Hisato Suzuki

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/6282474/publications.pdf

Version: 2024-02-01

		933447	888059
50	448	10	17
papers	citations	h-index	g-index

50	50	50	873
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Evaluation of Face2Gene using facial images of patients with congenital dysmorphic syndromes recruited in Japan. Journal of Human Genetics, 2019, 64, 789-794.	2.3	51
2	Clonal Expansion of Second-Hit Cells with Somatic Recombinations or C>T Transitions Form Porokeratosis in MVD or MVK Mutant Heterozygotes. Journal of Investigative Dermatology, 2019, 139, 2458-2466.e9.	0.7	49
3	Genotyping NUDT15 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
4	Poor outcomes in carriers of the RNF213 variant (p.Arg4810Lys) with pulmonary arterial hypertension. Journal of Heart and Lung Transplantation, 2020, 39, 103-112.	0.6	25
5	SOX17 Mutations in Japanese Patients with Pulmonary Arterial Hypertension. American Journal of Respiratory and Critical Care Medicine, 2018, 198, 1231-1233.	5.6	22
6	Truncating mutation in CSNK2B and myoclonic epilepsy. Human Mutation, 2017, 38, 1611-1612.	2.5	18
7	Schuursâ€Hoeijmakers syndrome in two patients from Japan. American Journal of Medical Genetics, Part A, 2019, 179, 341-343.	1.2	16
8	Genomic Comparison With Supercentenarians Identifies RNF213 as a Risk Gene for Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2018, 11, e002317.	3.6	14
9	Biallelic loss of OTUD7A causes severe muscular hypotonia, intellectual disability, and seizures. American Journal of Medical Genetics, Part A, 2021, 185, 1182-1186.	1.2	14
10	Severe Noonan syndrome phenotype associated with a germline Q71R MRAS variant: a recurrent substitution in RAS homologs in various cancers. American Journal of Medical Genetics, Part A, 2019, 179, 1628-1630.	1.2	13
11	Parallel detection of single nucleotide variants and copy number variants with exome analysis: Validation in a cohort of 700 undiagnosed patients. American Journal of Medical Genetics, Part A, 2020, 182, 2529-2532.	1.2	12
12	Preaxial polydactyly in an individual with Wiedemann-Steiner syndrome caused by a novel nonsense mutation in KMT2A. , 2017, 173, 2821-2825.		11
13	SATB2â€associated syndrome in patients from Japan: Linguistic profiles. American Journal of Medical Genetics, Part A, 2019, 179, 896-899.	1.2	11
14	Biallelic Mutations in the LSR Gene Cause a Novel Type of Infantile Intrahepatic Cholestasis. Journal of Pediatrics, 2020, 221, 251-254.	1.8	11
15	Role of chimeric transcript formation in the pathogenesis of birth defects. Congenital Anomalies (discontinued), 2021, 61, 76-81.	0.6	11
16	De novo NSF mutations cause early infantile epileptic encephalopathy. Annals of Clinical and Translational Neurology, 2019, 6, 2334-2339.	3.7	10
17	Consecutive medical exome analysis at a tertiary center: Diagnostic and healthâ€economic outcomes. American Journal of Medical Genetics, Part A, 2020, 182, 1601-1607.	1.2	10
18	IFT172 as the 19th gene causative of oralâ€facialâ€digital syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2510-2513.	1.2	9

