# Joris A Veltman

## List of Publications by Citations



| | | | |
|---|---|---|---|
| 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 | Transcriptome and genome sequencing uncovers functional variation in humans. *Nature*, **2013**, 501, 506-11 | 50.4 | 1323 |
| 257 | Diagnostic exome sequencing in persons with severe intellectual disability. *New England Journal of Medicine*, **2012**, 367, 1921-9 | 59.2 | 1102 |
| 256 | Mutations in a new member of the chromodomain gene family cause CHARGE syndrome. *Nature Genetics*, **2004**, 36, 955-7 | 36.3 | 923 |
| 255 | Genome sequencing identifies major causes of severe intellectual disability. *Nature*, **2014**, 511, 344-7 | 50.4 | 761 |
| 254 | A de novo paradigm for mental retardation. *Nature Genetics*, **2010**, 42, 1109-12 | 36.3 | 627 |
| 253 | Recurrent rearrangements of chromosome 1q21.1 and variable pediatric phenotypes. *New England Journal of Medicine*, **2008**, 359, 1685-99 | 59.2 | 587 |
| 252 | De novo mutations in human genetic disease. *Nature Reviews Genetics*, **2012**, 13, 565-75 | 30.1 | 551 |
| 251 | STAT1 mutations in autosomal dominant chronic mucocutaneous candidiasis. *New England Journal of Medicine*, **2011**, 365, 54-61 | 59.2 | 505 |
| 250 | Identification of common variants associated with human hippocampal and intracranial volumes. *Nature Genetics*, **2012**, 44, 552-61 | 36.3 | 498 |
| 249 | Diagnostic genome profiling in mental retardation. *American Journal of Human Genetics*, **2005**, 77, 606-16 | 11 | 467 |
| 248 | Genetic studies in intellectual disability and related disorders. *Nature Reviews Genetics*, **2016**, 17, 9-18 | 30.1 | 410 |
| 247 | Array-based comparative genomic hybridization for the genomewide detection of submicroscopic chromosomal abnormalities. *American Journal of Human Genetics*, **2003**, 73, 1261-70 | 11 | 384 |
| 246 | Disruption of the neurexin 1 gene is associated with schizophrenia. *Human Molecular Genetics*, **2009**, 18, 988-96 | 5.6 | 376 |
| 245 | De novo mutations of SETBP1 cause Schinzel-Giedion syndrome. *Nature Genetics*, **2010**, 42, 483-5 | 36.3 | 362 |
| 244 | A new chromosome 17q21.31 microdeletion syndrome associated with a common inversion polymorphism. *Nature Genetics*, **2006**, 38, 999-1001 | 36.3 | 355 |
| 243 | Disease gene identification strategies for exome sequencing. *European Journal of Human Genetics*, **2012**, 20, 490-7 | 5.3 | 344 |
| 242 | CHARGE syndrome: the phenotypic spectrum of mutations in the CHD7 gene. *Journal of Medical Genetics*, **2006**, 43, 306-14 | 5.8 | 324 |

