# 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 | Childhood amyotrophic lateral sclerosis caused by excess sphingolipid synthesis. Nature Medicine, 2021, 27, 1197-1204. | 15.2 | 96 |
| 2 | SH3TC2, a protein mutant in Charcotâ€“Marieâ€“Tooth neuropathy, links peripheral nerve myelination to endosomal recycling. Brain, 2010, 133, 2462-2474. | 3.7 | 82 |
| 3 | 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 |
| 4 | Myopathy in Marinescoâ€“SjÃ¶gren syndrome links endoplasmic reticulum chaperone dysfunction to nuclear envelope pathology. Acta Neuropathologica, 2014, 127, 761-777. | 3.9 | 51 |
| 5 | Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308. | 1.7 | 43 |
| 6 | 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 |
| 7 | Advances in the diagnosis of inherited neuromuscular diseases and implications for therapy development. Lancet Neurology, The, 2020, 19, 522-532. | 4.9 | 36 |
| 8 | 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 |
| 9 | 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 |
| 10 | 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 |
| 11 | 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 |
| 12 | Mutations in glycyl-tRNA synthetase impair mitochondrial metabolism in neurons. Human Molecular Genetics, 2018, 27, 2187-2204. | 1.4 | 26 |
| 13 | Mass spectrometryâ€based protein analysis to unravel the tissue pathophysiology in Duchenne muscular dystrophy. Proteomics - Clinical Applications, 2018, 12, 1700071. | 0.8 | 26 |
| 14 | 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 |
| 15 | 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 |
| 16 | Blood-derived biomarkers correlate with clinical progression in Duchenne muscular dystrophy. Journal of Neuromuscular Diseases, 2020, 7, 231-246. | 1.1 | 20 |
| 17 | Biochemical and pathological changes result from mutated Caveolin-3 in muscle. Skeletal Muscle, 2018, 8, 28. | 1.9 | 19 |
| 18 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | GFPT1 deficiency in muscle leads to myasthenia and myopathy in mice. Human Molecular Genetics, 2018, 27, 3218-3232. | 1.4 | 18 |
| 20 | 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 |
| 21 | 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 |
| 22 | Proteomic Profiling Unravels a Key Role of Specific Macrophage Subtypes in Sporadic Inclusion Body Myositis. Frontiers in Immunology, 2019, 10, 1040. | 2.2 | 17 |
| 23 | 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 |
| 24 | Multispectral optoacoustic tomography for non-invasive disease phenotyping in pediatric spinal muscular atrophy patients. Photoacoustics, 2022, 25, 100315. | 4.4 | 16 |
| 25 | 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 |
| 26 | 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 |
| 27 | Molecular pathophysiology of human MICU1 deficiency. Neuropathology and Applied Neurobiology, 2021, 47, 840-855. | 1.8 | 15 |
| 28 | MYO9A deficiency in motor neurons is associated with reduced neuromuscular agrin secretion. Human Molecular Genetics, 2018, 27, 1434-1446. | 1.4 | 14 |
| 29 | 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 |
| 30 | 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 |
| 31 | High diagnostic rate of trio exome sequencing in consanguineous families with neurogenetic diseases. Brain, 2022, 145, 1507-1518. | 3.7 | 14 |
| 32 | Muscle Pathology as a Diagnostic Clue to Allgrove Syndrome. Journal of Neuropathology and Experimental Neurology, 2017, 76, 337-341. | 0.9 | 13 |
| 33 | 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 |
| 34 | 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 |
| 35 | Diagnosing X-linked Myotubular Myopathy â€“ A German 20-year Follow Up Experience. Journal of Neuromuscular Diseases, 2021, 8, 79-90. | 1.1 | 12 |
