

# Joris A Veltman

## List of Publications by Year in descending order

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Version: 2024-02-01

258  
papers

31,550  
citations

4370

86  
h-index

5227

165  
g-index

281  
all docs

281  
docs citations

281  
times ranked

40879  
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. <i>Human Fertility</i> , 2022, 25, 939-946.	0.7	2
2	A de novo paradigm for male infertility. <i>Nature Communications</i> , 2022, 13, 154.	5.8	38
3	De novo mutations in children born after medical assisted reproduction. <i>Human Reproduction</i> , 2022, 37, 1360-1369.	0.4	12
4	Large-scale analyses of the X chromosome in 2,354 infertile men discover recurrently affected genes associated with spermatogenic failure. <i>American Journal of Human Genetics</i> , 2022, 109, 1458-1471.	2.6	10
5	Disease gene discovery in male infertility: past, present and future. <i>Human Genetics</i> , 2021, 140, 7-19.	1.8	50
6	A global approach to addressing the policy, research and social challenges of male reproductive health. <i>Human Reproduction Open</i> , 2021, 2021, hoab009.	2.3	19
7	Deleterious variants in X-linked CFAP47 induce asthenoteratozoospermia and primary male infertility. <i>American Journal of Human Genetics</i> , 2021, 108, 309-323.	2.6	74
8	Lack of evidence for a role of PIWIL1 variants in human male infertility. <i>Cell</i> , 2021, 184, 1941-1942.	13.5	11
9	Variants in GCNA, X-linked germ-cell genome integrity gene, identified in men with primary spermatogenic failure. <i>Human Genetics</i> , 2021, 140, 1169-1182.	1.8	27
10	Exome sequencing reveals variants in known and novel candidate genes for severe sperm motility disorders. <i>Human Reproduction</i> , 2021, 36, 2597-2611.	0.4	32
11	Differences in the number of de novo mutations between individuals are due to small family-specific effects and stochasticity. <i>Genome Research</i> , 2021, 31, 1513-1518.	2.4	6
12	Variant PNLDC1, Defective piRNA Processing, and Azoospermia. <i>New England Journal of Medicine</i> , 2021, 385, 707-719.	13.9	54
13	A systematic review of the validated monogenic causes of human male infertility: 2020 update and a discussion of emerging gene-disease relationships. <i>Human Reproduction Update</i> , 2021, 28, 15-29.	5.2	121
14	Bi-allelic Mutations in MIAP Are a Frequent Cause of Meiotic Arrest and Severely Impaired Spermatogenesis Leading to Male Infertility. <i>American Journal of Human Genetics</i> , 2020, 107, 342-351.	2.6	68
15	Programmed Cell Death 2-Like (Pcd2l) Is Required for Mouse Embryonic Development. <i>G3: Genes, Genomes, Genetics</i> , 2020, 10, 4449-4457.	0.8	2
16	Mutations in the ATPase Assembly Factor VMA21 Cause a Congenital Disorder of Glycosylation With Autophagic Liver Disease. <i>Hepatology</i> , 2020, 72, 1968-1986.	3.6	32
17	Exome sequencing reveals novel causes as well as new candidate genes for human globozoospermia. <i>Human Reproduction</i> , 2020, 35, 240-252.	0.4	37
18	Aberrant Expressions and Variant Screening of SEMA3D in Indonesian Hirschsprung Patients. <i>Frontiers in Pediatrics</i> , 2020, 8, 60.	0.9	7

