

L'essentielle Maladies Mitochondriales



1^{er} trimestre 2024



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MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

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Ataxie de Friedreich - *Friedreich ataxia*

1. Ahmad, I., Kamai, A., Zahra, S., Kapoor, H., Kumar Srivastava, A., & Faruq, M. (2024). Generation and characterization of iPSC lines from Friedreich's ataxia patient (FRDA) with GAA.TTC repeat expansion in the Frataxin (FXN) gene's first intron (IGIBi016-A) and a non-FRDA healthy control individual (IGIBi017-A). *Stem Cell Research*, 77, 103382. <https://doi.org/10.1016/j.scr.2024.103382>
2. Ahmad, I., Kapoor, H., Srivastava, A. K., & Faruq, M. (2024). Generation of two human induced pluripotent stem cell lines, IGIBi012-A and IGIBi013-A from Friedreich's ataxia (FRDA) patients with homozygous GAA repeat expansion in FXN gene. *Stem Cell Research*, 76, 103340. <https://doi.org/10.1016/j.scr.2024.103340>
3. Ast, T., Itoh, Y., Sadre, S., McCoy, J. G., Namkoong, G., Wengrod, J. C., Chicherin, I., Joshi, P. R., Kamenski, P., Suess, D. L. M., Amunts, A., & Mootha, V. K. (2024). METTL17 is an Fe-S cluster checkpoint for mitochondrial translation. *Molecular Cell*, 84(2), 359-374.e8. <https://doi.org/10.1016/j.molcel.2023.12.016>
4. Casey, H. L., Shah, V. V., Muzyka, D., McNames, J., El-Gohary, M., Sowalsky, K., Safarpour, D., Carlson-Kuhta, P., Schmahmann, J. D., Rosenthal, L. S., Perlman, S., Rummey, C., Horak, F. B., & Gomez, C. M. (2024). Standing Balance Conditions and Digital Sway Measures for Clinical Trials of Friedreich's Ataxia. *Movement Disorders: Official Journal of the Movement Disorder Society*, 39(6), 996-1005. <https://doi.org/10.1002/mds.29777>
5. Chang, J. C., Ryan, M. R., Stark, M. C., Liu, S., Purushothaman, P., Bolan, F., Johnson, C. A., Champe, M., Meng, H., Lawlor, M. W., Halawani, S., Ngaba, L. V., Lynch, D. R., Davis, C., Gonzalo-Gil, E., Lutz, C., Urbinati, F., Medicherla, B., & Fonck, C. (2024). AAV8 gene therapy reverses cardiac pathology and prevents early mortality in a mouse model of Friedreich's ataxia. *Molecular Therapy. Methods & Clinical Development*, 32(1), 101193. <https://doi.org/10.1016/j.omtm.2024.101193>
6. Cilenti, N. A., Tamaroff, J. G., Capiola, C. J., Faig, W., McBride, M. G., Paridon, S. M., O'Malley, S., Edelson, J. B., Lynch, D. R., McCormack, S. E., & Lin, K. Y. (2024). Cardiopulmonary exercise testing on adaptive equipment in children and adults with Friedreich ataxia. *Muscle & Nerve*, 69(5), 613-619. <https://doi.org/10.1002/mus.28085>
7. Clayton, R., Galas, T., Scherer, N., Farmer, J., Ruiz, N., Hamdani, M., Schecter, D., & Bettoun, D. (2024). Safety, pharmacokinetics, and pharmacodynamics of nomlabofusp (CTI-1601) in Friedreich's ataxia. *Annals of Clinical and Translational Neurology*, 11(3), 540-553. <https://doi.org/10.1002/acn3.51971>

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8. Di Pietro, G., Cioffi, E., Falco, P., Galosi, E., De Stefano, G., Di Stefano, G., Leone, C., Martines, V., Perotti, S., Casali, C., & Truini, A. (2024). Nerve ultrasound in Friedreich's Ataxia : Enlarged nerves as a biomarker of disease severity. *Clinical Neurophysiology: Official Journal of the International Federation of Clinical Neurophysiology*, 159, 75-80. <https://doi.org/10.1016/j.clinph.2024.01.004>
9. Dong, Y. N., Ngaba, L. V., An, J., Adeshina, M. W., Warren, N., Wong, J., & Lynch, D. R. (2024). A peptide derived from TID1S rescues frataxin deficiency and mitochondrial defects in FRDA cellular models. *Frontiers in Pharmacology*, 15, 1352311. <https://doi.org/10.3389/fphar.2024.1352311>
10. Doni, D., Cavallari, E., Noguera, M. E., Gentili, H. G., Cavion, F., Parisi, G., Fornasari, M. S., Sartori, G., Santos, J., Bellanda, M., Carbonera, D., Costantini, P., & Bortolus, M. (2024). Searching for Frataxin Function : Exploring the Analogy with Nqo15, the Frataxin-like Protein of Respiratory Complex I from *Thermus thermophilus*. *International Journal of Molecular Sciences*, 25(3), 1912. <https://doi.org/10.3390/ijms25031912>
11. Doni, D., Cavion, F., Bortolus, M., Baschiera, E., Muccioli, S., Tombesi, G., d'Ettorre, F., Ottaviani, D., Marchesan, E., Leanza, L., Greggio, E., Ziviani, E., Russo, A., Bellin, M., Sartori, G., Carbonera, D., Salviati, L., & Costantini, P. (2024). Correction : Human frataxin, the Friedreich ataxia deficient protein, interacts with mitochondrial respiratory chain. *Cell Death & Disease*, 15(1), 93. <https://doi.org/10.1038/s41419-024-06459-2>
12. Edzeamey, F. J., Ramchunder, Z., Pourzand, C., & Anjomani Virumouni, S. (2024). Emerging antioxidant therapies in Friedreich's ataxia. *Frontiers in Pharmacology*, 15, 1359618. <https://doi.org/10.3389/fphar.2024.1359618>
13. Fernández, A. G. (2024). Vestibular Pathology as Early Finding of Friedreich's Ataxia in a 16 Years Old Woman. *Indian Journal of Otolaryngology and Head and Neck Surgery: Official Publication of the Association of Otolaryngologists of India*, 76(1), 1247-1250. <https://doi.org/10.1007/s12070-023-04249-4>
14. Hopf, S., Tüscher, O., & Schuster, A. K. (2024). [Retinal OCT biomarkers and neurodegenerative diseases of the central nervous system beyond Alzheimer's disease]. *Die Ophthalmologie*, 121(2), 93-104. <https://doi.org/10.1007/s00347-023-01974-7>

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15. Iskusnykh, I. Y., Zakharova, A. A., Kryl'skii, E. D., & Popova, T. N. (2024). Aging, Neurodegenerative Disorders, and Cerebellum. *International Journal of Molecular Sciences*, 25(2), 1018. <https://doi.org/10.3390/ijms25021018>
16. Johnsson, M., Zetterberg, H., Blennow, K., & Lindberg, C. (2024). Clinical stage and plasma neurofilament concentration in adults with Friedreich ataxia. *Heliyon*, 10(1), e23347. <https://doi.org/10.1016/j.heliyon.2023.e23347>
17. Koeppen, A. H. (2024). Tissue Iron in Friedreich Ataxia. *Journal of Integrative Neuroscience*, 23(1), 4. <https://doi.org/10.31083/j.jin2301004>
18. Lynch, D. R., Perlman, S., & Schadt, K. (2024). Omaveloxolone for the treatment of Friedreich ataxia : Clinical trial results and practical considerations. *Expert Review of Neurotherapeutics*, 24(3), 251-258. <https://doi.org/10.1080/14737175.2024.2310617>
19. Lynch, D. R., Subramony, S., Lin, K. Y., Mathews, K., Perlman, S., Yoon, G., & Rummey, C. (2024). Characterization of Cardiac-Onset Initial Presentation in Friedreich Ataxia. *Pediatric Cardiology*. <https://doi.org/10.1007/s00246-024-03429-5>
20. Machado, D. S., Viana, C. F., Pedroso, J. L., Barsottini, O. G. P., Tomaselli, P. J., Marques, W., Rezende, T. J. R., Martinez, A. R. M., & França, M. C. (2024). Prevalence and Diagnostic Journey of Friedreich's Ataxia in the State of São Paulo, Brazil. *Cerebellum (London, England)*, 23(5), 1916-1922. <https://doi.org/10.1007/s12311-024-01687-w>
21. Malina, J., Huessler, E.-M., Jöckel, K.-H., Boog-Whiteside, E., Jeschonneck, N., Schröder, B., Schüle, R., Köhl, T., & Klebe, S. (2024). Development and validation of TreatHSP-QoL : A patient-reported outcome measure for health-related quality of life in hereditary spastic paraplegia. *Orphanet Journal of Rare Diseases*, 19(1), 2. <https://doi.org/10.1186/s13023-023-03012-w>
22. Maltby, C. J., Krans, A., Grudzien, S. J., Palacios, Y., Muiños, J., Suárez, A., Asher, M., Khurana, V., Barmada, S. J., Dijkstra, A. A., & Todd, P. K. (2023). AAGGG repeat expansions trigger RFC1-independent synaptic dysregulation in human CANVAS Neurons. *bioRxiv: The Preprint Server for Biology*, 2023.12.13.571345. <https://doi.org/10.1101/2023.12.13.571345>
23. Mishra, P., Sivakumar, A., Johnson, A., Pernaci, C., Warden, A. S., El-Hachem, L. R., Hansen, E., Badell-Grau, R. A., Khare, V., Ramirez, G., Gillette, S., Solis, A. B., Guo, P., Coufal, N., & Cherqui, S. (2024). Gene editing improves endoplasmic reticulum-mitochondrial contacts and unfolded protein response in Friedreich's ataxia iPSC-derived neurons. *Frontiers in Pharmacology*, 15, 1323491. <https://doi.org/10.3389/fphar.2024.1323491>

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24. Pelosi, L., Mulroy, E., Leadbetter, R., Rodrigues, M., & Roxburgh, R. (2024). Unique combinations of ultrasound and electrophysiological findings distinguish Friedreich's ataxia from other inherited ataxias. *Clinical Neurophysiology: Official Journal of the International Federation of Clinical Neurophysiology*, 161, 157-158. <https://doi.org/10.1016/j.clinph.2024.02.035>
25. Pietrangeli, P., Marcocci, L., Pennacchietti, V., Diop, A., Di Felice, M., Pagano, L., Malagrino, F., Toto, A., Brunori, M., & Gianni, S. (2024). The Mechanism of Folding of Human Frataxin in Comparison to the Yeast Homologue—Broad Energy Barriers and the General Properties of the Transition State. *Journal of Molecular Biology*, 436(10), 168555. <https://doi.org/10.1016/j.jmb.2024.168555>
26. Pilotto, F., Chellapandi, D. M., & Puccio, H. (2024). Omaveloxolone : A groundbreaking milestone as the first FDA-approved drug for Friedreich ataxia. *Trends in Molecular Medicine*, 30(2), 117-125. <https://doi.org/10.1016/j.molmed.2023.12.002>
27. Retraction : Very late-onset Friedreich's ataxia with rapid course mimicking as possible multiple system atrophy cerebellar type. (2024). *BMJ Case Reports*, 17(3), e242073.ret. <https://doi.org/10.1136/bcr-2021-242073.ret>
28. Rojsajakul, 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. (2023). Expression and processing of mature human frataxin after gene therapy in mice. *Research Square*, rs.3.rs-3788652. <https://doi.org/10.21203/rs.3.rs-3788652/v1>
29. Shen, M. M., Rummey, C., & Lynch, D. R. (2024). Phenotypic variation of FXN compound heterozygotes in a Friedreich ataxia cohort. *Annals of Clinical and Translational Neurology*, 11(5), 1110-1121. <https://doi.org/10.1002/acn3.52027>
30. Smith, F. M., & Kosman, D. J. (2023). Loss of filamentous actin, tight junction protein expression, and paracellular barrier integrity in frataxin-deficient human brain microvascular endothelial cells-implications for blood-brain barrier physiology in Friedreich's ataxia. *Frontiers in Molecular Biosciences*, 10, 1299201. <https://doi.org/10.3389/fmolb.2023.1299201>
31. Stavros, K. (2024). Genetic Myelopathies. *Continuum (Minneapolis, Minn.)*, 30(1), 119-132. <https://doi.org/10.1212/CON.0000000000001377>
32. Stovickova, L., Hansikova, H., Hanzalova, J., Musova, Z., Semjonov, V., Stovicek, P., Hadzic, H., Novotna, L., Simcik, M., Strnad, P., Serbina, A., Karamazovova, S., Schwabova Paulasova, J., Vyhnaek, M., Krsek, P., & Zumrova, A. (2024). Exploring mitochondrial biomarkers for Friedreich's ataxia : A multifaceted approach. *Journal of Neurology*, 271(6), 3439-3454. <https://doi.org/10.1007/s00415-024-12223-5>

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33. Trantham, S. J., Coker, M. A., Norman, S., Crowley, E., Berthy, J., Byrne, B. J., Subramony, S., Lou, X., & Corti, M. (2024). Perspectives of the Friedreich ataxia community on gene therapy clinical trials. *Molecular Therapy. Methods & Clinical Development*, 32(1), 101179. <https://doi.org/10.1016/j.omtm.2023.101179>
34. Traschütz, A., Fleszar, Z., Hengel, H., Klockgether, T., Erdlenbruch, F., Falkenburger, B. H., Klopstock, T., Öztöç-Çakmak, Ö., Pedrosa, J. L., Santorelli, F. M., Schöls, L., RFC1 Study Group, PREPARE Consortium, & Synofzik, M. (2024). FARS-ADL across Ataxias : Construct Validity, Sensitivity to Change, and Minimal Important Change. *Movement Disorders: Official Journal of the Movement Disorder Society*, 39(6), 965-974. <https://doi.org/10.1002/mds.29788>

