L'essentielle
Maladies Mitochondriales

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Veille bibliographique trimestrielle
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Cette veille, effectuée à partir de requêtes sur PubMed® sans prétendre à l'exhaustivité, présente une sélection de références d'articles médico-scientifiques concernant le domaine des maladies mitochondriales.

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Ataxie de Friedreich - *Friedreich ataxia*

1. Alemany-Perna, B., Tamarit, J., Cabiscol, E., Delaspre, F., Miguela, A., Huertas-Pons, J. M., Quiroga-Varela, A., Merchan Ruiz, M., López Domínguez, D., Ramió I Torrentà, L., Genís, D., & Ros, J. (2024). Calcitriol Treatment Is Safe and Increases Frataxin Levels in Friedreich Ataxia Patients. *Movement Disorders: Official Journal of the Movement Disorder Society*, *39*(7), 1099-1108. https://doi.org/10.1002/mds.29808

2. Balderas, E., Lee, S. H. J., Rai, N. K., Mollinedo, D. M., Duron, H. E., & Chaudhuri, D. (2024). Mitochondrial Calcium Regulation of Cardiac Metabolism in Health and Disease. *Physiology (Bethesda, Md.)*, *39*(5), 0. https://doi.org/10.1152/physiol.00014.2024

3. Boesch, S., & Indelicato, E. (2024). Approval of omaveloxolone for Friedreich ataxia. *Nature Reviews. Neurology*, *20*(6), 313-314. https://doi.org/10.1038/s41582-024-00957-9

4. Cantrell, A. C., Besanson, J., Williams, Q., Hoang, N., Edwards, K., Bishop, G. R., Chen, Y., Zeng, H., & Chen, J.-X. (2024). Ferrostatin-1 specifically targets mitochondrial iron-sulfur clusters and aconitase to improve cardiac function in Sirtuin 3 cardiomyocyte knockout mice. *Journal of Molecular and Cellular Cardiology*, *192*, 36-47. https://doi.org/10.1016/j.yjmcc.2024.05.003

5. Corben, L. A., Blomfield, E., Tai, G., Bilal, H., Harding, I. H., Georgiou-Karistianis, N., Delatycki, M. B., & Vogel, A. P. (2024). The Role of Verbal Fluency in the Cerebellar Cognitive Affective Syndrome Scale in Friedreich Ataxia. *Cerebellum (London, England)*, *23*(5), 1975-1980. https://doi.org/10.1007/s12311-024-01694-x

6. Da Conceição, L. M. A., Cabral, L. M., Pereira, G. R. C., & De Mesquita, J. F. (2024). An In Silico Analysis of Genetic Variants and Structural Modeling of the Human Frataxin Protein in Friedreich's Ataxia. *International Journal of Molecular Sciences*, *25*(11), 5796. https://doi.org/10.3390/ijms25115796

7. Dong, Y. N., Mercado-Ayón, E., Coulman, J., Flatley, L., Ngaba, L. V., Adeshina, M. W., & Lynch, D. R. (2024). The Regulation of the Disease-Causing Gene FXN. *Cells*, *13*(12), 1040. https://doi.org/10.3390/cells13121040

8. Ferreira, J. (2024). AAV gene therapy to treat Friedreich's ataxia cardiomyopathy. *Lab Animal*, *53*(4), 86. https://doi.org/10.1038/s41684-024-01351-0

9. Fichera, M., Nanetti, L., Monelli, A., Castaldo, A., Marchini, G., Neri, M., Vukaj, X., Marzorati, M., Porcelli, S., & Mariotti, C. (2024). Accelerometer-based measures in Friedreich ataxia : A longitudinal study on real-life activity. *Frontiers in Pharmacology*, *15*, 1342965. https://doi.org/10.3389/fphar.2024.1342965

10. Gnimpieba, E., Diing, D. M., Ailts, J., Cucak, A., Gakh, O., Isaya, G., Vitiello, S., Wang, S., Pierce, P., Cooper, A., Roux, K., Rogers, L. K., & Vitiello, P. F. (2024). Mapping Novel Frataxin Mitochondrial Networks Through Protein- Protein Interactions. *Research Square*, rs.3.rs-4259413. https://doi.org/10.21203/rs.3.rs-4259413/v1

11. Grander, M., Haschka, D., Indelicato, E., Kremser, C., Amprosi, M., Nachbauer, W., Henninger, B., Stefani, A., Högl, B., Fischer, C., Seifert, M., Kiechl, S., Weiss, G., & Boesch, S. (2024). Genetic Determined Iron Starvation Signature in Friedreich's Ataxia. *Movement Disorders: Official Journal of the Movement Disorder Society*, *39*(7), 1088-1098. https://doi.org/10.1002/mds.29819

12. Gunther, K., & Lynch, D. R. (2024). Pharmacotherapeutic strategies for Friedreich Ataxia : A review of the available data. *Expert Opinion on Pharmacotherapy*, *25*(5), 529-539. https://doi.org/10.1080/14656566.2024.2343782

13. Harding, I. H., Nur Karim, M. I., Selvadurai, L. P., Corben, L. A., Delatycki, M. B., Monti, S., Saccà, F., Georgiou-Karistianis, N., Cocozza, S., & Egan, G. F. (2024). Localized Changes in Dentate Nucleus Shape and Magnetic Susceptibility in Friedreich Ataxia. *Movement Disorders: Official Journal of the Movement Disorder Society*, *39*(7), 1109-1118. https://doi.org/10.1002/mds.29816

14. Hynes, S. M., Goldsberry, A., Henneghan, P. D., Murai, M., Shinde, A., Wells, J. A., Wu, L., Wu, T., Zahir, H., & Khan, S. (2024). Relative Bioavailability of Omaveloxolone When Capsules Are Sprinkled Over and Mixed in Applesauce Compared With Administration as Intact Omaveloxolone Capsules : A Phase 1, Randomized, Open-Label, Single-Dose, Crossover Study in Healthy Adults. *Journal of Clinical Pharmacology*, *64*(10), 1304-1311. https://doi.org/10.1002/jcph.2482

15. Indelicato, E., & Boesch, S. (2024). Reply to : « Advancing Understanding of Predictive Factors for Survival in Friedreich's Ataxia: A Review of Current Evidence and Future Directions ». *Movement Disorders: Official Journal of the Movement Disorder Society*, *39*(6), 1082. https://doi.org/10.1002/mds.29873

16. Jullian, E., Russi, M., Turki, E., Bouvelot, M., Tixier, L., Middendorp, S., Martin, E., & Monnier, V. (2024). Glial overexpression of Tspo extends lifespan and protects against frataxin deficiency in Drosophila. *Biochimie*, *224*, 71-79. https://doi.org/10.1016/j.biochi.2024.05.003

17. Kissell, J., Rochmann, C., Minini, P., Eichler, F., Stephen, C. D., Lau, H., Toro, C., Johnston, J. M., Krupnick, R., Hamed, A., & Cox, G. F. (2024). Clinical outcome assessments of disease burden and progression in late-onset GM2 gangliosidoses. *Molecular Genetics and Metabolism*, *142*(3), 108512. https://doi.org/10.1016/j.ymgme.2024.108512

18. Koka, M., Li, H., Akther, R., Perlman, S., Wong, D., Fogel, B. L., Lynch, D. R., & Chandran, V. (2024). Long non-coding RNA TUG1 is downregulated in Friedreich's ataxia. *Brain Communications*, *6*(3), fcae170. https://doi.org/10.1093/braincomms/fcae170

19. Lowit, A., Greenfield, J., Cutting, E., Wallis, R., & Hadjivassiliou, M. (2021). Symptom burden of people with progressive ataxia, and its wider impact on their friends and relatives : A cross-sectional study. *AMRC Open Research*, *3*, 28. https://doi.org/10.12688/amrcopenres.13036.1

20. Lynch, D. R., Sharma, S., Hearle, P., Greeley, N., Gunther, K., Keita, M., Strawser, C., Hauser, L., Park, C., Schadt, K., & Lin, K. Y. (2024). Characterization of clinical serum cardiac biomarker levels in individuals with Friedreich ataxia. *Journal of the Neurological Sciences*, *461*, 123053. https://doi.org/10.1016/j.jns.2024.123053

21. Mahale, R., Purushottam, M., Singh, R., Yelamanchi, R., Kamble, N., Holla, V., Pal, P. K., Jain, S., & Yadav, R. (2024). Revisiting Friedreich's Ataxia : Phenotypic and Imaging Characteristics. *Annals of Indian Academy of Neurology*, *27*(2), 152-157. https://doi.org/10.4103/aian.aian_1001_23

22. Mantle, D., & Hargreaves, I. P. (2024). Efficacy and Safety of Coenzyme Q10 Supplementation in Neonates, Infants and Children : An Overview. *Antioxidants (Basel, Switzerland)*, *13*(5), 530. https://doi.org/10.3390/antiox13050530

23. Miellet, S., Maddock, M., Napierala, J. S., Napierala, M., & Dottori, M. (2024). Generation of genetically modified Friedreich's ataxia induced pluripotent stem cell lines and isogenic control lines carrying an inducible neurogenin-2 expression cassette. *Stem Cell Research*, *79*, 103477. https://doi.org/10.1016/j.scr.2024.103477

24. Motamed-Gorji, N., Khalil, Y., Gonzalez-Robles, C., Khan, S., Mills, P., Garcia-Moreno, H., Ging, H., Tariq, A., Clayton, P. T., & Giunti, P. (2024). Elevated Bile Acid 3β,5α,6β-Trihydroxycholanoyl Glycine in a Subset of Adult Ataxias Including Niemann-Pick Type C. *Antioxidants (Basel, Switzerland)*, *13*(5), 561. https://doi.org/10.3390/antiox13050561

25. O'Brien, E. M., Neiswinter, N., Lin, K. Y., Lynch, D., Baldwin, K., Profeta, V., Flynn, J. M., & Muhly, W. T. (2024). Perioperative management and outcomes for posterior spinal fusion in patients with Friedreich ataxia : A single-center, retrospective study. *Paediatric Anaesthesia*, *34*(7), 654-661. https://doi.org/10.1111/pan.14896

26. Pasol, J., Uddin, M. S., Tekin, M., & Moore, H. P. (2024). Leber Hereditary Optic Neuropathy in 2 Sisters With Friedreich Ataxia. *Journal of Neuro-Ophthalmology: The Official Journal of the North American Neuro-Ophthalmology Society*. https://doi.org/10.1097/WNO.0000000000002143

27. Peverill, R. E., Lin, K. Y., Fogel, M. A., Cheung, M. M. H., Moir, W. S., Corben, L. A., Cahoon, G., & Delatycki, M. B. (2024). Insights into the effects of Friedreich ataxia on the left ventricle using T1 mapping and late gadolinium enhancement. *PloS One*, *19*(5), e0303969. https://doi.org/10.1371/journal.pone.0303969

28. Rojsajjakul, T., Selvan, N., De, B., Rosenberg, J. B., Kaminsky, S. M., Sondhi, D., Janki, P., Crystal, R. G., Mesaros, C., Khanna, R., & Blair, I. A. (2024). Expression and processing of mature human frataxin after gene therapy in mice. *Scientific Reports*, *14*(1), 8391. https://doi.org/10.1038/s41598-024-59060-0

29. Rudaks, L. I., Yeow, D., Ng, K., Deveson, I. W., Kennerson, M. L., & Kumar, K. R. (2024). An Update on the Adult-Onset Hereditary Cerebellar Ataxias : Novel Genetic Causes and New Diagnostic Approaches. *Cerebellum (London, England)*, *23*(5), 2152-2168. https://doi.org/10.1007/s12311-024-01703-z

30. Rummey, C., Perlman, S., Subramony, S. H., Farmer, J., & Lynch, D. R. (2024). Evaluating mFARS in pediatric Friedreich's ataxia : Insights from the FACHILD study. *Annals of Clinical and Translational Neurology*, *11*(5), 1290-1300. https://doi.org/10.1002/acn3.52057

31. Saini, A. K., Anil, N., Vijay, A. N., Mangla, B., Javed, S., Kumar, P., & Ahsan, W. (2024). Recent Advances in the Treatment Strategies of Friedreich's Ataxia : A Review of Potential Drug Candidates and their Underlying Mechanisms. *Current Pharmaceutical Design*, *30*(19), 1472-1489. https://doi.org/10.2174/0113816128288707240404051856

32. Schulz, V., Steinhilper, R., Oltmanns, J., Freibert, S.-A., Krapoth, N., Linne, U., Welsch, S., Hoock, M. H., Schünemann, V., Murphy, B. J., & Lill, R. (2024). Mechanism and structural dynamics of sulfur transfer during de novo [2Fe-2S] cluster assembly on ISCU2. *Nature Communications*, *15*(1), 3269. https://doi.org/10.1038/s41467-024-47310-8

33. Sciarretta, F., Zaccaria, F., Ninni, A., Ceci, V., Turchi, R., Apolloni, S., Milani, M., Della Valle, I., Tiberi, M., Chiurchiù, V., D'Ambrosi, N., Pedretti, S., Mitro, N., Volontè, C., Amadio, S., Aquilano, K., & Lettieri-Barbato, D. (2024). Frataxin deficiency shifts metabolism to promote reactive microglia via glucose catabolism. *Life Science Alliance*, *7*(7), e202402609. https://doi.org/10.26508/lsa.202402609

34. Seabury, J., Varma, A., Weinstein, J., Rosero, S. J., Engebrecht, C., Khosa, S., Zizzi, C., Wagner, E. S., Alexandrou, D., Cohen, B. L., Dilek, N., Heatwole, J. M., Lynch, D. R., Park, C. C., Wells, M., Subramony, S. H., & Heatwole, C. R. (2024). Friedreich Ataxia Caregiver-Reported Health Index : Development of a Novel, Disease-Specific Caregiver-Reported Outcome Measure. *Neurology. Clinical Practice*, *14*(3), e200303. https://doi.org/10.1212/CPJ.0000000000200300

