

# Fernando Scaglia

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/8857682/publications.pdf>

Version: 2024-02-01

117  
papers

7,651  
citations

50170

46  
h-index

56606

83  
g-index

122  
all docs

122  
docs citations

122  
times ranked

10974  
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical Spectrum, Morbidity, and Mortality in 113 Pediatric Patients With Mitochondrial Disease. <i>Pediatrics</i> , 2004, 114, 925-931.	1.0	431
2	MELAS syndrome: Clinical manifestations, pathogenesis, and treatment options. <i>Molecular Genetics and Metabolism</i> , 2015, 116, 4-12.	0.5	431
3	Diagnosis and management of mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. <i>Genetics in Medicine</i> , 2015, 17, 689-701.	1.1	414
4	GM1 gangliosidosis: Review of clinical, molecular, and therapeutic aspects. <i>Molecular Genetics and Metabolism</i> , 2008, 94, 391-396.	0.5	359
5	Use of Exome Sequencing for Infants in Intensive Care Units. <i>JAMA Pediatrics</i> , 2017, 171, e173438.	3.3	348
6	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2017, 101, 664-685.	2.6	337
7	Mitochondrial DNA Depletion Syndromes: Review and Updates of Genetic Basis, Manifestations, and Therapeutic Options. <i>Neurotherapeutics</i> , 2013, 10, 186-198.	2.1	255
8	Mitochondrial DNA maintenance defects. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2017, 1863, 1539-1555.	1.8	205
9	Reanalysis of Clinical Exome Sequencing Data. <i>New England Journal of Medicine</i> , 2019, 380, 2478-2480.	13.9	205
10	Lessons learned from additional research analyses of unsolved clinical exome cases. <i>Genome Medicine</i> , 2017, 9, 26.	3.6	184
11	Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. <i>Genetics in Medicine</i> , 2017, 19, 1380-1397.	1.1	173
12	Mitochondrial Cardiomyopathies. <i>Frontiers in Cardiovascular Medicine</i> , 2016, 3, 25.	1.1	149
13	Therapies for mitochondrial diseases and current clinical trials. <i>Molecular Genetics and Metabolism</i> , 2017, 122, 1-9.	0.5	140
14	Restoration of impaired nitric oxide production in MELAS syndrome with citrulline and arginine supplementation. <i>Molecular Genetics and Metabolism</i> , 2012, 105, 607-614.	0.5	127
15	Combined array CGH plus SNP genome analyses in a single assay for optimized clinical testing. <i>European Journal of Human Genetics</i> , 2014, 22, 79-87.	1.4	112
16	Guanidinoacetate methyltransferase (GAMT) deficiency: Outcomes in 48 individuals and recommendations for diagnosis, treatment and monitoring. <i>Molecular Genetics and Metabolism</i> , 2014, 111, 16-25.	0.5	111
17	Mitochondrial dynamics: Biological roles, molecular machinery, and related diseases. <i>Molecular Genetics and Metabolism</i> , 2018, 125, 315-321.	0.5	99
18	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. <i>American Journal of Human Genetics</i> , 2016, 98, 347-357.	2.6	98

