

Mitochondrial disorders : The Essential
MMITO-2025-3 from july 1st to september 30th, 2025

Mitochondrial disorders : The Essential



3rd quarter 2025



quarterly literature review

carried out by the AFM-Téléthon documentation department

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



Mitochondrial disorders : The Essential

MMITO-2025-3 from july 1st to september 30th, 2025

Sommaire

ATAXIE DE FRIEDREICH - <i>FRIEDREICH ATAXIA</i>	3
ATROPHIE OPTIQUE AUTOSOMIQUE DOMINANTE – <i>AUTOSOMAL DOMINANT OPTIC ATROPHY (ADOA)</i>	7
ENCEPHALOPATHIE MYO-NEURO-GASTROINTESTINALE - <i>MITOCHONDRIAL NEUROGASTROINTESTINAL ENCEPHALOMYOPATHY (MNGIE)</i>	8
MALADIE DE CHARCOT-MARIE-TOOTH D’ORIGINE MITOCHONDRIALE – <i>MITOCHONDRIAL CHARCOT-MARIE-TOOTH DISEASE</i>	9
MALADIES LIEES AU GENE ACO2- <i>ACO2-RELATED DISORDERS</i>	11
MALADIE LIEE AU GENE GFM1 – <i>GFM1-RELATED DISORDER</i>	11
MALADIES LIEES AU GENE POLG (INCLUS SANDO, SCAE) - <i>POLG-RELATED DISORDERS (INCLUDED SANDO, SCAE)</i>	11
MELAS	12
MERFF	15
NEUROPATHIE OPTIQUE HEREDITAIRE DE LEBER - <i>LEBER HEREDITARY OPTIC NEUROPATHY (LHON)</i>	15
OPHTALMOPLÉGIE EXTERNE PROGRESSIVE – <i>PROGRESSIVE EXTERNAL OPHTHALMOPLÉGIA (PEO)</i>	20
SYNDROME D'ALPERS-HUTTENLOCHER - <i>ALPERS-HUTTENLOCHER SYNDROME</i>	21
SYNDROME DE KEARNS-SAYRE – <i>KEARNS-SAYRE SYNDROME</i>	21
SYNDROME DE LEIGH - <i>LEIGH SYNDROME</i>	23
MALADIES MITOCHONDRIALES (AUTRES) – <i>MITOCHONDRIAL DISORDERS (OTHER)</i>	26



Mitochondrial disorders : The Essential
MMITO-2025-3 from july 1st to september 30th, 2025

Ataxie de Friedreich - *Friedreich ataxia*

Benkirane, M., Marelli, C., Choumert, A., Goizet, C., Patat, O., Ewencyk, C., Anheim, M., Mégarbané, A., Larrieu, L., Charlin, C., Ory Magne, F., Chaussenot, A., Fradin, M., Guissart, C., Pointaux, M., Cossée, M., Vincent, M.-C., Bergougnoux, A., Hersent, C., Bareil, C., Roubertie, A., Fluchère, F., Renaud, M., Kremer, L., Tranchant, C., Attarian, S., Odent, S., Laugel, V., Walther-Louvier, U., Desnoux, B., Bieth, E., Husson, I., Azulay, J. P., INTREP-AF consortium, Rivier, F., Doray, B., Durr, A., Aouinti, S., Molinari, N. et Koenig, M. (2025). **Type and position of repeat interruptions as determinants of disease severity and expansion size in Friedreich ataxia.** Genet Med 101588, doi:[10.1016/j.gim.2025.101588](https://doi.org/10.1016/j.gim.2025.101588).

Chen, L., Rizzo, G., Bulawa, C., Van Dijk, K. R. A., Henning, E. C., Martelli, A., Palmer, J., McIntosh, A., Pregel, M., Sun, P., Adewunmi, E., Aldridge, M., Chan, J., Gunn, R. N., Huiban, M., Listanco, A., Loudon, P. T., Moz, S., Passchier, J., Sauvage, L., Stewart, R., Wells, L., Rabiner, E. A., Charnas, L. R. et Festenstein, R. J. (2025). **Evaluation of Mitochondrial Complex 1 Density with [18F]BCPP-EF in a Murine Model and Individuals with Friedreich Ataxia.** J Nucl Med 66, 1434-1439, doi:[10.2967/jnumed.124.268698](https://doi.org/10.2967/jnumed.124.268698).

Correa-Arrieta, C., Castellar-Leones, S., Ruiz-Ospina, E., Villamil-Osorio, M., Bobadilla-Quesada, E. J. et Ortiz-Corredor, F. (2025). **Friedreich's Ataxia in Colombia: A Population-Based Study of Incidence and Socioeconomic Determinants.** Mov Disord, doi:[10.1002/mds.70033](https://doi.org/10.1002/mds.70033).

Dakin, A., Bogdanova-Mihaylova, P., Walsh, R. A., Murphy, S. M., Ward, D., Maher, N. et McCarthy, C. M. (2025). **Friedreich's ataxia: A case series, literature review, and recommendations for pregnancy.** Int J Gynaecol Obstet, doi:[10.1002/ijgo.70361](https://doi.org/10.1002/ijgo.70361).

