Sabine Krause

List of Publications by Year in descending order

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SARINE KDALLSE

#	Article	IF	CITATIONS
1	Health-related Quality of Life and Satisfaction with German Health Care Services in Patients with Charcot-Marie-Tooth Neuropathy. Journal of Neuromuscular Diseases, 2022, 9, 211-220.	2.6	4
2	Skeletal muscle provides the immunological micro-milieu for specific plasma cells in anti-synthetase syndrome-associated myositis. Acta Neuropathologica, 2022, 144, 353-372.	7.7	19
3	Slowly Progressive Limb-Girdle Weakness and HyperCKemia – Limb Girdle Muscular Dystrophy or Anti-3-Hydroxy-3-Methylglutaryl-CoA-Reductase-Myopathy?. Journal of Neuromuscular Diseases, 2022, , 1-8.	2.6	2
4	Expanding the clinical and molecular spectrum of <scp><i>ATP6V1A</i></scp> related metabolic cutis laxa. Journal of Inherited Metabolic Disease, 2021, 44, 972-986.	3.6	7
5	Delivery of oligonucleotideâ€based therapeutics: challenges and opportunities. EMBO Molecular Medicine, 2021, 13, e13243.	6.9	181
6	Late-onset neuromuscular disorders in the differential diagnosis of sarcopenia. BMC Neurology, 2021, 21, 241.	1.8	6
7	Congenital myopathy and epidermolysis bullosa due to PLEC variant. Neuromuscular Disorders, 2021, 31, 1212-1217.	0.6	4
8	A scalable, clinically severe pig model for Duchenne muscular dystrophy. DMM Disease Models and Mechanisms, 2021, 14, .	2.4	20
9	Antisense-Mediated Skipping of Dysferlin Exons in Control and Dysferlinopathy Patient-Derived Cells. Nucleic Acid Therapeutics, 2020, 30, 71-79.	3.6	4
10	Somatic gene editing ameliorates skeletal and cardiac muscle failure in pig and human models of Duchenne muscular dystrophy. Nature Medicine, 2020, 26, 207-214.	30.7	169
11	Cost of illness in Charcot-Marie-Tooth neuropathy. Neurology, 2019, 92, e2027-e2037.	1.1	17
12	Assessment of disease progression in dysferlinopathy. Neurology, 2019, 92, .	1.1	20
13	Teenage exercise is associated with earlier symptom onset in dysferlinopathy: a retrospective cohort study. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 1224-1226.	1.9	19
14	Muscle MRI in patients with dysferlinopathy: pattern recognition and implications for clinical trials. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 1071-1081.	1.9	81
15	Rare diagnosis of telethoninopathy (LGMD2G) in a Turkish patient. Neuromuscular Disorders, 2017, 27, 856-860.	0.6	15
16	The multifaceted clinical presentation of VCP-proteinopathy in a Greek family. Acta Myologica, 2017, 36, 203-206.	1.5	8
17	Differential expression and localization of Ankrd2 isoforms in human skeletal and cardiac muscles. Histochemistry and Cell Biology, 2016, 146, 569-584.	1.7	6
18	The immunoproteasomes are key to regulate myokines and MHC class I expression in idiopathic inflammatory myopathies. Journal of Autoimmunity, 2016, 75, 118-129.	6.5	34