35. Sureshkumar, S., Bandaranayake, C., Lv, J., Dent, C. I., Bhagat, P. K., Mukherjee, S., Sarwade, R., Atri, C., York, H. M., Tamizhselvan, P., Shamaya, N., Folini, G., Bergey, B. G., Yadav, A. S., Kumar, S., Grummisch, O. S., Saini, P., Yadav, R. K., Arumugam, S., … Balasubramanian, S. (2024). SUMO protease FUG1, histone reader AL3 and chromodomain protein LHP1 are integral to repeat expansion-induced gene silencing in Arabidopsis thaliana. *Nature Plants*, *10*(5), 749-759. https://doi.org/10.1038/s41477-024-01672-5

36. Vancheri, C., Quatrana, A., Morini, E., Mariotti, C., Mongelli, A., Fichera, M., Rufini, A., Condò, I., Testi, R., Novelli, G., Malisan, F., & Amati, F. (2024). An RNA-seq study in Friedreich ataxia patients identified hsa-miR-148a-3p as a putative prognostic biomarker of the disease. *Human Genomics*, *18*(1), 50. https://doi.org/10.1186/s40246-024-00602-y

37. Vogt, L., Quiroz, V., & Ebrahimi-Fakhari, D. (2024). Emerging therapies for childhood-onset movement disorders. *Current Opinion in Pediatrics*, *36*(3), 331-341. https://doi.org/10.1097/MOP.0000000000001354

38. Xia, F., & Su, C. (2024). Advancing Understanding of Predictive Factors for Survival in Friedreich's Ataxia : A Review of Current Evidence and Future Directions. *Movement Disorders: Official Journal of the Movement Disorder Society*, *39*(6), 1081-1082. https://doi.org/10.1002/mds.29869

39. Yetkin, N. A., Yetkin, M. F., Baran Ketencioğlu, B., Oymak, F. S., Gülmez, İ., Yilmaz, İ., & Tutar, N. (2023). Evaluation of diaphragm functions with diaphragm ultrasound and pulmonary function tests in individuals with Friedreich's ataxia. *Turkish Journal of Medical Sciences*, *53*(5), 1301-1311. https://doi.org/10.55730/1300-0144.5696

Atrophie optique autosomique dominante – *ADOA*

40. Borbolis, F., & Palikaras, K. (2024). Identifying therapeutic compounds for autosomal dominant optic atrophy (ADOA) through screening in the nematode C. elegans. *Methods in Cell Biology*, *188*, 89-108. https://doi.org/10.1016/bs.mcb.2024.04.004

41. Calcagni, A., Neveu, M. M., Jurkute, N., & Robson, A. G. (2024). Electrodiagnostic tests of the visual pathway and applications in neuro-ophthalmology. *Eye (London, England)*, *38*(12), 2392-2405. https://doi.org/10.1038/s41433-024-03154-6

42. Gao, Z., Zhu, X., Ren, H., Wang, Y., Hua, C., & Kong, X. (2024). Prenatal exome sequencing for morphologically normal fetus : Should we be doing it? *Prenatal Diagnosis*. https://doi.org/10.1002/pd.6624

43. Lambiri, D. W., & Levin, L. A. (2024). Maculopapillary Bundle Degeneration in Optic Neuropathies. *Current Neurology and Neuroscience Reports*, *24*(7), 203-218. https://doi.org/10.1007/s11910-024-01343-0

44. Lin, Y., Wang, D., Li, B., Wang, J., Xu, L., Sun, X., Ji, K., Yan, C., Liu, F., & Zhao, Y. (2024). Targeting DRP1 with Mdivi-1 to correct mitochondrial abnormalities in ADOA+ syndrome. *JCI Insight*, *9*(15), e180582. https://doi.org/10.1172/jci.insight.180582

45. Lombardo, M., Cusumano, A., Mancino, R., Aiello, F., Sorge, R. P., Nucci, C., & Cesareo, M. (2024). Short Wavelength Automated Perimetry, Standard Automated Perimetry, and Optical Coherence Tomography in Dominant Optic Atrophy. *Journal of Clinical Medicine*, *13*(7), 1971. https://doi.org/10.3390/jcm13071971

46. Martucci, M., Moretton, A., Tarrés-Solé, A., Ropars, V., Lambert, L., Vernet, P., Solà, M., Falkenberg, M., Farge, G., & van den Wildenberg, S. (2024). The mutation R107Q alters mtSSB ssDNA compaction ability and binding dynamics. *Nucleic Acids Research*, *52*(10), 5912-5927. https://doi.org/10.1093/nar/gkae354

47. Wang, E. H.-H., Lin, P.-H., Wu, P.-L., Kang, E. Y.-C., Liu, L., Yeh, L.-K., Chen, K.-J., Hsiao, M.-C., & Wang, N.-K. (2024). Genetic underpinnings explored : OPA1 deletion and complex phenotypes on chromosome 3q29. *BMC Medical Genomics*, *17*(1), 94. https://doi.org/10.1186/s12920-024-01850-6

Encéphalopathie myo-neuro-gastrointestinale - *MNGIE*

48. Du, J., Liu, F., Liu, X., Zhao, D., Wang, D., Sun, H., Yan, C., & Zhao, Y. (2024). Lysosomal dysfunction and overload of nucleosides in thymidine phosphorylase deficiency of MNGIE. *Journal of Translational Medicine*, *22*(1), 449. https://doi.org/10.1186/s12967-024-05275-8

49. Finsterer, J., & Strobl, W. (2024). Gastrointestinal involvement in neuromuscular disorders. *Journal of Gastroenterology and Hepatology*, *39*(10), 1982-1993. https://doi.org/10.1111/jgh.16650

50. Sifeddine, N., Elkhattabi, L., Ait El Cadi, C., Krami, A. M., Mounaji, K., El Khalfi, B., & Barakat, A. (2024). Insights from the SNP analysis of TYMP gene linking MNGIE. *Bioinformation*, *20*(3), 261-270. https://doi.org/10.6026/973206300200261

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

51. Abati, E., Rizzuti, M., Anastasia, A., Comi, G. P., Corti, S., & Rizzo, F. (2024). Charcot-Marie-Tooth type 2A in vivo models : Current updates. *Journal of Cellular and Molecular Medicine*, *28*(9), e18293. https://doi.org/10.1111/jcmm.18293

52. Ceprian, M., Juntas-Morales, R., Campbell, G., Walther-Louvier, U., Rivier, F., Camu, W., Esselin, F., Echaniz-Laguna, A., Stojkovic, T., Bouhour, F., Latour, P., & Tricaud, N. (2024). The Hexokinase 1 5'-UTR Mutation in Charcot-Marie-Tooth 4G Disease Alters Hexokinase 1 Binding to Voltage-Dependent Anion Channel-1 and Leads to Dysfunctional Mitochondrial Calcium Buffering. *International Journal of Molecular Sciences*, *25*(8), 4364. https://doi.org/10.3390/ijms25084364

53. Kumar, A., Larrea, D., Pero, M. E., Infante, P., Conenna, M., Shin, G. J., Van Elias, V., Grueber, W. B., Di Marcotullio, L., Area-Gomez, E., & Bartolini, F. (2024). MFN2 coordinates mitochondria motility with α-tubulin acetylation and this regulation is disrupted in CMT2A. *iScience*, *27*(6), 109994. https://doi.org/10.1016/j.isci.2024.109994

54. Li, Z., Zeng, S., Xie, Y., Li, X., Huang, S., Zhao, H., Cao, W., Liu, L., Wang, M., Gong, Q., Liu, J., Rong, P., & Zhang, R. (2024). Genetic and clinical profile of 15 Chinese families with GDAP1-related Charcot-Marie-Tooth disease and identification of H256R as a frequent mutation. *Journal of the Peripheral Nervous System: JPNS*, *29*(2), 232-242. https://doi.org/10.1111/jns.12628

55. Song, B., Cho, H., Yun, J., & Moon, I. J. (2024). Cochlear implantation in patients with Charcot-Marie-Tooth disease : Two cases with a review of the literature. *European Archives of Oto-Rhino-Laryngology: Official Journal of the European Federation of Oto-Rhino-Laryngological Societies (EUFOS): Affiliated with the German Society for Oto-Rhino-Laryngology - Head and Neck Surgery*, *281*(7), 3845-3851. https://doi.org/10.1007/s00405-024-08592-2

56. Subramanian, B., Williams, S., Karp, S., Hennino, M.-F., Jacas, S., Lee, M., Riella, C. V., Alper, S. L., Higgs, H. N., & Pollak, M. R. (2024). Missense Mutant Gain-of-Function Causes Inverted Formin 2 (INF2)-Related Focal Segmental Glomerulosclerosis (FSGS). *bioRxiv: The Preprint Server for Biology*, 2024.06.08.598088. https://doi.org/10.1101/2024.06.08.598088

57. Trajano, L. A. da S. N., Siqueira, P. B., Rodrigues, M. M. de S., Pires, B. R. B., da Fonseca, A. de S., & Mencalha, A. L. (2024). Does photobiomodulation alter mitochondrial dynamics? *Photochemistry and Photobiology*. https://doi.org/10.1111/php.13963

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

58. Penkl, M., Mayr, J. A., Feichtinger, R. G., Reilmann, R., Debus, O., Fobker, M., Penkl, A., Reunert, J., Rust, S., & Marquardt, T. (2024). Anaplerotic Therapy Using Triheptanoin in Two Brothers Suffering from Aconitase 2 Deficiency. *Metabolites*, *14*(4), 238. https://doi.org/10.3390/metabo14040238

Maladie liée au gène GFM1 – GFM1-related disorder

59. Cilleros-Holgado, P., Gómez-Fernández, D., Piñero-Pérez, R., Romero Domínguez, J. M., Talaverón-Rey, M., Reche-López, D., Suárez-Rivero, J. M., Álvarez-Córdoba, M., Romero-González, A., López-Cabrera, A., Oliveira, M. C. D., Rodríguez-Sacristan, A., & Sánchez-Alcázar, J. A. (2024). Polydatin and Nicotinamide Rescue the Cellular Phenotype of Mitochondrial Diseases by Mitochondrial Unfolded Protein Response (mtUPR) Activation. *Biomolecules*, *14*(5), 598. https://doi.org/10.3390/biom14050598

60. Key, J., Gispert, S., & Auburger, G. (2024). Knockout Mouse Studies Show That Mitochondrial CLPP Peptidase and CLPX Unfoldase Act in Matrix Condensates near IMM, as Fast Stress Response in Protein Assemblies for Transcript Processing, Translation, and Heme Production. *Genes*, *15*(6), 694. https://doi.org/10.3390/genes15060694

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

61. Ali, M. J. (2024). Alterations of lacrimal sac vasculature in lacrimal disorders : Chromo endoscopic assessment with changes in effective spectral response. *Journal Francais D'ophtalmologie*, *47*(5), 104133. https://doi.org/10.1016/j.jfo.2024.104133

62. Brañas Casas, R., Zuppardo, A., Risato, G., Dinarello, A., Celeghin, R., Fontana, C., Grelloni, E., Gilea, A. I., Viscomi, C., Rasola, A., Dalla Valle, L., Lodi, T., Baruffini, E., Facchinello, N., Argenton, F., & Tiso, N. (2024). Zebrafish polg2 knock-out recapitulates human POLG-disorders; implications for drug treatment. *Cell Death & Disease*, *15*(4), 281. https://doi.org/10.1038/s41419-024-06622-9

63. Cakmak, C., & Zempel, H. (2021). A perspective on human cell models for POLG-spectrum disorders : Advantages and disadvantages of CRISPR-Cas-based vs. patient-derived iPSC models. *Medizinische Genetik: Mitteilungsblatt Des Berufsverbandes Medizinische Genetik e.V*, *33*(3), 245-249. https://doi.org/10.1515/medgen-2021-2090

64. Guan, Q., Zhang, Y., Wang, Z.-K., Liu, X.-H., Zou, J., & Zhang, L.-L. (2024). Skeletal phenotypes and molecular mechanisms in aging mice. *Zoological Research*, *45*(4), 724-746. https://doi.org/10.24272/j.issn.2095-8137.2023.397

65. Hikmat, O., Naess, K., Engvall, M., Klingenberg, C., Rasmussen, M., Brodtkorb, E., Ostergaard, E., de Coo, I., Pias-Peleteiro, L., Isohanni, P., Uusimaa, J., Majamaa, K., Kärppä, M., Ortigoza-Escobar, J. D., Tangeraas, T., Berland, S., Harrison, E., Biggs, H., Horvath, R., … Bindoff, L. A. (2024). Status epilepticus in POLG disease : A large multinational study. *Journal of Neurology*, *271*(8), 5156-5164. https://doi.org/10.1007/s00415-024-12463-5

66. Illés, A., Pikó, H., Árvai, K., Donka, V., Szepesi, O., Kósa, J., Lakatos, P., & Beke, A. (2024). Screening of premature ovarian insufficiency associated genes in Hungarian patients with next generation sequencing. *BMC Medical Genomics*, *17*(1), 98. https://doi.org/10.1186/s12920-024-01873-z

67. Kristensen, E., Mathisen, L., Berland, S., Klingenberg, C., Brodtkorb, E., Rasmussen, M., Tangeraas, T., Bliksrud, Y. T., Rahman, S., Bindoff, L. A., & Hikmat, O. (2024). Epidemiology and natural history of POLG disease in Norway : A nationwide cohort study. *Annals of Clinical and Translational Neurology*, *11*(7), 1819-1830. https://doi.org/10.1002/acn3.52088

68. Liang, K. X., Chen, A., Kianian, A., Kristiansen, C. K., Yangzom, T., Furriol, J., Høyland, L. E., Ziegler, M., Kråkenes, T., Tzoulis, C., Fang, E. F., Sullivan, G. J., & Bindoff, L. A. (2024). Activation of Neurotoxic Astrocytes Due to Mitochondrial Dysfunction Triggered by POLG Mutation. *International Journal of Biological Sciences*, *20*(8), 2860-2880. https://doi.org/10.7150/ijbs.93445