Atrophie optique autosomique dominante – ADOA

35. Affortit, C., Coyat, C., Saidia, A. R., Ceccato, J.-C., Charif, M., Sarzi, E., Flamant, F., Guyot, R., Cazevielle, C., Puel, J.-L., Lenaers, G., & Wang, J. (2024). The human OPA1delTTAG mutation induces adult onset and progressive auditory neuropathy in mice. *Cellular and Molecular Life Sciences: CMLS*, 81(1), 80. <https://doi.org/10.1007/s00018-024-05115-4>
36. Amore, G., Romagnoli, M., Carbonelli, M., Cascavilla, M. L., De Negri, A. M., Carta, A., Parisi, V., Di Renzo, A., Schiavi, C., Lenzetti, C., Zenesini, C., Ormanbekova, D., Palombo, F., Fiorini, C., Caporali, L., Carelli, V., Barboni, P., & La Morgia, C. (2024). AFG3L2 and ACO2-Linked Dominant Optic Atrophy: Genotype-Phenotype Characterization Compared to OPA1 Patients. *American Journal of Ophthalmology*, 262, 114-124. <https://doi.org/10.1016/j.ajo.2024.01.011>
37. Battista, M., Coutinho, C. P., Berni, A., Borrelli, E., Galzignato, A., Lari, G., Checchin, L., Pizza, I. C., Brotto, L., Nucci, P., Bandello, F., Cascavilla, M. L., & Barboni, P. (2024). Sectorial Ganglion Cell Complex Thickness as Biomarker of Vision Outcome in Patients With Dominant Optic Atrophy. *Investigative Ophthalmology & Visual Science*, 65(1), 24. <https://doi.org/10.1167/iovs.65.1.24>
38. Gilhooley, M. J., Raoof, N., Yu-Wai-Man, P., & Moosajee, M. (2024). Inherited Optic Neuropathies : Real-World Experience in the Paediatric Neuro-Ophthalmology Clinic. *Genes*, 15(2), 188. <https://doi.org/10.3390/genes15020188>
39. Ladero, M., Reche-Sainz, J. A., & Gallardo, M. E. (2024). Hereditary Optic Neuropathies : A Systematic Review on the Interplay between Biomaterials and Induced Pluripotent Stem Cells. *Bioengineering (Basel, Switzerland)*, 11(1), 52. <https://doi.org/10.3390/bioengineering11010052>

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40. Ophthalmology Group of China Alliance for Rare Diseases/Beijing Society of Rare Disease Clinical Care and Accessibility & Neuro-ophthalmology Group of Ophthalmology Branch of Chinese Medical Association. (2024). [Chinese expert consensus on the clinical diagnosis and treatment of autosomal dominant optic atrophy (2024)]. *[Zhonghua Yan Ke Za Zhi] Chinese Journal of Ophthalmology*, 60(3), 226-233. <https://doi.org/10.3760/cma.j.cn112142-20231225-00308>
41. Shimabukuro, W., Chinen, Y., Imanaga, N., Yanagi, K., Kaname, T., & Nakanishi, K. (2024). Renal coloboma syndrome/dominant optic atrophy with severe retinal atrophy and de novo digenic mutations in PAX2 and OPA1. *Pediatric Nephrology (Berlin, Germany)*, 39(8), 2351-2353. <https://doi.org/10.1007/s00467-024-06347-z>
42. Zanfardino, P., Amati, A., Doccini, S., Cox, S. N., Tullo, A., Longo, G., D'Erchia, A., Picardi, E., Nesti, C., Santorelli, F. M., & Petruzzella, V. (2024). OPA1 mutation affects autophagy and triggers senescence in autosomal dominant optic atrophy plus fibroblasts. *Human Molecular Genetics*, 33(9), 768-786. <https://doi.org/10.1093/hmg/ddae008>

Encéphalopathie myo-neuro-gastrointestinale - MNGIE

43. Kubal, C. A., Mihaylov, P., Snook, R., Soma, D., Saeed, O., Rokop, Z., Lacerda, M., Graham, B. H., & Mangus, R. S. (2024). Successful Sequential Liver and Isolated Intestine Transplantation for Mitochondrial Neurogastrointestinal Encephalopathy Syndrome: A Case Report. *Annals of Transplantation*, 29, e941881. <https://doi.org/10.12659/AOT.941881>
44. Redha, N., Al-Sahlawi, Z., Hasan, H., Ghareeb, S., & Humaidan, H. (2024). Meningoencephalitis in a novel mutation in MNGIE (mitochondrial neurogastrointestinal encephalomyopathy) ending a familial diagnostic odyssey: A case series report. *Journal of Central Nervous System Disease*, 16, 11795735241241423. <https://doi.org/10.1177/11795735241241423>
45. Rust, D., Martinez, M., Lagana, S., Hirano, M., Kato, T., & Weiner, J. (2024). Multivisceral transplantation for the treatment of mitochondrial neurogastrointestinal encephalomyopathy. *Clinical Transplantation*, 38(1), e15230. <https://doi.org/10.1111/ctr.15230>

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

46. Alberti, C., Rizzo, F., Anastasia, A., Comi, G., Corti, S., & Abati, E. (2024). Charcot-Marie-tooth disease type 2A: An update on pathogenesis and therapeutic perspectives. *Neurobiology of Disease*, 193, 106467. <https://doi.org/10.1016/j.nbd.2024.106467>
47. Armirola-Ricaurte, C., Zonnekein, N., Koutsis, G., Amor-Barris, S., Pelayo-Negro, A. L., Atkinson, D., Efthymiou, S., Turchetti, V., Dinopoulos, A., Garcia, A., Karakaya, M., Moris, G., Polat, A. I., Yiş, U., Espinos, C., Van de Vondel, L., De Vriendt, E., Karadima, G., Wirth, B., ... Jordanova, A. (2024). Alternative splicing expands the clinical spectrum of NDUF56-related mitochondrial disorders. *Genetics in Medicine: Official Journal of the American College of Medical Genetics*, 26(6), 101117. <https://doi.org/10.1016/j.gim.2024.101117>
48. Cao, L., Yang, J., Zhang, X., Wang, X., Chen, Z., Tan, S., & Yang, J. (2023). Clinical, neurophysiological evaluation and genetic features of axonal Charcot-Marie-Tooth disease in a Chinese family. *Frontiers in Neurology*, 14, 1337065. <https://doi.org/10.3389/fneur.2023.1337065>
49. Doherty, C. M., Morrow, J. M., Zuccarino, R., Howard, P., Wastling, S., Pipis, M., Zafeiropoulos, N., Stephens, K. J., Grider, T., Feely, S. M. E., Nopoulous, P., Skorupinska, M., Milev, E., Nicolaisen, E., Dudzeic, M., McDowell, A., Dilek, N., Muntoni, F., Rossor, A. M., ... Reilly, M. M. (2024). Lower limb muscle MRI fat fraction is a responsive outcome measure in CMT X1, 1B and 2A. *Annals of Clinical and Translational Neurology*, 11(3), 607-617. <https://doi.org/10.1002/acn3.51979>
50. Elbert, A., Dixon, K., Shen, Y., Hamilton, S., Boerkoel, C. F., Jones, S. J., & Kanungo, A. K. (2024). Mitofusin 2 Variant Presenting With a Phenotype of Multiple System Atrophy of Cerebellar Subtype. *Neurology. Genetics*, 10(1), e200114. <https://doi.org/10.1212/NXG.0000000000200114>
51. Ferreira, T., Polavarapu, K., Olimpico, C., Paramonov, I., Lochmüller, H., & Horvath, R. (2024). Variants in mitochondrial disease genes are common causes of inherited peripheral neuropathies. *Journal of Neurology*, 271(6), 3546-3553. <https://doi.org/10.1007/s00415-024-12319-y>
52. Figueiredo, F. B., Tomaselli, P. J., Hallak, J., Mattiello-Sverzut, A. C., Covaleski, A. P. P. M., Sobreira, C. F. da R., de Paula Gouvêa, S., & Marques, W. (2024). Genetic diversity in hereditary axonal neuropathy: Analyzing 53 Brazilian children. *Journal of the Peripheral Nervous System: JPNS*, 29(1), 97-106. <https://doi.org/10.1111/jns.12617>

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53. Gangfuß, A., Rating, P., Ferreira, T., Hentschel, A., Marina, A. D., Kölbel, H., Sickmann, A., Abicht, A., Kraft, F., Ruck, T., Böhm, J., Schänzer, A., Schara-Schmidt, U., Neuhaus, T. M., Horvath, R., & Roos, A. (2024). A Homozygous NDUF6 Variant Associated with Neuropathy and Optic Atrophy. *Journal of Neuromuscular Diseases*, 11(2), 485-491. <https://doi.org/10.3233/JND-230181>
54. Geroldi, A., Ponti, C., Mammi, A., Patrone, S., Gotta, F., Trevisan, L., Sanguineri, F., Origone, P., Gaudio, A., La Barbera, A., Cataldi, M., Gemelli, C., Massucco, S., Schenone, A., Lanteri, P., Fiorillo, C., Grandis, M., Mandich, P., & Bellone, E. (2024). Early Onset Inherited Peripheral Neuropathies : The Experience of a Specialized Referral Center for Genetic Diagnosis Achievement. *Pediatric Neurology*, 154, 4-8. <https://doi.org/10.1016/j.pediatrneurol.2024.02.002>
55. Record, C. J., Pipis, M., Skorupinska, M., Blake, J., Poh, R., Polke, J. M., Eggleton, K., Nanji, T., Zuchner, S., Cortese, A., Houlden, H., Rossor, A. M., Laura, M., & Reilly, M. M. (2024). Whole genome sequencing increases the diagnostic rate in Charcot-Marie-Tooth disease. *Brain: A Journal of Neurology*, 147(9), 3144-3156. <https://doi.org/10.1093/brain/awae064>
56. Record, C. J., & Reilly, M. M. (2024). Lessons and pitfalls of whole genome sequencing. *Practical Neurology*, 24(4), 263-274. <https://doi.org/10.1136/pn-2023-004083>
57. Sharma, G., Zaman, M., Sabouny, R., Joel, M., Martens, K., Martino, D., de Koning, A. P. J., Pfeffer, G., & Shutt, T. E. (2021). Characterization of a novel variant in the HR1 domain of MFN2 in a patient with ataxia, optic atrophy and sensorineural hearing loss. *F1000Research*, 10, 606. <https://doi.org/10.12688/f1000research.53230.2>
58. Sun, X., Liu, X., Zhao, Q., Zhang, L., & Yuan, H. (2023). Quantified fat fraction as biomarker assessing disease severity in rare Charcot-Marie-Tooth subtypes. *Frontiers in Neurology*, 14, 1334976. <https://doi.org/10.3389/fneur.2023.1334976>
59. Xu, X., Lu, F., Du, S., & Zhang, L. (2024). A case report of peroneal muscle atrophy type 2A2 with central nervous system involvement as initial presentation. *BMC Pediatrics*, 24(1), 21. <https://doi.org/10.1186/s12887-023-04441-z>



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Maladies liées au gène ACO2 - ACO2-related disorders

60. Amore, G., Romagnoli, M., Carbonelli, M., Cascavilla, M. L., De Negri, A. M., Carta, A., Parisi, V., Di Renzo, A., Schiavi, C., Lenzetti, C., Zenesini, C., Ormanbekova, D., Palombo, F., Fiorini, C., Caporali, L., Carelli, V., Barboni, P., & La Morgia, C. (2024). AFG3L2 and ACO2-Linked Dominant Optic Atrophy: Genotype-Phenotype Characterization Compared to OPA1 Patients. *American Journal of Ophthalmology*, 262, 114-124. <https://doi.org/10.1016/j.ajo.2024.01.011>

Maladie liée au gène GFM1 – GFM1-related disorder

61. Aleksic, D., Jankovic, M. G., Todorovic, S., Kovacevic, M., & Borkovic, M. (2023). The first case of combined oxidative phosphorylation deficiency-1 due to a GFM1 mutation in the Serbian population : A case report and literature review. *The Turkish Journal of Pediatrics*, 65(6), 1018-1024. <https://doi.org/10.24953/turkjp.2022.1082>
62. Gao, A. W., Alam, G. E., Zhu, Y., Li, W., Katsyuba, E., Sulc, J., Li, T. Y., Li, X., Overmyer, K. A., Lalou, A., Mouchiroud, L., Sleiman, M. B., Cornaglia, M., Morel, J.-D., Houtkooper, R. H., Coon, J. J., & Auwerx, J. (2024). High-content phenotypic analysis of a C. elegans recombinant inbred population identifies genetic and molecular regulators of lifespan. *bioRxiv: The Preprint Server for Biology*, 2024.01.15.575638. <https://doi.org/10.1101/2024.01.15.575638>
63. Nayan, A., Mehta, S., Chakravarty, K., Mehta, S., & Lal, V. (2024). Generalized Dystonia as a Cardinal Manifestation of Combined Oxidative Phosphorylation Deficiency 1. *Movement Disorders Clinical Practice*, 11(6), 742-745. <https://doi.org/10.1002/mdc3.14027>



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

64. Beecher, G., Gavrilova, R. H., Mandrekar, J., & Naddaf, E. (2024). Mitochondrial myopathies diagnosed in adulthood: Clinico-genetic spectrum and long-term outcomes. *Brain Communications*, 6(2), fcae041. <https://doi.org/10.1093/braincomms/fcae041>
65. Chen, A., Yangzom, T., Hong, Y., Lundberg, B. C., Sullivan, G. J., Tzoulis, C., Bindoff, L. A., & Liang, K. X. (2024). Hallmark Molecular and Pathological Features of POLG Disease are Recapitulated in Cerebral Organoids. *Advanced Science (Weinheim, Baden-Wurttemberg, Germany)*, 11(18), e2307136. <https://doi.org/10.1002/advs.202307136>
66. Chen, Y., Ma, G., Gai, Y., Yang, Q., Liu, X., de Avila, J. M., Mao, S., Zhu, M.-J., & Du, M. (2024). AMPK Suppression Due to Obesity Drives Oocyte mtDNA Heteroplasmy via ATF5-POLG Axis. *Advanced Science (Weinheim, Baden-Wurttemberg, Germany)*, 11(20), e2307480. <https://doi.org/10.1002/advs.202307480>
67. Cohen, B. H., Chinnery, P. F., & Copeland, W. C. (1993). POLG-Related Disorders. 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/NBK26471/>
68. Hong, Y., Zhang, Z., Yangzom, T., Chen, A., Lundberg, B. C., Fang, E. F., Siller, R., Sullivan, G. J., Zeman, J., Tzoulis, C., Bindoff, L. A., & Liang, K. X. (2024). The NAD⁺ Precursor Nicotinamide Riboside Rescues Mitochondrial Defects and Neuronal Loss in iPSC derived Cortical Organoid of Alpers' Disease. *International Journal of Biological Sciences*, 20(4), 1194-1217. <https://doi.org/10.7150/ijbs.91624>
69. Liu, H., Gao, M., Sun, Q., Chen, S., Luo, Y., Yang, H., Li, Q., Li, J., & Yang, G. (2023). A case of mitochondrial myopathy and chronic progressive external ophthalmoplegia. *Zhong Nan Da Xue Xue Bao. Yi Xue Ban = Journal of Central South University. Medical Sciences*, 48(11), 1760-1768. <https://doi.org/10.11817/j.issn.1672-7347.2023.220605>
70. Reitering, J. C., & Mackay, D. D. (2024). Optic Neuropathy Associated with POLG Mutations : A Case Series and Literature Review. *Journal of Neuro-Ophthalmology: The Official Journal of the North American Neuro-Ophthalmology Society*. <https://doi.org/10.1097/WNO.0000000000002089>

