

# L'essentiel des Maladies Mitochondriales



**1<sup>er</sup> trimestre 2026**



## **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'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

---

### Sommaire

---

<b>ATAXIE DE FRIEDREICH - <i>FRIEDREICH ATAXIA</i></b> .....	<b>3</b>
<b>ATROPHIE OPTIQUE AUTOSOMIQUE DOMINANTE – <i>AUTOSOMAL DOMINANT OPTIC ATROPHY (ADOA)</i></b> .....	<b>7</b>
<b>ENCEPHALOPATHIE MYO-NEURO-GASTROINTESTINALE - <i>MITOCHONDRIAL NEUROGASTROINTESTINAL ENCEPHALOMYOPATHY (MNGIE)</i></b> .....	<b>8</b>
<b>MALADIE DE CHARCOT-MARIE-TOOTH D'ORIGINE MITOCHONDRIALE – <i>MITOCHONDRIAL CHARCOT-MARIE-TOOTH DISEASE</i></b> .....	<b>8</b>
<b>MALADIES LIEES AU GENE ACO2- <i>ACO2-RELATED DISORDERS</i></b> .....	<b>10</b>
<b>MALADIE LIEE AU GENE GFM1 – <i>GFM1-RELATED DISORDER</i></b> .....	<b>10</b>
<b>MALADIES LIEES AU GENE POLG (INCLUS SANDO, SCAE) - <i>POLG-RELATED DISORDERS (INCLUDED SANDO, SCAE)</i></b> .....	<b>10</b>
<b>MELAS</b> .....	<b>12</b>
<b>MERFF</b> .....	<b>15</b>
<b>NEUROPATHIE OPTIQUE HEREDITAIRE DE LEBER - <i>LEBER HEREDITARY OPTIC NEUROPATHY (LHON)</i></b> .....	<b>16</b>
<b>OPHTALMOPLÉGIE EXTERNE PROGRESSIVE – <i>PROGRESSIVE EXTERNAL OPHTHALMOPLÉGIA (PEO)</i></b> 19	
<b>SYNDROME D'ALPERS-HUTTENLOCHER - <i>ALPERS-HUTTENLOCHER SYNDROME</i></b> .....	<b>20</b>
<b>SYNDROME DE BARTH – <i>BARTH SYNDROME</i></b> .....	<b>20</b>
<b>SYNDROME DE KEARNS-SAYRE – <i>KEARNS-SAYRE SYNDROME</i></b> .....	<b>21</b>
<b>SYNDROME DE LEIGH - <i>LEIGH SYNDROME</i></b> .....	<b>22</b>
<b>MALADIES MITOCHONDRIALES (AUTRES) – <i>MITOCHONDRIAL DISORDERS (OTHER)</i></b> .....	<b>25</b>

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

---

### **Ataxie de Friedreich - Friedreich ataxia**

---

Annear, D. J., Vandeweyer, G. et Kooy, R. F. (2026). **CGG, CAG, and GAA: Genome-wide comparison of the disease linked trinucleotide short tandem repeats**. BMC Genomics 27, 302, doi:[10.1186/s12864-026-12651-9](https://doi.org/10.1186/s12864-026-12651-9).

Assali, A., Zaoui, F. et Bahoum, A. (2026). **Zygomatic Screws for Severe Open Bite Closure in a Young Friedreich's Ataxia Patient: A Case Report**. Case Rep Dent 2026, 4287988, doi:[10.1155/crid/4287988](https://doi.org/10.1155/crid/4287988).

Bachhuber, A. (2026). **[Trinucleotide repeat disorders]**. Radiologie (Heidelb) 66, 258-263, doi:[10.1007/s00117-026-01561-2](https://doi.org/10.1007/s00117-026-01561-2).

Baris, S., Dogan, M., Terali, K., Gezdirici, A., Erozu, R., Yucel, P. P., Kilic, H., Yavas, C., Yildirim, G. et Baris, I. (2026). **Biallelic Truncating DNAH14 Variant in Siblings with Neurodevelopmental Disorder and Predominant Ataxia: Clinical Report and Literature Review**. Int J Mol Sci 27, 575, doi:[10.3390/ijms27020575](https://doi.org/10.3390/ijms27020575).

Bernardi, E., López-Lombardía, Ó., Olmedo-Saura, G., Pagonabarraga, J., Kulisevsky, J. et Pérez-Pérez, J. (2026). **Hereditary Ataxias: From Pathogenesis and Clinical Features to Neuroimaging, Fluid, and Digital Biomarkers-A Scoping Review**. Int J Mol Sci 27, 881, doi:[10.3390/ijms27020881](https://doi.org/10.3390/ijms27020881).

Concepción, J., de Brun, C., Usmanov, G., Powell, L., Rogula, B., Dhand, A. et Schmahmann, J. D. (2026). **Personal Social Network Analysis in Cerebellar Ataxia: Exploring Correlations with Quality of Life and Functional Outcomes**. Cerebellum 25, 8, doi:[10.1007/s12311-025-01953-5](https://doi.org/10.1007/s12311-025-01953-5).

Dadsena, R., Romanzetti, S., Lischewski, S. A., Jing, Y., Timmann, D., Faber, J., Schulz, J. B., Reetz, K., Dogan, I., et FACROSS study group (2026). **Fronto-cerebellar connectivity disruptions and functional reorganization in Friedreich's Ataxia: A structural and resting-state fMRI study**. Neuroimage 332, 121872, doi:[10.1016/j.neuroimage.2026.121872](https://doi.org/10.1016/j.neuroimage.2026.121872).

de Oliveira, M. R. (2026). **Dimethyl fumarate and mitochondrial physiology: implications for neurological disorders**. Front Pharmacol 17, 1748360, doi:[10.3389/fphar.2026.1748360](https://doi.org/10.3389/fphar.2026.1748360).

Erdlenbruch, F. (2026). **[Friedreich's ataxia: The most common autosomal recessive inherited ataxia]**. MMW Fortschr Med 168, 40-42, doi:[10.1007/s15006-026-5619-2](https://doi.org/10.1007/s15006-026-5619-2).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Fleszar, Z., Thomas-Black, G., Garcia-Moreno, H., Cook, A. et Giunti, P. (2026). **An Exploration of Vitamin D Deficiency and Clinical Status in Friedreich's Ataxia Patients in the UK.** *Mov Disord Clin Pract*, doi:[10.1002/mdc3.70529](https://doi.org/10.1002/mdc3.70529).

Gaetz, W., Saleh, M. G., Birnbaum, C., Bloy, L., Roberts, T. P. L. et Lynch, D. R. (2026). **Myo-inositol elevation as an in vivo marker of reactive gliosis in pediatric Friedreich ataxia: evidence from HERMES-edited MR spectroscopy.** *Neuroimage Clin* 50, 103983, doi:[10.1016/j.nicl.2026.103983](https://doi.org/10.1016/j.nicl.2026.103983).

Ghika, N., Accolla, E., Girardin, F. et Theaudin, M. (2026). **[Pharmacotherapy of neuromuscular diseases: what's new in 2025].** *Rev Med Suisse* 22, 66-70, doi:[10.53738/REVMED.2026.22.945.48049](https://doi.org/10.53738/REVMED.2026.22.945.48049).

Grobe-Einsler, M., Borel, S., Buchholz, M., Sayah, S., Hilab, R., Pierron, L., Iskandar, A., Humphries, B., Ewencyk, C., Heinzmann, A., Atencio, M., Feldmann, K., Maas, V., Faber, J., Boesch, S., Indelicato, E., Reetz, K., Schulz, J. B., Bischoff, A. T., Klopstock, T., Schöls, L., Minnerop, M., Timmann, D., Davies, E. H., Klockgether, T., Durr, A., Xie, F. et Michalowsky, B. (2026). **Patient-reported, psychosocial and health economic outcomes in mild to moderate Friedreich's ataxia: baseline results of the PROFA study.** *Lancet Reg Health Eur* 61, 101552, doi:[10.1016/j.lanepe.2025.101552](https://doi.org/10.1016/j.lanepe.2025.101552).

Jee, E., Medha, M., Baek, H., Kim, J. et Kim, Y. (2026). **Mitochondrial iron overload is associated with lysosomal dysfunction-mediated mitophagy impairment in the heart of Friedreich's ataxia.** *Mitochondrion* 88, 102120, doi:[10.1016/j.mito.2026.102120](https://doi.org/10.1016/j.mito.2026.102120).

Jing, Y., Dogan, I., Dadsena, R., Faber, J., Schulz, J. B., Reetz, K., Romanzetti, S., et FACROSS study group (2026). **Multimodal Imaging Investigation of the Dentato-Thalamo-Cortical Pathway in Friedreich's Ataxia.** *Mov Disord* 41, 909-920, doi:[10.1002/mds.70179](https://doi.org/10.1002/mds.70179).

Kirboğa, K. K. et Küçüksille, E. U. (2026). **Integration of Evolutionary Analysis With RFdiffusion for De Novo Design of Aggregation-Resistant Frataxin.** *Proteins*, doi:[10.1002/prot.70114](https://doi.org/10.1002/prot.70114).

Kwa, F. A. A., Anjomani-Virmouni, S., Ramchunder, Z., Kendal, E. et Xiao, J. (2026). **Deciphering the missing links between Friedreich ataxia and multiple sclerosis for targeted drug development.** *Drug Discov Today* 31, 104644, doi:[10.1016/j.drudis.2026.104644](https://doi.org/10.1016/j.drudis.2026.104644).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Maddock, M. L., Miellet, S., Dongol, A., Hulme, A. J., Kennedy, C. K., Corben, L. A., Finol-Urdaneta, R. K., Nettel-Aguirre, A., Dionisi, C., Delatycki, M. B., Gottesfeld, J. M., Pandolfo, M., Soragni, E., Bidichandani, S. I., Lees, J. G., Lim, S. Y., Napierala, J. S., Napierala, M. et Dottori, M. (2026). **Friedreich ataxia transcriptomic dysregulation and identification of cell type-specific biomarkers: A systematic review and meta-analysis.** bioRxiv 2026.03.18.712785, doi:[10.64898/2026.03.18.712785](https://doi.org/10.64898/2026.03.18.712785).

Mehrban, A., Babamahmoodi, A., Hamidian, M. T., Amiri, B., Ramzani, P. et Karimi, M. (2026). **Coexistence of Friedreich's Ataxia and Esophageal Cancer: A Case Report.** Clin Case Rep 14, e72153, doi:[10.1002/ccr3.72153](https://doi.org/10.1002/ccr3.72153).

Miyashita, S., Zaki, A., Schlabach, K., Kim, G., Vierecke, J. K., Lynch, D., Tan, C., Hsich, E. et Alvarez, P. (2026). **Advanced Heart Failure in Friedreich's Ataxia: A Story of Challenges, Opportunities, and Hope.** JACC Case Rep 107391, doi:[10.1016/j.jaccas.2026.107391](https://doi.org/10.1016/j.jaccas.2026.107391).

Nguyen, H. T., Napierala, M. et Napierala, J. S. (2026). **Human pluripotent stem cell models of Friedreich's ataxia: innovations, considerations, and future perspectives.** Stem Cell Res Ther 17, 84, doi:[10.1186/s13287-025-04861-x](https://doi.org/10.1186/s13287-025-04861-x).

Pazos-Gil, M., Medina-Carbonero, M., Sanz-Alcázar, A., Portillo-Carrasquer, M., Oliveira-Jorge, L., Hernández, G., Sánchez, M., Delaspre, F., Cabisco, E., Ros, J. et Tamarit, J. (2026). **Pathological frataxin deficiency in mice causes tissue-specific alterations in iron homeostasis.** iScience 29, 114625, doi:[10.1016/j.isci.2025.114625](https://doi.org/10.1016/j.isci.2025.114625).

Petit, E., Beaubois-Gandoin, A., Durr, A., du Montcel, S. T. et Coarelli, G. (2026). **Quantifying Placebo Effects in Hereditary Ataxia Trials: A Meta-Analysis of Scale for the Assessment and Rating of Ataxia (SARA) Score Changes.** Mov Disord, doi:[10.1002/mds.70151](https://doi.org/10.1002/mds.70151).

Pignataro, M. F., Fernández, N. B., Garay-Alvarez, A., Pavan, M. F., Molina, R., Muñoz, I. G., Grossi, J., Noguera, M., Vila, A., García, A. E., Gentili, H. G., Rodríguez, N. A., Aran, M., Parreño, V., Bok, M., Hermoso, J. A., Ibañez, L. I. et Santos, J. (2026). **Nanobodies as tools for studying human frataxin biology.** Commun Biol 9, 181, doi:[10.1038/s42003-025-09458-x](https://doi.org/10.1038/s42003-025-09458-x).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Pontillo, G., Penna, S., Arrigoni, F., Bender, B., Boesch, S., Brunetti, A., Cendes, F., Chopra, S., Corben, L. A., Deistung, A., Delatycki, M. B., Diciotti, S., Dogan, I., Egan, G. F., França, M. C., Georgiou-Karistianis, N., Göricke, S. L., Henry, P.-G., Hernandez-Castillo, C. R., Hutter, D., Joers, J. M., Lenglet, C., Lindig, T., Lodi, R., Manners, D. N., Martinez, A. R. M., Martinuzzi, A., Marzi, C., Mascalchi, M., Nachbauer, W., Pane, C., Peruzzo, D., Pishardy, P. K., Reetz, K., Rezende, T. J. R., Romanzetti, S., Saccà, F., Schoels, L., Schulz, J. B., Stefani, A., Synofzik, M., Thomopoulos, S. I., Thompson, P. M., Timmann, D., Tonon, C., Vavla, M., Harding, I. H. et Coccozza, S. (2026). **Identification of Biological Subtypes of Friedreich Ataxia with Structural MRI-based Machine Learning.** *Radiology* 318, e251386, doi:[10.1148/radiol.251386](https://doi.org/10.1148/radiol.251386).

Portillo-Carrasquer, M., Sanz-Alcázar, A., Sánchez-López, B., Delaspre, F., Pazos-Gil, M., Oliveira-Jorge, L., Castells-Roca, L., Tamarit, J., Ros, J. et Cabiscol, E. (2026). **Targeting frataxin deficiency in DRG neurons and fibroblasts: omaveloxolone restores metabolic and iron balance to reduce ferroptosis.** *Biomed Pharmacother* 195, 119031, doi:[10.1016/j.biopha.2026.119031](https://doi.org/10.1016/j.biopha.2026.119031).

Raza, M. L., Rawalia, M. A., Fatima, Z. et Zehra, H. (2026). **The multifaceted nature of Friedreich ataxia: strategies for comprehensive patient care.** *Neurodegener Dis Manag* 1-9, doi:[10.1080/17582024.2026.2645501](https://doi.org/10.1080/17582024.2026.2645501).

Squillaci, G., Cotticelli, G. M., Carbone, V., Westfall, A. O., Wilson, R. B. et Morana, A. (2025). **Polyphenol-Enriched Fraction from Chestnut Shells as a Source of Bioactive Compounds for Friedreich Ataxia.** *Molecules* 31, 70, doi:[10.3390/molecules31010070](https://doi.org/10.3390/molecules31010070).

Yameogo, P., Gerhart, B. J., Sentmanat, M. F., Neilson, A., Cui, X., Verma, M., Lynch, D. R., Napierala, J. S. et Napierala, M. (2026). **Generation of Friedreich's ataxia induced pluripotent stem cells carrying the FXN c.165 + 5G>C splicing mutation.** *Stem Cell Res* 93, 103966, doi:[10.1016/j.scr.2026.103966](https://doi.org/10.1016/j.scr.2026.103966).

Zhang, Y., Ren, Y., Zhu, X., Liu, T., Han, R., Fang, Y., Zhao, Z., Mao, F., Wang, Y., Li, Xian et Li, Xiuhua (2025). **Research Progress on Idebenone in Neurodegenerative Diseases.** *Aging Med (Milton)* 8, 624-633, doi:[10.1002/agm2.70047](https://doi.org/10.1002/agm2.70047).

---

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

---

Kang, E. Y.-C., Tseng, Y.-J., Peng, W.-H., Hung, H.-C., Lin, P.-H., Montales, K. P., Sherman, E., Peregrin, J., Wang, E. H., Kang, C., Teng, Y.-C., Huang, C.-Y., Tsai, C.-L., Chang, I. Y.-F., Chen, J., Tezel, G., He, Y., Li, T.-D., Stiles, L., Shirihai, O., Tsang, S. H., Lai, C.-C., Tsai, C.-N., Lin, C.-S. et Wang, N.-K. (2026). **Disrupted energy metabolism is associated with retinal ganglion cell degeneration in autosomal dominant optic atrophy.** *Sci Adv* 12, eadx7815, doi:[10.1126/sciadv.adx7815](https://doi.org/10.1126/sciadv.adx7815).

Kawakita, M., Moteki, H., Nishio, S.-Y., Kobayashi, Y., Adachi, M., Okano, T., Yamazaki, H., Nakayama, J., Ohira, S., Ishino, T., Takumi, Y. et Usami, S.-I. (2026). **Frequency and Hearing Loss Phenotypes of OPA1 Variants in a Cohort of 18,475 Patients with Hearing Impairment.** *Genes (Basel)* 17, 341, doi:[10.3390/genes17030341](https://doi.org/10.3390/genes17030341).

Ovens, C. A., Grigg, J. R. et Fraser, C. L. (2026). **Visual Acuity Alone Is Not Enough: The Need for Multimodal Biomarkers in Dominant Optic Atrophy-Response.** *Clin Exp Ophthalmol* 54, 167-168, doi:[10.1111/ceo.70043](https://doi.org/10.1111/ceo.70043).

