

Svetlana Gorokhova

List of Publications by Year in descending order

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Version: 2024-02-01

27
papers

970
citations

933447

10
h-index

580821

25
g-index

31
all docs

31
docs citations

31
times ranked

1640
citing authors

#	ARTICLE	IF	CITATIONS
1	Missense mutations in desmin associated with familial cardiac and skeletal myopathy. <i>Nature Genetics</i> , 1998, 19, 402-403.	21.4	484
2	Loss of function of axonemal dynein <i>Mdnah5</i> causes primary ciliary dyskinesia and hydrocephalus. <i>Human Molecular Genetics</i> , 2002, 11, 715-721.	2.9	209
3	A novel family of transmembrane proteins interacting with β subunits of the Na,K-ATPase. <i>Human Molecular Genetics</i> , 2007, 16, 2394-2410.	2.9	45
4	Genetic variants in components of the NALCN-UNC80-UNC79 ion channel complex cause a broad clinical phenotype (NALCN channelopathies). <i>Human Genetics</i> , 2018, 137, 753-768.	3.8	38
5	Improving molecular diagnosis of distal myopathies by targeted next-generation sequencing: Table 1. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, 340-342.	1.9	20
6	Novel <i>CAPN3</i> variant associated with an autosomal dominant calpainopathy. <i>Neuropathology and Applied Neurobiology</i> , 2020, 46, 564-578.	3.2	20
7	Comparing targeted exome and whole exome approaches for genetic diagnosis of neuromuscular disorders. <i>Applied & Translational Genomics</i> , 2015, 7, 26-31.	2.1	18
8	Direct evidence for the interaction of stathmin along the length and the plus end of microtubules in cells. <i>FASEB Journal</i> , 2016, 30, 3202-3215.	0.5	17
9	Uncoupling of Molecular Maturation from Peripheral Target Innervation in Nociceptors Expressing a Chimeric TrkA/TrkC Receptor. <i>PLoS Genetics</i> , 2014, 10, e1004081.	3.5	11
10	Novel Pathogenic Variants in a French Cohort Widen the Mutational Spectrum of GNE Myopathy. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, 131-136.	2.6	11
11	Clinical massively parallel sequencing for the diagnosis of myopathies. <i>Revue Neurologique</i> , 2015, 171, 558-571.	1.5	11
12	Retrospective analysis and reclassification of DYSF variants in a large French series of dysferlinopathy patients. <i>Genetics in Medicine</i> , 2021, 23, 1574-1577.	2.4	11
13	Splicing impact of deep exonic missense variants in <i>CAPN3</i> explored systematically by minigene functional assay. <i>Human Mutation</i> , 2020, 41, 1797-1810.	2.5	9
14	An AI-Powered Blood Test to Detect Cancer Using NanoDSF. <i>Cancers</i> , 2021, 13, 1294.	3.7	9
15	Deep phenotyping of an international series of patients with late-onset dysferlinopathy. <i>European Journal of Neurology</i> , 2021, 28, 2092-2102.	3.3	9
16	<i>BET1</i> variants establish impaired vesicular transport as a cause for muscular dystrophy with epilepsy. <i>EMBO Molecular Medicine</i> , 2021, 13, e13787.	6.9	9
17	Growth charts in Kabuki syndrome 1. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 446-453.	1.2	7
18	Genetic Characterization of a French Cohort of GNE mutation negative inclusion body myopathy patients with exome sequencing. <i>Muscle and Nerve</i> , 2017, 56, 993-997.	2.2	6

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19	Discovering a new part of the phenotypic spectrum of Coffin-Siris syndrome in a fetal cohort. <i>Genetics in Medicine</i> , 2022, 24, 1753-1760.	2.4	6
20	First characterization of congenital myasthenic syndrome type 5 in North Africa. <i>Molecular Biology Reports</i> , 2021, 48, 6999-7006.	2.3	4
21	Spindle-Derived NT3 in Sensorimotor Connections: Principal Role at Later Stages. <i>Journal of Neuroscience</i> , 2009, 29, 10181-10183.	3.6	3
22	Significant contribution of intragenic deletions to ARID1B mutation spectrum. <i>Genetics in Medicine</i> , 2019, 21, 2654-2655.	2.4	3
23	Genetic Profile of Patients with Limb-Girdle Muscle Weakness in the Chilean Population. <i>Genes</i> , 2022, 13, 1076.	2.4	3
24	Identification of novel mutations by targeted NGS in Moroccan families clinically diagnosed with a neuromuscular disorder. <i>Clinica Chimica Acta</i> , 2022, 524, 51-58.	1.1	2
25	A new tool CovReport generates easy-to-understand sequencing coverage summary for diagnostic reports. <i>Scientific Reports</i> , 2020, 10, 6247.	3.3	1
26	Refining NGS diagnosis of muscular disorders. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2021, 92, 223-225.	1.9	1
27	Correspondence on "De novo variants in MED12 cause X-linked syndromic neurodevelopmental disorders in 18 females" by Polla et al.. <i>Genetics in Medicine</i> , 2021, 23, 2003-2004.	2.4	1