Li Liu

List of Publications by Year
in descending order

29 papers	2,847 citations	1170033 9 h-index	563245 28 g-index
30 all docs	30 docs citations	30 times ranked	4632 citing authors

#	Article	IF	Citations
1	Two De Novo Mosaic Variants Within the Same Site of PHEX Gene in a Girl with X-Linked Hypophosphatemic Rickets. Calcified Tissue International, 2022, 110, 266-271.	1.5	0
2	Mitochondrial FAD shortage in SLC25A32 deficiency affects folate-mediated one-carbon metabolism. Cellular and Molecular Life Sciences, 2022, 79, .	2.4	3
3	A Multicenter Survey of Type I Diabetes Mellitus in Chinese Children. Frontiers in Endocrinology, 2021, 12, 583114.	1.5	9
4	Molecular and clinical characteristics of monogenic diabetes mellitus in southern Chinese children with onset before 3 years of age. BMJ Open Diabetes Research and Care, 2020, 8, e001345.	1.2	6
5	Chinese family with Blau syndrome: Mutated NOD2 allele transmitted from the father with de novo somatic and germ line mosaicism. Journal of Dermatology, 2020, 47, e395.	0.6	2
6	Distinct severity of phenotype in Hajdu-Cheney syndrome: a case report and literature review. BMC Musculoskeletal Disorders, 2020, 21, 154.	0.8	9
7	Clinical and biochemical characteristics of patients with ornithine transcarbamylase deficiency. Clinical Biochemistry, 2020, 84, 63-72.	0.8	4
8	Novel mutations in a Chinese family with two patients with succinic semialdehyde dehydrogenase deficiency. Gynecological Endocrinology, 2020, 36, 929-933.	0.7	3
9	â€˜Isolatedâ€™ germline mosaicism in the phenotypically normal father of a girl with X-linked hypophosphatemic rickets. European Journal of Endocrinology, 2020, 182, K1-K6.	1.9	9
10	A novel homozygous spliceâ€site variant of NCAPD2 gene identified in two siblings with primary microcephaly: The second case report. Clinical Genetics, 2019, 96, 98-101.	1.0	2
11	Somatic and germline FOXP3 mosaicism in the mother of a boy with IPEX syndrome. European Journal of Immunology, 2018, 48, 885-887.	1.6	6
12	Pathogenicity analysis of variations and prenatal diagnosis in a hereditary coagulation factor XIII deficiency family. Hematology, 2018, 23, 501-509.	0.7	4
13	Clinical and molecular characteristics of patients with Gaucher disease in Southern China. Blood Cells, Molecules, and Diseases, 2018, 68, 30-34.	0.6	22
14	Early prenatal diagnosis of lysosomal storage disorders by enzymatic and molecular analysis. Prenatal Diagnosis, 2018, 38, 779-787.	1.1	6
15	Brain abnormalities in fucosidosis: transplantation or supportive therapy?. Metabolic Brain Disease, 2017, 32, 317-320.	1.4	21
16	Congenital cataract with LSS gene mutations: a new case report. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 1231-1235.	0.4	30
17	Clinical,biochemical and molecular analysis of five Chinese patients with Sandhoff disease. Metabolic Brain Disease, 2016, 31, 861-867.	1.4	10
18	Rapid quantification of metabolic intermediates in blood by liquid chromatography-tandem mass spectrometry to investigate congenital lactic acidosis. Analytica Chimica Acta, 2016, 942, 50-57.	2.6	7

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19	Combination of a Haploidentical Stem Cell Transplant With Umbilical Cord Blood for Cerebral X-Linked Adrenoleukodystrophy. Pediatric Neurology, 2015, 53, 163-165.e1.	1.0	6
20	First case report of medium-chain acyl-coenzyme A dehydrogenase deficiency in China. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 681-4.	0.4	5
21	Calcium-sensing receptor stimulates Cl^{â˜}- and SCFA-dependent but inhibits cAMP-dependent HCO₃^{â˜} secretion in colon. American Journal of Physiology - Renal Physiology, 2015, 308, G874-G883.	1.6	35
22	Williams-Beuren Syndrome: A Case Confirmed by Array-CGH Method. Iranian Journal of Pediatrics, 2015, 25, e247.	0.1	2
23	Status and trends of diabetes in Chinese children: analysis of data from 14 medical centers. World Journal of Pediatrics, 2013, 9, 127-134.	0.8	51
24	Molecular genetic assay of mucopolysaccharidosis IVA in South China. Gene, 2013, 532, 46-52.	1.0	8
25	Separation and identification of underivatized plasma acylcarnitine isomers using liquid chromatographyâ€“tandem mass spectrometry for the differential diagnosis of organic acidemias and fatty acid oxidation defects. Journal of Chromatography A, 2013, 1319, 97-106.	1.8	38
26	Measurement of free carnitine and acylcarnitines in plasma by HILIC-ESI-MS/MS without derivatization. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2013, 932, 12-18.	1.2	37
27	First case report of short-chain acyl-CoA dehydrogenase deficiency in China. Journal of Pediatric Endocrinology and Metabolism, 2012, 25, 795-7.	0.4	1
28	Evaluation of factors influencing accuracy in the analysis of succinylacetone in dried blood spots. Clinica Chimica Acta, 2012, 413, 1265-1269.	0.5	13
29	Global, regional, and national causes of child mortality in 2008: a systematic analysis. Lancet, The, 2010, 375, 1969-1987.	6.3	2,498