature, 2013, 501, 506-511.	27.8	1,857
110	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
111	A compound heterozygous mutation in DPAGT1 results in a congenital disorder of glycosylation with a relatively mild phenotype. European Journal of Human Genetics, 2013, 21, 844-849.	2.8	25
112	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
113	Point mutations as a source of de novo genetic disease. Current Opinion in Genetics and Development, 2013, 23, 257-263.	3.3	44
114	Challenges for implementing next-generation sequencing-based genome diagnostics: itâ€™s also the people, not just the machines. Personalized Medicine, 2013, 10, 473-484.	1.5	9
115	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
116	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
117	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
118	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
119	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
120	Novel PI3KÎ³ Mutation in a 44-Year-Old Man with Chronic Infections and Chronic Pelvic Pain. PLoS ONE, 2013, 8, e68118.	2.5	2
121	Genome and exome sequencing in the clinic: unbiased genomic approaches with a high diagnostic yield. Pharmacogenomics, 2012, 13, 511-514.	1.3	19
122	An integrated framework of personalized medicine: from individual genomes to participatory health care. Croatian Medical Journal, 2012, 53, 301-303.	0.7	25
123	A microduplication of the Rubinsteinâ€“Taybi region on 16p13.3 in a girl with a bilateral complete cleft lip and palate and severe mental retardation. Clinical Dysmorphology, 2012, 21, 204-207.	0.3	7
124	Gene identification in the congenital disorders of glycosylation type I by whole-exome sequencing. Human Molecular Genetics, 2012, 21, 4151-4161.	2.9	147
125	Nuclear ReceptorsNur77andNurr1Modulate Mesenchymal Stromal Cell Migration. Stem Cells and Development, 2012, 21, 228-238.	2.1	56
126	Mutations inDYNC1H1cause severe intellectual disability with neuronal migration defects. Journal of Medical Genetics, 2012, 49, 179-183.	3.2	151

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127	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
128	Mutations in ISPD cause Walker-Warburg syndrome and defective glycosylation of Î±-dystroglycan. Nature Genetics, 2012, 44, 581-585.	21.4	191
129	Common variants at 12q14 and 12q24 are associated with hippocampal volume. Nature Genetics, 2012, 44, 545-551.	21.4	212
130	Diagnostic Exome Sequencing in Persons with Severe Intellectual Disability. New England Journal of Medicine, 2012, 367, 1921-1929.	27.0	1,367
131	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
132	Nextâ€generation genetic testing for retinitis pigmentosa. Human Mutation, 2012, 33, 963-972.	2.5	258
133	Targeted Next Generation Sequencing Reveals a Novel Intragenic Deletion of the TPO Gene in a Family with Intellectual Disability. Archives of Medical Research, 2012, 43, 312-316.	3.3	8
134	Mutations in the chromatin modifier gene KANSL1 cause the 17q21.31 microdeletion syndrome. Nature Genetics, 2012, 44, 639-641.	21.4	194
135	De novo mutations in the actin genes ACTB and ACTG1 cause Baraitser-Winter syndrome. Nature Genetics, 2012, 44, 440-444.	21.4	237
136	Identification of common variants associated with human hippocampal and intracranial volumes. Nature Genetics, 2012, 44, 552-561.	21.4	594
137	Disruption of an EHMT1-Associated Chromatin-Modification Module Causes Intellectual Disability. American Journal of Human Genetics, 2012, 91, 73-82.	6.2	214
138	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
139	De novo diagnostics of patients with intellectual disability. BMC Proceedings, 2012, 6, .	1.6	0
140	Validation Study of Existing Gene Expression Signatures for Anti-TNF Treatment in Patients with Rheumatoid Arthritis. PLoS ONE, 2012, 7, e33199.	2.5	61
141	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
142	Disease gene identification strategies for exome sequencing. European Journal of Human Genetics, 2012, 20, 490-497.	2.8	412
143	De novo mutations in human genetic disease. Nature Reviews Genetics, 2012, 13, 565-575.	16.3	715
144	Structural Genomic Variation in Intellectual Disability. Methods in Molecular Biology, 2012, 838, 77-95.	0.9	10