Pajareeyapong, P., Buathong, S., Thammasarnsophon, S., Sathianvichitr, K., Rattanathamsakul, N., Eiamsamarng, A., Chirapapaisan, N. et Ngamsombat, C. (2026). **Volumetric brain analysis and associated retinal thinning in autosomal dominant optic atrophy patients.** *Neuroimage Rep* 6, 100314, doi:[10.1016/j.ynirp.2025.100314](https://doi.org/10.1016/j.ynirp.2025.100314).

Roberti, G., Calabrese, A., Valiante, M., Formicola, D., Lolli, C. et De Negri, A. M. (2026). **Concomitant dominant optic atrophy and juvenile glaucoma in two siblings with a novel OPA1 splicing variant.** *Doc Ophthalmol* 152, 97-102, doi:[10.1007/s10633-025-10079-2](https://doi.org/10.1007/s10633-025-10079-2).

Ronfini, M., Prando, V., Di Mauro, V., Dokshokova, L., Lazzeri, E., Costantini, I., Alàn, L., Riviere, E. A., Incensi, A., Olianti, C., La Morgia, C., Liguori, R., Sacconi, L., Donadio, V., Scorrano, L., Carelli, V., Mongillo, M. et Zaglia, T. (2026). **OPA1 Deficiency Impairs NGF Signaling and Drives Sympathetic Neurodegeneration.** *JACC Basic Transl Sci* 11, 101460, doi:[10.1016/j.jacbts.2025.101460](https://doi.org/10.1016/j.jacbts.2025.101460).

Ware, M. A., Li, H. C. et Micieli, J. (2026). **A Case Report of Unilateral OPA3-Related Dominant Optic Atrophy.** *Case Rep Ophthalmol* 17, 81-86, doi:[10.1159/000550003](https://doi.org/10.1159/000550003).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

---

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

---

Amalnath, D., Saibaba, J., Wadwekar, V., Nagarajan, K. et Senthilvelan, S. (2026). **Mitochondrial DNA Maintenance Defects: Clinical, Imaging, and Genetic Spectrum of Four Patients from a Single Tertiary Care Centre.** Ann Indian Acad Neurol, doi:[10.4103/aian.aian\\_843\\_25](https://doi.org/10.4103/aian.aian_843_25).

Capece, G., Caumo, L., Volta, S., Riguzzi, P., Sogus, E., Petrosino, A., Vianello, S., Sabbatini, D., Salviati, L., Manara, R., Viscomi, C., Sorarù, G., Bello, L. et Pegoraro, E. (2026). **Wernicke Encephalopathy Complicating a Distinctive POLG Phenotype With MNGIE-Like Features.** Eur J Neurol 33, e70554, doi:[10.1111/ene.70554](https://doi.org/10.1111/ene.70554).

Fernández, B., Pérez-Moreno, G., Martínez-Arribas, B., Vidal, A. E., Ruiz-Pérez, L. M. et González-Pacanowska, D. (2026). **DCTPP1 orchestrates dCTP pool dynamics and mtDNA stability in quiescent cells.** Cell Death Dis, doi:[10.1038/s41419-026-08632-1](https://doi.org/10.1038/s41419-026-08632-1).

Kural, I., M MombEEK, L. M. et Wilson, D. M. (2026). **Role of mitochondria in neuronal function and survival in the enteric and central nervous systems.** Cell Mol Life Sci 83, 129, doi:[10.1007/s00018-025-06053-5](https://doi.org/10.1007/s00018-025-06053-5).

---

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

---

Aynaashe, A. et Kursula, P. (2026). **Genetic commonalities between rare subtypes of ALS and CMT: insights into molecular mechanisms of neurodegeneration.** Amino Acids 58, 8, doi:[10.1007/s00726-026-03500-w](https://doi.org/10.1007/s00726-026-03500-w).

Baris, S., Ipek, R., Baris, S. T. et Baris, I. (2026). **Expanding the Phenotypic Spectrum of NDUFS6-Related Disease: From Neonatal Mitochondrial Encephalopathy to Childhood-Onset Axonal Neuropathy.** Int J Mol Sci 27, 1375, doi:[10.3390/ijms27031375](https://doi.org/10.3390/ijms27031375).

Burns, J., Timmerman, V., Laurá, M., Yiu, E. M., D'Antonio, M., Mukherjee-Clavin, B., De Winter, J. et Scherer, S. S. (2026). **Charcot-Marie-Tooth disease and related neuropathies.** Nat Rev Dis Primers 12, 3, doi:[10.1038/s41572-025-00679-2](https://doi.org/10.1038/s41572-025-00679-2).

Cantarero, L., Hoenicka, J. et Palau, F. (2026). **Unraveling GDAP1: Bridging Mitochondrial Biology and Peripheral Neuropathy.** Biomolecules 16, 280, doi:[10.3390/biom16020280](https://doi.org/10.3390/biom16020280).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Chrysostomaki, M., Chatzi, D., Kyriakoudi, S. A., Meditskou, S., Manthou, M. E., Gargani, S., Theotokis, P. et Dermitzakis, I. (2026). **Hereditary Polyneuropathies in the Era of Precision Medicine: Genetic Complexity and Emerging Strategies**. *Genes (Basel)* 17, 56, doi:[10.3390/genes17010056](https://doi.org/10.3390/genes17010056).

Epstein, L., Weiner, A. C., Macklin, B. L., Kelly, K. R., Conklin, B. R. et Engelhardt, B. E. (2026). **Using image classifiers to predict CMT2A disease-relevant mitochondrial motility phenotypes in iPSC motor neurons**. *bioRxiv* 2026.03.16.712192, doi:[10.64898/2026.03.16.712192](https://doi.org/10.64898/2026.03.16.712192).

Goret, M., Piccolo, G. et Laporte, J. (2026). **Muscle-Specific DNMT2 Overexpression Improves Charcot-Marie-Tooth Disease In Vivo and Reveals a Narrow Therapeutic Window in Skeletal Muscle**. *Int J Mol Sci* 27, 1471, doi:[10.3390/ijms27031471](https://doi.org/10.3390/ijms27031471).

Hughes, S. E., Mehta, A. H. et Stucken, E. Z. (2026). **Evaluating Cochlear Implantation Outcomes in Charcot-Marie-Tooth Disease: A Case Series Analysis of Genetic Profiles and Intervention Timing**. *Otol Neurotol* 47, e525-e528, doi:[10.1097/MAO.0000000000004815](https://doi.org/10.1097/MAO.0000000000004815).

Joaquim, M., Dohrn, M. F., Chevrollier, A. et Escobar-Henriques, M. (2026). **Mitofusin MFN2 acts as a molecular sensor preventing protein aggregation and mitophagy, with a protective effect against apoptosis in Charcot-Marie-Tooth type 2A disease**. *Autophagy Rep* 5, 2629624, doi:[10.1080/27694127.2026.2629624](https://doi.org/10.1080/27694127.2026.2629624).

Ozes, B., Tong, L., Moss, K., Myers, M., Ndengabaganizi, I. et Sahenk, Z. (2026). **AAVrh74.tMCK.NT-3 Surrogate Gene Therapy in a Mouse Model of CMT2A**. *Int J Mol Sci* 27, 1942, doi:[10.3390/ijms27041942](https://doi.org/10.3390/ijms27041942).

Pan, X., Xie, J., Li, Z., Xiang, Y., Yu, Y., Cai, Q., Xu, H., Wan, Y. et Xing, J. (2026). **Stress-Driven Selective Neuronal Vulnerability in Charcot-Marie-Tooth Disease: From Prodromal Pathology to Therapeutic Implications**. *Cells* 15, 271, doi:[10.3390/cells15030271](https://doi.org/10.3390/cells15030271).

Rocco, A., Laurini, C., Falzone, Y. M., Calzavara, S., Del Carro, U., Previtali, S. C. et Maltecca, F. (2026). **Expanding the AFG3L2 Spectrum: A Link to Axonal Neuropathy**. *Neurol Genet* 12, e200368, doi:[10.1212/NXG.0000000000200368](https://doi.org/10.1212/NXG.0000000000200368).

Sun, B., Li, T. J., Chen, Z. H., Ling, L., Cheng, H. M., Huang, X. S. et Wang, H. F. (2026). **[Genetic variant analysis in patients with autosomal recessive demyelinating Charcot-Marie-Tooth disease]**. *Zhonghua Nei Ke Za Zhi* 65, 78-84, doi:[10.3760/cma.j.cn112138-20250917-00552](https://doi.org/10.3760/cma.j.cn112138-20250917-00552).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

---

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

---

Pas de résultat ce trimestre

---

### Maladie liée au gène **GFM1** – GFM1-related disorder

---

Molina-Berenguer, M., Herrero-Martínez, D., Vallbona-Garcia, A., Vila-Julià, F., Cámara, Y., Vales, Á., González-Asequinolaza, G., Torres-Torronteras, J. et Martí, R. (2026). **Systemic delivery of AAV-GFM1 corrects COXPD1 molecular alterations in Gfm1R671C/- mice.** EMBO Mol Med, doi:[10.1038/s44321-026-00426-4](https://doi.org/10.1038/s44321-026-00426-4).

---

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

---

Bermejo-Guerrero, L., Restrepo-Vera, J. L., Martín-Jimenez, P., Navarro-Riquelme, M., Garrido-Moraga, R., Ochoa, L. E., González-Quintana, A., Hernandez-Lain, A., Castillo-Villalba, J., Morís, G., Muelas, N., García-Arumí, E., González, V., Martínez-Sáez, E., Vesperinas, A., Llansó, L., Juntas-Morales, R., Paradas, C., Kapetanovic, S., Blázquez, A., Martí, R., Arenas, J., Martín, M. A. et Domínguez-González, C. (2026). **Clinical Heterogeneity and Candidate Biomarkers in POLG-Related Mitochondrial Disease.** Neurol Genet 12, e200365, doi:[10.1212/NXG.000000000200365](https://doi.org/10.1212/NXG.000000000200365).

Capece, G., Caumo, L., Volta, S., Riguzzi, P., Sogus, E., Petrosino, A., Vianello, S., Sabbatini, D., Salviati, L., Manara, R., Viscomi, C., Sorarù, G., Bello, L. et Pegoraro, E. (2026). **Wernicke Encephalopathy Complicating a Distinctive POLG Phenotype With MNGIE-Like Features.** Eur J Neurol 33, e70554, doi:[10.1111/ene.70554](https://doi.org/10.1111/ene.70554).

Damiano, M., Lambrecq, V., Nguyen-Michel, V.-H., Marois, C., Rucheton, B., Mochel, F., Gourfinkel-An, I., Demeret, S. et Navarro, V. (2026). **EEG, clinical, and MRI features of status epilepticus associated with mitochondrial diseases.** J Neurol 273, 160, doi:[10.1007/s00415-026-13681-9](https://doi.org/10.1007/s00415-026-13681-9).

Dille, Y., Rampakakis, E., Aubert, G., Dassi, C., Mannherz, W., Berrahmoune, S., Srouf, M., Buhas, D., Agarwal, S. et Myers, K. A. (2026). **Short telomeres in mitochondrial DNA depletion disorders.** Mitochondrion 88, 102131, doi:[10.1016/j.mito.2026.102131](https://doi.org/10.1016/j.mito.2026.102131).

Efthymiou, E., Büchele, F., Mahendran, S., Stieglitz, L. et Balint, B. (2026). **POLG-Related Parkinsonism with Good Response to Deep Brain Stimulation.** Mov Disord Clin Pract, doi:[10.1002/mdc3.70588](https://doi.org/10.1002/mdc3.70588).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Franczak, E., Montgomery, M. M., Terwilliger, Z. S., Aruleba, R. T., Krassovskaia, P., Boykov, I. N., Hagen, J. T., Pacheco, E. A., Chrest, B. R., Zeczycki, T. N., Vandiver, K. J., Neufer, P. D., McClung, J. M. et Fisher-Wellman, K. H. (2026). **Accumulated mtDNA mutations are linked to specific impairments in NADH-linked respiration.** *iScience* 29, 115184, doi:[10.1016/j.isci.2026.115184](https://doi.org/10.1016/j.isci.2026.115184).

Fumini, V., Gilea, A. I., Tacchetto, E., Salviati, L., Baruffini, E. et Doimo, M. (2026). **Progressive cognitive impairment and ventricular tachycardia in a boy with biallelic POLG variants and a de novo RYR2 variation.** *Sci Rep*, doi:[10.1038/s41598-026-44913-7](https://doi.org/10.1038/s41598-026-44913-7).

Hsieh, Y.-H., Kautz, P., Nitsch, L., Giguelay, A. M., Liebold, J., Dimitrova, V., Contreras Castillo, S., Jungen, F., Zsurka, G., Trombly, G., Schuelke, M., Kunz, W. S., Lareau, C. A. et Ludwig, L. S. (2026). **Single-cell multi-omic analysis of mitochondrial mutational mosaicism and dynamics.** *Nat Commun* 17, 2532, doi:[10.1038/s41467-026-70399-y](https://doi.org/10.1038/s41467-026-70399-y).

Kane, M. (2026). **Valproic Acid Therapy and POLG Genotype.** In: **Medical Genetics Summaries**, Éd. Pratt, V. M., Scott, S. A., Pirmohamed, M., Esquivel, B., Kattman, B. L., et Malheiro, A. J. National Center for Biotechnology Information (US), Bethesda (MD), URL: <http://www.ncbi.nlm.nih.gov/books/NBK620296/> [Consulté le avril 2026].

Ratnaike, T., Ramanan, S., Elkhateeb, N., Narayanan, R., Yang, J., Arany, E. S., Mirchandani, M., Piper, R., Schon, K., Kule, M. E., Gilmartin, C., Lochmüller, A., Shaw, E., Horváth, R. et Chinnery, P. F. (2026). **Charting the phenotypic landscape of mitochondrial diseases through a systematic evaluation of pathogenic mitochondrial DNA and nuclear gene variants.** *Genet Med* 28, 101620, doi:[10.1016/j.gim.2025.101620](https://doi.org/10.1016/j.gim.2025.101620).

Sadiq, E., Liu, Y., Li, N., Zong, C., Li, X., Yu, T., Pu, Z., Alameri, L., Li, Z., Li, S., Wang, X. et Zhao, R. (2026). **Growth hormone enhances mitochondria biogenesis and endows mitochondrial thermogenesis in murine adipocytes.** *Mol Cell Endocrinol* 616, 112758, doi:[10.1016/j.mce.2026.112758](https://doi.org/10.1016/j.mce.2026.112758).

Sørensen, N. B., Stamenkovic, K., Steffensen, E. H. et Kofoed, I. R. (2026). **[Super-refractory status epilepticus caused by hereditary mitochondrial disease].** *Ugeskr Laeger* 188, V07250609, doi:[10.61409/V07250609](https://doi.org/10.61409/V07250609).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

---

### MELAS

---

Banerjee, S., Mondal, R., Deb, S., Shome, G., Chakraborty, S., Saha, C., Pandit, A., Sen, G., Bhattacharya, S., Sen, P., Bhattacharya, N. P., Roy, J., Chowdhury, A. et Benito-León, J. (2026). **Integrated molecular and clinical profiling of primary mitochondrial oxidative phosphorylation disorders in an Indian cohort: Insights from genetics, neuroimaging, and machine learning.** *Mitochondrion* 89, 102150, doi:[10.1016/j.mito.2026.102150](https://doi.org/10.1016/j.mito.2026.102150).

Damiano, M., Lambrecq, V., Nguyen-Michel, V.-H., Marois, C., Rucheton, B., Mochel, F., Gourfinkel-An, I., Demeret, S. et Navarro, V. (2026). **EEG, clinical, and MRI features of status epilepticus associated with mitochondrial diseases.** *J Neurol* 273, 160, doi:[10.1007/s00415-026-13681-9](https://doi.org/10.1007/s00415-026-13681-9).

Dodulík, J., Lazárová, M., Kapsová, E. et Václavík, J. (2026). **MELAS Syndrome Presenting with Hypertrophic Cardiomyopathy and Advanced Heart Failure: A Multisystem Diagnostic Challenge.** *J Clin Med* 15, 1109, doi:[10.3390/jcm15031109](https://doi.org/10.3390/jcm15031109).

Finsterer, J. (2026a). **Before diagnosing immunological cerebellitis in an m.3243A>G carrier, a cerebellar stroke-like lesion should be ruled out.** *Rinsho Shinkeigaku* 66, 190-191, doi:[10.5692/clinicalneuroi.cn-002162](https://doi.org/10.5692/clinicalneuroi.cn-002162).

Finsterer, J. (2026b). **Multidimensional MRI is only one piece of the puzzle in the diagnosis of stroke-like episodes in MELAS.** *Quant Imaging Med Surg* 16, 182, doi:[10.21037/qims-2025-aw-2171](https://doi.org/10.21037/qims-2025-aw-2171).

Finsterer, J. (2026c). **Sleep performance in MELAS is related not only to the syndrome, but also to several other endogenous and exogenous determinants.** *Sleep Breath* 30, 97, doi:[10.1007/s11325-026-03647-6](https://doi.org/10.1007/s11325-026-03647-6).

Finsterer, J. (2026d). **The Loss and Dysfunction of Smooth Muscle Cells in MELAS Are Not the Only Cause for Gastrointestinal Dysmotility.** *Neurogastroenterol Motil* 38, e70284, doi:[10.1111/nmo.70284](https://doi.org/10.1111/nmo.70284).

Gaddigoudar, M. S., Khera, R., Gupta, R. et Wander, A. (2026). **Atypical subacute sclerosing panencephalitis mimicking mitochondrial encephalopathy, lactic acidosis and stroke-like episodes.** *Neurol Sci* 47, 383, doi:[10.1007/s10072-026-08972-y](https://doi.org/10.1007/s10072-026-08972-y).

