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List of Publications by Year
in descending order

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Version: 2024-02-01

83 papers	5,330 citations	94433 37 h-index	91884 69 g-index
86 all docs	86 docs citations	86 times ranked	10057 citing authors

#	Article	IF	Citations
1	Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. New England Journal of Medicine, 2017, 376, 21-31.	27.0	565
2	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	6.2	348
3	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
4	Detection of clinically relevant exonic copy-number changes by array CGH. Human Mutation, 2010, 31, 1326-1342.	2.5	225
5	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. American Journal of Human Genetics, 2014, 95, 173-182.	6.2	219
6	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	27.0	205
7	Increased LIS1 expression affects human and mouse brain development. Nature Genetics, 2009, 41, 168-177.	21.4	199
8	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	8.2	184
9	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
10	Positive predictive value estimates for cell-free noninvasive prenatal screening from data of a large referral genetic diagnostic laboratory. American Journal of Obstetrics and Gynecology, 2017, 217, 691.e1-691.e6.	1.3	141
11	NAHR-mediated copy-number variants in a clinical population: Mechanistic insights into both genomic disorders and Mendelizing traits. Genome Research, 2013, 23, 1395-1409.	5.5	120
12	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
13	Reciprocal Crossovers and a Positional Preference for Strand Exchange in Recombination Events Resulting in Deletion or Duplication of Chromosome 17p11.2. American Journal of Human Genetics, 2003, 73, 1302-1315.	6.2	111
14	Clinical exome sequencing for fetuses with ultrasound abnormalities and a suspected Mendelian disorder. Genome Medicine, 2018, 10, 74.	8.2	105
15	Prenatal chromosomal microarray analysis in a diagnostic laboratory; experience with >1000 cases and review of the literature. Prenatal Diagnosis, 2012, 32, 351-361.	2.3	103
16	Mutations of RAI1, a PHD-containing protein, in nondeletion patients with Smith-Magenis syndrome. Human Genetics, 2004, 115, 515-524.	3.8	102
17	Genes in a Refined Smith-Magenis Syndrome Critical Deletion Interval on Chromosome 17p11.2 and the Syntenic Region of the Mouse. Genome Research, 2002, 12, 713-728.	5.5	101
18	Inactivation of Rai1 in mice recapitulates phenotypes observed in chromosome engineered mouse models for Smithâ€“Magenis syndrome. Human Molecular Genetics, 2005, 14, 983-995.	2.9	86

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19	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
20	Rai1 deficiency in mice causes learning impairment and motor dysfunction, whereas Rai1 heterozygous mice display minimal behavioral phenotypes. Human Molecular Genetics, 2007, 16, 1802-1813.	2.9	75
21	Mutations in the Chromatin Regulator Gene BRPF1 Cause Syndromic Intellectual Disability and Deficient Histone Acetylation. American Journal of Human Genetics, 2017, 100, 91-104.	6.2	72
22	Comparison of chromosome analysis and chromosomal microarray analysis: what is the value of chromosome analysis in todayâ€™s genomic array era?. Genetics in Medicine, 2013, 15, 450-457.	2.4	63
23	Deletions of recessive disease genes: CNV contribution to carrier states and disease-causing alleles. Genome Research, 2013, 23, 1383-1394.	5.5	62
24	Novel PIK3CD mutations affecting N-terminal residues of p110Î´ cause activated PI3KÎ´ syndrome (APDS) in humans. Journal of Allergy and Clinical Immunology, 2017, 140, 1152-1156.e10.	2.9	62
25	Rapid prenatal diagnosis using uncultured amniocytes and oligonucleotide array CGH. Prenatal Diagnosis, 2008, 28, 943-949.	2.3	57
26	A clinical survey of mosaic single nucleotide variants in disease-causing genes detected by exome sequencing. Genome Medicine, 2019, 11, 48.	8.2	55
27	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. American Journal of Human Genetics, 2017, 100, 676-688.	6.2	54
28	Novel EED mutation in patient with Weaver syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 541-545.	1.2	52
29	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	2.4	52
30	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
31	Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83.	8.2	50
32	Validation Studies for Single Circulating Trophoblast Genetic Testing as a Form of Noninvasive Prenatal Diagnosis. American Journal of Human Genetics, 2019, 105, 1262-1273.	6.2	47
33	RAI1 point mutations, CAG repeat variation, and SNP analysis in non-deletion Smithâ€“Magenis syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 2454-2463.	1.2	46
34	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
35	Mechanisms for Complex Chromosomal Insertions. PLoS Genetics, 2016, 12, e1006446.	3.5	45
36	De Novo Truncating Variants in SON Cause Intellectual Disability, Congenital Malformations, and Failure to Thrive. American Journal of Human Genetics, 2016, 99, 720-727.	6.2	45

