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	Heterogeneity of PNPT1 neuroimaging: mitochondriopathy, interferonopathy or both?. Journal of Medical Genetics, 2022, 59, 204-208.	3.2	6
2	LC-MS/MS Identification of Prolidase Deficiency: A Rare Cause of Infantile Hepatosplenomegaly. Clinical Chemistry, 2022, 68, 478-480.	3.2	2
3	Subclinical maculopathy and retinopathy in transcobalamin deficiency: a 10-year follow-up. Documenta Ophthalmologica, 2022, 144, 53-65.	2.2	1
4	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
5	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
6	Postauthorization safety study of betaine anhydrous. Journal of Inherited Metabolic Disease, 2022, 45, 719-733.	3.6	5
7	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
8	Covid-19: Possible trigger of SLC13A3 reversible leukoencephalopathy relapse?. Molecular Genetics and Metabolism, 2022, 136, 83-84.	1.1	2
9	Neonatal gene therapy achieves sustained disease rescue of maple syrup urine disease in mice. Nature Communications, 2022, 13, .	12.8	8
10	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
11	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
12	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
13	Phenotypic diversity of brain MRI patterns in mitochondrial aminoacyl-tRNA synthetase mutations. Molecular Genetics and Metabolism, 2021, 133, 222-229.	1.1	15
14	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
15	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
16	A Review of Multiple Mitochondrial Dysfunction Syndromes, Syndromes Associated with Defective Fe-S Protein Maturation. Biomedicines, 2021, 9, 989.	3.2	15
17	A noncoding RNA modulator potentiates phenylalanine metabolism in mice. Science, 2021, 373, 662-673.	12.6	42
18	Should patients with Phosphomannomutase 2-CDG (PMM2-CDG) be screened for adrenal insufficiency?. Molecular Genetics and Metabolism, 2021, 133, 397-399.	1.1	3

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19	Development and characterization of a mouse model for Acad9 deficiency. Molecular Genetics and Metabolism, 2021, 134, 156-163.	1.1	6
20	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
21	Clinical, neuroimaging and biochemical findings in patients and patient fibroblasts expressing ten novel GFM1 mutations. Human Mutation, 2020, 41, 397-402.	2.5	10
22	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
23	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
24	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
25	Inherited Disorders of Lysine Metabolism: A Review. Journal of Nutrition, 2020, 150, 2556S-2560S.	2.9	15
26	CUGC for lysinuric protein intolerance (LPI). European Journal of Human Genetics, 2020, 28, 1129-1134.	2.8	4
27	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
28	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
29	Early prediction of phenotypic severity in Citrullinemia Type 1. Annals of Clinical and Translational Neurology, 2019, 6, 1858-1871.	3.7	26
30	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
31	Diagnosis of â€˜possibleâ€™ mitochondrial disease: an existential crisis. Journal of Medical Genetics, 2019, 56, 123-130.	3.2	42
32	Betaine anhydrous in homocystinuria: results from the RoCH registry. Orphanet Journal of Rare Diseases, 2019, 14, 66.	2.7	18
33	Expanding and Underscoring the Hepatoâ€Encephalopathic Phenotype of QIL1/MIC13. Hepatology, 2019, 70, 1066-1070.	7.3	17
34	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
35	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
36	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

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37	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
38	SLC13A3 variants cause acute reversible leukoencephalopathy and Î±â€ketoglutarate accumulation. Annals of Neurology, 2019, 85, 385-395.	5.3	22
39	Title is missing!. , 2019, 14, e0224132.		0
40	Title is missing!. , 2019, 14, e0224132.		0
41	Title is missing!. , 2019, 14, e0224132.		0
42	Title is missing!. , 2019, 14, e0224132.		0
43	Long-term liver disease in methylmalonic and propionic acidemias. Molecular Genetics and Metabolism, 2018, 123, 433-440.	1.1	23
44	DNAJC12: A molecular chaperone involved in proteostasis, PKU, biogenic amines metabolism and beyond?. Molecular Genetics and Metabolism, 2018, 123, 285-286.	1.1	6
45	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
46	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
47	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
48	Mitochondrial double-stranded RNA triggers antiviral signalling in humans. Nature, 2018, 560, 238-242.	27.8	397
49	Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. American Journal of Human Genetics, 2017, 100, 257-266.	6.2	127
50	Paradoxical Inhibition of Glycolysis by Pioglitazone Opposes the Mitochondriopathy Caused by AIF Deficiency. EBioMedicine, 2017, 17, 75-87.	6.1	15
51	Antenatal nephromegaly and propionic acidemia: a case report. BMC Nephrology, 2017, 18, 110.	1.8	7
52	Phosphoglycerate dehydrogenase (PHGDH) deficiency without epilepsy mimicking primary microcephaly. American Journal of Medical Genetics, Part A, 2017, 173, 1936-1942.	1.2	8
53	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
54	Neurocognitive profiles in MSUD schoolâ€age patients. Journal of Inherited Metabolic Disease, 2017, 40, 377-383.	3.6	13

