

L'essentielle Maladies Mitochondriales
MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

L'essentielle Maladies Mitochondriales



4^{ème} trimestre 2024



Veille bibliographique trimestrielle réalisée par le pôle documentation de l'AFM-Téléthon

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



L'essentielle Maladies Mitochondriales
MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Sommaire

ATAXIE DE FRIEDREICH - <i>FRIEDREICH ATAXIA</i>	3
ATROPHIE OPTIQUE AUTOSOMIQUE DOMINANTE – <i>ADOA</i>	6
ENCEPHALOPATHIE MYO-NEURO-GASTROINTESTINALE - <i>MNGIE</i>	7
MALADIE DE CHARCOT-MARIE-TOOTH D’ORIGINE MITOCHONDRIALE – <i>MITOCHONDRIAL CMT</i>	8
MALADIES LIEES AU GENE ACO2 - <i>ACO2-RELATED DISORDERS</i>	9
MALADIE LIEE AU GENE GFM1 – <i>GFM1-RELATED DISORDER</i>	9
MALADIES LIEES AU GENE POLG (INCLUS SANDO, SCAE) - <i>POLG-RELATED DISORDERS</i>	9
MELAS	10
MERFF	12
NEUROPATHIE OPTIQUE HEREDITAIRE DE LEBER - <i>LHON</i>	13
OPHTALMOPLÉGIE EXTERNE PROGRESSIVE – <i>PEO</i>	15
SYNDROME D'ALPERS-HUTTENLOCHER - <i>ALPERS-HUTTENLOCHER SYNDROME</i>	16
SYNDROME DE KEARNS-SAYRE – <i>KEARNS-SAYRE SYNDROME</i>	16
SYNDROME DE LEIGH - <i>LEIGH SYNDROME</i>	17
MALADIES MITOCHONDRIALES (AUTRES) – <i>MITOCHONDRIAL DISORDERS (OTHER)</i>	18



L'essentielle Maladies Mitochondriales

MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Ataxie de Friedreich - *Friedreich ataxia*

Alemaný-Perna, B., Tamarit, J., Cabiscol, E. et Ros, J. (2024). **Reply to: « Calcitriol in Friedreich Ataxia »**. *Mov Disord* 39, 1654-1655, doi:[10.1002/mds.29971](https://doi.org/10.1002/mds.29971).

Bidichandani, S. I., Delatycki, M. B., Napierala, M. et Duquette, A. (2024). **Friedreich Ataxia**. In: **GeneReviews®**, Éd. Adam, M. P., Feldman, J., Mirzaa, G. M., Pagon, R. A., Wallace, S. E., et Amemiya, A. University of Washington, Seattle, Seattle (WA), URL: <http://www.ncbi.nlm.nih.gov/books/NBK1281/> [Consulté le janvier 2025].

Brito, R., Fabrício, J. V., Araujo, A., Sacchi, M., Baltar, A., Lima, F. A., Ribeiro, A. C., Sousa, B., Santos, C., Tanaka, C. et Monte-Silva, K. (2024). **Differential Effects of Cerebellar Transcranial Direct Current Stimulation with Gait Training on Functional Mobility, Balance, and Ataxia Symptoms**. *Cerebellum* 23, 2457-2467, doi:[10.1007/s12311-024-01750-6](https://doi.org/10.1007/s12311-024-01750-6).

Buchholz, M., Pfaff, M., Iskandar, A., Reetz, K., Schulz, J. B., Grobe-Einsler, M., Klockgether, T., Michalowsky, B., et EFACTS Study Group (2024). **Health-Related Quality of Life in Patients with Friedreich Ataxia Using Mobility Assistive Technologies: Limited Fit of the EQ-5D-3L Mobility Dimension**. *Neurol Ther*, doi:[10.1007/s40120-024-00694-7](https://doi.org/10.1007/s40120-024-00694-7).

Chartier, C., Godard, J., Durand, S., Humeau-Heurtier, A., Menetrier, E., Allain, P. et Besnard, J. (2024). **Combinations of physical and cognitive training for subcortical neurodegenerative diseases with physical, cognitive and behavioral symptoms: a systematic review**. *Neurol Sci* 45, 5571-5589, doi:[10.1007/s10072-024-07808-x](https://doi.org/10.1007/s10072-024-07808-x).

Ercanbrack, W. S., Dungan, A., Gaul, E., Ramirez, M., J DelVecchio, A., Grass, C. et Wingert, R. A. (2024). **Frataxin is essential for zebrafish embryogenesis and pronephros formation**. *Front Cell Dev Biol* 12, 1496244, doi:[10.3389/fcell.2024.1496244](https://doi.org/10.3389/fcell.2024.1496244).

Fitisemanu, F. M. et Padilla-Benavides, T. (2024). **Emerging perspectives of copper-mediated transcriptional regulation in mammalian cell development**. *Metallomics* 16, mfae046, doi:[10.1093/mtomcs/mfae046](https://doi.org/10.1093/mtomcs/mfae046).

Gerhart, B. J., Pellerin, D., Danzi, M. C., Zuchner, S., Brais, B., Matos-Rodrigues, G., Nussenzweig, A., Usdin, K., Park, C. C., Napierala, J. S., Lynch, D. R. et Napierala, M. (2024). **Assessment of the Clinical Interactions of GAA Repeat Expansions in FGF14 and FXN**. *Neurol Genet* 10, e200210, doi:[10.1212/NXG.0000000000200210](https://doi.org/10.1212/NXG.0000000000200210).

L'essentielle Maladies Mitochondriales

MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Gunawardene, A. N., Reyes, N., Valdes-Arias, D., Ortug, A., Martinez, J., Galor, A. et Moulton, E. A. (2024). **Abnormal visual cortex activity using functional magnetic resonance imaging in treatment resistant photophobia in Friedreich Ataxia.** Am J Ophthalmol Case Rep 36, 102213, doi:[10.1016/j.ajoc.2024.102213](https://doi.org/10.1016/j.ajoc.2024.102213).

Hayiroğlu, M. İ., Kalenderoğlu, K. et Gürkan, K. (2024). **Tachycardiomyopathy Treated With Ablation by Using 3D Mapping System in a Patient With Friedreich Ataxia.** Pacing Clin Electrophysiol, doi:[10.1111/pace.15125](https://doi.org/10.1111/pace.15125).

Hisey, J. A., Masnovi, C. et Mirkin, S. M. (2024). **Triplex H-DNA structure: the long and winding road from the discovery to its role in human disease.** NAR Mol Med 1, ugae024, doi:[10.1093/narmme/ugae024](https://doi.org/10.1093/narmme/ugae024).

Karamazovova, S., Stovickova, L., Jester, D. J., Matuskova, V., Paulasova-Schwabova, J., Kuzmiak, M., Zumrova, A., Andel, R. et Vyhnaček, M. (2024). **Exploring neuropsychiatric symptoms in Friedreich ataxia.** Sci Rep 14, 29076, doi:[10.1038/s41598-024-80258-9](https://doi.org/10.1038/s41598-024-80258-9).

Kwa, F. A. A. et Kendal, E. (2024). **Precision medicine and Friedreich ataxia: promoting equity, beneficence, and informed consent for novel gene therapies.** Int J Equity Health 23, 230, doi:[10.1186/s12939-024-02318-w](https://doi.org/10.1186/s12939-024-02318-w).

Lee, L., Flach, S., Xue, H., Arivelu, L., Golden, L., Kong, R. et Darpo, B. (2024). **Lack of Concentration-QTc Relationship and Cardiac Risk With Vatiquinone Therapeutic and Supratherapeutic Doses.** Clin Pharmacol Drug Dev 13, 1227-1238, doi:[10.1002/cpdd.1476](https://doi.org/10.1002/cpdd.1476).

Lenahan, A., Yano, S., Graham, B., Sen, K., et ACMG Therapeutics Committee5*documents@acmg.net (2023). **Omaveloxolone approved for patients aged 16 years and older with Friedreich ataxia (FRDA): A therapeutics bulletin of the American College of Medical Genetics and Genomics (ACMG).** Genet Med Open 1, 100832, doi:[10.1016/j.gimo.2023.100832](https://doi.org/10.1016/j.gimo.2023.100832).

Li, L., Scott, W. S., Khristich, A. N., Armenia, J. F. et Mirkin, S. M. (2024). **Recurrent DNA nicks drive massive expansions of (GAA)_n repeats.** Proc Natl Acad Sci U S A 121, e2413298121, doi:[10.1073/pnas.2413298121](https://doi.org/10.1073/pnas.2413298121).

Marullo, C., Croci, L., Giupponi, I., Rivoletti, C., Zuffetti, S., Bettegazzi, B., Cremona, O., Giunti, P., Ambrosi, A., Casoni, F., Consalez, G. G. et Codazzi, F. (2025). **Altered Ca²⁺ responses and antioxidant properties in Friedreich's ataxia-like cerebellar astrocytes.** J Cell Sci 138, jcs263446, doi:[10.1242/jcs.263446](https://doi.org/10.1242/jcs.263446).

L'essentielle Maladies Mitochondriales
MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Masnovi, C., Paleiov, Z., Dovrat, D., Baxter, L. K., Movafaghi, S., Aharoni, A. et Mirkin, S. M. (2024). **Stabilization of expandable DNA repeats by the replication factor Mcm10 promotes cell viability**. *Nat Commun* 15, 10532, doi:[10.1038/s41467-024-54977-6](https://doi.org/10.1038/s41467-024-54977-6).

McCormack, S. E., Weber, D. R. et Lynch, D. R. (2024). **Calcitriol in Friedreich Ataxia**. *Mov Disord* 39, 1653-1654, doi:[10.1002/mds.29970](https://doi.org/10.1002/mds.29970).

Mishra, R. K., Nunes, A. S., Enriquez, A., Profeta, V. R., Wells, M., Lynch, D. R. et Vaziri, A. (2024). **At-home wearable-based monitoring predicts clinical measures and biological biomarkers of disease severity in Friedreich's Ataxia**. *Commun Med (Lond)* 4, 217, doi:[10.1038/s43856-024-00653-1](https://doi.org/10.1038/s43856-024-00653-1).

Mutlu, M. B., Karakaya, T., Çelebi, H. B. G., Duymuş, F., Seyhan, S., Yılmaz, S., Yiş, U., Atik, T., Yetkin, M. F. et Gümüş, H. (2025). **Utility of Optical Genome Mapping in Repeat Disorders**. *Clin Genet* 107, 188-195, doi:[10.1111/cge.14633](https://doi.org/10.1111/cge.14633).

Naghipour, S., Corben, L. A., Hulme, A. J., Dottori, M., Delatycki, M. B., Lees, J. G. et Lim, S. Y. (2024). **Omaveloxolone for the Treatment of Friedreich Ataxia: Efficacy, Safety, and Future Perspectives**. *Mov Disord*, doi:[10.1002/mds.30070](https://doi.org/10.1002/mds.30070).

Ozaki, K., Yatsuka, Y., Oyazato, Y., Nishiyama, A., Nitta, K. R., Kishita, Y., Fushimi, T., Shimura, M., Noma, S., Sugiyama, Y., Tagami, M., Fukunaga, M., Kinoshita, H., Hirata, T., Suda, W., Murakawa, Y., Carninci, P., Ohtake, A., Murayama, K. et Okazaki, Y. (2024). **Biallelic GGGCC repeat expansion leading to NAXE-related mitochondrial encephalopathy**. *NPJ Genom Med* 9, 48, doi:[10.1038/s41525-024-00429-5](https://doi.org/10.1038/s41525-024-00429-5).

Peluzzo, T. M., Vieira, A. S., Matos, A. H. B., Silveira, C., Martin, M., Filho, O. R. C., Rezende, T. J. R., Martinez, A. R. M. et França, M. C. (2024). **Plasma miRNAs Correlate with Structural Brain and Cardiac Damage in Friedreich's Ataxia**. *Cerebellum* 24, 15, doi:[10.1007/s12311-024-01766-y](https://doi.org/10.1007/s12311-024-01766-y).

Petrillo, S., Perna, A., Quatrana, A., Silvestri, G., Bertini, E., Piemonte, F. et Santoro, M. (2024). **Differential Gene Expression in Late-Onset Friedreich Ataxia: A Comparative Transcriptomic Analysis Between Symptomatic and Asymptomatic Sisters**. *Int J Mol Sci* 25, 11615, doi:[10.3390/ijms252111615](https://doi.org/10.3390/ijms252111615).

Ramakrishnan, S., Shah, M. et Gupta, V. (2024). **Trinucleotide Repeat Disorders**. In: **StatPearls**, StatPearls Publishing, Treasure Island (FL), URL: <http://www.ncbi.nlm.nih.gov/books/NBK559254/> [Consulté le janvier 2025].

L'essentielle Maladies Mitochondriales
MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Salinas, L., Montgomery, C. B., Figueroa, F., Thai, P. N., Chiamvimonvat, N., Cortopassi, G. et Dedkova, E. N. (2024). **Sexual dimorphism in a mouse model of Friedreich's ataxia with severe cardiomyopathy.** Commun Biol 7, 1250, doi:[10.1038/s42003-024-06962-4](https://doi.org/10.1038/s42003-024-06962-4).

Sperelakis-Beedham, B., Gitiaux, C., Rajaoba, M., Magen, M., Derive, N., Chansard, J., de Sainte Agathe, J.-M., Maurin, M.-L., Assouline, Z., Barnerias, C., Desguerre, I., Steffann, J. et Barcia, G. (2025). **Uniparental IsoDisomy: a case study on a new mechanism of Friedreich ataxia.** Eur J Hum Genet 33, 137-140, doi:[10.1038/s41431-024-01728-2](https://doi.org/10.1038/s41431-024-01728-2).

Umrao, A., Pahuja, M. et Chatterjee, N. S. (2024). **Safety and efficacy of omaveloxolone v/s placebo for the treatment of Friedreich's ataxia in patients aged more than 16 years: a systematic review.** Orphanet J Rare Dis 19, 495, doi:[10.1186/s13023-024-03474-6](https://doi.org/10.1186/s13023-024-03474-6).

Venturiello, D., Tiberi, P. G., Perulli, F., Nardoiani, G., Guida, L., Barsali, C., Terrone, C., Cianca, A., Lustri, C., Sclafani, M., Tini, G., Barbato, E. et Musumeci, B. (2024). **Unveiling the Future of Cardiac Care: A Review of Gene Therapy in Cardiomyopathies.** Int J Mol Sci 25, 13147, doi:[10.3390/ijms252313147](https://doi.org/10.3390/ijms252313147).

