

Anders Oldfors

List of Publications by Year in descending order

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Version: 2024-02-01

110
papers

8,102
citations

147566

31
h-index

49773

87
g-index

144
all docs

144
docs citations

144
times ranked

9612
citing authors

#	ARTICLE	IF	CITATIONS
1	Proteomic characterisation of polyglucosan bodies in skeletal muscle in RBCK1 deficiency. <i>Neuropathology and Applied Neurobiology</i> , 2022, 48, .	1.8	13
2	Somatostatin Receptor Positron Emission Tomography/Computed Tomography in Giant Cell Myocarditis: A Promising Approach to Molecular Myocardial Inflammation Imaging. <i>Circulation: Cardiovascular Imaging</i> , 2022, 15, CIRCIMAGING121013551.	1.3	5
3	Cardiac involvement in immune-mediated necrotizing myopathy: insights from CMR and somatostatin receptor PET/CT. <i>European Heart Journal Cardiovascular Imaging</i> , 2022, 23, e237-e237.	0.5	2
4	Diagnosis, management, and outcome of cardiac sarcoidosis and giant cell myocarditis: a Swedish single center experience. <i>BMC Cardiovascular Disorders</i> , 2022, 22, 192.	0.7	16
5	Epidemiology, Survival, and Clinical Characteristics of Inclusion Body Myositis. <i>Annals of Neurology</i> , 2022, 92, 201-212.	2.8	10
6	Mitochondrial DNA variants in inclusion body myositis characterized by deep sequencing. <i>Brain Pathology</i> , 2021, 31, e12931.	2.1	17
7	Progressive external ophthalmoplegia associated with novel <i>MT</i> and <i>TN</i> mutations. <i>Acta Neurologica Scandinavica</i> , 2021, 143, 103-108.	1.0	11
8	251st ENMC international workshop: Polyglucosan storage myopathies 13-15 December 2019, Hoofddorp, the Netherlands. <i>Neuromuscular Disorders</i> , 2021, 31, 466-477.	0.3	4
9	The phenotypic variability and natural history of NARS2 associated disease. <i>European Journal of Paediatric Neurology</i> , 2021, 31, 31-37.	0.7	14
10	Functional analysis of a novel POLÎ3A mutation associated with a severe perinatal mitochondrial encephalomyopathy. <i>Neuromuscular Disorders</i> , 2021, 31, 348-358.	0.3	2
11	The localization of amyloid precursor protein to ependymal cilia in vertebrates and its role in ciliogenesis and brain development in zebrafish. <i>Scientific Reports</i> , 2021, 11, 19115.	1.6	4
12	Neuromuscular Disorders Special Issue 2021 Marking the 90th birthday of Victor Dubowitz, Founding Editor-in-Chief. <i>Neuromuscular Disorders</i> , 2021, 31, 919-920.	0.3	0
13	Expression pattern of mitochondrial respiratory chain enzymes in skeletal muscle of patients with mitochondrial myopathy associated with the homoplasmic m.14674T>C variant. <i>Brain Pathology</i> , 2021, , e13038.	2.1	4
14	Glycogenin is Dispensable for Glycogen Synthesis in Human Muscle, and Glycogenin Deficiency Causes Polyglucosan Storage. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, 557-566.	1.8	17
15	Pathogenic Variants in the Myosin Chaperone UNC-45B Cause Progressive Myopathy with Eccentric Cores. <i>American Journal of Human Genetics</i> , 2020, 107, 1078-1095.	2.6	24
16	Loss of supervillin causes myopathy with myofibrillar disorganization and autophagic vacuoles. <i>Brain</i> , 2020, 143, 2406-2420.	3.7	15
17	<i>RBCK1</i> -related disease: A rare multisystem disorder with polyglucosan storage, autoinflammation, recurrent infections, skeletal, and cardiac myopathy—Four additional patients and a review of the current literature. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 1002-1013.	1.7	23
18	COX deficiency and leukoencephalopathy due to a novel homozygous APOPT1/COA8 mutation. <i>Neurology: Genetics</i> , 2020, 6, e464.	0.9	9