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19	Progressive muscle proteome changes in a clinically relevant pig model of Duchenne muscular dystrophy. Scientific Reports, 2016, 6, 33362.	3.3	60
20	Insights into Muscle Degeneration from Heritable Inclusion Body Myopathies. Frontiers in Aging Neuroscience, 2015, 7, 13.	3.4	10
21	Integrative Analysis of MicroRNA and mRNA Data Reveals an Orchestrated Function of MicroRNAs in Skeletal Myocyte Differentiation in Response to TNF-α or IGF1. PLoS ONE, 2015, 10, e0135284.	2.5	21
22	Tumor Necrosis Factor Alpha and Insulin-Like Growth Factor 1 Induced Modifications of the Gene Expression Kinetics of Differentiating Skeletal Muscle Cells. PLoS ONE, 2015, 10, e0139520.	2.5	15
23	TNF-α and IGF1 modify the microRNA signature in skeletal muscle cell differentiation. Cell Communication and Signaling, 2015, 13, 4.	6.5	38
24	GNE myopathy in Roma patients homozygous for the p.I618T founder mutation. Neuromuscular Disorders, 2015, 25, 713-718.	0.6	32
25	Upregulation of Immunoproteasome Subunits in Myositis Indicates Active Inflammation with Involvement of Antigen Presenting Cells, CD8 T-Cells and IFNÎ ³ . PLoS ONE, 2014, 9, e104048.	2.5	33
26	The ubiquitin-selective chaperone Cdc48/p97 associates with Ubx3 to modulate monoubiquitylation of histone H2B. Nucleic Acids Research, 2014, 42, 10975-10986.	14.5	13
27	A new web-based method for automated analysis of muscle histology. BMC Musculoskeletal Disorders, 2013, 14, 26.	1.9	25
28	Proteomic characterization of aggregate components in an intrafamilial variable FHL1-associated myopathy. Neuromuscular Disorders, 2013, 23, 418-426.	0.6	25
29	Dystrophin-deficient pigs provide new insights into the hierarchy of physiological derangements of dystrophic muscle. Human Molecular Genetics, 2013, 22, 4368-4382.	2.9	134
30	<i>ANO5</i> Gene Analysis in a Large Cohort of Patients with Anoctaminopathy: Confirmation of Male Prevalence and High Occurrence of the Common Exon 5 Gene Mutation. Human Mutation, 2013, 34, 1111-1118.	2.5	64
31	In vitro supplementation with deoxynucleoside monophosphates rescues mitochondrial DNA depletion. Molecular Genetics and Metabolism, 2012, 107, 95-103.	1.1	31
32	Hexosamine Biosynthetic Pathway Mutations Cause Neuromuscular Transmission Defect. American Journal of Human Genetics, 2011, 88, 162-172.	6.2	153
33	A novel mutation in the myotilin gene (MYOT) causes a severe form of limb girdle muscular dystrophy 1A (LGMD1A). Journal of Neurology, 2011, 258, 1437-1444.	3.6	27
34	The phenotypic spectrum of neutral lipid storage myopathy due to mutations in the PNPLA2 gene. Journal of Neurology, 2011, 258, 1987-1997.	3.6	87
35	A founder mutation in Anoctamin 5 is a major cause of limb girdle muscular dystrophy. Brain, 2011, 134, 171-182.	7.6	254
36	Divergent Molecular Effects of Desmin Mutations on Protein Assembly in Myofibrillar Myopathy. Journal of Neuropathology and Experimental Neurology, 2010, 69, 415-424.	1.7	13