69. Lopriore, P., Palermo, G., Meli, A., Bellini, G., Benevento, E., Montano, V., Siciliano, G., Mancuso, M., & Ceravolo, R. (2024). Mitochondrial Parkinsonism : A Practical Guide to Genes and Clinical Diagnosis. *Movement Disorders Clinical Practice*, *11*(8), 948-965. https://doi.org/10.1002/mdc3.14148

70. Mancuso, M., Papadopoulou, M. T., Ng, Y. S., Ardissone, A., Bellusci, M., Bertini, E., Di Vito, L., Evangelista, T., Fons, C., Hikmat, O., Horvath, R., Klopstock, T., Kornblum, C., Lamperti, C., Licchetta, L., Molnar, M. J., Varhaug, K. N., O'Callaghan, M., Pressler, R. M., … Rahman, S. (2024). Management of seizures in patients with primary mitochondrial diseases : Consensus statement from the InterERNs Mitochondrial Working Group. *European Journal of Neurology*, *31*(7), e16275. https://doi.org/10.1111/ene.16275

71. Ramesh, R., Amanmahanya, C., Krishnamoorthy, V., Krishnan, V., Palani, S., & Narasimhan Ranganathan, L. (2024). Oculomasticatory rhythmic movements, insomnia and stroke-like episodes in a patient with POLG mutation. *BMJ Case Reports*, *17*(4), e259426. https://doi.org/10.1136/bcr-2023-259426

72. Ratnaike, T. E., Elkhateeb, N., Lochmüller, A., Gilmartin, C., Schon, K., Horváth, R., & Chinnery, P. F. (2024). Evidence for sodium valproate toxicity in mitochondrial diseases : A systematic analysis. *BMJ Neurology Open*, *6*(1), e000650. https://doi.org/10.1136/bmjno-2024-000650

73. Riccio, A. A., Brannon, A. J., Krahn, J. M., Bouvette, J., Williams, J. G., Borgnia, M. J., & Copeland, W. C. (2024). Coordinated DNA polymerization by Polγ and the region of LonP1 regulated proteolysis. *Nucleic Acids Research*, *52*(13), 7863-7875. https://doi.org/10.1093/nar/gkae539

74. Trease, A. J., Totusek, S., Lichter, E. Z., Stauch, K. L., & Fox, H. S. (2024). Mitochondrial DNA Instability Supersedes Parkin Mutations in Driving Mitochondrial Proteomic Alterations and Functional Deficits in Polg Mutator Mice. *International Journal of Molecular Sciences*, *25*(12), 6441. https://doi.org/10.3390/ijms25126441

75. VanPortfliet, J. J., Lei, Y., Martinez, C. G., Wong, J., Pflug, K., Sitcheran, R., Kneeland, S. C., Murray, S. A., McGuire, P. J., Cannon, C. L., & West, A. P. (2024). Caspase-11 drives macrophage hyperinflammation in models of Polg-related mitochondrial disease. *bioRxiv: The Preprint Server for Biology*, 2024.05.11.593693. https://doi.org/10.1101/2024.05.11.593693

76. Wang, C., Li, M., Liu, Z., Guo, Y., Liu, H., Zhao, P., & Acute Liver Failure Study Team. (2024). Genetic evaluation in indeterminate acute liver failure : A post hoc analysis. *Arab Journal of Gastroenterology: The Official Publication of the Pan-Arab Association of Gastroenterology*, *25*(2), 125-128. https://doi.org/10.1016/j.ajg.2024.03.004

MELAS

77. Argov, Z. (2024). Statins in hereditary myopathies : To give or not to give. *Neuromuscular Disorders: NMD*, *41*, 35-39. https://doi.org/10.1016/j.nmd.2024.06.004

78. Barros, C. D. S., Coutinho, A., & Tengan, C. H. (2024). Arginine Supplementation in MELAS Syndrome : What Do We Know about the Mechanisms? *International Journal of Molecular Sciences*, *25*(7), 3629. https://doi.org/10.3390/ijms25073629

79. Cai, H., Li, L.-M., Zhang, M., Zhou, Y., & Li, P. (2024). Case report : Late-onset MELAS syndrome with mtDNA 5783G>A mutation diagnosed by urinary sediment genetic testing. *Frontiers in Genetics*, *15*, 1367716. https://doi.org/10.3389/fgene.2024.1367716

80. Du, J., Liu, F., Liu, X., Zhao, D., Wang, D., Sun, H., Yan, C., & Zhao, Y. (2024). Lysosomal dysfunction and overload of nucleosides in thymidine phosphorylase deficiency of MNGIE. *Journal of Translational Medicine*, *22*(1), 449. https://doi.org/10.1186/s12967-024-05275-8

81. El Ouali, I., Naggar, A., Berrada, K., Jiddane, M., & Touarsa, F. (2024). A burning encephalitis : Fluid-attenuated inversion recovery-hyperintense lesions in Anti-myelin oligodendrocyte glycoprotein-associated encephalitis with seizures in anti-myelin oligodendrocyte glycoprotein-associated encephalitis with seizures-A case report and review of the literature. *SAGE Open Medical Case Reports*, *12*, 2050313X241261021. https://doi.org/10.1177/2050313X241261021

82. Finsterer, J. (2024a). Before Attributing Malnutrition in Melas to Superior Mesenterica Artery Syndrome, all Differentials must be Excluded. *European Journal of Case Reports in Internal Medicine*, *11*(4), 004388. https://doi.org/10.12890/2024_004388

83. Finsterer, J. (2024b). Diagnostic accuracy is required when analysing cohorts with mitochondrial disorders. *European Journal of Paediatric Neurology: EJPN: Official Journal of the European Paediatric Neurology Society*, *51*, 71-72. https://doi.org/10.1016/j.ejpn.2024.05.017

84. Finsterer, J., & Strobl, W. (2024). Gastrointestinal involvement in neuromuscular disorders. *Journal of Gastroenterology and Hepatology*, *39*(10), 1982-1993. https://doi.org/10.1111/jgh.16650

85. Huang, Q., Trumpff, C., Monzel, A. S., Rausser, S., Haahr, R., Devine, J., Liu, C. C., Kelly, C., Thompson, E., Kurade, M., Michelson, J., Shaulson, E. D., Li, S., Engelstad, K., Tanji, K., Lauriola, V., Wang, T., Wang, S., Zuraikat, F. M., … Picard, M. (2024). Psychobiological regulation of plasma and saliva GDF15 dynamics in health and mitochondrial diseases. *bioRxiv: The Preprint Server for Biology*, 2024.04.19.590241. https://doi.org/10.1101/2024.04.19.590241

86. Khanna, S., & Smith, B. T. (2024). Neovascular Glaucoma in MELAS syndrome. *American Journal of Ophthalmology Case Reports*, *34*, 102064. https://doi.org/10.1016/j.ajoc.2024.102064

87. Liufu, T., Zhao, X., Yu, M., Xie, Z., Meng, L., Lv, H., Zhang, W., Yuan, Y., Xing, G., Deng, J., & Wang, Z. (2024). Multiomics analysis reveals serine catabolism as a potential therapeutic target for MELAS. *FASEB Journal: Official Publication of the Federation of American Societies for Experimental Biology*, *38*(12), e23742. https://doi.org/10.1096/fj.202302286RRR

88. Momoh, R., & Kollamparambil, S. (2024). A Case Report of a Clinically Suspected Diagnosis of Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-Like Episodes (MELAS) Syndrome With Cardiac Impairment. *Cureus*, *16*(3), e56980. https://doi.org/10.7759/cureus.56980

89. Namani, S., Kavetsky, K., Lin, C.-Y., Maharjan, S., Gamper, H. B., Li, N.-S., Piccirilli, J. A., Hou, Y.-M., & Drndic, M. (2024). Unraveling RNA Conformation Dynamics in Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-like Episode Syndrome with Solid-State Nanopores. *ACS Nano*, *18*(26), 17240-17250. https://doi.org/10.1021/acsnano.4c04625

MERFF

90. Barbour, K., Tian, N., Yozawitz, E. G., Wolf, S., McGoldrick, P. E., Sands, T. T., Nelson, A., Basma, N., & Grinspan, Z. M. (2024). Population-based study of rare epilepsy incidence in a US urban population. *Epilepsia*, *65*(8), 2341-2353. https://doi.org/10.1111/epi.18029

91. Dupuis, H., Lemaitre, M., Jannin, A., Douillard, C., Espiard, S., & Vantyghem, M.-C. (2024). Lipomatoses. *Annales D'endocrinologie*, *85*(3), 231-247. https://doi.org/10.1016/j.ando.2024.05.003

92. Zhang, J., Zhou, J., Ji, C., Wu, D., & Wang, K. (2024). [Progressive myoclonic epilepsy : A retrospective study of newly-diagnosed adult patients from a single center]. *Zhonghua Yi Xue Yi Chuan Xue Za Zhi = Zhonghua Yixue Yichuanxue Zazhi = Chinese Journal of Medical Genetics*, *41*(4), 432-436. https://doi.org/10.3760/cma.j.cn511374-20230214-00073

Neuropathie optique héréditaire de Leber - *LHON*

93. Battista, M., Carelli, V., Bottazzi, L., Bandello, F., Cascavilla, M. L., & Barboni, P. (2024). Gene therapy for Leber hereditary optic neuropathy. *Expert Opinion on Biological Therapy*, *24*(6), 521-528. https://doi.org/10.1080/14712598.2024.2359015

94. Bocca, C., Kouassi-Nzoughet, J., Chao de la Barca, J. M., Bonneau, D., Verny, C., Gohier, P., Orssaud, C., & Reynier, P. (2024). Letter to the editor on : Prophylactic nicotinamide treatment protects from rotenone-induced neurodegeneration by increasing mitochondrial content and volume. *Acta Neuropathologica Communications*, *12*(1), 53. https://doi.org/10.1186/s40478-024-01768-1

95. Calcagni, A., Neveu, M. M., Jurkute, N., & Robson, A. G. (2024). Electrodiagnostic tests of the visual pathway and applications in neuro-ophthalmology. *Eye (London, England)*, *38*(12), 2392-2405. https://doi.org/10.1038/s41433-024-03154-6

96. de Muijnck, C., van Schooneveld, M. J., Plomp, A. S., Rodenburg, R. J., van Genderen, M. M., & Boon, C. J. F. (2024). Leber's hereditary optic neuropathy like disease in MT-ATP6 variant m.8969G>A. *American Journal of Ophthalmology Case Reports*, *34*, 102070. https://doi.org/10.1016/j.ajoc.2024.102070

97. Emperador, S., Habbane, M., López-Gallardo, E., Del Rio, A., Llobet, L., Mateo, J., Sanz-López, A. M., Fernández-García, M. J., Sánchez-Tocino, H., Benbunan-Ferreiro, S., Calabuig-Goena, M., Narvaez-Palazón, C., Fernández-Vega, B., González-Iglesias, H., Urreizti, R., Artuch, R., Pacheu-Grau, D., Bayona-Bafaluy, P., Montoya, J., & Ruiz-Pesini, E. (2024). Identification and characterization of a new pathologic mutation in a large Leber hereditary optic neuropathy pedigree. *Orphanet Journal of Rare Diseases*, *19*(1), 148. https://doi.org/10.1186/s13023-024-03165-2

98. Esteban-Vasallo, M. D., Domínguez-Berjón, M. F., Chalco-Orrego, J. P., & González Martín-Moro, J. (2024). Prevalence of Leber hereditary optic neuropathy in the Community of Madrid (Spain), estimation with a capture-recapture method. *Orphanet Journal of Rare Diseases*, *19*(1), 220. https://doi.org/10.1186/s13023-024-03225-7

99. Giannoccaro, M. P., Morelli, L., Ricciardiello, F., Donadio, V., Bartiromo, F., Tonon, C., Carbonelli, M., Amore, G., Carelli, V., Liguori, R., & La Morgia, C. (2024). Co-occurrence of glial fibrillary acidic protein astrocytopathy in a patient with Leber's hereditary optic neuropathy due to DNAJC30 mutations. *European Journal of Neurology*, *31*(9), e16344. https://doi.org/10.1111/ene.16344

100. Handzic, A., Donaldson, L., & Margolin, E. (2024). Preserved Ganglion Cell Analysis in a Case of Longstanding Leber Hereditary Optic Neuropathy : A Sign of Hibernating Neurons? *Journal of Neuro-Ophthalmology: The Official Journal of the North American Neuro-Ophthalmology Society*. https://doi.org/10.1097/WNO.0000000000002165

101. Iorga, R. E., Munteanu-Dănulescu, R. S., & Danielescu, C. (2024). A challenging differential diagnosis—Leber's Hereditary Optic Neuropathy. *Romanian Journal of Ophthalmology*, *68*(1), 65-71. https://doi.org/10.22336/rjo.2024.13

102. Joo, H. J., Moon, Y., & Jung, J. H. (2024). Variability of relationship between inner-retinal structural changes and visual dysfunction in optic neuropathy. *Scientific Reports*, *14*(1), 12069. https://doi.org/10.1038/s41598-024-62704-w

103. Kaur, G., Ganev, Y., Rodriguez, W., Tseng, S., Orozco, L., & Chand, P. (2024). Deep Brain Stimulation for Medication Refractory Tremor in Leber Optic Neuropathy Plus Syndrome. *Cureus*, *16*(4), e58255. https://doi.org/10.7759/cureus.58255

104. Lambiri, D. W., & Levin, L. A. (2024). Maculopapillary Bundle Degeneration in Optic Neuropathies. *Current Neurology and Neuroscience Reports*, *24*(7), 203-218. https://doi.org/10.1007/s11910-024-01343-0

105. Larin, I. I., Shatalova, R. O., Laktyushkin, V. S., Rybtsov, S. A., Lapshin, E. V., Shevyrev, D. V., Karabelsky, A. V., Moskalets, A. P., Klinov, D. V., & Ivanov, D. A. (2024). Deep Learning for Cell Migration in Nonwoven Materials and Evaluating Gene Transfer Effects following AAV6-ND4 Transduction. *Polymers*, *16*(9), 1187. https://doi.org/10.3390/polym16091187