Vo, A. T. T., Khan, U., Liopo, A. V., Mouli, K., Olson, K. R., McHugh, E. A., Tour, J. M., Pooparayil Manoj, M., Derry, P. J. et Kent, T. A. (2024). **Harshly Oxidized Activated Charcoal Enhances Protein Persulfidation with Implications for Neurodegeneration as Exemplified by Friedreich's Ataxia.** Nanomaterials (Basel) 14, 2007, doi:[10.3390/nano14242007](https://doi.org/10.3390/nano14242007).

Atrophie optique autosomique dominante – ADOA

Bureau, J., Manero, F., Baris, O., Bodin, A., Verny, C., Chevrollier, A., Lenaers, G. et Codron, P. (2024). **Opa1 and MT-Nd6 mutations induce early mitochondrial changes in the retina and prelaminar optic nerve of hereditary optic neuropathy mouse models.** Brain Commun 6, fcae404, doi:[10.1093/braincomms/fcae404](https://doi.org/10.1093/braincomms/fcae404).

de Muijnck, C., Haer-Wigman, L., van Everdingen, J. A. M., Lushchik, T., Heutinck, P. A. T., van Dooren, M. F., Kievit, A. J. A., Verhoeven, V. J. M., Simon, M. E. H., Wasmann, R. A., Notting, I. C., De Baere, E., Walraedt, S., De Zaeytijd, J., Van den Broeck, F., Leroy, B. P., Boon, C. J. F. et van Genderen, M. M. (2024). **Characteristics of autosomal dominant WFS1-associated optic neuropathy and its comparability to OPA1-associated autosomal dominant optic atrophy.** Sci Rep 14, 22956, doi:[10.1038/s41598-024-74364-X](https://doi.org/10.1038/s41598-024-74364-X).



L'essentielle Maladies Mitochondriales

MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Kleerekooper, I., Verschueren, D. V., Trip, S. A., Plant, G. T. et Petzold, A. (2024). **Ellipsoid Zone Reflectivity: Exploring its Potential as a Novel Non-Invasive Biomarker for Assessing Mitochondrial Function.** *Neuroophthalmology* 48, 417-428, doi:[10.1080/01658107.2024.2341769](https://doi.org/10.1080/01658107.2024.2341769).

Macuada, J., Molina-Riquelme, I., Vidal, G., Pérez-Bravo, N., Vásquez-Trincado, C., Aedo, G., Lagos, D., Yu-Wai-Man, P., Horvath, R., Rudge, T. J., Cartes-Saavedra, B. et Eisner, V. (2024). **OPA1 and disease-causing mutants perturb mitochondrial nucleoid distribution.** *Cell Death Dis* 15, 870, doi:[10.1038/s41419-024-07165-9](https://doi.org/10.1038/s41419-024-07165-9).

Wang, Chenghui, Zhang, L., Nie, Z., Liang, M., Liu, H., Yi, Q., Wang, Chunyan, Ai, C., Zhang, J., Gao, Y., Ji, Y. et Guan, M.-X. (2024). **Mutation of CRYAB encoding a conserved mitochondrial chaperone and antiapoptotic protein causes hereditary optic atrophy.** *JCI Insight* 10, e182209, doi:[10.1172/jci.insight.182209](https://doi.org/10.1172/jci.insight.182209).

Zheng, Y., Wang, P., Li, S., Long, Y., Jiang, Y., Guo, D., Jia, X., Liu, M., Zeng, Y., Xiao, X., Hejtmancik, J. F., Zhang, Q. et Sun, W. (2024). **Clinical and genetic landscape of optic atrophy in 826 families: insights from 50 nuclear genes.** *Brain* awae324, doi:[10.1093/brain/awae324](https://doi.org/10.1093/brain/awae324).

Encéphalopathie myo-neuro-gastrointestinale - MNGIE

Finsterer, J. et Mehri, S. (2024). **Letter re: Aseptic pleocytosis can only be classified as a phenotypic manifestation of MNGIE after exclusion of all differential causes.** *J Cent Nerv Syst Dis* 16, 11795735241292198, doi:[10.1177/11795735241292198](https://doi.org/10.1177/11795735241292198).

Hanna, A., Ing, E. B., Sundaram, A. N., Bril, V. et Sharma, R. A. (2024). **Late-Onset Mitochondrial Neurogastrointestinal Encephalopathy Presenting With Isolated Ophthalmic Findings.** *J Neuroophthalmol*, doi:[10.1097/WNO.0000000000002287](https://doi.org/10.1097/WNO.0000000000002287).

Sun, Y. H., Bai, X. Y., Guo, T., Fan, S. Y., Ruan, G. C., Zhou, W. X. et Yang, H. (2024). **Rare digestive disease: Mitochondrial neurogastrointestinal encephalomyopathy, review of the literature.** *J Dig Dis* 25, 624-631, doi:[10.1111/1751-2980.13317](https://doi.org/10.1111/1751-2980.13317).



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

Abedidoust, S., Badv, R.-S., Saliyani, A. et Azari-Yam, A. (2024). **Peripheral Neuropathy in Mitochondrial Trifunctional Protein Deficiency due to a Variant in HADHA Gene.** Iran J Pathol 19, 355-358, doi:[10.30699/IJP.2024.2010490.3163](https://doi.org/10.30699/IJP.2024.2010490.3163).

Guerra San Juan, I., Brunner, J. W., Eggan, K., Toonen, R. F. et Verhage, M. (2025). **KIF5A regulates axonal repair and time-dependent axonal transport of SFPQ granules and mitochondria in human motor neurons.** Neurobiol Dis 204, 106759, doi:[10.1016/j.nbd.2024.106759](https://doi.org/10.1016/j.nbd.2024.106759).

Kam, J. et Momoh, R. (2024). **Rhabdomyolysis in a Patient With a Possible Mitochondrial Pathogenic Variant in the Peri-operative Period: A Case Report.** Cureus 16, e73577, doi:[10.7759/cureus.73577](https://doi.org/10.7759/cureus.73577).

Labat-de-Hoz, L., Jiménez, M. Á., Correas, I. et Alonso, M. A. (2024). **Regulation of formin INF2 and its alteration in INF2-linked inherited disorders.** Cell Mol Life Sci 81, 463, doi:[10.1007/s00018-024-05499-3](https://doi.org/10.1007/s00018-024-05499-3).

Mohammed, S., Kifelew, S., Tsehayneh, F., Haile, A. T. et Gebrehiwot, E. H. (2024). **Genetically confirmed Charcot-Marie-Tooth disease type 2A manifesting with postural tremor: a case report.** J Med Case Rep 18, 571, doi:[10.1186/s13256-024-04945-x](https://doi.org/10.1186/s13256-024-04945-x).

Sisto, A., van Wermeskerken, T., Pancher, M., Gatto, P., Asselbergh, B., Assunção Carreira, Á. S., De Winter, V., Adami, V., Provenzani, A. et Timmerman, V. (2024). **Autophagy induction by piplartine ameliorates axonal degeneration caused by mutant HSPB1 and HSPB8 in Charcot-Marie-Tooth type 2 neuropathies.** Autophagy 1-28, doi:[10.1080/15548627.2024.2439649](https://doi.org/10.1080/15548627.2024.2439649).

Subramanian, B., Williams, S., Karp, S., Hennino, M.-F., Jacas, S., Lee, M., Riella, C. V., Alper, S. L., Higgs, H. N. et Pollak, M. R. (2024). **INF2 mutations cause kidney disease through a gain-of-function mechanism.** Sci Adv 10, eadr1017, doi:[10.1126/sciadv.adr1017](https://doi.org/10.1126/sciadv.adr1017).

Živković, S. A., Nowak, R. J. et DiCapua, D. (2024). **CMT2 and distal hereditary motor neuropathy associated with VRK1 variants: Case series.** Neuromuscul Disord 47, 105254, doi:[10.1016/j.nmd.2024.105254](https://doi.org/10.1016/j.nmd.2024.105254).

Maladies liées au gène ACO2 - ACO2-related disorders

Yang, W., Wang, S., Yang, K., Li, Y., Guo, Z., Huang, J., Wang, J. et Liao, S. (2024). **Transcriptome analyses reveal molecular mechanisms of novel compound heterozygous ACO2 variants causing infantile cerebellar retinal degeneration.** Front Cell Neurosci 18, 1492048, doi:[10.3389/fncel.2024.1492048](https://doi.org/10.3389/fncel.2024.1492048).

Zheng, Y., Wang, P., Li, S., Long, Y., Jiang, Y., Guo, D., Jia, X., Liu, M., Zeng, Y., Xiao, X., Hejtmancik, J. F., Zhang, Q. et Sun, W. (2024). **Clinical and genetic landscape of optic atrophy in 826 families: insights from 50 nuclear genes.** Brain awae324, doi:[10.1093/brain/awae324](https://doi.org/10.1093/brain/awae324).

Maladie liée au gène GFM1 – GFM1-related disorder

Pas de résultat sur la période

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

Carvalho Araújo, L., Lopes Tenório de Cerqueira, I. C. et de Aguiar Coelho Silva Madeiro, B. (2025). **Teaching Neurolmage: Notable MRI Findings in Sensory Ataxia Neuropathy, Dysarthria, and Ophthalmoplegia (SANDO), a POLG-Related Disorder.** Neurology 104, e210227, doi:[10.1212/WNL.0000000000210227](https://doi.org/10.1212/WNL.0000000000210227).

Hu, W., Shi, C., Guo, H. et Zhang, B. (2024). **POLG p.A962T Mutation Leads to Neuronal Mitochondrial Dysfunction That is Restored After Mitochondrial Transplantation.** Physiol Res 73, 801-808, doi:[10.33549/physiolres.935313](https://doi.org/10.33549/physiolres.935313).

Karaa, A., Bertini, E., Carelli, V., Cohen, B., Ennes, G. M., Falk, M. J., Goldstein, A., Gorman, G., Haas, R., Hirano, M., Klopstock, T., Koenig, M. K., Kornblum, C., Lamperti, C., Lehman, A., Longo, N., Molnar, M. J., Parikh, S., Phan, H., Pitceathly, R. D. S., Saneto, R., Scaglia, F., Servidei, S., Tarnopolsky, M., Toscano, A., Van Hove, J. L. K., Vissing, J., Vockley, J., Finman, J. S., Abbruscato, A., Brown, D. A., Sullivan, A., Shiffer, J. A., Mancuso, M., et MMPOWER-3 Trial Investigators (2024). **Genotype-specific effects of elamipretide in patients with primary mitochondrial myopathy: a post hoc analysis of the MMPOWER-3 trial.** Orphanet J Rare Dis 19, 431, doi:[10.1186/s13023-024-03421-5](https://doi.org/10.1186/s13023-024-03421-5).

Sun, H., Zhang, G., Li, N. et Bu, X. (2024). **Molecular diagnosis of patients with syndromic short stature identified by trio whole-exome sequencing.** Front Genet 15, 1399186, doi:[10.3389/fgene.2024.1399186](https://doi.org/10.3389/fgene.2024.1399186).



L'essentielle Maladies Mitochondriales

MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

MELAS

Akamatsu, S., Mitsuhashi, S., Soga, K., Mizukami, H., Shiraishi, M., Frith, M. C. et Yamano, Y. (2024). **Targeted nanopore sequencing using the Flongle device to identify mitochondrial DNA variants**. Sci Rep 14, 25161, doi:[10.1038/s41598-024-75749-8](https://doi.org/10.1038/s41598-024-75749-8).

Alten, B., Chu, C. J., Rost, N. S. et Walker, M. A. (2024). **Perfusion and Electrophysiological Changes in MELAS**. Ann Neurol, doi:[10.1002/ana.27176](https://doi.org/10.1002/ana.27176).

Anderson, E., Setty, S., Dahmen, M., Townsley, M. M., Augoustides, J. G. et Fernando, R. J. (2025). **Cardiac and Renal Transplantation in Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-like Symptoms: Anesthetic Challenges and Considerations**. J Cardiothorac Vasc Anesth 39, 301-308, doi:[10.1053/j.jvca.2024.09.138](https://doi.org/10.1053/j.jvca.2024.09.138).

Arriola-Montenegro, J., Mutschler, M., Cogswell, R., Alexy, T., John, R., Voeller, R., Humphreville, V., Aggarwal, A. et Maharaj, V. (2024). **Successful Simultaneous Heart-Kidney Transplant in a Patient With MT-TL1 MELAS Cardiomyopathy**. JACC Case Rep 29, 102523, doi:[10.1016/j.jaccas.2024.102523](https://doi.org/10.1016/j.jaccas.2024.102523).

Battaglia Parodi, M., Antropoli, A., Bianco, L., Arrigo, A., Del Fabbro, S., Carrera, P., Spiga, I., Catania, A., Lamperti, C., Bandello, F. et Mansour, A. (2024). **Multifocal vitelliform lesions associated with mitochondrial retinopathy**. Am J Ophthalmol Case Rep 36, 102181, doi:[10.1016/j.ajoc.2024.102181](https://doi.org/10.1016/j.ajoc.2024.102181).

Benbrahim, F. Z., El Haddad, S., Allali, N. et Chat, L. (2024). **Moyamoya syndrome secondary to MELAS syndrome in a child: A case report and literature revue**. Radiol Case Rep 19, 6347-6353, doi:[10.1016/j.radcr.2024.08.159](https://doi.org/10.1016/j.radcr.2024.08.159).

Chujo, T. et Tomizawa, K. (2024). **Mitochondrial tRNA modifications: functions, diseases caused by their loss, and treatment strategies**. RNA rna.080257.124, doi:[10.1261/rna.080257.124](https://doi.org/10.1261/rna.080257.124).

Chung, C.-Y., Singh, K., Sheshadri, P., Valdebenito, G. E., Chacko, A. R., Costa Besada, M. A., Liang, X. F., Kabir, L., Pitceathly, R. D. S., Szabadkai, G. et Duchon, M. R. (2024). **Inhibition of the PI3K-AKT-MTORC1 axis reduces the burden of the m.3243A>G mtDNA mutation by promoting mitophagy and improving mitochondrial function**. Autophagy 1-16, doi:[10.1080/15548627.2024.2437908](https://doi.org/10.1080/15548627.2024.2437908).

Feng, X., Li, Y., Zhao, Q. et Xu, S. (2024). **Neuronal Intranuclear Inclusion Disease Presenting with Acute-Onset Dementia and Cortical Edema: A Case Report**. Front Neurol 15, 1464991, doi:[10.3389/fneur.2024.1464991](https://doi.org/10.3389/fneur.2024.1464991).

L'essentielle Maladies Mitochondriales
MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Finsterer, J. (2024a). **Phenotypic Heterogeneity of the Mitochondrial DNA Variant m.13513 G > A**. J Pediatr Genet 13, 253-257, doi:[10.1055/s-0043-1768474](https://doi.org/10.1055/s-0043-1768474).

