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List of Publications by Year
in descending order

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65 papers	5,921 citations	201385 27 h-index	123241 61 g-index
66 all docs	66 docs citations	66 times ranked	11720 citing authors

#	Article	IF	Citations
1	Ancient human genome sequence of an extinct Palaeo-Eskimo. Nature, 2010, 463, 757-762.	13.7	750
2	Antagonism of microRNA-122 in mice by systemically administered LNA-antimiR leads to up-regulation of a large set of predicted target mRNAs in the liver. Nucleic Acids Research, 2008, 36, 1153-1162.	6.5	630
3	Altered MicroRNA Expression Confined to Specific Epithelial Cell Subpopulations in Breast Cancer. Cancer Research, 2007, 67, 11612-11620.	0.4	515
4	JARID2 regulates binding of the Polycomb repressive complexâ€‰2 to target genes in ES cells. Nature, 2010, 464, 306-310.	13.7	499
5	MicroRNA-138 regulates osteogenic differentiation of human stromal (mesenchymal) stem cells in vivo. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 6139-6144.	3.3	443
6	MicroRNA expression in the adult mouse central nervous system. Rna, 2008, 14, 432-444.	1.6	427
7	MicroRNA-31 functions as an oncogenic microRNA in mouse and human lung cancer cells by repressing specific tumor suppressors. Journal of Clinical Investigation, 2010, 120, 1298-1309.	3.9	353
8	Immature truncated O-glycophenotype of cancer directly induces oncogenic features. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, E4066-75.	3.3	251
9	A Functional Link between the Histone Demethylase PHF8 and the Transcription Factor ZNF711 in X-Linked Mental Retardation. Molecular Cell, 2010, 38, 165-178.	4.5	186
10	Jarid1b targets genes regulating development and is involved in neural differentiation. EMBO Journal, 2011, 30, 4586-4600.	3.5	183
11	Uncovering Growth-Suppressive MicroRNAs in Lung Cancer. Clinical Cancer Research, 2009, 15, 1177-1183.	3.2	167
12	RESTâ€“Mediated Recruitment of Polycomb Repressor Complexes in Mammalian Cells. PLoS Genetics, 2012, 8, e1002494.	1.5	140
13	Corpus callosum abnormalities, intellectual disability, speech impairment, and autism in patients with haploinsufficiency of ARID1B. Clinical Genetics, 2012, 82, 248-255.	1.0	126
14	Hypomorphic Mutations in PGAP2, Encoding a GPI-Anchor-Remodeling Protein, Cause Autosomal-Recessive Intellectual Disability. American Journal of Human Genetics, 2013, 92, 575-583.	2.6	87
15	The small RNA content of human sperm reveals pseudogene-derived piRNAs complementary to protein-coding genes. Rna, 2015, 21, 1085-1095.	1.6	83
16	Genome-wide detection of chromosomal rearrangements, indels, and mutations in circular chromosomes by short read sequencing. Genome Research, 2011, 21, 1388-1393.	2.4	79
17	Aberrant expression of miRâ€218 and miRâ€204 in human mesial temporal lobe epilepsy and hippocampal sclerosisâ€”Convergence on axonal guidance. Epilepsia, 2014, 55, 2017-2027.	2.6	71
18	Genome-wide Analysis of CDX2 Binding in Intestinal Epithelial Cells (Caco-2). Journal of Biological Chemistry, 2010, 285, 25115-25125.	1.6	68