106. Lin, X., Zhou, Y., & Xue, L. (2024). Mitochondrial complex I subunit MT-ND1 mutations affect disease progression. *Heliyon*, *10*(7), e28808. https://doi.org/10.1016/j.heliyon.2024.e28808

107. McAnnis, K. E., Ruiz-Montenegro, A., Davila, P. A., Laylani, N. A., & Lee, A. G. (2024). Leber Hereditary Optic Neuropathy Plus Phenotype With Double Point Mutations (m.11778 G>A and m.14484T>C) and Concurrent Myelin Oligodendrocyte Glycoprotein Antibody-Associated Disease (MOGAD). *Journal of Neuro-Ophthalmology: The Official Journal of the North American Neuro-Ophthalmology Society*. https://doi.org/10.1097/WNO.0000000000002153

108. Nagy, M. A., Cunnane, M. E., Juliano, A. F., Wiggs, J. L., Caruso, P. A., & Gaier, E. D. (2024). Optic Nerve T2 Signal Intensity and Caliber Reflect Clinical Severity in Genetic Optic Atrophy. *Journal of Neuro-Ophthalmology: The Official Journal of the North American Neuro-Ophthalmology Society*. https://doi.org/10.1097/WNO.0000000000002191

109. Pandya, B. U., Takyi, N. A., Vosoughi, A. R., Margolin, E. A., & Micieli, J. A. (2024). Novel Mutations in the ND5 Gene Associated With Leber Hereditary Optic Neuropathy. *Journal of Neuro-Ophthalmology: The Official Journal of the North American Neuro-Ophthalmology Society*, *44*(2), e227-e229. https://doi.org/10.1097/WNO.0000000000001796

110. Pasol, J., Uddin, M. S., Tekin, M., & Moore, H. P. (2024). Leber Hereditary Optic Neuropathy in 2 Sisters With Friedreich Ataxia. *Journal of Neuro-Ophthalmology: The Official Journal of the North American Neuro-Ophthalmology Society*. https://doi.org/10.1097/WNO.0000000000002143

111. Pasqualotto, B. A., Nelson, A., Deheshi, S., Sheldon, C. A., Vogl, A. W., & Rintoul, G. L. (2024). Impaired mitochondrial morphological plasticity and failure of mitophagy associated with the G11778A mutation of LHON. *Biochemical and Biophysical Research Communications*, *721*, 150119. https://doi.org/10.1016/j.bbrc.2024.150119

112. Pasqualotto, B. A., Tegeman, C., Frame, A. K., McPhedrain, R., Halangoda, K., Sheldon, C. A., & Rintoul, G. L. (2024). Galactose-replacement unmasks the biochemical consequences of the G11778A mitochondrial DNA mutation of LHON in patient-derived fibroblasts. *Experimental Cell Research*, *439*(1), 114075. https://doi.org/10.1016/j.yexcr.2024.114075

113. Rajagopalan, A., Jeste, S., Borchert, M. S., & Chang, M. Y. (2024). Autosomal Recessive Leber Hereditary Optic Neuropathy Triggered by Superior Mesenteric Artery Syndrome. *Journal of Neuro-Ophthalmology: The Official Journal of the North American Neuro-Ophthalmology Society*. https://doi.org/10.1097/WNO.0000000000002142

114. Rigobello, L., Lugli, F., Caporali, L., Bartocci, A., Fadanni, J., Zerbetto, F., Iommarini, L., Carelli, V., Ghelli, A. M., & Musiani, F. (2024). A computational study to assess the pathogenicity of single or combinations of missense variants on respiratory complex I. *International Journal of Biological Macromolecules*, *273*(Pt 2), 133086. https://doi.org/10.1016/j.ijbiomac.2024.133086

115. Roomets, E., & Mauring, L. (2024). Autosomal recessive leber hereditary optic neuropathy in a choroideremia carrier. A case report. *European Journal of Ophthalmology*, *34*(5), NP1-NP7. https://doi.org/10.1177/11206721241254408

116. Takai, Y., Yamagami, A., & Ishikawa, H. (2024). [Leber's hereditary optic neuropathy]. *Rinsho Shinkeigaku = Clinical Neurology*, *64*(5), 326-332. https://doi.org/10.5692/clinicalneurol.cn-001924

117. Xiao, D., Lhamo, T., Meng, Y., Xu, Y., & Chen, C. (2024). Peripapillary hyperreflective ovoid mass-like structures : Multimodal imaging and associated diseases. *Frontiers in Neurology*, *15*, 1379801. https://doi.org/10.3389/fneur.2024.1379801

118. Xie, Q., Wu, H., Long, H., Xiao, C., Qiu, J., Yu, W., Jiang, X., Liu, J., Zhang, S., Lyu, Q., Suo, L., & Kuang, Y. (2024). Secondary follicles enable efficient germline mtDNA base editing at hard-to-edit site. *Molecular Therapy. Nucleic Acids*, *35*(2), 102170. https://doi.org/10.1016/j.omtn.2024.102170

119. Yang, H. K., Seong, M.-W., Kim, J. Y., Park, S. S., & Hwang, J.-M. (2024). Poor visual prognosis of Asian patients with 3460 mitochondrial DNA mutation in Leber's hereditary optic neuropathy. *Canadian Journal of Ophthalmology. Journal Canadien D'ophtalmologie*, S0008-4182(24)00095-4. https://doi.org/10.1016/j.jcjo.2024.03.013

120. Yasin, A. L., Mendoza-Santiesteban, C. E., Khodeiry, M. M., & Lam, B. L. (2024). Atypical Late-Onset Leber Hereditary Optic Neuropathy (LHON) Associated With T14484C Mutation. *Journal of Neuro-Ophthalmology: The Official Journal of the North American Neuro-Ophthalmology Society*, *44*(2), e224-e226. https://doi.org/10.1097/WNO.0000000000001835

Ophtalmoplégie externe progressive – *PEO*

121. Dombi, E., Marinaki, T., Spingardi, P., Millar, V., Hadjichristou, N., Carver, J., Johnston, I. G., Fratter, C., & Poulton, J. (2024). Nucleoside supplements as treatments for mitochondrial DNA depletion syndrome. *Frontiers in Cell and Developmental Biology*, *12*, 1260496. https://doi.org/10.3389/fcell.2024.1260496

122. Moustaine, M. O., Azemour, Z., Mohammed, F., Benlanda, O., Nassik, H., & Karkouri, M. (2024). Management of Ptosis in Kearns-Sayre Syndrome : A Case Report and Literature Review. *Archives of Plastic Surgery*, *51*(2), 182-186. https://doi.org/10.1055/a-2207-7587

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

123. Barbour, K., Tian, N., Yozawitz, E. G., Wolf, S., McGoldrick, P. E., Sands, T. T., Nelson, A., Basma, N., & Grinspan, Z. M. (2024). Population-based study of rare epilepsy incidence in a US urban population. *Epilepsia*, *65*(8), 2341-2353. https://doi.org/10.1111/epi.18029

124. Dombi, E., Marinaki, T., Spingardi, P., Millar, V., Hadjichristou, N., Carver, J., Johnston, I. G., Fratter, C., & Poulton, J. (2024). Nucleoside supplements as treatments for mitochondrial DNA depletion syndrome. *Frontiers in Cell and Developmental Biology*, *12*, 1260496. https://doi.org/10.3389/fcell.2024.1260496

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

125. Moustaine, M. O., Azemour, Z., Mohammed, F., Benlanda, O., Nassik, H., & Karkouri, M. (2024). Management of Ptosis in Kearns-Sayre Syndrome : A Case Report and Literature Review. *Archives of Plastic Surgery*, *51*(2), 182-186. https://doi.org/10.1055/a-2207-7587

Syndrome de Leigh - *Leigh syndrome*

126. Alfaraidi, A. T., ALSulimani, N. K., & Garout, W. (2024). Incidental Finding of MEGDEL Syndrome at a Tertiary Care Center in Saudi Arabia. *Cureus*, *16*(3), e55308. https://doi.org/10.7759/cureus.55308

127. Amar, Z., Hussaini, H., Kachhadia, M. P., Samreen, I., Mohamed, A. S., & Nasir, H. (2024). Late-Onset Leigh Syndrome With Protracted Gastrointestinal Manifestations : A Rare Case Report. *Cureus*, *16*(5), e59669. https://doi.org/10.7759/cureus.59669

128. Ball, M., Thorburn, D. R., & Rahman, S. (1993). Mitochondrial DNA-Associated Leigh Syndrome Spectrum. 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/NBK1173/

129. Bernhardt, I., Frajman, L. E., Ryder, B., Andersen, E., Wilson, C., McKeown, C., Anderson, T., Coman, D., Vincent, A. L., Buchanan, C., Roxburgh, R., Pitt, J., De Hora, M., Christodoulou, J., Thorburn, D. R., Wilson, F., Drake, K. M., Leask, M., Yardley, A.-M., … Glamuzina, E. (2024). Further delineation of short-chain enoyl-CoA hydratase deficiency in the Pacific population. *Molecular Genetics and Metabolism*, *142*(3), 108508. https://doi.org/10.1016/j.ymgme.2024.108508

130. Galosi, S., Mancini, C., Commone, A., Calligari, P., Caputo, V., Nardecchia, F., Carducci, C., van den Heuvel, L. P., Pizzi, S., Bruselles, A., Niceta, M., Martinelli, S., Rodenburg, R. J., Tartaglia, M., & Leuzzi, V. (2024). Biallelic Variants of MRPS36 Cause a New Form of Leigh Syndrome. *Movement Disorders: Official Journal of the Movement Disorder Society*, *39*(7), 1225-1231. https://doi.org/10.1002/mds.29795

131. Gélinas, R., Lévesque, C., Thompson Legault, J., Rivard, M.-E., Villeneuve, L., Laprise, C., & Rioux, J. D. (2024). Human induced pluripotent stem cells (hiPSCs) derived cells reflect tissue specificity found in patients with Leigh syndrome French Canadian variant (LSFC). *Frontiers in Genetics*, *15*, 1375467. https://doi.org/10.3389/fgene.2024.1375467

132. Hanaford, A. R., Khanna, A., James, K., Truong, V., Liao, R., Chen, Y., Mulholland, M., Kayser, E.-B., Watanabe, K., Hsieh, E. S., Sedensky, M., Morgan, P. G., Kalia, V., Sarkar, S., & Johnson, S. C. (2024). Interferon-gamma contributes to disease progression in the Ndufs4(-/-) model of Leigh syndrome. *Neuropathology and Applied Neurobiology*, *50*(3), e12977. https://doi.org/10.1111/nan.12977

133. Kanemaru, E., Shimoda, K., Marutani, E., Morita, M., Miranda, M., Miyazaki, Y., Sinow, C., Sharma, R., Dong, F., Bloch, D. B., Akaike, T., & Ichinose, F. (2024). Exclusion of sulfide:quinone oxidoreductase from mitochondria causes Leigh-like disease in mice by impairing sulfide metabolism. *The Journal of Clinical Investigation*, *134*(15), e170994. https://doi.org/10.1172/JCI170994

134. Kayser, E.-B., Chen, Y., Mulholland, M., Truong, V., James, K., Hanaford, A., & Johnson, S. (2024). Evaluating the efficacy of vatiquinone in preclinical models of mitochondrial disease. *Research Square*, rs.3.rs-4202689. https://doi.org/10.21203/rs.3.rs-4202689/v1

135. Kistol, D., Tsygankova, P., Bostanova, F., Orlova, M., & Zakharova, E. (2024). New Case of Spinocerebellar Ataxia, Autosomal Recessive 4, Due to VPS13D Variants. *International Journal of Molecular Sciences*, *25*(10), 5127. https://doi.org/10.3390/ijms25105127

136. Kobayashi, M., Miyauchi, A., Jimbo, E. F., Oishi, N., Aoki, S., Watanabe, M., Yoshikawa, Y., Akiyama, Y., Yamagata, T., & Osaka, H. (2024). Synthetic aporphine alkaloids are potential therapeutics for Leigh syndrome. *Scientific Reports*, *14*(1), 11561. https://doi.org/10.1038/s41598-024-62445-w

137. Kowalska, A., Figura, M., Zawadka, M., & Koziorowski, D. (2024). Pyruvate dehydrogenase-E1α deficiency presenting as generalized dystonia : A genetic diagnosis with important clinical implications. *Clinical Neurology and Neurosurgery*, *241*, 108307. https://doi.org/10.1016/j.clineuro.2024.108307

138. Mahesan, A., Choudhary, P. K., Kamila, G., Rohil, A., Meena, A. K., Kumar, A., Jauhari, P., Chakrabarty, B., & Gulati, S. (2024). NDUFV1-Related Mitochondrial Complex-1 Disorders : A Retrospective Case Series and Literature Review. *Pediatric Neurology*, *155*, 91-103. https://doi.org/10.1016/j.pediatrneurol.2024.02.012

139. Meldau, S., Ackermann, S., Riordan, G., van der Watt, G. F., Spencer, C., Raga, S., Khan, K., Blackhurst, D. M., & van der Westhuizen, F. H. (2024). A novel mitochondrial DNA variant in MT-ND6 : M.14430A>C p.(Trp82Gly) identified in a patient with Leigh syndrome and complex I deficiency. *Molecular Genetics and Metabolism Reports*, *39*, 101078. https://doi.org/10.1016/j.ymgmr.2024.101078

140. Misceo, D., Strømme, P., Bitarafan, F., Chawla, M. S., Sheng, Y., Bach de Courtade, S. M., Eide, L., & Frengen, E. (2024). Biallelic NDUFA4 Deletion Causes Mitochondrial Complex IV Deficiency in a Patient with Leigh Syndrome. *Genes*, *15*(4), 500. https://doi.org/10.3390/genes15040500

141. Moreno-Loshuertos, R., & Fernández-Silva, P. (2024). Isolation of Mitochondria for Mitochondrial Supercomplex Analysis from Small Tissue and Cell Culture Samples. *Journal of Visualized Experiments: JoVE*, *207*. https://doi.org/10.3791/66771

