Joris A Veltman

List of Publications by Year in Descending Order

258 papers	25,023 citations	83 h-index	153 g-index
281 ext. papers	29,084 ext. citations	10.3 avg, IF	6.52 L-index

#	Paper	IF	Citations
258	A de novo paradigm for male infertility.. *Nature Communications*, **2022**, 13, 154	17.4	2
257	Lack of evidence for a role of PIWIL1 variants in human male infertility. *Cell*, **2021**, 184, 1941-1942	56.2	4
256	Variants in GCNA, X-linked germ-cell genome integrity gene, identified in men with primary spermatogenic failure. *Human Genetics*, **2021**, 140, 1169-1182	6.3	3
255	Exome sequencing reveals variants in known and novel candidate genes for severe sperm motility disorders. *Human Reproduction*, **2021**, 36, 2597-2611	5.7	6
254	Screening by single-molecule molecular inversion probes targeted sequencing panel of candidate genes of infertility in azoospermic infertile Jordanian males. *Human Fertility*, **2021**, 1-8	1.9	1
253	Disease gene discovery in male infertility: past, present and future. *Human Genetics*, **2021**, 140, 7-19	6.3	21
252	A global approach to addressing the policy, research and social challenges of male reproductive health. *Human Reproduction Open*, **2021**, 2021, hoab009	6.1	6
251	Deleterious variants in X-linked CFAP47 induce asthenoteratozoospermia and primary male infertility. *American Journal of Human Genetics*, **2021**, 108, 309-323	11	16
250	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	9.7	1
249	Variant , Defective piRNA Processing, and Azoospermia. *New England Journal of Medicine*, **2021**, 385, 707-719	59.2	8
248	A systematic review of the validated monogenic causes of human male infertility: 2020 update and a discussion of emerging gene-disease relationships. *Human Reproduction Update*, **2021**,	15.8	19
247	Programmed Cell Death 2-Like () Is Required for Mouse Embryonic Development. *G3: Genes, Genomes, Genetics*, **2020**, 10, 4449-4457	3.2	1
246	Mutations in the V-ATPase Assembly Factor VMA21 Cause a Congenital Disorder of Glycosylation With Autophagic Liver Disease. *Hepatology*, **2020**, 72, 1968-1986	11.2	20
245	Exome sequencing reveals novel causes as well as new candidate genes for human globozoospermia. *Human Reproduction*, **2020**, 35, 240-252	5.7	14
244	Aberrant Expressions and Variant Screening of in Indonesian Hirschsprung Patients. *Frontiers in Pediatrics*, **2020**, 8, 60	3.4	5
243	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	11	19
242	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	5.3	11

241	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,	2.8	11
240	Exome sequencing in routine diagnostics: a generic test for 254 patients with primary immunodeficiencies. *Genome Medicine*, **2019**, 11, 38	14.4	27
239	A systems genomics approach identifies as a susceptibility factor in recurrent vulvovaginal candidiasis. *Science Translational Medicine*, **2019**, 11,	17.5	25
238	MetaDome: Pathogenicity analysis of genetic variants through aggregation of homologous human protein domains. *Human Mutation*, **2019**, 40, 1030-1038	4.7	58
237	A systematic review and standardized clinical validity assessment of male infertility genes. *Human Reproduction*, **2019**, 34, 932-941	5.7	78
236	De Novo Mutations Reflect Development and Aging of the Human Germline. *Trends in Genetics*, **2019**, 35, 828-839	8.5	34
235	Front Cover, Volume 40, Issue 8. *Human Mutation*, **2019**, 40, i-i	4.7	18
234	The role of de novo mutations in adult-onset neurodegenerative disorders. *Acta Neuropathologica*, **2019**, 137, 183-207	14.3	29
233	Germline de novo mutation clusters arise during oocyte aging in genomic regions with high double-strand-break incidence. *Nature Genetics*, **2018**, 50, 487-492	36.3	35
