Mirjana Gusic

List of Publications by Year
in descending order

15	765	687363 13	888059 17
papers	citations	h-index	g-index

22	22	22	1567
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	8.2	85
2	Detection of aberrant splicing events in RNA-seq data using FRASER. Nature Communications, 2021, 12, 529.	12.8	78
3	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. Journal of Clinical Investigation, 2019, 130, 108-125.	8.2	65
4	Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy. American Journal of Human Genetics, 2017, 100, 151-159.	6.2	63
5	Quantification and discovery of sequence determinants of proteinâ€perâ€mRNA amount inÂ29Âhuman tissues. Molecular Systems Biology, 2019, 15, e8513.	7.2	63
6	ncRNAs: New Players in Mitochondrial Health and Disease?. Frontiers in Genetics, 2020, 11, 95.	2.3	58
7	Detection of aberrant gene expression events in RNA sequencing data. Nature Protocols, 2021, 16, 1276-1296.	12.0	58
8	OCR-Stats: Robust estimation and statistical testing of mitochondrial respiration activities using Seahorse XF Analyzer. PLoS ONE, 2018, 13, e0199938.	2.5	55
9	NDUFB8 Mutations Cause Mitochondrial Complex I Deficiency in Individuals with Leigh-like Encephalomyopathy. American Journal of Human Genetics, 2018, 102, 460-467.	6.2	40
10	Bi-allelic ADPRHL2 Mutations Cause Neurodegeneration with Developmental Delay, Ataxia, and Axonal Neuropathy. American Journal of Human Genetics, 2018, 103, 817-825.	6.2	40
11	Bi-Allelic UQCRFS1 Variants Are Associated with Mitochondrial Complex III Deficiency, Cardiomyopathy, and Alopecia Totalis. American Journal of Human Genetics, 2020, 106, 102-111.	6.2	36
12	Genetic basis of mitochondrial diseases. FEBS Letters, 2021, 595, 1132-1158.	2.8	36
13	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. Mitochondrion, 2017, 36, 15-20.	3.4	23
14	Biallelic Mutations in SLC1A2; an Additional Mode of Inheritance for SLC1A2-Related Epilepsy. Neuropediatrics, 2018, 49, 059-062.	0.6	14
15	Homozygous lossâ€ofâ€function variants of TASP1 , 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. Human Mutation, 2019, 40, 1985-1992.	2.5	10