L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

71. Saha, D., Kothari, S., Kulkarni, S. D., Thambiraja, M., Yennamalli, R. M., & Das, D. K. (2024). Genetic heterogeneity and respiratory chain enzyme analysis in pediatric Indian patients with mitochondrial disorder : Report of novel variants in POLG1 gene and their functional implication using molecular dynamic simulation. *Mitochondrion*, 76, 101870. <https://doi.org/10.1016/j.mito.2024.101870>

MELAS

72. Acquaah, J., Ferdinand, P., & Roffe, C. (2024). Adult-onset mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes (MELAS) : A diagnostic challenge. *BMJ Case Reports*, 17(2), e256306. <https://doi.org/10.1136/bcr-2023-256306>
73. Ambrogetti, R., Kavanagh, E., & ElTayeb, K. (2024). Late-onset mitochondrial encephalopathy with lactic acidosis and stroke-like episodes and the role of serial imaging. *BMJ Case Reports*, 17(2), e259102. <https://doi.org/10.1136/bcr-2023-259102>
74. Anders, A. G., Tidwell, E. D., Gadkari, V. V., Koutmos, M., & Ruotolo, B. T. (2024). Collision-Induced Unfolding Reveals Disease-Associated Stability Shifts in Mitochondrial Transfer Ribonucleic Acids. *Journal of the American Chemical Society*, 146(7), 4412-4420. <https://doi.org/10.1021/jacs.3c09230>
75. Beecher, G., Gavrilova, R. H., Mandrekar, J., & Naddaf, E. (2024). Mitochondrial myopathies diagnosed in adulthood: Clinico-genetic spectrum and long-term outcomes. *Brain Communications*, 6(2), fcae041. <https://doi.org/10.1093/braincomms/fcae041>
76. Du, Y., Zhu, J., Guo, Z., Wang, Z., Wang, Y., Hu, M., Zhang, L., Yang, Y., Wang, J., Huang, Y., Huang, P., Chen, M., Chen, B., & Yang, C. (2024). Metformin adverse event profile : A pharmacovigilance study based on the FDA Adverse Event Reporting System (FAERS) from 2004 to 2022. *Expert Review of Clinical Pharmacology*, 17(2), 189-201. <https://doi.org/10.1080/17512433.2024.2306223>
77. Ferreira, F., Gonçalves Bacelar, C., Lisboa-Gonçalves, P., Paulo, N., Quental, R., Nunes, A. T., Silva, R., & Tavares, I. (2023). Renal manifestations in adults with mitochondrial disease from the mtDNA m.3243A>G pathogenic variant. *Nefrologia*, 43 Suppl 2, 1-7. <https://doi.org/10.1016/j.nefro.2024.01.017>
78. Finsterer, J. (2024). Assessing the effect of oral glutamine on the MELAS phenotype requires appropriate study designs. *Neuroradiology*, 66(4), 459-460. <https://doi.org/10.1007/s00234-024-03284-4>



L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

79. Finsterer, J., & Mehri, S. (2024). Before blaming metformin as a trigger for stroke-like lesions in MELAS, alternative etiologies must be off the table. *Neurological Sciences: Official Journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology*, 45(7), 3523-3524. <https://doi.org/10.1007/s10072-024-07444-5>
80. Guerrero-Molina, M. P., & González de la Aleja, J. (2024). Reply : Assessing the effect of oral glutamine on the MELAS phenotype requires appropriate study designs. *Neuroradiology*, 66(4), 461-462. <https://doi.org/10.1007/s00234-024-03299-x>
81. Hämmerl, L., & Kraya, T. (2024). [Migraine and mitochondrial diseases : Energy deficit in the brain]. *Der Nervenarzt*, 95(2), 169-178. <https://doi.org/10.1007/s00115-024-01622-8>
82. Horná, S., Péč, M. J., Krivuš, J., Michalová, R., Sivák, Š., Galajda, P., & Mokáň, M. (2024). Gastrointestinal Complications of Mitochondrial Encephalomyopathy, Lactic Acidosis and Stroke-Like Episodes (MELAS) Syndrome Managed By Parenteral Nutrition. *European Journal of Case Reports in Internal Medicine*, 11(2), 004268. https://doi.org/10.12890/2024_004268
83. Kelly, C., Junker, A., Englestad, K., Hirano, M., Trumpff, C., & Picard, M. (2024). Perceived association of mood and symptom severity in adults with mitochondrial diseases. *medRxiv: The Preprint Server for Health Sciences*, 2024.02.02.24302076. <https://doi.org/10.1101/2024.02.02.24302076>
84. Kurzawa-Akanbi, M., Tzoumas, N., Corral-Serrano, J. C., Guarascio, R., Steel, D. H., Cheetham, M. E., Armstrong, L., & Lako, M. (2024). Pluripotent stem cell-derived models of retinal disease : Elucidating pathogenesis, evaluating novel treatments, and estimating toxicity. *Progress in Retinal and Eye Research*, 100, 101248. <https://doi.org/10.1016/j.preteyeres.2024.101248>
85. Lin, Y.-H., Lin, K.-L., Wang, X.-W., Lee, J.-J., Wang, F.-S., Wang, P.-W., Lan, M.-Y., Liou, C.-W., & Lin, T.-K. (2024). Miro1 improves the exogenous engraftment efficiency and therapeutic potential of mitochondria transfer using Wharton's jelly mesenchymal stem cells. *Mitochondrion*, 76, 101856. <https://doi.org/10.1016/j.mito.2024.101856>
86. Pia, S., & Lui, F. (2024). Melas Syndrome. In *StatPearls*. StatPearls Publishing. <http://www.ncbi.nlm.nih.gov/books/NBK532959/>
87. Sakamoto, N., Hamada, S., Takahashi, H., Satou, R., Suzuki, M., & Maeno, T. (2023). Improvement of Intestinal Pseudo-Obstruction by Total Parenteral Nutrition in a Young Woman With Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, and Stroke-Like Episodes : A Case Report. *Cureus*, 15(12), e50075. <https://doi.org/10.7759/cureus.50075>

L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

88. Shin, H. J., Na, J.-H., & Lee, Y.-M. (2024). A case of exacerbated encephalopathy with stroke-like episodes and lactic acidosis triggered by metformin in a patient with MELAS. *Neurological Sciences: Official Journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology*, 45(5), 2337-2339. <https://doi.org/10.1007/s10072-024-07343-9>
89. Sun, Y.-C., Chou, Y.-P., Ho, P.-H., Chen, X.-X., Chen, P.-Y., Chu, C.-H., & Lin, H.-C. (2024). Bilateral cochlear implants in a MELAS patient. *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(6), 3265-3268. <https://doi.org/10.1007/s00405-024-08532-0>
90. Walker, M. A., Li, S., Livak, K. J., Karaa, A., Wu, C. J., & Mootha, V. K. (2024). T cell activation contributes to purifying selection against the MELAS-associated m.3243A>G pathogenic variant in blood. *Journal of Inherited Metabolic Disease*, 47(4), 757-765. <https://doi.org/10.1002/jimd.12726>
91. Zhao, Y., Xu, Z., Yan, C., & Ji, K. (2024). Unraveling the Diagnostic Puzzle : Minor Stroke-Like Lesions and Normal Muscle Histopathology in MELAS Syndrome. *Stroke*, 55(4), e127-e130. <https://doi.org/10.1161/STROKEAHA.123.045984>
92. Zijun, L., Xu, Y., Yujia, Y., & Zhiqiang, X. (2024). Elderly onset of MELAS carried an M.3243A >G mutation in a female with deafness and visual deficits : A case report. *Clinical Case Reports*, 12(3), e8438. <https://doi.org/10.1002/ccr3.8438>

MERFF

93. Carmona Alexandrino, H., Ferreira, M. A., Ramalho, D., Jesus, N. R., & Oliveira, M. J. (2023). Endocrine Challenges in Myoclonic Epilepsy With Ragged Red Fibers Syndrome : A Case Report. *Cureus*, 15(12), e51114. <https://doi.org/10.7759/cureus.51114>
94. Zimmern, V., & Minassian, B. (2024). Progressive Myoclonus Epilepsy : A Scoping Review of Diagnostic, Phenotypic and Therapeutic Advances. *Genes*, 15(2), 171. <https://doi.org/10.3390/genes15020171>



Neuropathie optique héréditaire de Leber - LHON

95. Aleo, S. J., Del Dotto, V., Romagnoli, M., Fiorini, C., Capirossi, G., Peron, C., Maresca, A., Caporali, L., Capristo, M., Tropeano, C. V., Zanna, C., Ross-Cisneros, F. N., Sadun, A. A., Pignataro, M. G., Giordano, C., Fasano, C., Cavaliere, A., Porcelli, A. M., Tioli, G., ... Ghelli, A. M. (2024). Genetic variants affecting NQO1 protein levels impact the efficacy of idebenone treatment in Leber hereditary optic neuropathy. *Cell Reports. Medicine*, 5(2), 101383. <https://doi.org/10.1016/j.xcrm.2023.101383>
96. Andreeva, N. A., Murakhovskaya, Y. K., Krylova, T. D., Tsygankova, P. G., & Sheremet, N. L. (2023). [Rare pathogenic nucleotide variants of mitochondrial DNA associated with Leber's hereditary optic neuropathy]. *Vestnik Oftalmologii*, 139(6), 166-174. <https://doi.org/10.17116/oftalma2023139061166>
97. Barboni, M. T. S., Sustar Habjan, M., Petrovic Pajic, S., & Hawlina, M. (2024). Electroretinographic oscillatory potentials in Leber hereditary optic neuropathy. *Documenta Ophthalmologica. Advances in Ophthalmology*, 148(3), 133-143. <https://doi.org/10.1007/s10633-024-09968-9>
98. Barboni, P., Battista, M., Brotto, L., Nucci, P., Checchin, L., Bandello, F., Fiorini, C., Ormanbekova, D., Carelli, V., Cascavilla, M. L., & Caporali, L. (2024). Recurrence of Visual Loss in Recessive Leber Hereditary Optic Neuropathy : New Paradigm. *Journal of Neuro-Ophthalmology: The Official Journal of the North American Neuro-Ophthalmology Society*. <https://doi.org/10.1097/WNO.0000000000002117>
99. Blickhäuser, B., Stenton, S. L., Neuhofer, C. M., Floride, E., Nesbitt, V., Fratter, C., Koch, J., Kauffmann, B., Catarino, C., Schlieben, L. D., Kopajtich, R., Carelli, V., Sadun, A. A., McFarland, R., Fang, F., La Morgia, C., Paquay, S., Nassogne, M. C., Ghezzi, D., ... Prokisch, H. (2024). Digenic Leigh syndrome on the background of the m.11778G>A Leber hereditary optic neuropathy variant. *Brain: A Journal of Neurology*, 147(6), 1967-1974. <https://doi.org/10.1093/brain/awae057>
100. Gilhooley, M. J., Raoof, N., Yu-Wai-Man, P., & Moosajee, M. (2024). Inherited Optic Neuropathies : Real-World Experience in the Paediatric Neuro-Ophthalmology Clinic. *Genes*, 15(2), 188. <https://doi.org/10.3390/genes15020188>
101. Keshavan, N., Minczuk, M., Viscomi, C., & Rahman, S. (2024). Gene therapy for mitochondrial disorders. *Journal of Inherited Metabolic Disease*, 47(1), 145-175. <https://doi.org/10.1002/jimd.12699>
102. Ladero, M., Reche-Sainz, J. A., & Gallardo, M. E. (2024). Hereditary Optic Neuropathies : A Systematic Review on the Interplay between Biomaterials and Induced Pluripotent Stem Cells. *Bioengineering (Basel, Switzerland)*, 11(1), 52. <https://doi.org/10.3390/bioengineering11010052>



L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

103. Lapshin, E. V., Gershovich, Y. G., & Karabelsky, A. V. (2023). The Potential and Application of iPSCs in Gene and Cell Therapy for Retinopathies and Optic Neuropathies. *Acta Naturae*, 15(4), 56-64.
<https://doi.org/10.32607/actanaturae.25454>
104. Layrolle, P., Orssaud, C., Leleu, M., Payoux, P., & Chavanas, S. (2024). The Optic Nerve at Stake : Update on Environmental Factors Modulating Expression of Leber's Hereditary Optic Neuropathy. *Biomedicines*, 12(3), 584.
<https://doi.org/10.3390/biomedicines12030584>
105. Lee, D. K., Choi, Y. J., Lee, S. J., Kang, H. G., & Park, Y. R. (2024). Development of a deep learning model to distinguish the cause of optic disc atrophy using retinal fundus photography. *Scientific Reports*, 14(1), 5079. <https://doi.org/10.1038/s41598-024-55054-0>
106. Liu, Y.-C., Lin, Y.-K., Lin, Y.-T., Lin, C.-W., Lan, G.-Y., Su, Y.-C., Hu, F.-R., Chang, K.-H., Chen, V., Yeh, Y.-C., Chen, T.-C., & Yu, J. (2024). Injectable, Antioxidative, and Tissue-Adhesive Nanocomposite Hydrogel as a Potential Treatment for Inner Retina Injuries. *Advanced Science (Weinheim, Baden-Wurttemberg, Germany)*, 11(11), e2308635. <https://doi.org/10.1002/advs.202308635>
107. Murakhovskaya, Y. K., Andreeva, N. A., Tsygankova, P. G., Krylova, T. D., & Sheremet, N. L. (2023). [Long-term changes in morphological and functional parameters of the optic nerve in patients with various genetic variants of hereditary optic neuropathies]. *Vestnik Oftalmologii*, 139(6), 77-86.
<https://doi.org/10.17116/oftalma202313906177>
108. Nair, A. P., Selvakumar, A., Gopalarethinam, J., Abishek Kumar, B., Vellingiri, B., & Subramaniam, M. D. (2024). Epigenetic regulation of the nuclear genome associated with mitochondrial dysfunction in Leber's hereditary optic neuropathy (LHON). *Human Genome Variation*, 11(1), 6. <https://doi.org/10.1038/s41439-023-00258-5>
109. Otmani, A., Jóhannesson, G., Brautaset, R., Tribble, J. R., & Williams, P. A. (2024). Prophylactic nicotinamide treatment protects from rotenone-induced neurodegeneration by increasing mitochondrial content and volume. *Acta Neuropathologica Communications*, 12(1), 37. <https://doi.org/10.1186/s40478-024-01724-z>