142. Reynaud-Dulaurier, R., Clément, R., Yjjou, S., Cresson, C., Saoudi, Y., Faideau, M., & Decressac, M. (2024). The Blood-Brain Barrier Is Unaffected in the Ndufs4-/- Mouse Model of Leigh Syndrome. *International Journal of Molecular Sciences*, *25*(9), 4828. https://doi.org/10.3390/ijms25094828

143. Sasaki, M., Okanishi, T., Matsuoka, T., Yoshimura, A., Maruyama, S., Shiohama, T., Hoshino, H., Mori, T., Majima, H., Matsumoto, H., Kobayashi, S., Chiyonobu, T., Matsushige, T., Nakamura, K., Kubota, K., Tanaka, R., Fujita, T., Enoki, H., Suzuki, Y., … Maegaki, Y. (2024). Infantile Epileptic Spasms Syndrome Complicated by Leigh Syndrome and Leigh-Like Syndrome : A Retrospective, Nationwide, Multicenter Case Series. *Pediatric Neurology*, *157*, 29-38. https://doi.org/10.1016/j.pediatrneurol.2024.05.007

144. Tian, D., Cui, M., & Han, M. (2024). Bacterial muropeptides promote OXPHOS and suppress mitochondrial stress in mammals. *Cell Reports*, *43*(4), 114067. https://doi.org/10.1016/j.celrep.2024.114067

145. Wortmann, S. B., Feichtinger, R. G., Abela, L., van Gemert, L. A., Aubart, M., Dufeu-Berat, C.-M., Boddaert, N., de Coo, R., Stühn, L., Hebbink, J., Heinritz, W., Hildebrandt, J., Himmelreich, N., Korenke, C., Lehman, A., Leyland, T., Makowski, C., Martinez Marin, R. J., Marzin, P., … Willemsen, M. A. (2024). Clinical, Neuroimaging, and Metabolic Footprint of the Neurodevelopmental Disorder Caused by Monoallelic HK1 Variants. *Neurology. Genetics*, *10*(2), e200146. https://doi.org/10.1212/NXG.0000000000200146

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

146. Agostini, F., Pereyra, L., Dale, J., Yambire, K. F., Maglioni, S., Schiavi, A., Ventura, N., Milosevic, I., & Raimundo, N. (2024). Upregulation of cholesterol synthesis by lysosomal defects requires a functional mitochondrial respiratory chain. *The Journal of Biological Chemistry*, *300*(7), 107403. https://doi.org/10.1016/j.jbc.2024.107403

147. Aguilar, K., Jakubek, P., Zorzano, A., & Wieckowski, M. R. (2024). Primary mitochondrial diseases : The intertwined pathophysiology of bioenergetic dysregulation, oxidative stress and neuroinflammation. *European Journal of Clinical Investigation*, *54*(7), e14217. https://doi.org/10.1111/eci.14217

148. Antolínez-Fernández, Á., Esteban-Ramos, P., Fernández-Moreno, M. Á., & Clemente, P. (2024). Molecular pathways in mitochondrial disorders due to a defective mitochondrial protein synthesis. *Frontiers in Cell and Developmental Biology*, *12*, 1410245. https://doi.org/10.3389/fcell.2024.1410245

149. Argov, Z. (2024). Statins in hereditary myopathies : To give or not to give. *Neuromuscular Disorders: NMD*, *41*, 35-39. https://doi.org/10.1016/j.nmd.2024.06.004

150. Ball, M., Thorburn, D. R., & Rahman, S. (1993). Mitochondrial DNA-Associated Leigh Syndrome Spectrum. 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/NBK1173/

151. Barros, C. D. S., Coutinho, A., & Tengan, C. H. (2024). Arginine Supplementation in MELAS Syndrome : What Do We Know about the Mechanisms? *International Journal of Molecular Sciences*, *25*(7), 3629. https://doi.org/10.3390/ijms25073629

152. Battista, M., Carelli, V., Bottazzi, L., Bandello, F., Cascavilla, M. L., & Barboni, P. (2024). Gene therapy for Leber hereditary optic neuropathy. *Expert Opinion on Biological Therapy*, *24*(6), 521-528. https://doi.org/10.1080/14712598.2024.2359015

153. Bermejo-Guerrero, L., Hernández-Voth, A., Serrano-Lorenzo, P., Blázquez, A., Martin-Jimenez, P., Martin, M. A., & Domínguez-González, C. (2024). Remarkable clinical improvement with oral nucleoside treatment in a patient with adult-onset TK2 deficiency : A case report. *Mitochondrion*, *76*, 101879. https://doi.org/10.1016/j.mito.2024.101879

154. Bernardino Gomes, T. M., Vincent, A. E., Menger, K. E., Stewart, J. B., & Nicholls, T. J. (2024). Mechanisms and pathologies of human mitochondrial DNA replication and deletion formation. *The Biochemical Journal*, *481*(11), 683-715. https://doi.org/10.1042/BCJ20230262

155. Brañas Casas, R., Zuppardo, A., Risato, G., Dinarello, A., Celeghin, R., Fontana, C., Grelloni, E., Gilea, A. I., Viscomi, C., Rasola, A., Dalla Valle, L., Lodi, T., Baruffini, E., Facchinello, N., Argenton, F., & Tiso, N. (2024). Zebrafish polg2 knock-out recapitulates human POLG-disorders; implications for drug treatment. *Cell Death & Disease*, *15*(4), 281. https://doi.org/10.1038/s41419-024-06622-9

156. Brischigliaro, M., Sierra-Magro, A., Ahn, A., & Barrientos, A. (2024). Mitochondrial ribosome biogenesis and redox sensing. *FEBS Open Bio*, *14*(10), 1640-1655. https://doi.org/10.1002/2211-5463.13844

157. Burr, S. P., & Chinnery, P. F. (2024). Origins of tissue and cell-type specificity in mitochondrial DNA (mtDNA) disease. *Human Molecular Genetics*, *33*(R1), R3-R11. https://doi.org/10.1093/hmg/ddae059

158. Bury, A., Pyle, A., Vincent, A. E., Actis, P., & Hudson, G. (2024). Nanobiopsy investigation of the subcellular mtDNA heteroplasmy in human tissues. *Scientific Reports*, *14*(1), 13789. https://doi.org/10.1038/s41598-024-64455-0

159. Cai, H., Li, L.-M., Zhang, M., Zhou, Y., & Li, P. (2024). Case report : Late-onset MELAS syndrome with mtDNA 5783G>A mutation diagnosed by urinary sediment genetic testing. *Frontiers in Genetics*, *15*, 1367716. https://doi.org/10.3389/fgene.2024.1367716

160. Caron-Godon, C. A., Collington, E., Wolf, J. L., Coletta, G., & Glerum, D. M. (2024). More than Just Bread and Wine : Using Yeast to Understand Inherited Cytochrome Oxidase Deficiencies in Humans. *International Journal of Molecular Sciences*, *25*(7), 3814. https://doi.org/10.3390/ijms25073814

161. Chen, C., Tang, D., Xu, S., Xiang, L., Wang, B., Yao, Y., Li, Z., Lin, S., Li, S., Shi, X., Gu, C., & Gao, W. (2024). The promotion of non-small cell lung cancer progression by collagen and calcium binding EGF domain 1 is mediated through the regulation of ERK/JNK/P38 phosphorylation by reactive oxygen species. *Molecular Carcinogenesis*, *63*(8), 1467-1485. https://doi.org/10.1002/mc.23736

162. Chen, J., & Gao, L. (2024). SLC7A11-mediated cystine import protects against NDUFS7 deficiency-induced cell death in HEK293T cells. *Biochemical and Biophysical Research Communications*, *723*, 150178. https://doi.org/10.1016/j.bbrc.2024.150178

163. Chen, Y., Xu, R., Liu, Q., Zeng, Y., Chen, W., Liu, Y., Cao, Y., Liu, G., & Chen, Y. (2024). Rosmarinic acid ameliorated oxidative stress, neuronal injuries, and mitochondrial dysfunctions mediated by polyglutamine and α-synuclein in Caenorhabditis elegans models. *Molecular Neurobiology*. https://doi.org/10.1007/s12035-024-04206-4

164. Ciki, K., Alavanda, C., & Kara, M. (2024). Novel Mutation in the HSD17B10 Gene Accompanied by Dysmorphic Findings in Female Patients. *Molecular Syndromology*, *15*(3), 211-216. https://doi.org/10.1159/000535589

165. Cilleros-Holgado, P., Gómez-Fernández, D., Piñero-Pérez, R., Romero Domínguez, J. M., Talaverón-Rey, M., Reche-López, D., Suárez-Rivero, J. M., Álvarez-Córdoba, M., Romero-González, A., López-Cabrera, A., Oliveira, M. C. D., Rodríguez-Sacristan, A., & Sánchez-Alcázar, J. A. (2024). Polydatin and Nicotinamide Rescue the Cellular Phenotype of Mitochondrial Diseases by Mitochondrial Unfolded Protein Response (mtUPR) Activation. *Biomolecules*, *14*(5), 598. https://doi.org/10.3390/biom14050598

166. Constante, A. D., Abreu, S. M., & Trigo, C. (2024). Mitochondrial cardiomyopathy : A puzzle for the final diagnosis. *Cardiology in the Young*, *34*(6), 1393-1396. https://doi.org/10.1017/S1047951124025095

167. Corral-Sarasa, J., Martínez-Gálvez, J. M., González-García, P., Wendling, O., Jiménez-Sánchez, L., López-Herrador, S., Quinzii, C. M., Díaz-Casado, M. E., & López, L. C. (2024). 4-Hydroxybenzoic acid rescues multisystemic disease and perinatal lethality in a mouse model of mitochondrial disease. *Cell Reports*, *43*(5), 114148. https://doi.org/10.1016/j.celrep.2024.114148

168. Crameri, J. J., Palmer, C. S., Stait, T., Jackson, T. D., Lynch, M., Sinclair, A., Frajman, L. E., Compton, A. G., Coman, D., Thorburn, D. R., Frazier, A. E., & Stojanovski, D. (2024). Reduced Protein Import via TIM23 SORT Drives Disease Pathology in TIMM50-Associated Mitochondrial Disease. *Molecular and Cellular Biology*, *44*(6), 226-244. https://doi.org/10.1080/10985549.2024.2353652

169. Cwerman-Thibault, H., Malko-Baverel, V., Le Guilloux, G., Torres-Cuevas, I., Ratcliffe, E., Mouri, D., Mignon, V., Saubaméa, B., Boespflug-Tanguy, O., Gressens, P., & Corral-Debrinski, M. (2024). Harlequin mice exhibit cognitive impairment, severe loss of Purkinje cells and a compromised bioenergetic status due to the absence of Apoptosis Inducing Factor. *Biochimica Et Biophysica Acta. Molecular Basis of Disease*, *1870*(7), 167272. https://doi.org/10.1016/j.bbadis.2024.167272

170. Dobner, J., Nguyen, T., Pavez-Giani, M. G., Cyganek, L., Distelmaier, F., Krutmann, J., Prigione, A., & Rossi, A. (2024). mtDNA analysis using Mitopore. *Molecular Therapy. Methods & Clinical Development*, *32*(2), 101231. https://doi.org/10.1016/j.omtm.2024.101231

171. Dupuis, H., Lemaitre, M., Jannin, A., Douillard, C., Espiard, S., & Vantyghem, M.-C. (2024). Lipomatoses. *Annales D'endocrinologie*, *85*(3), 231-247. https://doi.org/10.1016/j.ando.2024.05.003

172. Eller, M. M., Zuberi, A. R., Fu, X., Burgess, S. C., Lutz, C. M., & Bailey, R. M. (2024). Valine and Inflammation Drive Epilepsy in a Mouse Model of ECHS1 Deficiency. *bioRxiv: The Preprint Server for Biology*, 2024.06.13.598697. https://doi.org/10.1101/2024.06.13.598697

173. Esteban-Vasallo, M. D., Domínguez-Berjón, M. F., Chalco-Orrego, J. P., & González Martín-Moro, J. (2024). Prevalence of Leber hereditary optic neuropathy in the Community of Madrid (Spain), estimation with a capture-recapture method. *Orphanet Journal of Rare Diseases*, *19*(1), 220. https://doi.org/10.1186/s13023-024-03225-7

174. Falkenberg, M., Larsson, N.-G., & Gustafsson, C. M. (2024). Replication and Transcription of Human Mitochondrial DNA. *Annual Review of Biochemistry*, *93*(1), 47-77. https://doi.org/10.1146/annurev-biochem-052621-092014

175. Finsterer, J. (2024). Diagnostic accuracy is required when analysing cohorts with mitochondrial disorders. *European Journal of Paediatric Neurology: EJPN: Official Journal of the European Paediatric Neurology Society*, *51*, 71-72. https://doi.org/10.1016/j.ejpn.2024.05.017

176. Finsterer, J., & Strobl, W. (2024). Gastrointestinal involvement in neuromuscular disorders. *Journal of Gastroenterology and Hepatology*, *39*(10), 1982-1993. https://doi.org/10.1111/jgh.16650

177. Fortin, O., Christoffel, K., Shoaib, A., Venkatesan, C., Cilli, K., Schroeder, J. W., Alves, C., Ganetzky, R. D., & Fraser, J. L. (2024). Characteristic Fetal Brain MRI Abnormalities in Pyruvate Dehydrogenase Complex Deficiency. *medRxiv: The Preprint Server for Health Sciences*, 2024.04.08.24303574. https://doi.org/10.1101/2024.04.08.24303574

178. Galosi, S., Mancini, C., Commone, A., Calligari, P., Caputo, V., Nardecchia, F., Carducci, C., van den Heuvel, L. P., Pizzi, S., Bruselles, A., Niceta, M., Martinelli, S., Rodenburg, R. J., Tartaglia, M., & Leuzzi, V. (2024). Biallelic Variants of MRPS36 Cause a New Form of Leigh Syndrome. *Movement Disorders: Official Journal of the Movement Disorder Society*, *39*(7), 1225-1231. https://doi.org/10.1002/mds.29795

