Manuel Schiff

List of Publications by Year
in descending order

106 papers	3,550 citations	136950 32 h-index	168389 53 g-index
116 all docs	116 docs citations	116 times ranked	5294 citing authors

#	Article	IF	Citations
1	Mitochondrial double-stranded RNA triggers antiviral signalling in humans. Nature, 2018, 560, 238-242.	27.8	397
2	Lysinuric protein intolerance (LPI): A multi organ disease by far more complex than a classic urea cycle disorder. Molecular Genetics and Metabolism, 2012, 106, 12-17.	1.1	221
3	Guidelines for diagnosis and management of the cobalaminâ€related remethylation disorders cblC, cblD, cblE, cblF, cblG, cblJ and MTHFR deficiency. Journal of Inherited Metabolic Disease, 2017, 40, 21-48.	3.6	206
4	Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. American Journal of Human Genetics, 2017, 100, 257-266.	6.2	127
5	Mitochondrial cytochrome c oxidase deficiency. Clinical Science, 2016, 130, 393-407.	4.3	121
6	Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency. American Journal of Human Genetics, 2016, 98, 1130-1145.	6.2	118
7	Electron transfer flavoprotein deficiency: Functional and molecular aspects. Molecular Genetics and Metabolism, 2006, 88, 153-158.	1.1	94
8	International clinical guidelines for the management of phosphomannomutase 2â€congenital disorders of glycosylation: Diagnosis, treatment and follow up. Journal of Inherited Metabolic Disease, 2019, 42, 5-28.	3.6	91
9	Clinical, laboratory and molecular findings and long-term follow-up data in 96 French patients with PMM2-CDG (phosphomannomutase 2-congenital disorder of glycosylation) and review of the literature. Journal of Medical Genetics, 2017, 54, 843-851.	3.2	88
10	SLC25A32 Mutations and Riboflavin-Responsive Exercise Intolerance. New England Journal of Medicine, 2016, 374, 795-797.	27.0	87
11	Pontocerebellar hypoplasia type 1. Neurology, 2013, 80, 438-446.	1.1	84
12	Progressive deafnessâ€“dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015.	5.3	63
13	Isolated remethylation disorders: do our treatments benefit patients?. Journal of Inherited Metabolic Disease, 2011, 34, 137-145.	3.6	62
14	Mutations in CYC1, Encoding Cytochrome c1 Subunit of Respiratory Chain Complex III, Cause Insulin-Responsive Hyperglycemia. American Journal of Human Genetics, 2013, 93, 384-389.	6.2	61
15	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. Orphanet Journal of Rare Diseases, 2018, 13, 120.	2.7	61
16	Impact of age at onset and newborn screening on outcome in organic acidurias. Journal of Inherited Metabolic Disease, 2016, 39, 341-353.	3.6	60
17	Identification of novel mutations in theSLC25A15gene in hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome: A clinical, molecular, and functional study. Human Mutation, 2009, 30, 741-748.	2.5	57
18	Treatment of Inherited Homocystinurias. Neuropediatrics, 2012, 43, 295-304.	0.6	56