L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

110. Peron, C., Cavaliere, A., Fasano, C., Iannielli, A., Spagnolo, M., Legati, A., Nicol Colombo, M., Rizzo, A., Sciacca, F. L., Carelli, V., Broccoli, V., Lamperti, C., & Tiranti, V. (2024). Generation of iPSCs from identical twin, one affected by LHON and one unaffected, both carrying a combination of two mitochondrial variants : M.14484 T>C and m.10680G>A. *Stem Cell Research*, 77, 103406. <https://doi.org/10.1016/j.scr.2024.103406>
111. Quigley, C., Stephenson, K. A. J., Kenna, P. F., & Cassidy, L. (2024). Peripapillary retinal nerve fibre layer thinning, perfusion changes and optic neuropathy in carriers of Leber hereditary optic neuropathy-associated mitochondrial variants. *BMJ Open Ophthalmology*, 9(1), e001295. <https://doi.org/10.1136/bmjophth-2023-001295>
112. Ricciotti, G., De Negri, A. M., & Carta, A. (2024). An atypical presentation of Leber's hereditary optic neuropathy in childhood. *Journal Francais D'ophtalmologie*, 47(3), 104037. <https://doi.org/10.1016/j.jfo.2023.104037>
113. Samuel Kim, U., & Yun, J. S. (2024). Clinical Characteristics of m.11778G>A Leber Hereditary Optic Neuropathy with Favorable Outcomes. *Seminars in Ophthalmology*, 39(4), 320-323. <https://doi.org/10.1080/08820538.2024.2323114>
114. Shemesh, A., Sood, G., Blair, K., & Margolin, E. (2024). Leber Hereditary Optic Neuropathy (LHON). In *StatPearls*. StatPearls Publishing. <http://www.ncbi.nlm.nih.gov/books/NBK482499/>
115. Siqueira, R. C. (2024). Photobiomodulation Using Light-Emitting Diode (LED) for Treatment of Retinal Diseases. *Clinical Ophthalmology (Auckland, N.Z.)*, 18, 215-225. <https://doi.org/10.2147/OPHTH.S441962>
116. Takai, Y., Iwasa, M., Yamagami, A., Inoue, K., Yasumoto, R., Ishikawa, H., & Wakakura, M. (2024). A Japanese Case of Leber's Hereditary Optic Neuropathy with the m.13051G>A Pathogenic Variant. *Neuro-Ophthalmology (Aeolus Press)*, 48(1), 51-55. <https://doi.org/10.1080/01658107.2023.2273480>
117. Xu, X., Zhou, H., Sun, M., Li, Y., Chen, B., Chen, X., Xu, Q., Yu-Wai-Man, P., & Wei, S. (2024). Neuroimaging changes in the pregeniculate visual pathway and chiasmal enlargement in Leber hereditary optic neuropathy. *The British Journal of Ophthalmology*, 108(9), 1313-1317. <https://doi.org/10.1136/bjo-2023-324628>
118. Yang, H. K., Seong, M.-W., & Hwang, J.-M. (2024). Mitochondrial DNA mutations in Korean patients with Leber's hereditary optic neuropathy. *Scientific Reports*, 14(1), 5702. <https://doi.org/10.1038/s41598-024-56215-x>

L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

119. Yang, Y.-P., Chang, Y.-L., Chiou, G.-Y., Lee, M.-S., Wu, Y.-R., Chen, P.-W., Lin, Y.-Y., Lai, W.-Y., Liu, Y.-H., Hwang, D.-K., & Chien, Y. (2024). Dysregulation of the circRNA_0087207/miR-548c-3p/PLSR1-TGFB2 axis in Leber hereditary optic neuropathy in vitro. *Journal of the Chinese Medical Association: JCMA*, 87(3), 261-266. <https://doi.org/10.1097/JCMA.0000000000001063>
120. Yu-Wai-Man, P., Carelli, V., Newman, N. J., Silva, M. J., Linden, A., Van Stavern, G., Szaflik, J. P., Banik, R., Lubiński, W., Pemp, B., Liao, Y. J., Subramanian, P. S., Misiuk-Hojto, M., Newman, S., Castillo, L., Kocięcki, J., Levin, M. H., Muñoz-Negrete, F. J., Yagan, A., ... LEROS Study Group. (2024). Therapeutic benefit of idebenone in patients with Leber hereditary optic neuropathy : The LEROS nonrandomized controlled trial. *Cell Reports. Medicine*, 5(3), 101437. <https://doi.org/10.1016/j.xcrm.2024.101437>
121. Zheng, Y., Wang, Y., Jiang, Y., Wang, J., Li, S., Xiao, X., Sun, W., Wang, P., Zhang, Q., & Jia, X. (2024). Variant and clinical landscape of Leber hereditary optic neuropathy based on 1516 families with mtDNA variants in a tertiary centre. *The British Journal of Ophthalmology*, 108(9), 1318-1327. <https://doi.org/10.1136/bjo-2023-323557>

Ophtalmoplégie externe progressive – PEO

122. Ali, A., Esmaeil, A., & Behbehani, R. (2024). Mitochondrial Chronic Progressive External Ophthalmoplegia. *Brain Sciences*, 14(2), 135. <https://doi.org/10.3390/brainsci14020135>
123. Beecher, G., Gavrilova, R. H., Mandrekar, J., & Naddaf, E. (2024). Mitochondrial myopathies diagnosed in adulthood: Clinico-genetic spectrum and long-term outcomes. *Brain Communications*, 6(2), fcae041. <https://doi.org/10.1093/braincomms/fcae041>
124. Breuss, A., Strasser, M., Nuoffer, J.-M., Klein, A., Perret-Hoigné, E., Felder, C., Stauffer, R., Wolf, P., Riener, R., & Gautschi, M. (2024). Nocturnal vestibular stimulation using a rocking bed improves a severe sleep disorder in a patient with mitochondrial disease. *Journal of Sleep Research*, e14153. <https://doi.org/10.1111/jsr.14153>
125. Cohen, B. H., Chinnery, P. F., & Copeland, W. C. (1993). POLG-Related Disorders. 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/NBK26471/>



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MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

126. Feng, Z., Lai, R., Wei, J., Liu, X., Chen, X., Liu, Y., Qin, W., Qin, X., & Kong, F. (2023). Have one's view of the important overshadowed by the trivial : Chronic progressive external ophthalmoplegia combined with unilateral facial nerve injury: a case report and literature review. *Frontiers in Neurology*, 14, 1268053. <https://doi.org/10.3389/fneur.2023.1268053>
127. Fraiman, P. H. A., Silva, T. Y. T., Pedroso, J. L., & Barsottini, O. G. P. (2024). Teaching Video Neurolmage : TWNK Mutation Causes Chronic Progressive External Ophthalmoplegia and Cerebellar Ataxia: A POLG-Like Phenotype. *Neurology*, 102(5), e209186. <https://doi.org/10.1212/WNL.0000000000209186>
128. Gogu, A. E., Jianu, D. C., Parv, F., Motoc, A. G. M., Axelerad, A., Stuparu, A. Z., & Gogu, A. A. (2023). Case report : Clinical profile, molecular genetics, and neuroimaging findings presenting in a patient with Kearns-Sayre syndrome associated with inherited thrombophilia. *Frontiers in Neurology*, 14, 1320757. <https://doi.org/10.3389/fneur.2023.1320757>
129. Liu, H., Gao, M., Sun, Q., Chen, S., Luo, Y., Yang, H., Li, Q., Li, J., & Yang, G. (2023). A case of mitochondrial myopathy and chronic progressive external ophthalmoplegia. *Zhong Nan Da Xue Xue Bao. Yi Xue Ban = Journal of Central South University. Medical Sciences*, 48(11), 1760-1768. <https://doi.org/10.11817/j.issn.1672-7347.2023.220605>
130. Maniyar, A. M. H., Singh, R. K., Ojha, P. T., Chaudhary, G. S., Mahto, A. P., & Shah, A. G. (2023). Myosin Myopathy Presenting as Chronic Progressive External Ophthalmoplegia. *Annals of Indian Academy of Neurology*, 26(6), 1024-1025. https://doi.org/10.4103/aian.aian_552_23
131. Menon, D., Nair, S. S., Radhakrishnan, N., Saraf, U. U., & Nair, M. (2023). Clinical Spectrum of Biopsy Proven Mitochondrial Myopathy. *Neurology India*, 71(6), 1192-1196. <https://doi.org/10.4103/0028-3886.391399>

L'essentielle Maladies Mitochondriales

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

132. Cohen, B. H., Chinnery, P. F., & Copeland, W. C. (1993). POLG-Related Disorders. 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/NBK26471/>
133. Hong, Y., Zhang, Z., Yangzom, T., Chen, A., Lundberg, B. C., Fang, E. F., Siller, R., Sullivan, G. J., Zeman, J., Tzoulis, C., Bindoff, L. A., & Liang, K. X. (2024). The NAD⁺ Precursor Nicotinamide Riboside Rescues Mitochondrial Defects and Neuronal Loss in iPSC derived Cortical Organoid of Alpers' Disease. *International Journal of Biological Sciences*, 20(4), 1194-1217. <https://doi.org/10.7150/ijbs.91624>

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

134. Feng, Z., Lai, R., Wei, J., Liu, X., Chen, X., Liu, Y., Qin, W., Qin, X., & Kong, F. (2023). Have one's view of the important overshadowed by the trivial : Chronic progressive external ophthalmoplegia combined with unilateral facial nerve injury: a case report and literature review. *Frontiers in Neurology*, 14, 1268053. <https://doi.org/10.3389/fneur.2023.1268053>
135. Finsterer, J. (2023). Diagnosing Kearns-Sayre syndrome requires clinical and genetic evidence. *Journal of Family Medicine and Primary Care*, 12(12), 3437-3438. https://doi.org/10.4103/jfmpc.jfmpc_1023_23
136. Gogu, A. E., Jianu, D. C., Parv, F., Motoc, A. G. M., Axelerad, A., Stuparu, A. Z., & Gogu, A. A. (2023). Case report: Clinical profile, molecular genetics, and neuroimaging findings presenting in a patient with Kearns-Sayre syndrome associated with inherited thrombophilia. *Frontiers in Neurology*, 14, 1320757. <https://doi.org/10.3389/fneur.2023.1320757>
137. Maddali, M. M., Munasinghe, T. D., Al Aamri, I., Al-Abri, I. A., & Al-Adawi, S. (2023). Propofol and Kearns-Sayre Syndrome: An idiographic approach. *Sultan Qaboos University Medical Journal*, 23(Spec Iss), 63-67. <https://doi.org/10.18295/squmj.12.2023.080>
138. Mehri, S., Zarrouk, S., & Finsterer, J. (2024). Re : Propofol in Triple Trouble Kearns-Sayre Syndrome, Dyggve-Melchior-Clausen Syndrome, and Chromosome-9 Inversion. *Sultan Qaboos University Medical Journal*, 24(1), 146-148. <https://doi.org/10.18295/squmj.2.2024.013>

L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

139. Yazdani, M. (2024). Cellular and Molecular Responses to Mitochondrial DNA Deletions in Kearns-Sayre Syndrome: Some Underlying Mechanisms. *Molecular Neurobiology*, 61(8), 5665-5679. <https://doi.org/10.1007/s12035-024-03938-7>

Syndrome de Leigh - Leigh syndrome

140. Abu Hanna, F., Zehavi, Y., Cohen-Barak, E., Khayat, M., Warwar, N., Shreter, R., Rodenburg, R. J., & Spiegel, R. (2024). Lack of mitochondrial complex I assembly factor NDUFAF2 results in a distinctive infantile-onset brainstem neurodegenerative disease with early lethality. *Orphanet Journal of Rare Diseases*, 19(1), 92. <https://doi.org/10.1186/s13023-024-03094-0>
141. Aguilar, K., Canal, C., Comes, G., Díaz-Clavero, S., Llanos, M. A., Quintana, A., Sanz, E., & Hidalgo, J. (2024). Interleukin-6-elicited chronic neuroinflammation may decrease survival but is not sufficient to drive disease progression in a mouse model of Leigh syndrome. *Journal of Inflammation (London, England)*, 21(1), 1. <https://doi.org/10.1186/s12950-023-00369-4>
142. Arbide, D., Elkhateeb, N., Goljan, E., Gonzalez, C. P., Maw, A., & Park, S.-M. (2024). A Novel Heterozygous De Novo MORC2 Missense Variant Causes an Early Onset and Severe Neurodevelopmental Disorder. *Case Reports in Genetics*, 2024, 5906936. <https://doi.org/10.1155/2024/5906936>
143. Armirola-Ricaurte, C., Zonnekein, N., Koutsis, G., Amor-Barris, S., Pelayo-Negro, A. L., Atkinson, D., Efthymiou, S., Turchetti, V., Dinopoulos, A., Garcia, A., Karakaya, M., Moris, G., Polat, A. I., Yiş, U., Espinos, C., Van de Vondel, L., De Vriendt, E., Karadima, G., Wirth, B., ... Jordanova, A. (2024). Alternative splicing expands the clinical spectrum of NDUFS6-related mitochondrial disorders. *Genetics in Medicine: Official Journal of the American College of Medical Genetics*, 26(6), 101117. <https://doi.org/10.1016/j.gim.2024.101117>
144. Blickhäuser, B., Stenton, S. L., Neuhofer, C. M., Floride, E., Nesbitt, V., Fratter, C., Koch, J., Kauffmann, B., Catarino, C., Schlieben, L. D., Kopajtich, R., Carelli, V., Sadun, A. A., McFarland, R., Fang, F., La Morgia, C., Paquay, S., Nassogne, M. C., Ghezzi, D., ... Prokisch, H. (2024). Digenic Leigh syndrome on the background of the m.11778G>A Leber hereditary optic neuropathy variant. *Brain: A Journal of Neurology*, 147(6), 1967-1974. <https://doi.org/10.1093/brain/awae057>



