Stephan Kemp

List of Publications by Year
in descending order

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106 papers	8,634 citations	70961 41 h-index	43802 91 g-index
111 all docs	111 docs citations	111 times ranked	9936 citing authors

#	Article	IF	Citations
1	Structure and Function of the ABCD1 Variant Database: 20 Years, 940 Pathogenic Variants, and 3400 Cases of Adrenoleukodystrophy. Cells, 2022, 11, 283.	1.8	23
2	Peroxisome Metabolism Contributes to PIEZO2-Mediated Mechanical Allodynia. Cells, 2022, 11, 1842.	1.8	2
3	Evolution of adrenoleukodystrophy model systems. Journal of Inherited Metabolic Disease, 2021, 44, 544-553.	1.7	5
4	<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.	1.7	39
5	Metabolic rerouting via SCD1 induction impacts X-linked adrenoleukodystrophy. Journal of Clinical Investigation, 2021, 131, .	3.9	17
6	The brain penetrant PPARÎ³ agonist leriglitazone restores multiple altered pathways in models of X-linked adrenoleukodystrophy. Science Translational Medicine, 2021, 13, .	5.8	24
7	iBRET Screen of the ABCD1 Peroxisomal Network and Mutation-Induced Network Perturbations. Journal of Proteome Research, 2021, 20, 4366-4380.	1.8	3
8	THE CONCISE GUIDE TO PHARMACOLOGY 2021/22: Transporters. British Journal of Pharmacology, 2021, 178, S412-S513.	2.7	114
9	Endocrine dysfunction in adrenoleukodystrophy. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2021, 182, 257-267.	1.0	11
10	Expanding Neonatal Bloodspot Screening: A Multi-Stakeholder Perspective. Frontiers in Pediatrics, 2021, 9, 706394.	0.9	13
11	Biochemical Studies in Fibroblasts to Interpret Variants of Unknown Significance in the ABCD1 Gene. Genes, 2021, 12, 1930.	1.0	6
12	Molecular Biomarkers for Adrenoleukodystrophy: An Unmet Need. Cells, 2021, 10, 3427.	1.8	14
13	Plasma NfL and GFAP as biomarkers of spinal cord degeneration in adrenoleukodystrophy. Annals of Clinical and Translational Neurology, 2020, 7, 2127-2136.	1.7	19
14	The variability conundrum in neurometabolic degenerative diseases. Molecular Genetics and Metabolism, 2020, 131, 367-369.	0.5	3
15	Postural Body Sway as Surrogate Outcome for Myelopathy in Adrenoleukodystrophy. Frontiers in Physiology, 2020, 11, 786.	1.3	3
16	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.	1.8	31
17	Vorinostat in the acute neuroinflammatory form of Xâ€linked adrenoleukodystrophy. Annals of Clinical and Translational Neurology, 2020, 7, 639-652.	1.7	19
18	Proteasome-dependent protein quality control of the peroxisomal membrane protein Pxa1p. Biochimica Et Biophysica Acta - Biomembranes, 2020, 1862, 183342.	1.4	3

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19	Adrenoleukodystrophy Newborn Screening in the Netherlands (SCAN Study): The X-Factor. Frontiers in Cell and Developmental Biology, 2020, 8, 499.	1.8	47
20	Multi-Omic Approach to Identify Phenotypic Modifiers Underlying Cerebral Demyelination in X-Linked Adrenoleukodystrophy. Frontiers in Cell and Developmental Biology, 2020, 8, 520.	1.8	14
21	Spinal cord atrophy as a measure of severity of myelopathy in adrenoleukodystrophy. Journal of Inherited Metabolic Disease, 2020, 43, 852-860.	1.7	13
22	Targeting foam cell formation in inflammatory brain diseases by the histone modifier MSâ€275. Annals of Clinical and Translational Neurology, 2020, 7, 2161-2177.	1.7	8
23	THE CONCISE GUIDE TO PHARMACOLOGY 2019/20: Transporters. British Journal of Pharmacology, 2019, 176, S397-S493.	2.7	166
24	Disease progression in women with X-linked adrenoleukodystrophy is slow. Orphanet Journal of Rare Diseases, 2019, 14, 30.	1.2	58
25	Longitudinal diffusion MRI as surrogate outcome measure for myelopathy in adrenoleukodystrophy. Neurology, 2019, 93, e2133-e2143.	1.5	14
26	Progression of myelopathy in males with adrenoleukodystrophy: towards clinical trial readiness. Brain, 2019, 142, 334-343.	3.7	43
27	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.	1.7	20
28	Intrathecal Adeno-Associated Viral Vector-Mediated Gene Delivery for Adrenomyeloneuropathy. Human Gene Therapy, 2019, 30, 544-555.	1.4	21
29	The Natural History of Adrenal Insufficiency in X-Linked Adrenoleukodystrophy: An International Collaboration. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 118-126.	1.8	102
30	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
31	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
32	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.	0.7	4
33	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.	0.4	14
34	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.	0.5	50
35	THE CONCISE GUIDE TO PHARMACOLOGY 2017/18: Overview. British Journal of Pharmacology, 2017, 174, S1-S16.	2.7	269
36	Lipid-induced endoplasmic reticulum stress in X-linked adrenoleukodystrophy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 2255-2265.	1.8	26

