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	Genetic diagnosis of Mendelian disorders via RNA sequencing. Nature Communications, 2017, 8, 15824.	12.8	432
2	Exome sequencing identifies ACAD9 mutations as a cause of complex I deficiency. Nature Genetics, 2010, 42, 1131-1134.	21.4	234
3	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
4	Loss-of-function mutations in MGME1 impair mtDNA replication and cause multisystemic mitochondrial disease. Nature Genetics, 2013, 45, 214-219.	21.4	198
5	Molecular diagnosis in mitochondrial complex I deficiency using exome sequencing. Journal of Medical Genetics, 2012, 49, 277-283.	3.2	182
6	Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918.	10.2	139
7	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
8	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
9	Mitochondrial Respiratory Dysfunction in Familiar Parkinsonism Associated with PINK1 Mutation. Neurochemical Research, 2008, 33, 2565-2574.	3.3	99
10	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
11	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
12	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
13	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
14	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
15	cAMP controls oxygen metabolism in mammalian cells. FEBS Letters, 2006, 580, 4539-4543.	2.8	60
16	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
17	OCR-Stats: Robust estimation and statistical testing of mitochondrial respiration activities using Seahorse XF Analyzer. PLoS ONE, 2018, 13, e0199938.	2.5	55
18	Mutations in PPCS, Encoding Phosphopantothenoylcysteine Synthetase, Cause Autosomal-Recessive Dilated Cardiomyopathy. American Journal of Human Genetics, 2018, 102, 1018-1030.	6.2	42

#	Article	IF	Citations
19	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
20	Impairment of Drosophila Orthologs of the Human Orphan Protein C19orf12 Induces Bang Sensitivity and Neurodegeneration. PLoS ONE, 2014, 9, e89439.	2.5	28
21	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
22	Rare causes of early-onset dystonia-parkinsonism with cognitive impairment: a de novo PSEN-1 mutation. Neurogenetics, 2017, 18, 175-178.	1.4	23
23	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
24	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
25	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
26	A comprehensive phenotypic characterization of a whole-body Wdr45 knock-out mouse. Mammalian Genome, 2021, 32, 332-349.	2.2	4
27	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
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