Florian S Eichler

List of Publications by Year
in descending order

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70	3,714	147801	133252
papers	citations	31 h-index	59 g-index

73	73	73	5161
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Hematopoietic Stem-Cell Gene Therapy for Cerebral Adrenoleukodystrophy. New England Journal of Medicine, 2017, 377, 1630-1638.	27.0	412
2	Cerebral White Matter. Annals of the New York Academy of Sciences, 2008, 1142, 266-309.	3.8	410
3	Hereditary Sensory Neuropathy Type 1 Is Caused by the Accumulation of Two Neurotoxic Sphingolipids. Journal of Biological Chemistry, 2010, 285, 11178-11187.	3.4	320
4	Case definition and classification of leukodystrophies and leukoencephalopathies. Molecular Genetics and Metabolism, 2015, 114, 494-500.	1.1	185
5	Oral l-serine supplementation reduces production of neurotoxic deoxysphingolipids in mice and humans with hereditary sensory autonomic neuropathy type 1. Journal of Clinical Investigation, 2011, 121, 4735-4745.	8.2	172
6	Serine and Lipid Metabolism in Macular Disease and Peripheral Neuropathy. New England Journal of Medicine, 2019, 381, 1422-1433.	27.0	166
7	Is microglial apoptosis an early pathogenic change in cerebral Xâ€linked adrenoleukodystrophy?. Annals of Neurology, 2008, 63, 729-742.	5.3	140
8	Proton MR Spectroscopic and Diffusion Tensor Brain MR Imaging in X-linked Adrenoleukodystrophy: Initial Experience. Radiology, 2002, 225, 245-252.	7.3	133
9	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
10	The genetic landscape of X-linked adrenoleukodystrophy: inheritance, mutations, modifier genes, and diagnosis. The Application of Clinical Genetics, 2015, 8, 109.	3.0	96
11	Overexpression of the Wild-Type SPT1 Subunit Lowers Desoxysphingolipid Levels and Rescues the Phenotype of HSAN1. Journal of Neuroscience, 2009, 29, 14646-14651.	3.6	87
12	Diagnosis, treatment, and clinical outcomes in 43 cases with cerebrotendinous xanthomatosis. Journal of Clinical Lipidology, 2018, 12, 1169-1178.	1.5	83
13	Randomized trial of <scp>l</scp> -serine in patients with hereditary sensory and autonomic neuropathy type 1. Neurology, 2019, 92, e359-e370.	1.1	83
14	Leukodystrophies. Neurologist, 2009, 15, 319-328.	0.7	82
15	Brain endothelial dysfunction in cerebral adrenoleukodystrophy. Brain, 2015, 138, 3206-3220.	7.6	61
16	Magnetic Resonance Imaging Detection of Lesion Progression in Adult Patients With X-linked Adrenoleukodystrophy. Archives of Neurology, 2007, 64, 659.	4.5	55
17	Arginine:glycine amidinotransferase (AGAT) deficiency: Clinical features and long term outcomes in 16 patients diagnosed worldwide. Molecular Genetics and Metabolism, 2015, 116, 252-259.	1.1	55
18	CSF1Rmosaicism in a family with hereditary diffuse leukoencephalopathy with spheroids. Brain, 2016, 139, 1666-1672.	7.6	53

