Andreas Roos

List of Publications by Year
in descending order

66 papers	1,107 citations	471061 17 h-index	500791 28 g-index
69 all docs	69 docs citations	69 times ranked	1657 citing authors

#	Article	IF	Citations
1	A de novo <scp>CSDE1</scp> variant causing neurodevelopmental delay, intellectual disability, neurologic and psychiatric symptoms in a child of consanguineous parents. American Journal of Medical Genetics, Part A, 2022, 188, 283-291.	0.7	1
2	High diagnostic rate of trio exome sequencing in consanguineous families with neurogenetic diseases. Brain, 2022, 145, 1507-1518.	3.7	14
3	Multispectral optoacoustic tomography for non-invasive disease phenotyping in pediatric spinal muscular atrophy patients. Photoacoustics, 2022, 25, 100315.	4.4	16
4	Novel insights into PORCN mutations, associated phenotypes and pathophysiological aspects. Orphanet Journal of Rare Diseases, 2022, 17, 29.	1.2	3
5	Identification of a novel homozygous synthesis of cytochrome c oxidase 2 variant in siblings with earlyâ€onset axonal Charcotâ€Marieâ€Tooth disease. Human Mutation, 2022, 43, 477-486.	1.1	3
6	NCAM1 and GDF15 are biomarkers of Charcot-Marie-Tooth disease in patients and mice. Brain, 2022, 145, 3999-4015.	3.7	12
7	Serum miRNAs as biomarkers for the rare types of muscular dystrophy. Neuromuscular Disorders, 2022, 32, 332-346.	0.3	5
8	Cathepsin D as biomarker in cerebrospinal fluid of nusinersenâ€treated patients with spinal muscular atrophy. European Journal of Neurology, 2022, 29, 2084-2096.	1.7	13
9	Circulating small RNA signatures differentiate accurately the subtypes of muscular dystrophies: small-RNA next-generation sequencing analytics and functional insights. RNA Biology, 2022, 19, 507-518.	1.5	1
10	Skeletal muscle provides the immunological micro-milieu for specific plasma cells in anti-synthetase syndrome-associated myositis. Acta Neuropathologica, 2022, 144, 353-372.	3.9	19
11	New Insights into the Neuromyogenic Spectrum of a Gain of Function Mutation in SPTLC1. Genes, 2022, 13, 893.	1.0	2
12	Endoplasmic reticulumâ€stress and unfolded protein responseâ€activation in immuneâ€mediated necrotizing myopathy. Brain Pathology, 2022, 32, .	2.1	7
13	Diagnosing X-linked Myotubular Myopathy â€“ A German 20-year Follow Up Experience. Journal of Neuromuscular Diseases, 2021, 8, 79-90.	1.1	12
14	Expanding the clinical and molecular spectrum of <scp>ATP6V1A</scp> related metabolic cutis laxa. Journal of Inherited Metabolic Disease, 2021, 44, 972-986.	1.7	7
15	Intellectual disability associated with craniofacial dysmorphism, cleft palate, and congenital heart defect due to a de novo <scp>MEIS2</scp> mutation: A clinical longitudinal study. American Journal of Medical Genetics, Part A, 2021, 185, 1216-1221.	0.7	9
16	Protein signature of human skin fibroblasts allows the study of the molecular etiology of rare neurological diseases. Orphanet Journal of Rare Diseases, 2021, 16, 73.	1.2	18
17	Molecular pathophysiology of human MICU1 deficiency. Neuropathology and Applied Neurobiology, 2021, 47, 840-855.	1.8	15
18	INPP5K and SIL1 associated pathologies with overlapping clinical phenotypes converge through dysregulation of PHGDH. Brain, 2021, 144, 2427-2442.	3.7	7

