Benedetta Ruzzenente

List of Publications by Year
in descending order

32 papers	2,044 citations	361413 20 h-index	434195 31 g-index

34 all docs	34 docs citations	34 times ranked	2972 citing authors

#	Article	IF	Citations
1	Novel FARS2 variants in patients with early onset encephalopathy with or without epilepsy associated with long survival. European Journal of Human Genetics, 2021, 29, 533-538.	2.8	8
2	Cerebral blood flow and acute episodes of Leigh syndrome in neurometabolic disorders. Developmental Medicine and Child Neurology, 2021, 63, 705-711.	2.1	6
3	Variants in the MIPEP gene presenting with complex neurological phenotype without cardiomyopathy, impair OXPHOS protein maturation and lead to a reduced OXPHOS abundance in patient cells. Molecular Genetics and Metabolism, 2021, 134, 267-273.	1.1	4
4	Linear Density Sucrose Gradients to Study Mitoribosomal Biogenesis in Tissue-Specific Knockout Mice. Methods in Molecular Biology, 2021, 2224, 47-60.	0.9	1
5	Biallelic <i>IARS2</i> mutations presenting as sideroblastic anemia. Haematologica, 2021, 106, 0-0.	3.5	3
6	Clinical, neuroimaging and biochemical findings in patients and patient fibroblasts expressing ten novel GFM1 mutations. Human Mutation, 2020, 41, 397-402.	2.5	10
7	Mutations in the MRPS28 gene encoding the small mitoribosomal subunit protein bS1m in a patient with intrauterine growth retardation, craniofacial dysmorphism and multisystemic involvement. Human Molecular Genetics, 2019, 28, 1445-1462.	2.9	19
8	Mitochondrial fusion is required for regulation of mitochondrial DNA replication. PLoS Genetics, 2019, 15, e1008085.	3.5	116
9	High predictive value of brain MRI imaging in primary mitochondrial respiratory chain deficiency. Journal of Medical Genetics, 2018, 55, 378-383.	3.2	21
10	Bi-allelic Mutations in the Mitochondrial Ribosomal Protein MRPS2 Cause Sensorineural Hearing Loss, Hypoglycemia, and Multiple OXPHOS Complex Deficiencies. American Journal of Human Genetics, 2018, 102, 685-695.	6.2	61
11	Inhibition of mitochondrial translation in fibroblasts from a patient expressing the KARS p.(Pro228Leu) variant and presenting with sensorineural deafness, developmental delay, and lactic acidosis. Human Mutation, 2018, 39, 2047-2059.	2.5	14
12	Biallelic Mutations in MRPS34 Lead to Instability of the Small Mitoribosomal Subunit and Leigh Syndrome. American Journal of Human Genetics, 2017, 101, 239-254.	6.2	83
13	LRPPRC-mediated folding of the mitochondrial transcriptome. Nature Communications, 2017, 8, 1532.	12.8	80
14	Mutations in Complex I Assembly Factor TMEM126B Result in Muscle Weakness and Isolated Complex I Deficiency. American Journal of Human Genetics, 2016, 99, 208-216.	6.2	51
15	<scp>CLPP</scp> coordinates mitoribosomal assembly through the regulation of <scp>ERAL</scp>1 levels. EMBO Journal, 2016, 35, 2566-2583.	7.8	123
16	Complex I assembly factor TMEM126B mutations result in muscle weakness and isolated complex I deficiency. Biochimica Et Biophysica Acta - Bioenergetics, 2016, 1857, e31-e32.	1.0	0
17	Mouse models for mitochondrial diseases. Human Molecular Genetics, 2016, 25, R115-R122.	2.9	24
18	Loss of LRPPRC causes ATP synthase deficiency. Human Molecular Genetics, 2014, 23, 2580-2592.	2.9	91

#	Article	IF	Citations
19	NSUN4 Is a Dual Function Mitochondrial Protein Required for Both Methylation of 12S rRNA and Coordination of Mitoribosomal Assembly. PLoS Genetics, 2014, 10, e1004110.	3.5	232
20	The Respiratory Chain Supercomplex Organization Is Independent of COX7a2l Isoforms. Cell Metabolism, 2014, 20, 1069-1075.	16.2	90
21	POLRMT does not transcribe nuclear genes. Nature, 2014, 514, E7-E11.	27.8	35
22	MTERF1 Binds mtDNA to Prevent Transcriptional Interference at the Light-Strand Promoter but Is Dispensable for rRNA Gene Transcription Regulation. Cell Metabolism, 2013, 17, 618-626.	16.2	93
23	MTERF3 Regulates Mitochondrial Ribosome Biogenesis in Invertebrates and Mammals. PLoS Genetics, 2013, 9, e1003178.	3.5	85
24	TWINKLE is an essential mitochondrial helicase required for synthesis of nascent D-loop strands and complete mtDNA replication. Human Molecular Genetics, 2013, 22, 1983-1993.	2.9	132
25	The Leucine-rich Pentatricopeptide Repeat-containing Protein (LRPPRC) Does Not Activate Transcription in Mammalian Mitochondria. Journal of Biological Chemistry, 2013, 288, 15510-15519.	3.4	27
26	LRPPRC is necessary for polyadenylation and coordination of translation of mitochondrial mRNAs. EMBO Journal, 2012, 31, 443-456.	7.8	264
27	MTERF4 Regulates Translation by Targeting the Methyltransferase NSUN4 to the Mammalian Mitochondrial Ribosome. Cell Metabolism, 2011, 13, 527-539.	16.2	221
28	The Bicoid Stability Factor Controls Polyadenylation and Expression of Specific Mitochondrial mRNAs in Drosophila melanogaster. PLoS Genetics, 2011, 7, e1002324.	3.5	55
29	LRPPRC is a mitochondrial matrix protein that is conserved in metazoans. Biochemical and Biophysical Research Communications, 2010, 398, 759-764.	2.1	49
30	Crystal Structures of Human and Murine Deoxyribonucleotidases:â€‰ Insights into Recognition of Substrates and Nucleotide Analogues. Biochemistry, 2007, 46, 13809-13818.	2.5	19
31	Structural Basis for Substrate Specificity of the Human Mitochondrial Deoxyribonucleotidase. Structure, 2005, 13, 1081-1088.	3.3	17
32	Mouse cytosolic and mitochondrial deoxyribonucleotidases: cDNA cloning of the mitochondrial enzyme, gene structures, chromosomal mapping and comparison with the human orthologs. Gene, 2002, 294, 109-117.	2.2	10