Masakazu Kohda

List of Publications by Year
in descending order

41 papers	1,593 citations	393982 19 h-index	301761 39 g-index
43 all docs	43 docs citations	43 times ranked	3041 citing authors

#	Article	IF	Citations
1	Immunohistochemical staining patterns of p53 predict the mutational status of TP53 in oral epithelial dysplasia. Modern Pathology, 2022, 35, 177-185.	2.9	13
2	Prenatal diagnosis of severe mitochondrial diseases caused by nuclear gene defects: a study in Japan. Scientific Reports, 2021, 11, 3531.	1.6	1
3	A high mutation load of m.14597A>G in MT-ND6 causes Leigh syndrome. Scientific Reports, 2021, 11, 11123.	1.6	8
4	Genome sequencing and RNAâ€seq analyses of mitochondrial complex I deficiency revealed Alu insertionâ€mediated deletion in NDUFV2. Human Mutation, 2021, 42, 1422-1428.	1.1	4
5	Long-term prognosis and genetic background of cardiomyopathy in 223 pediatric mitochondrial disease patients. International Journal of Cardiology, 2021, 341, 48-55.	0.8	14
6	Valine metabolites analysis in ECHS1 deficiency. Molecular Genetics and Metabolism Reports, 2021, 29, 100809.	0.4	9
7	A novel homozygous variant in MICOS13/QIL1 causes hepatoâ€encephalopathy with mitochondrial DNA depletion syndrome. Molecular Genetics & Genomic Medicine, 2020, 8, e1427.	0.6	12
8	Leigh Syndrome Due to NDUFV1 Mutations Initially Presenting as LBSL. Genes, 2020, 11, 1325.	1.0	8
9	Mortality of Japanese patients with Leigh syndrome: Effects of age at onset and genetic diagnosis. Journal of Inherited Metabolic Disease, 2020, 43, 819-826.	1.7	32
10	Ski3/TTC37 deficiency associated with trichohepatoenteric syndrome causes mitochondrial dysfunction in Drosophila. FEBS Letters, 2020, 594, 2168-2181.	1.3	4
11	Mitochondrial complex deficiency by novel compound heterozygous <scp>TMEM</scp>70 variants and correlation with developmental delay, undescended testicle, and left ventricular noncompaction in a Japanese patient: A case report. Clinical Case Reports (discontinued), 2019, 7, 553-557.	0.2	11
12	Mitochondrial ribosomal protein PTCD3 mutations cause oxidative phosphorylation defects with Leigh syndrome. Neurogenetics, 2019, 20, 9-25.	0.7	46
13	Cardiomyopathy in children with mitochondrial disease: Prognosis and genetic background. International Journal of Cardiology, 2019, 279, 115-121.	0.8	35
14	Leigh syndrome with spinal cord involvement due to a hemizygous NDUFA1 mutation. Brain and Development, 2018, 40, 498-502.	0.6	15
15	Characteristics of MUTYH variants in Japanese colorectal polyposis patients. International Journal of Clinical Oncology, 2018, 23, 497-503.	1.0	10
16	Barth Syndrome: Different Approaches to Diagnosis. Journal of Pediatrics, 2018, 193, 256-260.	0.9	14
17	Identification of the Coiled-Coil Domain as an Essential Methyl-CpG-Binding Domain Protein 3 Element for Preserving Lineage Commitment Potential of Embryonic Stem Cells. Stem Cells, 2018, 36, 1355-1367.	1.4	7
18	Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. American Journal of Human Genetics, 2018, 103, 221-231.	2.6	65