| | | | |
|---|---|---|---|
| 241 | CNTNAP2 gene dosage variation is associated with schizophrenia and epilepsy. *Molecular Psychiatry* , **2008**, 13, 261-6 | 15.1 | 272 |
| 240 | Exome sequencing identifies truncating mutations in human SERPINF1 in autosomal-recessive osteogenesis imperfecta. *American Journal of Human Genetics*, **2011**, 88, 362-71 | 11 | 270 |
| 239 | Meta-analysis of 2,104 trios provides support for 10 new genes for intellectual disability. *Nature Neuroscience*, **2016**, 19, 1194-6 | 25.5 | 258 |
| 238 | 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 |
| 237 | A recent bottleneck of Y chromosome diversity coincides with a global change in culture. *Genome Research*, **2015**, 25, 459-66 | 9.7 | 235 |
| 236 | Exome sequencing identifies WDR35 variants involved in Sensenbrenner syndrome. *American Journal of Human Genetics*, **2010**, 87, 418-23 | 11 | 233 |
| 235 | Next-generation genetic testing for retinitis pigmentosa. *Human Mutation*, **2012**, 33, 963-72 | 4.7 | 232 |
| 234 | 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 |
| 233 | Recurrent CNVs disrupt three candidate genes in schizophrenia patients. *American Journal of Human Genetics*, **2008**, 83, 504-10 | 11 | 220 |
| 232 | New insights into the generation and role of de novo mutations in health and disease. *Genome Biology*, **2016**, 17, 241 | 18.3 | 215 |
| 231 | De novo nonsense mutations in ASXL1 cause Bohring-Opitz syndrome. *Nature Genetics*, **2011**, 43, 729-31 | 36.3 | 198 |
| 230 | Multiple phenotypes in phosphoglucomutase 1 deficiency. *New England Journal of Medicine*, **2014**, 370, 533-42 | 59.2 | 197 |
| 229 | Unlocking Mendelian disease using exome sequencing. *Genome Biology*, **2011**, 12, 228 | 18.3 | 189 |
| 228 | Identification of tumor-specific molecular signatures in intracranial ependymoma and association with clinical characteristics. *Journal of Clinical Oncology*, **2006**, 24, 5223-33 | 2.2 | 187 |
| 227 | 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 |
| 226 | High-throughput analysis of subtelomeric chromosome rearrangements by use of array-based comparative genomic hybridization. *American Journal of Human Genetics*, **2002**, 70, 1269-76 | 11 | 181 |
| 225 | 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 |
| 224 | 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, 240-7 | 11 | 178 |

| | | | |
|---|---|---|---|
| 223 | Array-based comparative genomic hybridization for genome-wide screening of DNA copy number in bladder tumors. *Cancer Research*, **2003**, 63, 2872-80 | 10.1 | 178 |
| 222 | Common variants at 12q14 and 12q24 are associated with hippocampal volume. *Nature Genetics*, **2012**, 44, 545-51 | 36.3 | 175 |
| 221 | Parent-of-origin-specific signatures of de novo mutations. *Nature Genetics*, **2016**, 48, 935-9 | 36.3 | 174 |
| 220 | Parental somatic mosaicism is underrecognized and influences recurrence risk of genomic disorders. *American Journal of Human Genetics*, **2014**, 95, 173-82 | 11 | 172 |
| 219 | 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 | 10.1 | 169 |
| 218 | Mutations in ISPD cause Walker-Warburg syndrome and defective glycosylation of □dystroglycan. *Nature Genetics*, **2012**, 44, 581-5 | 36.3 | 168 |
| 217 | Disruption of an EHMT1-associated chromatin-modification module causes intellectual disability. *American Journal of Human Genetics*, **2012**, 91, 73-82 | 11 | 165 |
| 216 | Characterization of a recurrent 15q24 microdeletion syndrome. *Human Molecular Genetics*, **2007**, 16, 567-72 | 5.6 | 159 |
| 215 | Post-zygotic Point Mutations Are an Underrecognized Source of De Novo Genomic Variation. *American Journal of Human Genetics*, **2015**, 97, 67-74 | 11 | 158 |
| 214 | Clinical and molecular delineation of the 17q21.31 microdeletion syndrome. *Journal of Medical Genetics*, **2008**, 45, 710-20 | 5.8 | 156 |
| 213 | OFD1 is mutated in X-linked Joubert syndrome and interacts with LCA5-encoded lebercilin. *American Journal of Human Genetics*, **2009**, 85, 465-81 | 11 | 153 |
| 212 | Mutations in the chromatin modifier gene KANSL1 cause the 17q21.31 microdeletion syndrome. *Nature Genetics*, **2012**, 44, 639-41 | 36.3 | 149 |
| 211 | Mutations in LCA5, encoding the ciliary protein lebercilin, cause Leber congenital amaurosis. *Nature Genetics*, **2007**, 39, 889-95 | 36.3 | 148 |
| 210 | 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 |
| 209 | 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 |
| 208 | A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology. *Genetics in Medicine*, **2017**, 19, 1055-1063 | 8.1 | 140 |
| 207 | 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 |
| 206 | Mutations in DYNC1H1 cause severe intellectual disability with neuronal migration defects. *Journal of Medical Genetics*, **2012**, 49, 179-83 | 5.8 | 131 |

