

Hisato Suzuki

List of Publications by Year in descending order

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Version: 2024-02-01

50
papers

448
citations

933447

10
h-index

888059

17
g-index

50
all docs

50
docs citations

50
times ranked

873
citing authors

#	ARTICLE	IF	CITATIONS
1	Early diagnosis of lateral meningocele syndrome in an infant without neurological symptoms based on genomic analysis. <i>Child's Nervous System</i> , 2022, 38, 659-663.	1.1	3
2	Ketogenic Diet for KARS-Related Mitochondrial Dysfunction and Progressive Leukodystrophy. <i>Neuropediatrics</i> , 2022, 53, 065-068.	0.6	3
3	A patient with compound heterozygosity of <i>SMPD4</i> : Another example of utility of exome-based copy number analysis in autosomal recessive disorders. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 613-617.	1.2	3
4	BCS1L mutations produce Fanconi syndrome with developmental disability. <i>Journal of Human Genetics</i> , 2022, 67, 143-148.	2.3	5
5	Diagnosis of SLC25A46-related pontocerebellar hypoplasia in two siblings with fulminant neonatal course: role of postmortem CT and whole genomic analysis: a case report. <i>BMC Neurology</i> , 2022, 22, 20.	1.8	0
6	The p.Thr395Met missense variant of <i>NFIA</i> found in a patient with intellectual disability is a defective variant. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1184-1192.	1.2	5
7	Genome Analysis in Sick Neonates and Infants: High-yield Phenotypes and Contribution of Small Copy Number Variations. <i>Journal of Pediatrics</i> , 2022, 244, 38-48.e1.	1.8	8
8	RNF213-Associated Vascular Disease: A Concept Unifying Various Vasculopathies. <i>Life</i> , 2022, 12, 555.	2.4	5
9	Phenotypic overlap between cardioacrofacial dysplasia-2 and oral-facial-digital syndrome. <i>European Journal of Medical Genetics</i> , 2022, , 104512.	1.3	0
10	Deciphering complex rearrangements at the breakpoint of an apparently balanced reciprocal translocation t(4:18)(q31;q11.2)dn and at a cryptic deletion: Further evidence of <i>TLL1</i> as a causative gene for atrial septal defect. <i>American Journal of Medical Genetics, Part A</i> , 2022, ,	1.2	4
11	Precocious puberty in a case of Simpson-Golabi-Behmel syndrome with a de novo 240-kb deletion including GPC3. <i>Human Genome Variation</i> , 2022, 9, .	0.7	0
12	Role of chimeric transcript formation in the pathogenesis of birth defects. <i>Congenital Anomalies (discontinued)</i> , 2021, 61, 76-81.	0.6	11
13	Severe course with lethal hepatocellular injury and skeletal muscular dysgenesis in a neonate with infantile liver failure syndrome type 1 caused by novel <i>LARS1</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 866-870.	1.2	4
14	Fork-shaped mandibular incisors as a novel phenotype of LRP5-associated disorder. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1544-1549.	1.2	1
15	Establishing intellectual disability as the key feature of patients with biallelic <i>RNPC3</i> variants. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1836-1840.	1.2	3
16	Frequent FGFR3 and Ras Gene Mutations in Skin Tags or Acrochordons. <i>Journal of Investigative Dermatology</i> , 2021, 141, 2756-2760.e8.	0.7	2
17	Recurrent <i>NFIA</i> K125E substitution represents a loss-of-function allele: Sensitive in vitro and in vivo assays for nontruncating alleles. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2084-2093.	1.2	9
18	Noonan syndrome-like phenotype in a patient with heterozygous <i>ERF</i> truncating variant. <i>Congenital Anomalies (discontinued)</i> , 2021, 61, 226-230.	0.6	1

