Mitochondrial disorders : The Essentials

3rd quarter 2024

Quarterly literature follow-up
carried out by the AFM-Téléthon documentation department

This literature follow-up, carried out based on queries on PubMed® without claiming to be exhaustive, presents a selection of references to medical-scientific articles concerning the field of mitochondrial diseases.

Summary

Ataxie de Friedreich - *Friedreich ataxia*

1. Baille, G., Geoffre, N., Wissocq, A., Planté-Bordeneuve, P., Mutez, E., & Huin, V. (2024). Early-onset phenotype in a patient with an intermediate allele and a large SCA1 expansion : A case report. *BMC Neurology*, *24*(1), 348. https://doi.org/10.1186/s12883-024-03846-2

2. Beaudin, M., Dupre, N., & Manto, M. (2024). The importance of synthetic pharmacotherapy for recessive cerebellar ataxias. *Expert Review of Neurotherapeutics*, *24*(9), 897-912. https://doi.org/10.1080/14737175.2024.2376840

3. Cocozza, S., Bosticardo, S., Battocchio, M., Corben, L., Delatycki, M., Egan, G., Georgiou-Karistianis, N., Monti, S., Palma, G., Pane, C., Saccà, F., Schiavi, S., Selvadurai, L., Tranfa, M., Daducci, A., Brunetti, A., & Harding, I. H. (2024). Gradient of microstructural damage along the dentato-thalamo-cortical tract in Friedreich ataxia. *Annals of Clinical and Translational Neurology*, *11*(7), 1691-1702. https://doi.org/10.1002/acn3.52048

4. Della Valle, I., Milani, M., Rossi, S., Turchi, R., Tortolici, F., Nesci, V., Ferri, A., Valle, C., Lettieri-Barbato, D., Aquilano, K., Cozzolino, M., Apolloni, S., & D'Ambrosi, N. (2024). Loss of homeostatic functions in microglia from a murine model of Friedreich's ataxia. *Genes & Diseases*, *11*(6), 101178. https://doi.org/10.1016/j.gendis.2023.101178

5. Eisel, M. L. S., Burns, M., Ashizawa, T., Byrne, B., Corti, M., & Subramony, S. H. (2024). Emerging therapies in hereditary ataxias. *Trends in Molecular Medicine*, S1471-4914(24)00194-1. https://doi.org/10.1016/j.molmed.2024.07.008

6. Indelicato, E., Wanschitz, J., Löscher, W., & Boesch, S. (2024). Skeletal Muscle Involvement in Friedreich Ataxia. *International Journal of Molecular Sciences*, *25*(18), 9915. https://doi.org/10.3390/ijms25189915

7. Lai, Y., Diaz, N., Armbrister, R., Agoulnik, I., & Liu, Y. (2024). DNA Base Damage Repair Crosstalks with Chromatin Structures to Contract Expanded GAA Repeats in Friedreich's Ataxia. *Biomolecules*, *14*(7), 809. https://doi.org/10.3390/biom14070809

8. Lee, L., Okudaira, N., Murase, K., Kong, R., & Jones, H. M. (2024). Determination of Vatiquinone Drug-Drug Interactions, as CYP450 Perpetrator and Victim, Using Physiologically Based Pharmacokinetic (PBPK) Modeling and Simulation. *Journal of Clinical Pharmacology*. https://doi.org/10.1002/jcph.6133

9. Lee, L., Thoolen, M., Ma, J., Kaushik, D., Golden, L., & Kong, R. (2024). Effect of Itraconazole, a CYP3A4 Inhibitor, and Rifampin, a CYP3A4 Inducer, on the Pharmacokinetics of Vatiquinone. *Clinical Pharmacology in Drug Development*. https://doi.org/10.1002/cpdd.1461

10. Lehmer, M., & Zoncu, R. (2024). mTORC1 Signaling Inhibition Modulates Mitochondrial Function in Frataxin Deficiency. *bioRxiv: The Preprint Server for Biology*, 2024.08.06.606942. https://doi.org/10.1101/2024.08.06.606942

11. Louis, E. D., Kuo, S.-H., & Faust, P. L. (2024). Purkinje Cell Dendritic Swellings : A Postmortem Study of Essential Tremor and Other Cerebellar Degenerative Disorders. *Cerebellum (London, England)*. https://doi.org/10.1007/s12311-024-01739-1

12. Mathew, A. M., Bhuvanendran, S., Nair, R. S., & K Radhakrishnan, A. (2023). Exploring the anti-inflammatory activities, mechanism of action and prospective drug delivery systems of tocotrienol to target neurodegenerative diseases. *F1000Research*, *12*, 338. https://doi.org/10.12688/f1000research.131863.1

13. Mosbach, V., & Puccio, H. (2024). A multiple animal and cellular models approach to study frataxin deficiency in Friedreich Ataxia. *Biochimica Et Biophysica Acta. Molecular Cell Research*, *1871*(7), 119809. https://doi.org/10.1016/j.bbamcr.2024.119809

14. Nishiyama, M., Kalambogias, J., Imai, F., Yang, E., Lang, S., de Nooij, J. C., & Yoshida, Y. (2024). Anatomical and functional analysis of the corticospinal tract in an FRDA mouse model. *bioRxiv: The Preprint Server for Biology*, 2024.06.28.601178. https://doi.org/10.1101/2024.06.28.601178

15. Pall, A. E., Bond, S., Bailey, D. K., Stoj, C. S., Deschamps, I., Huggins, P., Parsons, J., Bradbury, M. J., Kosman, D. J., & Stemmler, T. L. (2024). ATH434, a promising iron-targeting compound for treating iron regulation disorders. *Metallomics: Integrated Biometal Science*, *16*(10), mfae044. https://doi.org/10.1093/mtomcs/mfae044

16. Paparella, G., Stragà, C., Pesenti, N., Dal Molin, V., Martorel, G. A., Merotto, V., Genova, C., Piazza, A., Piccoli, G., Panzeri, E., Rufini, A., Testi, R., & Martinuzzi, A. (2024). A Pilot Phase 2 Randomized Trial to Evaluate the Safety and Potential Efficacy of Etravirine in Friedreich Ataxia Patients. *Children (Basel, Switzerland)*, *11*(8), 958. https://doi.org/10.3390/children11080958

17. Pepin, X. J. H., Hynes, S. M., Zahir, H., Walker, D., Semmens, L. Q., & Suarez-Sharp, S. (2024). Understanding the mechanisms of food effect on omaveloxolone pharmacokinetics through physiologically based biopharmaceutics modeling. *CPT: Pharmacometrics & Systems Pharmacology*, *13*(10), 1771-1783. https://doi.org/10.1002/psp4.13221

18. Perry, C. E., Halawani, S. M., Mukherjee, S., Ngaba, L. V., Lieu, M., Lee, W. D., Davis, J. G., Adzika, G. K., Bebenek, A. N., Bazianos, D. D., Chen, B., Mercado-Ayon, E., Flatley, L. P., Suryawanshi, A. P., Ho, I., Rabinowitz, J. D., Serai, S. D., Biko, D. M., Tamaroff, J., … Baur, J. A. (2024). NAD+ precursors prolong survival and improve cardiac phenotypes in a mouse model of Friedreich's Ataxia. *JCI Insight*, *9*(16), e177152. https://doi.org/10.1172/jci.insight.177152

19. Quinlan, A., Rodan, L., Barkoudah, E., Tam, A., Saffari, A., Shammas, I., Ranatunga, W., Morava-Kozicz, E., Oglesbee, D., Berry, G., Ebrahimi-Fakhari, D., & Srivastava, S. (2024). Case Report of Friedreich's Ataxia and ALG1-Related Biochemical Abnormalities in a Patient With Progressive Spastic Paraplegia. *American Journal of Medical Genetics. Part A*, e63890. https://doi.org/10.1002/ajmg.a.63890

20. Rebs, S., & Streckfuss-Bömeke, K. (2023). How can we use stem cell-derived cardiomyocytes to understand the involvement of energetic metabolism in alterations of cardiac function? *Frontiers in Molecular Medicine*, *3*, 1222986. https://doi.org/10.3389/fmmed.2023.1222986

21. Sanz-Alcázar, A., Portillo-Carrasquer, M., Delaspre, F., Pazos-Gil, M., Tamarit, J., Ros, J., & Cabiscol, E. (2024). Deciphering the ferroptosis pathways in dorsal root ganglia of Friedreich ataxia models. The role of LKB1/AMPK, KEAP1, and GSK3β in the impairment of the NRF2 response. *Redox Biology*, *76*, 103339. https://doi.org/10.1016/j.redox.2024.103339

22. Scarabino, D., Veneziano, L., Nethisinghe, S., Mantuano, E., Fiore, A., Granata, G., Solanky, N., Zanni, G., Cavalcanti, F., Corbo, R. M., & Giunti, P. (2024). Unusual Age-Dependent Behavior of Leukocytes Telomere Length in Friedreich's Ataxia. *Movement Disorders: Official Journal of the Movement Disorder Society*. https://doi.org/10.1002/mds.29976

23. Scott, V., Delatycki, M. B., Tai, G., & Corben, L. A. (2024). New and Emerging Drug and Gene Therapies for Friedreich Ataxia. *CNS Drugs*, *38*(10), 791-805. https://doi.org/10.1007/s40263-024-01113-z

24. Sohns, E., Szmulewicz, D. J., & Tarnutzer, A. A. (2024). Oculomotor and Vestibular Deficits in Friedreich Ataxia—Systematic Review and Meta-Analysis of Quantitative Measurements. *Cerebellum (London, England)*. https://doi.org/10.1007/s12311-024-01716-8

25. Tamaroff, J., Nguyen, S., Wilson, N. E., Stefanovski, D., Xiao, R., Scattergood, T., Capiola, C., Schur, G. M., Dunn, J., Dedio, A., Wade, K., Shah, H., Sharma, R., Mootha, V. K., Kelly, A., Lin, K. Y., Lynch, D. R., Reddy, R., Rickels, M. R., & McCormack, S. E. (2024). Insulin sensitivity and insulin secretion in adults with Friedreich's Ataxia : The role of skeletal muscle. *The Journal of Clinical Endocrinology and Metabolism*, dgae545. https://doi.org/10.1210/clinem/dgae545

26. Theodosopoulou, P., Mavromati, M., & Paraskeva, A. (2024). Friedreich's Ataxia and Cesarean Delivery : A Case Report of Epidural Anesthesia With Ropivacaine. *Cureus*, *16*(6), e61776. https://doi.org/10.7759/cureus.61776

27. Vicente-Acosta, A., Herranz-Martín, S., Pazos, M. R., Galán-Cruz, J., Amores, M., Loria, F., & Díaz-Nido, J. (2024). Glial cell activation precedes neurodegeneration in the cerebellar cortex of the YG8-800 murine model of Friedreich ataxia. *Neurobiology of Disease*, *200*, 106631. https://doi.org/10.1016/j.nbd.2024.106631

28. Wu, L., Huang, F., Yang, L., Yang, L., Sun, Z., Zhang, J., Xia, S., Zhao, H., Ding, Y., Bian, D., & Li, K. (2024). Interplay of FXN expression and lipolysis in white adipocytes plays a critical role in insulin sensitivity in Friedreich's ataxia mouse model. *Scientific Reports*, *14*(1), 19876. https://doi.org/10.1038/s41598-024-71099-7

29. Wu, Y., Zhang, Y., Ge, L., He, S., Zhang, Y., Chen, D., Nie, Y., Zhu, M., & Pang, Q. (2024). RTA408 alleviates lipopolysaccharide-induced acute lung injury via inhibiting Bach1-mediated ferroptosis. *International Immunopharmacology*, *142*(Pt B), 113250. https://doi.org/10.1016/j.intimp.2024.113250

Atrophie optique autosomique dominante – *Autosomal dominant optic atrophy (ADOA)*

30. Costello, S. M., Schultz, A., Smith, D., Horan, D., Chaverra, M., Tripet, B., George, L., Bothner, B., Lefcort, F., & Copié, V. (2024). Metabolic Deficits in the Retina of a Familial Dysautonomia Mouse Model. *Metabolites*, *14*(8), 423. https://doi.org/10.3390/metabo14080423

31. García-López, M., Jiménez-Vicente, L., González-Jabardo, R., Dorado, H., Gómez-Manjón, I., Martín, M. Á., Ayuso, C., Arenas, J., & Gallardo, M. E. (2024). Creation of an Isogenic Human iPSC-Based RGC Model of Dominant Optic Atrophy Harboring the Pathogenic Variant c.1861C>T (p.Gln621Ter) in the OPA1 Gene. *International Journal of Molecular Sciences*, *25*(13), 7240. https://doi.org/10.3390/ijms25137240

32. Nitta, Y., Osaka, J., Maki, R., Hakeda-Suzuki, S., Suzuki, E., Ueki, S., Suzuki, T., & Sugie, A. (2024). Drosophila model to clarify the pathological significance of OPA1 in autosomal dominant optic atrophy. *eLife*, *12*, RP87880. https://doi.org/10.7554/eLife.87880

33. Rozanes, E., Ben-Arzi, A., Boas, H., Ehrenberg, M., Bialer, O., & Stiebel-Kalish, H. (2024). Family Planning in Genetic Optic Atrophies in Israel, a Case Series and a Discussion of Ethical Considerations. *Journal of Neuro-Ophthalmology: The Official Journal of the North American Neuro-Ophthalmology Society*. https://doi.org/10.1097/WNO.0000000000002232

34. Venkatesh, A., McKenty, T., Ali, S., Sonntag, D., Ravipaty, S., Cui, Y., Slate, D., Lin, Q., Christiansen, A., Jacobson, S., Kach, J., Lim, K. H., Srinivasan, V., Zinshteyn, B., Aznarez, I., Huryn, L. A., Li, Z., Hufnagel, R. B., Liau, G., … Hoger, J. (2024). Antisense Oligonucleotide STK-002 Increases OPA1 in Retina and Improves Mitochondrial Function in Autosomal Dominant Optic Atrophy Cells. *Nucleic Acid Therapeutics*, *34*(5), 221-233. https://doi.org/10.1089/nat.2024.0022

