Jing Zhang

List of Publications by Year in descending order

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227 papers	18,050 citations	17429 63 h-index	16164 124 g-index
233 all docs	233 docs citations	233 times ranked	21651 citing authors

#	Article	IF	Citations
1	Molecular Findings Among Patients Referred for Clinical Whole-Exome Sequencing. JAMA - Journal of the American Medical Association, 2014, 312, 1870.	3.8	1,171
2	Structural Variation in the Human Genome and its Role in Disease. Annual Review of Medicine, 2010, 61, 437-455.	5.0	1,015
3	Genome architecture, rearrangements and genomic disorders. Trends in Genetics, 2002, 18, 74-82.	2.9	815
4	Whole-Genome Sequencing in a Patient with Charcotâ€“Marieâ€“Tooth Neuropathy. New England Journal of Medicine, 2010, 362, 1181-1191.	13.9	698
5	Recurrent reciprocal 1q21.1 deletions and duplications associated with microcephaly or macrocephaly and developmental and behavioral abnormalities. Nature Genetics, 2008, 40, 1466-1471.	9.4	535
6	Genomic Disorders: Molecular Mechanisms for Rearrangements and Conveyed Phenotypes. PLoS Genetics, 2005, 1, e49.	1.5	496
7	Recurrent reciprocal 16p11.2 rearrangements associated with global developmental delay, behavioural problems, dysmorphism, epilepsy, and abnormal head size. Journal of Medical Genetics, 2010, 47, 332-341.	1.5	447
8	Chromosome Catastrophes Involve Replication Mechanisms Generating Complex Genomic Rearrangements. Cell, 2011, 146, 889-903.	13.5	391
9	Genomic and Genic Deletions of the FOX Gene Cluster on 16q24.1 and Inactivating Mutations of FOXF1 Cause Alveolar Capillary Dysplasia and Other Malformations. American Journal of Human Genetics, 2009, 84, 780-791.	2.6	389
10	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	3.3	348
11	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
12	Mutant chromatin remodeling protein SMARCAL1 causes Schimke immuno-osseous dysplasia. Nature Genetics, 2002, 30, 215-220.	9.4	297
13	Use of array CGH in the evaluation of dysmorphology, malformations, developmental delay, and idiopathic mental retardation. Current Opinion in Genetics and Development, 2007, 17, 182-192.	1.5	293
14	Development and validation of a CGH microarray for clinical cytogenetic diagnosis. Genetics in Medicine, 2005, 7, 422-432.	1.1	241
15	Somatic mosaicism: implications for disease and transmission genetics. Trends in Genetics, 2015, 31, 382-392.	2.9	234
16	Detection of clinically relevant exonic copy-number changes by array CGH. Human Mutation, 2010, 31, 1326-1342.	1.1	225
17	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. American Journal of Human Genetics, 2014, 95, 173-182.	2.6	219
18	Periaxin Mutations Cause Recessive Dejerine-Sottas Neuropathy. American Journal of Human Genetics, 2001, 68, 325-333.	2.6	205

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19	Alveolar Capillary Dysplasia. American Journal of Respiratory and Critical Care Medicine, 2011, 184, 172-179.	2.5	194
20	Clinical Implementation of Chromosomal Microarray Analysis: Summary of 2513 Postnatal Cases. PLoS ONE, 2007, 2, e327.	1.1	191
21	Position Effects Due to Chromosome Breakpoints that Map â^¼900 Kb Upstream and â^¼1.3 Mb Downstream of SOX9 in Two Patients with Campomelic Dysplasia. American Journal of Human Genetics, 2005, 76, 652-662.	2.6	178
22	A small recurrent deletion within 15q13.3 is associated with a range of neurodevelopmental phenotypes. Nature Genetics, 2009, 41, 1269-1271.	9.4	171
23	Genome Architecture Catalyzes Nonrecurrent Chromosomal Rearrangements. American Journal of Human Genetics, 2003, 72, 1101-1116.	2.6	167
24	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
25	Microarrayâ€based CGH detects chromosomal mosaicism not revealed by conventional cytogenetics. American Journal of Medical Genetics, Part A, 2007, 143A, 1679-1686.	0.7	158
