Fanny Mochel

List of Publications by Year
in descending order

113 papers	4,524 citations	94381 37 h-index	118793 62 g-index
116 all docs	116 docs citations	116 times ranked	6996 citing authors

#	Article	IF	Citations
1	Early Energy Deficit in Huntington Disease: Identification of a Plasma Biomarker Traceable during Disease Progression. PLoS ONE, 2007, 2, e647.	1.1	202
2	Case definition and classification of leukodystrophies and leukoencephalopathies. Molecular Genetics and Metabolism, 2015, 114, 494-500.	0.5	185
3	Energy deficit in Huntington disease: why it matters. Journal of Clinical Investigation, 2011, 121, 493-499.	3.9	182
4	Alteration of Fatty-Acid-Metabolizing Enzymes Affects Mitochondrial Form and Function in Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2012, 91, 1051-1064.	2.6	179
5	Cognitive and neuroimaging features and brain Î²-amyloidosis in individuals at risk of Alzheimer's disease (INSIGHT-preAD): a longitudinal observational study. Lancet Neurology, The, 2018, 17, 335-346.	4.9	161
6	Alteration of Ganglioside Biosynthesis Responsible for Complex Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2013, 93, 118-123.	2.6	151
7	Genetic Analysis of Inherited Leukodystrophies. JAMA Neurology, 2013, 70, 875.	4.5	147
8	Pyruvate carboxylase deficiency: clinical and biochemical response to anaplerotic diet therapy. Molecular Genetics and Metabolism, 2005, 84, 305-312.	0.5	127
9	Adult polyglucosan body disease: Natural History and Key Magnetic Resonance Imaging Findings. Annals of Neurology, 2012, 72, 433-441.	2.8	125
10	Two-site reproducibility of cerebellar and brainstem neurochemical profiles with short-echo, single-voxel MRS at 3T. Magnetic Resonance in Medicine, 2015, 73, 1718-1725.	1.9	117
11	Anaplerotic diet therapy in inherited metabolic disease: Therapeutic potential. Journal of Inherited Metabolic Disease, 2006, 29, 332-340.	1.7	113
12	Diagnosis, prognosis, and treatment of leukodystrophies. Lancet Neurology, The, 2019, 18, 962-972.	4.9	106
13	ATAD3 gene cluster deletions cause cerebellar dysfunction associated with altered mitochondrial DNA and cholesterol metabolism. Brain, 2017, 140, 1595-1610.	3.7	105
14	Early Alterations of Brain Cellular Energy Homeostasis in Huntington Disease Models. Journal of Biological Chemistry, 2012, 287, 1361-1370.	1.6	104
15	Disorders of phospholipids, sphingolipids and fatty acids biosynthesis: toward a new category of inherited metabolic diseases. Journal of Inherited Metabolic Disease, 2013, 36, 411-425.	1.7	85
16	Autosomal Recessive Cerebellar Ataxias: Paving the Way toward Targeted Molecular Therapies. Neuron, 2019, 101, 560-583.	3.8	83
17	Triheptanoin improves brain energy metabolism in patients with Huntington disease. Neurology, 2015, 84, 490-495.	1.5	82
18	Relapsing encephalopathy with cerebellar ataxia related to an <scp>ATP</scp>1A3 mutation. Developmental Medicine and Child Neurology, 2015, 57, 1183-1186.	1.1	78

