Giovanna Pellecchia

List of Publications by Year
in descending order

25	4,166	393982	580395
papers	citations	19 h-index	25 g-index

28	28	28	8090
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Convergence of Genes and Cellular Pathways Dysregulated in Autism Spectrum Disorders. American Journal of Human Genetics, 2014, 94, 677-694.	2.6	819
2	Whole genome sequencing resource identifies 18 new candidate genes for autism spectrum disorder. Nature Neuroscience, 2017, 20, 602-611.	7.1	691
3	Whole-genome sequencing of quartet families with autism spectrum disorder. Nature Medicine, 2015, 21, 185-191.	15.2	457
4	Improved diagnostic yield compared with targeted gene sequencing panels suggests a role for whole-genome sequencing as a first-tier genetic test. Genetics in Medicine, 2018, 20, 435-443.	1.1	404
5	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	1.7	295
6	Genome-wide characteristics of de novo mutations in autism. Npj Genomic Medicine, 2016, 1, 160271-1602710.	1.7	200
7	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.	2.6	156
8	Genome-wide detection of tandem DNA repeats that are expanded in autism. Nature, 2020, 586, 80-86.	13.7	155
9	Brain-expressed exons under purifying selection are enriched for de novo mutations in autism spectrum disorder. Nature Genetics, 2014, 46, 742-747.	9.4	149
10	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, 2752-2768.	1.4	140
11	A large data resource of genomic copy number variation across neurodevelopmental disorders. Npj Genomic Medicine, 2019, 4, 26.	1.7	118
12	Complete Disruption of Autism-Susceptibility Genes by Gene Editing Predominantly Reduces Functional Connectivity of Isogenic Human Neurons. Stem Cell Reports, 2018, 11, 1211-1225.	2.3	111
13	OTUD7A Regulates Neurodevelopmental Phenotypes in the 15q13.3 Microdeletion Syndrome. American Journal of Human Genetics, 2018, 102, 278-295.	2.6	81
14	A high-resolution copy-number variation resource for clinical and population genetics. Genetics in Medicine, 2015, 17, 747-752.	1.1	73
15	Identification of CDC25 as a Common Therapeutic Target for Triple-Negative Breast Cancer. Cell Reports, 2018, 23, 112-126.	2.9	58
16	The Personal Genome Project Canada: findings from whole genome sequences of the inaugural 56 participants. Cmaj, 2018, 190, E126-E136.	0.9	57
17	Mutations in RAB39B in individuals with intellectual disability, autism spectrum disorder, and macrocephaly. Molecular Autism, 2017, 8, 59.	2.6	49
18	Germline and somatic mutations in STXBP1 with diverse neurodevelopmental phenotypes. Neurology: Genetics, 2017, 3, e199.	0.9	41

#	Article	IF	Citations
19	Indexing Effects of Copy Number Variation on Genes Involved in Developmental Delay. Scientific Reports, 2016, 6, 28663.	1.6	35
20	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.	1.1	23
21	Genome-wide tandem repeat expansions contribute to schizophrenia risk. Molecular Psychiatry, 2022, 27, 3692-3698.	4.1	20
22	Clinical Genetic Risk Variants Inform a Functional Protein Interaction Network for Tetralogy of Fallot. Circulation Genomic and Precision Medicine, 2021, 14, e003410.	1.6	15
23	Single allele loss-of-function mutations select and sculpt conditional cooperative networks in breast cancer. Nature Communications, 2021, 12, 5238.	5.8	8
24	Using Next-Generation Sequencing Transcriptomics To Determine Markers of Post-traumatic Symptoms: Preliminary Findings from a Post-deployment Cohort of Soldiers. G3: Genes, Genomes, Genetics, 2019, 9, 463-471.	0.8	7
25	MG-132â€...Diagnostic utility of whole genome sequencing in paediatric medicine. Journal of Medical Genetics, 2015, 52, A12.1-A12.	1.5	1