Jing Zhang

List of Publications by Citations

221 papers	14,670 citations	60 h-index	115 g-index
233 ext. papers	16,701 ext. citations	7.2 avg, IF	6.12 L-index

#	Paper	IF	Citations
221	Molecular findings among patients referred for clinical whole-exome sequencing. *JAMA - Journal of the American Medical Association*, **2014**, 312, 1870-9	27.4	915
220	Structural variation in the human genome and its role in disease. *Annual Review of Medicine*, **2010**, 61, 437-55	17.4	827
219	Genome architecture, rearrangements and genomic disorders. *Trends in Genetics*, **2002**, 18, 74-82	8.5	703
218	Whole-genome sequencing in a patient with Charcot-Marie-Tooth neuropathy. *New England Journal of Medicine*, **2010**, 362, 1181-91	59.2	613
217	Recurrent reciprocal 1q21.1 deletions and duplications associated with microcephaly or macrocephaly and developmental and behavioral abnormalities. *Nature Genetics*, **2008**, 40, 1466-71	36.3	457
216	Genomic disorders: molecular mechanisms for rearrangements and conveyed phenotypes. *PLoS Genetics*, **2005**, 1, e49	6	424
215	Recurrent reciprocal 16p11.2 rearrangements associated with global developmental delay, behavioural problems, dysmorphism, epilepsy, and abnormal head size. *Journal of Medical Genetics*, **2010**, 47, 332-41	5.8	362
214	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, 780-91	11	328
213	Chromosome catastrophes involve replication mechanisms generating complex genomic rearrangements. *Cell*, **2011**, 146, 889-903	56.2	315
212	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, 633-49	11	307
211	Use of array CGH in the evaluation of dysmorphology, malformations, developmental delay, and idiopathic mental retardation. *Current Opinion in Genetics and Development*, **2007**, 17, 182-92	4.9	256
210	Mutant chromatin remodeling protein SMARCAL1 causes Schimke immuno-osseous dysplasia. *Nature Genetics*, **2002**, 30, 215-20	36.3	250
209	Development and validation of a CGH microarray for clinical cytogenetic diagnosis. *Genetics in Medicine*, **2005**, 7, 422-32	8.1	219
208	Use of Exome Sequencing for Infants in Intensive Care Units: Ascertainment of Severe Single-Gene Disorders and Effect on Medical Management. *JAMA Pediatrics*, **2017**, 171, e173438	8.3	215
207	Detection of clinically relevant exonic copy-number changes by array CGH. *Human Mutation*, **2010**, 31, 1326-42	4.7	195
206	Periaxin mutations cause recessive Dejerine-Sottas neuropathy. *American Journal of Human Genetics*, **2001**, 68, 325-33	11	184
205	Somatic mosaicism: implications for disease and transmission genetics. *Trends in Genetics*, **2015**, 31, 382-92	8.5	182