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37	C26:0-Carnitine Is a New Biomarker for X-Linked Adrenoleukodystrophy in Mice and Man. PLoS ONE, 2016, 11, e0154597.	1.1	33
38	Pathogenicity of novel ABCD1 variants: The need for biochemical testing in the era of advanced genetics. Molecular Genetics and Metabolism, 2016, 118, 123-127.	0.5	15
39	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.	1.8	15
40	Adrenoleukodystrophy â€“ neuroendocrine pathogenesis and redefinition of natural history. Nature Reviews Endocrinology, 2016, 12, 606-615.	4.3	189
41	Betulinic acid induces a novel cell death pathway that depends on cardiolipin modification. Oncogene, 2016, 35, 427-437.	2.6	57
42	Hematopoietic cell transplantation does not prevent myelopathy in Xâ€linked adrenoleukodystrophy: a retrospective study. Journal of Inherited Metabolic Disease, 2015, 38, 359-361.	1.7	54
43	Reply: Age-dependent penetrance among females with X-linked adrenoleukodystrophy. Brain, 2015, 138, e326-e326.	3.7	0
44	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.	1.2	26
45	X-linked adrenoleukodystrophy in women: a cross-sectional cohort study. Brain, 2014, 137, 693-706.	3.7	182
46	X-Linked Adrenoleukodystrophy: Pathogenesis and Treatment. Current Neurology and Neuroscience Reports, 2014, 14, 486.	2.0	127
47	Intellectual Disability and Hemizygous <scp>GPD</scp>2 Mutation. American Journal of Medical Genetics, Part A, 2013, 161, 1044-1050.	0.7	16
48	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
49	The X-ALD Mouse 2.0. Tijdschrift Voor Kindergeneeskunde, 2013, 81, 60-60.	0.0	0
50	Endoplasmic reticulum stress signaling in patients with X-linked adrenoleukodystrophy. Tijdschrift Voor Kindergeneeskunde, 2013, 81, 91-91.	0.0	0
51	ELOVL1 is a potential target for therapeutic intervention in X-linked adrenoleukodystrophy. Tijdschrift Voor Kindergeneeskunde, 2013, 81, 102-102.	0.0	0
52	Comment on the paper â€œEffect of statin treatment on adrenomyeloneuropathy with cerebral inflammation: A revisitâ€. Clinical Neurology and Neurosurgery, 2013, 115, 2401-2402.	0.6	0
53	Clinical utility gene card for: Adrenoleukodystrophy. European Journal of Human Genetics, 2012, 20, 1-3.	1.4	0
54	A novel defect of peroxisome division due to a homozygous non-sense mutation in thePEX11Î²gene. Journal of Medical Genetics, 2012, 49, 307-313.	1.5	127

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55	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.	1.7	39
56	X-linked adrenoleukodystrophy: Clinical, metabolic, genetic and pathophysiological aspects. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1465-1474.	1.8	217
57	X-linked adrenoleukodystrophy (X-ALD): clinical presentation and guidelines for diagnosis, follow-up and management. Orphanet Journal of Rare Diseases, 2012, 7, 51.	1.2	403
58	Bezafibrate for X-Linked Adrenoleukodystrophy. PLoS ONE, 2012, 7, e41013.	1.1	26
59	X-Linked Adrenoleukodystrophy: Molecular and Functional Analysis of the ABCD1 Gene in Argentinean Patients. PLoS ONE, 2012, 7, e52635.	1.1	13
60	Mammalian peroxisomal ABC transporters: from endogenous substrates to pathology and clinical significance. British Journal of Pharmacology, 2011, 164, 1753-1766.	2.7	93
61	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.	1.4	7
62	Fatty acid omegaâ€oxidation as a rescue pathway for fatty acid oxidation disorders in humans. FEBS Journal, 2011, 278, 182-194.	2.2	181
63	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.	1.7	41
64	Biochemical Aspects of Xâ€Linked Adrenoleukodystrophy. Brain Pathology, 2010, 20, 831-837.	2.1	83
65	Invariant NKT cells in adrenoleukodystrophy patients and mice. Journal of Neuroimmunology, 2010, 229, 204-211.	1.1	7
66	The role of ELOVL1 in very longâ€chain fatty acid homeostasis and Xâ€linked adrenoleukodystrophy. EMBO Molecular Medicine, 2010, 2, 90-97.	3.3	140
67	Valproic acid induces antioxidant effects in X-linked adrenoleukodystrophy. Human Molecular Genetics, 2010, 19, 2005-2014.	1.4	90
68	Lovastatin in X-Linked Adrenoleukodystrophy. New England Journal of Medicine, 2010, 362, 276-277.	13.9	58
69	Peroxisomes, lipid metabolism and lipotoxicity. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2010, 1801, 272-280.	1.2	135
70	Contiguous gene deletion of ELOVL7, ERCC8 and NDUFAF2 in a patient with a fatal multisystem disorder. Human Molecular Genetics, 2009, 18, 3365-3374.	1.4	30
71	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.	1.2	8
72	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.	0.5	0

