Dorothea Haas

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/4508769/publications.pdf

Version: 2024-02-01

26	859	567281	552781
papers	citations	15	26
		h-index	g-index

26	26	26	1358
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Sudden neonatal death in individuals with medium-chain acyl-coenzyme A dehydrogenase deficiency: limit of newborn screening. European Journal of Pediatrics, 2022, 181, 2415-2422.	2.7	3
2	Generation of two human iPSC lines, HMGUi003-A and MRIi028-A, carrying pathogenic biallelic variants in the PPCS gene. Stem Cell Research, 2022, 61, 102773.	0.7	2
3	Efficacy and safety of empagliflozin in glycogen storage disease type Ib: Data from an international questionnaire. Genetics in Medicine, 2022, 24, 1781-1788.	2.4	29
4	Longâ€term disease course of two patients with multiple sulfatase deficiency differs from metachromatic leukodystrophy in a broad cohort. JIMD Reports, 2021, 58, 80-88.	1.5	3
5	The spectrum of peripheral neuropathy in disorders of the mitochondrial trifunctional protein. Journal of Inherited Metabolic Disease, 2021, 44, 893-902.	3.6	12
6	Phenotypic diversity, disease progression, and pathogenicity of <scp>MVK</scp> missense variants in mevalonic aciduria. Journal of Inherited Metabolic Disease, 2021, 44, 1272-1287.	3.6	17
7	Health Outcomes of Infants with Vitamin B12 Deficiency Identified by Newborn Screening and Early Treated. Journal of Pediatrics, 2021, 235, 42-48.	1.8	17
8	Impact of glycogen storage disease type I on adult daily life: a survey. Orphanet Journal of Rare Diseases, 2021, 16, 371.	2.7	12
9	Differences of Phenylalanine Concentrations in Dried Blood Spots and in Plasma: Erythrocytes as a Neglected Component for This Observation. Metabolites, 2021, 11, 680.	2.9	3
10	Allelic phenotype values: a model for genotype-based phenotype prediction in phenylketonuria. Genetics in Medicine, 2019, 21, 580-590.	2.4	48
11	Extended diagnosis of purine and pyrimidine disorders from urine: LC MS/MS assay development and clinical validation. PLoS ONE, 2019, 14, e0212458.	2.5	25
12	Semisynthetic sensor proteins enable metabolic assays at the point of care. Science, 2018, 361, 1122-1126.	12.6	120
13	Mutations in PPCS, Encoding Phosphopantothenoylcysteine Synthetase, Cause Autosomal-Recessive Dilated Cardiomyopathy. American Journal of Human Genetics, 2018, 102, 1018-1030.	6.2	42
14	Amyloidosis Cutis Dyschromica, a Rare Cause of Hyperpigmentation: A New Case and Literature Review. Pediatrics, 2017, 139, e20160170.	2.1	10
15	Successful intrauterine treatment of a patient with cobalamin C defect. Molecular Genetics and Metabolism Reports, 2016, 6, 55-59.	1.1	17
16	Adenosine kinase deficiency: expanding the clinical spectrum and evaluating therapeutic options. Journal of Inherited Metabolic Disease, 2016, 39, 273-283.	3.6	55
17	A newly recognized syndrome of severe growth deficiency, microcephaly, intellectual disability, and characteristic facial features. European Journal of Medical Genetics, 2014, 57, 288-292.	1.3	1
18	Diagnosis and therapeutic monitoring of inborn errors of creatine metabolism and transport using liquid chromatographyâ€“tandem mass spectrometry in urine, plasma and CSF. Gene, 2014, 538, 188-194.	2.2	16

#	Article	IF	Citations
19	Prenatal Presentation and Diagnostic Evaluation of Suspected Smithâ€“<scp>L</scp>emliâ€“<scp>O</scp>pitz (<scp>RSH</scp>) Syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 1008-1011.	1.2	16
20	Differential diagnosis in patients with suspected bile acid synthesis defects. World Journal of Gastroenterology, 2012, 18, 1067.	3.3	38
21	Abnormal sterol metabolism in holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 102-108.	1.6	35
22	Plasma and thrombocyte levels of coenzyme Q₁₀ in children with Smithâ€Lemliâ€Opitz syndrome (SLOS) and the influence of HMGâ€CoA reductase inhibitors. BioFactors, 2008, 32, 191-197.	5.4	9
23	Mevalonate kinase deficiencies: from mevalonic aciduria to hyperimmunoglobulinemia D syndrome. Orphanet Journal of Rare Diseases, 2006, 1, 13.	2.7	150
24	Molecular analysis of the MVK and TNFRSF1A genes in patients with a clinical presentation typical of the hyperimmunoglobulinemia D with periodic fever syndrome: A low-penetrance TNFRSF1A variant in a heterozygous MVK carrier possibly influences the phenot. Arthritis and Rheumatism, 2004, 50, 1951-1958.	6.7	41
25	Mevalonate Kinase Deficiency: Enlarging the Clinical and Biochemical Spectrum. Pediatrics, 2003, 111, 258-261.	2.1	83
26	Inherited Disorders of Cholesterol Biosynthesis. Neuropediatrics, 2001, 32, 113-122.	0.6	55