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19	Elevation of brain-enriched miRNAs in cerebrospinal fluid of patients with acute ischemic stroke. Biomarker Research, 2017, 5, 24.	2.8	59
20	Profiling microRNAs in lung tissue from pigs infected with Actinobacillus pleuropneumoniae. BMC Genomics, 2012, 13, 459.	1.2	54
21	Nextâ€generation sequencing: proof of concept for antenatal prediction of the fetal <scp>K</scp>ell blood group phenotype from cellâ€free fetal <scp>DNA</scp> in maternal plasma. Transfusion, 2013, 53, 2892-2898.	0.8	51
22	Germline Chromothripsis Driven by L1-Mediated Retrotransposition and Alu/Alu Homologous Recombination. Human Mutation, 2016, 37, 385-395.	1.1	50
23	The strength of combined cytogenetic and mate-pair sequencing techniques illustrated by a germline chromothripsis rearrangement involving FOXP2. European Journal of Human Genetics, 2014, 22, 338-343.	1.4	46
24	Non-disjunction of chromosome 13. Human Molecular Genetics, 2007, 16, 2004-2010.	1.4	38
25	YKL-40 Is Differentially Expressed in Human Embryonic Stem Cells and in Cell Progeny of the Three Germ Layers. Journal of Histochemistry and Cytochemistry, 2012, 60, 188-204.	1.3	36
26	Haploinsufficiency of CELF4 at 18q12.2 is associated with developmental and behavioral disorders, seizures, eye manifestations, and obesity. European Journal of Human Genetics, 2012, 20, 1315-1319.	1.4	34
27	Accurate Breakpoint Mapping in Apparently Balanced Translocation Families with Discordant Phenotypes Using Whole Genome Mate-Pair Sequencing. PLoS ONE, 2017, 12, e0169935.	1.1	31
28	Identification of the BRD1 interaction network and its impact on mental disorder risk. Genome Medicine, 2016, 8, 53.	3.6	29
29	Risks and Recommendations in Prenatally Detected De Novo Balanced Chromosomal Rearrangements from Assessment of Long-Term Outcomes. American Journal of Human Genetics, 2018, 102, 1090-1103.	2.6	29
30	Replicative and non-replicative mechanisms in the formation of clustered CNVs are indicated by whole genome characterization. PLoS Genetics, 2018, 14, e1007780.	1.5	28
31	Genome-wide DNA methylation analysis of transient neonatal diabetes type 1 patients with mutations in ZFP57. BMC Medical Genetics, 2016, 17, 29.	2.1	27
32	The Hedgehog signaling pathway â€“ implications for drug targets in cancer and neurodegenerative disorders. Pharmacogenomics, 2003, 4, 411-429.	0.6	26
33	Neurodevelopmental disorders associated with dosage imbalance ofZBTB20correlate with the morbidity spectrum of ZBTB20 candidate target genes. Journal of Medical Genetics, 2014, 51, 605-613.	1.5	26
34	Cost-effective multiplexing before capture allows screening of 25â€‰000 clinically relevant SNPs in childhood acute lymphoblastic leukemia. Leukemia, 2011, 25, 1001-1006.	3.3	20
35	Regulatory variants of FOXG1 in the context of its topological domain organisation. European Journal of Human Genetics, 2018, 26, 186-196.	1.4	20
36	Aggressiveness of non-EMT breast cancer cells relies on FBXO11 activity. Molecular Cancer, 2018, 17, 171.	7.9	20

