Sinda Zarrouk Mahjoub

List of Publications by Year
in descending order

147 papers	600 citations	687220 13 h-index	677027 22 g-index
149 all docs	149 docs citations	149 times ranked	780 citing authors

#	Article	IF	Citations
1	Comment on ``Long-term results of palpebral fissure transfer with no lower eyelid spacer in chronic progressive external ophthalmoplegia''. American Journal of Ophthalmology, 2022, 236, 319.	1.7	0
2	Fatigue and Exercise Intolerance as Initial Manifestations of a Nonsyndromic Mitochondrial Disorder Due to the Variant m.3243A>G. Case Reports in Neurological Medicine, 2022, 2022, 1-3.	0.3	0
3	Left ventricular hypertrabeculation/noncompaction in hyperoxaluria. Turk Kardiyoloji Dernegi Arsivi, 2021, 43, 585-585.	0.6	0
4	Single nucleotide mtDNA polymorphisms may contribute to cancerogenesis in mitochondrial disorders. Environmental and Molecular Mutagenesis, 2019, 60, 463-464.	0.9	0
5	Headache in mitochondrial disorders. Clinical Neurology and Neurosurgery, 2018, 166, 44-49.	0.6	14
6	Frequency of Headache in Mitochondrial Disorders. Headache, 2018, 58, 313-314.	1.8	1
7	Ketogenic diet and avoidance of mitochondrion-toxic AEDs may improve the outcome of mitochondrial epilepsy. Clinical Neurology and Neurosurgery, 2018, 173, 202-203.	0.6	2
8	Re: Guy etÂal.: Gene therapy for Leber hereditary optic neuropathy: low-and medium-dose visual results (Ophthalmology . 2017;124:1621-1634). Ophthalmology, 2018, 125, e14-e15.	2.5	5
9	Phenotypic and Genotypic Heterogeneity of RRM2B Variants. Neuropediatrics, 2018, 49, 231-237.	0.3	12
10	Fibroblast growth-factor-21 is currently a weak biomarker for identifying mitochondrial and non-mitochondrial inborn errors of metabolism. Molecular Genetics and Metabolism Reports, 2018, 14, 1-2.	0.4	0
11	Low Heteroplasmy Rates of the m.8993T>G Variant May Not Be Pathogenic. American Journal of Kidney Diseases, 2018, 71, 759.	2.1	0
12	Polymegathism as a biomarker of mitochondrial disorders. Graefe's Archive for Clinical and Experimental Ophthalmology, 2018, 256, 1211-1212.	1.0	0
13	Phenotypic variability of MTO1-deficiency. Molecular Genetics and Metabolism Reports, 2018, 15, 28-29.	0.4	3
14	Phenotypic manifestations of the m.8969G>A variant. Neurogenetics, 2018, 19, 131-132.	0.7	0
15	Regional cerebral hyperperfusion: A biomarker of upcoming strokeâ€like episodes?. Journal of Magnetic Resonance Imaging, 2018, 47, 582-582.	1.9	0
16	The cerebellum is a common site of affection in Leigh syndrome. Metabolic Brain Disease, 2018, 33, 11-12.	1.4	0
17	The Cerebellum Is a Common Site of Affection in Leigh Syndrome. Pediatric Neurology, 2018, 78, e9.	1.0	1
18	Avoid mitochondrion-toxic antiepileptic drugs in glycine encephalopathy. Brain and Development, 2018, 40, 366.	0.6	0