204	Parental somatic mosaicism is underrecognized and influences recurrence risk of genomic disorders. *American Journal of Human Genetics*, **2014**, 95, 173-82	11	172
203	Clinical implementation of chromosomal microarray analysis: summary of 2513 postnatal cases. *PLoS ONE*, **2007**, 2, e327	3.7	172
202	Genome architecture catalyzes nonrecurrent chromosomal rearrangements. *American Journal of Human Genetics*, **2003**, 72, 1101-16	11	159
201	Position effects due to chromosome breakpoints that map approximately 900 Kb upstream and approximately 1.3 Mb downstream of SOX9 in two patients with campomelic dysplasia. *American Journal of Human Genetics*, **2005**, 76, 652-62	11	158
200	A small recurrent deletion within 15q13.3 is associated with a range of neurodevelopmental phenotypes. *Nature Genetics*, **2009**, 41, 1269-71	36.3	155
199	Alveolar capillary dysplasia. *American Journal of Respiratory and Critical Care Medicine*, **2011**, 184, 172-9	10.2	147
198	Complex rearrangements in patients with duplications of MECP2 can occur by fork stalling and template switching. *Human Molecular Genetics*, **2009**, 18, 2188-203	5.6	143
197	Microarray-based CGH detects chromosomal mosaicism not revealed by conventional cytogenetics. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1679-86	2.5	141
196	Prenatal diagnosis of chromosomal abnormalities using array-based comparative genomic hybridization. *Genetics in Medicine*, **2006**, 8, 719-27	8.1	139
195	Rare pathogenic microdeletions and tandem duplications are microhomology-mediated and stimulated by local genomic architecture. *Human Molecular Genetics*, **2009**, 18, 3579-93	5.6	126
194	Molecular-evolutionary mechanisms for genomic disorders. *Current Opinion in Genetics and Development*, **2002**, 12, 312-9	4.9	126
193	Genomic imbalances in neonates with birth defects: high detection rates by using chromosomal microarray analysis. *Pediatrics*, **2008**, 122, 1310-8	7.4	124
192	Assessing structural variation in a personal genome-towards a human reference diploid genome. *BMC Genomics*, **2015**, 16, 286	4.5	117
191	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	11	116
190	DNA sequence of human chromosome 17 and analysis of rearrangement in the human lineage. *Nature*, **2006**, 440, 1045-9	50.4	114
189	Small noncoding differentially methylated copy-number variants, including lncRNA genes, cause a lethal lung developmental disorder. *Genome Research*, **2013**, 23, 23-33	9.7	108
188	Structures and molecular mechanisms for common 15q13.3 microduplications involving CHRNA7: benign or pathological?. *Human Mutation*, **2010**, 31, 840-50	4.7	101
187	Emergence of a predominant clone of community-acquired Staphylococcus aureus among children in Houston, Texas. *Pediatric Infectious Disease Journal*, **2005**, 24, 201-6	3.4	99

186	Duplications of FOXG1 in 14q12 are associated with developmental epilepsy, mental retardation, and severe speech impairment. *European Journal of Human Genetics*, **2011**, 19, 102-7	5.3	96
185	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, 2242-51	2.5	96
184	Combined array CGH plus SNP genome analyses in a single assay for optimized clinical testing. *European Journal of Human Genetics*, **2014**, 22, 79-87	5.3	95
183	NAHR-mediated copy-number variants in a clinical population: mechanistic insights into both genomic disorders and Mendelizing traits. *Genome Research*, **2013**, 23, 1395-409	9.7	95
182	Clinical spectrum associated with recurrent genomic rearrangements in chromosome 17q12. *European Journal of Human Genetics*, **2010**, 18, 278-84	5.3	90
181	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, 713-28	9.7	88
180	Variability in clinical phenotype despite common chromosomal deletion in Smith-Magenis syndrome [del(17)(p11.2p11.2)]. *Genetics in Medicine*, **2003**, 5, 430-4	8.1	87
179	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, 4360-70	5.6	84
178	Phenotypic spectrum and genotype-phenotype correlations of NRXN1 exon deletions. *European Journal of Human Genetics*, **2012**, 20, 1240-7	5.3	82
177	Phenotypic manifestations of copy number variation in chromosome 16p13.11. *European Journal of Human Genetics*, **2011**, 19, 280-6	5.3	82
176	Novel FOXF1 mutations 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, 801-11	4.7	80
175	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**, 85, 537	11	78
174	Parent of origin, mosaicism, and recurrence risk: probabilistic modeling explains the broken symmetry of transmission genetics. *American Journal of Human Genetics*, **2014**, 95, 345-59	11	76
173	Structure and evolution of the Smith-Magenis syndrome repeat gene clusters, SMS-REPs. *Genome Research*, **2002**, 12, 729-38	9.7	72
172	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, 454-61	11	71
171	Insertional translocation detected using FISH confirmation of array-comparative genomic hybridization (aCGH) results. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1111-26	2.5	71
170	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. *Genetics in Medicine*, **2019**, 21, 816-825	8.1	71
169	The Alu-rich genomic architecture of SPAST predisposes to diverse and functionally distinct disease-associated CNV alleles. *American Journal of Human Genetics*, **2014**, 95, 143-61	11	66

