Alicia B Byrne

List of Publications by Year
in descending order

9 papers	159 citations	1307594 7 h-index	1474206 9 g-index
12 all docs	12 docs citations	12 times ranked	406 citing authors

#	Article	IF	Citations
1	Recurrent de novo missense variants in GNB2 can cause syndromic intellectual disability. Journal of Medical Genetics, 2022, 59, 511-516.	3.2	4
2	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	2.4	44
3	Pathogenic variants in MDFIC cause recessive central conducting lymphatic anomaly with lymphedema. Science Translational Medicine, 2022, 14, eabm4869.	12.4	14
4	Compound heterozygous variants in LAMC3 in association with posterior periventricular nodular heterotopia. BMC Medical Genomics, 2021, 14, 64.	1.5	5
5	Pseudodiastrophic dysplasia expands the known phenotypic spectrum of defects in proteoglycan biosynthesis. Journal of Medical Genetics, 2020, 57, 454-460.	3.2	8
6	Paternal mosaicism for a novel <scp>PBX1</scp> mutation associated with recurrent perinatal death: Phenotypic expansion of the <scp>PBX1</scp>â€related syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1273-1277.	1.2	12
7	Posterior Neocortex-Specific Regulation of Neuronal Migration by CEP85L Identifies Maternal Centriole-Dependent Activation of CDK5. Neuron, 2020, 106, 246-255.e6.	8.1	19
8	Identification and targeted management of a neurodegenerative disorder caused by biallelic mutations in SLC5A6. Npj Genomic Medicine, 2019, 4, 28.	3.8	16
9	matchbox: An open-source tool for patient matching via the Matchmaker Exchange. Human Mutation, 2018, 39, 1827-1834.	2.5	20