Fernando Scaglia

List of Publications by Year
in descending order

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117 papers	7,651 citations	50276 46 h-index	56724 83 g-index
122 all docs	122 docs citations	122 times ranked	10974 citing authors

#	Article	IF	Citations
1	A phenotypic expansion of <scp>TRNT1</scp> associated sideroblastic anemia with immunodeficiency, fevers, and developmental delay. American Journal of Medical Genetics, Part A, 2022, 188, 259-268.	1.2	5
2	Niacin therapy improves outcome and normalizes metabolic abnormalities in an NAXD-deficient patient. Brain, 2022, 145, e36-e40.	7.6	6
3	Community Consensus Guidelines to Support FAIR Data Standards in Clinical Research Studies in Primary Mitochondrial Disease. Genetics & Genomics Next, 2022, 3, 2100047.	1.5	1
4	TRMU deficiency: A broad clinical spectrum responsive to cysteine supplementation. Molecular Genetics and Metabolism, 2021, 132, 146-153.	1.1	13
5	11979 Using whole-exome and mtDNA sequencing to develop a testing algorithm for diagnosis of mitochondrial disease in Puerto Ricans. Journal of Clinical and Translational Science, 2021, 5, 106-106.	0.6	0
6	Acute Strokelike Presentation and Long-term Evolution of Diffusion Restriction Pattern in Ethylmalonic Encephalopathy. Journal of Child Neurology, 2021, 36, 841-852.	1.4	3
7	Expansion of the clinical phenotype of <scp>GALE</scp> deficiency. American Journal of Medical Genetics, Part A, 2021, 185, 3118-3121.	1.2	8
8	Effective Aspirin Treatment of Women at Risk for Preeclampsia Delays the Metabolic Clock of Gestation. Hypertension, 2021, 78, 1398-1410.	2.7	10
9	Hematologic presentation and the role of untargeted metabolomics analysis in monitoring treatment for riboflavin transporter deficiency. American Journal of Medical Genetics, Part A, 2020, 182, 2781-2787.	1.2	15
10	Clinical trials in mitochondrial disorders, an update. Molecular Genetics and Metabolism, 2020, 131, 1-13.	1.1	44
11	Mitochondrial energetic impairment in a patient with lateâ€onset glutaric acidemia Type 2. American Journal of Medical Genetics, Part A, 2020, 182, 2426-2431.	1.2	5
12	Severe Generalized Epidermolysis Bullosa Simplex in Two Hong Kong Children due to De Novo Variants in KRT14 and KRT5. Case Reports in Pediatrics, 2020, 2020, 1-5.	0.4	4
13	Clinical and biochemical improvement with galactose supplementation in SLC35A2-CDG. Genetics in Medicine, 2020, 22, 1102-1107.	2.4	42
14	Sequencing data of cell-free DNA fragments in living-related liver transplantation for inborn errors of metabolism. Data in Brief, 2020, 29, 105183.	1.0	1
15	Safety and efficacy of omaveloxolone in patients with mitochondrial myopathy. Neurology, 2020, 94, e687-e698.	1.1	38
16	Multiple mitochondrial dysfunctions syndrome 1: An unusual cause of developmental pulmonary hypertension. American Journal of Medical Genetics, Part A, 2020, 182, 755-761.	1.2	8
17	Successful liver transplantation in mitochondrial neurogastrointestinal encephalomyopathy (MNGIE). Molecular Genetics and Metabolism, 2020, 130, 58-64.	1.1	26
18	Mitochondrial diseases in North America. Neurology: Genetics, 2020, 6, e402.	1.9	38

