# 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 | 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 |
| 2 | 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 |
| 3 | Rare Loss-of-Function Variants in NPC1 Predispose to Human Obesity. Diabetes, 2017, 66, 935-947. | 0.6 | 54 |
| 4 | 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 |
| 5 | 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 |
| 6 | Diagnostic Application of Targeted Next-Generation Sequencing of 80 Genes Associated with Disorders of Sexual Development. Scientific Reports, 2017, 7, 44536. | 3.3 | 37 |
| 7 | 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 |
| 8 | 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 |
| 9 | Sidt2 regulates hepatocellular lipid metabolism through autophagy. Journal of Lipid Research, 2018, 59, 404-415. | 4.2 | 31 |
| 10 | 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 |
| 11 | 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 |
| 12 | Demographic characteristics and distribution of lysosomal storage disorder subtypes in Eastern China. Journal of Human Genetics, 2016, 61, 345-349. | 2.3 | 28 |
| 13 | 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 |
| 14 | Analysis of genetic mutations in Chinese patients with systemic primary carnitine deficiency. European Journal of Medical Genetics, 2014, 57, 571-575. | 1.3 | 27 |
| 15 | A rare form of Gaucher disease resulting from saposin C deficiency. Blood Cells, Molecules, and Diseases, 2018, 68, 60-65. | 1.4 | 26 |
| 16 | Chromosomal microarray analysis in developmental delay and intellectual disability with comorbid conditions. BMC Medical Genomics, 2018, 11, 49. | 1.5 | 26 |
| 17 | Successful newborn screening for Gaucher disease using fluorometric assay in China. Journal of Human Genetics, 2017, 62, 763-768. | 2.3 | 24 |
| 18 | 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 |

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|---|---|---|---|
| 19 | Three novel mutations of the FBN1 gene in Chinese children with acromelic dysplasia. Journal of Human Genetics, 2014, 59, 563-567. | 2.3 | 20 |
| 20 | Identification of five novel STAR variants in ten Chinese patients with congenital lipoid adrenal hyperplasia. Steroids, 2016, 108, 85-91. | 1.8 | 20 |
| 21 | A Study of Gene Expression Profiles of Cultured Embryonic Rat Neurons Induced by Phenylalanine. Metabolic Brain Disease, 2005, 20, 61-72. | 2.9 | 18 |
| 22 | 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 |
| 23 | 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 |
| 24 | 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 |
| 25 | Haplotype-based Noninvasive Prenatal Diagnosis of Hyperphenylalaninemia through Targeted Sequencing of Maternal Plasma. Scientific Reports, 2018, 8, 161. | 3.3 | 16 |
| 26 | High Detection Rate of Copy Number Variations Using Capture Sequencing Data: A Retrospective Study. Clinical Chemistry, 2020, 66, 455-462. | 3.2 | 16 |
| 27 | 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 |
| 28 | Prenatal diagnosis of methylmalonic aciduria from amniotic fluid using genetic and biochemical approaches. Prenatal Diagnosis, 2019, 39, 993-997. | 2.3 | 14 |
| 29 | Skeletal muscle-specific Sidt2 knockout in mice induced muscular dystrophy-like phenotype. Metabolism: Clinical and Experimental, 2018, 85, 259-270. | 3.4 | 12 |
| 30 | 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 |
| 31 | Ambroxol improves skeletal and hematological manifestations on a child with Gaucher disease. Journal of Human Genetics, 2020, 65, 345-349. | 2.3 | 11 |
| 32 | Purification and Proteomic Analysis of Lysosomal Integral Membrane Proteins. Methods in Molecular Biology, 2008, 432, 229-241. | 0.9 | 10 |
| 33 | 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 |
| 34 | 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 |
| 35 | 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 |
| 36 | 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 |

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|---|---|---|---|
| 37 | Clinical, Biochemical, and Molecular Analyses of Medium-Chain Acyl-CoA Dehydrogenase Deficiency in Chinese Patients. Frontiers in Genetics, 2021, 12, 577046. | 2.3 | 9 |
| 38 | 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 |
| 39 | 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 |
| 40 | 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 |
| 41 | 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 |
| 42 | 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 |
| 43 | 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 |
| 44 | 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 |
| 45 | 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 |
| 46 | 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 |
| 47 | Genotypes and phenotypes in 20 Chinese patients with type 2 Gaucher disease. Brain and Development, 2018, 40, 876-883. | 1.1 | 5 |
| 48 | 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 |
| 49 | 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 |
| 50 | 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 |
| 51 | 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 |
| 52 | Examining the blood amino acid status in pretherapeutic patients with Âhyperphenylalaninemia. Journal of Clinical Laboratory Analysis, 2020, 34, e23106. | 2.1 | 3 |
| 53 | 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 |
| 54 | 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 |

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|---|---|---|---|
| 55 | 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 |
| 56 | 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 |
| 57 | Potential Disease-Modifying Effects of Lithium Carbonate in Niemann-Pick Disease, Type C1. Frontiers in Pharmacology, 2021, 12, 667361. | 3.5 | 1 |
| 58 | 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 |