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19	De Novo Mutations Reflect Development and Aging of the Human Germline. Trends in Genetics, 2019, 35, 828-839.	2.9	80
20	Front Cover, Volume 40, Issue 8. Human Mutation, 2019, 40, i-i.	1.1	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.	1.4	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.	0.5	13
23	Exome sequencing in routine diagnostics: a generic test for 254 patients with primary immunodeficiencies. Genome Medicine, 2019, 11, 38.	3.6	49
24	A systems genomics approach identifies <i>SIGLEC15</i> as a susceptibility factor in recurrent vulvovaginal candidiasis. Science Translational Medicine, 2019, 11, .	5.8	38
25	MetaDome: Pathogenicity analysis of genetic variants through aggregation of homologous human protein domains. Human Mutation, 2019, 40, 1030-1038.	1.1	133
26	A systematic review and standardized clinical validity assessment of male infertility genes. Human Reproduction, 2019, 34, 932-941.	0.4	144
27	The role of de novo mutations in adult-onset neurodegenerative disorders. Acta Neuropathologica, 2019, 137, 183-207.	3.9	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.	9.4	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.	1.1	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.	1.4	32
31	Pathogenic variants in glutamyl-tRNA <sup>Gln</sup> amidotransferase subunits cause a lethal mitochondrial cardiomyopathy disorder. Nature Communications, 2018, 9, 4065.	5.8	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.	0.4	51
33	Upstream SLC2A1 translation initiation causes GLUT1 deficiency syndrome. European Journal of Human Genetics, 2017, 25, 771-774.	1.4	15
34	Rare NOX3 Variants Confer Susceptibility to Agranulocytosis During Thyrostatic Treatment of Graves' Disease. Clinical Pharmacology and Therapeutics, 2017, 102, 1017-1024.	2.3	12
35	Next-generation DNA sequencing identifies novel gene variants and pathways involved in specific language impairment. Scientific Reports, 2017, 7, 46105.	1.6	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.	2.6	56

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37	A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology. <i>Genetics in Medicine</i> , 2017, 19, 1055-1063.	1.1	220
38	<i>MST1R</i> mutation as a genetic cause of Lady Windermere syndrome. <i>European Respiratory Journal</i> , 2017, 49, 1601478.	3.1	18
39	Copy Number Variation in Syndromic Forms of Psychiatric Illness: The Emerging Value of Clinical Genetic Testing in Psychiatry. <i>American Journal of Psychiatry</i> , 2017, 174, 1036-1050.	4.0	16
40	Spatial Clustering of de Novo Missense Mutations Identifies Candidate Neurodevelopmental Disorder-Associated Genes. <i>American Journal of Human Genetics</i> , 2017, 101, 478-484.	2.6	84
41	Biallelic variants in <i>WARS2</i> encoding mitochondrial tryptophanyl-tRNA synthase in six individuals with mitochondrial encephalopathy. <i>Human Mutation</i> , 2017, 38, 1786-1795.	1.1	24
42	Aggregation of population-based genetic variation over protein domain homologues and its potential use in genetic diagnostics. <i>Human Mutation</i> , 2017, 38, 1454-1463.	1.1	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. <i>Human Mutation</i> , 2017, 38, 1592-1605.	1.1	45
44	Ultra-sensitive Sequencing Identifies High Prevalence of Clonal Hematopoiesis-Associated Mutations throughout Adult Life. <i>American Journal of Human Genetics</i> , 2017, 101, 50-64.	2.6	210
45	Detection of clinically relevant copy-number variants by exome sequencing in a large cohort of genetic disorders. <i>Genetics in Medicine</i> , 2017, 19, 667-675.	1.1	143
46	Exome sequencing of Pakistani consanguineous families identifies 30 novel candidate genes for recessive intellectual disability. <i>Molecular Psychiatry</i> , 2017, 22, 1604-1614.	4.1	118
47	Overlapping <i>SETBP1</i> gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. <i>PLoS Genetics</i> , 2017, 13, e1006683.	1.5	35
48	Different Balance of Wnt Signaling in Adult and Fetal Bone Marrow-Derived Mesenchymal Stromal Cells. <i>Stem Cells and Development</i> , 2016, 25, 934-947.	1.1	14
49	De novo loss-of-function mutations in <i>X-linked SMC1A</i> cause severe ID and therapy-resistant epilepsy in females: expanding the phenotypic spectrum. <i>Clinical Genetics</i> , 2016, 90, 413-419.	1.0	32
50	New insights into the generation and role of de novo mutations in health and disease. <i>Genome Biology</i> , 2016, 17, 241.	3.8	339
51	Is the \$1000 Genome as Near as We Think? A Cost Analysis of Next-Generation Sequencing. <i>Clinical Chemistry</i> , 2016, 62, 1458-1464.	1.5	126
52	The Cost-Effectiveness of whole-Exome Sequencing in Complex Paediatric Neurology. <i>Value in Health</i> , 2016, 19, A695.	0.1	1
53	Novel bioinformatic developments for exome sequencing. <i>Human Genetics</i> , 2016, 135, 603-614.	1.8	37
54	Immunologic defects in severe mucocutaneous HSV-2 infections: Response to IFN- $\gamma$ therapy. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 895-898.	1.5	6

