Veronika Boczonadi

List of Publications by Year
in descending order

26 papers	1,245 citations	361413 20 h-index	580821 25 g-index
27 all docs	27 docs citations	27 times ranked	2534 citing authors

#	Article	IF	Citations
1	Mitochondria: Impaired mitochondrial translation in human disease. International Journal of Biochemistry and Cell Biology, 2014, 48, 77-84.	2.8	158
2	EXOSC8 mutations alter mRNA metabolism and cause hypomyelination with spinal muscular atrophy and cerebellar hypoplasia. Nature Communications, 2014, 5, 4287.	12.8	120
3	Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency. American Journal of Human Genetics, 2016, 98, 1130-1145.	6.2	118
4	Synaptotagmin 2 Mutations Cause an Autosomal-Dominant Form of Lambert-Eaton Myasthenic Syndrome and Nonprogressive Motor Neuropathy. American Journal of Human Genetics, 2014, 95, 332-339.	6.2	96
5	Genetic heterogeneity of motor neuropathies. Neurology, 2017, 88, 1226-1234.	1.1	81
6	Mitochondrial DNA transcription and translation: clinical syndromes. Essays in Biochemistry, 2018, 62, 321-340.	4.7	72
7	Periplakin-dependent re-organisation of keratin cytoskeleton and loss of collective migration in keratin-8-downregulated epithelial sheets. Journal of Cell Science, 2006, 119, 5147-5159.	2.0	69
8	The role of <scp>tRNA</scp> synthetases in neurological and neuromuscular disorders. FEBS Letters, 2018, 592, 703-717.	2.8	68
9	Altered 2-thiouridylation impairs mitochondrial translation in reversible infantile respiratory chain deficiency. Human Molecular Genetics, 2013, 22, 4602-4615.	2.9	52
10	A novel mechanism causing imbalance of mitochondrial fusion and fission in human myopathies. Human Molecular Genetics, 2018, 27, 1186-1195.	2.9	52
11	Cytolinker cross-talk: Periplakin N-terminus interacts with plectin to regulate keratin organisation and epithelial migration. Experimental Cell Research, 2007, 313, 3579-3591.	2.6	43
12	Reversible infantile mitochondrial diseases. Journal of Inherited Metabolic Disease, 2015, 38, 427-435.	3.6	37
13	Altered RNA metabolism due to a homozygousRBM7mutation in a patient with spinal motor neuropathy. Human Molecular Genetics, 2016, 25, ddw149.	2.9	35
14	Mitochondrial oxodicarboxylate carrier deficiency is associated with mitochondrial DNA depletion and spinal muscular atrophyâ€“like disease. Genetics in Medicine, 2018, 20, 1224-1235.	2.4	31
15	Salbutamol modifies the neuromuscular junction in a mouse model of ColQ myasthenic syndrome. Human Molecular Genetics, 2019, 28, 2339-2351.	2.9	29
16	Mutations in glycyl-tRNA synthetase impair mitochondrial metabolism in neurons. Human Molecular Genetics, 2018, 27, 2187-2204.	2.9	26
17	Metabolic shift underlies recovery in reversible infantile respiratory chain deficiency. EMBO Journal, 2020, 39, e105364.	7.8	26
18	Clinical and functional characterisation of the combined respiratory chain defect in two sisters due to autosomal recessive mutations in MTFMT. Mitochondrion, 2013, 13, 743-748.	3.4	25

#	Article	IF	Citations
19	Scrib:Rac1 interactions are required for the morphogenesis of the ventricular myocardium. Cardiovascular Research, 2014, 104, 103-115.	3.8	25
20	Behr's Syndrome is Typically Associated with Disturbed Mitochondrial Translation and Mutations in the C12orf65 Gene. Journal of Neuromuscular Diseases, 2014, 1, 55-63.	2.6	20
21	Annexin A9 is a periplakin interacting partner in membraneâ€targeted cytoskeletal linker protein complexes. FEBS Letters, 2012, 586, 3090-3096.	2.8	18
22	Cysteine Supplementation May be Beneficial in a Subgroup of Mitochondrial Translation Deficiencies. Journal of Neuromuscular Diseases, 2016, 3, 363-379.	2.6	17
23	Investigating the role of the physiological isoform switch of cytochrome c oxidase subunits in reversible mitochondrial disease. International Journal of Biochemistry and Cell Biology, 2015, 63, 32-40.	2.8	14
24	Functional Analysis of Periplakin and Envoplakin, Cytoskeletal Linkers, and Cornified Envelope Precursor Proteins. Methods in Enzymology, 2016, 569, 309-329.	1.0	8
25	Amyloidâ€Î² in mitochondrial disease: mutation in a human metallopeptidase links amyloidotic neurodegeneration with mitochondrial processing. EMBO Molecular Medicine, 2016, 8, 173-175.	6.9	5
26	Mitochondrial Translation Deficiencies. , 2021, , 95-117.		0