Jing Zhang

List of Publications by Year
in descending order

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227 papers	18,050 citations	20036 63 h-index	18400 124 g-index
233 all docs	233 docs citations	233 times ranked	23640 citing authors

#	Article	IF	Citations
1	Exacerbation of mild lung disorders to lethal pulmonary hypoplasia by a noncoding hypomorphic <scp>SNV</scp> in a lungâ€specific enhancer in trans to the frameshifting <scp>TBX4</scp> variant. American Journal of Medical Genetics, Part A, 2022, 188, 1420-1425.	0.7	7
2	Parental mosaicism for apparent de novo genetic variants: Scope, detection, and counseling challenges. Prenatal Diagnosis, 2022, 42, 811-821.	1.1	8
3	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.	0.7	3
4	Ultra-conserved non-coding sequences within the FOXF1 enhancer are critical for human lung development. Genes and Diseases, 2022, 9, 1423-1426.	1.5	4
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.	1.4	6
6	Deciphering the complexity of simple chromosomal insertions by genome sequencing. Human Genetics, 2021, 140, 361-380.	1.8	15
7	Long Non-Coding RNA FENDRR: Gene Structure, Expression, and Biological Relevance. Genes, 2021, 12, 177.	1.0	13
8	Phenotypic expansion of the <scp>BPTF</scp>â€related neurodevelopmental disorder with dysmorphic facies and distal limb anomalies. American Journal of Medical Genetics, Part A, 2021, 185, 1366-1378.	0.7	8
9	Karyotyping as the first genomic approach. , 2021, , 17-34.		3
10	Potential interactions between the TBX4-FGF10 and SHH-FOXF1 signaling during human lung development revealed using ChIP-seq. Respiratory Research, 2021, 22, 26.	1.4	11
11	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.	0.9	5
12	Lungâ€specific distant enhancer cis regulates expression ofFOXF1and lncRNAFENDRR. Human Mutation, 2021, 42, 694-698.	1.1	10
13	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.	2.6	15
14	Perturbation of semaphorin and VEGF signaling in ACDMPV lungs due to FOXF1 deficiency. Respiratory Research, 2021, 22, 212.	1.4	11
15	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.	1.4	11
16	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
17	Clinical genomics and contextualizing genome variation in the diagnostic laboratory. Expert Review of Molecular Diagnostics, 2020, 20, 995-1002.	1.5	14
18	Parental somatic mosaicism for CNV deletions â€“ A need for more sensitive and precise detection methods in clinical diagnostics settings. Genomics, 2020, 112, 2937-2941.	1.3	14

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19	A de novo 2.2â€‰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.	0.7	12
20	Quantitative Assessment of Parental Somatic Mosaicism for Copyâ€Number Variant (CNV) Deletions. Current Protocols in Human Genetics, 2020, 106, e99.	3.5	7
21	Genotypeâ€“phenotype correlation in two Polish neonates with alveolar capillary dysplasia. BMC Pediatrics, 2020, 20, 320.	0.7	7
22	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.	1.2	13
23	Disruption of normal patterns of FOXF1 expression in a lethal disorder of lung development. Journal of Medical Genetics, 2020, 57, 296-300.	1.5	7
24	Heterozygous CTNNB1 and TBX4 variants in a patient with abnormal lung growth, pulmonary hypertension, microcephaly, and spasticity. Clinical Genetics, 2019, 96, 366-370.	1.0	14
25	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
26	A clinical survey of mosaic single nucleotide variants in disease-causing genes detected by exome sequencing. Genome Medicine, 2019, 11, 48.	3.6	55
27	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	5.8	43
28	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.	1.8	13
29	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.	0.7	2
30	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
31	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
32	Interchromosomal template-switching as a novel molecular mechanism for imprinting perturbations associated with Temple syndrome. Genome Medicine, 2019, 11, 25.	3.6	22
33	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	2.6	61
34	Novel parent-of-origin-specific differentially methylated loci on chromosome 16. Clinical Epigenetics, 2019, 11, 60.	1.8	18
35	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
36	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies syndrome due to disruption of BPTF in a 35â€yearâ€old man initially diagnosed with Silverâ€Russell syndrome. Clinical Genetics, 2019, 95, 534-536.	1.0	4

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37	Rare copy number variants contribute pathogenic alleles in patients with intestinal malrotation. Molecular Genetics & Genomic Medicine, 2019, 7, e549.	0.6	12
38	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
39	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
40	An estimation of the prevalence of genomic disorders using chromosomal microarray data. Journal of Human Genetics, 2018, 63, 795-801.	1.1	49
41	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
42	LINE- andAlu-containing genomic instability hotspot at 16q24.1 associated with recurrent and nonrecurrent CNV deletions causative for ACDMPV. Human Mutation, 2018, 39, 1916-1925.	1.1	14
43	Predicting human genes susceptible to genomic instability associated with Alu/Alu-mediated rearrangements. Genome Research, 2018, 28, 1228-1242.	2.4	74
44	SOX9 chromatin folding domains correlate with its real and putative distant cis-regulatory elements. Nucleus, 2017, 8, 182-187.	0.6	10
45	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
46	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. Cell, 2017, 168, 830-842.e7.	13.5	66
47	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
48	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
49	CRISPR/Cas9-mediated deletion of lncRNA Gm26878 in the distant Foxf1 enhancer region. Mammalian Genome, 2017, 28, 275-282.	1.0	14
50	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	3.3	348
51	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
52	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
53	Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83.	3.6	50
54	Mechanisms for Complex Chromosomal Insertions. PLoS Genetics, 2016, 12, e1006446.	1.5	45

