# Fernando Scaglia

List of Publications by Year
in descending order

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

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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 | Clinical Spectrum, Morbidity, and Mortality in 113 Pediatric Patients With Mitochondrial Disease. Pediatrics, 2004, 114, 925-931. | 2.1 | 431 |
| 2 | MELAS syndrome: Clinical manifestations, pathogenesis, and treatment options. Molecular Genetics and Metabolism, 2015, 116, 4-12. | 1.1 | 431 |
| 3 | Diagnosis and management of mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. Genetics in Medicine, 2015, 17, 689-701. | 2.4 | 414 |
| 4 | GM1 gangliosidosis: Review of clinical, molecular, and therapeutic aspects. Molecular Genetics and Metabolism, 2008, 94, 391-396. | 1.1 | 359 |
| 5 | Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438. | 6.2 | 348 |
| 6 | High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685. | 6.2 | 337 |
| 7 | Mitochondrial DNA Depletion Syndromes: Review and Updates of Genetic Basis, Manifestations, and Therapeutic Options. Neurotherapeutics, 2013, 10, 186-198. | 4.4 | 255 |
| 8 | Mitochondrial DNA maintenance defects. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 1539-1555. | 3.8 | 205 |
| 9 | Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480. | 27.0 | 205 |
| 10 | Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26. | 8.2 | 184 |
| 11 | 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 |
| 12 | Mitochondrial Cardiomyopathies. Frontiers in Cardiovascular Medicine, 2016, 3, 25. | 2.4 | 149 |
| 13 | Therapies for mitochondrial diseases and current clinical trials. Molecular Genetics and Metabolism, 2017, 122, 1-9. | 1.1 | 140 |
| 14 | 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 |
| 15 | 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 |
| 16 | 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 |
| 17 | Mitochondrial dynamics: Biological roles, molecular machinery, and related diseases. Molecular Genetics and Metabolism, 2018, 125, 315-321. | 1.1 | 99 |
| 18 | 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 |

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|---|---|---|---|
| 19 | 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 |
| 20 | 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 |
| 21 | Maternal systemic primary carnitine deficiency uncovered by newborn screening: Clinical, biochemical, and molecular aspects. Genetics in Medicine, 2010, 12, 19-24. | 2.4 | 91 |
| 22 | Disorders of carnitine biosynthesis and transport. Molecular Genetics and Metabolism, 2015, 116, 107-112. | 1.1 | 90 |
| 23 | 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 |
| 24 | Molybdenum cofactor deficiency. Molecular Genetics and Metabolism, 2016, 117, 1-4. | 1.1 | 86 |
| 25 | Citrulline and arginine utility in treating nitric oxide deficiency in mitochondrial disorders. Molecular Genetics and Metabolism, 2012, 107, 247-252. | 1.1 | 84 |
| 26 | 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 |
| 27 | Citrin deficiency, a perplexing global disorder. Molecular Genetics and Metabolism, 2009, 96, 44-49. | 1.1 | 81 |
| 28 | Primary and secondary alterations of neonatal carnitine metabolism. Seminars in Perinatology, 1999, 23, 152-161. | 2.5 | 80 |
| 29 | Transition to Next Generation Analysis of the Whole Mitochondrial Genome: A Summary of Molecular Defects. Human Mutation, 2013, 34, 882-893. | 2.5 | 79 |
| 30 | Utility of Oligonucleotide Arrayâ€“Based Comparative Genomic Hybridization for Detection of Target Gene Deletions. Clinical Chemistry, 2008, 54, 1141-1148. | 3.2 | 78 |
| 31 | Epimerase-Deficiency Galactosemia Is Not a Binary Condition. American Journal of Human Genetics, 2006, 78, 89-102. | 6.2 | 77 |
| 32 | 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 |
| 33 | The Mitochondrial Myopathy Encephalopathy, Lactic Acidosis with??Stroke-Like Episodes (MELAS) Syndrome. CNS Drugs, 2006, 20, 443-464. | 5.9 | 75 |
| 34 | 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 |
| 35 | Early-onset severe neuromuscular phenotype associated with compound heterozygosity for OPA1 mutations. Molecular Genetics and Metabolism, 2011, 103, 383-387. | 1.1 | 71 |
| 36 | Human mitochondrial transfer RNAs: Role of pathogenic mutation in disease. Muscle and Nerve, 2008, 37, 150-171. | 2.2 | 68 |