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19	Deep sequencing of mitochondrial DNA and characterization of a novel POLG mutation in a patient with arPEO. <i>Neurology: Genetics</i> , 2020, 6, e391.	0.9	8
20	Accurate mapping of mitochondrial DNA deletions and duplications using deep sequencing. <i>PLoS Genetics</i> , 2020, 16, e1009242.	1.5	41
21	Accurate mapping of mitochondrial DNA deletions and duplications using deep sequencing. , 2020, 16, e1009242.		0
22	Accurate mapping of mitochondrial DNA deletions and duplications using deep sequencing. , 2020, 16, e1009242.		0
23	Accurate mapping of mitochondrial DNA deletions and duplications using deep sequencing. , 2020, 16, e1009242.		0
24	Accurate mapping of mitochondrial DNA deletions and duplications using deep sequencing. , 2020, 16, e1009242.		0
25	Parvovirus B19 in Endomyocardial Biopsy of Patients With Idiopathic Dilated Cardiomyopathy: Foe or Bystander?. <i>Journal of Cardiac Failure</i> , 2019, 25, 60-63.	0.7	21
26	Danon disease presenting with early onset of hypertrophic cardiomyopathy and peripheral pigmentary retinal dystrophy in a female with a <i>de novo</i> novel mosaic mutation in the <i>LAMP2</i> gene. <i>Ophthalmic Genetics</i> , 2019, 40, 227-236.	0.5	5
27	<i>TANGO2</i> deficiency as a cause of neurodevelopmental delay with indirect effects on mitochondrial energy metabolism. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 898-908.	1.7	32
28	Deep morphological analysis of muscle biopsies from type III glycogenesis (GSDIII), debranching enzyme deficiency, revealed stereotyped vacuolar myopathy and autophagy impairment. <i>Acta Neuropathologica Communications</i> , 2019, 7, 167.	2.4	17
29	Functional characterization of GYG1 variants in two patients with myopathy and glycogenin-1 deficiency. <i>Neuromuscular Disorders</i> , 2019, 29, 951-960.	0.3	8
30	The Atrioventricular Junction: A Potential Niche Region for Progenitor Cells in the Adult Human Heart. <i>Stem Cells and Development</i> , 2019, 28, 1078-1088.	1.1	11
31	1st ENMC European meeting: The EURO-NMD pathology working group Recommended Standards for Muscle Pathology Amsterdam, The Netherlands, 7 December 2018. <i>Neuromuscular Disorders</i> , 2019, 29, 483-485.	0.3	27
32	Cardiomyopathy with lethal arrhythmias associated with inactivation of KLHL24. <i>Human Molecular Genetics</i> , 2019, 28, 1919-1929.	1.4	35
33	An AARS variant as the likely cause of Swedish type hereditary diffuse leukoencephalopathy with spheroids. <i>Acta Neuropathologica Communications</i> , 2019, 7, 188.	2.4	19
34	Long-term follow-up and characteristic pathological findings in severe nemaline myopathy due to LMOD3 mutations. <i>Neuromuscular Disorders</i> , 2019, 29, 108-113.	0.3	14
35	Cardiac arrest in Wilson's disease after curative liver transplantation: a life-threatening complication of myocardial copper excess?. <i>ESC Heart Failure</i> , 2019, 6, 228-231.	1.4	10
36	Association between muscle strength, histopathology, and magnetic resonance imaging in sporadic inclusion body myositis. <i>Acta Neurologica Scandinavica</i> , 2019, 139, 177-182.	1.0	7