#	ARTICLE	IF	CITATIONS
19	Recommendations for the Management of Strokelike Episodes in Patients With Mitochondrial Encephalomyopathy, Lactic Acidosis, and Strokelike Episodes. <i>JAMA Neurology</i> , 2016, 73, 591.	4.5	94
20	Effect of alternative pathway therapy on branched chain amino acid metabolism in urea cycle disorder patients. <i>Molecular Genetics and Metabolism</i> , 2004, 81, 79-85.	0.5	93
21	Maternal systemic primary carnitine deficiency uncovered by newborn screening: Clinical, biochemical, and molecular aspects. <i>Genetics in Medicine</i> , 2010, 12, 19-24.	1.1	91
22	Disorders of carnitine biosynthesis and transport. <i>Molecular Genetics and Metabolism</i> , 2015, 116, 107-112.	0.5	90
23	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. <i>American Journal of Human Genetics</i> , 2019, 104, 213-228.	2.6	90
24	Molybdenum cofactor deficiency. <i>Molecular Genetics and Metabolism</i> , 2016, 117, 1-4.	0.5	86
25	Citrulline and arginine utility in treating nitric oxide deficiency in mitochondrial disorders. <i>Molecular Genetics and Metabolism</i> , 2012, 107, 247-252.	0.5	84
26	Citrin Deficiency: A Novel Cause of Failure to Thrive That Responds to a High-Protein, Low-Carbohydrate Diet. <i>Pediatrics</i> , 2007, 119, e773-e777.	1.0	81
27	Citrin deficiency, a perplexing global disorder. <i>Molecular Genetics and Metabolism</i> , 2009, 96, 44-49.	0.5	81
28	Primary and secondary alterations of neonatal carnitine metabolism. <i>Seminars in Perinatology</i> , 1999, 23, 152-161.	1.1	80
29	Transition to Next Generation Analysis of the Whole Mitochondrial Genome: A Summary of Molecular Defects. <i>Human Mutation</i> , 2013, 34, 882-893.	1.1	79
30	Utility of Oligonucleotide Array-Based Comparative Genomic Hybridization for Detection of Target Gene Deletions. <i>Clinical Chemistry</i> , 2008, 54, 1141-1148.	1.5	78
31	Epimerase-Deficiency Galactosemia Is Not a Binary Condition. <i>American Journal of Human Genetics</i> , 2006, 78, 89-102.	2.6	77
32	Clinical Consequences of Urea Cycle Enzyme Deficiencies and Potential Links to Arginine and Nitric Oxide Metabolism. <i>Journal of Nutrition</i> , 2004, 134, 2775S-2782S.	1.3	76
33	The Mitochondrial Myopathy Encephalopathy, Lactic Acidosis with Stroke-Like Episodes (MELAS) Syndrome. <i>CNS Drugs</i> , 2006, 20, 443-464.	2.7	75
34	Mutations in the Chromatin Regulator Gene BRPF1 Cause Syndromic Intellectual Disability and Deficient Histone Acetylation. <i>American Journal of Human Genetics</i> , 2017, 100, 91-104.	2.6	72
35	Early-onset severe neuromuscular phenotype associated with compound heterozygosity for OPA1 mutations. <i>Molecular Genetics and Metabolism</i> , 2011, 103, 383-387.	0.5	71
36	Human mitochondrial transfer RNAs: Role of pathogenic mutation in disease. <i>Muscle and Nerve</i> , 2008, 37, 150-171.	1.0	68

#	ARTICLE	IF	CITATIONS
37	Abnormal neurological features predict poor survival and should preclude liver transplantation in patients with deoxyguanosine kinase deficiency. <i>Liver Transplantation</i> , 2008, 14, 1480-1485.	1.3	67
38	Mitochondrial cytopathies. <i>Cell Calcium</i> , 2016, 60, 199-206.	1.1	67
39	Impaired nitric oxide production in children with MELAS syndrome and the effect of arginine and citrulline supplementation. <i>Molecular Genetics and Metabolism</i> , 2016, 117, 407-412.	0.5	64
40	Mitochondrial myopathy, lactic acidosis, and sideroblastic anemia (MLASA) plus associated with a novel de novo mutation (m.8969G>A) in the mitochondrial encoded ATP6 gene. <i>Molecular Genetics and Metabolism</i> , 2014, 113, 207-212.	0.5	63
41	Missense variants in the middle domain of <i>DNM1L</i> in cases of infantile encephalopathy alter peroxisomes and mitochondria when assayed in <i>Drosophila</i> . <i>Human Molecular Genetics</i> , 2016, 25, 1846-1856.	1.4	62
42	Molecular spectrum of SLC22A5 (OCTN2) gene mutations detected in 143 subjects evaluated for systemic carnitine deficiency. <i>Human Mutation</i> , 2010, 31, E1632-E1651.	1.1	61
43	Insights into the Pathogenesis and Treatment of Cancer from Inborn Errors of Metabolism. <i>American Journal of Human Genetics</i> , 2011, 88, 402-421.	2.6	58
44	De Novo Mutations in SLC25A24 Cause a Craniosynostosis Syndrome with Hypertrichosis, Progeroid Appearance, and Mitochondrial Dysfunction. <i>American Journal of Human Genetics</i> , 2017, 101, 833-843.	2.6	56
45	Solid organ transplantation in primary mitochondrial disease: Proceed with caution. <i>Molecular Genetics and Metabolism</i> , 2016, 118, 178-184.	0.5	55
46	Compensating for central nervous system dysmyelination: Females with a proteolipid protein gene duplication and sustained clinical improvement. <i>Annals of Neurology</i> , 2001, 50, 747-754.	2.8	53
47	Novel homoplasmic mutation in the mitochondrial <i>rRNATyr</i> gene associated with atypical mitochondrial cytopathy presenting with focal segmental glomerulosclerosis. <i>American Journal of Medical Genetics Part A</i> , 2003, 123A, 172-178.	2.4	52
48	Untargeted metabolomic profiling reveals multiple pathway perturbations and new clinical biomarkers in urea cycle disorders. <i>Genetics in Medicine</i> , 2019, 21, 1977-1986.	1.1	47
49	Predominant cerebellar volume loss as a neuroradiologic feature of pediatric respiratory chain defects. <i>American Journal of Neuroradiology</i> , 2005, 26, 1675-80.	1.2	47
50	Clinical trials in mitochondrial disorders, an update. <i>Molecular Genetics and Metabolism</i> , 2020, 131, 1-13.	0.5	44
51	Clinical and biochemical improvement with galactose supplementation in SLC35A2-CDG. <i>Genetics in Medicine</i> , 2020, 22, 1102-1107.	1.1	42
52	Molecular and clinical characterization of the myopathic form of mitochondrial DNA depletion syndrome caused by mutations in the thymidine kinase (TK2) gene. <i>Molecular Genetics and Metabolism</i> , 2013, 110, 153-161.	0.5	40
53	Mitochondria: Role of citrulline and arginine supplementation in MELAS syndrome. <i>International Journal of Biochemistry and Cell Biology</i> , 2014, 48, 85-91.	1.2	40
54	Arginine and Citrulline for the Treatment of MELAS Syndrome. <i>FIRE Forum for International Research in Education</i> , 2017, 5, 232640981769739.	0.7	40