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145	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
146	Resolving the Breakpoints of the 17q21.31 Microdeletion Syndrome with Next-Generation Sequencing. American Journal of Human Genetics, 2012, 90, 599-613.	6.2	22
147	CantÃº Syndrome Is Caused by Mutations in ABCC9. American Journal of Human Genetics, 2012, 90, 1094-1101.	6.2	141
148	Analysis of genes regulated by the transcription factor LUMAN identifies ApoA4 as a target gene in dendritic cells. Molecular Immunology, 2012, 50, 66-73.	2.2	18
149	Amplified segment in the â€˜Down Syndrome critical regionâ€™ on HSA21 shared between Down syndrome and euploid AML-M0 excludes RUNX1, ERG and ETS2. British Journal of Haematology, 2012, 157, 197-200.	2.5	10
150	STAT1Mutations in Autosomal Dominant Chronic Mucocutaneous Candidiasis. New England Journal of Medicine, 2011, 365, 54-61.	27.0	614
151	De novo mutations in mental retardation. Genome Biology, 2011, 12, .	8.8	0
152	De novo nonsense mutations in ASXL1 cause Bohring-Opitz syndrome. Nature Genetics, 2011, 43, 729-731.	21.4	236
153	Unlocking Mendelian disease using exome sequencing. Genome Biology, 2011, 12, 228.	9.6	228
154	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
155	STAT1 Hyperphosphorylation and Defective IL12R/IL23R Signaling Underlie Defective Immunity in Autosomal Dominant Chronic Mucocutaneous Candidiasis. PLoS ONE, 2011, 6, e29248.	2.5	101
156	Intragenic deletion in DYRK1A leads to mental retardation and primary microcephaly. Clinical Genetics, 2011, 79, 296-299.	2.0	94
157	Common variants in DGKK are strongly associated with risk of hypospadias. Nature Genetics, 2011, 43, 48-50.	21.4	99
158	Homozygosity mapping in outbred families with mental retardation. European Journal of Human Genetics, 2011, 19, 597-601.	2.8	25
159	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
160	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
161	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
162	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

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163	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
164	Comprehensive genetic analysis of OEIS complex reveals no evidence for a recurrent microdeletion or duplication. American Journal of Medical Genetics, Part A, 2011, 155, 38-49.	1.2	20
165	A novel Xp22.11 deletion causing a syndrome of craniosynostosis and periventricular nodular heterotopia. American Journal of Medical Genetics, Part A, 2011, 155, 3144-3147.	1.2	5
166	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
167	Association of the Alzheimer's GeneSORL1With Hippocampal Volume in Young, Healthy Adults. American Journal of Psychiatry, 2011, 168, 1083-1089.	7.2	58
168	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
169	Diagnostic Genome Profiling in Mental Retardation. Advances in Neurobiology, 2011, , 177-194.	1.8	0
170	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
171	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
172	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
173	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
174	Exome Sequencing Identifies WDR35 Variants Involved in Sensenbrenner Syndrome. American Journal of Human Genetics, 2010, 87, 418-423.	6.2	260
175	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
176	Massively parallel sequencing of ataxia genes after array-based enrichment. Human Mutation, 2010, 31, 494-499.	2.5	86
177	Recurrent deletion of ZNF630 at Xp11.23 is not associated with mental retardation. American Journal of Medical Genetics, Part A, 2010, 152A, 638-645.	1.2	5
178	High density gene expression microarrays and gene ontology analysis for identifying processes in implanted tissue engineering constructs. Biomaterials, 2010, 31, 8299-8312.	11.4	19
179	The CASPR2 cell adhesion molecule functions as a tumor suppressor gene in glioma. Oncogene, 2010, 29, 6138-6148.	5.9	28
180	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

