

# Hideo Sasai

## List of Publications by Year in descending order

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

Version: 2024-02-01

48  
papers

421  
citations

759233

12  
h-index

888059

17  
g-index

50  
all docs

50  
docs citations

50  
times ranked

503  
citing authors

#	ARTICLE	IF	CITATIONS
1	Long QT as a first sign for propionic acidemia in a 10-year-old girl. <i>Pediatrics International</i> , 2022, 64, .	0.5	0
2	MCAD deficiency caused by compound heterozygous pathogenic variants in ACADM. <i>Human Genome Variation</i> , 2022, 9, 2.	0.7	3
3	Clinical manifestation and long-term outcome of citrin deficiency: Report from a nationwide study in Japan. <i>Journal of Inherited Metabolic Disease</i> , 2022, 45, 431-444.	3.6	11
4	The frequencies of very long-chain acyl-CoA dehydrogenase deficiency genetic variants in Japan have changed since the implementation of expanded newborn screening. <i>Molecular Genetics and Metabolism</i> , 2022, 136, 74-79.	1.1	5
5	Founder genetic variants of <i>ABCC4</i> and <i>ABCC11</i> in the Japanese population are not associated with the development of subacute myelo-optic neuropathy (SMON). <i>Molecular Genetics &amp; Genomic Medicine</i> , 2022, 10, e1845.	1.2	2
6	Immediate postnatal central hypothyroidism caused by maternal Graves' disease: Importance of early screening. <i>Clinical Case Reports (discontinued)</i> , 2022, 10, .	0.5	1
7	Endoscopic third ventriculostomy for hydrocephalus in a patient with achondroplasia: a case report and literature review. <i>Child's Nervous System</i> , 2021, 37, 3907-3911.	1.1	3
8	Propranolol for infantile hemangiomas with hyperinsulinemic hypoglycemia. <i>Pediatrics International</i> , 2021, 63, 724-725.	0.5	1
9	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. <i>International Journal of Neonatal Screening</i> , 2021, 7, 35.	3.2	4
10	In vitro functional analysis of four variants of human asparagine synthetase. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 1226-1234.	3.6	7
11	Advanced Diagnostic System and Introduction of Newborn Screening of Adrenoleukodystrophy and Peroxisomal Disorders in Japan. <i>International Journal of Neonatal Screening</i> , 2021, 7, 58.	3.2	11
12	A short sequence within <i>Alu</i> Sx induces downstream exon skipping in an <i>ACAT1</i> minigene model. <i>International Journal of Transgender Health</i> , 2021, 14, 869-873.	2.3	1
13	A female patient with GSD IXc developing multiple and recurrent hepatocellular carcinoma: a case report and literature review. <i>Human Genome Variation</i> , 2021, 8, 45.	0.7	2
14	Novel HADHB mutations in a patient with mitochondrial trifunctional protein deficiency. <i>Human Genome Variation</i> , 2020, 7, 10.	0.7	2
15	Neonatal bacteremia caused by <i>emm</i> type 80 group A <i>Streptococcus</i> : A case report. <i>Pediatrics International</i> , 2020, 62, 1305-1306.	0.5	0
16	Retrospective evaluations revealed pre-symptomatic citrulline concentrations measured by newborn screening were significantly low in late-onset ornithine transcarbamylase deficiency patients. <i>Clinica Chimica Acta</i> , 2020, 510, 633-637.	1.1	2
17	Short-chain enoyl-CoA hydratase deficiency causes prominent ketoacidosis with normal plasma lactate levels: A case report. <i>Molecular Genetics and Metabolism Reports</i> , 2020, 25, 100672.	1.1	5
18	Deficiency of 3-hydroxybutyrate dehydrogenase (BDH1) in mice causes low ketone body levels and fatty liver during fasting. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 960-968.	3.6	21

