Stephan Kemp

List of Publications by Year
in descending order

106 papers	8,634 citations	71102 41 h-index	43889 91 g-index
111 all docs	111 docs citations	111 times ranked	9936 citing authors

#	Article	IF	Citations
1	Histone deacetylases (HDACs): characterization of the classical HDAC family. Biochemical Journal, 2003, 370, 737-749.	3.7	2,671
2	X-linked adrenoleukodystrophy (X-ALD): clinical presentation and guidelines for diagnosis, follow-up and management. Orphanet Journal of Rare Diseases, 2012, 7, 51.	2.7	403
3	THE CONCISE GUIDE TO PHARMACOLOGY 2017/18: Overview. British Journal of Pharmacology, 2017, 174, S1-S16.	5.4	269
4	ABCD1 mutations and the X-linked adrenoleukodystrophy mutation database: Role in diagnosis and clinical correlations. Human Mutation, 2001, 18, 499-515.	2.5	261
5	Gene redundancy and pharmacological gene therapy: Implications for X-linked adrenoleukodystrophy. Nature Medicine, 1998, 4, 1261-1268.	30.7	237
6	X-linked adrenoleukodystrophy: Clinical, metabolic, genetic and pathophysiological aspects. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1465-1474.	3.8	217
7	Deletion of the serine 34 codon from the major peripheral myelin protein P0 gene in Charcotâ€“Marieâ€“Tooth disease type 1B. Nature Genetics, 1993, 5, 35-39.	21.4	215
8	Adrenoleukodystrophy â€“ neuroendocrine pathogenesis and redefinition of natural history. Nature Reviews Endocrinology, 2016, 12, 606-615.	9.6	189
9	X-linked adrenoleukodystrophy in women: a cross-sectional cohort study. Brain, 2014, 137, 693-706.	7.6	182
10	Early oxidative damage underlying neurodegeneration in X-adrenoleukodystrophy. Human Molecular Genetics, 2008, 17, 1762-1773.	2.9	181
11	Fatty acid omegaâ€oxidation as a rescue pathway for fatty acid oxidation disorders in humans. FEBS Journal, 2011, 278, 182-194.	4.7	181
12	THE CONCISE GUIDE TO PHARMACOLOGY 2019/20: Transporters. British Journal of Pharmacology, 2019, 176, S397-S493.	5.4	166
13	X-linked adrenoleukodystrophy: genes, mutations, and phenotypes. Neurochemical Research, 1999, 24, 521-535.	3.3	162
14	The role of ELOVL1 in very longâ€chain fatty acid homeostasis and Xâ€linked adrenoleukodystrophy. EMBO Molecular Medicine, 2010, 2, 90-97.	6.9	140
15	Analysis of very long-chain fatty acids using electrospray ionization mass spectrometry. Molecular Genetics and Metabolism, 2003, 79, 189-196.	1.1	138
16	Peroxisomes, lipid metabolism and lipotoxicity. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2010, 1801, 272-280.	2.4	135
17	A novel defect of peroxisome division due to a homozygous non-sense mutation in thePEX11Î²gene. Journal of Medical Genetics, 2012, 49, 307-313.	3.2	127
18	X-Linked Adrenoleukodystrophy: Pathogenesis and Treatment. Current Neurology and Neuroscience Reports, 2014, 14, 486.	4.2	127

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19	THE CONCISE GUIDE TO PHARMACOLOGY 2021/22: Transporters. British Journal of Pharmacology, 2021, 178, S412-S513.	5.4	114
20	The Natural History of Adrenal Insufficiency in X-Linked Adrenoleukodystrophy: An International Collaboration. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 118-126.	3.6	102
21	The peroxisomal ABC transporter family. Pflugers Archiv European Journal of Physiology, 2007, 453, 719-734.	2.8	95
22	Mammalian peroxisomal ABC transporters: from endogenous substrates to pathology and clinical significance. British Journal of Pharmacology, 2011, 164, 1753-1766.	5.4	93