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19	Autosomal dominant cerebellar ataxias: Imaging biomarkers with high effect sizes. NeuroImage: Clinical, 2018, 19, 858-867.	1.4	78
20	Triheptanoin dramatically reduces paroxysmal motor disorder in patients with GLUT1 deficiency. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 550-553.	0.9	73
21	Long-term cardiac prognosis and risk stratification in 260 adults presenting with mitochondrial diseases. European Heart Journal, 2015, 36, 2886-2893.	1.0	71
22	GLUT1 deficiency syndrome: An update. Revue Neurologique, 2014, 170, 91-99.	0.6	67
23	Plasma oxysterols: biomarkers for diagnosis and treatment in spastic paraplegia type 5. Brain, 2018, 141, 72-84.	3.7	67
24	In vivo neurometabolic profiling in patients with spinocerebellar ataxia types 1, 2, 3, and 7. Movement Disorders, 2015, 30, 662-670.	2.2	63
25	Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy. American Journal of Human Genetics, 2017, 100, 151-159.	2.6	63
26	Inhibition of Lysosome Membrane Recycling Causes Accumulation of Gangliosides that Contribute to Neurodegeneration. Cell Reports, 2018, 23, 3813-3826.	2.9	63
27	New practical definitions for the diagnosis of autosomal recessive spastic ataxia of <scp>C</scp>harlevoixâ€“<scp>S</scp>aguenay. Annals of Neurology, 2015, 78, 871-886.	2.8	62
28	Natural history of cerebrotendinous xanthomatosis: a paediatric disease diagnosed in adulthood. Orphanet Journal of Rare Diseases, 2016, 11, 41.	1.2	55
29	An overview of inborn errors of complex lipid biosynthesis and remodelling. Journal of Inherited Metabolic Disease, 2015, 38, 3-18.	1.7	54
30	Phenotype, treatment practice and outcome in the cobalaminâ€dependent remethylation disorders and MTHFR deficiency: Data from the Eâ€HOD registry. Journal of Inherited Metabolic Disease, 2019, 42, 333-352.	1.7	53
31	Dietary anaplerotic therapy improves peripheral tissue energy metabolism in patients with Huntington's disease. European Journal of Human Genetics, 2010, 18, 1057-1060.	1.4	50
32	Metabolic and Organelle Morphology Defects in Mice and Human Patients Define Spinocerebellar Ataxia Type 7 as a Mitochondrial Disease. Cell Reports, 2019, 26, 1189-1202.e6.	2.9	49
33	Loss of paraplegin drives spasticity rather than ataxia in a cohort of 241 patients with SPG7. Neurology, 2019, 92, e2679-e2690.	1.5	49
34	Altered Dopamine and Serotonin Metabolism in Motorically Asymptomatic R6/2 Mice. PLoS ONE, 2011, 6, e18336.	1.1	45
35	Abnormal response to cortical activation in early stages of Huntington disease. Movement Disorders, 2012, 27, 907-910.	2.2	44
36	The clinical spectrum of inherited diseases involved in the synthesis and remodeling of complex lipids. A tentative overview. Journal of Inherited Metabolic Disease, 2015, 38, 19-40.	1.7	44

