Cheryl Cytrynbaum

List of Publications by Year
in descending order

28	4,425	394421 19	501196 28
papers	citations	h-index	g-index

28	28	28	8735
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Functional impact of global rare copy number variation in autism spectrum disorders. Nature, 2010, 466, 368-372.	27.8	1,803
2	Whole genome sequencing resource identifies 18 new candidate genes for autism spectrum disorder. Nature Neuroscience, 2017, 20, 602-611.	14.8	691
3	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.	2.4	404
4	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	3.8	295
5	Practical guidelines for managing adults with 22q11.2 deletion syndrome. Genetics in Medicine, 2015, 17, 599-609.	2.4	222
6	CHARGE and Kabuki Syndromes: Gene-Specific DNA Methylation Signatures Identify Epigenetic Mechanisms Linking These Clinically Overlapping Conditions. American Journal of Human Genetics, 2017, 100, 773-788.	6.2	166
7	Elastin: mutational spectrum in supravalvular aortic stenosis. European Journal of Human Genetics, 2000, 8, 955-963.	2.8	147
8	A large data resource of genomic copy number variation across neurodevelopmental disorders. Npj Genomic Medicine, 2019, 4, 26.	3.8	118
9	Velo-cardio-facial syndrome: Implications of microdeletion 22q11 for schizophrenia and mood disorders. American Journal of Medical Genetics Part A, 2001, 105, 354-362.	2.4	91
10	De Novo Variants in the ATPase Module of MORC2 Cause a Neurodevelopmental Disorder with Growth Retardation and Variable Craniofacial Dysmorphism. American Journal of Human Genetics, 2020, 107, 352-363.	6.2	64
11	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. American Journal of Human Genetics, 2020, 106, 596-610.	6.2	59
12	Anatomy of DNA methylation signatures: Emerging insights and applications. American Journal of Human Genetics, 2021, 108, 1359-1366.	6.2	36
13	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	1.2	34
14	De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. American Journal of Human Genetics, 2020, 107, 311-324.	6.2	32
15	Chitayat-Hall and Schaaf-Yang syndromes:a common aetiology: expanding the phenotype of MAGEL2-related disorders. Journal of Medical Genetics, 2018, 55, 316-321.	3.2	31
16	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. American Journal of Human Genetics, 2021, 108, 1053-1068.	6.2	31
17	Expanding the neurodevelopmental phenotypes of individuals with de novo KMT2A variants. Npj Genomic Medicine, 2019, 4, 9.	3.8	29
18	Communicating complex genomic information: A counselling approach derived from research experience with Autism Spectrum Disorder. Patient Education and Counseling, 2018, 101, 352-361.	2.2	27

#	Article	IF	Citations
19	New insights into DNA methylation signatures: SMARCA2 variants in Nicolaides-Baraitser syndrome. BMC Medical Genomics, 2019, 12, 105.	1.5	25
20	An increased prevalence of thyroid disease in children with 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1560-1564.	1.2	24
21	Genome-wide copy number variation analysis identifies novel candidate loci associated with pediatric obesity. European Journal of Human Genetics, 2018, 26, 1588-1596.	2.8	23
22	HIST1H1E heterozygous proteinâ€truncating variants cause a recognizable syndrome with intellectual disability and distinctive facial gestalt: A study to clarify the HIST1H1E syndrome phenotype in 30 individuals. American Journal of Medical Genetics, Part A, 2019, 179, 2049-2055.	1.2	16
23	Genomic imbalance in the centromeric 11p15 imprinting center in three families: Further evidence of a role for IC2 as a cause of Russellâ€“Silver syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2731-2739.	1.2	15
24	Mild Angelman syndrome phenotype due to a mosaic methylation imprinting defect. American Journal of Medical Genetics, Part A, 2015, 167, 1565-1569.	1.2	14
25	Epigenetic signatures in overgrowth syndromes: Translational opportunities. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 491-501.	1.6	10
26	The Clinician-reported Genetic testing Utility InDEx (C-GUIDE): Preliminary evidence of validity and reliability. Genetics in Medicine, 2022, 24, 430-438.	2.4	8
27	Novel heterozygous variants in PXDN cause different anterior segment dysgenesis phenotypes in monozygotic twins. Ophthalmic Genetics, 2021, 42, 624-630.	1.2	5
28	The utility of <scp>DNA</scp> methylation signatures in directing genome sequencing workflow: Kabuki syndrome and <scp>CDK13</scp>â€related disorder. American Journal of Medical Genetics, Part A, 2022, 188, 1368-1375.	1.2	5