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List of Publications by Year in Descending Order

70 papers	7,768 citations	34 h-index	88 g-index
92 ext. papers	10,369 ext. citations	11.7 avg, IF	5.21 L-index

#	Paper	IF	Citations
70	Schizophrenia Risk Mediated by microRNA Target Genes Overlapped by Genome-Wide Rare Copy Number Variation in 22q11.2 Deletion Syndrome.. *Frontiers in Genetics*, **2022**, 13, 812183	4.5	0
69	FAN1 exo- not endo-nuclease pausing on disease-associated slipped-DNA repeats: A mechanism of repeat instability. *Cell Reports*, **2021**, 37, 110078	10.6	0
68	Genomic imbalances in the placenta are associated with poor fetal growth. *Molecular Medicine*, **2021**, 27, 3	6.2	5
67	Genome sequencing broadens the range of contributing variants with clinical implications in schizophrenia. *Translational Psychiatry*, **2021**, 11, 84	8.6	5
66	Rare and low frequency genomic variants impacting neuronal functions modify the Dup7q11.23 phenotype. *Orphanet Journal of Rare Diseases*, **2021**, 16, 6	4.2	3
65	Genome sequencing identifies rare tandem repeat expansions and copy number variants in Lennox-Gastaut syndrome. *Brain Communications*, **2021**, 3, fcab207	4.5	2
64	Sleep phenotype of individuals with autism spectrum disorder bearing mutations in the PER2 circadian rhythm gene. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1120-1130	2.5	1
63	Segregating patterns of copy number variations in extended autism spectrum disorder (ASD) pedigrees. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2020**, 183, 268-276	3.5	3
62	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. *Cell*, **2020**, 180, 568-584.e23	56.2	578
61	ExpansionHunter Denovo: a computational method for locating known and novel repeat expansions in short-read sequencing data. *Genome Biology*, **2020**, 21, 102	18.3	29
60	Genes and Pathways Implicated in Tetralogy of Fallot Revealed by Ultra-Rare Variant Burden Analysis in 231 Genome Sequences. *Frontiers in Genetics*, **2020**, 11, 957	4.5	8
59	Genome Sequencing as a Diagnostic Test in Children With Unexplained Medical Complexity. *JAMA Network Open*, **2020**, 3, e2018109	10.4	13
58	Genetics of Epileptic Networks: from Focal to Generalized Genetic Epilepsies. *Current Neurology and Neuroscience Reports*, **2020**, 20, 46	6.6	6
57	Genome-wide detection of tandem DNA repeats that are expanded in autism. *Nature*, **2020**, 586, 80-86	50.4	58
56	Synaptic Dysfunction in Human Neurons With Autism-Associated Deletions in PTCHD1-AS. *Biological Psychiatry*, **2020**, 87, 139-149	7.9	32
55	A framework for an evidence-based gene list relevant to autism spectrum disorder. *Nature Reviews Genetics*, **2020**, 21, 367-376	30.1	30
54	Expanding the neurodevelopmental phenotypes of individuals with de novo variants. *Npj Genomic Medicine*, **2019**, 4, 9	6.2	10

