Vandana A Gupta

List of Publications by Year
in descending order

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32	1,426	361045	414034
papers	citations	20 h-index	32 g-index

33	33	33	2568
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Exome Sequencing and Functional Validation in Zebrafish Identify GTDC2 Mutations as a Cause of Walker-Warburg Syndrome. American Journal of Human Genetics, 2012, 91, 541-547.	2.6	167
2	Leiomodin-3 dysfunction results in thin filament disorganization and nemaline myopathy. Journal of Clinical Investigation, 2014, 124, 4693-4708.	3.9	153
3	Identification of KLHL41 Mutations Implicates BTB-Kelch-Mediated Ubiquitination as an Alternate Pathway to Myofibrillar Disruption in Nemaline Myopathy. American Journal of Human Genetics, 2013, 93, 1108-1117.	2.6	147
4	Kelch proteins: emerging roles in skeletal muscle development and diseases. Skeletal Muscle, 2014, 4, 11.	1.9	119
5	The zebrafish dag1 mutant: a novel genetic model for dystroglycanopathies. Human Molecular Genetics, 2011, 20, 1712-1725.	1.4	101
6	POMK mutations disrupt muscle development leading to a spectrum of neuromuscular presentations. Human Molecular Genetics, 2014, 23, 5781-5792.	1.4	72
7	Role of water structure on phase separation in polyelectrolyteâ€“polyethyleneglycol based aqueous two-phase systems. Polymer, 2002, 43, 3387-3390.	1.8	58
8	Mutationâ€specific effects on thin filament length in thin filament myopathy. Annals of Neurology, 2016, 79, 959-969.	2.8	54
9	A Splice Site Mutation in Laminin-Î±2 Results in a Severe Muscular Dystrophy and Growth Abnormalities in Zebrafish. PLoS ONE, 2012, 7, e43794.	1.1	48
10	GOLGA2, encoding a master regulator of golgi apparatus, is mutated in a patient with a neuromuscular disorder. Human Genetics, 2016, 135, 245-251.	1.8	38
11	Myotubularin-Deficient Myoblasts Display Increased Apoptosis, Delayed Proliferation, and Poor Cell Engraftment. American Journal of Pathology, 2012, 181, 961-968.	1.9	37
12	Î±â€Actininâ€2 deficiency results in sarcomeric defects in zebrafish that cannot be rescued by Î±â€actininâ€3 revealing functional differences between sarcomeric isoforms. FASEB Journal, 2012, 26, 1892-1908.	0.2	34
13	Mutation of the mitochondrial carrier SLC25A42 causes a novel form of mitochondrial myopathy in humans. Human Genetics, 2016, 135, 21-30.	1.8	34
14	RNA helicase, DDX27 regulates skeletal muscle growth and regeneration by modulation of translational processes. PLoS Genetics, 2018, 14, e1007226.	1.5	34
15	Prednisolone rescues Duchenne muscular dystrophy phenotypes in human pluripotent stem cellâ€“derived skeletal muscle in vitro. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	32
16	ACTN2 mutations cause â€œMultiple structured Core Diseaseâ€•(MsCD). Acta Neuropathologica, 2019, 137, 501-519.	3.9	31
17	Bridging integrator 1 (Bin1) deficiency in zebrafish results in centronuclear myopathy. Human Molecular Genetics, 2014, 23, 3566-3578.	1.4	28
18	Modification of Msx1 by SUMO-1. Biochemical and Biophysical Research Communications, 2006, 345, 74-77.	1.0	25

#	ARTICLE	IF	CITATIONS
19	ZBTB42 mutation defines a novel lethal congenital contracture syndrome (LCCS6). Human Molecular Genetics, 2014, 23, 6584-6593.	1.4	25
20	An integrated clinical program and crowdsourcing strategy for genomic sequencing and Mendelian disease gene discovery. Npj Genomic Medicine, 2018, 3, 21.	1.7	24
21	Loss of Catalytically Inactive Lipid Phosphatase Myotubularin-related Protein 12 Impairs Myotubularin Stability and Promotes Centronuclear Myopathy in Zebrafish. PLoS Genetics, 2013, 9, e1003583.	1.5	22
22	Dysregulation of NRAP degradation by KLHL41 contributes to pathophysiology in nemaline myopathy. Human Molecular Genetics, 2019, 28, 2549-2560.	1.4	22
23	A Network of Transcription Factors Operates during Early Tooth Morphogenesis. Molecular and Cellular Biology, 2013, 33, 3099-3112.	1.1	19
24	Novel Recessive TNNT1 Congenital Coreâ€Rod Myopathy in French Canadians. Annals of Neurology, 2020, 87, 568-583.	2.8	19
25	Homozygous TRPV4 mutation causes congenital distal spinal muscular atrophy and arthrogryposis. Neurology: Genetics, 2019, 5, e312.	0.9	15
26	Transgenic zebrafish model of DUX4 misexpression reveals a developmental role in FSHD pathogenesis. Human Molecular Genetics, 2019, 28, 320-331.	1.4	14
27	An open source microcontroller based flume for evaluating swimming performance of larval, juvenile, and adult zebrafish. PLoS ONE, 2018, 13, e0199712.	1.1	13
28	Mullerian-Inhibiting Substance Induces Gro-Î² Expression in Breast Cancer Cells through a Nuclear Factor-ÎºBâ€“Dependent and Smad1-Dependent Mechanism. Cancer Research, 2007, 67, 2747-2756.	0.4	12
29	A novel early onset phenotype in a zebrafish model of merosin deficient congenital muscular dystrophy. PLoS ONE, 2017, 12, e0172648.	1.1	9
30	Biâ€allelic loss of function variants in <scp>GOLGA2</scp> are associated with a complex neurological phenotype: Report of a second family. Clinical Genetics, 2021, 100, 748-751.	1.0	8
31	<scp>hnRNP L</scp> is essential for myogenic differentiation and modulates myotonic dystrophy pathologies. Muscle and Nerve, 2021, 63, 928-940.	1.0	7
32	246th ENMC International Workshop: Protein aggregate myopathies 24â€“26 May 2019, Hoofddorp, The Netherlands. Neuromuscular Disorders, 2021, 31, 158-166.	0.3	5