

# Gerald B Schaefer

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/2986838/publications.pdf>

Version: 2024-02-01

12  
papers

173  
citations

1478505

6  
h-index

1588992

8  
g-index

12  
all docs

12  
docs citations

12  
times ranked

518  
citing authors

#	ARTICLE	IF	CITATIONS
1	Multimodal imaging of an RPGR carrier female. <i>Ophthalmic Genetics</i> , 2021, 42, 312-316.	1.2	4
2	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor $\beta^2$ Signaling. <i>Biological Psychiatry</i> , 2020, 87, 100-112.	1.3	42
3	Constitutive activation of the PI3K $\rightarrow$ AKT pathway and cardiovascular abnormalities in an individual with Kosaki overgrowth syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1047-1052.	1.2	18
4	A parent $\rightarrow$ of $\rightarrow$ origin analysis of paternal genetic variants and increased risk of conotruncal heart defects. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 609-617.	1.2	7
5	McArdle Disease Presenting With Muscle Pain in a Teenage Girl: The Role of Whole-Exome Sequencing in Neurogenetic Disorders. <i>Seminars in Pediatric Neurology</i> , 2018, 26, 50-51.	2.0	0
6	Additional <i>de novo</i> missense genetic variants in <i>NALCN</i> associated with <i>CLIFAHDD</i> syndrome. <i>Clinical Genetics</i> , 2017, 91, 929-931.	2.0	10
7	Alu-mediated deletion of PIGL in a Patient with CHIME syndrome. , 2017, 173, 1378-1382.		14
8	The role of IQSEC2 in syndromic intellectual disability: Narrowing the diagnostic odyssey. , 2017, 173, 2814-2820.		14
9	A novel approach in pediatric telegenetic services: geneticist, pediatrician and genetic counselor team. <i>Genetics in Medicine</i> , 2017, 19, 1260-1267.	2.4	43
10	What is the role of clinical genetics in the patient-centered medical home?: A commentary from the Medical Home Workgroup of the Heartland Regional Genetics and Newborn Screening Collaborative. <i>Genetics in Medicine</i> , 2016, 18, 440-442.	2.4	4
11	A Newborn With Complex Skeletal Abnormalities, Joint Contractures, and Bilateral Corneal Clouding With Sclerocornea. <i>Seminars in Pediatric Neurology</i> , 2014, 21, 84-87.	2.0	17
12	Editorial Comment: An Unusual Cause of Peroneal Neuropathy. <i>Seminars in Pediatric Neurology</i> , 2014, 21, 82-83.	2.0	0