Hideo Sasai

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/1295049/publications.pdf

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48	421	759233	888059
papers	citations	12 h-index	17 g-index

50	50	50	503
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Biallelic GALM pathogenic variants cause a novel type of galactosemia. Genetics in Medicine, 2019, 21, 1286-1294.	2.4	40
2	Recent advances in understanding beta-ketothiolase (mitochondrial acetoacetyl-CoA thiolase, T2) deficiency. Journal of Human Genetics, 2019, 64, 99-111.	2.3	30
3	Newborn screening for carnitine palmitoyltransferase II deficiency using (C16 + C18:1)/C2: Evaluation of additional indices for adequate sensitivity and lower false-positivity. Molecular Genetics and Metabolism, 2017, 122, 67-75.	1.1	26
4	Deficiency of 3â€hydroxybutyrate dehydrogenase (BDH1) in mice causes low ketone body levels and fatty liver during fasting. Journal of Inherited Metabolic Disease, 2020, 43, 960-968.	3.6	21
5	Beta-Ketothiolase Deficiency. FIRE Forum for International Research in Education, 2016, 4, 232640981663664.	0.7	18
6	Refractory Chronic Pleurisy Caused by Helicobacter equorum-Like Bacterium in a Patient with X-Linked Agammaglobulinemia. Journal of Clinical Microbiology, 2011, 49, 3432-3435.	3.9	16
7	Exon 10 skipping in ACAT1 caused by a novel c.949G>A mutation located at an exonic splice enhancer site. Molecular Medicine Reports, 2016, 14, 4906-4910.	2.4	15
8	Characterization and outcome of 41 patients with betaâ€ketothiolase deficiency: 10 yearsâ€™ experience of a medical center in northern Vietnam. Journal of Inherited Metabolic Disease, 2017, 40, 395-401.	3.6	15
9	A Transient Lesion in the Corpus Callosum During Rotavirus Infection. Pediatric Neurology, 2009, 41, 467-469.	2.1	13
10	Congenital inner ear malformations without sensorineural hearing loss in children. International Journal of Pediatric Otorhinolaryngology, 2009, 73, 1484-1487.	1.0	13
11	Clinical and Mutational Characterizations of Ten Indian Patients with Beta-Ketothiolase Deficiency. JIMD Reports, 2017, 35, 59-65.	1.5	13
12	Intronic antisense Alu elements have a negative splicing effect on the inclusion of adjacent downstream exons. Gene, 2018, 664, 84-89.	2.2	13
13	Late-onset ornithine transcarbamylase deficiency caused by a somatic mosaic mutation. Human Genome Variation, 2018, 5, 22.	0.7	13
14	A Japanese case of mitochondrial 3â€hydroxyâ€3â€methylglutarylâ€CoA synthase deficiency who presented with severe metabolic acidosis and fatty liver without hypoglycemia. JIMD Reports, 2019, 48, 19-25.	1.5	13
15	Acute cerebellitis associated with rotavirus infection. World Journal of Pediatrics, 2013, 9, 87-89.	1.8	12
16	Successive MRI Findings of Reversible Cerebral White Matter Lesions in a Patient with Cystathionine <i>Î²</i>-Synthase Deficiency. Tohoku Journal of Experimental Medicine, 2015, 237, 323-327.	1.2	12
17	Heterozygous carriers of succinylâ€CoA:3â€oxoacid CoA transferase deficiency can develop severe ketoacidosis. Journal of Inherited Metabolic Disease, 2017, 40, 845-852.	3.6	12
18	Japanese Male Siblings with 2-Methyl-3-Hydroxybutyryl-CoA Dehydrogenase Deficiency (HSD10 Disease) Without Neurological Regression. JIMD Reports, 2016, 32, 81-85.	1.5	11