35. Wang, J., Wang, M., Moshiri, A., Harris, R. A., Raveendran, M., Nguyen, T., Kim, S., Young, L., Wang, K., Wiseman, R., O'Connor, D. H., Johnson, Z., Martinez, M., Montague, M. J., Sayers, K., Lyke, M., Vallender, E., Stout, T., Li, Y., … Chen, R. (2024). Genetic diversity of 1,845 rhesus macaques improves genetic variation interpretation and identifies disease models. *Nature Communications*, *15*(1), 5658. https://doi.org/10.1038/s41467-024-49922-6

36. Yang, T.-H., Kang, E. Y.-C., Lin, P.-H., Yu, B. B.-C., Wang, J. H.-H., Chen, V., & Wang, N.-K. (2024). Mitochondria in Retinal Ganglion Cells : Unraveling the Metabolic Nexus and Oxidative Stress. *International Journal of Molecular Sciences*, *25*(16), 8626. https://doi.org/10.3390/ijms25168626

Encéphalopathie myo-neuro-gastrointestinale - *Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE)*

37. Pizzamiglio, C., Hanna, M. G., & Pitceathly, R. D. S. (2024). Primary mitochondrial diseases. *Handbook of Clinical Neurology*, *204*, 53-76. https://doi.org/10.1016/B978-0-323-99209-1.00004-1

Maladie de Charcot-Marie-Tooth d'origine mitochondriale – *Mitochondrial Charcot-Marie-Tooth*

38. Abati, E., Gagliardi, D., Manini, A., Del Bo, R., Ronchi, D., Meneri, M., Beretta, F., Sarno, A., Rizzo, F., Monfrini, E., Di Fonzo, A., Pellecchia, M. T., Brusati, A., Silani, V., Comi, G. P., Ratti, A., Verde, F., Ticozzi, N., & Corti, S. (2024). Investigating the prevalence of MFN2 mutations in amyotrophic lateral sclerosis : Insights from an Italian cohort. *Brain Communications*, *6*(5), fcae312. https://doi.org/10.1093/braincomms/fcae312

39. Armirola-Ricaurte, C., Morant, L., Adant, I., Hamed, S. A., Pipis, M., Efthymiou, S., Amor-Barris, S., Atkinson, D., Van de Vondel, L., Tomic, A., de Vriendt, E., Zuchner, S., Ghesquiere, B., Hanna, M., Houlden, H., Lunn, M. P., Reilly, M. M., Rasic, V. M., & Jordanova, A. (2024). Biallelic variants in COX18 cause a mitochondrial disorder primarily manifesting as peripheral neuropathy. *medRxiv: The Preprint Server for Health Sciences*, 2024.07.03.24309787. https://doi.org/10.1101/2024.07.03.24309787

40. Bird, T. D. (1993). Charcot-Marie-Tooth Hereditary Neuropathy Overview. In M. P. Adam, J. Feldman, G. M. Mirzaa, R. A. Pagon, S. E. Wallace, & A. Amemiya (Éds.), *GeneReviews®*. University of Washington, Seattle. http://www.ncbi.nlm.nih.gov/books/NBK1358/

41. Cozzi, M., Magri, S., Tedesco, B., Patelli, G., Ferrari, V., Casarotto, E., Chierichetti, M., Pramaggiore, P., Cornaggia, L., Piccolella, M., Galbiati, M., Rusmini, P., Crippa, V., Mandrioli, J., Pareyson, D., Pisciotta, C., D'Arrigo, S., Ratti, A., Nanetti, L., … Poletti, A. (2024). Altered molecular and cellular mechanisms in KIF5A-associated neurodegenerative or neurodevelopmental disorders. *Cell Death & Disease*, *15*(9), 692. https://doi.org/10.1038/s41419-024-07096-5

42. Karaś, K., Pastwińska, J., Sałkowska, A., Karwaciak, I., & Ratajewski, M. (2024). Epigenetic regulation of the human GDAP1 gene. *Biochemistry and Biophysics Reports*, *40*, 101827. https://doi.org/10.1016/j.bbrep.2024.101827

43. Molitor, A., Lederle, A., Radosavljevic, M., Sapuru, V., Zavorka Thomas, M. E., Yang, J., Shirin, M., Collin-Bund, V., Jerabkova-Roda, K., Miao, Z., Bernard, A., Rolli, V., Grenot, P., Castro, C. N., Rosenzwajg, M., Lewis, E. G., Person, R., Esperón-Moldes, U.-S., Kaare, M., … Bahram, S. (2024). A pleiotropic recurrent dominant ITPR3 variant causes a complex multisystemic disease. *Science Advances*, *10*(37), eado5545. https://doi.org/10.1126/sciadv.ado5545

44. Tran, Q. T. H., Kondo, N., Ueda, H., Matsuo, Y., & Tsukaguchi, H. (2024). Altered Endoplasmic Reticulum Integrity and Organelle Interactions in Living Cells Expressing INF2 Variants. *International Journal of Molecular Sciences*, *25*(18), 9783. https://doi.org/10.3390/ijms25189783

45. Weigele, J., Zhang, L., Franco, A., Cartier, E., & Dorn, G. W. (2024). Sensory-Motor Neuropathy in Mfn2 T105M Knock-in Mice and Its Reversal by a Novel Piperine-Derived Mitofusin Activator. *The Journal of Pharmacology and Experimental Therapeutics*, *391*(2), 361-374. https://doi.org/10.1124/jpet.124.002258

46. Yang, H., Zhang, V. W., Ai, L., & Wu, L. (2024). A Novel m.1636A > G Variant in Mitochondrial TV Gene Might Cause New Phenotype of Mitochondrial Disease in a 2-Year Old Chinese Boy. *Molecular Neurobiology*. https://doi.org/10.1007/s12035-024-04472-2

47. Zhang, Y., Ma, L., Wang, Z., Gao, C., Yang, L., Li, M., Tang, X., Yuan, H., Pang, D., & Ouyang, H. (2024). Mfn2R364W, Mfn2G176S, and Mfn2H165R mutations drive Charcot-Marie-Tooth type 2A disease by inducing apoptosis and mitochondrial oxidative phosphorylation damage. *International Journal of Biological Macromolecules*, *278*(Pt 1), 134673. https://doi.org/10.1016/j.ijbiomac.2024.134673

Maladies liées au gène ACO2 - *ACO2-related disorders*

No results

Maladie liée au gène GFM1 – GFM1-related disorder

No results

Maladies liées au gène POLG (inclus SANDO, SCAE) - *POLG-related disorders (included SANDO, SCAE)*

48. Bayat, M., Harbo, T., Anzabi, M., Bayat, A., & Thomsen, J. L. S. (2024). POLG-related mitochondrial disease mimicking autoimmune encephalitis. *Journal of Neurology*, *271*(10), 7021-7023. https://doi.org/10.1007/s00415-024-12641-5

49. Cheyne, I., Boryszewski, B., Chang, W., & Mikaszewska-Sokolewicz, M. (2024). A DNA Polymerase Subunit Gamma (POLG) Mutation Imposing a Difficult Differential Diagnosis of Hepatic Encephalopathy in a Newborn : A Case Report. *Cureus*, *16*(7), e65239. https://doi.org/10.7759/cureus.65239

50. Levee, V., Sivaganesh, K., Schaeffer, A., & Karunaratne, K. (2024). POLG epilepsy presenting as new-onset refractory status epilepticus (NORSE) in pregnancy. *Practical Neurology*, pn-2024-004232. https://doi.org/10.1136/pn-2024-004232

51. Mabry, C. J., Weindel, C. G., Stranahan, L. W., Martinez, E. L., VanPortfliet, J. J., West, A. P., Patrick, K. L., & Watson, R. O. (2024). Necrosis drives susceptibility to Mycobacterium tuberculosis in POLG mtDNA mutator mice. *bioRxiv: The Preprint Server for Biology*, 2024.07.17.603991. https://doi.org/10.1101/2024.07.17.603991

52. Pekeles, H., Berrahmoune, S., Dassi, C., Cheung, A. C. T., Gagnon, T., Waters, P. J., Eberhard, R., Buhas, D., & Myers, K. A. (2024). Safety and efficacy of deoxycytidine/deoxythymidine combination therapy in POLG-related disorders : 6-month interim results of an open-label, single arm, phase 2 trial. *EClinicalMedicine*, *74*, 102740. https://doi.org/10.1016/j.eclinm.2024.102740

53. Pizzamiglio, C., Hanna, M. G., & Pitceathly, R. D. S. (2024). Primary mitochondrial diseases. *Handbook of Clinical Neurology*, *204*, 53-76. https://doi.org/10.1016/B978-0-323-99209-1.00004-1

54. Rathore, A., Arunachal, G., Mahale, R. R., Padmanabha, H., & Mailankody, P. (2024). « The phenotypic conundrum of Trp748Ser variant in POLG gene : A report of two patients ». *Acta Neurologica Belgica*. https://doi.org/10.1007/s13760-024-02640-8

55. Rötig, A., Gaignard, P., Barcia, G., Assouline, Z., Berat, C.-M., Barth, M., Damaj, L., Laborde, N., Abi-Warde, M.-T., Chabrol, B., De Lonlay, P., Desguerre, I., Goldenberg, A., Gonzales, E., Jacquemin, E., Amati-Bonneau, P., Bonneau, D., Abadie, V., Bonnemains, C., … Schiff, M. (2024). Distinct Clinical Courses and Shortened Lifespans in Childhood-Onset DNA Polymerase Gamma Deficiency. *Neurology. Genetics*, *10*(4), e200167. https://doi.org/10.1212/NXG.0000000000200167

56. Singh, F., Wilhelm, L., Prescott, A. R., Ostacolo, K., Zhao, J.-F., Ogmundsdottir, M. H., & Ganley, I. G. (2024). PINK1 regulated mitophagy is evident in skeletal muscles. *Autophagy Reports*, *3*(1), 2326402. https://doi.org/10.1080/27694127.2024.2326402

57. Sturgis, J., Singh, R., Caron, Q. R., Samuels, I. S., Shiju, T. M., Mukkara, A., Freedman, P., & Bonilha, V. L. (2024). Modeling aging and retinal degeneration with mitochondrial DNA mutation burden. *Aging Cell*, e14282. https://doi.org/10.1111/acel.14282

58. Zhao, Y., Zhao, X., Ji, K., Wang, J., Zhao, Y., Lin, J., Gang, Q., Yu, M., Yuan, Y., Jiang, H., Sun, C., Fang, F., Yan, C., & Wang, Z. (2024). The clinical and genetic spectrum of mitochondrial diseases in China : A multicenter retrospective cross-sectional study. *Clinical Genetics*, *106*(6), 733-744. https://doi.org/10.1111/cge.14605

MELAS

59. Burattini, M., Pisano, A., Medeghini, V., Rossi, S., Luciani, G. B., D'Amati, G., & Miragoli, M. (2024). MELAS syndrome : Insights into cardiac mechanics. *Vascular Pharmacology*, *155*, 107303. https://doi.org/10.1016/j.vph.2024.107303

60. Dhawan, S., Musa, A. H., & Mantripragada, K. (2024). Novel Mitochondrial Cytopathy Causing Mitochondrial Encephalomyopathy With Lactic Acidosis and Stroke-Like Episodes Syndrome and Tubulointerstitial Nephropathy. *Cureus*, *16*(8), e66722. https://doi.org/10.7759/cureus.66722

61. Finsterer, J. (2024). Cortical atrophy is a common phenotypic feature in MELAS patients. *Asian Journal of Surgery*, S1015-9584(24)01748-2. https://doi.org/10.1016/j.asjsur.2024.08.010

62. Finsterer, J., & Mehri, S. (2024a). Cluster Analysis to Specify the MELAS Genotype and Phenotype Must Consider Genetic Background Peculiarities and Disease Progression. *AJNR. American Journal of Neuroradiology*, *45*(8), E31. https://doi.org/10.3174/ajnr.A8323

63. Finsterer, J., & Mehri, S. (2024b). MIDD Patients Should Not Be Confused with MELAS Patients, Even Though Both Carry the m.3243A>G Variant. *Diabetes & Metabolism Journal*, *48*(4), 816-817. https://doi.org/10.4093/dmj.2024.0065

64. Gunawardena, K., Praveenan, S., Dissanayake, V. H. W., & Ratnayake, P. (2024). Mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes with coexisting nemaline myopathy : A case report. *Journal of Medical Case Reports*, *18*(1), 420. https://doi.org/10.1186/s13256-024-04723-9

65. Jia, Y., Jiao, L., & Wang, Y. (2024). X-linked agammaglobulinaemic progressive encephalopathy mimicking mitochondrial encephalomyopathy-latic axidosis-stroke like episode. *Neurological Sciences: Official Journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology*, *45*(10), 5109-5110. https://doi.org/10.1007/s10072-024-07698-z

66. Khan, Z., & Khan, A. (2024). An Interesting Case of Wolf-Parkinson-White Syndrome in a Young Patient With Sensorineural Deafness. *Cureus*, *16*(6), e62928. https://doi.org/10.7759/cureus.62928

67. Mahesan, A., Kamila, G., Sundaram, M., Kumar, A., Jauhari, P., Chakrabarty, B., & Gulati, S. (2024). MELAS Syndrome : Rare Early Presentation of a Known Stroke Mimic. *Neurology India*, *72*(3), 645-646. https://doi.org/10.4103/neurol-india.Neurol-India-D-24-00065

68. Majamaa, K., Kärppä, M., & Moilanen, J. S. (2024). Neurological manifestations in adult patients with the m.3243A>G variant in mitochondrial DNA. *BMJ Neurology Open*, *6*(2), e000825. https://doi.org/10.1136/bmjno-2024-000825

