Vilma-Lotta Lehtokari

List of Publications by Year
in descending order

22	1,296	567247 15	677123 22
papers	citations	h-index	g-index

24	24	24	1146
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Nemaline Myopathy with Minicores Caused by Mutation of the CFL2 Gene Encoding the Skeletal Muscle Actinâ€“Binding Protein, Cofilin-2. American Journal of Human Genetics, 2007, 80, 162-167.	6.2	213
2	Mutations in KLHL40 Are a Frequent Cause of Severe Autosomal-Recessive Nemaline Myopathy. American Journal of Human Genetics, 2013, 93, 6-18.	6.2	186
3	Distal myopathy caused by homozygous missense mutations in the nebulin gene. Brain, 2007, 130, 1465-1476.	7.6	130
4	Identification of 45 novel mutations in the nebulin gene associated with autosomal recessive nemaline myopathy. Human Mutation, 2006, 27, 946-956.	2.5	112
5	Mutation Update: The Spectra of Nebulin Variants and Associated Myopathies. Human Mutation, 2014, 35, 1418-1426.	2.5	107
6	Complete genomic structure of the human nebulin gene and identification of alternatively spliced transcripts. European Journal of Human Genetics, 2004, 12, 744-751.	2.8	101
7	Cap disease caused by heterozygous deletion of the Î²-tropomyosin gene TPM2. Neuromuscular Disorders, 2007, 17, 433-442.	0.6	90
8	Nemaline myopathy caused by mutations in the nebulin gene may present as a distal myopathy. Neuromuscular Disorders, 2011, 21, 556-562.	0.6	56
9	The exon 55 deletion in the nebulin gene â€“ One single founder mutation with world-wide occurrence. Neuromuscular Disorders, 2009, 19, 179-181.	0.6	54
10	Mutationâ€specific effects on thin filament length in thin filament myopathy. Annals of Neurology, 2016, 79, 959-969.	5.3	54
11	Identification of a founder mutation in TPM3 in nemaline myopathy patients of Turkish origin. European Journal of Human Genetics, 2008, 16, 1055-1061.	2.8	36
12	A recurrent copy number variation of the NEB triplicate region: only revealed by the targeted nemaline myopathy CGH array. European Journal of Human Genetics, 2016, 24, 574-580.	2.8	32
13	Dominantly inherited distal nemaline/cap myopathy caused by a large deletion in the nebulin gene. Neuromuscular Disorders, 2019, 29, 97-107.	0.6	31
14	An Extended Targeted Copy Number Variation Detection Array Including 187 Genes for the Diagnostics of Neuromuscular Disorders. Journal of Neuromuscular Diseases, 2018, 5, 307-314.	2.6	17
15	MoBiDiC Prioritization Algorithm, a Free, Accessible, and Efficient Pipeline for Single-Nucleotide Variant Annotation and Prioritization for Next-Generation Sequencing Routine Molecular Diagnosis. Journal of Molecular Diagnostics, 2018, 20, 465-473.	2.8	13
16	Bilateral foot-drop as predominant symptom in nebulin (NEB) gene related â€œcore-rodâ€•congenital myopathy. European Journal of Medical Genetics, 2015, 58, 556-561.	1.3	12
17	A nebulin superâ€•repeat panel reveals stronger actin binding toward the ends of the superâ€•repeat region. Muscle and Nerve, 2019, 59, 116-121.	2.2	10
18	Congenital asymmetric distal myopathy with hemifacial weakness caused by a heterozygous large de novo mosaic deletion in nebulin. Neuromuscular Disorders, 2021, 31, 539-545.	0.6	9

#	Article	IF	Citations
19	Clinically variable nemaline myopathy in a three-generation family caused by mutation of the skeletal muscle alpha-actin gene. Neuromuscular Disorders, 2018, 28, 323-326.	0.6	6
20	A custom ddPCR method for the detection of copy number variations in the nebulin triplicate region. PLoS ONE, 2022, 17, e0267793.	2.5	2
21	A commentary on identification of the rare compound heterozygous variants in the NEB gene in a Korean family with intellectual disability, epilepsy and early-childhood-onset generalized muscle weakness. Journal of Human Genetics, 2015, 60, 161-162.	2.3	1
22	Array Comparative Genomic Hybridisation and Droplet Digital PCR Uncover Recurrent Copy Number Variation of the TTN Segmental Duplication Region. Genes, 2022, 13, 905.	2.4	0