Anders Oldfors

List of Publications by Year
in descending order

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110 papers	8,102 citations	136950 32 h-index	48315 88 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. Neuropathology and Applied Neurobiology, 2022, 48, .	3.2	13
2	Somatostatin Receptor Positron Emission Tomography/Computed Tomography in Giant Cell Myocarditis: A Promising Approach to Molecular Myocardial Inflammation Imaging. Circulation: Cardiovascular Imaging, 2022, 15, CIRCIMAGING121013551.	2.6	5
3	Cardiac involvement in immune-mediated necrotizing myopathy: insights from CMR and somatostatin receptor PET/CT. European Heart Journal Cardiovascular Imaging, 2022, 23, e237-e237.	1.2	2
4	Diagnosis, management, and outcome of cardiac sarcoidosis and giant cell myocarditis: a Swedish single center experience. BMC Cardiovascular Disorders, 2022, 22, 192.	1.7	16
5	Epidemiology, Survival, and Clinical Characteristics of Inclusion Body Myositis. Annals of Neurology, 2022, 92, 201-212.	5.3	10
6	Mitochondrial DNA variants in inclusion body myositis characterized by deep sequencing. Brain Pathology, 2021, 31, e12931.	4.1	17
7	Progressive external ophthalmoplegia associated with novel MT â€•TN mutations. Acta Neurologica Scandinavica, 2021, 143, 103-108.	2.1	11
8	251st ENMC international workshop: Polyglucosan storage myopathies 13â€“15 December 2019, Hoofddorp, the Netherlands. Neuromuscular Disorders, 2021, 31, 466-477.	0.6	4
9	The phenotypic variability and natural history of NARS2 associated disease. European Journal of Paediatric Neurology, 2021, 31, 31-37.	1.6	14
10	Functional analysis of a novel POLÎ³A mutation associated with a severe perinatal mitochondrial encephalomyopathy. Neuromuscular Disorders, 2021, 31, 348-358.	0.6	2
11	The localization of amyloid precursor protein to ependymal cilia in vertebrates and its role in ciliogenesis and brain development in zebrafish. Scientific Reports, 2021, 11, 19115.	3.3	4
12	Neuromuscular Disorders Special Issue 2021 Marking the 90th birthday of Victor Dubowitz, Founding Editor-in-Chief. Neuromuscular Disorders, 2021, 31, 919-920.	0.6	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. Brain Pathology, 2021, , e13038.	4.1	4
14	Glycogenin is Dispensable for Glycogen Synthesis in Human Muscle, and Glycogenin Deficiency Causes Polyglucosan Storage. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 557-566.	3.6	17
15	Pathogenic Variants in the Myosin Chaperone UNC-45B Cause Progressive Myopathy with Eccentric Cores. American Journal of Human Genetics, 2020, 107, 1078-1095.	6.2	24
16	Loss of supervillin causes myopathy with myofibrillar disorganization and autophagic vacuoles. Brain, 2020, 143, 2406-2420.	7.6	15
17	<scp>RBCK1</scp>â€related disease: A rare multisystem disorder with polyglucosan storage, autoâ€inflammation, recurrent infections, skeletal, and cardiac myopathyâ€”Four additional patients and a review of the current literature. Journal of Inherited Metabolic Disease, 2020, 43, 1002-1013.	3.6	23
18	COX deficiency and leukoencephalopathy due to a novel homozygous APOPT1/COA8 mutation. Neurology: Genetics, 2020, 6, e464.	1.9	9

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19	Deep sequencing of mitochondrial DNA and characterization of a novel POLG mutation in a patient with arPEO. Neurology: Genetics, 2020, 6, e391.	1.9	8
20	Accurate mapping of mitochondrial DNA deletions and duplications using deep sequencing. PLoS Genetics, 2020, 16, e1009242.	3.5	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?. Journal of Cardiac Failure, 2019, 25, 60-63.	1.7	21
26	Danon disease presenting with early onset of hypertrophic cardiomyopathy and peripheral pigmentary retinal dystrophy in a female with a de novo novel mosaic mutation in the LAMP2 gene. Ophthalmic Genetics, 2019, 40, 227-236.	1.2	5
27	TANGO2 deficiency as a cause of neurodevelopmental delay with indirect effects on mitochondrial energy metabolism. Journal of Inherited Metabolic Disease, 2019, 42, 898-908.	3.6	32
28	Deep morphological analysis of muscle biopsies from type III glycogenesis (GSDIII), debranching enzyme deficiency, revealed stereotyped vacuolar myopathy and autophagy impairment. Acta Neuropathologica Communications, 2019, 7, 167.	5.2	17
