

Kazuki Yamazawa

List of Publications by Year in descending order

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Version: 2024-02-01

30
papers

1,354
citations

687363

13
h-index

526287

27
g-index

30
all docs

30
docs citations

30
times ranked

2366
citing authors

#	ARTICLE	IF	CITATIONS
1	A novel pathogenic variant of the FH gene in a family with hereditary leiomyomatosis and renal cell carcinoma. <i>Human Genome Variation</i> , 2022, 9, 3.	0.7	3
2	Pathogenic Copy Number and Sequence Variants in Children Born SGA With Short Stature Without Imprinting Disorders. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, e3121-e3133.	3.6	7
3	Loss of imprinting of the human-specific imprinted gene <i>ZNF597</i> causes prenatal growth retardation and dysmorphic features: implications for phenotypic overlap with Silver-Russell syndrome. <i>Journal of Medical Genetics</i> , 2021, 58, 427-432.	3.2	6
4	Role of Imprinting Disorders in Short Children Born SGA and Silver-Russell Syndrome Spectrum. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, 802-813.	3.6	16
5	Metacarpophalangeal pattern profile analysis for a 3-month-old infant with Feingold syndrome 2. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 952-954.	1.2	0
6	A Japanese boy with double diagnoses of 2p15p16.1 microdeletion syndrome and RP2-associated retinal disorder. <i>Human Genome Variation</i> , 2021, 8, 46.	0.7	0
7	Compound heterozygous variants in the ABCG8 gene in a Japanese girl with sitosterolemia. <i>Human Genome Variation</i> , 2020, 7, 25.	0.7	3
8	Genome-wide methylation analysis in Silver-Russell syndrome, Temple syndrome, and Prader-Willi syndrome. <i>Clinical Epigenetics</i> , 2020, 12, 159.	4.1	7
9	Contribution of gene mutations to Silver-Russell syndrome phenotype: multigene sequencing analysis in 92 etiology-unknown patients. <i>Clinical Epigenetics</i> , 2020, 12, 86.	4.1	29
10	A Familial Case of a Whole Germline <i>CDC73</i> Deletion Discordant for Primary Hyperparathyroidism. <i>Hormone Research in Paediatrics</i> , 2019, 92, 56-63.	1.8	1
11	Molecular and clinical analyses of two patients with UPD(16)mat detected by screening 94 patients with Silver-Russell syndrome phenotype of unknown aetiology. <i>Journal of Medical Genetics</i> , 2019, 56, 413-418.	3.2	23
12	High-level heteroplasmy for the m.7445A>G mitochondrial DNA mutation can cause progressive sensorineural hearing loss in infancy. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2018, 108, 125-131.	1.0	6
13	Spontaneous intramural duodenal hematoma as the manifestation of Noonan syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 496-498.	1.2	4
14	Genetic heterogeneity of patients with suspected Silver-Russell syndrome: genome-wide copy number analysis in 82 patients without imprinting defects. <i>Clinical Epigenetics</i> , 2017, 9, 52.	4.1	15
15	H1foo Has a Pivotal Role in Qualifying Induced Pluripotent Stem Cells. <i>Stem Cell Reports</i> , 2016, 6, 825-833.	4.8	40
16	Allele-specific binding of ZFP57 in the epigenetic regulation of imprinted and non-imprinted monoallelic expression. <i>Genome Biology</i> , 2015, 16, 112.	8.8	150
17	In utero undernourishment perturbs the adult sperm methylome and intergenerational metabolism. <i>Science</i> , 2014, 345, 1255903.	12.6	535
18	A Pediatric Case of Antibiotic-Associated Hemorrhagic Colitis Caused by <i>Klebsiella Oxytoca</i> . <i>Global Pediatric Health</i> , 2014, 1, 2333794X1455052.	0.7	4

#	ARTICLE	IF	CITATIONS
19	Molecular and Clinical Studies in 138 Japanese Patients with Silver-Russell Syndrome. <i>PLoS ONE</i> , 2013, 8, e60105.	2.5	55
20	Mosaic upd(7)mat in a patient with Silver-Russell syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 465-468.	1.2	7
21	Neonatal intestinal volvulus and preduodenal portal vein associated with situs ambiguus: Report of a case. <i>Surgery Today</i> , 2011, 41, 726-729.	1.5	3
22	Androgenetic/biparental mosaicism in a girl with Beckwith-Wiedemann syndrome-like and upd(14)pat-like phenotypes. <i>Journal of Human Genetics</i> , 2011, 56, 91-93.	2.3	25
23	Uniparental disomy and human disease: An overview. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2010, 154C, 329-334.	1.6	151
24	Molecular and clinical findings and their correlations in Silver-Russell syndrome: implications for a positive role of IGF2 in growth determination and differential imprinting regulation of the IGF2-H19 domain in bodies and placentas. <i>Journal of Molecular Medicine</i> , 2008, 86, 1171-1181.	3.9	54
25	Monozygotic female twins discordant for Silver-Russell syndrome and hypomethylation of the H19-DMR. <i>Journal of Human Genetics</i> , 2008, 53, 950-955.	2.3	44
26	Placental hypoplasia in maternal uniparental disomy for chromosome 7. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 514-516.	1.2	11
27	Mutation and Polymorphism Analyses of <i>INS3</i> and <i>LGR8</i> ; and <i>GREAT</i> in 62 Japanese Patients with Cryptorchidism. <i>Hormone Research in Paediatrics</i> , 2007, 67, 73-76.	1.8	20
28	Silver-Russell syndrome in a girl born after in vitro fertilization: partial hypermethylation at the differentially methylated region of PEG1/MEST. <i>Journal of Assisted Reproduction and Genetics</i> , 2007, 24, 131-136.	2.5	121
29	Hypонатremia, Hypophosphatemia, and Hypouricemia in a Girl With Macrophage Activation Syndrome. <i>Pediatrics</i> , 2006, 118, 2557-2560.	2.1	14
30	Frequency and clinical characteristics of distinct etiologies in patients with Silver-Russell syndrome diagnosed based on the Netchine-Harbison clinical scoring system. <i>Journal of Human Genetics</i> , 0, , .	2.3	0