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List of Publications by Year
in descending order

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76 papers	5,788 citations	94381 37 h-index	85498 71 g-index
79 all docs	79 docs citations	79 times ranked	9162 citing authors

#	Article	IF	Citations
1	A DNA Replication Mechanism for Generating Nonrecurrent Rearrangements Associated with Genomic Disorders. Cell, 2007, 131, 1235-1247.	13.5	756
2	Mechanisms underlying structural variant formation in genomic disorders. Nature Reviews Genetics, 2016, 17, 224-238.	7.7	526
3	Characterization of Potocki-Lupski Syndrome (dup(17)(p11.2p11.2)) and Delineation of a Dosage-Sensitive Critical Interval That Can Convey an Autism Phenotype. American Journal of Human Genetics, 2007, 80, 633-649.	2.6	340
4	Mechanisms for recurrent and complex human genomic rearrangements. Current Opinion in Genetics and Development, 2012, 22, 211-220.	1.5	289
5	Complex human chromosomal and genomic rearrangements. Trends in Genetics, 2009, 25, 298-307.	2.9	239
6	Inverted genomic segments and complex triplication rearrangements are mediated by inverted repeats in the human genome. Nature Genetics, 2011, 43, 1074-1081.	9.4	184
7	Complex rearrangements in patients with duplications of MECP2 can occur by fork stalling and template switching. Human Molecular Genetics, 2009, 18, 2188-2203.	1.4	165
8	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	1.1	161
9	Identifying Genes Whose Mutant Transcripts Cause Dominant Disease Traits by Potential Gain-of-Function Alleles. American Journal of Human Genetics, 2018, 103, 171-187.	2.6	160
10	ARMC4 Mutations Cause Primary Ciliary Dyskinesia with Randomization of Left/Right Body Asymmetry. American Journal of Human Genetics, 2013, 93, 357-367.	2.6	150
11	Heterozygous Truncating Variants in POMP Escape Nonsense-Mediated Decay and Cause a Unique Immune Dysregulatory Syndrome. American Journal of Human Genetics, 2018, 102, 1126-1142.	2.6	128
12	Replicative mechanisms for CNV formation are error prone. Nature Genetics, 2013, 45, 1319-1326.	9.4	125
13	Copy-Number Variation Contributes to the Mutational Load of Bardet-Biedl Syndrome. American Journal of Human Genetics, 2016, 99, 318-336.	2.6	112
14	DVL1 Frameshift Mutations Clustering in the Penultimate Exon Cause Autosomal-Dominant Robinow Syndrome. American Journal of Human Genetics, 2015, 96, 612-622.	2.6	110
15	Homozygous and hemizygous CNV detection from exome sequencing data in a Mendelian disease cohort. Nucleic Acids Research, 2017, 45, gkw1237.	6.5	98
16	DVL3 Alleles Resulting in a â˜1 Frameshift of the Last Exon Mediate Autosomal-Dominant Robinow Syndrome. American Journal of Human Genetics, 2016, 98, 553-561.	2.6	88
17	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. American Journal of Human Genetics, 2018, 102, 27-43.	2.6	88
18	From cytogenetics to cytogenomics: whole-genome sequencing as a first-line test comprehensively captures the diverse spectrum of disease-causing genetic variation underlying intellectual disability. Genome Medicine, 2019, 11, 68.	3.6	88

