

# Xiumin Wang

## List of Publications by Year in descending order

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Version: 2024-02-01

50  
papers

878  
citations

567281

15  
h-index

526287

27  
g-index

54  
all docs

54  
docs citations

54  
times ranked

1864  
citing authors

#	ARTICLE	IF	CITATIONS
1	Evaluation of three read-depth based CNV detection tools using whole-exome sequencing data. <i>Molecular Cytogenetics</i> , 2017, 10, 30.	0.9	87
2	CXCL16/CXCR6 chemokine signaling mediates breast cancer progression by pERK1/2-dependent mechanisms. <i>Oncotarget</i> , 2015, 6, 14165-14178.	1.8	77
3	Proband-only medical exome sequencing as a cost-effective first-tier genetic diagnostic test for patients without prior molecular tests and clinical diagnosis in a developing country: the China experience. <i>Genetics in Medicine</i> , 2018, 20, 1045-1053.	2.4	64
4	The feedback loop between miR-124 and TGF- $\beta$ 2 pathway plays a significant role in non-small cell lung cancer metastasis. <i>Carcinogenesis</i> , 2016, 37, 333-343.	2.8	55
5	TriGlycerides and high-density lipoprotein cholesterol ratio compared with homeostasis model assessment insulin resistance indexes in screening for metabolic syndrome in the chinese obese children: a cross section study. <i>BMC Pediatrics</i> , 2015, 15, 138.	1.7	39
6	The effects of intrauterine undernutrition on pancreas ghrelin and insulin expression in neonate rats. <i>Journal of Endocrinology</i> , 2007, 194, 121-129.	2.6	37
7	Association between bisphenol a exposure and idiopathic central precocious puberty (ICPP) among school-aged girls in Shanghai, China. <i>Environment International</i> , 2018, 115, 410-416.	10.0	37
8	Novel pathogenic ACAN variants in non-syndromic short stature patients. <i>Clinica Chimica Acta</i> , 2017, 469, 126-129.	1.1	35
9	Prevalence of precocious puberty among Chinese children: a school population-based study. <i>Endocrine</i> , 2021, 72, 573-581.	2.3	35
10	Description of the molecular and phenotypic spectrum of Wiedemann-Steiner syndrome in Chinese patients. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 178.	2.7	30
11	Molecular and phenotypic spectrum of Noonan syndrome in Chinese patients. <i>Clinical Genetics</i> , 2019, 96, 290-299.	2.0	29
12	Clinical and Molecular Characterization of Patients with Fructose 1,6-Bisphosphatase Deficiency. <i>International Journal of Molecular Sciences</i> , 2017, 18, 857.	4.1	27
13	Association between Dietary Patterns and Precocious Puberty in Children: A Population-Based Study. <i>International Journal of Endocrinology</i> , 2018, 2018, 1-7.	1.5	27
14	Clinical and mutation profile of pediatric patients with RASopathy-associated hypertrophic cardiomyopathy: results from a Chinese cohort. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 29.	2.7	27
15	Difference of Precocious Puberty Between Before and During the COVID-19 Pandemic: A Cross-Sectional Study Among Shanghai School-Aged Girls. <i>Frontiers in Endocrinology</i> , 2022, 13, 839895.	3.5	21
16	Biallelic mutations in GPD1 gene in a Chinese boy mainly presented with obesity, insulin resistance, fatty liver, and short stature. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 3189-3194.	1.2	19
17	Hypermethylated in cancer 1(HIC1) suppresses non-small cell lung cancer progression by targeting interleukin-6/Stat3 pathway. <i>Oncotarget</i> , 2016, 7, 30350-30364.	1.8	17
18	Clinical Profiles and Genetic Spectra of 814 Chinese Children With Short Stature. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, 972-985.	3.6	17