168	A Mild PUM1 Mutation Is Associated with Adult-Onset Ataxia, whereas Haploinsufficiency Causes Developmental Delay and Seizures. *Cell*, **2018**, 172, 924-936.e11	56.2	65
167	Role of genomic architecture in PLP1 duplication causing Pelizaeus-Merzbacher disease. *Human Molecular Genetics*, **2006**, 15, 2250-65	5.6	64
166	Redefined genomic architecture in 15q24 directed by patient deletion/duplication breakpoint mapping. *Human Genetics*, **2009**, 126, 589-602	6.3	63
165	Alu-mediated diverse and complex pathogenic copy-number variants within human chromosome 17 at p13.3. *Human Molecular Genetics*, **2015**, 24, 4061-77	5.6	62
164	Pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins. *Human Genetics*, **2016**, 135, 569-586	6.3	62
163	Observation and prediction of recurrent human translocations mediated by NAHR between nonhomologous chromosomes. *Genome Research*, **2011**, 21, 33-46	9.7	61
162	Copy number gain at Xp22.31 includes complex duplication rearrangements and recurrent triplications. *Human Molecular Genetics*, **2011**, 20, 1975-88	5.6	61
161	SOX9cre1, a cis-acting regulatory element located 1.1 Mb upstream of SOX9, mediates its enhancement through the SHH pathway. *Human Molecular Genetics*, **2007**, 16, 1143-56	5.6	60
160	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	11	58
159	Sotos syndrome common deletion is mediated by directly oriented subunits within inverted Sos-REP low-copy repeats. *Human Molecular Genetics*, **2005**, 14, 535-42	5.6	56
158	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. *Cell*, **2017**, 168, 830-842.e7	56.2	53
157	Genome-wide analyses of LINE-LINE-mediated nonallelic homologous recombination. *Nucleic Acids Research*, **2015**, 43, 2188-98	20.1	52
156	The phenotype of recurrent 10q22q23 deletions and duplications. *European Journal of Human Genetics*, **2011**, 19, 400-8	5.3	52
155	Deletions of recessive disease genes: CNV contribution to carrier states and disease-causing alleles. *Genome Research*, **2013**, 23, 1383-94	9.7	51
154	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, 343-6	5.3	50
153	Male-to-female sex reversal associated with an approximately 250 kb deletion upstream of NR0B1 (DAX1). *Human Genetics*, **2007**, 122, 63-70	6.3	50
152	Mutational and genotype-phenotype correlation analyses in 28 Polish patients with Cornelia de Lange syndrome. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1531-41	2.5	50
151	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. *American Journal of Human Genetics*, **2017**, 100, 352-363	11	49

150	Exon deletions of the EP300 and CREBBP genes in two children with Rubinstein-Taybi syndrome detected by aCGH. *European Journal of Human Genetics*, **2011**, 19, 43-9	5.3	49
149	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, 857-65	11	48
148	Microdeletion and microduplication syndromes. *Methods in Molecular Biology*, **2012**, 838, 29-75	1.4	47
147	Comparison of chromosome analysis and chromosomal microarray analysis: what is the value of chromosome analysis in today genomic array era?. *Genetics in Medicine*, **2013**, 15, 450-7	8.1	47
146	Somatic mosaicism detected by exon-targeted, high-resolution aCGH in 10,362 consecutive cases. *European Journal of Human Genetics*, **2014**, 22, 969-78	5.3	46
145	Human endogenous retroviral elements promote genome instability via non-allelic homologous recombination. *BMC Biology*, **2014**, 12, 74	7.3	46
144	Population bottlenecks as a potential major shaping force of human genome architecture. *PLoS Genetics*, **2007**, 3, e119	6	46
143	Predicting human genes susceptible to genomic instability associated with /-mediated rearrangements. *Genome Research*, **2018**, 28, 1228-1242	9.7	44
142	Rare DNA copy number variants in cardiovascular malformations with extracardiac abnormalities. *European Journal of Human Genetics*, **2013**, 21, 173-81	5.3	42
141	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**, 164A, 2013-9	2.5	42
140	Inverted low-copy repeats and genome instability--a genome-wide analysis. *Human Mutation*, **2013**, 34, 210-20	4.7	41
139	Novel 9q34.11 gene deletions encompassing combinations of four Mendelian disease genes: STXBP1, SPTAN1, ENG, and TOR1A. *Genetics in Medicine*, **2012**, 14, 868-76	8.1	41
138	Severe mental retardation, seizures, and hypotonia due to deletions of MEF2C. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2010**, 153B, 1042-51	3.5	41
137	Genomic and Epigenetic Complexity of the FOXF1 Locus in 16q24.1: Implications for Development and Disease. *Current Genomics*, **2015**, 16, 107-16	2.6	41
136	Early-onset seizures due to mosaic exonic deletions of CDKL5 in a male and two females. *Genetics in Medicine*, **2011**, 13, 447-52	8.1	40
135	Absence of heterozygosity due to template switching during replicative rearrangements. *American Journal of Human Genetics*, **2015**, 96, 555-64	11	39
134	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, 760-71	3.5	39
133	Alu-specific microhomology-mediated deletions in CDKL5 in females with early-onset seizure disorder. *Neurogenetics*, **2009**, 10, 363-9	3	39