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37	Autosomal dominant nemaline myopathy caused by a novel α-tropomyosin 3 mutation. Journal of Neurology, 2010, 257, 658-660.	3.6	29
38	Late-onset autosomal dominant limb girdle muscular dystrophy and Paget's disease of bone unlinked to the VCP gene locus. Journal of the Neurological Sciences, 2010, 291, 79-85.	0.6	15
39	The p.G154S mutation of the alpha-B crystallin gene (CRYAB) causes late-onset distal myopathy. Neuromuscular Disorders, 2010, 20, 255-259.	0.6	81
40	Novel missense mutation p.A310P in the GNE gene in autosomal-recessive hereditary inclusion-body myopathy/distal myopathy with rimmed vacuoles in an Italian family. Neuromuscular Disorders, 2010, 20, 335-336.	0.6	11
41	McArdle disease and sporadic inclusion body myositis. Neuropathology and Applied Neurobiology, 2009, 35, 442-445.	3.2	2
42	Severe nemaline myopathy associated with consecutive mutations E74D and H75Y on a single ACTA1 allele. Neuromuscular Disorders, 2009, 19, 481-484.	0.6	16
43	Valosin containing protein associated inclusion body myopathy: abnormal vacuolization, autophagy and cell fusion in myoblasts. Neuromuscular Disorders, 2009, 19, 766-772.	0.6	59
44	5′ Trans-Splicing Repair of the PLEC1 Gene. Journal of Investigative Dermatology, 2008, 128, 568-574.	0.7	64
45	UDP-N-Acetylglucosamine 2-Epimerase/N-Acetylmannosamine Kinase (GNE) Binds to Alpha-Actinin 1: Novel Pathways in Skeletal Muscle?. PLoS ONE, 2008, 3, e2477.	2.5	71
46	GNE protein expression and subcellular distribution are unaltered in HIBM. Neurology, 2007, 69, 655-659.	1.1	40
47	Pathological consequences of VCP mutations on human striated muscle. Brain, 2007, 130, 381-393.	7.6	148
48	The ubiquitin-selective chaperone CDC-48/p97 links myosin assembly to human myopathy. Nature Cell Biology, 2007, 9, 379-390.	10.3	135
49	Characterization of hereditary inclusion body myopathy myoblasts: possible primary impairment of apoptotic events. Cell Death and Differentiation, 2007, 14, 1916-1924.	11.2	58
50	Brain imaging and neuropsychology in late-onset dementia due to a novel mutation (R93C) of valosin-containing protein. , 2007, 26, 232-240.		25
51	Influence of UDP-GlcNAc 2-Epimerase/ManNAc Kinase Mutant Proteins on Hereditary Inclusion Body Myopathyâ€. Biochemistry, 2006, 45, 2968-2977.	2.5	58
52	CHRND mutation causes a congenital myasthenic syndrome by impairing co-clustering of the acetylcholine receptor with rapsyn. Brain, 2006, 129, 2784-2793.	7.6	34
53	Impaired receptor clustering in congenital myasthenic syndrome with novel RAPSN mutations. Neurology, 2006, 67, 1159-1164.	1.1	34
54	Immunoproteasome subunit LMP2 expression is deregulated in Sjogren's syndrome but not in other autoimmune disorders. Annals of the Rheumatic Diseases, 2006, 65, 1021-1027.	0.9	57

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55	Localization of UDP-GlcNAc 2-epimerase/ManAc kinase (GNE) in the Golgi complex and the nucleus of mammalian cells. Experimental Cell Research, 2005, 304, 365-379.	2.6	72
56	No overall hyposialylation in hereditary inclusion body myopathy myoblasts carrying the homozygous M712T GNE mutation. Biochemical and Biophysical Research Communications, 2005, 328, 221-226.	2.1	93
57	Missense mutations of ACTA1 cause dominant congenital myopathy with cores. Journal of Medical Genetics, 2004, 41, 842-848.	3.2	110
58	Analysis of HLA class I and II alleles in sporadic inclusion-body myositis. Journal of Neurology, 2003, 250, 1313-1317.	3.6	38
59	Antigen processing and presentation in human muscle: cathepsin S is critical for MHC class II expression and upregulated in inflammatory myopathies. Journal of Neuroimmunology, 2003, 138, 132-143.	2.3	44
60	A novel homozygous missense mutation in the GNE gene of a patient with quadriceps-sparing hereditary inclusion body myopathy associated with muscle inflammation. Neuromuscular Disorders, 2003, 13, 830-834.	0.6	65
61	Ultrastructural Analysis of Transcription and Splicing in the Cell Nucleus after Bromo-UTP Microinjection. Molecular Biology of the Cell, 1999, 10, 211-223.	2.1	228
62	Localization of hepatitis delta virus RNA in the nucleus of human cells. Rna, 1998, 4, 680-693.	3.5	33
63	Structural Requirements for RNA Editing in Glutamate Receptor Pre-mRNAs by Recombinant Double-stranded RNA Adenosine Deaminase. Journal of Biological Chemistry, 1996, 271, 12221-12226.	3.4	146
64	Cloning of cDNAs encoding mammalian double-stranded RNA-specific adenosine deaminase. Molecular and Cellular Biology, 1995, 15, 1389-1397.	2.3	252
65	Isolation of genomic and cDNA clones encoding bovine poly(A) binding protein II. Nucleic Acids Research, 1995, 23, 4034-4041.	14.5	92
66	Immunodetection of Poly(A) Binding Protein II in the Cell Nucleus. Experimental Cell Research, 1994, 214, 75-82.	2.6	123