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55	Understanding the Psychosocial Effects of WES Test Results on Parents of Children with Rare Diseases. <i>Journal of Genetic Counseling</i> , 2016, 25, 1207-1214.	0.9	73
56	Meta-analysis of 2,104 trios provides support for 10 new genes for intellectual disability. <i>Nature Neuroscience</i> , 2016, 19, 1194-1196.	7.1	407
57	Chromosomal abnormalities in hepatic cysts point to novel polycystic liver disease genes. <i>European Journal of Human Genetics</i> , 2016, 24, 1707-1714.	1.4	14
58	De Novo Mutations in SON Disrupt RNA Splicing of Genes Essential for Brain Development and Metabolism, Causing an Intellectual-Disability Syndrome. <i>American Journal of Human Genetics</i> , 2016, 99, 711-719.	2.6	81
59	ATP6AP1 deficiency causes an immunodeficiency with hepatopathy, cognitive impairment and abnormal protein glycosylation. <i>Nature Communications</i> , 2016, 7, 11600.	5.8	110
60	Sequence variation between 462 human individuals fine-tunes functional sites of RNA processing. <i>Scientific Reports</i> , 2016, 6, 32406.	1.6	28
61	Parent-of-origin-specific signatures of de novo mutations. <i>Nature Genetics</i> , 2016, 48, 935-939.	9.4	266
62	De novo loss-of-function mutations in WAC cause a recognizable intellectual disability syndrome and learning deficits in <i>Drosophila</i> . <i>European Journal of Human Genetics</i> , 2016, 24, 1145-1153.	1.4	34
63	TRIO loss of function is associated with mild intellectual disability and affects dendritic branching and synapse function. <i>Human Molecular Genetics</i> , 2016, 25, 892-902.	1.4	94
64	Influence of paternal age on ongoing pregnancy rate at eight weeks' gestation in assisted reproduction. <i>Reproductive BioMedicine Online</i> , 2016, 32, 96-103.	1.1	21
65	CCDC115 Deficiency Causes a Disorder of Golgi Homeostasis with Abnormal Protein Glycosylation. <i>American Journal of Human Genetics</i> , 2016, 98, 310-321.	2.6	88
66	TMEM199 Deficiency Is a Disorder of Golgi Homeostasis Characterized by Elevated Aminotransferases, Alkaline Phosphatase, and Cholesterol and Abnormal Glycosylation. <i>American Journal of Human Genetics</i> , 2016, 98, 322-330.	2.6	73
67	Genetic studies in intellectual disability and related disorders. <i>Nature Reviews Genetics</i> , 2016, 17, 9-18.	7.7	614
68	Missense variants in AIMP1 gene are implicated in autosomal recessive intellectual disability without neurodegeneration. <i>European Journal of Human Genetics</i> , 2016, 24, 392-399.	1.4	17
69	LRP5 variants may contribute to ADPKD. <i>European Journal of Human Genetics</i> , 2016, 24, 237-242.	1.4	28
70	Evaluating a counselling strategy for diagnostic WES in paediatric neurology: an exploration of parents' information and communication needs. <i>Clinical Genetics</i> , 2016, 89, 244-250.	1.0	22
71	A Next-Generation Framework: Deciding On The Role Of Costs In The Clinical Use Of Targeted Gene Panels, Exome And Genome Sequencing. <i>Value in Health</i> , 2015, 18, A352.	0.1	2
72	Comparison of Exome and Genome Sequencing Technologies for the Complete Capture of Protein-Coding Regions. <i>Human Mutation</i> , 2015, 36, 815-822.	1.1	156

