Yousuke Higuchi

List of Publications by Year
in descending order

21 papers	126 citations	1307366 7 h-index	1281743 11 g-index
21 all docs	21 docs citations	21 times ranked	233 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.	0.7	2
2	The clinical course of Rathkeâ€™s cleft cysts in pediatric patients: impact on growth and pubertal development. Clinical Pediatric Endocrinology, 2022, 31, 38-43.	0.4	3
3	A 9â€yearâ€old boy with severe motor and intellectual disabilities and prolonged abdominal distension. Journal of Paediatrics and Child Health, 2022, 58, 361-361.	0.4	0
4	A 4â€monthâ€old female infant with breast development and virilisation. Journal of Paediatrics and Child Health, 2022, , .	0.4	0
5	A 9â€yearâ€old boy with severe motor and intellectual disabilities and prolonged abdominal distension. Journal of Paediatrics and Child Health, 2021, , .	0.4	0
6	Genetic analysis in Japanese patients with osteogenesis imperfecta: Genotype and phenotype spectra in 96 probands. Molecular Genetics & Genomic Medicine, 2021, 9, e1675.	0.6	5
7	Infantile pyknocytosis with marked hemolytic anemia. Pediatrics and Neonatology, 2021, 62, 563-564.	0.3	0
8	A 9-year-old girl with Kawasaki disease and pulmonary nodules. Clinical Rheumatology, 2020, 39, 3139-3140.	1.0	5
9	Effect of growth hormone therapy on thyroid function in isolated growth hormone deficient and short small for gestational age children: a two-year study, including on assessment of the usefulness of the thyrotropin-releasing hormone (TRH) stimulation test. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 1417-1423.	0.4	5
10	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.	0.4	0
11	Novel heterozygous mutation in <i>TBX1</i> in an infant with hypocalcemic seizures. Clinical Pediatric Endocrinology, 2018, 27, 159-164.	0.4	13
12	Neonatal-Onset Hereditary Coproporphyria: A New Variant of Hereditary Coproporphyria. JIMD Reports, 2017, 37, 99-106.	0.7	1
13	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.	0.4	8
14	Clinical Epidemiology and Treatment of Febrile and Afebrile Convulsions With Mild Gastroenteritis: A Multicenter Study. Pediatric Neurology, 2017, 67, 78-84.	1.0	18
15	Sorafenib treatment for papillary thyroid carcinoma with diffuse lung metastases in a child with autism spectrum disorder: a case report. BMC Cancer, 2017, 17, 775.	1.1	6
16	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.	0.4	22
17	A novel mutation p.Ser348Cys in FGFR3 causes achondroplasia. American Journal of Medical Genetics, Part A, 2016, 170, 1370-1372.	0.7	10
18	HDR syndrome in a Japanese girl with biliary atresia: a case report. BMC Pediatrics, 2016, 16, 14.	0.7	8

#	Article	IF	Citations
19	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.	0.4	4
20	Urinary Cross-linked N-terminal Telopeptide of Type I Collagen Levels of Infants with Osteogenesis Imperfecta and Healthy Infants. Acta Medica Okayama, 2016, 70, 435-439.	0.1	1
21	The identification of a novel splicing mutation in C1qB in a Japanese family with C1q deficiency: a case report. Pediatric Rheumatology, 2013, 11, 41.	0.9	15