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	A Comprehensive Genomic Analysis Reveals the Genetic Landscape of Mitochondrial Respiratory Chain Complex Deficiencies. PLoS Genetics, 2016, 12, e1005679.	1.5	236
2	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
3	ATAD3 gene cluster deletions cause cerebellar dysfunction associated with altered mitochondrial DNA and cholesterol metabolism. Brain, 2017, 140, 1595-1610.	3.7	105
4	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
5	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
6	Frequent loss of imprinting ofIGF2 andMEST in lung adenocarcinoma. Molecular Carcinogenesis, 2001, 31, 184-191.	1.3	83
7	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
8	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
9	Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. American Journal of Human Genetics, 2018, 103, 221-231.	2.6	65
10	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
11	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
12	Intra-mitochondrial Methylation Deficiency Due to Mutations in SLC25A26. American Journal of Human Genetics, 2015, 97, 761-768.	2.6	58
13	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
14	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
15	Mitochondrial ribosomal protein PTCD3 mutations cause oxidative phosphorylation defects with Leigh syndrome. Neurogenetics, 2019, 20, 9-25.	0.7	46
16	Cardiomyopathy in children with mitochondrial disease: Prognosis and genetic background. International Journal of Cardiology, 2019, 279, 115-121.	0.8	35
17	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
18	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

#	Article	IF	Citations
19	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
20	Leigh syndrome with spinal cord involvement due to a hemizygous NDUFA1 mutation. Brain and Development, 2018, 40, 498-502.	0.6	15
21	Barth Syndrome: Different Approaches to Diagnosis. Journal of Pediatrics, 2018, 193, 256-260.	0.9	14
22	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
23	Myocerebrohepatopathy spectrum disorder due to POLG mutations: A clinicopathological report. Brain and Development, 2015, 37, 719-724.	0.6	13
24	Immunohistochemical staining patterns of p53 predict the mutational status of TP53 in oral epithelial dysplasia. Modern Pathology, 2022, 35, 177-185.	2.9	13
25	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
26	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
27	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
28	Characteristics of MUTYH variants in Japanese colorectal polyposis patients. International Journal of Clinical Oncology, 2018, 23, 497-503.	1.0	10
29	A quantitatively-modeled homozygosity mapping algorithm, qHomozygosityMapping, utilizing whole genome single nucleotide polymorphism genotyping data. BMC Bioinformatics, 2010, 11, S5.	1.2	9
30	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
31	Valine metabolites analysis in ECHS1 deficiency. Molecular Genetics and Metabolism Reports, 2021, 29, 100809.	0.4	9
32	Leigh Syndrome Due to NDUFV1 Mutations Initially Presenting as LBSL. Genes, 2020, 11, 1325.	1.0	8
33	A high mutation load of m.14597A>G in MT-ND6 causes Leigh syndrome. Scientific Reports, 2021, 11, 11123.	1.6	8
34	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
35	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
36	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

#	Article	IF	Citations
37	A novel mutation in TAZ causes mitochondrial respiratory chain disorder without cardiomyopathy. Journal of Human Genetics, 2017, 62, 539-547.	1.1	5
38	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
39	Ski3/TTC37 deficiency associated with trichohepatoenteric syndrome causes mitochondrial dysfunction in Drosophila. FEBS Letters, 2020, 594, 2168-2181.	1.3	4
40	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
41	Prenatal diagnosis of severe mitochondrial diseases caused by nuclear gene defects: a study in Japan. Scientific Reports, 2021, 11, 3531.	1.6	1