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37	A strategy for multimodal data integration: application to biomarkers identification in spinocerebellar ataxia. Briefings in Bioinformatics, 2018, 19, 1356-1369.	3.2	44
38	Treatment with chenodeoxycholic acid in cerebrotendinous xanthomatosis: clinical, neurophysiological, and quantitative brain structural outcomes. Journal of Inherited Metabolic Disease, 2018, 41, 799-807.	1.7	43
39	Lipids in the Physiopathology of Hereditary Spastic Paraplegias. Frontiers in Neuroscience, 2020, 14, 74.	1.4	40
40	Plasma neurofilament light chain predicts cerebellar atrophy and clinical progression in spinocerebellar ataxia. Neurobiology of Disease, 2021, 153, 105311.	2.1	39
41	Cerebellar ataxia with elevated cerebrospinal free sialic acid (CAFSA). Brain, 2009, 132, 801-809.	3.7	37
42	Triheptanoin for the treatment of brain energy deficit: A 14â€year experience. Journal of Neuroscience Research, 2017, 95, 2236-2243.	1.3	37
43	Regional Brain and Spinal Cord Volume Loss in Spinocerebellar Ataxia Type 3. Movement Disorders, 2021, 36, 2273-2281.	2.2	37
44	Multi-omics signature of brain amyloid deposition in asymptomatic individuals at-risk for Alzheimer's disease: The INSIGHT-preAD study. EBioMedicine, 2019, 47, 518-528.	2.7	36
45	Proposal for a simplified classification of IMD based on a pathophysiological approach: A practical guide for clinicians. Journal of Inherited Metabolic Disease, 2019, 42, 706-727.	1.7	35
46	Adult-Onset Leukoencephalopathy with Axonal Spheroids and Pigmented Glia: An MRI Study of 16 French Cases. American Journal of Neuroradiology, 2018, 39, 1657-1661.	1.2	31
47	Factors in the disease severity of ATP1A3 mutations: Impairment, misfolding, and allele competition. Neurobiology of Disease, 2019, 132, 104577.	2.1	31
48	A diagnostic flow chart forPOLG-related diseases based on signs sensitivity and specificity. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 646-654.	0.9	30
49	A simple blood test expedites the diagnosis of glucose transporter type 1 deficiency syndrome. Annals of Neurology, 2017, 82, 133-138.	2.8	30
50	Quantification ofin vivo³¹P NMR brain spectra using LCModel. NMR in Biomedicine, 2015, 28, 633-641.	1.6	27
51	Evolution of structural neuroimaging biomarkers in a series of adult patients with Niemann-Pick type C under treatment. Orphanet Journal of Rare Diseases, 2017, 12, 22.	1.2	27
52	MRI of neurodegeneration with brain iron accumulation. Current Opinion in Neurology, 2020, 33, 462-473.	1.8	27
53	Abnormal glycogen in astrocytes is sufficient to cause adult polyglucosan body disease. Gene, 2013, 515, 376-379.	1.0	26
54	Imaging and spectroscopic approaches to probe brain energy metabolism dysregulation in neurodegenerative diseases. Journal of Cerebral Blood Flow and Metabolism, 2017, 37, 1927-1943.	2.4	24

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55	Haematopoietic stem cell transplantation in CSF1R-related adult-onset leukoencephalopathy with axonal spheroids and pigmented glia. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, jnnp-2019-320701.	0.9	24
56	Mutations in DDHD1 , encoding a phospholipase A1, is a novel cause of retinopathy and neurodegeneration with brain iron accumulation. European Journal of Medical Genetics, 2017, 60, 639-642.	0.7	22
57	Low cancer prevalence in polyglutamine expansion diseases. Neurology, 2017, 88, 1114-1119.	1.5	21
58	Long-term follow-up in an open-label trial of triheptanoin in GLUT1 deficiency syndrome: a sustained dramatic effect. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 1291-1293.	0.9	21
59	Elevated CSF N -acetylaspartylglutamate in patients with free sialic acid storage diseases. Neurology, 2010, 74, 302-305.	1.5	20
60	Longâ€term outcome of methylmalonic aciduria after kidney, liver, or combined liverâ€kidney transplantation: The French experience. Journal of Inherited Metabolic Disease, 2020, 43, 234-243.	1.7	20
61	Elevated CSF N-acetylaspartylglutamate suggests specific molecular diagnostic abnormalities in patients with white matter diseases. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2010, 1802, 1112-1117.	1.8	19
62	Free sialic acid storage disease without sialuria. Annals of Neurology, 2009, 65, 753-757.	2.8	18
63	Peak hyperammonemia and atypical acute liver failure: The eruption of an urea cycle disorder during hyperemesis gravidarum. Journal of Hepatology, 2018, 68, 185-192.	1.8	18
64	A doubleâ€blind, placeboâ€controlled trial of triheptanoin in adult polyglucosan body disease and openâ€label, longâ€term outcome. Journal of Inherited Metabolic Disease, 2018, 41, 877-883.	1.7	17
65	Lipids and synaptic functions. Journal of Inherited Metabolic Disease, 2018, 41, 1117-1122.	1.7	17
66	No effect of triheptanoin on exercise performance in McArdle disease. Annals of Clinical and Translational Neurology, 2019, 6, 1949-1960.	1.7	17
67	<scp>Adultâ€</scp>onset diagnosis of urea cycle disorders: Results of a French cohort of 71 patients. Journal of Inherited Metabolic Disease, 2021, 44, 1199-1214.	1.7	17
68	Consensus clinical management guideline for betaâ€propeller proteinâ€associated neurodegeneration. Developmental Medicine and Child Neurology, 2021, 63, 1402-1409.	1.1	17
69	The phenotype of adult versus pediatric patients with inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2018, 41, 753-756.	1.7	16
70	Differential Expression of Interferon-Alpha Protein Provides Clues to Tissue Specificity Across Type I Interferonopathies. Journal of Clinical Immunology, 2021, 41, 603-609.	2.0	16
71	Expanded neurochemical profile in the early stage of Huntington disease using proton magnetic resonance spectroscopy. NMR in Biomedicine, 2018, 31, e3880.	1.6	14
72	Ureaplasma parvum causes hyperammonemia presenting as refractory status epilepticus after kidney transplant. Journal of Critical Care, 2020, 57, 79-83.	1.0	14