132	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, 983-93	3.5	39
131	Identification of novel candidate disease genes from de novo exonic copy number variants. *Genome Medicine*, **2017**, 9, 83	14.4	38
130	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	11	37
129	Diagnostic implications of genetic copy number variation in epilepsy plus. *Epilepsia*, **2019**, 60, 689-706	6.4	37
128	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, 474-7	5.3	37
127	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, 165-79	4.7	36
126	Deletions in chromosome 6p22.3-p24.3, including ATXN1, are associated with developmental delay and autism spectrum disorders. *Molecular Cytogenetics*, **2012**, 5, 17	2	36
125	Delineation of a deletion region critical for corpus callosal abnormalities in chromosome 1q43-q44. *European Journal of Human Genetics*, **2012**, 20, 176-9	5.3	36
124	Ovotestes and XY sex reversal in a female with an interstitial 9q33.3-q34.1 deletion encompassing NR5A1 and LMX1B causing features of Genitopatellar syndrome. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1071-81	2.5	36
123	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	5.3	35
122	Recurrent HERV-H-mediated 3q13.2-q13.31 deletions cause a syndrome of hypotonia and motor, language, and cognitive delays. *Human Mutation*, **2013**, 34, 1415-23	4.7	35
121	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, 840-50	5.8	35
120	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, 258-61	5.3	35
119	Interstitial deletion of 6q25.2-q25.3: a novel microdeletion syndrome associated with microcephaly, developmental delay, dysmorphic features and hearing loss. *European Journal of Human Genetics*, **2009**, 17, 573-81	5.3	34
118	Branchiootorenal syndrome and oculoauriculovertebral spectrum features associated with duplication of SIX1, SIX6, and OTX2 resulting from a complex chromosomal rearrangement. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2480-9	2.5	33
117	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	3.6	32
116	Phenotypic expansion of TBX4 mutations to include acinar dysplasia of the lungs. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2440-4	2.5	32
115	The complex behavioral phenotype of 15q13.3 microdeletion syndrome. *Genetics in Medicine*, **2016**, 18, 1111-1118	8.1	32

114	Application of array comparative genomic hybridization in 256 patients with developmental delay or intellectual disability. *Journal of Applied Genetics*, **2014**, 55, 125-44	2.5	32
113	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	11	32
112	Incidental copy-number variants identified by routine genome testing in a clinical population. *Genetics in Medicine*, **2013**, 15, 45-54	8.1	32
111	Mechanisms for Complex Chromosomal Insertions. *PLoS Genetics*, **2016**, 12, e1006446	6	31
110	Application of custom-designed oligonucleotide array CGH in 145 patients with autistic spectrum disorders. *European Journal of Human Genetics*, **2013**, 21, 620-5	5.3	30
109	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, 777-81	5.8	30
108	Duplication of Xq26.2-q27.1, including SOX3, in a mother and daughter with short stature and dyslalia. *American Journal of Medical Genetics, Part A*, **2005**, 138, 11-7	2.5	30
107	A clinical survey of mosaic single nucleotide variants in disease-causing genes detected by exome sequencing. *Genome Medicine*, **2019**, 11, 48	14.4	29
106	CHRNA7 triplication associated with cognitive impairment and neuropsychiatric phenotypes in a three-generation pedigree. *European Journal of Human Genetics*, **2014**, 22, 1071-6	5.3	29
105	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, 179-91		29
104	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	14.4	27
103	Neurodevelopmental and neurobehavioral characteristics in males and females with CDKL5 duplications. *European Journal of Human Genetics*, **2015**, 23, 915-21	5.3	27
102	PTCH1 duplication in a family with microcephaly and mild developmental delay. *European Journal of Human Genetics*, **2009**, 17, 267-71	5.3	27
101	Challenges in clinical interpretation of microduplications detected by array CGH analysis. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1089-100	2.5	27
100	Hominoid lineage specific amplification of low-copy repeats on 22q11.2 (LCR22s) associated with velo-cardio-facial/digeorge syndrome. *Human Molecular Genetics*, **2007**, 16, 2560-71	5.6	27
99	The S52F FOXF1 Mutation Inhibits STAT3 Signaling and Causes Alveolar Capillary Dysplasia. *American Journal of Respiratory and Critical Care Medicine*, **2019**, 200, 1045-1056	10.2	26
98	Molecular analysis of a constitutional complex genome rearrangement with 11 breakpoints involving chromosomes 3, 11, 12, and 21 and a approximately 0.5-Mb submicroscopic deletion in a patient with mild mental retardation. *Human Genetics*, **2005**, 118, 267-75	6.3	26
97	Cytogenetic and molecular characterization of two isodicentric Y chromosomes. *American Journal of Medical Genetics Part A*, **2001**, 101, 20-5		26