29	Functional characterization of GYG1 variants in two patients with myopathy and glycogenin-1 deficiency. Neuromuscular Disorders, 2019, 29, 951-960.	0.6	8
30	The Atrioventricular Junction: A Potential Niche Region for Progenitor Cells in the Adult Human Heart. Stem Cells and Development, 2019, 28, 1078-1088.	2.1	11
31	1st ENMC European meeting: The EURO-NMD pathology working group Recommended Standards for Muscle Pathology Amsterdam, The Netherlands, 7 December 2018. Neuromuscular Disorders, 2019, 29, 483-485.	0.6	27
32	Cardiomyopathy with lethal arrhythmias associated with inactivation of KLHL24. Human Molecular Genetics, 2019, 28, 1919-1929.	2.9	35
33	An AARS variant as the likely cause of Swedish type hereditary diffuse leukoencephalopathy with spheroids. Acta Neuropathologica Communications, 2019, 7, 188.	5.2	19
34	Long-term follow-up and characteristic pathological findings in severe nemaline myopathy due to LMOD3 mutations. Neuromuscular Disorders, 2019, 29, 108-113.	0.6	14
35	Cardiac arrest in Wilson's disease after curative liver transplantation: a lifeâ€threatening complication of myocardial copper excess?. ESC Heart Failure, 2019, 6, 228-231.	3.1	10
36	Association between muscle strength, histopathology, and magnetic resonance imaging in sporadic inclusion body myositis. Acta Neurologica Scandinavica, 2019, 139, 177-182.	2.1	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. European Journal of Human Genetics, 2019, 27, 331-335.	2.8	17
38	Oxygen consumption in platelets as an adjunct diagnostic method for pediatric mitochondrial disease. Pediatric Research, 2018, 83, 455-465.	2.3	4
39	Tubular aggregates in congenital myasthenic syndrome. Neuromuscular Disorders, 2018, 28, 174-175.	0.6	1
40	Loss of Sarcomeric Scaffolding as a Common Baseline Histopathologic Lesion in Titin-Related Myopathies. Journal of Neuropathology and Experimental Neurology, 2018, 77, 1101-1114.	1.7	22
41	Some DNM2 mutations cause extremely severe congenital myopathy and phenocopy myotubular myopathy. Acta Neuropathologica Communications, 2018, 6, 93.	5.2	14
42	A novel complex neurological phenotype due to a homozygous mutation in FDX2. Brain, 2018, 141, 2289-2298.	7.6	29
43	Hypoxic cardiac fibroblasts from failing human hearts decrease cardiomyocyte beating frequency in an ALOX15 dependent manner. PLoS ONE, 2018, 13, e0202693.	2.5	16
44	Carey-Fineman-Ziter syndrome with mutations in the myomaker gene and muscle fiber hypertrophy. Neurology: Genetics, 2018, 4, e254.	1.9	18
45	Muscle pathology in Vici syndromeâ€“A case study with a novel mutation in EPG5 and a summary of the literature. Neuromuscular Disorders, 2017, 27, 771-776.	0.6	21
46	Grand paternal inheritance of X-linked myotubular myopathy due to mosaicism, and identification of necklace fibers in an asymptomatic male. Neuromuscular Disorders, 2017, 27, 843-847.	0.6	6
47	Is Glycogenin Essential for Glycogen Synthesis?. Cell Metabolism, 2017, 26, 12-14.	16.2	9
48	Cardiomyopathy as presenting sign of glycogeninâ€1 deficiencyâ€”report of three cases and review of the literature. Journal of Inherited Metabolic Disease, 2017, 40, 139-149.	3.6	32
49	Leukoencephalopathy due to Complex II Deficiency and Bi-Allelic SDHB Mutations: Further Cases and Implications for Genetic Counselling. JIMD Reports, 2016, 33, 69-77.	1.5	17
50	Start codon mutation of GYG1 causing late-onset polyglucosan body myopathy with nemaline rods. Journal of Neurology, 2016, 263, 2133-2135.	3.6	17
51	A new early-onset neuromuscular disorder associated with kyphoscoliosis peptidase (KY) deficiency. European Journal of Human Genetics, 2016, 24, 1771-1777.	2.8	17
52	Histopathological changes in skeletal muscle associated with chronic ischaemia. Apmis, 2016, 124, 935-941.	2.0	9
53	A novel neuromuscular form of glycogen storage disease type IV with arthrogryposis, spinal stiffness and rare polyglucosan bodies in muscle. Neuromuscular Disorders, 2016, 26, 681-687.	0.6	9
54	Novel myopathy in a newborn with Shwachmanâ€“Diamond syndrome and review of neonatal presentation. American Journal of Medical Genetics, Part A, 2016, 170, 1155-1164.	1.2	9

