Xiumin Wang

List of Publications by Year
in descending order

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50	878	643344	591227
papers	citations	15 h-index	27 g-index

54	54	54	1949
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinical Profiles and Genetic Spectra of 814 Chinese Children With Short Stature. Journal of Clinical Endocrinology and Metabolism, 2022, 107, 972-985.	1.8	17
2	Molecular and Phenotypic Expansion of AlstrÃ¶m Syndrome in Chinese Patients. Frontiers in Genetics, 2022, 13, 808919.	1.1	5
3	Study of novel NARS2 variants in patient of combined oxidative phosphorylation deficiency 24. Translational Pediatrics, 2022, 11, 448-457.	0.5	6
4	Evaluating the variety of GNAS inactivation disorders and their clinical manifestations in 11 Chinese children. BMC Endocrine Disorders, 2022, 22, 70.	0.9	3
5	A novel CEP57 variant associated with mosaic variegated aneuploidy syndrome in a Chinese female presenting with short stature, microcephaly, brachydactyly, and small teeth. Molecular Genetics & Genomic Medicine, 2022, , e1951.	0.6	2
6	Difference of Precocious Puberty Between Before and During the COVID-19 Pandemic: A Cross-Sectional Study Among Shanghai School-Aged Girls. Frontiers in Endocrinology, 2022, 13, 839895.	1.5	21
7	Growth Curves of Chinese Children with Androgen Insensitivity Syndrome: A Multicenter Registry Study. Journal of Personalized Medicine, 2022, 12, 771.	1.1	0
8	Clinical and genetic analysis in a Chinese cohort of children and adolescents with diabetes/persistent hyperglycemia. Journal of Diabetes Investigation, 2021, 12, 48-62.	1.1	3
9	Prevalence of precocious puberty among Chinese children: a school population-based study. Endocrine, 2021, 72, 573-581.	1.1	35
10	Identification of SOFT syndrome caused by a pathogenic homozygous splicing variant of POC1A: a case report. BMC Medical Genomics, 2021, 14, 207.	0.7	5
11	A novel homozygous variant of COL2A1 in a Chinese male with type II collagenopathy: a case report. BMC Medical Genomics, 2021, 14, 201.	0.7	0
12	The phenotypic spectrum of Kabuki syndrome in patients of Chinese descent: A case series. American Journal of Medical Genetics, Part A, 2020, 182, 640-651.	0.7	8
13	A novel PIK3R1 mutation of SHORT syndrome in a Chinese female with diffuse thyroid disease: a case report and review of literature. BMC Medical Genetics, 2020, 21, 215.	2.1	7
14	TRPS1 mutation detection in Chinese patients with Trichoâ€rhinoâ€phalangeal syndrome and identification of four novel mutations. Molecular Genetics & Genomic Medicine, 2020, 8, e1417.	0.6	5
15	Clinical and molecular analysis in a cohort of Chinese children with Cornelia de Lange syndrome. Scientific Reports, 2020, 10, 21224.	1.6	3
16	Compound heterozygous variants of the COG6 gene in a Chinese patient with deficiency of subunit 6 of the conserved oligomeric Golgi complex (COG6-CDG). European Journal of Medical Genetics, 2019, 62, 44-46.	0.7	12
17	TRMA syndrome with a severe phenotype, cerebral infarction, and novel compound heterozygous SLC19A2 mutation: a case report. BMC Pediatrics, 2019, 19, 233.	0.7	6
18	Molecular and phenotypic spectrum of Noonan syndrome in Chinese patients. Clinical Genetics, 2019, 96, 290-299.	1.0	29