| | | | |
|---|---|---|---|
| 205 | 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 |
| 204 | Rare pathogenic microdeletions and tandem duplications are microhomology-mediated and stimulated by local genomic architecture. *Human Molecular Genetics*, **2009**, 18, 3579-93 | 5.6 | 126 |
| 203 | Genomic microarrays in mental retardation: a practical workflow for diagnostic applications. *Human Mutation*, **2009**, 30, 283-92 | 4.7 | 126 |
| 202 | Gene identification in the congenital disorders of glycosylation type I by whole-exome sequencing. *Human Molecular Genetics*, **2012**, 21, 4151-61 | 5.6 | 126 |
| 201 | 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 |
| 200 | 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 |
| 199 | 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 |
| 198 | Identification of disease genes by whole genome CGH arrays. *Human Molecular Genetics*, **2005**, 14 Spec No. 2, R215-23 | 5.6 | 121 |
| 197 | Cant syndrome is caused by mutations in ABCC9. *American Journal of Human Genetics*, **2012**, 90, 1094-101 | 11 | 112 |
| 196 | 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 |
| 195 | Array-based comparative genomic hybridization for the differential diagnosis of renal cell cancer. *Cancer Research*, **2002**, 62, 957-60 | 10.1 | 112 |
| 194 | Functional differences between mesenchymal stem cell populations are reflected by their transcriptome. *Stem Cells and Development*, **2010**, 19, 481-90 | 4.4 | 109 |
| 193 | Homozygous and heterozygous disruptions of ANK3: at the crossroads of neurodevelopmental and psychiatric disorders. *Human Molecular Genetics*, **2013**, 22, 1960-70 | 5.6 | 108 |
| 192 | Comparison of Exome and Genome Sequencing Technologies for the Complete Capture of Protein-Coding Regions. *Human Mutation*, **2015**, 36, 815-22 | 4.7 | 107 |
| 191 | Role of gain of 12p in germ cell tumour development. *Apmis*, **2003**, 111, 161-71; discussion 172-3 | 3.4 | 106 |
| 190 | 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 |
| 189 | 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 |
| 188 | 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, 813-9 | 11 | 98 |

| | | | |
|---|---|---|---|
| 187 | STAT1 hyperphosphorylation and defective IL12R/IL23R signaling underlie defective immunity in autosomal dominant chronic mucocutaneous candidiasis. *PLoS ONE*, **2011**, 6, e29248 | 3.7 | 93 |
| 186 | Genotype-phenotype mapping of chromosome 18q deletions by high-resolution array CGH: an update of the phenotypic map. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1858-67 | 2.5 | 93 |
| 185 | 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 |
| 184 | Definition of a critical region on chromosome 18 for congenital aural atresia by arrayCGH. *American Journal of Human Genetics*, **2003**, 72, 1578-84 | 11 | 92 |
| 183 | Detection of clinically relevant copy number variants with whole-exome sequencing. *Human Mutation*, **2013**, 34, 1439-48 | 4.7 | 89 |
| 182 | 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 | 4.5 | 87 |
| 181 | Common variants in DGKK are strongly associated with risk of hypospadias. *Nature Genetics*, **2011**, 43, 48-50 | 36.3 | 86 |
| 180 | Homozygosity mapping in patients with cone-rod dystrophy: novel mutations and clinical characterizations **2010**, 51, 5943-51 | | 86 |
| 179 | Identification of novel mutations in patients with Leber congenital amaurosis and juvenile RP by genome-wide homozygosity mapping with SNP microarrays. *Investigative Ophthalmology and Visual Science*, **2007**, 48, 5690-8 | | 85 |
| 178 | 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 |
| 177 | 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 |
| 176 | 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, 1794-801 | 9.2 | 84 |
| 175 | ATP6AP1 deficiency causes an immunodeficiency with hepatopathy, cognitive impairment and abnormal protein glycosylation. *Nature Communications*, **2016**, 7, 11600 | 17.4 | 83 |
| 174 | Massively parallel sequencing of ataxia genes after array-based enrichment. *Human Mutation*, **2010**, 31, 494-9 | 4.7 | 79 |
| 173 | A systematic review and standardized clinical validity assessment of male infertility genes. *Human Reproduction*, **2019**, 34, 932-941 | 5.7 | 78 |
| 172 | De novo diagnostics of patients with intellectual disability. *BMC Proceedings*, **2012**, 6, | 2.3 | 78 |
| 171 | Chromosomal copy number changes in patients with non-syndromic X linked mental retardation detected by array CGH. *Journal of Medical Genetics*, **2006**, 43, 362-70 | 5.8 | 78 |
| 170 | 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 |

