

# Christalena Sofocleous

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/10121148/publications.pdf>

Version: 2024-02-01

8  
papers

68  
citations

1684188  
5  
h-index

1588992  
8  
g-index

8  
all docs

8  
docs citations

8  
times ranked

153  
citing authors

#	ARTICLE	IF	CITATIONS
1	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
2	The Greek Registry of Shwachman Diamond Syndrome: Molecular and clinical data. <i>Pediatric Blood and Cancer</i> , 2017, 64, e26630.	1.5	12
3	Compound heterozygosity of a paternal submicroscopic deletion and a maternal missense mutation in <i>POR</i> gene: Antley-Bixler syndrome phenotype in three sibling fetuses. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2016, 106, 536-541.	1.6	11
4	A novel large deletion of the ICR1 region including H19 and putative enhancer elements. <i>BMC Medical Genetics</i> , 2015, 16, 30.	2.1	9
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	CantÃ Syndrome Associated with Ovarian Agenesis. <i>Molecular Syndromology</i> , 2017, 8, 206-210.	0.8	2
7	Does splenectomy influence the development of Hypothyroidism in Transfusion Dependent Thalassemia Patients? A retrospective study.. <i>Mediterranean Journal of Hematology and Infectious Diseases</i> , 2019, 11, e2019064.	1.3	1
8	Heterozygosity of the Complex Corfu $\beta^+$ Thalassemic Allele (HBD Deletion and HBB:c.92+5G>A) Revisited. <i>Biology</i> , 2022, 11, 432.	2.8	1