Jing Zhang

List of Publications by Year in Descending Order

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	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
220	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
219	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
218	Lung-specific distant enhancer cis regulates expression of FOXF1 and lncRNA FENDRR. *Human Mutation*, **2021**, 42, 694-698	4.7	4
217	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
216	Deciphering the complexity of simple chromosomal insertions by genome sequencing. *Human Genetics*, **2021**, 140, 361-380	6.3	4
215	Long Non-Coding RNA : Gene Structure, Expression, and Biological Relevance. *Genes*, **2021**, 12,	4.2	9
214	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
213	Karyotyping as the first genomic approach **2021**, 17-34		
212	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
211	Perturbation of semaphorin and VEGF signaling in ACDMPV lungs due to FOXF1 deficiency. *Respiratory Research*, **2021**, 22, 212	7.3	2
210	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
209	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
208	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
207	Quantitative Assessment of Parental Somatic Mosaicism for Copy-Number Variant (CNV) Deletions. *Current Protocols in Human Genetics*, **2020**, 106, e99	3.2	3
206	Genotype-phenotype correlation in two Polish neonates with alveolar capillary dysplasia. *BMC Pediatrics*, **2020**, 20, 320	2.6	3
205	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

204	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
203	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
202	Clinical genomics and contextualizing genome variation in the diagnostic laboratory. *Expert Review of Molecular Diagnostics*, **2020**, 20, 995-1002	3.8	7
201	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
200	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
199	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
198	Interchromosomal template-switching as a novel molecular mechanism for imprinting perturbations associated with Temple syndrome. *Genome Medicine*, **2019**, 11, 25	14.4	14
197	Diagnostic implications of genetic copy number variation in epilepsy plus. *Epilepsia*, **2019**, 60, 689-706	6.4	37
196	Novel parent-of-origin-specific differentially methylated loci on chromosome 16. *Clinical Epigenetics*, **2019**, 11, 60	7.7	12
195	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
194	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
193	A clinical survey of mosaic single nucleotide variants in disease-causing genes detected by exome sequencing. *Genome Medicine*, **2019**, 11, 48	14.4	29
192	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. *Nature Communications*, **2019**, 10, 4679	17.4	21
191	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
190	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
189	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
188	Rare copy number variants contribute pathogenic alleles in patients with intestinal malrotation. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e549	2.3	7
187	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

186	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
185	An estimation of the prevalence of genomic disorders using chromosomal microarray data. *Journal of Human Genetics*, **2018**, 63, 795-801	4.3	22
184	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
183	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
182	Predicting human genes susceptible to genomic instability associated with /-mediated rearrangements. *Genome Research*, **2018**, 28, 1228-1242	9.7	44
181	SOX9 chromatin folding domains correlate with its real and putative distant cis-regulatory elements. *Nucleus*, **2017**, 8, 182-187	3.9	8
180	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
179	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. *Cell*, **2017**, 168, 830-842.e7	36.2	53
178	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
177	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
176	CRISPR/Cas9-mediated deletion of lncRNA Gm26878 in the distant Foxf1 enhancer region. *Mammalian Genome*, **2017**, 28, 275-282	3.2	13
175	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
174	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
173	Identification of novel candidate disease genes from de novo exonic copy number variants. *Genome Medicine*, **2017**, 9, 83	14.4	38
172	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
171	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
170	Narrowing the distant enhancer region on 16q24.1 critical for ACDMPV. *Clinical Epigenetics*, **2016**, 8, 112	7.7	17
169	Lethal lung hypoplasia and vascular defects in mice with conditional Foxf1 overexpression. *Biology Open*, **2016**, 5, 1595-1606	2.2	16