96	6q22.1 microdeletion and susceptibility to pediatric epilepsy. *European Journal of Human Genetics*, **2015**, 23, 173-9	5.3	25
95	Increased STAG2 dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. *Human Molecular Genetics*, **2015**, 24, 7171-81	5.6	24
94	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	3.7	24
93	Clinical and molecular-cytogenetic evaluation of a family with partial Jacobsen syndrome without thrombocytopenia caused by an approximately 5 Mb deletion del(11)(q24.3). *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2449-54	2.5	23
92	Small marker chromosomes in two patients with segmental aneusomy for proximal 17p. *Human Genetics*, **2004**, 115, 1-7	6.3	23
91	An estimation of the prevalence of genomic disorders using chromosomal microarray data. *Journal of Human Genetics*, **2018**, 63, 795-801	4.3	22
90	Detection of 1Mb microdeletions and microduplications in a single cell using custom oligonucleotide arrays. *Prenatal Diagnosis*, **2012**, 32, 10-20	3.2	22
89	HERV-mediated genomic rearrangement of EYA1 in an individual with branchio-oto-renal syndrome. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2854-60	2.5	22
88	Phenotypic findings due to trisomy 7p15.3-pter including the TWIST locus. *American Journal of Medical Genetics Part A*, **2001**, 103, 56-62		22
87	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	6.3	21
86	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. *Nature Communications*, **2019**, 10, 4679	17.4	21
85	A girl with deletion 9q22.1-q22.32 including the PTCH and ROR2 genes identified by genome-wide array-CGH. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1885-9	2.5	21
84	Clinical, Histopathological, and Molecular Diagnostics in Lethal Lung Developmental Disorders. *American Journal of Respiratory and Critical Care Medicine*, **2019**, 200, 1093-1101	10.2	20
83	Expanding the genotype-phenotype correlation in subtelomeric 19p13.3 microdeletions using high resolution clinical chromosomal microarray analysis. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2953-63	2.5	20
82	AT-rich repeats associated with chromosome 22q11.2 rearrangement disorders shape human genome architecture on Yq12. *Genome Research*, **2007**, 17, 451-60	9.7	20
81	Novel FOXF1 deep intronic deletion causes lethal lung developmental disorder, alveolar capillary dysplasia with misalignment of pulmonary veins. *Human Mutation*, **2013**, 34, 1467-71	4.7	19
80	Chromosome conformation capture-on-chip analysis of long-range cis-interactions of the SOX9 promoter. *Chromosome Research*, **2013**, 21, 781-8	4.4	19
79	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, 191-5	4	19

