

Dorothea Haas

List of Publications by Year in descending order

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

Version: 2024-02-01

26
papers

859
citations

567281

15
h-index

552781

26
g-index

26
all docs

26
docs citations

26
times ranked

1358
citing authors

#	ARTICLE	IF	CITATIONS
1	Mevalonate kinase deficiencies: from mevalonic aciduria to hyperimmunoglobulinemia D syndrome. Orphanet Journal of Rare Diseases, 2006, 1, 13.	2.7	150
2	Semisynthetic sensor proteins enable metabolic assays at the point of care. Science, 2018, 361, 1122-1126.	12.6	120
3	Mevalonate Kinase Deficiency: Enlarging the Clinical and Biochemical Spectrum. Pediatrics, 2003, 111, 258-261.	2.1	83
4	Inherited Disorders of Cholesterol Biosynthesis. Neuropediatrics, 2001, 32, 113-122.	0.6	55
5	Adenosine kinase deficiency: expanding the clinical spectrum and evaluating therapeutic options. Journal of Inherited Metabolic Disease, 2016, 39, 273-283.	3.6	55
6	Allelic phenotype values: a model for genotype-based phenotype prediction in phenylketonuria. Genetics in Medicine, 2019, 21, 580-590.	2.4	48
7	Mutations in PPCS, Encoding Phosphopantothencycysteine Synthetase, Cause Autosomal-Recessive Dilated Cardiomyopathy. American Journal of Human Genetics, 2018, 102, 1018-1030.	6.2	42
8	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
9	Differential diagnosis in patients with suspected bile acid synthesis defects. World Journal of Gastroenterology, 2012, 18, 1067.	3.3	38
10	Abnormal sterol metabolism in holoprosencephaly. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 102-108.	1.6	35
11	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
12	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
13	Successful intrauterine treatment of a patient with cobalamin C defect. Molecular Genetics and Metabolism Reports, 2016, 6, 55-59.	1.1	17
14	Phenotypic diversity, disease progression, and pathogenicity of <i>MVK</i> missense variants in mevalonic aciduria. Journal of Inherited Metabolic Disease, 2021, 44, 1272-1287.	3.6	17
15	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
16	Prenatal Presentation and Diagnostic Evaluation of Suspected Smith-Lemli-Opitz (RSH) Syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 1008-1011.	1.2	16
17	Diagnosis and therapeutic monitoring of inborn errors of creatine metabolism and transport using liquid chromatography-tandem mass spectrometry in urine, plasma and CSF. Gene, 2014, 538, 188-194.	2.2	16
18	The spectrum of peripheral neuropathy in disorders of the mitochondrial trifunctional protein. Journal of Inherited Metabolic Disease, 2021, 44, 893-902.	3.6	12

#	ARTICLE	IF	CITATIONS
19	Impact of glycogen storage disease type I on adult daily life: a survey. Orphanet Journal of Rare Diseases, 2021, 16, 371.	2.7	12
20	Amyloidosis Cutis Dyschromica, a Rare Cause of Hyperpigmentation: A New Case and Literature Review. Pediatrics, 2017, 139, e20160170.	2.1	10
21	Plasma and thrombocyte levels of coenzyme Q ₁₀ in children with Smith-Lemli-Opitz syndrome (SLOS) and the influence of HMG-CoA reductase inhibitors. BioFactors, 2008, 32, 191-197.	5.4	9
22	Long-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
23	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
24	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
25	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
26	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