#	ARTICLE	IF	CITATIONS
55	SLC35A2â€œCDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. <i>Human Mutation</i> , 2019, 40, 908-925.	1.1	39
56	Safety and efficacy of omaveloxolone in patients with mitochondrial myopathy. <i>Neurology</i> , 2020, 94, e687-e698.	1.5	38
57	Mitochondrial diseases in North America. <i>Neurology: Genetics</i> , 2020, 6, e402.	0.9	38
58	An Integrated Approach to the Diagnosis and Prospective Management of Partial Ornithine Transcarbamylase Deficiency. <i>Pediatrics</i> , 2002, 109, 150-152.	1.0	37
59	Practice patterns of mitochondrial disease physicians in North America. Part 1: Diagnostic and clinical challenges. <i>Mitochondrion</i> , 2014, 14, 26-33.	1.6	36
60	Liver transplantation in propionic and methylmalonic acidemia: A single center study with literature review. <i>Molecular Genetics and Metabolism</i> , 2019, 128, 431-443.	0.5	36
61	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. <i>American Journal of Human Genetics</i> , 2017, 100, 343-351.	2.6	35
62	Clinical and molecular characterization of de novo loss of function variants in <i>HNRNPU</i> . <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2680-2689.	0.7	34
63	Genotype-phenotype correlations in individuals with pathogenic <i>RERE</i> variants. <i>Human Mutation</i> , 2018, 39, 666-675.	1.1	34
64	Deoxyguanosine kinase deficiency presenting as neonatal hemochromatosis. <i>Molecular Genetics and Metabolism</i> , 2011, 103, 262-267.	0.5	33
65	Glucose metabolism derangements in adults with the MELAS m.3243A>G mutation. <i>Mitochondrion</i> , 2014, 18, 63-69.	1.6	32
66	Biallelic mutations in the ferredoxin reductase gene cause novel mitochondriopathy with optic atrophy. <i>Human Molecular Genetics</i> , 2017, 26, 4937-4950.	1.4	32
67	Functional Characterization of the Carnitine Transporter Defective in Primary Carnitine Deficiency. <i>Archives of Biochemistry and Biophysics</i> , 1999, 364, 99-106.	1.4	30
68	Improvement of regressive autism symptoms in a child with <i>TMLHE</i> deficiency following carnitine supplementation. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2162-2167.	0.7	30
69	Improved clinical outcome following liver transplant in patients with ethylmalonic encephalopathy. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1015-1019.	0.7	30
70	Mitochondrial DNA replication: clinical syndromes. <i>Essays in Biochemistry</i> , 2018, 62, 297-308.	2.1	27
71	Successful liver transplantation in mitochondrial neurogastrointestinal encephalomyopathy (MNGIE). <i>Molecular Genetics and Metabolism</i> , 2020, 130, 58-64.	0.5	26
72	Milder clinical course of Type IV 3-methylglutaconic aciduria due to a novel mutation in TMEM70. <i>Molecular Genetics and Metabolism</i> , 2010, 101, 282-285.	0.5	25