26	Prenatal diagnosis of chromosomal abnormalities using array-based comparative genomic hybridization. Genetics in Medicine, 2006, 8, 719-727.	1.1	154
27	Assessing structural variation in a personal genomeâ€”towards a human reference diploid genome. BMC Genomics, 2015, 16, 286.	1.2	153
28	Molecular-evolutionary mechanisms for genomic disorders. Current Opinion in Genetics and Development, 2002, 12, 312-319.	1.5	151
29	Rare pathogenic microdeletions and tandem duplications are microhomology-mediated and stimulated by local genomic architecture. Human Molecular Genetics, 2009, 18, 3579-3593.	1.4	143
30	Genomic Imbalances in Neonates With Birth Defects: High Detection Rates by Using Chromosomal Microarray Analysis. Pediatrics, 2008, 122, 1310-1318.	1.0	137
31	DNA sequence of human chromosome 17 and analysis of rearrangement in the human lineage. Nature, 2006, 440, 1045-1049.	13.7	130
32	Small noncoding differentially methylated copy-number variants, including lncRNA genes, cause a lethal lung developmental disorder. Genome Research, 2013, 23, 23-33.	2.4	127
33	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. Genetics in Medicine, 2019, 21, 816-825.	1.1	127
34	The Breakpoint Region of the Most Common Isochromosome, i(17q), in Human Neoplasia Is Characterized by a Complex Genomic Architecture with Large, Palindromic, Low-Copy Repeats. American Journal of Human Genetics, 2004, 74, 1-10.	2.6	122
35	NAHR-mediated copy-number variants in a clinical population: Mechanistic insights into both genomic disorders and Mendelizing traits. Genome Research, 2013, 23, 1395-1409.	2.4	120
36	Emergence of a Predominant Clone of Community-Acquired Staphylococcus aureus Among Children in Houston, Texas. Pediatric Infectious Disease Journal, 2005, 24, 201-206.	1.1	116

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37	Clinical spectrum associated with recurrent genomic rearrangements in chromosome 17q12. European Journal of Human Genetics, 2010, 18, 278-284.	1.4	114
38	Identification of chromosome abnormalities in subtelomeric regions by microarray analysis: A study of 5,380 cases. American Journal of Medical Genetics, Part A, 2008, 146A, 2242-2251.	0.7	113
39	Combined array CGH plus SNP genome analyses in a single assay for optimized clinical testing. European Journal of Human Genetics, 2014, 22, 79-87.	1.4	112
40	Structures and molecular mechanisms for common 15q13.3 microduplications involving CHRNA7: benign or pathological?. Human Mutation, 2010, 31, 840-850.	1.1	111
41	Variability in clinical phenotype despite common chromosomal deletion in Smith-Magenis syndrome [del(17)(p11.2p11.2)]. Genetics in Medicine, 2003, 5, 430-434.	1.1	104
42	Duplications of FOXG1 in 14q12 are associated with developmental epilepsy, mental retardation, and severe speech impairment. European Journal of Human Genetics, 2011, 19, 102-107.	1.4	104
43	Parent of Origin, Mosaicism, and Recurrence Risk: Probabilistic Modeling Explains the Broken Symmetry of Transmission Genetics. American Journal of Human Genetics, 2014, 95, 345-359.	2.6	103
44	A Mild PUM1 Mutation Is Associated with Adult-Onset Ataxia, whereas Haploinsufficiency Causes Developmental Delay and Seizures. Cell, 2018, 172, 924-936.e11.	13.5	103
45	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.	2.4	101
46	Use of array CGH to detect exonic copy number variants throughout the genome in autism families detects a novel deletion in TMLHE. Human Molecular Genetics, 2011, 20, 4360-4370.	1.4	101
47	Phenotypic spectrum and genotypeâ€“phenotype correlations of NRXN1 exon deletions. European Journal of Human Genetics, 2012, 20, 1240-1247.	1.4	99