Finsterer, J. (2024b). « **Strokes** » in **MELAS** are Actually Stroke-like Episodes Due To **Metabolic Pathophysiology**. Neurol India 72, 1287, doi:[10.4103/neurol-india.Neurol-India-D-24-00572](https://doi.org/10.4103/neurol-india.Neurol-India-D-24-00572).

Gao, R., Gu, L., Zuo, W. et Wang, P. (2024). **Long-term prognostic factors and outcomes in mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes: a clinical and biochemical marker analysis**. Front Neurol 15, 1491283, doi:[10.3389/fneur.2024.1491283](https://doi.org/10.3389/fneur.2024.1491283).

Kamps, R. et Robinson, E. L. (2024). **LNC-ing Genetics in Mitochondrial Disease**. Noncoding RNA 10, 57, doi:[10.3390/ncrna10060057](https://doi.org/10.3390/ncrna10060057).

Kawano, Y., Taniguchi, A., Narita, Y., Kagawa, K., Harada, T. et Shindo, A. (2024). **Effective Management of Chronic Intestinal Pseudo-Obstruction in MELAS Using Acotiamide: A Case Report**. Case Rep Neurol 16, 288-293, doi:[10.1159/000541012](https://doi.org/10.1159/000541012).

Lessard, L. E. R., Girard, E., Streichenberger, N., Petiot, P., Acquaviva, C., Pagan, C., Mulligan, P., Bouhour, F., Schaeffer, L. et Jacquier, A. (2024). **Mitochondrial disorders are associated with morphological neuromuscular junction defects**. Neuromuscul Disord 45, 105235, doi:[10.1016/j.nmd.2024.105235](https://doi.org/10.1016/j.nmd.2024.105235).

Liu, Q., Wang, Z., Shi, J., Wang, W., Wen, C., Zhu, Y., Chen, X., Xing, X. et Su, Y. (2024). **MELAS Presenting as Bilateral Symmetric Occipital and Temporal Cortices Lesions: A Case Report and Literature Review**. Neurologist, doi:[10.1097/NRL.0000000000000588](https://doi.org/10.1097/NRL.0000000000000588).

Liu, Z., Xie, Y., Lou, X., Zeng, X., Zhang, L., Yu, M., Wang, J., Li, J., Shen, D., Li, H., Zhao, S., Zhou, Y., Fang, H., Lyu, J., Yuan, Y., Wang, Z., Jin, L. et Fang, F. (2024). **A novel m.5906G > a variant in MT-CO1 causes MELAS/Leigh overlap syndrome**. Mol Genet Genomics 299, 102, doi:[10.1007/s00438-024-02181-y](https://doi.org/10.1007/s00438-024-02181-y).

Maharjan, S., Gamper, H., Yamaki, Y., Christian, T., Henley, R. Y., Li, N.-S., Suzuki, Takeo, Suzuki, Tsutomu, Piccirilli, J. A., Wanunu, M., Seifert, E., Wallace, D. C. et Hou, Y.-M. (2024). **Post-transcriptional methylation of mitochondrial-tRNA differentially contributes to mitochondrial pathology**. Nat Commun 15, 9008, doi:[10.1038/s41467-024-53318-x](https://doi.org/10.1038/s41467-024-53318-x).

Mehri, S. et Finsterer, J. (2024). **To prevent sudden death in m.3243A>G carriers, comprehensive neurologic, cardiac, and pulmological examinations are required**. Cardiol Young 1-2, doi:[10.1017/S1047951124036187](https://doi.org/10.1017/S1047951124036187).

L'essentielle Maladies Mitochondriales
MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Orsucci, D., Caldarazzo Ienco, E., Lopriore, P. et Mancuso, M. (2025). **Disease registries and rare disorders: The virtuous example of mitochondrial medicine**. Exp Neurol 384, 115073, doi:[10.1016/j.expneurol.2024.115073](https://doi.org/10.1016/j.expneurol.2024.115073).

Shi, Y., Dong, G., Pan, H., Tai, H., Zhou, Y., Wang, A., Niu, S., Chen, B., Wang, X. et Zhang, Z. (2024). **Stroke-like episodes in patients with adult-onset neuronal intranuclear inclusion disease and patients with late-onset MELAS: A comparative study**. Ann Clin Transl Neurol 11, 3125-3136, doi:[10.1002/acn3.52219](https://doi.org/10.1002/acn3.52219).

Smeitink, J., van Es, J., Bosman, B., Janssen, M. C. H., Klopstock, T., Gorman, G., Vissing, J., Ruiterkamp, G., Edgar, C. J., Abbink, E. J., van Maanen, R., Pogoryelova, O., Stendel, C., Bischoff, A., Karin, I., Munshi, M., Kümmel, A., Burgert, L., Verhaak, C. et Renkema, H. (2024). **Phase 2b program with sonlicromanol in patients with mitochondrial disease due to m.3243A>G mutation**. Brain awae277, doi:[10.1093/brain/awae277](https://doi.org/10.1093/brain/awae277).

Sunada, Y. (2024). **[Taurine for Mitochondrial Diseases]**. Brain Nerve 76, 1127-1135, doi:[10.11477/mf.1416202748](https://doi.org/10.11477/mf.1416202748).

Weerasinghe, N., Weerasinghe, M., Dissanayake, K., Hordagoda, H. et Peiris, J. (2024). **Unveiling the Mitochondrial Mystery: A Case of Mitochondrial Encephalopathy With Lactic Acidosis and Stroke-Like Episodes**. Cureus 16, e72397, doi:[10.7759/cureus.72397](https://doi.org/10.7759/cureus.72397).

Xu, S., Jiang, J., Chang, L., Zhang, B., Zhu, X. et Niu, F. (2024). **Multisystem clinicopathologic and genetic analysis of MELAS**. Orphanet J Rare Dis 19, 487, doi:[10.1186/s13023-024-03511-4](https://doi.org/10.1186/s13023-024-03511-4).

MERFF

Kamps, R. et Robinson, E. L. (2024). **LNC-ing Genetics in Mitochondrial Disease**. Noncoding RNA 10, 57, doi:[10.3390/ncrna10060057](https://doi.org/10.3390/ncrna10060057).

Orsucci, D., Caldarazzo Ienco, E., Lopriore, P. et Mancuso, M. (2025). **Disease registries and rare disorders: The virtuous example of mitochondrial medicine**. Exp Neurol 384, 115073, doi:[10.1016/j.expneurol.2024.115073](https://doi.org/10.1016/j.expneurol.2024.115073).

Wang, M. Z., Jiang, H. F., Song, T. Y., Xu, C. L., Li, H., Song, M. H. et Fang, F. (2024). **[Clinical characteristics of children with MT-TK gene m.8344A>G variation]**. Zhonghua Er Ke Za Zhi 62, 1056-1063, doi:[10.3760/cma.j.cn112140-20240516-00337](https://doi.org/10.3760/cma.j.cn112140-20240516-00337).



Neuropathie optique héréditaire de Leber - LHON

Abuhamaid, A. I., Alsaffar, G. S., Madan, Z. H. et Wahba, Y. (2024). **Childhood Blindness: A Rare Case of Leber Hereditary Optic Neuropathy in a 16-Year-Old Egyptian Patient.** Cureus 16, e71619, doi:[10.7759/cureus.71619](https://doi.org/10.7759/cureus.71619).

Bassi, S. T., Newman, N. J., Chen, J. J., Tisavipat, N. Y., Mollan, S. P., Moss, H. E. et Milea, D. (2024). **Recent advances in neuro-ophthalmology.** Indian J Ophthalmol 72, 1544-1559, doi:[10.4103/IJO.IJO_594_24](https://doi.org/10.4103/IJO.IJO_594_24).

Bayona-Bafaluy, P., Sanz-Pons, J., Esteban, O., Bueno-Borghi, L. et Ruiz-Pesini, E. (2024). **Risk Factors Associated With Leber Hereditary Optic Neuropathy due to Rare Mutations in Mitochondrial DNA-Encoded Respiratory Complex I Subunits.** Clin Genet, doi:[10.1111/cge.14683](https://doi.org/10.1111/cge.14683).

Bureau, J., Manero, F., Baris, O., Bodin, A., Verny, C., Chevrollier, A., Lenaers, G. et Codron, P. (2024). **Opa1 and MT-Nd6 mutations induce early mitochondrial changes in the retina and prelaminar optic nerve of hereditary optic neuropathy mouse models.** Brain Commun 6, fcae404, doi:[10.1093/braincomms/fcae404](https://doi.org/10.1093/braincomms/fcae404).

Chen, B. S. et Newman, N. J. (2025). **Clinical trials in Leber hereditary optic neuropathy: outcomes and opportunities.** Curr Opin Neurol 38, 79-86, doi:[10.1097/WCO.0000000000001343](https://doi.org/10.1097/WCO.0000000000001343).

Chen, X. et Lin, P. (2024). **Leber hereditary optic neuropathy with disc haemorrhage.** Eye (Lond), doi:[10.1038/s41433-024-03453-y](https://doi.org/10.1038/s41433-024-03453-y).

Chien, Y., Yang, Y.-P., Lin, T.-C., Chiou, G.-Y., Yarmishyn, A. A., Wang, C.-H., Ching, L.-J., Lin, Y.-Y., Chen, S.-J., Hwang, D.-K. et Hsu, C.-C. (2024). **Reprogramming patient-iPSC specific retinal organoids for deciphering epigenetic modifications of RNA methylation.** J Chin Med Assoc, doi:[10.1097/JCMA.0000000000001198](https://doi.org/10.1097/JCMA.0000000000001198).

Chkioua, L., Amri, Y., Sahli, C., Nasri, T., Miladi, M. O., Massoud, T., Laradi, S., Ghorbel, M. et Ben Abdennebi, H. (2024). **Respiratory Chain Complex I Deficiency in Leber Hereditary Optic Neuropathy: Insights from Ophthalmologic and Molecular Investigations in Tunisia.** BMC Genomics 25, 1133, doi:[10.1186/s12864-024-11060-0](https://doi.org/10.1186/s12864-024-11060-0).

Finsterer, J. (2024a). **DNAJC30 variants can also manifest phenotypically as Leigh/LHON overlap syndrome.** Neurol Neurochir Pol, doi:[10.5603/pjnns.103221](https://doi.org/10.5603/pjnns.103221).

Finsterer, J. (2024b). **Genetic family studies and prospective evaluation for multisystem involvement are needed in LHON patients.** Rom J Ophthalmol 68, 338-339, doi:[10.22336/rjo.2024.62](https://doi.org/10.22336/rjo.2024.62).

L'essentielle Maladies Mitochondriales
MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Finsterer, J. (2024c). **Phenotypic Heterogeneity of the Mitochondrial DNA Variant m.13513 G > A**. J Pediatr Genet 13, 253-257, doi:[10.1055/s-0043-1768474](https://doi.org/10.1055/s-0043-1768474).

Francomme, R., Lenoble, Q., Smirnov, V. et Boucart, M. (2024). **Visual Functions in Patients With Leber Hereditary Optic Neuropathy (LHON)**. J Neuroophthalmol, doi:[10.1097/WNO.0000000000002237](https://doi.org/10.1097/WNO.0000000000002237).

Hassan, A. K., Mohsen, M. et Abu Serhan, H. (2024). **Efficacy of Intravitreal rAAV2-ND4 Injection in Treated Versus Fellow Eyes with Leber's Hereditary Optic Neuropathy: A Meta-Analysis**. Neuroophthalmology 48, 391-400, doi:[10.1080/01658107.2024.2360413](https://doi.org/10.1080/01658107.2024.2360413).

Hedström, J., Nilsson, M., Engvall, M., Williams, P. A. et Venkataraman, A. P. (2024). **Ganglion Cell Complex Thickness and Visual Function in Chronic Leber Hereditary Optic Neuropathy**. Invest Ophthalmol Vis Sci 65, 4, doi:[10.1167/iovs.65.12.4](https://doi.org/10.1167/iovs.65.12.4).

Kamps, R. et Robinson, E. L. (2024). **LNC-ing Genetics in Mitochondrial Disease**. Noncoding RNA 10, 57, doi:[10.3390/ncrna10060057](https://doi.org/10.3390/ncrna10060057).

La Morgia, C., Cascavilla, M. L., De Negri, A. M., Romano, M., Canalini, F., Rossi, S., Centonze, D. et Filippi, M. (2024). **Recognizing Leber's Hereditary Optic Neuropathy to avoid delayed diagnosis and misdiagnosis**. Front Neurol 15, 1466275, doi:[10.3389/fneur.2024.1466275](https://doi.org/10.3389/fneur.2024.1466275).

Murakhovskaya, Y. K., Sheremet, N. L., Eliseeva, D. D. et Bryukhov, V. V. (2024). **[Brain magnetic resonance imaging features in Leber's hereditary optic neuropathy]**. Vestn Oftalmol 140, 146-153, doi:[10.17116/oftalma2024140051146](https://doi.org/10.17116/oftalma2024140051146).

Newman, N. J., Biousse, V., Yu-Wai-Man, P., Carelli, V., Vignal-Clermont, C., Montestruc, F., Taiel, M. et Sahel, J.-A. (2025). **Meta-analysis of treatment outcomes for patients with m.11778G>A MT-ND4 Leber hereditary optic neuropathy**. Surv Ophthalmol 70, 283-295, doi:[10.1016/j.survophthal.2024.10.002](https://doi.org/10.1016/j.survophthal.2024.10.002).

Ozir, M. A., Nordin, M. H., Hashim, S. E., Adzahar, S., Ahmad, M. A., Ng, K. S. et Wan Hitam, W.-H. (2024). **Leber Hereditary Optic Neuropathy With Significant Visual Recovery: An MT-ND6 Mutation in a Malay Patient**. Cureus 16, e71210, doi:[10.7759/cureus.71210](https://doi.org/10.7759/cureus.71210).

Scholl, H. P. N. et György, B. (2024). **Single-Eye Gene Therapy for Leber Hereditary Optic Neuropathy**. JAMA Ophthalmol, doi:[10.1001/jamaophthalmol.2024.5618](https://doi.org/10.1001/jamaophthalmol.2024.5618).

Şenol, H. B., Soydemir, D., Polat, A. İ., Aydın, A., Hız, A. S. et Yiş, U. (2025). **An Unusual Presentation of Leber Hereditary Optic Neuropathy-Plus Case Caused by a Novel DNAJC30 Variant**. Am J Med Genet A 197, e63902, doi:[10.1002/ajmg.a.63902](https://doi.org/10.1002/ajmg.a.63902).