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37	Mitochondrial complex IV deficiency caused by a novel frameshift variant in MT-CO2 associated with myopathy and perturbed acylcarnitine profile. <i>European Journal of Human Genetics</i> , 2019, 27, 331-335.	1.4	17
38	Oxygen consumption in platelets as an adjunct diagnostic method for pediatric mitochondrial disease. <i>Pediatric Research</i> , 2018, 83, 455-465.	1.1	4
39	Tubular aggregates in congenital myasthenic syndrome. <i>Neuromuscular Disorders</i> , 2018, 28, 174-175.	0.3	1
40	Loss of Sarcomeric Scaffolding as a Common Baseline Histopathologic Lesion in Titin-Related Myopathies. <i>Journal of Neuropathology and Experimental Neurology</i> , 2018, 77, 1101-1114.	0.9	22
41	Some DNM2 mutations cause extremely severe congenital myopathy and phenocopy myotubular myopathy. <i>Acta Neuropathologica Communications</i> , 2018, 6, 93.	2.4	14
42	A novel complex neurological phenotype due to a homozygous mutation in FDX2. <i>Brain</i> , 2018, 141, 2289-2298.	3.7	29
43	Hypoxic cardiac fibroblasts from failing human hearts decrease cardiomyocyte beating frequency in an ALOX15 dependent manner. <i>PLoS ONE</i> , 2018, 13, e0202693.	1.1	16
44	Carey-Fineman-Ziter syndrome with mutations in the myomaker gene and muscle fiber hypertrophy. <i>Neurology: Genetics</i> , 2018, 4, e254.	0.9	18
45	Muscle pathology in Vici syndrome – A case study with a novel mutation in EPG5 and a summary of the literature. <i>Neuromuscular Disorders</i> , 2017, 27, 771-776.	0.3	21
46	Grand paternal inheritance of X-linked myotubular myopathy due to mosaicism, and identification of necklace fibers in an asymptomatic male. <i>Neuromuscular Disorders</i> , 2017, 27, 843-847.	0.3	6
47	Is Glycogenin Essential for Glycogen Synthesis?. <i>Cell Metabolism</i> , 2017, 26, 12-14.	7.2	9
48	Cardiomyopathy as presenting sign of glycogenin1 deficiency – report of three cases and review of the literature. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 139-149.	1.7	32
49	Leukoencephalopathy due to Complex II Deficiency and Bi-Allelic SDHB Mutations: Further Cases and Implications for Genetic Counselling. <i>JIMD Reports</i> , 2016, 33, 69-77.	0.7	17
50	Start codon mutation of GYG1 causing late-onset polyglucosan body myopathy with nemaline rods. <i>Journal of Neurology</i> , 2016, 263, 2133-2135.	1.8	17
51	A new early-onset neuromuscular disorder associated with kyphoscoliosis peptidase (KY) deficiency. <i>European Journal of Human Genetics</i> , 2016, 24, 1771-1777.	1.4	17
52	Histopathological changes in skeletal muscle associated with chronic ischaemia. <i>Apms</i> , 2016, 124, 935-941.	0.9	9
53	A novel neuromuscular form of glycogen storage disease type IV with arthrogryposis, spinal stiffness and rare polyglucosan bodies in muscle. <i>Neuromuscular Disorders</i> , 2016, 26, 681-687.	0.3	9
54	Novel myopathy in a newborn with Shwachman – Diamond syndrome and review of neonatal presentation. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1155-1164.	0.7	9

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55	A novel MYH2 mutation in family members presenting with congenital myopathy, ophthalmoplegia and facial weakness. <i>Journal of Neurology</i> , 2016, 263, 1427-1433.	1.8	14
56	Broad phenotypic variability in patients with complex I deficiency due to mutations in NDUFS1 and NDUFV1. <i>Mitochondrion</i> , 2015, 21, 33-40.	1.6	30
57	Glycogen pathways in disease: new developments in a classical field of medical genetics. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 483-487.	1.7	20
58	Early onset cardiomyopathy in females with Danon disease. <i>Neuromuscular Disorders</i> , 2015, 25, 493-501.	0.3	32
59	Mitochondrial pathology in inclusion body myositis. <i>Neuromuscular Disorders</i> , 2015, 25, 281-288.	0.3	31
60	A novel dynamin-2 gene mutation associated with a late-onset centronuclear myopathy with necklace fibres. <i>Neuromuscular Disorders</i> , 2015, 25, 345-348.	0.3	15
61	New cardiac and skeletal protein aggregate myopathy associated with combined MuRF1 and MuRF3 mutations. <i>Human Molecular Genetics</i> , 2015, 24, 3638-3650.	1.4	51
62	Muscle pathology and whole-body MRI in a polyglucosan myopathy associated with a novel glycogenin-1 mutation. <i>Neuromuscular Disorders</i> , 2015, 25, 780-785.	0.3	28
63	Whole exome sequencing reveals mutations in <i>NARS2</i> and <i>PARS2</i> , encoding the mitochondrial asparaginyl-tRNA synthetase and prolyl-tRNA synthetase, in patients with Alpers syndrome. <i>Molecular Genetics & Genomic Medicine</i> , 2015, 3, 59-68.	0.6	87
64	Polyglucosan storage myopathies. <i>Molecular Aspects of Medicine</i> , 2015, 46, 85-100.	2.7	63
65	Hereditary myopathy with early respiratory failure is associated with misfolding of the titin fibronectin III 119 subdomain. <i>Neuromuscular Disorders</i> , 2014, 24, 373-379.	0.3	17
66	LC-MS/MS characterization of combined glycogenin-1 and glycogenin-2 enzymatic activities reveals their self-glucosylation preferences. <i>Biochimica Et Biophysica Acta - Proteins and Proteomics</i> , 2014, 1844, 398-405.	1.1	11
67	B3GALNT2 is a gene associated with congenital muscular dystrophy with brain malformations. <i>European Journal of Human Genetics</i> , 2014, 22, 707-710.	1.4	27
68	Recessive myosin myopathy with external ophthalmoplegia associated with MYH2 mutations. <i>European Journal of Human Genetics</i> , 2014, 22, 801-808.	1.4	33
69	A new muscle glycogen storage disease associated with glycogenin-1 deficiency. <i>Annals of Neurology</i> , 2014, 76, 891-898.	2.8	72
70	New insights in the field of muscle glycogenoses. <i>Current Opinion in Neurology</i> , 2013, 26, 544-553.	1.8	44
71	Polyglucosan body myopathy caused by defective ubiquitin ligase RBCK1. <i>Annals of Neurology</i> , 2013, 74, 914-919.	2.8	132
72	Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. <i>Journal of Neurology</i> , 2012, 259, 838-850.	1.8	72

