Akira Ohtake

List of Publications by Year
in descending order

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78 papers	2,213 citations	236925 25 h-index	254184 43 g-index
86 all docs	86 docs citations	86 times ranked	3715 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	ATAD3 gene cluster deletions cause cerebellar dysfunction associated with altered mitochondrial DNA and cholesterol metabolism. Brain, 2017, 140, 1595-1610.	7.6	105
5	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
6	COQ4 Mutations Cause a Broad Spectrum of Mitochondrial Disorders Associated with CoQ10 Deficiency. American Journal of Human Genetics, 2015, 96, 309-317.	6.2	86
7	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	8.2	85
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.	3.6	78
9	Biallelic IARS Mutations Cause Growth Retardation with Prenatal Onset, Intellectual Disability, Muscular Hypotonia, and Infantile Hepatopathy. American Journal of Human Genetics, 2016, 99, 414-422.	6.2	73
10	Guidance for Pediatric Familial Hypercholesterolemia 2017. Journal of Atherosclerosis and Thrombosis, 2018, 25, 539-553.	2.0	68
11	Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. American Journal of Human Genetics, 2018, 103, 221-231.	6.2	65
12	Intra-mitochondrial Methylation Deficiency Due to Mutations in SLC25A26. American Journal of Human Genetics, 2015, 97, 761-768.	6.2	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.	6.2	58
14	MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. EBioMedicine, 2018, 30, 86-93.	6.1	47
15	Loss of the Mitochondrial Fatty Acid Î²-Oxidation Protein Medium-Chain Acyl-Coenzyme A Dehydrogenase Disrupts Oxidative Phosphorylation Protein Complex Stability and Function. Scientific Reports, 2018, 8, 153.	3.3	47
16	Mitochondrial ribosomal protein PTCD3 mutations cause oxidative phosphorylation defects with Leigh syndrome. Neurogenetics, 2019, 20, 9-25.	1.4	46
17	Recent topics: the diagnosis, molecular genesis, and treatment of mitochondrial diseases. Journal of Human Genetics, 2019, 64, 113-125.	2.3	44
18	ANKRD11 variants cause variable clinical features associated with KBG syndrome and Coffinâ€“Siris-like syndrome. Journal of Human Genetics, 2017, 62, 741-746.	2.3	43

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19	A girl with West syndrome and autistic features harboring a de novo TBL1XR1 mutation. Journal of Human Genetics, 2014, 59, 581-583.	2.3	42
20	Mutations in COA7 cause spinocerebellar ataxia with axonal neuropathy. Brain, 2018, 141, 1622-1636.	7.6	38
21	Cardiomyopathy in children with mitochondrial disease: Prognosis and genetic background. International Journal of Cardiology, 2019, 279, 115-121.	1.7	35
22	Fatal Perinatal Mitochondrial Cardiac Failure Caused by Recurrent De Novo Duplications in the ATAD3 Locus. Med, 2021, 2, 49-73.e10.	4.4	33
23	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
24	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
25	Biallelic variants in LIG3 cause a novel mitochondrial neurogastrointestinal encephalomyopathy. Brain, 2021, 144, 1451-1466.	7.6	28
26	Newborn screening for carnitine palmitoyltransferase II deficiency using (C16 + C18:1)/C2: Evaluation of additional indices for adequate sensitivity and lower false-positivity. Molecular Genetics and Metabolism, 2017, 122, 67-75.	1.1	26
27	The first case in Asia of 2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency (HSD10 disease) with atypical presentation. Journal of Human Genetics, 2014, 59, 609-614.	2.3	24
28	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
29	Characteristic MRI features of chronic inflammatory demyelinating polyradiculoneuropathy. Brain and Development, 2015, 37, 894-896.	1.1	21
30	New MTâ€ND6 and NDUFA1 mutations in mitochondrial respiratory chain disorders. Annals of Clinical and Translational Neurology, 2014, 1, 361-369.	3.7	19
31	An infant case of diffuse cerebrospinal lesions and cardiomyopathy caused by a BOLA3 mutation. Brain and Development, 2018, 40, 484-488.	1.1	19
32	Effects of 5-aminolevulinic acid and sodium ferrous citrate on fibroblasts from individuals with mitochondrial diseases. Scientific Reports, 2019, 9, 10549.	3.3	19
33	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
34	Clinical manifestations and enzymatic activities of mitochondrial respiratory chain complexes in Pearson marrow-pancreas syndrome with 3-methylglutaconic aciduria: a case report and literature review. European Journal of Pediatrics, 2015, 174, 1593-1602.	2.7	17
35	Efficacy and Safety of Pitavastatin in Children and Adolescents with Familial Hypercholesterolemia in Japan and Europe. Journal of Atherosclerosis and Thrombosis, 2018, 25, 422-429.	2.0	17
36	Molecular diagnosis of mitochondrial respiratory chain disorders in <scp>J</scp>apan: Focusing on mitochondrial <scp>DNA</scp> depletion syndrome. Pediatrics International, 2014, 56, 180-187.	0.5	15

