Mireille Cossée

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/5234839/publications.pdf Version: 2024-02-01



#	Article	IF	CITATIONS
1	A National French Consensus on Gene List for the Diagnosis of Charcot–Marie–Tooth Disease and Related Disorders Using Next-Generation Sequencing. Genes, 2022, 13, 318.	2.4	4
2	Long-Reads Sequencing Strategy to Localize Variants in TTN Repeated Domains. Journal of Molecular Diagnostics, 2022, 24, 719-726.	2.8	0
3	Clinical phenotype and loss of the slow skeletal muscle troponin T in three new patients with recessive TNNT1 nemaline myopathy. Journal of Medical Genetics, 2021, 58, 602-608.	3.2	11
4	Evaluating next-generation sequencing in neuromuscular diseases with neonatal respiratory distress. European Journal of Paediatric Neurology, 2021, 31, 78-87.	1.6	8
5	Association between prophylactic angiotensin-converting enzyme inhibitors and overall survival in Duchenne muscular dystrophy—analysis of registry data. European Heart Journal, 2021, 42, 1976-1984.	2.2	25
6	Novel dominant distal titinopathy phenotype associated with copy number variation. Annals of Clinical and Translational Neurology, 2021, 8, 1906-1912.	3.7	3
7	An Integrated Clinical-Biological Approach to Identify Interindividual Variability and Atypical Phenotype-Genotype Correlations in Myopathies: Experience on A Cohort of 156 Families. Genes, 2021, 12, 1199.	2.4	8
8	The genomic and clinical landscape of fetal akinesia. Genetics in Medicine, 2020, 22, 511-523.	2.4	35
9	ASCâ€∎ Is a Cell Cycle Regulator Associated with Severe and Mild Forms of Myopathy. Annals of Neurology, 2020, 87, 217-232.	5.3	12
10	The importance of an integrated genotype-phenotype strategy to unravel the molecular bases of titinopathies. Neuromuscular Disorders, 2020, 30, 877-887.	0.6	18
11	Novel <i>CAPN3</i> variant associated with an autosomal dominant calpainopathy. Neuropathology and Applied Neurobiology, 2020, 46, 564-578.	3.2	20
12	A new congenital multicore titinopathy associated with fast myosin heavy chain deficiency. Annals of Clinical and Translational Neurology, 2020, 7, 846-854.	3.7	8
13	Corrélations phénotype-génotype dans les titinopathies. Les Cahiers De Myologie, 2020, , 16-20.	0.0	1
14	A National French consensus on gene lists for the diagnosis of myopathies using next-generation sequencing. European Journal of Human Genetics, 2019, 27, 349-352.	2.8	27
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	A Reliable Targeted Next-Generation Sequencing Strategy for Diagnosis of Myopathies and Muscular Dystrophies, Especially for the Giant Titin and Nebulin Genes. Journal of Molecular Diagnostics, 2018, 20, 533-549.	2.8	28
17	Muscle involvement in limb-girdle muscular dystrophy with GMPPB deficiency (LGMD2T). Neurology: Genetics, 2016, 2, e112.	1.9	29
18	Implementation of a Reliable Next-Generation Sequencing Strategy for Molecular Diagnosis of Dystrophinopathies. Journal of Molecular Diagnostics, 2016, 18, 731-740.	2.8	24

#	Article	lF	CITATIONS
19	Detection of TRIM32 deletions in LGMD patients analyzed by a combined strategy of CGH array and massively parallel sequencing. European Journal of Human Genetics, 2015, 23, 929-934.	2.8	21
20	Custom oligonucleotide array-based CGH: a reliable diagnostic tool for detection of exonic copy-number changes in multiple targeted genes. European Journal of Human Genetics, 2013, 21, 977-987.	2.8	20
21	Genotype-phenotype analysis in 2,405 patients with a dystrophinopathy using the UMD-DMD database: a model of nationwide knowledgebase. Human Mutation, 2009, 30, 934-945.	2.5	309
22	Detection of exonic copy-number changes using a highly efficient oligonucleotide-based comparative genomic hybridization-array method. Human Mutation, 2008, 29, 1083-1090.	2.5	51