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	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. Science, 2020, 370, .	12.6	1,749
2	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
3	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
4	The Evolutionary Origin of Peroxisomes: An ER-Peroxisome Connection. Molecular Biology and Evolution, 2006, 23, 838-845.	8.9	152
5	Antioxidants halt axonal degeneration in a mouse model of Xâ€adrenoleukodystrophy. Annals of Neurology, 2011, 70, 84-92.	5.3	122
6	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
7	Deregulation of purine metabolism in Alzheimer's disease. Neurobiology of Aging, 2015, 36, 68-80.	3.1	108
8	Neuroinflammatory Signals in Alzheimer Disease and APP/PS1 Transgenic Mice. Journal of Neuropathology and Experimental Neurology, 2015, 74, 319-344.	1.7	105
9	PeroxisomeDB 2.0: an integrative view of the global peroxisomal metabolome. Nucleic Acids Research, 2010, 38, D800-D805.	14.5	103
10	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
11	A key role for the peroxisomalABCD2transporter in fatty acid homeostasis. American Journal of Physiology - Endocrinology and Metabolism, 2009, 296, E211-E221.	3.5	91
12	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
13	Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. Journal of Clinical Investigation, 2019, 129, 1240-1256.	8.2	68
14	PeroxisomeDB: a database for the peroxisomal proteome, functional genomics and disease. Nucleic Acids Research, 2007, 35, D815-D822.	14.5	65
15	Functional genomic analysis unravels a metabolic-inflammatory interplay in adrenoleukodystrophy. Human Molecular Genetics, 2012, 21, 1062-1077.	2.9	62
16	Shorter androgen receptor polyQ alleles protect against life-threatening COVID-19 disease in European males. EBioMedicine, 2021, 65, 103246.	6.1	52
17	Parkinsonism and spastic paraplegia type 7: Expanding the spectrum of mitochondrial Parkinsonism. Movement Disorders, 2019, 34, 1547-1561.	3.9	44
18	Phytanic acid, a novel activator of uncoupling protein-1 gene transcription and brown adipocyte differentiation. Biochemical Journal, 2002, 362, 61-69.	3.7	43

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19	Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308.	3.6	43
20	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
21	Allelic Expression Imbalance Promoting a Mutant PEX6 Allele Causes Zellweger Spectrum Disorder. American Journal of Human Genetics, 2017, 101, 965-976.	6.2	41
22	The chlorophyll-derived metabolite phytanic acid induces white adipocyte differentiation. International Journal of Obesity, 2002, 26, 1277-1280.	3.4	39
23	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
24	Altered glycolipid and glycerophospholipid signaling drive inflammatory cascades in adrenomyeloneuropathy. Human Molecular Genetics, 2015, 24, ddv375.	2.9	37
25	Biomarker Identification, Safety, and Efficacy of High-Dose Antioxidants for Adrenomyeloneuropathy: a Phase II Pilot Study. Neurotherapeutics, 2019, 16, 1167-1182.	4.4	31
26	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
27	Targeted activation of <scp>CREB</scp> in reactive astrocytes is neuroprotective in focal acute cortical injury. Glia, 2016, 64, 853-874.	4.9	27
28	Uniparental disomy of chromosome 16 unmasks recessive mutations of FA2H /SPG35 in 4 families. Neurology, 2016, 87, 186-191.	1.1	27
29	Complete loss of KCNA1 activity causes neonatal epileptic encephalopathy and dyskinesia. Journal of Medical Genetics, 2020, 57, 132-137.	3.2	26
30	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
31	<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
32	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
33	Case Report: Benign Infantile Seizures Temporally Associated With COVID-19. Frontiers in Pediatrics, 2020, 8, 507.	1.9	23
34	Phytanic acid, a novel activator of uncoupling protein-1 gene transcription and brown adipocyte differentiation. Biochemical Journal, 2002, 362, 61.	3.7	22
35	Epigenomic signature of adrenoleukodystrophy predicts compromised oligodendrocyte differentiation. Brain Pathology, 2018, 28, 902-919.	4.1	21
36	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

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37	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
38	Biallelic PI4KA variants cause a novel neurodevelopmental syndrome with hypomyelinating leukodystrophy. Brain, 2021, 144, 2659-2669.	7.6	19
39	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
40	Truncating variants in UBAP1 associated with childhoodâ€onset nonsyndromic hereditary spastic paraplegia. Human Mutation, 2020, 41, 632-640.	2.5	15
41	Expanding the clinical and genetic spectrum of PCYT2-related disorders. Brain, 2020, 143, e76-e76.	7.6	14
42	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
43	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
44	A novel mutation in the GFAP gene expands the phenotype of Alexander disease. Journal of Medical Genetics, 2019, 56, 846-849.	3.2	9
45	Phylogenomic Evidence for a Myxococcal Contribution to the Mitochondrial Fatty Acid Beta-Oxidation. PLoS ONE, 2011, 6, e21989.	2.5	7
46	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
47	Modulation of mitochondrial and inflammatory homeostasis through RIP140 is neuroprotective in an adrenoleukodystrophy mouse model. Neuropathology and Applied Neurobiology, 2022, 48, .	3.2	6
48	Systematic Collaborative Reanalysis of Genomic Data Improves Diagnostic Yield in Neurologic Rare Diseases. Journal of Molecular Diagnostics, 2022, 24, 529-542.	2.8	6
49	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
50	Reply to: â€œMitochondrial Parkinsonism due to SPG7/Paraplegin variants with secondary mtDNA depletionâ€• Movement Disorders, 2019, 34, 1932-1933.	3.9	0