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19	Childhood amyotrophic lateral sclerosis caused by excess sphingolipid synthesis. Nature Medicine, 2021, 27, 1197-1204.	15.2	96
20	Exome reanalysis and proteomic profiling identified TRIP4 as a novel cause of cerebellar hypoplasia and spinal muscular atrophy (PCH1). European Journal of Human Genetics, 2021, 29, 1348-1353.	1.4	10
21	Muscular and Molecular Pathology Associated with SPATA5 Deficiency in a Child with EHLMRS. International Journal of Molecular Sciences, 2021, 22, 7835.	1.8	4
22	Dysregulation of GSK3Î²-Target Proteins in Skin Fibroblasts of Myotonic Dystrophy Type 1 (DM1) Patients. Journal of Neuromuscular Diseases, 2021, 8, 603-619.	1.1	2
23	Clinical Course, Myopathology and Challenge of Therapeutic Intervention in Pediatric Patients with Autoimmune-Mediated Necrotizing Myopathy. Children, 2021, 8, 721.	0.6	7
24	Autosomal recessive variants in TUBGCP2 alter the Î³-tubulin ring complex leading to neurodevelopmental disease. IScience, 2021, 24, 101948.	1.9	6
25	Intracellular Lipid Accumulation and Mitochondrial Dysfunction Accompanies Endoplasmic Reticulum Stress Caused by Loss of the Co-chaperone DNAJC3. Frontiers in Cell and Developmental Biology, 2021, 9, 710247.	1.8	13
26	Homozygous WASHC4 variant in two sisters causes a syndromic phenotype defined by dysmorphisms, intellectual disability, profound developmental disorder, and skeletal muscle involvement. Journal of Pathology, 2021, , .	2.1	5
27	Three Individuals with PURA Syndrome in a Cohort of Patients with Neuromuscular Disease. Neuropediatrics, 2021, 52, 390-393.	0.3	7
28	Noninvasive Imaging in Pediatric Spinal Muscular Atrophy Patients Using Multispectral Optoacoustic Tomography: A Proof-of-Concept Study. Neuropediatrics, 2021, 52, .	0.3	0
29	Phenotypical and Myopathological Consequences of Compound Heterozygous Missense and Nonsense Variants in SLC18A3. Cells, 2021, 10, 3481.	1.8	1
30	Severe neurodevelopmental disease caused by a homozygous TLK2 variant. European Journal of Human Genetics, 2020, 28, 383-387.	1.4	6
31	Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308.	1.7	43
32	Further evidence for POMK as candidate gene for WWS with meningoencephalocele. Orphanet Journal of Rare Diseases, 2020, 15, 242.	1.2	4
33	Long Term Follow-Up on Pediatric Cases With Congenital Myasthenic Syndromesâ€"A Retrospective Single Centre Cohort Study. Frontiers in Human Neuroscience, 2020, 14, 560860.	1.0	14
34	Blood-derived biomarkers correlate with clinical progression in Duchenne muscular dystrophy. Journal of Neuromuscular Diseases, 2020, 7, 231-246.	1.1	20
35	Advances in the diagnosis of inherited neuromuscular diseases and implications for therapy development. Lancet Neurology, The, 2020, 19, 522-532.	4.9	36
36	Immunofluorescence-Based Analysis of Caveolin-3 in the Diagnostic Management of Neuromuscular Diseases. Methods in Molecular Biology, 2020, 2169, 197-216.	0.4	5