Di Folco, C., Dubec-Fleury, C., Träschütz, A., Kessler, C., Reich, S., Gagnon, C., Lessard, I., Rodrigue, X., Coccozza, S., Satolli, S., Santorelli, F. M., Durr, A., Heinzmann, A., van de Warrenburg, B. P., Willemse, I. H. J., Başak, A. N., Vural, A., Brais, B., Klebe, S., Horvath, R., PROSPAX Consortium, Schüle, R., Synofzik, M. et Tezenas du Montcel, S. (2025). **Spastic Ataxia Composite (SPAXCOM): A Scale to Evaluate the Progression of Subjects with Spasticity and Ataxia.** Mov Disord, doi:[10.1002/mds.70006](https://doi.org/10.1002/mds.70006).

Mitochondrial disorders : The Essential

MMITO-2025-3 from July 1st to September 30th, 2025

Erdmann, H., Schaub, A., Lucas, M. C., Scholz, V., Benet-Pages, A., Becker, K., Dineiger, C., Mayer, V., van Buren, I., Breithausen, E., Akbari, K., Cordts, I., Sauer, M., Schneider, C., Krakowsky, R., Schnabel, F., Dunker, K., Fabritius, L., Gerb, J., Grabova, D., Möhwald, K., Näher, M., Steinmetz, K., Thiessen, F., Jäck, A., Schneider-Gold, C., Zittel, S., Petersen, C., Schreyer, I., Mämecke, L., Wilfling, S., Wunderlich, G., Brenner, D., Hellenbroich, Y., Muhle, K., Huchtemann, T., Claus, I., Klopstock, T., Strupp, M., Levin, J., Höglinger, G., Huppert, D., Becker-Bense, S., Filippopoulos, F., Kilpert, F., Leitão, E., Kaya, S., Depienne, C., Schöberl, F., Neuhaus, T., Holinski-Feder, E., Zwergal, A. et Abicht, A. (2025). **Repeat-associated ataxias in a German patient cohort analysed by targeted parallel long-read sequencing.** *Brain* awaf318, doi:[10.1093/brain/awaf318](https://doi.org/10.1093/brain/awaf318).

Eshaghi, K., Rao, P. H., Shen, M. M. et Lynch, D. R. (2025). **Genetic and Phenotypic Variability in Siblings With Friedreich Ataxia.** *Neurol Genet* 11, e200234, doi:[10.1212/NXG.0000000000200234](https://doi.org/10.1212/NXG.0000000000200234).

Giacich, M., Naef, V., Santorelli, F. M. et Damiani, D. (2025). **Roots of Progress: Uncovering Cerebellar Ataxias Using iPSC Models.** *Biomedicine* 13, 2121, doi:[10.3390/biomedicine13092121](https://doi.org/10.3390/biomedicine13092121).

Heleven, E., Vyhnaek, M., Karamazovová, S., Van Overwalle, F. et Naeije, G. (2025). **False Beliefs, True Deficits: Investigating Social Cognition in Friedreich Ataxia.** *Cerebellum* 24, 128, doi:[10.1007/s12311-025-01886-z](https://doi.org/10.1007/s12311-025-01886-z).

Hemachudha, P., Anukoolwittaya, P., Pongpitakmetha, T., Joyjinda, Y., Ruchisrisarod, C., Saraya, A. W., Rattanawong, W., Thanapornsungsuth, P. et Hemachudha, T. (2025). **Understanding the genetics and neurology: an overview of adult neurogenetics.** *Asian Biomed (Res Rev News)* 19, 196-208, doi:[10.2478/abm-2025-0022](https://doi.org/10.2478/abm-2025-0022).

Hu, Y., Gao, L., Lee, L., Cherry, J. J. et Kong, R. (2025). **Characterizing Population Pharmacokinetics of Vatiquinone in Healthy Volunteers and Patients with Friedreich's Ataxia.** *Pharmaceuticals (Basel)* 18, 1339, doi:[10.3390/ph18091339](https://doi.org/10.3390/ph18091339).

Jiménez-Jiménez, F. J., Alonso-Navarro, H., García-Martín, E., Cárcamo-Fonfría, A., Martín-Gómez, M. A. et Agúndez, J. A. G. (2025). **Oxidative Stress and Antioxidant Therapies in Friedreich's Ataxia.** *Cells* 14, 1406, doi:[10.3390/cells14181406](https://doi.org/10.3390/cells14181406).

Karamazovova, S., Laczó, M., Matuskova, V., Svecova, N., Stovickova, L., Blichova, Z., Paulasova Schwabova, J., Kuzmiak, M., Laczó, J. et Vyhnaek, M. (2025). **Spatial perspective taking is impaired in spinocerebellar ataxias and Friedreich ataxia.** *Sci Rep* 15, 31126, doi:[10.1038/s41598-025-16302-z](https://doi.org/10.1038/s41598-025-16302-z).