78	Complex translocation disrupting TCF4 and altering TCF4 isoform expression segregates as mild autosomal dominant intellectual disability. *Orphanet Journal of Rare Diseases*, **2016**, 11, 62	4.2	19
77	Characterization of chromosomal abnormalities in pregnancy losses reveals critical genes and loci for human early development. *Human Mutation*, **2017**, 38, 669-677	4.7	18
76	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**, 140, 442-52	2.5	18
75	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, 175-80	2.5	18
74	Kabuki syndrome-like features associated with a small ring chromosome X and XIST gene expression. *American Journal of Medical Genetics Part A*, **2001**, 102, 286-92		18
73	Narrowing the distant enhancer region on 16q24.1 critical for ACDMPV. *Clinical Epigenetics*, **2016**, 8, 112	7.7	17
72	TGFBR2 deletion in a 20-month-old female with developmental delay and microcephaly. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1442-7	2.5	17
71	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, 167-75	2.5	17
70	Evidence against ZNF469 being causative for keratoconus in Polish patients. *Acta Ophthalmologica*, **2016**, 94, 289-94	3.7	17
69	Lethal lung hypoplasia and vascular defects in mice with conditional Foxf1 overexpression. *Biology Open*, **2016**, 5, 1595-1606	2.2	16
68	Small genomic rearrangements involving FMR1 support the importance of its gene dosage for normal neurocognitive function. *Neurogenetics*, **2012**, 13, 333-9	3	16
67	Fusion of large-scale genomic knowledge and frequency data computationally prioritizes variants in epilepsy. *PLoS Genetics*, **2013**, 9, e1003797	6	16
66	Recurrent partial rhombencephalosynapsis and holoprosencephaly in siblings with a mutation of ZIC2. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1574-80	2.5	16
65	Cornelia de Lange syndrome case due to genomic rearrangements including NIPBL. *European Journal of Medical Genetics*, **2010**, 53, 378-82	2.6	16
64	Small rare recurrent deletions and reciprocal duplications in 2q21.1, including brain-specific ARHGEF4 and GPR148. *Human Molecular Genetics*, **2012**, 21, 3345-55	5.6	16
63	Interchromosomal template-switching as a novel molecular mechanism for imprinting perturbations associated with Temple syndrome. *Genome Medicine*, **2019**, 11, 25	14.4	14
62	Prenatal Diagnosis of Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins. *Journal of Pediatrics*, **2016**, 170, 317-8	3.6	14
61	Molecular cytogenetic characterization of a familial der(1)del(1)(p36.33)dup(1)(p36.33p36.22) with variable phenotype. *American Journal of Medical Genetics, Part A*, **2005**, 139A, 136-40	2.5	14

60	CRISPR/Cas9-mediated deletion of lncRNA Gm26878 in the distant Foxf1 enhancer region. *Mammalian Genome*, **2017**, 28, 275-282	3.2	13
59	SOX12 and NRSN2 are candidate genes for 20p13 subtelomeric deletions associated with developmental delay. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2013**, 162B, 832-40	3.5	13
58	Variants in SKP1, PROB1, and IL17B genes at keratoconus 5q31.1-q35.3 susceptibility locus identified by whole-exome sequencing. *European Journal of Human Genetics*, **2017**, 25, 73-78	5.3	13
57	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, e427-32	3	13
56	Variable phenotypic presentation of a novel FOXF1 missense mutation in a single family. *Pediatric Pulmonology*, **2016**, 51, 921-7	3.5	13
55	Novel parent-of-origin-specific differentially methylated loci on chromosome 16. *Clinical Epigenetics*, **2019**, 11, 60	7.7	12
54	Parental somatic mosaicism for CNV deletions - A need for more sensitive and precise detection methods in clinical diagnostics settings. *Genomics*, **2020**, 112, 2937-2941	4.3	11
53	Molecular and clinical analyses of 16q24.1 duplications involving FOXF1 identify an evolutionarily unstable large minisatellite. *BMC Medical Genetics*, **2014**, 15, 128	2.1	11
52	Clinical improvement of the aggressive neurobehavioral phenotype in a patient with a deletion of PITX3 and the absence of L-DOPA in the cerebrospinal fluid. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2012**, 159B, 236-42	3.5	11
51	Autistic features with speech delay in a girl with an approximately 1.5-Mb deletion in 6q16.1, including GPR63 and FUT9. *Clinical Genetics*, **2009**, 75, 199-202	4	11
50	Early recurrence in standard-risk medulloblastoma patients with the common idic(17)(p11.2) rearrangement. *Neuro-Oncology*, **2012**, 14, 831-40	1	11
49	Complex balanced translocation t(1;5;7)(p32.1;q14.3;p21.3) and two microdeletions del(1)(p31.1p31.1) and del(7)(p14.1p14.1) in a patient with features of Greig cephalopolysyndactyly and mental retardation. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 2738-43	2.5	11
48	Mosaicism for r(X) and der(X)del(X)(p11.23)dup(X)(p11.21p11.22) provides insight into the possible mechanism of rearrangement. *Molecular Cytogenetics*, **2008**, 1, 16	2	11
47	De novo three-way chromosome translocation 46,XY,t(4;6;21)(p16;p21.1;q21) in a male with cleidocranial dysplasia. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 453-8	2.5	11
46	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	8.1	11
45	Copy number variants in patients with intellectual disability affect the regulation of ARX transcription factor gene. *Human Genetics*, **2015**, 134, 1163-82	6.3	9
44	Highly Sensitive Blocker Displacement Amplification and Droplet Digital PCR Reveal Low-Level Parental FOXF1 Somatic Mosaicism in Families with Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins. *Journal of Molecular Diagnostics*, **2020**, 22, 447-456	5.1	9
43	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, 681-8	5.8	9