#	ARTICLE	IF	CITATIONS
73	New insights in nutritional management and amino acid supplementation in urea cycle disorders. <i>Molecular Genetics and Metabolism</i> , 2010, 100, S72-S76.	0.5	24
74	Atypical presentation of Leigh syndrome associated with a Leber hereditary optic neuropathy primary mitochondrial DNA mutation. <i>Molecular Genetics and Metabolism</i> , 2011, 103, 153-160.	0.5	24
75	LIPT1 deficiency presenting as early infantile epileptic encephalopathy, Leigh disease, and secondary pyruvate dehydrogenase complex deficiency. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1184-1189.	0.7	24
76	De novo missense variant in the GTPase effector domain (GED) of <i>DNM1L</i> leads to static encephalopathy and seizures. <i>Journal of Physical Education and Sports Management</i> , 2019, 5, a003673.	0.5	24
77	Tetrasomy 13q mosaicism associated with phylloid hypomelanosis and precocious puberty. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 993-996.	0.7	22
78	Adult liver disorders caused by inborn errors of metabolism: Review and update. <i>Molecular Genetics and Metabolism</i> , 2015, 114, 1-10.	0.5	21
79	Milder clinical and biochemical phenotypes associated with the c.482G > A (p.Arg161Gln) pathogenic variant in cobalamin C disease: Implications for management and screening. <i>Molecular Genetics and Metabolism</i> , 2017, 122, 60-66.	0.5	20
80	The Xp contiguous deletion syndrome and autism. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1138-1148.	0.7	19
81	Corneal clouding, cataract, and colobomas with a novel missense mutation in <i>B4GALT7</i> – a review of eye anomalies in the linkeropathy syndromes. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2711-2718.	0.7	19
82	DYRK1A-related intellectual disability: a syndrome associated with congenital anomalies of the kidney and urinary tract. <i>Genetics in Medicine</i> , 2019, 21, 2755-2764.	1.1	19
83	Case report and novel treatment of an autosomal recessive Leigh syndrome caused by short-chain enoyl-CoA hydratase deficiency. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 803-807.	0.7	18
84	The effect of citrulline and arginine supplementation on lactic acidemia in MELAS syndrome. <i>Meta Gene</i> , 2013, 1, 8-14.	0.3	17
85	L-Cysteine supplementation prevents liver transplantation in a patient with TRMU deficiency. <i>Molecular Genetics and Metabolism Reports</i> , 2019, 19, 100453.	0.4	16
86	Hematologic presentation and the role of untargeted metabolomics analysis in monitoring treatment for riboflavin transporter deficiency. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2781-2787.	0.7	15
87	Analysis of fragment size distribution of cell-free DNA: A potential non-invasive marker to monitor graft damage in living-related liver transplantation for inborn errors of metabolism. <i>Molecular Genetics and Metabolism</i> , 2019, 127, 45-50.	0.5	14
88	Neonatal presentation of ventricular tachycardia and a Reye-like syndrome episode associated with disturbed mitochondrial energy metabolism. <i>BMC Pediatrics</i> , 2002, 2, 12.	0.7	13
89	TRMU deficiency: A broad clinical spectrum responsive to cysteine supplementation. <i>Molecular Genetics and Metabolism</i> , 2021, 132, 146-153.	0.5	13
90	Side Effects and Behavioral Outcomes Following High-Dose Carnitine Supplementation Among Young Males With Autism Spectrum Disorder: A Pilot Study. <i>Global Pediatric Health</i> , 2019, 6, 2333794X1983069.	0.3	12