232	Somatic variants in autosomal dominant genes are a rare cause of sporadic Alzheimer's disease. *Alzheimer's and Dementia*, **2018**, 14, 1632-1639	1.2	32
231	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	3.5	20
230	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. *European Journal of Human Genetics*, **2018**, 26, 54-63	5.3	23
229	Pathogenic variants in glutamyl-tRNA amidotransferase subunits cause a lethal mitochondrial cardiomyopathy disorder. *Nature Communications*, **2018**, 9, 4065	17.4	24
228	Upstream SLC2A1 translation initiation causes GLUT1 deficiency syndrome. *European Journal of Human Genetics*, **2017**, 25, 771-774	5.3	11
227	Rare NOX3 Variants Confer Susceptibility to Agranulocytosis During Thyrostatic Treatment of Graves' Disease. *Clinical Pharmacology and Therapeutics*, **2017**, 102, 1017-1024	6.1	9
226	Next-generation DNA sequencing identifies novel gene variants and pathways involved in specific language impairment. *Scientific Reports*, **2017**, 7, 46105	4.9	49
225	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	11	36
224	A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology. *Genetics in Medicine*, **2017**, 19, 1055-1063	8.1	140

223	MST1R mutation as a genetic cause of Lady Windermere syndrome. *European Respiratory Journal*, **2017**, 49,	13.6	10
222	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	11.9	13
221	Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. *PLoS Genetics*, **2017**, 13, e1006683	6	17
220	Spatial Clustering of de Novo Missense Mutations Identifies Candidate Neurodevelopmental Disorder-Associated Genes. *American Journal of Human Genetics*, **2017**, 101, 478-484	11	50
219	Biallelic variants in WARS2 encoding mitochondrial tryptophanyl-tRNA synthase in six individuals with mitochondrial encephalopathy. *Human Mutation*, **2017**, 38, 1786-1795	4.7	15
218	Aggregation of population-based genetic variation over protein domain homologues and its potential use in genetic diagnostics. *Human Mutation*, **2017**, 38, 1454-1463	4.7	21
217	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	4.7	32
216	Ultra-sensitive Sequencing Identifies High Prevalence of Clonal Hematopoiesis-Associated Mutations throughout Adult Life. *American Journal of Human Genetics*, **2017**, 101, 50-64	11	122
215	Detection of clinically relevant copy-number variants by exome sequencing in a large cohort of genetic disorders. *Genetics in Medicine*, **2017**, 19, 667-675	8.1	98
214	Exome sequencing of Pakistani consanguineous families identifies 30 novel candidate genes for recessive intellectual disability. *Molecular Psychiatry*, **2017**, 22, 1604-1614	15.1	69
213	Missense variants in AIMP1 gene are implicated in autosomal recessive intellectual disability without neurodegeneration. *European Journal of Human Genetics*, **2016**, 24, 392-9	5.3	14
212	LRP5 variants may contribute to ADPKD. *European Journal of Human Genetics*, **2016**, 24, 237-42	5.3	19
211	Evaluating a counselling strategy for diagnostic WES in paediatric neurology: an exploration of parentsPinformation and communication needs. *Clinical Genetics*, **2016**, 89, 244-50	4	20
210	Chromosomal abnormalities in hepatic cysts point to novel polycystic liver disease genes. *European Journal of Human Genetics*, **2016**, 24, 1707-1714	5.3	9
209	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	11	44
208	ATP6AP1 deficiency causes an immunodeficiency with hepatopathy, cognitive impairment and abnormal protein glycosylation. *Nature Communications*, **2016**, 7, 11600	17.4	83
207	Sequence variation between 462 human individuals fine-tunes functional sites of RNA processing. *Scientific Reports*, **2016**, 6, 32406	4.9	21