Mitochondrial disorders : The Essential

MMITO-2025-3 from July 1st to September 30th, 2025

Lee, L., Murase, K., Thoolen, M., Giannousis, P., Kaushik, D., Golden, L. et Kong, R. (2025). **Enhanced Vatiquinone Bioavailability With Fatty Meals and Optimal Dosing Schedule.** Biopharm Drug Dispos, doi:[10.1002/bdd.70015](https://doi.org/10.1002/bdd.70015).

Li, L., Scott, W. S. et Mirkin, S. M. (2025). **Emerging drivers of DNA repeat expansions.** Biochem Soc Trans 53, 995-1010, doi:[10.1042/BST20253067](https://doi.org/10.1042/BST20253067).

McGarrell, N. T., Green, M. E. et McCully, K. K. (2025). **Muscle Endurance Training in a Person with Friedreich's Ataxia.** Muscles 4, 1, doi:[10.3390/muscles4010001](https://doi.org/10.3390/muscles4010001).

Omaveloxolone (Skyclarys): Indication: For the treatment of Friedreich's ataxia in patients 16 years of age and older: Reimbursement Recommendation (2025). Canadian Agency for Drugs and Technologies in Health, Ottawa (ON), URL: <http://www.ncbi.nlm.nih.gov/books/NBK617230/>.

Oubre, B., Yang, F., Luddy, A. C., Manohar, R., Soja, N. N., Stephen, C. D., Schmahmann, J. D., Kulkarni, D., White, L., Patel, S. et Gupta, A. S. (2025). **A Digital Measure of Eye Movements During Reading Sensitively Captures Oculomotor and Speech Dysfunction, Early Changes, and Disease Progression in Ataxias.** Ann Neurol, doi:[10.1002/ana.78039](https://doi.org/10.1002/ana.78039).

Perlman, S., Zhang, S., Setyawan, J., Yang, H., Jiang, A., Hua, Q., Boudreau, J., Khan, S., Bajaj, A., Lynch, D. R. et Lawson, R. (2025). **The clinical burden of Friedreich ataxia in the United States: A retrospective claims database analysis.** J Neurol Sci 475, 123594, doi:[10.1016/j.jns.2025.123594](https://doi.org/10.1016/j.jns.2025.123594).

Petrillo, S., Mongelli, A., Castaldo, A., Sarro, L., Azzarelli, S., Ronco, R., Castellotti, B., Gellera, C., Piemonte, F. et Mariotti, C. (2025). **Impact of age on neurofilament light chain in Friedreich ataxia: a 1-year longitudinal study.** Brain Commun 7, fcac331, doi:[10.1093/braincomms/fcac331](https://doi.org/10.1093/braincomms/fcac331).

Phang, M. W. L., Hisam, N. S. M., Supandi, F., Cheng, P. G., Lim, S. H., Lim, L. W. et Wong, K. H. (2025). **The Tiger Milk Medicinal Mushroom Lignosus rhinocerus (Agaricomycetes) Mitigates Oxidative Damage in a Cellular Model Mimicking Friedreich's Ataxia.** Int J Med Mushrooms 27, 63-87, doi:[10.1615/IntJMedMushrooms.2025059734](https://doi.org/10.1615/IntJMedMushrooms.2025059734).

Portillo-Carrasquer, M., Sanz-Alcázar, A., Delaspre, F., Pazos-Gil, M., Oliveira-Jorge, L., Vergara, C., Rodríguez-Pascau, L., Pizcueta, P., Tamarit, J., Ros, J. et Cabisco, E. (2025). **Leriglitazone improves iron homeostasis and ferroptotic markers in frataxin-deficient dorsal root ganglia neurons.** Biomed Pharmacother 192, 118553, doi:[10.1016/j.biopha.2025.118553](https://doi.org/10.1016/j.biopha.2025.118553).

Mitochondrial disorders : The Essential
MMITO-2025-3 from july 1st to september 30th, 2025

Pretegiani, E., Garces, P., Antoniadis, C. A., Sobanska, A., Kovacs, N., Ying, S. H., Gupta, A. S., Perlman, S., Szmulewicz, D. J., Pane, C., Németh, A. H., Jardim, L. B., Coarelli, G., Kuzmiak, M., Milovanovic, A., Träschütz, A. et Tarnutzer, A. A. (2025). **Best Oculomotor Endpoints for Clinical Trials in Hereditary Ataxias: A Systematic Review and Consensus by the Ataxia Global Initiative Working Group on Digital-Motor Biomarkers.** Cerebellum 24, 141, doi:[10.1007/s12311-025-01894-z](https://doi.org/10.1007/s12311-025-01894-z).

Rotar, M. et Leonardis, L. (2025). **Sensory nerve action potential reappearance after omaveloxolone treatment in patients with Friedreich ataxia.** Clin Neurophysiol 178, 2110974, doi:[10.1016/j.clinph.2025.2110974](https://doi.org/10.1016/j.clinph.2025.2110974).

