Vittoria Petruzzella

List of Publications by Year
in descending order

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64 papers	3,242 citations	201575 27 h-index	155592 55 g-index
65 all docs	65 docs citations	65 times ranked	4614 citing authors

#	Article	IF	Citations
1	Bi-allelic variants in MTMR5/SBF1 cause Charcot-Marie-Tooth type 4B3 featuring mitochondrial dysfunction. BMC Medical Genomics, 2021, 14, 157.	0.7	2
2	Tackling Dysfunction of Mitochondrial Bioenergetics in the Brain. International Journal of Molecular Sciences, 2021, 22, 8325.	1.8	5
3	RCC1L (WBSCR16) isoforms coordinate mitochondrial ribosome assembly through their interaction with GTPases. PLoS Genetics, 2020, 16, e1008923.	1.5	18
4	CMT2A Harboring Mitofusin 2 Mutation with Optic Nerve Atrophy and Normal Visual Acuity . International Medical Case Reports Journal, 2020, Volume 13, 41-45.	0.3	12
5	Fishing in the Cell Powerhouse: Zebrafish as A Tool for Exploration of Mitochondrial Defects Affecting the Nervous System. International Journal of Molecular Sciences, 2019, 20, 2409.	1.8	16
6	TRIM8 Blunts the Pro-proliferative Action of Î”Np63Î± in a p53 Wild-Type Background. Frontiers in Oncology, 2019, 9, 1154.	1.3	8
7	Mitochondrial DNA copy number in affected and unaffected LHON mutation carriers. BMC Research Notes, 2018, 11, 911.	0.6	25
8	Author Response: Increased mtDNA Copy Number Protects Against LHON. , 2018, 59, 331.		1
9	Leberâ€™s hereditary optic neuropathy, intellectual disability and epilepsy presenting with variable penetrance associated to the m.3460Gâ€‰›A mutation and a heteroplasmic expansion of the microsatellite in MTRNR1 gene â€“ case report. BMC Medical Genetics, 2018, 19, 129.	2.1	7
10	Author Response: Do High mtDNA Copy Numbers Truly Prevent LHON Manifestations?. , 2017, 58, 4077.		0
11	High Mitochondrial DNA Copy Number Is a Protective Factor From Vision Loss in Heteroplasmic Leber's Hereditary Optic Neuropathy (LHON). , 2017, 58, 2193.		37
12	Leber's hereditary optic neuropathy (LHON) in an Apulian cohort of subjects. Acta Myologica, 2017, 36, 163-177.	1.5	7
13	Mitochondrial DNA copy number differentiates the Leberâ€™s hereditary optic neuropathy affected individuals from the unaffected mutation carriers. Brain, 2016, 139, e1-e1.	3.7	39
14	Mitochondrial DNA variants and risk of familial breast cancer: An exploratory study. International Journal of Oncology, 2014, 44, 1691-1698.	1.4	33
15	Pontocerebellar hypoplasia type 6 caused by mutations in RARS2: definition of the clinical spectrum and molecular findings in five patients. Journal of Inherited Metabolic Disease, 2013, 36, 43-53.	1.7	70
16	Mitochondrial genome aberrations in skeletal muscle of patients with motor neuron disease. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 261-266.	1.1	14
17	HmtDB, a genomic resource for mitochondrion-based human variability studies. Nucleic Acids Research, 2012, 40, D1150-D1159.	6.5	82
18	Deep sequencing unearths Nuclear mitochondrial Sequences under Leber's hereditary optic neuropathy-associated false heteroplasmic mitochondrial DNA variants. Human Molecular Genetics, 2012, 21, 3753-3764.	1.4	15