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55	A novel MYH2 mutation in family members presenting with congenital myopathy, ophthalmoplegia and facial weakness. Journal of Neurology, 2016, 263, 1427-1433.	3.6	14
56	Broad phenotypic variability in patients with complex I deficiency due to mutations in NDUFS1 and NDUFV1. Mitochondrion, 2015, 21, 33-40.	3.4	30
57	Glycogen pathways in disease: new developments in a classical field of medical genetics. Journal of Inherited Metabolic Disease, 2015, 38, 483-487.	3.6	20
58	Early onset cardiomyopathy in females with Danon disease. Neuromuscular Disorders, 2015, 25, 493-501.	0.6	32
59	Mitochondrial pathology in inclusion body myositis. Neuromuscular Disorders, 2015, 25, 281-288.	0.6	31
60	A novel dynamin-2 gene mutation associated with a late-onset centronuclear myopathy with necklace fibres. Neuromuscular Disorders, 2015, 25, 345-348.	0.6	15
61	New cardiac and skeletal protein aggregate myopathy associated with combined MuRF1 and MuRF3 mutations. Human Molecular Genetics, 2015, 24, 3638-3650.	2.9	51
62	Muscle pathology and whole-body MRI in a polyglucosan myopathy associated with a novel glycogenin-1 mutation. Neuromuscular Disorders, 2015, 25, 780-785.	0.6	28
63	Whole exome sequencing reveals mutations inNARS2andPARS2, encoding the mitochondrial asparaginyl-tRNA synthetase and prolyl-tRNA synthetase, in patients with Alpers syndrome. Molecular Genetics & Genomic Medicine, 2015, 3, 59-68.	1.2	87
64	Polyglucosan storage myopathies. Molecular Aspects of Medicine, 2015, 46, 85-100.	6.4	63
65	Hereditary myopathy with early respiratory failure is associated with misfolding of the titin fibronectin III 119 subdomain. Neuromuscular Disorders, 2014, 24, 373-379.	0.6	17
66	LCâ€“MS/MS characterization of combined glycogenin-1 and glycogenin-2 enzymatic activities reveals their self-glucosylation preferences. Biochimica Et Biophysica Acta - Proteins and Proteomics, 2014, 1844, 398-405.	2.3	11
67	B3GALNT2 is a gene associated with congenital muscular dystrophy with brain malformations. European Journal of Human Genetics, 2014, 22, 707-710.	2.8	27
68	Recessive myosin myopathy with external ophthalmoplegia associated with MYH2 mutations. European Journal of Human Genetics, 2014, 22, 801-808.	2.8	33
69	A new muscle glycogen storage disease associated with glycogeninâ€1 deficiency. Annals of Neurology, 2014, 76, 891-898.	5.3	72
70	New insights in the field of muscle glycogenoses. Current Opinion in Neurology, 2013, 26, 544-553.	3.6	44
71	Polyglucosan body myopathy caused by defective ubiquitin ligase RBCK1. Annals of Neurology, 2013, 74, 914-919.	5.3	132
72	Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. Journal of Neurology, 2012, 259, 838-850.	3.6	72

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73	Phenotypic and genotypic variability in Alpers syndrome. European Journal of Paediatric Neurology, 2012, 16, 379-389.	1.6	22
74	Glycogenin-1 Deficiency and Inactivated Priming of Glycogen Synthesis. New England Journal of Medicine, 2010, 362, 1203-1210.	27.0	95
75	Changes in specific gravity as a sign of disturbed brain maturation in protein-deprived rats. Acta Neurologica Scandinavica, 2009, 73, 76-77.	2.1	1
76	Thick Filament Diseases. Advances in Experimental Medicine and Biology, 2008, 642, 78-91.	1.6	21
77	Tropomyosin mutations responsible for muscle weakness in inherited skeletal muscle diseases. , 2008, , 20-21.		0
78	Hereditary myosin myopathies. Neuromuscular Disorders, 2007, 17, 355-367.	0.6	117
79	Cardiomyopathy and Exercise Intolerance in Muscle Glycogen Storage Disease 0. New England Journal of Medicine, 2007, 357, 1507-1514.	27.0	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. Antiviral Therapy, 2006, 11, 601-608.	1.0	48
81	Diagnosis, pathogenesis and treatment of inclusion body myositis. Current Opinion in Neurology, 2005, 18, 497-503.	3.6	15
82	Premature ageing in mice expressing defective mitochondrial DNA polymerase. Nature, 2004, 429, 417-423.	27.8	2,318
83	Mitochondrial Encephalomyopathies. Journal of Neuropathology and Experimental Neurology, 2003, 62, 217-227.	1.7	38