#	ARTICLE	IF	CITATIONS
91	Nuclear Gene Defects in Mitochondrial Disorders. <i>Methods in Molecular Biology</i> , 2012, 837, 17-34.	0.4	11
92	Biallelic variants in <i>COX4I1</i> associated with a novel phenotype resembling Leigh syndrome with developmental regression, intellectual disability, and seizures. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2138-2143.	0.7	11
93	Characterization of the renal phenotype in <i>RMND1</i> related mitochondrial disease. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2019, 7, e973.	0.6	10
94	Effective Aspirin Treatment of Women at Risk for Preeclampsia Delays the Metabolic Clock of Gestation. <i>Hypertension</i> , 2021, 78, 1398-1410.	1.3	10
95	Molecular bases of hearing loss in multi-systemic mitochondrial cytopathy. <i>Genetics in Medicine</i> , 2006, 8, 641-652.	1.1	9
96	Multiple mitochondrial dysfunctions syndrome 1: An unusual cause of developmental pulmonary hypertension. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 755-761.	0.7	8
97	Expansion of the clinical phenotype of <i>GALE</i> deficiency. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3118-3121.	0.7	8
98	Differential utilization of systemic and enteral ammonia for urea synthesis in control subjects and ornithine transcarbamylase deficiency carriers. <i>American Journal of Clinical Nutrition</i> , 2003, 78, 749-755.	2.2	7
99	The expanding neurological phenotype of <i>DNM1L</i> -related disorders. <i>Brain</i> , 2018, 141, e28-e28.	3.7	7
100	Niacin therapy improves outcome and normalizes metabolic abnormalities in an <i>NAXD</i> -deficient patient. <i>Brain</i> , 2022, 145, e36-e40.	3.7	6
101	Methionyl-tRNA Formyltransferase ( <i>MTFMT</i> ) Deficiency Mimicking Acquired Demyelinating Disease. <i>Journal of Child Neurology</i> , 2016, 31, 215-219.	0.7	5
102	Mitochondrial energetic impairment in a patient with late-onset glutaric acidemia Type 2. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2426-2431.	0.7	5
103	A phenotypic expansion of <i>TRNT1</i> associated sideroblastic anemia with immunodeficiency, fevers, and developmental delay. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 259-268.	0.7	5
104	<i>GNAI1</i> brain somatic pathogenic variant in an individual with phacomatosis pigmentovascularis. <i>Neurology: Genetics</i> , 2019, 5, e366.	0.9	4
105	Severe Generalized Epidermolysis Bullosa Simplex in Two Hong Kong Children due to <i>De Novo</i> Variants in <i>KRT14</i> and <i>KRT5</i> . <i>Case Reports in Pediatrics</i> , 2020, 2020, 1-5.	0.2	4
106	Early onset and severe clinical course associated with the m.5540G>A mutation in <i>MT - TW</i> . <i>Molecular Genetics and Metabolism Reports</i> , 2014, 1, 61-65.	0.4	3
107	Acute Strokelike Presentation and Long-term Evolution of Diffusion Restriction Pattern in Ethylmalonic Encephalopathy. <i>Journal of Child Neurology</i> , 2021, 36, 841-852.	0.7	3
108	Sequencing data of cell-free DNA fragments in living-related liver transplantation for inborn errors of metabolism. <i>Data in Brief</i> , 2020, 29, 105183.	0.5	1

#	ARTICLE	IF	CITATIONS
109	Community Consensus Guidelines to Support FAIR Data Standards in Clinical Research Studies in Primary Mitochondrial Disease. <i>Genetics &amp; Genomics Next</i> , 2022, 3, 2100047.	0.8	1
110	Deletion (9) (p13.1 p21.1). <i>American Journal of Medical Genetics Part A</i> , 2000, 91, 113-115.	2.4	0
111	In reply to: Proving pathogenicity?when evolution is not enough. <i>American Journal of Medical Genetics Part A</i> , 2004, 131A, 109-110.	2.4	0
112	Rhombencephalosynapsis is a malformation deserving of further study. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2902-2902.	0.7	0
113	Obstetric and Infant Outcomes Following Planned Maternal Third Trimester Exposure to Tenofovir 1% Vaginal Gel. <i>AIDS Research and Human Retroviruses</i> , 2014, 30, A267-A268.	0.5	0
114	11979 Using whole-exome and mtDNA sequencing to develop a testing algorithm for diagnosis of mitochondrial disease in Puerto Ricans. <i>Journal of Clinical and Translational Science</i> , 2021, 5, 106-106.	0.3	0
115	Phenylbutyrate reduces plasma leucine concentrations without affecting the flux of leucine. <i>FASEB Journal</i> , 2007, 21, A335.	0.2	0
116	Mitochondrial immunofluorescence assay as an adjunctive tool in the diagnosis of mitochondrial myopathy. <i>FASEB Journal</i> , 2008, 22, 708.20.	0.2	0
117	Mitochondrial disorders. , 2020, , 283-304.		0