L'essentielle Maladies Mitochondriales

MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Warwick, A. M., Bomze, H. M., Wang, L., Hao, Y., Stinnett, S. S. et Gospe, S. M. (2024). **Hypoxia-mediated rescue of retinal ganglion cells deficient in mitochondrial complex I is independent of the hypoxia-inducible factor pathway.** Sci Rep 14, 24114, doi:[10.1038/s41598-024-75916-x](https://doi.org/10.1038/s41598-024-75916-x).

Yu-Wai-Man, P., Newman, N. J., Biousse, V., Carelli, V., Moster, M. L., Vignal-Clermont, C., Klopstock, T., Sadun, A. A., Sergott, R. C., Hage, R., Degli Esposti, S., La Morgia, C., Priglinger, C., Karanja, R., Taiel, M., Sahel, J.-A., et LHON Study Group (2024). **Five-Year Outcomes of Lenadogene Nolparvovec Gene Therapy in Leber Hereditary Optic Neuropathy.** JAMA Ophthalmol, doi:[10.1001/jamaophthalmol.2024.5375](https://doi.org/10.1001/jamaophthalmol.2024.5375).

Zheng, Y., Wang, P., Li, S., Long, Y., Jiang, Y., Guo, D., Jia, X., Liu, M., Zeng, Y., Xiao, X., Hejtmancik, J. F., Zhang, Q. et Sun, W. (2024). **Clinical and genetic landscape of optic atrophy in 826 families: insights from 50 nuclear genes.** Brain awae324, doi:[10.1093/brain/awae324](https://doi.org/10.1093/brain/awae324).

Ophtalmoplégie externe progressive – PEO

Ansari, S. et Koenig, M. K. (2024). **Expanded-Access Use of Elamipretide Improves Quality of Life in Patients With Rare Mitochondrial Disorders Characterized by Ophthalmic Symptoms: A Case Series.** Clin Case Rep 12, e9591, doi:[10.1002/ccr3.9591](https://doi.org/10.1002/ccr3.9591).

Cicchinelli, M., Primiano, G., Servidei, S., Ardito, M., Percio, A., Urbani, A. et Iavarone, F. (2024). **Resolving Phenotypic Variability in Mitochondrial Diseases: Preliminary Findings of a Proteomic Approach.** Int J Mol Sci 25, 10731, doi:[10.3390/ijms251910731](https://doi.org/10.3390/ijms251910731).

Finsterer, J. (2024). **Phenotypic Heterogeneity of the Mitochondrial DNA Variant m.13513 G > A.** J Pediatr Genet 13, 253-257, doi:[10.1055/s-0043-1768474](https://doi.org/10.1055/s-0043-1768474).

Karaa, A., Bertini, E., Carelli, V., Cohen, B., Ennes, G. M., Falk, M. J., Goldstein, A., Gorman, G., Haas, R., Hirano, M., Klopstock, T., Koenig, M. K., Kornblum, C., Lamperti, C., Lehman, A., Longo, N., Molnar, M. J., Parikh, S., Phan, H., Pitceathly, R. D. S., Saneto, R., Scaglia, F., Servidei, S., Tarnopolsky, M., Toscano, A., Van Hove, J. L. K., Vissing, J., Vockley, J., Finman, J. S., Abbruscato, A., Brown, D. A., Sullivan, A., Shiffer, J. A., Mancuso, M., et MMPOWER-3 Trial Investigators (2024). **Genotype-specific effects of elamipretide in patients with primary mitochondrial myopathy: a post hoc analysis of the MMPOWER-3 trial.** Orphanet J Rare Dis 19, 431, doi:[10.1186/s13023-024-03421-5](https://doi.org/10.1186/s13023-024-03421-5).

L'essentielle Maladies Mitochondriales
MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Karimi, N., Ghahvehchian, H., Keyhani, A., Manavishad, A., Compton, C. J., Clark, J. D., West, N. L. et Kashkouli, M. B. (2024). **Type and Frequency of Misdiagnosis and Time Lag to Diagnosis in Patients with Chronic Progressive External Ophthalmoplegia.** J Ophthalmic Vis Res 19, 334-339, doi:[10.18502/jovr.v19i3.13998](https://doi.org/10.18502/jovr.v19i3.13998).

Kleerekooper, I., Verschueren, D. V., Trip, S. A., Plant, G. T. et Petzold, A. (2024). **Ellipsoid Zone Reflectivity: Exploring its Potential as a Novel Non-Invasive Biomarker for Assessing Mitochondrial Function.** Neuroophthalmology 48, 417-428, doi:[10.1080/01658107.2024.2341769](https://doi.org/10.1080/01658107.2024.2341769).

Lessard, L. E. R., Girard, E., Streichenberger, N., Petiot, P., Acquaviva, C., Pagan, C., Mulligan, P., Bouhour, F., Schaeffer, L. et Jacquier, A. (2024). **Mitochondrial disorders are associated with morphological neuromuscular junction defects.** Neuromuscul Disord 45, 105235, doi:[10.1016/j.nmd.2024.105235](https://doi.org/10.1016/j.nmd.2024.105235).

Lilley, T., Camera, D. M. et Kwa, F. A. A. (2024). **Repairing muscle with broccoli-derived sulforaphane: A preclinical evaluation for the treatment of mitochondrial myopathies.** Drug Discov Today 30, 104283, doi:[10.1016/j.drudis.2024.104283](https://doi.org/10.1016/j.drudis.2024.104283).

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

Shukla, V., Webb, P., AlMohaimed, B., Lee, J. et Boelman, C. (2024). **Rhythmic high-amplitude delta with superimposed spikes (RHADS): a treatment dilemma.** Oxf Med Case Reports 2024, omae114, doi:[10.1093/omcr/omae114](https://doi.org/10.1093/omcr/omae114).

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

Amergoolov, I. I., Khruleva, Y. I., Pavlova, M. G., Likhodey, N. V., Sulaev, A. M., Surkova, E. V., Sych, Y. P., Kalashnikova, M. F., Arustamyan, A. S., Martirosyan, G. A. et Lew-Gor, S. T. (2024). **Endocrine disorders in Kearns-Sayre syndrome with different severity of symptoms: two case reports and a literature review.** Eur J Transl Myol 34, 12897, doi:[10.4081/ejtm.2024.12897](https://doi.org/10.4081/ejtm.2024.12897).

Greenberg-Kushnir, N., Grossmann, L. D., Barg, A. A., Schiby, G., Mardoukh, C., Varda-Bloom, N., Marcu-Malina, V., Anikster, Y., Gruber, N., Lahav, E., Bolquier, Y., Bar, D., Bielora, B., Toren, A. et Jacoby, E. (2025). **Long-term hematopoietic dysfunction in patients with large-scale mitochondrial DNA deletion syndromes.** Pediatr Blood Cancer 72, e31383, doi:[10.1002/pbc.31383](https://doi.org/10.1002/pbc.31383).

Kamps, R. et Robinson, E. L. (2024). **LNC-ing Genetics in Mitochondrial Disease.** Noncoding RNA 10, 57, doi:[10.3390/ncrna10060057](https://doi.org/10.3390/ncrna10060057).

L'essentielle Maladies Mitochondriales
MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Lessard, L. E. R., Girard, E., Streichenberger, N., Petiot, P., Acquaviva, C., Pagan, C., Mulligan, P., Bouhour, F., Schaeffer, L. et Jacquier, A. (2024). **Mitochondrial disorders are associated with morphological neuromuscular junction defects.** Neuromuscul Disord 45, 105235, doi:[10.1016/j.nmd.2024.105235](https://doi.org/10.1016/j.nmd.2024.105235).

Leung, V., Wong, J. G. et Grigg, J. R. (2024). **Anti-VEGF therapy for proliferative diabetic retinopathy in Kearns-Sayre syndrome.** Doc Ophthalmol, doi:[10.1007/s10633-024-09999-2](https://doi.org/10.1007/s10633-024-09999-2).

Syndrome de Leigh - *Leigh syndrome*

Amergoolov, I. I., Khruleva, Y. I., Pavlova, M. G., Likhodey, N. V., Sulaev, A. M., Surkova, E. V., Sych, Y. P., Kalashnikova, M. F., Arustamyan, A. S., Martirosyan, G. A. et Lew-Gor, S. T. (2024). **Endocrine disorders in Kearns-Sayre syndrome with different severity of symptoms: two case reports and a literature review.** Eur J Transl Myol 34, 12897, doi:[10.4081/ejtm.2024.12897](https://doi.org/10.4081/ejtm.2024.12897).

Greenberg-Kushnir, N., Grossmann, L. D., Barg, A. A., Schiby, G., Mardoukh, C., Varda-Bloom, N., Marcu-Malina, V., Anikster, Y., Gruber, N., Lahav, E., Bolkier, Y., Bar, D., Bielorai, B., Toren, A. et Jacoby, E. (2025). **Long-term hematopoietic dysfunction in patients with large-scale mitochondrial DNA deletion syndromes.** Pediatr Blood Cancer 72, e31383, doi:[10.1002/pbc.31383](https://doi.org/10.1002/pbc.31383).

Kamps, R. et Robinson, E. L. (2024). **LNC-ing Genetics in Mitochondrial Disease.** Noncoding RNA 10, 57, doi:[10.3390/ncrna10060057](https://doi.org/10.3390/ncrna10060057).

Lessard, L. E. R., Girard, E., Streichenberger, N., Petiot, P., Acquaviva, C., Pagan, C., Mulligan, P., Bouhour, F., Schaeffer, L. et Jacquier, A. (2024). **Mitochondrial disorders are associated with morphological neuromuscular junction defects.** Neuromuscul Disord 45, 105235, doi:[10.1016/j.nmd.2024.105235](https://doi.org/10.1016/j.nmd.2024.105235).

Leung, V., Wong, J. G. et Grigg, J. R. (2024). **Anti-VEGF therapy for proliferative diabetic retinopathy in Kearns-Sayre syndrome.** Doc Ophthalmol, doi:[10.1007/s10633-024-09999-2](https://doi.org/10.1007/s10633-024-09999-2).



L'essentielle Maladies Mitochondriales

MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

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

Adelizzi, A., Giri, A., Di Donfrancesco, A., Boito, S., Prigione, A., Bottani, E., Bollati, V., Tiranti, V., Persico, N. et Brunetti, D. (2024). **Correction to: Fetal and obstetrics manifestations of mitochondrial diseases.** J Transl Med 22, 1110, doi:[10.1186/s12967-024-05862-9](https://doi.org/10.1186/s12967-024-05862-9).

Ahmed, N. (2024). **Anaesthetic management of an infant with MEGD(H)EL syndrome undergoing cochlear implant.** BMC Anesthesiol 24, 428, doi:[10.1186/s12871-024-02812-2](https://doi.org/10.1186/s12871-024-02812-2).

Akamatsu, S., Mitsuhashi, S., Soga, K., Mizukami, H., Shiraishi, M., Frith, M. C. et Yamano, Y. (2024). **Targeted nanopore sequencing using the Flongle device to identify mitochondrial DNA variants.** Sci Rep 14, 25161, doi:[10.1038/s41598-024-75749-8](https://doi.org/10.1038/s41598-024-75749-8).

Akar, H. T., Akduman, H., Kolkiran, A., Taşadelen, E. et Aycan, N. (2024). **The rare reason for massive lactic aciduria and mitochondrial disorders: combined oxidative phosphorylation deficiency type 23 (COXPD23).** Z Geburtshilfe Neonatol, doi:[10.1055/a-2465-3661](https://doi.org/10.1055/a-2465-3661).

Ambrose, A., Bahl, S., Sharma, S., Zhang, D., Hung, C., Jain-Ghai, S., Chan, A. et Mercimek-Andrews, S. (2024). **Genetic landscape of primary mitochondrial diseases in children and adults using molecular genetics and genomic investigations of mitochondrial and nuclear genome.** Orphanet J Rare Dis 19, 424, doi:[10.1186/s13023-024-03437-x](https://doi.org/10.1186/s13023-024-03437-x).

Amergoolov, I. I., Khruleva, Y. I., Pavlova, M. G., Likhodey, N. V., Sulaev, A. M., Surkova, E. V., Sych, Y. P., Kalashnikova, M. F., Arustamyan, A. S., Martirosyan, G. A. et Lew-Gor, S. T. (2024). **Endocrine disorders in Kearns-Sayre syndrome with different severity of symptoms: two case reports and a literature review.** Eur J Transl Myol 34, 12897, doi:[10.4081/ejtm.2024.12897](https://doi.org/10.4081/ejtm.2024.12897).

Ansari, S. et Koenig, M. K. (2024). **Expanded-Access Use of Elamipretide Improves Quality of Life in Patients With Rare Mitochondrial Disorders Characterized by Ophthalmic Symptoms: A Case Series.** Clin Case Rep 12, e9591, doi:[10.1002/ccr3.9591](https://doi.org/10.1002/ccr3.9591).

Baldo, M. S., Azevedo, L., Coelho, M. P., Martins, E. et Vilarinho, L. (2024). **A Comprehensive Approach to the Diagnosis of Leigh Syndrome Spectrum.** Diagnostics (Basel) 14, 2133, doi:[10.3390/diagnostics14192133](https://doi.org/10.3390/diagnostics14192133).



L'essentielle Maladies Mitochondriales

MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Ball, M., Bouffler, S. E., Barnett, C. B., Freckmann, M.-L., Hunter, M. F., Kamien, B., Kassahn, K. S., Lunke, S., Patel, C. V., Pinner, J., Roscioli, T., Sandaradura, S. A., Scott, H. S., Tan, T. Y., Wallis, M., Compton, A. G., Thorburn, D. R., Stark, Z. et Christodoulou, J. (2025). **Critically unwell infants and children with mitochondrial disorders diagnosed by ultrarapid genomic sequencing.** Genet Med 27, 101293, doi:[10.1016/j.gim.2024.101293](https://doi.org/10.1016/j.gim.2024.101293).

Bangel, K. A., Lim, A. Z., Blain, A., Ng, Y. S., Winder, A., Bulmer, J., Gorman, G., Baker, M. et McFarland, R. (2024). **TRANscraniel direct current stimulation for FOcal Refractory epilepsy in mitochondrial disease (TRANSFORM): delayed-start, randomised, double-blinded, placebo-controlled study.** BMC Neurol 24, 407, doi:[10.1186/s12883-024-03907-6](https://doi.org/10.1186/s12883-024-03907-6).

Bayrak, H., Sezer, A. et Kılıç, M. (2024). **RMND1 Mutation Case Report and Literature Review.** Mol Syndromol 15, 487-494, doi:[10.1159/000538930](https://doi.org/10.1159/000538930).

Begelman, D. V., Dixit, B., Truong, C., King, C. D., Watson, M. A., Schilling, B., Brand, M. D. et Boominathan, A. (2024). **Exogenous expression of ATP8, a mitochondrial encoded protein, from the nucleus in vivo.** Mol Ther Methods Clin Dev 32, 101372, doi:[10.1016/j.omtm.2024.101372](https://doi.org/10.1016/j.omtm.2024.101372).