23	Inactivation of the peroxisomal ABCD2 transporter in the mouse leads to late-onset ataxia involving mitochondria, Golgi and endoplasmic reticulum damage. Human Molecular Genetics, 2005, 14, 3565-3577.	2.9	90
24	Valproic acid induces antioxidant effects in X-linked adrenoleukodystrophy. Human Molecular Genetics, 2010, 19, 2005-2014.	2.9	90
25	Elongation of very long-chain fatty acids is enhanced in X-linked adrenoleukodystrophy. Molecular Genetics and Metabolism, 2005, 84, 144-151.	1.1	83
26	Biochemical Aspects of Xâ€Linked Adrenoleukodystrophy. Brain Pathology, 2010, 20, 831-837.	4.1	83
27	A very long-chain acyl-CoA synthetase-deficient mouse and its relevance to X-linked adrenoleukodystrophy. Human Molecular Genetics, 2003, 12, 1145-1154.	2.9	82
28	X-linked adrenoleukodystrophy: Very long-chain fatty acid metabolism, ABC half-transporters and the complicated route to treatment. Molecular Genetics and Metabolism, 2007, 90, 268-276.	1.1	73
29	Identification of a Two Base Pair Deletion in Five Unrelated Families with Adrenoleukodystrophy: A Possible Hot Spot for Mutations. Biochemical and Biophysical Research Communications, 1994, 202, 647-653.	2.1	70
30	Ï‰-Oxidation of Very Long-chain Fatty Acids in Human Liver Microsomes. Journal of Biological Chemistry, 2006, 281, 13180-13187.	3.4	69
31	Pharmacological induction of peroxisomes in peroxisome biogenesis disorders. Annals of Neurology, 2000, 47, 286-296.	5.3	67
32	Lovastatin in X-Linked Adrenoleukodystrophy. New England Journal of Medicine, 2010, 362, 276-277.	27.0	58
33	Disease progression in women with X-linked adrenoleukodystrophy is slow. Orphanet Journal of Rare Diseases, 2019, 14, 30.	2.7	58
34	Betulinic acid induces a novel cell death pathway that depends on cardiolipin modification. Oncogene, 2016, 35, 427-437.	5.9	57
35	Connexin32 gene mutations in X-linked dominant Charcot-Marie-Tooth disease (CMTX1). Human Genetics, 1997, 99, 501-505.	3.8	55
36	Hematopoietic cell transplantation does not prevent myelopathy in Xâ€linked adrenoleukodystrophy: a retrospective study. Journal of Inherited Metabolic Disease, 2015, 38, 359-361.	3.6	54

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37	Comparison of C26:0-carnitine and C26:0-lysophosphatidylcholine as diagnostic markers in dried blood spots from newborns and patients with adrenoleukodystrophy. Molecular Genetics and Metabolism, 2017, 122, 209-215.	1.1	50
38	Role of very-long-chain acyl-coenzyme A synthetase in X-linked adrenoleukodystrophy. Annals of Neurology, 1999, 46, 409-412.	5.3	48
39	Adrenoleukodystrophy Newborn Screening in the Netherlands (SCAN Study): The X-Factor. Frontiers in Cell and Developmental Biology, 2020, 8, 499.	3.7	47
40	The novel histone deacetylase inhibitor BL1521 inhibits proliferation and induces apoptosis in neuroblastoma cells. Biochemical Pharmacology, 2004, 68, 1279-1288.	4.4	46
41	Characterization of the human Ï‰â€oxidation pathway for Ï‰â€hydroxyâ€veryâ€longâ€chain fatty acids. FASEB Journal, 2008, 22, 2064-2071.	0.5	46
42	Progression of myelopathy in males with adrenoleukodystrophy: towards clinical trial readiness. Brain, 2019, 142, 334-343.	7.6	43
43	Conservation of targeting but divergence in function and quality control of peroxisomal ABC transporters: an analysis using cross-kingdom expression. Biochemical Journal, 2011, 436, 547-557.	3.7	41
