Huiwen Zhang

List of Publications by Year
in descending order

58	1,074	430874	526287
papers	citations	18 h-index	27 g-index

64	64	64	1454
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinical, genetic profile and therapy evaluation of 55 children and 5 adults with sitosterolemia. Journal of Clinical Lipidology, 2022, 16, 40-51.	1.5	10
2	Genome sequencing demonstrates high diagnostic yield in children with undiagnosed global developmental delay/intellectual disability: A prospective study. Human Mutation, 2022, 43, 568-581.	2.5	12
3	Diagnosis and followâ€up of glycogen storage disease (GSD) type VI from the largest GSD center in China. Human Mutation, 2022, 43, 557-567.	2.5	2
4	The Follow-Up of Chinese Patients in cblC Type Methylmalonic Acidemia Identified Through Expanded Newborn Screening. Frontiers in Genetics, 2022, 13, 805599.	2.3	7
5	Investigation of <scp> GALNS </scp> variants and genotypeâ€“phenotype correlations in a large cohort of patients with mucopolysaccharidosis type <scp>IVA</scp>. Journal of Inherited Metabolic Disease, 2022, , .	3.6	2
6	Diagnostic yield of additional exome sequencing after the detection of long continuous stretches of homozygosity (LCSH) in SNP arrays. Journal of Human Genetics, 2021, 66, 409-417.	2.3	3
7	A rare mutation c.1663Gâ€‰>â€‰A (p.A555T) in the MMUT gene associated with mild clinical and biochemical phenotypes of methylmalonic acidemia in 30 Chinese patients. Orphanet Journal of Rare Diseases, 2021, 16, 22.	2.7	5
8	Chromosomal microarray analysis in fetuses with high-risk prenatal indications: A retrospective study in China. Taiwanese Journal of Obstetrics and Gynecology, 2021, 60, 299-304.	1.3	7
9	Clinical, Biochemical, and Molecular Analyses of Medium-Chain Acyl-CoA Dehydrogenase Deficiency in Chinese Patients. Frontiers in Genetics, 2021, 12, 577046.	2.3	9
10	Clinical, biochemical, and genotypeâ€phenotype correlations of 118 patients with Niemannâ€Pick disease Types A/B. Human Mutation, 2021, 42, 614-625.	2.5	18
11	Value of amniotic fluid homocysteine assay in prenatal diagnosis of combined methylmalonic acidemia and homocystinuria, cobalamin C type. Orphanet Journal of Rare Diseases, 2021, 16, 125.	2.7	8
12	Potential Disease-Modifying Effects of Lithium Carbonate in Niemann-Pick Disease, Type C1. Frontiers in Pharmacology, 2021, 12, 667361.	3.5	1
13	Predominant cellular mitochondrial dysfunction in the TOP3A gene-caused Bloom syndrome-like disorder. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2021, 1867, 166106.	3.8	7
14	Rapid detection of twenty-nine common Chinese glucose-6-phosphate dehydrogenase variants using a matrix-assisted laser desorption/ionization-time of flight mass spectrometry assay on dried blood spots. Clinical Biochemistry, 2021, 94, 27-34.	1.9	4
15	One-year follow-up of thyroid function in 23 infants with Prader-Willi syndrome at a single center in China. Intractable and Rare Diseases Research, 2021, 10, 198-201.	0.9	0
16	Different mutations in the MMUT gene are associated with the effect of vitamin B12 in a cohort of 266 Chinese patients with mutâ€type methylmalonic acidemia: A retrospective study. Molecular Genetics & Genomic Medicine, 2021, 9, e1822.	1.2	10
17	An open label, multicenter clinical trial that investigated the efficacy and safety of leuprorelin treatment of central precocious puberty in Chinese children. Medicine (United States), 2021, 100, e28158.	1.0	2
18	Noninvasive prenatal diagnosis of cobalamin C (cblC) deficiency through target region sequencing of cellâ€free DNA in maternal plasma. Prenatal Diagnosis, 2020, 40, 324-332.	2.3	8