#	ARTICLE	IF	CITATIONS
19	Japanese patients with mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency: <i>in vitro</i> functional analysis of five novel HMGCS2 mutations. <i>Experimental and Therapeutic Medicine</i> , 2020, 20, 1-1.	1.8	3
20	A Japanese case of mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency who presented with severe metabolic acidosis and fatty liver without hypoglycemia. <i>JIMD Reports</i> , 2019, 48, 19-25.	1.5	13
21	Primary carnitine deficiency with severe acute hepatitis following rotavirus gastroenteritis. <i>Journal of Infection and Chemotherapy</i> , 2019, 25, 913-916.	1.7	4
22	Biallelic GALM pathogenic variants cause a novel type of galactosemia. <i>Genetics in Medicine</i> , 2019, 21, 1286-1294.	2.4	40
23	Recent advances in understanding beta-ketothiolase (mitochondrial acetoacetyl-CoA thiolase, T2) deficiency. <i>Journal of Human Genetics</i> , 2019, 64, 99-111.	2.3	30
24	Beta-ketothiolase deficiency: A case with unusual presentation of nonketotic hypoglycemic episodes due to coexistent probable secondary carnitine deficiency. <i>JIMD Reports</i> , 2019, 46, 23-27.	1.5	4
25	Intronic antisense Alu elements have a negative splicing effect on the inclusion of adjacent downstream exons. <i>Gene</i> , 2018, 664, 84-89.	2.2	13
26	Late-onset ornithine transcarbamylase deficiency caused by a somatic mosaic mutation. <i>Human Genome Variation</i> , 2018, 5, 22.	0.7	13
27	Characterization and outcome of 41 patients with beta-ketothiolase deficiency: 10 years' experience of a medical center in northern Vietnam. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 395-401.	3.6	15
28	Single nucleotide substitution T to A in the polypyrimidine stretch at the splice acceptor site of intron 9 causes exon 10 skipping in the <i>ACAT1</i> gene. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2017, 5, 177-184.	1.2	5
29	Clinical and Mutational Characterizations of Ten Indian Patients with Beta-Ketothiolase Deficiency. <i>JIMD Reports</i> , 2017, 35, 59-65.	1.5	13
30	Newborn screening for carnitine palmitoyltransferase II deficiency using (C16 + C18:1)/C2: Evaluation of additional indices for adequate sensitivity and lower false-positivity. <i>Molecular Genetics and Metabolism</i> , 2017, 122, 67-75.	1.1	26
31	Heterozygous carriers of succinyl-CoA:3-oxoacid CoA transferase deficiency can develop severe ketoacidosis. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 845-852.	3.6	12
32	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. <i>Molecular Medicine Reports</i> , 2017, 15, 3879-3884.	2.4	6
33	Exon 10 skipping in <i>ACAT1</i> caused by a novel c.949G>A mutation located at an exonic splice enhancer site. <i>Molecular Medicine Reports</i> , 2016, 14, 4906-4910.	2.4	15
34	Effectiveness of Medium-Chain Triglyceride Oil Therapy in Two Japanese Citrin-Deficient Siblings: Evaluation Using Oral Glucose Tolerance Tests. <i>Tohoku Journal of Experimental Medicine</i> , 2016, 240, 323-328.	1.2	9
35	Beta-Ketothiolase Deficiency. <i>FIRE Forum for International Research in Education</i> , 2016, 4, 232640981663664.	0.7	18
36	A Turkish Patient With Succinyl-CoA:3-Oxoacid CoA Transferase Deficiency Mimicking Diabetic Ketoacidosis. <i>FIRE Forum for International Research in Education</i> , 2016, 4, 232640981665128.	0.7	6

#	ARTICLE	IF	CITATIONS
37	Japanese Male Siblings with 2-Methyl-3-Hydroxybutyryl-CoA Dehydrogenase Deficiency (HSD10 Disease) Without Neurological Regression. <i>JIMD Reports</i> , 2016, 32, 81-85.	1.5	11
38	Successive MRI Findings of Reversible Cerebral White Matter Lesions in a Patient with Cystathionine Synthase Deficiency. <i>Tohoku Journal of Experimental Medicine</i> , 2015, 237, 323-327.	1.2	12
39	Spondylocostal dysostosis with tetralogy of Fallot and herniation of the spleen through the diaphragm. <i>Congenital Anomalies (discontinued)</i> , 2014, 54, 189-192.	0.6	1
40	Acute cerebellitis associated with rotavirus infection. <i>World Journal of Pediatrics</i> , 2013, 9, 87-89.	1.8	12
41	Peripheral blood stem cell transplantation in a significant body weight difference between a smaller donor and a larger recipient: A case report. <i>Transfusion and Apheresis Science</i> , 2013, 49, 504-506.	1.0	0
42	Paclitaxel-based chemotherapy for aggressive kaposiform hemangioendothelioma of the temporomastoid region: Case report and review of the literature. <i>Head and Neck</i> , 2013, 35, E258-61.	2.0	10
43	Successful treatment of pediatric immune thrombocytopenic purpura associated with ulcerative colitis. <i>Pediatrics International</i> , 2011, 53, 771-773.	0.5	4
44	Refractory Chronic Pleurisy Caused by Helicobacter equorum-Like Bacterium in a Patient with X-Linked Agammaglobulinemia. <i>Journal of Clinical Microbiology</i> , 2011, 49, 3432-3435.	3.9	16
45	Diffuse large B-cell lymphoma presenting with osteolytic lesions in the bilateral femur. <i>European Journal of Haematology</i> , 2009, 83, 502-502.	2.2	2
46	Translocation (1;10)(p34;p15) in infant acute myeloid leukemia with extramedullary infiltration in multiple sites. <i>Cancer Genetics and Cytogenetics</i> , 2009, 192, 86-89.	1.0	0
47	A Transient Lesion in the Corpus Callosum During Rotavirus Infection. <i>Pediatric Neurology</i> , 2009, 41, 467-469.	2.1	13
48	Congenital inner ear malformations without sensorineural hearing loss in children. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2009, 73, 1484-1487.	1.0	13