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19	Mitochondrial disorders. , 2020, , 283-304.		0
20	Biallelic variants in COX4I1 associated with a novel phenotype resembling Leigh syndrome with developmental regression, intellectual disability, and seizures. American Journal of Medical Genetics, Part A, 2019, 179, 2138-2143.	1.2	11
21	DYRK1A-related intellectual disability: a syndrome associated with congenital anomalies of the kidney and urinary tract. Genetics in Medicine, 2019, 21, 2755-2764.	2.4	19
22	Characterization of the renal phenotype in RMND1 â€related mitochondrial disease. Molecular Genetics & Genomic Medicine, 2019, 7, e973.	1.2	10
23	Liver transplantation in propionic and methylmalonic acidemia: A single center study with literature review. Molecular Genetics and Metabolism, 2019, 128, 431-443.	1.1	36
24	Untargeted metabolomic profiling reveals multiple pathway perturbations and new clinical biomarkers in urea cycle disorders. Genetics in Medicine, 2019, 21, 1977-1986.	2.4	47
25	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	27.0	205
26	Side Effects and Behavioral Outcomes Following High-Dose Carnitine Supplementation Among Young Males With Autism Spectrum Disorder: A Pilot Study. Global Pediatric Health, 2019, 6, 2333794X1983069.	0.7	12
27	Case report and novel treatment of an autosomal recessive Leigh syndrome caused by shortâ€chain enoylâ€CoA hydratase deficiency. American Journal of Medical Genetics, Part A, 2019, 179, 803-807.	1.2	18
28	De novo missense variant in the GTPase effector domain (GED) of DNM1L leads to static encephalopathy and seizures. Journal of Physical Education and Sports Management, 2019, 5, a003673.	1.2	24
29	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. Molecular Genetics and Metabolism, 2019, 127, 45-50.	1.1	14
30	Improved clinical outcome following liver transplant in patients with ethylmalonic encephalopathy. American Journal of Medical Genetics, Part A, 2019, 179, 1015-1019.	1.2	30
31	L-Cysteine supplementation prevents liver transplantation in a patient with TRMU deficiency. Molecular Genetics and Metabolism Reports, 2019, 19, 100453.	1.1	16
32	SLC35A2â€CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	2.5	39
33	GNA11 brain somatic pathogenic variant in an individual with phacomatosis pigmentovascularis. Neurology: Genetics, 2019, 5, e366.	1.9	4
34	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. American Journal of Human Genetics, 2019, 104, 213-228.	6.2	90
35	LIPT1 deficiency presenting as early infantile epileptic encephalopathy, Leigh disease, and secondary pyruvate dehydrogenase complex deficiency. American Journal of Medical Genetics, Part A, 2018, 176, 1184-1189.	1.2	24
36	Genotype-phenotype correlations in individuals with pathogenicREREvariants. Human Mutation, 2018, 39, 666-675.	2.5	34

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37	The expanding neurological phenotype of DNM1L-related disorders. Brain, 2018, 141, e28-e28.	7.6	7
38	Mitochondrial dynamics: Biological roles, molecular machinery, and related diseases. Molecular Genetics and Metabolism, 2018, 125, 315-321.	1.1	99
39	Mitochondrial DNA replication: clinical syndromes. Essays in Biochemistry, 2018, 62, 297-308.	4.7	27
40	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. American Journal of Human Genetics, 2017, 100, 343-351.	6.2	35
41	Mitochondrial DNA maintenance defects. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 1539-1555.	3.8	205
42	Arginine and Citrulline for the Treatment of MELAS Syndrome. FIRE Forum for International Research in Education, 2017, 5, 232640981769739.	0.7	40
43	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	8.2	184
44	Mutations in the Chromatin Regulator Gene BRPF1 Cause Syndromic Intellectual Disability and Deficient Histone Acetylation. American Journal of Human Genetics, 2017, 100, 91-104.	6.2	72
45	Therapies for mitochondrial diseases and current clinical trials. Molecular Genetics and Metabolism, 2017, 122, 1-9.	1.1	140
46	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	6.2	348
47	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
48	De Novo Mutations in SLC25A24 Cause a Craniosynostosis Syndrome with Hypertrichosis, Progeroid Appearance, and Mitochondrial Dysfunction. American Journal of Human Genetics, 2017, 101, 833-843.	6.2	56
49	Biallelic mutations in the ferredoxin reductase gene cause novel mitochondriopathy with optic atrophy. Human Molecular Genetics, 2017, 26, 4937-4950.	2.9	32
50	Milder clinical and biochemical phenotypes associated with the c.482G > A (p.Arg161Gln) pathogenic variant in cobalamin C disease: Implications for management and screening. Molecular Genetics and Metabolism, 2017, 122, 60-66.	1.1	20
51	Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. Genetics in Medicine, 2017, 19, 1380-1397.	2.4	173
52	Clinical and molecular characterization of de novo loss of function variants in HNRNPU. American Journal of Medical Genetics, Part A, 2017, 173, 2680-2689.	1.2	34
53	Mitochondrial Cardiomyopathies. Frontiers in Cardiovascular Medicine, 2016, 3, 25.	2.4	149
54	Corneal clouding, cataract, and colobomas with a novel missense mutation in B4GALT7â€"a review of eye anomalies in the linkeropathy syndromes. American Journal of Medical Genetics, Part A, 2016, 170, 2711-2718.	1.2	19