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37	Epigenetic remodelling and dysregulation of DLGAP4 is linked with early-onset cerebellar ataxia. Human Molecular Genetics, 2014, 23, 6163-6176.	1.4	19
38	Very short DNA segments can be detected and handled by the repair machinery during germline chromothriptic chromosome reassembly. Human Mutation, 2018, 39, 709-716.	1.1	19
39	Validation of Genome-Wide Intervertebral Disk Calcification Associations in Dachshund and Further Investigation of the Chromosome 12 Susceptibility Locus. Frontiers in Genetics, 2012, 3, 225.	1.1	18
40	Integrative analysis of genomic variants reveals new associations of candidate haploinsufficient genes with congenital heart disease. PLoS Genetics, 2021, 17, e1009679.	1.5	17
41	Diagnostic pitfalls in vitamin B6â€dependent epilepsy caused by mutations in the PLPBP gene. JIMD Reports, 2019, 50, 1-8.	0.7	16
42	Phenotypic subregions within the split-hand/foot malformation 1 locus. Human Genetics, 2016, 135, 345-357.	1.8	15
43	Position effect, cryptic complexity, and direct gene disruption as disease mechanisms in de novo apparently balanced translocation cases. PLoS ONE, 2018, 13, e0205298.	1.1	14
44	Aberrant brain microRNA target and miRISC gene expression in the anx/anx anorexia mouse model. Gene, 2012, 497, 181-190.	1.0	12
45	Haploinsufficiency of ARHGAP42 is associated with hypertension. European Journal of Human Genetics, 2019, 27, 1296-1303.	1.4	12
46	GLI1 Is Involved in Cell Cycle Regulation and Proliferation of NT2 Embryonal Carcinoma Stem Cells. DNA and Cell Biology, 2008, 27, 251-256.	0.9	11
47	Cryptic breakpoint identified by whole-genome mate-pair sequencing in a rare paternally inherited complex chromosomal rearrangement. Molecular Cytogenetics, 2018, 11, 34.	0.4	11
48	Trisomy 13 due to rea(13q;13q) is caused by i(13) and not rob(13;13)(q10;q10) in the majority of cases. , 2005, 132A, 310-313.		9
49	Dysregulation of FOXG1 by ring chromosome 14. Molecular Cytogenetics, 2015, 8, 24.	0.4	8
50	Partial USH2A deletions contribute to Usher syndrome in Denmark. European Journal of Human Genetics, 2015, 23, 1646-1651.	1.4	8
51	Biparental inheritance of chromosomal abnormalities in male twins with non-syndromic mental retardation. European Journal of Medical Genetics, 2011, 54, e383-e388.	0.7	7
52	Case report: â€˜AARS2 leukodystrophyâ€™. Molecular Genetics and Metabolism Reports, 2021, 28, 100782.	0.4	7
53	The segregation of different submicroscopic imbalances underlying the clinical variability associated with a familial karyotypically balanced translocation. Molecular Cytogenetics, 2015, 8, 106.	0.4	6
54	Evaluation of two methods for generating cRNA for microarray experiments from nanogram amounts of total RNA. Analytical Biochemistry, 2006, 358, 111-119.	1.1	5

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55	Sequence and expression analysis of gaps in human chromosome 20. Nucleic Acids Research, 2012, 40, 6660-6672.	6.5	5
56	Enrichment of megabase-sized DNA molecules for single-molecule optical mapping and next-generation sequencing. Scientific Reports, 2017, 7, 17893.	1.6	5
57	Mutation analysis of the Sonic hedgehog promoter and putative enhancer elements in Parkinson's disease patients. Molecular Brain Research, 2004, 126, 207-211.	2.5	4
58	Multigenic truncation of the semaphorinâ€“plexin pathway by a germline chromothriptic rearrangement associated with Moebius syndrome. Human Mutation, 2019, 40, 1057-1062.	1.1	4
59	Paroxysmal Cranial Dyskinesia and Nailâ€Patella Syndrome Caused by a Novel Variant in the LMX1B Gene. Movement Disorders, 2020, 35, 2343-2347.	2.2	2
60	Mitochondrial dysfunction induced by variation in the non-coding genome â€“ A proposed workflow to improve diagnostics. Mitochondrion, 2020, 53, 255-259.	1.6	1
61	A splice-site variant in the lncRNA gene cosegregates in the large Volkmann cataract family. Molecular Vision, 2019, 25, 1-11.	1.1	1
62	9p21.3 Microdeletion involving CDKN2A/2B in a young patient with multiple primary cancers and review of the literature. Journal of Physical Education and Sports Management, 2022, 8, a006164.	0.5	1
63	International Breakpoint Mapping Consortium (IBMC). Systematic Mapping of Chromosomal Breakpoints in the Context of Phenotypes and Nuclear Genome Organization. Cancer Genetics, 2015, 208, 359-360.	0.2	0
64	Re-Examination of Danish Carriers of Balanced Chromosomal Inversions. Cancer Genetics, 2016, 209, 231.	0.2	0
65	Systematic Mapping of Chromosomal Breakpoints in the Context of Phenotypes and Nuclear Genome Organization. Cancer Genetics, 2016, 209, 301.	0.2	0