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37	KCTD7 deficiency defines a distinct neurodegenerative disorder with a conserved autophagyâ€lysosome defect. Annals of Neurology, 2018, 84, 766-780.	5.3	42
38	Copy number variant and runs of homozygosity detection by microarrays enabled more precise molecular diagnoses in 11,020 clinical exome cases. Genome Medicine, 2019, 11, 30.	8.2	42
39	Pathogenic variants in USP7 cause a neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies. Genetics in Medicine, 2019, 21, 1797-1807.	2.4	41
40	CHRNA7 triplication associated with cognitive impairment and neuropsychiatric phenotypes in a three-generation pedigree. European Journal of Human Genetics, 2014, 22, 1071-1076.	2.8	37
41	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
42	CNVs cause autosomal recessive genetic diseases with or without involvement of SNV/indels. Genetics in Medicine, 2020, 22, 1633-1641.	2.4	36
43	6q22.1 microdeletion and susceptibility to pediatric epilepsy. European Journal of Human Genetics, 2015, 23, 173-179.	2.8	35
44	Genotype-phenotype correlations in individuals with pathogenicREREvariants. Human Mutation, 2018, 39, 666-675.	2.5	34
45	Loss of Oxidation Resistance 1, OXR1, Is Associated with an Autosomal-Recessive Neurological Disease with Cerebellar Atrophy and Lysosomal Dysfunction. American Journal of Human Genetics, 2019, 105, 1237-1253.	6.2	34
46	Neurodevelopmental and neurobehavioral characteristics in males and females with CDKL5 duplications. European Journal of Human Genetics, 2015, 23, 915-921.	2.8	32
47	A homozygous deletion of 8q24.3 including the NIBP gene associated with severe developmental delay, dysgenesis of the corpus callosum, and dysmorphic facial features. American Journal of Medical Genetics, Part A, 2010, 152A, 1268-1272.	1.2	29
48	Detection of â‰¥1â€‰Mb microdeletions and microduplications in a single cell using custom oligonucleotide arrays. Prenatal Diagnosis, 2012, 32, 10-20.	2.3	29
49	Novel applications of array comparative genomic hybridization in molecular diagnostics. Expert Review of Molecular Diagnostics, 2018, 18, 531-542.	3.1	28
50	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	6.2	27
51	Bi-allelic Mutations in NADSYN1 Cause Multiple Organ Defects and Expand the Genotypic Spectrum of Congenital NAD Deficiency Disorders. American Journal of Human Genetics, 2020, 106, 129-136.	6.2	27
52	4p16.3 microdeletions and microduplications detected by chromosomal microarray analysis: New insights into mechanisms and critical regions. American Journal of Medical Genetics, Part A, 2016, 170, 2540-2550.	1.2	25
53	Whole exome sequencing identifies the first STRADA point mutation in a patient with polyhydramnios, megalencephaly, and symptomatic epilepsy syndrome (PMSE). American Journal of Medical Genetics, Part A, 2016, 170, 2181-2185.	1.2	23
54	PSTPIP1â€associated myeloidâ€related proteinemia inflammatory syndrome: A rare cause of childhood neutropenia associated with systemic inflammation and hyperzincemia. Pediatric Blood and Cancer, 2019, 66, e27439.	1.5	23

