Yoshihito Kishita

List of Publications by Year
in descending order

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41 papers	1,432 citations	430874 18 h-index	345221 36 g-index
46 all docs	46 docs citations	46 times ranked	2333 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.	3.5	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.	6.2	123
3	Metabolic and chemical regulation of tRNA modification associated with taurine deficiency and human disease. Nucleic Acids Research, 2018, 46, 1565-1583.	14.5	110
4	Deficiency of <scp>ECHS</scp>1 causes mitochondrial encephalopathy with cardiac involvement. Annals of Clinical and Translational Neurology, 2015, 2, 492-509.	3.7	90
5	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	8.2	85
6	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.	3.6	78
7	Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. American Journal of Human Genetics, 2018, 103, 221-231.	6.2	65
8	Intra-mitochondrial Methylation Deficiency Due to Mutations in SLC25A26. American Journal of Human Genetics, 2015, 97, 761-768.	6.2	58
9	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.	6.2	58
10	Mitochondrial ribosomal protein PTCD3 mutations cause oxidative phosphorylation defects with Leigh syndrome. Neurogenetics, 2019, 20, 9-25.	1.4	46
11	Cardiomyopathy in children with mitochondrial disease: Prognosis and genetic background. International Journal of Cardiology, 2019, 279, 115-121.	1.7	35
12	Loss of Trxâ€2 enhances oxidative stressâ€dependent phenotypes in Drosophila. FEBS Letters, 2010, 584, 3398-3401.	2.8	34
13	Impaired energy metabolism in a Drosophila model of mitochondrial aconitase deficiency. Biochemical and Biophysical Research Communications, 2013, 433, 145-150.	2.1	33
14	Fatal Perinatal Mitochondrial Cardiac Failure Caused by Recurrent De Novo Duplications in the ATAD3 Locus. Med, 2021, 2, 49-73.e10.	4.4	33
15	Mortality of Japanese patients with Leigh syndrome: Effects of age at onset and genetic diagnosis. Journal of Inherited Metabolic Disease, 2020, 43, 819-826.	3.6	32
16	Clinical and molecular basis of hepatocerebral mitochondrial DNA depletion syndrome in Japan: evaluation of outcomes after liver transplantation. Orphanet Journal of Rare Diseases, 2020, 15, 169.	2.7	29
17	<scp>MSH</scp>1 maintains organelle genome stability and genetically interacts with <scp>RECA</scp> and <scp>RECG</scp> in the moss Physcomitrella patens. Plant Journal, 2017, 91, 455-465.	5.7	27
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.	1.7	23

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19	Impaired fatty acid oxidation in a Drosophila model of mitochondrial trifunctional protein (MTP) deficiency. Biochemical and Biophysical Research Communications, 2012, 419, 344-349.	2.1	20
20	A simple method for sequencing the whole human mitochondrial genome directly from samples and its application to genetic testing. Scientific Reports, 2019, 9, 17411.	3.3	20
21	Effects of 5-aminolevulinic acid and sodium ferrous citrate on fibroblasts from individuals with mitochondrial diseases. Scientific Reports, 2019, 9, 10549.	3.3	19
22	A homozygous variant in <scp>NDUFA8</scp> is associated with developmental delay, microcephaly, and epilepsy due to mitochondrial complex I deficiency. Clinical Genetics, 2020, 98, 155-165.	2.0	18
23	Leigh syndrome with spinal cord involvement due to a hemizygous NDUFA1 mutation. Brain and Development, 2018, 40, 498-502.	1.1	15
24	Barth Syndrome: Different Approaches to Diagnosis. Journal of Pediatrics, 2018, 193, 256-260.	1.8	14
25	Long-term prognosis and genetic background of cardiomyopathy in 223 pediatric mitochondrial disease patients. International Journal of Cardiology, 2021, 341, 48-55.	1.7	14
26	A novel homozygous variant in MICOS13/QIL1 causes hepatoâ€encephalopathy with mitochondrial DNA depletion syndrome. Molecular Genetics & Genomic Medicine, 2020, 8, e1427.	1.2	12
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.	0.5	11
28	Neonatal-onset mitochondrial disease: clinical features, molecular diagnosis and prognosis. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2022, 107, 329-334.	2.8	9
29	Valine metabolites analysis in ECHS1 deficiency. Molecular Genetics and Metabolism Reports, 2021, 29, 100809.	1.1	9
30	Early infantile-onset Leigh syndrome complicated with infantile spasms associated with the m.9185â€Tâ€>â€C variant in the MT-ATP6 gene: Expanding the clinical spectrum. Brain and Development, 2020, 42, 69-72.	1.1	8
31	Leigh Syndrome Due to NDUFV1 Mutations Initially Presenting as LBSL. Genes, 2020, 11, 1325.	2.4	8
32	A high mutation load of m.14597A>G in MT-ND6 causes Leigh syndrome. Scientific Reports, 2021, 11, 11123.	3.3	8
33	A case report of adult-onset COQ8B nephropathy presenting focal segmental glomerulosclerosis with granular swollen podocytes. BMC Nephrology, 2020, 21, 376.	1.8	7
34	NAD(P)HX dehydratase protein-truncating mutations are associated with neurodevelopmental disorder exacerbated by acute illness. Brain, 2020, 143, e54-e54.	7.6	7
35	A novel mutation in TAZ causes mitochondrial respiratory chain disorder without cardiomyopathy. Journal of Human Genetics, 2017, 62, 539-547.	2.3	5
36	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.	1.7	4

#	Article	IF	Citations
37	Ski3/TTC37 deficiency associated with trichohepatoenteric syndrome causes mitochondrial dysfunction in Drosophila. FEBS Letters, 2020, 594, 2168-2181.	2.8	4
38	Genome sequencing and RNAâ€seq analyses of mitochondrial complex I deficiency revealed Alu insertionâ€mediated deletion in NDUFV2. Human Mutation, 2021, 42, 1422-1428.	2.5	4
39	Trigenic ADH5/ALDH2/ADGRV1 mutations in myelodysplasia with Usher syndrome. Heliyon, 2021, 7, e07804.	3.2	2
40	Development of Leigh syndrome with a high probability of cardiac manifestations in infantile-onset patients with m.14453GÂ>ÂA. Mitochondrion, 2022, 63, 1-8.	3.4	2
41	Prenatal diagnosis of severe mitochondrial diseases caused by nuclear gene defects: a study in Japan. Scientific Reports, 2021, 11, 3531.	3.3	1