Herrero San Martin, A., Arias, E. M., Dominguez Gonzalez, C., Morales Conejo, M. et Diaz Cambriles, T. (2026). **Sleep in MELAS syndrome.** *Sleep Breath* 30, 45, doi:[10.1007/s11325-026-03611-4](https://doi.org/10.1007/s11325-026-03611-4).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Hoang, D., Pestronk, A. et Kafaie, J. (2026). **Progressive Myopathy and Respiratory Failure in a 7-Year-Old Boy With m.3251A>G MT-TL1 Mutation.** J Clin Neuromuscul Dis 27, 89-95, doi:[10.1097/CND.0000000000000547](https://doi.org/10.1097/CND.0000000000000547).

Ito, Y., Ochi, C., Yamanishi, Y., Takashima, H., Hashiguchi, A. et Nagai, M. (2026). **A case of recurrent cerebellitis leading to the diagnosis of mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes (MELAS).** Rinsho Shinkeigaku 66, 192-193, doi:[10.5692/clinicalneurolog.cn-002200](https://doi.org/10.5692/clinicalneurolog.cn-002200).

Kyriakopoulos, A. M., McCullough, P. A. et Seneff, S. (2026). **Taurine intake ameliorates lactic acidosis and hyperferritinemia occurring after mRNA SARS-CoV-2 vaccination in a patient with  $\beta$ -thalassemia trait: a case report and review of literature.** J Med Case Rep 20, 108, doi:[10.1186/s13256-026-05844-z](https://doi.org/10.1186/s13256-026-05844-z).

Lee, K., Ahn, D., Jung, G. E. et Lee, W. (2026). **Middle-aged woman presenting with new-onset status epilepticus and family history of MELAS syndrome but negative mtDNA testing.** Epileptic Disord, doi:[10.1002/epd2.70236](https://doi.org/10.1002/epd2.70236).

Li, J., Wang, X., Guo, Y., Wang, W., Wu, S. et Sun, J. (2026). **Fahr Syndrome, Hypoparathyroidism and Mitochondrial Encephalomyopathy With Lactic Acidosis and Stroke-Like Episodes (MELAS) Syndrome.** AACE Endocrinol Diabetes 13, 102-106, doi:[10.1016/j.aed.2025.09.007](https://doi.org/10.1016/j.aed.2025.09.007).

Lin, Y.-H., Wang, X.-W., Li, Y.-A., Chen, T.-Y., Lian, W.-S., Wang, F.-S., Lan, M.-Y., Liou, C.-W. et Lin, T.-K. (2026). **Dysregulated iron homeostasis Drives mitochondrial Injury and ferroptosis susceptibility in MELAS fibroblasts.** Mitochondrion 88, 102132, doi:[10.1016/j.mito.2026.102132](https://doi.org/10.1016/j.mito.2026.102132).

Liu, Q., Yang, L., Liu, P., Wang, X., Peng, K., Liu, Qi et Zhang, K. (2026). **A case of delayed acute intestinal pseudo-obstruction after MELAS crisis in mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes.** Front Med (Lausanne) 13, 1760790, doi:[10.3389/fmed.2026.1760790](https://doi.org/10.3389/fmed.2026.1760790).

Liu, X., Zhang, Z.-S. et Tao, Q.-Q. (2026). **Neuronal intranuclear inclusion disease: a diagnostic pitfall for MELAS.** BMC Neurol 26, 178, doi:[10.1186/s12883-026-04705-y](https://doi.org/10.1186/s12883-026-04705-y).

Lu, Y.-H., Niu, D.-M. et Lin, W.-S. (2025). **Low-Frequency Photoparoxysmal Responses in a Patient With MELAS.** Rev Neurol 80, 40724, doi:[10.31083/RN40724](https://doi.org/10.31083/RN40724).

Luo, Y., Chu, Q., Yu, T., Lu, X., Wang, Y., Li, J., Wu, L. et Shen, Y. (2026). **Migratory vasodilatation of cerebral arteries in MELAS episodes: a case report and literature review.** Front Immunol 17, 1706012, doi:[10.3389/fimmu.2026.1706012](https://doi.org/10.3389/fimmu.2026.1706012).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Machida, A., Shirai, K., Oyama, J., Usui, E. et Kakuta, T. (2026). **MERRF/MELAS overlap syndrome mimicking paradoxical cerebral embolism due to patent foramen ovale.** *Neurol Sci* 47, 395, doi:[10.1007/s10072-026-09012-5](https://doi.org/10.1007/s10072-026-09012-5).

Maresca, A., Moresco, M., Amore, G., La Morgia, C., Valentino, M. L., Capirossi, G., Sacchetti, G., Carelli, V., Vedeler, C., Bindoff, L. A. et Varhaug, K. N. (2026). **Biomarking MELAS with neurofilament light chain and circulating cell free mitochondrial DNA.** *Mol Genet Metab* 147, 109753, doi:[10.1016/j.ymgme.2026.109753](https://doi.org/10.1016/j.ymgme.2026.109753).

Nagao, S., Aoki, M., Yatsuga, S., Takeshita, S., Tsuboi, Y. et Uesugi, N. (2026). **Loss of Myofilaments in Gastrointestinal Smooth Muscle: A Novel Pathological Finding in MELAS-Associated Chronic Intestinal Pseudo-Obstruction.** *Neurogastroenterol Motil* 38, e70226, doi:[10.1111/nmo.70226](https://doi.org/10.1111/nmo.70226).

Nilsén, V., Bojstedt, J. et Nordström, J. (2026). **Case Report: Simultaneous pancreas-kidney transplantation in MELAS: first reported case with 5-year follow-up.** *Front Transplant* 5, 1737352, doi:[10.3389/frtra.2026.1737352](https://doi.org/10.3389/frtra.2026.1737352).

Purandare, N., Pasupathi, V., Padhan, D., Rai, S., Grossman, L. I. et Aras, S. (2026). **Transcriptional activation by MNRR1 is effected by recruiting p300 and can be induced by minimal peptides.** *Mitochondrion* 88, 102119, doi:[10.1016/j.mito.2026.102119](https://doi.org/10.1016/j.mito.2026.102119).

Shu, J., Zhou, Y., Xu, Z., Wei, W., Zhao, L. et Chang, J. (2026). **MELAS syndrome complicated by anti-GFAP autoantibody positivity: a case report and literature review.** *BMC Neurol* 26, 244, doi:[10.1186/s12883-026-04745-4](https://doi.org/10.1186/s12883-026-04745-4).

Sideris, D., Lee, H., Olson, L., Nallaparaju, K., Okuyama, K., Ciavarri, J., Lafyatis, R., Larsen, M., Lin, B., Alfaras, I., Kennerdell, J., Finkel, T., Liu, Y., Chen, B. et Lyu, L. (2026). **Suppression of interferon signaling via small-molecule modulation of TFAM.** *Elife* 14, RP108742, doi:[10.7554/eLife.108742](https://doi.org/10.7554/eLife.108742).

Skinner, O. S., Miranda, M., Dong, F., Struhl, T., Walker, M. A., Schleifer, G., Henke, M. T., Clardy, J., Hirano, M., De Vivo, D. C., Schon, E. A., Engelstad, K., Siegmund, S. E., Laprise, C., Des Rosiers, C., Mootha, V. K. et Sharma, R. (2026). **4,5-dihydroxyhexanoic acid is a robust circulating and urine marker of mitochondrial disease and its severity.** *bioRxiv* 2026.02.10.705117, doi:[10.64898/2026.02.10.705117](https://doi.org/10.64898/2026.02.10.705117).

Varga, L., Borecka, S., Skopkova, M., Rambani, V., Sklenar, M., Cipkova, K., Kickova, T., Ugorova, D., Kabatova, Z., Stanik, J., Profant, M. et Gasperikova, D. (2026). **Screening for Maternally Inherited Diabetes and Deafness in Large Cohorts of Hearing Impaired and Diabetic Patients.** *Ear Hear* 47, 863-876, doi:[10.1097/AUD.0000000000001780](https://doi.org/10.1097/AUD.0000000000001780).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Yang, F., Yao, R., Chang, G., Hu, J., Feng, B., Wang, L., Hu, F., Huang, Y., Wu, S., Yu, T., Ding, Y. et Wang, X. (2026). **Clinical and genetic spectrum of pediatric mitochondrial disorders in China: insights from a 47-case genetically confirmed cohort.** Orphanet J Rare Dis 21, 158, doi:[10.1186/s13023-025-04180-7](https://doi.org/10.1186/s13023-025-04180-7).

Yu, Q., Wang, R., Sun, C., Hu, B., Liu, X., Yang, L., Lin, J., Li, Y. et Geng, D. (2026). **Dynamic functional connectivity changes in the triple networks in patients with mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes.** Quant Imaging Med Surg 16, 139, doi:[10.21037/qims-2025-807](https://doi.org/10.21037/qims-2025-807).

Zeng, Y., Yang, Y., Wang, W., Peng, Q., Bai, Y., Yu, X., Yang, S. et Ren, L. (2026). **TSPO-PET highlights an atypical mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes (MELAS) phenotype.** Neurol Sci 47, 372, doi:[10.1007/s10072-026-08998-2](https://doi.org/10.1007/s10072-026-08998-2).

Zhang, X., Lu, X. P., Xu, T. T., Ren, L. J., Xu, J. X. et Song, C. D. (2026). **[Two cases of mitochondrial encephalomyopathy with lactic acidosis and stroke like episodes complicated by Henoch-Schönlein purpura nephritis].** Zhonghua Er Ke Za Zhi 64, 213-215, doi:[10.3760/cma.j.cn112140-20250808-00731](https://doi.org/10.3760/cma.j.cn112140-20250808-00731).

Zhou, S., Tian, Y., Liu, F., Wu, E. et Feng, X. (2026). **Mitochondrial Myopathy, Lactic Acidosis, and Stroke-Like Episodes Combined with Diabetes: A Case Report.** Endocr Metab Immune Disord Drug Targets, doi:[10.2174/0118715303419754251125093050](https://doi.org/10.2174/0118715303419754251125093050).

Zhuang, X., Wang, Jiayin, Wang, Jianing, Lin, Y., Yan, C. et Ji, K. (2026). **A novel tRNASer(AGY) 12244G > a variant impairs mitochondrial function and presents with classical MELAS phenotype.** Neurol Sci 47, 264, doi:[10.1007/s10072-026-08875-y](https://doi.org/10.1007/s10072-026-08875-y).

---

### MERFF

Cherian, A. et Divya, K. P. (2026). **Progressive Myoclonic Epilepsies - A Pragmatic Review.** Neurol India 74, 175-183, doi:[10.4103/neurol-india.Neurol-India-D-25-00075](https://doi.org/10.4103/neurol-india.Neurol-India-D-25-00075).

Damiano, M., Lambrecq, V., Nguyen-Michel, V.-H., Marois, C., Rucheton, B., Mochel, F., Gourfinkel-An, I., Demeret, S. et Navarro, V. (2026). **EEG, clinical, and MRI features of status epilepticus associated with mitochondrial diseases.** J Neurol 273, 160, doi:[10.1007/s00415-026-13681-9](https://doi.org/10.1007/s00415-026-13681-9).

Machida, A., Shirai, K., Oyama, J., Usui, E. et Kakuta, T. (2026). **MERRF/MELAS overlap syndrome mimicking paradoxical cerebral embolism due to patent foramen ovale.** Neurol Sci 47, 395, doi:[10.1007/s10072-026-09012-5](https://doi.org/10.1007/s10072-026-09012-5).

---

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

---

Ajmera, P., Guion, D., Barnes, S., Sadun, A. A. et Alexandrova, A. N. (2026). **Molecular Mechanism of Mitochondrial Complex I Disruption by m.14484T>C Underlying Leber Hereditary Optic Neuropathy.** bioRxiv 2026.01.28.701874, doi:[10.64898/2026.01.28.701874](https://doi.org/10.64898/2026.01.28.701874).

Banerjee, S., Mondal, R., Deb, S., Shome, G., Chakraborty, S., Saha, C., Pandit, A., Sen, G., Bhattacharya, S., Sen, P., Bhattacharya, N. P., Roy, J., Chowdhury, A. et Benito-León, J. (2026). **Integrated molecular and clinical profiling of primary mitochondrial oxidative phosphorylation disorders in an Indian cohort: Insights from genetics, neuroimaging, and machine learning.** Mitochondrion 89, 102150, doi:[10.1016/j.mito.2026.102150](https://doi.org/10.1016/j.mito.2026.102150).

Clarke, J. E., Yu, A., Kawakibi, T., Le, M., Pack, D., Alves, C. et Acharya, J. (2026). **Diagnostic and Imaging Features of Leber Hereditary Optic Neuropathy: An Individual Participant Data Meta-Analysis.** AJNR Am J Neuroradiol ajnr.A9240, doi:[10.3174/ajnr.A9240](https://doi.org/10.3174/ajnr.A9240).

Cyrino, L. G., Saldaña Lagos, B. J., Varalta Martins, P. H., Bonato Cavalcanti, J. F., Ariello, L. E. et Monteiro, M. L. R. (2026). **Visual Loss from Leber's Optic Neuropathy Presenting in a 76-Year-Old Man with the 14484 Mutations.** Neuroophthalmology 50, 159-166, doi:[10.1080/01658107.2025.2487842](https://doi.org/10.1080/01658107.2025.2487842).

De Napoli, G., Ferraro, D., Fiore, A., Covelli, A., Bianchi, A., Fabiani, C., De Stefano, N., Ulivelli, M. et Cortese, R. (2026). **Atypical clinical and MRI features in Leber hereditary optic neuropathy: a case series of four patients.** Neurol Sci 47, 82, doi:[10.1007/s10072-025-08645-2](https://doi.org/10.1007/s10072-025-08645-2).

Fang, L., Fu, K., Yang, M., Xu, Y., Ukamaka, E. S., Qu, D., Huang, T. et Hu, J. (2026). **Oxidative stress imbalance and cellular damage mediated by the ND4 G11778A mutation.** Sci Rep 16, 10122, doi:[10.1038/s41598-026-40061-0](https://doi.org/10.1038/s41598-026-40061-0).

Iwaki, Y., Ueda, K., Mori, S., Sakamoto, M., Yamada-Nakanishi, Y., Kondo, M. et Nakamura, M. (2026). **Difference in pupillary response to red and blue color stimuli between glaucoma and Leber hereditary optic neuropathy patients with comparable central visual dysfunction.** Doc Ophthalmol, doi:[10.1007/s10633-025-10071-w](https://doi.org/10.1007/s10633-025-10071-w).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

KamaliZonouzi, S. et Micieli, J. (2026). **Leber Hereditary Optic Neuropathy-Associated Novel Mutation in MT-RNR2 Gene: A Case Report.** *Case Rep Ophthalmol* 17, 75-80, doi:[10.1159/000550116](https://doi.org/10.1159/000550116).

Kawano, H., Nakazawa, M., Kawano, K., Terasaki, H. et Sakamoto, T. (2026). **A Case Report of Late-Onset Leber's Hereditary Optic Neuropathy Diagnosed Following Vision Loss After Cataract Surgery.** *Neuroophthalmology* 50, 180-185, doi:[10.1080/01658107.2025.2495295](https://doi.org/10.1080/01658107.2025.2495295).

Krasniakova, M., Pansell, T. et Gustafsson, J. (2026). **Impact of individualized colored spectacle filters on photophobia and visual comfort in central visual field defect patients: a one-year study.** *Sci Rep* 16, 10504, doi:[10.1038/s41598-026-45302-w](https://doi.org/10.1038/s41598-026-45302-w).

Leonel Boone, D., Cavalcanti Costa, M., Candeias da Silva, C., Arb Saba Rodrigues Pinto, R., de Carvalho Aguiar, P., Harumi Tengan, C., Borges, V. et Ballalai Ferraz, H. (2026). **Progressive generalized dystonia caused by a mutation in the MT-ND6 gene.** *Parkinsonism Relat Disord* 145, 108239, doi:[10.1016/j.parkreldis.2026.108239](https://doi.org/10.1016/j.parkreldis.2026.108239).

Li, H., Ai, C., Jin, X., Wang, J., Yu, J., Gao, Y., Wallace, D. C. et Guan, M.-X. (2026). **Optic neuropathy arising from the synergy between YARS2 and mitochondrial COX1 mutations.** *J Genet Genomics* S1673-8527(26)00047-0, doi:[10.1016/j.jgg.2026.02.003](https://doi.org/10.1016/j.jgg.2026.02.003).

Mikulenaite, P., Vilkeviciute, A., Stramkauskaite, A., Povilaityte, I., Jurkute, N. et Liutkeviciene, R. (2026). **Leber Hereditary Optic Neuropathy Caused by the Rare MT-ND1 m.3394T>C Mutation: A Case With Favorable Visual Prognosis and a Literature Review.** *Cureus* 18, e103261, doi:[10.7759/cureus.103261](https://doi.org/10.7759/cureus.103261).

Nair, A. P., Janaki P, A., Gopalarethinam, J., Kumar B, A., Vellingiri, B. et Subramaniam, M. D. (2026). **Mesenchymal stem cell mitochondrial transfer effectively protects Leber's Hereditary Optic Neuropathy (LHON) mutant cells from mitochondrial damage.** *Acta Histochem* 128, 152331, doi:[10.1016/j.acthis.2026.152331](https://doi.org/10.1016/j.acthis.2026.152331).

Oh, S.-Y., Park, J., Choi, J.-H., Shin, J. et Lee, H.-J. (2026). **Chronic Progressive External Ophthalmoplegia Associated With the m.14484T>C Leber Hereditary Optic Neuropathy Mutation.** *J Neuroophthalmol*, doi:[10.1097/WNO.0000000000002439](https://doi.org/10.1097/WNO.0000000000002439).