53	Length of Uninterrupted CAG, Independent of Polyglutamine Size, Results in Increased Somatic Instability, Hastening Onset of Huntington Disease. *American Journal of Human Genetics*, **2019**, 104, 1116-1126	11	67
52	Glutaminase Deficiency Caused by Short Tandem Repeat Expansion in. *New England Journal of Medicine*, **2019**, 380, 1433-1441	59.2	31
51	Pathogenic WDFY3 variants cause neurodevelopmental disorders and opposing effects on brain size. *Brain*, **2019**, 142, 2617-2630	11.2	17
50	A large data resource of genomic copy number variation across neurodevelopmental disorders. *Npj Genomic Medicine*, **2019**, 4, 26	6.2	54
49	or human iPSC-derived neurons from individuals with autism develop hyperactive neuronal networks. *ELife*, **2019**, 8,	8.9	41
48	The Genetics of Autism Spectrum Disorders **2019**, 112-128		6
47	Predictive impact of rare genomic copy number variations in siblings of individuals with autism spectrum disorders. *Nature Communications*, **2019**, 10, 5519	17.4	13
46	Altered TAOK2 activity causes autism-related neurodevelopmental and cognitive abnormalities through RhoA signaling. *Molecular Psychiatry*, **2019**, 24, 1329-1350	15.1	70
45	The Personal Genome Project Canada: findings from whole genome sequences of the inaugural 56 participants. *Cmaj*, **2018**, 190, E126-E136	3.5	37
44	A Comprehensive Workflow for Read Depth-Based Identification of Copy-Number Variation from Whole-Genome Sequence Data. *American Journal of Human Genetics*, **2018**, 102, 142-155	11	97
43	A genome-wide linkage study of autism spectrum disorder and the broad autism phenotype in extended pedigrees. *Journal of Neurodevelopmental Disorders*, **2018**, 10, 20	4.6	10
42	Complete Disruption of Autism-Susceptibility Genes by Gene Editing Predominantly Reduces Functional Connectivity of Isogenic Human Neurons. *Stem Cell Reports*, **2018**, 11, 1211-1225	8	58
41	CNTN6 mutations are risk factors for abnormal auditory sensory perception in autism spectrum disorders. *Molecular Psychiatry*, **2017**, 22, 625-633	15.1	40
40	Genome and Transcriptome Assembly of the Canadian Beaver (). *G3: Genes, Genomes, Genetics*, **2017**, 7, 755-773	3.2	13
39	Whole genome sequencing resource identifies 18 new candidate genes for autism spectrum disorder. *Nature Neuroscience*, **2017**, 20, 602-611	25.5	427
38	Variable phenotype expression in a family segregating microdeletions of the and autism spectrum disorder susceptibility genes. *Npj Genomic Medicine*, **2017**, 2,	6.2	17
37	Mutations in in individuals with intellectual disability, autism spectrum disorder, and macrocephaly. *Molecular Autism*, **2017**, 8, 59	6.5	22
36	Germline and somatic mutations in with diverse neurodevelopmental phenotypes. *Neurology: Genetics*, **2017**, 3, e199	3.8	28

35	DIXDC1 Phosphorylation and Control of Dendritic Morphology Are Impaired by Rare Genetic Variants. *Cell Reports*, **2016**, 17, 1892-1904	10.6	21
34	Genome-wide characteristics of mutations in autism. *Npj Genomic Medicine*, **2016**, 1, 160271-1602710	6.2	126
33	Indexing Effects of Copy Number Variation on Genes Involved in Developmental Delay. *Scientific Reports*, **2016**, 6, 28663	4.9	26
32	Whole Genome Sequencing Expands Diagnostic Utility and Improves Clinical Management in Pediatric Medicine. *Npj Genomic Medicine*, **2016**, 1,	6.2	208
31	Genome-wide DNA methylation identifies trophoblast invasion-related genes: Claudin-4 and Fucosyltransferase IV control mobility via altering matrix metalloproteinase activity. *Molecular Human Reproduction*, **2015**, 21, 452-65	4.4	9
30	Compound heterozygous mutations in the noncoding RNU4ATAC cause Roifman Syndrome by disrupting minor intron splicing. *Nature Communications*, **2015**, 6, 8718	17.4	74
29	Molecular Diagnostic Yield of Chromosomal Microarray Analysis and Whole-Exome Sequencing in Children With Autism Spectrum Disorder. *JAMA - Journal of the American Medical Association*, **2015**, 314, 895-903	27.4	241
28	ISDN2014_0253: High resolution genomic analyses of a clinically defined autism spectrum disorder cohort. *International Journal of Developmental Neuroscience*, **2015**, 47, 76-76	2.7	1
27	Using extended pedigrees to identify novel autism spectrum disorder (ASD) candidate genes. *Human Genetics*, **2015**, 134, 191-201	6.3	16
26	RNA splicing. The human splicing code reveals new insights into the genetic determinants of disease. *Science*, **2015**, 347, 1254806	33.3	748
25	Whole-genome sequencing of quartet families with autism spectrum disorder. *Nature Medicine*, **2015**, 21, 185-91	50.5	353
24	Brain-expressed exons under purifying selection are enriched for de novo mutations in autism spectrum disorder. *Nature Genetics*, **2014**, 46, 742-7	36.3	121
23	The Database of Genomic Variants: a curated collection of structural variation in the human genome. *Nucleic Acids Research*, **2014**, 42, D986-92	20.1	764
22	Disruption of the ASTN2/TRIM32 locus at 9q33.1 is a risk factor in males for autism spectrum disorders, ADHD and other neurodevelopmental phenotypes. *Human Molecular Genetics*, **2014**, 23, 2752-68	5.6	104
21	Synaptic, transcriptional and chromatin genes disrupted in autism. *Nature*, **2014**, 515, 209-15	50.4	1581
20	Development of a high-resolution Y-chromosome microarray for improved male infertility diagnosis. *Fertility and Sterility*, **2014**, 101, 1079-1085.e3	4.8	24
19	Performance of high-throughput sequencing for the discovery of genetic variation across the complete size spectrum. *G3: Genes, Genomes, Genetics*, **2014**, 4, 63-5	3.2	26
18	Detection of clinically relevant genetic variants in autism spectrum disorder by whole-genome sequencing. *American Journal of Human Genetics*, **2013**, 93, 249-63	11	345

