Patrick Frosk

List of Publications by Year
in descending order

31 papers	1,732 citations	516710 16 h-index	501196 28 g-index
31 all docs	31 docs citations	31 times ranked	3355 citing authors

#	Article	IF	Citations
1	Utility of wholeâ€exome sequencing for those near the end of the diagnostic odyssey: time to address gaps in care. Clinical Genetics, 2016, 89, 275-284.	2.0	323
2	Limb-Girdle Muscular Dystrophy Type 2H Associated with Mutation in TRIM32, a Putative E3-Ubiquitinâ€“Ligase Gene. American Journal of Human Genetics, 2002, 70, 663-672.	6.2	227
3	An siRNA-based functional genomics screen for theÂidentification of regulators of ciliogenesis and ciliopathyÂgenes. Nature Cell Biology, 2015, 17, 1074-1087.	10.3	215
4	TMEM237 Is Mutated in Individuals with a Joubert Syndrome Related Disorder and Expands the Role of the TMEM Family at the Ciliary Transition Zone. American Journal of Human Genetics, 2011, 89, 713-730.	6.2	178
5	Neuropathological, biochemical and molecular findings in a glutaric acidemia type 1 cohort. Brain, 2005, 128, 711-722.	7.6	137
6	Cardiac and respiratory failure in limb-girdle muscular dystrophy 2I. Annals of Neurology, 2004, 56, 738-741.	5.3	110
7	Commonality ofTRIM32mutation in causing sarcotubular myopathy and LGMD2H. Annals of Neurology, 2005, 57, 591-595.	5.3	96
8	The Interaction of Piasy with Trim32, an E3-Ubiquitin Ligase Mutated in Limb-girdle Muscular Dystrophy Type 2H, Promotes Piasy Degradation and Regulates UVB-induced Keratinocyte Apoptosis through NFÎºB. Journal of Biological Chemistry, 2006, 281, 25850-25866.	3.4	77
9	The most common mutation inFKRP causing limb girdle muscular dystrophy type 2I (LGMD2I) may have occurred only once and is present in Hutterites and other populations. Human Mutation, 2005, 25, 38-44.	2.5	71
10	A truncating mutation in CEP55 is the likely cause of MARCH, a novel syndrome affecting neuronal mitosis. Journal of Medical Genetics, 2017, 54, 490-501.	3.2	45
11	Multisystem inflammation and susceptibility to viral infections in human ZNFX1 deficiency. Journal of Allergy and Clinical Immunology, 2021, 148, 381-393.	2.9	40
12	Characterization of a Unique Form of Arrhythmic Cardiomyopathy Caused by Recessive Mutation in LEMD2. JACC Basic To Translational Science, 2019, 4, 204-221.	4.1	37
13	Hutterite brothers both affected with two forms of limb girdle muscular dystrophy: LGMD2H and LGMD2I. European Journal of Human Genetics, 2005, 13, 978-982.	2.8	31
14	The Canadian Rare Diseases Models and Mechanisms (RDMM) Network: Connecting Understudied Genes to Model Organisms. American Journal of Human Genetics, 2020, 106, 143-152.	6.2	30
15	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.	12.8	28
16	SCN1A Mutation Associated With Intractable Myoclonic Epilepsy and Migraine Headache. Journal of Child Neurology, 2013, 28, 389-391.	1.4	18
17	A novel CCBE1 mutation leading to a mild form of hennekam syndrome: case report and review of the literature. BMC Medical Genetics, 2015, 16, 28.	2.1	14
18	A homozygous canonical splice acceptor site mutation in PRUNE1 is responsible for a rare childhood neurodegenerative disease in Manitoba Cree families. American Journal of Medical Genetics, Part A, 2019, 179, 206-218.	1.2	12

#	Article	IF	Citations
19	Compound heterozygous variants in SHQ1 are associated with a spectrum of neurological features, including early-onset dystonia. Human Molecular Genetics, 2022, 31, 614-624.	2.9	12
20	The importance of functional validation after nextâ€generation sequencing: evaluation of a novel <scp>CARD</scp>11 variant. Pediatric Allergy and Immunology, 2018, 29, 663-668.	2.6	8
21	The Use of Ancestral Haplotypes in the Molecular Diagnosis of Familial Breast Cancer. Genetic Testing and Molecular Biomarkers, 2007, 11, 208-215.	1.7	5
22	Recurrent Posterior Circulation Stroke in an Infant With Basilar Artery Aneurysm. Journal of Child Neurology, 2009, 24, 1019-1020.	1.4	3
23	Acquired Microcephaly in a Patient with HECW2 Mutation. Canadian Journal of Neurological Sciences, 2021, 48, 435-437.	0.5	3
24	Further delineation of BCAP31-linked intellectual disability: description of 17 new families with LoF and missense variants. European Journal of Human Genetics, 2021, 29, 1405-1417.	2.8	3
25	Heat-Labile Serotyping of Two Campylobacter jejuni Strains Isolated from Patients with Guillain-Barrè Syndrome and Belonging to Serotype O19 (Penner) . Journal of Clinical Microbiology, 2000, 38, 2021-2022.	3.9	3
26	Heterozygous intragenic deletions of FREM1 are not associated with trigonocephaly. Clinical Dysmorphology, 2021, 30, 83-88.	0.3	3
27	Scientific correspondence. Neuropathology and Applied Neurobiology, 2014, 40, 946-950.	3.2	2
28	Origin of a Prenatal Mosaic Supernumerary Neocentromeric Derivative Chromosome 13 Determined by QF-PCR. Fetal Diagnosis and Therapy, 2013, 33, 75-78.	1.4	1
29	Cover Image, Volume 179A, Number 2, February 2019. American Journal of Medical Genetics, Part A, 2019, 179, i.	1.2	0
30	Limb Girdle Muscular Dystrophy Type 2H. , 2009, , 1184-1185.		0
31	Limb Girdle Muscular Dystrophy Type 2B and Miyoshi Myopathy. , 2009, , 1181-1182.		0