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	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
2	Restless Legs Syndrome in X-linked adrenoleukodystrophy. Sleep Medicine, 2022, 91, 31-34.	1.6	5
3	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
4	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
5	Peroxisome Metabolism Contributes to PIEZO2-Mediated Mechanical Allodynia. Cells, 2022, 11, 1842.	4.1	2
6	Practical Approaches and Knowledge Gaps in the Care for Children With Leukodystrophies. Journal of Child Neurology, 2021, 36, 65-78.	1.4	6
7	Unusual Behaviors in a 7-year-old Boy. Pediatrics in Review, 2021, 42, S122-S125.	0.4	0
8	Beyond gait and balance: urinary and bowel dysfunction in X-linked adrenoleukodystrophy. Orphanet Journal of Rare Diseases, 2021, 16, 14.	2.7	7
9	<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
10	Neurofilament light chain as a potential biomarker for monitoring neurodegeneration in X-linked adrenoleukodystrophy. Nature Communications, 2021, 12, 1816.	12.8	33
11	Metabolic rerouting via SCD1 induction impacts X-linked adrenoleukodystrophy. Journal of Clinical Investigation, 2021, 131, .	8.2	17
12	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
13	Gait Difficulties and Postural Instability in Adrenoleukodystrophy. Frontiers in Neurology, 2021, 12, 684102.	2.4	2
14	Neutralizing Antibody Evasion and Transduction with Purified Extracellular Vesicle-Enveloped Adeno-Associated Virus Vectors. Human Gene Therapy, 2021, 32, 1457-1470.	2.7	16
15	A Longitudinal Analysis of Early Lesion Growth in Presymptomatic Patients with Cerebral Adrenoleukodystrophy. American Journal of Neuroradiology, 2021, 42, 1904-1911.	2.4	2
16	Endocrine dysfunction in adrenoleukodystrophy. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2021, 182, 257-267.	1.8	11
17	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
18	Clinical myopathy in patients with nephropathic cystinosis. Muscle and Nerve, 2020, 61, 74-80.	2.2	15

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19	Clinical trial readiness study of distal myopathy and dysphagia in nephropathic cystinosis. Muscle and Nerve, 2020, 62, 681-687.	2.2	5
20	The Changing Face of Adrenoleukodystrophy. Endocrine Reviews, 2020, 41, 577-593.	20.1	38
21	Clinical and radiographic course of arrested cerebral adrenoleukodystrophy. Neurology, 2020, 94, e2499-e2507.	1.1	21
22	Quantitative oculomotor and nonmotor assessments in late-onset GM2 gangliosidosis. Neurology, 2020, 94, e705-e717.	1.1	17
23	Serine and Lipid Metabolism in Macular Disease and Peripheral Neuropathy. New England Journal of Medicine, 2019, 381, 1422-1433.	27.0	166
24	MRI brain lesions in asymptomatic boys with X-linked adrenoleukodystrophy. Neurology, 2019, 92, e1698-e1708.	1.1	41
25	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
26	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
27	Intrathecal Adeno-Associated Viral Vector-Mediated Gene Delivery for Adrenomyeloneuropathy. Human Gene Therapy, 2019, 30, 544-555.	2.7	21
28	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
29	Natural history of neurological abnormalities in cerebrotendinous xanthomatosis. Journal of Inherited Metabolic Disease, 2018, 41, 647-656.	3.6	41
30	Complex care of individuals with multiple sulfatase deficiency: Clinical cases and consensus statement. Molecular Genetics and Metabolism, 2018, 123, 337-346.	1.1	31
31	Gene Therapy for Cerebral Adrenoleukodystrophy. New England Journal of Medicine, 2018, 378, 490-491.	27.0	2
32	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
33	Diagnosis, treatment, and clinical outcomes in 43 cases with cerebrotendinous xanthomatosis. Journal of Clinical Lipidology, 2018, 12, 1169-1178.	1.5	83
34	Hematopoietic Stem-Cell Gene Therapy for Cerebral Adrenoleukodystrophy. New England Journal of Medicine, 2017, 377, 1630-1638.	27.0	412
35	Microglial dysfunction as a key pathological change in adrenomyeloneuropathy. Annals of Neurology, 2017, 82, 813-827.	5.3	37
36	Revised consensus statement on the preventive and symptomatic care of patients with leukodystrophies. Molecular Genetics and Metabolism, 2017, 122, 18-32.	1.1	42