179. Georgiou, T., Petrou, P. P., Malekkou, A., Ioannou, I., Gavatha, M., Skordis, N., Nicolaidou, P., Savvidou, I., Athanasiou, E., Ourani, S., Papamichael, E., Vogazianos, M., Dionysiou, M., Mavrikiou, G., Grafakou, O., Tanteles, G. A., Anastasiadou, V., & Drousiotou, A. (2024). Inherited metabolic disorders in Cyprus. *Molecular Genetics and Metabolism Reports*, *39*, 101083. https://doi.org/10.1016/j.ymgmr.2024.101083

180. Gervasoni, J., Primiano, A., Cicchinelli, M., Santucci, L., Servidei, S., Urbani, A., Primiano, G., & Iavarone, F. (2024). Mitochondrial Biomarkers in the Omics Era : A Clinical-Pathophysiological Perspective. *International Journal of Molecular Sciences*, *25*(9), 4855. https://doi.org/10.3390/ijms25094855

181. Giannoccaro, M. P., Morelli, L., Ricciardiello, F., Donadio, V., Bartiromo, F., Tonon, C., Carbonelli, M., Amore, G., Carelli, V., Liguori, R., & La Morgia, C. (2024). Co-occurrence of glial fibrillary acidic protein astrocytopathy in a patient with Leber's hereditary optic neuropathy due to DNAJC30 mutations. *European Journal of Neurology*, *31*(9), e16344. https://doi.org/10.1111/ene.16344

182. Gómez González, C., Del Campo Cano, I., Isabel Fernández-Avila, A., Paz Suárez-Mier, M., José Sagastizábal, M., Álvarez García-Rovés, R., Méndez Fernández, I., Vilches, S., Centeno Jiménez, M., Siles Sánchez-Manjavacas, A., Usano Carrasco, A., Gonzalez-Vioque, E., Pablo Ochoa, J., Medrano, C., González López, E., García-Pavía, P., Bermejo, J., & Angeles Espinosa Castro, M. (2024). Sudden cardiac death triggered by minimal alcohol consumption in the context of novel PPA2 mutations in 2 unrelated families. *Gene*, *916*, 148437. https://doi.org/10.1016/j.gene.2024.148437

183. Gupta, V., Jolly, B., Bhoyar, R. C., Divakar, M. K., Jain, A., Mishra, A., Senthivel, V., Imran, M., Scaria, V., & Sivasubbu, S. (2024). Spectrum of rare and common mitochondrial DNA variations from 1029 whole genomes of self-declared healthy individuals from India. *Computational Biology and Chemistry*, *112*, 108118. https://doi.org/10.1016/j.compbiolchem.2024.108118

184. Han, X., Li, H., Deng, J., Zhuo, X., Liu, Z., Xu, M., Feng, W., Chen, S., & Fang, F. (2024). Genotype and Phenotype Characteristics of 58 Cases of Mitochondrial Epilepsy with Nuclear DNA Mutations in Children. *Neurological Sciences: Official Journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology*, *45*(11), 5465-5480. https://doi.org/10.1007/s10072-024-07586-6

185. Hanaford, A. R., Khanna, A., James, K., Truong, V., Liao, R., Chen, Y., Mulholland, M., Kayser, E.-B., Watanabe, K., Hsieh, E. S., Sedensky, M., Morgan, P. G., Kalia, V., Sarkar, S., & Johnson, S. C. (2024). Interferon-gamma contributes to disease progression in the Ndufs4(-/-) model of Leigh syndrome. *Neuropathology and Applied Neurobiology*, *50*(3), e12977. https://doi.org/10.1111/nan.12977

186. Haque, S., Crawley, K., Davis, R., Schofield, D., Shrestha, R., & Sue, C. M. (2024). Clinical drivers of hospitalisation in patients with mitochondrial diseases. *BMJ Neurology Open*, *6*(1), e000717. https://doi.org/10.1136/bmjno-2024-000717

187. Hassani, S., & Esmaeili, A. (2024). The neuroprotective effects of ferulic acid in toxin-induced models of Parkinson's disease : A review. *Ageing Research Reviews*, *97*, 102299. https://doi.org/10.1016/j.arr.2024.102299

188. Heath, O., Hammerl, E., Spitzinger, A., & Wortmann, S. B. (2024). Ending an Odyssey? The Psychosocial Experiences of Parents after the Genetic Diagnosis of a Mitochondrial Disease in Children. *Journal of Personalized Medicine*, *14*(5), 523. https://doi.org/10.3390/jpm14050523

189. Hikmat, O., Naess, K., Engvall, M., Klingenberg, C., Rasmussen, M., Brodtkorb, E., Ostergaard, E., de Coo, I., Pias-Peleteiro, L., Isohanni, P., Uusimaa, J., Majamaa, K., Kärppä, M., Ortigoza-Escobar, J. D., Tangeraas, T., Berland, S., Harrison, E., Biggs, H., Horvath, R., … Bindoff, L. A. (2024). Status epilepticus in POLG disease : A large multinational study. *Journal of Neurology*, *271*(8), 5156-5164. https://doi.org/10.1007/s00415-024-12463-5

190. Huang, D., Li, Y., Han, J., Zuo, H., Liu, H., & Chen, Z. (2024). Xbp1 promotes odontoblastic differentiation through modulating mitochondrial homeostasis. *FASEB Journal: Official Publication of the Federation of American Societies for Experimental Biology*, *38*(7), e23600. https://doi.org/10.1096/fj.202400186R

191. Huang, Q., Trumpff, C., Monzel, A. S., Rausser, S., Haahr, R., Devine, J., Liu, C. C., Kelly, C., Thompson, E., Kurade, M., Michelson, J., Shaulson, E. D., Li, S., Engelstad, K., Tanji, K., Lauriola, V., Wang, T., Wang, S., Zuraikat, F. M., … Picard, M. (2024). Psychobiological regulation of plasma and saliva GDF15 dynamics in health and mitochondrial diseases. *bioRxiv: The Preprint Server for Biology*, 2024.04.19.590241. https://doi.org/10.1101/2024.04.19.590241

192. Hughes, L. A., Rackham, O., & Filipovska, A. (2024). Illuminating mitochondrial translation through mouse models. *Human Molecular Genetics*, *33*(R1), R61-R79. https://doi.org/10.1093/hmg/ddae020

193. Indelicato, E., Schlieben, L. D., Stenton, S. L., Boesch, S., Skorvanek, M., Necpal, J., Jech, R., Winkelmann, J., Prokisch, H., & Zech, M. (2024). Dystonia and mitochondrial disease : The movement disorder connection revisited in 900 genetically diagnosed patients. *Journal of Neurology*, *271*(7), 4685-4692. https://doi.org/10.1007/s00415-024-12447-5

194. Iwami, K., Kano, T., Mizushima, K., Yaguchi, H., Nishino, I., & Houzen, H. (2024). [Diagnosis of anti-melanoma differentiation-associated gene 5 antibody-positive dermatomyositis led by sarcoplasmic myxovirus resistance protein A expression on muscle pathology]. *Rinsho Shinkeigaku = Clinical Neurology*, *64*(7), 480-485. https://doi.org/10.5692/clinicalneurol.cn-001963

195. Jiao, Q., Xiang, L., & Chen, Y. (2024). Mitochondrial transplantation : A promising therapy for mitochondrial disorders. *International Journal of Pharmaceutics*, *658*, 124194. https://doi.org/10.1016/j.ijpharm.2024.124194

196. Kanemaru, E., Shimoda, K., Marutani, E., Morita, M., Miranda, M., Miyazaki, Y., Sinow, C., Sharma, R., Dong, F., Bloch, D. B., Akaike, T., & Ichinose, F. (2024). Exclusion of sulfide:quinone oxidoreductase from mitochondria causes Leigh-like disease in mice by impairing sulfide metabolism. *The Journal of Clinical Investigation*, *134*(15), e170994. https://doi.org/10.1172/JCI170994

197. Kang, Y., Hepojoki, J., Maldonado, R. S., Mito, T., Terzioglu, M., Manninen, T., Kant, R., Singh, S., Othman, A., Verma, R., Uusimaa, J., Wartiovaara, K., Kareinen, L., Zamboni, N., Nyman, T. A., Paetau, A., Kipar, A., Vapalahti, O., & Suomalainen, A. (2024). Ancestral allele of DNA polymerase gamma modifies antiviral tolerance. *Nature*, *628*(8009), 844-853. https://doi.org/10.1038/s41586-024-07260-z

198. Kaufman, O., Donnelly, C., Cork, E., Fiel, M. I., Chu, J., & Ganesh, J. (2024). Shwachman-Diamond syndrome mimicking mitochondrial hepatopathy. *JPGN Reports*, *5*(2), 213-217. https://doi.org/10.1002/jpr3.12064

199. Kaur, G., Ganev, Y., Rodriguez, W., Tseng, S., Orozco, L., & Chand, P. (2024). Deep Brain Stimulation for Medication Refractory Tremor in Leber Optic Neuropathy Plus Syndrome. *Cureus*, *16*(4), e58255. https://doi.org/10.7759/cureus.58255

200. Kayser, E.-B., Chen, Y., Mulholland, M., Truong, V., James, K., Hanaford, A., & Johnson, S. (2024). Evaluating the efficacy of vatiquinone in preclinical models of mitochondrial disease. *Research Square*, rs.3.rs-4202689. https://doi.org/10.21203/rs.3.rs-4202689/v1

201. Kobayashi, M., Miyauchi, A., Jimbo, E. F., Oishi, N., Aoki, S., Watanabe, M., Yoshikawa, Y., Akiyama, Y., Yamagata, T., & Osaka, H. (2024). Synthetic aporphine alkaloids are potential therapeutics for Leigh syndrome. *Scientific Reports*, *14*(1), 11561. https://doi.org/10.1038/s41598-024-62445-w

202. Kousa, A., Ahmed, R., & Alasmar, P. D. (2024). Syrian child carrying multiple pathogenic variants in MBOAT7 and MT-TS1 genes : A case report on neurodevelopmental phenotypes and mitochondrial inheritance. *Annals of Medicine and Surgery (2012)*, *86*(5), 3086-3089. https://doi.org/10.1097/MS9.0000000000001941

203. Kowalska, A., Figura, M., Zawadka, M., & Koziorowski, D. (2024). Pyruvate dehydrogenase-E1α deficiency presenting as generalized dystonia : A genetic diagnosis with important clinical implications. *Clinical Neurology and Neurosurgery*, *241*, 108307. https://doi.org/10.1016/j.clineuro.2024.108307

204. Küçükcongar Yavaş, A., Basan, H., Dinçer, S., Bilginer Gürbüz, B., & Kasapkara, Ç. S. (2024). Mitochondrial phosphate-carrier deficiency mimicking infantile-onset Pompe disease. *American Journal of Medical Genetics. Part A*, *194*(9), e63643. https://doi.org/10.1002/ajmg.a.63643

205. Kuin, R. E. M., Groothuis, J. T., Buit, P., Janssen, M. C. H., & Knuijt, S. (2024). Dysarthria and dysphagia in patients with mitochondrial diseases. *Molecular Genetics and Metabolism*, *142*(3), 108510. https://doi.org/10.1016/j.ymgme.2024.108510

206. Kumar, A., Choudhary, A., & Munshi, A. (2024). Epigenetic reprogramming of mtDNA and its etiology in mitochondrial diseases. *Journal of Physiology and Biochemistry*. https://doi.org/10.1007/s13105-024-01032-z

207. Launay, N., Lopez-Erauskin, J., Bianchi, P., Guha, S., Parameswaran, J., Coppa, A., Torreni, L., Schlüter, A., Fourcade, S., Paredes-Fuentes, A. J., Artuch, R., Casasnovas, C., Ruiz, M., & Pujol, A. (2024). Imbalanced mitochondrial dynamics contributes to the pathogenesis of X-linked adrenoleukodystrophy. *Brain: A Journal of Neurology*, *147*(6), 2069-2084. https://doi.org/10.1093/brain/awae038

208. Li, J., Hou, F., Lv, N., Zhao, R., Zhang, L., Yue, C., Nie, M., & Chen, L. (2024). From Rare Disorders of Kidney Tubules to Acute Renal Injury : Progress and Prospective. *Kidney Diseases (Basel, Switzerland)*, *10*(2), 153-166. https://doi.org/10.1159/000536423

209. Li, T., Aziz, T., Li, G., Zhang, L., Yao, J., & Jia, S. (2024). A zebrafish tufm mutant model for the COXPD4 syndrome of aberrant mitochondrial function. *Journal of Genetics and Genomics = Yi Chuan Xue Bao*, *51*(9), 922-933. https://doi.org/10.1016/j.jgg.2024.05.009

210. Liang, D., Zhu, L., Zhu, Y., Huang, M., Lin, Y., Li, H., Hu, P., Zhang, J., Shen, B., & Xu, Z. (2024). A PCR-independent approach for mtDNA enrichment and next-generation sequencing : Comprehensive evaluation and clinical application. *Journal of Translational Medicine*, *22*(1), 386. https://doi.org/10.1186/s12967-024-05213-8

211. Liang, K. X., Chen, A., Kianian, A., Kristiansen, C. K., Yangzom, T., Furriol, J., Høyland, L. E., Ziegler, M., Kråkenes, T., Tzoulis, C., Fang, E. F., Sullivan, G. J., & Bindoff, L. A. (2024). Activation of Neurotoxic Astrocytes Due to Mitochondrial Dysfunction Triggered by POLG Mutation. *International Journal of Biological Sciences*, *20*(8), 2860-2880. https://doi.org/10.7150/ijbs.93445

212. Lin, Y., Wang, J., Xu, R., Xu, Z., Wang, Y., Pan, S., Zhang, Y., Tao, Q., Zhao, Y., Yan, C., Cao, Z., & Ji, K. (2024). HiFi long-read amplicon sequencing for full-spectrum variants of human mtDNA. *BMC Genomics*, *25*(1), 538. https://doi.org/10.1186/s12864-024-10433-9

213. Liu, D., Zhou, X., He, Y., & Zhao, J. (2024). The Roles of CircRNAs in Mitochondria. *Journal of Cancer*, *15*(9), 2759-2769. https://doi.org/10.7150/jca.92111