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19	Ambroxol improves skeletal and hematological manifestations on a child with Gaucher disease. Journal of Human Genetics, 2020, 65, 345-349.	2.3	11
20	Examining the blood amino acid status in pretherapeutic patients with Âhyperphenylalaninemia. Journal of Clinical Laboratory Analysis, 2020, 34, e23106.	2.1	3
21	Biochemical and genetic approaches to the prenatal diagnosis of propionic acidemia in 78 pregnancies. Orphanet Journal of Rare Diseases, 2020, 15, 276.	2.7	7
22	Prenatal Diagnosis of Glutaric Acidemia I Based on Amniotic Fluid Samples in 42 Families Using Genetic and Biochemical Approaches. Frontiers in Genetics, 2020, 11, 496.	2.3	9
23	High Detection Rate of Copy Number Variations Using Capture Sequencing Data: A Retrospective Study. Clinical Chemistry, 2020, 66, 455-462.	3.2	16
24	Prenatal diagnosis of methylmalonic aciduria from amniotic fluid using genetic and biochemical approaches. Prenatal Diagnosis, 2019, 39, 993-997.	2.3	14
25	In vitro residual activities in 20 variants of phenylalanine hydroxylase and genotype-phenotype correlation in phenylketonuria patients. Gene, 2019, 707, 239-245.	2.2	7
26	Sidt2 regulates hepatocellular lipid metabolism through autophagy. Journal of Lipid Research, 2018, 59, 404-415.	4.2	31
27	Haplotype-based Noninvasive Prenatal Diagnosis of Hyperphenylalaninemia through Targeted Sequencing of Maternal Plasma. Scientific Reports, 2018, 8, 161.	3.3	16
28	Mutation spectrum of hyperphenylalaninemia candidate genes and the genotype-phenotype correlation in the Chinese population. Clinica Chimica Acta, 2018, 481, 132-138.	1.1	24
29	A rare form of Gaucher disease resulting from saposin C deficiency. Blood Cells, Molecules, and Diseases, 2018, 68, 60-65.	1.4	26
30	Genotypes and phenotypes in 20 Chinese patients with type 2 Gaucher disease. Brain and Development, 2018, 40, 876-883.	1.1	5
31	Chromosomal microarray analysis in developmental delay and intellectual disability with comorbid conditions. BMC Medical Genomics, 2018, 11, 49.	1.5	26
32	Skeletal muscle-specific Sidt2 knockout in mice induced muscular dystrophy-like phenotype. Metabolism: Clinical and Experimental, 2018, 85, 259-270.	3.4	12
33	Genetic Evaluation of 114 Chinese Short Stature Children in the Next Generation Era: a Single Center Study. Cellular Physiology and Biochemistry, 2018, 49, 295-305.	1.6	28
34	Rare Loss-of-Function Variants in NPC1 Predispose to Human Obesity. Diabetes, 2017, 66, 935-947.	0.6	54
35	Successful newborn screening for Gaucher disease using fluorometric assay in China. Journal of Human Genetics, 2017, 62, 763-768.	2.3	24
36	Diagnostic Application of Targeted Next-Generation Sequencing of 80 Genes Associated with Disorders of Sexual Development. Scientific Reports, 2017, 7, 44536.	3.3	37