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19	Is vatiquinone truly beneficial for Leigh syndrome?. Brain and Development, 2018, 40, 443.	0.6	1
20	Comment to the manuscript by Bacalhau etÂal. on â€œIn silico prediction is insufficient to assess pathogenicity of mtDNA variantsâ€• European Journal of Medical Genetics, 2018, 61, 98-99.	0.7	0
21	Anti-mitochondrial M2 Antibodies and Myopathy. Internal Medicine, 2018, 57, 1187-1187.	0.3	1
22	Involvement of the Spinal Cord in Mitochondrial Disorders. Journal of Neurosciences in Rural Practice, 2018, 09, 245-251.	0.3	23
23	Increased mtDNA Copy Number Does Not Protect Against LHON. , 2018, 59, 330.		3
24	Penetrance of the LHON Mutation m.11778G>A May Depend on Factors Other Than the Haplotype or Heteroplasmy Rate. , 2018, 59, 381.		1
25	MELAS can be psychiatric and neurological. ENeurologicalSci, 2018, 11, 3-4.	0.5	2
26	BAG3 -related myofibrillar myopathy requiring heart transplantation for restrictive cardiomyopathy. Molecular Genetics and Metabolism Reports, 2018, 15, 65-66.	0.4	3
27	MicroRNAs are inappropriate for characterising hearing impairment in mitochondrial disorders. Orphanet Journal of Rare Diseases, 2018, 13, 87.	1.2	0
28	Cerebral imaging in paediatric mitochondrial disorders. Neuroradiology Journal, 2018, 31, 596-608.	0.6	8
29	Biomarkers for Detecting Mitochondrial Disorders. Journal of Clinical Medicine, 2018, 7, 16.	1.0	30
30	Modified Yarham and Smith scores for pathogenicity assessment of mtDNA tRNA variants. Neuromuscular Disorders, 2018, 28, 373-374.	0.3	0
31	Causes of low muscle coenzyme-Q levels beyond primary coenzyme-Q-deficiency. Molecular Genetics and Metabolism Reports, 2018, 15, 96-97.	0.4	0
32	A beneficial effect of l -arginine for stroke-like episodes is currently unsupported. Molecular Genetics and Metabolism Reports, 2018, 15, 67-68.	0.4	3
33	Gastrointestinal Involvement in m.3243A>G-associated MELAS. Internal Medicine, 2018, 57, 769-770.	0.3	5
34	Ophthalmologic involvement in Leigh syndrome. Acta Ophthalmologica, 2017, 95, e75-e76.	0.6	0
35	Phenotypic heterogeneity of a compound heterozygous SUCLA2 mutation. Molecular Genetics and Metabolism Reports, 2017, 10, 31-32.	0.4	1
36	Levels of nitric oxide pathway parameters may depend on heteroplasmy rates of the m.3243A>G mutation. International Journal of Cardiology, 2017, 229, 26.	0.8	1

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37	ND2 mutation with minimal coenzyme-Q responsive manifestations. Molecular Genetics and Metabolism Reports, 2017, 10, 83.	0.4	0
38	Low blood heteroplasmy-rate may cause late-onset MELAS. Molecular Genetics and Metabolism Reports, 2017, 10, 100.	0.4	1
39	Compound heterozygous fukutin mutation-related non-compaction. International Journal of Cardiology, 2017, 233, 101.	0.8	1
40	Death in Pediatric Mitochondrial Disorders. Pediatric Neurology, 2017, 73, e1.	1.0	2
41	A MELAS phenotype is not necessarily MELAS. Tropical Doctor, 2017, 47, 191-192.	0.2	0
42	Unilateral Ptosis and Homolateral Hemifacial Weakness in Chronic Progressive External Ophthalmoplegia. Neuro-Ophthalmology, 2017, 41, 165-166.	0.4	1
43	Epilepsy in MELAS. Pediatric Neurology, 2017, 67, e7-e8.	1.0	1
44	Epilepsia partialis continua in MELAS/Leigh overlap syndrome. Brain and Development, 2017, 39, 365.	0.6	0
45	Macroangiopathy is a typical phenotypic manifestation of MELAS. Metabolic Brain Disease, 2017, 32, 977-979.	1.4	3
46	MELAS/Leigh overlap syndrome due to the ND6 mutation m.10158T>C. Brain and Development, 2017, 39, 724.	0.6	1
47	A commentary on homozygous p.(Glu87Lys) variant in ISCA1 is associated with a multiple mitochondrial dysfunctions syndrome. Journal of Human Genetics, 2017, 62, 865-866.	1.1	5
48	Mitochondrial myopathy, dysmorphism, exercise-induced vomiting and tachycardia the mutation m.4831G > A. Molecular Genetics and Metabolism Reports, 2017, 11, 74.	0.4	0
49	Hypogonadism in mitochondrial disorders. Journal of Pediatrics, 2017, 187, 334.	0.9	0
50	Clozapine for mitochondrial psychosis. Molecular Genetics and Metabolism Reports, 2017, 10, 50.	0.4	1
51	Phenotypic spectrum of DARS2 mutations. Journal of the Neurological Sciences, 2017, 376, 117-118.	0.3	5
52	Phenotypic and genotypic features in pediatric and adult mitochondrial disorders. Muscle and Nerve, 2017, 56, E48.	1.0	0
53	NDUFS4â€related Leigh syndrome in Hutterites. American Journal of Medical Genetics, Part A, 2017, 173, 1450-1451.	0.7	5
54	Mitochondrial disorders due to tRNA(Pro) mutations. Neuromuscular Disorders, 2017, 27, 791.	0.3	1