44	Bezafibrate lowers very longâ€chain fatty acids in Xâ€linked adrenoleukodystrophy fibroblasts by inhibiting fatty acid elongation. Journal of Inherited Metabolic Disease, 2012, 35, 1137-1145.	3.6	39
45	<scp>MRI</scp> surveillance of boys with Xâ€linked adrenoleukodystrophy identified by newborn screening: Metaâ€analysis and consensus guidelines. Journal of Inherited Metabolic Disease, 2021, 44, 728-739.	3.6	39
46	Method for Measurement of Peroxisomal Very-Long-Chain Fatty Acid Î²-Oxidation in Human Skin Fibroblasts Using Stable-Isotope-Labeled Tetracosanoic Acid. Clinical Chemistry, 2004, 50, 1824-1826.	3.2	38
47	Gene expression profiling in response to the histone deacetylase inhibitor BL1521 in neuroblastoma. Experimental Cell Research, 2005, 309, 451-467.	2.6	38
48	Evidence for two enzymatic pathways for Ï‰-oxidation of docosanoic acid in rat liver microsomes. Journal of Lipid Research, 2005, 46, 1001-1008.	4.2	37
49	No evidence for â€˜skewedâ€™ inactivation of the X-chromosome as cause of Leber's hereditary optic neuropathy in female carriers. Human Genetics, 1996, 97, 500-505.	3.8	35
50	C26:0-Carnitine Is a New Biomarker for X-Linked Adrenoleukodystrophy in Mice and Man. PLoS ONE, 2016, 11, e0154597.	2.5	33
51	Accumulation of very long-chain fatty acids does not affect mitochondrial function in adrenoleukodystrophy protein deficiency. Human Molecular Genetics, 2005, 14, 1127-1137.	2.9	32
52	Comparison of the Diagnostic Performance of C26:0-Lysophosphatidylcholine and Very Long-Chain Fatty Acids Analysis for Peroxisomal Disorders. Frontiers in Cell and Developmental Biology, 2020, 8, 690.	3.7	31
53	Contiguous gene deletion of ELOVL7, ERCC8 and NDUFAF2 in a patient with a fatal multisystem disorder. Human Molecular Genetics, 2009, 18, 3365-3374.	2.9	30
54	Mouse Very Long-chain Acyl-CoA Synthetase in X-linked Adrenoleukodystrophy. Journal of Biological Chemistry, 2002, 277, 28765-28773.	3.4	26

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55	Bezafibrate for X-Linked Adrenoleukodystrophy. PLoS ONE, 2012, 7, e41013.	2.5	26
56	Enzymatic characterization of ELOVL1, a key enzyme in very long-chain fatty acid synthesis. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2015, 1851, 231-237.	2.4	26
57	Lipid-induced endoplasmic reticulum stress in X-linked adrenoleukodystrophy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 2255-2265.	3.8	26
58	The brain penetrant PPARÎ³ agonist leriglitazone restores multiple altered pathways in models of X-linked adrenoleukodystrophy. Science Translational Medicine, 2021, 13, .	12.4	24
59	Structure and Function of the ABCD1 Variant Database: 20 Years, 940 Pathogenic Variants, and 3400 Cases of Adrenoleukodystrophy. Cells, 2022, 11, 283.	4.1	23
60	Intrathecal Adeno-Associated Viral Vector-Mediated Gene Delivery for Adrenomyeloneuropathy. Human Gene Therapy, 2019, 30, 544-555.	2.7	21
61	Methionine metabolism and phenotypic variability in X-linked adrenoleukodystrophy. Neurology, 2006, 66, 442-443.	1.1	20
62	Translational Metabolism: A multidisciplinary approach towards precision diagnosis of inborn errors of metabolism in the omics era. Journal of Inherited Metabolic Disease, 2019, 42, 197-208.	3.6	20
63	Plasma NfL and GFAP as biomarkers of spinal cord degeneration in adrenoleukodystrophy. Annals of Clinical and Translational Neurology, 2020, 7, 2127-2136.	3.7	19