42	Evidence for involvement of TRE-2 (USP6) oncogene, low-copy repeat and acrocentric heterochromatin in two families with chromosomal translocations. *Human Genetics*, **2006**, 120, 227-37	6.3	9
41	A girl with duplication 17p10-p12 associated with a dicentric chromosome. *American Journal of Medical Genetics Part A*, **2004**, 124A, 173-8		9
40	Long Non-Coding RNA : Gene Structure, Expression, and Biological Relevance. *Genes*, **2021**, 12,	4.2	9
39	SOX9 chromatin folding domains correlate with its real and putative distant cis-regulatory elements. *Nucleus*, **2017**, 8, 182-187	3.9	8
38	LINE- and Alu-containing genomic instability hotspot at 16q24.1 associated with recurrent and nonrecurrent CNV deletions causative for ACDMPV. *Human Mutation*, **2018**, 39, 1916-1925	4.7	8
37	Association of rare non-coding SNVs in the lung-specific FOXF1 enhancer with a mitigation of the lethal ACDMPV phenotype. *Human Genetics*, **2019**, 138, 1301-1311	6.3	8
36	A de novo 1.58 Mb deletion, including MAP2K6 and mapping 1.28 Mb upstream to SOX9, identified in a patient with Pierre Robin sequence and osteopenia with multiple fractures. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 1842-50	2.5	8
35	Head bobber: an insertional mutation causes inner ear defects, hyperactive circling, and deafness. *JARO - Journal of the Association for Research in Otolaryngology*, **2012**, 13, 335-49	3.3	7
34	Cryptic unbalanced translocation t(17;18)(p13.2;q22.3) identified by subtelomeric FISH and defined by array-based comparative genomic hybridization in a patient with mental retardation and dysmorphic features. *American Journal of Medical Genetics, Part A*, **2005**, 137, 88-93	2.5	7
33	Clinical genomics and contextualizing genome variation in the diagnostic laboratory. *Expert Review of Molecular Diagnostics*, **2020**, 20, 995-1002	3.8	7
32	Rare copy number variants contribute pathogenic alleles in patients with intestinal malrotation. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e549	2.3	7
31	A de novo 2.2 Mb recurrent 17q23.1q23.2 deletion unmasks novel putative regulatory non-coding SNVs associated with lethal lung hypoplasia and pulmonary hypertension: a case report. *BMC Medical Genomics*, **2020**, 13, 34	3.7	6
30	Intragenic deletions of the IGF1 receptor gene in five individuals with psychiatric phenotypes and developmental delay. *European Journal of Human Genetics*, **2013**, 21, 1304-7	5.3	6
29	Interphase FISH screening for the LCR-mediated common rearrangement of isochromosome 17q in primary myelofibrosis. *American Journal of Hematology*, **2005**, 79, 309-13	7.1	6
28	Heterozygous CTNNB1 and TBX4 variants in a patient with abnormal lung growth, pulmonary hypertension, microcephaly, and spasticity. *Clinical Genetics*, **2019**, 96, 366-370	4	5
27	Screening and familial characterization of copy-number variations in NR5A1 in 46,XY disorders of sex development and premature ovarian failure. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2487-94	2.5	5
26	CAV3 mutation in a patient with transient hyperCKemia and myalgia. *Neurologia I Neurochirurgia Polska*, **2016**, 50, 468-473	1	5
25	Disruption of normal patterns of FOXF1 expression in a lethal disorder of lung development. *Journal of Medical Genetics*, **2020**, 57, 296-300	5.8	4

