Kosei Hasegawa

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3234188/publications.pdf

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35	407	933447	794594
papers	citations	10 h-index	19 g-index

37	37	37	677
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A novel pathogenic variant p.<scp>Asp797Val</scp> in <scp>IFIH1</scp> in a Japanese boy with overlapping <scp>Singletonâ€Merten</scp> syndrome and <scp>Aicardiâ€GoutiÃ¨res</scp> syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 249-252.	1.2	2
2	Transient central diabetes insipidus after cranioplasty for craniosynostosis in an infant with septo-optic dysplasia. Clinical Pediatric Endocrinology, 2022, 31, 50-53.	0.8	0
3	The clinical course of Rathkeâ€™s cleft cysts in pediatric patients: impact on growth and pubertal development. Clinical Pediatric Endocrinology, 2022, 31, 38-43.	0.8	3
4	Thioredoxin interacting protein protects mice from fasting induced liver steatosis by activating ER stress and its downstream signaling pathways. Scientific Reports, 2022, 12, 4819.	3.3	4
5	Preclinical diagnosis and identification of the chimeric CYP11B1/CYP11B2 gene in two pediatric cases of a Japanese family with glucocorticoid-remediable aldosteronism. Hypertension Research, 2021, 44, 891-893.	2.7	0
6	A pediatric case of extrapontine myelinolysis due to rapid fluctuation of sodium concentration after craniopharyngioma surgery. Journal of the Japanese Society of Intensive Care Medicine, 2021, 28, 227-229.	0.0	0
7	Genetic analysis in Japanese patients with osteogenesis imperfecta: Genotype and phenotype spectra in 96 probands. Molecular Genetics & Genomic Medicine, 2021, 9, e1675.	1.2	5
8	Residual endogenous insulin secretion in Japanese children with type 1A diabetes. Clinical Pediatric Endocrinology, 2021, 30, 27-33.	0.8	5
9	Pyridoxal in the Cerebrospinal Fluid May Be a Better Indicator of Vitamin B6â€“dependent Epilepsy Than Pyridoxal 5â€²-Phosphate. Pediatric Neurology, 2020, 113, 33-41.	2.1	6
10	A case of pyridoxine-dependent epilepsy with novel ALDH7A1 mutations. Oxford Medical Case Reports, 2020, 2020, omaa008.	0.4	4
11	Clinical Practice Guidelines for Hypophosphatasia*. Clinical Pediatric Endocrinology, 2020, 29, 9-24.	0.8	28
12	Clinical Practice Guidelines for Achondroplasia*. Clinical Pediatric Endocrinology, 2020, 29, 25-42.	0.8	19
13	Laboratory changes during adrenocorticotropic hormone therapy associated with renal calcified lesions. Pediatrics International, 2020, 62, 587-592.	0.5	1
14	Achondroplasia. , 2019, , 145-154.		0
15	Novel <i>AVPR2</i> variant in a male infant with nephrogenic diabetes insipidus who showed delayed head control. Clinical Pediatric Endocrinology, 2019, 28, 155-158.	0.8	0
16	A Case of Nager Syndrome Diagnosed Before Birth. Acta Medica Okayama, 2019, 73, 273-277.	0.2	2
17	Incidence rate and characteristics of symptomatic vitamin D deficiency in children: a nationwide survey in Japan. Endocrine Journal, 2018, 65, 593-599.	1.6	12
18	Novel heterozygous mutation in <i>TBX1</i> in an infant with hypocalcemic seizures. Clinical Pediatric Endocrinology, 2018, 27, 159-164.	0.8	13

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19	Neonatal-Onset Hereditary Coproporphyria: A New Variant of Hereditary Coproporphyria. JIMD Reports, 2017, 37, 99-106.	1.5	1
20	Three cases of Japanese acromicric/geleophysic dysplasia with FBN1 mutations: a comparison of clinical and radiological features. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 117-121.	0.9	8
21	A novel mutation in the COL2A1 gene in a patient with Stickler syndrome type 1: a case report and review of the literature. Journal of Medical Case Reports, 2017, 11, 237.	0.8	22
22	Acanthosis nigricans in a Japanese boy with hypochondroplasia due to a K650T mutation in <i>FGFR3</i>. Clinical Pediatric Endocrinology, 2017, 26, 223-228.	0.8	7
23	A novel mutation p.Ser348Cys in FGFR3 causes achondroplasia. American Journal of Medical Genetics, Part A, 2016, 170, 1370-1372.	1.2	10
24	HDR syndrome in a Japanese girl with biliary atresia: a case report. BMC Pediatrics, 2016, 16, 14.	1.7	8
25	Changes in facial appearance from neonate to adult in 3-M syndrome patient with novel CUL7 gene mutations. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 241-6.	0.9	4
26	Japanese familial case with metaphyseal dysplasia, Schmid Type caused by the p.T555P mutation in the COL10A1 gene. Clinical Pediatric Endocrinology, 2015, 24, 33-36.	0.8	4
27	Hypophosphatemic osteomalacia and bone sclerosis caused by a novel homozygous mutation of the FAM20C gene in an elderly man with a mild variant of Raine syndrome. Bone, 2014, 67, 56-62.	2.9	59
28	Mutation spectrum of COL1A1 and COL1A2 genes in Indian patients with osteogenesis imperfecta. American Journal of Medical Genetics, Part A, 2014, 164, 1482-1489.	1.2	24
29	Children with shortâ€limbed short stature in pediatric endocrinological services in <scp>J</scp>apan. Pediatrics International, 2014, 56, 809-812.	0.5	7
30	Novel and recurrent PORCN gene mutations in almost unilateral and typical focal dermal hypoplasia patients. European Journal of Dermatology, 2013, 23, 64-67.	0.6	7
31	Urinary N-telopeptides of type I collagen in healthy children. Pediatrics International, 2010, 52, 398-401.	0.5	6
32	Heterozygous Orthodenticle Homeobox 2 Mutations Are Associated with Variable Pituitary Phenotype. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 756-764.	3.6	98
33	Growth of infants with osteogenesis imperfecta treated with bisphosphonate. Pediatrics International, 2009, 51, 54-58.	0.5	11
34	Impaired pyridinoline cross-link formation in patients with osteogenesis imperfecta. Journal of Bone and Mineral Metabolism, 2008, 26, 394-399.	2.7	5
35	Mutations in type I collagen genes in Japanese osteogenesis imperfecta patients. Pediatrics International, 2007, 49, 564-569.	0.5	21