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73	From genes to genomes in the clinic. <i>Genome Medicine</i> , 2015, 7, 78.	3.6	13
74	Post-zygotic Point Mutations Are an Underrecognized Source of De Novo Genomic Variation. <i>American Journal of Human Genetics</i> , 2015, 97, 67-74.	2.6	215
75	A missense mutation underlies defective <scp>SOCS</scp>4 function in a family with autoimmunity. <i>Journal of Internal Medicine</i> , 2015, 278, 203-210.	2.7	6
76	The diagnostic pathway in complex paediatric neurology: A cost analysis. <i>European Journal of Paediatric Neurology</i> , 2015, 19, 233-239.	0.7	40
77	Next-generation sequencing-based genome diagnostics across clinical genetics centers: implementation choices and their effects. <i>European Journal of Human Genetics</i> , 2015, 23, 1142-1150.	1.4	56
78	Homozygous mutation of STXBP5L explains an autosomal recessive infantile-onset neurodegenerative disorder. <i>Human Molecular Genetics</i> , 2015, 24, 2000-2010.	1.4	25
79	Exome sequencing and whole genome sequencing for the detection of copy number variation. <i>Expert Review of Molecular Diagnostics</i> , 2015, 15, 1023-1032.	1.5	73
80	Exome Sequencing in an Admixed Isolated Population Indicates NFXL1 Variants Confer a Risk for Specific Language Impairment. <i>PLoS Genetics</i> , 2015, 11, e1004925.	1.5	50
81	Whole-genome sequencing reveals important role for TBK1 and OPTN mutations in frontotemporal lobar degeneration without motor neuron disease. <i>Acta Neuropathologica</i> , 2015, 130, 77-92.	3.9	267
82	A recent bottleneck of Y chromosome diversity coincides with a global change in culture. <i>Genome Research</i> , 2015, 25, 459-466.	2.4	348
83	Standardized phenotyping enhances Mendelian disease gene identification. <i>Nature Genetics</i> , 2015, 47, 1222-1224.	9.4	17
84	Cell-Free RNA Is a Reliable Fetoplacental Marker in Noninvasive Fetal Sex Determination. <i>Clinical Chemistry</i> , 2015, 61, 1515-1523.	1.5	11
85	Advantages and Disadvantages of Different Implementation Strategies of Non-Invasive Prenatal Testing in Down Syndrome Screening Programmes. <i>Public Health Genomics</i> , 2015, 18, 260-271.	0.6	5
86	Patient experiences with gene panels based on exome sequencing in clinical diagnostics: high acceptance and low distress. <i>Clinical Genetics</i> , 2015, 87, 319-326.	1.0	23
87	De novo mutations in beta-catenin (CTNNB1) appear to be a frequent cause of intellectual disability: expanding the mutational and clinical spectrum. <i>Human Genetics</i> , 2015, 134, 97-109.	1.8	93
88	Heterozygous germline mutations in A2ML1 are associated with a disorder clinically related to Noonan syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 317-324.	1.4	61
89	Exome Sequencing Identifies Three Novel Candidate Genes Implicated in Intellectual Disability. <i>PLoS ONE</i> , 2014, 9, e112687.	1.1	23
90	Whole-exome sequencing reveals <i>LRP5</i> mutations and canonical Wnt signaling associated with hepatic cystogenesis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 5343-5348.	3.3	79