Rummey, C., Perlman, S., Subramony, S. H., Corti, M., Farmer, J. et Lynch, D. R. (2025). **Disease Progression in Children With Friedreich Ataxia: Functional Performance and Other Outcome Assessments in the FACHILD Study.** J Child Neurol 8830738251353475, doi:[10.1177/08830738251353475](https://doi.org/10.1177/08830738251353475).

Saha, S., Jha, A., Yadaw, M. et Tiwari, B. (2025). **Hypertrophic cardiomyopathy with ataxic gait: a cardiac clue to a neurologic diagnosis.** BMJ Case Rep 18, e265662, doi:[10.1136/bcr-2025-265662](https://doi.org/10.1136/bcr-2025-265662).

Sureshkumar, P., Kumar, S. S., Cheriyan, J. et Masood, A. (2025). **Friedreich Ataxia and Related Diabetes: Therapeutic Approach Targeting Mitochondrial Dysfunction.** JCEM Case Rep 3, luaf215, doi:[10.1210/jcemcr/luaf215](https://doi.org/10.1210/jcemcr/luaf215).

Talaverón-Rey, M., Reche-López, D., Povea-Cabello, S., Álvarez-Córdoba, M., Gómez-Fernández, D., Romero-González, A., Cilleros-Holgado, P., Romero-Domínguez, J. M., López-Cabrera, A., Piñero-Pérez, R., González-Granero, S., García-Verdugo, J. M. et Sánchez-Alcázar, J. A. (2025). **Alpha-lipoic acid supplementation improves pathological alterations in cellular models of Friedreich ataxia.** Orphanet J Rare Dis 20, 453, doi:[10.1186/s13023-025-03990-z](https://doi.org/10.1186/s13023-025-03990-z).

Wirth, T., Faber, J., Depienne, C., Roze, E., Honnorat, J., Meissner, W. G., Giunti, P., Tranchant, C., Klockgether, T. et Anheim, M. (2025). **Progress and challenges in sporadic late-onset cerebellar ataxias.** Nat Rev Neurol, doi:[10.1038/s41582-025-01136-0](https://doi.org/10.1038/s41582-025-01136-0).

Yameogo, P., Aguilar, S., Prakash, T. P., Rigo, F., Lynch, D. R., Napierala, J. S. et Napierala, M. (2025). **Antisense oligonucleotide therapy for patients with Friedreich's ataxia carrying the c.165+5G>C splicing mutation.** Mol Ther Nucleic Acids 36, 102617, doi:[10.1016/j.omtn.2025.102617](https://doi.org/10.1016/j.omtn.2025.102617).

Zesiewicz, T. A. (2025). **Ataxia.** Continuum (Minneap Minn) 31, 1093-1119, doi:[10.1212/cont.0000000000001599](https://doi.org/10.1212/cont.0000000000001599).



Mitochondrial disorders : The Essential
MMITO-2025-3 from july 1st to september 30th, 2025

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

Becchi, G., Whitehead, M., Harvey, J. P., Sladen, P. E., Dushti, M., Chapple, J. P., Yu-Wai-Man, P. et Cheetham, M. E. (2025). **CRISPRa-Mediated Increase of OPA1 Expression in Dominant Optic Atrophy**. Int J Mol Sci 26, 6364, doi:[10.3390/ijms26136364](https://doi.org/10.3390/ijms26136364).

Britton, J. O. T., Yu-Wai-Man, P. et Chen, B. S. (2025). **Technological advances in the diagnosis and management of inherited optic neuropathies**. Front Neurol 16, 1609033, doi:[10.3389/fneur.2025.1609033](https://doi.org/10.3389/fneur.2025.1609033).

Champigny, C., Botella, M., Atamena, D., Bullich, S., Coustham, C., Guiard, B., Belenguer, P. et Davezac, N. (2025). **A High-Fat Diet Induces Oxidative Stress in OPA1+/- Mouse Cortices: A Critical Double Challenge**. Antioxidants (Basel) 14, 876, doi:[10.3390/antiox14070876](https://doi.org/10.3390/antiox14070876).

Chen, B. S., Seikus, C., Ferguson, J., Tadić, V., Horton, M., Yu-Wai-Man, P. et Archer, S. (2025). « **Adrift From the World** »: Exploring the Lived Experiences of Individuals Affected by an Inherited Optic Neuropathy in the United Kingdom-A Qualitative Study. Value Health S1098-3015(25)02490-8, doi:[10.1016/j.jval.2025.07.023](https://doi.org/10.1016/j.jval.2025.07.023).

Harvey, J. P., Hong, E. H. et Yu-Wai-Man, P. (2025). **Natural History and Biomarker Challenges in Dominant Optic Atrophy: Implications for Therapeutic Studies**. Clin Exp Ophthalmol 53, 597-599, doi:[10.1111/ceo.14583](https://doi.org/10.1111/ceo.14583).