168	Prenatal Diagnosis of Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins. *Journal of Pediatrics*, **2016**, 170, 317-8	3.6	14
167	The complex behavioral phenotype of 15q13.3 microdeletion syndrome. *Genetics in Medicine*, **2016**, 18, 1111-1118	8.1	32
166	Mechanisms for Complex Chromosomal Insertions. *PLoS Genetics*, **2016**, 12, e1006446	6	31
165	Evidence against ZNF469 being causative for keratoconus in Polish patients. *Acta Ophthalmologica*, **2016**, 94, 289-94	3.7	17
164	Variable phenotypic presentation of a novel FOXF1 missense mutation in a single family. *Pediatric Pulmonology*, **2016**, 51, 921-7	3.5	13
163	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
162	Pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins. *Human Genetics*, **2016**, 135, 569-586	6.3	62
161	CAV3 mutation in a patient with transient hyperCKemia and myalgia. *Neurologia I Neurochirurgia Polska*, **2016**, 50, 468-473	1	5
160	One pedigree we all may have come from - did Adam and Eve have the chromosome 2 fusion?. *Molecular Cytogenetics*, **2016**, 9, 72	2	4
159	Somatic mosaicism: implications for disease and transmission genetics. *Trends in Genetics*, **2015**, 31, 382-92	8.5	182
158	Neurodevelopmental and neurobehavioral characteristics in males and females with CDKL5 duplications. *European Journal of Human Genetics*, **2015**, 23, 915-21	5.3	27
157	Absence of heterozygosity due to template switching during replicative rearrangements. *American Journal of Human Genetics*, **2015**, 96, 555-64	11	39
156	Assessing structural variation in a personal genome-towards a human reference diploid genome. *BMC Genomics*, **2015**, 16, 286	4.5	117
155	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
154	6q22.1 microdeletion and susceptibility to pediatric epilepsy. *European Journal of Human Genetics*, **2015**, 23, 173-9	5.3	25
153	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
152	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
151	Co-segregation of Freiberg© 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

150	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
149	Increased STAG2 dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. *Human Molecular Genetics*, **2015**, 24, 7171-81	5.6	24
148	Genome-wide analyses of LINE-LINE-mediated nonallelic homologous recombination. *Nucleic Acids Research*, **2015**, 43, 2188-98	20.1	52
147	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
146	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
145	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
144	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
143	Parental somatic mosaicism is underrecognized and influences recurrence risk of genomic disorders. *American Journal of Human Genetics*, **2014**, 95, 173-82	11	172
142	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
141	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
140	Human endogenous retroviral elements promote genome instability via non-allelic homologous recombination. *BMC Biology*, **2014**, 12, 74	7.3	46
139	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
138	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
137	Molecular findings among patients referred for clinical whole-exome sequencing. *JAMA - Journal of the American Medical Association*, **2014**, 312, 1870-9	27.4	915
136	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
135	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
134	Multiple samples aCGH analysis for rare CNVs detection. *Journal of Clinical Bioinformatics*, **2013**, 3, 12		2
133	Functional performance of aCGH design for clinical cytogenetics. *Computers in Biology and Medicine* , **2013**, 43, 775-85	7	4

132	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
131	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
130	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
129	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
128	Rare DNA copy number variants in cardiovascular malformations with extracardiac abnormalities. *European Journal of Human Genetics*, **2013**, 21, 173-81	5.3	42
127	Inverted low-copy repeats and genome instability--a genome-wide analysis. *Human Mutation*, **2013**, 34, 210-20	4.7	41
126	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
125	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
124	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
123	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
122	Chromosome conformation capture-on-chip analysis of long-range cis-interactions of the SOX9 promoter. *Chromosome Research*, **2013**, 21, 781-8	4.4	19
121	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
120	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
119	Fusion of large-scale genomic knowledge and frequency data computationally prioritizes variants in epilepsy. *PLoS Genetics*, **2013**, 9, e1003797	6	16
118	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
117	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
116	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
115	Incidental copy-number variants identified by routine genome testing in a clinical population. *Genetics in Medicine*, **2013**, 15, 45-54	8.1	32