| | | | |
|---|---|---|---|
| 169 | Intragenic deletion in DYRK1A leads to mental retardation and primary microcephaly. *Clinical Genetics*, **2011**, 79, 296-9 | 4 | 74 |
| 168 | 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, 1430-8 | 2.5 | 74 |
| 167 | Genome-wide array-based comparative genomic hybridization reveals multiple amplification targets and novel homozygous deletions in pancreatic carcinoma cell lines. *Cancer Research*, **2004**, 64, 3052-9 | 10.1 | 74 |
| 166 | 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 |
| 165 | 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 |
| 164 | Exome sequencing of Pakistani consanguineous families identifies 30 novel candidate genes for recessive intellectual disability. *Molecular Psychiatry*, **2017**, 22, 1604-1614 | 15.1 | 69 |
| 163 | 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 |
| 162 | 12p-amplicon structure analysis in testicular germ cell tumors of adolescents and adults by array CGH. *Oncogene*, **2003**, 22, 7695-701 | 9.2 | 68 |
| 161 | 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 |
| 160 | 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, 254-61 | 11 | 67 |
| 159 | MLL2 mutation detection in 86 patients with Kabuki syndrome: a genotype-phenotype study. *Clinical Genetics*, **2013**, 84, 539-45 | 4 | 66 |
| 158 | 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 |
| 157 | CCDC115 Deficiency Causes a Disorder of Golgi Homeostasis with Abnormal Protein Glycosylation. *American Journal of Human Genetics*, **2016**, 98, 310-21 | 11 | 65 |
| 156 | The 2q23.1 microdeletion syndrome: clinical and behavioural phenotype. *European Journal of Human Genetics*, **2010**, 18, 163-70 | 5.3 | 65 |
| 155 | Reduced purifying selection prevails over positive selection in human copy number variant evolution. *Genome Research*, **2008**, 18, 1711-23 | 9.7 | 64 |
| 154 | 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 |
| 153 | Mobster: accurate detection of mobile element insertions in next generation sequencing data. *Genome Biology*, **2014**, 15, 488 | 18.3 | 62 |
| 152 | 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 |

| | | | |
|---|---|---|---|
| 151 | Molecular karyotyping of patients with unexplained mental retardation by SNP arrays: a multicenter study. *Human Mutation*, **2009**, 30, 1082-92 | 4.7 | 61 |
| 150 | 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 |
| 149 | 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 |
| 148 | MetaDome: Pathogenicity analysis of genetic variants through aggregation of homologous human protein domains. *Human Mutation*, **2019**, 40, 1030-1038 | 4.7 | 58 |
| 147 | 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 |
| 146 | Mutations in the interleukin receptor IL11RA cause autosomal recessive Crouzon-like craniosynostosis. *Molecular Genetics & Genomic Medicine*, **2013**, 1, 223-37 | 2.3 | 56 |
| 145 | Anomalies of the CD8+ T cell pool in haemochromatosis: HLA-A3-linked expansions of CD8+CD28- T cells. *Clinical and Experimental Immunology*, **1997**, 107, 548-54 | 6.2 | 56 |
| 144 | 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 |
| 143 | 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 |
| 142 | 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 |
| 141 | Homozygosity mapping reveals mutations of GRXCR1 as a cause of autosomal-recessive nonsyndromic hearing impairment. *American Journal of Human Genetics*, **2010**, 86, 138-47 | 11 | 52 |
| 140 | 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 |
| 139 | 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 |
| 138 | Spatial Clustering of de Novo Missense Mutations Identifies Candidate Neurodevelopmental Disorder-Associated Genes. *American Journal of Human Genetics*, **2017**, 101, 478-484 | 11 | 50 |
| 137 | Next-generation DNA sequencing identifies novel gene variants and pathways involved in specific language impairment. *Scientific Reports*, **2017**, 7, 46105 | 4.9 | 49 |
| 136 | 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 |
| 135 | Nuclear receptors Nur77 and Nurr1 modulate mesenchymal stromal cell migration. *Stem Cells and Development*, **2012**, 21, 228-38 | 4.4 | 48 |
| 134 | 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 |