206	Parent-of-origin-specific signatures of de novo mutations. *Nature Genetics*, **2016**, 48, 935-9	36.3	174

205	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, 1145-53	5.3	23
204	TRIO loss of function is associated with mild intellectual disability and affects dendritic branching and synapse function. *Human Molecular Genetics*, **2016**, 25, 892-902	5.6	56
203	Influence of paternal age on ongoing pregnancy rate at eight weeksPgestation in assisted reproduction. *Reproductive BioMedicine Online*, **2016**, 32, 96-103	4	11
202	CCDC115 Deficiency Causes a Disorder of Golgi Homeostasis with Abnormal Protein Glycosylation. *American Journal of Human Genetics*, **2016**, 98, 310-21	11	65
201	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, 322-30	11	53
200	Genetic studies in intellectual disability and related disorders. *Nature Reviews Genetics*, **2016**, 17, 9-18	30.1	410
199	Different Balance of Wnt Signaling in Adult and Fetal Bone Marrow-Derived Mesenchymal Stromal Cells. *Stem Cells and Development*, **2016**, 25, 934-47	4.4	7
198	De novo loss-of-function mutations in X-linked SMC1A cause severe ID and therapy-resistant epilepsy in females: expanding the phenotypic spectrum. *Clinical Genetics*, **2016**, 90, 413-419	4	20
197	New insights into the generation and role of de novo mutations in health and disease. *Genome Biology*, **2016**, 17, 241	18.3	215
196	Is the $1000 Genome as Near as We Think? A Cost Analysis of Next-Generation Sequencing. *Clinical Chemistry*, **2016**, 62, 1458-1464	5.5	93
195	Novel bioinformatic developments for exome sequencing. *Human Genetics*, **2016**, 135, 603-14	6.3	27
194	Immunologic defects in severe mucocutaneous HSV-2 infections: Response to IFN-□therapy. *Journal of Allergy and Clinical Immunology*, **2016**, 138, 895-898	11.5	4
193	Understanding the Psychosocial Effects of WES Test Results on Parents of Children with Rare Diseases. *Journal of Genetic Counseling*, **2016**, 25, 1207-1214	2.5	48
192	Meta-analysis of 2,104 trios provides support for 10 new genes for intellectual disability. *Nature Neuroscience*, **2016**, 19, 1194-6	25.5	258
191	Homozygous mutation of STXBP5L explains an autosomal recessive infantile-onset neurodegenerative disorder. *Human Molecular Genetics*, **2015**, 24, 2000-10	5.6	14
190	Exome sequencing and whole genome sequencing for the detection of copy number variation. *Expert Review of Molecular Diagnostics*, **2015**, 15, 1023-32	3.8	52
189	Exome sequencing in an admixed isolated population indicates NFXL1 variants confer a risk for specific language impairment. *PLoS Genetics*, **2015**, 11, e1004925	6	32
188	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	14.3	222

187	A recent bottleneck of Y chromosome diversity coincides with a global change in culture. *Genome Research*, **2015**, 25, 459-66	9.7	235
186	Standardized phenotyping enhances Mendelian disease gene identification. *Nature Genetics*, **2015**, 47, 1222-4	36.3	14
185	Cell-Free RNA Is a Reliable Fetoplacental Marker in Noninvasive Fetal Sex Determination. *Clinical Chemistry*, **2015**, 61, 1515-23	5.5	9
184	Advantages and Disadvantages of Different Implementation Strategies of Non-Invasive Prenatal Testing in Down Syndrome Screening Programmes. *Public Health Genomics*, **2015**, 18, 260-71	1.9	4
183	Patient experiences with gene panels based on exome sequencing in clinical diagnostics: high acceptance and low distress. *Clinical Genetics*, **2015**, 87, 319-26	4	19
182	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	6.3	62
181	Heterozygous germline mutations in A2ML1 are associated with a disorder clinically related to Noonan syndrome. *European Journal of Human Genetics*, **2015**, 23, 317-24	5.3	48
180	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	3.3	2
