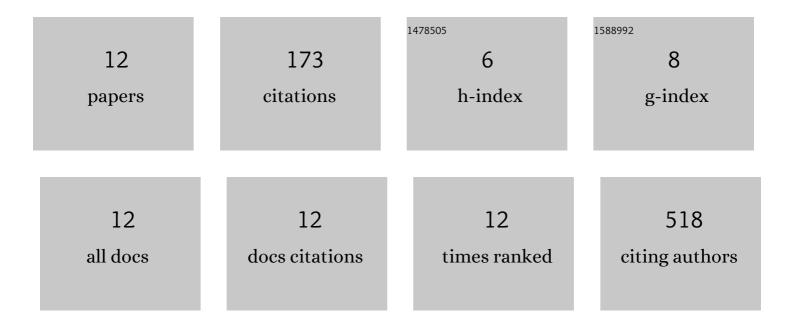
Gerald B Schaefer

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/2986838/publications.pdf Version: 2024-02-01



#	Article	IF	CITATIONS
1	Multimodal imaging of an RPGR carrier female. Ophthalmic Genetics, 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 β Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
3	Constitutive activation of the PI3Kâ€AKT pathway and cardiovascular abnormalities in an individual with Kosaki overgrowth syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 1047-1052.	1.2	18
4	A parentâ€ofâ€origin analysis of paternal genetic variants and increased risk of conotruncal heart defects. American Journal of Medical Genetics, Part A, 2018, 176, 609-617.	1.2	7
5	McArdle Disease Presenting With Muscle Pain in a Teenage Cirl: The Role of Whole-Exome Sequencing in Neurogenetic Disorders. Seminars in Pediatric Neurology, 2018, 26, 50-51.	2.0	0
6	Additional <i>de novo</i> missense genetic variants in <i><scp>NALCN</scp></i> associated with <scp>CLIFAHDD</scp> syndrome. Clinical Genetics, 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. Genetics in Medicine, 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. Genetics in Medicine, 2016, 18, 440-442.	2.4	4
11	A Newborn With Complex Skeletal Abnormalities, Joint Contractures, and Bilateral Corneal Clouding With Sclerocornea. Seminars in Pediatric Neurology, 2014, 21, 84-87.	2.0	17
12	Editorial Comment: An Unusual Cause of Peroneal Neuropathy. Seminars in Pediatric Neurology, 2014, 21, 82-83.	2.0	0