Kyriaki Kekou

List of Publications by Year
in descending order

14	692	1684188 5	1281871 11
papers	citations	h-index	g-index

14	14	14	1151
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Homozygosity of the Zâ€2 polymorphic variant in the aldose reductase gene promoter confers increased risk for neuropathy in children and adolescents with Type 1 diabetes. Pediatric Diabetes, 2022, 23, 104-114.	2.9	1
2	Orofacial Muscle Weakening in Facioscapulohumeral Muscular Dystrophy (FSHD) Patients. Children, 2022, 9, 96.	1.5	1
3	Orofacial Manifestations Associated with Muscular Dystrophies: A Review. , 2022, 35, 67-73.		5
4	Phenotypeâ€driven variant filtration strategy in exome sequencing toward a high diagnostic yield and identification of 85 novel variants in 400 patients with rare Mendelian disorders. American Journal of Medical Genetics, Part A, 2021, 185, 2561-2571.	1.2	24
5	Evaluation of Genotypes and Epidemiology of Spinal Muscular Atrophy in Greece: A Nationwide Study Spanning 24 Years. Journal of Neuromuscular Diseases, 2020, 7, 247-256.	2.6	8
6	Single amino acid loss in the dystrophin protein associated with a mild clinical phenotype. Muscle and Nerve, 2017, 55, 46-50.	2.2	5
7	Clinical Outcomes in Duchenne Muscular Dystrophy: A Study of 5345 Patients from the TREAT-NMD DMD Global Database. Journal of Neuromuscular Diseases, 2017, 4, 293-306.	2.6	125
8	Author's Reply: Myotonic dystrophy: The occurrence of early-onset cataract. Neurology India, 2017, 65, 923.	0.4	0
9	Analysis of a founder mutation in the TH gene in a cohort of greek patients with Parkinson's disease. Movement Disorders, 2016, 31, 1753-1754.	3.9	2
10	A dynamic trinucleotide repeat (TNR) expansion in the DMD gene. Molecular and Cellular Probes, 2016, 30, 254-260.	2.1	3
11	Myotonic dystrophy type 2 presenting as inflammatory myopathy. Neurology India, 2016, 64, 1051.	0.4	0
12	The TREAT-NMD DMD Global Database: Analysis of More than 7,000 Duchenne Muscular Dystrophy Mutations. Human Mutation, 2015, 36, 395-402.	2.5	507
13	A simplified approach for FSHD molecular testing. Clinica Chimica Acta, 2014, 429, 96-103.	1.1	3
14	Screening Human Genes for Small Alterations Performing an Enzymatic Cleavage Mismatched Analysis (ECMA) Protocol. Molecular Biotechnology, 2013, 55, 1-9.	2.4	8