48	Phenotypic manifestations of copy number variation in chromosome 16p13.11. European Journal of Human Genetics, 2011, 19, 280-286.	1.4	97
49	NovelFOXF1Mutations in Sporadic and Familial Cases of Alveolar Capillary Dysplasia with Misaligned Pulmonary Veins Imply a Role for its DNA Binding Domain. Human Mutation, 2013, 34, 801-811.	1.1	97
50	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. American Journal of Human Genetics, 2019, 104, 213-228.	2.6	90
51	The Alu-Rich Genomic Architecture of SPAST Predisposes to Diverse and Functionally Distinct Disease-Associated CNV Alleles. American Journal of Human Genetics, 2014, 95, 143-161.	2.6	87
52	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	2.6	86
53	Structure and Evolution of the Smith-Magenis Syndrome Repeat Gene Clusters, SMS-REPs. Genome Research, 2002, 12, 729-738.	2.4	85
54	Identification of a Recurrent Microdeletion at 17q23.1q23.2 Flanked by Segmental Duplications Associated with Heart Defects and Limb Abnormalities. American Journal of Human Genetics, 2010, 86, 454-461.	2.6	85

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55	Insertional translocation detected using FISH confirmation of arrayâ€comparative genomic hybridization (aCGH) results. American Journal of Medical Genetics, Part A, 2010, 152A, 1111-1126.	0.7	85
56	Pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins. Human Genetics, 2016, 135, 569-586.	1.8	85
57	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
58	Genome-wide analyses of LINEâ€“LINE-mediated nonallelic homologous recombination. Nucleic Acids Research, 2015, 43, 2188-2198.	6.5	79
59	Copy number gain at Xp22.31 includes complex duplication rearrangements and recurrent triplications. Human Molecular Genetics, 2011, 20, 1975-1988.	1.4	74
60	Predicting human genes susceptible to genomic instability associated with Alu/Alu-mediated rearrangements. Genome Research, 2018, 28, 1228-1242.	2.4	74
61	Role of genomic architecture in PLP1 duplication causing Pelizaeus-Merzbacher disease. Human Molecular Genetics, 2006, 15, 2250-2265.	1.4	73
62	Observation and prediction of recurrent human translocations mediated by NAHR between nonhomologous chromosomes. Genome Research, 2011, 21, 33-46.	2.4	72
63	SOX9cre1, a cis-acting regulatory element located 1.1ÂMb upstream of SOX9, mediates its enhancement through the SHH pathway. Human Molecular Genetics, 2007, 16, 1143-1156.	1.4	68
64	Severe mental retardation, seizures, and hypotonia due to deletions of MEF2C. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 1042-1051.	1.1	68
65	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. Cell, 2017, 168, 830-842.e7.	13.5	66
66	Redefined genomic architecture in 15q24 directed by patient deletion/duplication breakpoint mapping. Human Genetics, 2009, 126, 589-602.	1.8	65
67	Sotos syndrome common deletion is mediated by directly oriented subunits within inverted Sos-REP low-copy repeats. Human Molecular Genetics, 2005, 14, 535-542.	1.4	64
68	The phenotype of recurrent 10q22q23 deletions and duplications. European Journal of Human Genetics, 2011, 19, 400-408.	1.4	63
69	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.	1.1	63
70	Deletions of recessive disease genes: CNV contribution to carrier states and disease-causing alleles. Genome Research, 2013, 23, 1383-1394.	2.4	62
71	Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. American Journal of Human Genetics, 2017, 101, 503-515.	2.6	61
72	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	2.6	61

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73	Human endogenous retroviral elements promote genome instability via non-allelic homologous recombination. BMC Biology, 2014, 12, 74.	1.7	60