64	Vorinostat in the acute neuroinflammatory form of Xâ€linked adrenoleukodystrophy. Annals of Clinical and Translational Neurology, 2020, 7, 639-652.	3.7	19
65	The cystathionine beta-synthase variant c.844_845ins68 protects against CNS demyelination in X-linked adrenoleukodystrophy. Human Mutation, 2006, 27, 1063-1064.	2.5	18
66	Metabolic rerouting via SCD1 induction impacts X-linked adrenoleukodystrophy. Journal of Clinical Investigation, 2021, 131, .	8.2	17
67	Cholesterol-deprivation increases mono-unsaturated very long-chain fatty acids in skin fibroblasts from patients with X-linked adrenoleukodystrophy. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2008, 1781, 105-111.	2.4	16
68	Intellectual Disability and Hemizygous <scp>GPD</scp>2 Mutation. American Journal of Medical Genetics, Part A, 2013, 161, 1044-1050.	1.2	16
69	Pathogenicity of novel ABCD1 variants: The need for biochemical testing in the era of advanced genetics. Molecular Genetics and Metabolism, 2016, 118, 123-127.	1.1	15
70	CYP4F2 affects phenotypic outcome in adrenoleukodystrophy by modulating the clearance of very long-chain fatty acids. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2016, 1862, 1861-1870.	3.8	15
71	Method for Measurement of Peroxisomal Very Long-Chain Fatty Acid Beta-Oxidation and De Novo C26:0 Synthesis Activity in Living Cells Using Stable-Isotope Labeled Docosanoic Acid. Methods in Molecular Biology, 2017, 1595, 45-54.	0.9	14
72	Longitudinal diffusion MRI as surrogate outcome measure for myelopathy in adrenoleukodystrophy. Neurology, 2019, 93, e2133-e2143.	1.1	14

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73	Multi-Omic Approach to Identify Phenotypic Modifiers Underlying Cerebral Demyelination in X-Linked Adrenoleukodystrophy. Frontiers in Cell and Developmental Biology, 2020, 8, 520.	3.7	14
74	Molecular Biomarkers for Adrenoleukodystrophy: An Unmet Need. Cells, 2021, 10, 3427.	4.1	14
75	Two intronic mutations in the adrenoleukodystrophy gene. Human Mutation, 1995, 6, 272-273.	2.5	13
76	Mutational Analysis and the Pathogenesis of Variant X-linked Adrenoleukodystrophy Phenotypes. Archives of Neurology, 1999, 56, 273.	4.5	13
77	X-Linked Adrenoleukodystrophy: Molecular and Functional Analysis of the ABCD1 Gene in Argentinean Patients. PLoS ONE, 2012, 7, e52635.	2.5	13
78	Spinal cord atrophy as a measure of severity of myelopathy in adrenoleukodystrophy. Journal of Inherited Metabolic Disease, 2020, 43, 852-860.	3.6	13
79	Expanding Neonatal Bloodspot Screening: A Multi-Stakeholder Perspective. Frontiers in Pediatrics, 2021, 9, 706394.	1.9	13
80	The gene for X-linked myotubular myopathy is located in an 8 Mb region at the border of Xq27.3 and Xq28. Neuromuscular Disorders, 1994, 4, 455-461.	0.6	11
81	Endocrine dysfunction in adrenoleukodystrophy. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2021, 182, 257-267.	1.8	11
82	A novel cell model to study the function of the adrenoleukodystrophy-related protein. Biochemical and Biophysical Research Communications, 2006, 341, 150-157.	2.1	8
83	Enzymatic diagnosis of SjÃ¶gren-Larsson syndrome using electrospray ionization mass spectrometry. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2009, 877, 451-455.	2.3	8
84	Targeting foam cell formation in inflammatory brain diseases by the histone modifier MSâ€275. Annals of Clinical and Translational Neurology, 2020, 7, 2161-2177.	3.7	8