24	Functional performance of aCGH design for clinical cytogenetics. *Computers in Biology and Medicine* , **2013**, 43, 775-85	7	4
23	Reply to Amor et al. *European Journal of Human Genetics*, **2012**, 20, 597-597	5.3	4
22	Lung-specific distant enhancer cis regulates expression of FOXF1 and lncRNA FENDRR. *Human Mutation*, **2021**, 42, 694-698	4.7	4
21	One pedigree we all may have come from - did Adam and Eve have the chromosome 2 fusion?. *Molecular Cytogenetics*, **2016**, 9, 72	2	4
20	Deciphering the complexity of simple chromosomal insertions by genome sequencing. *Human Genetics*, **2021**, 140, 361-380	6.3	4
19	Quantitative Assessment of Parental Somatic Mosaicism for Copy-Number Variant (CNV) Deletions. *Current Protocols in Human Genetics*, **2020**, 106, e99	3.2	3
18	Genotype-phenotype correlation in two Polish neonates with alveolar capillary dysplasia. *BMC Pediatrics*, **2020**, 20, 320	2.6	3
17	Alagille syndrome associated with a paracentric inversion 20p12.2p13 disrupting the JAG1 gene. *American Journal of Medical Genetics Part A*, **2001**, 103, 166-71		3
16	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies syndrome due to disruption of BPTF in a 35-year-old man initially diagnosed with Silver-Russell syndrome. *Clinical Genetics*, **2019**, 95, 534-536	4	3
15	Phenotypic expansion of the BPTF-related neurodevelopmental disorder with dysmorphic facies and distal limb anomalies. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1366-1378	2.5	3
14	Multiple samples aCGH analysis for rare CNVs detection. *Journal of Clinical Bioinformatics*, **2013**, 3, 12		2
13	Potential interactions between the TBX4-FGF10 and SHH-FOXF1 signaling during human lung development revealed using ChIP-seq. *Respiratory Research*, **2021**, 22, 26	7.3	2
12	Perturbation of semaphorin and VEGF signaling in ACDMPV lungs due to FOXF1 deficiency. *Respiratory Research*, **2021**, 22, 212	7.3	2
11	A recurrent 8 bp frameshifting indel in FOXF1 defines a novel mutation hotspot associated with alveolar capillary dysplasia with misalignment of pulmonary veins. *American Journal of Medical Genetics, Part A*, **2019**, 179, 2272-2276	2.5	1
10	Co-segregation of Freiberg‐Q infraction with a familial translocation t(5;7)(p13.3;p22.2) ascertained by a child with cri du chat syndrome and brachydactyly type A1B. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 445-9	2.5	1
9	A de novo deletion of CALN1 in a male with a bilateral diaphragmatic defect does not definitely cause this malformation. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1196-201	2.5	1
8	Complex genomic rearrangement of chromosome 16p13.3 detected by array comparative genomic hybridization in a patient with multiple congenital anomalies, dysmorphic craniofacial features, and developmental delay. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 2589-92	2.5	1
7	Variants in FLRT3 and SLC35E2B identified using exome sequencing in seven high myopia families from Central Europe. *Advances in Medical Sciences*, **2021**, 66, 192-198	2.8	1

6	Do paternal deletions involving the FOXF1 locus on chromosome 16q24.1 manifest with more severe non-lung anomalies?. *European Journal of Medical Genetics*, **2022**, 65, 104519	2.6	1
5	Transcriptome and Immunohistochemical Analyses in TBX4- and FGF10-Deficient Lungs Imply TMEM100 as a Mediator of Human Lung Development. *American Journal of Respiratory Cell and Molecular Biology*, **2022**, 66, 694-697	5.7	1
4	Haploinsufficiency of the Sin3/HDAC corepressor complex member SIN3B causes a syndromic intellectual disability/autism spectrum disorder. *American Journal of Human Genetics*, **2021**, 108, 929-941	11	0
3	Detection of low-level parental somatic mosaicism for clinically relevant SNVs and indels identified in a large exome sequencing dataset.. *Human Genomics*, **2021**, 15, 72	6.8	0
2	Gene, Genomic, and Chromosomal Disorders **2012**, 187-195		
1	Karyotyping as the first genomic approach **2021**, 17-34		