74	Male-to-female sex reversal associated with an â¯¼250Âkb deletion upstream of NR0B1 (DAX1). Human Genetics, 2007, 122, 63-70.	1.8	59
75	Recurrent Distal 7q11.23 Deletion Including HIP1 and YWHAG Identified in Patients with Intellectual Disabilities, Epilepsy, and Neurobehavioral Problems. American Journal of Human Genetics, 2010, 87, 857-865.	2.6	58
76	Intragenic rearrangements in NRXN1 in three families with autism spectrum disorder, developmental delay, and speech delay. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 983-993.	1.1	58
77	Microdeletion and Microduplication Syndromes. Methods in Molecular Biology, 2012, 838, 29-75.	0.4	58
78	Detection of copy-number variation in AUTS2 gene by targeted exonic array CGH in patients with developmental delay and autistic spectrum disorders. European Journal of Human Genetics, 2013, 21, 343-346.	1.4	56
79	Phenotypic expansion of TBX4 mutations to include acinar dysplasia of the lungs. American Journal of Medical Genetics, Part A, 2016, 170, 2440-2444.	0.7	56
80	Mutational and genotypeâ€“phenotype correlation analyses in 28 Polish patients with Cornelia de Lange syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 1531-1541.	0.7	55
81	Population Bottlenecks as a Potential Major Shaping Force of Human Genome Architecture. PLoS Genetics, 2007, 3, e119.	1.5	55
82	A clinical survey of mosaic single nucleotide variants in disease-causing genes detected by exome sequencing. Genome Medicine, 2019, 11, 48.	3.6	55
83	Exon deletions of the EP300 and CREBBP genes in two children with Rubinsteinâ€“Taybi syndrome detected by aCGH. European Journal of Human Genetics, 2011, 19, 43-49.	1.4	54
84	Novel 9q34.11 gene deletions encompassing combinations of four Mendelian disease genes: STXBP1, SPTAN1, ENG, and TOR1A. Genetics in Medicine, 2012, 14, 868-876.	1.1	51
85	Somatic mosaicism detected by exon-targeted, high-resolution aCGH in 10â€‰362 consecutive cases. European Journal of Human Genetics, 2014, 22, 969-978.	1.4	51
86	The S52F FOXF1 Mutation Inhibits STAT3 Signaling and Causes Alveolar Capillary Dysplasia. American Journal of Respiratory and Critical Care Medicine, 2019, 200, 1045-1056.	2.5	51
87	Genomic and Epigenetic Complexity of the FOXF1 Locus in 16q24.1: Implications for Development and Disease. Current Genomics, 2015, 16, 107-116.	0.7	51
88	Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83.	3.6	50
89	Rare DNA copy number variants in cardiovascular malformations with extracardiac abnormalities. European Journal of Human Genetics, 2013, 21, 173-181.	1.4	49
90	An estimation of the prevalence of genomic disorders using chromosomal microarray data. Journal of Human Genetics, 2018, 63, 795-801.	1.1	49

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91	Application of array comparative genomic hybridization in 102 patients with epilepsy and additional neurodevelopmental disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 760-771.	1.1	48
92	Inverted Low-Copy Repeats and Genome Instability-A Genome-Wide Analysis. Human Mutation, 2013, 34, 210-220.	1.1	48
93	Infants with Atypical Presentations of Alveolar Capillary Dysplasia with Misalignment of the Pulmonary Veins Who Underwent Bilateral Lung Transplantation. Journal of Pediatrics, 2018, 194, 158-164.e1.	0.9	48
94	Clinical, Histopathological, and Molecular Diagnostics in Lethal Lung Developmental Disorders. American Journal of Respiratory and Critical Care Medicine, 2019, 200, 1093-1101.	2.5	47
95	Two deletions overlapping a distant FOXF1 enhancer unravel the role of lncRNA LINC01081 in etiology of alveolar capillary dysplasia with misalignment of pulmonary veins. American Journal of Medical Genetics, Part A, 2014, 164, 2013-2019.	0.7	46