Okrent Smolar, A. L., Viswanath, R., Bomze, H. M., Hao, Y., Stinnett, S. S. et Gospe, S. M. (2026). **Pharmacological Depletion of Retinal Mononuclear Phagocytes Is Neuroprotective in a Mouse Model of Mitochondrial Optic Neuropathy.** *Invest Ophthalmol Vis Sci* 67, 6, doi:[10.1167/iovs.67.2.6](https://doi.org/10.1167/iovs.67.2.6).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Sánchez-Fernández, B., Zamorano-González, P., Martín-Montañez, E., Alba-Linero, C., Rius-Díaz, F., García-Fernandez, M., Luque-Aranda, R. et García-Basterra, I. (2025). **Oxidative Stress and Inflammatory Biomarkers in Aqueous Humor and Blood of Patients with Leber's Hereditary Optic Neuropathy.** *Antioxidants (Basel)* 15, 51, doi:[10.3390/antiox15010051](https://doi.org/10.3390/antiox15010051).

Sherratt-Mayhew, S., Page, C., Lowe, M., Mollan, S. P. et Berman, G. (2026). **Infographic: Randomized trial of bilateral gene therapy injection for m.11778 G > A MT-ND4 Leber optic neuropathy (REFLECT Trial).** *Eye (Lond)*, doi:[10.1038/s41433-026-04266-x](https://doi.org/10.1038/s41433-026-04266-x).

Takai, Y., Yamagami, A., Iwasa, M., Inoue, K., Yasumoto, R., Ishikawa, H. et Wakakura, M. (2026). **Five-year post-onset visual acuity trajectories in Japanese Leber hereditary optic neuropathy: a longitudinal analysis by age, sex, and mtDNA mutation.** *Can J Ophthalmol* S0008-4182(26)00079-7, doi:[10.1016/j.jcjo.2026.02.011](https://doi.org/10.1016/j.jcjo.2026.02.011).

Wang, D., Yuan, J., Liu, H.-L., Yuan, H., Ma, N., Chen, M.-L., Li, B., Jie, H. et Zhang, T. (2025). **Comparison of thickness changes in retinal nerve fibre layer in Leber's hereditary optic neuropathy patients with 11778, 14484 and 3460 mutations.** *Transl Pediatr* 14, 3420-3428, doi:[10.21037/tp-2025-589](https://doi.org/10.21037/tp-2025-589).

Wang, J., Li, J., Dai, L., Zhu, X., Long, Y., Meng, X., Li, S., Huang, X. et Guo, H. (2026). **A multistage cost-effective strategy for the molecular diagnosis of unexplained vision loss patients: practice in inherited ocular fundus disease.** *Mol Genet Genomics* 301, 78, doi:[10.1007/s00438-025-02321-y](https://doi.org/10.1007/s00438-025-02321-y).

Wu, J., Pan, C., Zhu, R., Huang, X., Lou, X. et Yang, L. (2026). **A Heteroplasmic MT-CO2 m.8024G > A Variant Is Associated with Mitochondrial Bioenergetic Deficiency and Optic Atrophy.** *Mol Neurobiol* 63, 485, doi:[10.1007/s12035-026-05774-3](https://doi.org/10.1007/s12035-026-05774-3).

Wu, J.-Y., Ye, S., Yin, T.-L., Zhang, S., Zheng, D.-F., Fu, J.-Y., Ma, G.-W. et Fan, D.-S. (2025). **Amyotrophic Lateral Sclerosis With Concurrent LHON-associated m.14484T>C Mutation: A Case Report and Literature Review.** *Rev Neurol* 80, 44110, doi:[10.31083/RN44110](https://doi.org/10.31083/RN44110).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

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

Banerjee, S., Mondal, R., Deb, S., Shome, G., Chakraborty, S., Saha, C., Pandit, A., Sen, G., Bhattacharya, S., Sen, P., Bhattacharya, N. P., Roy, J., Chowdhury, A. et Benito-León, J. (2026). **Integrated molecular and clinical profiling of primary mitochondrial oxidative phosphorylation disorders in an Indian cohort: Insights from genetics, neuroimaging, and machine learning.** *Mitochondrion* 89, 102150, doi:[10.1016/j.mito.2026.102150](https://doi.org/10.1016/j.mito.2026.102150).

Lang, S. H., Cottingham, N., Donnelly, C., Risen, S., Muncher, R. M., Brewer, E. D., Saland, J. M., Benchimol, C., Scaglia, F. et Ganesh, J. (2026). **Outcomes of kidney transplantation in three patients with single large-scale mitochondrial DNA deletion syndromes.** *Mol Genet Metab* 147, 109731, doi:[10.1016/j.ymgme.2026.109731](https://doi.org/10.1016/j.ymgme.2026.109731).

Lopriore, P., Ünlütürk, Z., Klopstock, T., Karaa, A., Rouzier, C., Domínguez-González, C., Lamperti, C., Mancuso, M., Twinkle-Related Disorders International Consortium for Trial Readiness (TReDIC), Cecchi, G., Montano, V., Siciliano, G., Nicoletta, V., Maioli, M., Primiano, G., Servidei, S., La Morgia, C., Carelli, V., Valentino, M. L., Caporali, L., Arena, I. G., Musumeci, O., Lopergolo, D., Malandrini, A., Gallus, G. N., Filosto, M., Bello, L., Pegoraro, E., Comi, G. P., Magri, F., Ronchi, D., Di Fonzo, A., Percetti, M., Azzimonti, M., Büchner, B., Prokisch, H., Bermejo-Guerrero, L., Procaccio, V., Gaignard, P., Echaniz-Laguna, A., Schiff, M., Rötig, A., Toutain, A., Paquis-Flucklinger, V., Morel, G., Robin, S., Nadaj-Pakleza, A., Chanson, J.-B., Chaussenot, A., Ait-El-Mkadem Saadi, S., Trimouille, A., Tranchant, C., Salort-Campana, E., Bieth, E., Sacconi, S., Duval, F., Restrepo Vera, J. L., Molnar, M. J., Vissing, J., Haas, R., Larson, A., Enns, G. M., Parikh, S., Goldstein, A. et Hirano, M. (2026). **Clinical and Genotypic Spectrum of Twinkle-Related Disorders: Insights From a Multinational Cohort Study.** *Neurology* 106, e214401, doi:[10.1212/WNL.0000000000214401](https://doi.org/10.1212/WNL.0000000000214401).

Martín-Jimenez, P., Bermejo-Guerrero, L., Ochoa, L. E., Navarro-Riquelme, M., Garrido-Moraga, R., Hernández-Laín, A., Hernández-Voth, A., González Quintana, A., Bermejo-Moriñigo, A., González-Méndez, V., Martín-Arriscado Arroba, C., Smirnov, D., Konstantinovskiy, N., Arenas, J., Martín, M. Á., Blázquez, A. et Domínguez-González, C. (2026). **Exploring Outcome Measures for Mitochondrial Myopathies; Insights From a Longitudinal Study on TK2 Deficiency.** *J Inherit Metab Dis* 49, e70147, doi:[10.1002/jimd.70147](https://doi.org/10.1002/jimd.70147).



## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Mayer, D., Kartsonaki, E., Wilder-Smith, E., Schaller, A., Frank, S., Bohlhalter, S. et Mihaylova, V. (2026). **A Novel Truncating Pathogenic Variant in RRM2B in a Kurdish Family With Autosomal-Dominant Chronic Progressive External Ophthalmoplegia Plus (PEOA5).** J Clin Neuromuscul Dis 27, 108-111, doi:[10.1097/CND.0000000000000514](https://doi.org/10.1097/CND.0000000000000514).

Oh, S.-Y., Park, J., Choi, J.-H., Shin, J. et Lee, H.-J. (2026). **Chronic Progressive External Ophthalmoplegia Associated With the m.14484T>C Leber Hereditary Optic Neuropathy Mutation.** J Neuroophthalmol, doi:[10.1097/WNO.0000000000002439](https://doi.org/10.1097/WNO.0000000000002439).

Pajareeyapong, P., Buathong, S., Thammasarnsophon, S., Sathianvichitr, K., Rattanathamsakul, N., Eiamsamarng, A., Chirapapaisan, N. et Ngamsombat, C. (2026). **Volumetric brain analysis and associated retinal thinning in autosomal dominant optic atrophy patients.** Neuroimage Rep 6, 100314, doi:[10.1016/j.ynirp.2025.100314](https://doi.org/10.1016/j.ynirp.2025.100314).

---

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

---

Pas de résultat ce trimestre

---

### Syndrome de Barth – *Barth syndrome*

---

AlShaer, D., Al Musaimi, O., Albericio, F. et de la Torre, B. G. (2026). **2025 FDA TIDES (Peptides and Oligonucleotides) Harvest.** Pharmaceuticals (Basel) 19, 244, doi:[10.3390/ph19020244](https://doi.org/10.3390/ph19020244).

Chan, J. Z., Berdeklis, A. N., Liu, M. R., Rahman, F. A., Tomczewski, M. V., Graham, M. Q., Musa, M., Cocco, A. D., Stark, K. D., Quadrilatero, J. et Duncan, R. E. (2026). **Effects of Cannabidiol on TFAZZIN-Deficient B-Lymphoblastoid Cells.** FASEB J 40, e71674, doi:[10.1096/fj.202503384R](https://doi.org/10.1096/fj.202503384R).

Ferreira, C., Pierre, G., Thompson, R. et Vernon, H. (2026). **Barth Syndrome.** In: **GeneReviews**<sup>®</sup>, Éd. Adam, M. P., Bick, S., 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/NBK247162/> [Consulté le avril 2026].

Low, Y. C., McKnight, C. L., Elliott, D. A., Thorburn, D. R. et Frazier, A. E. (2026). **Generation of a pluripotent embryonic stem cell TFAZZIN hESC model (WAE009-A-3H) of Barth syndrome.** Stem Cell Res 93, 103948, doi:[10.1016/j.scr.2026.103948](https://doi.org/10.1016/j.scr.2026.103948).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Matias, C., Snider, P. L., Sierra Potchanant, E. A., Huot, J. R., Raghav, R., Chin, M. T., Conway, S. J. et Brault, J. J. (2026). **Deficient Cardiolipin Remodelling Alters Muscle Fibre Composition and Neuromuscular Connectivity in Barth Syndrome.** *J Cachexia Sarcopenia Muscle* 17, e70246, doi:[10.1002/jcsm.70246](https://doi.org/10.1002/jcsm.70246).

Senoo, N., Sheridan, M. S., Wohlfarter, Y., Primrose, M. T., Tampakakis, E., Keller, M. A. et Claypool, S. M. (2026). **Disturbed mitochondrial maturation in cardiolipin remodeling-deficient cardiomyocytes.** *iScience* 29, 115111, doi:[10.1016/j.isci.2026.115111](https://doi.org/10.1016/j.isci.2026.115111).

Singer, E. S., Smith, J., Lin, R., Morrish, A. M., Lal, S., Irving, C., Casey, C., King, I., Weintraub, R. G. et Bagnall, R. D. (2026). **Case Report: Deletion in the 5' untranslated region of TFAZZIN in a boy with Barth syndrome.** *Front Cardiovasc Med* 13, 1766067, doi:[10.3389/fcvm.2026.1766067](https://doi.org/10.3389/fcvm.2026.1766067).

Stockbridge, N. et Southworth, M. R. (2026). **Letter to the Editor: Battle Won but War Lost for Barth Syndrome Patients.** *Ther Innov Regul Sci*, doi:[10.1007/s43441-026-00951-x](https://doi.org/10.1007/s43441-026-00951-x).

Tahir, A., Imtiaz, E. et Mahato, R. K. (2026). **3D bioprinted myocardium patches for rare Barth syndrome: TAZ mutation correction in cardioblasts.** *Ann Med Surg (Lond)* 88, 1096-1097, doi:[10.1097/MS9.0000000000004456](https://doi.org/10.1097/MS9.0000000000004456).

---

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

---

Lang, S. H., Cottingham, N., Donnelly, C., Risen, S., Muncher, R. M., Brewer, E. D., Saland, J. M., Benchimol, C., Scaglia, F. et Ganesh, J. (2026). **Outcomes of kidney transplantation in three patients with single large-scale mitochondrial DNA deletion syndromes.** *Mol Genet Metab* 147, 109731, doi:[10.1016/j.ymgme.2026.109731](https://doi.org/10.1016/j.ymgme.2026.109731).

Shahab, S. H. et Habib, F. (2026). **CRISPR prime editing of mitochondrial heteroplasmy in rare Kearns-Sayre syndrome: ocular and cardiac synergies.** *Ann Med Surg (Lond)* 88, 1019-1020, doi:[10.1097/MS9.0000000000004366](https://doi.org/10.1097/MS9.0000000000004366).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

---

### Syndrome de Leigh - Leigh syndrome

---

Banerjee, S., Mondal, R., Deb, S., Shome, G., Chakraborty, S., Saha, C., Pandit, A., Sen, G., Bhattacharya, S., Sen, P., Bhattacharya, N. P., Roy, J., Chowdhury, A. et Benito-León, J. (2026). **Integrated molecular and clinical profiling of primary mitochondrial oxidative phosphorylation disorders in an Indian cohort: Insights from genetics, neuroimaging, and machine learning.** *Mitochondrion* 89, 102150, doi:[10.1016/j.mito.2026.102150](https://doi.org/10.1016/j.mito.2026.102150).

Bennett, N., Soto-Muniz, Y., Meng, J. X., Lee, M., Yang, J., Flanigan, W. R., Pico, A. R., Jain, I. H. et Nakamura, K. (2025). **Genetic regulators of neuronal survival across metabolic environments.** *bioRxiv* 2025.12.19.695350, doi:[10.64898/2025.12.19.695350](https://doi.org/10.64898/2025.12.19.695350).

Bowen, A. B., Nwanze, C., Alves, C., Rodan, L., Pinto, A. L., Walker, M. A., Anselm, I. et Pearl, P. L. (2026). **Epilepsy Phenotype and EEG Finding of Rhythmic High-Amplitude Delta With Superimposed Spikes (RHADS) in Succinate Dehydrogenase Deficiency.** *JIMD Rep* 67, e70072, doi:[10.1002/jmd2.70072](https://doi.org/10.1002/jmd2.70072).

Du, S., Long, Q., Zhou, Y., Fu, J., Wu, H., Yang, L., Xie, Y., Ding, Y., Zhang, M., Guo, J., Wang, M., Lin, J., Hu, M., Zhang, Jian, Yao, D., Li, W., Bao, F., Xiang, G., Wu, Y., Huang, Y., Liang, H., Wang, R., Li, H., Chen, B., Li, C., Wang, J., Zhang, Jiwei, Qin, D., Sun, J., Zhu, Y., Sun, F., Wang, W., Lu, G., Chan, W.-Y., Zhao, H., Liu, C. et Liu, X. (2026). **Transplantation of encapsulated mitochondria alleviates dysfunction in mitochondrial and Parkinson's disease models.** *Cell* S0092-8674(26)00230-8, doi:[10.1016/j.cell.2026.02.023](https://doi.org/10.1016/j.cell.2026.02.023).

Faideau, M., Clément, R., Rigollet, S., Benegiamo, G., Cresson, C., Blot, B., Reynaud-Dulaurier, R., Yjjou, S., Aprahamian, F., Durand, S., Delalande, A., Barbier, E. L., Stupar, V., Auwerx, J. et Decressac, M. (2026). **Ultrasound-assisted gene therapy mitigates Leigh syndrome pathology.** *Brain* awag026, doi:[10.1093/brain/awag026](https://doi.org/10.1093/brain/awag026).

Fogle, K. J., Lindley, S. K., Satterfield, S. L., Amsalu, B. A., Figura, J. R., Eicher, S. L., Scherz, L. A. et Palladino, M. J. (2026). **The Citric Acid Cycle Modulates Neurologic Health and Is a Therapeutic Target of Dietary and Genetic Modification in Metabolic Disease.** *Genes (Basel)* 17, 192, doi:[10.3390/genes17020192](https://doi.org/10.3390/genes17020192).

Fouché, B. R., Khumalo, S. G., Koopman, W. J. H. et Venter, M. (2026). **Leigh Syndrome Pathomechanism Involves Region-Specific Innate Immune Activation in Ndufs4 Knockout Mice.** *Cell Mol Neurobiol* 46, 42, doi:[10.1007/s10571-026-01681-2](https://doi.org/10.1007/s10571-026-01681-2).

Gerrie, S. K., Navallas, M., Marie, E., Inarejos Clemente, E., Castle, A. M. R. et Branson, H. M. (2025). **Congenital Inborn Errors of Metabolism: Clinical and Imaging Pearls.** *Radiographics* 45, e240053, doi:[10.1148/rg.240053](https://doi.org/10.1148/rg.240053).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Jin, Z.-Z., Wang, X.-S., Tang, H.-B. et Lv, Y. (2025). **Leigh Syndrome Complicated by Takotsubo Cardiomyopathy: A Case Report and Literature Review.** *Neuro Endocrinol Lett* 46, 417-422.

Khumalo, S. G., Naicker, P., Lindeque, J. Z. et Venter, M. (2026). **SWATH-MS reveals tissue-specific proteomic changes in a Leigh syndrome mouse model.** *Mol Genet Metab* 147, 109715, doi:[10.1016/j.ymgme.2025.109715](https://doi.org/10.1016/j.ymgme.2025.109715).

Kuwabara, T., Tamura, T., Takakura, M., Fujii, T., Ozeki, K. et Akiyama, K. (2026). **Remimazolam-based anesthesia with intraoperative motor evoked potential monitoring in a patient with Leigh syndrome undergoing scoliosis surgery: a case report.** *JA Clin Rep* 12, 19, doi:[10.1186/s40981-026-00851-x](https://doi.org/10.1186/s40981-026-00851-x).

