Svetlana Gorokhova

List of Publications by Year
in descending order

27	970	933447 10	580821 25
papers	citations	h-index	g-index

31	31	31	1640
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Identification of novel mutations by targeted NGS in Moroccan families clinically diagnosed with a neuromuscular disorder. Clinica Chimica Acta, 2022, 524, 51-58.	1.1	2
2	Discovering a new part of the phenotypic spectrum of Coffin-Siris syndrome in a fetal cohort. Genetics in Medicine, 2022, 24, 1753-1760.	2.4	6
3	Genetic Profile of Patients with Limb-Girdle Muscle Weakness in the Chilean Population. Genes, 2022, 13, 1076.	2.4	3
4	Refining NGS diagnosis of muscular disorders. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 223-225.	1.9	1
5	An AI-Powered Blood Test to Detect Cancer Using NanoDSF. Cancers, 2021, 13, 1294.	3.7	9
6	Deep phenotyping of an international series of patients with lateâ€onset dysferlinopathy. European Journal of Neurology, 2021, 28, 2092-2102.	3.3	9
7	Retrospective analysis and reclassification of DYSF variants in a large French series of dysferlinopathy patients. Genetics in Medicine, 2021, 23, 1574-1577.	2.4	11
8	Correspondence on â€œDe novo variants in MED12 cause X-linked syndromic neurodevelopmental disorders in 18 femalesâ€•by Polla et al.. Genetics in Medicine, 2021, 23, 2003-2004.	2.4	1
9	First characterization of congenital myasthenic syndrome type 5 in North Africa. Molecular Biology Reports, 2021, 48, 6999-7006.	2.3	4
10	BET1 variants establish impaired vesicular transport as a cause for muscular dystrophy with epilepsy. EMBO Molecular Medicine, 2021, 13, e13787.	6.9	9
11	Growth charts in Kabuki syndrome 1. American Journal of Medical Genetics, Part A, 2020, 182, 446-453.	1.2	7
12	Splicing impact of deep exonic missense variants in CAPN3 explored systematically by minigene functional assay. Human Mutation, 2020, 41, 1797-1810.	2.5	9
13	Novel CAPN3 variant associated with an autosomal dominant calpainopathy. Neuropathology and Applied Neurobiology, 2020, 46, 564-578.	3.2	20
14	A new tool CovReport generates easy-to-understand sequencing coverage summary for diagnostic reports. Scientific Reports, 2020, 10, 6247.	3.3	1
15	Significant contribution of intragenic deletions to ARID1B mutation spectrum. Genetics in Medicine, 2019, 21, 2654-2655.	2.4	3
16	Genetic variants in components of the NALCNâ€“UNC80â€“UNC79 ion channel complex cause a broad clinical phenotype (NALCN channelopathies). Human Genetics, 2018, 137, 753-768.	3.8	38
17	Genetic Characterization of a French Cohort of GNEâ€mutation negative inclusion body myopathy patients with exome sequencing. Muscle and Nerve, 2017, 56, 993-997.	2.2	6
18	Direct evidence for the interaction of stathmin along the length and the plus end of microtubules in cells. FASEB Journal, 2016, 30, 3202-3215.	0.5	17

#	Article	IF	Citations
19	Improving molecular diagnosis of distal myopathies by targeted next-generation sequencing: TableÂ1. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 340-342.	1.9	20
20	Novel Pathogenic Variants in a French Cohort Widen the Mutational Spectrum ofÂGNE Myopathy. Journal of Neuromuscular Diseases, 2015, 2, 131-136.	2.6	11
21	Comparing targeted exome and whole exome approaches for genetic diagnosis of neuromuscular disorders. Applied & Translational Genomics, 2015, 7, 26-31.	2.1	18
22	Clinical massively parallel sequencing for the diagnosis of myopathies. Revue Neurologique, 2015, 171, 558-571.	1.5	11
23	Uncoupling of Molecular Maturation from Peripheral Target Innervation in Nociceptors Expressing a Chimeric TrkA/TrkC Receptor. PLoS Genetics, 2014, 10, e1004081.	3.5	11
24	Spindle-Derived NT3 in Sensorimotor Connections: Principal Role at Later Stages. Journal of Neuroscience, 2009, 29, 10181-10183.	3.6	3
25	A novel family of transmembrane proteins interacting with Î² subunits of the Na,K-ATPase. Human Molecular Genetics, 2007, 16, 2394-2410.	2.9	45
26	Loss of function of axonemal dynein Mdnah5 causes primary ciliary dyskinesia and hydrocephalus. Human Molecular Genetics, 2002, 11, 715-721.	2.9	209
27	Missense mutations in desmin associated with familial cardiac and skeletal myopathy. Nature Genetics, 1998, 19, 402-403.	21.4	484