96	Interstitial deletion of 6q25.2â€“q25.3: a novel microdeletion syndrome associated with microcephaly, developmental delay, dysmorphic features and hearing loss. European Journal of Human Genetics, 2009, 17, 573-581.	1.4	45
97	Early-onset seizures due to mosaic exonic deletions of CDKL5 in a male and two females. Genetics in Medicine, 2011, 13, 447-452.	1.1	45
98	Recurrent deletions and reciprocal duplications of 10q11.21q11.23 including CHAT and SLC18A3 are likely mediated by complex low-copy repeats. Human Mutation, 2012, 33, 165-179.	1.1	45
99	Absence of Heterozygosity Due to Template Switching during Replicative Rearrangements. American Journal of Human Genetics, 2015, 96, 555-564.	2.6	45
100	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.	1.4	45
101	Mechanisms for Complex Chromosomal Insertions. PLoS Genetics, 2016, 12, e1006446.	1.5	45
102	The complex behavioral phenotype of 15q13.3 microdeletion syndrome. Genetics in Medicine, 2016, 18, 1111-1118.	1.1	45
103	Alu-specific microhomology-mediated deletions in CDKL5 in females with early-onset seizure disorder. Neurogenetics, 2009, 10, 363-369.	0.7	44
104	Ovotestes and XY sex reversal in a female with an interstitial9q33.3-q34.1 deletion encompassingNR5A1 andLMX1B causing features of genitopatellar syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1071-1081.	0.7	43
105	Int22h-1/int22h-2-mediated Xq28 rearrangements: intellectual disability associated with duplications and in utero male lethality with deletions. Journal of Medical Genetics, 2011, 48, 840-850.	1.5	43
106	Deletions in chromosome 6p22.3-p24.3, including ATXN1, are associated with developmental delay and autism spectrum disorders. Molecular Cytogenetics, 2012, 5, 17.	0.4	43
107	TM4SF20 Ancestral Deletion and Susceptibility to a Pediatric Disorder of Early Language Delay and Cerebral White Matter Hyperintensities. American Journal of Human Genetics, 2013, 93, 197-210.	2.6	43
108	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	5.8	43

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109	Branchiootorenal syndrome and oculoauriculovertebral spectrum features associated with duplication ofSIX1,SIX6, andOTX2resulting from a complex chromosomal rearrangement. American Journal of Medical Genetics, Part A, 2008, 146A, 2480-2489.	0.7	42
110	Delineation of a deletion region critical for corpus callosal abnormalities in chromosome 1q43â€“q44. European Journal of Human Genetics, 2012, 20, 176-179.	1.4	42
111	A familial case of alveolar capillary dysplasia with misalignment of pulmonary veins supports paternal imprinting of FOXF1 in human. European Journal of Human Genetics, 2013, 21, 474-477.	1.4	42
112	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.	3.6	42
113	A syndrome of short stature, microcephaly and speech delay is associated with duplications reciprocal to the common Sotos syndrome deletion. European Journal of Human Genetics, 2010, 18, 258-261.	1.4	41
114	Recurrent microdeletions of 15q25.2 are associated with increased risk of congenital diaphragmatic hernia, cognitive deficits and possibly Diamond-Blackfan anaemia. Journal of Medical Genetics, 2010, 47, 777-781.	1.5	40
115	Recurrent HERV-H-Mediated 3q13.2-q13.31 Deletions Cause a Syndrome of Hypotonia and Motor, Language, and Cognitive Delays. Human Mutation, 2013, 34, 1415-1423.	1.1	40
116	Interstitial deletion 9q22.32-q33.2 associated with additional familial translocation t(9;17)(q34.11;p11.2) in a patient with Gorlin-Goltz syndrome and features of Nail-Patella syndrome. , 2004, 124A, 179-191.		38
117	Application of custom-designed oligonucleotide array CGH in 145 patients with autistic spectrum disorders. European Journal of Human Genetics, 2013, 21, 620-625.	1.4	37
118	Incidental copy-number variants identified by routine genome testing in a clinical population. Genetics in Medicine, 2013, 15, 45-54.	1.1	37