#	Article	IF	Citations
181	The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype. European Journal of Human Genetics, 2010, 18, 163-170.	2.8	71
182	De novo mutations of SETBP1 cause Schinzel-Giedion syndrome. Nature Genetics, 2010, 42, 483-485.	21.4	417
183	A de novo paradigm for mental retardation. Nature Genetics, 2010, 42, 1109-1112.	21.4	751
184	Understanding variable expressivity in microdeletion syndromes. Nature Genetics, 2010, 42, 192-193.	21.4	67
185	Homozygosity Mapping in Patients with Coneâ€“Rod Dystrophy: Novel Mutations and Clinical Characterizations. , 2010, 51, 5943.		92
186	Recurrent Inversion Events at 17q21.31 Microdeletion Locus Are Linked to the <i>MAPT</i> H2 Haplotype. Cytogenetic and Genome Research, 2010, 129, 275-279.	1.1	27
187	Accurate Distinction of Pathogenic from Benign CNVs in Mental Retardation. PLoS Computational Biology, 2010, 6, e1000752.	3.2	46
188	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
189	Periventricular Heterotopia in Common Microdeletion Syndromes. Molecular Syndromology, 2010, 1, 35-41.	0.8	30
190	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
191	Functional Differences Between Mesenchymal Stem Cell Populations Are Reflected by Their Transcriptome. Stem Cells and Development, 2010, 19, 481-490.	2.1	124
192	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
193	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
194	Forging Links between Human Mental Retardationâ€“Associated CNVs and Mouse Gene Knockout Models. PLoS Genetics, 2009, 5, e1000531.	3.5	40
195	Disruption of the neurexin 1 gene is associated with schizophrenia. Human Molecular Genetics, 2009, 18, 988-996.	2.9	424
196	Genomic microarrays in mental retardation: A practical workflow for diagnostic applications. Human Mutation, 2009, 30, 283-292.	2.5	136
197	Molecular karyotyping of patients with unexplained mental retardation by SNP arrays: A multicenter study. Human Mutation, 2009, 30, 1082-1092.	2.5	65
198	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

#	Article	IF	Citations
199	Transchromosomic cell model of Down syndrome shows aberrant migration, adhesion and proteome response to extracellular matrix. Proteome Science, 2009, 7, 31.	1.7	16
200	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
201	Recurrent CNVs Disrupt Three Candidate Genes in Schizophrenia Patients. American Journal of Human Genetics, 2008, 83, 504-510.	6.2	248
202	CNTNAP2 gene dosage variation is associated with schizophrenia and epilepsy. Molecular Psychiatry, 2008, 13, 261-266.	7.9	300
203	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	27.0	663
204	Mapping of 5q35 chromosomal rearrangements within a genomically unstable region. Journal of Medical Genetics, 2008, 45, 672-678.	3.2	7
205	Reduced purifying selection prevails over positive selection in human copy number variant evolution. Genome Research, 2008, 18, 1711-1723.	5.5	73
206	Clinical and molecular delineation of the 17q21.31 microdeletion syndrome. Journal of Medical Genetics, 2008, 45, 710-720.	3.2	191
207	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
208	Characterization of a recurrent 15q24 microdeletion syndrome. Human Molecular Genetics, 2007, 16, 567-572.	2.9	173
209	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
210	High-resolution genomic microarrays for X-linked mental retardation. Genetics in Medicine, 2007, 9, 560-565.	2.4	18
211	Whole-Genome Array Comparative Genome Hybridization. Journal of Molecular Diagnostics, 2007, 9, 277.	2.8	12
212	Variation of CNV distribution in five different ethnic populations. Cytogenetic and Genome Research, 2007, 118, 19-30.	1.1	46
213	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
214	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
215	Mutations in LCA5, encoding the ciliary protein lebercilin, cause Leber congenital amaurosis. Nature Genetics, 2007, 39, 889-895.	21.4	186
216	Diagnostics: Genomic copy number analysis in mental retardation: finding the needles in the haystack. European Journal of Human Genetics, 2007, 15, 1-2.	2.8	5