Tachibana, M., Hayashi, T., Igawa, Y., Mizobuchi, K., Miyasaka, Y., Tsuchihashi, T. et Shinoda, K. (2025). **Case of autosomal dominant optic atrophy with relatively good visual function**. BMC Ophthalmol 25, 443, doi:[10.1186/s12886-025-04276-5](https://doi.org/10.1186/s12886-025-04276-5).



Mitochondrial disorders : The Essential
MMITO-2025-3 from july 1st to september 30th, 2025

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

Bax, B. E. et Uçar, S. K. (2025). **Untargeted Plasma Metabolomics Extends the Biomarker Profile of Mitochondrial Neurogastrointestinal Encephalomyopathy.** Int J Mol Sci 26, 9107, doi:[10.3390/ijms26189107](https://doi.org/10.3390/ijms26189107).

Brevini, T., Swift, L., Reynolds, H., Ong, J., Stopforth, R. J., Malam, Y., Spiers, H. V. M., Jovanovic, E., Scaravaglio, M., Vakeeswarasarma, V., Heslop, J., Emmett, E., Andreatta, B., Athanasiadis, E., Cacciottolo, T. M., Fuchs, C. D., Trauner, M., Hosgood, S. A., See, T. C., Pantelis, P., Foukaneli, D., Gelson, W. T. H., Gibbs, P., Allinson, K., Pettigrew, G. J., Duckworth, A., Paterson, A. L., Gorgoulis, V. G., Thompson, R. J., Kaser, A., Kosmoliaptsis, V., Chinnery, P. F., Marti, R., van den Ameele, J., Webb, G. J., Watson, C. J. E., Butler, A. J. et Sampaziotis, F. (2025). **Successful AAV8 gene therapy on hepatic ex situ machine perfusion for mitochondrial neurogastrointestinal encephalomyopathy.** J Hepatol 83, 1218-1225, doi:[10.1016/j.jhep.2025.07.022](https://doi.org/10.1016/j.jhep.2025.07.022).

Patel, M., Keener, K., LaWall, G. et Jha, P. (2025). **Mitochondrial Neurogastrointestinal Encephalomyopathy Presenting with Peripheral Neuropathy and Hearing Loss.** WMJ 124, 287-290.

Xu, X., Xia, J., Xu, F., Wang, M., Yang, L. et Chen, X. (2025). **Mitochondrial neurogastrointestinal encephalomyopathy in china: a novel TYMP variant and comprehensive clinical-genetic insights.** Orphanet J Rare Dis 20, 439, doi:[10.1186/s13023-025-03962-3](https://doi.org/10.1186/s13023-025-03962-3).

Zhang, X.-Y., Zhu, Q.-L., Ruan, G.-C. et Li, W.-B. (2025). **[Clinical and Intestinal Ultrasound Findings in Mitochondrial Neurogastrointestinal Encephalomyopathy: Report of One Case].** Zhongguo Yi Xue Ke Xue Yuan Xue Bao, doi:[10.3881/j.issn.1000-503X.16801](https://doi.org/10.3881/j.issn.1000-503X.16801).

Zioudi, A., Gouiza, I., Galai, S., Hechmi, M., Klaa, H., Miladi, Z., Ben Younes, T., Benrhouma, H., Ben Youssef-Turki, I., Omar, S., Lenaers, G., Kefi, R., Gouider-Khouja, N. et Kraoua, I. (2025). **A Tunisian POLG mutation expands the clinical spectrum of POLG-related disorders.** Mitochondrion 85, 102071, doi:[10.1016/j.mito.2025.102071](https://doi.org/10.1016/j.mito.2025.102071).

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

Armirola-Ricaurte, C., Morant, L., Adant, I., Hamed, S. A., Pipis, M., Efthymiou, S., Amor-Barris, S., Atkinson, D., Van de Vondel, L., Tomic, A., Seneca, S., de Vriendt, E., Zuchner, S., Ghesquiere, B., Hanna, M., Houlden, H., Lunn, M. P., Reilly, M. M., Rasic, V. M. et Jordanova, A. (2025). **Biallelic variants in COX18 cause a mitochondrial disorder primarily manifesting as peripheral neuropathy.** Brain awaf300, doi:[10.1093/brain/awaf300](https://doi.org/10.1093/brain/awaf300).

Bora, P., Zaman, M., Oviedo, S., Kutseikin, S., Madrazo, N., Mathur, P., Pannikkat, M., Krasny, S., Chu, A., Johnson, K. A., Grotjahn, D. A., Shutt, T. E. et Wiseman, R. L. (2025). **Drug Repurposing Screen Identifies an HRI Activating Compound that Promotes Adaptive Mitochondrial Remodeling in MFN2-deficient Cells.** bioRxiv 2025.06.23.660251, doi:[10.1101/2025.06.23.660251](https://doi.org/10.1101/2025.06.23.660251).

Correction to «Digenesis in Charcot-Marie-Tooth Disease: Impact of Combined Mutations in the MFN2 and GDAP1 Genes » (2025). 30, e70045, doi:[10.1111/jns.70045](https://doi.org/10.1111/jns.70045).