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91	Mobster: accurate detection of mobile element insertions in next generation sequencing data. <i>Genome Biology</i> , 2014, 15, 488.	3.8	86
92	The effect of enamel matrix derivative (Emdogain®) on gene expression profiles of human primary alveolar bone cells. <i>Journal of Tissue Engineering and Regenerative Medicine</i> , 2014, 8, 463-472.	1.3	17
93	Exome sequencing identifies a de novo <i>SCN2A</i> mutation in a patient with intractable seizures, severe intellectual disability, optic atrophy, muscular hypotonia, and brain abnormalities. <i>Epilepsia</i> , 2014, 55, e25-9.	2.6	58
94	Diagnostic serum glycosylation profile in patients with intellectual disability as a result of <i>MAN1B1</i> deficiency. <i>Brain</i> , 2014, 137, 1030-1038.	3.7	41
95	Clinical exome sequencing in daily practice: 1,000 patients and beyond. <i>Genome Medicine</i> , 2014, 6, 2.	3.6	23
96	Platform comparison of detecting copy number variants with microarrays and whole-exome sequencing. <i>Genomics Data</i> , 2014, 2, 144-146.	1.3	13
97	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. <i>American Journal of Human Genetics</i> , 2014, 95, 173-182.	2.6	219
98	Multiple Phenotypes in Phosphoglucomutase 1 Deficiency. <i>New England Journal of Medicine</i> , 2014, 370, 533-542.	13.9	236
99	Mutations Affecting the SAND Domain of <i>DEAF1</i> Cause Intellectual Disability with Severe Speech Impairment and Behavioral Problems. <i>American Journal of Human Genetics</i> , 2014, 94, 649-661.	2.6	59
100	Genome sequencing identifies major causes of severe intellectual disability. <i>Nature</i> , 2014, 511, 344-347.	13.7	996
101	Detection of Clinically Relevant Copy Number Variants with Whole-Exome Sequencing. <i>Human Mutation</i> , 2013, 34, 1439-1448.	1.1	105
102	Pathogenic or not? Assessing the clinical relevance of copy number variants. <i>Clinical Genetics</i> , 2013, 84, 415-421.	1.0	44
103	A different balance in wnt-signaling in adult and fetal bone marrow-derived MSC. <i>Experimental Hematology</i> , 2013, 41, S75.	0.2	0
104	Identification of pathogenic gene variants in small families with intellectually disabled siblings by exome sequencing. <i>Journal of Medical Genetics</i> , 2013, 50, 802-811.	1.5	93
105	A Post-Hoc Comparison of the Utility of Sanger Sequencing and Exome Sequencing for the Diagnosis of Heterogeneous Diseases. <i>Human Mutation</i> , 2013, 34, 1721-1726.	1.1	303
106	Homozygous and heterozygous disruptions of <i>ANK3</i> : at the crossroads of neurodevelopmental and psychiatric disorders. <i>Human Molecular Genetics</i> , 2013, 22, 1960-1970.	1.4	137
107	Breast cancer size estimation with MRI in <i>BRCA</i> mutation carriers and other high risk patients. <i>European Journal of Radiology</i> , 2013, 82, 1416-1422.	1.2	18
108	<i>MLL2</i> mutation detection in 86 patients with Kabuki syndrome: a genotype-phenotype study. <i>Clinical Genetics</i> , 2013, 84, 539-545.	1.0	85