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55	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
56	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
57	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
58	Progressive deafnessâ€“dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015.	5.3	63
59	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
60	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
61	Abnormal Glycosylation Profile and High Alpha-Fetoprotein in a Patient with Twinkle Variants. JIMD Reports, 2016, 29, 109-113.	1.5	5
62	Idebenone in Friedreich ataxia and Leberâ€™s hereditary optic neuropathy: close mechanisms, similar therapy?: Table 1. Brain, 2016, 139, e39-e39.	7.6	8
63	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
64	Transient neonatal renal failure and massive polyuria in MEGDEL syndrome. Molecular Genetics and Metabolism Reports, 2016, 7, 8-10.	1.1	4
65	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
66	Mitochondrial cytochrome c oxidase deficiency. Clinical Science, 2016, 130, 393-407.	4.3	121
67	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
68	SLC25A32 Mutations and Riboflavin-Responsive Exercise Intolerance. New England Journal of Medicine, 2016, 374, 795-797.	27.0	87
69	Drug development for mitochondrial disease: recent progress, current challenges, and future prospects. Expert Opinion on Orphan Drugs, 2016, 4, 83-92.	0.8	2
70	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
71	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
72	QIL1 mutation causes MICOS disassembly and early onset fatal mitochondrial encephalopathy with liver disease. ELife, 2016, 5, .	6.0	46

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73	Disorders of Amino Acid Transport at the Cell Membrane. , 2016, , 363-371.		0
74	High homocysteine induces betaine depletion. Bioscience Reports, 2015, 35, .	2.4	20
75	Autism and inborn errors of metabolism: how much is enough?. Developmental Medicine and Child Neurology, 2015, 57, 788-789.	2.1	9
76	ANT2-defective fibroblasts exhibit normal mitochondrial bioenergetics. Molecular Genetics and Metabolism Reports, 2015, 3, 43-46.	1.1	4
77	Eyes on MEGDEL: Distinctive Basal Ganglia Involvement in Dystonia Deafness Syndrome. Neuropediatrics, 2015, 46, 098-103.	0.6	34
78	Autism and Medical Comorbidities. Key Issues in Mental Health, 2015, , 20-33.	0.6	2
79	Angelman syndrome and isovaleric acidemia: What is the link?. Molecular Genetics and Metabolism Reports, 2015, 3, 36-38.	1.1	4
80	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
81	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
82	Molecular and cellular pathology of very-long-chain acyl-CoA dehydrogenase deficiency. Molecular Genetics and Metabolism, 2013, 109, 21-27.	1.1	49
83	Pontocerebellar hypoplasia type 1. Neurology, 2013, 80, 438-446.	1.1	84
84	Treatment of Inherited Homocystinurias. Neuropediatrics, 2012, 43, 295-304.	0.6	56
85	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
86	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
87	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
88	Therapies in inborn errors of oxidative metabolism. Trends in Endocrinology and Metabolism, 2012, 23, 488-495.	7.1	19
89	Neonatal cardiomyopathies and metabolic crises due to oxidative phosphorylation defects. Seminars in Fetal and Neonatal Medicine, 2011, 16, 216-221.	2.3	30
90	Should Metabolic Diseases Be Systematically Screened in Nonsyndromic Autism Spectrum Disorders?. PLoS ONE, 2011, 6, e21932.	2.5	31

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91	Mouse Studies to Shape Clinical Trials for Mitochondrial Diseases: High Fat Diet in Harlequin Mice. PLoS ONE, 2011, 6, e28823.	2.5	28
92	Mitochondrial response to controlled nutrition in health and disease. Nutrition Reviews, 2011, 69, 65-75.	5.8	45
93	Isolated remethylation disorders: do our treatments benefit patients?. Journal of Inherited Metabolic Disease, 2011, 34, 137-145.	3.6	62
94	Pericallosal lipoma and middle cerebral artery aneurysm: a coincidence?. Pediatric Radiology, 2010, 40, 1417-1420.	2.0	9
95	Should transcobalamin deficiency be treated aggressively?. Journal of Inherited Metabolic Disease, 2010, 33, 223-229.	3.6	26
96	Genetic background influences mitochondrial function: modeling mitochondrial disease for therapeutic development. Trends in Molecular Medicine, 2010, 16, 210-217.	6.7	31
97	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
98	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
99	A case of pyruvate carboxylase deficiency with atypical clinical and neuroradiological presentation. Molecular Genetics and Metabolism, 2006, 87, 175-177.	1.1	20
100	Electron transfer flavoprotein deficiency: Functional and molecular aspects. Molecular Genetics and Metabolism, 2006, 88, 153-158.	1.1	94
101	A novel gross deletion caused by non-homologous recombination of the PDHX gene in a patient with pyruvate dehydrogenase deficiencyâˆ†. Molecular Genetics and Metabolism, 2006, 89, 106-110.	1.1	13
102	Leigh's disease due to a new mutation in thePDHXgene. Annals of Neurology, 2006, 59, 709-714.	5.3	38
103	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
104	Neonatal Hyperammonemia: The N-carbamoyl-L-glutamic Acid Test. Journal of Pediatrics, 2005, 147, 260-262.	1.8	35
105	Ear and kidney malformations with renal failure in an infant: what is the link?. Nephrology Dialysis Transplantation, 2003, 18, 1673-1674.	0.7	0
106	Very longâ€term outcomes in 23 patients with <scp>cblA</scp> type methylmalonic acidemia. Journal of Inherited Metabolic Disease, 0, , .	3.6	2