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	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
2	A novel Twinkle gene mutation in autosomal dominant progressive external ophthalmoplegia. Neuromuscular Disorders, 2003, 13, 568-572.	0.6	41
3	Prevalence of Headache in Patients With Mitochondrial Disease: A Crossâ€Sectional Study. Headache, 2018, 58, 45-52.	3.9	39
4	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
5	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
6	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
7	FGF-21 as a Potential Biomarker for Mitochondrial Diseases. Current Medicinal Chemistry, 2018, 25, 2070-2081.	2.4	26
8	Clinico-genetic findings in 509 frontotemporal dementia patients. Molecular Psychiatry, 2021, 26, 5824-5832.	7.9	23
9	Intrathecal nusinersen administration in adult spinal muscular atrophy patients with complex spinal anatomy. Therapeutic Advances in Neurological Disorders, 2020, 13, 175628641988761.	3.5	21
10	Decreased water T₂ in fatty infiltrated skeletal muscles of patients with neuromuscular diseases. NMR in Biomedicine, 2019, 32, e4111.	2.8	20
11	Understanding mitochondrial DNA maintenance disorders at the single muscle fibre level. Nucleic Acids Research, 2019, 47, 7430-7443.	14.5	16
12	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
13	Bi-allelic truncating mutations in VWA1 cause neuromyopathy. Brain, 2021, 144, 574-583.	7.6	16
14	A Nation-Wide, Multi-Center Study on the Quality of Life of ALS Patients in Germany. Brain Sciences, 2021, 11, 372.	2.3	15
15	Phenotype of carnitine palmitoyltransferase II (CPT II) deficiency: A questionnaire-based survey. Journal of Clinical Neuroscience, 2019, 59, 32-36.	1.5	13
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	Peripheral neuropathy in patients with CPEO associated with single and multiple mtDNA deletions. Neurology: Genetics, 2016, 2, e113.	1.9	12
18	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

#	Article	IF	Citations
19	Defective phosphatidylethanolamine biosynthesis leads to a broad ataxia-spasticity spectrum. Brain, 2021, 144, e30-e30.	7.6	12
20	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
21	Radiation dose reduction for CT-guided intrathecal nusinersen administration in adult patients with spinal muscular atrophy. Scientific Reports, 2020, 10, 3406.	3.3	7
22	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
23	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
24	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
25	Bilateral thoracic disc herniation with abdominal wall paresis: a case report. Acta Neurochirurgica, 2020, 162, 2055-2059.	1.7	6
26	Characterization of cognitive impairment in adult polyglucosan body disease. Journal of Neurology, 2022, 269, 2854-2861.	3.6	6
27	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
28	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
29	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
30	A Novel <scp>NPTX1 de novo</scp> Variant in a Lateâ€Onset Ataxia Patient. Movement Disorders, 2022, 37, 1319-1321.	3.9	2
31	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
32	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