| | | | |
|---|---|---|---|
| 133 | 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 |
| 132 | Loss of a small region around the PTEN locus is a major chromosome 10 alteration in prostate cancer xenografts and cell lines. *Genes Chromosomes and Cancer*, **2004**, 39, 171-84 | 5 | 47 |
| 131 | Chromosome instability as an indicator of malignant progression in laryngeal mucosa. *Journal of Clinical Oncology*, **2000**, 18, 1644-51 | 2.2 | 46 |
| 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 | Mutations in TPRN cause a progressive form of autosomal-recessive nonsyndromic hearing loss. *American Journal of Human Genetics*, **2010**, 86, 479-84 | 11 | 45 |
| 128 | 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 |
| 127 | Variation of CNV distribution in five different ethnic populations. *Cytogenetic and Genome Research* , **2007**, 118, 19-30 | 1.9 | 43 |
| 126 | Accurate distinction of pathogenic from benign CNVs in mental retardation. *PLoS Computational Biology*, **2010**, 6, e1000752 | 5 | 42 |
| 125 | High resolution profiling of X chromosomal aberrations by array comparative genomic hybridisation. *Journal of Medical Genetics*, **2004**, 41, 425-32 | 5.8 | 42 |
| 124 | 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 |
| 123 | Point mutations as a source of de novo genetic disease. *Current Opinion in Genetics and Development*, **2013**, 23, 257-63 | 4.9 | 40 |
| 122 | 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 |
| 121 | Human mitochondrial complex I deficiency: investigating transcriptional responses by microarray. *Neuropediatrics*, **2003**, 34, 14-22 | 1.6 | 40 |
| 120 | Pathogenic or not? Assessing the clinical relevance of copy number variants. *Clinical Genetics*, **2013**, 84, 415-21 | 4 | 39 |
| 119 | Forging links between human mental retardation-associated CNVs and mouse gene knockout models. *PLoS Genetics*, **2009**, 5, e1000531 | 6 | 39 |
| 118 | Molecular characterisation of patients with subtelomeric 22q abnormalities using chromosome specific array-based comparative genomic hybridisation. *European Journal of Human Genetics*, **2005**, 13, 1019-24 | 5.3 | 39 |
| 117 | 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 |
| 116 | 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, 367-76 | 5 | 38 |

| | | | |
|---|---|---|---|
| 115 | Diagnostic genome profiling: unbiased whole genome or targeted analysis?. *Journal of Molecular Diagnostics*, **2006**, 8, 534-7; discussion 537-9 | 5.1 | 37 |
| 114 | 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 |
| 113 | Ovotestes and XY sex reversal in a female with an interstitial 9q33.3-q34.1 deletion encompassing NR5A1 and LMX1B causing features of Genitopatellar syndrome. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1071-81 | 2.5 | 36 |
| 112 | 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 |
| 111 | Diagnostic serum glycosylation profile in patients with intellectual disability as a result of MAN1B1 deficiency. *Brain*, **2014**, 137, 1030-8 | 11.2 | 35 |
| 110 | The diagnostic pathway in complex paediatric neurology: a cost analysis. *European Journal of Paediatric Neurology*, **2015**, 19, 233-9 | 3.8 | 35 |
| 109 | De Novo Mutations Reflect Development and Aging of the Human Germline. *Trends in Genetics*, **2019**, 35, 828-839 | 8.5 | 34 |
| 108 | Chromosomal breakpoint mapping by arrayCGH using flow-sorted chromosomes. *BioTechniques*, **2003**, 35, 1066-70 | 2.5 | 34 |
| 107 | Exome sequencing in an admixed isolated population indicates NFXL1 variants confer a risk for specific language impairment. *PLoS Genetics*, **2015**, 11, e1004925 | 6 | 32 |
| 106 | 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 |
| 105 | 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 |
| 104 | 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 |
| 103 | The role of de novo mutations in adult-onset neurodegenerative disorders. *Acta Neuropathologica*, **2019**, 137, 183-207 | 14.3 | 29 |
| 102 | Exome sequencing in routine diagnostics: a generic test for 254 patients with primary immunodeficiencies. *Genome Medicine*, **2019**, 11, 38 | 14.4 | 27 |
| 101 | Periventricular heterotopia in common microdeletion syndromes. *Molecular Syndromology*, **2010**, 1, 35-41 | 1.5 | 27 |
| 100 | Genomic microarrays in clinical diagnosis. *Current Opinion in Pediatrics*, **2006**, 18, 598-603 | 3.2 | 27 |
| 99 | Novel bioinformatic developments for exome sequencing. *Human Genetics*, **2016**, 135, 603-14 | 6.3 | 27 |
| 98 | Novel candidate tumour suppressor gene loci on chromosomes 11q23-24 and 22q13 involved in human insulinoma tumourigenesis. *Journal of Pathology*, **2006**, 210, 450-8 | 9.4 | 26 |