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19	Alu-mediated diverse and complex pathogenic copy-number variants within human chromosome 17 at p13.3. Human Molecular Genetics, 2015, 24, 4061-4077.	1.4	83
20	Recurrent CNVs and SNVs at the NPHP1 Locus Contribute Pathogenic Alleles to Bardet-Biedl Syndrome. American Journal of Human Genetics, 2014, 94, 745-754.	2.6	80
21	Copy number gain at Xp22.31 includes complex duplication rearrangements and recurrent triplications. Human Molecular Genetics, 2011, 20, 1975-1988.	1.4	74
22	Megabase Length Hypermutation Accompanies Human Structural Variation at 17p11.2. Cell, 2019, 176, 1310-1324.e10.	13.5	73
23	Microhomology-Mediated Mechanisms Underlie Non-Recurrent Disease-Causing Microdeletions of the FOXL2 Gene or Its Regulatory Domain. PLoS Genetics, 2013, 9, e1003358.	1.5	72
24	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. Cell, 2017, 168, 830-842.e7.	13.5	66
25	The Phylogeography of African Brazilians. Human Heredity, 2008, 65, 23-32.	0.4	62
26	Genomic disorders: A window into human gene and genome evolution. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 1765-1771.	3.3	60
27	Whole-Genome Sequencing of Cytogenetically Balanced Chromosome Translocations Identifies Potentially Pathological Gene Disruptions and Highlights the Importance of Microhomology in the Mechanism of Formation. Human Mutation, 2017, 38, 180-192.	1.1	58
28	Complex Genomic Rearrangements at the PLP1 Locus Include Triplication and Quadruplication. PLoS Genetics, 2015, 11, e1005050.	1.5	57
29	Pre- and Post-Columbian Gene and Cultural Continuity: The Case of the Gaucho from Southern Brazil. Human Heredity, 2007, 64, 160-171.	0.4	55
30	No association found between gr/gr deletions and infertility in Brazilian males. Molecular Human Reproduction, 2006, 12, 269-273.	1.3	54
31	Alu-specific microhomology-mediated deletion of the final exon of SPAST in three unrelated subjects with hereditary spastic paraplegia. Genetics in Medicine, 2011, 13, 582-592.	1.1	53
32	Characterization of molecular and cellular phenotypes associated with a heterozygous CNTNAP2 deletion using patient-derived hiPSC neural cells. NPJ Schizophrenia, 2015, 1, .	2.0	52
33	The Behavioral Phenotype in <scp>MECP</scp>2 Duplication Syndrome: A Comparison With Idiopathic Autism. Autism Research, 2013, 6, 42-50.	2.1	50
34	Inverted Low-Copy Repeats and Genome Instability-A Genome-Wide Analysis. Human Mutation, 2013, 34, 210-220.	1.1	48
35	Copy number variation at the breakpoint region of isochromosome 17q. Genome Research, 2008, 18, 1724-1732.	2.4	47
36	Absence of Heterozygosity Due to Template Switching during Replicative Rearrangements. American Journal of Human Genetics, 2015, 96, 555-564.	2.6	45

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37	Mechanisms for Complex Chromosomal Insertions. PLoS Genetics, 2016, 12, e1006446.	1.5	45
38	REST Final-Exon-Truncating Mutations Cause Hereditary Gingival Fibromatosis. American Journal of Human Genetics, 2017, 101, 149-156.	2.6	44
39	Dosage Changes of a Segment at 17p13.1 Lead to Intellectual Disability and Microcephaly as a Result of Complex Genetic Interaction of Multiple Genes. American Journal of Human Genetics, 2014, 95, 565-578.	2.6	40
40	High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population. American Journal of Human Genetics, 2021, 108, 1981-2005.	2.6	38
41	CHRNA7 triplication associated with cognitive impairment and neuropsychiatric phenotypes in a three-generation pedigree. European Journal of Human Genetics, 2014, 22, 1071-1076.	1.4	37
42	Comprehensive structural variation genome map of individuals carrying complex chromosomal rearrangements. PLoS Genetics, 2019, 15, e1007858.	1.5	36
43	Nonrecurrent 17p11.2p12 Rearrangement Events that Result in Two Concomitant Genomic Disorders: The PMP22-RAI1 Contiguous Gene Duplication Syndrome. American Journal of Human Genetics, 2015, 97, 691-707.	2.6	33
44	Structural variation of the human genome: mechanisms, assays, and role in male infertility. Systems Biology in Reproductive Medicine, 2011, 57, 3-16.	1.0	32
45	Targeted Treatment of Individuals With Psychosis Carrying a Copy Number Variant Containing a Genomic Triplication of the Glycine Decarboxylase Gene. Biological Psychiatry, 2019, 86, 523-535.	0.7	32
46	NIPBL rearrangements in Cornelia de Lange syndrome: evidence for replicative mechanism and genotypeâ€“phenotype correlation. Genetics in Medicine, 2012, 14, 313-322.	1.1	30
47	Exonic duplication CNV of NDRG1 associated with autosomal-recessive HMSN-Lom/CMT4D. Genetics in Medicine, 2014, 16, 386-394.	1.1	30
48	Genitourinary Defects Associated with Genomic Deletions in 2p15 Encompassing OTX1. PLoS ONE, 2014, 9, e107028.	1.1	29
49	Dual molecular diagnosis contributes to atypical Praderâ€“Willi phenotype in monozygotic twins. American Journal of Medical Genetics, Part A, 2017, 173, 2451-2455.	0.7	26
50	Marker chromosome genomic structure and temporal origin implicate a chromoanasynthesis event in a family with pleiotropic psychiatric phenotypes. Human Mutation, 2018, 39, 939-946.	1.1	26
51	Distinct patterns of complex rearrangements and a mutational signature of microhomeology are frequently observed in PLP1 copy number gain structural variants. Genome Medicine, 2019, 11, 80.	3.6	24
52	Interchromosomal template-switching as a novel molecular mechanism for imprinting perturbations associated with Temple syndrome. Genome Medicine, 2019, 11, 25.	3.6	22
53	Efficient CNV breakpoint analysis reveals unexpected structural complexity and correlation of dosage-sensitive genes with clinical severity in genomic disorders. Human Molecular Genetics, 2017, 26, 1927-1941.	1.4	20
54	A large CRISPR-induced bystander mutation causes immune dysregulation. Communications Biology, 2019, 2, 70.	2.0	19

