

# 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	Confirming the contribution and genetic spectrum of de novo mutation in infantile spasms: Evidence from a Chinese cohort. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2021, 9, e1689.	1.2	6
2	CNV profiles of Chinese pediatric patients with developmental disorders. <i>Genetics in Medicine</i> , 2021, 23, 669-678.	2.4	17
3	A familial Sonic Hedgehog (SHH) stop-gain mutation associated with agenesis of the corpus callosum, mild intellectual disability and facial dysmorphism. <i>Brain and Development</i> , 2020, 42, 771-774.	1.1	2
4	Neurodevelopmental trajectory and modifiers of 16p11.2 microdeletion: A follow-up study of four Chinese children carriers. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1485.	1.2	3
5	A rare case of acquired immunodeficiency associated with myelodysplastic syndrome. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2019, 7, e923.	1.2	1
6	F74EXPLORING THE PATHOGENICITY OF DE NOVO VARIANTS TO EARLY-ONSET NEURO-DEVELOPMENTAL DISORDERS IN CHILDREN. <i>European Neuropsychopharmacology</i> , 2019, 29, S1150.	0.7	0
7	Disruption of TCF4 regulatory networks leads to abnormal cortical development and mental disabilities. <i>Molecular Psychiatry</i> , 2019, 24, 1235-1246.	7.9	63
8	TBX6-associated congenital scoliosis (TACS) as a clinically distinguishable subtype of congenital scoliosis: further evidence supporting the compound inheritance and TBX6 gene dosage model. <i>Genetics in Medicine</i> , 2019, 21, 1548-1558.	2.4	60
9	Response to Brosens et al. <i>Genetics in Medicine</i> , 2018, 20, 1479-1480.	2.4	0
10	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. <i>BMC Medical Genetics</i> , 2018, 19, 21.	2.1	11
11	Comparative analysis of serum proteome in congenital scoliosis patients with <i>homozygous TBX6</i> haploinsufficiency – a first report pointing to lipid metabolism. <i>Journal of Cellular and Molecular Medicine</i> , 2018, 22, 533-545.	3.6	16
12	RET somatic mutations are underrecognized in Hirschsprung disease. <i>Genetics in Medicine</i> , 2018, 20, 770-777.	2.4	24
13	A complex intragenic rearrangement of ERCC8 in Chinese siblings with Cockayne syndrome. <i>Scientific Reports</i> , 2017, 7, 44271.	3.3	7
14	Rare Deleterious <i>PARD3</i> Variants in the aPKC-Binding Region are Implicated in the Pathogenesis of Human Cranial Neural Tube Defects Via Disrupting Apical Tight Junction Formation. <i>Human Mutation</i> , 2017, 38, 378-389.	2.5	29
15	Further defining the critical genes for the 4q21 microdeletion disorder. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 120-125.	1.2	17
16	Clinical and molecular genetic characterization of familial MECP2 duplication syndrome in a Chinese family. <i>BMC Medical Genetics</i> , 2017, 18, 131.	2.1	10
17	Engineering microdeletions and microduplications by targeting segmental duplications with CRISPR. <i>Nature Neuroscience</i> , 2016, 19, 517-522.	14.8	72
18	Genome-wide copy number variant analysis for congenital ventricular septal defects in Chinese Han population. <i>BMC Medical Genomics</i> , 2015, 9, 2.	1.5	20

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19	Annular pancreas in Trichorhinophalangeal syndrome type II with 8q23.3-q24.12 interstitial deletion. <i>Molecular Cytogenetics</i> , 2015, 8, 95.	0.9	4
20	<i>TBX6</i> Null Variants and a Common Hypomorphic Allele in Congenital Scoliosis. <i>New England Journal of Medicine</i> , 2015, 372, 341-350.	27.0	239
21	Fragile X syndrome screening in Chinese children with unknown intellectual developmental disorder. <i>BMC Pediatrics</i> , 2015, 15, 77.	1.7	11
22	Recurrent 8q13.2-13.3 microdeletions associated with Branchio-oto-renal syndrome are mediated by human endogenous retroviral (HERV) sequence blocks. <i>BMC Medical Genetics</i> , 2014, 15, 90.	2.1	10
23	PCMT1 gene polymorphisms, maternal folate metabolism, and neural tube defects: a case-control study in a population with relatively low folate intake. <i>Genes and Nutrition</i> , 2013, 8, 581-587.	2.5	8
24	Increased genome instability in human DNA segments with self-chains: homology-induced structural variations via replicative mechanisms. <i>Human Molecular Genetics</i> , 2013, 22, 2642-2651.	2.9	22
25	Molecular Analysis of a Deletion Hotspot in the NRXN1 Region Reveals the Involvement of Short Inverted Repeats in Deletion CNVs. <i>American Journal of Human Genetics</i> , 2013, 92, 375-386.	6.2	42
26	A novel approach for copy number variation analysis by combining multiplex PCR with matrix-assisted laser desorption ionization time-of-flight mass spectrometry. <i>Journal of Biotechnology</i> , 2013, 166, 6-11.	3.8	7
27	Detection of Copy Number Variants Reveals Association of Cilia Genes with Neural Tube Defects. <i>PLoS ONE</i> , 2013, 8, e54492.	2.5	27
28	Association Study of PARD3 Gene Polymorphisms With Neural Tube Defects in a Chinese Han Population. <i>Reproductive Sciences</i> , 2012, 19, 764-771.	2.5	8
29	Altered Methylation of the DNA Repair Gene MGMT Is Associated with Neural Tube Defects. <i>Journal of Molecular Neuroscience</i> , 2012, 47, 42-51.	2.3	18
30	Intra-family phenotypic heterogeneity of 16p11.2 deletion carriers in a three-generation Chinese family. <i>Journal of Medical Genetics</i> , 2011, 156, 225-232.		38
31	Antidiabetic effect of flavones from <i>Cirsium japonicum</i> DC in diabetic rats. <i>Archives of Pharmacal Research</i> , 2010, 33, 353-362.	6.3	65
32	Novel function of RECS1 as a negative regulator of TNF- $\alpha$ -induced NF- $\kappa$ B activation. <i>Molecular and Cellular Biochemistry</i> , 2010, 337, 317-317.	3.1	0
33	Deletions of <i>NRXN1</i> (neurexin-1) predispose to a wide spectrum of developmental disorders. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2010, 153B, 937-947.	1.7	217
34	Global DNA hypomethylation is associated with NTD-affected pregnancy: A case-control study. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2010, 88, 575-581.	1.6	51
35	Molecular cloning and functional analysis of Chinese sturgeon ( <i>Acipenser sinensis</i> ) growth hormone receptor. <i>Science in China Series C: Life Sciences</i> , 2009, 52, 911-921.	1.3	7
36	Genomic Dissection of Population Substructure of Han Chinese and Its Implication in Association Studies. <i>American Journal of Human Genetics</i> , 2009, 85, 762-774.	6.2	338

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37	The Effect of Plasma Lead on Anembryonic Pregnancy. Annals of the New York Academy of Sciences, 2008, 1140, 184-189.	3.8	9