114	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
113	Deletions of recessive disease genes: CNV contribution to carrier states and disease-causing alleles. *Genome Research*, **2013**, 23, 1383-94	9.7	51
112	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
111	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
110	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
109	Small genomic rearrangements involving FMR1 support the importance of its gene dosage for normal neurocognitive function. *Neurogenetics*, **2012**, 13, 333-9	3	16
108	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
107	Detection of 1Mb microdeletions and microduplications in a single cell using custom oligonucleotide arrays. *Prenatal Diagnosis*, **2012**, 32, 10-20	3.2	22
106	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
105	Phenotypic spectrum and genotype-phenotype correlations of NRXN1 exon deletions. *European Journal of Human Genetics*, **2012**, 20, 1240-7	5.3	82
104	Microdeletion and microduplication syndromes. *Methods in Molecular Biology*, **2012**, 838, 29-75	1.4	47
103	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
102	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
101	Early recurrence in standard-risk medulloblastoma patients with the common idic(17)(p11.2) rearrangement. *Neuro-Oncology*, **2012**, 14, 831-40	1	11
100	Reply to Amor et al. *European Journal of Human Genetics*, **2012**, 20, 597-597	5.3	4
99	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
98	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
97	Gene, Genomic, and Chromosomal Disorders **2012**, 187-195		

96	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
95	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
94	Chromosome catastrophes involve replication mechanisms generating complex genomic rearrangements. *Cell*, **2011**, 146, 889-903	56.2	315
93	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
92	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
91	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
90	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
89	Phenotypic manifestations of copy number variation in chromosome 16p13.11. *European Journal of Human Genetics*, **2011**, 19, 280-6	5.3	82
88	The phenotype of recurrent 10q22q23 deletions and duplications. *European Journal of Human Genetics*, **2011**, 19, 400-8	5.3	52
87	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
86	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
85	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
84	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
83	Observation and prediction of recurrent human translocations mediated by NAHR between nonhomologous chromosomes. *Genome Research*, **2011**, 21, 33-46	9.7	61
82	Alveolar capillary dysplasia. *American Journal of Respiratory and Critical Care Medicine*, **2011**, 184, 172-9	10.2	147
81	Copy number gain at Xp22.31 includes complex duplication rearrangements and recurrent triplications. *Human Molecular Genetics*, **2011**, 20, 1975-88	5.6	61
80	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
79	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

78	Clinical spectrum associated with recurrent genomic rearrangements in chromosome 17q12. *European Journal of Human Genetics*, **2010**, 18, 278-84	5.3	90
77	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
76	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
75	Cornelia de Lange syndrome case due to genomic rearrangements including NIPBL. *European Journal of Medical Genetics*, **2010**, 53, 378-82	2.6	16
74	Whole-genome sequencing in a patient with Charcot-Marie-Tooth neuropathy. *New England Journal of Medicine*, **2010**, 362, 1181-91	59.2	613
73	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
72	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
71	Structural variation in the human genome and its role in disease. *Annual Review of Medicine*, **2010**, 61, 437-55	17.4	827
70	Structures and molecular mechanisms for common 15q13.3 microduplications involving CHRNA7: benign or pathological?. *Human Mutation*, **2010**, 31, 840-50	4.7	101
69	Detection of clinically relevant exonic copy-number changes by array CGH. *Human Mutation*, **2010**, 31, 1326-42	4.7	195
68	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
67	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
66	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
65	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
64	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
63	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
62	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
61	Redefined genomic architecture in 15q24 directed by patient deletion/duplication breakpoint mapping. *Human Genetics*, **2009**, 126, 589-602	6.3	63

60	Alu-specific microhomology-mediated deletions in CDKL5 in females with early-onset seizure disorder. *Neurogenetics*, **2009**, 10, 363-9	3	39
59	PTCH1 duplication in a family with microcephaly and mild developmental delay. *European Journal of Human Genetics*, **2009**, 17, 267-71	5.3	27
58	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
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