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109	Transcriptome and genome sequencing uncovers functional variation in humans. <i>Nature</i> , 2013, 501, 506-511.	13.7	1,857
110	Mutations in the interleukin receptor <i>IL11RA</i> cause autosomal recessive Crouzon-like craniosynostosis. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2013, 1, 223-237.	0.6	70
111	A compound heterozygous mutation in DPAGT1 results in a congenital disorder of glycosylation with a relatively mild phenotype. <i>European Journal of Human Genetics</i> , 2013, 21, 844-849.	1.4	25
112	Mutations in BICD2, which Encodes a Golgin and Important Motor Adaptor, Cause Congenital Autosomal-Dominant Spinal Muscular Atrophy. <i>American Journal of Human Genetics</i> , 2013, 92, 946-954.	2.6	150
113	Point mutations as a source of de novo genetic disease. <i>Current Opinion in Genetics and Development</i> , 2013, 23, 257-263.	1.5	44
114	Challenges for implementing next-generation sequencing-based genome diagnostics: it's also the people, not just the machines. <i>Personalized Medicine</i> , 2013, 10, 473-484.	0.8	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. <i>Human Molecular Genetics</i> , 2013, 22, 656-667.	1.4	75
116	Exome sequencing identifies <i>DYNC2H1</i> mutations as a common cause of asphyxiating thoracic dystrophy (Jeune syndrome) without major polydactyly, renal or retinal involvement. <i>Journal of Medical Genetics</i> , 2013, 50, 309-323.	1.5	127
117	<i>GATAD2B</i> loss-of-function mutations cause a recognisable syndrome with intellectual disability and are associated with learning deficits and synaptic undergrowth in <i>Drosophila</i> . <i>Journal of Medical Genetics</i> , 2013, 50, 507-514.	1.5	63
118	<i>ZNF408</i> is mutated in familial exudative vitreoretinopathy and is crucial for the development of zebrafish retinal vasculature. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 9856-9861.	3.3	144
119	Exome sequencing identifies putative drivers of progression of transient myeloproliferative disorder to AMKL in infants with Down syndrome. <i>Blood</i> , 2013, 122, 554-561.	0.6	72
120	Novel PI3K <sup>Î³</sup> Mutation in a 44-Year-Old Man with Chronic Infections and Chronic Pelvic Pain. <i>PLoS ONE</i> , 2013, 8, e68118.	1.1	2
121	Genome and exome sequencing in the clinic: unbiased genomic approaches with a high diagnostic yield. <i>Pharmacogenomics</i> , 2012, 13, 511-514.	0.6	19
122	An integrated framework of personalized medicine: from individual genomes to participatory health care. <i>Croatian Medical Journal</i> , 2012, 53, 301-303.	0.2	25
123	A microduplication of the Rubinstein-Taybi region on 16p13.3 in a girl with a bilateral complete cleft lip and palate and severe mental retardation. <i>Clinical Dysmorphology</i> , 2012, 21, 204-207.	0.1	7
124	Gene identification in the congenital disorders of glycosylation type I by whole-exome sequencing. <i>Human Molecular Genetics</i> , 2012, 21, 4151-4161.	1.4	147
125	Nuclear Receptors <i>Nur77</i> and <i>Nurr1</i> Modulate Mesenchymal Stromal Cell Migration. <i>Stem Cells and Development</i> , 2012, 21, 228-238.	1.1	56
126	Mutations in <i>DYNC1H1</i> cause severe intellectual disability with neuronal migration defects. <i>Journal of Medical Genetics</i> , 2012, 49, 179-183.	1.5	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. <i>Nature Genetics</i> , 2012, 44, 797-802.	9.4	175
128	Mutations in ISPD cause Walker-Warburg syndrome and defective glycosylation of Î±-dystroglycan. <i>Nature Genetics</i> , 2012, 44, 581-585.	9.4	191
129	Common variants at 12q14 and 12q24 are associated with hippocampal volume. <i>Nature Genetics</i> , 2012, 44, 545-551.	9.4	212
130	Diagnostic Exome Sequencing in Persons with Severe Intellectual Disability. <i>New England Journal of Medicine</i> , 2012, 367, 1921-1929.	13.9	1,367
131	Mutations in DDHD2, Encoding an Intracellular Phospholipase A1, Cause a Recessive Form of Complex Hereditary Spastic Paraplegia. <i>American Journal of Human Genetics</i> , 2012, 91, 1073-1081.	2.6	159
132	Next-Generation genetic testing for retinitis pigmentosa. <i>Human Mutation</i> , 2012, 33, 963-972.	1.1	258
133	Targeted Next Generation Sequencing Reveals a Novel Intragenic Deletion of the TPO Gene in a Family with Intellectual Disability. <i>Archives of Medical Research</i> , 2012, 43, 312-316.	1.5	8
134	Mutations in the chromatin modifier gene KANSL1 cause the 17q21.31 microdeletion syndrome. <i>Nature Genetics</i> , 2012, 44, 639-641.	9.4	194
135	De novo mutations in the actin genes ACTB and ACTG1 cause Baraitser-Winter syndrome. <i>Nature Genetics</i> , 2012, 44, 440-444.	9.4	237
136	Identification of common variants associated with human hippocampal and intracranial volumes. <i>Nature Genetics</i> , 2012, 44, 552-561.	9.4	594
137	Disruption of an EHMT1-Associated Chromatin-Modification Module Causes Intellectual Disability. <i>American Journal of Human Genetics</i> , 2012, 91, 73-82.	2.6	214
138	Recurrent De Novo Mutations in PACS1 Cause Defective Cranial-Neural-Crest Migration and Define a Recognizable Intellectual-Disability Syndrome. <i>American Journal of Human Genetics</i> , 2012, 91, 1122-1127.	2.6	96
139	De novo diagnostics of patients with intellectual disability. <i>BMC Proceedings</i> , 2012, 6, .	1.8	0
140	Validation Study of Existing Gene Expression Signatures for Anti-TNF Treatment in Patients with Rheumatoid Arthritis. <i>PLoS ONE</i> , 2012, 7, e33199.	1.1	61
141	Trisomy for Synaptojanin1 in Down syndrome is functionally linked to the enlargement of early endosomes. <i>Human Molecular Genetics</i> , 2012, 21, 3156-3172.	1.4	92
142	Disease gene identification strategies for exome sequencing. <i>European Journal of Human Genetics</i> , 2012, 20, 490-497.	1.4	412
143	De novo mutations in human genetic disease. <i>Nature Reviews Genetics</i> , 2012, 13, 565-575.	7.7	715
144	Structural Genomic Variation in Intellectual Disability. <i>Methods in Molecular Biology</i> , 2012, 838, 77-95.	0.4	10

