Lianshu Han

List of Publications by Year
in descending order

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45	967	567281 15	477307 29
papers	citations	h-index	g-index

53	53	53	1517
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinical validation of cutoff target ranges in newborn screening of metabolic disorders by tandem mass spectrometry: A worldwide collaborative project. Genetics in Medicine, 2011, 13, 230-254.	2.4	308
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	Diagnostic Application of Targeted Next-Generation Sequencing of 80 Genes Associated with Disorders of Sexual Development. Scientific Reports, 2017, 7, 44536.	3.3	37
4	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
5	Newborn screening in China: phenylketonuria, congenital hypothyroidism and expanded screening. Annals of the Academy of Medicine, Singapore, 2008, 37, 107-4.	0.4	34
6	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
7	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
8	Demographic characteristics and distribution of lysosomal storage disorder subtypes in Eastern China. Journal of Human Genetics, 2016, 61, 345-349.	2.3	28
9	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
10	Analysis of genetic mutations in Chinese patients with systemic primary carnitine deficiency. European Journal of Medical Genetics, 2014, 57, 571-575.	1.3	27
11	A rare form of Gaucher disease resulting from saposin C deficiency. Blood Cells, Molecules, and Diseases, 2018, 68, 60-65.	1.4	26
12	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
13	Three novel mutations of the FBN1 gene in Chinese children with acromelic dysplasia. Journal of Human Genetics, 2014, 59, 563-567.	2.3	20
14	Identification of five novel STAR variants in ten Chinese patients with congenital lipoid adrenal hyperplasia. Steroids, 2016, 108, 85-91.	1.8	20
15	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
16	Identification of the Plasma Metabolomics as Early Diagnostic Markers between Biliary Atresia and Neonatal Hepatitis Syndrome. PLoS ONE, 2014, 9, e85694.	2.5	17
17	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
18	Haplotype-based Noninvasive Prenatal Diagnosis of Hyperphenylalaninemia through Targeted Sequencing of Maternal Plasma. Scientific Reports, 2018, 8, 161.	3.3	16

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19	High Detection Rate of Copy Number Variations Using Capture Sequencing Data: A Retrospective Study. Clinical Chemistry, 2020, 66, 455-462.	3.2	16
20	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
21	Prenatal diagnosis of methylmalonic aciduria from amniotic fluid using genetic and biochemical approaches. Prenatal Diagnosis, 2019, 39, 993-997.	2.3	14
22	Clinical and molecular characteristics of 69 Chinese patients with ornithine transcarbamylase deficiency. Orphanet Journal of Rare Diseases, 2020, 15, 340.	2.7	12
23	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
24	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
25	Clinical, Biochemical, and Molecular Analyses of Medium-Chain Acyl-CoA Dehydrogenase Deficiency in Chinese Patients. Frontiers in Genetics, 2021, 12, 577046.	2.3	9
26	Insights into the molecular mechanisms of methylmalonic acidemia using microarray technology. International Journal of Clinical and Experimental Medicine, 2015, 8, 8866-79.	1.3	9
27	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
28	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
29	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
30	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
31	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
32	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
33	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
34	Analysis of Amino Acid Patterns With Nutrition Regimens in Preterm Infants With Extrauterine Growth Retardation. Frontiers in Pediatrics, 2020, 8, 184.	1.9	6
35	Noninvasive Prenatal Testing of Methylmalonic Acidemia cblC Type Using the cSMART Assay for MMACHC Gene Mutations. Frontiers in Genetics, 2021, 12, 750719.	2.3	6
36	Genotypes and phenotypes in 20 Chinese patients with type 2 Gaucher disease. Brain and Development, 2018, 40, 876-883.	1.1	5

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37	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
38	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
39	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
40	Different Pattern of Cardiovascular Impairment in Methylmalonic Acidaemia Subtypes. Frontiers in Pediatrics, 2022, 10, 810495.	1.9	3
41	Prenatal Diagnosis of Isovaleric Acidemia From Amniotic Fluid Using Genetic and Biochemical Approaches. Frontiers in Genetics, 0, 13, .	2.3	3
42	Dynamic changes in blood amino acid concentrations in preterm infants in different nutritional periods. Asia Pacific Journal of Clinical Nutrition, 2020, 29, 803-812.	0.4	2
43	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
44	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
45	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