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	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
2	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
3	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
4	Novel heterozygous variants in PXDN cause different anterior segment dysgenesis phenotypes in monozygotic twins. Ophthalmic Genetics, 2021, 42, 624-630.	1.2	5
5	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
6	Anatomy of DNA methylation signatures: Emerging insights and applications. American Journal of Human Genetics, 2021, 108, 1359-1366.	6.2	36
7	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
8	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
9	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
10	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
11	New insights into DNA methylation signatures: SMARCA2 variants in Nicolaides-Baraitser syndrome. BMC Medical Genomics, 2019, 12, 105.	1.5	25
12	A large data resource of genomic copy number variation across neurodevelopmental disorders. Npj Genomic Medicine, 2019, 4, 26.	3.8	118
13	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
14	Expanding the neurodevelopmental phenotypes of individuals with de novo KMT2A variants. Npj Genomic Medicine, 2019, 4, 9.	3.8	29
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	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
17	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
18	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

#	Article	IF	Citations
19	Whole genome sequencing resource identifies 18 new candidate genes for autism spectrum disorder. Nature Neuroscience, 2017, 20, 602-611.	14.8	691
20	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
21	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	3.8	295
22	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
23	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
24	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
25	Practical guidelines for managing adults with 22q11.2 deletion syndrome. Genetics in Medicine, 2015, 17, 599-609.	2.4	222
26	Functional impact of global rare copy number variation in autism spectrum disorders. Nature, 2010, 466, 368-372.	27.8	1,803
27	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
28	Elastin: mutational spectrum in supravalvular aortic stenosis. European Journal of Human Genetics, 2000, 8, 955-963.	2.8	147