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55	Onset of MELAS due to the m.3243A > G mutation is early if the large phenotypic variability is considered. Molecular Genetics and Metabolism Reports, 2017, 10, 23.	0.4	2
56	Differentiating acute ischemic lesions from mitochondrial strokeâ€like lesions on 3D pseudoâ€continuous arterial spin labelling. Journal of Magnetic Resonance Imaging, 2017, 45, 937-937.	1.9	0
57	Phenotypic heterogeneity of MELAS. Molecular Genetics and Metabolism Reports, 2017, 10, 18-19.	0.4	4
58	Variants m.13276G > A in the MT-ND5 and m.8241T > G in the MT-CO2 gene are not responsible for maternally inherited diabetes and deafness. Journal of Diabetes and Its Complications, 2017, 31, 1638.	1.2	0
59	Pathogenicity of the LHON variant m.3472T > C is uncertain. Journal of the Neurological Sciences, 2017, 382, 164-165.	0.3	0
60	Serious assessment of mtDNA variants is a prerequisite to confirm pathogenicity. Journal of the Neurological Sciences, 2017, 381, 341-342.	0.3	0
61	Can <scp>MR</scp> spectroscopy and muscle biopsy findings be correlated with <scp>MELAS</scp> and <scp>CPEO</scp>?. CNS Neuroscience and Therapeutics, 2017, 23, 846-847.	1.9	0
62	Reduced Bone Mineral Density in m.3243A>G Carriers May Be Multifactorial. Journal of Bone and Mineral Research, 2017, 32, 2315-2316.	3.1	0
63	Leigh-like syndrome due to OPA1 mutations. European Journal of Paediatric Neurology, 2017, 21, 921-922.	0.7	1
64	Affection of immune cells by a C10orf2 mutation manifesting as mitochondrial myopathy and transient sensory transverse syndrome. Acta Neurologica Belgica, 2017, 117, 969-970.	0.5	6
65	Striatal necrosis due to the m.14459G>A mutation. Journal of the Neurological Sciences, 2017, 380, 281-282.	0.3	0
66	Mitochondrial cardioencephalopathy due to a COQ4 mutation. Molecular Genetics and Metabolism Reports, 2017, 13, 7-8.	0.4	1
67	Mitochondrial multiorgan disorder syndrome (MIMODS) due to a compound heterozygous mutation in the ACAD9 gene. Molecular Genetics and Metabolism Reports, 2017, 13, 31-32.	0.4	3
68	Involvement of the cerebral veins in MELAS syndrome?. Neuroradiology, 2017, 59, 833-834.	1.1	1
69	Correspondence. Retina, 2017, 37, e88-e89.	1.0	0
70	Axonal hyperexcitability due to Schwann cell involvement in chronic progressive external ophthalmoplegia. Clinical Neurophysiology, 2017, 128, 2096-2097.	0.7	1
71	Costs for mitochondrial medicine will remain high as long as mitochondrial disorders are misdiagnosed. Molecular Genetics and Metabolism Reports, 2017, 13, 41.	0.4	0
72	Treating mitochondrial disorders requires full exploitation of available therapeutic options. Molecular Genetics and Metabolism Reports, 2017, 13, 97-98.	0.4	1