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55	Solid organ transplantation in primary mitochondrial disease: Proceed with caution. Molecular Genetics and Metabolism, 2016, 118, 178-184.	1.1	55
56	Missense variants in the middle domain ofDNM1Lin cases of infantile encephalopathy alter peroxisomes and mitochondria when assayed inDrosophila. Human Molecular Genetics, 2016, 25, 1846-1856.	2.9	62
57	Mitochondrial cytopathies. Cell Calcium, 2016, 60, 199-206.	2.4	67
58	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. American Journal of Human Genetics, 2016, 98, 347-357.	6.2	98
59	Impaired nitric oxide production in children with MELAS syndrome and the effect of arginine and citrulline supplementation. Molecular Genetics and Metabolism, 2016, 117, 407-412.	1.1	64
60	Recommendations for the Management of Strokelike Episodes in Patients With Mitochondrial Encephalomyopathy, Lactic Acidosis, and Strokelike Episodes. JAMA Neurology, 2016, 73, 591.	9.0	94
61	Molybdenum cofactor deficiency. Molecular Genetics and Metabolism, 2016, 117, 1-4.	1.1	86
62	Methionyl-tRNA Formyltransferase (MTFMT) Deficiency Mimicking Acquired Demyelinating Disease. Journal of Child Neurology, 2016, 31, 215-219.	1.4	5
63	Improvement of regressive autism symptoms in a child with TMLHE deficiency following carnitine supplementation. American Journal of Medical Genetics, Part A, 2015, 167, 2162-2167.	1.2	30
64	Adult liver disorders caused by inborn errors of metabolism: Review and update. Molecular Genetics and Metabolism, 2015, 114, 1-10.	1.1	21
65	Diagnosis and management of mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. Genetics in Medicine, 2015, 17, 689-701.	2.4	414
66	MELAS syndrome: Clinical manifestations, pathogenesis, and treatment options. Molecular Genetics and Metabolism, 2015, 116, 4-12.	1.1	431
67	Disorders of carnitine biosynthesis and transport. Molecular Genetics and Metabolism, 2015, 116, 107-112.	1.1	90
68	Obstetric and Infant Outcomes Following Planned Maternal Third Trimester Exposure to Tenofovir 1% Vaginal Gel. AIDS Research and Human Retroviruses, 2014, 30, A267-A268.	1.1	0
69	Glucose metabolism derangements in adults with the MELAS m.3243A>G mutation. Mitochondrion, 2014, 18, 63-69.	3.4	32
70	Early onset and severe clinical course associated with the m.5540G>A mutation in MT - TW. Molecular Genetics and Metabolism Reports, 2014, 1, 61-65.	1.1	3
71	Combined array CGH plus SNP genome analyses in a single assay for optimized clinical testing. European Journal of Human Genetics, 2014, 22, 79-87.	2.8	112
72	Practice patterns of mitochondrial disease physicians in North America. Part 1: Diagnostic and clinical challenges. Mitochondrion, 2014, 14, 26-33.	3.4	36

