Koh-ichiro

List of Publications by Year
in descending order

38 papers	932 citations	516710 16 h-index	454955 30 g-index
39 all docs	39 docs citations	39 times ranked	1854 citing authors

#	Article	IF	Citations
1	Proteasome assembly defect due to a proteasome subunit beta type 8 (PSMB8) mutation causes the autoinflammatory disorder, Nakajo-Nishimura syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 14914-14919.	7.1	288
2	LRP5, low-density-lipoprotein-receptor-related protein 5, is a determinant for bone mineral density. Journal of Human Genetics, 2004, 49, 80-86.	2.3	126
3	Nonsense mutation in CFAP43 causes normal-pressure hydrocephalus with ciliary abnormalities. Neurology, 2019, 92, e2364-e2374.	1.1	65
4	Familial Mediterranean fever is no longer a rare disease in Japan. Arthritis Research and Therapy, 2016, 18, 175.	3.5	63
5	Paroxysmal kinesigenic choreoathetosis (PKC): confirmation of linkage to 16p11-q21, but unsuccessful detection of mutations among 157 genes at the PKC-critical region in seven PKC families. Journal of Human Genetics, 2007, 52, 334-341.	2.3	50
6	Novel compound heterozygous DNA ligase IV mutations in an adolescent with a slowly-progressing radiosensitive-severe combined immunodeficiency. Clinical Immunology, 2015, 160, 255-260.	3.2	29
7	Genomeâ€wide association study of HPVâ€associated cervical cancer in Japanese women. Journal of Medical Virology, 2014, 86, 1153-1158.	5.0	27
8	Uniparental disomy analysis in trios using genome-wide SNP array and whole-genome sequencing data imply segmental uniparental isodisomy in general populations. Gene, 2013, 512, 267-274.	2.2	26
9	Effects of HLA-DRB1 alleles on susceptibility and clinical manifestations in Japanese patients with adult onset Stillâ€™s disease. Arthritis Research and Therapy, 2017, 19, 199.	3.5	25
10	Agile parallel bioinformatics workflow management using Pwrake. BMC Research Notes, 2011, 4, 331.	1.4	20
11	Japanese founder duplications/triplications involving BHLHA9 are associated with split-hand/foot malformation with or without long bone deficiency and Gollop-Wolfgang complex. Orphanet Journal of Rare Diseases, 2014, 9, 125.	2.7	20
12	Analysis of clinical symptoms and <scp>ABCC</scp>6 mutations in 76 Japanese patients with pseudoxanthoma elasticum. Journal of Dermatology, 2017, 44, 644-650.	1.2	20
13	Detection of de novo single nucleotide variants in offspring of atomic-bomb survivors close to the hypocenter by whole-genome sequencing. Journal of Human Genetics, 2018, 63, 357-363.	2.3	20
14	Heterozygous missense variant of the proteasome subunit Î²-type 9 causes neonatal-onset autoinflammation and immunodeficiency. Nature Communications, 2021, 12, 6819.	12.8	20
15	Expression of the Snurfâ€“Snrpn IC transcript in the oocyte and its putative role in the imprinting establishment of the mouse 7C imprinting domain. Journal of Human Genetics, 2006, 51, 236-243.	2.3	18
16	Genotype Analyses in the Japanese and Belarusian Populations Reveal Independent Effects of rs965513 and rs1867277 but Do Not Support the Role of FOXE1 Polyalanine Tract Length in Conferring Risk for Papillary Thyroid Carcinoma. Thyroid, 2017, 27, 224-235.	4.5	18
17	Genome-wide linkage analysis and mutation analysis of hereditary congenital blepharoptosis in a Japanese family. Journal of Human Genetics, 2008, 53, 34-41.	2.3	14
18	Clinical and histological findings of autosomal dominant renalâ€limited disease with LMX1B mutation. Nephrology, 2016, 21, 765-773.	1.6	12

#	Article	IF	Citations
19	Detailed analysis of 26 cases of 1q partial duplication/triplication syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 908-917.	1.2	11
20	Targeting Adaptive IRE1Î± Signaling and PLK2 in Multiple Myeloma: Possible Anti-Tumor Mechanisms of KIRA8 and Nilotinib. International Journal of Molecular Sciences, 2020, 21, 6314.	4.1	9
21	Itpr1 regulates the formation of anterior eye segment tissues derived from neural crest cells. Development (Cambridge), 2021, 148, .	2.5	9
22	Bile extracellular vesicles from end-stage liver disease patients show altered microRNA content. Hepatology International, 2021, 15, 821-830.	4.2	7
23	KN3014, a piperidine-containing small compound, inhibits auto-secretion of IL-1Î² from PBMCs in a patient with Muckleâ€“Wells syndrome. Scientific Reports, 2020, 10, 13562.	3.3	6
24	BCS1L mutations produce Fanconi syndrome with developmental disability. Journal of Human Genetics, 2022, 67, 143-148.	2.3	5
25	Auto-immune disorders in a child with PIK3CD variant and 22q13 deletion. European Journal of Medical Genetics, 2018, 61, 631-633.	1.3	4
26	Identification of a novel CCDC22 mutation in a patient with severe Epsteinâ€“Barr virus-associated hemophagocytic lymphohistiocytosis and aggressive natural killer cell leukemia. International Journal of Hematology, 2019, 109, 744-750.	1.6	4
27	Expression of unfolded protein response markers in the pheochromocytoma with Waardenburg syndrome: a case report. BMC Endocrine Disorders, 2020, 20, 90.	2.2	4
28	Brothers with novel compound heterozygous mutations in COL27A1 causing dental and genital abnormalities. European Journal of Medical Genetics, 2021, 64, 104125.	1.3	3
29	A case of ezetimibe-effective hypercholesterolemia with a novel heterozygous variant in <i>ABCG5</i>. Endocrine Journal, 2020, 67, 1099-1105.	1.6	3
30	Clonal dynamics in a case of acute monoblastic leukemia that later developed myeloproliferative neoplasm. International Journal of Hematology, 2018, 108, 213-217.	1.6	2
31	Large deletion in 6q containing the TNFAIP3 gene associated with autoimmune lymphoproliferative syndrome. Clinical Immunology, 2022, 235, 108853.	3.2	1
32	Sphenoethmoidal meningoencephalocele with variable hypopituitarism: A case report and review of literature. Clinical Pediatric Endocrinology, 2020, 29, 183-187.	0.8	1
33	Atypical phenotype without fever in a Japanese family with an autosomal dominant transmission of familial Mediterranean fever due to heterozygous MEFV Thr577Asn mutations. Clinical and Experimental Rheumatology, 2019, 37 Suppl 121, 161-162.	0.8	1
34	No mutation in RASâ€MAPK pathway genes in 30 patients with Kabuki syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 1893-1896.	1.2	0
35	A unique missense variant in the E1A-binding protein P400 gene is implicated in schizophrenia by whole-exome sequencing and mutant mouse models. Translational Psychiatry, 2021, 11, 132.	4.8	0
36	Clonal Evolution Underlying the Progression from Myelodysplastic Syndromes to Ph-Positive Acute Lymphoblastic Leukemia. Blood, 2016, 128, 5514-5514.	1.4	0

#	Article	IF	Citations
37	Next-generation sequencing of the whole MEFV gene in Japanese patients with familial Mediterranean fever: a case-control association study. Clinical and Experimental Rheumatology, 2020, 38 Suppl 127, 35-41.	0.8	0
38	Simultaneous monitoring of oxygen and carbon dioxide for Pittâ€Hopkins syndrome. Pediatrics International, 2022, 64, e15180.	0.5	0