119	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
120	Application of array comparative genomic hybridization in 256 patients with developmental delay or intellectual disability. Journal of Applied Genetics, 2014, 55, 125-144.	1.0	37
121	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.	1.8	36
122	Challenges in clinical interpretation of microduplications detected by array CGH analysis. American Journal of Medical Genetics, Part A, 2010, 152A, 1089-1100.	0.7	35
123	6q22.1 microdeletion and susceptibility to pediatric epilepsy. European Journal of Human Genetics, 2015, 23, 173-179.	1.4	35
124	Complex translocation disrupting TCF4 and altering TCF4 isoform expression segregates as mild autosomal dominant intellectual disability. Orphanet Journal of Rare Diseases, 2016, 11, 62.	1.2	35
125	Duplication of Xq26.2-q27.1, includingSOX3, in a mother and daughter with short stature and dyslalia. American Journal of Medical Genetics, Part A, 2005, 138A, 11-17.	0.7	34
126	PTCH1 duplication in a family with microcephaly and mild developmental delay. European Journal of Human Genetics, 2009, 17, 267-271.	1.4	34

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127	Hominoid lineage specific amplification of low-copy repeats on 22q11.2 (LCR22s) associated with velo-cardio-facial/digeorge syndrome. Human Molecular Genetics, 2007, 16, 2560-2571.	1.4	32
128	HERVâ€mediated genomic rearrangement of EYA1 in an individual with branchioâ€otoâ€renal syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 2854-2860.	0.7	32
129	Neurodevelopmental and neurobehavioral characteristics in males and females with CDKL5 duplications. European Journal of Human Genetics, 2015, 23, 915-921.	1.4	32
130	Comparative Analyses of Lung Transcriptomes in Patients with Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins and in Foxf1 Heterozygous Knockout Mice. PLoS ONE, 2014, 9, e94390.	1.1	31
131	Cytogenetic and molecular characterization of two isodicentric Y chromosomes. American Journal of Medical Genetics Part A, 2001, 101, 20-25.	2.4	30
132	AT-rich repeats associated with chromosome 22q11.2 rearrangement disorders shape human genome architecture on Yq12. Genome Research, 2007, 17, 451-460.	2.4	30
133	Clinical and molecularâ€cytogenetic evaluation of a family with partial Jacobsen syndrome without thrombocytopenia caused by an â¼45 Mb deletion del(11)(q24.3). American Journal of Medical Genetics, Part A, 2008, 146A, 2449-2454.	0.7	30
134	Low-level parental somatic mosaic SNVs in exomes from a large cohort of trios with diverse suspected Mendelian conditions. Genetics in Medicine, 2020, 22, 1768-1776.	1.1	30
135	Phenotypic findings due to trisomy 7p15.3-pter including theTWIST locus. American Journal of Medical Genetics Part A, 2001, 103, 56-62.	2.4	29
136	Detection of â‰¥1â€‰Mb microdeletions and microduplications in a single cell using custom oligonucleotide arrays. Prenatal Diagnosis, 2012, 32, 10-20.	1.1	29
137	Molecular analysis of a constitutional complex genome rearrangement with 11 breakpoints involving chromosomes 3, 11, 12, and 21 and a â¼40.5-Mb submicroscopic deletion in a patient with mild mental retardation. Human Genetics, 2005, 118, 267-275.	1.8	28
138	IncreasedSTAG2dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. Human Molecular Genetics, 2015, 24, 7171-7181.	1.4	28
139	Characterization of chromosomal abnormalities in pregnancy losses reveals critical genes and loci for human early development. Human Mutation, 2017, 38, 669-677.	1.1	28
140	Expanding the genotypeâ€“phenotype correlation in subtelomeric 19p13.3 microdeletions using high resolution clinical chromosomal microarray analysis. American Journal of Medical Genetics, Part A, 2013, 161, 2953-2963.	0.7	25
