Robert Kopajtich

List of Publications by Year
in descending order

32 papers	1,787 citations	361413 20 h-index	414414 32 g-index
36 all docs	36 docs citations	36 times ranked	3733 citing authors

#	Article	IF	Citations
1	Genetic diagnosis of Mendelian disorders via RNA sequencing. Nature Communications, 2017, 8, 15824.	12.8	432
2	ELAC2 Mutations Cause a Mitochondrial RNA Processing Defect Associated with Hypertrophic Cardiomyopathy. American Journal of Human Genetics, 2013, 93, 211-223.	6.2	127
3	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
4	Absence of BiP Co-chaperone DNAJC3 Causes Diabetes Mellitus and Multisystemic Neurodegeneration. American Journal of Human Genetics, 2014, 95, 689-697.	6.2	100
5	Impaired complex I repair causes recessive Leberâ€™s hereditary optic neuropathy. Journal of Clinical Investigation, 2021, 131, .	8.2	89
6	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	8.2	85
7	TRMT5 Mutations Cause a Defect in Post-transcriptional Modification of Mitochondrial tRNA Associated with Multiple Respiratory-Chain Deficiencies. American Journal of Human Genetics, 2015, 97, 319-328.	6.2	83
8	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
9	Phenotypic spectrum of eleven patients and five novel MTFMT mutations identified by exome sequencing and candidate gene screening. Molecular Genetics and Metabolism, 2014, 111, 342-352.	1.1	65
10	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. Journal of Clinical Investigation, 2019, 130, 108-125.	8.2	65
11	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
12	OCR-Stats: Robust estimation and statistical testing of mitochondrial respiration activities using Seahorse XF Analyzer. PLoS ONE, 2018, 13, e0199938.	2.5	55
13	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. American Journal of Human Genetics, 2016, 99, 674-682.	6.2	48
14	The diagnosis of inborn errors of metabolism by an integrative â€œmultiâ€•omicsâ€•approach: A perspective encompassing genomics, transcriptomics, and proteomics. Journal of Inherited Metabolic Disease, 2020, 43, 25-35.	3.6	47
15	Defining clinical subgroups and genotypeâ€“phenotype correlations in NBAS-associated disease across 110 patients. Genetics in Medicine, 2020, 22, 610-621.	2.4	46
16	RINT1 Bi-allelic Variations Cause Infantile-Onset Recurrent Acute Liver Failure and Skeletal Abnormalities. American Journal of Human Genetics, 2019, 105, 108-121.	6.2	39
17	Bi-allelic Mutations in Phe-tRNA Synthetase Associated with a Multi-system Pulmonary Disease Support Non-translational Function. American Journal of Human Genetics, 2018, 103, 100-114.	6.2	34
18	Mutations inELAC2associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3â€²â€end processing. Human Mutation, 2019, 40, 1731-1748.	2.5	31

#	Article	IF	Citations
19	Mutation in ITCH Gene Can Cause Syndromic Multisystem Autoimmune Disease With Acute Liver Failure. Pediatrics, 2019, 143, .	2.1	29
20	Biallelic variants inWARS2encoding mitochondrial tryptophanyl-tRNA synthase in six individuals with mitochondrial encephalopathy. Human Mutation, 2017, 38, 1786-1795.	2.5	24
21	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. Genetics in Medicine, 2020, 22, 1863-1873.	2.4	19
22	De novo stop-loss variants in CLDN11 cause hypomyelinating leukodystrophy. Brain, 2021, 144, 411-419.	7.6	12
23	Variants in Mitochondrial <scp>ATP</scp> Synthase Cause Variable Neurologic Phenotypes. Annals of Neurology, 2022, 91, 225-237.	5.3	12
24	PRUNE1 Deficiency: Expanding the Clinical and Genetic Spectrum. Neuropediatrics, 2018, 49, 330-338.	0.6	11
25	Expanding the clinical and genetic spectrum of FDXR deficiency by functional validation of variants of uncertain significance. Human Mutation, 2021, 42, 310-319.	2.5	11
26	Protonation-Dependent Sequencing of 5-Formylcytidine in RNA. Biochemistry, 2022, 61, 535-544.	2.5	10
27	Analysis of Mitochondrial RNA-Processing Defects in Patient-Derived Tissues by qRT-PCR and RNAseq. Methods in Molecular Biology, 2017, 1567, 379-390.	0.9	8
28	AOPEP variants as a novel cause of recessive dystonia: Generalized dystonia and dystonia-parkinsonism. Parkinsonism and Related Disorders, 2022, 97, 52-56.	2.2	7
29	Myopathic mitochondrial DNA depletion syndrome associated with biallelic variants in LIG3. Brain, 2021, 144, e74-e74.	7.6	5
30	Identification of a Novel m.3955G>A Variant in MT-ND1 Associated with Leigh Syndrome. Mitochondrion, 2021, 62, 13-23.	3.4	3
31	Suleiman-El-Hattab syndrome: a histone modification disorder caused by TASP1 deficiency. Human Molecular Genetics, 2022, 31, 3083-3094.	2.9	3
32	Identification of a Novel Variant in MT-CO3 Causing MELAS. Frontiers in Genetics, 2021, 12, 638749.	2.3	2