

# Sabine Krause

## List of Publications by Year in descending order

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Version: 2024-02-01

66  
papers

3,901  
citations

136950

32  
h-index

128289

60  
g-index

69  
all docs

69  
docs citations

69  
times ranked

6741  
citing authors

#	ARTICLE	IF	CITATIONS
1	A founder mutation in Anoctamin 5 is a major cause of limb girdle muscular dystrophy. <i>Brain</i> , 2011, 134, 171-182.	7.6	254
2	Cloning of cDNAs encoding mammalian double-stranded RNA-specific adenosine deaminase. <i>Molecular and Cellular Biology</i> , 1995, 15, 1389-1397.	2.3	252
3	Ultrastructural Analysis of Transcription and Splicing in the Cell Nucleus after Bromo-UTP Microinjection. <i>Molecular Biology of the Cell</i> , 1999, 10, 211-223.	2.1	228
4	Delivery of oligonucleotide-based therapeutics: challenges and opportunities. <i>EMBO Molecular Medicine</i> , 2021, 13, e13243.	6.9	181
5	Somatic gene editing ameliorates skeletal and cardiac muscle failure in pig and human models of Duchenne muscular dystrophy. <i>Nature Medicine</i> , 2020, 26, 207-214.	30.7	169
6	Hexosamine Biosynthetic Pathway Mutations Cause Neuromuscular Transmission Defect. <i>American Journal of Human Genetics</i> , 2011, 88, 162-172.	6.2	153
7	Pathological consequences of VCP mutations on human striated muscle. <i>Brain</i> , 2007, 130, 381-393.	7.6	148
8	Structural Requirements for RNA Editing in Glutamate Receptor Pre-mRNAs by Recombinant Double-stranded RNA Adenosine Deaminase. <i>Journal of Biological Chemistry</i> , 1996, 271, 12221-12226.	3.4	146
9	The ubiquitin-selective chaperone CDC-48/p97 links myosin assembly to human myopathy. <i>Nature Cell Biology</i> , 2007, 9, 379-390.	10.3	135
10	Dystrophin-deficient pigs provide new insights into the hierarchy of physiological derangements of dystrophic muscle. <i>Human Molecular Genetics</i> , 2013, 22, 4368-4382.	2.9	134
11	Immunodetection of Poly(A) Binding Protein II in the Cell Nucleus. <i>Experimental Cell Research</i> , 1994, 214, 75-82.	2.6	123
12	Missense mutations of ACTA1 cause dominant congenital myopathy with cores. <i>Journal of Medical Genetics</i> , 2004, 41, 842-848.	3.2	110
13	No overall hyposialylation in hereditary inclusion body myopathy myoblasts carrying the homozygous M712T GNE mutation. <i>Biochemical and Biophysical Research Communications</i> , 2005, 328, 221-226.	2.1	93
14	Isolation of genomic and cDNA clones encoding bovine poly(A) binding protein II. <i>Nucleic Acids Research</i> , 1995, 23, 4034-4041.	14.5	92
15	The phenotypic spectrum of neutral lipid storage myopathy due to mutations in the PNPLA2 gene. <i>Journal of Neurology</i> , 2011, 258, 1987-1997.	3.6	87
16	The p.G154S mutation of the alpha-B crystallin gene (CRYAB) causes late-onset distal myopathy. <i>Neuromuscular Disorders</i> , 2010, 20, 255-259.	0.6	81
17	Muscle MRI in patients with dysferlinopathy: pattern recognition and implications for clinical trials. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 1071-1081.	1.9	81
18	Localization of UDP-GlcNAc 2-epimerase/ManAc kinase (GNE) in the Golgi complex and the nucleus of mammalian cells. <i>Experimental Cell Research</i> , 2005, 304, 365-379.	2.6	72

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19	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
20	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
21	5â€² Trans-Splicing Repair of the PLEC1 Gene. Journal of Investigative Dermatology, 2008, 128, 568-574.	0.7	64
22	<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
23	Progressive muscle proteome changes in a clinically relevant pig model of Duchenne muscular dystrophy. Scientific Reports, 2016, 6, 33362.	3.3	60
24	Valosin containing protein associated inclusion body myopathy: abnormal vacuolization, autophagy and cell fusion in myoblasts. Neuromuscular Disorders, 2009, 19, 766-772.	0.6	59
25	Influence of UDP-GlcNAc 2-Epimerase/ManNAc Kinase Mutant Proteins on Hereditary Inclusion Body Myopathyâ€. Biochemistry, 2006, 45, 2968-2977.	2.5	58
26	Characterization of hereditary inclusion body myopathy myoblasts: possible primary impairment of apoptotic events. Cell Death and Differentiation, 2007, 14, 1916-1924.	11.2	58
27	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
28	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
29	GNE protein expression and subcellular distribution are unaltered in HIBM. Neurology, 2007, 69, 655-659.	1.1	40
30	Analysis of HLA class I and II alleles in sporadic inclusion-body myositis. Journal of Neurology, 2003, 250, 1313-1317.	3.6	38
31	TNF-Î± and IGF1 modify the microRNA signature in skeletal muscle cell differentiation. Cell Communication and Signaling, 2015, 13, 4.	6.5	38
32	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
33	Impaired receptor clustering in congenital myasthenic syndrome with novel RAPSN mutations. Neurology, 2006, 67, 1159-1164.	1.1	34
34	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
35	Localization of hepatitis delta virus RNA in the nucleus of human cells. Rna, 1998, 4, 680-693.	3.5	33
36	Upregulation of Immunoproteasome Subunits in Myositis Indicates Active Inflammation with Involvement of Antigen Presenting Cells, CD8 T-Cells and IFNÎ³. PLoS ONE, 2014, 9, e104048.	2.5	33