#	Article	IF	Citations
217	Complex chromosome 17p rearrangements associated with low-copy repeats in two patients with congenital anomalies. Human Genetics, 2007, 121, 697-709.	3.8	26
218	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
219	Diagnostic Genome Profiling. Journal of Molecular Diagnostics, 2006, 8, 534-537.	2.8	44
220	Genomic microarrays in clinical diagnosis. Current Opinion in Pediatrics, 2006, 18, 598-603.	2.0	38
221	A novel 2.3â€‰Mb microduplication of 12q24.21q24.23 detected by genome-wide tiling-path resolution array comparative genomic hybridization in a girl with syndromic mental retardation. Clinical Dysmorphology, 2006, 15, 133-137.	0.3	8
222	Novel candidate tumour suppressor gene loci on chromosomes 11q23â€“24 and 22q13 involved in human insulinoma tumourigenesis. Journal of Pathology, 2006, 210, 450-458.	4.5	31
223	A new chromosome 17q21.31 microdeletion syndrome associated with a common inversion polymorphism. Nature Genetics, 2006, 38, 999-1001.	21.4	418
224	Holoprosencephaly and preaxial polydactyly associated with a 1.24ÂMb duplication encompassing FBXW11 at 5q35.1. Journal of Human Genetics, 2006, 51, 721-726.	2.3	18
225	Interstitial 2.2 Mb deletion at 9q34 in a patient with mental retardation but without classical features of the 9q subtelomeric deletion syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 618-623.	1.2	21
226	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
227	Molecular parameters associated with insulinoma progression: chromosomal instability versus p53 and CK19 status. Cytogenetic and Genome Research, 2006, 115, 289-297.	1.1	23
228	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
229	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
230	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
231	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
232	CHARGE syndrome: the phenotypic spectrum of mutations in the CHD7 gene. Journal of Medical Genetics, 2005, 43, 306-314.	3.2	382
233	Diagnostic Genome Profiling in Mental Retardation. American Journal of Human Genetics, 2005, 77, 606-616.	6.2	514
234	Identification of disease genes by whole genome CGH arrays. Human Molecular Genetics, 2005, 14, R215-R223.	2.9	140

#	Article	IF	Citations
235	P49: Identification ofÂrecurrent chromosomal aberrations inÂdifferent types ofÂhuman germ cell tumours using array CGH. European Journal of Medical Genetics, 2005, 48, 511-512.	1.3	0
236	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
237	A novel microdeletion, del(2)(q22.3q23.3) in a mentally retarded patient, detected by array-based comparative genomic hybridization. Clinical Genetics, 2004, 65, 429-432.	2.0	22
238	Mutations in a new member of the chromodomain gene family cause CHARGE syndrome. Nature Genetics, 2004, 36, 955-957.	21.4	1,098
239	Claes Lundsteenâ€"in Memoriam. European Journal of Human Genetics, 2004, 12, 603-603.	2.8	0
240	Chromosome 22q11 deletion and pachygyria characterized by array-based comparative genomic hybridization. American Journal of Medical Genetics Part A, 2004, 131A, 322-324.	2.4	15
241	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
242	High resolution profiling of X chromosomal aberrations by array comparative genomic hybridisation. Journal of Medical Genetics, 2004, 41, 425-432.	3.2	50
243	Role of gain of 12p in germ cell tumour development. Apmis, 2003, 111, 161-170.	2.0	126
244	12p-Amplicon structure analysis in testicular germ cell tumors of adolescents and adults by array CGH. Oncogene, 2003, 22, 7695-7701.	5.9	72
245	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
246	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
247	Human Mitochondrial Complex I Deficiency: Investigating Transcriptional Responses by Microarray. Neuropediatrics, 2003, 34, 14-22.	0.6	44
248	Chromosomal breakpoint mapping by arrayCGH using flow-sorted chromosomes. BioTechniques, 2003, 35, 1066-1070.	1.8	36
249	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
250	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
251	Array-based comparative genomic hybridization for the differential diagnosis of renal cell cancer. Cancer Research, 2002, 62, 957-60.	0.9	132
252	Specific steps in aneuploidization correlate with loss of heterozygosity of 9p21, 17p13 and 18q21 in the progression of pre-malignant laryngeal lesions. International Journal of Cancer, 2001, 91, 193-199.	5.1	3

#	Article	IF	Citations
253	Mapping of resection margins of oral cancer for p53 overexpression and chromosome instability to detect residual (pre)malignant cells. Journal of Pathology, 2001, 193, 66-72.	4.5	26
254	Specific steps in aneuploidization correlate with loss of heterozygosity of 9p21, 17p13 and 18q21 in the progression of preâ€malignant laryngeal lesions. International Journal of Cancer, 2001, 91, 193-199.	5.1	22
255	Chromosome Instability as an Indicator of Malignant Progression in Laryngeal Mucosa. Journal of Clinical Oncology, 2000, 18, 1644-1651.	1.6	48
256	Double-target fluorescence in situ hybridization distinguishes multiple genetically aberrant clones in head and neck squamous cell carcinoma. , 1998, 34, 113-120.		15
257	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
258	Detection of chromosomal aberrations in cytologic brush specimens from head and neck squamous cell carcinoma. , 1997, 81, 309-314.		19