Emily Fassi

List of Publications by Year
in descending order

11	831	933447 10	1281871 11
papers	citations	h-index	g-index

12	12	12	2354
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
2	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
3	The Exome Clinic and the role of medical genetics expertise in the interpretation of exome sequencing results. Genetics in Medicine, 2017, 19, 1040-1048.	2.4	85
4	De novo mutation screening in childhood-onset cerebellar atrophy identifies gain-of-function mutations in the CACNA1G calcium channel gene. Brain, 2018, 141, 1998-2013.	7.6	67
5	Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. American Journal of Human Genetics, 2019, 104, 815-834.	6.2	59
6	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. Genetics in Medicine, 2019, 21, 2723-2733.	2.4	48
7	Widening of the genetic and clinical spectrum of Lambâ€“Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. Genetics in Medicine, 2020, 22, 524-537.	2.4	21
8	Mutations in the <scp>PH</scp> Domain of <scp>DNM</scp>1 are associated with a nonepileptic phenotype characterized by developmental delay and neurobehavioral abnormalities. Molecular Genetics & Genomic Medicine, 2018, 6, 294-300.	1.2	19
9	Missense variants in TAF1 and developmental phenotypes: Challenges of determining pathogenicity. Human Mutation, 2020, 41, 449-464.	2.5	17
10	Breast cancer polygenic risk scores in the clinical cancer genetic counseling setting: Current practices and impact on patient management. Journal of Genetic Counseling, 2021, 30, 588-597.	1.6	14
11	Genetic counseling in CHARGE syndrome: Diagnostic evaluation through follow up. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 407-416.	1.6	13