Michael Yourshaw

List of Publications by Year
in descending order

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	EPCAMmutation update: Variants associated with congenital tufting enteropathy and Lynch syndrome. Human Mutation, 2019, 40, 142-161.	2.5	51
2	System for Informatics in the Molecular Pathology Laboratory. Journal of Molecular Diagnostics, 2018, 20, 522-532.	2.8	8
3	Loss of function of SLC25A46 causes lethal congenital pontocerebellar hypoplasia. Brain, 2016, 139, 2877-2890.	7.6	74
4	Loss of ADAM17 is associated with severe multiorgan dysfunction. Human Pathology, 2015, 46, 923-928.	2.0	31
5	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
6	Clinical Exome Sequencing for Genetic Identification of Rare Mendelian Disorders. JAMA - Journal of the American Medical Association, 2014, 312, 1880.	7.4	842
7	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
8	Congenital Proprotein Convertase 1/3 Deficiency Causes Malabsorptive Diarrhea and Other Endocrinopathies in a Pediatric Cohort. Gastroenterology, 2013, 145, 138-148.	1.3	131
9	Pontocerebellar hypoplasia type 1. Neurology, 2013, 80, 438-446.	1.1	84
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	Rare Genomic Variants Link Bipolar Disorder with Anxiety Disorders to CREB-Regulated Intracellular Signaling Pathways. Frontiers in Psychiatry, 2013, 4, 154.	2.6	54
12	Functional Consequences of a Novel Variant of PCSK1. PLoS ONE, 2013, 8, e55065.	2.5	24
13	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