Anya Revah-Politi

List of Publications by Year
in descending order

16	373	1040056 9	940533 16
papers	citations	h-index	g-index

17	17	17	884
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The clinical and molecular spectrum of QRICH1 associated neurodevelopmental disorder. Human Mutation, 2022, 43, 266-282.	2.5	7
2	Diagnostic sequencing to support genetically stratified medicine in a tertiary care setting. Genetics in Medicine, 2022, 24, 862-869.	2.4	4
3	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617.	6.2	16
4	Biâ€allelic KARS1 pathogenic variants affecting functions of cytosolic and mitochondrial isoforms are associated with a progressive and multisystem disease. Human Mutation, 2021, 42, 745-761.	2.5	7
5	Genetic testing in individuals with cerebral palsy. Developmental Medicine and Child Neurology, 2021, 63, 1448-1455.	2.1	19
6	Truncating variants in the SHANK1 gene are associated with a spectrum of neurodevelopmental disorders. Genetics in Medicine, 2021, 23, 1912-1921.	2.4	5
7	ZTTK syndrome: Clinical and molecular findings ofÂ15 cases and a review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 3740-3753.	1.2	11
8	Causal Genetic Variants in Stillbirth. New England Journal of Medicine, 2020, 383, 1107-1116.	27.0	67
9	Dominant mutations in ITPR3 cause Charcotâ€Marieâ€Tooth disease. Annals of Clinical and Translational Neurology, 2020, 7, 1962-1972.	3.7	9
10	Evaluation of the cost and effectiveness of diverse recruitment methods for a genetic screening study. Genetics in Medicine, 2019, 21, 2371-2380.	2.4	10
11	Heterozygous loss-of-function variants of MEIS2 cause a triad of palatal defects, congenital heart defects, and intellectual disability. European Journal of Human Genetics, 2019, 27, 278-290.	2.8	30
12	De novo mutations in the GTP/GDP-binding region of RALA, a RAS-like small GTPase, cause intellectual disability and developmental delay. PLoS Genetics, 2018, 14, e1007671.	3.5	16
13	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. Annals of Neurology, 2018, 84, 788-795.	5.3	44
14	Refining the phenotype associated with GNB1 mutations: Clinical data on 18 newly identified patients and review of the literature. American Journal of Medical Genetics, Part A, 2018, 176, 2259-2275.	1.2	47
15	Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. American Journal of Human Genetics, 2018, 103, 221-231.	6.2	65
16	Lossâ€ofâ€function variants in NFIA provide further support that NFIA is a critical gene in 1p32â€p31 deletion syndrome: A four patient series. American Journal of Medical Genetics, Part A, 2017, 173, 3158-3164.	1.2	16