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	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
2	Epilepsy in mitochondrial disorders. Seizure: the Journal of the British Epilepsy Association, 2012, 21, 316-321.	0.9	73
3	Leber’s hereditary optic neuropathy is multiorgan not mono-organ. Clinical Ophthalmology, 2016, Volume 10, 2187-2190.	0.9	40
4	Biomarkers for Detecting Mitochondrial Disorders. Journal of Clinical Medicine, 2018, 7, 16.	1.0	30
5	The Eye on Mitochondrial Disorders. Journal of Child Neurology, 2016, 31, 652-662.	0.7	23
6	Involvement of the Spinal Cord in Mitochondrial Disorders. Journal of Neurosciences in Rural Practice, 2018, 09, 245-251.	0.3	23
7	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
8	Management of epilepsy in MERRF syndrome. Seizure: the Journal of the British Epilepsy Association, 2017, 50, 166-170.	0.9	22
9	Mitochondrial multiorgan disorder syndrome score generated from definite mitochondrial disorders. Neuropsychiatric Disease and Treatment, 2017, Volume 13, 2569-2579.	1.0	18
10	Transition m.3308T>C in the ND1 Gene Is Associated with Left Ventricular Hypertrabeculation/Noncompaction. Cardiology, 2011, 118, 153-158.	0.6	17
11	Considerations about the genetics of left ventricular hypertrabeculation/non-compaction. Cardiology in the Young, 2015, 25, 1435-1437.	0.4	17
12	Leukoencephalopathies in Mitochondrial Disorders: Clinical and MRI Findings. Journal of Neuroimaging, 2012, 22, e1-11.	1.0	14
13	Headache in mitochondrial disorders. Clinical Neurology and Neurosurgery, 2018, 166, 44-49.	0.6	14
14	Phenotypic and Genotypic Heterogeneity of RRM2B Variants. Neuropediatrics, 2018, 49, 231-237.	0.3	12
15	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
16	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
17	Cerebral imaging in paediatric mitochondrial disorders. Neuroradiology Journal, 2018, 31, 596-608.	0.6	8
18	Mitochondrial Disorders May Mimic Amyotrophic Lateral Sclerosis at Onset. Sultan Qaboos University Medical Journal, 2016, 16, e92-95.	0.3	8

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19	Treatment of muscle weakness in neuromuscular disorders. Expert Review of Neurotherapeutics, 2016, 16, 1383-1395.	1.4	6
20	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
21	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
22	Phenotypic spectrum of DARS2 mutations. Journal of the Neurological Sciences, 2017, 376, 117-118.	0.3	5
23	NDUFS4â€related Leigh syndrome in Hutterites. American Journal of Medical Genetics, Part A, 2017, 173, 1450-1451.	0.7	5
24	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
25	Gastrointestinal Involvement in m.3243A>G-associated MELAS. Internal Medicine, 2018, 57, 769-770.	0.3	5
26	Phenotypic heterogeneity of MELAS. Molecular Genetics and Metabolism Reports, 2017, 10, 18-19.	0.4	4
27	Pulmonary hypertension in mitochondrial disorders. Brain and Development, 2013, 35, 466.	0.6	3
28	Is chronic fatigue syndrome truly associated with haplogroups or mtDNA single nucleotide polymorphisms?. Journal of Translational Medicine, 2016, 14, 182.	1.8	3
29	In the heart of MELAS syndrome. International Journal of Cardiology, 2016, 214, 157-158.	0.8	3
30	Spectrum of Movement Disorders in Mitochondrial Disorders. JAMA Neurology, 2016, 73, 1253.	4.5	3
31	Cerebral involvement in mitochondrial disorders on imaging. Child's Nervous System, 2016, 32, 2059-2060.	0.6	3
32	Macroangiopathy is a typical phenotypic manifestation of MELAS. Metabolic Brain Disease, 2017, 32, 977-979.	1.4	3
33	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
34	Why does Leigh syndrome respond to immunotherapy?. Molecular Genetics and Metabolism Reports, 2017, 11, 90-91.	0.4	3
35	Phenotypic variability of MTO1-deficiency. Molecular Genetics and Metabolism Reports, 2018, 15, 28-29.	0.4	3
36	Increased mtDNA Copy Number Does Not Protect Against LHON. , 2018, 59, 330.		3