| 36 | NCAM1 and GDF15 are biomarkers of Charcot-Marie-Tooth disease in patients and mice. Brain, 2022, 145, 3999-4015. | 3.7 | 12 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Modulation of Agrin and RhoA Pathways Ameliorates Movement Defects and Synapse Morphology in MYO9A-Depleted Zebrafish. Cells, 2019, 8, 848. | 1.8 | 10 |
| 38 | 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 |
| 39 | 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 |
| 40 | 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 |
| 41 | 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 |
| 42 | INPP5K and SIL1 associated pathologies with overlapping clinical phenotypes converge through dysregulation of PHGDH. Brain, 2021, 144, 2427-2442. | 3.7 | 7 |
| 43 | Clinical Course, Myopathology and Challenge of Therapeutic Intervention in Pediatric Patients with Autoimmune-Mediated Necrotizing Myopathy. Children, 2021, 8, 721. | 0.6 | 7 |
| 44 | Three Individuals with PURA Syndrome in a Cohort of Patients with Neuromuscular Disease. Neuropediatrics, 2021, 52, 390-393. | 0.3 | 7 |
| 45 | Endoplasmic reticulumâ€stress and unfolded protein responseâ€activation in immuneâ€mediated necrotizing myopathy. Brain Pathology, 2022, 32, . | 2.1 | 7 |
| 46 | 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 |
| 47 | Severe neurodevelopmental disease caused by a homozygous TLK2 variant. European Journal of Human Genetics, 2020, 28, 383-387. | 1.4 | 6 |
| 48 | Autosomal recessive variants in TUBGCP2 alter the Î³-tubulin ring complex leading to neurodevelopmental disease. IScience, 2021, 24, 101948. | 1.9 | 6 |
| 49 | Identification of Cellular Pathogenicity Markers for SIL1 Mutations Linked to Marinesco-SjÃ¶gren Syndrome. Frontiers in Neurology, 2019, 10, 562. | 1.1 | 5 |
| 50 | Immunofluorescence-Based Analysis of Caveolin-3 in the Diagnostic Management of Neuromuscular Diseases. Methods in Molecular Biology, 2020, 2169, 197-216. | 0.4 | 5 |
| 51 | 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 |
| 52 | 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 |
| 53 | Serum miRNAs as biomarkers for the rare types of muscular dystrophy. Neuromuscular Disorders, 2022, 32, 332-346. | 0.3 | 5 |
| 54 | Intragenic DOK7 deletion detected by whole-genome sequencing in congenital myasthenic syndromes. Neurology: Genetics, 2017, 3, e152. | 0.9 | 4 |

| # | Article | IF | Citations |
|---|---|---|---|
| 55 | Multifocal demyelinating motor neuropathy and hamartoma syndrome associated with a de novo PTEN mutation. Neurology, 2018, 90, e1842-e1848. | 1.5 | 4 |
| 56 | Further evidence for POMK as candidate gene for WWS with meningoencephalocele. Orphanet Journal of Rare Diseases, 2020, 15, 242. | 1.2 | 4 |
| 57 | Muscular and Molecular Pathology Associated with SPATA5 Deficiency in a Child with EHLMRS. International Journal of Molecular Sciences, 2021, 22, 7835. | 1.8 | 4 |
| 58 | Novel insights into PORCN mutations, associated phenotypes and pathophysiological aspects. Orphanet Journal of Rare Diseases, 2022, 17, 29. | 1.2 | 3 |
| 59 | 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 |
| 60 | 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 |
| 61 | SIL1-negative Marinesco-SjÃ¶gren syndrome: First report of two sibs from India. Journal of Pediatric Neurosciences, 2014, 9, 291. | 0.2 | 2 |
| 62 | New Insights into the Neuromyogenic Spectrum of a Gain of Function Mutation in SPTLC1. Genes, 2022, 13, 893. | 1.0 | 2 |
| 63 | 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 |
| 64 | Phenotypical and Myopathological Consequences of Compound Heterozygous Missense and Nonsense Variants in SLC18A3. Cells, 2021, 10, 3481. | 1.8 | 1 |
| 65 | 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 |
| 66 | Noninvasive Imaging in Pediatric Spinal Muscular Atrophy Patients Using Multispectral Optoacoustic Tomography: A Proof-of-Concept Study. Neuropediatrics, 2021, 52, . | 0.3 | 0 |