Gerald B Schaefer

List of Publications by Year
in descending order

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

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12	173	1478505 6	1588992 8
papers	citations	h-index	g-index

12	12	12	518
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A novel approach in pediatric telegenetic services: geneticist, pediatrician and genetic counselor team. Genetics in Medicine, 2017, 19, 1260-1267.	2.4	43
2	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² 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 Newborn With Complex Skeletal Abnormalities, Joint Contractures, and Bilateral Corneal Clouding With Sclerocornea. Seminars in Pediatric Neurology, 2014, 21, 84-87.	2.0	17
5	Alu -mediated deletion of PIGL in a Patient with CHIME syndrome. , 2017, 173, 1378-1382.		14
6	The role of IQSEC2 in syndromic intellectual disability: Narrowing the diagnostic odyssey. , 2017, 173, 2814-2820.		14
7	Additional de novo missense genetic variants in <scp>NALCN</scp> associated with <scp>CLIFAHDD</scp> syndrome. Clinical Genetics, 2017, 91, 929-931.	2.0	10
8	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
9	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
10	Multimodal imaging of an RPGR carrier female. Ophthalmic Genetics, 2021, 42, 312-316.	1.2	4
11	Editorial Comment: An Unusual Cause of Peroneal Neuropathy. Seminars in Pediatric Neurology, 2014, 21, 82-83.	2.0	0
12	McArdle Disease Presenting With Muscle Pain in a Teenage Girl: The Role of Whole-Exome Sequencing in Neurogenetic Disorders. Seminars in Pediatric Neurology, 2018, 26, 50-51.	2.0	0