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37	GNE myopathy in Roma patients homozygous for the p.I618T founder mutation. <i>Neuromuscular Disorders</i> , 2015, 25, 713-718.	0.6	32
38	In vitro supplementation with deoxynucleoside monophosphates rescues mitochondrial DNA depletion. <i>Molecular Genetics and Metabolism</i> , 2012, 107, 95-103.	1.1	31
39	Autosomal dominant nemaline myopathy caused by a novel $\beta$ -tropomyosin 3 mutation. <i>Journal of Neurology</i> , 2010, 257, 658-660.	3.6	29
40	A novel mutation in the myotilin gene (MYOT) causes a severe form of limb girdle muscular dystrophy 1A (LGMD1A). <i>Journal of Neurology</i> , 2011, 258, 1437-1444.	3.6	27
41	A new web-based method for automated analysis of muscle histology. <i>BMC Musculoskeletal Disorders</i> , 2013, 14, 26.	1.9	25
42	Proteomic characterization of aggregate components in an intrafamilial variable FHL1-associated myopathy. <i>Neuromuscular Disorders</i> , 2013, 23, 418-426.	0.6	25
43	Brain imaging and neuropsychology in late-onset dementia due to a novel mutation (R93C) of valosin-containing protein. , 2007, 26, 232-240.		25
44	Integrative Analysis of MicroRNA and mRNA Data Reveals an Orchestrated Function of MicroRNAs in Skeletal Myocyte Differentiation in Response to TNF- $\alpha$ or IGF1. <i>PLoS ONE</i> , 2015, 10, e0135284.	2.5	21
45	Assessment of disease progression in dysferlinopathy. <i>Neurology</i> , 2019, 92, .	1.1	20
46	A scalable, clinically severe pig model for Duchenne muscular dystrophy. <i>DMM Disease Models and Mechanisms</i> , 2021, 14, .	2.4	20
47	Teenage exercise is associated with earlier symptom onset in dysferlinopathy: a retrospective cohort study. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 1224-1226.	1.9	19
48	Skeletal muscle provides the immunological micro-milieu for specific plasma cells in anti-synthetase syndrome-associated myositis. <i>Acta Neuropathologica</i> , 2022, 144, 353-372.	7.7	19
49	Cost of illness in Charcot-Marie-Tooth neuropathy. <i>Neurology</i> , 2019, 92, e2027-e2037.	1.1	17
50	Severe nemaline myopathy associated with consecutive mutations E74D and H75Y on a single ACTA1 allele. <i>Neuromuscular Disorders</i> , 2009, 19, 481-484.	0.6	16
51	Late-onset autosomal dominant limb girdle muscular dystrophy and Paget's disease of bone unlinked to the VCP gene locus. <i>Journal of the Neurological Sciences</i> , 2010, 291, 79-85.	0.6	15
52	Tumor Necrosis Factor Alpha and Insulin-Like Growth Factor 1 Induced Modifications of the Gene Expression Kinetics of Differentiating Skeletal Muscle Cells. <i>PLoS ONE</i> , 2015, 10, e0139520.	2.5	15
53	Rare diagnosis of telethoninopathy (LGMD2G) in a Turkish patient. <i>Neuromuscular Disorders</i> , 2017, 27, 856-860.	0.6	15
54	Divergent Molecular Effects of Desmin Mutations on Protein Assembly in Myofibrillar Myopathy. <i>Journal of Neuropathology and Experimental Neurology</i> , 2010, 69, 415-424.	1.7	13

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55	The ubiquitin-selective chaperone Cdc48/p97 associates with Ubx3 to modulate monoubiquitylation of histone H2B. <i>Nucleic Acids Research</i> , 2014, 42, 10975-10986.	14.5	13
56	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. <i>Neuromuscular Disorders</i> , 2010, 20, 335-336.	0.6	11
57	Insights into Muscle Degeneration from Heritable Inclusion Body Myopathies. <i>Frontiers in Aging Neuroscience</i> , 2015, 7, 13.	3.4	10
58	The multifaceted clinical presentation of VCP-proteinopathy in a Greek family. <i>Acta Myologica</i> , 2017, 36, 203-206.	1.5	8
59	Expanding the clinical and molecular spectrum of <i>ATP6V1A</i> related metabolic cutis laxa. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 972-986.	3.6	7
60	Differential expression and localization of Ankrd2 isoforms in human skeletal and cardiac muscles. <i>Histochemistry and Cell Biology</i> , 2016, 146, 569-584.	1.7	6
61	Late-onset neuromuscular disorders in the differential diagnosis of sarcopenia. <i>BMC Neurology</i> , 2021, 21, 241.	1.8	6
62	Antisense-Mediated Skipping of Dysferlin Exons in Control and Dysferlinopathy Patient-Derived Cells. <i>Nucleic Acid Therapeutics</i> , 2020, 30, 71-79.	3.6	4
63	Health-related Quality of Life and Satisfaction with German Health Care Services in Patients with Charcot-Marie-Tooth Neuropathy. <i>Journal of Neuromuscular Diseases</i> , 2022, 9, 211-220.	2.6	4
64	Congenital myopathy and epidermolysis bullosa due to PLEC variant. <i>Neuromuscular Disorders</i> , 2021, 31, 1212-1217.	0.6	4
65	McArdle disease and sporadic inclusion body myositis. <i>Neuropathology and Applied Neurobiology</i> , 2009, 35, 442-445.	3.2	2
66	Slowly Progressive Limb-Girdle Weakness and HyperCKemia – Limb Girdle Muscular Dystrophy or Anti-3-Hydroxy-3-Methylglutaryl-CoA-Reductase-Myopathy?. <i>Journal of Neuromuscular Diseases</i> , 2022, , 1-8.	2.6	2