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37	BAG3 -related myofibrillar myopathy requiring heart transplantation for restrictive cardiomyopathy. Molecular Genetics and Metabolism Reports, 2018, 15, 65-66.	0.4	3
38	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
39	Lamin A/C Mutations Do Not Cause Left Ventricular Hypertrabeculation/Noncompaction. Texas Heart Institute Journal, 2015, 42, 301-302.	0.1	3
40	Pathogenicity of the Transition m.3308T>C in Left Ventricular Hypertrabeculation/Noncompaction. Cardiology, 2012, 122, 116-118.	0.6	2
41	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
42	Left ventricular noncompaction associated with a compound heterozygous MYBPC3 mutation. European Journal of Medical Genetics, 2014, 57, 349.	0.7	2
43	Peculiarities of progressive external ophthalmoplegia due to single mitochondrial DNA deletions. Journal of the Formosan Medical Association, 2016, 115, 1099-1100.	0.8	2
44	Double trouble progressive external ophthalmoplegia and Huntington's disease. Molecular Genetics and Metabolism Reports, 2016, 7, 77.	0.4	2
45	Acute heart failure from noncompaction requiring emergency heart transplantation. Revista Portuguesa De Cardiologia, 2016, 35, 507-508.	0.2	2
46	Fukutin mutations in Fukuyama congenital muscular dystrophy do not cause noncompaction. International Journal of Cardiology, 2016, 225, 75-76.	0.8	2
47	Re: Feuer etÂal.: Gene therapy for Leber hereditary optic neuropathy: initial results (Ophthalmology) Tj ETQq1 1 0.784314 rgBT /Overl	2.5	2
48	Corneal Involvement in Kearnsâ€“Sayre Syndrome Responsive to Coenzyme-Q?. Cornea, 2016, 35, e39.	0.9	2
49	Mitochondrial neuropathy affects peripheral and cranial nerves and is primary or secondary or both. Neuromuscular Disorders, 2016, 26, 548-549.	0.3	2
50	Death in Pediatric Mitochondrial Disorders. Pediatric Neurology, 2017, 73, e1.	1.0	2
51	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
52	Ophthalmologic involvement in mitochondrial disorders. Ophthalmic Genetics, 2017, 38, 298-298.	0.5	2
53	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
54	MELAS can be psychiatric and neurological. ENeurologicalSci, 2018, 11, 3-4.	0.5	2

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55	Increased prevalence of non-compaction. European Journal of Internal Medicine, 2014, 25, e20.	1.0	1
56	Diagnosing Kearns-Sayre Syndrome Requires Genetic Confirmation. Chinese Medical Journal, 2016, 129, 2267-2268.	0.9	1
57	Diagnosis of Kearns-Sayre Syndrome Requires Comprehensive Work-up. Chinese Medical Journal, 2016, 129, 2518-2519.	0.9	1
58	Diagnose Kearnsâ€“Sayre syndrome genetically and investigate the phenotype comprehensively. Oxford Medical Case Reports, 2016, 2016, omw059.	0.2	1
59	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
60	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
61	Managing seizures in mitochondrial disorders. Expert Opinion on Orphan Drugs, 2016, 4, 197-204.	0.5	1
62	MELAS syndrome due to the m.3291T>C mutation. Molecular Genetics and Metabolism Reports, 2016, 7, 50.	0.4	1
63	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
64	Nephrocalcinosis and retinal dystrophy, rare manifestations of MPV17-related mitochondrial depletion syndrome?. Molecular Genetics and Metabolism Reports, 2016, 9, 18.	0.4	1
65	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
66	Hearing impairment in m.3243A>G carriers requires comprehensive work- and follow-up. Clinical Neurology and Neurosurgery, 2016, 150, 199-200.	0.6	1
67	Prevalence and Outcome of Mitochondrial Epilepsy. Annals of Neurology, 2016, 80, 313-314.	2.8	1
68	Arrhythmias in MELAS syndrome. Molecular Genetics and Metabolism Reports, 2016, 7, 54.	0.4	1
69	Phenotypic spectrum of the m.8344A>G mutation. Journal of Neurology, 2016, 263, 1452-1453.	1.8	1
70	Phenotypic heterogeneity of a compound heterozygous SUCLA2 mutation. Molecular Genetics and Metabolism Reports, 2017, 10, 31-32.	0.4	1
71	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
72	Low blood heteroplasmy-rate may cause late-onset MELAS. Molecular Genetics and Metabolism Reports, 2017, 10, 100.	0.4	1