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37	A new case of malonylâ€CoA decarboxylase deficiency with mild clinical features. American Journal of Medical Genetics, Part A, 2016, 170, 1347-1351.	1.2	7
38	21-Hydroxylase deficiency-induced congenital adrenal hyperplasia in 230 Chinese patients: Genotypeâ€“phenotype correlation and identification of nine novel mutations. Steroids, 2016, 108, 47-55.	1.8	33
39	Demographic characteristics and distribution of lysosomal storage disorder subtypes in Eastern China. Journal of Human Genetics, 2016, 61, 345-349.	2.3	28
40	Identification of five novel STAR variants in ten Chinese patients with congenital lipoid adrenal hyperplasia. Steroids, 2016, 108, 85-91.	1.8	20
41	SIDT2 is involved in the NAADP-mediated release of calcium from insulin secretory granules. Journal of Molecular Endocrinology, 2016, 56, 249-259.	2.5	18
42	Biochemical, molecular and outcome analysis of eight chinese asymptomatic individuals with methyl malonic acidemia detected through newborn screening. American Journal of Medical Genetics, Part A, 2015, 167, 2300-2305.	1.2	15
43	A pilot study of gene testing of genetic bone dysplasia using targeted next-generation sequencing. Journal of Human Genetics, 2015, 60, 769-776.	2.3	16
44	Spectrum Analysis of Common Inherited Metabolic Diseases in Chinese Patients Screened and Diagnosed by Tandem Mass Spectrometry. Journal of Clinical Laboratory Analysis, 2015, 29, 162-168.	2.1	34
45	Determination of 7-ketocholesterol in plasma by LC-MS for rapid diagnosis of acid SMase-deficient Niemann-Pick disease. Journal of Lipid Research, 2014, 55, 338-343.	4.2	59
46	Three novel mutations of the FBN1 gene in Chinese children with acromelic dysplasia. Journal of Human Genetics, 2014, 59, 563-567.	2.3	20
47	Diagnosis of Niemann-Pick disease type C with 7-ketocholesterol screening followed by NPC1/NPC2 gene mutation confirmation in Chinese patients. Orphanet Journal of Rare Diseases, 2014, 9, 82.	2.7	46
48	Analysis of genetic mutations in Chinese patients with systemic primary carnitine deficiency. European Journal of Medical Genetics, 2014, 57, 571-575.	1.3	27
49	Identification of a distinct mutation spectrum in the SMPD1 gene of Chinese patients with acid sphingomyelinase-deficient Niemann-Pick disease. Orphanet Journal of Rare Diseases, 2013, 8, 15.	2.7	29
50	Maternal origin of a de novo microdeletion spanning the ERCC6 gene in a classic form of the Cockayne syndrome. European Journal of Medical Genetics, 2011, 54, e389-e393.	1.3	10
51	Analysis of the IDS Gene in 38 Patients with Hunter Syndrome: The c.879G>A (p.Gln293Gln) Synonymous Variation in a Female Create Exonic Splicing. PLoS ONE, 2011, 6, e22951.	2.5	44
52	Two novel missense mutations in the aspartoacylase gene in a Chinese patient with congenital Canavan disease. Brain and Development, 2010, 32, 879-882.	1.1	127
53	A Large Deletion of <i>PROP1</i> Gene in Patients with Combined Pituitary Hormone Deficiency from Two Unrelated Chinese Pedigrees. Hormone Research in Paediatrics, 2010, 74, 98-105.	1.8	8
54	Identification of proteins in the ceroid-like autofluorescent aggregates from liver lysosomes of Beige, a mouse model for human Chediakâ€“Higashi syndrome. Molecular Genetics and Metabolism, 2010, 99, 389-395.	1.1	3

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55	Recurring G12S Mutation of HRAS in a Chinese Child with Costello Syndrome with High Alkaline Phosphatase Level. Biochemical Genetics, 2009, 47, 868-872.	1.7	3
56	Purification and Proteomic Analysis of Lysosomal Integral Membrane Proteins. Methods in Molecular Biology, 2008, 432, 229-241.	0.9	10
57	Lysosomal Membranes from Beige Mice Contain Higher Than Normal Levels of Endoplasmic Reticulum Proteins. Journal of Proteome Research, 2007, 6, 240-249.	3.7	28
58	A Study of Gene Expression Profiles of Cultured Embryonic Rat Neurons Induced by Phenylalanine. Metabolic Brain Disease, 2005, 20, 61-72.	2.9	18