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73	Phenotypic and genotypic variability in Alpers syndrome. <i>European Journal of Paediatric Neurology</i> , 2012, 16, 379-389.	0.7	22
74	Glycogenin-1 Deficiency and Inactivated Priming of Glycogen Synthesis. <i>New England Journal of Medicine</i> , 2010, 362, 1203-1210.	13.9	95
75	Changes in specific gravity as a sign of disturbed brain maturation in protein-deprived rats. <i>Acta Neurologica Scandinavica</i> , 2009, 73, 76-77.	1.0	1
76	Thick Filament Diseases. <i>Advances in Experimental Medicine and Biology</i> , 2008, 642, 78-91.	0.8	21
77	Tropomyosin mutations responsible for muscle weakness in inherited skeletal muscle diseases. , 2008, , 20-21.		0
78	Hereditary myosin myopathies. <i>Neuromuscular Disorders</i> , 2007, 17, 355-367.	0.3	117
79	Cardiomyopathy and Exercise Intolerance in Muscle Glycogen Storage Disease 0. <i>New England Journal of Medicine</i> , 2007, 357, 1507-1514.	13.9	123
80	Mitochondrial (Mt)Dna Changes in Tissue May Not be Reflected by Depletion of Mtdna in Peripheral Blood Mononuclear Cells in HIV-Infected Patients. <i>Antiviral Therapy</i> , 2006, 11, 601-608.	0.6	48
81	Diagnosis, pathogenesis and treatment of inclusion body myositis. <i>Current Opinion in Neurology</i> , 2005, 18, 497-503.	1.8	15
82	Premature ageing in mice expressing defective mitochondrial DNA polymerase. <i>Nature</i> , 2004, 429, 417-423.	13.7	2,318
83	Mitochondrial Encephalomyopathies. <i>Journal of Neuropathology and Experimental Neurology</i> , 2003, 62, 217-227.	0.9	38
84	Ageing muscle: clonal expansions of mitochondrial DNA point mutations and deletions cause focal impairment of mitochondrial function. <i>Neuromuscular Disorders</i> , 2002, 12, 484-493.	0.3	180
85	The incidence of mitochondrial encephalomyopathies in childhood: Clinical features and morphological, biochemical, and DNA abnormalities. <i>Annals of Neurology</i> , 2001, 49, 377-383.	2.8	311
86	The incidence of mitochondrial encephalomyopathies in childhood: Clinical features and morphological, biochemical, and DNA abnormalities. <i>Annals of Neurology</i> , 2001, 49, 377-383.	2.8	7
87	Impaired insulin secretion and β -cell loss in tissue-specific knockout mice with mitochondrial diabetes. <i>Nature Genetics</i> , 2000, 26, 336-340.	9.4	417
88	Dilated cardiomyopathy and atrioventricular conduction blocks induced by heart-specific inactivation of mitochondrial DNA gene expression. <i>Nature Genetics</i> , 1999, 21, 133-137.	9.4	393
89	A novel heteroplasmic point mutation in the mitochondrial tRNA ^{Lys} gene in a sporadic case of mitochondrial encephalomyopathy: De novo mutation and no transmission to the offspring. , 1999, 13, 203-209.		21
90	Mitochondrial transcription factor A is necessary for mtDNA maintenance and embryogenesis in mice. <i>Nature Genetics</i> , 1998, 18, 231-236.	9.4	1,377