179	Comparison of Exome and Genome Sequencing Technologies for the Complete Capture of Protein-Coding Regions. *Human Mutation*, **2015**, 36, 815-22	4.7	107
178	From genes to genomes in the clinic. *Genome Medicine*, **2015**, 7, 78	14.4	11
177	Post-zygotic Point Mutations Are an Underrecognized Source of De Novo Genomic Variation. *American Journal of Human Genetics*, **2015**, 97, 67-74	11	158
176	A missense mutation underlies defective SOCS4 function in a family with autoimmunity. *Journal of Internal Medicine*, **2015**, 278, 203-10	10.8	6
175	The diagnostic pathway in complex paediatric neurology: a cost analysis. *European Journal of Paediatric Neurology*, **2015**, 19, 233-9	3.8	35
174	Next-generation sequencing-based genome diagnostics across clinical genetics centers: implementation choices and their effects. *European Journal of Human Genetics*, **2015**, 23, 1142-50	5.3	41
173	Exome sequencing identifies a de novo SCN2A mutation in a patient with intractable seizures, severe intellectual disability, optic atrophy, muscular hypotonia, and brain abnormalities. *Epilepsia*, **2014**, 55, e25-9	6.4	47
172	Diagnostic serum glycosylation profile in patients with intellectual disability as a result of MAN1B1 deficiency. *Brain*, **2014**, 137, 1030-8	11.2	35
171	Clinical exome sequencing in daily practice: 1,000 patients and beyond. *Genome Medicine*, **2014**, 6, 2	14.4	22
170	Platform comparison of detecting copy number variants with microarrays and whole-exome sequencing. *Genomics Data*, **2014**, 2, 144-146		10

169	Parental somatic mosaicism is underrecognized and influences recurrence risk of genomic disorders. *American Journal of Human Genetics*, **2014**, 95, 173-82	11	172
168	Multiple phenotypes in phosphoglucomutase 1 deficiency. *New England Journal of Medicine*, **2014**, 370, 533-42	59.2	197
167	Mutations affecting the SAND domain of DEAF1 cause intellectual disability with severe speech impairment and behavioral problems. *American Journal of Human Genetics*, **2014**, 94, 649-61	11	51
166	Genome sequencing identifies major causes of severe intellectual disability. *Nature*, **2014**, 511, 344-7	50.4	761
165	Exome sequencing identifies three novel candidate genes implicated in intellectual disability. *PLoS ONE*, **2014**, 9, e112687	3.7	16
164	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, 5343-8	11.5	61
163	Mobster: accurate detection of mobile element insertions in next generation sequencing data. *Genome Biology*, **2014**, 15, 488	18.3	62
162	The effect of enamel matrix derivative (Emdogain®) on gene expression profiles of human primary alveolar bone cells. *Journal of Tissue Engineering and Regenerative Medicine*, **2014**, 8, 463-72	4.4	16
161	Detection of clinically relevant copy number variants with whole-exome sequencing. *Human Mutation*, **2013**, 34, 1439-48	4.7	89
160	Pathogenic or not? Assessing the clinical relevance of copy number variants. *Clinical Genetics*, **2013**, 84, 415-21	4	39
159	Identification of pathogenic gene variants in small families with intellectually disabled siblings by exome sequencing. *Journal of Medical Genetics*, **2013**, 50, 802-11	5.8	70
158	A post-hoc comparison of the utility of sanger sequencing and exome sequencing for the diagnosis of heterogeneous diseases. *Human Mutation*, **2013**, 34, 1721-6	4.7	240
157	Homozygous and heterozygous disruptions of ANK3: at the crossroads of neurodevelopmental and psychiatric disorders. *Human Molecular Genetics*, **2013**, 22, 1960-70	5.6	108
156	Breast cancer size estimation with MRI in BRCA mutation carriers and other high risk patients. *European Journal of Radiology*, **2013**, 82, 1416-22	4.7	14
