Virginia Barone

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/1818395/publications.pdf

Version: 2024-02-01

40 papers	2,913 citations	236925 25 h-index	361022 35 g-index
40 all docs	40 docs citations	40 times ranked	2795 citing authors

#	Article	IF	Citations
1	Point mutations affecting the tyrosine kinase domain of the RET proto-oncogene in Hirschsprung's disease. Nature, 1994, 367, 377-378.	27.8	722
2	Loss of function effect of RET mutations causing Hirschsprung disease. Nature Genetics, 1995, 10, 35-40.	21.4	225
3	Increased Susceptibility to Cortical Spreading Depression in the Mouse Model of Familial Hemiplegic Migraine Type 2. PLoS Genetics, 2011, 7, e1002129.	3.5	179
4	Binding of an ankyrin-1 isoform to obscurin suggests a molecular link between the sarcoplasmic reticulum and myofibrils in striated muscles. Journal of Cell Biology, 2003, 160, 245-253.	5.2	177
5	Deletion of the ryanodine receptor type 3 (RyR3) impairs forms of synaptic plasticity and spatial learning. EMBO Journal, 1999, 18, 5264-5273.	7.8	161
6	Frequency of RET mutations in long- and short-segment Hirschsprung disease. Human Mutation, 1997, 9, 243-249.	2.5	138
7	Requirement for the ryanodine receptor type 3for efficient contraction in neonatal skeletal muscles. EMBO Journal, 1997, 16, 6956-6963.	7.8	126
8	A novel mutation in the ATP1A2 gene causes alternating hemiplegia of childhood. Journal of Medical Genetics, 2004, 41, 621-628.	3.2	100
9	Close linkage with the RET protooncogene and boundaries of deletion mutations in autosomal dominant Hirschsprung disease. Human Molecular Genetics, 1993, 2, 1803-1808.	2.9	88
10	Contribution of Ryanodine Receptor Type 3 to Ca2+ Sparks in Embryonic Mouse Skeletal Muscle. Biophysical Journal, 1999, 77, 1394-1403.	0.5	72
11	Molecular cloning and functional characterization of a GABA/betaine transporter from human kidney. FEBS Letters, 1995, 373, 229-233.	2.8	66
12	The Sarcoplasmic Reticulum: An Organized Patchwork of Specialized Domains. Traffic, 2008, 9, 1044-1049.	2.7	66
13	Intracellular Ca2+ release channels in evolution. Current Opinion in Genetics and Development, 2000, 10, 662-667.	3.3	62
14	Heterogeneity and Low Detection Rate of RET Mutations in Hirschsprung Disease. European Journal of Human Genetics, 1994, 2, 272-280.	2.8	60
15	Two de novo mutations in the Na,K-ATPase gene ATP1A2 associated with pure familial hemiplegic migraine. European Journal of Human Genetics, 2006, 14, 555-560.	2.8	56
16	The gas 5 gene shows four alternative splicing patterns without coding for a protein. Gene, 2000, 256, 13-17.	2.2	55
17	A Mutation in theCASQ1Gene Causes a Vacuolar Myopathy with Accumulation of Sarcoplasmic Reticulum Protein Aggregates. Human Mutation, 2014, 35, 1163-1170.	2.5	53
18	Homer proteins and InsP3 receptors co-localise in the longitudinal sarcoplasmic reticulum of skeletal muscle fibres. Cell Calcium, 2002, 32, 193-200.	2.4	52

#	Article	IF	Citations
19	Molecular interactions with obscurin are involved in the localization of muscle-specific small ankyrin1 isoforms to subcompartments of the sarcoplasmic reticulum. Experimental Cell Research, 2006, 312, 3546-3558.	2.6	51
20	Identification and characterization of three novel mutations in theCASQ1gene in four patients with tubular aggregate myopathy. Human Mutation, 2017, 38, 1761-1773.	2.5	51
21	Structure and mutation analysis of the glycogen storage disease type 1b gene. FEBS Letters, 1998, 436, 247-250.	2.8	50
22	Organization of junctional sarcoplasmic reticulum proteins in skeletal muscle fibers. Journal of Muscle Research and Cell Motility, 2015, 36, 501-515.	2.0	40
23	Contractile impairment and structural alterations of skeletal muscles from knockout mice lacking type 1 and type 3 ryanodine receptors. FEBS Letters, 1998, 422, 160-164.	2.8	39
24	Characterization of chloride and cation channels in cultured human keratinocytes. Pflugers Archiv European Journal of Physiology, 1991, 418, 18-25.	2.8	36
25	A novel FLNC frameshift and an OBSCN variant in a family with distal muscular dystrophy. PLoS ONE, 2017, 12, e0186642.	2.5	29
26	Voltage-controlled Ca2+release in normal and ryanodine receptor type 3 (RyR3)-deficient mouse myotubes. Journal of Physiology, 1998, 513, 3-9.	2.9	24
27	Exclusion of linkage between RET and neuronal intestinal dysplasia type B. , 1996, 62, 195-198.		23
28	A forskolin and verapamil sensitive K+ current in human tracheal cells. Biochemical and Biophysical Research Communications, 1991, 179, 1155-1160.	2.1	18
29	Testing an Attachment-Based Parenting Intervention-VIPP-FC/A in Adoptive Families with Post-institutionalized Children: Do Maternal Sensitivity and Genetic Markers Count?. Frontiers in Psychology, 2018, 9, 156.	2.1	16
30	Overexpression of YAP1 induces immortalization of normal human keratinocytes by blocking clonal evolution. Histochemistry and Cell Biology, 2010, 134, 265-276.	1.7	15
31	Yip1B isoform is localized at ERâ€“Golgi intermediate and cis-Golgi compartments and is not required for maintenance of the Golgi structure in skeletal muscle. Histochemistry and Cell Biology, 2015, 143, 235-243.	1.7	14
32	New Insights into the Pathophysiology of Primary and Secondary Lymphedema: Histopathological Studies on Human Lymphatic Collecting Vessels. Lymphatic Research and Biology, 2020, 18, 502-509.	1.1	14
33	Prevalence and Parental Origin of de novo RET Mutations in Hirschsprung's Disease. European Journal of Human Genetics, 1996, 4, 356-358.	2.8	13
34	Chemical Characterisation and Antihypertensive Effects of Locular Gel and Serum of Lycopersicum esculentum L. var. â€œCamoneâ€•Tomato in Spontaneously Hypertensive Rats. Molecules, 2020, 25, 3758.	3.8	11
35	Lymphatic Collecting Vessels in Health and Disease: A Review of Histopathological Modifications in Lymphedema. Lymphatic Research and Biology, 2022, , .	1.1	7
36	Examining the Impact of Maternal Individual Features on Childrenâ€™s Behavioral Problems in Adoptive Families: The Role of Maternal Temperament and Neurobiological Markers. International Journal of Environmental Research and Public Health, 2018, 15, 196.	2.6	3

#	Article	IF	Citations
37	Ryanodine-Sensitive Calcium Release Channels. , 2000, , 205-219.		1
38	Identification of a Calsequestrin-1 Mutation in a Human Vacuolar Myopathy. Biophysical Journal, 2015, 108, 503a.	0.5	0
39	A Novel FLNC Frameshift and an OBSCN Variant in a Family with Distal Muscular Distrophy. Biophysical Journal, 2018, 114, 136a.	0.5	0
40	Light-Induced Smooth Endoplasmic Reticulum Rearrangement in a Unique Interlaced Compartmental Pattern in Macaca mulatta RPE. , 2021, 62, 32.		0