Michael Yourshaw

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/1250515/publications.pdf

Version: 2024-02-01

13 papers	1,604 citations	759233 12 h-index	1125743 13 g-index
13 all docs	13 docs citations	13 times ranked	4282 citing authors

#	Article	IF	Citations
1	Clinical Exome Sequencing for Genetic Identification of Rare Mendelian Disorders. JAMA - Journal of the American Medical Association, 2014, 312, 1880.	7.4	842
2	Mutations in the RNA exosome component gene EXOSC3 cause pontocerebellar hypoplasia and spinal motor neuron degeneration. Nature Genetics, 2012, 44, 704-708.	21.4	216
3	Congenital Proprotein Convertase 1/3 Deficiency Causes Malabsorptive Diarrhea and Other Endocrinopathies in a Pediatric Cohort. Gastroenterology, 2013, 145, 138-148.	1.3	131
4	Pontocerebellar hypoplasia type 1. Neurology, 2013, 80, 438-446.	1.1	84
5	Loss of function of SLC25A46 causes lethal congenital pontocerebellar hypoplasia. Brain, 2016, 139, 2877-2890.	7.6	74
6	Rare Genomic Variants Link Bipolar Disorder with Anxiety Disorders to CREB-Regulated Intracellular Signaling Pathways. Frontiers in Psychiatry, 2013, 4, 154.	2.6	54
7	EPCAMmutation update: Variants associated with congenital tufting enteropathy and Lynch syndrome. Human Mutation, 2019, 40, 142-161.	2.5	51
8	Rich annotation of DNA sequencing variants by leveraging the Ensembl Variant Effect Predictor with plugins. Briefings in Bioinformatics, 2015, 16, 255-264.	6.5	41
9	Loss of ADAM17 is associated with severe multiorgan dysfunction. Human Pathology, 2015, 46, 923-928.	2.0	31
10	Exome Sequencing Finds a Novel PCSK1 Mutation in a Child With Generalized Malabsorptive Diarrhea and Diabetes Insipidus. Journal of Pediatric Gastroenterology and Nutrition, 2013, 57, 759-767.	1.8	29
11	Functional Consequences of a Novel Variant of PCSK1. PLoS ONE, 2013, 8, e55065.	2.5	24
12	A Novel Familial Mutation in the PCSK1 Gene That Alters the Oxyanion Hole Residue of Proprotein Convertase 1/3 and Impairs Its Enzymatic Activity. PLoS ONE, 2014, 9, e108878.	2.5	19
13	System for Informatics in the Molecular Pathology Laboratory. Journal of Molecular Diagnostics, 2018, 20, 522-532.	2.8	8