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19	Respiratory chain complex I, a main regulatory target of the cAMP/PKA pathway is defective in different human diseases. FEBS Letters, 2012, 586, 568-577.	1.3	75
20	The Oxidative Phosphorylation System in Mammalian Mitochondria. Advances in Experimental Medicine and Biology, 2012, 942, 3-37.	0.8	198
21	Mitochondrial DNA metabolism in early development of zebrafish (Danio rerio). Biochimica Et Biophysica Acta - Bioenergetics, 2012, 1817, 1002-1011.	0.5	78
22	Mitochondrial genome large rearrangements in the skeletal muscle of a patient with <scp>PMA</scp>. European Journal of Neurology, 2012, 19, e63-4.	1.7	9
23	Dysfunction of Mitochondrial Respiratory Chain Complex I in Neurological Disorders: Genetics and Pathogenetic Mechanisms. Advances in Experimental Medicine and Biology, 2012, 942, 371-384.	0.8	12
24	Bilateral progressive visual loss in an epileptic, mentally retarded boy. Middle East African Journal of Ophthalmology, 2011, 18, 67.	0.5	5
25	Late-onset Leber hereditary optic neuropathy mimicking Susacâ€™s syndrome. Journal of Neurology, 2010, 257, 1999-2003.	1.8	14
26	A new locus on 3p23â€“p25 for an autosomal-dominant limb-girdle muscular dystrophy, LGMD1H. European Journal of Human Genetics, 2010, 18, 636-641.	1.4	27
27	Pathogenetic mechanisms in hereditary dysfunctions of complex I of the respiratory chain in neurological diseases. Biochimica Et Biophysica Acta - Bioenergetics, 2009, 1787, 502-517.	0.5	33
28	Molecular analysis in a family presenting with a mild form of late-onset autosomal dominant chronic progressive external ophthalmoplegia. Neuromuscular Disorders, 2009, 19, 423-426.	0.3	8
29	Bilateral striatal necrosis, dystonia and multiple mitochondrial DNA deletions: Case study and effect of deep brain stimulation. Movement Disorders, 2008, 23, 114-118.	2.2	30
30	The regulation of PTC containing transcripts of the human NDUFS4 gene of complex I of respiratory chain and the impact of pathological mutations. Biochimie, 2008, 90, 1452-1460.	1.3	11
31	The NDUFB11 gene is not a modifier in Leber hereditary optic neuropathy. Biochemical and Biophysical Research Communications, 2007, 355, 181-187.	1.0	13
32	1.5 Electron Transport. Structure, Redox-Coupled Protonmotive Activity, and Pathological Disorders of Respiratory Chain Complexes. , 2007, , 93-118.		8
33	Prefrontal-Hippocampal Coupling During Memory Processing Is Modulated by COMT Val158Met Genotype. Biological Psychiatry, 2006, 60, 1250-1258.	0.7	153
34	Prefrontal dysfunction in schizophrenia controlling for COMT Val158Met genotype and working memory performance. Psychiatry Research - Neuroimaging, 2006, 147, 221-226.	0.9	53
35	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.	1.6	128
36	Additive Effects of Genetic Variation in Dopamine Regulating Genes on Working Memory Cortical Activity in Human Brain. Journal of Neuroscience, 2006, 26, 3918-3922.	1.7	208