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73	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. Molecular Genetics and Metabolism, 2014, 113, 207-212.	1.1	63
74	Mitochondria: Role of citrulline and arginine supplementation in MELAS syndrome. International Journal of Biochemistry and Cell Biology, 2014, 48, 85-91.	2.8	40
75	Guanidinoacetate methyltransferase (GAMT) deficiency: Outcomes in 48 individuals and recommendations for diagnosis, treatment and monitoring. Molecular Genetics and Metabolism, 2014, 111, 16-25.	1.1	111
76	The effect of citrulline and arginine supplementation on lactic acidemia in MELAS syndrome. Meta Gene, 2013, 1, 8-14.	0.6	17
77	Molecular and clinical characterization of the myopathic form of mitochondrial DNA depletion syndrome caused by mutations in the thymidine kinase (TK2) gene. Molecular Genetics and Metabolism, 2013, 110, 153-161.	1.1	40
78	Transition to Next Generation Analysis of the Whole Mitochondrial Genome: A Summary of Molecular Defects. Human Mutation, 2013, 34, 882-893.	2.5	79
79	Mitochondrial DNA Depletion Syndromes: Review and Updates of Genetic Basis, Manifestations, and Therapeutic Options. Neurotherapeutics, 2013, 10, 186-198.	4.4	255
80	Citrulline and arginine utility in treating nitric oxide deficiency in mitochondrial disorders. Molecular Genetics and Metabolism, 2012, 107, 247-252.	1.1	84
81	Restoration of impaired nitric oxide production in MELAS syndrome with citrulline and arginine supplementation. Molecular Genetics and Metabolism, 2012, 105, 607-614.	1.1	127
82	Nuclear Gene Defects in Mitochondrial Disorders. Methods in Molecular Biology, 2012, 837, 17-34.	0.9	11
83	Atypical presentation of Leigh syndrome associated with a Leber hereditary optic neuropathy primary mitochondrial DNA mutation. Molecular Genetics and Metabolism, 2011, 103, 153-160.	1.1	24
84	Deoxyguanosine kinase deficiency presenting as neonatal hemochromatosis. Molecular Genetics and Metabolism, 2011, 103, 262-267.	1.1	33
85	Early-onset severe neuromuscular phenotype associated with compound heterozygosity for OPA1 mutations. Molecular Genetics and Metabolism, 2011, 103, 383-387.	1.1	71
86	Insights into the Pathogenesis and Treatment of Cancer from Inborn Errors of Metabolism. American Journal of Human Genetics, 2011, 88, 402-421.	6.2	58
87	Rhombencephalosynapsis is a malformation deserving of further study. American Journal of Medical Genetics, Part A, 2011, 155, 2902-2902.	1.2	0
88	Molecular spectrum of SLC22A5 (OCTN2) gene mutations detected in 143 subjects evaluated for systemic carnitine deficiency. Human Mutation, 2010, 31, E1632-E1651.	2.5	61
89	Maternal systemic primary carnitine deficiency uncovered by newborn screening: Clinical, biochemical, and molecular aspects. Genetics in Medicine, 2010, 12, 19-24.	2.4	91
90	New insights in nutritional management and amino acid supplementation in urea cycle disorders. Molecular Genetics and Metabolism, 2010, 100, S72-S76.	1.1	24

