Agatha Schluter

List of Publications by Year
in descending order

50 papers	4,314 citations	201674 27 h-index	197818 49 g-index
52 all docs	52 docs citations	52 times ranked	10355 citing authors

#	Article	IF	Citations
1	Modulation of mitochondrial and inflammatory homeostasis through RIP140 is neuroprotective in an adrenoleukodystrophy mouse model. Neuropathology and Applied Neurobiology, 2022, 48, .	3.2	6
2	Diagnosis of Genetic White Matter Disorders by Singleton Whole-Exome and Genome Sequencing Using Interactome-Driven Prioritization. Neurology, 2022, , 10.1212/WNL.0000000000013278.	1.1	13
3	Systematic Collaborative Reanalysis of Genomic Data Improves Diagnostic Yield in Neurologic Rare Diseases. Journal of Molecular Diagnostics, 2022, 24, 529-542.	2.8	6
4	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. EBioMedicine, 2021, 65, 103246.	6.1	52
5	SARS-CoV-2â€“related MIS-C: A key to the viral and genetic causes of Kawasaki disease?. Journal of Experimental Medicine, 2021, 218, .	8.5	100
6	Biallelic PI4KA variants cause a novel neurodevelopmental syndrome with hypomyelinating leukodystrophy. Brain, 2021, 144, 2659-2669.	7.6	19
7	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. Science Immunology, 2021, 6, .	11.9	267
8	Bi-allelic variants in the mitochondrial RNase P subunit PRORP cause mitochondrial tRNA processing defects and pleiotropic multisystem presentations. American Journal of Human Genetics, 2021, 108, 2195-2204.	6.2	26
9	Complete loss of KCNA1 activity causes neonatal epileptic encephalopathy and dyskinesia. Journal of Medical Genetics, 2020, 57, 132-137.	3.2	26
10	Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308.	3.6	43
11	Truncating variants in UBAP1 associated with childhoodâ€onset nonsyndromic hereditary spastic paraplegia. Human Mutation, 2020, 41, 632-640.	2.5	15
12	A deep intronic splice variant advises reexamination of presumably dominant SPG7 Cases. Annals of Clinical and Translational Neurology, 2020, 7, 105-111.	3.7	17
13	Impairment of the mitochondrial one-carbon metabolism enzyme SHMT2 causes a novel brain and heart developmental syndrome. Acta Neuropathologica, 2020, 140, 971-975.	7.7	24
14	A novel hypomorphic splice variant in EIF2B5 gene is associated with mild ovarioleukodystrophy. Annals of Clinical and Translational Neurology, 2020, 7, 1574-1579.	3.7	3
15	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. Science, 2020, 370, .	12.6	1,749
16	Case Report: Benign Infantile Seizures Temporally Associated With COVID-19. Frontiers in Pediatrics, 2020, 8, 507.	1.9	23
17	Expanding the clinical and genetic spectrum of PCYT2-related disorders. Brain, 2020, 143, e76-e76.	7.6	14
18	Highâ€dose biotin restores redox balance, energy and lipid homeostasis, and axonal health in a model of adrenoleukodystrophy. Brain Pathology, 2020, 30, 945-963.	4.1	11