McManus, M. J., Zhu, Y., Alves, C., Kohli, N., Prada-Dacasa, P., Sanchez-Benito, L., Sanz, E., Yee, I., Robinson, L., Sheldon, M., McHugh, W. J., Ranganathan, A., Meng, J., Duncan, N., Grönberg, A., Wallace, D. C., Piel, S., Karlsson, M., Moss, S. J., Webster, L., Hansson, M. J., Elmér, E., Ehinger, J. K., Quintana, A. et Kilbaugh, T. J. (2026). **The succinate prodrug NV354 prevents brain lesions and late-stage motor dysfunction in mitochondrial complex I deficiency.** *iScience* 29, 114717, doi:[10.1016/j.isci.2026.114717](https://doi.org/10.1016/j.isci.2026.114717).

Pinho, B. R., Martins, V., Chacko, A. R., Nogueira, C., Duchon, M. R. et Oliveira, J. M. A. (2026). **Targeting mitochondrial deubiquitinase USP30 to induce mitophagy in heteroplasmic mitochondrial diseases.** *Pharmacol Rep* 78, 519-534, doi:[10.1007/s43440-026-00829-7](https://doi.org/10.1007/s43440-026-00829-7).

Shamriz, O., Bar-On, Z., Yosef, O., Cohen-Daniel, L., Sheer, A., Reuven, O., Salaymeh, W., Saragovi, A., Somech, R., Lev, A., Mor-Shaked, H., Tal, Y., Fattal-Valevski, A., Edvardson, S. et Berger, M. (2025). **NDUFS4, a mitochondrial complex I subunit, is essential for T-cell metabolic fitness and immune function.** *Front Immunol* 16, 1734203, doi:[10.3389/fimmu.2025.1734203](https://doi.org/10.3389/fimmu.2025.1734203).

Shen, L. (2025). **The Path to Precision Medicine in Leigh Syndrome Spectrum: A Four-Decade Chronicle of Genetic Discovery and Targeted Treatment.** *Front Biosci (Schol Ed)* 17, 45427, doi:[10.31083/FBS45427](https://doi.org/10.31083/FBS45427).

Shen, L. (2026). **Generative AI Accelerates Genotype-Phenotype Characterization of a 1600-Case Leigh Syndrome Virtual Cohort from Published Literature.** *Biology (Basel)* 15, 334, doi:[10.3390/biology15040334](https://doi.org/10.3390/biology15040334).

Stephan, A., Graca, F. A., Poudel, S., Fu, Y., Wang, Y.-D., Labelle, M. et Demontis, F. (2026). **Largely Distinct Post-Translational Modifications Differentiate Skeletal Muscle Wasting Caused by Cancer, Dexamethasone and Aging.** *J Cachexia Sarcopenia Muscle* 17, e70220, doi:[10.1002/jcsm.70220](https://doi.org/10.1002/jcsm.70220).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Tada, H., Ichimoto, K., Murayama, K., Goto, T. et Takanashi, J.-I. (2026). **New Neuroimaging Findings in Enoyl-CoA Hydratase Short-Chain 1 (ECHS1) Deficiency.** *Cureus* 18, e102392, doi:[10.7759/cureus.102392](https://doi.org/10.7759/cureus.102392).

Tan, N. B., Gautschi, M., Raum, M., Hock, D. H., Kopajtich, R., Wang, J., Qian, X., Sharma, T., Green, T. E., Nuoffer, J.-M., Bell, K. M., Pospieszny, K., Stait, T., Pike, C., Cao, M., White, S. M., Thorburn, D. R., Brunet, T., Wagner, M., Müller-Felber, W., Zeng, L., Klopstock, T., Schaller, A., Liu, J., Stroud, D. A. et Prokisch, H. (2026). **Bi-allelic variants in NDUFA5 cause a mitochondriopathy with complex I deficiency.** *Am J Hum Genet* S0002-9297(26)00113-8, doi:[10.1016/j.ajhg.2026.03.003](https://doi.org/10.1016/j.ajhg.2026.03.003).

Terburgh, K., Sweeney, N. et Louw, R. (2026). **Energy Metabolism Under Stress: Late-Stage Leigh Syndrome Reveals Profound Cardiometabolic Perturbations in Ndufs4 KO Mice.** *J Inherit Metab Dis* 49, e70142, doi:[10.1002/jimd.70142](https://doi.org/10.1002/jimd.70142).

Vissac, A., Sarda, E., Roux, C.-J., Schiff, M., Bérat, C.-M., Bouchereau, J., Arnoux, J.-B., de Puyraimond, C., Gaschignard, M., Mekdade, T., Cucinotta, U., Mayer, C., Barbier, V., Benoist, J. F., Oualha, M., Capito, C., Chardot, C., Dao, M., Servais, A., Francoz, C., Dehoux, L., Lacaille, F., Desguerre, I., Boddaert, N., de Lonlay, P. et Brassier, A. (2026). **Acute Neurological Complications After Transplantation in Methylmalonic Acidemia: A 35-Patient French Cohort.** *J Inherit Metab Dis* 49, e70165, doi:[10.1002/jimd.70165](https://doi.org/10.1002/jimd.70165).

Yan, H.-M., Quan, Y., Zhou, Y., Jiang, L., Zhang, L.-Y., Wan, Z.-Q. et Xi, H. (2026). **[Decreased plasma citrulline is a biochemical marker in newborn screening for MT-ATP6-associated mitochondrial disease: two case reports and a literature review].** *Zhongguo Dang Dai Er Ke Za Zhi* 28, 250-256, doi:[10.7499/j.issn.1008-8830.2505143](https://doi.org/10.7499/j.issn.1008-8830.2505143).

Yang, F., Yao, R., Chang, G., Hu, J., Feng, B., Wang, L., Hu, F., Huang, Y., Wu, S., Yu, T., Ding, Y. et Wang, X. (2026). **Clinical and genetic spectrum of pediatric mitochondrial disorders in China: insights from a 47-case genetically confirmed cohort.** *Orphanet J Rare Dis* 21, 158, doi:[10.1186/s13023-025-04180-7](https://doi.org/10.1186/s13023-025-04180-7).

Zhou, Y., Li, K., Zhu, R., Ma, X., Ye, X., Mao, M., Li, D., Zeng, X., Chen, Z., Wu, J., Jin, L., Tang, X., Yang, Y., Lyu, J. et Lou, X. (2026). **COG5 deficiency disrupts cellular copper homeostasis and underlies the impaired mitochondrial OXPHOS function.** *PLoS Genet* 22, e1012076, doi:[10.1371/journal.pgen.1012076](https://doi.org/10.1371/journal.pgen.1012076).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Zink, A., Dai, D.-F., Wittich, A., Henke, M.-T., Pedrotti, G., Heiduschka, S., Santamaria, G., Pentimalli, T. M., Brueser, C., Notopoulou, S., Umar, A. R., Zhaivoron, A., Petersilie, L., Jerred, C., Bergmans, J., Neu, L. A., Schumacher, F., Keller-Findeisen, J., Rybak-Wolf, A., Stach, D., Reinshagen, J., Haferkamp, U., Krieg, K., Zaliani, A., Euro, L., Di Donfrancesco, A., Santanatoglia, C., Cappelozza, E., Suarez Cubero, M., Pavez-Giani, M., Bakumenko, O., Meierhofer, D., Foley, A., Morales-Gonzalez, S., Tolle, I., Herebian, D., Bonesso, D., Cecchetto, G., Wong, S. N., Moresco, M., Maresca, A., Decimo, I., De Sanctis, F., Adamo, A., Adjobo-Hermans, M. J. W., Duchi, R., Barandalla, M., Scaglia, M., Perota, A., Galli, C., Kleuser, B., Cyganek, L., Mühlhausen, C., Schlotawa, L., Tiranti, V., Mayatepek, E., Szabo, I., La Morgia, C., Klopstock, T., Carelli, V., Distelmaier, F., Rossi, A., Rajewsky, N., Ullah, G., Jakobs, S., Rose, C. R., Petrakis, S., Edenhofer, F., Koopman, W. J. H., Lisowski, P., Suomalainen, A., Brunetti, D., Del Sol, A., Bottani, E., Pless, O., Schuelke, M. et Prigione, A. (2026). **Pluripotent stem-cell-based screening uncovers sildenafil as a mitochondrial disease therapy.** *Cell* 189, 1656-1679.e42, doi:[10.1016/j.cell.2026.02.008](https://doi.org/10.1016/j.cell.2026.02.008).

---

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

---

Abbas, A. A., Monagel, D. A. et Althubaiti, S. J. (2026). **A Case of Mitochondrial Myopathy, Lactic Acidosis and Sideroblastic Anemia (MLASA Syndrome) and Long QT Interval in a 10-Year-Old Saudi Child.** *Saudi J Med Med Sci* 14, 96-99, doi:[10.4103/sjmms.sjmms\\_716\\_25](https://doi.org/10.4103/sjmms.sjmms_716_25).

Abe, S., Miyagi, M., Saito, W., Tamaki, R., Inoue, T., Hagiwara, K., Inoue, G., Okazaki, K. et Takaso, M. (2026). **Surgical Treatment for Spinal Scoliosis in Patient With Mitochondrial Disease After Heart Transplantation: A Case Report and Literature Review.** *Case Rep Orthop* 2026, 8814759, doi:[10.1155/cro/8814759](https://doi.org/10.1155/cro/8814759).

Acikgoz, N. B., Demir, G. U., Yildiz, Y., Duz, M. B., Sener, N., Alpat, S., Kesici, S., Ertugrul, I., Yalnizoglu, D., Utine, G. E. et Kiper, P. O. S. (2026). **Homozygous MGME1 Variant in Turkish Siblings: First Reported Case With Successful Heart Transplantation, Expanding the Clinical Spectrum of MGME1 -Related Mitochondrial Disease.** *Am J Med Genet A* 200, 1128-1133, doi:[10.1002/ajmga.70049](https://doi.org/10.1002/ajmga.70049).

Adrian, A. E., Boucher, N. J., McVary, K. T., DeFranco, D. B. et Ricke, W. A. (2026). **Mitochondrial dysfunction in urologic disease.** *Prostate Cancer Prostatic Dis*, doi:[10.1038/s41391-026-01097-5](https://doi.org/10.1038/s41391-026-01097-5).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Ahmadian, S., Lindsey, P. J., Ummelen, M., Hopman, A., van Zandvoort, M. A. M. J., Smeets, H. J. M. et van Tienen, F. H. J. (2026). **Fusion Between Control Mesoangioblasts and mtDNA-Mutant Myotubes Preserves Myotube Morphology and Mitochondrial Network Organization.** *Int J Mol Sci* 27, 1357, doi:[10.3390/ijms27031357](https://doi.org/10.3390/ijms27031357).

Algariri, E. S., Nordin, F., Ng, M. H., Mohd Idris, I., Abdul Karim, N., Tye, G. J. et Wan Kamarul Zaman, W. S. (2026). **Mesenchymal Stromal Cells and Extracellular Vesicles: A Novel Therapeutic Paradigm for Mitochondrial Dysfunctions.** *Int J Mol Sci* 27, 1981, doi:[10.3390/ijms27041981](https://doi.org/10.3390/ijms27041981).

Alreshidi, N. F., Al-Dhelaan, R. A., Yousuf, A., Almuraikhi, N., Alanazi, H. G., Alsufayan, T., Alsahly, M. B. et Alazzam, M. B. (2026). **Evaluation of metformin's effect on 5-fluorouracil-induced cardiotoxicity through cellular protection.** *Daru* 34, 11, doi:[10.1007/s40199-025-00589-4](https://doi.org/10.1007/s40199-025-00589-4).

Alshamrani, F. J., Alajmi, M. S., Almuslim, N. I., Alsubaie, M. M., Fardan, G. M. et Alabdali, M. M. (2026). **Identifying NDUF A12 mutation in a Saudi family: An unusual presentation of mitochondrial Complex I deficiency mimicking as idiopathic intracranial hypertension in a patient with papilledema and visual loss.** *J Family Community Med* 33, 47-52, doi:[10.4103/jfcm.jfcm\\_322\\_25](https://doi.org/10.4103/jfcm.jfcm_322_25).

Amalnath, D., Saibaba, J., Wadwekar, V., Nagarajan, K. et Senthilvelan, S. (2026). **Mitochondrial DNA Maintenance Defects: Clinical, Imaging, and Genetic Spectrum of Four Patients from a Single Tertiary Care Centre.** *Ann Indian Acad Neurol*, doi:[10.4103/aian.aian\\_843\\_25](https://doi.org/10.4103/aian.aian_843_25).

Asfour, M. A., Nedimyer Horner, J., Gupta, K., Silva, G., Chandra, T. et Rossi, G. (2025). **Mitochondrial Disorder in a Child With Brainstem Lesions Mimicking Thiamine Deficiency.** *Cureus* 17, e98909, doi:[10.7759/cureus.98909](https://doi.org/10.7759/cureus.98909).

Ball, M., Baker, N., Lim, S. C., Casauria, S., Lunke, S., Compton, A. G., Thorburn, D. R., Christodoulou, J. et Stark, Z. (2026). **Mainstreaming genomic testing for mitochondrial disease in Australia.** *Eur J Hum Genet*, doi:[10.1038/s41431-026-02053-6](https://doi.org/10.1038/s41431-026-02053-6).

Banerjee, S., Mondal, R., Deb, S., Shome, G., Chakraborty, S., Saha, C., Pandit, A., Sen, G., Bhattacharya, S., Sen, P., Bhattacharya, N. P., Roy, J., Chowdhury, A. et Benito-León, J. (2026). **Integrated molecular and clinical profiling of primary mitochondrial oxidative phosphorylation disorders in an Indian cohort: Insights from genetics, neuroimaging, and machine learning.** *Mitochondrion* 89, 102150, doi:[10.1016/j.mito.2026.102150](https://doi.org/10.1016/j.mito.2026.102150).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Barca, E., Jacoby, N., Naini, A., Miller, M. L., Emmanuele, V., Winfree, C. J., Tadesse, S., Tanji, K. et Hirano, M. (2026). **Rhabdomyolysis due to mtDNA pathogenic variants: Report of a subject with a novel MT-CO3 variant and review of the literature.** *Mol Genet Metab* 147, 109767, doi:[10.1016/j.ymgme.2026.109767](https://doi.org/10.1016/j.ymgme.2026.109767).

Barriocanal-Casado, E., Stopka, S. A., Pesini, A., Aristizabal-Henao, J. J., Karmacharya, S., Van Cura, D., Zhang, R., Nickerson, K. R., Grover, K., Zavidij, O., Wessel, S. R., Narain, N. R., Modur, V., Gesta, S., Kiebish, M. A. et Quinzii, C. M. (2026). **Spatial mapping of CoQ10 repletion by BPM31510 in a genetic mouse model (Coq4F147C) of coenzyme Q deficiency.** *J Lipid Res* 67, 100987, doi:[10.1016/j.jlr.2026.100987](https://doi.org/10.1016/j.jlr.2026.100987).

Beltrán-Camacho, L., Vallejo-Mudarra, M., Pozuelo-Sánchez, I., García-Caballero, C., Rojas-Torres, M., Lazaro, I., Sala-Vila, A., Valentín-Aragón, L., Caballero-Villarraso, J., Opazo-Rios, L., Leiva-Cepas, F., Mas-Fontao, S., Egido, J., Villalba, J. M., Durán-Ruiz, M. C. et Moreno, J. A. (2026). **Loss of energy homeostasis contributes to hepatic damage development in sickle cell disease.** *Mol Metab* 106, 102348, doi:[10.1016/j.molmet.2026.102348](https://doi.org/10.1016/j.molmet.2026.102348).

Biswas, S. R., Tomsick, P. L., Pickrell, A. M. et Morton, P. D. (2026). **Linking neurogenesis, oligodendrogenesis, and myelination defects to neurodevelopmental disruption in primary mitochondrial disorders.** *FEBS Lett*, doi:[10.1002/1873-3468.70335](https://doi.org/10.1002/1873-3468.70335).

Borden, C., Chaudhry, I., Maditz, R., Mazzola, S., Kent, R. S., Tomaszewski, K. et Wang, X. (2026). **Mitochondrial DNA Analysis Should Be Considered in the Genetic Assessment of Focal Segmental Glomerulosclerosis or Unexplained Chronic Kidney Disease: A Case Report.** *Kidney Med* 8, 101233, doi:[10.1016/j.xkme.2025.101233](https://doi.org/10.1016/j.xkme.2025.101233).

Braun, N., Patil, M. K., Rohan, T. Z., Milosavljevic, S., Sakunchotpanit, G., Iriarte, C. et Nambudiri, V. E. (2026). **Pediatric Conditions for Which Skin Biopsies of Clinically Normal Skin Have Diagnostic Yield: A Review for the Pediatric Dermatologist.** *Pediatr Dermatol*, doi:[10.1111/pde.70154](https://doi.org/10.1111/pde.70154).

Chen, B. et Mayer, J. U. (2026). **Emerging frontiers in the mitochondrial regulation of dendritic cell biology.** *Redox Biol* 90, 104032, doi:[10.1016/j.redox.2026.104032](https://doi.org/10.1016/j.redox.2026.104032).

Chetu, N., Onsod, P., Rerkamnuaychoke, B., Siriboonpiputtana, T. et Chareonsirisuthigul, T. (2026). **Evaluation of MASSARRAY technique in detecting mitochondrial disease mutations.** *Clin Chim Acta* 583, 120820, doi:[10.1016/j.cca.2026.120820](https://doi.org/10.1016/j.cca.2026.120820).

Costa-Borges, N. et Wells, D. (2026). **Revisiting the promise and pitfalls of mitochondrial replacement therapies.** *Hum Reprod* 41, 479-482, doi:[10.1093/humrep/deag020](https://doi.org/10.1093/humrep/deag020).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Courtois, S., Angelini, C., Preud'homme, J., Le Quang, M., Dumon, E., Dulucq, S., Aladjidi, N., Kannengiesser, C., Barat, P., Naudion, S., Espil, C., Martin-Negrier, M.-L. et Trimouille, A. (2026). **Phenotypic description and functional characterization of the mitochondrial disease associated with the SFXN4 gene.** *Mitochondrion* 88, 102136, doi:[10.1016/j.mito.2026.102136](https://doi.org/10.1016/j.mito.2026.102136).