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|---|---|---|---|
| 37 | 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 |
| 38 | Mitochondrial cytopathies. Cell Calcium, 2016, 60, 199-206. | 2.4 | 67 |
| 39 | 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 |
| 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. Molecular Genetics and Metabolism, 2014, 113, 207-212. | 1.1 | 63 |
| 41 | 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 |
| 42 | 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 |
| 43 | 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 |
| 44 | 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 |
| 45 | Solid organ transplantation in primary mitochondrial disease: Proceed with caution. Molecular Genetics and Metabolism, 2016, 118, 178-184. | 1.1 | 55 |
| 46 | 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 |
| 47 | 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 |
| 48 | 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 |
| 49 | Predominant cerebellar volume loss as a neuroradiologic feature of pediatric respiratory chain defects. American Journal of Neuroradiology, 2005, 26, 1675-80. | 2.4 | 47 |
| 50 | Clinical trials in mitochondrial disorders, an update. Molecular Genetics and Metabolism, 2020, 131, 1-13. | 1.1 | 44 |
| 51 | Clinical and biochemical improvement with galactose supplementation in SLC35A2-CDG. Genetics in Medicine, 2020, 22, 1102-1107. | 2.4 | 42 |
| 52 | 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 |
| 53 | Mitochondria: Role of citrulline and arginine supplementation in MELAS syndrome. International Journal of Biochemistry and Cell Biology, 2014, 48, 85-91. | 2.8 | 40 |
| 54 | Arginine and Citrulline for the Treatment of MELAS Syndrome. FIRE Forum for International Research in Education, 2017, 5, 232640981769739. | 0.7 | 40 |

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|---|---|---|---|
| 55 | SLC35A2â€CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925. | 2.5 | 39 |
| 56 | Safety and efficacy of omaveloxolone in patients with mitochondrial myopathy. Neurology, 2020, 94, e687-e698. | 1.1 | 38 |
| 57 | Mitochondrial diseases in North America. Neurology: Genetics, 2020, 6, e402. | 1.9 | 38 |
| 58 | An Integrated Approach to the Diagnosis and Prospective Management of Partial Ornithine Transcarbamylase Deficiency. Pediatrics, 2002, 109, 150-152. | 2.1 | 37 |
| 59 | Practice patterns of mitochondrial disease physicians in North America. Part 1: Diagnostic and clinical challenges. Mitochondrion, 2014, 14, 26-33. | 3.4 | 36 |
| 60 | 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 |
| 61 | 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 |
| 62 | 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 |
| 63 | Genotype-phenotype correlations in individuals with pathogenicREREvariants. Human Mutation, 2018, 39, 666-675. | 2.5 | 34 |
| 64 | Deoxyguanosine kinase deficiency presenting as neonatal hemochromatosis. Molecular Genetics and Metabolism, 2011, 103, 262-267. | 1.1 | 33 |
| 65 | Glucose metabolism derangements in adults with the MELAS m.3243A>G mutation. Mitochondrion, 2014, 18, 63-69. | 3.4 | 32 |
| 66 | Biallelic mutations in the ferredoxin reductase gene cause novel mitochondriopathy with optic atrophy. Human Molecular Genetics, 2017, 26, 4937-4950. | 2.9 | 32 |
| 67 | Functional Characterization of the Carnitine Transporter Defective in Primary Carnitine Deficiency. Archives of Biochemistry and Biophysics, 1999, 364, 99-106. | 3.0 | 30 |
| 68 | 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 |
| 69 | 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 |
| 70 | Mitochondrial DNA replication: clinical syndromes. Essays in Biochemistry, 2018, 62, 297-308. | 4.7 | 27 |
| 71 | Successful liver transplantation in mitochondrial neurogastrointestinal encephalomyopathy (MNGIE). Molecular Genetics and Metabolism, 2020, 130, 58-64. | 1.1 | 26 |
| 72 | 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 |