69. Matsuura, A., Tozawa, T., Moroto, M., Miyamoto, Y., Kawabe, Y., Zuiki, M., Hasegawa, T., Kayaki, T., Yano, N., Yoshida, T., Chiyonobu, T., Morimoto, M., & Iehara, T. (2024). Alternating cerebral edema and arterial dilations in Molybdenum cofactor deficiency type-A. *Journal of Inherited Metabolic Disease*. https://doi.org/10.1002/jimd.12775

70. Papatya Çakır, E. D., Ersıoy, M., Çakır Biçer, N., & Gedikbaşı, A. (2024). Endocrine Disorders in Children with Primary Mitochondrial Diseases : Single-Center Experience. *Journal of Clinical Research in Pediatric Endocrinology*. https://doi.org/10.4274/jcrpe.galenos.2024.2024-1-11

71. Pizzamiglio, C., Hanna, M. G., & Pitceathly, R. D. S. (2024). Primary mitochondrial diseases. *Handbook of Clinical Neurology*, *204*, 53-76. https://doi.org/10.1016/B978-0-323-99209-1.00004-1

72. Ravi, V., Osouli Meinagh, S., & Bavarsad Shahripour, R. (2024). Reviewing migraine-associated pathophysiology and its impact on elevated stroke risk. *Frontiers in Neurology*, *15*, 1435208. https://doi.org/10.3389/fneur.2024.1435208

73. Xu, S., Qu, L. J., Chen, X., Zhu, X. L., & Niu, F. N. (2024). [Clinical, imaging, and pathological characteristics of 35 cases of mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes syndrome]. *Zhonghua Nei Ke Za Zhi*, *63*(7), 674-679. https://doi.org/10.3760/cma.j.cn112138-20231227-00411

74. Yen, H.-C., Hsu, C.-T., Wu, S.-Y., Kan, C.-C., Chang, C.-W., Chang, H.-M., Chien, Y.-A., Wei, Y.-H., & Wu, C.-Y. (2024). Alterations in coenzyme Q10 status in a cybrid line harboring the 3243A>G mutation of mitochondrial DNA is associated with abnormal mitochondrial bioenergetics and dysregulated mitochondrial biogenesis. *Biochimica Et Biophysica Acta. Bioenergetics*, *1865*(4), 149492. https://doi.org/10.1016/j.bbabio.2024.149492

75. Zhao, X., Yu, M., Zhang, W., Hou, Y., Yuan, Y., & Wang, Z. (2024). Demographic characteristics, diagnostic challenges, treatment patterns, and caregiver burden of mitochondrial diseases : A retrospective cross-sectional study. *Orphanet Journal of Rare Diseases*, *19*(1), 287. https://doi.org/10.1186/s13023-024-03289-5

76. Zhao, Y., Zhao, X., Ji, K., Wang, J., Zhao, Y., Lin, J., Gang, Q., Yu, M., Yuan, Y., Jiang, H., Sun, C., Fang, F., Yan, C., & Wang, Z. (2024). The clinical and genetic spectrum of mitochondrial diseases in China : A multicenter retrospective cross-sectional study. *Clinical Genetics*, *106*(6), 733-744. https://doi.org/10.1111/cge.14605

77. Zheng, Y., Wu, H., Zhang, M., Huang, B., Yang, J., Liu, C., Wang, H., & Du, K. (2024). Fahr's syndrome as the initial imaging characteristics of MELAS syndrome with a possible seizure activity and cardiac arrest : A case report. *Frontiers in Genetics*, *15*, 1393158. https://doi.org/10.3389/fgene.2024.1393158

MERFF

78. Giaj Levra, A., & Amata, F. (2024). Myoclonic Epilepsy With Ragged Red Fiber Cardiomyopathy : A Case Report and Brief Review of Literature. *Cureus*, *16*(8), e66745. https://doi.org/10.7759/cureus.66745

79. Nordli, D. R., Natarus, D., Yano, S., & Nordli, D. R. (2024). Eye closure sensitivity as a novel EEG marker of MERRF. *Epilepsia Open*, *9*(5), 2000-2002. https://doi.org/10.1002/epi4.13006

80. Pizzamiglio, C., Hanna, M. G., & Pitceathly, R. D. S. (2024). Primary mitochondrial diseases. *Handbook of Clinical Neurology*, *204*, 53-76. https://doi.org/10.1016/B978-0-323-99209-1.00004-1

Neuropathie optique héréditaire de Leber - *Leber hereditary optic neuropathy (LHON)*

81. Alorainy, J., Alorfi, Y., Karanjia, R., & Badeeb, N. (2024). A Comprehensive Review of Leber Hereditary Optic Neuropathy and Its Association with Multiple Sclerosis-Like Phenotypes Known as Harding's Disease. *Eye and Brain*, *16*, 17-24. https://doi.org/10.2147/EB.S470184

82. Biotti, D., Varenne, F., Girardie, P., Ciron, J., Freeman, S., & Bonneville, F. (2024). Extensive Optic Tracts Involvement in the Acute Phase of Leber Hereditary Optic Neuropathy. *Neurology*, *103*(3), e209680. https://doi.org/10.1212/WNL.0000000000209680

83. Bušányová, B., Vajter, M., Kelifová, S., Lišková, P., Miková, H., Breciková, K., Žigmond, J., Rogalewicz, V., Tichopád, A., Višňanský, M., & Šarkanová, I. (2024). Leber hereditary optic neuropathy in Czechia and Slovakia : Quality of life and costs from patient perspective. *Heliyon*, *10*(11), e32296. https://doi.org/10.1016/j.heliyon.2024.e32296

84. Chen, Y., Wei, X., Ci, X., Ji, Y., & Zhang, J. (2024). Dysregulation of mitochondria, apoptosis and mitophagy in Leber's hereditary optic neuropathy with MT-ND1 3635G>A mutation. *Gene*, *930*, 148853. https://doi.org/10.1016/j.gene.2024.148853

85. Costello, S. M., Schultz, A., Smith, D., Horan, D., Chaverra, M., Tripet, B., George, L., Bothner, B., Lefcort, F., & Copié, V. (2024). Metabolic Deficits in the Retina of a Familial Dysautonomia Mouse Model. *Metabolites*, *14*(8), 423. https://doi.org/10.3390/metabo14080423

86. Esmaeil, A., Ali, A., & Behbehani, R. (2022). Leber's hereditary optic neuropathy : Update on current diagnosis and treatment. *Frontiers in Ophthalmology*, *2*, 1077395. https://doi.org/10.3389/fopht.2022.1077395

87. Ezan, P., Hardy, E., Bemelmans, A., Taiel, M., Dossi, E., & Rouach, N. (2024). Retinal damage promotes mitochondrial transfer in the visual system of a mouse model of Leber hereditary optic neuropathy. *Neurobiology of Disease*, *201*, 106681. https://doi.org/10.1016/j.nbd.2024.106681

88. Hawlina, M., Kovač, L., Breciková, K., Žigmond, J., Rogalewicz, V., Tichopád, A., Višňanský, M., & Šarkanová, I. (2024). Leber hereditary optic neuropathy in Slovenia : Quality of life and costs from patient perspective. *Orphanet Journal of Rare Diseases*, *19*(1), 318. https://doi.org/10.1186/s13023-024-03329-0

89. Hernansanz-Agustín, P., Morales-Vidal, C., Calvo, E., Natale, P., Martí-Mateos, Y., Jaroszewicz, S. N., Cabrera-Alarcón, J. L., Acín-Pérez, R., López-Montero, I., Vázquez, J., & Enríquez, J. A. (2024). A transmitochondrial sodium gradient controls membrane potential in mammalian mitochondria. *Cell*, S0092-8674(24)00974-7. https://doi.org/10.1016/j.cell.2024.08.045

90. Hotta, Y., Torii, K., & Takayama, M. (2024). Ocular genetics in the Japanese population. *Japanese Journal of Ophthalmology*, *68*(5), 401-418. https://doi.org/10.1007/s10384-024-01109-8

91. Jin, L., Gan, D., He, W., Wu, N., Xiang, S., Wei, Y., Eriani, G., Ji, Y., Guan, M.-X., & Wang, M. (2024). Mitochondrial tRNAGlu 14693A > G Mutation, an « Enhancer » to the Phenotypic Expression of Leber's Hereditary Optic Neuropathy. *Advanced Science (Weinheim, Baden-Wurttemberg, Germany)*, *11*(41), e2401856. https://doi.org/10.1002/advs.202401856

92. Jørstad, Ø. K., Skaar, S., Strand, H., Røsby, O., Brokstad, R. T., & Rønning, P. A. (2024). Leber Hereditary Optic Neuropathy Case Report : Clinical Presentation and Treatment with Idebenone Reinforce the Evidence for m.3866T>C as a Causative Variant. *Case Reports in Ophthalmology*, *15*(1), 513-517. https://doi.org/10.1159/000539445

93. Kumar, S., Kaur, R., Malik, M. A., Angmo, D., & Kaur, J. (2023). Extranuclear DNA Variations in the Susceptibility of Glaucoma. *Middle East African Journal of Ophthalmology*, *30*(2), 113-120. https://doi.org/10.4103/meajo.meajo_132_23

94. Masuda, Y., Ishikawa, H., Ishikawa, H., Kezuka, T., Miyazaki, A., Matsumoto, K., Gomi, F., Mimura, O., Shikishima, K., Nakano, T., & Terao, M. (2024). Assessment of objective visual function following idebenone administration in patients with leber hereditary optic neuropathy. *Japanese Journal of Ophthalmology*, *68*(5), 548-555. https://doi.org/10.1007/s10384-024-01077-z

95. Papatya Çakır, E. D., Ersıoy, M., Çakır Biçer, N., & Gedikbaşı, A. (2024). Endocrine Disorders in Children with Primary Mitochondrial Diseases : Single-Center Experience. *Journal of Clinical Research in Pediatric Endocrinology*. https://doi.org/10.4274/jcrpe.galenos.2024.2024-1-11

96. Peng, S.-Y., Chen, C.-Y., Chen, H., Yang, Y.-P., Wang, M.-L., Tsai, F.-T., Chien, C.-S., Weng, P.-Y., Tsai, E.-T., Wang, I.-C., Hsu, C.-C., Lin, T.-C., Hwang, D.-K., Chen, S.-J., Chiou, S.-H., Chiao, C.-C., & Chien, Y. (2024). Inhibition of angiogenesis by the secretome from iPSC-derived retinal ganglion cells with Leber's hereditary optic neuropathy-like phenotypes. *Biomedicine & Pharmacotherapy = Biomedecine & Pharmacotherapie*, *178*, 117270. https://doi.org/10.1016/j.biopha.2024.117270

97. Pietraszkiewicz, A., Ayesha, A., Digre, K. B., Warner, J. E., Seay, M. D., Crum, A. V., & Katz, B. J. (2024). Diagnostic dilemma : Leber's hereditary optic neuropathy in a 70-year-Old woman. *American Journal of Ophthalmology Case Reports*, *36*, 102143. https://doi.org/10.1016/j.ajoc.2024.102143

98. Pizzamiglio, C., Hanna, M. G., & Pitceathly, R. D. S. (2024). Primary mitochondrial diseases. *Handbook of Clinical Neurology*, *204*, 53-76. https://doi.org/10.1016/B978-0-323-99209-1.00004-1

99. Rozanes, E., Ben-Arzi, A., Boas, H., Ehrenberg, M., Bialer, O., & Stiebel-Kalish, H. (2024). Family Planning in Genetic Optic Atrophies in Israel, a Case Series and a Discussion of Ethical Considerations. *Journal of Neuro-Ophthalmology: The Official Journal of the North American Neuro-Ophthalmology Society*. https://doi.org/10.1097/WNO.0000000000002232

100. Vrisakis, J.-L., Fraser, C. L., Shahnam, A., Nindra, U., & Grimison, P. (2024). A case for the use of chemotherapy in hereditary mitochondrial optic neuropathies : Successful administration of cisplatin/etoposide in a male patient with testicular seminoma and Leber's hereditary optic neuropathy. *Clinical Case Reports*, *12*(7), e9045. https://doi.org/10.1002/ccr3.9045

101. Wang, D., Yuan, J., Liu, H.-L., Li, S.-L., Ma, N., Chen, M.-L., Yuan, H., Jie, H., Li, B., & Zhang, T. (2024). Clinical follow-up investigation on thickness changes in the peripapillary retinal nerve fibre layer of patients with Leber hereditary optic neuropathy. *BMC Ophthalmology*, *24*(1), 348. https://doi.org/10.1186/s12886-024-03625-0

102. Wang, R., Bao, F., Lu, M., Jia, X., Xiao, J., Wu, Y., Zhang, Q., & Liu, X. (2024). MSC-mediated mitochondrial transfer restores mitochondrial DNA and function in neural progenitor cells of Leber's hereditary optic neuropathy. *Science China. Life Sciences*, *67*(11), 2511-2519. https://doi.org/10.1007/s11427-024-2647-8

103. Xu, J., Li, Y., Yao, S., Jin, X., Yang, M., Guo, Q., Qiu, R., & Lei, B. (2024). Preservation of Mitochondrial Function by SkQ1 in Skin Fibroblasts Derived from Patients with Leber's Hereditary Optic Neuropathy Is Associated with the PINK1/PRKN-Mediated Mitophagy. *Biomedicines*, *12*(9), 2020. https://doi.org/10.3390/biomedicines12092020

104. Zhang, J., Li, W., Liu, Z., Chen, Y., Wei, X., Peng, L., Xu, M., & Ji, Y. (2024). Defective post-transcriptional modification of tRNA disrupts mitochondrial homeostasis in Leber's hereditary optic neuropathy. *The Journal of Biological Chemistry*, *300*(9), 107728. https://doi.org/10.1016/j.jbc.2024.107728

Ophtalmoplégie externe progressive – *Progressive external ophthalmoplegia (PEO)*