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91	Upregulation of Fas/Fas ligand in inclusion body myositis. <i>Annals of Neurology</i> , 1998, 43, 127-130.	2.8	25
92	Down-Regulation of Mitochondrial Transcription Factor a During Spermatogenesis in Humans. <i>Human Molecular Genetics</i> , 1997, 6, 185-1991.	1.4	75
93	Analysis of multiple mitochondrial DNA deletions in inclusion body myositis. , 1997, 10, 381-386.		53
94	Fatal mitochondrial myopathy, lactic acidosis, and complex I deficiency associated with a heteroplasmic Aâ†’G mutation at position 3251 in the mitochondrial tRNA ^{Leu} (UUR) gene. <i>Human Genetics</i> , 1996, 97, 269-273.	1.8	20
95	A single mouse gene encodes the mitochondrial transcription factor A and a testisâ€“specific nuclear HMG-box protein. <i>Nature Genetics</i> , 1996, 13, 296-302.	9.4	145
96	Clonal expansion of mitochondrial DNA with multiple deletions in autosomal dominant progressive external ophthalmoplegia. <i>Annals of Neurology</i> , 1996, 40, 707-713.	2.8	96
97	Anticipation of autosomal dominant progressive external ophthalmoplegia with hypogonadism. , 1996, 19, 1561-1569.		27
98	Mitochondrial DNA Deletions in Muscle Fibers in Inclusion Body Myositis. <i>Journal of Neuropathology and Experimental Neurology</i> , 1995, 54, 581-587.	0.9	103
99	Pathogenetic aspects of the A8344G mutation of mitochondrial DNA associated with MERRF syndrome and multiple symmetric lipomas. <i>Muscle and Nerve</i> , 1995, 18, S102-S106.	1.0	59
100	De novo mutation in the mitochondrial ATP synthase subunit 6 gene (T8993G) with rapid segregation resulting in Leigh syndrome in the offspring. <i>Human Genetics</i> , 1995, 96, 290-4.	1.8	35
101	Tissue distribution and disease manifestations of the tRNA ^{Lys} A?G(8344) mitochondrial DNA mutation in a case of myoclonus epilepsy and ragged red fibres. <i>Acta Neuropathologica</i> , 1995, 90, 328-333.	3.9	4
102	Restricted use of T cell receptor V genes in endomysial infiltrates of patients with inflammatory myopathies. <i>European Journal of Immunology</i> , 1994, 24, 2659-2663.	1.6	56
103	Duchenne muscular dystrophy and spinal muscular atrophy type I segregating in the same family. <i>Clinical Genetics</i> , 1994, 45, 97-103.	1.0	2
104	Mitochondrial ATP-Synthase Deficiency in a Child with 3-Methylglutaconic Aciduria. <i>Pediatric Research</i> , 1992, 32, 731-736.	1.1	65
105	Mitochondrial encephalomyopathies in childhood. I. Biochemical and morphologic investigations. <i>Journal of Pediatrics</i> , 1991, 119, 242-250.	0.9	128
106	Leber's hereditary optic neuropathy and complex I deficiency in muscle. <i>Annals of Neurology</i> , 1991, 30, 701-708.	2.8	155
107	Xâ€“linked myotubular myopathy: a linkage study. <i>Clinical Genetics</i> , 1990, 37, 335-340.	1.0	34
108	THE EFFECT OF Î²â€“ADRENERGIC BLOCKADE ON THE MYOPATHIC CHANGES IN EXPERIMENTAL HYPERTHYROIDISM IN RATS. <i>Acta Pathologica, Microbiologica, Et Immunologica Scandinavica Section A, Pathology</i> , 1986, 94A, 91-99.	0.3	5

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109	The effect of undernutrition on spinal ganglion nerve cell size. Acta Neuropathologica, 1977, 39, 75-80.	3.9	4
110	Paranodal myelin retraction in protein deficient and normal rats. Acta Neuropathologica, 1977, 40, 249-252.	3.9	5