#	Article	IF	Citations
19	CNOT2 haploinsufficiency causes a neurodevelopmental disorder with characteristic facial features. American Journal of Medical Genetics, Part A, 2019, 179, 2506-2509.	1.2	9
20	Recurrent <scp>NFIA K125E</scp> substitution represents a lossâ€ofâ€function allele: Sensitive in vitro and in vivo assays forÂnontruncating alleles. American Journal of Medical Genetics, Part A, 2021, 185, 2084-2093.	1.2	9
21	CNOT2 as the critical gene for phenotypes of 12q15 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 659-662.	1.2	8
22	Genome Analysis in Sick Neonates and Infants: High-yield Phenotypes and Contribution of Small Copy Number Variations. Journal of Pediatrics, 2022, 244, 38-48.e1.	1.8	8
23	Ablepharon and craniosynostosis in a patient with a localized TWIST1 basic domain substitution. American Journal of Medical Genetics, Part A, 2018, 176, 2777-2780.	1.2	7
24	Noninvasive diagnosis of TRIT1â€related mitochondrial disorder by measuring i⁶A37 and ms²i⁶A37 modifications in tRNAs from blood and urine samples. American Journal of Medical Genetics, Part A, 2019, 179, 1609-1614.	1.2	6
25	Haploinsufficiency of NCOR1 associated with autism spectrum disorder, scoliosis, and abnormal palatogenesis. American Journal of Medical Genetics, Part A, 2018, 176, 2466-2469.	1.2	5
26	A case of autism spectrum disorder with cleft lip and palate carrying a mutation in exon 8 of AUTS2. Clinical Case Reports (discontinued), 2019, 7, 2059-2063.	0.5	5
27	BCS1L mutations produce Fanconi syndrome with developmental disability. Journal of Human Genetics, 2022, 67, 143-148.	2.3	5
28	The p.<scp>Thr395Met</scp> missense variant of <scp>NFIA</scp> found in a patient with intellectual disability is a defective variant. American Journal of Medical Genetics, Part A, 2022, 188, 1184-1192.	1.2	5
29	RNF213-Associated Vascular Disease: A Concept Unifying Various Vasculopathies. Life, 2022, 12, 555.	2.4	5
30	A novel BBS10 mutation identified in a patient with Bardetâ€“Biedl syndrome with a violent emotional outbreak. Human Genome Variation, 2017, 4, 17033.	0.7	4
31	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
32	Severe course with lethal hepatocellular injury and skeletal muscular dysgenesis in a neonate with infantile liver failure syndrome type 1 caused by novel <scp>LARS1</scp> mutations. American Journal of Medical Genetics, Part A, 2021, 185, 866-870.	1.2	4
33	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 <scp> TLL1 </scp> as a causative gene for atrial septal defect. American Journal of Medical Genetics, Part A, 2022, , .	1.2	4
34	A novel missense PTEN mutation identified in a patient with macrocephaly and developmental delay. Human Genome Variation, 2019, 6, 25.	0.7	3
35	Establishing intellectual disability as the key feature of patients with biallelic <scp>RNPC3</scp> variants. American Journal of Medical Genetics, Part A, 2021, 185, 1836-1840.	1.2	3
36	Early diagnosis of lateral meningocele syndrome in an infant without neurological symptoms based on genomic analysis. Child's Nervous System, 2022, 38, 659-663.	1.1	3

#	Article	IF	Citations
37	Ketogenic Diet for KARS-Related Mitochondrial Dysfunction and Progressive Leukodystrophy. Neuropediatrics, 2022, 53, 065-068.	0.6	3
38	A patient with compound heterozygosity of <scp>SMPD4</scp>: Another example of utility of exomeâ€based copy number analysis in autosomal recessive disorders. American Journal of Medical Genetics, Part A, 2022, 188, 613-617.	1.2	3
39	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. Travel Medicine and Infectious Disease, 2021, 44, 102210.	3.0	3
40	Infantile neuroaxonal dystrophy in a pair of Malaysian siblings with progressive cerebellar atrophy: Description of an expanded phenotype with novel PLA2G6 variants. Journal of Clinical Neuroscience, 2020, 71, 289-292.	1.5	2
41	Novel ARX mutation identified in infantile spasm syndrome patient. Human Genome Variation, 2020, 7, 9.	0.7	2
42	Frequent FGFR3 and Ras Gene Mutations in Skin Tags or Acrochordons. Journal of Investigative Dermatology, 2021, 141, 2756-2760.e8.	0.7	2
43	Japanese pathogenic variant database: DPV. Translational Science of Rare Diseases, 2018, 3, 133-137.	1.5	1
44	De novo 2q36.3q37.1 deletion encompassing TRIP12 and NPPC yields distinct phenotypes. Human Genome Variation, 2020, 7, 19.	0.7	1
45	Forkâ€shaped mandibular incisors as a novel phenotype of LRP5 â€associated disorder. American Journal of Medical Genetics, Part A, 2021, 185, 1544-1549.	1.2	1
46	Noonan syndromeâ€like phenotype in a patient with heterozygous <scp>ERF</scp> truncating variant. Congenital Anomalies (discontinued), 2021, 61, 226-230.	0.6	1
47	Diagnosis of SLC25A46-related pontocerebellar hypoplasia in two siblings with fulminant neonatal course: role of postmortem CT and whole genomic analysis: a case report. BMC Neurology, 2022, 22, 20.	1.8	0
48	Phenotypic overlap between cardioacrofacial dysplasia-2 and oral-facial-digital syndrome. European Journal of Medical Genetics, 2022, , 104512.	1.3	0
49	Precocious puberty in a case of Simpsonâ€“Golabiâ€“Behmel syndrome with a de novo 240-kb deletion including GPC3. Human Genome Variation, 2022, 9, .	0.7	0
50	De novo non-synonymous CTR9 variants are associated with motor delay and macrocephaly: human genetic and zebrafish experimental evidence. Human Molecular Genetics, 0, , .	2.9	0