105. Chen, Y., Yan, L., Xue, J., Li, H., Wu, L., Zheng, J., & Di, Y. (2024). [Clinical features and genetic analysis of child with Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6 due to variant of DNA2 gene]. *Zhonghua Yi Xue Yi Chuan Xue Za Zhi = Zhonghua Yixue Yichuanxue Zazhi = Chinese Journal of Medical Genetics*, *41*(10), 1238-1242. https://doi.org/10.3760/cma.j.cn511374-20231020-00211

106. Leister, N., Wendt, S., Hedergott, A., Heindl, L. M., Rokohl, A. C., Stoll, S. E., Gordon, E., Böttiger, B. W., Fricke, J., & Schick, V. C. (2024). Anaesthesia Concepts in Patients with Chronic Progressive External Ophthalmoplegia Undergoing Ophthalmic Surgery-A Retrospective Cohort Analysis. *Journal of Clinical Medicine*, *13*(16), 4710. https://doi.org/10.3390/jcm13164710

107. Zhao, Y., Zhao, X., Ji, K., Wang, J., Zhao, Y., Lin, J., Gang, Q., Yu, M., Yuan, Y., Jiang, H., Sun, C., Fang, F., Yan, C., & Wang, Z. (2024). The clinical and genetic spectrum of mitochondrial diseases in China : A multicenter retrospective cross-sectional study. *Clinical Genetics*, *106*(6), 733-744. https://doi.org/10.1111/cge.14605

Syndrome d'Alpers-Huttenlocher - *Alpers-Huttenlocher syndrome*

108. Andrade-Machado, R., Abushanab, E., Patel, N. D., & Singh, A. (2024). Differentiating rhythmic high-amplitude delta with superimposed (poly) spikes from extreme delta brushes : Limitations of standardized nomenclature and implications for patient management. *World Journal of Pediatrics: WJP*, *20*(8), 764-773. https://doi.org/10.1007/s12519-024-00816-z

109. Pekeles, H., Berrahmoune, S., Dassi, C., Cheung, A. C. T., Gagnon, T., Waters, P. J., Eberhard, R., Buhas, D., & Myers, K. A. (2024). Safety and efficacy of deoxycytidine/deoxythymidine combination therapy in POLG-related disorders : 6-month interim results of an open-label, single arm, phase 2 trial. *EClinicalMedicine*, *74*, 102740. https://doi.org/10.1016/j.eclinm.2024.102740

Syndrome de Kearns-Sayre – *Kearns-Sayre syndrome*

110. Benoit, M., Forêt, F., Ntwali, F., & Higny, J. (2024). Kearns-Sayre Syndrome : A rare mitochondrial cytopathy complicated with complete heart block in a teenager. *Acta Cardiologica*, 1-3. https://doi.org/10.1080/00015385.2024.2359291

111. Finsterer, J. (2024). Syncope in Kearns-Sayre syndrome may not only be due to AV-block, but may also have other causes due to the multiorgan nature of the disease. *Acta Cardiologica*, 1-2. https://doi.org/10.1080/00015385.2024.2404774

112. Li, Y., Zhang, H., Yu, C., Dong, X., Yang, F., Wang, M., Wen, Z., Su, M., Li, B., & Yang, L. (2024). New Insights into Mitochondria in Health and Diseases. *International Journal of Molecular Sciences*, *25*(18), 9975. https://doi.org/10.3390/ijms25189975

113. Pizzamiglio, C., Hanna, M. G., & Pitceathly, R. D. S. (2024). Primary mitochondrial diseases. *Handbook of Clinical Neurology*, *204*, 53-76. https://doi.org/10.1016/B978-0-323-99209-1.00004-1

114. Traunero, A., Baldo, F., Magnolato, A., Di Leo, G., Barbi, E., & Bruno, I. (2024). Administration of bicarbonates through percutaneous gastrostomy with continuous nocturnal infusion in a patient with Kearns-Sayre disease : A life changing therapeutical paradigm. *Italian Journal of Pediatrics*, *50*(1), 132. https://doi.org/10.1186/s13052-024-01696-9

Syndrome de Leigh - *Leigh syndrome*

115. Akar, H. T., Sayar, E., Sarıtaş Nakip, Ö., Sönmez, E., Özkan, M. B., & Olgaç, A. (2024). Leigh Syndrome due to MT-ATP6 Variants : A Case Presentation and the Review of the Literature. *Molecular Syndromology*, *15*(4), 333-338. https://doi.org/10.1159/000536676

116. Alagarsamy, K. N., Srivastava, A., & Dhingra, S. (2024). A Method for Producing Induced Pluripotent Stem Cell-Derived Cardiomyocytes from Leigh Syndrome Patients for Its Application in Disease Modeling and Drug Validation. *Methods in Molecular Biology (Clifton, N.J.)*, *2835*, 121-133. https://doi.org/10.1007/978-1-0716-3995-5_11

117. Baker, E. K. (2024). Blonde hair, blue eyes. *American Journal of Medical Genetics. Part C, Seminars in Medical Genetics*, e32106. https://doi.org/10.1002/ajmg.c.32106

118. Brischigliaro, M., Krüger, A., Moran, J. C., Antonicka, H., Ahn, A., Shoubridge, E. A., Rorbach, J., & Barrientos, A. (2024). The human mitochondrial translation factor TACO1 alleviates mitoribosome stalling at polyproline stretches. *Nucleic Acids Research*, *52*(16), 9710-9726. https://doi.org/10.1093/nar/gkae645

119. Buchanan, G. F. (2024). Leigh has no Brakes : Impaired Inhibition in a Mouse Model of Leigh Syndrome Leads to Enhanced Seizure Sensitivity. *Epilepsy Currents*, *24*(4), 298-300. https://doi.org/10.1177/15357597241257031

120. Di Nottia, M., Rizza, T., Baruffini, E., Nesti, C., Torraco, A., Diodato, D., Martinelli, D., Dal Canto, F., Gilea, A. I., Zoccola, M., Siri, B., Dionisi-Vici, C., Bertini, E., Santorelli, F. M., Goffrini, P., & Carrozzo, R. (2024). Severe mitochondrial encephalomyopathy caused by de novo variants in OPA1 gene. *Frontiers in Genetics*, *15*, 1437959. https://doi.org/10.3389/fgene.2024.1437959

121. Dong, Q., Yin, X., Fan, S., Zhong, S., Yang, W., Chen, K., Wang, Q., Ma, X., Mahlatsi, R. L., Yang, Y., Lyu, J., Fang, H., & Wang, Y. (2024). IARS2 mutations lead to Leigh syndrome with a combined oxidative phosphorylation deficiency. *Orphanet Journal of Rare Diseases*, *19*(1), 305. https://doi.org/10.1186/s13023-024-03310-x

122. Dupré, M., Warne, R., Shipman, P., Kava, M., Ghia, T., Loughman, L., & Lakshmanan, R. (2024). Cranial and spinal nerve enhancement in SURF1-associated Leigh syndrome. *Pediatric Radiology*, *54*(10), 1758-1762. https://doi.org/10.1007/s00247-024-06005-4

123. Dzwilewski, K., Chojnowski, K., Krygier, M., Zawadzka, M., Chylińska, M., & Mazurkiewicz-Bełdzińska, M. (2024). Effects of idebenone treatment in a patient with DNAJC30-associated Leigh Syndrome. *Neurologia I Neurochirurgia Polska*, *58*(4), 468-470. https://doi.org/10.5603/pjnns.100423

124. Gana, S., Rossetto, G., Garau, J., Vacchini, V., Ferraro, F., Rognone, E., Pichiecchio, A., Gasperini, S., Valente, E. M., & Orcesi, S. (2024). « Relapsing-Remitting » Ataxia and Unexpected Brain Imaging in a Child with HIBCH Deficiency. *Movement Disorders Clinical Practice*. https://doi.org/10.1002/mdc3.14190

125. Göppert-Asadollahpour, S., Wohlwend, D., & Friedrich, T. (2024). Structural robustness of the NADH binding site in NADH:ubiquinone oxidoreductase (complex I). *Biochimica Et Biophysica Acta. Bioenergetics*, *1865*(4), 149491. https://doi.org/10.1016/j.bbabio.2024.149491

126. Haschke, A. M., Diecke, S., & Schuelke, M. (2024). Characterization of two iPSC lines from patients with maternally inherited leigh (MILS) and neuropathy, ataxia, and retinitis pigmentosa (NARP) syndrome carrying the MT-ATP6 m.8993 T>G mutation at different degrees of heteroplasmy. *Stem Cell Research*, *81*, 103547. https://doi.org/10.1016/j.scr.2024.103547

127. Huff, A., Oliveira, L. M., Karlen-Amarante, M., Ebiala, F., Ramirez, J. M., & Kalume, F. (2024). Ndufs4 inactivation in glutamatergic neurons reveals swallow-breathing discoordination in a mouse model of Leigh Syndrome. *bioRxiv: The Preprint Server for Biology*, 2024.09.11.612506. https://doi.org/10.1101/2024.09.11.612506

128. Lavorato, M., Iadarola, D., Remes, C., Kaur, P., Broxton, C., Mathew, N. D., Xiao, R., Seiler, C., Nakamaru-Ogiso, E., Anderson, V. E., & Falk, M. J. (2024). Dldhcri3 zebrafish exhibit altered mitochondrial ultrastructure, morphology, and dysfunction partially rescued by probucol or thiamine. *JCI Insight*, *9*(18), e178973. https://doi.org/10.1172/jci.insight.178973

129. Li, Y., Zhang, H., Yu, C., Dong, X., Yang, F., Wang, M., Wen, Z., Su, M., Li, B., & Yang, L. (2024). New Insights into Mitochondria in Health and Diseases. *International Journal of Molecular Sciences*, *25*(18), 9975. https://doi.org/10.3390/ijms25189975

130. Li, Y.-X., Wang, D.-J., Zhou, M.-B., Sun, H.-X., Hong, S.-Q., Jiang, L., & Guo, Y. (2024). [Leigh syndrome caused by the mitochondrial m.8993T>G mutation with hypocitrullinemia : A report of four cases and literature review]. *Zhongguo Dang Dai Er Ke Za Zhi = Chinese Journal of Contemporary Pediatrics*, *26*(9), 940-945. https://doi.org/10.7499/j.issn.1008-8830.2404036

131. Lo, C.-H., Liu, Z., Chen, S., Lin, F., Berneshawi, A. R., Yu, C. Q., Koo, E. B., Kowal, T. J., Ning, K., Hu, Y., Wang, W.-J., Liao, Y. J., & Sun, Y. (2024). Primary cilia formation requires the Leigh syndrome-associated mitochondrial protein NDUFAF2. *The Journal of Clinical Investigation*, *134*(13), e175560. https://doi.org/10.1172/JCI175560

132. Luo, Y., Xu, Y., Ahmad, F., Feng, G., & Huang, Y. (2024). Characterization of Shy1, the Schizosaccharomyces pombe homolog of human SURF1. *Scientific Reports*, *14*(1), 21678. https://doi.org/10.1038/s41598-024-72681-9

133. Nakai, R., Varnum, S., Field, R. L., Shi, H., Giwa, R., Jia, W., Krysa, S. J., Cohen, E. F., Borcherding, N., Saneto, R. P., Tsai, R. C., Suganuma, M., Ohta, H., Yokota, T., & Brestoff, J. R. (2024). Mitochondria transfer-based therapies reduce the morbidity and mortality of Leigh syndrome. *Nature Metabolism*, *6*(10), 1886-1896. https://doi.org/10.1038/s42255-024-01125-5

134. Nakashima, K., Bo, R., Awano, H., Nishiyama, M., & Iijima, K. (2024). Response to : Is pancreatitis actually a phenotypic manifestation of Leigh syndrome? *Pediatrics International: Official Journal of the Japan Pediatric Society*, *66*(1), e15783. https://doi.org/10.1111/ped.15783

135. Narra, R. K., & Vemuri, R. (2024). Distinct Imaging Markers of Leigh's Disease Linked to SURF1 Mutation : A Pediatric Case Study. *The American Journal of Case Reports*, *25*, e944514. https://doi.org/10.12659/AJCR.944514

136. Newstead, S. M., & Finsterer, J. (2024). Hyperkinesias in Leigh-like Syndrome with Complex-I Deficiency Due to m.10191T>C in MT-ND3. *Annals of African Medicine*, *23*(3), 512-513. https://doi.org/10.4103/aam.aam_32_23

137. Papatya Çakır, E. D., Ersıoy, M., Çakır Biçer, N., & Gedikbaşı, A. (2024). Endocrine Disorders in Children with Primary Mitochondrial Diseases : Single-Center Experience. *Journal of Clinical Research in Pediatric Endocrinology*. https://doi.org/10.4274/jcrpe.galenos.2024.2024-1-11

138. Pizzamiglio, C., Hanna, M. G., & Pitceathly, R. D. S. (2024). Primary mitochondrial diseases. *Handbook of Clinical Neurology*, *204*, 53-76. https://doi.org/10.1016/B978-0-323-99209-1.00004-1

139. Puighermanal, E., Luna-Sánchez, M., Gella, A., van der Walt, G., Urpi, A., Royo, M., Tena-Morraja, P., Appiah, I., de Donato, M. H., Menardy, F., Bianchi, P., Esteve-Codina, A., Rodríguez-Pascau, L., Vergara, C., Gómez-Pallarès, M., Marsicano, G., Bellocchio, L., Martinell, M., Sanz, E., … Quintana, A. (2024). Cannabidiol ameliorates mitochondrial disease via PPARγ activation in preclinical models. *Nature Communications*, *15*(1), 7730. https://doi.org/10.1038/s41467-024-51884-8

140. Puvabanditsin, S., Lee, I., Cordero, N., Target, K., Park, S. Y., & Mehta, R. (2024). A Fatal Case of 3-Hydroxyisobutyryl-CoA Hydrolase Deficiency in a Term Infant with Severe High Anion Gap Acidosis and Review of the Literature. *Case Reports in Genetics*, *2024*, 8099373. https://doi.org/10.1155/2024/8099373