155	MLL2 mutation detection in 86 patients with Kabuki syndrome: a genotype-phenotype study. *Clinical Genetics*, **2013**, 84, 539-45	4	66
154	Transcriptome and genome sequencing uncovers functional variation in humans. *Nature*, **2013**, 501, 506-11	50.4	1323
153	Mutations in the interleukin receptor IL11RA cause autosomal recessive Crouzon-like craniosynostosis. *Molecular Genetics & Genomic Medicine*, **2013**, 1, 223-37	2.3	56
152	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, 844-9	5.3	18

151	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, 946-54	11	122
150	Point mutations as a source of de novo genetic disease. *Current Opinion in Genetics and Development*, **2013**, 23, 257-63	4.9	40
149	Challenges for implementing next-generation sequencing-based genome diagnostics: it's also the people, not just the machines. *Personalized Medicine*, **2013**, 10, 473-484	2.2	9
148	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, 656-67	5.6	66
147	Exome sequencing identifies DYNC2H1 mutations as a common cause of asphyxiating thoracic dystrophy (Jeune syndrome) without major polydactyly, renal or retinal involvement. *Journal of Medical Genetics*, **2013**, 50, 309-23	5.8	103
146	GATAD2B loss-of-function mutations cause a recognisable syndrome with intellectual disability and are associated with learning deficits and synaptic undergrowth in Drosophila. *Journal of Medical Genetics*, **2013**, 50, 507-14	5.8	51
145	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, 9856-61	11.5	112
144	Exome sequencing identifies putative drivers of progression of transient myeloproliferative disorder to AMKL in infants with Down syndrome. *Blood*, **2013**, 122, 554-61	2.2	60
143	Novel PI3K□ mutation in a 44-year-old man with chronic infections and chronic pelvic pain. *PLoS ONE*, **2013**, 8, e68118	3.7	2
142	Analysis of genes regulated by the transcription factor LUMAN identifies ApoA4 as a target gene in dendritic cells. *Molecular Immunology*, **2012**, 50, 66-73	4.3	17
141	Amplified segment in the 'Down syndrome critical region' on HSA21 shared between Down syndrome and euploid AML-M0 excludes RUNX1, ERG and ETS2. *British Journal of Haematology*, **2012**, 157, 197-200	4.5	9
140	Diagnostic exome sequencing in persons with severe intellectual disability. *New England Journal of Medicine*, **2012**, 367, 1921-9	59.2	1102
139	Mutations in DDHD2, encoding an intracellular phospholipase A(1), cause a recessive form of complex hereditary spastic paraplegia. *American Journal of Human Genetics*, **2012**, 91, 1073-81	11	128
138	Next-generation genetic testing for retinitis pigmentosa. *Human Mutation*, **2012**, 33, 963-72	4.7	232
137	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, 312-6	6.6	7
136	Mutations in the chromatin modifier gene KANSL1 cause the 17q21.31 microdeletion syndrome. *Nature Genetics*, **2012**, 44, 639-41	36.3	149
135	De novo mutations in the actin genes ACTB and ACTG1 cause Baraitser-Winter syndrome. *Nature Genetics*, **2012**, 44, 440-4, S1-2	36.3	181
134	Identification of common variants associated with human hippocampal and intracranial volumes. *Nature Genetics*, **2012**, 44, 552-61	36.3	498

133	Disruption of an EHMT1-associated chromatin-modification module causes intellectual disability. *American Journal of Human Genetics*, **2012**, 91, 73-82	11	165
132	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, 1122-7	11	60
131	De novo diagnostics of patients with intellectual disability. *BMC Proceedings*, **2012**, 6,	2.3	78
130	Validation study of existing gene expression signatures for anti-TNF treatment in patients with rheumatoid arthritis. *PLoS ONE*, **2012**, 7, e33199	3.7	45
129	Impact of Genomewide Structural Variation on Gene Discovery **2012**, 443-470		