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73	Compound heterozygous fukutin mutation-related non-compaction. International Journal of Cardiology, 2017, 233, 101.	0.8	1
74	Unilateral Ptosis and Homolateral Hemifacial Weakness in Chronic Progressive External Ophthalmoplegia. Neuro-Ophthalmology, 2017, 41, 165-166.	0.4	1
75	Epilepsy in MELAS. Pediatric Neurology, 2017, 67, e7-e8.	1.0	1
76	MELAS/Leigh overlap syndrome due to the ND6 mutation m.10158T>C. Brain and Development, 2017, 39, 724.	0.6	1
77	Clozapine for mitochondrial psychosis. Molecular Genetics and Metabolism Reports, 2017, 10, 50.	0.4	1
78	Mitochondrial disorders due to tRNA(Pro) mutations. Neuromuscular Disorders, 2017, 27, 791.	0.3	1
79	Leigh-like syndrome due to OPA1 mutations. European Journal of Paediatric Neurology, 2017, 21, 921-922.	0.7	1
80	Mitochondrial cardioencephalopathy due to a COQ4 mutation. Molecular Genetics and Metabolism Reports, 2017, 13, 7-8.	0.4	1
81	Involvement of the cerebral veins in MELAS syndrome?. Neuroradiology, 2017, 59, 833-834.	1.1	1
82	Axonal hyperexcitability due to Schwann cell involvement in chronic progressive external ophthalmoplegia. Clinical Neurophysiology, 2017, 128, 2096-2097.	0.7	1
83	Treating mitochondrial disorders requires full exploitation of available therapeutic options. Molecular Genetics and Metabolism Reports, 2017, 13, 97-98.	0.4	1
84	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
85	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
86	Frequency of Headache in Mitochondrial Disorders. Headache, 2018, 58, 313-314.	1.8	1
87	The Cerebellum Is a Common Site of Affection in Leigh Syndrome. Pediatric Neurology, 2018, 78, e9.	1.0	1
88	Is vatiquinone truly beneficial for Leigh syndrome?. Brain and Development, 2018, 40, 443.	0.6	1
89	Anti-mitochondrial M2 Antibodies and Myopathy. Internal Medicine, 2018, 57, 1187-1187.	0.3	1
90	Penetrance of the LHON Mutation m.11778G>A May Depend on Factors Other Than the Haplotype or Heteroplasmy Rate. , 2018, 59, 381.		1

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91	Acquired non-compaction in integrin-myopathy. Orphanet Journal of Rare Diseases, 2013, 8, 183.	1.2	0
92	Mitochondrial tRNA mutations manifest not only as hypertrophic cardiomyopathy but also as noncompaction. Human Pathology, 2014, 45, 1790-1791.	1.1	0
93	Implications of non-compaction in association with respiratory chain complex-I deficiency. Cardiology in the Young, 2015, 25, 1022-1023.	0.4	0
94	Unusual Myocardial Late Gadolinium Enhancement in Isolated Noncompaction Cardiomyopathy. Echocardiography, 2015, 32, 723-724.	0.3	0
95	Compound heterozygous myotonic dystrophy type 1. American Journal of Medical Genetics, Part A, 2015, 167, 1952-1953.	0.7	0
96	Impaired Hearing in Mitochondrial Disorders. Chinese Medical Journal, 2015, 128, 1839.	0.9	0
97	Hypertrabeculation Is Noncompaction Also in Athletes. American Journal of Cardiology, 2015, 116, 831.	0.7	0
98	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
99	Management of juvenile stroke from noncompaction with muscular dystrophy. International Journal of Cardiology, 2015, 184, 371-372.	0.8	0
100	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
101	Consider a Nongenetic Pathogenesis of Noncompaction. Pediatric Cardiology, 2015, 36, 1548-1549.	0.6	0
102	Noncompaction in Fabry's disease. Revista Portuguesa De Cardiologia, 2015, 34, 299-300.	0.2	0
103	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
104	Nitric Oxide Deficiency Triggering Strokelike Episodes. JAMA Neurology, 2016, 73, 1029.	4.5	0
105	Noncompaction in Duchenne Muscular Dystrophy. Internal Medicine, 2016, 55, 1241-1241.	0.3	0
106	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
107	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
108	Noncompaction with valve abnormalities is rarely associated with neurologic or genetic disease. International Journal of Cardiology, 2016, 202, 627-628.	0.8	0