141. Voges, T.-S., Lim, E. B., MacKenzie, A., Mudler, K., DeSouza, R., Onyejekwe, N. E., & Johnston, S. D. (2024). Phenotypic assessment of Cox10 variants and their implications for Leigh Syndrome. *BMC Research Notes*, *17*(1), 228. https://doi.org/10.1186/s13104-024-06879-5

142. Watanabe, M., & Sasaki, N. (2024). Mechanisms and Future Research Perspectives on Mitochondrial Diseases Associated with Isoleucyl-tRNA Synthetase Gene Mutations. *Genes*, *15*(7), 894. https://doi.org/10.3390/genes15070894

143. Yeole, M., Majethia, P., Siddiqui, S., Girisha, K. M., Shukla, A., Radhakrishnan, P., & Bhat, V. (2024). Bi-Allelic Splicing Variant, c.153-2A > C in TOMM7 Is Associated With Leigh Syndrome. *American Journal of Medical Genetics. Part A*, e63892. https://doi.org/10.1002/ajmg.a.63892

144. Zhao, X., Yu, M., Zhang, W., Hou, Y., Yuan, Y., & Wang, Z. (2024). Demographic characteristics, diagnostic challenges, treatment patterns, and caregiver burden of mitochondrial diseases : A retrospective cross-sectional study. *Orphanet Journal of Rare Diseases*, *19*(1), 287. https://doi.org/10.1186/s13023-024-03289-5

145. Zhao, Y., Zhao, X., Ji, K., Wang, J., Zhao, Y., Lin, J., Gang, Q., Yu, M., Yuan, Y., Jiang, H., Sun, C., Fang, F., Yan, C., & Wang, Z. (2024). The clinical and genetic spectrum of mitochondrial diseases in China : A multicenter retrospective cross-sectional study. *Clinical Genetics*, *106*(6), 733-744. https://doi.org/10.1111/cge.14605

Maladies mitochondriales (autres) – *Mitochondrial disorders (other)*

146. 26th Annual Symposium of the United Mitochondrial Disease Foundation - Mitochondrial Medicine 2023—Abstracts. (2023). *Therapeutic Advances in Rare Disease*, *4*, 26330040231199665. https://doi.org/10.1177/26330040231199665

147. Aaltio, J., Euro, L., Tynninen, O., Vu, H. S., Ni, M., DeBerardinis, R. J., Suomalainen, A., & Isohanni, P. (2024). Niacin supplementation in a child with novel MTTN variant m.5670A>G causing early onset mitochondrial myopathy and NAD+ deficiency. *Neuromuscular Disorders: NMD*, *43*, 14-19. https://doi.org/10.1016/j.nmd.2024.07.005

148. Akar, H. T., Sayar, E., Sarıtaş Nakip, Ö., Sönmez, E., Özkan, M. B., & Olgaç, A. (2024). Leigh Syndrome due to MT-ATP6 Variants : A Case Presentation and the Review of the Literature. *Molecular Syndromology*, *15*(4), 333-338. https://doi.org/10.1159/000536676

149. Alagarsamy, K. N., Srivastava, A., & Dhingra, S. (2024). A Method for Producing Induced Pluripotent Stem Cell-Derived Cardiomyocytes from Leigh Syndrome Patients for Its Application in Disease Modeling and Drug Validation. *Methods in Molecular Biology (Clifton, N.J.)*, *2835*, 121-133. https://doi.org/10.1007/978-1-0716-3995-5_11

150. Al-Amrani, F., Al-Thihli, K., Al-Ajmi, E., Al-Futaisi, A., & Al-Murshedi, F. (2024). Transient response to high-dose niacin therapy in a patient with NAXE deficiency. *JIMD Reports*, *65*(4), 212-225. https://doi.org/10.1002/jmd2.12425

151. Alessia, A., Anastasia, G., Alessia, D. D., Simona, B., Alessandro, P., Emanuela, B., Valentina, B., Valeria, T., Nicola, P., & Dario, B. (2024). Fetal and obstetrics manifestations of mitochondrial diseases. *Journal of Translational Medicine*, *22*(1), 853. https://doi.org/10.1186/s12967-024-05633-6

152. Alorainy, J., Alorfi, Y., Karanjia, R., & Badeeb, N. (2024). A Comprehensive Review of Leber Hereditary Optic Neuropathy and Its Association with Multiple Sclerosis-Like Phenotypes Known as Harding's Disease. *Eye and Brain*, *16*, 17-24. https://doi.org/10.2147/EB.S470184

153. Al-Suhaimi, E., AlQuwaie, R., AlSaqabi, R., Winarni, D., Dewi, F. R. P., AlRubaish, A. A., Shehzad, A., & Elaissari, A. (2024). Hormonal orchestra : Mastering mitochondria's role in health and disease. *Endocrine*. https://doi.org/10.1007/s12020-024-03967-1

154. Armirola-Ricaurte, C., Morant, L., Adant, I., Hamed, S. A., Pipis, M., Efthymiou, S., Amor-Barris, S., Atkinson, D., Van de Vondel, L., Tomic, A., de Vriendt, E., Zuchner, S., Ghesquiere, B., Hanna, M., Houlden, H., Lunn, M. P., Reilly, M. M., Rasic, V. M., & Jordanova, A. (2024). Biallelic variants in COX18 cause a mitochondrial disorder primarily manifesting as peripheral neuropathy. *medRxiv: The Preprint Server for Health Sciences*, 2024.07.03.24309787. https://doi.org/10.1101/2024.07.03.24309787

155. Barbato, A., Gori, G., Sacchini, M., Pochiero, F., Bargiacchi, S., Traficante, G., Palazzo, V., Tiberi, L., Bianchini, C., Mei, D., Parrini, E., Pisano, T., Procopio, E., Guerrini, R., Peron, A., & Stagi, S. (2024). Endocrinological features and epileptic encephalopathy in COX deficiency due to SCO1 mutations : Case series and review of literature. *Endocrine Connections*, *13*(10), e240221. https://doi.org/10.1530/EC-24-0221

156. Bayat, M., Harbo, T., Anzabi, M., Bayat, A., & Thomsen, J. L. S. (2024). POLG-related mitochondrial disease mimicking autoimmune encephalitis. *Journal of Neurology*, *271*(10), 7021-7023. https://doi.org/10.1007/s00415-024-12641-5

157. Berti, B., Verrigni, D., Nasca, A., Di Nottia, M., Leone, D., Torraco, A., Rizza, T., Bellacchio, E., Legati, A., Palermo, C., Marchet, S., Lamperti, C., Novelli, A., Mercuri, E. M., Bertini, E. S., Pane, M., Ghezzi, D., & Carrozzo, R. (2024). De Novo DNM1L Mutation in a Patient with Encephalopathy, Cardiomyopathy and Fatal Non-Epileptic Paroxysmal Refractory Vomiting. *International Journal of Molecular Sciences*, *25*(14), 7782. https://doi.org/10.3390/ijms25147782

158. Bharadwaj, A. (2024). A Review over Mitochondrial Diseases Due to mtDNA Mutations : Recent Advances and Remedial Aspects. *Infectious Disorders Drug Targets*. https://doi.org/10.2174/0118715265304029240801092834

159. Bowen, A. B., Rapalino, O., Jaimes, C., Ratai, E.-M., Zhong, Y., Thiele, E. A., Kritzer, A., Ganetzky, R. D., Gold, N. B., & Walker, M. A. (2024). Prenatal molecular diagnosis of pyruvate dehydrogenase complex deficiency enables rapid initiation of ketogenic diet. *American Journal of Medical Genetics. Part A*, *194*(12), e63825. https://doi.org/10.1002/ajmg.a.63825

160. Brady, S., Poulton, J., & Muller, S. (2024). Inclusion body myositis : Correcting impaired mitochondrial and lysosomal autophagy as a potential therapeutic strategy. *Autoimmunity Reviews*, *23*(11), 103644. https://doi.org/10.1016/j.autrev.2024.103644

161. Bušányová, B., Vajter, M., Kelifová, S., Lišková, P., Miková, H., Breciková, K., Žigmond, J., Rogalewicz, V., Tichopád, A., Višňanský, M., & Šarkanová, I. (2024). Leber hereditary optic neuropathy in Czechia and Slovakia : Quality of life and costs from patient perspective. *Heliyon*, *10*(11), e32296. https://doi.org/10.1016/j.heliyon.2024.e32296

162. Caldwell, J. L., Clarke, J. D., Smith, C. E. R., Pinali, C., Quinn, C. J., Pearman, C. M., Adomaviciene, A., Radcliffe, E. J., Watkins, A., Horn, M. A., Bode, E. F., Madders, G. W. P., Eisner, M., Eisner, D. A., Trafford, A. W., & Dibb, K. M. (2024). Restoring Atrial T-Tubules Augments Systolic Ca Upon Recovery From Heart Failure. *Circulation Research*, *135*(7), 739-754. https://doi.org/10.1161/CIRCRESAHA.124.324601

163. Cha, J. H., Lee, S.-H., Yun, Y., Choi, W. H., Koo, H., Jung, S. H., Chae, H. B., Lee, D. H., Lee, S. J., Jo, D. H., Kim, J. H., Song, J.-J., Chae, J.-H., Lee, J. H., Park, J., Kang, J. Y., Bae, S., & Lee, S.-Y. (2024). Discovery of novel disease-causing mutation in SSBP1 and its correction using adenine base editor to improve mitochondrial function. *Molecular Therapy. Nucleic Acids*, *35*(3), 102257. https://doi.org/10.1016/j.omtn.2024.102257

164. Chandrasekhar, V., Yelkur, P., & K, V. (2024). Infantile Fructose-1,6-Bisphosphatase Deficiency Masquerading as Mitochondriopathy. *Cureus*, *16*(8), e67291. https://doi.org/10.7759/cureus.67291

165. Chen, S., He, T., Chen, J., Wen, D., Wang, C., Huang, W., Yang, Z., Yang, M., Li, M., Huang, S., Huang, Z., & Zhu, H. (2024). Betaine delays age-related muscle loss by mitigating Mss51-induced impairment in mitochondrial respiration via Yin Yang1. *Journal of Cachexia, Sarcopenia and Muscle*, *15*(5), 2104-2117. https://doi.org/10.1002/jcsm.13558

166. Chen, S., Li, Q., Shi, H., Li, F., Duan, Y., & Guo, Q. (2024). New insights into the role of mitochondrial dynamics in oxidative stress-induced diseases. *Biomedicine & Pharmacotherapy = Biomedecine & Pharmacotherapie*, *178*, 117084. https://doi.org/10.1016/j.biopha.2024.117084

167. Cheyne, I., Boryszewski, B., Chang, W., & Mikaszewska-Sokolewicz, M. (2024). A DNA Polymerase Subunit Gamma (POLG) Mutation Imposing a Difficult Differential Diagnosis of Hepatic Encephalopathy in a Newborn : A Case Report. *Cureus*, *16*(7), e65239. https://doi.org/10.7759/cureus.65239

168. Coronel, R., García-Moreno, E., Siendones, E., Barrero, M. J., Martínez-Delgado, B., Santos-Ocaña, C., Liste, I., & Cascajo-Almenara, M. V. (2024). Brain organoid as a model to study the role of mitochondria in neurodevelopmental disorders : Achievements and weaknesses. *Frontiers in Cellular Neuroscience*, *18*, 1403734. https://doi.org/10.3389/fncel.2024.1403734

169. Correia, S. P., Moedas, M. F., Taylor, L. S., Naess, K., Lim, A. Z., McFarland, R., Kazior, Z., Rumyantseva, A., Wibom, R., Engvall, M., Bruhn, H., Lesko, N., Végvári, Á., Käll, L., Trost, M., Alston, C. L., Freyer, C., Taylor, R. W., Wedell, A., & Wredenberg, A. (2024). Quantitative proteomics of patient fibroblasts reveal biomarkers and diagnostic signatures of mitochondrial disease. *JCI Insight*, *9*(20), e178645. https://doi.org/10.1172/jci.insight.178645

170. Čunátová, K., & Fernández-Vizarra, E. (2024). Pathological variants in nuclear genes causing mitochondrial complex III deficiency : An update. *Journal of Inherited Metabolic Disease*. https://doi.org/10.1002/jimd.12751

171. Čunátová, K., Vrbacký, M., Puertas-Frias, G., Alán, L., Vanišová, M., Saucedo-Rodríguez, M. J., Houštěk, J., Fernández-Vizarra, E., Neužil, J., Pecinová, A., Pecina, P., & Mráček, T. (2024). Mitochondrial translation is the primary determinant of secondary mitochondrial complex I deficiencies. *iScience*, *27*(8), 110560. https://doi.org/10.1016/j.isci.2024.110560

172. de Miguel-Sánchez, C. J., Gómez, G. L., Hidalgo, R. L., Álvarez, I. C., Encinar, A. B., Blanco, J. L. M., Lotto, F. A., Rodríguez, M. I. E., & Sánchez, S. P. (2024). Multiple symmetric lipomatosis as a marker of mitochondrial disease. Case report and review of the literature. *Neurological Sciences: Official Journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology*. https://doi.org/10.1007/s10072-024-07710-6

173. Dhawan, S., Musa, A. H., & Mantripragada, K. (2024). Novel Mitochondrial Cytopathy Causing Mitochondrial Encephalomyopathy With Lactic Acidosis and Stroke-Like Episodes Syndrome and Tubulointerstitial Nephropathy. *Cureus*, *16*(8), e66722. https://doi.org/10.7759/cureus.66722

174. Di Nottia, M., Rizza, T., Baruffini, E., Nesti, C., Torraco, A., Diodato, D., Martinelli, D., Dal Canto, F., Gilea, A. I., Zoccola, M., Siri, B., Dionisi-Vici, C., Bertini, E., Santorelli, F. M., Goffrini, P., & Carrozzo, R. (2024). Severe mitochondrial encephalomyopathy caused by de novo variants in OPA1 gene. *Frontiers in Genetics*, *15*, 1437959. https://doi.org/10.3389/fgene.2024.1437959