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37	Unusual clinical presentation of a patient carrying a novel single 1.8 kb deletion of mitochondrial DNA. Functional Neurology, 2006, 21, 39-41.	1.3	6
38	Mutations in structural genes of complex I associated with neurological diseases. Italian Journal of Biochemistry, 2006, 55, 254-62.	0.3	10
39	Mutations in theNDUFS4gene of mitochondrial complex I alter stability of the splice variants. FEBS Letters, 2005, 579, 3770-3776.	1.3	19
40	Interaction of COMT Val^{108/158}Met Genotype and Olanzapine Treatment on Prefrontal Cortical Function in Patients With Schizophrenia. American Journal of Psychiatry, 2004, 161, 1798-1805.	4.0	281
41	Respiratory Complex I in Brain Development and Genetic Disease. Neurochemical Research, 2004, 29, 547-560.	1.6	13
42	Clinical heterogeneity in patients with mutations in the NDUFS4 gene of mitochondrial complex I. Journal of Inherited Metabolic Disease, 2003, 26, 813-815.	1.7	59
43	Cerebellar ataxia as atypical manifestation of the 3243A>G MELAS mutation. Clinical Genetics, 2003, 65, 64-65.	1.0	15
44	Differential expression ofATPAF1andATPAF2genes encoding F1-ATPase assembly proteins in mouse tissues. FEBS Letters, 2003, 551, 42-46.	1.3	8
45	Pathological Mutations of the Human NDUFS4 Gene of the 18-kDa (AQDQ) Subunit of Complex I Affect the Expression of the Protein and the Assembly and Function of the Complex. Journal of Biological Chemistry, 2003, 278, 44161-44167.	1.6	120
46	Mutations in human nuclear genes encoding for subunits of mitochondrial respiratory complex I: the NDUFS4 gene. Gene, 2002, 286, 149-154.	1.0	54
47	Mitochondrial Disease Mimicking Polymyositis: A Case Report. Clinical Rheumatology, 2002, 21, 411-414.	1.0	5
48	The NADH: ubiquinone oxidoreductase (complex I) of the mammalian respiratory chain and the cAMP cascade. Journal of Bioenergetics and Biomembranes, 2002, 34, 1-10.	1.0	57
49	Complex I and the cAMP Cascade in Human Physiopathology. Bioscience Reports, 2002, 22, 3-16.	1.1	38
50	A nonsense mutation in the NDUFS4 gene encoding the 18 kDa (AQDQ) subunit of complex I abolishes assembly and activity of the complex in a patient with Leigh-like syndrome. Human Molecular Genetics, 2001, 10, 529-535.	1.4	120
51	Alteration of mitochondrial DNA and RNA level in human fibroblasts with impaired vitamin B12 coenzyme synthesis. FEBS Letters, 1998, 432, 173-178.	1.3	9
52	A Novel Insertion Mutation (A169i) in the CLN1 Gene Is Associated with Infantile Neuronal Ceroid Lipofuscinosis in an Italian Patient. Biochemical and Biophysical Research Communications, 1998, 245, 519-522.	1.0	19
53	Identification and Characterization of Human cDNAs Specific to BCS1, PET112, SCO1, COX15, and COX11, Five Genes Involved in the Formation and Function of the Mitochondrial Respiratory Chain. Genomics, 1998, 54, 494-504.	1.3	144
54	Disorders of nuclear-mitochondrial intergenomic signalling. Journal of Bioenergetics and Biomembranes, 1997, 29, 121-130.	1.0	16

#	Article	IF	Citations
55	Decrease of D-loop frequency in heart and cerebral hemispheres mitochondrial DNA of aged rat. Molecular and Chemical Neuropathology, 1995, 24, 193-202.	1.0	6
56	New Morphological Approaches to the Study of Mitochondrial Encephalomyopathies. Brain Pathology, 1992, 2, 113-119.	2.1	67
57	Is a point mutation in the mitochondrial ND2 gene associated with alzheimer's disease?. Biochemical and Biophysical Research Communications, 1992, 186, 491-497.	1.0	52
58	Age-Dependent Structural Variations in Rat Brain Mitochondrial DNA. Annals of the New York Academy of Sciences, 1992, 673, 194-199.	1.8	13
59	In vivo effect of acetyl-l-carnitine on succinate oxidation, adenine nucleotide pool and lipid composition of synaptic and non-synaptic mitochondria from cerebral hemispheres of senescent rats. Archives of Gerontology and Geriatrics, 1992, 14, 131-144.	1.4	31
60	Lipid Composition in Synaptic and Nonsynaptic Mitochondria from Rat Brains and Effect of Aging. Journal of Neurochemistry, 1992, 59, 487-491.	2.1	65
61	Molecular analysis of the muscle pathology associated with mitochondrial DNA deletions. Nature Genetics, 1992, 1, 359-367.	9.4	156
62	Reduced synthesis of mtRNA in isolated mitochondria of senescent rat brain. Biochemical and Biophysical Research Communications, 1991, 176, 645-653.	1.0	76
63	Reduced transcription of mitochondrial DNA in the senescent rat. Tissue dependence and effect of l-carnitine. FEBS Journal, 1990, 187, 501-506.	0.2	180
64	Faithful and highly efficient RNA synthesis in isolated mitochondria from rat liver. Current Genetics, 1988, 14, 477-482.	0.8	21