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145	Mutations in C8orf37, Encoding a Ciliary Protein, are Associated with Autosomal-Recessive Retinal Dystrophies with Early Macular Involvement. <i>American Journal of Human Genetics</i> , 2012, 90, 102-109.	2.6	82
146	Resolving the Breakpoints of the 17q21.31 Microdeletion Syndrome with Next-Generation Sequencing. <i>American Journal of Human Genetics</i> , 2012, 90, 599-613.	2.6	22
147	CantÃ© Syndrome Is Caused by Mutations in ABCC9. <i>American Journal of Human Genetics</i> , 2012, 90, 1094-1101.	2.6	141
148	Analysis of genes regulated by the transcription factor LUMAN identifies ApoA4 as a target gene in dendritic cells. <i>Molecular Immunology</i> , 2012, 50, 66-73.	1.0	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. <i>British Journal of Haematology</i> , 2012, 157, 197-200.	1.2	10
150	<i>STAT1</i> Mutations in Autosomal Dominant Chronic Mucocutaneous Candidiasis. <i>New England Journal of Medicine</i> , 2011, 365, 54-61.	13.9	614
151	De novo mutations in mental retardation. <i>Genome Biology</i> , 2011, 12, .	3.8	0
152	De novo nonsense mutations in ASXL1 cause Bohring-Opitz syndrome. <i>Nature Genetics</i> , 2011, 43, 729-731.	9.4	236
153	Unlocking Mendelian disease using exome sequencing. <i>Genome Biology</i> , 2011, 12, 228.	13.9	228
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