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55	Genetic and molecular mechanism for distinct clinical phenotypes conveyed by allelic truncating mutations implicated in FBN1. Molecular Genetics & Genomic Medicine, 2020, 8, e1023.	0.6	19
56	Complex genomic rearrangements: an underestimated cause of rare diseases. Trends in Genetics, 2022, 38, 1134-1146.	2.9	19
57	The role of combined SNV and CNV burden in patients with distal symmetric polyneuropathy. Genetics in Medicine, 2016, 18, 443-451.	1.1	18
58	Novel parent-of-origin-specific differentially methylated loci on chromosome 16. Clinical Epigenetics, 2019, 11, 60.	1.8	18
59	Mechanisms for the Generation of Two Quadruplications Associated with Split-Hand Malformation. Human Mutation, 2016, 37, 160-164.	1.1	16
60	Alu-Alu mediated intragenic duplications in IFT81 and MATN3 are associated with skeletal dysplasias. Human Mutation, 2018, 39, 1456-1467.	1.1	16
61	Novel pathogenic genomic variants leading to autosomal dominant and recessive Robinow syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3593-3600.	0.7	16
62	Optimization of a multiplex minisequencing protocol for population studies and medical genetics. Genetics and Molecular Research, 2005, 4, 115-25.	0.3	16
63	Xq22 deletions and correlation with distinct neurological disease traits in females: Further evidence for a contiguous gene syndrome. Human Mutation, 2020, 41, 150-168.	1.1	15
64	Clinical genomics and contextualizing genome variation in the diagnostic laboratory. Expert Review of Molecular Diagnostics, 2020, 20, 995-1002.	1.5	14
65	Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability. Human Genetics and Genomics Advances, 2022, 3, 100074.	1.0	14
66	Cytogenetically visible inversions are formed by multiple molecular mechanisms. Human Mutation, 2020, 41, 1979-1998.	1.1	12
67	Divergent Levels of Marker Chromosomes in an hiPSC-Based Model ofÂPsychosis. Stem Cell Reports, 2017, 8, 519-528.	2.3	11
68	Chimeric transcripts resulting from complex duplications in chromosome Xq28. Human Genetics, 2016, 135, 253-256.	1.8	8
69	Extremity anomalies associated with Robinow syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3584-3592.	0.7	8
70	Phenotypic and mutational spectrum of ROR2 â€related Robinow syndrome. Human Mutation, 2022, 43, 900-918.	1.1	8
71	Xq26.3 Duplication in a Boy With Motor Delay and Low Muscle Tone Refines the X-Linked Acrogigantism Genetic Locus. Journal of the Endocrine Society, 2018, 2, 1100-1108.	0.1	7
72	Craniofacial phenotypes associated with <scp>Robinow</scp> syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3606-3612.	0.7	7

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73	Characterization of the Robinow syndrome skeletal phenotype, bone microâ€architecture, and genotypeâ€“phenotype correlations with the osteosclerotic form. American Journal of Medical Genetics, Part A, 2020, 182, 2632-2640.	0.7	5
74	Chromoanagenesis Event Underlies a de novo Pericentric and Multiple Paracentric Inversions in a Single Chromosome Causing Coffinâ€“Siris Syndrome. Frontiers in Genetics, 2021, 12, 708348.	1.1	5
75	Neurocognitive, adaptive, and psychosocial functioning in individuals with Robinow syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3576-3583.	0.7	4
76	Inside Back Cover, Volume 41, Issue 1. Human Mutation, 2020, 41, ii.	1.1	0