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37	Leigh syndrome with spinal cord involvement due to a hemizygous NDUFA1 mutation. Brain and Development, 2018, 40, 498-502.	1.1	15
38	Compound heterozygous GFM2 mutations with Leigh syndrome complicated by arthrogryposis multiplex congenita. Journal of Human Genetics, 2015, 60, 509-513.	2.3	14
39	Barth Syndrome: Different Approaches to Diagnosis. Journal of Pediatrics, 2018, 193, 256-260.	1.8	14
40	Valine-restricted diet for patients with ECHS1 deficiency: Divergent clinical outcomes in two Japanese siblings. Brain and Development, 2021, 43, 308-313.	1.1	14
41	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
42	Non-Hodgkin Lymphoma in a Patient With Cardiofaciocutaneous Syndrome. Journal of Pediatric Hematology/Oncology, 2011, 33, e342-e346.	0.6	13
43	Myocerebrohepatopathy spectrum disorder due to POLG mutations: A clinicopathological report. Brain and Development, 2015, 37, 719-724.	1.1	13
44	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
45	HDR: a statistical two-step approach successfully identifies disease genes in autosomal recessive families. Journal of Human Genetics, 2016, 61, 959-963.	2.3	11
46	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
47	Valine metabolites analysis in ECHS1 deficiency. Molecular Genetics and Metabolism Reports, 2021, 29, 100809.	1.1	9
48	Detection of novel Fabry diseaseâ€associated pathogenic variants in Japanese patients by newborn and highâ€risk screening. Molecular Genetics & Genomic Medicine, 2020, 8, e1502.	1.2	8
49	Leigh Syndrome Due to NDUFV1 Mutations Initially Presenting as LBSL. Genes, 2020, 11, 1325.	2.4	8
50	Bile Acid Synthesis Disorders in Japan: Long-Term Outcome and Chenodeoxycholic Acid Treatment. Digestive Diseases and Sciences, 2021, 66, 3885-3892.	2.3	8
51	A high mutation load of m.14597A>G in MT-ND6 causes Leigh syndrome. Scientific Reports, 2021, 11, 11123.	3.3	8
52	Identification of Cryptic Novel Î±-Galactosidase A Gene Mutations: Abnormal mRNA Splicing and Large Deletions. JIMD Reports, 2015, 30, 63-72.	1.5	7
53	Successful treatment of infantile-onset ACAD9-related cardiomyopathy with a combination of sodium pyruvate, beta-blocker, and coenzyme Q₁₀. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 1181-1185.	0.9	7
54	NAD(P)HX dehydratase protein-truncating mutations are associated with neurodevelopmental disorder exacerbated by acute illness. Brain, 2020, 143, e54-e54.	7.6	7