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|---|---|---|---|
| 73 | New insights in nutritional management and amino acid supplementation in urea cycle disorders. Molecular Genetics and Metabolism, 2010, 100, S72-S76. | 1.1 | 24 |
| 74 | 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 |
| 75 | 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 |
| 76 | 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 |
| 77 | Tetrasomy 13q mosaicism associated with phylloid hypomelanosis and precocious puberty. American Journal of Medical Genetics, Part A, 2009, 149A, 993-996. | 1.2 | 22 |
| 78 | Adult liver disorders caused by inborn errors of metabolism: Review and update. Molecular Genetics and Metabolism, 2015, 114, 1-10. | 1.1 | 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. Molecular Genetics and Metabolism, 2017, 122, 60-66. | 1.1 | 20 |
| 80 | The Xp contiguous deletion syndrome and autism. American Journal of Medical Genetics, Part A, 2009, 149A, 1138-1148. | 1.2 | 19 |
| 81 | 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 |
| 82 | 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 |
| 83 | 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 |
| 84 | The effect of citrulline and arginine supplementation on lactic acidemia in MELAS syndrome. Meta Gene, 2013, 1, 8-14. | 0.6 | 17 |
| 85 | L-Cysteine supplementation prevents liver transplantation in a patient with TRMU deficiency. Molecular Genetics and Metabolism Reports, 2019, 19, 100453. | 1.1 | 16 |
| 86 | 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 |
| 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. Molecular Genetics and Metabolism, 2019, 127, 45-50. | 1.1 | 14 |
| 88 | 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 |
| 89 | TRMU deficiency: A broad clinical spectrum responsive to cysteine supplementation. Molecular Genetics and Metabolism, 2021, 132, 146-153. | 1.1 | 13 |
| 90 | 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 |

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|---|---|---|---|
| 91 | Nuclear Gene Defects in Mitochondrial Disorders. Methods in Molecular Biology, 2012, 837, 17-34. | 0.9 | 11 |
| 92 | 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 |
| 93 | Characterization of the renal phenotype in RMND1 â€related mitochondrial disease. Molecular Genetics & Genomic Medicine, 2019, 7, e973. | 1.2 | 10 |
| 94 | Effective Aspirin Treatment of Women at Risk for Preeclampsia Delays the Metabolic Clock of Gestation. Hypertension, 2021, 78, 1398-1410. | 2.7 | 10 |
| 95 | Molecular bases of hearing loss in multi-systemic mitochondrial cytopathy. Genetics in Medicine, 2006, 8, 641-652. | 2.4 | 9 |
| 96 | 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 |
| 97 | Expansion of the clinical phenotype of <scp>GALE</scp> deficiency. American Journal of Medical Genetics, Part A, 2021, 185, 3118-3121. | 1.2 | 8 |
| 98 | 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 |
| 99 | The expanding neurological phenotype of DNM1L-related disorders. Brain, 2018, 141, e28-e28. | 7.6 | 7 |
| 100 | Niacin therapy improves outcome and normalizes metabolic abnormalities in an NAXD-deficient patient. Brain, 2022, 145, e36-e40. | 7.6 | 6 |
| 101 | Methionyl-tRNA Formyltransferase (MTFMT) Deficiency Mimicking Acquired Demyelinating Disease. Journal of Child Neurology, 2016, 31, 215-219. | 1.4 | 5 |
| 102 | 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 |
| 103 | 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 |
| 104 | GNA11 brain somatic pathogenic variant in an individual with phacomatosis pigmentovascularis. Neurology: Genetics, 2019, 5, e366. | 1.9 | 4 |
| 105 | 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 |
| 106 | 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 |
| 107 | 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 |
| 108 | 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 |

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|---|---|---|---|
| 109 | 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 |
| 110 | Deletion (9) (p13.1 p21.1). American Journal of Medical Genetics Part A, 2000, 91, 113-115. | 2.4 | 0 |
| 111 | In reply to: Proving pathogenicity?when evolution is not enough. American Journal of Medical Genetics Part A, 2004, 131A, 109-110. | 2.4 | 0 |
| 112 | Rhombencephalosynapsis is a malformation deserving of further study. American Journal of Medical Genetics, Part A, 2011, 155, 2902-2902. | 1.2 | 0 |
| 113 | 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 |
| 114 | 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 |
| 115 | Phenylbutyrate reduces plasma leucine concentrations without affecting the flux of leucine. FASEB Journal, 2007, 21, A335. | 0.5 | 0 |
| 116 | Mitochondrial immunofluorescence assay as an adjunctive tool in the diagnosis of mitochondrial myopathy. FASEB Journal, 2008, 22, 708.20. | 0.5 | 0 |
| 117 | Mitochondrial disorders. , 2020, , 283-304. | | 0 |