A Reghan Foley

List of Publications by Year
in descending order

18	1,312	759233 12	839539 18
papers	citations	h-index	g-index

18	18	18	2881
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. Science Translational Medicine, 2017, 9, .	12.4	516
2	The Role of PIEZO2 in Human Mechanosensation. New England Journal of Medicine, 2016, 375, 1355-1364.	27.0	293
3	Genetic regulatory variation in populations informs transcriptome analysis in rare disease. Science, 2019, 366, 351-356.	12.6	99
4	Natural history of pulmonary function in collagen VI-related myopathies. Brain, 2013, 136, 3625-3633.	7.6	85
5	Position of Glycine Substitutions in the Triple Helix ofCOL6A1,COL6A2, andCOL6A3is Correlated with Severity and Mode of Inheritance in Collagen VI Myopathies. Human Mutation, 2013, 34, 1558-1567.	2.5	79
6	Extracellular matrix-driven congenital muscular dystrophies. Matrix Biology, 2018, 71-72, 188-204.	3.6	44
7	A recurrent COL6A1 pseudoexon insertion causes muscular dystrophy and is effectively targeted by splice-correction therapies. JCI Insight, 2019, 4, .	5.0	33
8	Results of a two-year pilot study of clinical outcome measures in collagen VI- and laminin alpha2-related congenital muscular dystrophies. Neuromuscular Disorders, 2015, 25, 43-54.	0.6	30
9	Dominant collagen XII mutations cause a distal myopathy. Annals of Clinical and Translational Neurology, 2019, 6, 1980-1988.	3.7	24
10	Longitudinal changes in clinical outcome measures in COL6-related dystrophies and LAMA2-related dystrophies. Neurology, 2019, 93, e1932-e1943.	1.1	23
11	<scp>GGPS1</scp> Mutations Cause Muscular Dystrophy/Hearing Loss/Ovarian Insufficiency Syndrome. Annals of Neurology, 2020, 88, 332-347.	5.3	22
12	Adult MTM1-related myopathy carriers. Neurology, 2019, 93, e1535-e1542.	1.1	18
13	Pathogenic variants in TNNC2 cause congenital myopathy due to an impaired force response to calcium. Journal of Clinical Investigation, 2021, 131, .	8.2	11
14	Association of Initial Maximal Motor Ability With Long-term Functional Outcome in Patients With COL6-Related Dystrophies. Neurology, 2021, 96, e1413-e1424.	1.1	10
15	Transcriptome analysis of collagen VIâ€related muscular dystrophy muscle biopsies. Annals of Clinical and Translational Neurology, 2021, 8, 2184-2198.	3.7	10
16	Spectrum of Clinical Features in X-Linked Myotubular Myopathy Carriers. Neurology, 2021, 97, e501-e512.	1.1	9
17	MuscleViz: Free Open-Source Software for Muscle Weakness Visualization. Journal of Neuromuscular Diseases, 2019, 6, 263-266.	2.6	3
18	Cross-sectional Neuromuscular Phenotyping Study of Patients With Arhinia With SMCHD1 Variants. Neurology, 2022, 98, .	1.1	3