Cui, J., Li, M., Wang, L., Li, F., Ruan, K., Lv, M. et Shi, Y. (2026). **Molecular mechanism underlying the specific RNA recognition of mitochondrial helicase DDX28 and its critical role in mitoribosomal biogenesis.** *Structure* S0969-2126(26)00051-1, doi:[10.1016/j.str.2026.02.009](https://doi.org/10.1016/j.str.2026.02.009).

Curtabbi, A., Jaroszewicz, S. N., Sanz-Cortés, R., Acín-Pérez, R., Prymidis, D., Cherevatenko, M., Martínez-de-Mena, R., Esteban-Amo, M. J., de la Fuente, M. A., Frezza, C. et Enríquez, J. A. (2026). **Ectopic expression of cytosolic DHODH uncouples de novo pyrimidine biosynthesis from mitochondrial electron transport.** *Nat Metab* 8, 454-466, doi:[10.1038/s42255-026-01454-7](https://doi.org/10.1038/s42255-026-01454-7).

D'Amato, G., Gentile, M., Carella, R., Giannini, A., Faienza, M. F. et Tummolo, A. (2026). **The Ketogenic Diet in the Neonatal Intensive Care Setting: The Case of a Preterm Newborn With Mitochondrial DNA Depletion Syndrome Type 13 (MTDPS13).** *Case Rep Genet* 2026, 6492770, doi:[10.1155/crig/6492770](https://doi.org/10.1155/crig/6492770).

Damiano, M., Lambrecq, V., Nguyen-Michel, V.-H., Marois, C., Rucheton, B., Mochel, F., Gourfinkel-An, I., Demeret, S. et Navarro, V. (2026). **EEG, clinical, and MRI features of status epilepticus associated with mitochondrial diseases.** *J Neurol* 273, 160, doi:[10.1007/s00415-026-13681-9](https://doi.org/10.1007/s00415-026-13681-9).

Dicorato, M. M., De Sario, G., Carella, M. C., Guaricci, A. I., Ciccone, M. M., Forleo, C., D'Amato, G. et Faienza, M. F. (2026). **Genetic Mutations Underlying Growth Impairment and Cardiomyopathies in Children: Molecular Mechanisms, Clinical Implications and Targeted Therapies.** *Genes (Basel)* 17, 355, doi:[10.3390/genes17030355](https://doi.org/10.3390/genes17030355).

Digitale Selvaggio, L., Allosso, F., Errico, M., Grande, G., Yousaf, M., Torella, A., Nigro, V., Pasquali, D., et Telethon Undiagnosed Diseases Program Study Group (2026). **RTN4IP1 mutation and endocrine failure: clinical features and possible benefits of coenzyme Q10.** *Endocr Connect* 15, e250768, doi:[10.1530/EC-25-0768](https://doi.org/10.1530/EC-25-0768).

Dille, Y., Rampakakis, E., Aubert, G., Dassi, C., Mannherz, W., Berrahmoune, S., Srour, M., Buhas, D., Agarwal, S. et Myers, K. A. (2026). **Short telomeres in mitochondrial DNA depletion disorders.** *Mitochondrion* 88, 102131, doi:[10.1016/j.mito.2026.102131](https://doi.org/10.1016/j.mito.2026.102131).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Dobešová, D., Prídavok, M., Brumarová, R., Kvasnička, A., Pisklák, B., Ivanovová, E., Brennerová, K., Šaligová, J., Potočňáková, L., Drobnáková, S., Potočňáková, J. et Friedecký, D. (2025). **Comprehensive metabolomic/lipidomic characterization of patients with mitochondrial ATP synthase, short-chain acyl-CoA dehydrogenase and combined variant deficiencies.** *Heliyon* 11, e42797, doi:[10.1016/j.heliyon.2025.e42797](https://doi.org/10.1016/j.heliyon.2025.e42797).

Doudin, A. A. A., Omran, W. N. I., Alkomi, S. M., Rjoub, A., Shoubaki, H. et Abu Mufreh, S. T. (2026). **A case report of combined oxidative phosphorylation deficiency 35 (COXPD35) in Palestine caused by novel compound heterozygous TRIT1 variants.** *Medicine (Baltimore)* 105, e47885, doi:[10.1097/MD.00000000000047885](https://doi.org/10.1097/MD.00000000000047885).

Du, S., Long, Q., Zhou, Y., Fu, J., Wu, H., Yang, L., Xie, Y., Ding, Y., Zhang, M., Guo, J., Wang, M., Lin, J., Hu, M., Zhang, Jian, Yao, D., Li, W., Bao, F., Xiang, G., Wu, Y., Huang, Y., Liang, H., Wang, R., Li, H., Chen, B., Li, C., Wang, J., Zhang, Jiwei, Qin, D., Sun, J., Zhu, Y., Sun, F., Wang, W., Lu, G., Chan, W.-Y., Zhao, H., Liu, C. et Liu, X. (2026). **Transplantation of encapsulated mitochondria alleviates dysfunction in mitochondrial and Parkinson's disease models.** *Cell* S0092-8674(26)00230-8, doi:[10.1016/j.cell.2026.02.023](https://doi.org/10.1016/j.cell.2026.02.023).

Durmus, H., Gedikbaşı, A., Ceylaner, S., Çakar, A., Mergen, S., Kıyan, E. et Parman, Y. G. (2026). **Late-onset TK2 deficiency in adults: Long-term clinical outcomes of deoxynucleoside therapy.** *Mitochondrion* 89, 102138, doi:[10.1016/j.mito.2026.102138](https://doi.org/10.1016/j.mito.2026.102138).

Fernández, B., Pérez-Moreno, G., Martínez-Arribas, B., Vidal, A. E., Ruiz-Pérez, L. M. et González-Pacanowska, D. (2026). **DCTPP1 orchestrates dCTP pool dynamics and mtDNA stability in quiescent cells.** *Cell Death Dis*, doi:[10.1038/s41419-026-08632-1](https://doi.org/10.1038/s41419-026-08632-1).

Finsterer, J. (2025). **Mitochondrial Disorders Should Be Considered as a Differential Diagnosis of Thiamine Deficiency.** *HCA Healthc J Med* 6, 499-500, doi:[10.36518/2689-0216.2014](https://doi.org/10.36518/2689-0216.2014).

Finsterer, J. (2026a). **Before Diagnosing Encephalomyeloradiculoneuropathy, Infectious and Immune Encephalitis, Lymphomas, and Mitochondrial Disorders Must Be Ruled Out.** *J Child Neurol* 8830738261420613, doi:[10.1177/08830738261420613](https://doi.org/10.1177/08830738261420613).

Finsterer, J. (2026b). **Mitochondrial disorders with renal involvement and kidney transplantation require genotypic and phenotypic characterisation.** *Pediatr Nephrol*, doi:[10.1007/s00467-026-07178-w](https://doi.org/10.1007/s00467-026-07178-w).

Fontaine, F., Pénicaud, R. et Allouche, S. (2026). **Mitochondrial Dysfunctions in Human Primary Coenzyme Q10 Deficiencies.** *Biomolecules* 16, 302, doi:[10.3390/biom16020302](https://doi.org/10.3390/biom16020302).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Gab-Allah, M. A., Hwang, H., Kim, Mingyu, Tran, N.-T., Kim, B. J., Kim, Minyoung, Han, J. H., Kim, Y., Choi, B. Y. et Kim, J. (2026). **Detection and localization of single-nucleotide mutations in synthetic oligonucleotides by ultra-high-performance liquid chromatography coupled with high-resolution tandem mass spectrometry.** J Pharm Biomed Anal 273, 117384, doi:[10.1016/j.jpba.2026.117384](https://doi.org/10.1016/j.jpba.2026.117384).

Gabandé-Rodríguez, E., Gómez de Las Heras, M. M., Ramírez-Ruiz de Erenchun, P., Simó, C., García-Cañas, V., Inohara, N., Berenguer-López, I., Enríquez-Zarralanga, V., Fernández-Almeida, Á., Oller, J., Soto-Heredero, G., Carrasco, E., Delgado-Pulido, S., Escrig-Larena, J. I., Francos-Quijorna, I., Justo-Méndez, R., Aranda, J. F., Poulton, J., Lechuga-Vieco, A. V., Enríquez, J. A., Núñez, G. et Mittelbrunn, M. (2026a). **Butyrate extends health and lifespan in mice with mitochondrial deficiency.** bioRxiv 2026.01.13.699287, doi:[10.64898/2026.01.13.699287](https://doi.org/10.64898/2026.01.13.699287).

Gabandé-Rodríguez, E., Gómez de Las Heras, M. M., Ramírez-Ruiz de Erenchun, P., Simó, C., García-Cañas, V., Inohara, N., Berenguer-López, I., Enríquez-Zarralanga, V., Fernández-Almeida, Á., Oller, J., Soto-Heredero, G., Carrasco, E., Vázquez-Muñoz, C., Delgado-Pulido, S., Escrig-Larena, J. I., Francos-Quijorna, I., Justo-Méndez, R., Aranda, J. F., Poulton, J., Lechuga-Vieco, A. V., Enríquez, J. A., Núñez, G. et Mittelbrunn, M. (2026b). **Butyrate extends health and lifespan in mice with mitochondrial deficiency.** Nat Commun, doi:[10.1038/s41467-026-70547-4](https://doi.org/10.1038/s41467-026-70547-4).

Garrido-Moraga, R., Serrano-Lorenzo, P., Esteban-Amo, M. J., Bellusci, M., de la Fuente, M. Á., Arenas, J., González-Quintana, A., Ugalde, C., Simarro, M. et Martín, M. A. (2026). **Impact of compound heterozygous SDHA variants on mitochondrial function in pediatric with neurological disease.** Mitochondrion 89, 102149, doi:[10.1016/j.mito.2026.102149](https://doi.org/10.1016/j.mito.2026.102149).

Giovagnoli, A. R., Lamperti, C., Mihai, I. R. et Catania, A. (2026). **Expanding the cognitive spectrum of mitochondrial diseases.** Neurol Sci 47, 228, doi:[10.1007/s10072-026-08827-6](https://doi.org/10.1007/s10072-026-08827-6).

Gong, X., Liu, Y. et Liang, H. (2026). **Case Report: Sengers syndrome caused by a novel 7.6 kb AGK deletion misdiagnosed as isolated congenital cataract.** Front Pediatr 14, 1714952, doi:[10.3389/fped.2026.1714952](https://doi.org/10.3389/fped.2026.1714952).

Haham Zarbib, Y., Huri Ohev-Shalom, S., Lyskov, S. K., Mazor, Y., Anekstein-Spigel, M., Shalva, N., Spiegel, R., Staretz-Chacham, O., Manor, J., Saada, A., Rock, R., Anikster, Y. et Yardeni, T. (2025). **Bioenergetic Signatures of DLD Deficiency: Dissecting PDHc- and  $\alpha$ -KGDHc-Linked Defects.** Antioxidants (Basel) 15, 19, doi:[10.3390/antiox15010019](https://doi.org/10.3390/antiox15010019).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Han, S., Jang, Y., Joo, H. J., Ka, H. I., Mun, S. H., Nguyen, H.-A., Park, D., Jung, Y., Ko, D., Sohn, B. R., Sonn, S. K., Oh, G. T., Choi, Y.-C., Park, S.-Y., Kim, S.-E. et Yang, Y. (2026). **CTRP1 regulates skeletal muscle differentiation through quality control of mitochondrial dynamics and function.** *Mol Ther* 34, 2427-2445, doi:[10.1016/j.ymthe.2025.12.063](https://doi.org/10.1016/j.ymthe.2025.12.063).

Hautakangas, M.-R., Niskanen, T., Vieira, P., Helander, H., Portaankorva, A. M., Rantala, H., Hinttala, R. et Uusimaa, J. (2026). **Muscle biopsy and mitochondrial disease criteria as diagnostic tools for paediatric patients presenting with neuromuscular phenotypes: highlighting the role of secondary mitochondrial dysfunction.** *Neuromuscul Disord* 61, 106370, doi:[10.1016/j.nmd.2026.106370](https://doi.org/10.1016/j.nmd.2026.106370).

Heath, O., Del Caño-Ochoa, F., Baris, S., Carozzo, R., Coman, D., Distelmaier, F., Ellaway, C., Feichtinger, R. G., Finocchi, A., Guerrero-Castillo, S., Halligan, R., Hannibal, I., Kritzer, A., Lichter-Konecki, U., Merkevicius, K., Panis, B., Pitceathly, R. D. S., Pizzamiglio, C., Iwanicka-Pronicka, K., Rahman, S., Seltzer, L., Siepermann, M., Tal, G., Wevers, R. A., Ziętkiewicz, S., Ramón-Maiques, S., Mayr, J. A. et Wortmann, S. B. (2026). **From genotype to outcome: Zygosity-specific insights in 63 cases of CLPB-related mitochondrial disease.** *Mol Genet Metab* 147, 109752, doi:[10.1016/j.ymgme.2026.109752](https://doi.org/10.1016/j.ymgme.2026.109752).

Hoang, D., Pestronk, A. et Kafaie, J. (2026). **Progressive Myopathy and Respiratory Failure in a 7-Year-Old Boy With m.3251A>G MT-TL1 Mutation.** *J Clin Neuromuscul Dis* 27, 89-95, doi:[10.1097/CND.0000000000000547](https://doi.org/10.1097/CND.0000000000000547).

Horne, J. T., Allen, N. E., Paul, S. S., Walker, J. et Sue, C. (2026). **MITO-VATION: Feasibility of a technology-supported structured home exercise program in Mitochondrial Disease.** *PLOS Digit Health* 5, e0001257, doi:[10.1371/journal.pdig.0001257](https://doi.org/10.1371/journal.pdig.0001257).

Hu, X., Wu, Y., Chen, Q., Chen, Z. et Li, Q. (2026). **Insights into the cell type-specific susceptibility of kidney cells to ferroptosis.** *Drug Discov Today* 31, 104598, doi:[10.1016/j.drudis.2026.104598](https://doi.org/10.1016/j.drudis.2026.104598).

Hu, Y., Chen, H., Ding, C., Zhang, S., Chen, Q., Sun, H. et Yang, Q. (2026). **Fumarate loss destabilizes mitochondria and activates cGAS-STING in OLP.** *Biomed Pharmacother* 197, 119127, doi:[10.1016/j.biopha.2026.119127](https://doi.org/10.1016/j.biopha.2026.119127).

Jeejo, M., Mathew, M., Mohanakumar, K. P. et Rajamma, U. (2026). **Mitochondrial dysfunction and mitochondrial unfolded protein response (UPRmt): unravelling their roles in autism spectrum disorder pathogenesis.** *Mitochondrion* 89, 102148, doi:[10.1016/j.mito.2026.102148](https://doi.org/10.1016/j.mito.2026.102148).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Jeenkeawpiam, K., Srifa, P., Nokchan, N., Khongcharoen, N., Binkasem, A. et Sangkhathat, S. (2026). **MitoGEx: An Integrated Platform for Streamlined Human Mitochondrial Genome Analysis**. *Genes (Basel)* 17, 338, doi:[10.3390/genes17030338](https://doi.org/10.3390/genes17030338).

Jiménez-Sánchez, L., Ruiz-López, P., González-García, P., Purhonen, J., Martínez-Gálvez, J. M., López-Herrador, S., Corral-Sarasa, J., Díaz-Casado, M. E., Venegas, C., Santos-Pérez, I., Olivieri, E., Rojas, A. L. et López, L. C. (2026). **N-Acetylglucosamine Selectively Attenuates Neuroinflammation in a Mouse Model of Mitochondrial Dysfunction**. *Acta Physiol (Oxf)* 242, e70179, doi:[10.1111/apha.70179](https://doi.org/10.1111/apha.70179).

Khan, F. A., Kirmani, S., Memon, M. K. Y. et Akhtar, S. (2025). **Risk of sudden cardiac death due to inorganic pyrophosphate A2 deficiency in a Pakistani infant: Diagnostic and management challenges in low-resource settings**. *Ann Pediatr Cardiol* 18, 517-520, doi:[10.4103/apc.apc.152.25](https://doi.org/10.4103/apc.apc.152.25).

Kilich, G., Jadhav, T., Maurer, K., Breen, C., Jammihal, T., Schindewolf, E., Ganetzky, R. D., Vanderver, A., Skraban, C., Rajagopalan, R. et Sullivan, K. E. (2026). **Novel Biallelic LIG3 Mutations Causing Lethal Phenotype With Immunodeficiency**. *Am J Med Genet A*, doi:[10.1002/ajmg.a.70104](https://doi.org/10.1002/ajmg.a.70104).

Kim, T. Y., Jang, E.-H., Bae, Y.-H., Hur, E.-M. et Lee, B. D. (2026). **LRRK2 controls COX assembly through regulation of redox status of mitochondrial copper chaperones**. *Redox Biol* 90, 104061, doi:[10.1016/j.redox.2026.104061](https://doi.org/10.1016/j.redox.2026.104061).

Kozul, K.-L., AlAsmari, A., Alharby, E., Zakzouk, R., Yan, Y., Mushiba, A., Alhamad, A., Harrelson, E., Ayach, M., Cho, K., Zahid, H., Vitorino, F. D. L., Searfoss, R., Liu, X., Saleh, M. A., Latif, M., Wei, L., Aldawood, A., Alsuhaibani, L., Bafail, M. A., Menezes, T., Samman, M., Pletcher, H., Sandokji, I., Borhan, W., Locketto, T., Alamri, A., Mudayfin, W., Syed, M., Shriver, L., Garcia, B., Faqeih, E., Patti, G., Niemi, N. et Almontashiri, N. (2026). **Recessive PPTC7 deficiency triggers excessive mitophagy to cause a severe inborn error of metabolism with hypomyelinating leukodystrophy**. *Res Sq* rs.3.rs-8815446, doi:[10.21203/rs.3.rs-8815446/v1](https://doi.org/10.21203/rs.3.rs-8815446/v1).