Estévez-Arias, B., Sarv, S., Bonello-Palot, N., Carrera-García, L., Ortez, C., Expósito-Escudero, J., Yubero, D., Muchart, J., Delmont, E., Öiglane-Shlik, E., Meren, T., Puusepp, S., Murumets, Ü., Salomons, G. S., Udd, B., Väli, L., Cantarero, L., Bönnemann, C. G., Nascimento, A., Ramón-Maiques, S., Öunap, K., Hoenicka, J., Natera-de Benito, D. et Palau, F. (2025). **Biallelic Variants in the DARS2 Gene as a Novel Cause of Axonal Charcot-Marie-Tooth Disease.** Ann Neurol, doi:[10.1002/ana.78005](https://doi.org/10.1002/ana.78005).

Giovenale, A. M. G., Ferrone, I., Tomaselli, S., Mazzoni, M., Ruotolo, G., Turco, E. M., Torres, B., De Luca, A., Zanfardino, P., Vulcano, E., Ferrari, D., Damiani, D., Santorelli, F. M., Pennuto, M., Vescovi, A. L., Petruzzella, V. et Rosati, J. (2025). **Generation and characterization of a patient-derived iPSC line, CSSi022-A (15666), with a pathogenic MFN2 mutation causing Charcot-Marie-Tooth disease type 2A.** Stem Cell Res 88, 103817, doi:[10.1016/j.scr.2025.103817](https://doi.org/10.1016/j.scr.2025.103817).

Girolimetti, G., Marini, F., Calvani, R., Coelho-Júnior, H. J., Gervasoni, J., Santucci, L., Tozza, S., Manganelli, F., Saveri, P., Pareyson, D., Marzetti, E., Bucci, C., Picca, A. et Guerra, F. (2025). **Circulating mitochondrial components and metabolic and inflammatory markers in Charcot-Marie-Tooth type 2B.** Neurobiol Dis 214, 107051, doi:[10.1016/j.nbd.2025.107051](https://doi.org/10.1016/j.nbd.2025.107051).

Habib, S., Anadani, N. A. et Samkutty, D. (2025). **Co-occurrence of Charcot-Marie-Tooth disease type 2A and multiple sclerosis: A case report.** SAGE Open Med Case Rep 13, 2050313X251357782, doi:[10.1177/2050313X251357782](https://doi.org/10.1177/2050313X251357782).

Mitochondrial disorders : The Essential
MMITO-2025-3 from july 1st to september 30th, 2025

Joaquim, M., Bulimaga, M.-B., Mohn, M. A., Plouzenec, S., Osinski, L., Altin, S., Mahabir, E., Chevrollier, A. et Escobar-Henriques, M. (2025). **Charcot-Marie-Tooth type 2A variants of mitofusin 2 sensitize cells to apoptotic cell death.** J Cell Sci 138, jcs263691, doi:[10.1242/jcs.263691](https://doi.org/10.1242/jcs.263691).

Kim, J. W., Nam, S. H., Lee, G. S., Chung, H. Y., Kim, E. Y., Han, J. P., Jang, J.-H., Choi, B.-O. et Yeom, S. C. (2025). **Estradiol rescues male hydroxyl radical-mediated Charcot-Marie-Tooth 2Z by Morc2a stabilization through autophagy inhibition in a murine model.** Acta Neuropathol 150, 13, doi:[10.1007/s00401-025-02922-2](https://doi.org/10.1007/s00401-025-02922-2).

Liu, Y., Yan, C., Cao, B., Kong, D., Li, J., Li, W., Guo, Y., Yuan, Z., Gao, Y., Zhang, Y., Sui, R., Chen, G., Hao, X. et Chen, Q. (2025). **Modulating mitochondrial dynamics in CMT2A: a multifaceted platform for drug discovery and evaluation.** Biophys Rep 11, 143-155, doi:[10.52601/bpr.2024.240037](https://doi.org/10.52601/bpr.2024.240037).

Manini, A., Brusati, A., Grassano, M., Scacciatella, G., Peverelli, S., Spagliardi, J., Pensato, V., Doretti, A., Vasta, R., Manera, U., Canosa, A., Brunetti, M., Gentilini, D., Messina, S., Verde, F., Moglia, C., Morelli, C., Dalla Bella, E., Keagle, P. J., Landers, J. E., Gellera, C., Lauria Pinter, G., Chiò, A., Ratti, A., Calvo, A., Silani, V. et Ticozzi, N. (2025). **Whole genome sequencing analysis in primary lateral sclerosis (PLS) patients reveals mutations in neurological diseases-causing genes.** J Neurol 272, 587, doi:[10.1007/s00415-025-13328-1](https://doi.org/10.1007/s00415-025-13328-1).

Ravanbod, M., Mohammadi, M., Ghasemi, A., Jabbarzadeh, S., Okhovat, A. A., Khani, M., Elahi, E., Ramezani, M., Nafissi, S. et Alavi, A. (2025). **GDAP1-Related Charcot-Marie-Tooth Disease: Axonal or Demyelinating Subtype? Autosomal Recessive or Autosomal Dominant Inheritance?** J Peripher Nerv Syst 30, e70046, doi:[10.1111/jns.70046](https://doi.org/10.1111/jns.70046).

