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List of Publications by Year
in descending order

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68 papers	2,878 citations	159358 30 h-index	168136 53 g-index
70 all docs	70 docs citations	70 times ranked	4500 citing authors

#	Article	IF	Citations
1	Efficient mitochondrial biogenesis drives incomplete penetrance in Leberâ€™s hereditary optic neuropathy. Brain, 2014, 137, 335-353.	3.7	229
2	Oestrogens ameliorate mitochondrial dysfunction in Leberâ€™s hereditary optic neuropathy. Brain, 2011, 134, 220-234.	3.7	208
3	Cardiac involvement in mitochondrial DNA disease: clinical spectrum, diagnosis, and management. European Heart Journal, 2012, 33, 3023-3033.	1.0	182
4	Induction of Mitochondrial Biogenesis Is a Maladaptive Mechanism in Mitochondrial Cardiomyopathies. Journal of the American College of Cardiology, 2007, 50, 1362-1369.	1.2	164
5	A homoplasmic mitochondrial transfer Ribonucleic Acid mutation as a cause of maternally inherited hypertrophic cardiomyopathy. Journal of the American College of Cardiology, 2003, 41, 1786-1796.	1.2	161
6	Pathogenic expression of homoplasmic mtDNA mutations needs a complex nuclearâ€“mitochondrial interaction. Trends in Genetics, 2003, 19, 257-262.	2.9	137
7	Inhibition of glucose-6-phosphate dehydrogenase sensitizes cisplatin-resistant cells to death. Oncotarget, 2015, 6, 30102-30114.	0.8	101
8	Gastrointestinal Dysmotility in Mitochondrial Neurogastrointestinal Encephalomyopathy Is Caused by Mitochondrial DNA Depletion. American Journal of Pathology, 2008, 173, 1120-1128.	1.9	100
9	Pantothenate kinase-associated neurodegeneration: altered mitochondria membrane potential and defective respiration in Pank2 knock-out mouse model. Human Molecular Genetics, 2012, 21, 5294-5305.	1.4	87
10	Pantethine treatment is effective in recovering the disease phenotype induced by ketogenic diet in a pantothenate kinase-associated neurodegeneration mouse model. Brain, 2014, 137, 57-68.	3.7	78
11	Impaired mitochondrial biogenesis is a common feature to myocardial hypertrophy and end-stage ischemic heart failure. Cardiovascular Pathology, 2016, 25, 103-112.	0.7	77
12	Enhanced ROS production by NADPH oxidase is correlated to changes in antioxidant enzyme activity in human heart failure. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2010, 1802, 331-338.	1.8	76
13	Myocyte Transdifferentiation. Archives of Pathology and Laboratory Medicine, 2000, 124, 287-290.	1.2	70
14	Isoleucyl-tRNA synthetase levels modulate the penetrance of a homoplasmic m.4277T>C mitochondrial tRNAIle mutation causing hypertrophic cardiomyopathy. Human Molecular Genetics, 2012, 21, 85-100.	1.4	67
15	Mitochondrial Neurogastrointestinal Encephalomyopathy: Evidence of Mitochondrial DNA Depletion in the Small Intestine. Gastroenterology, 2006, 130, 893-901.	0.6	63
16	Targeting estrogen receptor Î² as preventive therapeutic strategy for Leber's hereditary optic neuropathy. Human Molecular Genetics, 2015, 24, ddv396.	1.4	62
17	NADPH oxidase-dependent redox signaling in human heart failure: Relationship between the left and right ventricle. Journal of Molecular and Cellular Cardiology, 2007, 42, 826-834.	0.9	59
18	Small-fibre neuropathy related to bulbar and spinal-onset in patients with ALS. Journal of Neurology, 2015, 262, 1014-1018.	1.8	57