128	Trisomy for synaptojanin1 in Down syndrome is functionally linked to the enlargement of early endosomes. *Human Molecular Genetics*, **2012**, 21, 3156-72	5.6	76
127	Disease gene identification strategies for exome sequencing. *European Journal of Human Genetics*, **2012**, 20, 490-7	5.3	344
126	De novo mutations in human genetic disease. *Nature Reviews Genetics*, **2012**, 13, 565-75	30.1	551
125	Structural genomic variation in intellectual disability. *Methods in Molecular Biology*, **2012**, 838, 77-95	1.4	8
124	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, 102-9	11	68
123	Resolving the breakpoints of the 17q21.31 microdeletion syndrome with next-generation sequencing. *American Journal of Human Genetics*, **2012**, 90, 599-613	11	18
122	Cant□syndrome is caused by mutations in ABCC9. *American Journal of Human Genetics*, **2012**, 90, 1094-101	11	112
121	An integrated framework of personalized medicine: from individual genomes to participatory health care. *Croatian Medical Journal*, **2012**, 53, 301-3	1.6	21
120	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. *Clinical Dysmorphology*, **2012**, 21, 204-207	0.9	7
119	Gene identification in the congenital disorders of glycosylation type I by whole-exome sequencing. *Human Molecular Genetics*, **2012**, 21, 4151-61	5.6	126
118	Nuclear receptors Nur77 and Nurr1 modulate mesenchymal stromal cell migration. *Stem Cells and Development*, **2012**, 21, 228-38	4.4	48
117	Mutations in DYNC1H1 cause severe intellectual disability with neuronal migration defects. *Journal of Medical Genetics*, **2012**, 49, 179-83	5.8	131
116	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	36.3	147

115	Mutations in ISPD cause Walker-Warburg syndrome and defective glycosylation of □dystroglycan. *Nature Genetics*, **2012**, 44, 581-5	36.3	168
114	Common variants at 12q14 and 12q24 are associated with hippocampal volume. *Nature Genetics*, **2012**, 44, 545-51	36.3	175
113	STAT1 mutations in autosomal dominant chronic mucocutaneous candidiasis. *New England Journal of Medicine*, **2011**, 365, 54-61	59.2	505
112	De novo nonsense mutations in ASXL1 cause Bohring-Opitz syndrome. *Nature Genetics*, **2011**, 43, 729-31	36.3	198
111	Unlocking Mendelian disease using exome sequencing. *Genome Biology*, **2011**, 12, 228	18.3	189
110	Heterozygous mutations of FREM1 are associated with an increased risk of isolated metopic craniosynostosis in humans and mice. *PLoS Genetics*, **2011**, 7, e1002278	6	67
109	STAT1 hyperphosphorylation and defective IL12R/IL23R signaling underlie defective immunity in autosomal dominant chronic mucocutaneous candidiasis. *PLoS ONE*, **2011**, 6, e29248	3.7	93
108	Intragenic deletion in DYRK1A leads to mental retardation and primary microcephaly. *Clinical Genetics*, **2011**, 79, 296-9	4	74
107	Common variants in DGKK are strongly associated with risk of hypospadias. *Nature Genetics*, **2011**, 43, 48-50	36.3	86
106	Homozygosity mapping in outbred families with mental retardation. *European Journal of Human Genetics*, **2011**, 19, 597-601	5.3	23
105	Exome sequencing identifies truncating mutations in human SERPINF1 in autosomal-recessive osteogenesis imperfecta. *American Journal of Human Genetics*, **2011**, 88, 362-71	11	270
104	Chondrodysplasia and abnormal joint development associated with mutations in IMPAD1, encoding the Golgi-resident nucleotide phosphatase, gPAPP. *American Journal of Human Genetics*, **2011**, 88, 608-15	11	73
103	Ciliopathies with skeletal anomalies and renal insufficiency due to mutations in the IFT-A gene WDR19. *American Journal of Human Genetics*, **2011**, 89, 634-43	11	180
102	Disruption of teashirt zinc finger homeobox 1 is associated with congenital aural atresia in humans. *American Journal of Human Genetics*, **2011**, 89, 813-9	11	32