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19	A novel mutation in TAZ causes mitochondrial respiratory chain disorder without cardiomyopathy. Journal of Human Genetics, 2017, 62, 539-547.	1.1	5
20	Clinical validity of biochemical and molecular analysis in diagnosing Leigh syndrome: a study of 106 Japanese patients. Journal of Inherited Metabolic Disease, 2017, 40, 685-693.	1.7	78
21	Biallelic C1QBP Mutations Cause Severe Neonatal-, Childhood-, or Later-Onset Cardiomyopathy Associated with Combined Respiratory-Chain Deficiencies. American Journal of Human Genetics, 2017, 101, 525-538.	2.6	58
22	HDR-del: A tool based on Hamming distance for prioritizing pathogenic chromosomal deletions in exome sequencing. Human Mutation, 2017, 38, 1796-1800.	1.1	6
23	ATAD3 gene cluster deletions cause cerebellar dysfunction associated with altered mitochondrial DNA and cholesterol metabolism. Brain, 2017, 140, 1595-1610.	3.7	105
24	Identification of a Japanese Lynch syndrome patient with large deletion in the 3â€² region of theEPCAMgene. Japanese Journal of Clinical Oncology, 2016, 46, hyv172.	0.6	7
25	A Comprehensive Genomic Analysis Reveals the Genetic Landscape of Mitochondrial Respiratory Chain Complex Deficiencies. PLoS Genetics, 2016, 12, e1005679.	1.5	236
26	Dried blood spots for newborn screening allows easy determination of a high heteroplasmy rate in severe infantile cardiomyopathy. International Journal of Cardiology, 2016, 221, 446-449.	0.8	4
27	HDR: a statistical two-step approach successfully identifies disease genes in autosomal recessive families. Journal of Human Genetics, 2016, 61, 959-963.	1.1	11
28	Rapid detection of germline mutations for hereditary gastrointestinal polyposis/cancers using HaloPlex target enrichment and high-throughput sequencing technologies. Familial Cancer, 2016, 15, 553-562.	0.9	21
29	Rapidly progressive infantile cardiomyopathy with mitochondrial respiratory chain complex V deficiency due to loss of ATPase 6 and 8 protein. International Journal of Cardiology, 2016, 207, 203-205.	0.8	23
30	Deficiency of <scp>ECHS</scp>1 causes mitochondrial encephalopathy with cardiac involvement. Annals of Clinical and Translational Neurology, 2015, 2, 492-509.	1.7	90
31	COQ4 Mutations Cause a Broad Spectrum of Mitochondrial Disorders Associated with CoQ10 Deficiency. American Journal of Human Genetics, 2015, 96, 309-317.	2.6	86
32	Myocerebrohepatopathy spectrum disorder due to POLG mutations: A clinicopathological report. Brain and Development, 2015, 37, 719-724.	0.6	13
33	Intra-mitochondrial Methylation Deficiency Due to Mutations in SLC25A26. American Journal of Human Genetics, 2015, 97, 761-768.	2.6	58
34	Mutations in GTPBP3 Cause a Mitochondrial Translation Defect Associated with Hypertrophic Cardiomyopathy, Lactic Acidosis, and Encephalopathy. American Journal of Human Genetics, 2014, 95, 708-720.	2.6	123
35	A Genome-Wide Association Study for Diabetic Retinopathy in a Japanese Population: Potential Association with a Long Intergenic Non-Coding RNA. PLoS ONE, 2014, 9, e111715.	1.1	81
36	Homozygosity Mapping on Homozygosity Haplotype Analysis to Detect Recessive Disease-Causing Genes from a Small Number of Unrelated, Outbred Patients. PLoS ONE, 2011, 6, e25059.	1.1	9

#	Article	IF	Citations
37	A quantitatively-modeled homozygosity mapping algorithm, qHomozygosityMapping, utilizing whole genome single nucleotide polymorphism genotyping data. BMC Bioinformatics, 2010, 11, S5.	1.2	9
38	Knockdown of COPA, Identified by Loss-of-Function Screen, Induces Apoptosis and Suppresses Tumor Growth in Mesothelioma Mouse Model. Genomics, 2010, 95, 210-216.	1.3	59
39	Homozygosity Haplotype Allows a Genomewide Search for the Autosomal Segments Shared among Patients. American Journal of Human Genetics, 2007, 80, 1090-1102.	2.6	59
40	Association of the HTRA1 gene variant with age-related macular degeneration in the Japanese population. Journal of Human Genetics, 2007, 52, 636-641.	1.1	55
41	Frequent loss of imprinting ofIGF2 andMEST in lung adenocarcinoma. Molecular Carcinogenesis, 2001, 31, 184-191.	1.3	83