#	Article	IF	Citations
19	Complex I assembly function and fatty acid oxidation enzyme activity of ACAD9 both contribute to disease severity in ACAD9 deficiency. Human Molecular Genetics, 2015, 24, 3238-3247.	2.9	53
20	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.	3.6	53
21	Age at disease onset and peak ammonium level rather than interventional variables predict the neurological outcome in urea cycle disorders. Journal of Inherited Metabolic Disease, 2016, 39, 661-672.	3.6	52
22	Clinical pattern, mutations and in vitro residual activity in 33 patients with severe 5, 10 methylenetetrahydrofolate reductase (MTHFR) deficiency. Journal of Inherited Metabolic Disease, 2016, 39, 115-124.	3.6	52
23	Molecular and cellular pathology of very-long-chain acyl-CoA dehydrogenase deficiency. Molecular Genetics and Metabolism, 2013, 109, 21-27.	1.1	49
24	QIL1 mutation causes MICOS disassembly and early onset fatal mitochondrial encephalopathy with liver disease. ELife, 2016, 5, .	6.0	46
25	Mitochondrial response to controlled nutrition in health and disease. Nutrition Reviews, 2011, 69, 65-75.	5.8	45
26	Further delineation of the 17p13.3 microdeletion involving YWHAE but distal to PAFAH1B1: Four additional patients. European Journal of Medical Genetics, 2010, 53, 303-308.	1.3	44
27	Evolutionarily conserved susceptibility of the mitochondrial respiratory chain to SDHI pesticides and its consequence on the impact of SDHIs on human cultured cells. PLoS ONE, 2019, 14, e0224132.	2.5	43
28	Diagnosis of â€˜possibleâ€™ mitochondrial disease: an existential crisis. Journal of Medical Genetics, 2019, 56, 123-130.	3.2	42
29	A noncoding RNA modulator potentiates phenylalanine metabolism in mice. Science, 2021, 373, 662-673.	12.6	42
30	Leigh's disease due to a new mutation in thePDHXgene. Annals of Neurology, 2006, 59, 709-714.	5.3	38
31	Neonatal Hyperammonemia: The N-carbamoyl-L-glutamic Acid Test. Journal of Pediatrics, 2005, 147, 260-262.	1.8	35
32	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disordersâ€”A successful strategy for clinical research of rare diseases. Journal of Inherited Metabolic Disease, 2019, 42, 93-106.	3.6	35
33	Eyes on MEGDEL: Distinctive Basal Ganglia Involvement in Dystonia Deafness Syndrome. Neuropediatrics, 2015, 46, 098-103.	0.6	34
34	New insights into the clinical and molecular spectrum of the novel CYFIP2-related neurodevelopmental disorder and impairment of the WRC-mediated actin dynamics. Genetics in Medicine, 2021, 23, 543-554.	2.4	32
35	Genetic background influences mitochondrial function: modeling mitochondrial disease for therapeutic development. Trends in Molecular Medicine, 2010, 16, 210-217.	6.7	31
36	Should Metabolic Diseases Be Systematically Screened in Nonsyndromic Autism Spectrum Disorders?. PLoS ONE, 2011, 6, e21932.	2.5	31

#	Article	IF	Citations
37	Efficacy and safety of i.v. sodium benzoate in urea cycle disorders: a multicentre retrospective study. Orphanet Journal of Rare Diseases, 2016, 11, 127.	2.7	31
38	Neonatal cardiomyopathies and metabolic crises due to oxidative phosphorylation defects. Seminars in Fetal and Neonatal Medicine, 2011, 16, 216-221.	2.3	30
39	Mouse Studies to Shape Clinical Trials for Mitochondrial Diseases: High Fat Diet in Harlequin Mice. PLoS ONE, 2011, 6, e28823.	2.5	28
40	Amlexanox provides a potential therapy for nonsense mutations in the lysosomal storage disorder Aspartylglucosaminuria. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 668-675.	3.8	27
41	Should transcobalamin deficiency be treated aggressively?. Journal of Inherited Metabolic Disease, 2010, 33, 223-229.	3.6	26
42	Early prediction of phenotypic severity in Citrullinemia Type 1. Annals of Clinical and Translational Neurology, 2019, 6, 1858-1871.	3.7	26
43	An LCâ€“MS/MS-Based Method for the Quantification of Pyridox(am)ine 5â€²-Phosphate Oxidase Activity in Dried Blood Spots from Patients with Epilepsy. Analytical Chemistry, 2017, 89, 8892-8900.	6.5	24
44	Long-term liver disease in methylmalonic and propionic acidemias. Molecular Genetics and Metabolism, 2018, 123, 433-440.	1.1	23
45	SLC13A3 variants cause acute reversible leukoencephalopathy and Î±â€ketoglutarate accumulation. Annals of Neurology, 2019, 85, 385-395.	5.3	22
46	A case of pyruvate carboxylase deficiency with atypical clinical and neuroradiological presentation. Molecular Genetics and Metabolism, 2006, 87, 175-177.	1.1	20
47	High homocysteine induces betaine depletion. Bioscience Reports, 2015, 35, .	2.4	20
48	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.	3.6	20
49	Therapies in inborn errors of oxidative metabolism. Trends in Endocrinology and Metabolism, 2012, 23, 488-495.	7.1	19
50	Efficacy and safety of D,L-3-hydroxybutyrate (D,L-3-HB) treatment in multiple acyl-CoA dehydrogenase deficiency. Genetics in Medicine, 2020, 22, 908-916.	2.4	19
51	Long-term safety and outcomes in hereditary tyrosinaemia type 1 with nitisinone treatment: a 15-year non-interventional, multicentre study. Lancet Diabetes and Endocrinology,the, 2021, 9, 427-435.	11.4	19
52	Heterogeneity of follow-up procedures in French and Belgian patients with treated hereditary tyrosinemia type 1: results of a questionnaire and proposed guidelines. Journal of Inherited Metabolic Disease, 2012, 35, 823-829.	3.6	18
53	Diagnostic contribution of metabolic workup for neonatal inherited metabolic disorders in the absence of expanded newborn screening. Scientific Reports, 2019, 9, 14098.	3.3	18
54	Betaine anhydrous in homocystinuria: results from the RoCH registry. Orphanet Journal of Rare Diseases, 2019, 14, 66.	2.7	18