| | | | |
|---|---|---|---|
| 97 | A systems genomics approach identifies as a susceptibility factor in recurrent vulvovaginal candidiasis. *Science Translational Medicine*, **2019**, 11, | 17.5 | 25 |
| 96 | 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 | 9.4 | 25 |
| 95 | Complex chromosome 17p rearrangements associated with low-copy repeats in two patients with congenital anomalies. *Human Genetics*, **2007**, 121, 697-709 | 6.3 | 24 |
| 94 | Pathogenic variants in glutamyl-tRNA amidotransferase subunits cause a lethal mitochondrial cardiomyopathy disorder. *Nature Communications*, **2018**, 9, 4065 | 17.4 | 24 |
| 93 | 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 |
| 92 | Homozygosity mapping in outbred families with mental retardation. *European Journal of Human Genetics*, **2011**, 19, 597-601 | 5.3 | 23 |
| 91 | 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 |
| 90 | Clinical exome sequencing in daily practice: 1,000 patients and beyond. *Genome Medicine*, **2014**, 6, 2 | 14.4 | 22 |
| 89 | The CASPR2 cell adhesion molecule functions as a tumor suppressor gene in glioma. *Oncogene*, **2010**, 29, 6138-48 | 9.2 | 22 |
| 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 | Molecular parameters associated with insulinoma progression: chromosomal instability versus p53 and CK19 status. *Cytogenetic and Genome Research*, **2006**, 115, 289-97 | 1.9 | 22 |
| 86 | Sequence variation between 462 human individuals fine-tunes functional sites of RNA processing. *Scientific Reports*, **2016**, 6, 32406 | 4.9 | 21 |
| 85 | 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 |
| 84 | An integrated framework of personalized medicine: from individual genomes to participatory health care. *Croatian Medical Journal*, **2012**, 53, 301-3 | 1.6 | 21 |
| 83 | A novel microdeletion, del(2)(q22.3q23.3) in a mentally retarded patient, detected by array-based comparative genomic hybridization. *Clinical Genetics*, **2004**, 65, 429-32 | 4 | 21 |
| 82 | Disease gene discovery in male infertility: past, present and future. *Human Genetics*, **2021**, 140, 7-19 | 6.3 | 21 |
| 81 | 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 |
| 80 | 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 |

| | | | |
|---|---|---|---|
| 79 | 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 |
| 78 | 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 |
| 77 | LRP5 variants may contribute to ADPKD. *European Journal of Human Genetics*, **2016**, 24, 237-42 | 5.3 | 19 |
| 76 | 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 |
| 75 | 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 |
| 74 | 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 |
| 73 | Front Cover, Volume 40, Issue 8. *Human Mutation*, **2019**, 40, i-i | 4.7 | 18 |
| 72 | 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 |
| 71 | Resolving the breakpoints of the 17q21.31 microdeletion syndrome with next-generation sequencing. *American Journal of Human Genetics*, **2012**, 90, 599-613 | 11 | 18 |
| 70 | High density gene expression microarrays and gene ontology analysis for identifying processes in implanted tissue engineering constructs. *Biomaterials*, **2010**, 31, 8299-312 | 15.6 | 18 |
| 69 | Detection of chromosomal aberrations in cytologic brush specimens from head and neck squamous cell carcinoma. *Cancer*, **1997**, 81, 309-14 | 6.4 | 18 |
| 68 | Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. *PLoS Genetics*, **2017**, 13, e1006683 | 6 | 17 |
| 67 | 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 |
| 66 | 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**, 140, 618-23 | 2.5 | 17 |
| 65 | 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, 193-9 | 7.5 | 17 |
| 64 | Exome sequencing identifies three novel candidate genes implicated in intellectual disability. *PLoS ONE*, **2014**, 9, e112687 | 3.7 | 16 |
| 63 | 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 |
| 62 | High-resolution genomic microarrays for X-linked mental retardation. *Genetics in Medicine*, **2007**, 9, 560-5 | 3.1 | 16 |