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73	Acute but transient neurological deterioration revealing adult polyglucosan body disease. Journal of the Neurological Sciences, 2013, 324, 179-182.	0.3	13
74	Expert opinion on diagnosing, treating and managing patients with cerebrotendinous xanthomatosis (CTX): a modified Delphi study. Orphanet Journal of Rare Diseases, 2021, 16, 353.	1.2	13
75	Implication of folate deficiency in CYP2U1 loss of function. Journal of Experimental Medicine, 2021, 218, .	4.2	13
76	Education and training in adult metabolic medicine: Results of an international survey. JIMD Reports, 2019, 49, 63-69.	0.7	12
77	Deciphering the natural history of SCA7 in children. European Journal of Neurology, 2020, 27, 2267-2276.	1.7	12
78	Making a â€˜JUMPâ€™ from paediatric to adult healthcare: A transitional program for young adults with chronic neurological disease. Journal of the Neurological Sciences, 2018, 395, 77-83.	0.3	11
79	Targeted versus untargeted omics â€” the CAFSA story. Journal of Inherited Metabolic Disease, 2018, 41, 447-456.	1.7	10
80	Multiparametric characterization of white matter alterations in early stage Huntington disease. Scientific Reports, 2021, 11, 13101.	1.6	9
81	Evaluation of CSF1R â€related adult onset leukoencephalopathy with axonal spheroids and pigmented glia diagnostic criteria. European Journal of Neurology, 2022, 29, 329-334.	1.7	8
82	Subspecialty training in adult inherited metabolic diseases: a call to action for unmet needs. Lancet Diabetes and Endocrinology,the, 2019, 7, 82-84.	5.5	7
83	Transition from ketogenic diet to triheptanoin in patients with GLUT1 deficiency syndrome. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 444-445.	0.9	7
84	Multiparametric in vivo analyses of the brain and spine identify structural and metabolic biomarkers in men with adrenomyeloneuropathy. NeuroImage: Clinical, 2021, 29, 102566.	1.4	7
85	Influence of early identification and therapy on longâ€term outcomes in earlyâ€onset <scp>MTHFR</scp> deficiency. Journal of Inherited Metabolic Disease, 2022, 45, 848-861.	1.7	7
86	Severe transient myopathy in a patient with progressive multiple sclerosis and high-dose biotin. Neurology, 2019, 92, 1060-1062.	1.5	6
87	Outline of metabolic diseases in adult neurology. Revue Neurologique, 2015, 171, 531-538.	0.6	5
88	A high prevalence of arterial hypertension in patients with mitochondrial diseases. Journal of Inherited Metabolic Disease, 2020, 43, 478-485.	1.7	5
89	Triheptanoin â€“ Novel therapeutic approach for the ultra-rare disease mitochondrial malate dehydrogenase deficiency. Molecular Genetics and Metabolism Reports, 2021, 29, 100814.	0.4	5
90	Adult-onset Generalized Dystonia as the Main Manifestation of MEGDEL Syndrome. Tremor and Other Hyperkinetic Movements, 2018, 8, 554.	1.1	5