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73	Management of epilepsy in MERRF syndrome. Seizure: the Journal of the British Epilepsy Association, 2017, 50, 166-170.	0.9	22
74	Do lesional perfusion abnormalities on arterial spin labeling truly contribute to the diagnosis of Leigh syndrome?. Pediatric Radiology, 2017, 47, 124-125.	1.1	1
75	Why does Leigh syndrome respond to immunotherapy?. Molecular Genetics and Metabolism Reports, 2017, 11, 90-91.	0.4	3
76	Ophthalmologic involvement in mitochondrial disorders. Ophthalmic Genetics, 2017, 38, 298-298.	0.5	2
77	POLG1 mutations in bipolar disorders. Psychiatry and Clinical Neurosciences, 2017, 71, 569-569.	1.0	0
78	Mitochondrial multiorgan disorder syndrome score generated from definite mitochondrial disorders. Neuropsychiatric Disease and Treatment, 2017, Volume 13, 2569-2579.	1.0	18
79	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, and Stroke-like Episodes (MELAS) due to a m.10158T>C ND3 Mutation with a Normal Muscle Biopsy. Internal Medicine, 2017, 56, 2693-2693.	0.3	0
80	Letter to the Editor: Maternally Inherited Diabetes and Deafness is Not Only Biorgan but Multiorgan. Acta Medica Portuguesa, 2017, 30, 665.	0.2	0
81	Psychological morbidity in Leber’s hereditary optic neuropathy depends on phenotypic, social, economic, and genetic factors. Clinical Ophthalmology, 2017, Volume 11, 959-962.	0.9	1
82	Heteroplasmy of the m.3243A>G Mutation May Influence Phenotypic Heterogeneity. Internal Medicine, 2017, 56, 3123-3123.	0.3	0
83	Diagnosing Kearns-Sayre Syndrome Requires Genetic Confirmation. Chinese Medical Journal, 2016, 129, 2267-2268.	0.9	1
84	Detection of the mutation may guide treatment of heart and muscle in Duchenne muscular dystrophy. International Medical Case Reports Journal, 2016, 9, 55.	0.3	0
85	Diagnosis of Kearns-Sayre Syndrome Requires Comprehensive Work-up. Chinese Medical Journal, 2016, 129, 2518-2519.	0.9	1
86	Leber’s hereditary optic neuropathy is multiorgan not mono-organ. Clinical Ophthalmology, 2016, Volume 10, 2187-2190.	0.9	40
87	Diagnose Kearnsâ€“Sayre syndrome genetically and investigate the phenotype comprehensively. Oxford Medical Case Reports, 2016, 2016, omw059.	0.2	1
88	Is chronic fatigue syndrome truly associated with haplogroups or mtDNA single nucleotide polymorphisms?. Journal of Translational Medicine, 2016, 14, 182.	1.8	3
89	Nitric Oxide Deficiency Triggering Strokelike Episodes. JAMA Neurology, 2016, 73, 1029.	4.5	0
90	Noncompaction in Duchenne Muscular Dystrophy. Internal Medicine, 2016, 55, 1241-1241.	0.3	0