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55	Evidence against <scp>ZNF</scp>469 being causative for keratoconus in Polish patients. Acta Ophthalmologica, 2016, 94, 289-294.	0.6	20
56	Variable phenotypic presentation of a novelFOXF1missense mutation in a single family. Pediatric Pulmonology, 2016, 51, 921-927.	1.0	16
57	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
58	Pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins. Human Genetics, 2016, 135, 569-586.	1.8	85
59	CAV3 mutation in a patient with transient hyperCKemia and myalgia. Neurologia I Neurochirurgia Polska, 2016, 50, 468-473.	0.6	8
60	One pedigree we all may have come from â€“ did Adam and Eve have the chromosome 2 fusion?. Molecular Cytogenetics, 2016, 9, 72.	0.4	6
61	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
62	Narrowing the FOXF1 distant enhancer region on 16q24.1 critical for ACDMPV. Clinical Epigenetics, 2016, 8, 112.	1.8	19
63	Lethal lung hypoplasia and vascular defects in mice with conditionalFoxf1overexpression. Biology Open, 2016, 5, 1595-1606.	0.6	20
64	Prenatal Diagnosis of Alveolar Capillary Dysplasia with Misalignment ofÂPulmonary Veins. Journal of Pediatrics, 2016, 170, 317-318.	0.9	17
65	The complex behavioral phenotype of 15q13.3 microdeletion syndrome. Genetics in Medicine, 2016, 18, 1111-1118.	1.1	45
66	A de novo 1.58â€‰Mb deletion, including MAP2K6 and mapping 1.28â€‰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, 167, 1842-1850.	0.7	9
67	Coâ€segregation of Freiberg's 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, 167, 445-449.	0.7	1
68	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
69	IncreasedSTAG2dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. Human Molecular Genetics, 2015, 24, 7171-7181.	1.4	28
70	Genome-wide analyses of LINEâ€“LINE-mediated nonallelic homologous recombination. Nucleic Acids Research, 2015, 43, 2188-2198.	6.5	79
71	Somatic mosaicism: implications for disease and transmission genetics. Trends in Genetics, 2015, 31, 382-392.	2.9	234
72	Neurodevelopmental and neurobehavioral characteristics in males and females with CDKL5 duplications. European Journal of Human Genetics, 2015, 23, 915-921.	1.4	32

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73	Absence of Heterozygosity Due to Template Switching during Replicative Rearrangements. American Journal of Human Genetics, 2015, 96, 555-564.	2.6	45
74	Assessing structural variation in a personal genomeâ€"towards a human reference diploid genome. BMC Genomics, 2015, 16, 286.	1.2	153
75	Copy number variants in patients with intellectual disability affect the regulation of ARX transcription factor gene. Human Genetics, 2015, 134, 1163-1182.	1.8	14
76	6q22.1 microdeletion and susceptibility to pediatric epilepsy. European Journal of Human Genetics, 2015, 23, 173-179.	1.4	35
77	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
78	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
79	Human endogenous retroviral elements promote genome instability via non-allelic homologous recombination. BMC Biology, 2014, 12, 74.	1.7	60
80	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
81	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
82	Molecular Findings Among Patients Referred for Clinical Whole-Exome Sequencing. JAMA - Journal of the American Medical Association, 2014, 312, 1870.	3.8	1,171
83	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
84	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
85	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
86	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
87	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. American Journal of Human Genetics, 2014, 95, 173-182.	2.6	219
88	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
89	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
90	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

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91	Multiple samples aCGH analysis for rare CNVs detection. Journal of Clinical Bioinformatics, 2013, 3, 12.	1.2	2
92	Functional performance of aCGH design for clinical cytogenetics. Computers in Biology and Medicine, 2013, 43, 775-785.	3.9	4
93	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
94	SOX12 and NRSN2 are candidate genes for 20p13 subtelomeric deletions associated with developmental delay. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2013, 162, 832-840.	1.1	15
95	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
96	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
97	Rare DNA copy number variants in cardiovascular malformations with extracardiac abnormalities. European Journal of Human Genetics, 2013, 21, 173-181.	1.4	49
98	Inverted Low-Copy Repeats and Genome Instability-A Genome-Wide Analysis. Human Mutation, 2013, 34, 210-220.	1.1	48
99	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
100	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
101	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
102	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
103	Chromosome conformation capture-on-chip analysis of long-range cis-interactions of the SOX9 promoter. Chromosome Research, 2013, 21, 781-788.	1.0	23
104	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
105	Intragenic deletions of the IGF1 receptor gene in five individuals with psychiatric phenotypes and developmental delay. European Journal of Human Genetics, 2013, 21, 1304-1307.	1.4	8
106	Fusion of Large-Scale Genomic Knowledge and Frequency Data Computationally Prioritizes Variants in Epilepsy. PLoS Genetics, 2013, 9, e1003797.	1.5	22
107	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
108	Screening and familial characterization of copyâ€number variations in NR5A1 in 46,XY disorders of sex development and premature ovarian failure. American Journal of Medical Genetics, Part A, 2013, 161, 2487-2494.	0.7	12