175. Dong, Q., Yin, X., Fan, S., Zhong, S., Yang, W., Chen, K., Wang, Q., Ma, X., Mahlatsi, R. L., Yang, Y., Lyu, J., Fang, H., & Wang, Y. (2024). IARS2 mutations lead to Leigh syndrome with a combined oxidative phosphorylation deficiency. *Orphanet Journal of Rare Diseases*, *19*(1), 305. https://doi.org/10.1186/s13023-024-03310-x

176. Dupré, M., Warne, R., Shipman, P., Kava, M., Ghia, T., Loughman, L., & Lakshmanan, R. (2024). Cranial and spinal nerve enhancement in SURF1-associated Leigh syndrome. *Pediatric Radiology*, *54*(10), 1758-1762. https://doi.org/10.1007/s00247-024-06005-4

177. Dzwilewski, K., Chojnowski, K., Krygier, M., Zawadzka, M., Chylińska, M., & Mazurkiewicz-Bełdzińska, M. (2024). Effects of idebenone treatment in a patient with DNAJC30-associated Leigh Syndrome. *Neurologia I Neurochirurgia Polska*, *58*(4), 468-470. https://doi.org/10.5603/pjnns.100423

178. Erb, C., Erb, C., Kazakov, A., Kapanova, G., & Weisser, B. (2024). Lifestyle Changes in Aging and their Potential Impact on POAG. *Klinische Monatsblatter Fur Augenheilkunde*. https://doi.org/10.1055/a-2372-3505

179. Esmaeil, A., Ali, A., & Behbehani, R. (2022). Leber's hereditary optic neuropathy : Update on current diagnosis and treatment. *Frontiers in Ophthalmology*, *2*, 1077395. https://doi.org/10.3389/fopht.2022.1077395

180. Esteban-Amo, M. J., Jiménez-Cuadrado, P., Serrano-Lorenzo, P., de la Fuente, M. Á., & Simarro, M. (2024). Succinate Dehydrogenase and Human Disease : Novel Insights into a Well-Known Enzyme. *Biomedicines*, *12*(9), 2050. https://doi.org/10.3390/biomedicines12092050

181. Fabra, M. A., Paredes-Fuentes, A. J., Torralba Carnerero, M., Moreno Férnandez de Ayala, D. J., Arroyo Luque, A., Sánchez Cuesta, A., Staiano, C., Sanchez-Pintos, P., Luz Couce, M., Tomás, M., Marco-Hernández, A. V., Orellana, C., Martínez, F., Roselló, M., Caro, A., Oltra Soler, J. S., Monfort, S., Sánchez, A., Rausell, D., … Santos-Ocaña, C. (2024). New variants expand the neurological phenotype of COQ7 deficiency. *Journal of Inherited Metabolic Disease*. https://doi.org/10.1002/jimd.12776

182. Falabella, M., Pizzamiglio, C., Tabara, L. C., Munro, B., Abdel-Hamid, M. S., Sonmezler, E., Macken, W. L., Lu, S., Tilokani, L., Flannery, P. J., Patel, N., Pope, S. A. S., Heales, S. J. R., Hammadi, D. B. H., Alston, C. L., Taylor, R. W., Lochmuller, H., Woodward, C. E., Labrum, R., … Pitceathly, R. D. S. (2024). Biallelic PTPMT1 variants disrupt cardiolipin metabolism and lead to a neurodevelopmental syndrome. *Brain: A Journal of Neurology*, awae268. https://doi.org/10.1093/brain/awae268

183. Figuccia, S., Izzo, R., Legati, A., Nasca, A., Goffrini, P., Ghezzi, D., & Ceccatelli Berti, C. (2024). Investigation in yeast of novel variants in mitochondrial aminoacyl-tRNA synthetases WARS2, NARS2, and RARS2 genes associated with mitochondrial diseases. *Human Molecular Genetics*, *33*(18), 1630-1641. https://doi.org/10.1093/hmg/ddae104

184. Finsterer, J. (2024). Involvement of the Endocrine System is Common in Mitochondrial Disorders and Requires Long-term Comprehensive Investigations. *Journal of Clinical Research in Pediatric Endocrinology*. https://doi.org/10.4274/jcrpe.galenos.2024.2024-8-5

185. Forini, F., Levantini, E., Bramanti, E., Santorelli, F. M., Ali, A., Lionetti, V., & Rizzo, M. (2024). Editorial : Mitochondrial plasticity and quality control in health and disease. *Frontiers in Cell and Developmental Biology*, *12*, 1468818. https://doi.org/10.3389/fcell.2024.1468818

186. Ghannoum, M., Waters, P. J., Hovda, K. E., Choquette, G., Elgstøen, K. B. P., Nygaard, I., Rootwelt, H., Hickey, D., Yazdani, M., & Bourque, D. K. (2024). Can endogenous ethylene glycol production occur in humans? A detailed investigation of adult monozygotic twin sisters. *Clinical Toxicology (Philadelphia, Pa.)*, 1-9. https://doi.org/10.1080/15563650.2024.2401076

187. Giaj Levra, A., & Amata, F. (2024). Myoclonic Epilepsy With Ragged Red Fiber Cardiomyopathy : A Case Report and Brief Review of Literature. *Cureus*, *16*(8), e66745. https://doi.org/10.7759/cureus.66745

188. Gonçalves, F. P., Tavares, I., Silva, R., Nunes, A. T., Pereira, L., Campos, A., Pinto, J., Lopes, A., Simões, M., Grazina, M., Fogo, A. B., & Oliveira, J. P. (2024). Homozygosity for a Rare FASTKD2 Variant Resulting in an Adult Onset Autosomal Recessive Mitochondrial Podocytopathy. *American Journal of Kidney Diseases: The Official Journal of the National Kidney Foundation*, S0272-6386(24)00897-7. https://doi.org/10.1053/j.ajkd.2024.05.018

189. Grama, A., Benţa, G., Niculae, A. S., Mititelu, A., Simu, C., Fufezan, O., Stephenne, X., Reding, R., de Magnee, C., Tambucci, R., Sokal, E., & Pop, T. L. (2024). Favorable Outcome after Liver Transplantation in an Infant with Liver Failure Due to Deoxyguanosine Kinase Deficiency. *Journal of Clinical Medicine*, *13*(18), 5356. https://doi.org/10.3390/jcm13185356

190. Gürkan, Ö. E., Öztürk, H., Kaya, C., Kaya, N. G., Bozbulut, N. E., Can, A., Ceylan, K., Aksu, A. Ü., Düztaş, D. T., Sarı, S., Dalgıç, B., Kapısız, A., Coşkun, D., Emmez, G., İnan, G., Akdulum, İ., Pampal, H. K., Eryılmaz, N. C., Erel, S., … Sönmez, K. (2024). A Classification for Gastric Outlet Obstruction in Childhood : Extending Beyond Infantile Hypertrophic Pyloric Stenosis. *The Turkish Journal of Gastroenterology: The Official Journal of Turkish Society of Gastroenterology*, *35*(3), 255-261. https://doi.org/10.5152/tjg.2024.23202

191. Hammann, N., Lenz, D., Bianzano, A., Husain, R. A., Forman, E., Bernstein, J. A., Dattner, T., Engelen, M., Hanson-Kahn, A. K., Isidor, B., Kotzaeridou, U., Tietze, A., Trollmann, R., Weiß, C., Wolffenbuttel, B. H. R., Kölker, S., Hoffmann, G. F., Crushell, E., Staufner, C., … Harting, I. (2024). MRI in LARS1 deficiency-Spectrum, patterns, and correlation with acute neurological deterioration. *Journal of Inherited Metabolic Disease*. https://doi.org/10.1002/jimd.12764

192. Handley, S., Anwer, A. G., Knab, A., Bhargava, A., & Goldys, E. M. (2024). AutoMitoNetwork : Software for analyzing mitochondrial networks in autofluorescence images to enable label-free cell classification. *Cytometry. Part A: The Journal of the International Society for Analytical Cytology*, *105*(9), 688-703. https://doi.org/10.1002/cyto.a.24889

193. Haque, S., Crawley, K., Schofield, D., Shrestha, R., Davis, R., & Sue, C. M. (2024). Social provisions in patients with mitochondrial diseases. *BMJ Neurology Open*, *6*(2), e000770. https://doi.org/10.1136/bmjno-2024-000770

194. Haque, S., Crawley, K., Schofield, D., Shrestha, R., & Sue, C. M. (2024). Cascade testing in mitochondrial diseases : A cross-sectional retrospective study. *BMC Neurology*, *24*(1), 343. https://doi.org/10.1186/s12883-024-03850-6

195. Haschke, A. M., Diecke, S., & Schuelke, M. (2024). Characterization of two iPSC lines from patients with maternally inherited leigh (MILS) and neuropathy, ataxia, and retinitis pigmentosa (NARP) syndrome carrying the MT-ATP6 m.8993 T>G mutation at different degrees of heteroplasmy. *Stem Cell Research*, *81*, 103547. https://doi.org/10.1016/j.scr.2024.103547

196. Hawlina, M., Kovač, L., Breciková, K., Žigmond, J., Rogalewicz, V., Tichopád, A., Višňanský, M., & Šarkanová, I. (2024). Leber hereditary optic neuropathy in Slovenia : Quality of life and costs from patient perspective. *Orphanet Journal of Rare Diseases*, *19*(1), 318. https://doi.org/10.1186/s13023-024-03329-0

197. Houghton, D., Ng, Y. S., Jackson, M. A., Stefanetti, R., Hynd, P., Mac Aogáin, M., Stewart, C. J., Lamb, C. A., Bright, A., Feeney, C., Newman, J., Turnbull, D. M., McFarland, R., Blain, A. P., & Gorman, G. S. (2022). Phase II Feasibility Study of the Efficacy, Tolerability, and Impact on the Gut Microbiome of a Low-Residue (Fiber) Diet in Adult Patients With Mitochondrial Disease. *Gastro Hep Advances*, *1*(4), 666-677. https://doi.org/10.1016/j.gastha.2022.03.007

198. Huff, A., Oliveira, L. M., Karlen-Amarante, M., Ebiala, F., Ramirez, J. M., & Kalume, F. (2024). Ndufs4 inactivation in glutamatergic neurons reveals swallow-breathing discoordination in a mouse model of Leigh Syndrome. *bioRxiv: The Preprint Server for Biology*, 2024.09.11.612506. https://doi.org/10.1101/2024.09.11.612506

199. Hui, L., Hayman, P., Buckland, A., Fahey, M. C., Mackey, D. A., Mallett, A. J., Schweitzer, D. R., Stuart, C. P., Yau, W. Y., & Christodoulou, J. (2024). Pregnancy in women with mitochondrial disease-A literature review and suggested guidance for preconception and pregnancy care. *The Australian & New Zealand Journal of Obstetrics & Gynaecology*. https://doi.org/10.1111/ajo.13874

200. Ishikawa, K., Miyata, D., Hattori, S., Tani, H., Kuriyama, T., Wei, F.-Y., Miyakawa, T., & Nakada, K. (2024). Accumulation of mitochondrial DNA with a point mutation in tRNALeu(UUR) gene induces brain dysfunction in mice. *Pharmacological Research*, *208*, 107374. https://doi.org/10.1016/j.phrs.2024.107374

201. Islak, C., Bağcılar, Ö., Selçuk, H. H., Saltık, S., Korkmazer, B., Zubarioğlu, T., Arslan, S., Üstündag, A., & Kızılkılıç, O. (2024). A New Perspective On Arterioectatic Spinal Angiopathy with a Reversible Pattern : Cause or Consequence? *Clinical Neuroradiology*. https://doi.org/10.1007/s00062-024-01451-x

202. Jiang, H., Xu, C., Duan, R., Liu, Z., Ren, X., Li, J., Chen, C., Wang, H., Han, T., Tian, X., Duan, X., Song, M., Li, T., & Fang, F. (2024). Phenotypic spectrum of iron-sulfur cluster assembly gene IBA57 mutations : C.286 T > C identified as a hotspot mutation in Chinese patients with a stable natural history. *Journal of Human Genetics*. https://doi.org/10.1038/s10038-024-01291-0

203. Jing, S., Yao, Q., Wu, M., & Li, Y. (2024). Case Report : Lethal mitochondrial cardiomyopathy linked to a compound heterozygous variant of PARS2. *Frontiers in Cardiovascular Medicine*, *11*, 1446055. https://doi.org/10.3389/fcvm.2024.1446055

204. Jørstad, Ø. K., Skaar, S., Strand, H., Røsby, O., Brokstad, R. T., & Rønning, P. A. (2024). Leber Hereditary Optic Neuropathy Case Report : Clinical Presentation and Treatment with Idebenone Reinforce the Evidence for m.3866T>C as a Causative Variant. *Case Reports in Ophthalmology*, *15*(1), 513-517. https://doi.org/10.1159/000539445

205. Joyce, E., Ali, M., Richard, G., Kelly, S., Martin, F., Conlon, P. J., Whelehan, A., Ng, Y. S., & Lefter, S. (2024). Adult presentation of a rare mitochondrial tRNA Val gene mutation-an expanding clinical phenotype. *European Journal of Neurology*, *31*(10), e16405. https://doi.org/10.1111/ene.16405

206. Kaur, P., Nazeer, N., Gurjar, V., Tiwari, R., & Mishra, P. K. (2024). Nanophotonic waveguide-based sensing of circulating cell-free mitochondrial DNA : Implications for personalized medicine. *Drug Discovery Today*, *29*(8), 104086. https://doi.org/10.1016/j.drudis.2024.104086

207. Keshavan, N., Mhaldien, L., Gilmour, K., & Rahman, S. (2024). Interferon Stimulated Gene Expression Is a Biomarker for Primary Mitochondrial Disease. *Annals of Neurology*. https://doi.org/10.1002/ana.27081

208. Keshavan, N., & Rahman, S. (2024). Natural history of deoxyguanosine kinase deficiency. *Molecular Genetics and Metabolism*, *143*(1-2), 108554. https://doi.org/10.1016/j.ymgme.2024.108554