Krupa-Nurcek, S., Semań, T., Szczupak, M., Kobak, J., Mędrzycka-Dąbrowska, W. et Widenka, K. (2026). **Mechanisms Linking Oxidative Stress and Sarcopenia in Cardiovascular Diseases: A Scoping Review**. *Antioxidants (Basel)* 15, 184, doi:[10.3390/antiox15020184](https://doi.org/10.3390/antiox15020184).

Kural, I., M Mombeek, L. M. et Wilson, D. M. (2026). **Role of mitochondria in neuronal function and survival in the enteric and central nervous systems**. *Cell Mol Life Sci* 83, 129, doi:[10.1007/s00018-025-06053-5](https://doi.org/10.1007/s00018-025-06053-5).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Kwan, J. Y., Lantz, C. I., Korobeynikov, V. A., Snyder, A., Huang, X., Haselhuhn, T., Dore, K. N., Madruga, A., Danielian, L. E., Schindler, A. B., Chia, R., Rasheed, M., Crook, J., Szabo, M., Portley, M., Sherer, C. M., King, M. C., Huang, T.-H., Kosa, P., Bielekova, B., Ward, M. E., Grunseich, C., Shneider, N. A., Traynor, B. J. et Narendra, D. P. (2026). **Clinical and biochemical characterization of amyotrophic lateral sclerosis in a CHCHD10 R15L family.** *Brain* awag115, doi:[10.1093/brain/awag115](https://doi.org/10.1093/brain/awag115).

Lang, S. H., Cottingham, N., Donnelly, C., Risen, S., Muncher, R. M., Brewer, E. D., Saland, J. M., Benchimol, C., Scaglia, F. et Ganesh, J. (2026). **Outcomes of kidney transplantation in three patients with single large-scale mitochondrial DNA deletion syndromes.** *Mol Genet Metab* 147, 109731, doi:[10.1016/j.ymgme.2026.109731](https://doi.org/10.1016/j.ymgme.2026.109731).

Leeuwenberg, K. E., IntHout, J., Groothuis, J. T., Cup, E., Smeitink, J., Mul, K. et Janssen, M. C. H. (2026). **The goal attainment scale in primary mitochondrial disease: Construct validity and lessons learned from a randomized controlled trial.** *Mol Genet Metab* 148, 109875, doi:[10.1016/j.ymgme.2026.109875](https://doi.org/10.1016/j.ymgme.2026.109875).

Lewis, A., Forti, R. M., Ko, T. S., Elmér, E., McManus, M. J., Yodh, A. G., Kilbaugh, T. J. et Baker, W. B. (2026). **Optical and Microdialysis Monitoring of Succinate Prodrug Treatment in a Rotenone-Induced Model of Mitochondrial Dysfunction in Swine.** *Metabolites* 16, 65, doi:[10.3390/metabo16010065](https://doi.org/10.3390/metabo16010065).

Li, H., Ai, C., Jin, X., Wang, J., Yu, J., Gao, Y., Wallace, D. C. et Guan, M.-X. (2026). **Optic neuropathy arising from the synergy between YARS2 and mitochondrial COX1 mutations.** *J Genet Genomics* S1673-8527(26)00047-0, doi:[10.1016/j.jgg.2026.02.003](https://doi.org/10.1016/j.jgg.2026.02.003).

Liu, C. C., Kurade, M., Monzel, A. S., Kelly, C., Juster, R.-P., Trumpff, C., Hirano, M. et Picard, M. (2026). **Immune Cell Mitochondrial Phenotypes Are Largely Preserved in Mitochondrial Diseases and Do Not Reflect Disease Severity.** *Neurol Genet* 12, e200343, doi:[10.1212/NXG.0000000000200343](https://doi.org/10.1212/NXG.0000000000200343).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Lopriore, P., Ünlütürk, Z., Klopstock, T., Karaa, A., Rouzier, C., Domínguez-González, C., Lamperti, C., Mancuso, M., Twinkle-Related Disorders International Consortium for Trial Readiness (TReDIC), Cecchi, G., Montano, V., Siciliano, G., Nicoletta, V., Maioli, M., Primiano, G., Servidei, S., La Morgia, C., Carelli, V., Valentino, M. L., Caporali, L., Arena, I. G., Musumeci, O., Lopergolo, D., Malandrini, A., Gallus, G. N., Filosto, M., Bello, L., Pegoraro, E., Comi, G. P., Magri, F., Ronchi, D., Di Fonzo, A., Percetti, M., Azzimonti, M., Büchner, B., Prokisch, H., Bermejo-Guerrero, L., Procaccio, V., Gaignard, P., Echaniz-Laguna, A., Schiff, M., Rötig, A., Toutain, A., Paquis-Flucklinger, V., Morel, G., Robin, S., Nadaj-Pakleza, A., Chanson, J.-B., Chaussenot, A., Ait-El-Mkadem Saadi, S., Trimouille, A., Tranchant, C., Salort-Campana, E., Bieth, E., Sacconi, S., Duval, F., Restrepo Vera, J. L., Molnar, M. J., Vissing, J., Haas, R., Larson, A., Enns, G. M., Parikh, S., Goldstein, A. et Hirano, M. (2026). **Clinical and Genotypic Spectrum of Twinkle-Related Disorders: Insights From a Multinational Cohort Study.** *Neurology* 106, e214401, doi:[10.1212/WNL.0000000000214401](https://doi.org/10.1212/WNL.0000000000214401).

Low, Y. C., McKnight, C. L., Stojanovski, D., Thorburn, D. R. et Frazier, A. E. (2026). **Generation of a pluripotent human AGK knockout embryonic stem cell model (WAE009-A-3C) of Sengers syndrome.** *Stem Cell Res* 90, 103895, doi:[10.1016/j.scr.2025.103895](https://doi.org/10.1016/j.scr.2025.103895).

Luan, Z. et Liang, Z. (2025). **Diagnostic challenges in mitochondrial encephalomyopathy with m.10158T>C mutation: a case report and literature review.** *Front Integr Neurosci* 19, 1709380, doi:[10.3389/fnint.2025.1709380](https://doi.org/10.3389/fnint.2025.1709380).

Makwikwi, T., Schoonen, M., Smuts, I. et van der Westhuizen, F. H. (2026). **Bridging gaps in mitochondrial disease diagnosis: the role of advanced biomarker discovery.** *J Mol Med (Berl)* 104, 36, doi:[10.1007/s00109-026-02646-0](https://doi.org/10.1007/s00109-026-02646-0).

Mandia, D., Metodiev, M. D., Benoist, J.-F., Gaignard, P., Ruzzenente, B., Zuchner, S., Beijer, D., Fernandez-Eulate, G., Rötig, A., Lamari, F., Rucheton, B., Rouzier, C., Navarro, L. R., Saadi, S. A.-E.-M., Cintas, P., Maquet, J., Masingue, M., Shor, N. et Nadjar, Y. (2026). **MRPS Genes Causing Leukoencephalopathy With Profound Cerebral Folate Deficiency in Adults.** *J Inher Metab Dis* 49, e70139, doi:[10.1002/jimd.70139](https://doi.org/10.1002/jimd.70139).

Marchais, M., Pennisi, A., Pierga, A., Lepelley, A., Cagnard, N., Bole, C., Nitschke, P., Hamici, M., Rieux-Laucat, F., Schiff, M., Munnich, A. et Rötig, A. (2026). **No Correlation Between Interferon Signaling and Cytosolic Mitochondrial DNA/RNA Leakage in Cultured Skin Fibroblasts of Patients With Mitochondrial Diseases.** *Eur J Immunol* 56, e70176, doi:[10.1002/eji.70176](https://doi.org/10.1002/eji.70176).



## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Martín-Jimenez, P., Bermejo-Guerrero, L., Ochoa, L. E., Navarro-Riquelme, M., Garrido-Moraga, R., Hernández-Lain, A., Hernández-Voth, A., González Quintana, A., Bermejo-Moriñigo, A., González-Méndez, V., Martín-Arriscado Arroba, C., Smirnov, D., Konstantinovskiy, N., Arenas, J., Martín, M. Á., Blázquez, A. et Domínguez-González, C. (2026). **Exploring Outcome Measures for Mitochondrial Myopathies; Insights From a Longitudinal Study on TK2 Deficiency.** *J Inherit Metab Dis* 49, e70147, doi:[10.1002/jimd.70147](https://doi.org/10.1002/jimd.70147).

McStay, G. P. (2026). **Mitochondrial complex assembly in epilepsy of primary mitochondrial disease origin.** *Seizure* S1059-1311(26)00050-6, doi:[10.1016/j.seizure.2026.02.016](https://doi.org/10.1016/j.seizure.2026.02.016).

Meshrkey, F., Scheulin, K. M., Saikia, B., Stabach, J., Rao, R. R., West, F. D. et Iyer, S. (2026). **Impaired mitochondrial morphology and respiratory dysfunction in human induced pluripotent stem cells with mitochondrial tRNA mutations (m.3243A>G and m.14739G>A).** *Orphanet J Rare Dis* 21, 73, doi:[10.1186/s13023-026-04201-z](https://doi.org/10.1186/s13023-026-04201-z).

Messina, M., Ganetzky, R., Ferreira, C. R., Blau, N. et Rahman, S. (2026). **Clinical and biochemical footprints of primary mitochondrial disorders: proposed nosology.** *Mol Genet Metab* 147, 109704, doi:[10.1016/j.ymgme.2025.109704](https://doi.org/10.1016/j.ymgme.2025.109704).

**Mitochondrial diseases** (2026). 44, 38, doi:[10.1038/s41587-025-02973-6](https://doi.org/10.1038/s41587-025-02973-6).

Mohammadi, P., Basovic, L. et McGraw, C. M. (2026). **Super-Refractory Status Epilepticus (SRSE) in a Patient With Compound Heterozygous OPA1 Variants: Case Report and Literature Review.** *Ann Clin Transl Neurol*, doi:[10.1002/acn3.70287](https://doi.org/10.1002/acn3.70287).

Mohr, J., Frederiksen, A. L., Duno, M., Hermann, A. P., Juul, T. M. et Nielsen, S. R. (2026). **m.10010T>C Mitochondrial Disease: A Case Report With Hypoparathyroidism and Review of the Literature.** *Am J Med Genet A*, doi:[10.1002/ajmg.a.70134](https://doi.org/10.1002/ajmg.a.70134).

Mohsin, M. M., Azher, M., Asghar, F., Rahiman, H. H., Dube, R., Kar, Subhranshu Sekhar, Bahutair, S. N. M., Goud, B. K. M. et Kar, Swayam Siddha (2026). **Mitochondrial DNA Modification in Assisted Reproduction: Concept to Practice-A Narrative Review.** *Int J Mol Sci* 27, 2890, doi:[10.3390/ijms27062890](https://doi.org/10.3390/ijms27062890).

Morozov, Y. M. et Rakic, P. (2026). **Accomplishments of « Old-Fashioned » Electron Microscopy in the Period of Dominance of Immunofluorescent Methods.** *Int J Mol Sci* 27, 2803, doi:[10.3390/ijms27062803](https://doi.org/10.3390/ijms27062803).

Motoi, H., Watanabe, R., Shirai, A., Sakata, Y., Kuroda, Y., Watanabe, Y. et Ito, S. (2026). **Coenzyme Q10 Supplementation in a Child with Biallelic COQ8A Variants: A Case Report.** *Case Rep Neurol* 18, 121-127, doi:[10.1159/000550495](https://doi.org/10.1159/000550495).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Nakano, A., Masuya, T., Akisada, S., Ishikawa-Fukuda, M., Mitsuoka, K., Miyoshi, H., Murai, M. et Yokoyama, K. (2026). **Structures of respiratory supercomplexes and ATP synthase oligomers in mammalian mitochondrial inner membrane.** Nat Commun, doi:[10.1038/s41467-026-70578-x](https://doi.org/10.1038/s41467-026-70578-x).

Ngoh, S. H. et Gudi, A. (2026). **First case of schizophrenia and OCD in TK2-related mitochondrial DNA depletion myopathy: a case report.** BMC Psychiatry 26, 323, doi:[10.1186/s12888-026-07905-5](https://doi.org/10.1186/s12888-026-07905-5).

Orsucci, D., Caldarazzo lenco, E., Giuntini, M. et Vista, M. (2025). **The Clinical Burden of Inherited Neurometabolic Disorders in Adults-A Territorial Care Approach.** J Clin Med 15, 146, doi:[10.3390/jcm15010146](https://doi.org/10.3390/jcm15010146).

Paiva Coelho, M., Rodrigues, J. E., Costa, T., Dias, A., Graça, I. C. R., Rocha, H., Vilarinho, L., Martins, E. et Gil, A. M. (2026). **NMR-based urinary biomarkers in pediatric primary mitochondrial disorders and chronic kidney disease: shared mitochondrial dysfunction, diverging biosignatures.** Metabolomics 22, 17, doi:[10.1007/s11306-025-02363-8](https://doi.org/10.1007/s11306-025-02363-8).

Pan, X., Munan, S., Zuckerman, A. L., Pon, A., Thompson, N. B., Perera, R. M., Shrestha, N., Violante, S., Cross, J. R., Goodman, R. P., Shah, H. et Cracan, V. (2026). **A genetically encoded bifunctional enzyme mitigates redox imbalance and lipotoxicity.** Nat Metab 8, 350-370, doi:[10.1038/s42255-025-01450-3](https://doi.org/10.1038/s42255-025-01450-3).

Pasturino, F., De Wert, G. M. W. R., Herbrand, C. et Smeets, H. J. M. (2026). **From disease prevention to fertility treatment: rethinking the use of mitochondrial donation for oocyte-related infertility in the light of safety and efficacy.** Hum Reprod 41, 483-487, doi:[10.1093/humrep/deag027](https://doi.org/10.1093/humrep/deag027).

Pecheritsyna, S., Ermert, M. E., Podhumljak, E., Pennings, B., Zondag, R., Iannetti, E. F., Renkema, H. et Smeitink, J. (2026). **Sensitivity of primary mitochondrial disease fibroblasts to ferroptosis: The role of intracellular iron.** Mitochondrion 87, 102112, doi:[10.1016/j.mito.2026.102112](https://doi.org/10.1016/j.mito.2026.102112).

Perelló, M. A., Bellafont, F. J., Pujol-Ayach, E. et Alegret, M. (2026). **Anaesthetic management on a pediatric patient with Sengers syndrome. Case report.** Rev Esp Anesthesiol Reanim (Engl Ed) 502024, doi:[10.1016/j.redare.2026.502024](https://doi.org/10.1016/j.redare.2026.502024).

Pinho, B. R., Martins, V., Chacko, A. R., Nogueira, C., Duchon, M. R. et Oliveira, J. M. A. (2026). **Targeting mitochondrial deubiquitinase USP30 to induce mitophagy in heteroplasmic mitochondrial diseases.** Pharmacol Rep 78, 519-534, doi:[10.1007/s43440-026-00829-7](https://doi.org/10.1007/s43440-026-00829-7).

**L'essentiel des Maladies Mitochondriales**

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Plaza Enriquez, L., Ibrahim, R., Ayari, L., Gavrilova, R. et Kudva, Y. (2026). **Comprehensive Phenotype and Treatment Description of Mitochondrial Diabetes: Insights From a Large Cohort Study.** *Endocr Pract* S1530-891X(26)00027-3, doi:[10.1016/j.eprac.2026.01.010](https://doi.org/10.1016/j.eprac.2026.01.010).

Putko, B. N., Milone, M. et Liewluck, T. (2026). **Statins in Genetic Myopathies: A Retrospective Analysis of Safety and Tolerability.** *Neurol Clin Pract* 16, e200573, doi:[10.1212/CPJ.0000000000200573](https://doi.org/10.1212/CPJ.0000000000200573).

Ratnaike, T., Ramanan, S., Elkhateeb, N., Narayanan, R., Yang, J., Arany, E. S., Mirchandani, M., Piper, R., Schon, K., Kule, M. E., Gilmartin, C., Lochmüller, A., Shaw, E., Horváth, R. et Chinnery, P. F. (2026). **Charting the phenotypic landscape of mitochondrial diseases through a systematic evaluation of pathogenic mitochondrial DNA and nuclear gene variants.** *Genet Med* 28, 101620, doi:[10.1016/j.gim.2025.101620](https://doi.org/10.1016/j.gim.2025.101620).

Ruiz-Martinez, D.-A., Vega-Peniche, E.-R., Quiñonez-Pacheco, Y., Laviada-Molina, H., Medina-Campos, C.-A. et Campos-Garcia, F.-J. (2026). **Ethylmalonic encephalopathy caused by biallelic truncating variants in ETHE1: A case report.** *SAGE Open Med Case Rep* 14, 2050313X251412221, doi:[10.1177/2050313X251412221](https://doi.org/10.1177/2050313X251412221).

Russo, V., Hudelo, J., Marcel, M., Florence, J., Soulat, G., Manka, R., Pontana, F., Dacher, J. N., Toupin, S., Nazarian, S., Nigro, G., Wahbi, K. et Pezel, T. (2026). **Role of Cardiovascular Magnetic Resonance in Diagnosis and Management of Muscular Dystrophies.** *J Cardiovasc Magn Reson* 102693, doi:[10.1016/j.jocmr.2026.102693](https://doi.org/10.1016/j.jocmr.2026.102693).