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109	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
110	Incidental copy-number variants identified by routine genome testing in a clinical population. Genetics in Medicine, 2013, 15, 45-54.	1.1	37
111	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
112	Deletions of recessive disease genes: CNV contribution to carrier states and disease-causing alleles. Genome Research, 2013, 23, 1383-1394.	2.4	62
113	Early recurrence in standard-risk medulloblastoma patients with the common idic(17)(p11.2) rearrangement. Neuro-Oncology, 2012, 14, 831-840.	0.6	13
114	Reply to Amor et al. European Journal of Human Genetics, 2012, 20, 597-597.	1.4	4
115	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
116	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
117	Gene, Genomic, and Chromosomal Disorders. , 2012, , 187-195.		0
118	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
119	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, 681-688.	1.5	11
120	Small genomic rearrangements involving FMR1 support the importance of its gene dosage for normal neurocognitive function. Neurogenetics, 2012, 13, 333-339.	0.7	21
121	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
122	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
123	Clinical improvement of the aggressive neurobehavioral phenotype in a patient with a deletion of PITX3 and the absence of <scp>L</scp>â€DOPA in the cerebrospinal fluid. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 236-242.	1.1	14
124	Phenotypic spectrum and genotypeâ€“phenotype correlations of NRXN1 exon deletions. European Journal of Human Genetics, 2012, 20, 1240-1247.	1.4	99
125	Microdeletion and Microduplication Syndromes. Methods in Molecular Biology, 2012, 838, 29-75.	0.4	58
126	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

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127	Head Bobber: An Insertional Mutation Causes Inner Ear Defects, Hyperactive Circling, and Deafness. JARO - Journal of the Association for Research in Otolaryngology, 2012, 13, 335-349.	0.9	8
128	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
129	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
130	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
131	Chromosome Catastrophes Involve Replication Mechanisms Generating Complex Genomic Rearrangements. Cell, 2011, 146, 889-903.	13.5	391
132	Efficient Multiple Samples aCGH Analysis for Rare CNVs Detection. , 2011, , .		0
133	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
134	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
135	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
136	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
137	Phenotypic manifestations of copy number variation in chromosome 16p13.11. European Journal of Human Genetics, 2011, 19, 280-286.	1.4	97
138	The phenotype of recurrent 10q22q23 deletions and duplications. European Journal of Human Genetics, 2011, 19, 400-408.	1.4	63
139	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, 155, 1196-1201.	0.7	1
140	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
141	Recurrent partial rhombencephalosynapsis and holoprosencephaly in siblings with a mutation of ZIC2. American Journal of Medical Genetics, Part A, 2011, 155, 1574-1580.	0.7	17
142	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, 155, 2589-2592.	0.7	1
143	Observation and prediction of recurrent human translocations mediated by NAHR between nonhomologous chromosomes. Genome Research, 2011, 21, 33-46.	2.4	72
144	Alveolar Capillary Dysplasia. American Journal of Respiratory and Critical Care Medicine, 2011, 184, 172-179.	2.5	194

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145	Copy number gain at Xp22.31 includes complex duplication rearrangements and recurrent triplications. Human Molecular Genetics, 2011, 20, 1975-1988.	1.4	74
146	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
147	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
148	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
149	Structural Variation in the Human Genome and its Role in Disease. Annual Review of Medicine, 2010, 61, 437-455.	5.0	1,015
150	Structures and molecular mechanisms for common 15q13.3 microduplications involving CHRNA7: benign or pathological?. Human Mutation, 2010, 31, 840-850.	1.1	111
151	Detection of clinically relevant exonic copy-number changes by array CGH. Human Mutation, 2010, 31, 1326-1342.	1.1	225
152	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
153	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
154	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
155	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
156	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
157	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
158	Clinical spectrum associated with recurrent genomic rearrangements in chromosome 17q12. European Journal of Human Genetics, 2010, 18, 278-284.	1.4	114
159	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
160	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
161	Cornelia de Lange syndrome case due to genomic rearrangements including NIPBL. European Journal of Medical Genetics, 2010, 53, 378-382.	0.7	18
162	Whole-Genome Sequencing in a Patient with Charcotâ€“Marieâ€“Tooth Neuropathy. New England Journal of Medicine, 2010, 362, 1181-1191.	13.9	698

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