| | | | |
|---|---|---|---|
| 61 | Deleterious variants in X-linked CFAP47 induce asthenoteratozoospermia and primary male infertility. *American Journal of Human Genetics*, **2021**, 108, 309-323 | 11 | 16 |
| 60 | Biallelic variants in WARS2 encoding mitochondrial tryptophanyl-tRNA synthase in six individuals with mitochondrial encephalopathy. *Human Mutation*, **2017**, 38, 1786-1795 | 4.7 | 15 |
| 59 | Holoprosencephaly and preaxial polydactyly associated with a 1.24 Mb duplication encompassing FBXW11 at 5q35.1. *Journal of Human Genetics*, **2006**, 51, 721-726 | 4.3 | 15 |
| 58 | Chromosome 22q11 deletion and pachygyria characterized by array-based comparative genomic hybridization. *American Journal of Medical Genetics Part A*, **2004**, 131, 322-4 | | 15 |
| 57 | 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 |
| 56 | Homozygous mutation of STXBP5L explains an autosomal recessive infantile-onset neurodegenerative disorder. *Human Molecular Genetics*, **2015**, 24, 2000-10 | 5.6 | 14 |
| 55 | Standardized phenotyping enhances Mendelian disease gene identification. *Nature Genetics*, **2015**, 47, 1222-4 | 36.3 | 14 |
| 54 | Exome sequencing reveals novel causes as well as new candidate genes for human globozoospermia. *Human Reproduction*, **2020**, 35, 240-252 | 5.7 | 14 |
| 53 | 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 |
| 52 | 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 |
| 51 | 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 |
| 50 | Transchromosomic cell model of Down syndrome shows aberrant migration, adhesion and proteome response to extracellular matrix. *Proteome Science*, **2009**, 7, 31 | 2.6 | 12 |
| 49 | Upstream SLC2A1 translation initiation causes GLUT1 deficiency syndrome. *European Journal of Human Genetics*, **2017**, 25, 771-774 | 5.3 | 11 |
| 48 | 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 |
| 47 | 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 |
| 46 | Influence of paternal age on ongoing pregnancy rate at eight weeksPgestation in assisted reproduction. *Reproductive BioMedicine Online*, **2016**, 32, 96-103 | 4 | 11 |
| 45 | From genes to genomes in the clinic. *Genome Medicine*, **2015**, 7, 78 | 14.4 | 11 |
| 44 | Double-target fluorescence in situ hybridization distinguishes multiple genetically aberrant clones in head and neck squamous cell carcinoma. *Cytometry*, **1998**, 34, 113-20 | | 11 |