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19	New insights into 5 $\alpha$ -reductase type 2 deficiency based on a multi-centre study: regional distribution and genotype-phenotype profiling of <i>SRD5A2</i> in 190 Chinese patients. <i>Journal of Medical Genetics</i> , 2019, 56, 685-692.	3.2	16
20	New insights from unbiased panel and whole-exome sequencing in a large Chinese cohort with disorders of sex development. <i>European Journal of Endocrinology</i> , 2019, 181, 311-323.	3.7	13
21	Nine cases of childhood adrenal tumour presenting with hypertension and a review of the literature. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2007, 96, 930-934.	1.5	12
22	Compound heterozygous variants of the <i>COG6</i> gene in a Chinese patient with deficiency of subunit 6 of the conserved oligomeric Golgi complex (COG6-CDG). <i>European Journal of Medical Genetics</i> , 2019, 62, 44-46.	1.3	12
23	SOPH Syndrome with Growth Hormone Deficiency, Normal Bone Age, and Novel Compound Heterozygous Mutations in <i>NBAS</i> . <i>Fetal and Pediatric Pathology</i> , 2018, 37, 404-410.	0.7	11
24	Exome Sequencing Identifies <i>De Novo DYNC1H1</i> Mutations Associated With Distal Spinal Muscular Atrophy and Malformations of Cortical Development. <i>Journal of Child Neurology</i> , 2017, 32, 379-386.	1.4	10
25	Novel compound heterozygous variants in the <i>LHCGR</i> gene identified in a subject with Leydig cell hypoplasia type 1. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2018, 31, 239-245.	0.9	9
26	The phenotypic spectrum of Kabuki syndrome in patients of Chinese descent: A case series. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 640-651.	1.2	8
27	Effects of Long-Term High-Fat/High-Energy and High-Protein Diets on Insulin and Ghrelin Expression in Developing Rats. <i>Endocrine Research</i> , 2012, 37, 97-109.	1.2	7
28	Causal variants screened by whole exome sequencing in a patient with maternal uniparental isodisomy of chromosome 10 and a complicated phenotype. <i>Experimental and Therapeutic Medicine</i> , 2016, 11, 2247-2253.	1.8	7
29	Identification and analysis of the genetic causes in nine unrelated probands with syndromic craniosynostosis. <i>Gene</i> , 2018, 641, 144-150.	2.2	7
30	Clinical and molecular genetic characterization of two patients with mutations in the phosphoglucomutase 1 ( <i>PGM1</i> ) gene. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2018, 31, 781-788.	0.9	7
31	A novel <i>PIK3R1</i> mutation of SHORT syndrome in a Chinese female with diffuse thyroid disease: a case report and review of literature. <i>BMC Medical Genetics</i> , 2020, 21, 215.	2.1	7
32	TRMA syndrome with a severe phenotype, cerebral infarction, and novel compound heterozygous <i>SLC19A2</i> mutation: a case report. <i>BMC Pediatrics</i> , 2019, 19, 233.	1.7	6
33	Study of novel <i>NARS2</i> variants in patient of combined oxidative phosphorylation deficiency 24. <i>Translational Pediatrics</i> , 2022, 11, 448-457.	1.2	6
34	Clinical characteristics and follow-up of 5 young Chinese males with gonadotropin-releasing hormone deficiency caused by mutations in the <i>KAL1</i> gene. <i>Meta Gene</i> , 2016, 7, 64-69.	0.6	5
35	<i>De Novo</i> Mutation of <i>KAT6B</i> Gene Causing Atypical Say-Barber-Biesecker-Young-Simpson Syndrome or Genitopatellar Syndrome. <i>Fetal and Pediatric Pathology</i> , 2017, 36, 130-138.	0.7	5
36	Increased transactivation and impaired repression of $\beta$ -catenin-mediated transcription associated with a novel <i>SOX3</i> missense mutation in an X-linked hypopituitarism pedigree with modest growth failure. <i>Molecular and Cellular Endocrinology</i> , 2018, 478, 133-140.	3.2	5

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37	<i>TRPS1</i> mutation detection in Chinese patients with Tricho-rhino-phalangeal syndrome and identification of four novel mutations. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1417.	1.2	5
38	Identification of SOFT syndrome caused by a pathogenic homozygous splicing variant of POC1A: a case report. <i>BMC Medical Genomics</i> , 2021, 14, 207.	1.5	5
39	Molecular and Phenotypic Expansion of Alström Syndrome in Chinese Patients. <i>Frontiers in Genetics</i> , 2022, 13, 808919.	2.3	5
40	Turner syndrome caused by rare complex structural abnormalities involving chromosome X. <i>Experimental and Therapeutic Medicine</i> , 2017, 14, 2265-2270.	1.8	4
41	Growth Pattern in Chinese Children With $\beta$ -Reductase Type 2 Deficiency: A Retrospective Multicenter Study. <i>Frontiers in Pharmacology</i> , 2019, 10, 173.	3.5	4
42	Biallelic ERBB3 loss-of-function variants are associated with a novel multisystem syndrome without congenital contracture. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 265.	2.7	4
43	Targeted exome sequencing identified a novel mutation hotspot and a deletion in Chinese primary hypertrophic osteoarthropathy patients. <i>Clinica Chimica Acta</i> , 2018, 487, 264-269.	1.1	3
44	Clinical and molecular analysis in a cohort of Chinese children with Cornelia de Lange syndrome. <i>Scientific Reports</i> , 2020, 10, 21224.	3.3	3
45	Clinical and genetic analysis in a Chinese cohort of children and adolescents with diabetes/persistent hyperglycemia. <i>Journal of Diabetes Investigation</i> , 2021, 12, 48-62.	2.4	3
46	Evaluating the variety of GNAS inactivation disorders and their clinical manifestations in 11 Chinese children. <i>BMC Endocrine Disorders</i> , 2022, 22, 70.	2.2	3
47	A rare unbalanced Y:autosome translocation in a Turner syndrome patient. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2018, 31, 349-353.	0.9	2
48	A novel <i>CEP57</i> variant associated with mosaic variegated aneuploidy syndrome in a Chinese female presenting with short stature, microcephaly, brachydactyly, and small teeth. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2022, , e1951.	1.2	2
49	A novel homozygous variant of COL2A1 in a Chinese male with type II collagenopathy: a case report. <i>BMC Medical Genomics</i> , 2021, 14, 201.	1.5	0
50	Growth Curves of Chinese Children with Androgen Insensitivity Syndrome: A Multicenter Registry Study. <i>Journal of Personalized Medicine</i> , 2022, 12, 771.	2.5	0