

Mirjana Gusic

List of Publications by Year in descending order

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Version: 2024-02-01

15
papers

765
citations

687363

13
h-index

888059

17
g-index

22
all docs

22
docs citations

22
times ranked

1567
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. <i>Genome Medicine</i> , 2022, 14, 38.	8.2	85
2	Detection of aberrant splicing events in RNA-seq data using FRASER. <i>Nature Communications</i> , 2021, 12, 529.	12.8	78
3	Detection of aberrant gene expression events in RNA sequencing data. <i>Nature Protocols</i> , 2021, 16, 1276-1296.	12.0	58
4	Genetic basis of mitochondrial diseases. <i>FEBS Letters</i> , 2021, 595, 1132-1158.	2.8	36
5	Bi-Allelic UQCRCFS1 Variants Are Associated with Mitochondrial Complex III Deficiency, Cardiomyopathy, and Alopecia Totalis. <i>American Journal of Human Genetics</i> , 2020, 106, 102-111.	6.2	36
6	ncRNAs: New Players in Mitochondrial Health and Disease?. <i>Frontiers in Genetics</i> , 2020, 11, 95.	2.3	58
7	Homozygous loss-of-function variants of <i>TASP1</i> , a gene encoding an activator of the histone methyltransferases KMT2A and KMT2D, cause a syndrome of developmental delay, happy demeanor, distinctive facial features, and congenital anomalies. <i>Human Mutation</i> , 2019, 40, 1985-1992.	2.5	10
8	Quantification and discovery of sequence determinants of protein-coding mRNA amount in human tissues. <i>Molecular Systems Biology</i> , 2019, 15, e8513.	7.2	63
9	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. <i>Journal of Clinical Investigation</i> , 2019, 130, 108-125.	8.2	65
10	NDUFB8 Mutations Cause Mitochondrial Complex I Deficiency in Individuals with Leigh-like Encephalomyopathy. <i>American Journal of Human Genetics</i> , 2018, 102, 460-467.	6.2	40
11	Biallelic Mutations in SLC1A2; an Additional Mode of Inheritance for SLC1A2-Related Epilepsy. <i>Neuropediatrics</i> , 2018, 49, 059-062.	0.6	14
12	Bi-allelic ADPRHL2 Mutations Cause Neurodegeneration with Developmental Delay, Ataxia, and Axonal Neuropathy. <i>American Journal of Human Genetics</i> , 2018, 103, 817-825.	6.2	40
13	OCR-Stats: Robust estimation and statistical testing of mitochondrial respiration activities using Seahorse XF Analyzer. <i>PLoS ONE</i> , 2018, 13, e0199938.	2.5	55
14	Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy. <i>American Journal of Human Genetics</i> , 2017, 100, 151-159.	6.2	63
15	Characterization of a Leber's hereditary optic neuropathy (LHON) family harboring two primary LHON mutations m.11778G > A and m.14484T > C of the mitochondrial DNA. <i>Mitochondrion</i> , 2017, 36, 15-20.	3.4	23