214. Liu, N., Liang, Y., Wei, T., Huang, X., Zhang, T., & Tang, M. (2024). ROS-mediated NRF2/p-ERK1/2 signaling-involved mitophagy contributes to macrophages activation induced by CdTe quantum dots. *Toxicology*, *505*, 153825. https://doi.org/10.1016/j.tox.2024.153825

215. Liu, N., Pang, B., Kang, L., Li, D., Jiang, X., & Zhou, C.-M. (2024). TUFM in health and disease : Exploring its multifaceted roles. *Frontiers in Immunology*, *15*, 1424385. https://doi.org/10.3389/fimmu.2024.1424385

216. Liufu, T., Zhao, X., Yu, M., Xie, Z., Meng, L., Lv, H., Zhang, W., Yuan, Y., Xing, G., Deng, J., & Wang, Z. (2024). Multiomics analysis reveals serine catabolism as a potential therapeutic target for MELAS. *FASEB Journal: Official Publication of the Federation of American Societies for Experimental Biology*, *38*(12), e23742. https://doi.org/10.1096/fj.202302286RRR

217. Lopes, E. C., Shi, F., Sawant, A., Ibrahim, M., Gomez-Jenkins, M., Hu, Z., Manchiraju, P., Bhatt, V., Wang, W., Hinrichs, C. S., Wallace, D. C., Su, X., Rabinowitz, J. D., Chan, C. S., Guo, J. Y., Ganesan, S., Lattime, E. C., & White, E. (2024). RESPIRATION DEFECTS LIMIT SERINE SYNTHESIS REQUIRED FOR LUNG CANCER GROWTH AND SURVIVAL. *bioRxiv: The Preprint Server for Biology*, 2024.05.28.596339. https://doi.org/10.1101/2024.05.28.596339

218. Lopriore, P., Palermo, G., Meli, A., Bellini, G., Benevento, E., Montano, V., Siciliano, G., Mancuso, M., & Ceravolo, R. (2024). Mitochondrial Parkinsonism : A Practical Guide to Genes and Clinical Diagnosis. *Movement Disorders Clinical Practice*, *11*(8), 948-965. https://doi.org/10.1002/mdc3.14148

219. Lopriore, P., Vista, M., Tessa, A., Giuntini, M., Caldarazzo Ienco, E., Mancuso, M., Siciliano, G., Santorelli, F. M., & Orsucci, D. (2024). Primary Coenzyme Q10 Deficiency-Related Ataxias. *Journal of Clinical Medicine*, *13*(8), 2391. https://doi.org/10.3390/jcm13082391

220. Male, A. J., Koohi, N., Holmes, S. L., Pitceathly, R. D. S., & Kaski, D. (2024). Acceptability of Audiovestibular Assessment in the Home-A Patient Survey. *Audiology Research*, *14*(3), 545-553. https://doi.org/10.3390/audiolres14030045

221. Mallawaarachchi, A. C., Fowles, L., Wardrop, L., Wood, A., O'Shea, R., Biros, E., Harris, T., Alexander, S. I., Bodek, S., Boudville, N., Burke, J., Burnett, L., Casauria, S., Chadban, S., Chakera, A., Crafter, S., Dai, P., De Fazio, P., Faull, R., … Mallett, A. J. (2024). Genomic Testing in Patients with Kidney Failure of an Unknown Cause : A National Australian Study. *Clinical Journal of the American Society of Nephrology: CJASN*, *19*(7), 887-897. https://doi.org/10.2215/CJN.0000000000000464

222. Mancuso, M., Papadopoulou, M. T., Ng, Y. S., Ardissone, A., Bellusci, M., Bertini, E., Di Vito, L., Evangelista, T., Fons, C., Hikmat, O., Horvath, R., Klopstock, T., Kornblum, C., Lamperti, C., Licchetta, L., Molnar, M. J., Varhaug, K. N., O'Callaghan, M., Pressler, R. M., … Rahman, S. (2024). Management of seizures in patients with primary mitochondrial diseases : Consensus statement from the InterERNs Mitochondrial Working Group. *European Journal of Neurology*, *31*(7), e16275. https://doi.org/10.1111/ene.16275

223. Marin-Valencia, I., Kocabas, A., Rodriguez-Navas, C., Miloushev, V. Z., González-Rodríguez, M., Lees, H., Henry, K. E., Vaynshteyn, J., Longo, V., Deh, K., Eskandari, R., Mamakhanyan, A., Berishaj, M., & Keshari, K. R. (2024). Imaging brain glucose metabolism in vivo reveals propionate as a major anaplerotic substrate in pyruvate dehydrogenase deficiency. *Cell Metabolism*, *36*(6), 1394-1410.e12. https://doi.org/10.1016/j.cmet.2024.05.002

224. Meldau, S., Ackermann, S., Riordan, G., van der Watt, G. F., Spencer, C., Raga, S., Khan, K., Blackhurst, D. M., & van der Westhuizen, F. H. (2024). A novel mitochondrial DNA variant in MT-ND6 : M.14430A>C p.(Trp82Gly) identified in a patient with Leigh syndrome and complex I deficiency. *Molecular Genetics and Metabolism Reports*, *39*, 101078. https://doi.org/10.1016/j.ymgmr.2024.101078

225. Messina, M., Vaz, F. M., & Rahman, S. (2024). Mitochondrial membrane synthesis, remodelling and cellular trafficking. *Journal of Inherited Metabolic Disease*. https://doi.org/10.1002/jimd.12766

226. Misceo, D., Strømme, P., Bitarafan, F., Chawla, M. S., Sheng, Y., Bach de Courtade, S. M., Eide, L., & Frengen, E. (2024). Biallelic NDUFA4 Deletion Causes Mitochondrial Complex IV Deficiency in a Patient with Leigh Syndrome. *Genes*, *15*(4), 500. https://doi.org/10.3390/genes15040500

227. Moisoi, N. (2024). Mitochondrial proteases modulate mitochondrial stress signalling and cellular homeostasis in health and disease. *Biochimie*, S0300-9084(24)00141-X. https://doi.org/10.1016/j.biochi.2024.06.005

228. Momoh, R., & Kollamparambil, S. (2024). A Case Report of a Clinically Suspected Diagnosis of Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-Like Episodes (MELAS) Syndrome With Cardiac Impairment. *Cureus*, *16*(3), e56980. https://doi.org/10.7759/cureus.56980

229. Morcillo, P., Kabra, K., Velasco, K., Cordero, H., Jennings, S., Yun, T. D., Larrea, D., Akman, H. O., & Schon, E. A. (2024). Aberrant ER-mitochondria communication is a common pathomechanism in mitochondrial disease. *Cell Death & Disease*, *15*(6), 405. https://doi.org/10.1038/s41419-024-06781-9

230. Moustaine, M. O., Azemour, Z., Mohammed, F., Benlanda, O., Nassik, H., & Karkouri, M. (2024). Management of Ptosis in Kearns-Sayre Syndrome : A Case Report and Literature Review. *Archives of Plastic Surgery*, *51*(2), 182-186. https://doi.org/10.1055/a-2207-7587

231. Neuhofer, C. M., & Prokisch, H. (2024). Digenic Inheritance in Rare Disorders and Mitochondrial Disease-Crossing the Frontier to a More Comprehensive Understanding of Etiology. *International Journal of Molecular Sciences*, *25*(9), 4602. https://doi.org/10.3390/ijms25094602

232. Oharazawa, A., Maimaituxun, G., Watanabe, K., Nishiyasu, T., & Fujii, N. (2024). Metabolome analyses of skin dialysate : Insights into skin interstitial fluid biomarkers. *Journal of Dermatological Science*, *114*(3), 141-147. https://doi.org/10.1016/j.jdermsci.2024.04.001

233. Olimpio, C., Paramonov, I., Matalonga, L., Laurie, S., Schon, K., Polavarapu, K., Kirschner, J., Schara-Schmidt, U., Lochmüller, H., Chinnery, P. F., & Horvath, R. (2024). Increased Diagnostic Yield by Reanalysis of Whole Exome Sequencing Data in Mitochondrial Disease. *Journal of Neuromuscular Diseases*, *11*(4), 767-775. https://doi.org/10.3233/JND-240020

234. Othonicar, M. F., Garcia, G. S., & Oliveira, M. T. (2024). The alternative enzymes-bearing tunicates lack multiple widely distributed genes coding for peripheral OXPHOS subunits. *Biochimica Et Biophysica Acta. Bioenergetics*, *1865*(3), 149046. https://doi.org/10.1016/j.bbabio.2024.149046

235. Ozlu, C., Messahel, S., Minassian, B., & Kayani, S. (2024). Mitochondrial encephalopathies and myopathies : Our tertiary center's experience. *European Journal of Paediatric Neurology: EJPN: Official Journal of the European Paediatric Neurology Society*, *50*, 31-40. https://doi.org/10.1016/j.ejpn.2024.03.005

236. Pennings, G., Heindryckx, B., Stoop, D., & Mertes, H. (2024). Attitude of Belgian women towards enucleated egg donation for treatment of mitochondrial diseases and infertility. *Reproductive Biomedicine Online*, *49*(3), 104101. https://doi.org/10.1016/j.rbmo.2024.104101

237. Pickup, E., Moore, S. A., Suwannarat, P., Grant, C., Ah Mew, N., Gropman, A., & Sen, K. (2024). Expedited Exome Reanalysis Following Deep Phenotyping and Muscle Biopsy in Suspected Mitochondrial Disorder. *Pediatric Neurology*, *156*, 178-181. https://doi.org/10.1016/j.pediatrneurol.2024.04.007

238. Pieri, M., D'Andria Ursoleo, J., Di Prima, A. L., Bugo, S., Barucco, G., Licheri, M., Losiggio, R., Frau, G., Monaco, F., & Collaborators. (2024). Remimazolam for anesthesia and sedation in pediatric patients : A scoping review. *Journal of Anesthesia*, *38*(5), 692-710. https://doi.org/10.1007/s00540-024-03358-w

239. Qiu, J., Wu, H., Xie, Q., Zhou, Y., Gao, Y., Liu, J., Jiang, X., Suo, L., & Kuang, Y. (2024). Harnessing accurate mitochondrial DNA base editing mediated by DdCBEs in a predictable manner. *Frontiers in Bioengineering and Biotechnology*, *12*, 1372211. https://doi.org/10.3389/fbioe.2024.1372211

240. Rackham, O., Saurer, M., Ban, N., & Filipovska, A. (2024). Unique architectural features of mammalian mitochondrial protein synthesis. *Trends in Cell Biology*, S0962-8924(24)00097-7. https://doi.org/10.1016/j.tcb.2024.05.001

241. Ramesh, R., Amanmahanya, C., Krishnamoorthy, V., Krishnan, V., Palani, S., & Narasimhan Ranganathan, L. (2024). Oculomasticatory rhythmic movements, insomnia and stroke-like episodes in a patient with POLG mutation. *BMJ Case Reports*, *17*(4), e259426. https://doi.org/10.1136/bcr-2023-259426

242. Ratnaike, T. E., Elkhateeb, N., Lochmüller, A., Gilmartin, C., Schon, K., Horváth, R., & Chinnery, P. F. (2024). Evidence for sodium valproate toxicity in mitochondrial diseases : A systematic analysis. *BMJ Neurology Open*, *6*(1), e000650. https://doi.org/10.1136/bmjno-2024-000650

243. Ray, S., Parmar, U., Saxena, V., & Dias, R. (2024). Delayed awakening after sevoflurane anesthesia for MRI brain in a child with undiagnosed mitochondrial disorder. *Journal of Anaesthesiology, Clinical Pharmacology*, *40*(1), 162-163. https://doi.org/10.4103/joacp.joacp_231_22

244. Reynaud-Dulaurier, R., Clément, R., Yjjou, S., Cresson, C., Saoudi, Y., Faideau, M., & Decressac, M. (2024). The Blood-Brain Barrier Is Unaffected in the Ndufs4-/- Mouse Model of Leigh Syndrome. *International Journal of Molecular Sciences*, *25*(9), 4828. https://doi.org/10.3390/ijms25094828

245. Rezaei, M., Dourandish, Z., Kiani Mehr, G., Ghorbani, A., & Fatehi, F. (2024). Cavitating leukodystrophy in a case of mitochondrial complex III deficiency due to LYRM7 mutation. *Acta Neurologica Belgica*, *124*(4), 1409-1410. https://doi.org/10.1007/s13760-024-02529-6

246. Riccio, A. A., Brannon, A. J., Krahn, J. M., Bouvette, J., Williams, J. G., Borgnia, M. J., & Copeland, W. C. (2024). Coordinated DNA polymerization by Polγ and the region of LonP1 regulated proteolysis. *Nucleic Acids Research*, *52*(13), 7863-7875. https://doi.org/10.1093/nar/gkae539

247. Rouzier, C., Pion, E., Chaussenot, A., Bris, C., Ait-El-Mkadem Saadi, S., Desquiret-Dumas, V., Gueguen, N., Fragaki, K., Amati-Bonneau, P., Barcia, G., Gaignard, P., Steffann, J., Pennisi, A., Bonnefont, J.-P., Lebigot, E., Bannwarth, S., Francou, B., Rucheton, B., Sternberg, D., … Procaccio, V. (2024). Primary mitochondrial disorders and mimics : Insights from a large French cohort. *Annals of Clinical and Translational Neurology*, *11*(6), 1478-1491. https://doi.org/10.1002/acn3.52062

248. Sartorelli, J., Longo, D., Travaglini, L., Orlando, V., D'Amico, A., Bertini, E., & Nicita, F. (2024). Acute Ophthalmoplegia with Wernicke-Like MRI Pattern in a Patient with HPDL-Related Disorder. *Movement Disorders Clinical Practice*, *11*(9), 1160-1162. https://doi.org/10.1002/mdc3.14153

249. Selvanathan, A., Teo, J., & Parayil Sankaran, B. (2024). Hematologic Manifestations in Primary Mitochondrial Diseases. *Journal of Pediatric Hematology/Oncology*, *46*(5), e338-e347. https://doi.org/10.1097/MPH.0000000000002890