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19	Adenoassociated Virus Serotype 9-Mediated Gene Therapy for X-Linked Adrenoleukodystrophy. Molecular Therapy, 2015, 23, 824-834.	8.2	51
20	Disease specific therapies in leukodystrophies and leukoencephalopathies. Molecular Genetics and Metabolism, 2015, 114, 527-536.	1.1	45
21	Revised consensus statement on the preventive and symptomatic care of patients with leukodystrophies. Molecular Genetics and Metabolism, 2017, 122, 18-32.	1.1	42
22	Natural history of neurological abnormalities in cerebrotendinous xanthomatosis. Journal of Inherited Metabolic Disease, 2018, 41, 647-656.	3.6	41
23	MRI brain lesions in asymptomatic boys with X-linked adrenoleukodystrophy. Neurology, 2019, 92, e1698-e1708.	1.1	41
24	<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
25	Immune Response in Leukodystrophies. Pediatric Neurology, 2007, 37, 235-244.	2.1	38
26	The Changing Face of Adrenoleukodystrophy. Endocrine Reviews, 2020, 41, 577-593.	20.1	38
27	Altered sphingoid base profiles in type 1 compared to type 2 diabetes. Lipids in Health and Disease, 2014, 13, 161.	3.0	37
28	Microglial dysfunction as a key pathological change in adrenomyeloneuropathy. Annals of Neurology, 2017, 82, 813-827.	5.3	37
29	Hematopoietic Stem Cell Transplantation in the Leukodystrophies: A Systematic Review of the Literature. Neuropediatrics, 2014, 45, 169-174.	0.6	35
30	Seven-Tesla Proton Magnetic Resonance Spectroscopic Imaging in Adult X-Linked Adrenoleukodystrophy. Archives of Neurology, 2008, 65, 1488.	4.5	34
31	Magnetic Resonance Imaging Evaluation of Possible Neonatal Sinovenous Thrombosis. Pediatric Neurology, 2007, 37, 317-323.	2.1	33
32	Natural history and biomarkers in hereditary sensory neuropathy type 1. Muscle and Nerve, 2015, 51, 489-495.	2.2	33
33	Neurofilament light chain as a potential biomarker for monitoring neurodegeneration in X-linked adrenoleukodystrophy. Nature Communications, 2021, 12, 1816.	12.8	33
34	Complex care of individuals with multiple sulfatase deficiency: Clinical cases and consensus statement. Molecular Genetics and Metabolism, 2018, 123, 337-346.	1.1	31
35	CSF and Blood Levels of GFAP in Alexander Disease. ENeuro, 2015, 2, ENEURO.0080-15.2015.	1.9	30
36	Neurocognitive clinical outcome assessments for inborn errors of metabolism and other rare conditions. Molecular Genetics and Metabolism, 2016, 118, 65-69.	1.1	28

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37	Metachromatic Leukodystrophy. Journal of Child Neurology, 2016, 31, 1457-1463.	1.4	26
38	The Landscape of Hematopoietic Stem Cell Transplant and Gene Therapy for X-Linked Adrenoleukodystrophy. Current Treatment Options in Neurology, 2019, 21, 61.	1.8	25
39	ABCD1 dysfunction alters white matter microvascular perfusion. Brain, 2017, 140, 3139-3152.	7.6	24
40	Adult-Onset Leukoencephalopathy With Axonal Spheroids and Pigmented Glia: Review of Clinical Manifestations as Foundations for Therapeutic Development. Frontiers in Neurology, 2021, 12, 788168.	2.4	24
41	Intrathecal Adeno-Associated Viral Vector-Mediated Gene Delivery for Adrenomyeloneuropathy. Human Gene Therapy, 2019, 30, 544-555.	2.7	21
42	Clinical and radiographic course of arrested cerebral adrenoleukodystrophy. Neurology, 2020, 94, e2499-e2507.	1.1	21
43	Proton Magnetic Resonance Spectroscopy and Diffusion-Weighted Imaging in Isolated Sulfite Oxidase Deficiency. Journal of Child Neurology, 2006, 21, 801-805.	1.4	20
44	5,10-methenyltetrahydrofolate synthetase deficiency causes a neurometabolic disorder associated with microcephaly, epilepsy, and cerebral hypomyelination. Molecular Genetics and Metabolism, 2018, 125, 118-126.	1.1	18
45	Substrate Availability of Mutant SPT Alters Neuronal Branching and Growth Cone Dynamics in Dorsal Root Ganglia. Journal of Neuroscience, 2015, 35, 13713-13719.	3.6	17
46	Quantitative oculomotor and nonmotor assessments in late-onset GM2 gangliosidosis. Neurology, 2020, 94, e705-e717.	1.1	17
47	Metabolic rerouting via SCD1 induction impacts X-linked adrenoleukodystrophy. Journal of Clinical Investigation, 2021, 131, .	8.2	17
48	The AAV9 Variant Capsid AAV-F Mediates Widespread Transgene Expression in Nonhuman Primate Spinal Cord After Intrathecal Administration. Human Gene Therapy, 2022, 33, 61-75.	2.7	16
49	Neutralizing Antibody Evasion and Transduction with Purified Extracellular Vesicle-Enveloped Adeno-Associated Virus Vectors. Human Gene Therapy, 2021, 32, 1457-1470.	2.7	16
50	Clinical myopathy in patients with nephropathic cystinosis. Muscle and Nerve, 2020, 61, 74-80.	2.2	15
51	Endocrine dysfunction in adrenoleukodystrophy. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2021, 182, 257-267.	1.8	11
52	The natural history of Canavan disease: 23 new cases and comparison with patients from literature. Orphanet Journal of Rare Diseases, 2021, 16, 227.	2.7	9
53	Inherited or acquired metabolic disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2016, 135, 603-636.	1.8	7
54	Beyond gait and balance: urinary and bowel dysfunction in X-linked adrenoleukodystrophy. Orphanet Journal of Rare Diseases, 2021, 16, 14.	2.7	7