L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

145. Borna, N. N., Kishita, Y., Shimura, M., Murayama, K., Ohtake, A., & Okazaki, Y. (2024). Identification of a novel MT-ND3 variant and restoring mitochondrial function by allotopic expression of MT-ND3 gene. *Mitochondrion*, 76, 101858. <https://doi.org/10.1016/j.mito.2024.101858>
146. Cho, S.-I., Lim, K., Hong, S., Lee, J., Kim, A., Lim, C. J., Ryou, S., Lee, J. M., Mok, Y. G., Chung, E., Kim, S., Han, S., Cho, S.-M., Kim, J., Kim, E.-K., Nam, K.-H., Oh, Y., Choi, M., An, T. H., ... Kim, J.-S. (2024). Engineering TALE-linked deaminases to facilitate precision adenine base editing in mitochondrial DNA. *Cell*, 187(1), 95-109.e26. <https://doi.org/10.1016/j.cell.2023.11.035>
147. Christen, M., Gregor, A., Gutierrez-Quintana, R., Bongers, J., Rupp, A., Penderis, J., Shelton, G. D., Jagannathan, V., Zweier, C., & Leeb, T. (2024). NDUF57 variant in dogs with Leigh syndrome and its functional validation in a *Drosophila melanogaster* model. *Scientific Reports*, 14(1), 2975. <https://doi.org/10.1038/s41598-024-53314-7>
148. Gangfuß, A., Rating, P., Ferreira, T., Hentschel, A., Marina, A. D., Kölbel, H., Sickmann, A., Abicht, A., Kraft, F., Ruck, T., Böhm, J., Schänzer, A., Schara-Schmidt, U., Neuhaus, T. M., Horvath, R., & Roos, A. (2024). A Homozygous NDUF56 Variant Associated with Neuropathy and Optic Atrophy. *Journal of Neuromuscular Diseases*, 11(2), 485-491. <https://doi.org/10.3233/JND-230181>
149. Gouiza, I., Hechmi, M., Zioudi, A., Dallali, H., Kheriji, N., Charif, M., Le Mao, M., Galai, S., Kraoua, L., Ben Youssef-Turki, I., Kraoua, I., Lenaers, G., & Kefi, R. (2023). Expanding the genetic spectrum of mitochondrial diseases in Tunisia : Novel variants revealed by whole-exome sequencing. *Frontiers in Genetics*, 14, 1259826. <https://doi.org/10.3389/fgene.2023.1259826>
150. Gurusamy, U., Ramadesikan, S., Marhabaie, M., Colwell, C. M., Hunter, J. M., Leung, M. L., Mardis, E. R., White, P., Manickam, M., Wilson, R. K., & Koboldt, D. C. (2023). Biallelic variants in HTRA2 cause 3-methylglutaconic aciduria mitochondrial disorder : Case report and literature review. *Frontiers in Genetics*, 14, 1298574. <https://doi.org/10.3389/fgene.2023.1298574>
151. Hai, B., Guntin, J., Rao, A. G., & Ata, M. (2024). Case 321 : Leigh Syndrome. *Radiology*, 310(1), e222509. <https://doi.org/10.1148/radiol.222509>
152. Ichinose, F., & Hindle, A. (2024). Sulfide catabolism in hibernation and neuroprotection. *Nitric Oxide: Biology and Chemistry*, 146, 19-23. <https://doi.org/10.1016/j.niox.2024.03.002>

L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

153. Ipas, H., Gouws, E. B., Abell, N. S., Chiou, P.-C., Devanathan, S. K., Hervé, S., Lee, S., Mercado, M., Reinsborough, C., Halabelian, L., Arrowsmith, C. H., & Xhemaççe, B. (2024). ChemRAP uncovers specific mRNA translation regulation via RNA 5' phosphomethylation. *EMBO Reports*, 25(3), 1570-1588. <https://doi.org/10.1038/s44319-024-00059-z>
154. Maalej, M., Sfaihi, L., Fersi, O.-A., Khabou, B., Ammar, M., Felhi, R., Kharrat, M., Chouchen, J., Kammoun, T., Tlili, A., & Fakhfakh, F. (2024). Molecular and in silico investigation of a novel ECHS1 gene mutation in a consanguine family with short-chain enoyl-CoA hydratase deficiency and Mt-DNA depletion: Effect on trimer assembly and catalytic activity. *Metabolic Brain Disease*, 39(4), 611-623. <https://doi.org/10.1007/s11011-024-01343-6>
155. Matthews, E., Whittle, E. F., Khan, F., McEntagart, M., & Carroll, C. J. (2024). Leigh syndrome with developmental regression and ataxia due to a novel splicing variant in the PMPCB gene. *Journal of Human Genetics*, 69(6), 283-285. <https://doi.org/10.1038/s10038-024-01226-9>
156. Ou-Yang, C.-H., Chen, P.-S., & Lin, C.-H. (2024). Generation of a human induced pluripotent stem cell line NTUHi004-A from a patient with Leigh syndrome harboring a homozygous missense mutation c.836 T > G (p.Met279Arg) in NDUFAF5 gene. *Stem Cell Research*, 76, 103379. <https://doi.org/10.1016/j.scr.2024.103379>
157. Pooja, P. S., Greeshma, R., Joy, E. K., & Swamy, S. P. (2024). Auditory Brainstem Response in a Child with Mitochondrial Disorder-Leigh Syndrome. *Indian Journal of Otolaryngology and Head and Neck Surgery: Official Publication of the Association of Otolaryngologists of India*, 76(1), 1014-1017. <https://doi.org/10.1007/s12070-023-03971-3>
158. Rambani, V., Kolnikova, M., Cagalinec, M., Skopkova, M., & Gasperikova, D. (2023). PMPCA-Related Encephalopathy: Novel Variants, Phenotype Extension, and Mitochondrial Morphology. *Neurology. Genetics*, 9(6), e200106. <https://doi.org/10.1212/NXG.000000000200106>
159. Sabharwal, A., Gupta, V., Kv, S., Kumar Manokaran, R., Verma, A., Mishra, A., Bhoyar, R. C., Jain, A., Sivadas, A., Rawat, S., Jolly, B., Mohanty, S., Gulati, S., Gupta, N., Kabra, M., Scaria, V., & Sivasubbu, S. (2024). Whole genome sequencing followed by functional analysis of genomic deletion encompassing ERCC8 and NDUFAF2 genes in a non-consanguineous Indian family reveals dysfunctional mitochondrial bioenergetics leading to infant mortality. *Mitochondrion*, 75, 101844. <https://doi.org/10.1016/j.mito.2024.101844>

L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

160. Savvidou, A., Sofou, K., Eklund, E. A., Aronsson, J., & Darin, N. (2024). Manifestations of X-linked pyruvate dehydrogenase complex deficiency in female PDHA1 carriers. *European Journal of Neurology*, 31(7), e16283. <https://doi.org/10.1111/ene.16283>
161. Soares, M. P., Travessa, A. M., Custódio, S., Pereira, C., Pinto, P., & Sousa, A. B. (2024). Uniparental Disomy as a Mechanism for Combined Oxidative Phosphorylation Deficiency Associated with MRPS34 Gene. *Endocrine, Metabolic & Immune Disorders Drug Targets*. <https://doi.org/10.2174/0118715303283767231120113921>
162. Sonsalla, G., Malpartida, A. B., Riedemann, T., Gusic, M., Rusha, E., Bulli, G., Najas, S., Janjic, A., Hersbach, B. A., Smialowski, P., Drukker, M., Enard, W., Prehn, J. H. M., Prokisch, H., Götz, M., & Masserdotti, G. (2024). Direct neuronal reprogramming of NDUFS4 patient cells identifies the unfolded protein response as a novel general reprogramming hurdle. *Neuron*, 112(7), 1117-1132.e9. <https://doi.org/10.1016/j.neuron.2023.12.020>
163. Spencer, K. A., Howe, M. N., Mulholland, M. T., Truong, V., Liao, R. W., Chen, Y., Setha, M., Snell, J. C., Hanaford, A., James, K., Morgan, P. G., Sedensky, M. M., & Johnson, S. C. (2024). Impact of dietary ketosis on volatile anesthesia toxicity in a model of Leigh syndrome. *Paediatric Anaesthesia*, 34(5), 467-476. <https://doi.org/10.1111/pan.14855>
164. Yang, N., Chen, L., Zhang, Y., Wu, X., Hao, Y., Yang, F., Yang, Z., & Liang, J. (2024). Novel NARS2 variants in a patient with early-onset status epilepticus : Case study and literature review. *BMC Pediatrics*, 24(1), 96. <https://doi.org/10.1186/s12887-024-04553-0>
165. Yin, Z., Agip, A.-N. A., Bridges, H. R., & Hirst, J. (2024). Structural insights into respiratory complex I deficiency and assembly from the mitochondrial disease-related ndufs4-/- mouse. *The EMBO Journal*, 43(2), 225-249. <https://doi.org/10.1038/s44318-023-00001-4>
166. Zhang, J., Gan, J., & Wang, J. (2024). A case of Leigh syndrome presented with paroxysmal body swing. *Heliyon*, 10(1), e23137. <https://doi.org/10.1016/j.heliyon.2023.e23137>
167. Zhou, F., Yi, G., Liu, X., Sheng, W., Shu, J., Li, D., & Cai, C. (2024). A Pair of Compound Heterozygous IARS2 Variants Manifesting West Syndrome and Electrolyte Disorders in a Chinese Patient. *Global Medical Genetics*, 11(1), 25-28. <https://doi.org/10.1055/s-0043-1778091>

Maladies mitochondriales (autres) – Mitochondrial disorders (other)

168. Abadie, J. M. (2024). A Two-Genome Portrayal of Mitochondrial Disorders : A Review with Clinical Presentations. *Frontiers in Bioscience (Scholar Edition)*, 16(1), 7. <https://doi.org/10.31083/j.fbs1601007>
169. Abu Hanna, F., Zehavi, Y., Cohen-Barak, E., Khayat, M., Warwar, N., Shreter, R., Rodenburg, R. J., & Spiegel, R. (2024). Lack of mitochondrial complex I assembly factor NDUFAF2 results in a distinctive infantile-onset brainstem neurodegenerative disease with early lethality. *Orphanet Journal of Rare Diseases*, 19(1), 92. <https://doi.org/10.1186/s13023-024-03094-0>
170. Aguilar, K., Canal, C., Comes, G., Díaz-Clavero, S., Llanos, M. A., Quintana, A., Sanz, E., & Hidalgo, J. (2024). Interleukin-6-elicited chronic neuroinflammation may decrease survival but is not sufficient to drive disease progression in a mouse model of Leigh syndrome. *Journal of Inflammation (London, England)*, 21(1), 1. <https://doi.org/10.1186/s12950-023-00369-4>
171. Aimo, A., Morfino, P., Arzilli, C., Vergaro, G., Spini, V., Fabiani, I., Castiglione, V., Rapezzi, C., & Emdin, M. (2024). Disease features and management of cardiomyopathies in women. *Heart Failure Reviews*, 29(3), 663-674. <https://doi.org/10.1007/s10741-024-10386-x>
172. Aleksic, D., Jankovic, M. G., Todorovic, S., Kovacevic, M., & Borkovic, M. (2023). The first case of combined oxidative phosphorylation deficiency-1 due to a GFM1 mutation in the Serbian population : A case report and literature review. *The Turkish Journal of Pediatrics*, 65(6), 1018-1024. <https://doi.org/10.24953/turkjpeds.2022.1082>
173. Ali, A., Esmaeil, A., & Behbehani, R. (2024). Mitochondrial Chronic Progressive External Ophthalmoplegia. *Brain Sciences*, 14(2), 135. <https://doi.org/10.3390/brainsci14020135>
174. Alix, J. J. P., Plesia, M., Schooling, C. N., Dudgeon, A. P., Kendall, C. A., Kadirkamanathan, V., McDermott, C. J., Gorman, G. S., Taylor, R. W., Mead, R. J., Shaw, P. J., & Day, J. C. (2023). Non-negative matrix factorisation of Raman spectra finds common patterns relating to neuromuscular disease across differing equipment configurations, preclinical models and human tissue. *Journal of Raman Spectroscopy: JRS*, 54(3), 258-268. <https://doi.org/10.1002/jrs.6480>
175. Alves, C. A. P. F., & Whitehead, M. T. (2024). Advancing the neuroimaging diagnosis and understanding of mitochondrial disorders. *Neurotherapeutics: The Journal of the American Society for Experimental NeuroTherapeutics*, 21(1), e00324. <https://doi.org/10.1016/j.neurot.2024.e00324>

L'essentielle Maladies Mitochondriales

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176. Ambrogetti, R., Kavanagh, E., & ElTayeb, K. (2024). Late-onset mitochondrial encephalopathy with lactic acidosis and stroke-like episodes and the role of serial imaging. *BMJ Case Reports*, 17(2), e259102. <https://doi.org/10.1136/bcr-2023-259102>
177. Armirola-Ricaurte, C., Zonnekeijn, N., Koutsis, G., Amor-Barris, S., Pelayo-Negro, A. L., Atkinson, D., Efthymiou, S., Turchetti, V., Dinopoulos, A., Garcia, A., Karakaya, M., Moris, G., Polat, A. I., Yiş, U., Espinos, C., Van de Vondel, L., De Vriendt, E., Karadima, G., Wirth, B., ... Jordanova, A. (2024). Alternative splicing expands the clinical spectrum of NDUFS6-related mitochondrial disorders. *Genetics in Medicine: Official Journal of the American College of Medical Genetics*, 26(6), 101117. <https://doi.org/10.1016/j.gim.2024.101117>
178. Askari, H., Rabiei, F., Lohrasbi, F., Ghadir, S., Mehdipour Arbastan, A., & Ghasemi-Kasman, M. (2024). AMP-activated protein kinase as a mediator of mitochondrial dysfunction of multiple sclerosis in animal models : A systematic review. *Journal of Cellular Physiology*, 239(5), e31230. <https://doi.org/10.1002/jcp.31230>
179. Averina, O. A., Kuznetsova, S. A., Permyakov, O. A., & Sergiev, P. V. (2023). Animal Models of Mitochondrial Diseases Associated with Nuclear Gene Mutations. *Acta Naturae*, 15(4), 4-22. <https://doi.org/10.32607/actanaturae.25442>
180. Balicza, P., Gezsi, A., Fedor, M., Sagi, J. C., Gal, A., Varga, N. A., & Molnar, M. J. (2023). Multilevel evidence of MECP2-associated mitochondrial dysfunction and its therapeutic implications. *Frontiers in Psychiatry*, 14, 1301272. <https://doi.org/10.3389/fpsyt.2023.1301272>
181. Balmaceda, V., Komlódi, T., Szibor, M., Gnaiger, E., Moore, A. L., Fernandez-Vizarra, E., & Viscomi, C. (2024). The striking differences in the bioenergetics of brain and liver mitochondria are enhanced in mitochondrial disease. *Biochimica Et Biophysica Acta. Molecular Basis of Disease*, 1870(3), 167033. <https://doi.org/10.1016/j.bbadis.2024.167033>
182. Bilgin, H., & Bozaci, A. E. (2024). The evaluation of inherited metabolic diseases presenting with rhabdomyolysis from Turkey : Single center experience. *Molecular Genetics and Metabolism Reports*, 39, 101070. <https://doi.org/10.1016/j.ymgmr.2024.101070>