Sellem, C. H., Bounaix, N., Logerais, M., Renaud, A., d'Elia, M. A., Richard, J., Almyre, C., Becker, G., Rivron, J., Hoarau, A., Gueguen, N., Desquirit-Dumas, V., Inisan, A., Belal, S., Mellinger, A., Godard, F., Paquis-Flucklinger, V., Baris, O. R., Azoulay, S., Delahodde, A., Tribouillard-Tanvier, D., Bonnefoy, N. et Procaccio, V. (2026). **Screening strategy using a filamentous fungus model to repurpose drugs for mitochondrial complex I deficiencies.** *Life Sci* 390, 124251, doi:[10.1016/j.lfs.2026.124251](https://doi.org/10.1016/j.lfs.2026.124251).

Şenol Ersak, A., Çağırın, T., Koçyiğit, A., Çıkmış, K., Yıldız, Y., Aypar, E., Ketenci İşlek, S., Şimşek Kiper, P. Ö., Haliloğlu, G. et Dursun, A. (2026). **Hypertrophic cardiomyopathy as a novel phenotypic feature of NSUN3-related mitochondrial disease: a case report with review of the literature.** *J Pediatr Endocrinol Metab* 39, 372-380, doi:[10.1515/jpem-2025-0578](https://doi.org/10.1515/jpem-2025-0578).

Shakerdi, L. A. (2026). **Metabolic Myopathies and HyperCKemia in Adulthood: A Clinical Approach to Diagnosis and Management.** *J Clin Med* 15, 2070, doi:[10.3390/jcm15052070](https://doi.org/10.3390/jcm15052070).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Sharma, R., Schimmenti, L. A., Smith, B., Pinto E Vairo, F., Selcen, D. et Dhamija, R. (2026). **MSTO1-related mitochondrial myopathy and ataxia syndrome: Case series and literature review.** *Neuromuscul Disord* 60, 106364, doi:[10.1016/j.nmd.2026.106364](https://doi.org/10.1016/j.nmd.2026.106364).

Shen, L. (2026). **Generative AI Accelerates Genotype-Phenotype Characterization of a 1600-Case Leigh Syndrome Virtual Cohort from Published Literature.** *Biology (Basel)* 15, 334, doi:[10.3390/biology15040334](https://doi.org/10.3390/biology15040334).

Sideris, D., Lee, H., Olson, L., Nallaparaju, K., Okuyama, K., Ciavarri, J., Lafyatis, R., Larsen, M., Lin, B., Alfaras, I., Kennerdell, J., Finkel, T., Liu, Y., Chen, B. et Lyu, L. (2026). **Suppression of interferon signaling via small-molecule modulation of TFAM.** *Elife* 14, RP108742, doi:[10.7554/eLife.108742](https://doi.org/10.7554/eLife.108742).

Silva, S. P., Pinto, M., Costa, D., Azevedo, A. et Dourado, E. (2026). **Non-inflammatory mimickers of myositis: a guide for rheumatologists.** *Clin Exp Rheumatol* 44, 353-367, doi:[10.55563/clinexprheumatol/v0joqy](https://doi.org/10.55563/clinexprheumatol/v0joqy).

Silva-Araújo, E. R., Toscano, A. E., Cavalcanti Bezerra Gouveia, H. J., Pontes, P. B., Dos Santos-Júnior, J. P., Dos Santos-Júnior, O. H., Teixeira Beltrão de Lemos, M. D., de Oliveira Melo, N. C., Viana de Melo, J., Padrón-Hernández, E. et Manhães-de-Castro, R. (2026). **Unraveling Riboflavin-Mediated Mitochondrial Modulation as a Therapeutic Pathway in Neurological Disorders: An Integrative Systematic Review.** *J Nutr* 101427, doi:[10.1016/j.tjnut.2026.101427](https://doi.org/10.1016/j.tjnut.2026.101427).

Singh, P., Tahavvori, A., Kuschner, C. E., Espin, B. B., Kazmi, J., Ramos, S. V., Yin, T., Hayashida, K., Ito-Hagiwara, K., Endo, Y., Yoshioka, K., Hagiwara, J., Sohi, A., Oropallo, A., Haddad, G., Li, T., Becker, L. B. et Kim, J. (2026). **Mitochondrial Transplantation as a Therapeutic Strategy for Inherited Mitochondrial Diseases.** *Adv Sci (Weinh)* 13, e23368, doi:[10.1002/advs.202523368](https://doi.org/10.1002/advs.202523368).

Skinner, O. S., Miranda, M., Dong, F., Struhl, T., Walker, M. A., Schleifer, G., Henke, M. T., Clardy, J., Hirano, M., De Vivo, D. C., Schon, E. A., Engelstad, K., Siegmund, S. E., Laprise, C., Des Rosiers, C., Mootha, V. K. et Sharma, R. (2026). **4,5-dihydroxyhexanoic acid is a robust circulating and urine marker of mitochondrial disease and its severity.** *bioRxiv* 2026.02.10.705117, doi:[10.64898/2026.02.10.705117](https://doi.org/10.64898/2026.02.10.705117).

Smirnova, A., Urtis, M. et Arbustini, E. (2026). **Applications of genetic testing in cardiovascular disease.** *Curr Opin Cardiol* 41, 109-119, doi:[10.1097/HCO.0000000000001281](https://doi.org/10.1097/HCO.0000000000001281).

Sørensen, N. B., Stamenkovic, K., Steffensen, E. H. et Kofoed, I. R. (2026). **[Super-refractory status epilepticus caused by hereditary mitochondrial disease].** *Ugeskr Laeger* 188, V07250609, doi:[10.61409/V07250609](https://doi.org/10.61409/V07250609).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Sun, S., Cao, H., Li, X. et Liao, H. (2025). **NFS1 Plays a Critical Role in Regulating Ferroptosis Homeostasis**. *Biomolecules* 16, 32, doi:[10.3390/biom16010032](https://doi.org/10.3390/biom16010032).

Szabo, M., Lagos, D., Cross, E., Collier, J. et Horvath, R. (2026). **Mitochondrial DNA release and inflammation in mitochondrial disease pathogenesis**. *Brain* awag037, doi:[10.1093/brain/awag037](https://doi.org/10.1093/brain/awag037).

Tafakhori, A., Sarvestani, Z., Kariminejad, A., Tajsharghi, H., Seo, G. H., Ryu, S. W. et Heydari Havadaragh, S. (2026). **Homozygous FDXR variant in twin sisters with spastic paraparesis followed by acute progressive flaccid quadriplegia**. *BMC Neurol* 26, 202, doi:[10.1186/s12883-026-04752-5](https://doi.org/10.1186/s12883-026-04752-5).

Tan, N. B., Gautschi, M., Raum, M., Hock, D. H., Kopajtich, R., Wang, J., Qian, X., Sharma, T., Green, T. E., Nuoffer, J.-M., Bell, K. M., Pospieszny, K., Stait, T., Pike, C., Cao, M., White, S. M., Thorburn, D. R., Brunet, T., Wagner, M., Müller-Felber, W., Zeng, L., Klopstock, T., Schaller, A., Liu, J., Stroud, D. A. et Prokisch, H. (2026). **Bi-allelic variants in NDUF5 cause a mitochondrialopathy with complex I deficiency**. *Am J Hum Genet* S0002-9297(26)00113-8, doi:[10.1016/j.ajhg.2026.03.003](https://doi.org/10.1016/j.ajhg.2026.03.003).

Tao, X., Jiang, W. et Tang, H. (2026). **A Mysterious Case of Diffuse Severe Hepatic Steatosis in a Thin Teenager**. *Gastroenterology* S0016-5085(26)00255-6, doi:[10.1053/j.gastro.2026.02.043](https://doi.org/10.1053/j.gastro.2026.02.043).

Thoma, F., Hagen, J., Rathberger, R., Padovani, F., Hörl, D., Schmoller, K. M. et Osman, C. (2026). **Local mitochondrial physiology defined by mtDNA quality guides purifying selection**. *PLoS Genet* 22, e1011836, doi:[10.1371/journal.pgen.1011836](https://doi.org/10.1371/journal.pgen.1011836).

Tinker, R. J., Jacob, N., Syed, M. G., Kelkar, J., Donnelly, C., Elsharkawi, I., Ganesh, J., Gelb, B. D., Pejaver, V., Kozicz, T. et Morava, E. (2026). **Drivers of Diagnostic Delay in Mitochondrial Disease: Missed Recognition of Canonical Features**. *JIMD Rep* 67, e70068, doi:[10.1002/jmd2.70068](https://doi.org/10.1002/jmd2.70068).

Varga, L., Borecka, S., Skopkova, M., Rambani, V., Sklenar, M., Cipkova, K., Kickova, T., Ugorova, D., Kabatova, Z., Stanik, J., Profant, M. et Gasperikova, D. (2026). **Screening for Maternally Inherited Diabetes and Deafness in Large Cohorts of Hearing Impaired and Diabetic Patients**. *Ear Hear* 47, 863-876, doi:[10.1097/AUD.0000000000001780](https://doi.org/10.1097/AUD.0000000000001780).

Verbal, F., Rubilar, N., Marileo, A. M., Fierro, H., Ramirez-Molina, O. G., Pinto-Leon, A., Yevénes, G. E., Fuentealba, J. et Panes-Fernández, J. (2026). **Current perspectives on circadian regulation of mitochondrial dynamics in mood disorders and perioperative stress**. *Front Pharmacol* 17, 1723748, doi:[10.3389/fphar.2026.1723748](https://doi.org/10.3389/fphar.2026.1723748).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Villafan-Bernal, J. R., Martínez-Hernández, A., García-Ortiz, H., Contreras-Cubas, C., Guerrero-Contreras, I., Frías-Cabrera, J. L., Centeno-Cruz, F., Morales Rivera, M. I., Hernández, J. R., Carnevale, A., Barajas-Olmos, F. et Orozco, L. (2026). **Correlation of MLASA2 Clinical Phenotype and Survival with Mt-TyrRS Protein Damage: Linking Systematic Review, Meta-Analysis and 3D Hotspot Mapping**. *Curr Issues Mol Biol* 48, 95, doi:[10.3390/cimb48010095](https://doi.org/10.3390/cimb48010095).

Villeneuve-Cloutier, N., Warman-Chardon, J. et Bourque, D. K. (2026). **Expanding the Phenotype of TUFM-Related Combined Oxidative Phosphorylation Deficiency 4**. *Am J Med Genet A*, doi:[10.1002/ajmg.a.70136](https://doi.org/10.1002/ajmg.a.70136).

Wang, D., Zhang, G., Fang, X., Liu, F. et Wang, L. (2025). **Case Report: Myoclonic and tremulous movements associated with COQ8A-related coenzyme Q10 deficiency type 4**. *Front Genet* 16, 1682085, doi:[10.3389/fgene.2025.1682085](https://doi.org/10.3389/fgene.2025.1682085).

Wang, J., Yu, X., Si, W., Lou, J., Zhuang, X., Li, B., Zhao, Y., Shi, Y., Yan, C., Ji, K. et Lin, Y. (2026). **Single-Cell Sequencing and Transcriptomic Sequencing Exploration of the Mechanisms Governing Lactylation in Mitochondrial Diseases**. *FASEB J* 40, e71644, doi:[10.1096/fj.202504230R](https://doi.org/10.1096/fj.202504230R).

Wang, Y., Su, X., Chen, Yu, Chen, Yijian, Shi, C., Liu, F., Ye, Y., Sun, P., Tan, M., Yu, M., Wang, Ya, Xie, S., Liu, J., Yan, Q., Sun, Q., Neculai, D., Liu, W., Shao, J., Liu, Y., Lin, W. et Lin, A. (2026). **A CRISPR-based mitochondrial gene therapy tool derived by engineering guide RNAs**. *Cell Rep* 45, 116958, doi:[10.1016/j.celrep.2026.116958](https://doi.org/10.1016/j.celrep.2026.116958).

Wu, Y., Fu, L., Xu, X., Chan, P. S. et Wu, R. (2026). **Systematic and Quantitative Investigation of Newly Synthesized Proteins Reveals Distinct Ion Homeostasis and Mitochondrial Changes between Cuproptosis and Ferroptosis in Human Cells**. *Anal Chem* 98, 8309-8320, doi:[10.1021/acs.analchem.5c07257](https://doi.org/10.1021/acs.analchem.5c07257).

Xu, C., Jin, H., Li, J., Liu, Z., Zhang, W., Zhou, J., Duan, R., Liu, Y., Song, M., Zhang, Z., Li, T., Shen, D., Zou, Y., Wang, J., Li, H., Jiang, H. et Fang, F. (2026). **Deficiency of the NAD(P)HX metabolic repair system: a treatable mitochondrial disease**. *Orphanet J Rare Dis* 21, 63, doi:[10.1186/s13023-026-04218-4](https://doi.org/10.1186/s13023-026-04218-4).

Xu, J., Zhang, X., Hua, Y., Yang, L., Shi, D. et Qiu, S. (2026). **Multiple Mitochondrial Dysfunction Syndrome Caused by IBA57 Gene Mutation: A Case Report and Literature Review**. *Mol Genet Genomic Med* 14, e70200, doi:[10.1002/mgg3.70200](https://doi.org/10.1002/mgg3.70200).

Yagasaki, H., Narusawa, H., Watanabe, D., Sano, F., Fujioka, K., Mizorogi, S., Kaga, Y. et Inukai, T. (2026). **Nutrient and endocrine factors affecting impaired growth in pediatric mitochondrial diseases**. *Endocrinol Diabetes Metab Case Rep* 2026, EDM250140, doi:[10.1530/EDM-25-0140](https://doi.org/10.1530/EDM-25-0140).

## L'essentiel des Maladies Mitochondriales

MMITO-2026-1 du 1<sup>er</sup> janvier au 31 mars 2026

Yan, H.-M., Quan, Y., Zhou, Y., Jiang, L., Zhang, L.-Y., Wan, Z.-Q. et Xi, H. (2026). **[Decreased plasma citrulline is a biochemical marker in newborn screening for MT-ATP6-associated mitochondrial disease: two case reports and a literature review]**. Zhongguo Dang Dai Er Ke Za Zhi 28, 250-256, doi:[10.7499/j.issn.1008-8830.2505143](https://doi.org/10.7499/j.issn.1008-8830.2505143).

Yang, F., Yao, R., Chang, G., Hu, J., Feng, B., Wang, L., Hu, F., Huang, Y., Wu, S., Yu, T., Ding, Y. et Wang, X. (2026). **Clinical and genetic spectrum of pediatric mitochondrial disorders in China: insights from a 47-case genetically confirmed cohort**. Orphanet J Rare Dis 21, 158, doi:[10.1186/s13023-025-04180-7](https://doi.org/10.1186/s13023-025-04180-7).

Zhang, X., Li, K., Tan, L., Chen, W., Gao, S., Liu, C., Pan, S., He, J., Liu, N., Wan, G., Dong, W., Kong, W., Shen, B., Qi, X. et Ma, Y. (2026). **One-step generation of heritable mitochondrial DNA multiplex-engineered rats using DddA-derived cytosine base editor**. Animal Model Exp Med, doi:[10.1002/ame2.70154](https://doi.org/10.1002/ame2.70154).

Zhou, Y., Li, K., Zhu, R., Ma, X., Ye, X., Mao, M., Li, D., Zeng, X., Chen, Z., Wu, J., Jin, L., Tang, X., Yang, Y., Lyu, J. et Lou, X. (2026). **COG5 deficiency disrupts cellular copper homeostasis and underlies the impaired mitochondrial OXPHOS function**. PLoS Genet 22, e1012076, doi:[10.1371/journal.pgen.1012076](https://doi.org/10.1371/journal.pgen.1012076).

Zieniuk, B., Wierzchowska, K., Jasińska, K., Kobus, J., Piotrowicz, A., Uğur, Ş. et Fabiszewska, A. (2026). **Yeast as a Model for Human Disease**. Int J Mol Sci 27, 1632, doi:[10.3390/ijms27041632](https://doi.org/10.3390/ijms27041632).

Zink, A., Dai, D.-F., Wittich, A., Henke, M.-T., Pedrotti, G., Heiduschka, S., Santamaria, G., Pentimalli, T. M., Brueser, C., Notopoulou, S., Umar, A. R., Zhaivoron, A., Petersilie, L., Jerred, C., Bergmans, J., Neu, L. A., Schumacher, F., Keller-Findeisen, J., Rybak-Wolf, A., Stach, D., Reinshagen, J., Haferkamp, U., Krieg, K., Zaliani, A., Euro, L., Di Donfrancesco, A., Santanatoglia, C., Cappellozza, E., Suarez Cubero, M., Pavez-Giani, M., Bakumenko, O., Meierhofer, D., Foley, A., Morales-Gonzalez, S., Tolle, I., Herebian, D., Bonesso, D., Cecchetto, G., Wong, S. N., Moresco, M., Maresca, A., Decimo, I., De Sanctis, F., Adamo, A., Adjobo-Hermans, M. J. W., Duchi, R., Barandalla, M., Scaglia, M., Perota, A., Galli, C., Kleuser, B., Cyganek, L., Mülhhausen, C., Schlotawa, L., Tiranti, V., Mayatepek, E., Szabo, I., La Morgia, C., Klopstock, T., Carelli, V., Distelmaier, F., Rossi, A., Rajewsky, N., Ullah, G., Jakobs, S., Rose, C. R., Petrakis, S., Edenhofer, F., Koopman, W. J. H., Lisowski, P., Suomalainen, A., Brunetti, D., Del Sol, A., Bottani, E., Pless, O., Schuelke, M. et Prigione, A. (2026). **Pluripotent stem-cell-based screening uncovers sildenafil as a mitochondrial disease therapy**. Cell 189, 1656-1679.e42, doi:[10.1016/j.cell.2026.02.008](https://doi.org/10.1016/j.cell.2026.02.008).