Shumeri, E., Mandorah, E., Martini, N., Boyer, A., Halbert, C., Puma, A., Chaussenot, A., Delmont, E., N'guyen, K., Attarian, S. et Bonello-Palot, N. (2025). **Digenesis in Charcot-Marie-Tooth Disease: Impact of Combined Mutations in the MFN2 and GDAP1 Genes.** J Peripher Nerv Syst 30, e70044, doi:[10.1111/jns.70044](https://doi.org/10.1111/jns.70044).

Zanfardino, P., Amati, A., Doccini, S., Girolamo, F., Tullo, A., Longo, G., Santorelli, F. M. et Petruzzella, V. (2025). **Selective mitophagy activation and protein aggregate accumulation in MTMR5/SBF1-deficient fibroblasts.** Life Sci 381, 123995, doi:[10.1016/j.lfs.2025.123995](https://doi.org/10.1016/j.lfs.2025.123995).

Zhang, L., Pouya, A., Kopetzky, J., Bitar, S., Wolf, C., Bello, F. D., Gomez-Zepeda, D., Tenzer, S. et Methner, A. (2025). **Increased BNIP3-mediated mitophagy attenuates GDAP1 loss of function - implications for Charcot-Marie-Tooth disease 4A.** Neurobiol Dis 213, 107019, doi:[10.1016/j.nbd.2025.107019](https://doi.org/10.1016/j.nbd.2025.107019).

Mitochondrial disorders : The Essential
MMITO-2025-3 from July 1st to September 30th, 2025

Maladies liées au gène ACO2 - ACO2-related disorders

Pas de résultat ce trimestre

Maladie liée au gène GFM1 – GFM1-related disorder

Pas de résultat ce trimestre

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

Bérat, C.-M., Hully, M., Rötig, A., Barcia, G., Assouline, Z., Abi-Warde, M.-T., Barnerias, C., Payen, E., Jaroussie, M., Gaignard, P., Lebigot, E., Roubertie, A., Boddaert, N., Roux, C.-J., de Lonlay, P., Desguerre, I., Munnich, A., Schiff, M. et Gitiaux, C. (2025). **Childhood POLG-related disorders: Focus on polyradiculoneuropathy**. *Mol Genet Metab* 146, 109213, doi:[10.1016/j.ymgme.2025.109213](https://doi.org/10.1016/j.ymgme.2025.109213).

Jha, S. et Jog, M. (2025). **Late Adulthood Onset POLG Disease Presenting as Spastic Paraplegia: A Pathophysiological Continuum?** *Acta Neurol Belg*, doi:[10.1007/s13760-025-02878-w](https://doi.org/10.1007/s13760-025-02878-w).

Rossi, V., Brooks, D., Dai, H., Mizerik, E., Salazar, K., Davila-Williams, D., Ben-Moshe, Y., Lalani, S. R., Elsea, S. H., Gijavanekar, C., Scott, D. A., Machol, K., Bekheirnia, M. R. et Scaglia, F. (2025). **A Rare Molecular Diagnosis in a Patient With Hepatocerebral Syndrome Contributes to the Expansion of the Phenotypic Spectrum of POLG2 - Related Mitochondrial Disorder**. *Am J Med Genet A* 197, e64177, doi:[10.1002/ajmg.a.64177](https://doi.org/10.1002/ajmg.a.64177).

Sangeeth, T. A., Viswanathan, L. G., Dv, S., Rao, S., Nagappa, M., Saini, J. et Sinha, S. (2025). **Bilateral epilepsy partialis continua in POLG related mitochondrial disease**. *Seizure* 131, 200-202, doi:[10.1016/j.seizure.2025.07.004](https://doi.org/10.1016/j.seizure.2025.07.004).

Zioudi, A., Gouiza, I., Galai, S., Hechmi, M., Klaa, H., Miladi, Z., Ben Younes, T., Benrhouma, H., Ben Youssef-Turki, I., Omar, S., Lenaers, G., Kefi, R., Gouider-Khouja, N. et Kraoua, I. (2025). **A Tunisian POLG mutation expands the clinical spectrum of POLG-related disorders**. *Mitochondrion* 85, 102071, doi:[10.1016/j.mito.2025.102071](https://doi.org/10.1016/j.mito.2025.102071).

Mitochondrial disorders : The Essential
MMITO-2025-3 from july 1st to september 30th, 2025

MELAS

Burg, L., Yoon, H., Peng, M., Germano, P., Reese Gretzmacher, E., Xiao, R., Anderson, V. E., Nakamaru-Ogiso, E. et Falk, M. J. (2025). **Zagociguat prevented stressor-induced neuromuscular dysfunction, improved mitochondrial physiology, and increased exercise capacity in diverse mitochondrial respiratory chain disease zebrafish models.** Front Pharmacol 16, 1588426, doi:[10.3389/fphar.2025.1588426](https://doi.org/10.3389/fphar.2025.1588426).

