

Alicia B Byrne

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/6291290/publications.pdf>

Version: 2024-02-01

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