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109	Ophthalmologic involvement in Leigh syndrome. Acta Ophthalmologica, 2017, 95, e75-e76.	0.6	0
110	ND2 mutation with minimal coenzyme-Q responsive manifestations. Molecular Genetics and Metabolism Reports, 2017, 10, 83.	0.4	0
111	A MELAS phenotype is not necessarily MELAS. Tropical Doctor, 2017, 47, 191-192.	0.2	0
112	Epilepsia partialis continua in MELAS/Leigh overlap syndrome. Brain and Development, 2017, 39, 365.	0.6	0
113	Mitochondrial myopathy, dysmorphism, exercise-induced vomiting and tachycardia the mutation m.4831G > A. Molecular Genetics and Metabolism Reports, 2017, 11, 74.	0.4	0
114	Hypogonadism in mitochondrial disorders. Journal of Pediatrics, 2017, 187, 334.	0.9	0
115	Phenotypic and genotypic features in pediatric and adult mitochondrial disorders. Muscle and Nerve, 2017, 56, E48.	1.0	0
116	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
117	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
118	Pathogenicity of the LHON variant m.3472T > C is uncertain. Journal of the Neurological Sciences, 2017, 382, 164-165.	0.3	0
119	Serious assessment of mtDNA variants is a prerequisite to confirm pathogenicity. Journal of the Neurological Sciences, 2017, 381, 341-342.	0.3	0
120	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
121	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
122	Striatal necrosis due to the m.14459G>A mutation. Journal of the Neurological Sciences, 2017, 380, 281-282.	0.3	0
123	Correspondence. Retina, 2017, 37, e88-e89.	1.0	0
124	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
125	POLG1 mutations in bipolar disorders. Psychiatry and Clinical Neurosciences, 2017, 71, 569-569.	1.0	0
126	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

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127	Letter to the Editor: Maternally Inherited Diabetes and Deafness is Not Only Biorgan but Multiorgan. Acta Medica Portuguesa, 2017, 30, 665.	0.2	0
128	Heteroplasmy of the m.3243A>G Mutation May Influence Phenotypic Heterogeneity. Internal Medicine, 2017, 56, 3123-3123.	0.3	0
129	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
130	Low Heteroplasmy Rates of the m.8993T>G Variant May Not Be Pathogenic. American Journal of Kidney Diseases, 2018, 71, 759.	2.1	0
131	Polymegathism as a biomarker of mitochondrial disorders. Graefe's Archive for Clinical and Experimental Ophthalmology, 2018, 256, 1211-1212.	1.0	0
132	Phenotypic manifestations of the m.8969G>A variant. Neurogenetics, 2018, 19, 131-132.	0.7	0
133	Regional cerebral hyperperfusion: A biomarker of upcoming strokeâ€like episodes?. Journal of Magnetic Resonance Imaging, 2018, 47, 582-582.	1.9	0
134	The cerebellum is a common site of affection in Leigh syndrome. Metabolic Brain Disease, 2018, 33, 11-12.	1.4	0
135	Avoid mitochondrion-toxic antiepileptic drugs in glycine encephalopathy. Brain and Development, 2018, 40, 366.	0.6	0
136	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
137	MicroRNAs are inappropriate for characterising hearing impairment in mitochondrial disorders. Orphanet Journal of Rare Diseases, 2018, 13, 87.	1.2	0
138	Modified Yarham and Smith scores for pathogenicity assessment of mtDNA tRNA variants. Neuromuscular Disorders, 2018, 28, 373-374.	0.3	0
139	Causes of low muscle coenzyme-Q levels beyond primary coenzyme-Q-deficiency. Molecular Genetics and Metabolism Reports, 2018, 15, 96-97.	0.4	0
140	Single nucleotide mtDNA polymorphisms may contribute to cancerogenesis in mitochondrial disorders. Environmental and Molecular Mutagenesis, 2019, 60, 463-464.	0.9	0
141	Left ventricular hypertrabeculation/noncompaction in hyperoxaluria. Turk Kardiyoloji Dernegi Arsivi, 2021, 43, 585-585.	0.6	0
142	Uniform criteria for diagnosing noncompaction by cMRI and echocardiography are warranted. Anatolian Journal of Cardiology, 2015, 15, 959-960.	0.5	0
143	Noncompaction on scleroderma. Turk Kardiyoloji Dernegi Arsivi, 2016, 44, 190-1.	0.6	0
144	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

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145	Pathogenicity of the Homoplasmic m.8701A>G Variant Requires Confirmation. Chinese Medical Journal, 2016, 129, 1889-1890.	0.9	0
146	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
147	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