Marcus Deschauer

List of Publications by Year
in descending order

32	644	687363 13	610901 24
papers	citations	h-index	g-index

33	33	33	953
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Characterization of cognitive impairment in adult polyglucosan body disease. Journal of Neurology, 2022, 269, 2854-2861.	3.6	6
2	Clonal hematopoiesis as a pitfall in germline variant interpretation in the context of Mendelian disorders. Human Molecular Genetics, 2022, 31, 2386-2395.	2.9	7
3	A Novel <scp>NPTX1 de novo</scp> Variant in a Lateâ€Onset Ataxia Patient. Movement Disorders, 2022, 37, 1319-1321.	3.9	2
4	Adultâ€Onset Neurodegeneration in Nucleotide Excision Repair Disorders (<scp>NERD_{ND}</scp>): Time to Move Beyond the Skin. Movement Disorders, 2022, 37, 1707-1718.	3.9	7
5	GFPT1-Associated Congenital Myasthenic Syndrome Mimicking a Glycogen Storage Disease â€“ Diagnostic Pitfalls in Myopathology Solved by Next-Generation-Sequencing. Journal of Neuromuscular Diseases, 2022, , 1-9.	2.6	1
6	Defective phosphatidylethanolamine biosynthesis leads to a broad ataxia-spasticity spectrum. Brain, 2021, 144, e30-e30.	7.6	12
7	A Nation-Wide, Multi-Center Study on the Quality of Life of ALS Patients in Germany. Brain Sciences, 2021, 11, 372.	2.3	15
8	Serum creatine kinase and creatinine in adult spinal muscular atrophy under nusinersen treatment. Annals of Clinical and Translational Neurology, 2021, 8, 1049-1063.	3.7	29
9	Regional variation of thigh muscle fat infiltration in patients with neuromuscular diseases compared to healthy controls. Quantitative Imaging in Medicine and Surgery, 2021, 11, 2610-2621.	2.0	7
10	Quantitative Muscle MRI in Patients with Neuromuscular Diseasesâ€”Association of Muscle Proton Density Fat Fraction with Semi-Quantitative Grading of Fatty Infiltration and Muscle Strength at the Thigh Region. Diagnostics, 2021, 11, 1056.	2.6	9
11	Informal Caregiving in Amyotrophic Lateral Sclerosis (ALS): A High Caregiver Burden and Drastic Consequences on Caregiversâ€™ Lives. Brain Sciences, 2021, 11, 748.	2.3	30
12	Combined Treatment With Pembrolizumab and Allogenic BK Virus-Specific T Cells in Progressive Multifocal Leukoencephalopathy. Neurology: Neuroimmunology and NeuroInflammation, 2021, 8, e1042.	6.0	5
13	Bi-allelic truncating mutations in VWA1 cause neuromyopathy. Brain, 2021, 144, 574-583.	7.6	16
14	Clinico-genetic findings in 509 frontotemporal dementia patients. Molecular Psychiatry, 2021, 26, 5824-5832.	7.9	23
15	Brain Iron and Metabolic Abnormalities in C19orf12 Mutation Carriers: A 7.0 Tesla MRI Study in Mitochondrial Membrane Proteinâ€“Associated Neurodegeneration. Movement Disorders, 2020, 35, 142-150.	3.9	16
16	Water T 2 Mapping in Fatty Infiltrated Thigh Muscles of Patients With Neuromuscular Diseases Using a T 2 â€Prepared 3D Turbo Spin Echo With SPAIR. Journal of Magnetic Resonance Imaging, 2020, 51, 1727-1736.	3.4	13
17	Bi-allelic HPDL Variants Cause a Neurodegenerative Disease Ranging from Neonatal Encephalopathy to Adolescent-Onset Spastic Paraplegia. American Journal of Human Genetics, 2020, 107, 364-373.	6.2	30
18	Bilateral thoracic disc herniation with abdominal wall paresis: a case report. Acta Neurochirurgica, 2020, 162, 2055-2059.	1.7	6

#	Article	IF	Citations
19	Nusinersen in adults with 5q spinal muscular atrophy: a non-interventional, multicentre, observational cohort study. Lancet Neurology, The, 2020, 19, 317-325.	10.2	196
20	Progressive external ophthalmoplegia due to a recurrent de novo m.15990C>T MT-TP (mt-tRNAPro) gene variant. Neuromuscular Disorders, 2020, 30, 346-350.	0.6	4
21	Intrathecal nusinersen administration in adult spinal muscular atrophy patients with complex spinal anatomy. Therapeutic Advances in Neurological Disorders, 2020, 13, 175628641988761.	3.5	21
22	Radiation dose reduction for CT-guided intrathecal nusinersen administration in adult patients with spinal muscular atrophy. Scientific Reports, 2020, 10, 3406.	3.3	7
23	Regional Variation of Thigh Muscle Composition in Healthy Controls and Patients with Myotonic Dystrophy Type 2, Limb Girdle Muscular Dystrophy Type 2A, and Pompeâ€™s Disease. , 2020, 24, .		0
24	Patient-Reported Prevalence of Non-motor Symptoms Is Low in Adult Patients Suffering From 5q Spinal Muscular Atrophy. Frontiers in Neurology, 2019, 10, 1098.	2.4	12
25	Understanding mitochondrial DNA maintenance disorders at the single muscle fibre level. Nucleic Acids Research, 2019, 47, 7430-7443.	14.5	16
26	Decreased water T₂ in fatty infiltrated skeletal muscles of patients with neuromuscular diseases. NMR in Biomedicine, 2019, 32, e4111.	2.8	20
27	Phenotype of carnitine palmitoyltransferase II (CPT II) deficiency: A questionnaire-based survey. Journal of Clinical Neuroscience, 2019, 59, 32-36.	1.5	13
28	Prevalence of Headache in Patients With Mitochondrial Disease: A Crossâ€Sectional Study. Headache, 2018, 58, 45-52.	3.9	39
29	FGF-21 as a Potential Biomarker for Mitochondrial Diseases. Current Medicinal Chemistry, 2018, 25, 2070-2081.	2.4	26
30	Peripheral neuropathy in patients with CPEO associated with single and multiple mtDNA deletions. Neurology: Genetics, 2016, 2, e113.	1.9	12
31	Status Epilepticus After Subthalamic Deep Brain Stimulation Surgery in a Patient with Parkinson's Disease. World Neurosurgery, 2016, 96, 614.e1-614.e6.	1.3	3
32	A novel Twinkle gene mutation in autosomal dominant progressive external ophthalmoplegia. Neuromuscular Disorders, 2003, 13, 568-572.	0.6	41