| | | | |
|---|---|---|---|
| 43 | MST1R mutation as a genetic cause of Lady Windermere syndrome. *European Respiratory Journal*, **2017**, 49, | 13.6 | 10 |
| 42 | Platform comparison of detecting copy number variants with microarrays and whole-exome sequencing. *Genomics Data*, **2014**, 2, 144-146 | | 10 |
| 41 | Rare NOX3 Variants Confer Susceptibility to Agranulocytosis During Thyrostatic Treatment of GravesPDisease. *Clinical Pharmacology and Therapeutics*, **2017**, 102, 1017-1024 | 6.1 | 9 |
| 40 | Cell-Free RNA Is a Reliable Fetoplacental Marker in Noninvasive Fetal Sex Determination. *Clinical Chemistry*, **2015**, 61, 1515-23 | 5.5 | 9 |
| 39 | Chromosomal abnormalities in hepatic cysts point to novel polycystic liver disease genes. *European Journal of Human Genetics*, **2016**, 24, 1707-1714 | 5.3 | 9 |
| 38 | Amplified segment in the PDown syndrome critical regionPon 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 |
| 37 | Challenges for implementing next-generation sequencing-based genome diagnostics: itP also the people, not just the machines. *Personalized Medicine*, **2013**, 10, 473-484 | 2.2 | 9 |
| 36 | Whole-genome array comparative genome hybridization: the preferred diagnostic choice in postnatal clinical cytogenetics. *Journal of Molecular Diagnostics*, **2007**, 9, 277 | 5.1 | 9 |
| 35 | Structural genomic variation in intellectual disability. *Methods in Molecular Biology*, **2012**, 838, 77-95 | 1.4 | 8 |
| 34 | Variant , Defective piRNA Processing, and Azoospermia. *New England Journal of Medicine*, **2021**, 385, 707-719 | 59.2 | 8 |
| 33 | 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 |
| 32 | 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 |
| 31 | A novel 2.3 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 | 0.9 | 7 |
| 30 | 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 |
| 29 | A missense mutation underlies defective SOCS4 function in a family with autoimmunity. *Journal of Internal Medicine*, **2015**, 278, 203-10 | 10.8 | 6 |
| 28 | Mapping of 5q35 chromosomal rearrangements within a genomically unstable region. *Journal of Medical Genetics*, **2008**, 45, 672-8 | 5.8 | 6 |
| 27 | Exome sequencing reveals variants in known and novel candidate genes for severe sperm motility disorders. *Human Reproduction*, **2021**, 36, 2597-2611 | 5.7 | 6 |
| 26 | A global approach to addressing the policy, research and social challenges of male reproductive health. *Human Reproduction Open*, **2021**, 2021, hoab009 | 6.1 | 6 |

| | | | |
|---|---|---|---|
| 25 | Aberrant Expressions and Variant Screening of in Indonesian Hirschsprung Patients. *Frontiers in Pediatrics*, **2020**, 8, 60 | 3.4 | 5 |
| 24 | 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 |
| 23 | Recurrent deletion of ZNF630 at Xp11.23 is not associated with mental retardation. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 638-45 | 2.5 | 5 |
| 22 | 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 |
| 21 | Genomic copy number analysis in mental retardation: finding the needles in the haystack. *European Journal of Human Genetics*, **2007**, 15, 1-2 | 5.3 | 4 |
| 20 | Lack of evidence for a role of PIWIL1 variants in human male infertility. *Cell*, **2021**, 184, 1941-1942 | 56.2 | 4 |
| 19 | 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 |
| 18 | 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 |
| 17 | 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 |
| 16 | A de novo paradigm for male infertility.. *Nature Communications*, **2022**, 13, 154 | 17.4 | 2 |
| 15 | Novel PI3K mutation in a 44-year-old man with chronic infections and chronic pelvic pain. *PLoS ONE*, **2013**, 8, e68118 | 3.7 | 2 |
| 14 | Programmed Cell Death 2-Like () Is Required for Mouse Embryonic Development. *G3: Genes, Genomes, Genetics*, **2020**, 10, 4449-4457 | 3.2 | 1 |
| 13 | 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 | 7.5 | 1 |
| 12 | A systematic review and standardized clinical validity assessment of male infertility genes | | 1 |
| 11 | Next-generation sequencing identifies novel gene variants and pathways involved in specific language impairment | | 1 |
| 10 | Stochasticity explains differences in the number of de novo mutations between families | | 1 |
| 9 | Identifying long indels in exome sequencing data of patients with intellectual disability | | 1 |
| 8 | MetaDome: Pathogenicity analysis of genetic variants through aggregation of homologous human protein domains | | 1 |

| | | | |
|---|---|---|---|
| 7 | Biallelic mutations in M1AP are a frequent cause of meiotic arrest leading to male infertility | | 1 |
| 6 | Meta-analysis of 2,104 trios provides support for 10 novel candidate genes for intellectual disability | | 1 |
| 5 | 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 |
| 4 | 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 |
| 3 | Impact of Genomewide Structural Variation on Gene Discovery **2012**, 443-470 | | |
| 2 | P49: Identification of´recurrent chromosomal aberrations in´different types of´human germ cell tumours using array CGH. *European Journal of Medical Genetics*, **2005**, 48, 511-512 | 2.6 | |
| 1 | Diagnostic Genome Profiling in Mental Retardation. *Advances in Neurobiology*, **2011**, 177-194 | 2.1 | |