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37	Modulation of the Acetylcholine Receptor Clustering Pathway Improves Neuromuscular Junction Structure and Muscle Strength in a Mouse Model of Congenital Myasthenic Syndrome. Frontiers in Molecular Neuroscience, 2020, 13, 594220.	1.4	5
38	Modulation of Agrin and RhoA Pathways Ameliorates Movement Defects and Synapse Morphology in MYO9A-Depleted Zebrafish. Cells, 2019, 8, 848.	1.8	10
39	Identification of Cellular Pathogenicity Markers for SIL1 Mutations Linked to Marinesco-SjÃ¶gren Syndrome. Frontiers in Neurology, 2019, 10, 562.	1.1	5
40	Proteomic Profiling Unravels a Key Role of Specific Macrophage Subtypes in Sporadic Inclusion Body Myositis. Frontiers in Immunology, 2019, 10, 1040.	2.2	17
41	Identification of Candidate Protein Markers in Skeletal Muscle of Laminin-211-Deficient CMD Type 1A-Patients. Frontiers in Neurology, 2019, 10, 470.	1.1	14
42	SIL1 deficiency causes degenerative changes of peripheral nerves and neuromuscular junctions in fish, mice and human. Neurobiology of Disease, 2019, 124, 218-229.	2.1	7
43	The beta-adrenergic agonist salbutamol modulates neuromuscular junction formation in zebrafish models of human myasthenic syndromes. Human Molecular Genetics, 2018, 27, 1556-1564.	1.4	28
44	MYO9A deficiency in motor neurons is associated with reduced neuromuscular agrin secretion. Human Molecular Genetics, 2018, 27, 1434-1446.	1.4	14
45	Mitochondrial oxodicarboxylate carrier deficiency is associated with mitochondrial DNA depletion and spinal muscular atrophyâ€“like disease. Genetics in Medicine, 2018, 20, 1224-1235.	1.1	31
46	Mutations in glycyl-tRNA synthetase impair mitochondrial metabolism in neurons. Human Molecular Genetics, 2018, 27, 2187-2204.	1.4	26
47	PFN2 and GAMT as common molecular determinants of axonal Charcot-Marie-Tooth disease. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 870-878.	0.9	16
48	Clinical and research strategies for limbâ€girdle congenital myasthenic syndromes. Annals of the New York Academy of Sciences, 2018, 1412, 102-112.	1.8	17
49	Multifocal demyelinating motor neuropathy and hamartoma syndrome associated with a de novo PTEN mutation. Neurology, 2018, 90, e1842-e1848.	1.5	4
50	Tracking Effects of SIL1 Increase: Taking a Closer Look Beyond the Consequences of Elevated Expression Level. Molecular Neurobiology, 2018, 55, 2524-2546.	1.9	15
51	Mass spectrometryâ€based protein analysis to unravel the tissue pathophysiology in Duchenne muscular dystrophy. Proteomics - Clinical Applications, 2018, 12, 1700071.	0.8	26
52	Intersection of Proteomics and Genomics to â€œSolve the Unsolvedâ€• in Rare Disorders such as Neurodegenerative and Neuromuscular Diseases. Proteomics - Clinical Applications, 2018, 12, 1700073.	0.8	33
53	Neuromuscular Junction Changes in a Mouse Model of Charcot-Marie-Tooth Disease Type 4C. International Journal of Molecular Sciences, 2018, 19, 4072.	1.8	24
54	Comprehensive RNA-Sequencing Analysis in Serum and Muscle Reveals Novel Small RNA Signatures with Biomarker Potential for DMD. Molecular Therapy - Nucleic Acids, 2018, 13, 1-15.	2.3	41

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55	Biochemical and pathological changes result from mutated Caveolin-3 in muscle. Skeletal Muscle, 2018, 8, 28.	1.9	19
56	GFPT1 deficiency in muscle leads to myasthenia and myopathy in mice. Human Molecular Genetics, 2018, 27, 3218-3232.	1.4	18
57	Mutations in INPP5K , Encoding a Phosphoinositide 5-Phosphatase, Cause Congenital Muscular Dystrophy with Cataracts and Mild Cognitive Impairment. American Journal of Human Genetics, 2017, 100, 523-536.	2.6	67
58	Intragenic DOK7 deletion detected by whole-genome sequencing in congenital myasthenic syndromes. Neurology: Genetics, 2017, 3, e152.	0.9	4
59	Muscle Pathology as a Diagnostic Clue to Allgrove Syndrome. Journal of Neuropathology and Experimental Neurology, 2017, 76, 337-341.	0.9	13
60	MRC Centre Neuromuscular Biobank (Newcastle and London): Supporting and facilitating rare and neuromuscular disease research worldwide. Neuromuscular Disorders, 2017, 27, 1054-1064.	0.3	15
61	The Caveolin-3 G56S sequence variant of unknown significance: Muscle biopsy findings and functional cell biological analysis. Proteomics - Clinical Applications, 2017, 11, 1600007.	0.8	6
62	Cellular Signature of SIL1 Depletion: Disease Pathogenesis due to Alterations in Protein Composition Beyond the ER Machinery. Molecular Neurobiology, 2016, 53, 5527-5541.	1.9	30
63	Inverted formin 2â€related Charcotâ€Marieâ€Tooth disease: extension of the mutational spectrum and pathological findings in Schwann cells and axons. Journal of the Peripheral Nervous System, 2015, 20, 52-59.	1.4	21
64	Myopathy in Marinescoâ€“SjÃ¶gren syndrome links endoplasmic reticulum chaperone dysfunction to nuclear envelope pathology. Acta Neuropathologica, 2014, 127, 761-777.	3.9	51
65	SIL1-negative Marinesco-SjÃ¶gren syndrome: First report of two sibs from India. Journal of Pediatric Neurosciences, 2014, 9, 291.	0.2	2
66	SH3TC2, a protein mutant in Charcotâ€“Marieâ€“Tooth neuropathy, links peripheral nerve myelination to endosomal recycling. Brain, 2010, 133, 2462-2474.	3.7	82