250. Shen, J., Xie, P., Wang, J., Yang, F., Li, S., Jiang, H., Wu, X., Zhou, F., & Li, J. (2024). Nlrp6 protects from corticosterone-induced NSPC ferroptosis by modulating RIG-1/MAVS-mediated mitophagy. *Redox Biology*, *73*, 103196. https://doi.org/10.1016/j.redox.2024.103196

251. Shinkai, R., Honda, T., Watanabe, R., Ooka, M., Kitsuda, K., Hirata, Y., & Ishikura, K. (2024). Fetal bradycardia as the initial symptom of mitochondrial disease : A case report. *Pediatrics International: Official Journal of the Japan Pediatric Society*, *66*(1), e15771. https://doi.org/10.1111/ped.15771

252. Singh, D. (2024). Beyond the membrane : Exploring non-viral methods for mitochondrial gene delivery. *Mitochondrion*, *78*, 101922. https://doi.org/10.1016/j.mito.2024.101922

253. Slade, L., Deane, C. S., Szewczyk, N. J., Etheridge, T., & Whiteman, M. (2024). Hydrogen sulfide supplementation as a potential treatment for primary mitochondrial diseases. *Pharmacological Research*, *203*, 107180. https://doi.org/10.1016/j.phrs.2024.107180

254. Sniezek Carney, O., Harris, K. W., Wohlfarter, Y., Lee, K., Butschek, G., Anzmann, A., Claypool, S. M., Hamacher-Brady, A., Keller, M., & Vernon, H. J. (2024). Stem cell models of TAFAZZIN deficiency reveal novel tissue-specific pathologies in Barth Syndrome. *bioRxiv: The Preprint Server for Biology*, 2024.04.28.591534. https://doi.org/10.1101/2024.04.28.591534

255. Song, N., Mei, S., Wang, X., Hu, G., & Lu, M. (2024). Focusing on mitochondria in the brain : From biology to therapeutics. *Translational Neurodegeneration*, *13*(1), 23. https://doi.org/10.1186/s40035-024-00409-w

256. Southwell, N., Manzo, O., Bacman, S., Zhao, D., Sayles, N. M., Dash, J., Fujita, K., D'Aurelio, M., Di Lorenzo, A., Manfredi, G., & Kawamata, H. (2024). High fat diet ameliorates mitochondrial cardiomyopathy in CHCHD10 mutant mice. *EMBO Molecular Medicine*, *16*(6), 1352-1378. https://doi.org/10.1038/s44321-024-00067-5

257. Steenberge, L. H., Rogers, S., Sung, A. Y., Fan, J., & Pagliarini, D. J. (2024). Coenzyme Q4 is a functional substitute for coenzyme Q10 and can be targeted to the mitochondria. *The Journal of Biological Chemistry*, *300*(5), 107269. https://doi.org/10.1016/j.jbc.2024.107269

258. Suiter, E., Baiker, K., Kaczmarska, A., Christen, M., Leeb, T., Ororbia, A., Anselmi, C., Minguez, J., & Gutierrez-Quintana, R. (2024). Novel MRI and histopathological findings in a young Bullmastiff cross dog with mitochondrial fission encephalopathy. *Veterinary Radiology & Ultrasound: The Official Journal of the American College of Veterinary Radiology and the International Veterinary Radiology Association*, *65*(5), 463-468. https://doi.org/10.1111/vru.13342

259. Sun, G., Huang, W., Wang, L., Wu, J., Zhao, G., Ren, H., Liu, L., & Kong, X. (2024). Molecular findings in patients for whole exome sequencing and mitochondrial genome assessment. *Clinica Chimica Acta; International Journal of Clinical Chemistry*, *561*, 119774. https://doi.org/10.1016/j.cca.2024.119774

260. Sung, A. Y., Guerra, R. M., Steenberge, L. H., Alston, C. L., Murayama, K., Okazaki, Y., Shimura, M., Prokisch, H., Ghezzi, D., Torraco, A., Carrozzo, R., Rötig, A., Taylor, R. W., Keck, J. L., & Pagliarini, D. J. (2024). Systematic analysis of NDUFAF6 in complex I assembly and mitochondrial disease. *Nature Metabolism*, *6*(6), 1128-1142. https://doi.org/10.1038/s42255-024-01039-2

261. Takai, Y., Yamagami, A., & Ishikawa, H. (2024). [Leber's hereditary optic neuropathy]. *Rinsho Shinkeigaku = Clinical Neurology*, *64*(5), 326-332. https://doi.org/10.5692/clinicalneurol.cn-001924

262. Tavasoli, A., Kachuei, M., Talebi, S., & Eghdami, S. (2024). Complex mitochondrial disease caused by the mutation of COX10 in a toddler : A case-report study. *Annals of Medicine and Surgery (2012)*, *86*(6), 3753-3756. https://doi.org/10.1097/MS9.0000000000002096

263. Thompson, W. R., Manuel, R., Abbruscato, A., Carr, J., Campbell, J., Hornby, B., Vaz, F. M., & Vernon, H. J. (2024). Long-term efficacy and safety of elamipretide in patients with Barth syndrome : 168-week open-label extension results of TAZPOWER. *Genetics in Medicine: Official Journal of the American College of Medical Genetics*, *26*(7), 101138. https://doi.org/10.1016/j.gim.2024.101138

264. Tian, D., Cui, M., & Han, M. (2024). Bacterial muropeptides promote OXPHOS and suppress mitochondrial stress in mammals. *Cell Reports*, *43*(4), 114067. https://doi.org/10.1016/j.celrep.2024.114067

265. Tong, Q., Miao, Y., & Yin, H. (2024). Echocardiographic manifestations of mitochondrial disease with GTPBP3 gene mutations : A case report. *Medicine*, *103*(18), e37847. https://doi.org/10.1097/MD.0000000000037847

266. Tyczyńska, M., Gędek, M., Brachet, A., Stręk, W., Flieger, J., Teresiński, G., & Baj, J. (2024). Trace Elements in Alzheimer's Disease and Dementia : The Current State of Knowledge. *Journal of Clinical Medicine*, *13*(8), 2381. https://doi.org/10.3390/jcm13082381

267. Van Hove, J. L. K., Friederich, M. W., Hock, D. H., Stroud, D. A., Caruana, N. J., Christians, U., Schniedewind, B., Michel, C. R., Reisdorph, R., Lopez Gonzalez, E. D. J., Brenner, C., Donovan, T. E., Lee, J. C., Chatfield, K. C., Larson, A. A., Baker, P. R., McCandless, S. E., & Moore Burk, M. F. (2024). ACAD9 treatment with bezafibrate and nicotinamide riboside temporarily stabilizes cardiomyopathy and lactic acidosis. *Mitochondrion*, *78*, 101905. https://doi.org/10.1016/j.mito.2024.101905

268. VanPortfliet, J. J., Lei, Y., Martinez, C. G., Wong, J., Pflug, K., Sitcheran, R., Kneeland, S. C., Murray, S. A., McGuire, P. J., Cannon, C. L., & West, A. P. (2024). Caspase-11 drives macrophage hyperinflammation in models of Polg-related mitochondrial disease. *bioRxiv: The Preprint Server for Biology*, 2024.05.11.593693. https://doi.org/10.1101/2024.05.11.593693

269. Vaz, F. M., Ferdinandusse, S., Salomons, G. S., & Wanders, R. J. A. (2024). Disorders of fatty acid homeostasis. *Journal of Inherited Metabolic Disease*. https://doi.org/10.1002/jimd.12734

270. Vela-Sebastián, A., Bayona-Bafaluy, P., & Pacheu-Grau, D. (2024). ISR pathway contribution to tissue specificity of mitochondrial diseases. *Trends in Endocrinology and Metabolism: TEM*, *35*(10), 851-853. https://doi.org/10.1016/j.tem.2024.05.001

271. Wai, T. (2024). Is mitochondrial morphology important for cellular physiology? *Trends in Endocrinology and Metabolism: TEM*, *35*(10), 854-871. https://doi.org/10.1016/j.tem.2024.05.005

272. Wal, P., Wal, A., Vig, H., Mahmood, D., & Khan, M. M. U. (2024). Potential Applications of Mitochondrial Therapy with a Focus on Parkinson's Disease and Mitochondrial Transplantation. *Advanced Pharmaceutical Bulletin*, *14*(1), 147-160. https://doi.org/10.34172/apb.2024.019

273. Wang, Y., Lilienfeldt, N., & Hekimi, S. (2024). Understanding coenzyme Q. *Physiological Reviews*, *104*(4), 1533-1610. https://doi.org/10.1152/physrev.00040.2023

274. Wang, Y., Shi, Y., Shao, Y., Lu, X., Zhang, H., & Miao, C. (2024). S100A8/A9hi neutrophils induce mitochondrial dysfunction and PANoptosis in endothelial cells via mitochondrial complex I deficiency during sepsis. *Cell Death & Disease*, *15*(6), 462. https://doi.org/10.1038/s41419-024-06849-6

275. Wang, Y., Yang, J.-S., Zhao, M., Chen, J.-Q., Xie, H.-X., Yu, H.-Y., Liu, N.-H., Yi, Z.-J., Liang, H.-L., Xing, L., & Jiang, H.-L. (2024). Mitochondrial endogenous substance transport-inspired nanomaterials for mitochondria-targeted gene delivery. *Advanced Drug Delivery Reviews*, *211*, 115355. https://doi.org/10.1016/j.addr.2024.115355

276. Wu, Z., Bezwada, D., Cai, F., Harris, R. C., Ko, B., Sondhi, V., Pan, C., Vu, H. S., Nguyen, P. T., Faubert, B., Cai, L., Chen, H., Martin-Sandoval, M., Do, D., Gu, W., Zhang, Y., Zhang, Y., Brooks, B., Kelekar, S., … DeBerardinis, R. J. (2024). Electron transport chain inhibition increases cellular dependence on purine transport and salvage. *Cell Metabolism*, *36*(7), 1504-1520.e9. https://doi.org/10.1016/j.cmet.2024.05.014

277. Xu, H., Ma, K., Gao, Y., Song, Q., Chen, C., Xu, X., Peng, J., & Sun, Y. (2024). Clinical characteristics of a case of multiple mitochondrial dysfunction syndrome 3. *Molecular Genetics & Genomic Medicine*, *12*(6), e2485. https://doi.org/10.1002/mgg3.2485

278. Xu, Y., Hong, Z., Yu, S., Huang, R., Li, K., Li, M., Xie, S., & Zhu, L. (2024). Fresh Insights Into SLC25A26 : Potential New Therapeutic Target for Cancers: A Review. *Oncology Reviews*, *18*, 1379323. https://doi.org/10.3389/or.2024.1379323

279. Yazicioglu, Y. F., Mitchell, R. J., & Clarke, A. J. (2024). Mitochondrial control of lymphocyte homeostasis. *Seminars in Cell & Developmental Biology*, *161-162*, 42-53. https://doi.org/10.1016/j.semcdb.2024.03.002

280. Yin, X., Dong, Q., Fan, S., Yang, L., Li, H., Jin, Y., Laurentinah, M. R., Chen, X., Sysa, A., Fang, H., Lyu, J., Yu, Y., & Wang, Y. (2024). A novel pathogenic mitochondrial DNA variant m.4344T>C in tRNAGln causes developmental delay. *Journal of Human Genetics*, *69*(8), 381-389. https://doi.org/10.1038/s10038-024-01254-5

281. Yuan, T., Kumar, S., Skinner, M. E., Victor-Joseph, R., Abuaita, M., Keijer, J., Zhang, J., Kunkel, T. J., Liu, Y., Petrunak, E. M., Saunders, T. L., Lieberman, A. P., Stuckey, J. A., Neamati, N., Al-Murshedi, F., Alfadhel, M., Spelbrink, J. N., Rodenburg, R., de Boer, V. C. J., & Lombard, D. B. (2024). Human SIRT5 variants with reduced stability and activity do not cause neuropathology in mice. *iScience*, *27*(6), 109991. https://doi.org/10.1016/j.isci.2024.109991

282. Zhai, N., Hu, J., & Yan, H. (2024). Recurrent mitochondrial disease caused by the "m.3243A> G" mutation : A case report. *Asian Journal of Surgery*, S1015-9584(24)01160-6. https://doi.org/10.1016/j.asjsur.2024.05.251

283. Zhang, F., Zhang, L., Hu, G., Chen, X., Liu, H., Li, C., Guo, X., Huang, C., Sun, F., Li, T., Cui, Z., Guo, Y., Yan, W., Xia, Y., Liu, Z., Lin, Z., Duan, W., Lu, L., Wang, X., … Tao, L. (2024). Rectifying METTL4-Mediated N6-Methyladenine Excess in Mitochondrial DNA Alleviates Heart Failure. *Circulation*, *150*(18), 1441-1458. https://doi.org/10.1161/CIRCULATIONAHA.123.068358

284. Zhou, W., Wang, N., Dong, S., Huan, Z., Sui, L., & Ge, X. (2024). PRG4 mitigates hemorrhagic shock-induced cardiac injury by inhibiting mitochondrial dysregulation, oxidative stress and NLRP3-mediated pyroptosis. *International Immunopharmacology*, *137*, 112507. https://doi.org/10.1016/j.intimp.2024.112507

285. Zhu, X.-X., Meng, X.-Y., Zhang, A.-Y., Zhao, C.-Y., Chang, C., Chen, T.-X., Huang, Y.-B., Xu, J.-P., Fu, X., Cai, W.-W., Hou, B., Du, B., Zheng, G.-L., Zhang, J.-R., Lu, Q.-B., Bai, N., Han, Z.-J., Bao, N., Qiu, L.-Y., & Sun, H.-J. (2024). Vaccarin alleviates septic cardiomyopathy by potentiating NLRP3 palmitoylation and inactivation. *Phytomedicine: International Journal of Phytotherapy and Phytopharmacology*, *131*, 155771. https://doi.org/10.1016/j.phymed.2024.155771