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19	Biallelic loss of <i>OTUD7A</i> causes severe muscular hypotonia, intellectual disability, and seizures. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1182-1186.	1.2	14
20	Multiple introductions of SARS-CoV-2 B.1.1.214 lineages from mainland Japan preceded the third wave of the COVID-19 epidemic in Hokkaido. <i>Travel Medicine and Infectious Disease</i> , 2021, 44, 102210.	3.0	3
21	Infantile neuroaxonal dystrophy in a pair of Malaysian siblings with progressive cerebellar atrophy: Description of an expanded phenotype with novel <i>PLA2G6</i> variants. <i>Journal of Clinical Neuroscience</i> , 2020, 71, 289-292.	1.5	2
22	Poor outcomes in carriers of the <i>RNF213</i> variant (p.Arg4810Lys) with pulmonary arterial hypertension. <i>Journal of Heart and Lung Transplantation</i> , 2020, 39, 103-112.	0.6	25
23	Parallel detection of single nucleotide variants and copy number variants with exome analysis: Validation in a cohort of 700 undiagnosed patients. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2529-2532.	1.2	12
24	Consecutive medical exome analysis at a tertiary center: Diagnostic and health-economic outcomes. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1601-1607.	1.2	10
25	De novo 2q36.3q37.1 deletion encompassing <i>TRIP12</i> and <i>NPPC</i> yields distinct phenotypes. <i>Human Genome Variation</i> , 2020, 7, 19.	0.7	1
26	Novel <i>ARX</i> mutation identified in infantile spasm syndrome patient. <i>Human Genome Variation</i> , 2020, 7, 9.	0.7	2
27	Biallelic Mutations in the <i>LSR</i> Gene Cause a Novel Type of Infantile Intrahepatic Cholestasis. <i>Journal of Pediatrics</i> , 2020, 221, 251-254.	1.8	11
28	Clonal Expansion of Second-Hit Cells with Somatic Recombinations or C>T Transitions Form Porokeratosis in MVD or MVK Mutant Heterozygotes. <i>Journal of Investigative Dermatology</i> , 2019, 139, 2458-2466.e9.	0.7	49
29	<i>IFT172</i> as the 19th gene causative of oral-facial-digital syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2510-2513.	1.2	9
30	A case of autism spectrum disorder with cleft lip and palate carrying a mutation in exon 8 of <i>AUTS2</i> . <i>Clinical Case Reports (discontinued)</i> , 2019, 7, 2059-2063.	0.5	5
31	De novo <i>NSF</i> mutations cause early infantile epileptic encephalopathy. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 2334-2339.	3.7	10
32	<i>CNOT2</i> haploinsufficiency causes a neurodevelopmental disorder with characteristic facial features. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2506-2509.	1.2	9
33	Severe Noonan syndrome phenotype associated with a germline Q71R <i>MRAS</i> variant: a recurrent substitution in <i>RAS</i> homologs in various cancers. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1628-1630.	1.2	13
34	A novel missense <i>PTEN</i> mutation identified in a patient with macrocephaly and developmental delay. <i>Human Genome Variation</i> , 2019, 6, 25.	0.7	3
35	Noninvasive diagnosis of <i>TRIT1</i> -related mitochondrial disorder by measuring ⁶ A37 and ² <i>ms</i> ⁶ <i>A37</i> modifications in tRNAs from blood and urine samples. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1609-1614.	1.2	6
36	Evaluation of Face2Gene using facial images of patients with congenital dysmorphic syndromes recruited in Japan. <i>Journal of Human Genetics</i> , 2019, 64, 789-794.	2.3	51

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37	Medical genetics and genomic medicine in Japan. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 166-169.	1.6	4
38	SATB2-associated syndrome in patients from Japan: Linguistic profiles. American Journal of Medical Genetics, Part A, 2019, 179, 896-899.	1.2	11
39	<i>CNOT2</i> as the critical gene for phenotypes of 12q15 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 659-662.	1.2	8
40	Schuurs-Hoeijmakers syndrome in two patients from Japan. American Journal of Medical Genetics, Part A, 2019, 179, 341-343.	1.2	16
41	Japanese pathogenic variant database: DPV. Translational Science of Rare Diseases, 2018, 3, 133-137.	1.5	1
42	Ablepharon and craniosynostosis in a patient with a localized <i>TWIST1</i> basic domain substitution. American Journal of Medical Genetics, Part A, 2018, 176, 2777-2780.	1.2	7
43	Genomic Comparison With Supercentenarians Identifies <i>RNF213</i> as a Risk Gene for Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2018, 11, e002317.	3.6	14
44	Haploinsufficiency of <i>NCOR1</i> associated with autism spectrum disorder, scoliosis, and abnormal palatogenesis. American Journal of Medical Genetics, Part A, 2018, 176, 2466-2469.	1.2	5
45	<i>SOX17</i> Mutations in Japanese Patients with Pulmonary Arterial Hypertension. American Journal of Respiratory and Critical Care Medicine, 2018, 198, 1231-1233.	5.6	22
46	A novel BBS10 mutation identified in a patient with Bardet-Biedl syndrome with a violent emotional outbreak. Human Genome Variation, 2017, 4, 17033.	0.7	4
47	Preaxial polydactyly in an individual with Wiedemann-Steiner syndrome caused by a novel nonsense mutation in <i>KMT2A</i> . , 2017, 173, 2821-2825.		11
48	Truncating mutation in <i>CSNK2B</i> and myoclonic epilepsy. Human Mutation, 2017, 38, 1611-1612.	2.5	18
49	Genotyping <i>NUDT15</i> can predict the dose reduction of 6-MP for children with acute lymphoblastic leukemia especially at a preschool age. Journal of Human Genetics, 2016, 61, 797-801.	2.3	25
50	<i>De novo</i> non-synonymous <i>CTR9</i> variants are associated with motor delay and macrocephaly: human genetic and zebrafish experimental evidence. Human Molecular Genetics, 0, , .	2.9	0