Xiaoli Chen

List of Publications by Year
in descending order

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37 papers	1,479 citations	516710 16 h-index	395702 33 g-index
40 all docs	40 docs citations	40 times ranked	3553 citing authors

#	Article	IF	Citations
1	Genomic Dissection of Population Substructure of Han Chinese and Its Implication in Association Studies. American Journal of Human Genetics, 2009, 85, 762-774.	6.2	338
2	TBX6 Null Variants and a Common Hypomorphic Allele in Congenital Scoliosis. New England Journal of Medicine, 2015, 372, 341-350.	27.0	239
3	Deletions of NRXN1 (neurexinâ€1) predispose to a wide spectrum of developmental disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 937-947.	1.7	217
4	Engineering microdeletions and microduplications by targeting segmental duplications with CRISPR. Nature Neuroscience, 2016, 19, 517-522.	14.8	72
5	Antidiabetic effect of flavones from Cirsium japonicum DC in diabetic rats. Archives of Pharmacal Research, 2010, 33, 353-362.	6.3	65
6	Disruption of TCF4 regulatory networks leads to abnormal cortical development and mental disabilities. Molecular Psychiatry, 2019, 24, 1235-1246.	7.9	63
7	TBX6-associated congenital scoliosis (TACS) as a clinically distinguishable subtype of congenital scoliosis: further evidence supporting the compound inheritance and TBX6 gene dosage model. Genetics in Medicine, 2019, 21, 1548-1558.	2.4	60
8	Global DNA hypomethylation is associated with NTDâ€affected pregnancy: A caseâ€control study. Birth Defects Research Part A: Clinical and Molecular Teratology, 2010, 88, 575-581.	1.6	51
9	Molecular Analysis of a Deletion Hotspot in the NRXN1 Region Reveals the Involvement of Short Inverted Repeats in Deletion CNVs. American Journal of Human Genetics, 2013, 92, 375-386.	6.2	42
10	Intra-family phenotypic heterogeneity of 16p11.2 deletion carriers in a three-generation Chinese family. , 2011, 156, 225-232.		38
11	Rare DeleteriousPARD3Variants in the aPKC-Binding Region are Implicated in the Pathogenesis of Human Cranial Neural Tube Defects Via Disrupting Apical Tight Junction Formation. Human Mutation, 2017, 38, 378-389.	2.5	29
12	Detection of Copy Number Variants Reveals Association of Cilia Genes with Neural Tube Defects. PLoS ONE, 2013, 8, e54492.	2.5	27
13	RET somatic mutations are underrecognized in Hirschsprung disease. Genetics in Medicine, 2018, 20, 770-777.	2.4	24
14	Increased genome instability in human DNA segments with self-chains: homology-induced structural variations via replicative mechanisms. Human Molecular Genetics, 2013, 22, 2642-2651.	2.9	22
15	Genome-wide copy number variant analysis for congenital ventricular septal defects in Chinese Han population. BMC Medical Genomics, 2015, 9, 2.	1.5	20
16	Altered Methylation of the DNA Repair Gene MGMT Is Associated with Neural Tube Defects. Journal of Molecular Neuroscience, 2012, 47, 42-51.	2.3	18
17	Further defining the critical genes for the 4q21 microdeletion disorder. American Journal of Medical Genetics, Part A, 2017, 173, 120-125.	1.2	17
18	CNV profiles of Chinese pediatric patients with developmental disorders. Genetics in Medicine, 2021, 23, 669-678.	2.4	17

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19	Comparative analysis of serum proteome in congenital scoliosis patients with <scp>TBX</scp>6 haploinsufficiency â€“ a first report pointing to lipid metabolism. Journal of Cellular and Molecular Medicine, 2018, 22, 533-545.	3.6	16
20	Fragile X syndrome screening in Chinese children with unknown intellectual developmental disorder. BMC Pediatrics, 2015, 15, 77.	1.7	11
21	Identification of two novel null variants in CLN8 by targeted next-generation sequencing: first report of a Chinese patient with neuronal ceroid lipofuscinosis due to CLN8 variants. BMC Medical Genetics, 2018, 19, 21.	2.1	11
22	Recurrent 8q13.2-13.3 microdeletions associated with Branchio-oto-renal syndrome are mediated by human endogenous retroviral (HERV) sequence blocks. BMC Medical Genetics, 2014, 15, 90.	2.1	10
23	Clinical and molecular genetic characterization of familial MECP2 duplication syndrome in a Chinese family. BMC Medical Genetics, 2017, 18, 131.	2.1	10
24	The Effect of Plasma Lead on Anembryonic Pregnancy. Annals of the New York Academy of Sciences, 2008, 1140, 184-189.	3.8	9
25	Association Study of PARD3 Gene Polymorphisms With Neural Tube Defects in a Chinese Han Population. Reproductive Sciences, 2012, 19, 764-771.	2.5	8
26	PCMT1 gene polymorphisms, maternal folate metabolism, and neural tube defects: a caseâ€“control study in a population with relatively low folate intake. Genes and Nutrition, 2013, 8, 581-587.	2.5	8
27	Molecular cloning and functional analysis of Chinese sturgeon (Acipenser sinensis) growth hormone receptor. Science in China Series C: Life Sciences, 2009, 52, 911-921.	1.3	7
28	A novel approach for copy number variation analysis by combining multiplex PCR with matrix-assisted laser desorption ionization time-of-flight mass spectrometry. Journal of Biotechnology, 2013, 166, 6-11.	3.8	7
29	A complex intragenic rearrangement of ERCC8 in Chinese siblings with Cockayne syndrome. Scientific Reports, 2017, 7, 44271.	3.3	7
30	Confirming the contribution and genetic spectrum of de novo mutation in infantile spasms: Evidence from a Chinese cohort. Molecular Genetics & Genomic Medicine, 2021, 9, e1689.	1.2	6
31	Annular pancreas in Trichorhinophalangeal syndrome type II with 8q23.3-q24.12 interstitial deletion. Molecular Cytogenetics, 2015, 8, 95.	0.9	4
32	Neurodevelopmental trajectory and modifiers of 16p11.2 microdeletion: A followâ€up study of four Chinese children carriers. Molecular Genetics & Genomic Medicine, 2020, 8, e1485.	1.2	3
33	A familial Sonic Hedgehog (SHH) stop-gain mutation associated with agenesis of the corpus callosum, mild intellectual disability and facial dysmorphism. Brain and Development, 2020, 42, 771-774.	1.1	2
34	A rare case of acquired immunodeficiency associated with myelodysplastic syndrome. Molecular Genetics & Genomic Medicine, 2019, 7, e923.	1.2	1
35	Novel function of RECS1 as a negative regulator of TNF-Î±-induced NF-ÎºB activation. Molecular and Cellular Biochemistry, 2010, 337, 317-317.	3.1	0
36	Response to Brosens et al. Genetics in Medicine, 2018, 20, 1479-1480.	2.4	0

#	Article	IF	Citations
37	F74EXPLORING THE PATHOGENICITY OF DE NOVO VARIANTS TO EARLY-ONSET NEURO-DEVELOPMENTAL DISORDERS IN CHILDREN. European Neuropsychopharmacology, 2019, 29, S1150.	0.7	0