Benbrahim, F. Z., El Haddad, S., Allali, N. et Chat, L. (2024). **Moyamoya syndrome secondary to MELAS syndrome in a child: A case report and literature revue.** Radiol Case Rep 19, 6347-6353, doi:[10.1016/j.radcr.2024.08.159](https://doi.org/10.1016/j.radcr.2024.08.159).

Bercu, L., Amati-Bonneau, P., Desquirit-Dumas, V., Procaccio, V. et Maillot, F. (2025). **Uncommon case of mitochondrial disease: Mild amyotrophy of the legs and symmetrical lipomatosis of the arms.** J Inherit Metab Dis 48, e12820, doi:[10.1002/jimd.12820](https://doi.org/10.1002/jimd.12820).

Brügel, M., Kiesel, A.-S., Haack, T. B. et Peralta, S. (2024). **Mutations in mitochondrial ATAD3 gene and disease, lessons from in vivo models.** Front Neurosci 18, 1496142, doi:[10.3389/fnins.2024.1496142](https://doi.org/10.3389/fnins.2024.1496142).

Bulduk, B. K., Tortajada, J., Torres-Egurrola, L., Valiente-Pallejà, A., Martínez-Leal, R., Vilella, E., Torrell, H., Muntané, G. et Martorell, L. (2025). **High frequency of mitochondrial DNA rearrangements in the peripheral blood of adults with intellectual disability.** J Intellect Disabil Res 69, 137-152, doi:[10.1111/jir.13197](https://doi.org/10.1111/jir.13197).

Büyükyılmaz, G., İnözü, M. et Çavdarlı, B. (2024). **A case with short stature and proteinuria: atypical presentation of a family with m.3243A>G mutation.** Turk J Pediatr 66, 490-498, doi:[10.24953/turkjpediatr.2024.4702](https://doi.org/10.24953/turkjpediatr.2024.4702).

L'essentielle Maladies Mitochondriales

MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Cafferati Beltrame, L., Sgobba, M. N., Laera, L., Scaglione, V., Todisco, S., Barile, S., Francavilla, A. L., De Luca, D. I., Montaruli, M., Porcelli, V., Guerra, L., De Grassi, A., Volpicella, M. et Pierri, C. L. (2024). **Combined in silico/in vitro approaches for identifying modulators of the activity of the p.Tyr110Cys Carnitine O-Acetyltransferase (CRAT) variant associated to an early onset case of Leigh syndrome.** Acta Pharmacol Sin, doi:[10.1038/s41401-024-01435-0](https://doi.org/10.1038/s41401-024-01435-0).

Campbell, T., Slone, J., Metzger, H., Liu, W., Sacharow, S., Yang, A., Moosajee, M., La Morgia, C., Carelli, V., Palombo, F., Lines, M. A., Innes, A. M., Levy, R. J., Neilson, D., Longo, N. et Huang, T. (2024). **Clinical study of ferredoxin-reductase-related mitochondriopathy: Genotype-phenotype correlation and proposal of ancestry-based carrier screening in the Mexican population.** Genet Med Open 2, 100841, doi:[10.1016/j.gimo.2023.100841](https://doi.org/10.1016/j.gimo.2023.100841).

Chanoine, J.-P., Thompson, D. M. et Lehman, A. (2024). **Diabetes Mellitus Associated with Maternally Inherited Diabetes and Deafness (MIDD): From Pathogenic Variant to Phenotype.** Diabetes db240515, doi:[10.2337/db24-0515](https://doi.org/10.2337/db24-0515).

Chatel, B., Varlet, I., Ogier, A. C., Pecchi, E., Bernard, M., Gondin, J., Westerblad, H., Bendahan, D. et Gineste, C. (2025). **Cyclosporine A Delays the Terminal Disease Stage in the Tfam KO Mitochondrial Myopathy Mouse Model Without Improving Mitochondrial Energy Production.** Muscle Nerve 71, 265-274, doi:[10.1002/mus.28315](https://doi.org/10.1002/mus.28315).

Chen, B., Zhang, C., Yuan, Y., Wang, Z., Cui, T., Dong, G., Pan, H., Zhang, Z. et Li, W. (2024). **Novel PNPLA8 variants associated with primary ovarian insufficiency, tremors, cerebellar ataxia and limb weakness: a case report and literature review.** J Neurol 272, 78, doi:[10.1007/s00415-024-12838-8](https://doi.org/10.1007/s00415-024-12838-8).

Chujo, T. et Tomizawa, K. (2024). **Mitochondrial tRNA modifications: functions, diseases caused by their loss, and treatment strategies.** RNA rna.080257.124, doi:[10.1261/rna.080257.124](https://doi.org/10.1261/rna.080257.124).

Cicchinelli, M., Primiano, G., Servidei, S., Ardito, M., Percio, A., Urbani, A. et Iavarone, F. (2024). **Resolving Phenotypic Variability in Mitochondrial Diseases: Preliminary Findings of a Proteomic Approach.** Int J Mol Sci 25, 10731, doi:[10.3390/ijms251910731](https://doi.org/10.3390/ijms251910731).

Del Prado, L., Jaraíz-Rodríguez, M., Agro, M., Zamora-Dorta, M., Azpiazu, N., Calleja, M., Lopez-Manzaneda, M., de Juan-Sanz, J., Fernández-Rodrigo, A., Esteban, J. A., Girona, M., Quintana, A. et Balsa, E. (2024). **Compensatory activity of the PC-ME1 metabolic axis underlies differential sensitivity to mitochondrial complex I inhibition.** Nat Commun 15, 8682, doi:[10.1038/s41467-024-52968-1](https://doi.org/10.1038/s41467-024-52968-1).



L'essentielle Maladies Mitochondriales

MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

de Vreugd, A., Zimmermann, F. A., Steinbrücker, K., de Vries, M. C., de Boer, L., Janssen, M. C., Huemer, M. et Wortmann, S. B. (2025). **Vaccine safety in children with genetically confirmed mitochondrial disease.** Immunol Lett 271, 106946, doi:[10.1016/j.imlet.2024.106946](https://doi.org/10.1016/j.imlet.2024.106946).

Di Stadio, A., Frohman, E. M., Messineo, D., Brenner, M. J. et Bernitsas, E. (2024). **The bidirectional brain-cochlea axis: a scaffold for neurologic disease-associated hearing loss.** Brain Commun 6, fcae403, doi:[10.1093/braincomms/fcae403](https://doi.org/10.1093/braincomms/fcae403).

Donis, R., Patel, K. A., Wakeling, M. N., Johnson, M. B., Amoli, M. M., Yildiz, M., Akçay, T., Aspi, I., Yong, J., Yaghootkar, H., Weedon, M. N., Hattersley, A. T., Flanagan, S. E. et De Franco, E. (2024). **A homozygous TARS2 variant is a novel cause of syndromic neonatal diabetes.** Diabet Med e15471, doi:[10.1111/dme.15471](https://doi.org/10.1111/dme.15471).

El Fissi, N., Rosenberger, F. A., Chang, K., Wilhalm, A., Barton-Owen, T., Hansen, F. M., Golder, Z., Alsina, D., Wedell, A., Mann, M., Chinnery, P. F., Freyer, C. et Wredenberg, A. (2024). **Preventing excessive autophagy protects from the pathology of mtDNA mutations in Drosophila melanogaster.** Nat Commun 15, 10719, doi:[10.1038/s41467-024-55559-2](https://doi.org/10.1038/s41467-024-55559-2).

El Guessabi, S., Abouqal, R., Ibrahim, A., Zouiri, G., Sfifou, F., Finsterer, J. et Kriouile, Y. (2024). **Diagnostic accuracy of the lactate stress test for detecting mitochondrial disorders: Systematic review and meta-analysis.** Heliyon 10, e39648, doi:[10.1016/j.heliyon.2024.e39648](https://doi.org/10.1016/j.heliyon.2024.e39648).

Fałek, O., Wesół-Kucharska, D., Starostecka, E., Rokicki, D., Fortecka-Piestrzeniewicz, K., Kępczyński, Ł., Piekutowska-Abramczuk, D., Ciara, E. et Maroszyńska, I. (2024). **Family Occurrence of an m.3303C>T Point Mutation in the MT-TL1 Gene, Which Induces Cardiomyopathy Syndrome with/without Skeletal Muscle Myopathy.** Genes (Basel) 15, 1289, doi:[10.3390/genes15101289](https://doi.org/10.3390/genes15101289).

Fernández, A. C., Estrella, J., Oglesbee, D., Larson, A. A. et Van Hove, J. L. K. (2025). **The clinical utility in hospital-wide use of growth differentiation factor 15 as a biomarker for mitochondrial DNA-related disorders.** J Inherit Metab Dis 48, e12821, doi:[10.1002/jimd.12821](https://doi.org/10.1002/jimd.12821).

Ferrera, G., Derderian, K., Izzo, R., Gnutti, B., Legati, A., Zorzi, G., Lamantea, E., Iuso, A. et Ardisson, A. (2024). **WDR45-related encephalopathy mimicking Leigh syndrome associated with complex I deficiency: a case report.** Eur J Hum Genet, doi:[10.1038/s41431-024-01745-1](https://doi.org/10.1038/s41431-024-01745-1).

L'essentielle Maladies Mitochondriales
MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Finsterer, J. (2024a). **Genetic family studies and prospective evaluation for multisystem involvement are needed in LHON patients.** Rom J Ophthalmol 68, 338-339, doi:[10.22336/rjo.2024.62](https://doi.org/10.22336/rjo.2024.62).

Finsterer, J. (2024b). **Incomplete Unilateral Horner's Syndrome Due to Superior Vena Cava Compression Syndrome Caused by Small Cell Lung Carcinoma.** Cureus 16, e68662, doi:[10.7759/cureus.68662](https://doi.org/10.7759/cureus.68662).

Finsterer, J. (2024c). **Leigh Syndrome Caused by Compound Heterozygous Variants c.1162A_C and c.1138G_C in the NDUFV1 Gene: A Case Report.** Cureus 16, e71127, doi:[10.7759/cureus.71127](https://doi.org/10.7759/cureus.71127).

Finsterer, J. (2024d). **Phenotypic Heterogeneity of the Mitochondrial DNA Variant m.13513 G > A.** J Pediatr Genet 13, 253-257, doi:[10.1055/s-0043-1768474](https://doi.org/10.1055/s-0043-1768474).

Finsterer, J. et Mehri, S. (2024). **The Causality Spectrum of Dropped Head Syndrome is Broad and Includes Myopathy, Neurodegenerative Disorders, and Varia.** Noro Psikiyatr Ars 61, 382-383, doi:[10.29399/npa.28520](https://doi.org/10.29399/npa.28520).

Franco, P. C., Patrocinio, M., Costa-Riquetto, A. D., Santomauro, A. C., Gomes, L. G. et Teles, M. G. (2024). **Phenotypic and molecular reanalysis of a cohort of patients with monogenic diabetes reveals a case of partial lipodystrophy due to the A8344G mutation in the mitochondrial DNA.** Arch Endocrinol Metab 68, e230084, doi:[10.20945/2359-4292-2023-0084](https://doi.org/10.20945/2359-4292-2023-0084).

Fu, Y., Land, M., Cui, R., Kavlashvili, T., Kim, M., Lieber, T., Ryu, K. W., DeBitetto, E., Masilionis, I., Saha, R., Takizawa, M., Baker, D., Tigano, M., Reznik, E., Sharma, R., Chaligne, R., Thompson, C. B., Pe'er, D. et Sfeir, A. (2024). **Engineering mtDNA Deletions by Reconstituting End-Joining in Human Mitochondria.** bioRxiv 2024.10.15.618543, doi:[10.1101/2024.10.15.618543](https://doi.org/10.1101/2024.10.15.618543).

Gaini, R., Chamberlin, G., Wang, S.-H. J. et Morena, J. (2024). **Very-late-onset multiple Acyl-coenzyme a dehydrogenase deficiency with elevated GDF-15 and Aldolase: a case report.** Neuromuscul Disord 45, 105213, doi:[10.1016/j.nmd.2024.105213](https://doi.org/10.1016/j.nmd.2024.105213).

Gauthier, T., Puel, S., Rocher, O., Oswald, I. P. et Puel, O. (2024). **A precursor of Aflatoxin B1, Versicolorin A, impairs the mitochondrial function of human intestinal Caco-2 cells.** Environ Int 193, 109107, doi:[10.1016/j.envint.2024.109107](https://doi.org/10.1016/j.envint.2024.109107).

Geal Dor, M., Gross, M. et Adelman, C. (2024). **Follow-Up of Neonatal Hearing Screening in the Risk Factor Group for Hearing Loss: Results from a Tertiary Medical Center.** Children (Basel) 11, 1336, doi:[10.3390/children11111336](https://doi.org/10.3390/children11111336).

L'essentielle Maladies Mitochondriales
MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

George-Sankoh, I., MacMullen, L. E., Chinwalla, A. T., Taylor, D., Ganetzky, R. D., Stanley, K., McCormick, E. M., Zolkipli-Cunningham, Z. et Falk, M. J. (2024). **MMFP-Tableau: enabling precision mitochondrial medicine through integration, visualization, and analytics of clinical and research health system electronic data.** JAMIA Open 7, ooae134, doi:[10.1093/jamiaopen/ooae134](https://doi.org/10.1093/jamiaopen/ooae134).

Gholami, M., Ghelichkhani, Z., Aghakhani, R., Klionsky, D. J., Motaghinejad, O., Motaghinejad, M., Koohi, M. K. et Hassan, J. (2024). **Minocycline Acts as a Neuroprotective Agent Against Tramadol-Induced Neurodegeneration: Behavioral and Molecular Evidence.** Int J Prev Med 15, 47, doi:[10.4103/ijpvm.ijpvm_10_24](https://doi.org/10.4103/ijpvm.ijpvm_10_24).

Glevicka, M., Komlosi, M., Szantova, M., Gvozdjakova, A., Kucharska, J. et Sumbalova, Z. (2024). **Perspective targeted diagnosis and therapy of mitochondrial bioenergetics across different diagnoses.** Bratisl Lek Listy 125, 693-700, doi:[10.4149/BLL_2024_105](https://doi.org/10.4149/BLL_2024_105).

Goolab, S., Terburgh, K., du Plessis, C., Scholefield, J. et Louw, R. (2025). **CRISPR-Cas9 mediated knockout of NDUF54 in human iPSCs: A model for mitochondrial complex I deficiency.** Biochim Biophys Acta Mol Basis Dis 1871, 167569, doi:[10.1016/j.bbadis.2024.167569](https://doi.org/10.1016/j.bbadis.2024.167569).