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19	The peroxisomal fatty acid transporter ABCD1/PMP-4 is required in the C. elegans hypodermis for axonal maintenance: A worm model for adrenoleukodystrophy. Free Radical Biology and Medicine, 2020, 152, 797-809.	2.9	19
20	<scp>HNRNPH1</scp>â€related syndromic intellectual disability: Seven additional cases suggestive of a distinct syndromic neurodevelopmental syndrome. Clinical Genetics, 2020, 98, 91-98.	2.0	25
21	Parkinsonism and spastic paraplegia type 7: Expanding the spectrum of mitochondrial Parkinsonism. Movement Disorders, 2019, 34, 1547-1561.	3.9	44
22	Biomarker Identification, Safety, and Efficacy of High-Dose Antioxidants for Adrenomyeloneuropathy: a Phase II Pilot Study. Neurotherapeutics, 2019, 16, 1167-1182.	4.4	31
23	A novel mutation in the GFAP gene expands the phenotype of Alexander disease. Journal of Medical Genetics, 2019, 56, 846-849.	3.2	9
24	Reply to: â€œMitochondrial Parkinsonism due to SPG7/Paraplegin variants with secondary mtDNA depletionâ€. Movement Disorders, 2019, 34, 1932-1933.	3.9	0
25	Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. Journal of Clinical Investigation, 2019, 129, 1240-1256.	8.2	68
26	Epigenomic signature of adrenoleukodystrophy predicts compromised oligodendrocyte differentiation. Brain Pathology, 2018, 28, 902-919.	4.1	21
27	Genetic Variants in HSD17B3, SMAD3, and IPO11 Impact Circulating Lipids in Response to Fenofibrate in Individuals With Type 2 Diabetes. Clinical Pharmacology and Therapeutics, 2018, 103, 712-721.	4.7	30
28	Allelic Expression Imbalance Promoting a Mutant PEX6 Allele Causes Zellweger Spectrum Disorder. American Journal of Human Genetics, 2017, 101, 965-976.	6.2	41
29	Targeted activation of <scp>CREB</scp> in reactive astrocytes is neuroprotective in focal acute cortical injury. Glia, 2016, 64, 853-874.	4.9	27
30	Uniparental disomy of chromosome 16 unmasks recessive mutations of FA2H /SPG35 in 4 families. Neurology, 2016, 87, 186-191.	1.1	27
31	Neuroinflammatory Signals in Alzheimer Disease and APP/PS1 Transgenic Mice. Journal of Neuropathology and Experimental Neurology, 2015, 74, 319-344.	1.7	105
32	Altered glycolipid and glycerophospholipid signaling drive inflammatory cascades in adrenomyeloneuropathy. Human Molecular Genetics, 2015, 24, ddv375.	2.9	37
33	Deregulation of purine metabolism in Alzheimer's disease. Neurobiology of Aging, 2015, 36, 68-80.	3.1	108
34	ABCD2 Alters Peroxisome Proliferator-Activated ReceptorÎ±Signaling In Vitro, but Does Not Impair Responses to Fenofibrate Therapy in a Mouse Model of Diet-Induced Obesity. Molecular Pharmacology, 2014, 86, 505-513.	2.3	7
35	Oxidative stress regulates the ubiquitinâ€“proteasome system and immunoproteasome functioning in a mouse model of X-adrenoleukodystrophy. Brain, 2013, 136, 891-904.	7.6	39
36	Functional Genomics Reveals Dysregulation of Cortical Olfactory Receptors in Parkinson Disease: Novel Putative Chemoreceptors in the Human Brain. Journal of Neuropathology and Experimental Neurology, 2013, 72, 524-539.	1.7	111

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37	Functional genomic analysis unravels a metabolic-inflammatory interplay in adrenoleukodystrophy. Human Molecular Genetics, 2012, 21, 1062-1077.	2.9	62
38	Phylogenomic Evidence for a Myxococcal Contribution to the Mitochondrial Fatty Acid Beta-Oxidation. PLoS ONE, 2011, 6, e21989.	2.5	7
39	Antioxidants halt axonal degeneration in a mouse model of Xâ€adrenoleukodystrophy. Annals of Neurology, 2011, 70, 84-92.	5.3	122
40	Oxidative Damage Compromises Energy Metabolism in the Axonal Degeneration Mouse Model of X-Adrenoleukodystrophy. Antioxidants and Redox Signaling, 2011, 15, 2095-2107.	5.4	78
41	PeroxisomeDB 2.0: an integrative view of the global peroxisomal metabolome. Nucleic Acids Research, 2010, 38, D800-D805.	14.5	103
42	A key role for the peroxisomalABCD2transporter in fatty acid homeostasis. American Journal of Physiology - Endocrinology and Metabolism, 2009, 296, E211-E221.	3.5	91
43	PeroxisomeDB: a database for the peroxisomal proteome, functional genomics and disease. Nucleic Acids Research, 2007, 35, D815-D822.	14.5	65
44	RCAN3, a novel calcineurin inhibitor that down-regulates NFAT-dependent cytokine gene expression. Biochimica Et Biophysica Acta - Molecular Cell Research, 2007, 1773, 330-341.	4.1	41
45	The Evolutionary Origin of Peroxisomes: An ER-Peroxisome Connection. Molecular Biology and Evolution, 2006, 23, 838-845.	8.9	152
46	Phytanic acid, a novel activator of uncoupling protein-1 gene transcription and brown adipocyte differentiation. Biochemical Journal, 2002, 362, 61.	3.7	22
47	Phytanic acid, a novel activator of uncoupling protein-1 gene transcription and brown adipocyte differentiation. Biochemical Journal, 2002, 362, 61-69.	3.7	43
48	Phytanic acid, but not pristanic acid, mediates the positive effects of phytol derivatives on brown adipocyte differentiation. FEBS Letters, 2002, 517, 83-86.	2.8	19
49	The chlorophyll-derived metabolite phytanic acid induces white adipocyte differentiation. International Journal of Obesity, 2002, 26, 1277-1280.	3.4	39
50	Peroxisome Proliferator-activated Receptor Î± Activates Transcription of the Brown Fat Uncoupling Protein-1 Gene. Journal of Biological Chemistry, 2001, 276, 1486-1493.	3.4	302