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19	Nonischemic left ventricular scar and cardiac sudden death in the young. Human Pathology, 2016, 58, 78-89.	1.1	52
20	Sudden cardiac death in younger adults: autopsy diagnosis as a tool for preventive medicine. Human Pathology, 2006, 37, 794-801.	1.1	49
21	Homozygous <scp>NOTCH</scp> 3 null mutation and impaired <scp>NOTCH</scp> 3 signaling in recessive earlyâ€onset arteriopathy and cavitating leukoencephalopathy. EMBO Molecular Medicine, 2015, 7, 848-858.	3.3	48
22	AAV-mediated Liver-specific MPV17 Expression Restores mtDNA Levels and Prevents Diet-induced Liver Failure. Molecular Therapy, 2014, 22, 10-17.	3.7	47
23	Does the epidermal nerve fibre density measured by skin biopsy in patients with peripheral neuropathies correlate with neuropathic pain?. Pain, 2014, 155, 828-832.	2.0	47
24	Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE): Position paper on diagnosis, prognosis, and treatment by the <scp>MNGIE</scp> International Network. Journal of Inherited Metabolic Disease, 2021, 44, 376-387.	1.7	47
25	The isolated carboxyâ€terminal domain of human mitochondrial leucylâ€<scp>tRNA</scp> synthetase rescues the pathological phenotype of mitochondrial <scp>tRNA</scp> mutations in human cells. EMBO Molecular Medicine, 2014, 6, 169-182.	3.3	43
26	Monoamine Oxidase Is Overactivated in Left and Right Ventricles from Ischemic Hearts: An Intriguing Therapeutic Target. Oxidative Medicine and Cellular Longevity, 2016, 2016, 1-10.	1.9	37
27	Morphologic evidence of diffuse vascular damage in human and in the experimental model of ethylmalonic encephalopathy. Journal of Inherited Metabolic Disease, 2012, 35, 451-458.	1.7	35
28	A novel LAMP2 mutation associated with severe cardiac hypertrophy and microvascular remodeling in a female with Danon disease: a case report and literature review. Cardiovascular Pathology, 2016, 25, 423-431.	0.7	34
29	FATAL CONGENITAL MYOPATHY AND GASTROINTESTINAL PSEUDO-OBSTRUCTION DUE TO POLG1 MUTATIONS. Neurology, 2009, 72, 1103-1105.	1.5	33
30	Cardiomyopathies due to homoplasmic mitochondrial tRNA mutations: morphologic and molecular features. Human Pathology, 2013, 44, 1262-1270.	1.1	32
31	Pathogenesis of the deafness-associated A1555G mitochondrial DNA mutation. Biochemical and Biophysical Research Communications, 2002, 293, 521-529.	1.0	30
32	Is Apoptosis a Diagnostic Marker of Acute Myocardial Infarction?. Archives of Pathology and Laboratory Medicine, 2000, 124, 827-831.	1.2	28
33	Myositis in primary SjÃ¶gren's syndrome: data from a multicentre cohort. Clinical and Experimental Rheumatology, 2015, 33, 457-64.	0.4	27
34	Laryngeal Sensitivity in Patients with Amyotrophic Lateral Sclerosis. Frontiers in Neurology, 2016, 7, 212.	1.1	22
35	New insight into the mechanisms of ectopic fat deposition improvement after bariatric surgery. Scientific Reports, 2019, 9, 17315.	1.6	22
36	Neuromuscular magnetic stimulation counteracts muscle decline in ALS patients: results of a randomized, double-blind, controlled study. Scientific Reports, 2019, 9, 2837.	1.6	21

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37	Short peptides from leucyl-tRNA synthetase rescue disease-causing mitochondrial tRNA point mutations. Human Molecular Genetics, 2016, 25, 903-915.	1.4	19
38	Melanopsin-expressing retinal ganglion cells are resistant to cell injury, but not always. Mitochondrion, 2017, 36, 77-84.	1.6	18
39	Cerebral Mitochondrial Microangiopathy Leads to Leukoencephalopathy in Mitochondrial Neurogastrointestinal Encephalopathy. American Journal of Neuroradiology, 2018, 39, 427-434.	1.2	18
40	Isolated Distal Myopathy of the Upper Limbs Associated With Mitochondrial DNA Depletion and Polymerase Î³ Mutations. Archives of Neurology, 2010, 67, 1144-6.	4.9	16
41	Oncocytic glioblastoma: a glioblastoma showing oncocytic changes and increased mitochondrial DNA copy number. Human Pathology, 2013, 44, 1867-1876.	1.1	15
42	Revisiting the issue of mitochondrial DNA content in optic mitochondriopathies. Neurology, 2012, 79, 1517-1519.	1.5	13
43	Severe early onset ethylmalonic encephalopathy with West syndrome. Metabolic Brain Disease, 2015, 30, 1537-1545.	1.4	13
44	Novel compound mutations in the mitochondrial translation elongation factor (TSFM) gene cause severe cardiomyopathy with myocardial fibro-adipose replacement. Scientific Reports, 2019, 9, 5108.	1.6	12
45	Blue rubber bleb nevus syndrome and pulmonary hypertension: an unusual association. Cardiovascular Pathology, 2004, 13, 317-322.	0.7	11
46	Evaluation of Gastrointestinal mtDNA Depletion in Mitochondrial Neurogastrointestinal Encephalomyopathy (MNGIE). Methods in Molecular Biology, 2011, 755, 223-232.	0.4	11
47	Distribution of eosinophils in the gastrointestinal tract of children with no organic disease. Annals of Gastroenterology, 2020, 33, 508-515.	0.4	11
48	Pathologic Evidence of Arrhythmogenic Cardiomyopathy and Myocarditis in Two Siblings. Cardiovascular Pathology, 1998, 7, 39-46.	0.7	10
49	Novel Perspectives in Redox Biology and Pathophysiology of Failing Myocytes: Modulation of the Intramyocardial Redox Milieu for Therapeutic Interventionsâ€"A Review Article from the Working Group of Cardiac Cell Biology, Italian Society of Cardiology. Oxidative Medicine and Cellular Longevity, 2016, 2016, 1-13.	1.9	10
50	Afferent Nerve Ending Density in the Human Laryngeal Mucosa: Potential Implications on Endoscopic Evaluation of Laryngeal Sensitivity. Dysphagia, 2015, 30, 139-144.	1.0	9
51	Anti-aminoacyl-tRNA synthetase-related myositis and dermatomyositis: clues for differential diagnosis on muscle biopsy. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2018, 472, 477-487.	1.4	9
52	Evidence of enteric angiopathy and neuromuscular hypoxia in patients with mitochondrial neurogastrointestinal encephalomyopathy. American Journal of Physiology - Renal Physiology, 2021, 320, G768-G779.	1.6	9
53	Myocardial fibrosis: morphologic patterns and role of imaging in diagnosis and prognostication. Cardiovascular Pathology, 2022, 56, 107391.	0.7	9
54	Exogenous peptides are able to penetrate human cell and mitochondrial membranes, stabilize mitochondrial tRNA structures, and rescue severe mitochondrial defects. FASEB Journal, 2020, 34, 7675-7686.	0.2	6