101	Pyrosequencing of 16S rRNA gene amplicons to study the microbiota in the gastrointestinal tract of carp (Cyprinus carpio L.). *AMB Express*, **2011**, 1, 41	4.1	136
100	Comprehensive genetic analysis of OEIS complex reveals no evidence for a recurrent microdeletion or duplication. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 38-49	2.5	13
99	A novel Xp22.11 deletion causing a syndrome of craniosynostosis and periventricular nodular heterotopia. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 3144-7	2.5	5
98	Whole-exome sequencing detects somatic mutations of IDH1 in metaphyseal chondromatosis with D-2-hydroxyglutaric aciduria (MC-HGA). *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2609-16	2.5	40

97	Association of the Alzheimer's gene SORL1 with hippocampal volume in young, healthy adults. *American Journal of Psychiatry*, **2011**, 168, 1083-9	11.9	52
96	De novo copy number variants associated with intellectual disability have a paternal origin and age bias. *Journal of Medical Genetics*, **2011**, 48, 776-8	5.8	84
95	Diagnostic Genome Profiling in Mental Retardation. *Advances in Neurobiology*, **2011**, 177-194	2.1	
94	The CASPR2 cell adhesion molecule functions as a tumor suppressor gene in glioma. *Oncogene*, **2010**, 29, 6138-48	9.2	22
93	Trisomic dose of several chromosome 21 genes perturbs haematopoietic stem and progenitor cell differentiation in Down's syndrome. *Oncogene*, **2010**, 29, 6102-14	9.2	38
92	The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype. *European Journal of Human Genetics*, **2010**, 18, 163-70	5.3	65
91	De novo mutations of SETBP1 cause Schinzel-Giedion syndrome. *Nature Genetics*, **2010**, 42, 483-5	36.3	362
90	A de novo paradigm for mental retardation. *Nature Genetics*, **2010**, 42, 1109-12	36.3	627
89	Homozygosity mapping in patients with cone-rod dystrophy: novel mutations and clinical characterizations **2010**, 51, 5943-51		86
88	Recurrent inversion events at 17q21.31 microdeletion locus are linked to the MAPT H2 haplotype. *Cytogenetic and Genome Research*, **2010**, 129, 275-9	1.9	22
87	Accurate distinction of pathogenic from benign CNVs in mental retardation. *PLoS Computational Biology*, **2010**, 6, e1000752	5	42
86	Genome-wide profiling of p63 DNA-binding sites identifies an element that regulates gene expression during limb development in the 7q21 SHFM1 locus. *PLoS Genetics*, **2010**, 6, e1001065	6	142
85	Periventricular heterotopia in common microdeletion syndromes. *Molecular Syndromology*, **2010**, 1, 35-41	1.5	27
84	Genetic variation in CACNA1C, a gene associated with bipolar disorder, influences brainstem rather than gray matter volume in healthy individuals. *Biological Psychiatry*, **2010**, 68, 586-8	7.9	84
83	Functional differences between mesenchymal stem cell populations are reflected by their transcriptome. *Stem Cells and Development*, **2010**, 19, 481-90	4.4	109
82	Genomic microarrays in mental retardation: from copy number variation to gene, from research to diagnosis. *Journal of Medical Genetics*, **2010**, 47, 289-97	5.8	121
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6	Next-generation sequencing identifies novel gene variants and pathways involved in specific language impairment	1
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4	Identifying long indels in exome sequencing data of patients with intellectual disability	1
3	MetaDome: Pathogenicity analysis of genetic variants through aggregation of homologous human protein domains	1
2	Biallelic mutations in M1AP are a frequent cause of meiotic arrest leading to male infertility	1
1	Meta-analysis of 2,104 trios provides support for 10 novel candidate genes for intellectual disability	1