209. Khaghani, F., Hemmati, M., Ebrahimi, M., & Salmaninejad, A. (2024). Emerging Multi-omic Approaches to the Molecular Diagnosis of Mitochondrial Disease and Available Strategies for Treatment and Prevention. *Current Genomics*, *25*(5), 358-379. https://doi.org/10.2174/0113892029308327240612110334

210. Khaliulin, I., Hamoudi, W., & Amal, H. (2024). The multifaceted role of mitochondria in autism spectrum disorder. *Molecular Psychiatry*. https://doi.org/10.1038/s41380-024-02725-z

211. Landoni, J. C., Erkul, S., Laalo, T., Goffart, S., Kivelä, R., Skube, K., Nieminen, A. I., Wickström, S. A., Stewart, J., & Suomalainen, A. (2024). Overactive mitochondrial DNA replication disrupts perinatal cardiac maturation. *Nature Communications*, *15*(1), 8066. https://doi.org/10.1038/s41467-024-52164-1

212. Lasham, J., Djurabekova, A., Kolypetris, G., Zickermann, V., Vonck, J., & Sharma, V. (2024). Assessment of amino acid charge states based on cryo-electron microscopy and molecular dynamics simulations of respiratory complex I. *Biochimica Et Biophysica Acta. Bioenergetics*, *1866*(1), 149512. https://doi.org/10.1016/j.bbabio.2024.149512

213. Lavorato, M., Iadarola, D., Remes, C., Kaur, P., Broxton, C., Mathew, N. D., Xiao, R., Seiler, C., Nakamaru-Ogiso, E., Anderson, V. E., & Falk, M. J. (2024). Dldhcri3 zebrafish exhibit altered mitochondrial ultrastructure, morphology, and dysfunction partially rescued by probucol or thiamine. *JCI Insight*, *9*(18), e178973. https://doi.org/10.1172/jci.insight.178973

214. Lehmer, M., & Zoncu, R. (2024). mTORC1 Signaling Inhibition Modulates Mitochondrial Function in Frataxin Deficiency. *bioRxiv: The Preprint Server for Biology*, 2024.08.06.606942. https://doi.org/10.1101/2024.08.06.606942

215. Leister, N., Wendt, S., Hedergott, A., Heindl, L. M., Rokohl, A. C., Stoll, S. E., Gordon, E., Böttiger, B. W., Fricke, J., & Schick, V. C. (2024). Anaesthesia Concepts in Patients with Chronic Progressive External Ophthalmoplegia Undergoing Ophthalmic Surgery-A Retrospective Cohort Analysis. *Journal of Clinical Medicine*, *13*(16), 4710. https://doi.org/10.3390/jcm13164710

216. Levee, V., Sivaganesh, K., Schaeffer, A., & Karunaratne, K. (2024). POLG epilepsy presenting as new-onset refractory status epilepticus (NORSE) in pregnancy. *Practical Neurology*, pn-2024-004232. https://doi.org/10.1136/pn-2024-004232

217. Leyfer, D., & Fetterman, J. L. (2023). Beyond MitoCarta-expanding the list of candidate proteins involved in mitochondrial functions using a biological network approach. *NAR Genomics and Bioinformatics*, *5*(4), lqad107. https://doi.org/10.1093/nargab/lqad107

218. Li, J., Fang, X., Cui, D., Ma, Z., Yang, J., Niu, Y., Liu, H., & Xiang, P. (2024). Mechanistic insights into cadmium exacerbating 2-Ethylhexyl diphenyl phosphate-induced human keratinocyte toxicity : Oxidative damage, cell apoptosis, and tight junction disruption. *Ecotoxicology and Environmental Safety*, *283*, 116858. https://doi.org/10.1016/j.ecoenv.2024.116858

219. Li, R., Ma, M., Chen, W., & Qiu, Z. (2024). Classic ketogenic diet-induced ketoacidosis in the treatment of pyruvate dehydrogenase deficiency : A case report and literature review. *BMC Pediatrics*, *24*(1), 603. https://doi.org/10.1186/s12887-024-05054-w

220. Li, Y.-X., Wang, D.-J., Zhou, M.-B., Sun, H.-X., Hong, S.-Q., Jiang, L., & Guo, Y. (2024). [Leigh syndrome caused by the mitochondrial m.8993T>G mutation with hypocitrullinemia : A report of four cases and literature review]. *Zhongguo Dang Dai Er Ke Za Zhi = Chinese Journal of Contemporary Pediatrics*, *26*(9), 940-945. https://doi.org/10.7499/j.issn.1008-8830.2404036

221. Lillo, R., Meucci, M. C., Malara, S., Primiano, G., Servidei, S., Lombardo, A., Grandinetti, M., Massetti, M., Lanza, G. A., Limongelli, G., & Graziani, F. (2024). Early cardiac mechanics abnormalities in patients with mitochondrial diseases. *Mitochondrion*, *78*, 101940. https://doi.org/10.1016/j.mito.2024.101940

222. Lin, Y., Wang, J., Zhuang, X., Zhao, Y., Wang, W., Wang, D., Zhao, Y., Yan, C., & Ji, K. (2024). Queuine ameliorates impaired mitochondrial function caused by mt-tRNAAsn variants. *Journal of Translational Medicine*, *22*(1), 780. https://doi.org/10.1186/s12967-024-05574-0

223. Machado, C. M., de-Souza-Ferreira, E., Silva, G. F. S., Pimentel, F. S. A., De-Souza, E. A., Silva-Rodrigues, T., Gandara, A. C. P., Zeidler, J. D., Fernandes-Siqueira, L. O., De-Queiroz, A. L. F. V., Andrade-Silva, L., Victória-Martins, K., Fernandes-Carvalho, C., Chini, E. N., Passos, J. F., Da Poian, A. T., Montero-Lomelí, M., Galina, A., & Masuda, C. A. (2024). Galactose-1-phosphate inhibits cytochrome c oxidase and causes mitochondrial dysfunction in classic galactosemia. *Biochimica Et Biophysica Acta. Molecular Basis of Disease*, *1870*(7), 167340. https://doi.org/10.1016/j.bbadis.2024.167340

224. Majamaa, K., Kärppä, M., & Moilanen, J. S. (2024). Neurological manifestations in adult patients with the m.3243A>G variant in mitochondrial DNA. *BMJ Neurology Open*, *6*(2), e000825. https://doi.org/10.1136/bmjno-2024-000825

225. Malik, M. S., Chang, Y.-Y., Liu, Y.-C., Le, V. T., & Ou, Y.-Y. (2024). MCNN_MC : Computational Prediction of Mitochondrial Carriers and Investigation of Bongkrekic Acid Toxicity Using Protein Language Models and Convolutional Neural Networks. *Journal of Chemical Information and Modeling*. https://doi.org/10.1021/acs.jcim.4c00961

226. Manuelli, M., Migliorelli, A., Bianchini, C., Stomeo, F., Pelucchi, S., Genovese, E., Monzani, D., Palma, S., & Ciorba, A. (2024). Unilateral Hearing Loss and Auditory Asymmetry in Mitochondrial Disease : A Scoping Review. *Journal of Clinical Medicine*, *13*(17), 5044. https://doi.org/10.3390/jcm13175044

227. Mello, D. F., Perez, L., Bergemann, C. M., Morton, K. S., Ryde, I. T., & Meyer, J. N. (2024). Comprehensive characterization of mitochondrial bioenergetics at different larval stages reveals novel insights about the developmental metabolism of Caenorhabditis elegans. *bioRxiv: The Preprint Server for Biology*, 2024.06.26.600841. https://doi.org/10.1101/2024.06.26.600841

228. Mora-Romero, B., Capelo-Carrasco, N., Pérez-Moreno, J. J., Alvarez-Vergara, M. I., Trujillo-Estrada, L., Romero-Molina, C., Martinez-Marquez, E., Morano-Catalan, N., Vizuete, M., Lopez-Barneo, J., Nieto-Gonzalez, J. L., Garcia-Junco-Clemente, P., Vitorica, J., Gutierrez, A., Macias, D., Rosales-Nieves, A. E., & Pascual, A. (2024). Microglia mitochondrial complex I deficiency during development induces glial dysfunction and early lethality. *Nature Metabolism*, *6*(8), 1479-1491. https://doi.org/10.1038/s42255-024-01081-0

229. Motegi, M., Sakurai, Y., Mio, Y., & Ohashi, T. (2024). Cochlear Implantation for Isoleucyl-tRNA Synthetase Mutation-Associated Mitochondrial Disease : A Case Report. *Cureus*, *16*(8), e67760. https://doi.org/10.7759/cureus.67760

230. Nakai, R., Varnum, S., Field, R. L., Shi, H., Giwa, R., Jia, W., Krysa, S. J., Cohen, E. F., Borcherding, N., Saneto, R. P., Tsai, R. C., Suganuma, M., Ohta, H., Yokota, T., & Brestoff, J. R. (2024). Mitochondria transfer-based therapies reduce the morbidity and mortality of Leigh syndrome. *Nature Metabolism*, *6*(10), 1886-1896. https://doi.org/10.1038/s42255-024-01125-5

231. Navarro Capone, R., Bofill, A., López, L., & Astroza Eulufi, G. (2024). [Mitochondrial diseases and urolithiasis : Is there an association? Literature review about 2 cases]. *Revista Medica De Chile*, *152*(1), 111-118. https://doi.org/10.4067/s0034-98872024000100111

232. Newstead, S. M., & Finsterer, J. (2024). Hyperkinesias in Leigh-like Syndrome with Complex-I Deficiency Due to m.10191T>C in MT-ND3. *Annals of African Medicine*, *23*(3), 512-513. https://doi.org/10.4103/aam.aam_32_23

233. Nguyen-Dien, G. T., Townsend, B., Kulkarni, P. G., Kozul, K.-L., Ooi, S. S., Eldershaw, D. N., Weeratunga, S., Liu, M., Jones, M. J., Millard, S. S., Ng, D. C., Pagano, M., Bonfim-Melo, A., Schneider, T., Komander, D., Lazarou, M., Collins, B. M., & Pagan, J. K. (2024). PPTC7 antagonizes mitophagy by promoting BNIP3 and NIX degradation via SCFFBXL4. *EMBO Reports*, *25*(8), 3324-3347. https://doi.org/10.1038/s44319-024-00181-y

234. Pannier, E., Sekri, A., Roux, N., Vasiljevic, A., El Khattabi, L., Chatron, N., Grotto, S., Menzella, D., Grangé, G., Thébault, F., Massardier, J., Fourrage, C., Lohmann, L., Tsatsaris, V., Putoux, A., Boutaud, L., & Attié-Bitach, T. (2024). Prenatal diagnosis of SLC25A24 Fontaine progeroid syndrome : Description of the fetal phenotype, genotype and detection of parental mosaicism. *Birth Defects Research*, *116*(7), e2380. https://doi.org/10.1002/bdr2.2380

235. Papatya Çakır, E. D., Ersıoy, M., Çakır Biçer, N., & Gedikbaşı, A. (2024). Endocrine Disorders in Children with Primary Mitochondrial Diseases : Single-Center Experience. *Journal of Clinical Research in Pediatric Endocrinology*. https://doi.org/10.4274/jcrpe.galenos.2024.2024-1-11

236. Pekeles, H., Berrahmoune, S., Dassi, C., Cheung, A. C. T., Gagnon, T., Waters, P. J., Eberhard, R., Buhas, D., & Myers, K. A. (2024). Safety and efficacy of deoxycytidine/deoxythymidine combination therapy in POLG-related disorders : 6-month interim results of an open-label, single arm, phase 2 trial. *EClinicalMedicine*, *74*, 102740. https://doi.org/10.1016/j.eclinm.2024.102740

237. Pelayo, G., Paiva Coelho, M., Correia, J., Bandeira, A., Nogueira, C., Vilarinho, L., & Martins, E. (2024). Phenotyping mitochondrial glutamyl-tRNA synthetase deficiency (EARS2) : A case series and systematic literature review. *Neurobiology of Disease*, *200*, 106644. https://doi.org/10.1016/j.nbd.2024.106644

238. Pieles, G., Steward, C., Dabner, L., Collet, L., Culliford, L., Sheehan, K., Ellis, L., Damin, M., Sammut, E., Duarte, N., Burgess, O., Wadey, C., Williams, C., Crosby, J., Groves, S., Searle, A., Amulic, B., Rice, C., Bucciarelli-Ducci, C., … Reeves, B. C. (2024). *Bezafibrate as treatment in males for Barth syndrome : CARDIOMAN, a double-blind, placebo-controlled crossover RCT*. National Institute for Health and Care Research. http://www.ncbi.nlm.nih.gov/books/NBK606703/

239. Pietraszkiewicz, A., Ayesha, A., Digre, K. B., Warner, J. E., Seay, M. D., Crum, A. V., & Katz, B. J. (2024). Diagnostic dilemma : Leber's hereditary optic neuropathy in a 70-year-Old woman. *American Journal of Ophthalmology Case Reports*, *36*, 102143. https://doi.org/10.1016/j.ajoc.2024.102143

240. Pinto Payares, D. V., Spooner, L., Vosters, J., Dominguez, S., Patrick, L., Harris, A., & Kanungo, S. (2024). A systematic review on the role of mitochondrial dysfunction/disorders in neurodevelopmental disorders and psychiatric/behavioral disorders. *Frontiers in Psychiatry*, *15*, 1389093. https://doi.org/10.3389/fpsyt.2024.1389093

241. Pizzamiglio, C., Hanna, M. G., & Pitceathly, R. D. S. (2024). Primary mitochondrial diseases. *Handbook of Clinical Neurology*, *204*, 53-76. https://doi.org/10.1016/B978-0-323-99209-1.00004-1

242. Prasun, P., & Kohli, U. (2024). Making a case for mitochondria in hypertrophic cardiomyopathy. *Future Cardiology*, *20*(4), 179-182. https://doi.org/10.1080/14796678.2024.2360355