85	Invariant NKT cells in adrenoleukodystrophy patients and mice. Journal of Neuroimmunology, 2010, 229, 204-211.	2.3	7
86	X-linked adrenomyeloneuropathy due to a novel missense mutation in the ABCD1 start codon presenting as demyelinating neuropathy. Journal of the Peripheral Nervous System, 2011, 16, 353-355.	3.1	7
87	Biochemical Studies in Fibroblasts to Interpret Variants of Unknown Significance in the ABCD1 Gene. Genes, 2021, 12, 1930.	2.4	6
88	Evolution of adrenoleukodystrophy model systems. Journal of Inherited Metabolic Disease, 2021, 44, 544-553.	3.6	5
89	Stability of the ABCD1 Protein with a Missense Mutation: A Novel Approach to Finding Therapeutic Compounds for X-Linked Adrenoleukodystrophy. JIMD Reports, 2018, 44, 23-31.	1.5	4
90	Laboratory Diagnosis of Peroxisomal Disorders in the -Omics Era and the Continued Importance of Biomarkers and Biochemical Studies. FIRE Forum for International Research in Education, 2018, 6, 232640981881028.	0.7	3

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91	The variability conundrum in neurometabolic degenerative diseases. Molecular Genetics and Metabolism, 2020, 131, 367-369.	1.1	3
92	Postural Body Sway as Surrogate Outcome for Myelopathy in Adrenoleukodystrophy. Frontiers in Physiology, 2020, 11, 786.	2.8	3
93	Proteasome-dependent protein quality control of the peroxisomal membrane protein Pxa1p. Biochimica Et Biophysica Acta - Biomembranes, 2020, 1862, 183342.	2.6	3
94	iBRET Screen of the ABCD1 Peroxisomal Network and Mutation-Induced Network Perturbations. Journal of Proteome Research, 2021, 20, 4366-4380.	3.7	3
95	No evidence for 'skewed' inactivation of the X-chromosome as cause of Leber's hereditary optic neuropathy in female carriers. Human Genetics, 1996, 97, 500-505.	3.8	2
96	Peroxisome Metabolism Contributes to PIEZO2-Mediated Mechanical Allodynia. Cells, 2022, 11, 1842.	4.1	2
97	Pharmacological induction of peroxisomes in peroxisome biogenesis disorders. , 2000, 47, 286.		1
98	Corrigendum to â€œElongation of very long-chain fatty acids is enhanced in X-linked adrenoleukodystrophyâ€•[Mol. Genet. Metab. 84 (2005) 144â€“151]. Molecular Genetics and Metabolism, 2008, 93, 350.	1.1	0
99	Clinical utility gene card for: Adrenoleukodystrophy. European Journal of Human Genetics, 2012, 20, 1-3.	2.8	0
100	Functional studies on c.1347C>T, a polymorphism modulating phenotypic outcome in X-linked adrenoleukodystrophy. Tijdschrift Voor Kindergeneeskunde, 2013, 81, 10-10.	0.0	0
101	The X-ALD Mouse 2.0. Tijdschrift Voor Kindergeneeskunde, 2013, 81, 60-60.	0.0	0
102	Endoplasmic reticulum stress signaling in patients with X-linked adrenoleukodystrophy. Tijdschrift Voor Kindergeneeskunde, 2013, 81, 91-91.	0.0	0
103	ELOVL1 is a potential target for therapeutic intervention in X-linked adrenoleukodystrophy. Tijdschrift Voor Kindergeneeskunde, 2013, 81, 102-102.	0.0	0
104	Comment on the paper â€œEffect of statin treatment on adrenomyeloneuropathy with cerebral inflammation: A revisitâ€• Clinical Neurology and Neurosurgery, 2013, 115, 2401-2402.	1.4	0
105	Reply: Age-dependent penetrance among females with X-linked adrenoleukodystrophy. Brain, 2015, 138, e326-e326.	7.6	0
106	ABCD subfamily of peroxisomal ABC transporters (version 2019.4) in the IUPHAR/BPS Guide to Pharmacology Database. IUPHAR/BPS Guide To Pharmacology CITE, 2019, 2019, .	0.2	0