#	Article	IF	Citations
55	Expanding and Underscoring the Hepatoâ€Encephalopathic Phenotype of QIL1/MIC13. Hepatology, 2019, 70, 1066-1070.	7.3	17
56	Long-term follow-up of metachronous marrow-kidney transplantation in severe type II sialidosis: what does success mean?. Nephrology Dialysis Transplantation, 2005, 20, 2563-2565.	0.7	15
57	How can cobalamin injections be spaced in long-term therapy for inborn errors of vitamin B12 absorption?. Molecular Genetics and Metabolism, 2012, 107, 66-71.	1.1	15
58	Paradoxical Inhibition of Glycolysis by Pioglitazone Opposes the Mitochondriopathy Caused by AIF Deficiency. EBioMedicine, 2017, 17, 75-87.	6.1	15
59	Inherited Disorders of Lysine Metabolism: A Review. Journal of Nutrition, 2020, 150, 2556S-2560S.	2.9	15
60	Phenotypic diversity of brain MRI patterns in mitochondrial aminoacyl-tRNA synthetase mutations. Molecular Genetics and Metabolism, 2021, 133, 222-229.	1.1	15
61	A Review of Multiple Mitochondrial Dysfunction Syndromes, Syndromes Associated with Defective Fe-S Protein Maturation. Biomedicines, 2021, 9, 989.	3.2	15
62	A novel gross deletion caused by non-homologous recombination of the PDHX gene in a patient with pyruvate dehydrogenase deficiencyâ˜†. Molecular Genetics and Metabolism, 2006, 89, 106-110.	1.1	13
63	Neurocognitive profiles in MSUD schoolâ€age patients. Journal of Inherited Metabolic Disease, 2017, 40, 377-383.	3.6	13
64	Diagnostic approach to neurotransmitter monoamine disorders: experience from clinical, biochemical, and genetic profiles. Journal of Inherited Metabolic Disease, 2018, 41, 129-139.	3.6	12
65	Clinical, neuroimaging and biochemical findings in patients and patient fibroblasts expressing ten novel GFM1 mutations. Human Mutation, 2020, 41, 397-402.	2.5	10
66	Quantitative analysis of the natural history of prolidase deficiency: description of 17 families and systematic review of published cases. Genetics in Medicine, 2021, 23, 1604-1615.	2.4	10
67	Pericallosal lipoma and middle cerebral artery aneurysm: a coincidence?. Pediatric Radiology, 2010, 40, 1417-1420.	2.0	9
68	Autism and inborn errors of metabolism: how much is enough?. Developmental Medicine and Child Neurology, 2015, 57, 788-789.	2.1	9
69	Idebenone in Friedreich ataxia and Leberâ€™s hereditary optic neuropathy: close mechanisms, similar therapy?: Table 1. Brain, 2016, 139, e39-e39.	7.6	8
70	Phosphoglycerate dehydrogenase (PHGDH) deficiency without epilepsy mimicking primary microcephaly. American Journal of Medical Genetics, Part A, 2017, 173, 1936-1942.	1.2	8
71	Wide clinical spectrum in ALG8-CDG: clues from molecular findings suggest an explanation for a milder phenotype in the first-described patient. Pediatric Research, 2019, 85, 384-389.	2.3	8
72	New insights into carnitineâ€acylcarnitine translocase deficiency from 23 cases: Management challenges and potential therapeutic approaches. Journal of Inherited Metabolic Disease, 2021, 44, 903-915.	3.6	8