243. Puighermanal, E., Luna-Sánchez, M., Gella, A., van der Walt, G., Urpi, A., Royo, M., Tena-Morraja, P., Appiah, I., de Donato, M. H., Menardy, F., Bianchi, P., Esteve-Codina, A., Rodríguez-Pascau, L., Vergara, C., Gómez-Pallarès, M., Marsicano, G., Bellocchio, L., Martinell, M., Sanz, E., … Quintana, A. (2024). Cannabidiol ameliorates mitochondrial disease via PPARγ activation in preclinical models. *Nature Communications*, *15*(1), 7730. https://doi.org/10.1038/s41467-024-51884-8

244. Rebs, S., & Streckfuss-Bömeke, K. (2023). How can we use stem cell-derived cardiomyocytes to understand the involvement of energetic metabolism in alterations of cardiac function? *Frontiers in Molecular Medicine*, *3*, 1222986. https://doi.org/10.3389/fmmed.2023.1222986

245. Riccio, A. A., Bouvette, J., Pedersen, L. C., Somai, S., Dutcher, R. C., Borgnia, M. J., & Copeland, W. C. (2024). Structures of the mitochondrial single-stranded DNA binding protein with DNA and DNA polymerase γ. *Nucleic Acids Research*, *52*(17), 10329-10340. https://doi.org/10.1093/nar/gkae670

246. Rius, R., Compton, A. G., Baker, N. L., Balasubramaniam, S., Best, S., Bhattacharya, K., Boggs, K., Boughtwood, T., Braithwaite, J., Bratkovic, D., Bray, A., Brion, M.-J., Burke, J., Casauria, S., Chong, B., Coman, D., Cowie, S., Cowley, M., de Silva, M. G., … Thorburn, D. R. (2024). The Australian Genomics Mitochondrial Flagship : A national program delivering mitochondrial diagnoses. *Genetics in Medicine: Official Journal of the American College of Medical Genetics*, 101271. https://doi.org/10.1016/j.gim.2024.101271

247. Rötig, A., Gaignard, P., Barcia, G., Assouline, Z., Berat, C.-M., Barth, M., Damaj, L., Laborde, N., Abi-Warde, M.-T., Chabrol, B., De Lonlay, P., Desguerre, I., Goldenberg, A., Gonzales, E., Jacquemin, E., Amati-Bonneau, P., Bonneau, D., Abadie, V., Bonnemains, C., … Schiff, M. (2024). Distinct Clinical Courses and Shortened Lifespans in Childhood-Onset DNA Polymerase Gamma Deficiency. *Neurology. Genetics*, *10*(4), e200167. https://doi.org/10.1212/NXG.0000000000200167

248. Ru, Y., Deng, X., Chen, J., Zhang, L., Xu, Z., Lv, Q., Long, S., Huang, Z., Kong, M., Guo, J., & Jiang, M. (2024). Maternal age enhances purifying selection on pathogenic mutations in complex I genes of mammalian mtDNA. *Nature Aging*, *4*(9), 1211-1230. https://doi.org/10.1038/s43587-024-00672-6

249. Rubalcava-Gracia, D., Bubb, K., Levander, F., Burr, S. P., August, A. V., Chinnery, P. F., Koolmeister, C., & Larsson, N.-G. (2024). LRPPRC and SLIRP synergize to maintain sufficient and orderly mammalian mitochondrial translation. *Nucleic Acids Research*, *52*(18), 11266-11282. https://doi.org/10.1093/nar/gkae662

250. Sen, K., Harrar, D., Pariseau, N., Tucker, K., Keenan, J., Zhang, A., & Gropman, A. (2024). Seizure Characteristics and EEG Features in Intoxication Type and Energy Deficiency Neurometabolic Disorders in the Pediatric Intensive Care Unit : Single-Center Experience Over 10 Years. *Neurocritical Care*. https://doi.org/10.1007/s12028-024-02073-4

251. Sepulveda, C. J., Walsh, E., Carlin, K., & Saneto, R. P. (2024). Using a Previsit Questionnaire for Initial Visits in a Pediatric Mitochondrial Clinic : Perspectives of Parents, a Specialty Physician, and a Clinical Coordinator. *Journal of Child Neurology*, *39*(13-14), 453-460. https://doi.org/10.1177/08830738241278388

252. Sharma, S., Mahadevan, A., Narayanappa, G., Debnath, M., Govindaraj, P., Shivaram, S., Seshagiri, D. V., Siram, R., Shroti, A., Bindu, P. S., Chickabasaviah, Y. T., Taly, A. B., & Nagappa, M. (2024). Exploring the evidence for mitochondrial dysfunction and genetic abnormalities in the etiopathogenesis of tropical ataxic neuropathy. *Journal of Neurogenetics*, *38*(2), 27-34. https://doi.org/10.1080/01677063.2024.2373363

253. Stańczyk, M., Szubart, N., Maslanka, R., & Zadrag-Tecza, R. (2024). Mitochondrial Dysfunctions : Genetic and Cellular Implications Revealed by Various Model Organisms. *Genes*, *15*(9), 1153. https://doi.org/10.3390/genes15091153

254. Sun, L., Li, X., Li, H., & Peng, L. (2024). Treating hyperglycaemia in a patient with maternally inherited diabetes and deafness with an inhibitor of dipeptidyl peptidase-4 : A case report and two-year follow-up. *Acta Diabetologica*. https://doi.org/10.1007/s00592-024-02366-2

255. Sun, L., Ren, Y., Ma, Y., Zhu, Y., Wu, Y., Wang, S., Nie, L., Xiao, H., Jiang, Y., & Wang, F. (2024). A de novo novel variant in the MT-TD gene is associated with prominent extra-neurologic manifestations. *Clinical Genetics*, *106*(6), 745-749. https://doi.org/10.1111/cge.14594

256. Tashima, K., Hayashi, M., Oyoshi, T., Uemura, J., Korematsu, S., & Hirata, N. (2024). Anesthesia management for percutaneous mitral valve repair in a patient with mitochondrial cardiomyopathy and low cardiac function : A case report. *JA Clinical Reports*, *10*(1), 49. https://doi.org/10.1186/s40981-024-00734-z

257. Tauchmannová, K., Pecinová, A., Houštěk, J., & Mráček, T. (2024). Variability of Clinical Phenotypes Caused by Isolated Defects of Mitochondrial ATP Synthase. *Physiological Research*, *73*(Suppl 1), S243-S278. https://doi.org/10.33549/physiolres.935407

258. Traunero, A., Baldo, F., Magnolato, A., Di Leo, G., Barbi, E., & Bruno, I. (2024). Administration of bicarbonates through percutaneous gastrostomy with continuous nocturnal infusion in a patient with Kearns-Sayre disease : A life changing therapeutical paradigm. *Italian Journal of Pediatrics*, *50*(1), 132. https://doi.org/10.1186/s13052-024-01696-9

259. Tsai, N.-C., Liou, C.-W., Cheng, Y.-H., Lien, H.-T., Lin, T.-L., Lin, T.-K., Lan, M.-Y., Hung, P.-L., Wang, T.-J., Lee, C.-H., Liang, Y.-C., & Lan, K.-C. (2024). The establishment of a molecular diagnostic platform for mitochondrial diseases : From conventional to next-generation sequencing. *Biomedical Journal*, 100770. https://doi.org/10.1016/j.bj.2024.100770

260. Tsuruno, T., Tateiwa, H., Hashimoto, Y., Katsumata, Y., & Kawano, T. (2024). Remimazolam Anesthesia for a Pediatric Patient With Glutaric Aciduria Type I : A Case Report. *Cureus*, *16*(8), e66612. https://doi.org/10.7759/cureus.66612

261. Urzì, C., Meyer, C., Mathis, D., Vermathen, P., & Nuoffer, J.-M. (2024). Intra- and extracellular real-time analysis of perfused fibroblasts using an NMR bioreactor : A pilot study. *Journal of Inherited Metabolic Disease*. https://doi.org/10.1002/jimd.12794

262. Walker, B. R., Theard, L.-M., Pinto, M., Rodriguez-Silva, M., Bacman, S. R., & Moraes, C. T. (2024). Restoration of defective oxidative phosphorylation to a subset of neurons prevents mitochondrial encephalopathy. *EMBO Molecular Medicine*, *16*(9), 2210-2232. https://doi.org/10.1038/s44321-024-00111-4

263. Wang, R., Bao, F., Lu, M., Jia, X., Xiao, J., Wu, Y., Zhang, Q., & Liu, X. (2024). MSC-mediated mitochondrial transfer restores mitochondrial DNA and function in neural progenitor cells of Leber's hereditary optic neuropathy. *Science China. Life Sciences*, *67*(11), 2511-2519. https://doi.org/10.1007/s11427-024-2647-8

264. Watanabe, M., & Sasaki, N. (2024). Mechanisms and Future Research Perspectives on Mitochondrial Diseases Associated with Isoleucyl-tRNA Synthetase Gene Mutations. *Genes*, *15*(7), 894. https://doi.org/10.3390/genes15070894

265. Wei, L.-Y., Chen, X.-Q., Huang, L., Shan, Q.-W., & Tang, Q. (2024). Liver transplantation for mitochondrial DNA depletion syndrome caused by MPV17 deficiency : A case report and literature review. *Frontiers in Surgery*, *11*, 1348806. https://doi.org/10.3389/fsurg.2024.1348806

266. Xu, H. N., Morrow, R. M., Feng, M., Zhao, H., Wallace, D., & Li, L. Z. (2024). Optical redox imaging of ANT1-deficient muscles. *Journal of Innovative Optical Health Sciences*, *17*(1), 2350032. https://doi.org/10.1142/s1793545823500323

267. Yan, M., Zhang, Q., Chen, Y., Zhu, C., Wang, D., Tan, J., He, B., Li, Q., Deng, X., & Wan, Y. (2024). α-Synuclein-mediated mitochondrial translocation of cofilin-1 leads to oxidative stress and cell apoptosis in PD. *Frontiers in Neuroscience*, *18*, 1420507. https://doi.org/10.3389/fnins.2024.1420507

268. Yang, H., Zhang, V. W., Ai, L., & Wu, L. (2024). A Novel m.1636A > G Variant in Mitochondrial TV Gene Might Cause New Phenotype of Mitochondrial Disease in a 2-Year Old Chinese Boy. *Molecular Neurobiology*. https://doi.org/10.1007/s12035-024-04472-2

269. Yang, J., Wang, Y., Wang, G., Guo, Z., Li, X., Lu, J., Tu, H., Li, S., Wan, J., Guan, G., & Chen, L. (2024). Leptin A deficiency affecting the mitochondrial dynamics of aged oocytes in medaka (Oryzias latipes). *Molecular and Cellular Endocrinology*, *593*, 112345. https://doi.org/10.1016/j.mce.2024.112345

270. Yildirim, R. M., & Seli, E. (2024). Mitochondria as therapeutic targets in assisted reproduction. *Human Reproduction (Oxford, England)*, *39*(10), 2147-2159. https://doi.org/10.1093/humrep/deae170

271. Yu, K.-K., Li, K., Wang, H.-Y., Li, X.-L., Wu, S.-X., Xu, W.-M., Liu, Y.-H., Wu, C.-F., Yu, X.-Q., & Bao, J.-K. (2024). Construction of Near-Infrared Probes with Remarkable Large Stokes Shift Based on a Novel Purine Platform for the Visualization of mtG4 Upregulation during Mitochondrial Disorder in Somatic Cells and Human Sperms. *Analytical Chemistry*, *96*(29), 11915-11922. https://doi.org/10.1021/acs.analchem.4c01638

272. Zachos, K. A., Gamboa, J. A., Dewji, A. S., Lee, J., Brijbassi, S., & Andreazza, A. C. (2024). The interplay between mitochondria, the gut microbiome and metabolites and their therapeutic potential in primary mitochondrial disease. *Frontiers in Pharmacology*, *15*, 1428242. https://doi.org/10.3389/fphar.2024.1428242

273. Zhang, S., Zhou, Y., Shen, J., Wang, Y., Xia, J., Li, C., Liu, W., Hayat, K., & Qian, M. (2024). Early-Life Exposure to 4-Hydroxy-4'-Isopropoxydiphenylsulfone Induces Behavioral Deficits Associated with Autism Spectrum Disorders in Mice. *Environmental Science & Technology*, *58*(36), 15984-15996. https://doi.org/10.1021/acs.est.4c04760

274. Zhang, X., Yi, Z., Tang, W., & Wei, W. (2024). Streamlined process for effective and strand-selective mitochondrial base editing using mitoBEs. *Biophysics Reports*, *10*(4), 191-200. https://doi.org/10.52601/bpr.2024.240010

275. Zhao, M., Shen, Z., Zheng, Z., Xu, Y., Zhang, J., Liu, J., Peng, S., Wan, J., Qin, J.-J., & Wang, M. (2024). Cardiomyocyte LGR6 alleviates ferroptosis in diabetic cardiomyopathy via regulating mitochondrial biogenesis. *Metabolism: Clinical and Experimental*, *159*, 155979. https://doi.org/10.1016/j.metabol.2024.155979

276. Zhao, X., Yu, M., Zhang, W., Hou, Y., Yuan, Y., & Wang, Z. (2024). Demographic characteristics, diagnostic challenges, treatment patterns, and caregiver burden of mitochondrial diseases : A retrospective cross-sectional study. *Orphanet Journal of Rare Diseases*, *19*(1), 287. https://doi.org/10.1186/s13023-024-03289-5

277. Zhao, Y., Zhao, X., Ji, K., Wang, J., Zhao, Y., Lin, J., Gang, Q., Yu, M., Yuan, Y., Jiang, H., Sun, C., Fang, F., Yan, C., & Wang, Z. (2024). The clinical and genetic spectrum of mitochondrial diseases in China : A multicenter retrospective cross-sectional study. *Clinical Genetics*, *106*(6), 733-744. https://doi.org/10.1111/cge.14605