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91	Postauthorization safety study of betaine anhydrous. Journal of Inherited Metabolic Disease, 2022, 45, 719-733.	1.7	5
92	GJA1 Variants Cause Spastic Paraplegia Associated with Cerebral Hypomyelination. American Journal of Neuroradiology, 2019, 40, 788-791.	1.2	4
93	Domino liver transplantation: the risk of disease recurrence. Clinics and Research in Hepatology and Gastroenterology, 2019, 43, 510-512.	0.7	4
94	Quantitative neuroimaging biomarkers in a series of 20 adult patients with POLG mutations. Mitochondrion, 2019, 45, 22-28.	1.6	4
95	Clinical and molecular characterization of adult patients with lateâ€onset <scp>MTHFR</scp> deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 777-786.	1.7	4
96	Cervical Spinal Cord Degeneration in Spinocerebellar Ataxia Type 7. American Journal of Neuroradiology, 2021, 42, 1735-1739.	1.2	4
97	Adult-onset Generalized Dystonia as the Main Manifestation of MEGDEL Syndrome. Tremor and Other Hyperkinetic Movements, 2020, 8, 554.	1.1	4
98	Dopamine and serotonin levels in cerebrospinal fluid during episodes ofÂKleine-Levin syndrome. Sleep Medicine, 2017, 36, 184-185.	0.8	3
99	Usefulness of diagnostic tools in a GLUT1 deficiency syndrome patient with 2 inherited mutations. Brain and Development, 2019, 41, 808-811.	0.6	3
100	Phenotype, treatment practice and outcome in the cobalamin-dependent remethylation disorders and MTHFR deficiency: data from the E-HOD registry. Journal of Inherited Metabolic Disease, 2018, , .	1.7	2
101	Demyelinating motor neuropathy associated with a homozygous <scp>GPT2</scp> pathogenic variant. Muscle and Nerve, 2021, 63, E41-E44.	1.0	2
102	Dysfunctional Homozygous VRK1-D263G Variant Impairs the Assembly of Cajal Bodies and DNA Damage Response in Hereditary Spastic Paraplegia. Neurology: Genetics, 2021, 7, e624.	0.9	2
103	Nuclear Magnetic Resonance of Cerebrospinal Fluid: The Neurometabolome. , 0, , 257-270.		1
104	Unexplicated hyperammonemic encephalopathy: remember the old urinary diversions!. Acta Neurologica Belgica, 2016, 116, 677-678.	0.5	1
105	Focal epilepsy as the revealing symptom of 5,10-methylenetetrahydrofolate reductase deficiency in a young adult. Revue Neurologique, 2018, 174, 173-175.	0.6	1
106	Multimodal neurometabolic investigation of the effects of zolpidem on leukoencephalopathyâ€related apathy. European Journal of Neurology, 2020, 27, 2297-2302.	1.7	1
107	Hypomyelinating leukodystrophies in adults. European Journal of Neurology, 2021, 28, 733-734.	1.7	1
108	Diagnostic Procedures. , 2016, , 91-107.		1

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109	Adult Cerebellar Ataxia, Axonal Neuropathy, and Sensory Impairments Caused by Biallelic SCO2 Variants. Neurology: Genetics, 2021, 7, e630.	0.9	1
110	KIF1C Variants Are Associated with Hypomyelination, Ataxia, Tremor, and Dystonia in Fraternal Twins. Tremor and Other Hyperkinetic Movements, 2019, 9, .	1.1	1
111	A Multimodal Omics Exploration of the Motor and Non-Motor Symptoms of Parkinsonâ€™s Disease. International Journal of Translational Medicine, 2022, 2, 97-112.	0.1	1
112	Diagnostic Procedures: Functional Tests and Post-mortem Protocol. , 2012, , 87-102.		0
113	Reply. Annals of Neurology, 2013, 73, 318-318.	2.8	0