84	Ageing muscle: clonal expansions of mitochondrial DNA point mutations and deletions cause focal impairment of mitochondrial function. Neuromuscular Disorders, 2002, 12, 484-493.	0.6	180
85	The incidence of mitochondrial encephalomyopathies in childhood: Clinical features and morphological, biochemical, and DNA abnormalities. Annals of Neurology, 2001, 49, 377-383.	5.3	311
86	The incidence of mitochondrial encephalomyopathies in childhood: Clinical features and morphological, biochemical, and DNA abnormalities. Annals of Neurology, 2001, 49, 377-383.	5.3	7
87	Impaired insulin secretion and Î²-cell loss in tissue-specific knockout mice with mitochondrial diabetes. Nature Genetics, 2000, 26, 336-340.	21.4	417
88	Dilated cardiomyopathy and atrioventricular conduction blocks induced by heart-specific inactivation of mitochondrial DNA gene expression. Nature Genetics, 1999, 21, 133-137.	21.4	393
89	A novel heteroplasmic point mutation in the mitochondrial tRNALys gene in a sporadic case of mitochondrial encephalomyopathy: De novo mutation and no transmission to the offspring. Human Mutation, 1999, 13, 203-209.	2.5	21
90	Mitochondrial transcription factor A is necessary for mtDNA maintance and embryogenesis in mice. Nature Genetics, 1998, 18, 231-236.	21.4	1,377

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91	Upregulation of Fas/Fas ligand in inclusion body myositis. Annals of Neurology, 1998, 43, 127-130.	5.3	25
92	Down-Regulation of Mitochondrial Transcription Factor a During Spermatogenesis in Humans. Human Molecular Genetics, 1997, 6, 185-1991.	2.9	75
93	Analysis of multiple mitochondrial DNA deletions in inclusion body myositis. Human Mutation, 1997, 10, 381-386.	2.5	53
94	Fatal mitochondrial myopathy, lactic acidosis, and complex I deficiency associated with a heteroplasmic Aâ†'G mutation at position 3251 in the mitochondrial tRNALeu(UUR) gene. Human Genetics, 1996, 97, 269-273.	3.8	20
95	A single mouse gene encodes the mitochondrial transcription factor A and a testisâ€"specific nuclear HMG-box protein. Nature Genetics, 1996, 13, 296-302.	21.4	145
96	Clonal expansion of mitochondrial DNA with multiple deletions in autosomal dominant progressive external ophthalmoplegia. Annals of Neurology, 1996, 40, 707-713.	5.3	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. Journal of Neuropathology and Experimental Neurology, 1995, 54, 581-587.	1.7	103
99	Pathogenetic aspects of the A8344G mutation of mitochondrial DNA associated with MERRF syndrome and multiple symmetric lipomas. Muscle and Nerve, 1995, 18, S102-S106.	2.2	59
100	De novo mutation in the mitochondrial ATP synthase subunit 6 gene (T8993G) with rapid segregation resulting in Leigh syndrome in the offspring. Human Genetics, 1995, 96, 290-4.	3.8	35
101	Tissue distribution and disease manifestations of the tRNALys A?G(8344) mitochondrial DNA mutation in a case of myoclonus epilepsy and ragged red fibres. Acta Neuropathologica, 1995, 90, 328-333.	7.7	4
102	Restricted use of T cell receptor V genes in endomysial infiltrates of patients with inflammatory myopathies. European Journal of Immunology, 1994, 24, 2659-2663.	2.9	56
103	Duchenne muscular dystrophy and spinal muscular atrophy type I segregating in the same family. Clinical Genetics, 1994, 45, 97-103.	2.0	2
104	Mitochondrial ATP-Synthase Deficiency in a Child with 3-Methylglutaconic Aciduria. Pediatric Research, 1992, 32, 731-736.	2.3	65
105	Mitochondrial encephalomyopathies in childhood. I. Biochemical and morphologic investigations. Journal of Pediatrics, 1991, 119, 242-250.	1.8	128
106	Leber's hereditary optic neuropathy and complex I deficiency in muscle. Annals of Neurology, 1991, 30, 701-708.	5.3	155
107	Xâ€linked myotubular myopathy: a linkage study. Clinical Genetics, 1990, 37, 335-340.	2.0	34
108	THE EFFECT OF Î²â€ADRENERGIC BLOCKADE ON THE MYOPATHIC CHANGES IN EXPERIMENTAL HYPERTHYROIDISM IN RATS. Acta Pathologica, Microbiologica, Et Immunologica Scandinavica Section A, Pathology, 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.	7.7	4
110	Paranodal myelin retraction in protein deficient and normal rats. Acta Neuropathologica, 1977, 40, 249-252.	7.7	5