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183. Blickhäuser, B., Stenton, S. L., Neuhofer, C. M., Floride, E., Nesbitt, V., Fratter, C., Koch, J., Kauffmann, B., Catarino, C., Schlieben, L. D., Kopajtich, R., Carelli, V., Sadun, A. A., McFarland, R., Fang, F., La Morgia, C., Paquay, S., Nassogne, M. C., Ghezzi, D., ... Prokisch, H. (2024). Digenic Leigh syndrome on the background of the m.11778G>A Leber hereditary optic neuropathy variant. *Brain: A Journal of Neurology*, 147(6), 1967-1974. <https://doi.org/10.1093/brain/awae057>
184. Borna, N. N., Kishita, Y., Shimura, M., Murayama, K., Ohtake, A., & Okazaki, Y. (2024). Identification of a novel MT-ND3 variant and restoring mitochondrial function by allotopic expression of MT-ND3 gene. *Mitochondrion*, 76, 101858. <https://doi.org/10.1016/j.mito.2024.101858>
185. Braun, E. (2024). Mitochondrial replacement techniques for treating infertility. *Journal of Medical Ethics*, jme-2023-109660. <https://doi.org/10.1136/jme-2023-109660>
186. Breuss, A., Strasser, M., Nuoffer, J.-M., Klein, A., Perret-Hoigné, E., Felder, C., Stauffer, R., Wolf, P., Riener, R., & Gautschi, M. (2024). Nocturnal vestibular stimulation using a rocking bed improves a severe sleep disorder in a patient with mitochondrial disease. *Journal of Sleep Research*, e14153. <https://doi.org/10.1111/jsr.14153>
187. Bruni, F. (2024). Human mtDNA-Encoded Long ncRNAs : Knotty Molecules and Complex Functions. *International Journal of Molecular Sciences*, 25(3), 1502. <https://doi.org/10.3390/ijms25031502>
188. Byrne, M. M., Sanchez-Lechuga, B., Ng, N., & Byrne, B. (2024). Obstetric complications in women with mitochondrial disease. *Acta Diabetologica*, 61(2), 265-266. <https://doi.org/10.1007/s00592-023-02225-6>
189. Calame, D. G., & Emrick, L. T. (2024). Functional genomics and small molecules in mitochondrial neurodevelopmental disorders. *Neurotherapeutics: The Journal of the American Society for Experimental NeuroTherapeutics*, 21(1), e00316. <https://doi.org/10.1016/j.neurot.2024.e00316>
190. Cao, S., Wei, Y., Yue, Y., Chen, Y., Liao, S., Li, A., Liu, P., Xiong, A., & Zeng, H. (2024). Targeting ferroptosis unveils a new era for traditional Chinese medicine : A scientific metrology study. *Frontiers in Pharmacology*, 15, 1366852. <https://doi.org/10.3389/fphar.2024.1366852>

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MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

191. Cardon, I., Grobecker, S., Jenne, F., Jahner, T., Rupprecht, R., Milenkovic, V. M., & Wetzel, C. H. (2024). Serotonin effects on human iPSC-derived neural cell functions : From mitochondria to depression. *Molecular Psychiatry*, 29(9), 2689-2700. <https://doi.org/10.1038/s41380-024-02538-0>
192. Cardon, I., Grobecker, S., Kücükoktay, S., Bader, S., Jahner, T., Nothdurfter, C., Koschitzki, K., Berneburg, M., Weber, B. H. F., Stöhr, H., Höring, M., Liebisch, G., Braun, F., Rothammer-Hampl, T., Riemenschneider, M. J., Rupprecht, R., Milenkovic, V. M., & Wetzel, C. H. (2024). Mitochondrial and Cellular Function in Fibroblasts, Induced Neurons, and Astrocytes Derived from Case Study Patients : Insights into Major Depression as a Mitochondria-Associated Disease. *International Journal of Molecular Sciences*, 25(2), 963. <https://doi.org/10.3390/ijms25020963>
193. Carmona Alexandrino, H., Ferreira, M. A., Ramalho, D., Jesus, N. R., & Oliveira, M. J. (2023). Endocrine Challenges in Myoclonic Epilepsy With Ragged Red Fibers Syndrome : A Case Report. *Cureus*, 15(12), e51114. <https://doi.org/10.7759/cureus.51114>
194. Chang, L.-Y., Chao, Y.-L., Chiu, C.-C., Chen, P.-L., & Lin, H. Y.-H. (2024). Mitochondrial Signaling, the Mechanisms of AKI-to-CKD Transition and Potential Treatment Targets. *International Journal of Molecular Sciences*, 25(3), 1518. <https://doi.org/10.3390/ijms25031518>
195. Chang, X., Niu, S., Guo, M., Shang, M., Guo, S., Mou, X., Wu, T., Tang, M., & Xue, Y. (2024). Silver nanoparticles induced synaptic degeneration via Ca²⁺/CaMKII signal and Drp1-dependent mitochondrial disorder in HT22 cells. *Food and Chemical Toxicology: An International Journal Published for the British Industrial Biological Research Association*, 186, 114577. <https://doi.org/10.1016/j.fct.2024.114577>
196. Che, L., Wu, Y., Sheng, M., Xu, J., Yu, W., & Weng, Y. (2024). Intraoperative management during liver transplantation in the child with mitochondrial depletion syndrome : A case report. *International Journal of Surgery Case Reports*, 116, 109432. <https://doi.org/10.1016/j.ijscr.2024.109432>
197. Chen, A., Yangzom, T., Hong, Y., Lundberg, B. C., Sullivan, G. J., Tzoulis, C., Bindoff, L. A., & Liang, K. X. (2024). Hallmark Molecular and Pathological Features of POLG Disease are Recapitulated in Cerebral Organoids. *Advanced Science (Weinheim, Baden-Wurttemberg, Germany)*, 11(18), e2307136. <https://doi.org/10.1002/advs.202307136>

L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

198. Cheng, Y., Huang, P., Zou, Q., Tian, H., Cheng, Q., & Ding, H. (2024). Nicotinamide mononucleotide alleviates seizures via modulating SIRT1-PGC-1 α mediated mitochondrial fusion and fission. *Journal of Neurochemistry*. <https://doi.org/10.1111/jnc.16041>
199. Chin, H.-L., Lai, P. S., & Tay, S. K. H. (2024). A clinical approach to diagnosis and management of mitochondrial myopathies. *Neurotherapeutics: The Journal of the American Society for Experimental NeuroTherapeutics*, 21(1), e00304. <https://doi.org/10.1016/j.neurot.2023.11.001>
200. Chinnery, P. F. (2023). Precision mitochondrial medicine. *Cambridge Prisms. Precision Medicine*, 1, e6. <https://doi.org/10.1017/pcm.2022.8>
201. Citrigno, L., Qualtieri, A., Cerantonio, A., De Benedittis, S., Gallo, O., Di Palma, G., Spadafora, P., & Cavalcanti, F. (2024). Genomics landscape of mitochondrial DNA variations in patients from South Italy affected by mitochondriopathies. *Journal of the Neurological Sciences*, 457, 122869. <https://doi.org/10.1016/j.jns.2024.122869>
202. Deng, Y., Chen, Q., Wang, T., Wang, S., Li, R., Wang, Y., Zhang, J., Gan, J., & Guo, M. (2024). Myocardial Ischemia/Reperfusion Injury : Mechanism and Targeted Treatment for Ferroptosis. *Anatolian Journal of Cardiology*, 28(3), 133-141. <https://doi.org/10.14744/AnatoJCardiol.2023.3606>
203. Doni, D., Cavallari, E., Noguera, M. E., Gentili, H. G., Cavion, F., Parisi, G., Fornasari, M. S., Sartori, G., Santos, J., Bellanda, M., Carbonera, D., Costantini, P., & Bortolus, M. (2024). Searching for Frataxin Function : Exploring the Analogy with Nqo15, the Frataxin-like Protein of Respiratory Complex I from *Thermus thermophilus*. *International Journal of Molecular Sciences*, 25(3), 1912. <https://doi.org/10.3390/ijms25031912>
204. Eghbalsaid, S., Lawler, C., Petersen, B., Hajiye, R. A., Bischoff, S. R., & Frankenberg, S. (2024). CRISPR/Cas9-mediated base editors and their prospects for mitochondrial genome engineering. *Gene Therapy*, 31(5-6), 209-223. <https://doi.org/10.1038/s41434-023-00434-w>
205. Ferreira, F., Gonçalves Bacelar, C., Lisboa-Gonçalves, P., Paulo, N., Quental, R., Nunes, A. T., Silva, R., & Tavares, I. (2023). Renal manifestations in adults with mitochondrial disease from the mtDNA m.3243A>G pathogenic variant. *Nefrologia*, 43 Suppl 2, 1-7. <https://doi.org/10.1016/j.nefro.2024.01.017>

L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

206. Ferreira, T., Polavarapu, K., Olimpio, C., Paramonov, I., Lochmüller, H., & Horvath, R. (2024). Variants in mitochondrial disease genes are common causes of inherited peripheral neuropathies. *Journal of Neurology*, 271(6), 3546-3553. <https://doi.org/10.1007/s00415-024-12319-y>
207. Finsterer, J. (2024). MPV17-related mtDNA depletion syndrome complicated by postreperfusion syndrome during liver transplantation. *International Journal of Surgery Case Reports*, 117, 109553. <https://doi.org/10.1016/j.ijscr.2024.109553>
208. Finsterer, J., Mehri, S., & Zarrouk, S. (2024). Sudden death in non-syndromic mitochondrial disorders due to m.3243A > G may not only be cardiogenic. *Cardiology in the Young*, 34(4), 935-936. <https://doi.org/10.1017/S1047951124000234>
209. Gangfuß, A., Rating, P., Ferreira, T., Hentschel, A., Marina, A. D., Kölbel, H., Sickmann, A., Abicht, A., Kraft, F., Ruck, T., Böhm, J., Schänzer, A., Schara-Schmidt, U., Neuhaus, T. M., Horvath, R., & Roos, A. (2024). A Homozygous NDUF6 Variant Associated with Neuropathy and Optic Atrophy. *Journal of Neuromuscular Diseases*, 11(2), 485-491. <https://doi.org/10.3233/JND-230181>
210. Gao, Z., Santos, R. B., Rupert, J., Van Drunen, R., Yu, Y., Eckel-Mahan, K., & Kolonin, M. G. (2024). Endothelial-specific telomerase inactivation causes telomere-independent cell senescence and multi-organ dysfunction characteristic of aging. *Aging Cell*, 23(6), e14138. <https://doi.org/10.1111/accel.14138>
211. Gerard, A., Mizerik, E., Mohila, C. A., AlAwami, S., Hunter, J. V., Kearney, D. L., Lalani, S. R., & Scaglia, F. (2024). Intracranial calcifications simulating Aicardi-Goutières syndrome in PARS2-related mitochondrial disease. *American Journal of Medical Genetics. Part A*, 194(7), e63589. <https://doi.org/10.1002/ajmg.a.63589>
212. Gouiza, I., Hechmi, M., Zioudi, A., Dallali, H., Kheriji, N., Charif, M., Le Mao, M., Galai, S., Kraoua, L., Ben Youssef-Turki, I., Kraoua, I., Lenaers, G., & Kefi, R. (2023). Expanding the genetic spectrum of mitochondrial diseases in Tunisia : Novel variants revealed by whole-exome sequencing. *Frontiers in Genetics*, 14, 1259826. <https://doi.org/10.3389/fgene.2023.1259826>
213. Gowda, V. K., Bylappa, A. Y., Kinhal, U., Srinivasan, V. M., & Vamyanmane, D. K. (2023). Mitochondrial Complex I Deficiency Masquerading as Stroke-Like Episode Clinically and as Alexander Disease Radiologically Following Chicken Pox. *Annals of Indian Academy of Neurology*, 26(6), 977-979. https://doi.org/10.4103/aian.aian_339_23



L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

214. Granat, L., Knorr, D. Y., Ranson, D. C., Chakrabarty, R. P., Chandel, N. S., & Bateman, J. M. (2024). A *Drosophila* model of mitochondrial disease phenotypic heterogeneity. *Biology Open*, 13(2), bio060278. <https://doi.org/10.1242/bio.060278>
215. Gropman, A., & Chandra, B. (2024). Mitochondrial disorders : Emerging paradigms and the road ahead to personalized medicine. *Neurotherapeutics: The Journal of the American Society for Experimental NeuroTherapeutics*, 21(1), e00332. <https://doi.org/10.1016/j.neurot.2024.e00332>
216. Gropman, A. L., Uittenbogaard, M. N., & Chiaramello, A. E. (2024). Challenges and opportunities to bridge translational to clinical research for personalized mitochondrial medicine. *Neurotherapeutics: The Journal of the American Society for Experimental NeuroTherapeutics*, 21(1), e00311. <https://doi.org/10.1016/j.neurot.2023.e00311>
217. Gurusamy, U., Ramadesikan, S., Marhabaie, M., Colwell, C. M., Hunter, J. M., Leung, M. L., Mardis, E. R., White, P., Manickam, M., Wilson, R. K., & Koboldt, D. C. (2023). Biallelic variants in HTRA2 cause 3-methylglutaconic aciduria mitochondrial disorder : Case report and literature review. *Frontiers in Genetics*, 14, 1298574. <https://doi.org/10.3389/fgene.2023.1298574>
218. Hämmerl, L., & Kraya, T. (2024). [Migraine and mitochondrial diseases : Energy deficit in the brain]. *Der Nervenarzt*, 95(2), 169-178. <https://doi.org/10.1007/s00115-024-01622-8>
219. Hong, Y., Zhang, Z., Yangzom, T., Chen, A., Lundberg, B. C., Fang, E. F., Siller, R., Sullivan, G. J., Zeman, J., Tzoulis, C., Bindoff, L. A., & Liang, K. X. (2024). The NAD⁺ Precursor Nicotinamide Riboside Rescues Mitochondrial Defects and Neuronal Loss in iPSC derived Cortical Organoid of Alpers' Disease. *International Journal of Biological Sciences*, 20(4), 1194-1217. <https://doi.org/10.7150/ijbs.91624>
220. Jagadish, S., Calhoun, A. R. U. L., & Ganganna, S. T. (2024). Recurrent super-refractory status epilepticus and stroke like episode in a patient with Behr syndrome secondary to biallelic variants in OPA1 gene. *Epilepsy & Behavior Reports*, 25, 100652. <https://doi.org/10.1016/j.ebr.2024.100652>
221. Kardah, H., Al-Zoubi, H., Odeh, Z., Joumaa, R., & Alasmar, D. (2024). A case report of molybdenum cofactor deficiency type A : The first case diagnosed in Syria. *Annals of Medicine and Surgery* (2012), 86(3), 1762-1765. <https://doi.org/10.1097/MS9.0000000000001778>