17	Hypoxia alters the epigenetic profile in cultured human placental trophoblasts. *Epigenetics*, **2013**, 8, 192-202	3.7	69
16	Widespread DNA hypomethylation at gene enhancer regions in placentas associated with early-onset pre-eclampsia. *Molecular Human Reproduction*, **2013**, 19, 697-708	4.4	150
15	Genome-Wide Mapping **2013**, 71-109		
14	The autism sequencing consortium: large-scale, high-throughput sequencing in autism spectrum disorders. *Neuron*, **2012**, 76, 1052-6	13.9	124
13	PP065. dNK and dNK-CM mediated alterations of DNA methylation in extravillous cytotrophoblasts (EVTS). *Pregnancy Hypertension*, **2012**, 2, 277	2.6	0
12	Patterns of placental development evaluated by X chromosome inactivation profiling provide a basis to evaluate the origin of epigenetic variation. *Human Reproduction*, **2012**, 27, 1745-53	5.7	34
11	The utility of quantitative methylation assays at imprinted genes for the diagnosis of fetal and placental disorders. *Clinical Genetics*, **2011**, 79, 169-75	4	30
10	Review: A high capacity of the human placenta for genetic and epigenetic variation: implications for assessing pregnancy outcome. *Placenta*, **2011**, 32 Suppl 2, S136-41	3.4	37
9	Genome-wide mapping of imprinted differentially methylated regions by DNA methylation profiling of human placentas from triploidies. *Epigenetics and Chromatin*, **2011**, 4, 10	5.8	62
8	Extensive epigenetic reprogramming in human somatic tissues between fetus and adult. *Epigenetics and Chromatin*, **2011**, 4, 7	5.8	51
7	Evidence for widespread changes in promoter methylation profile in human placenta in response to increasing gestational age and environmental/stochastic factors. *BMC Genomics*, **2011**, 12, 529	4.5	142
6	DNA methylation profiling of human placentas reveals promoter hypomethylation of multiple genes in early-onset preeclampsia. *European Journal of Human Genetics*, **2010**, 18, 1006-12	5.3	164
5	Evaluating DNA methylation and gene expression variability in the human term placenta. *Placenta*, **2010**, 31, 1070-7	3.4	68
4	Human placental-specific epipolymorphism and its association with adverse pregnancy outcomes. *PLoS ONE*, **2009**, 4, e7389	3.7	54
3	Hypermethylation of RASSF1A in human and rhesus placentas. *American Journal of Pathology*, **2007**, 170, 941-50	5.8	118
2	Large-scale exome sequencing study implicates both developmental and functional changes in the neurobiology of autism		21
1	Length of uninterrupted CAG repeats, independent of polyglutamine size, results in increased somatic instability and hastened age of onset in Huntington disease		4