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55	Reply: Mitochondrial DNA copy number differentiates the Leberâ€™s hereditary optic neuropathy affected individuals from the unaffected mutation carriers. Brain, 2016, 139, e2-e2.	3.7	5
56	Diagnostic Value of Persistently Low Positive TGA-IgA Titers in Symptomatic Children With Suspected Celiac Disease. Journal of Pediatric Gastroenterology and Nutrition, 2021, 72, 712-717.	0.9	5
57	The phenotypic expression of mitochondrial tRNA-mutations can be modulated by either mitochondrial leucyl-tRNA synthetase or the C-terminal domain thereof. Frontiers in Genetics, 2015, 6, 113.	1.1	4
58	A new double immunohistochemistry method to detect mucosal anti-transglutaminase IgA deposits in coeliac children. Digestive and Liver Disease, 2022, 54, 200-206.	0.4	3
59	Myopathy Complicating Lupus Pregnancy. Journal of Clinical Rheumatology, 2013, 19, 132-133.	0.5	2
60	Coronary atherosclerosis and sudden cardiac death in the young: another face of the culprit, another way of striking?. International Journal of Cardiology, 2018, 264, 28-29.	0.8	2
61	Peculiarities of Prevalence and Morphology of Congenital Heart Disease Detected In Utero. Cardiovascular Pathology, 1998, 7, 251-259.	0.7	1
62	Diagnosis of Primary Cardiac T-cell Lymphoma: Feasibility and Safety of Endomyocardial Biopsy Guided by Pre-acquired Cardiovascular Magnetic Resonance. European Journal of Case Reports in Internal Medicine, 2019, 8, 002427.	0.2	1
63	Outcome of Surgical Resection and Chemotherapy Versus Chemotherapy Alone for the Treatment of Isolated Primary Adrenal Lymphoma: A Retrospective Cohort Study of 16 Consecutive Patients. Anticancer Research, 2021, 41, 2647-2652.	0.5	1
64	Resection for Internal Jugular Vein Thrombosis and Cervical Lymph Nodes' Involvement from Gastric Cancer. Anticancer Research, 2020, 40, 2889-2893.	0.5	1
65	Mitochondrial tRNA mutations manifest not only as hypertrophic cardiomyopathy but also as noncompactionâ€”reply. Human Pathology, 2014, 45, 1791-1792.	1.1	0
66	FRI0500â€…Evaluation of the Role of Fractalkine Chemokine CX3CL1 and Its Receptor CX3CR1 in Inflammatory Myopathies: Table 1. Annals of the Rheumatic Diseases, 2014, 73, 568.1-568.	0.5	0
67	AB0624â€…High Levels of Proinflammatory Biomarkers in Patients with Idiopathic Inflammatory Myopathies. Annals of the Rheumatic Diseases, 2014, 73, 1012.1-1012.	0.5	0
68	Pathology of endomyocardial biopsy. Diagnostic Histopathology, 2018, 24, 433-444.	0.2	0