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91	Posterior Spinal Instrumented Fusion for Idiopathic Scoliosis in Patients with Multisystemic Neurodegenerative Disorder: A Report of Two Cases. Journal of Orthopaedic Surgery, 2016, 24, 428-428.	0.4	0
92	Letter by Finsterer and Zarrouk-Mahjoub Regarding Article, â€œCombination of Whole Genome Sequencing, Linkage, and Functional Studies Implicates a Missense Mutation in Titin as a Cause of Autosomal Dominant Cardiomyopathy With Features of Left Ventricular Noncompactionâ€• Circulation: Cardiovascular Genetics, 2016, 9, 579-579.	5.1	1
93	Peculiarities of progressive external ophthalmoplegia due to single mitochondrial DNA deletions. Journal of the Formosan Medical Association, 2016, 115, 1099-1100.	0.8	2
94	Double trouble progressive external ophthalmoplegia and Huntington's disease. Molecular Genetics and Metabolism Reports, 2016, 7, 77.	0.4	2
95	Unusual recovery of respiratory chain complex-III deficiency upon G-tube feeding and a cocktail. Molecular Genetics and Metabolism Reports, 2016, 7, 92.	0.4	0
96	Affection of the frontal lobe in Leigh syndrome due to the m.8993 T > G mutation. Journal of the Neurological Sciences, 2016, 366, 249-250.	0.3	1
97	Managing seizures in mitochondrial disorders. Expert Opinion on Orphan Drugs, 2016, 4, 197-204.	0.5	1
98	In the heart of MELAS syndrome. International Journal of Cardiology, 2016, 214, 157-158.	0.8	3
99	MELAS syndrome due to the m.3291T>C mutation. Molecular Genetics and Metabolism Reports, 2016, 7, 50.	0.4	1
100	Acute heart failure from noncompaction requiring emergency heart transplantation. Revista Portuguesa De Cardiologia, 2016, 35, 507-508.	0.2	2
101	Assessment of the phenotype genotype variability and correlation in m.3243A > G mutation carriers requires prospective studies. Molecular Genetics and Metabolism Reports, 2016, 8, 33.	0.4	1
102	Nephrocalcinosis and retinal dystrophy, rare manifestations of MPV17-related mitochondrial depletion syndrome?. Molecular Genetics and Metabolism Reports, 2016, 9, 18.	0.4	1
103	Mitochondrial vasculopathy due to the m.3243A>G mutation is not restricted to the carotid artery. Molecular Genetics and Metabolism Reports, 2016, 9, 34.	0.4	1
104	Fukutin mutations in Fukuyama congenital muscular dystrophy do not cause noncompaction. International Journal of Cardiology, 2016, 225, 75-76.	0.8	2
105	Spectrum of Movement Disorders in Mitochondrial Disorders. JAMA Neurology, 2016, 73, 1253.	4.5	3
106	Cerebral involvement in mitochondrial disorders on imaging. Child's Nervous System, 2016, 32, 2059-2060.	0.6	3
107	Hearing impairment in m.3243A>G carriers requires comprehensive work- and follow-up. Clinical Neurology and Neurosurgery, 2016, 150, 199-200.	0.6	1
108	Re: Feuer etÂal.: Gene therapy for Leber hereditary optic neuropathy: initial results (Ophthalmology) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5	2.5	2

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109	Treatment of muscle weakness in neuromuscular disorders. Expert Review of Neurotherapeutics, 2016, 16, 1383-1395.	1.4	6
110	Prevalence and Outcome of Mitochondrial Epilepsy. Annals of Neurology, 2016, 80, 313-314.	2.8	1
111	Corneal Involvement in Kearnsâ€“Sayre Syndrome Responsive to Coenzyme-Q?. Cornea, 2016, 35, e39.	0.9	2
112	Mitochondrial neuropathy affects peripheral and cranial nerves and is primary or secondary or both. Neuromuscular Disorders, 2016, 26, 548-549.	0.3	2
113	Arrhythmias in MELAS syndrome. Molecular Genetics and Metabolism Reports, 2016, 7, 54.	0.4	1
114	Phenotypic spectrum of the m.8344A>G mutation. Journal of Neurology, 2016, 263, 1452-1453.	1.8	1
115	Noncompaction with valve abnormalities is rarely associated with neurologic or genetic disease. International Journal of Cardiology, 2016, 202, 627-628.	0.8	0
116	The Eye on Mitochondrial Disorders. Journal of Child Neurology, 2016, 31, 652-662.	0.7	23
117	Mitochondrial Disorders May Mimic Amyotrophic Lateral Sclerosis at Onset. Sultan Qaboos University Medical Journal, 2016, 16, e92-95.	0.3	8
118	Noncompaction on scleroderma. Turk Kardiyoloji Dernegi Arsivi, 2016, 44, 190-1.	0.6	0
119	Transient symmetric T2-hyperintensities of basal ganglia and brainstem not only point to Leigh syndrome. Annals of Indian Academy of Neurology, 2016, 19, 419.	0.2	0
120	Pathogenicity of the Homoplasmic m.8701A>G Variant Requires Confirmation. Chinese Medical Journal, 2016, 129, 1889-1890.	0.9	0
121	Implications of non-compaction in association with respiratory chain complex-I deficiency. Cardiology in the Young, 2015, 25, 1022-1023.	0.4	0
122	Considerations about the genetics of left ventricular hypertrabeculation/non-compaction. Cardiology in the Young, 2015, 25, 1435-1437.	0.4	17
123	Unusual Myocardial Late Gadolinium Enhancement in Isolated Noncompaction Cardiomyopathy. Echocardiography, 2015, 32, 723-724.	0.3	0
124	Compound heterozygous myotonic dystrophy type 1. American Journal of Medical Genetics, Part A, 2015, 167, 1952-1953.	0.7	0
125	Impaired Hearing in Mitochondrial Disorders. Chinese Medical Journal, 2015, 128, 1839.	0.9	0
126	Hypertrabeculation Is Noncompaction Also in Athletes. American Journal of Cardiology, 2015, 116, 831.	0.7	0