Gospodaryov, D. V. (2024). **Alternative NADH dehydrogenase: A complex I backup, a drug target, and a tool for mitochondrial gene therapy.** Biochim Biophys Acta Bioenerg 1866, 149529, doi:[10.1016/j.bbabbio.2024.149529](https://doi.org/10.1016/j.bbabbio.2024.149529).

Grifagni, D., Doni, D., Susini, B., Fonseca, B. M., Louro, R. O., Costantini, P. et Ciofi-Baffoni, S. (2024). **Unraveling the molecular determinants of a rare human mitochondrial disorder caused by the P144L mutation of FDX2.** Protein Sci 33, e5197, doi:[10.1002/pro.5197](https://doi.org/10.1002/pro.5197).

Gu, Y.-Y., Zhao, X.-R., Zhang, N., Yang, Y., Yi, Y., Shao, Q.-H., Liu, M.-X. et Zhang, X.-L. (2024). **Mitochondrial dysfunction as a therapeutic strategy for neurodegenerative diseases: Current insights and future directions.** Ageing Res Rev 102, 102577, doi:[10.1016/j.arr.2024.102577](https://doi.org/10.1016/j.arr.2024.102577).

Gupta, N., Aggarwal, B., Mishra, A., Chowdhury, M. R., Gulati, S., Kumar, A. et Kabra, M. (2024). **Clinico-Radiological and Genotypic Spectrum of Nuclear Mitochondriopathies.** Indian J Pediatr, doi:[10.1007/s12098-024-05266-z](https://doi.org/10.1007/s12098-024-05266-z).

Gupta, Y., Heintzmann, R., Costa, C., Jesus, R. et Pinho, E. (2024). **Deep learning-enhanced automated mitochondrial segmentation in FIB-SEM images using an entropy-weighted ensemble approach.** PLoS One 19, e0313000, doi:[10.1371/journal.pone.0313000](https://doi.org/10.1371/journal.pone.0313000).



L'essentielle Maladies Mitochondriales

MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Habib, C., Tal, G., Weiss, K., Magen, D. et Pollack, S. (2024). **PDSS1 mutations-associated steroid-resistant nephrotic syndrome: case report and review of literature.** *Pediatr Nephrol*, doi:[10.1007/s00467-024-06596-y](https://doi.org/10.1007/s00467-024-06596-y).

Hajhashemy, Z., Golpour-Hamedani, S., Eshaghian, N., Sadeghi, O., Khorvash, F. et Askari, G. (2024). **Practical supplements for prevention and management of migraine attacks: a narrative review.** *Front Nutr* 11, 1433390, doi:[10.3389/fnut.2024.1433390](https://doi.org/10.3389/fnut.2024.1433390).

Hassaan, H. M., Pyle, A., Almenabawy, N., Robertson, F. M., Elkhateeb, N., Girgis, M. Y., Mahmoud, I. G. E. D., Amer, F., Samaha, M., Shaheen, Y., ElNaggar, W., Abdoh, D., Mehaney, D. A., Meguid, I. E. A., Taylor, R. W., McFarland, R. et Selim, L. (2025). **Clinical and Genetic Spectrum of Patients With Mitochondrial Disease in a Pediatric Egyptian Cohort: Novel Variants and Phenotypic Expansion.** *Am J Med Genet A* 197, e63881, doi:[10.1002/ajmg.a.63881](https://doi.org/10.1002/ajmg.a.63881).

Hedberg-Oldfors, C., Lindgren, U., Visuttijai, K., Shen, Y., Ilinca, A., Nordström, S., Lindberg, C. et Oldfors, A. (2024). **Lipid storage myopathy associated with sertraline treatment is an acquired mitochondrial disorder with respiratory chain deficiency.** *Acta Neuropathol* 148, 73, doi:[10.1007/s00401-024-02830-x](https://doi.org/10.1007/s00401-024-02830-x).

Henke, M.-T., Prigione, A. et Schuelke, M. (2024). **Disease models of Leigh syndrome: From yeast to organoids.** *J Inherit Metab Dis* 47, 1292-1321, doi:[10.1002/jimd.12804](https://doi.org/10.1002/jimd.12804).

Hernández-Camacho, J. D., Vicente-García, C., Ardila-García, L., Padilla-Campos, A., López-Lluch, G., Santos-Ocaña, C., Zammit, P. S., Carvajal, J. J., Navas, P. et Fernández-Ayala, D. J. M. (2024). **Prenatal and progressive coenzyme Q10 administration to mitigate muscle dysfunction in mitochondrial disease.** *J Cachexia Sarcopenia Muscle* 15, 2402-2416, doi:[10.1002/jcsm.13574](https://doi.org/10.1002/jcsm.13574).

Hu, W., Shi, C., Guo, H. et Zhang, B. (2024). **POLG p.A962T Mutation Leads to Neuronal Mitochondrial Dysfunction That is Restored After Mitochondrial Transplantation.** *Physiol Res* 73, 801-808, doi:[10.33549/physiolres.935313](https://doi.org/10.33549/physiolres.935313).

Huff, A., Oliveira, L. M., Karlen-Amarante, M., Ebiala, F., Ramirez, J. M. et Kalume, F. (2024). **Ndufs4 inactivation in glutamatergic neurons reveals swallow-breathing discoordination in a mouse model of Leigh syndrome.** *Exp Neurol* 385, 115123, doi:[10.1016/j.expneurol.2024.115123](https://doi.org/10.1016/j.expneurol.2024.115123).



L'essentielle Maladies Mitochondriales

MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Imasawa, T., Murayama, K., Hirano, D. et Nozu, K. (2024). **Comprehensive review of mitochondrial nephropathy-a renal phenotype in mitochondrial disease: causative genes, clinical and pathological features, diagnosis, prognosis, and treatment.** Clin Exp Nephrol, doi:[10.1007/s10157-024-02554-y](https://doi.org/10.1007/s10157-024-02554-y).

Jacobs, H. T., Rustin, P., Bénit, P., Davidi, D. et Terzioglu, M. (2024). **Mitochondria: great balls of fire.** FEBS J 291, 5327-5341, doi:[10.1111/febs.17316](https://doi.org/10.1111/febs.17316).

Kam, J. et Momoh, R. (2024). **Rhabdomyolysis in a Patient With a Possible Mitochondrial Pathogenic Variant in the Peri-operative Period: A Case Report.** Cureus 16, e73577, doi:[10.7759/cureus.73577](https://doi.org/10.7759/cureus.73577).

Kamps, R. et Robinson, E. L. (2024). **LNC-ing Genetics in Mitochondrial Disease.** Noncoding RNA 10, 57, doi:[10.3390/ncrna10060057](https://doi.org/10.3390/ncrna10060057).

Karaa, A., Bertini, E., Carelli, V., Cohen, B., Ennes, G. M., Falk, M. J., Goldstein, A., Gorman, G., Haas, R., Hirano, M., Klopstock, T., Koenig, M. K., Kornblum, C., Lamperti, C., Lehman, A., Longo, N., Molnar, M. J., Parikh, S., Phan, H., Pitceathly, R. D. S., Saneto, R., Scaglia, F., Servidei, S., Tarnopolsky, M., Toscano, A., Van Hove, J. L. K., Vissing, J., Vockley, J., Finman, J. S., Abbruscato, A., Brown, D. A., Sullivan, A., Shiffer, J. A., Mancuso, M., et MMPOWER-3 Trial Investigators (2024). **Genotype-specific effects of elamipretide in patients with primary mitochondrial myopathy: a post hoc analysis of the MMPOWER-3 trial.** Orphanet J Rare Dis 19, 431, doi:[10.1186/s13023-024-03421-5](https://doi.org/10.1186/s13023-024-03421-5).

Karuntu, J. S., Almushattat, H., Nguyen, X.-T.-A., Plomp, A. S., Wanders, R. J. A., Hoyng, C. B., van Schooneveld, M. J., Schalijs-Delfos, N. E., Brands, M. M., Leroy, B. P., van Karnebeek, C. D. M., Bergen, A. A., van Genderen, M. M. et Boon, C. J. F. (2024). **Syndromic Retinitis Pigmentosa.** Prog Retin Eye Res 101324, doi:[10.1016/j.preteyeres.2024.101324](https://doi.org/10.1016/j.preteyeres.2024.101324).

Kawano, Y., Taniguchi, A., Narita, Y., Kagawa, K., Harada, T. et Shindo, A. (2024). **Effective Management of Chronic Intestinal Pseudo-Obstruction in MELAS Using Acotiamide: A Case Report.** Case Rep Neurol 16, 288-293, doi:[10.1159/000541012](https://doi.org/10.1159/000541012).

Kayani, S., Daescu, V., Dahshi, H., Messahel, S., Woleban, K., Minassian, B. A., Ling, Q. et Gray, S. J. (2024). **SURF1 Deficiency: Expanding on Disease Phenotype and Assessing Disease Burden by Describing Clinical and Biochemical Phenotype.** Am J Med Genet A e63947, doi:[10.1002/ajmg.a.63947](https://doi.org/10.1002/ajmg.a.63947).



L'essentielle Maladies Mitochondriales

MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Kelly, C., Trumpff, C., Acosta, C., Assuras, S., Baker, J., Basarrate, S., Behnke, A., Bo, K., Bobba-Alves, N., Champagne, F. A., Conklin, Q., Cross, M., De Jager, P., Engelstad, K., Epel, E., Franklin, S. G., Hirano, M., Huang, Q., Junker, A., Juster, R.-P., Kapri, D., Kirschbaum, C., Kurade, M., Lauriola, V., Li, S., Liu, C. C., Liu, G., McEwen, B., McGill, M. A., McIntyre, K., Monzel, A. S., Michelson, J., Prather, A. A., Puterman, E., Rosales, X. Q., Shapiro, P. A., Shire, D., Slavich, G. M., Sloan, R. P., Smith, J. L. M., Spann, M., Spicer, J., Sturm, G., Tepler, S., de Schotten, M. T., Wager, T. D., Picard, M., et MiSBIE Study Group (2024). **A platform to map the mind-mitochondria connection and the hallmarks of psychobiology: the MiSBIE study.** Trends Endocrinol Metab 35, 884-901, doi:[10.1016/j.tem.2024.08.006](https://doi.org/10.1016/j.tem.2024.08.006).

Kim, S., Park, S. G., Kim, J., Hong, S., Cho, S.-M., Lim, S.-Y., Kim, E.-K., Ju, S., Lee, S. B., Kim, S. P., Jeong, T. Y., Oh, Y., Han, S., Kim, H.-R., Lee, T. C., Kim, H.-C., Yoon, W. K., An, T. H., Oh, K.-J., Nam, K.-H., Lee, S., Kim, K., Seong, J. K. et Lee, H. (2024). **Comprehensive phenotypic assessment of nonsense mutations in mitochondrial ND5 in mice.** Exp Mol Med 56, 2395-2408, doi:[10.1038/s12276-024-01333-9](https://doi.org/10.1038/s12276-024-01333-9).

Kingren, M. S., Keeble, A. R., Galvan-Lara, A. M., Ogle, J. M., Ungvári, Z., St Clair, D. K., Butterfield, T. A., Owen, A. M., Fry, C. S., Patel, S. P. et Saito, H. (2024). **Post-sepsis chronic muscle weakness can be prevented by pharmacological protection of mitochondria.** Mol Med 30, 221, doi:[10.1186/s10020-024-00982-w](https://doi.org/10.1186/s10020-024-00982-w).

Kleerekooper, I., Verschueren, D. V., Trip, S. A., Plant, G. T. et Petzold, A. (2024). **Ellipsoid Zone Reflectivity: Exploring its Potential as a Novel Non-Invasive Biomarker for Assessing Mitochondrial Function.** Neuroophthalmology 48, 417-428, doi:[10.1080/01658107.2024.2341769](https://doi.org/10.1080/01658107.2024.2341769).

Koohi, N., Holmes, S., Male, A., Bamiou, D.-E., Dudzic, M. M., Ramdharry, G. M., Pizzamiglio, C., Hanna, M. G., Pitceathly, R. D. S. et Kaski, D. (2024). **Beyond the cochlea: exploring the multifaceted nature of hearing loss in primary mitochondrial diseases.** Brain Commun 6, fcae374, doi:[10.1093/braincomms/fcae374](https://doi.org/10.1093/braincomms/fcae374).

Korandová, Z., Pecina, P., Pecinová, A., Koňářková, E., Tesařová, M., Houštěk, J., Hansíková, H., Ptáčková, H., Zeman, J., Honzík, T. et Mráček, T. (2025). **Cryopreserved PBMCs can be used for the analysis of mitochondrial respiration and serve as a diagnostic tool for mitochondrial diseases.** Anal Biochem 698, 115745, doi:[10.1016/j.ab.2024.115745](https://doi.org/10.1016/j.ab.2024.115745).

Korotkevich, E., Conrad, D. N., Gartner, Z. J. et O'Farrell, P. H. (2024). **Selection promotes age-dependent degeneration of the mitochondrial genome.** bioRxiv 2024.09.27.615276, doi:[10.1101/2024.09.27.615276](https://doi.org/10.1101/2024.09.27.615276).

L'essentielle Maladies Mitochondriales

MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Kuusik, B. et Mithal, D. S. (2025). **Updates on neurologic manifestations of mitochondrial disease.** Curr Opin Pediatr 37, 107-111, doi:[10.1097/MOP.0000000000001418](https://doi.org/10.1097/MOP.0000000000001418).

Lancaster, M. S., Hafen, P., Law, A. S., Matias, C., Meyer, T., Fischer, K., Miller, M., Hao, C., Gillespie, P., McKinzie, D., Brault, J. J. et Graham, B. H. (2024). **Sucla2 Knock-Out in Skeletal Muscle Yields Mouse Model of Mitochondrial Myopathy With Muscle Type-Specific Phenotypes.** J Cachexia Sarcopenia Muscle 15, 2729-2742, doi:[10.1002/jcsm.13617](https://doi.org/10.1002/jcsm.13617).

Lessard, L. E. R., Girard, E., Streichenberger, N., Petiot, P., Acquaviva, C., Pagan, C., Mulligan, P., Bouhour, F., Schaeffer, L. et Jacquier, A. (2024). **Mitochondrial disorders are associated with morphological neuromuscular junction defects.** Neuromuscul Disord 45, 105235, doi:[10.1016/j.nmd.2024.105235](https://doi.org/10.1016/j.nmd.2024.105235).

Li, B.-G., Wu, W.-J., Wang, L.-H., Wang, X., Liu, C., Du, Y.-K., Li, B.-C., Hu, J.-T. et Sun, S.-Z. (2024). **Identification of a novel pathogenic gene, NDUFA3, in Leigh Syndrome through whole exome sequencing.** Neurogenetics 26, 13, doi:[10.1007/s10048-024-00782-8](https://doi.org/10.1007/s10048-024-00782-8).