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91	Milder clinical course of Type IV 3-methylglutaconic aciduria due to a novel mutation in TMEM70. Molecular Genetics and Metabolism, 2010, 101, 282-285.	1.1	25
92	Tetrasomy 13q mosaicism associated with phylloid hypomelanosis and precocious puberty. American Journal of Medical Genetics, Part A, 2009, 149A, 993-996.	1.2	22
93	The Xp contiguous deletion syndrome and autism. American Journal of Medical Genetics, Part A, 2009, 149A, 1138-1148.	1.2	19
94	Citrin deficiency, a perplexing global disorder. Molecular Genetics and Metabolism, 2009, 96, 44-49.	1.1	81
95	Human mitochondrial transfer RNAs: Role of pathogenic mutation in disease. Muscle and Nerve, 2008, 37, 150-171.	2.2	68
96	Abnormal neurological features predict poor survival and should preclude liver transplantation in patients with deoxyguanosine kinase deficiency. Liver Transplantation, 2008, 14, 1480-1485.	2.4	67
97	GM1 gangliosidosis: Review of clinical, molecular, and therapeutic aspects. Molecular Genetics and Metabolism, 2008, 94, 391-396.	1.1	359
98	Utility of Oligonucleotide Arrayâ€"Based Comparative Genomic Hybridization for Detection of Target Gene Deletions. Clinical Chemistry, 2008, 54, 1141-1148.	3.2	78
99	Mitochondrial immunofluorescence assay as an adjunctive tool in the diagnosis of mitochondrial myopathy. FASEB Journal, 2008, 22, 708.20.	0.5	0
100	Citrin Deficiency: A Novel Cause of Failure to Thrive That Responds to a High-Protein, Low-Carbohydrate Diet. Pediatrics, 2007, 119, e773-e777.	2.1	81
101	Phenylbutyrate reduces plasma leucine concentrations without affecting the flux of leucine. FASEB Journal, 2007, 21, A335.	0.5	0
102	The Mitochondrial Myopathy Encephalopathy, Lactic Acidosis with??Stroke-Like Episodes (MELAS) Syndrome. CNS Drugs, 2006, 20, 443-464.	5.9	75
103	Epimerase-Deficiency Galactosemia Is Not a Binary Condition. American Journal of Human Genetics, 2006, 78, 89-102.	6.2	77
104	Molecular bases of hearing loss in multi-systemic mitochondrial cytopathy. Genetics in Medicine, 2006, 8, 641-652.	2.4	9
105	Predominant cerebellar volume loss as a neuroradiologic feature of pediatric respiratory chain defects. American Journal of Neuroradiology, 2005, 26, 1675-80.	2.4	47
106	Clinical Consequences of Urea Cycle Enzyme Deficiencies and Potential Links to Arginine and Nitric Oxide Metabolism. Journal of Nutrition, 2004, 134, 2775S-2782S.	2.9	76
107	Clinical Spectrum, Morbidity, and Mortality in 113 Pediatric Patients With Mitochondrial Disease. Pediatrics, 2004, 114, 925-931.	2.1	431
108	In reply to: Proving pathogenicity?when evolution is not enough. American Journal of Medical Genetics Part A, 2004, 131A, 109-110.	2.4	0

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109	Effect of alternative pathway therapy on branched chain amino acid metabolism in urea cycle disorder patients. Molecular Genetics and Metabolism, 2004, 81, 79-85.	1.1	93
110	Novel homoplasmic mutation in the mitochondrialtRNATyr gene associated with atypical mitochondrial cytopathy presenting with focal segmental glomerulosclerosis. American Journal of Medical Genetics Part A, 2003, 123A, 172-178.	2.4	52
111	Differential utilization of systemic and enteral ammonia for urea synthesis in control subjects and ornithine transcarbamylase deficiency carriers. American Journal of Clinical Nutrition, 2003, 78, 749-755.	4.7	7
112	An Integrated Approach to the Diagnosis and Prospective Management of Partial Ornithine Transcarbamylase Deficiency. Pediatrics, 2002, 109, 150-152.	2.1	37
113	Neonatal presentation of ventricular tachycardia and a Reye-like syndrome episode associated with disturbed mitochondrial energy metabolism. BMC Pediatrics, 2002, 2, 12.	1.7	13
114	Compensating for central nervous system dysmyelination: Females with a proteolipid protein gene duplication and sustained clinical improvement. Annals of Neurology, 2001, 50, 747-754.	5.3	53
115	Deletion (9) (p13.1 p21.1). American Journal of Medical Genetics Part A, 2000, 91, 113-115.	2.4	0
116	Primary and secondary alterations of neonatal carnitine metabolism. Seminars in Perinatology, 1999, 23, 152-161.	2.5	80
117	Functional Characterization of the Carnitine Transporter Defective in Primary Carnitine Deficiency. Archives of Biochemistry and Biophysics, 1999, 364, 99-106.	3.0	30