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55	Practical Approaches and Knowledge Gaps in the Care for Children With Leukodystrophies. Journal of Child Neurology, 2021, 36, 65-78.	1.4	6
56	Clinical trial readiness study of distal myopathy and dysphagia in nephropathic cystinosis. Muscle and Nerve, 2020, 62, 681-687.	2.2	5
57	X-linked adrenoleukodystrophy in a chimpanzee due to an ABCD1 mutation reported in multiple unrelated humans. Molecular Genetics and Metabolism, 2017, 122, 130-133.	1.1	5
58	Restless Legs Syndrome in X-linked adrenoleukodystrophy. Sleep Medicine, 2022, 91, 31-34.	1.6	5
59	Case 38-2017. New England Journal of Medicine, 2017, 377, 2376-2385.	27.0	4
60	Gene Therapy for Cerebral Adrenoleukodystrophy. New England Journal of Medicine, 2018, 378, 490-491.	27.0	2
61	Gait Difficulties and Postural Instability in Adrenoleukodystrophy. Frontiers in Neurology, 2021, 12, 684102.	2.4	2
62	A Longitudinal Analysis of Early Lesion Growth in Presymptomatic Patients with Cerebral Adrenoleukodystrophy. American Journal of Neuroradiology, 2021, 42, 1904-1911.	2.4	2
63	Longitudinal dysphagia assessment in adult patients with nephropathic cystinosis using the Modified Barium Swallow Impairment Profile. Muscle and Nerve, 2022, 66, 223-226.	2.2	2
64	Feasibility of simultaneous highâ€resolution anatomical and quantitative <scp>magnetic resonance</scp> imaging of sciatic nerves in patients with <scp>Charcotâ€“Marieâ€“Tooth</scp> type <scp>1A</scp> (<scp>CMT1A</scp>) at <scp>7T</scp>. Muscle and Nerve, 2022, 66, 206-211.	2.2	2
65	Peroxisome Metabolism Contributes to PIEZO2-Mediated Mechanical Allodynia. Cells, 2022, 11, 1842.	4.1	2
66	Therapeutics of X-linked adrenoleukodystrophy. Drug Discovery Today: Therapeutic Strategies, 2008, 5, 237-242.	0.5	1
67	Hematopoietic Stem Cell Transplantation in the Leukodystrophies: A Systematic Review of the Literature. Neuropediatrics, 2014, 45, e1-e1.	0.6	0
68	Teaching Neuro Images : Vanishing white matter ovarioleukodystrophy. Neurology, 2016, 86, e248.	1.1	0
69	Unusual Behaviors in a 7-year-old Boy. Pediatrics in Review, 2021, 42, S122-S125.	0.4	0
70	Neonatal adrenoleukodystrophy with longâ€term survival. FASEB Journal, 2007, 21, A401.	0.5	0