

# Kyriaki Kekou

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/10409386/publications.pdf>

Version: 2024-02-01

14  
papers

692  
citations

1684188  
5  
h-index

1281871  
11  
g-index

14  
all docs

14  
docs citations

14  
times ranked

1151  
citing authors

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