Li, S.-X., He, N., Liao, J.-X., Lu, X.-G., Hu, W.-G., Liu, X.-R., Liao, W.-P., Song, X.-W. et Li, B. (2024). **Association of LONP1 gene with epilepsy and the sub-regional effect.** Sci Rep 14, 25575, doi:[10.1038/s41598-024-77039-9](https://doi.org/10.1038/s41598-024-77039-9).

Lilley, T., Camera, D. M. et Kwa, F. A. A. (2024). **Repairing muscle with broccoli-derived sulforaphane: A preclinical evaluation for the treatment of mitochondrial myopathies.** Drug Discov Today 30, 104283, doi:[10.1016/j.drudis.2024.104283](https://doi.org/10.1016/j.drudis.2024.104283).

Lin, H.-P., Petersen, J. D., Gilsrud, A. J., Madruga, A., D'Silva, T. M., Huang, X., Shammas, M. K., Randolph, N. P., Johnson, K. R., Li, Y., Jones, D. R., Pacold, M. E. et Narendra, D. P. (2024). **DELE1 maintains muscle proteostasis to promote growth and survival in mitochondrial myopathy.** EMBO J 43, 5548-5585, doi:[10.1038/s44318-024-00242-x](https://doi.org/10.1038/s44318-024-00242-x).

Liu, J., Tang, R., Zheng, J. et Luo, K. (2024). **Targeting ferroptosis reveals a new strategy for breast cancer treatment: a bibliometric study.** Discov Oncol 15, 679, doi:[10.1007/s12672-024-01569-x](https://doi.org/10.1007/s12672-024-01569-x).

Liu, Q., Wang, Z., Shi, J., Wang, W., Wen, C., Zhu, Y., Chen, X., Xing, X. et Su, Y. (2024). **MELAS Presenting as Bilateral Symmetric Occipital and Temporal Cortices Lesions: A Case Report and Literature Review.** Neurologist, doi:[10.1097/NRL.0000000000000588](https://doi.org/10.1097/NRL.0000000000000588).



L'essentiel le Maladies Mitochondriales

MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Ma, Y., Li, G., Li, L., Zong, J., Liu, W., Zhang, R. et Liu, S. (2025). **Two novel heterozygous HPDL variants in a Chinese family with a neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities.** Gene 934, 149018, doi:[10.1016/j.gene.2024.149018](https://doi.org/10.1016/j.gene.2024.149018).

Maharjan, S., Gamper, H., Yamaki, Y., Christian, T., Henley, R. Y., Li, N.-S., Suzuki, Takeo, Suzuki, Tsutomu, Piccirilli, J. A., Wanunu, M., Seifert, E., Wallace, D. C. et Hou, Y.-M. (2024). **Post-transcriptional methylation of mitochondrial-tRNA differentially contributes to mitochondrial pathology.** Nat Commun 15, 9008, doi:[10.1038/s41467-024-53318-x](https://doi.org/10.1038/s41467-024-53318-x).

Manickam, A., Fathima, K., Bagri, V. A., Ganesh Prabu, A. V. P. et R, V. P. (2024). **Balancing Anesthesia in a Child With Mitochondrial Disease: A Case Report.** Cureus 16, e70756, doi:[10.7759/cureus.70756](https://doi.org/10.7759/cureus.70756).

Massuyama, B. K., Viagi, A. M., Claudino de Queiroga, R. C., Camurugy da Hora, R. P., Procaci, V. R., Reis Rosa, A. B., Tonholo Silva, T. Y., Povoas Barsottini, O. G. et Pedroso, J. L. (2024). **NDUFAF5 variants cause early onset Leigh syndrome.** Parkinsonism Relat Disord 131, 107227, doi:[10.1016/j.parkreldis.2024.107227](https://doi.org/10.1016/j.parkreldis.2024.107227).

Mehri, S. et Finsterer, J. (2024). **To prevent sudden death in m.3243A>G carriers, comprehensive neurologic, cardiac, and pulmological examinations are required.** Cardiol Young 1-2, doi:[10.1017/S1047951124036187](https://doi.org/10.1017/S1047951124036187).

Mello, D. F., Perez, L., Bergemann, C. M., Morton, K. S., Ryde, I. T. et Meyer, J. N. (2024). **Comprehensive characterization of mitochondrial bioenergetics at different larval stages reveals novel insights about the developmental metabolism of Caenorhabditis elegans.** PLoS One 19, e0306849, doi:[10.1371/journal.pone.0306849](https://doi.org/10.1371/journal.pone.0306849).

Meng, M., Li, X., Zhou, S., Shi, X., Shen, X. et Chang, G. (2024). **β-Carotene Ameliorates LPS-Induced Endoplasmic Reticulum Stress and Mitochondrial Disorder by Targeting ORAI1 in Bovine Mammary Epithelial Cells.** J Agric Food Chem 72, 26733-26745, doi:[10.1021/acs.jafc.4c06875](https://doi.org/10.1021/acs.jafc.4c06875).

Mickelsson, N., Hirvonen, J. et Martikainen, M. H. (2024). **Clinical features and treatment of stroke-like episodes in mitochondrial disease: a cohort-based study.** J Neurol 272, 47, doi:[10.1007/s00415-024-12745-y](https://doi.org/10.1007/s00415-024-12745-y).

Miller, W. L., Pandey, A. V. et Flück, C. E. (2024). **Disordered electron transfer: New forms of defective steroidogenesis and mitochondriopathy.** J Clin Endocrinol Metab dgae815, doi:[10.1210/clinem/dgae815](https://doi.org/10.1210/clinem/dgae815).

L'essentielle Maladies Mitochondriales

MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Murakhovskaya, Y. K., Sheremet, N. L., Eliseeva, D. D. et Bryukhov, V. V. (2024). **[Brain magnetic resonance imaging features in Leber's hereditary optic neuropathy]**. *Vestn Oftalmol* 140, 146-153, doi:[10.17116/oftalma2024140051146](https://doi.org/10.17116/oftalma2024140051146).

Neikirk, K., Harris, C., Le, H., Oliver, A., Shao, B., Liu, K., Beasley, H. K., Jamison, S., Ishimwe, J. A., Kirabo, A. et Hinton, A. (2024). **Air pollutants as modulators of mitochondrial quality control in cardiovascular disease**. *Physiol Rep* 12, e70118, doi:[10.14814/phy2.70118](https://doi.org/10.14814/phy2.70118).

Neugebauer, J., Reinson, K., Bellusci, M., Park, J. H., Hikmat, O., Bertini, E., Schiff, M., MetabERN PM-MD Consortium authors et Rahman, S. (2025). **Current global vitamin and cofactor prescribing practices for primary mitochondrial diseases: Results of a European reference network survey**. *J Inherit Metab Dis* 48, e12805, doi:[10.1002/jimd.12805](https://doi.org/10.1002/jimd.12805).

Newman, N. J., Biousse, V., Yu-Wai-Man, P., Carelli, V., Vignal-Clermont, C., Montestruc, F., Taiel, M. et Sahel, J.-A. (2025). **Meta-analysis of treatment outcomes for patients with m.11778G>A MT-ND4 Leber hereditary optic neuropathy**. *Surv Ophthalmol* 70, 283-295, doi:[10.1016/j.survophthal.2024.10.002](https://doi.org/10.1016/j.survophthal.2024.10.002).

Orsucci, D., Caldarazzo Ienco, E., Lopriore, P. et Mancuso, M. (2025). **Disease registries and rare disorders: The virtuous example of mitochondrial medicine**. *Exp Neurol* 384, 115073, doi:[10.1016/j.expneurol.2024.115073](https://doi.org/10.1016/j.expneurol.2024.115073).

Ozaki, K., Yatsuka, Y., Oyazato, Y., Nishiyama, A., Nitta, K. R., Kishita, Y., Fushimi, T., Shimura, M., Noma, S., Sugiyama, Y., Tagami, M., Fukunaga, M., Kinoshita, H., Hirata, T., Suda, W., Murakawa, Y., Carninci, P., Ohtake, A., Murayama, K. et Okazaki, Y. (2024). **Biallelic GGGCC repeat expansion leading to NAXE-related mitochondrial encephalopathy**. *NPJ Genom Med* 9, 48, doi:[10.1038/s41525-024-00429-5](https://doi.org/10.1038/s41525-024-00429-5).

Ozir, M. A., Nordin, M. H., Hashim, S. E., Adzahar, S., Ahmad, M. A., Ng, K. S. et Wan Hitam, W.-H. (2024). **Leber Hereditary Optic Neuropathy With Significant Visual Recovery: An MT-ND6 Mutation in a Malay Patient**. *Cureus* 16, e71210, doi:[10.7759/cureus.71210](https://doi.org/10.7759/cureus.71210).

Pan, P., Zhou, N., Sun, Y., Chen, Z., Han, J. et Zhou, W. (2024). **The Spectrum of clinical manifestations in newborns with the COQ4 mutation: case series and literature review**. *Front Pediatr* 12, 1410133, doi:[10.3389/fped.2024.1410133](https://doi.org/10.3389/fped.2024.1410133).

Papatya Çakır, E. D., Ersoy, M., Çakır Biçer, N. et Gedikbaşı, A. (2024). **In Response to: « Involvement of the Endocrine System is Common in Mitochondrial Disorders and Requires Long-term Comprehensive Investigations »**. *J Clin Res Pediatr Endocrinol* 16, 516-518, doi:[10.4274/jcrpe.galenos.2024.2024-10-2](https://doi.org/10.4274/jcrpe.galenos.2024.2024-10-2).



L'essentielle Maladies Mitochondriales

MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Pizzamiglio, C., Stefanetti, R. J., McFarland, R., Thomas, N., Ransley, G., Hugerth, M., Grönberg, A., Serrano, S. S., Elmer, E., Hanna, M. G., Hansson, M. J., Gorman, G. S. et Pitceathly, R. D. S. (2025). **Optimizing rare disorder trials: a phase 1a/1b randomized study of KL1333 in adults with mitochondrial disease.** *Brain* 148, 39-46, doi:[10.1093/brain/awae308](https://doi.org/10.1093/brain/awae308).

Popovic, M., Isermann, L., Geißen, S., Senft, K., Georgomanolis, T., Baldus, S., Frezza, C. et Trifunovic, A. (2024). **Tissue-specific adaptations to cytochrome c oxidase deficiency shape physiological outcomes.** *Biochim Biophys Acta Mol Basis Dis* 1871, 167567, doi:[10.1016/j.bbadis.2024.167567](https://doi.org/10.1016/j.bbadis.2024.167567).

Rimskaya, B., Shebanov, N., Entelis, N. et Mazunin, I. (2024). **Enzymatic tools for mitochondrial genome manipulation.** *Biochimie* S0300-9084(24)00239-6, doi:[10.1016/j.biochi.2024.10.013](https://doi.org/10.1016/j.biochi.2024.10.013).

Ropert, B., Bannwarth, S., Genin, E. C., Vaillant-Beuchot, L., Lacas-Gervais, S., Hounoum, B. M., Bernardin, A., Dinh, N., Mauri-Crouzet, A., D'Elia, M.-A., Augé, G., Lespinasse, F., Di Giorgio, A., Meira, W., Bonnefoy, N., Monassier, L., Schiff, M., Sago, L., Kilinc, D., Brau, F., Redeker, V., Bohl, D., Tribouillard-Tanvier, D., Procaccio, V., Azoulay, S., Ricci, J.-E., Delahodde, A. et Paquis-Flucklinger, V. (2024). **Nifuroxazide rescues the deleterious effects due to CHCHD10-associated MICOS defects in disease models.** *Brain* awae348, doi:[10.1093/brain/awae348](https://doi.org/10.1093/brain/awae348).

Sambuughin, N., Mungunsukh, O., Klein, M. G., Ren, M., Bedocs, P., Kazman, J. B., Cofer, K., Friel, L. P., McNally, B., Kwon, K., Haigney, M. C., Leggit, J. C., Pazgier, M., Deuster, P. A. et O'Connor, F. G. (2024). **Genetics of Exertional Heat Illness: Revealing New Associations and Expanding Heterogeneity.** *Int J Mol Sci* 25, 11269, doi:[10.3390/ijms252011269](https://doi.org/10.3390/ijms252011269).

Santos Gonzalez, F., Hock, D. H., Thorburn, D. R., Mordaunt, D., Williamson, N. A., Ang, C.-S., Stroud, D. A., Christodoulou, J. et Goranitis, I. (2024). **A micro-costing study of mass-spectrometry based quantitative proteomics testing applied to the diagnostic pipeline of mitochondrial and other rare disorders.** *Orphanet J Rare Dis* 19, 443, doi:[10.1186/s13023-024-03462-w](https://doi.org/10.1186/s13023-024-03462-w).

Santos Silva, C., Nunes Vicente, B., Martins, B., Fonseca, A. C., Coelho, P., Roque, R., Cota de Medeiros, F., Oliveira Santos, M. et de Carvalho, M. (2024). **Ptoxis in human immunodeficiency virus-infected patients under long-term antiretroviral treatment.** *Clin Neurol Neurosurg* 249, 108690, doi:[10.1016/j.clineuro.2024.108690](https://doi.org/10.1016/j.clineuro.2024.108690).

Schierbaum, L., Quiroz, V., Tam, A., Zubair, U., Tochen, L., Srouji, R., Yang, K. et Ebrahimi-Fakhari, D. (2024). **Biallelic Variants in COQ4 Cause Childhood-Onset Pure Hereditary Spastic Paraplegia.** *Mov Disord Clin Pract* 11, 1620-1624, doi:[10.1002/mdc3.14226](https://doi.org/10.1002/mdc3.14226).

L'essentielle Maladies Mitochondriales

MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Sen, K., Vera, A. Z., Puronurmi, A., Gropman, A., Wongkittichote, P., Ganetzky, R., Autio, K. et Kastaniotis, A. (2025). **Biallelic Variants in LIPT2 as a Cause of Infantile-Onset Dystonia: Expanding the Clinical and Molecular Spectrum**. *Pediatr Neurol* 162, 32-39, doi:[10.1016/j.pediatrneurol.2024.09.013](https://doi.org/10.1016/j.pediatrneurol.2024.09.013).

Shi, Y., Dong, G., Pan, H., Tai, H., Zhou, Y., Wang, A., Niu, S., Chen, B., Wang, X. et Zhang, Z. (2024). **Stroke-like episodes in patients with adult-onset neuronal intranuclear inclusion disease and patients with late-onset MELAS: A comparative study**. *Ann Clin Transl Neurol* 11, 3125-3136, doi:[10.1002/acn3.52219](https://doi.org/10.1002/acn3.52219).

