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	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
2	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
3	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	8.2	85
4	Fatal Perinatal Mitochondrial Cardiac Failure Caused by Recurrent De Novo Duplications in the ATAD3 Locus. Med, 2021, 2, 49-73.e10.	4.4	33
5	Prenatal diagnosis of severe mitochondrial diseases caused by nuclear gene defects: a study in Japan. Scientific Reports, 2021, 11, 3531.	3.3	1
6	A high mutation load of m.14597A>G in MT-ND6 causes Leigh syndrome. Scientific Reports, 2021, 11, 11123.	3.3	8
7	Trigenic ADH5/ALDH2/ADGRV1 mutations in myelodysplasia with Usher syndrome. Heliyon, 2021, 7, e07804.	3.2	2
8	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
9	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
10	Valine metabolites analysis in ECHS1 deficiency. Molecular Genetics and Metabolism Reports, 2021, 29, 100809.	1.1	9
11	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
12	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
13	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
14	A case report of adult-onset COQ8B nephropathy presenting focal segmental glomerulosclerosis with granular swollen podocytes. BMC Nephrology, 2020, 21, 376.	1.8	7
15	Leigh Syndrome Due to NDUFV1 Mutations Initially Presenting as LBSL. Genes, 2020, 11, 1325.	2.4	8
16	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
17	NAD(P)HX dehydratase protein-truncating mutations are associated with neurodevelopmental disorder exacerbated by acute illness. Brain, 2020, 143, e54-e54.	7.6	7
18	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

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19	Ski3/TTC37 deficiency associated with trichohepatoenteric syndrome causes mitochondrial dysfunction in Drosophila. FEBS Letters, 2020, 594, 2168-2181.	2.8	4
20	Effects of 5-aminolevulinic acid and sodium ferrous citrate on fibroblasts from individuals with mitochondrial diseases. Scientific Reports, 2019, 9, 10549.	3.3	19
21	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
22	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
23	Mitochondrial ribosomal protein PTCD3 mutations cause oxidative phosphorylation defects with Leigh syndrome. Neurogenetics, 2019, 20, 9-25.	1.4	46
24	Cardiomyopathy in children with mitochondrial disease: Prognosis and genetic background. International Journal of Cardiology, 2019, 279, 115-121.	1.7	35
25	Leigh syndrome with spinal cord involvement due to a hemizygous NDUFA1 mutation. Brain and Development, 2018, 40, 498-502.	1.1	15
26	Metabolic and chemical regulation of tRNA modification associated with taurine deficiency and human disease. Nucleic Acids Research, 2018, 46, 1565-1583.	14.5	110
27	Barth Syndrome: Different Approaches to Diagnosis. Journal of Pediatrics, 2018, 193, 256-260.	1.8	14
28	Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. American Journal of Human Genetics, 2018, 103, 221-231.	6.2	65
29	A novel mutation in TAZ causes mitochondrial respiratory chain disorder without cardiomyopathy. Journal of Human Genetics, 2017, 62, 539-547.	2.3	5
30	<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
31	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
32	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
33	A Comprehensive Genomic Analysis Reveals the Genetic Landscape of Mitochondrial Respiratory Chain Complex Deficiencies. PLoS Genetics, 2016, 12, e1005679.	3.5	236
34	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
35	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
36	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

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37	Intra-mitochondrial Methylation Deficiency Due to Mutations in SLC25A26. American Journal of Human Genetics, 2015, 97, 761-768.	6.2	58
38	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
39	Impaired energy metabolism in a Drosophila model of mitochondrial aconitase deficiency. Biochemical and Biophysical Research Communications, 2013, 433, 145-150.	2.1	33
40	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
41	Loss of Trxâ€2 enhances oxidative stressâ€dependent phenotypes in Drosophila. FEBS Letters, 2010, 584, 3398-3401.	2.8	34