L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

222. Kelly, C., Junker, A., Englestad, K., Hirano, M., Trumpff, C., & Picard, M. (2024). Perceived association of mood and symptom severity in adults with mitochondrial diseases. *medRxiv: The Preprint Server for Health Sciences*, 2024.02.02.24302076.
<https://doi.org/10.1101/2024.02.02.24302076>
223. Kemperman, R. H. J., Ganetzky, R. D., & Master, S. R. (2024). Development and validation of a multiplexed LC-MS/MS ketone body assay for clinical diagnostics. *Journal of Mass Spectrometry and Advances in the Clinical Lab*, 31, 49-58.
<https://doi.org/10.1016/j.jmsacl.2024.01.004>
224. Keshavan, N., Minczuk, M., Viscomi, C., & Rahman, S. (2024). Gene therapy for mitochondrial disorders. *Journal of Inherited Metabolic Disease*, 47(1), 145-175.
<https://doi.org/10.1002/jimd.12699>
225. Khaksari, K., Chen, W.-L., Chanvanichtrakool, M., Taylor, A., Kotla, R., & Gropman, A. L. (2024). Applications of near-infrared spectroscopy in epilepsy, with a focus on mitochondrial disorders. *Neurotherapeutics: The Journal of the American Society for Experimental NeuroTherapeutics*, 21(1), e00323.
<https://doi.org/10.1016/j.neurot.2024.e00323>
226. Kojima, F., Okamoto, Y., Ando, M., Higuchi, Y., Hobara, T., Yuan, J., Yoshimura, A., Hashiguchi, A., Matsuura, E., & Takashima, H. (2024). A novel homozygous HPDL variant in Japanese siblings with autosomal recessive hereditary spastic paraplegia : Case report and literature review. *Neurogenetics*, 25(2), 149-156.
<https://doi.org/10.1007/s10048-024-00746-y>
227. Ladero, M., Reche-Sainz, J. A., & Gallardo, M. E. (2024). Hereditary Optic Neuropathies : A Systematic Review on the Interplay between Biomaterials and Induced Pluripotent Stem Cells. *Bioengineering (Basel, Switzerland)*, 11(1), 52.
<https://doi.org/10.3390/bioengineering11010052>
228. Lahham, E. E., Hasassneh, J. J., Adawi, D. O., & Ismail, M. K. (2023). Variants in the SARS2 gene cause HUPRA syndrome with atypical features : Two case reports and review of the literature. *Oxford Medical Case Reports*, 2023(11), omad119.
<https://doi.org/10.1093/omcr/omad119>
229. Lan, X., Ao, W. L., & Li, J. (2024). Preimplantation genetic testing as a preventive strategy for the transmission of mitochondrial DNA disorders. *Systems Biology in Reproductive Medicine*, 70(1), 38-51.
<https://doi.org/10.1080/19396368.2024.2306389>

L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

230. Layrolle, P., Orssaud, C., Leleu, M., Payoux, P., & Chavanas, S. (2024). The Optic Nerve at Stake : Update on Environmental Factors Modulating Expression of Leber's Hereditary Optic Neuropathy. *Biomedicines*, 12(3), 584.
<https://doi.org/10.3390/biomedicines12030584>
231. Li, F., Xiang, R., Liu, Y., Hu, G., Jiang, Q., & Jia, T. (2024). Approaches and challenges in identifying, quantifying, and manipulating dynamic mitochondrial genome variations. *Cellular Signalling*, 117, 111123.
<https://doi.org/10.1016/j.cellsig.2024.111123>
232. Liang, K. X. (2024). The application of brain organoid for drug discovery in mitochondrial diseases. *The International Journal of Biochemistry & Cell Biology*, 170, 106556. <https://doi.org/10.1016/j.biocel.2024.106556>
233. Lin, Y.-H., Lin, K.-L., Wang, X.-W., Lee, J.-J., Wang, F.-S., Wang, P.-W., Lan, M.-Y., Liou, C.-W., & Lin, T.-K. (2024). Miro1 improves the exogenous engraftment efficiency and therapeutic potential of mitochondria transfer using Wharton's jelly mesenchymal stem cells. *Mitochondrion*, 76, 101856.
<https://doi.org/10.1016/j.mito.2024.101856>
234. MacMullen, L. E., George-Sankoh, I., Stanley, K., McCormick, E. M., Muraresku, C. C., Goldstein, A., Zolkipli-Cunningham, Z., & Falk, M. J. (2024). Bridging the clinical-research gap : Harnessing an electronic data capture, integration, and visualization platform to systematically assess prospective patient-reported outcomes in mitochondrial medicine. *Molecular Genetics and Metabolism*, 142(1), 108348.
<https://doi.org/10.1016/j.ymgme.2024.108348>
235. Male, A. J., Holmes, S. L., Koohi, N., Dudziec, M., Hanna, M. G., Ramdharry, G. M., Pizzamiglio, C., Pitceathly, R. D. S., & Kaski, D. (2024). A diagnostic framework to identify vestibular involvement in multi-sensory neurological disease. *European Journal of Neurology*, 31(5), e16216. <https://doi.org/10.1111/ene.16216>
236. Martikainen, M. H., & Majamaa, K. (2024). Incidence and prevalence of mtDNA-related adult mitochondrial disease in Southwest Finland, 2009-2022 : An observational, population-based study. *BMJ Neurology Open*, 6(1), e000546.
<https://doi.org/10.1136/bmjno-2023-000546>
237. Mashkovskaia, A. V., Mariasina, S. S., Serebryakova, M. V., Rubtsova, M. P., Dontsova, O. A., & Sergiev, P. V. (2023). Testing a Hypothesis of 12S rRNA Methylation by Putative METTL17 Methyltransferase. *Acta Naturae*, 15(4), 75-82.
<https://doi.org/10.32607/actanaturae.25441>

L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

238. Menon, D., Nair, S. S., Radhakrishnan, N., Saraf, U. U., & Nair, M. (2023). Clinical Spectrum of Biopsy Proven Mitochondrial Myopathy. *Neurology India*, 71(6), 1192-1196. <https://doi.org/10.4103/0028-3886.391399>
239. Miyauchi, A., Watanabe, C., Yamada, N., Jimbo, E. F., Kobayashi, M., Ohishi, N., Nagayoshi, A., Aoki, S., Kishita, Y., Ohtake, A., Ohno, N., Takahashi, M., Yamagata, T., & Osaka, H. (2024). Apomorphine is a potent inhibitor of ferroptosis independent of dopaminergic receptors. *Scientific Reports*, 14(1), 4820. <https://doi.org/10.1038/s41598-024-55293-1>
240. Moorthy, R., Bhattamisra, S. K., Pandey, M., Mayuren, J., Kow, C. S., & Candasamy, M. (2024). Mitochondria and diabetes : Insights and potential therapies. *Expert Review of Endocrinology & Metabolism*, 19(2), 141-154. <https://doi.org/10.1080/17446651.2024.2307526>
241. Mourokh, L., & Friedman, J. (2024). Mitochondria at the Nanoscale : Physics Meets Biology-What Does It Mean for Medicine? *International Journal of Molecular Sciences*, 25(5), 2835. <https://doi.org/10.3390/ijms25052835>
242. Muñoz-Oreja, M., Sandoval, A., Bruland, O., Perez-Rodriguez, D., Fernandez-Pelayo, U., de Arbina, A. L., Villar-Fernandez, M., Hernández-Eguiaz, H., Hernández, I., Park, Y., Goicoechea, L., Pascual-Frías, N., Garcia-Ruiz, C., Fernandez-Checa, J., Martí-Carrera, I., Gil-Bea, F. J., Hasan, M. T., Gegg, M. E., Bredrup, C., ... Holt, I. J. (2024). Elevated cholesterol in ATAD3 mutants is a compensatory mechanism that leads to membrane cholesterol aggregation. *Brain: A Journal of Neurology*, 147(5), 1899-1913. <https://doi.org/10.1093/brain/awae018>
243. Na, J.-H., & Lee, Y.-M. (2024). Therapeutic outcome of patients with Lennox-Gastaut syndrome with mitochondrial respiratory chain complex I deficiency. *Frontiers in Neurology*, 15, 1305404. <https://doi.org/10.3389/fneur.2024.1305404>
244. Nagy-Grócz, G., Spekter, E., & Vécsei, L. (2024). Kynurenines, Neuronal Excitotoxicity, and Mitochondrial Oxidative Stress : Role of the Intestinal Flora. *International Journal of Molecular Sciences*, 25(3), 1698. <https://doi.org/10.3390/ijms25031698>
245. Nair, L. S., Nurul Jain, J. M., Dalal, A., & Ranganath, P. (2024). Etiologic Spectrum of Pediatric-Onset Leukodystrophies and Genetic Leukoencephalopathies : The Five-Year Experience of a Tertiary Care Center in Southern India. *Pediatric Neurology*, 152, 130-152. <https://doi.org/10.1016/j.pediatrneurol.2023.12.027>

L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

246. Nardecchia, F., Carrozzo, R., Innocenti, A., Torracco, A., Zaccaria, V., Rizza, T., Pisani, F., Bertini, E., & Leuzzi, V. (2024). Biallelic variants in GTPBP3 : New patients, phenotypic spectrum, and outcome. *Annals of Clinical and Translational Neurology*, 11(3), 819-825. <https://doi.org/10.1002/acn3.51980>
247. Nesci, S., & Romeo, G. (2024). H⁺-slip correlated to rotor free-wheeling as cause of F1FO-ATPase dysfunction in primary mitochondrial disorders. *Medicinal Research Reviews*, 44(3), 1183-1188. <https://doi.org/10.1002/med.22013>
248. Nogueira, C., Pereira, C., Silva, L., Laranjeira, M., Lopes, A., Neiva, R., Rodrigues, E., Campos, T., Martins, E., Bandeira, A., Coelho, M., Magalhães, M., Damásio, J., Gaspar, A., Janeiro, P., Gomes, A. L., Ferreira, A. C., Jacinto, S., Vieira, J. P., ... Vilarinho, L. (2024). The genetic landscape of mitochondrial diseases in the next-generation sequencing era : A Portuguese cohort study. *Frontiers in Cell and Developmental Biology*, 12, 1331351. <https://doi.org/10.3389/fcell.2024.1331351>
249. Ouyang, Y., Jeong, M.-Y., Cunningham, C. N., Berg, J. A., Toshniwal, A. G., Hughes, C. E., Seiler, K., Van Vranken, J. G., Cluntun, A. A., Lam, G., Winter, J. M., Akdogan, E., Dove, K. K., Nowinski, S. M., West, M., Odorizzi, G., Gygi, S. P., Dunn, C. D., Winge, D. R., & Rutter, J. (2024). Phosphate starvation signaling increases mitochondrial membrane potential through respiration-independent mechanisms. *eLife*, 13, e84282. <https://doi.org/10.7554/eLife.84282>
250. Papadopoulou-Legbelou, K., Ntounpara, M., Kavga, M., Kotanidou, E. P., Papoulidis, I., Galli-Tsinopoulou, A., & Fotoulaki, M. (2024). Genital Abnormalities and Growth Retardation as Early Signs of Dilated Cardiomyopathy with Ataxia Syndrome. *Case Reports in Genetics*, 2024, 8860889. <https://doi.org/10.1155/2024/8860889>
251. Paripović, A., Maver, A., Stajić, N., Putnik, J., Ostojić, S., Alimpić, B., Ilić, N., & Sarajlija, A. (2023). Expanding the Phenotypic Spectrum : Chronic Kidney Disease in a Patient with Combined Oxidative Phosphorylation Defect 21. *Balkan Journal of Medical Genetics: BJMG*, 26(2), 59-64. <https://doi.org/10.2478/bjmg-2023-0016>
252. Pia, S., & Lui, F. (2024). Melas Syndrome. In *StatPearls*. StatPearls Publishing. <http://www.ncbi.nlm.nih.gov/books/NBK532959/>
253. Pooja, P. S., Greeshma, R., Joy, E. K., & Swamy, S. P. (2024). Auditory Brainstem Response in a Child with Mitochondrial Disorder-Leigh Syndrome. *Indian Journal of Otolaryngology and Head and Neck Surgery: Official Publication of the Association of Otolaryngologists of India*, 76(1), 1014-1017. <https://doi.org/10.1007/s12070-023-03971-3>



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254. Pszczółowska, M., Walczak, K., Miśków, W., Mroziak, M., Chojdak-Łukasiewicz, J., & Leszek, J. (2024). Mitochondrial disorders leading to Alzheimer's disease- perspectives of diagnosis and treatment. *GeroScience*, 46(3), 2977-2988. <https://doi.org/10.1007/s11357-024-01118-y>
255. Ramesh, R., Hariharan, S., & Sundar, L. (2023). Stroke-Like Episodes and Epilepsy in a Patient with COQ8A-Related Coenzyme Q10 Deficiency. *Annals of Indian Academy of Neurology*, 26(6), 980-982. https://doi.org/10.4103/aian.aian_511_23
256. Rehman, A., Mujahid, M., Saba, T., & Jeon, G. (2024). Optimised stacked machine learning algorithms for genomics and genetics disorder detection in the healthcare industry. *Functional & Integrative Genomics*, 24(1), 23. <https://doi.org/10.1007/s10142-024-01289-z>
257. Rho, E. H., Baek, S. I., Lee, H., Seong, M.-W., Chae, J.-H., Park, K. S., & Kwak, S. H. (2024). Clinical Characteristics of Diabetes in People with Mitochondrial DNA 3243A>G Mutation in Korea. *Diabetes & Metabolism Journal*, 48(3), 482-486. <https://doi.org/10.4093/dmj.2023.0078>
258. Rogers, R. S., Sharma, R., Shah, H. B., Skinner, O. S., Guo, X. A., Panda, A., Gupta, R., Durham, T. J., Shaughnessy, K. B., Mayers, J. R., Hibbert, K. A., Baron, R. M., Thompson, B. T., & Mootha, V. K. (2024). Circulating N-lactoyl-amino acids and N-formyl-methionine reflect mitochondrial dysfunction and predict mortality in septic shock. *Metabolomics: Official Journal of the Metabolomic Society*, 20(2), 36. <https://doi.org/10.1007/s11306-024-02089-z>
259. Sabharwal, A., Gupta, V., Kv, S., Kumar Manokaran, R., Verma, A., Mishra, A., Bhoyar, R. C., Jain, A., Sivadas, A., Rawat, S., Jolly, B., Mohanty, S., Gulati, S., Gupta, N., Kabra, M., Scaria, V., & Sivasubbu, S. (2024). Whole genome sequencing followed by functional analysis of genomic deletion encompassing ERCC8 and NDUF2 genes in a non-consanguineous Indian family reveals dysfunctional mitochondrial bioenergetics leading to infant mortality. *Mitochondrion*, 75, 101844. <https://doi.org/10.1016/j.mito.2024.101844>
260. Saha, D., Kothari, S., Kulkarni, S. D., Thambiraja, M., Yennamalli, R. M., & Das, D. K. (2024). Genetic heterogeneity and respiratory chain enzyme analysis in pediatric Indian patients with mitochondrial disorder : Report of novel variants in POLG1 gene and their functional implication using molecular dynamic simulation. *Mitochondrion*, 76, 101870. <https://doi.org/10.1016/j.mito.2024.101870>
261. Saha, L. K., & Pommier, Y. (2024). TOP3A coupling with replication forks and repair of TOP3A cleavage complexes. *Cell Cycle (Georgetown, Tex.)*, 23(2), 115-130. <https://doi.org/10.1080/15384101.2024.2314440>