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73	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.	1.2	16
74	Early oxidative damage underlying neurodegeneration in X-adrenoleukodystrophy. Human Molecular Genetics, 2008, 17, 1762-1773.	1.4	181
75	Characterization of the human Ï‰â€oxidation pathway for Ï‰â€hydroxyâ€veryâ€longâ€chain fatty acids. FASEB Journal, 2008, 22, 2064-2071.	0.2	46
76	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.	0.5	73
77	The peroxisomal ABC transporter family. Pflugers Archiv European Journal of Physiology, 2007, 453, 719-734.	1.3	95
78	A novel cell model to study the function of the adrenoleukodystrophy-related protein. Biochemical and Biophysical Research Communications, 2006, 341, 150-157.	1.0	8
79	The cystathionine beta-synthase variant c.844_845ins68 protects against CNS demyelination in X-linked adrenoleukodystrophy. Human Mutation, 2006, 27, 1063-1064.	1.1	18
80	Ï‰-Oxidation of Very Long-chain Fatty Acids in Human Liver Microsomes. Journal of Biological Chemistry, 2006, 281, 13180-13187.	1.6	69
81	Methionine metabolism and phenotypic variability in X-linked adrenoleukodystrophy. Neurology, 2006, 66, 442-443.	1.5	20
82	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.	1.4	90
83	Evidence for two enzymatic pathways for Ï‰-oxidation of docosanoic acid in rat liver microsomes. Journal of Lipid Research, 2005, 46, 1001-1008.	2.0	37
84	Accumulation of very long-chain fatty acids does not affect mitochondrial function in adrenoleukodystrophy protein deficiency. Human Molecular Genetics, 2005, 14, 1127-1137.	1.4	32
85	Gene expression profiling in response to the histone deacetylase inhibitor BL1521 in neuroblastoma. Experimental Cell Research, 2005, 309, 451-467.	1.2	38
86	Elongation of very long-chain fatty acids is enhanced in X-linked adrenoleukodystrophy. Molecular Genetics and Metabolism, 2005, 84, 144-151.	0.5	83
87	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.	1.5	38
88	The novel histone deacetylase inhibitor BL1521 inhibits proliferation and induces apoptosis in neuroblastoma cells. Biochemical Pharmacology, 2004, 68, 1279-1288.	2.0	46
89	Histone deacetylases (HDACs): characterization of the classical HDAC family. Biochemical Journal, 2003, 370, 737-749.	1.7	2,671
90	Analysis of very long-chain fatty acids using electrospray ionization mass spectrometry. Molecular Genetics and Metabolism, 2003, 79, 189-196.	0.5	138

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91	A very long-chain acyl-CoA synthetase-deficient mouse and its relevance to X-linked adrenoleukodystrophy. Human Molecular Genetics, 2003, 12, 1145-1154.	1.4	82
92	Mouse Very Long-chain Acyl-CoA Synthetase in X-linked Adrenoleukodystrophy. Journal of Biological Chemistry, 2002, 277, 28765-28773.	1.6	26
93	ABCD1 mutations and the X-linked adrenoleukodystrophy mutation database: Role in diagnosis and clinical correlations. Human Mutation, 2001, 18, 499-515.	1.1	261
94	Pharmacological induction of peroxisomes in peroxisome biogenesis disorders. Annals of Neurology, 2000, 47, 286-296.	2.8	67
95	Pharmacological induction of peroxisomes in peroxisome biogenesis disorders. , 2000, 47, 286.		1
96	Mutational Analysis and the Pathogenesis of Variant X-linked Adrenoleukodystrophy Phenotypes. Archives of Neurology, 1999, 56, 273.	4.9	13
97	X-linked adrenoleukodystrophy: genes, mutations, and phenotypes. Neurochemical Research, 1999, 24, 521-535.	1.6	162
98	Role of very-long-chain acyl-coenzyme A synthetase in X-linked adrenoleukodystrophy. Annals of Neurology, 1999, 46, 409-412.	2.8	48
99	Gene redundancy and pharmacological gene therapy: Implications for X-linked adrenoleukodystrophy. Nature Medicine, 1998, 4, 1261-1268.	15.2	237
100	Connexin32 gene mutations in X-linked dominant Charcot-Marie-Tooth disease (CMTX1). Human Genetics, 1997, 99, 501-505.	1.8	55
101	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.	1.8	35
102	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.	1.8	2
103	Two intronic mutations in the adrenoleukodystrophy gene. Human Mutation, 1995, 6, 272-273.	1.1	13
104	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.	1.0	70
105	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.	0.3	11
106	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.	9.4	215