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55	Comparison of three whole genome amplification methods for detection of genomic aberrations in single cells. Prenatal Diagnosis, 2016, 36, 823-830.	2.3	22
56	Identification of a RAI1-associated disease network through integration of exome sequencing, transcriptomics, and 3D genomics. Genome Medicine, 2016, 8, 105.	8.2	20
57	Atlas-CNV: a validated approach to call single-exon CNVs in the eMERGESeq gene panel. Genetics in Medicine, 2019, 21, 2135-2144.	2.4	19
58	CRIPT exonic deletion and a novel missense mutation in a female with short stature, dysmorphic features, microcephaly, and pigmentary abnormalities. American Journal of Medical Genetics, Part A, 2016, 170, 2206-2211.	1.2	16
59	GARSâ€related disease in infantile spinal muscular atrophy: Implications for diagnosis and treatment. American Journal of Medical Genetics, Part A, 2020, 182, 1167-1176.	1.2	15
60	Amish nemaline myopathy and dilated cardiomyopathy caused by a homozygous contiguous gene deletion of TNNT1 and TNNI3 in a Mennonite child. European Journal of Medical Genetics, 2019, 62, 103567.	1.3	14
61	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
62	Two de novo novel mutations in one SHANK3 allele in a patient with autism and moderate intellectual disability. , 2018, 176, 973-979.		13
63	Biallelic mutation of FBXL7 suggests a novel form of Hennekam syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 189-194.	1.2	13
64	CSNK2B: A broad spectrum of neurodevelopmental disability and epilepsy severity. Epilepsia, 2021, 62, e103-e109.	5.1	13
65	Retrospective analysis of a clinical exome sequencing cohort reveals the mutational spectrum and identifies candidate diseaseâ€“associated loci for BAFopathies. Genetics in Medicine, 2022, 24, 364-373.	2.4	12
66	Microarray-Based Comparative Genomic Hybridization Using Sex-Matched Reference DNA Provides Greater Sensitivity for Detection of Sex Chromosome Imbalances than Array-Comparative Genomic Hybridization with Sex-Mismatched Reference DNA. Journal of Molecular Diagnostics, 2009, 11, 226-237.	2.8	11
67	Co-occurrence of recurrent duplications of the DiGeorge syndrome region on both chromosome 22 homologues due to inherited and de novo events. Journal of Medical Genetics, 2012, 49, 681-688.	3.2	11
68	Integrated sequencing and array comparative genomic hybridization in familial Parkinson disease. Neurology: Genetics, 2020, 6, e498.	1.9	11
69	BAZ2B haploinsufficiency as a cause of developmental delay, intellectual disability, and autism spectrum disorder. Human Mutation, 2020, 41, 921-925.	2.5	11
70	Homozygous lossâ€ofâ€function variants of TASP1 , a gene encoding an activator of the histone methyltransferases KMT2A and KMT2D, cause a syndrome of developmental delay, happy demeanor, distinctive facial features, and congenital anomalies. Human Mutation, 2019, 40, 1985-1992.	2.5	10
71	RCL1 copy number variants are associated with a range of neuropsychiatric phenotypes. Molecular Psychiatry, 2021, 26, 1706-1718.	7.9	10
72	De novo apparent loss-of-function mutations in PRR12 in three patients with intellectual disability and iris abnormalities. Human Genetics, 2018, 137, 257-264.	3.8	8

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73	Quantitative Assessment of Parental Somatic Mosaicism for Copyâ€Number Variant (CNV) Deletions. Current Protocols in Human Genetics, 2020, 106, e99.	3.5	7
74	<scp>PPP3CA</scp> truncating variants clustered in the regulatory domain cause earlyâ€onset refractory epilepsy. Clinical Genetics, 2021, 100, 227-233.	2.0	7
75	Clinical characterization of individuals with the distal 1q21.1 microdeletion. American Journal of Medical Genetics, Part A, 2021, 185, 1388-1398.	1.2	6
76	Haploinsufficiency of PRR12 causes a spectrum of neurodevelopmental, eye, and multisystem abnormalities. Genetics in Medicine, 2021, 23, 1234-1245.	2.4	6
77	<scp>LMOD2</scp>â€related dilated cardiomyopathy presenting in late infancy. American Journal of Medical Genetics, Part A, 2022, 188, 1858-1862.	1.2	5
78	Nephronophthisis due to a novel DCDC2 variant in a patient from Africanâ€Caribbean descent: A case report. American Journal of Medical Genetics, Part A, 2020, 182, 527-531.	1.2	4
79	Contribution of uniparental disomy in a clinical trio exome cohort of 2675 patients. Molecular Genetics & Genomic Medicine, 2021, 9, e1792.	1.2	4
80	A rare description of pure partial trisomy of 16q12.2q24.3 and review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 2903-2912.	1.2	2
81	Incidental Finding in Copy Number Variation (CNV) Analysis. Current Genetic Medicine Reports, 2014, 2, 179-181.	1.9	1
82	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
83	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