141	Small marker chromosomes in two patients with segmental aneusomy for proximal 17p. Human Genetics, 2004, 115, 1-7.	1.8	24
142	A girl with deletion 9q22.1â€“q22.32 including thePTCHandROR2genes identified by genomeâ€wide arrayâ€CGH. American Journal of Medical Genetics, Part A, 2007, 143A, 1885-1889.	0.7	24
143	Disruption of the SCN2A and SCN3A genes in a patient with mental retardation, neurobehavioral and psychiatric abnormalities, and a history of infantile seizures. Clinical Genetics, 2011, 80, 191-195.	1.0	24
144	Kabuki syndrome-like features associated with a small ring chromosome X andXIST gene expression. American Journal of Medical Genetics Part A, 2001, 102, 286-292.	2.4	23

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145	Chromosome conformation capture-on-chip analysis of long-range cis-interactions of the SOX9 promoter. Chromosome Research, 2013, 21, 781-788.	1.0	23
146	Small rare recurrent deletions and reciprocal duplications in 2q21.1, including brain-specific ARHGEF4 and GPR148. Human Molecular Genetics, 2012, 21, 3345-3355.	1.4	22
147	Fusion of Large-Scale Genomic Knowledge and Frequency Data Computationally Prioritizes Variants in Epilepsy. PLoS Genetics, 2013, 9, e1003797.	1.5	22
148	Interchromosomal template-switching as a novel molecular mechanism for imprinting perturbations associated with Temple syndrome. Genome Medicine, 2019, 11, 25.	3.6	22
149	Minimal phenotype in a girl with trisomy 15q due to t(X;15)(q22.3;q11.2) translocation. American Journal of Medical Genetics, Part A, 2006, 140A, 442-452.	0.7	21
150	16q24.1 microdeletion in a premature newborn: Usefulness of array-based comparative genomic hybridization in persistent pulmonary hypertension of the newborn. Pediatric Critical Care Medicine, 2011, 12, e427-e432.	0.2	21
151	Small genomic rearrangements involving FMR1 support the importance of its gene dosage for normal neurocognitive function. Neurogenetics, 2012, 13, 333-339.	0.7	21
152	Trisomy 17p10-p12 due to mosaic supernumerary marker chromosome: Delineation of molecular breakpoints and clinical phenotype, and comparison to other proximal 17p segmental duplications. American Journal of Medical Genetics, Part A, 2005, 138A, 175-180.	0.7	20
153	Novel FOXF1 Deep Intronic Deletion Causes Lethal Lung Developmental Disorder, Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins. Human Mutation, 2013, 34, 1467-1471.	1.1	20
154	Evidence against <scp>ZNF</scp>469 being causative for keratoconus in Polish patients. Acta Ophthalmologica, 2016, 94, 289-294.	0.6	20
155	Lethal lung hypoplasia and vascular defects in mice with conditionalFoxf1overexpression. Biology Open, 2016, 5, 1595-1606.	0.6	20
156	Molecular cytogenetic characterization of eight small supernumerary marker chromosomes originating from chromosomes 2, 4, 8,18, and 21 in three patients. Journal of Applied Genetics, 2007, 48, 167-175.	1.0	19
157	Narrowing the FOXF1 distant enhancer region on 16q24.1 critical for ACDMPV. Clinical Epigenetics, 2016, 8, 112.	1.8	19
158	Variants in SKP1, PROB1, and IL17B genes at keratoconus 5q31.1â€“q35.3 susceptibility locus identified by whole-exome sequencing. European Journal of Human Genetics, 2017, 25, 73-78.	1.4	19
159	Cornelia de Lange syndrome case due to genomic rearrangements including NIPBL. European Journal of Medical Genetics, 2010, 53, 378-382.	0.7	18
160	TGFBR2 deletion in a 20â€monthâ€old female with developmental delay and microcephaly. American Journal of Medical Genetics, Part A, 2011, 155, 1442-1447.	0.7	18
161	Novel parent-of-origin-specific differentially methylated loci on chromosome 16. Clinical Epigenetics, 2019, 11, 60.	1.8	18
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