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55	Possible mitochondrial dysfunction in a patient with deafness, dystonia, and cerebral hypomyelination (DDCH) due to BCAP31 Mutation. Molecular Genetics & Genomic Medicine, 2020, 8, e1129.	1.2	7
56	Expression analysis of two mutant human ornithine transcarbamylases in COS-7 cells. Journal of Human Genetics, 1998, 43, 54-58.	2.3	6
57	Fever of Unknown Origin as the Initial Manifestation ofÂValproate-Induced Fanconi Syndrome. Pediatric Neurology, 2014, 51, 846-849.	2.1	6
58	Changes of lipoproteins in phenylalanine hydroxylase-deficient children during the first year of life. Clinica Chimica Acta, 2014, 433, 1-4.	1.1	6
59	HDR-del: A tool based on Hamming distance for prioritizing pathogenic chromosomal deletions in exome sequencing. Human Mutation, 2017, 38, 1796-1800.	2.5	6
60	A novel mutation in TAZ causes mitochondrial respiratory chain disorder without cardiomyopathy. Journal of Human Genetics, 2017, 62, 539-547.	2.3	5
61	Mitochondrial respiratory chain complex IV deficiency complicated with chronic intestinal pseudoâ€obstruction in a neonate. Pediatrics International, 2016, 58, 651-655.	0.5	4
62	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
63	First Japanese case of maternal phenylketonuria treated with sapropterin dihydrochloride and the normal growth and development of the child. Molecular Genetics and Metabolism Reports, 2019, 21, 100526.	1.1	4
64	Ski3/TTC37 deficiency associated with trichohepatoenteric syndrome causes mitochondrial dysfunction in Drosophila. FEBS Letters, 2020, 594, 2168-2181.	2.8	4
65	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
66	Leigh syndrome-like MRI changes in a patient with biallelic HPDL variants treated with ketogenic diet. Molecular Genetics and Metabolism Reports, 2021, 29, 100800.	1.1	4
67	A rapid screening with direct sequencing from blood samples for the diagnosis of Leigh syndrome. Molecular Genetics and Metabolism Reports, 2014, 1, 133-138.	1.1	2
68	Need for strict clinical management of patients with carnitine palmitoyltransferase II deficiency: Experience with two cases detected by expanded newborn screening. Molecular Genetics and Metabolism Reports, 2020, 24, 100611.	1.1	2
69	Macroscopic Characteristics of the Native Liver in Children With MPV17â€Related Mitochondrial DNA Depletion Syndrome: An Indication for Performing Liver Transplantation?. Liver Transplantation, 2022, 28, 497-500.	2.4	2
70	A Japanese family with P102L Gerstmannâ€“StrÃ¤usslerâ€“Scheinker disease with a variant Creutzfeldt-Jakob disease-like phenotype among the siblings: A case report. ENeurologicalSci, 2021, 25, 100380.	1.3	2
71	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
72	Preoperative urinary tract obstruction in scoliosis patients. Pediatrics International, 2017, 59, 48-52.	0.5	1

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73	Reply to the â€œLetter to the Editorâ€•from Dr. J Finsterer and colleagues. Neurogenetics, 2019, 20, 55-56.	1.4	1
74	Prenatal diagnosis of severe mitochondrial diseases caused by nuclear gene defects: a study in Japan. Scientific Reports, 2021, 11, 3531.	3.3	1
75	A case of ATR-X syndrome with mitochondrial respiratory chain dysfunction. European Journal of Medical Genetics, 2021, 64, 104251.	1.3	1
76	Reply to the letter: â€œThe diagnostic value of MRI in pediatric chronic inflammatory demyelinating polyradiculoneuropathyâ€• Brain and Development, 2016, 38, 174.	1.1	0
77	Nationwide epidemiological survey of holoprosencephaly in Japan. Pediatrics International, 2020, 62, 593-599.	0.5	0
78	Clinical heterogeneity in patients with m.4412Gâ€¯>â€¯A MT-TM mutation and different heteroplasmy levels. Mitochondrion, 2021, 59, 214-215.	3.4	0