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127	Mitochondrial toxicity of cardiac drugs and its relevance to mitochondrial disorders. Expert Opinion on Drug Metabolism and Toxicology, 2015, 11, 15-24.	1.5	22
128	El trasfondo genÃ©tico de la hipertrabeculaciÃ³n/miocardiopatÃa no compactada ventricular izquierda sigue sin estar claro. Revista Espanola De Cardiologia, 2015, 68, 166-167.	0.6	0
129	Focal and Generalized Seizures May Occur in Mitochondrial Encephalomyopathy, Lactic Acidosis, and Strokelike Episodes (MELAS) Patients. Journal of Child Neurology, 2015, 30, 1553-1554.	0.7	11
130	Management of juvenile stroke from noncompaction with muscular dystrophy. International Journal of Cardiology, 2015, 184, 371-372.	0.8	0
131	Noncompaction with dysmorphism, mental retardation, general wasting, and hypogonadism requires neurologic and sophisticated cytogenetic investigations. Anatolian Journal of Cardiology, 2015, 15, 433-434.	0.5	0
132	Consider a Nongenetic Pathogenesis of Noncompaction. Pediatric Cardiology, 2015, 36, 1548-1549.	0.6	0
133	Noncompaction in Fabry's disease. Revista Portuguesa De Cardiologia, 2015, 34, 299-300.	0.2	0
134	Lamin A/C Mutations Do Not Cause Left Ventricular Hypertrabeculation/Noncompaction. Texas Heart Institute Journal, 2015, 42, 301-302.	0.1	3
135	Uniform criteria for diagnosing noncompaction by cMRI and echocardiography are warranted. Anatolian Journal of Cardiology, 2015, 15, 959-960.	0.5	0
136	Consider a Mitochondrial Disorder when Left Ventricular Hypertrabeculation/Noncompaction is Associated with Renal Cysts. Texas Heart Institute Journal, 2014, 41, 677-678.	0.1	2
137	Left ventricular noncompaction associated with a compound heterozygous MYBPC3 mutation. European Journal of Medical Genetics, 2014, 57, 349.	0.7	2
138	Mitochondrial tRNA mutations manifest not only as hypertrophic cardiomyopathy but also as noncompaction. Human Pathology, 2014, 45, 1790-1791.	1.1	0
139	Increased prevalence of non-compaction. European Journal of Internal Medicine, 2014, 25, e20.	1.0	1
140	Acquired non-compaction in integrin-myopathy. Orphanet Journal of Rare Diseases, 2013, 8, 183.	1.2	0
141	Pulmonary hypertension in mitochondrial disorders. Brain and Development, 2013, 35, 466.	0.6	3
142	Grant et al. 1926 did not provide the first description of left ventricular hypertrabeculation/noncompaction. International Journal of Cardiology, 2013, 169, e51-e52.	0.8	8
143	Pathogenicity of the Transition m.3308T>C in Left Ventricular Hypertrabeculation/Noncompaction. Cardiology, 2012, 122, 116-118.	0.6	2
144	Mitochondrial toxicity of antiepileptic drugs and their tolerability in mitochondrial disorders. Expert Opinion on Drug Metabolism and Toxicology, 2012, 8, 71-79.	1.5	93

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145	Epilepsy in mitochondrial disorders. Seizure: the Journal of the British Epilepsy Association, 2012, 21, 316-321.	0.9	73
146	Leukoencephalopathies in Mitochondrial Disorders: Clinical and MRI Findings. Journal of Neuroimaging, 2012, 22, e1-11.	1.0	14
147	Transition m.3308T>C in the ND1 Gene Is Associated with Left Ventricular Hypertrabeculation/Noncompaction. Cardiology, 2011, 118, 153-158.	0.6	17