Ankita Patel

List of Publications by Year
in descending order

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34 papers	2,783 citations	331670 21 h-index	454955 30 g-index
35 all docs	35 docs citations	35 times ranked	7244 citing authors

#	Article	IF	Citations
1	Parental somatic mosaicism for CNV deletions â€“ A need for more sensitive and precise detection methods in clinical diagnostics settings. Genomics, 2020, 112, 2937-2941.	2.9	14
2	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	6.2	86
3	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. Cell, 2017, 168, 830-842.e7.	28.9	66
4	Characterization of chromosomal abnormalities in pregnancy losses reveals critical genes and loci for human early development. Human Mutation, 2017, 38, 669-677.	2.5	28
5	Haploinsufficiency of the E3 ubiquitin-protein ligase gene TRIP12 causes intellectual disability with or without autism spectrum disorders, speech delay, and dysmorphic features. Human Genetics, 2017, 136, 377-386.	3.8	36
6	The importance of phase analysis in multiexon copy number variation detected by aCGH in autosomal recessive disorder loci. , 2017, 173, 2485-2488.		7
7	Chromosomal Microarrays: Understanding Genetics of Neurodevelopmental Disorders and Congenital Anomalies. Journal of Pediatric Genetics, 2017, 06, 042-050.	0.7	24
8	Universal Prenatal Chromosomal Microarray Analysis: Additive Value and Clinical Dilemmas in Fetuses with a Normal Karyotype. American Journal of Perinatology, 2017, 34, 340-348.	1.4	21
9	Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83.	8.2	50
10	Mechanisms for Complex Chromosomal Insertions. PLoS Genetics, 2016, 12, e1006446.	3.5	45
11	Triploidy mosaicism (45,X/68,XX) in an infant presenting with failure to thrive. American Journal of Medical Genetics, Part A, 2016, 170, 694-698.	1.2	6
12	Contribution of genomic copy-number variations in prenatal oral clefts: a multicenter cohort study. Genetics in Medicine, 2016, 18, 1052-1055.	2.4	25
13	Application of DNA Microarray to Clinical Diagnostics. Methods in Molecular Biology, 2016, 1368, 111-132.	0.9	10
14	Recurrent deletions and duplications of chromosome 2q11.2 and 2q13 are associated with variable outcomes. American Journal of Medical Genetics, Part A, 2015, 167, 2664-2673.	1.2	42
15	Adult Low-Hypodiploid Acute B-Lymphoblastic Leukemia WithIKZF3Deletion andTP53Mutation. American Journal of Clinical Pathology, 2015, 144, 263-270.	0.7	10
16	Neurodevelopmental and neurobehavioral characteristics in males and females with CDKL5 duplications. European Journal of Human Genetics, 2015, 23, 915-921.	2.8	32
17	Clinical characterization of int22h1/int22h2-mediated Xq28 duplication/deletion: new cases and literature review. BMC Medical Genetics, 2015, 16, 12.	2.1	37
18	USP7 Acts as a Molecular Rheostat to Promote WASH-Dependent Endosomal Protein Recycling and Is Mutated in a Human Neurodevelopmental Disorder. Molecular Cell, 2015, 59, 956-969.	9.7	175

#	Article	IF	Citations
19	Delineation of candidate genes responsible for structural brain abnormalities in patients with terminal deletions of chromosome 6q27. European Journal of Human Genetics, 2015, 23, 54-60.	2.8	45
20	Molecular Findings Among Patients Referred for Clinical Whole-Exome Sequencing. JAMA - Journal of the American Medical Association, 2014, 312, 1870.	7.4	1,171
21	Combined array CGH plus SNP genome analyses in a single assay for optimized clinical testing. European Journal of Human Genetics, 2014, 22, 79-87.	2.8	112
22	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. American Journal of Human Genetics, 2014, 95, 173-182.	6.2	219
23	Incidental Finding in Copy Number Variation (CNV) Analysis. Current Genetic Medicine Reports, 2014, 2, 179-181.	1.9	1
24	Somatic mosaicism detected by exon-targeted, high-resolution aCGH in 10â€‰362 consecutive cases. European Journal of Human Genetics, 2014, 22, 969-978.	2.8	51
25	Variable levels of tissue mosaicism can confound the interpretation of chromosomal microarray results from peripheral blood. European Journal of Medical Genetics, 2014, 57, 264-266.	1.3	1
26	Clinically silent clonal cytogenetic abnormalities arising in patients treated for lymphoid neoplasms. Leukemia Research, 2014, 38, 896-900.	0.8	8
27	PGM3 Mutations Cause a Congenital Disorder of Glycosylation with Severe Immunodeficiency and Skeletal Dysplasia. American Journal of Human Genetics, 2014, 95, 96-107.	6.2	148
28	Genitourinary Defects Associated with Genomic Deletions in 2p15 Encompassing OTX1. PLoS ONE, 2014, 9, e107028.	2.5	29
29	Is It Time for Arraycgh to Be the First Line Test for Detection of Chromosome Abnormalities in Hematological Disorders-Example Multiple Myeloma. Blood, 2011, 118, 2543-2543.	1.4	1
30	Detection of clinically relevant exonic copy-number changes by array CGH. Human Mutation, 2010, 31, 1326-1342.	2.5	225
31	De novo terminal 22q12.3q13.3 duplication with pituitary hypoplasia (Am J Med Genet Part A) Tj ETQq1 1 0.784314 rgBT /Overlock 1	1.2	0
32	Validation of a targeted DNA microarray for the clinical evaluation of recurrent abnormalities in chronic lymphocytic leukemia. American Journal of Hematology, 2008, 83, 540-546.	4.1	54
33	Clinical Evaluation of a Custom Genome Wide 44K Oligoarray for Copy Number Changes in Acute Myeloid Leukemia. Blood, 2008, 112, 4869-4869.	1.4	0
34	Chromosomal Microarrays: Understanding Genetics of Neurodevelopmental Disorders and Congenital Anomalies. Journal of Pediatric Genetics, 0, 06, .	0.7	0