L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

262. Sakamoto, N., Hamada, S., Takahashi, H., Satou, R., Suzuki, M., & Maeno, T. (2023). Improvement of Intestinal Pseudo-Obstruction by Total Parenteral Nutrition in a Young Woman With Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, and Stroke-Like Episodes : A Case Report. *Cureus*, 15(12), e50075. <https://doi.org/10.7759/cureus.50075>
263. Sercel, A. J., Sturm, G., Gallagher, D., St-Onge, M.-P., Kempes, C. P., Pontzer, H., Hirano, M., & Picard, M. (2024). Hypermetabolism and energetic constraints in mitochondrial disorders. *Nature Metabolism*, 6(2), 192-195. <https://doi.org/10.1038/s42255-023-00968-8>
264. Shafaei Pishabad, Z., & Ledgerwood, E. C. (2024). The Y49H cytochrome c variant enhances megakaryocytic maturation of K-562 cells. *Biochimica Et Biophysica Acta. Molecular Basis of Disease*, 1870(5), 167134. <https://doi.org/10.1016/j.bbadis.2024.167134>
265. Shan, Z., Li, S., Gao, Y., Jian, C., Ti, X., Zuo, H., Wang, Y., Zhao, G., Wang, Y., & Zhang, Q. (2024). mtDNA extramitochondrial replication mediates mitochondrial defect effects. *iScience*, 27(2), 108970. <https://doi.org/10.1016/j.isci.2024.108970>
266. Shayota, B. J. (2024). Biomarkers of mitochondrial disorders. *Neurotherapeutics: The Journal of the American Society for Experimental NeuroTherapeutics*, 21(1), e00325. <https://doi.org/10.1016/j.neurot.2024.e00325>
267. Shemesh, A., Sood, G., Blair, K., & Margolin, E. (2024). Leber Hereditary Optic Neuropathy (LHON). In *StatPearls*. StatPearls Publishing. <http://www.ncbi.nlm.nih.gov/books/NBK482499/>
268. Shen, L., Falk, M. J., & Gai, X. (2024). MSeqDR Quick-Mitome (QM) : Combining Phenotype-Guided Variant Interpretation and Machine Learning Classifiers to Aid Primary Mitochondrial Disease Genetic Diagnosis. *Current Protocols*, 4(1), e955. <https://doi.org/10.1002/cpz1.955>
269. Singh, D. (2024). Organelle Targeted Drug Delivery : Key Challenges, Recent Advancements and Therapeutic Implications. *Endocrine, Metabolic & Immune Disorders Drug Targets*, 24(13), 1480-1487. <https://doi.org/10.2174/0118715303282573240112104035>
270. Snyder, H. E., Jain, P., RamachandranNair, R., Jones, K. C., & Whitney, R. (2024). Genetic Advancements in Infantile Epileptic Spasms Syndrome and Opportunities for Precision Medicine. *Genes*, 15(3), 266. <https://doi.org/10.3390/genes15030266>

L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

271. Song, M., Chen, S., Zhang, M., Hu, S., Lei, W., & Yu, M. (2024). Generation of a human induced pluripotent stem cell line harboring heteroplasmic m.3243A > G mutation in MT-TL1 gene. *Stem Cell Research*, 77, 103387. <https://doi.org/10.1016/j.scr.2024.103387>
272. Song, M., Ye, L., Yan, Y., Li, X., Han, X., Hu, S., & Yu, M. (2024). Mitochondrial diseases and mtDNA editing. *Genes & Diseases*, 11(3), 101057. <https://doi.org/10.1016/j.gendis.2023.06.026>
273. Sonuç Kartal, G., Koç Yekedüz, M., Köse, E., & Eminoğlu, F. T. (2024). Two Turkish patients with Primary Coenzyme Q10 Deficiency-7 : Case report and literature review. *Journal of Pediatric Endocrinology & Metabolism: JPEM*, 37(3), 260-270. <https://doi.org/10.1515/jpem-2023-0490>
274. Spencer, K. A., Howe, M. N., Mulholland, M. T., Truong, V., Liao, R. W., Chen, Y., Setha, M., Snell, J. C., Hanaford, A., James, K., Morgan, P. G., Sedensky, M. M., & Johnson, S. C. (2024). Impact of dietary ketosis on volatile anesthesia toxicity in a model of Leigh syndrome. *Paediatric Anaesthesia*, 34(5), 467-476. <https://doi.org/10.1111/pan.14855>
275. Sun, F., Fang, M., Zhang, H., Song, Q., Li, S., Li, Y., Jiang, S., & Yang, L. (2024). Drp1 : Focus on Diseases Triggered by the Mitochondrial Pathway. *Cell Biochemistry and Biophysics*, 82(2), 435-455. <https://doi.org/10.1007/s12013-024-01245-5>
276. Sun, Y.-C., Chou, Y.-P., Ho, P.-H., Chen, X.-X., Chen, P.-Y., Chu, C.-H., & Lin, H.-C. (2024). Bilateral cochlear implants in a MELAS patient. *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(6), 3265-3268. <https://doi.org/10.1007/s00405-024-08532-0>
277. Takai, Y., Iwasa, M., Yamagami, A., Inoue, K., Yasumoto, R., Ishikawa, H., & Wakakura, M. (2024). A Japanese Case of Leber's Hereditary Optic Neuropathy with the m.13051G>A Pathogenic Variant. *Neuro-Ophthalmology (Aeolus Press)*, 48(1), 51-55. <https://doi.org/10.1080/01658107.2023.2273480>
278. Vincent, A. E., Chen, C., Gomes, T. B., Di Leo, V., Laalo, T., Pabis, K., Capaldi, R., Marusich, M. F., McDonald, D., Filby, A., Fuller, A., Lehmann Urban, D., Zierz, S., Deschauer, M., Turnbull, D., Reeve, A. K., & Lawless, C. (2024). A stagewise response to mitochondrial dysfunction in mitochondrial DNA maintenance disorders. *Biochimica Et Biophysica Acta. Molecular Basis of Disease*, 1870(5), 167131. <https://doi.org/10.1016/j.bbadis.2024.167131>

L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

279. Walimbe, A. S., Machol, K., Kralik, S. F., Mizerik, E. A., Gofin, Y., Bekheirnia, M. R., Gijavanekar, C., Elsea, S. H., Emrick, L. T., & Scaglia, F. (2024). Expanded clinical phenotype and untargeted metabolomics analysis in RARS2-related mitochondrial disorder : A case report. *BMC Neurology*, 24(1), 87. <https://doi.org/10.1186/s12883-024-03571-w>
280. Wang, B., Yu, W., Zhang, W., Zhang, M., Niu, Y., Jin, X., Zhang, J., Sun, D., Li, H., Zhang, Z., Luo, Q., Cheng, X., Niu, J., Cai, G., Chen, X., & Chen, Y. (2024). Enhanced TRPC3 transcription through AT1R/PKA/CREB signaling contributes to mitochondrial dysfunction in renal tubular epithelial cells in D-galactose-induced accelerated aging mice. *Aging Cell*, 23(6), e14130. <https://doi.org/10.1111/ace1.14130>
281. Wang, Y., He, J., Dong, F., Shou, W., Feng, X., Yang, Y., Li, C., Wang, J., Li, B., & Xiao, S. (2024). A novel mutation in GTPBP3 causes combined oxidative phosphorylation deficiency 23 by affecting pre-mRNA splicing. *Heliyon*, 10(6), e27199. <https://doi.org/10.1016/j.heliyon.2024.e27199>
282. Wesół-Kucharska, D., Greczan, M., Kaczor, M., Ehmke Vel Emczyńska-Seliga, E., Hajdacka, M., Czekuć-Kryśkiewicz, E., Piekutowska-Abramczuk, D., Halat-Wolska, P., Ciara, E., Jaworski, M., Jezela-Stanek, A., & Rokicki, D. (2024). Efficacy and Safety of Ketogenic Diet Treatment in Pediatric Patients with Mitochondrial Disease. *Nutrients*, 16(6), 812. <https://doi.org/10.3390/nu16060812>
283. Westerlund, E., Marelsson, S. E., Karlsson, M., Sjövall, F., Chamkha, I., Åsander Frostner, E., Lundgren, J., Fellman, V., Eklund, E. A., Steding-Ehrenborg, K., Darin, N., Paul, G., Hansson, M. J., Ehinger, J. K., & Elmér, E. (2024). Correlation of mitochondrial respiration in platelets, peripheral blood mononuclear cells and muscle fibers. *Heliyon*, 10(5), e26745. <https://doi.org/10.1016/j.heliyon.2024.e26745>
284. Wongkittichote, P., Pantano, C., He, M., Hong, X., & Demczko, M. M. (2024). Clinical, biochemical and molecular characterization of a new case with FDX2-related mitochondrial disorder : Potential biomarkers and treatment options. *JIMD Reports*, 65(2), 102-109. <https://doi.org/10.1002/jmd2.12408>
285. Xia, Y.-M., Guan, Y.-Q., Liang, J.-F., & Wu, W.-D. (2024). TAK-242 improves sepsis-associated acute kidney injury in rats by inhibiting the TLR4/NF-κB signaling pathway. *Renal Failure*, 46(1), 2313176. <https://doi.org/10.1080/0886022X.2024.2313176>
286. Yazdani, M. (2024). Cellular and Molecular Responses to Mitochondrial DNA Deletions in Kearns-Sayre Syndrome : Some Underlying Mechanisms. *Molecular Neurobiology*, 61(8), 5665-5679. <https://doi.org/10.1007/s12035-024-03938-7>

L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

287. Yin, Z., Agip, A.-N. A., Bridges, H. R., & Hirst, J. (2024). Structural insights into respiratory complex I deficiency and assembly from the mitochondrial disease-related ndufs4^{-/-} mouse. *The EMBO Journal*, 43(2), 225-249. <https://doi.org/10.1038/s44318-023-00001-4>
288. Yu-Wai-Man, P., Carelli, V., Newman, N. J., Silva, M. J., Linden, A., Van Stavern, G., Szaflik, J. P., Banik, R., Lubiński, W., Pemp, B., Liao, Y. J., Subramanian, P. S., Misiuk-Hojto, M., Newman, S., Castillo, L., Kocięcki, J., Levin, M. H., Muñoz-Negrete, F. J., Yagan, A., ... LEROS Study Group. (2024). Therapeutic benefit of idebenone in patients with Leber hereditary optic neuropathy : The LEROS nonrandomized controlled trial. *Cell Reports. Medicine*, 5(3), 101437. <https://doi.org/10.1016/j.xcrm.2024.101437>
289. Zanfardino, P., Amati, A., Doccini, S., Cox, S. N., Tullo, A., Longo, G., D'Erchia, A., Picardi, E., Nesti, C., Santorelli, F. M., & Petruzzella, V. (2024). OPA1 mutation affects autophagy and triggers senescence in autosomal dominant optic atrophy plus fibroblasts. *Human Molecular Genetics*, 33(9), 768-786. <https://doi.org/10.1093/hmg/ddae008>
290. Zhang, J., Gan, J., & Wang, J. (2024). A case of Leigh syndrome presented with paroxysmal body swing. *Heliyon*, 10(1), e23137. <https://doi.org/10.1016/j.heliyon.2023.e23137>
291. Zhang, J.-H., Eriani, G., & Zhou, X.-L. (2024). Pathophysiology of human mitochondrial tRNA metabolism. *Trends in Endocrinology and Metabolism: TEM*, 35(4), 285-289. <https://doi.org/10.1016/j.tem.2024.01.002>
292. Zhang, L.-W., Feng, H.-Q., Fu, S.-B., & Sun, D.-J. (2024). Low Selenium and Low Protein Exacerbate Myocardial Damage in Keshan Disease by Affecting the PINK1/Parkin-mediated Mitochondrial Autophagy Pathway. *Current Medical Science*, 44(1), 93-101. <https://doi.org/10.1007/s11596-024-2834-x>
293. Zhang, Z., Bie, X., Chen, Z., Liu, J., Xie, Z., Li, X., Xiao, M., Zhang, Q., Zhang, Y., Yang, Y., & Li, D. (2024). A novel variant of DNMT1L expanding the clinical phenotypic spectrum: A case report and literature review. *BMC Pediatrics*, 24(1), 104. <https://doi.org/10.1186/s12887-023-04442-y>
294. Zhao, Y., Xu, Z., Yan, C., & Ji, K. (2024). Unraveling the Diagnostic Puzzle : Minor Stroke-Like Lesions and Normal Muscle Histopathology in MELAS Syndrome. *Stroke*, 55(4), e127-e130. <https://doi.org/10.1161/STROKEAHA.123.045984>

L'essentielle Maladies Mitochondriales

MMITO-2024-1 du 1^{er} janvier au 31 mars 2024

295. Zheng, Q., Liu, H., Gao, Y., Cao, G., Wang, Y., & Li, Z. (2024). Ameliorating Mitochondrial Dysfunction for the Therapy of Parkinson's Disease. *Small (Weinheim an Der Bergstrasse, Germany)*, 20(29), e2311571.
<https://doi.org/10.1002/sml.202311571>
296. Zhou, F., Yi, G., Liu, X., Sheng, W., Shu, J., Li, D., & Cai, C. (2024). A Pair of Compound Heterozygous IARS2 Variants Manifesting West Syndrome and Electrolyte Disorders in a Chinese Patient. *Global Medical Genetics*, 11(1), 25-28.
<https://doi.org/10.1055/s-0043-1778091>