#	Article	IF	Citations
73	Neonatal gene therapy achieves sustained disease rescue of maple syrup urine disease in mice. Nature Communications, 2022, 13, .	12.8	8
74	Antenatal nephromegaly and propionic acidemia: a case report. BMC Nephrology, 2017, 18, 110.	1.8	7
75	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.	3.6	7
76	An unfortunate challenge: Ketogenic diet for the treatment of Lennoxâ€“Gastaut syndrome in tyrosinemia type 1. European Journal of Paediatric Neurology, 2016, 20, 674-677.	1.6	6
77	DNAJC12: A molecular chaperone involved in proteostasis, PKU, biogenic amines metabolism and beyond?. Molecular Genetics and Metabolism, 2018, 123, 285-286.	1.1	6
78	Heterogeneity of PNPT1 neuroimaging: mitochondriopathy, interferonopathy or both?. Journal of Medical Genetics, 2022, 59, 204-208.	3.2	6
79	Development and characterization of a mouse model for Acad9 deficiency. Molecular Genetics and Metabolism, 2021, 134, 156-163.	1.1	6
80	Determinants of Quality of Life in Children with Inborn Errors of Metabolism Receiving a Restricted Diet. Journal of Pediatrics, 2022, 242, 192-200.e3.	1.8	6
81	Abnormal Glycosylation Profile and High Alpha-Fetoprotein in a Patient with Twinkle Variants. JIMD Reports, 2016, 29, 109-113.	1.5	5
82	Postauthorization safety study of betaine anhydrous. Journal of Inherited Metabolic Disease, 2022, 45, 719-733.	3.6	5
83	ANT2-defective fibroblasts exhibit normal mitochondrial bioenergetics. Molecular Genetics and Metabolism Reports, 2015, 3, 43-46.	1.1	4
84	Angelman syndrome and isovaleric acidemia: What is the link?. Molecular Genetics and Metabolism Reports, 2015, 3, 36-38.	1.1	4
85	Transient neonatal renal failure and massive polyuria in MEGDEL syndrome. Molecular Genetics and Metabolism Reports, 2016, 7, 8-10.	1.1	4
86	CUGC for lysinuric protein intolerance (LPI). European Journal of Human Genetics, 2020, 28, 1129-1134.	2.8	4
87	Determining factors of the cognitive outcome in early treated PKU: A study of 39 pediatric patients. Molecular Genetics and Metabolism Reports, 2019, 20, 100498.	1.1	3
88	Should patients with Phosphomannomutase 2-CDG (PMM2-CDG) be screened for adrenal insufficiency?. Molecular Genetics and Metabolism, 2021, 133, 397-399.	1.1	3
89	Autism and Medical Comorbidities. Key Issues in Mental Health, 2015, , 20-33.	0.6	2
90	Doubling diet fat on sugar ratio in children with mitochondrial OXPHOS disorders: Effects of a randomized trial on resting energy expenditure, diet induced thermogenesis and body composition. Clinical Nutrition, 2016, 35, 1414-1422.	5.0	2

#	Article	IF	Citations
91	Drug development for mitochondrial disease: recent progress, current challenges, and future prospects. Expert Opinion on Orphan Drugs, 2016, 4, 83-92.	0.8	2
92	Limited benefits of presymptomatic cord blood transplantation in neurovisceral acid sphingomyelinase deficiency (ASMD) intermediate type. European Journal of Paediatric Neurology, 2017, 21, 907-911.	1.6	2
93	LC-MS/MS Identification of Prolidase Deficiency: A Rare Cause of Infantile Hepatosplenomegaly. Clinical Chemistry, 2022, 68, 478-480.	3.2	2
94	Enlargement of the Optic Chiasm: A Novel Imaging Finding in Glutaric Aciduria Type 1. American Journal of Neuroradiology, 2021, 42, 1722-1726.	2.4	2
95	What are the clues for an inherited metabolic disorder in Reye syndrome? A single Centre study of 58 children. Molecular Genetics and Metabolism, 2022, 135, 320-326.	1.1	2
96	Covid-19: Possible trigger of SLC13A3 reversible leukoencephalopathy relapse?. Molecular Genetics and Metabolism, 2022, 136, 83-84.	1.1	2
97	Very longâ€term outcomes in 23 patients with <scp>cblA</scp> type methylmalonic acidemia. Journal of Inherited Metabolic Disease, 0, , .	3.6	2
98	Subclinical maculopathy and retinopathy in transcobalamin deficiency: a 10-year follow-up. Documenta Ophthalmologica, 2022, 144, 53-65.	2.2	1
99	Ear and kidney malformations with renal failure in an infant: what is the link?. Nephrology Dialysis Transplantation, 2003, 18, 1673-1674.	0.7	0
100	Long-Term Follow-up of a Child With Putative Remethylation Disorder Who Presented With Severe Anemia as a Neonate. Journal of Pediatric Hematology/Oncology, 2020, 42, 332-333.	0.6	0
101	Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency and progressive retinopathy: one case report followed by ERGs, VEPs, EOG over a 17-year period. Documenta Ophthalmologica, 2021, 142, 371-380.	2.2	0
102	Disorders of Amino Acid Transport at the Cell Membrane. , 2016, , 363-371.		0
103	Title is missing!. , 2019, 14, e0224132.		0
104	Title is missing!. , 2019, 14, e0224132.		0
105	Title is missing!. , 2019, 14, e0224132.		0
106	Title is missing!. , 2019, 14, e0224132.		0