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19	New insights into 5Î±-reductase type 2 deficiency based on a multi-centre study: regional distribution and genotypeâ€“phenotype profiling ofSRD5A2in 190 Chinese patients. Journal of Medical Genetics, 2019, 56, 685-692.	1.5	16
20	Growth Pattern in Chinese Children With 5Î±-Reductase Type 2 Deficiency: A Retrospective Multicenter Study. Frontiers in Pharmacology, 2019, 10, 173.	1.6	4
21	Clinical and mutation profile of pediatric patients with RASopathy-associated hypertrophic cardiomyopathy: results from a Chinese cohort. Orphanet Journal of Rare Diseases, 2019, 14, 29.	1.2	27
22	Biallelic ERBB3 loss-of-function variants are associated with a novel multisystem syndrome without congenital contracture. Orphanet Journal of Rare Diseases, 2019, 14, 265.	1.2	4
23	New insights from unbiased panel and whole-exome sequencing in a large Chinese cohort with disorders of sex development. European Journal of Endocrinology, 2019, 181, 311-323.	1.9	13
24	Association between bisphenol a exposure and idiopathic central precocious puberty (ICPP) among school-aged girls in Shanghai, China. Environment International, 2018, 115, 410-416.	4.8	37
25	Novel compound heterozygous variants in the LHCGR gene identified in a subject with Leydig cell hypoplasia type 1. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 239-245.	0.4	9
26	A rare unbalanced Y:autosome translocation in a Turner syndrome patient. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 349-353.	0.4	2
27	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. Genetics in Medicine, 2018, 20, 1045-1053.	1.1	64
28	Identification and analysis of the genetic causes in nine unrelated probands with syndromic craniosynostosis. Gene, 2018, 641, 144-150.	1.0	7
29	SOPH Syndrome with Growth Hormone Deficiency, Normal Bone Age, and Novel Compound Heterozygous Mutations in NBAS. Fetal and Pediatric Pathology, 2018, 37, 404-410.	0.4	11
30	Targeted exome sequencing identified a novel mutation hotspot and a deletion in Chinese primary hypertrophic osteoarthropathy patients. Clinica Chimica Acta, 2018, 487, 264-269.	0.5	3
31	Description of the molecular and phenotypic spectrum of Wiedemann-Steiner syndrome in Chinese patients. Orphanet Journal of Rare Diseases, 2018, 13, 178.	1.2	30
32	Association between Dietary Patterns and Precocious Puberty in Children: A Population-Based Study. International Journal of Endocrinology, 2018, 2018, 1-7.	0.6	27
33	Increased transactivation and impaired repression of Î²-catenin-mediated transcription associated with a novel SOX3 missense mutation in an X-linked hypopituitarism pedigree with modest growth failure. Molecular and Cellular Endocrinology, 2018, 478, 133-140.	1.6	5
34	Clinical and molecular genetic characterization of two patients with mutations in the phosphoglucomutase 1 (PGM1) gene. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 781-788.	0.4	7
35	Exome Sequencing Identifies De Novo DYNC1H1 Mutations Associated With Distal Spinal Muscular Atrophy and Malformations of Cortical Development. Journal of Child Neurology, 2017, 32, 379-386.	0.7	10
36	De NovoMutation ofKAT6BGene Causing Atypical Sayâ€“Barberâ€“Bieseckerâ€“Youngâ€“Simpson Syndrome or Genitopatellar Syndrome. Fetal and Pediatric Pathology, 2017, 36, 130-138.	0.4	5

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37	Novel pathogenic ACAN variants in non-syndromic short stature patients. Clinica Chimica Acta, 2017, 469, 126-129.	0.5	35
38	Turner syndrome caused by rare complex structural abnormalities involving chromosome X. Experimental and Therapeutic Medicine, 2017, 14, 2265-2270.	0.8	4
39	Biallelic mutations in GPD1 gene in a Chinese boy mainly presented with obesity, insulin resistance, fatty liver, and short stature. American Journal of Medical Genetics, Part A, 2017, 173, 3189-3194.	0.7	19
40	Clinical and Molecular Characterization of Patients with Fructose 1,6-Bisphosphatase Deficiency. International Journal of Molecular Sciences, 2017, 18, 857.	1.8	27
41	Evaluation of three read-depth based CNV detection tools using whole-exome sequencing data. Molecular Cytogenetics, 2017, 10, 30.	0.4	87
42	Causal variants screened by whole exome sequencing in a patient with maternal uniparental isodisomy of chromosome 10 and a complicated phenotype. Experimental and Therapeutic Medicine, 2016, 11, 2247-2253.	0.8	7
43	Clinical characteristics and follow-up of 5 young Chinese males with gonadotropin-releasing hormone deficiency caused by mutations in the KAL1 gene. Meta Gene, 2016, 7, 64-69.	0.3	5
44	The feedback loop between miR-124 and TGF-Î² pathway plays a significant role in non-small cell lung cancer metastasis. Carcinogenesis, 2016, 37, 333-343.	1.3	55
45	Hypermethylated in cancer 1(HIC1) suppresses non-small cell lung cancer progression by targeting interleukin-6/Stat3 pathway. Oncotarget, 2016, 7, 30350-30364.	0.8	17
46	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. BMC Pediatrics, 2015, 15, 138.	0.7	39
47	CXCL16/CXCR6 chemokine signaling mediates breast cancer progression by pERK1/2-dependent mechanisms. Oncotarget, 2015, 6, 14165-14178.	0.8	77
48	Effects of Long-Term High-Fat/High-Energy and High-Protein Diets on Insulin and Ghrelin Expression in Developing Rats. Endocrine Research, 2012, 37, 97-109.	0.6	7
49	The effects of intrauterine undernutrition on pancreas ghrelin and insulin expression in neonate rats. Journal of Endocrinology, 2007, 194, 121-129.	1.2	37
50	Nine cases of childhood adrenal tumour presenting with hypertension and a review of the literature. Acta Paediatrica, International Journal of Paediatrics, 2007, 96, 930-934.	0.7	12