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19	Advanced Diagnostic System and Introduction of Newborn Screening of Adrenoleukodystrophy and Peroxisomal Disorders in Japan. International Journal of Neonatal Screening, 2021, 7, 58.	3.2	11
20	Clinical manifestation and longâ€term outcome of citrin deficiency: Report from a nationwide study in Japan. Journal of Inherited Metabolic Disease, 2022, 45, 431-444.	3.6	11
21	Paclitaxelâ€based chemotherapy for aggressive kaposiform hemangioendothelioma of the temporomastoid region: Case report and review of the literature. Head and Neck, 2013, 35, E258-61.	2.0	10
22	Effectiveness of Medium-Chain Triglyceride Oil Therapy in Two Japanese Citrin-Deficient Siblings: Evaluation Using Oral Glucose Tolerance Tests. Tohoku Journal of Experimental Medicine, 2016, 240, 323-328.	1.2	9
23	In vitro functional analysis of four variants of human asparagine synthetase. Journal of Inherited Metabolic Disease, 2021, 44, 1226-1234.	3.6	7
24	A Turkish Patient With Succinyl-CoA:3-Oxoacid CoA Transferase Deficiency Mimicking Diabetic Ketoacidosis. FIRE Forum for International Research in Education, 2016, 4, 232640981665128.	0.7	6
25	A novel mutation (c.121-13T>A) in the polypyrimidine tract of the splice acceptor site of intron 2 causes exon 3 skipping in mitochondrial acetoacetyl-CoA thiolase gene. Molecular Medicine Reports, 2017, 15, 3879-3884.	2.4	6
26	Singleâ€nucleotide substitution T to A in the polypyrimidine stretch at the splice acceptor site of intron 9 causes exon 10 skipping in the <scp>ACAT</scp>1 gene. Molecular Genetics & Genomic Medicine, 2017, 5, 177-184.	1.2	5
27	Short-chain enoyl-CoA hydratase deficiency causes prominent ketoacidosis with normal plasma lactate levels: A case report. Molecular Genetics and Metabolism Reports, 2020, 25, 100672.	1.1	5
28	The frequencies of very long-chain acyl-CoA dehydrogenase deficiency genetic variants in Japan have changed since the implementation of expanded newborn screening. Molecular Genetics and Metabolism, 2022, 136, 74-79.	1.1	5
29	Successful treatment of pediatric immune thrombocytopenic purpura associated with ulcerative colitis. Pediatrics International, 2011, 53, 771-773.	0.5	4
30	Primary carnitine deficiency with severe acute hepatitis following rotavirus gastroenteritis. Journal of Infection and Chemotherapy, 2019, 25, 913-916.	1.7	4
31	Current Perspectives on Neonatal Screening for Propionic Acidemia in Japan: An Unexpectedly High Incidence of Patients with Mild Disease Caused by a Common PCCB Variant. International Journal of Neonatal Screening, 2021, 7, 35.	3.2	4
32	Betaâ€ketothiolase deficiency: A case with unusual presentation of nonketotic hypoglycemic episodes due to coexistent probable secondary carnitine deficiency. JIMD Reports, 2019, 46, 23-27.	1.5	4
33	Endoscopic third ventriculostomy for hydrocephalus in a patient with achondroplasia: a case report and literature review. Child's Nervous System, 2021, 37, 3907-3911.	1.1	3
34	Japanese patients with mitochondrial 3â€‘hydroxyâ€‘3â€‘methylglutarylâ€‘CoA synthase deficiency: In vitro functional analysis of five novel HMGCS2 mutations. Experimental and Therapeutic Medicine, 2020, 20, 1-1.	1.8	3
35	MCAD deficiency caused by compound heterozygous pathogenic variants in ACADM. Human Genome Variation, 2022, 9, 2.	0.7	3
36	Diffuse large B-cell lymphoma presenting with osteolytic lesions in the bilateral femur. European Journal of Haematology, 2009, 83, 502-502.	2.2	2

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37	Novel HADHB mutations in a patient with mitochondrial trifunctional protein deficiency. Human Genome Variation, 2020, 7, 10.	0.7	2
38	Retrospective evaluations revealed pre-symptomatic citrulline concentrations measured by newborn screening were significantly low in late-onset ornithine transcarbamylase deficiency patients. Clinica Chimica Acta, 2020, 510, 633-637.	1.1	2
39	A female patient with GSD IXc developing multiple and recurrent hepatocellular carcinoma: a case report and literature review. Human Genome Variation, 2021, 8, 45.	0.7	2
40	Founder genetic variants of ABCC4 and ABCC11 in the Japanese population are not associated with the development of subacute myeloâ€opticoâ€neuropathy (SMON). Molecular Genetics & Genomic Medicine, 2022, 10, e1845.	1.2	2
41	Spondylocostal dysostosis with tetralogy of <scp>F</scp>allot and herniation of the spleen through the diaphragm. Congenital Anomalies (discontinued), 2014, 54, 189-192.	0.6	1
42	Propranolol for infantile hemangiomas with hyperinsulinemic hypoglycemia. Pediatrics International, 2021, 63, 724-725.	0.5	1
43	A short sequence within AluSx induces downstream exon skipping in an ACAT1 minigene model. International Journal of Transgender Health, 2021, 14, 869-873.	2.3	1
44	Immediate postnatal central hypothyroidism caused by maternal Graves' disease: Importance of early screening. Clinical Case Reports (discontinued), 2022, 10, .	0.5	1
45	Translocation (1;10)(p34;p15) in infant acute myeloid leukemia with extramedullary infiltration in multiple sites. Cancer Genetics and Cytogenetics, 2009, 192, 86-89.	1.0	0
46	Peripheral blood stem cell transplantation in a significant body weight difference between a smaller donor and a larger recipient: A case report. Transfusion and Apheresis Science, 2013, 49, 504-506.	1.0	0
47	Neonatal bacteremia caused by emm type 80 group A Streptococcus: A case report. Pediatrics International, 2020, 62, 1305-1306.	0.5	0
48	Long QT as a first sign for propionic acidemia in a 10â€yearâ€old girl. Pediatrics International, 2022, 64, .	0.5	0