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37	Case 38-2017. New England Journal of Medicine, 2017, 377, 2376-2385.	27.0	4
38	ABCD1 dysfunction alters white matter microvascular perfusion. Brain, 2017, 140, 3139-3152.	7.6	24
39	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
40	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
41	Neurocognitive clinical outcome assessments for inborn errors of metabolism and other rare conditions. Molecular Genetics and Metabolism, 2016, 118, 65-69.	1.1	28
42	CSF1Rmosaicism in a family with hereditary diffuse leukoencephalopathy with spheroids. Brain, 2016, 139, 1666-1672.	7.6	53
43	Metachromatic Leukodystrophy. Journal of Child Neurology, 2016, 31, 1457-1463.	1.4	26
44	Teaching Neuro Images : Vanishing white matter ovarioleukodystrophy. Neurology, 2016, 86, e248.	1.1	0
45	The genetic landscape of X-linked adrenoleukodystrophy: inheritance, mutations, modifier genes, and diagnosis. The Application of Clinical Genetics, 2015, 8, 109.	3.0	96
46	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
47	Natural history and biomarkers in hereditary sensory neuropathy type 1. Muscle and Nerve, 2015, 51, 489-495.	2.2	33
48	Disease specific therapies in leukodystrophies and leukoencephalopathies. Molecular Genetics and Metabolism, 2015, 114, 527-536.	1.1	45
49	Case definition and classification of leukodystrophies and leukoencephalopathies. Molecular Genetics and Metabolism, 2015, 114, 494-500.	1.1	185
50	Adenoassociated Virus Serotype 9-Mediated Gene Therapy for X-Linked Adrenoleukodystrophy. Molecular Therapy, 2015, 23, 824-834.	8.2	51
51	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
52	Brain endothelial dysfunction in cerebral adrenoleukodystrophy. Brain, 2015, 138, 3206-3220.	7.6	61
53	CSF and Blood Levels of GFAP in Alexander Disease. ENeuro, 2015, 2, ENEURO.0080-15.2015.	1.9	30
54	Altered sphingoid base profiles in type 1 compared to type 2 diabetes. Lipids in Health and Disease, 2014, 13, 161.	3.0	37

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55	Hematopoietic Stem Cell Transplantation in the Leukodystrophies: A Systematic Review of the Literature. Neuropediatrics, 2014, 45, 169-174.	0.6	35
56	Hematopoietic Stem Cell Transplantation in the Leukodystrophies: A Systematic Review of the Literature. Neuropediatrics, 2014, 45, e1-e1.	0.6	0
57	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
58	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
59	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
60	Leukodystrophies. Neurologist, 2009, 15, 319-328.	0.7	82
61	Is microglial apoptosis an early pathogenic change in cerebral Xâ€linked adrenoleukodystrophy?. Annals of Neurology, 2008, 63, 729-742.	5.3	140
62	Cerebral White Matter. Annals of the New York Academy of Sciences, 2008, 1142, 266-309.	3.8	410
63	Therapeutics of X-linked adrenoleukodystrophy. Drug Discovery Today: Therapeutic Strategies, 2008, 5, 237-242.	0.5	1
64	Seven-Tesla Proton Magnetic Resonance Spectroscopic Imaging in Adult X-Linked Adrenoleukodystrophy. Archives of Neurology, 2008, 65, 1488.	4.5	34
65	Magnetic Resonance Imaging Detection of Lesion Progression in Adult Patients With X-linked Adrenoleukodystrophy. Archives of Neurology, 2007, 64, 659.	4.5	55
66	Immune Response in Leukodystrophies. Pediatric Neurology, 2007, 37, 235-244.	2.1	38
67	Magnetic Resonance Imaging Evaluation of Possible Neonatal Sinovenous Thrombosis. Pediatric Neurology, 2007, 37, 317-323.	2.1	33
68	Neonatal adrenoleukodystrophy with longâ€term survival. FASEB Journal, 2007, 21, A401.	0.5	0
69	Proton Magnetic Resonance Spectroscopy and Diffusion-Weighted Imaging in Isolated Sulfite Oxidase Deficiency. Journal of Child Neurology, 2006, 21, 801-805.	1.4	20
70	Proton MR Spectroscopic and Diffusion Tensor Brain MR Imaging in X-linked Adrenoleukodystrophy: Initial Experience. Radiology, 2002, 225, 245-252.	7.3	133