Arcangela Iuso

List of Publications by Year
in descending order

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28 papers	2,512 citations	331670 21 h-index	526287 27 g-index
28 all docs	28 docs citations	28 times ranked	5239 citing authors

#	Article	IF	Citations
1	Generation of two human iPSC lines, HMGUi003-A and MRIi028-A, carrying pathogenic biallelic variants in the PPCS gene. Stem Cell Research, 2022, 61, 102773.	0.7	2
2	A comprehensive phenotypic characterization of a whole-body Wdr45 knock-out mouse. Mammalian Genome, 2021, 32, 332-349.	2.2	4
3	Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918.	10.2	139
4	Lossâ€ofâ€Function Variants in <scp>HOPS</scp> Complex Genes <scp>VPS16</scp> and <scp>VPS41</scp> Cause Early Onset Dystonia Associated with Lysosomal Abnormalities. Annals of Neurology, 2020, 88, 867-877.	5.3	70
5	Arabidopsis thaliana alternative dehydrogenases: a potential therapy for mitochondrial complex I deficiency? Perspectives and pitfalls. Orphanet Journal of Rare Diseases, 2019, 14, 236.	2.7	11
6	OCR-Stats: Robust estimation and statistical testing of mitochondrial respiration activities using Seahorse XF Analyzer. PLoS ONE, 2018, 13, e0199938.	2.5	55
7	Mutations in PPCS, Encoding Phosphopantothenoylcysteine Synthetase, Cause Autosomal-Recessive Dilated Cardiomyopathy. American Journal of Human Genetics, 2018, 102, 1018-1030.	6.2	42
8	Bi-allelic mutations in TRAPPC2L result in a neurodevelopmental disorder and have an impact on RAB11 in fibroblasts. Journal of Medical Genetics, 2018, 55, 753-764.	3.2	39
9	A Homozygous Splice Site Mutation in SLC25A42, Encoding the Mitochondrial Transporter of Coenzyme A, Causes Metabolic Crises and Epileptic Encephalopathy. JIMD Reports, 2018, 44, 1-7.	1.5	15
10	Genetic diagnosis of Mendelian disorders via RNA sequencing. Nature Communications, 2017, 8, 15824.	12.8	432
11	Assessing Mitochondrial Bioenergetics in Isolated Mitochondria from Various Mouse Tissues Using Seahorse XF96 Analyzer. Methods in Molecular Biology, 2017, 1567, 217-230.	0.9	64
12	Rare causes of early-onset dystonia-parkinsonism with cognitive impairment: a de novo PSEN-1 mutation. Neurogenetics, 2017, 18, 175-178.	1.4	23
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	Absence of the Autophagy Adaptor SQSTM1/p62 Causes Childhood-Onset Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy. American Journal of Human Genetics, 2016, 99, 735-743.	6.2	99
15	Disturbed mitochondrial and peroxisomal dynamics due to loss of MFF causes Leigh-like encephalopathy, optic atrophy and peripheral neuropathy. Journal of Medical Genetics, 2016, 53, 270-278.	3.2	105
16	Bi-allelic Truncating Mutations in TANGO2 Cause Infancy-Onset Recurrent Metabolic Crises with Encephalocardiomyopathy. American Journal of Human Genetics, 2016, 98, 358-362.	6.2	77
17	Mutations of C19orf12, coding for a transmembrane glycine zipper containing mitochondrial protein, cause mis-localization of the protein, inability to respond to oxidative stress and increased mitochondrial Ca2+. Frontiers in Genetics, 2015, 6, 185.	2.3	57
18	Impairment of Drosophila Orthologs of the Human Orphan Protein C19orf12 Induces Bang Sensitivity and Neurodegeneration. PLoS ONE, 2014, 9, e89439.	2.5	28

#	Article	IF	Citations
19	Loss-of-function mutations in MGME1 impair mtDNA replication and cause multisystemic mitochondrial disease. Nature Genetics, 2013, 45, 214-219.	21.4	198
20	Mutation screening of 75 candidate genes in 152 complex I deficiency cases identifies pathogenic variants in 16 genes includingNDUFB9. Journal of Medical Genetics, 2012, 49, 83-89.	3.2	78
21	Molecular diagnosis in mitochondrial complex I deficiency using exome sequencing. Journal of Medical Genetics, 2012, 49, 277-283.	3.2	182
22	Cellular rescue-assay aids verification of causative DNA-variants in mitochondrial complex I deficiency. Molecular Genetics and Metabolism, 2011, 103, 161-166.	1.1	23
23	Absence of an Orphan Mitochondrial Protein, C19orf12, Causes a Distinct Clinical Subtype of Neurodegeneration with Brain Iron Accumulation. American Journal of Human Genetics, 2011, 89, 543-550.	6.2	224
24	Exome sequencing identifies ACAD9 mutations as a cause of complex I deficiency. Nature Genetics, 2010, 42, 1131-1134.	21.4	234
25	Mitochondrial Respiratory Dysfunction in Familiar Parkinsonism Associated with PINK1 Mutation. Neurochemical Research, 2008, 33, 2565-2574.	3.3	99
26	cAMP controls oxygen metabolism in mammalian cells. FEBS Letters, 2006, 580, 4539-4543.	2.8	60
27	Dysfunctions of Cellular Oxidative Metabolism in Patients with Mutations in the NDUFS1 and NDUFS4 Genes of Complex I. Journal of Biological Chemistry, 2006, 281, 10374-10380.	3.4	128
28	Novel phosphopantothenoylcysteine synthetase (<scp> PPCS </scp>) mutations with prominent neuromuscular features: Expanding the phenotypical spectrum of <scp> PPCS </scp> â€related disorders. American Journal of Medical Genetics, Part A, 0, , .	1.2	1