Slapnik, B., Šket, R., Črepinšek, K., Tesovnik, T., Bizjan, B. J. et Kovač, J. (2024). **The quality and detection limits of mitochondrial heteroplasmy by long read nanopore sequencing**. *Sci Rep* 14, 26778, doi:[10.1038/s41598-024-78270-0](https://doi.org/10.1038/s41598-024-78270-0).

Smeitink, J., van Es, J., Bosman, B., Janssen, M. C. H., Klopstock, T., Gorman, G., Vissing, J., Ruiterkamp, G., Edgar, C. J., Abbink, E. J., van Maanen, R., Pogoryelova, O., Stendel, C., Bischoff, A., Karin, I., Munshi, M., Kümmel, A., Burgert, L., Verhaak, C. et Renkema, H. (2024). **Phase 2b program with sonlicromanol in patients with mitochondrial disease due to m.3243A>G mutation**. *Brain* awae277, doi:[10.1093/brain/awae277](https://doi.org/10.1093/brain/awae277).

Snizek Carney, O., Harris, K. W., Wohlfarter, Y., Lee, K., Butschek, G., Anzmann, A. F., Hamacher-Brady, A., Keller, M. A. et Vernon, H. J. (2024). **Stem cell models of TFAZZIN deficiency reveal novel tissue-specific pathologies in Barth syndrome**. *Hum Mol Genet* ddae152, doi:[10.1093/hmg/ddae152](https://doi.org/10.1093/hmg/ddae152).

Starosta, R. T., Larson, A. A., Meeks, N. J. L., Gracie, S., Friederich, M. W., Gaughan, S. M., Baker, P. R., Knupp, K. G., Michel, C. R., Reisdorph, R., Hock, D. H., Stroud, D. A., Wood, T. et Van Hove, J. L. K. (2024). **An integrated multi-omics approach allowed ultra-rapid diagnosis of a deep intronic pathogenic variant in PDHX and precision treatment in a neonate critically ill with lactic acidosis**. *Mitochondrion* 79, 101973, doi:[10.1016/j.mito.2024.101973](https://doi.org/10.1016/j.mito.2024.101973).

Stefanetti, R. J., Newman, J., Blain, A. P., Chisari, D., Gorman, G. S. et Rance, G. (2024). **Auditory and vestibular function in mitochondrial patients harbouring the m.3243A>G variant**. *Brain Commun* 6, fcae361, doi:[10.1093/braincomms/fcae361](https://doi.org/10.1093/braincomms/fcae361).

Sugasawa, T., Nguyen, K. D. M., Otani, N., Maehara, K., Kamiya, F., Hirokawa, A., Takemasa, T., Watanabe, K., Nishi, T., Sato, K., Shimmura, S., Takahashi, Y. et Kanki, Y. (2024). **Whole Mitochondrial DNA Sequencing Using Fecal Samples from Domestic Dogs**. *Animals (Basel)* 14, 2872, doi:[10.3390/ani14192872](https://doi.org/10.3390/ani14192872).

Sunada, Y. (2024). **[Taurine for Mitochondrial Diseases]**. *Brain Nerve* 76, 1127-1135, doi:[10.11477/mf.1416202748](https://doi.org/10.11477/mf.1416202748).



L'essentielle Maladies Mitochondriales

MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Tavoulari, S., Lacabanne, D., Pereira, G. C., Thangaratnarajah, C., King, M. S., He, J., Chowdhury, S. R., Tilokani, L., Palmer, S. M., Prudent, J., Walker, J. E. et Kunji, E. R. S. (2024). **Distinct roles for the domains of the mitochondrial aspartate/glutamate carrier citrin in organellar localization and substrate transport.** Mol Metab 90, 102047, doi:[10.1016/j.molmet.2024.102047](https://doi.org/10.1016/j.molmet.2024.102047).

Tsalouchidou, P.-E., Juenemann, C., Hahn, W., Zahnert, F., Möller, L., Hakel, L., Kemmling, A., Menzler, K., Simon, O. J., Timmermann, L., Knake, S. et Bernhard, F. (2024). **Adult-onset status epilepticus in patients with COQ8A coenzyme Q10 deficiency: A case series.** Epilepsy Behav Rep 28, 100716, doi:[10.1016/j.ebr.2024.100716](https://doi.org/10.1016/j.ebr.2024.100716).

Tsygankova, P., Chistol, D., Krylova, T., Bychkov, I., Tabakov, V., Markova, T., Dadali, E. et Zakharova, E. (2024). **A New Case of Mitochondrial RNA Helicase SUPV3L1-Associated Neurodegenerative Disease: Ataxia, Spasticity, Optic Atrophy, and Skin Hypopigmentation (ASOASH).** Genes (Basel) 15, 1406, doi:[10.3390/genes15111406](https://doi.org/10.3390/genes15111406).

Valentín Gesé, G. et Hällberg, B. M. (2024). **Structural basis of 3'-tRNA maturation by the human mitochondrial RNase Z complex.** EMBO J 43, 6573-6590, doi:[10.1038/s44318-024-00297-w](https://doi.org/10.1038/s44318-024-00297-w).

Van Haute, L., Páleníková, P., Tang, J. X., Nash, P. A., Simon, M. T., Pyle, A., Oláhová, M., Powell, C. A., Rebelo-Guiomar, P., Stover, A., Champion, M., Deshpande, C., Baple, E. L., Stals, K. L., Ellard, S., Anselem, O., Molac, C., Petrilli, G., Loeuillet, L., Grotto, S., Attie-Bitach, T., Abdenur, J. E., Taylor, R. W. et Minczuk, M. (2025). **Pathogenic PDE12 variants impair mitochondrial RNA processing causing neonatal mitochondrial disease.** EMBO Mol Med 17, 193-210, doi:[10.1038/s44321-024-00172-5](https://doi.org/10.1038/s44321-024-00172-5).

Wahedi, A., Sudhakar, S., Lam, A., Ciancio, J. I. R., Mills, P., Gissen, P., Gardham, A., Kapadia, J., Hassell, J., Heales, S. et Rahman, S. (2024). **Clinical Features, Biochemistry, Imaging, and Treatment Response in a Single-Center Cohort With Coenzyme Q10 Biosynthesis Disorders.** Neurol Genet 10, e200209, doi:[10.1212/NXG.0000000000200209](https://doi.org/10.1212/NXG.0000000000200209).

Wang, H., Miranda, M., Marutani, E., Lichtenegger, P., Wojtkiewicz, G. R., Ichinose, F. et Mootha, V. K. (2024). **Therapeutic hypoxia for mitochondrial disease via enhancement of hemoglobin affinity and inhibition of HIF-2α.** J Clin Invest 134, e185569, doi:[10.1172/JCI185569](https://doi.org/10.1172/JCI185569).

Wang, M., Min, M., Duan, H., Mai, J. et Liu, X. (2024). **The role of macrophage and adipocyte mitochondrial dysfunction in the pathogenesis of obesity.** Front Immunol 15, 1481312, doi:[10.3389/fimmu.2024.1481312](https://doi.org/10.3389/fimmu.2024.1481312).



L'essentielle Maladies Mitochondriales

MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Wang, M. Z., Jiang, H. F., Song, T. Y., Xu, C. L., Li, H., Song, M. H. et Fang, F. (2024). **[Clinical characteristics of children with MT-TK gene m.8344A>G variation]**. Zhonghua Er Ke Za Zhi 62, 1056-1063, doi:[10.3760/cma.j.cn112140-20240516-00337](https://doi.org/10.3760/cma.j.cn112140-20240516-00337).

Wang, Y., Hu, L.-F., Liu, N.-H., Yang, J.-S., Xing, L., Jeong, J.-H., Li, L. et Jiang, H.-L. (2024). **Mitophagy-Enhanced Nanoparticle-Engineered Mitochondria Restore Homeostasis of Mitochondrial Pool for Alleviating Pulmonary Fibrosis**. ACS Nano 18, 32705-32722, doi:[10.1021/acsnano.4c10328](https://doi.org/10.1021/acsnano.4c10328).

Wischhof, L., Mathew, A. J., Bonaguro, L., Beyer, M., Ehninger, D., Nicotera, P. et Bano, D. (2024). **Mitochondrial complex I inhibition enhances astrocyte responsiveness to pro-inflammatory stimuli**. Sci Rep 14, 27182, doi:[10.1038/s41598-024-78434-y](https://doi.org/10.1038/s41598-024-78434-y).

Xie, M., Gu, S., Liu, Y., Yang, H., Wang, Y., Yin, W., Hong, Y., Lu, W., He, C., Li, L., Zhao, L., Zhang, J., Liu, H., Lan, T., Li, S. et Wang, Q. (2025). **2-Hydroxyisobutyric acid targeted binding to MT-ND3 boosts mitochondrial respiratory chain homeostasis in hippocampus to rescue diabetic cognitive impairment**. Redox Biol 79, 103446, doi:[10.1016/j.redox.2024.103446](https://doi.org/10.1016/j.redox.2024.103446).

Xie, Y., Li, K., Yang, L., Zeng, X., Chen, Z., Ma, X., Zhang, L., Zhou, Y., Jin, L., Yang, Y. et Lou, X. (2024). **Expanding the phenotypic and genetic spectrum of GTPBP3 deficiency: findings from nine Chinese pedigrees**. Orphanet J Rare Dis 19, 488, doi:[10.1186/s13023-024-03469-3](https://doi.org/10.1186/s13023-024-03469-3).

Xu, S., Jiang, J., Chang, L., Zhang, B., Zhu, X. et Niu, F. (2024). **Multisystem clinicopathologic and genetic analysis of MELAS**. Orphanet J Rare Dis 19, 487, doi:[10.1186/s13023-024-03511-4](https://doi.org/10.1186/s13023-024-03511-4).

Yamashita, S. (2024). **Late-onset primary muscle diseases mimicking sarcopenia**. Geriatr Gerontol Int 24, 1099-1110, doi:[10.1111/ggi.15000](https://doi.org/10.1111/ggi.15000).

Yang, Y., Davis, I., Altman, R. A. et Liu, A. (2024). **α -Amino- β -carboxymuconate- ϵ -semialdehyde decarboxylase catalyzes enol/keto tautomerization of oxaloacetate**. J Biol Chem 300, 107878, doi:[10.1016/j.jbc.2024.107878](https://doi.org/10.1016/j.jbc.2024.107878).

Yu, C., Tigano, M. et Seifert, E. L. (2025). **PDE12 mediated pruning of the poly-A tail of mitochondrial DNA-encoded tRNAs is essential for survival**. EMBO Mol Med 17, 3-5, doi:[10.1038/s44321-024-00171-6](https://doi.org/10.1038/s44321-024-00171-6).

Zeng, L., Zhu, L., Fu, S., Li, Y. et Hu, K. (2024). **Mitochondrial Dysfunction-Molecular Mechanisms and Potential Treatment approaches of Hepatocellular Carcinoma**. Mol Cell Biochem, doi:[10.1007/s11010-024-05144-4](https://doi.org/10.1007/s11010-024-05144-4).



L'essentielle Maladies Mitochondriales

MMITO-2024-4 du 1^{er} octobre au 31 décembre 2024

Zhang, X., Zhang, B., Zhang, C., Sun, G. et Sun, X. (2024). **Author Correction: Trib1 deficiency causes brown adipose respiratory chain depletion and mitochondrial disorder.** Cell Death Dis 15, 747, doi:[10.1038/s41419-024-07066-x](https://doi.org/10.1038/s41419-024-07066-x).

Zhang, Z., Yu, R., Shi, Q., Wu, Z.-J., Li, Qingchun, Mu, J., Chen, B., Shi, J., Ni, R., Wu, L., Li, Qiaoli, Fu, J., Li, R., Sun, X., Wang, J., He, L., Kuang, Y., Sang, Q. et Wang, L. (2024). **COX15 deficiency causes oocyte ferroptosis.** Proc Natl Acad Sci U S A 121, e2406174121, doi:[10.1073/pnas.2406174121](https://doi.org/10.1073/pnas.2406174121).

Zhou, B.-Q., Li, S.-P., Wang, X., Meng, X.-Y., Deng, J.-R., Xing, J.-L., Wang, J.-G. et Xu, K. (2024). **Dual-localization signals enhance mitochondrial targeted presentation of engineered proteins.** Yi Chuan 46, 937-946, doi:[10.16288/j.ycz.24-171](https://doi.org/10.16288/j.ycz.24-171).

Zhou, X., Wu, J., He, Q., Wang, B., Xu, X., Zhao, X., Gao, M. et Yan, B. (2025). **Short-chain chlorinated paraffins induce liver injury in mice through mitochondrial disorders and disruption of cholesterol-bile acid pathway.** Environ Pollut 364, 125323, doi:[10.1016/j.envpol.2024.125323](https://doi.org/10.1016/j.envpol.2024.125323).

Zhou, Y., Zeng, X., Zhang, L., Yin, X., Ma, X., Li, K., Qiu, P., Lou, X., Jin, L., Wang, Y., Yang, Y. et Shen, T. (2024). **Biallelic variants in the NDUFAF6 cause mitochondrial respiratory complex assembly defects associated with Leigh syndrome in probands.** Mol Genet Metab Rep 41, 101168, doi:[10.1016/j.ymgmr.2024.101168](https://doi.org/10.1016/j.ymgmr.2024.101168).

Zhu, T., Gao, P., Ma, Y., Yang, P., Cao, Z., Gao, J., Du, J., Jiang, H. et Zhang, X. (2024). **Mitochondrial FIS1 As a Novel Drug Target for the Treatment of Erectile Dysfunction: A Multi-Omic and Epigenomic Association Study.** World J Mens Health, doi:[10.5534/wjmh.240131](https://doi.org/10.5534/wjmh.240131).

Zou, R., Shi, W., Chen, M., Zhang, M., Wu, D., Li, H., Zhou, H., Li, Y., Lu, W., Li, C. et Fan, X. (2024). **Phosphoglycerate mutase 1-mediated dephosphorylation and degradation of Dusp1 disrupt mitochondrial quality control and exacerbate endotoxemia-induced myocardial dysfunction.** Theranostics 14, 7488-7504, doi:[10.7150/thno.102647](https://doi.org/10.7150/thno.102647).

Zweers, H. E. E., Kroesen, S. H., Beerlink, G., Buit, E., Gerrits, K., Dorhout, A., van Wegberg, A. M. J., Janssen, M. C. H., Wortmann, S. B., Timmers, S. et Saris, C. G. J. (2024). **Ketogenic diet in adult patients with mitochondrial myopathy.** Mol Genet Metab 143, 108610, doi:[10.1016/j.ymgme.2024.108610](https://doi.org/10.1016/j.ymgme.2024.108610).