Dumbuya, J. S., Tian, C., Deng, L., Ahmad, B., Chen, X. et Lu, J. (2025). **Clinical features, disease burden and impact on quality of life in participants with mitochondrial encephalomyopathy.** Front Neurol 16, 1585906, doi:[10.3389/fneur.2025.1585906](https://doi.org/10.3389/fneur.2025.1585906).

Finsterer, Josef (2025a). **Autosomal recessive Alport syndrome should be diagnosed in an m.3243A>G carrier only if a pathogenic COL4A3 variant has been detected.** Intern Med, doi:[10.2169/internalmedicine.6230-25](https://doi.org/10.2169/internalmedicine.6230-25).

Finsterer, Josef (2025b). **Comments on: « Stroke-Like Episode, Aphasia, and Hearing Loss in MELAS ».** J Clin Neurol 21, 470-471, doi:[10.3988/jcn.2025.0239](https://doi.org/10.3988/jcn.2025.0239).

Finsterer, Josef (2025c). **Neuroimaging in Mitochondrial Encephalomyopathy with Lactic Acidosis with Stroke-Like Episodes (MELAS): Correspondence.** Indian J Pediatr, doi:[10.1007/s12098-025-05742-0](https://doi.org/10.1007/s12098-025-05742-0).

Finsterer, J. (2025). **The spectrum of ophthalmologic abnormalities in MELAS is broader than expected.** Arch Soc Esp Oftalmol (Engl Ed) S2173-5794(25)00135-5, doi:[10.1016/j.oftale.2025.08.001](https://doi.org/10.1016/j.oftale.2025.08.001).

Gillespie, H., Ng, Y. S., Wood, K. M., Hopton, S., Alston, C. L., Blakely, E. L., Thompson, N., Taylor, R. W., Browning, A. C., McFarland, R. et Sayer, J. A. (2025). **Severe clinical manifestation of mitochondrial disease due to the m.3243A>T variant: a case report of early-onset, multi-organ involvement and premature death.** J Rare Dis (Berlin) 4, 47, doi:[10.1007/s44162-025-00110-0](https://doi.org/10.1007/s44162-025-00110-0).

Huang, X., Pan, C.-H., Yin, F., Peng, J. et Yang, L. (2025). **The Role of Estrogen in Mitochondrial Disease.** Cell Mol Neurobiol 45, 68, doi:[10.1007/s10571-025-01592-8](https://doi.org/10.1007/s10571-025-01592-8).

Ito, Y., Ochi, C., Yamanishi, Y., Takashima, H., Hashiguchi, A. et Nagai, M. (2025). **[A case of recurrent cerebellitis leading to the diagnosis of mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes (MELAS)].** Rinsho Shinkeigaku 65, 601-606, doi:[10.5692/clinicalneuro.2025-002117](https://doi.org/10.5692/clinicalneuro.2025-002117).

Li, Y., Ding, Y., Jin, E. et Yin, H. (2025). **Case Report: Abnormal pupils caused by the mitochondrial MT-TL1 gene m.3243A>G mutation.** Front Pediatr 13, 1573886, doi:[10.3389/fped.2025.1573886](https://doi.org/10.3389/fped.2025.1573886).



Mitochondrial disorders : The Essential

MMITO-2025-3 from july 1st to september 30th, 2025

Liao, Y., Xu, D. et Li, L. (2025). **A case of mitochondrial encephalomyopathy remarkable presenting with refractory shock.** Zhong Nan Da Xue Xue Bao Yi Xue Ban 50, 511-516, doi:[10.11817/j.issn.1672-7347.2025.240250](https://doi.org/10.11817/j.issn.1672-7347.2025.240250).

Liu, J. Y., Koenig, M. K., Riggs-Harpur, K. et Numan, M. T. (2025). **Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-Like Episodes (MELAS) Patients with m.3243A > G have High Prevalence of Wolff-Parkinson-White Syndrome.** Pediatr Cardiol, doi:[10.1007/s00246-025-03985-4](https://doi.org/10.1007/s00246-025-03985-4).

Liu, X.-H., Shen, X. et Zhong, Y.-S. (2025). **Retinal multimodal-imaging and functional tests in a mitochondrial disease with focal and segmental glomerulosclerosis.** Int J Ophthalmol 18, 1770-1776, doi:[10.18240/ijo.2025.09.19](https://doi.org/10.18240/ijo.2025.09.19).

Lubotzky, A. (2025). **Re: Comments on « Stroke-Like Episode, Aphasia, and Hearing Loss in MELAS ».** J Clin Neurol 21, 472-473, doi:[10.3988/jcn.2025.0246](https://doi.org/10.3988/jcn.2025.0246).

McCully, K. K., Bossie, H. M. et Kendall, F. D. (2024). **Noninvasive Assessments of Mitochondrial Capacity in People with Mitochondrial Myopathies.** Muscles 3, 393-403, doi:[10.3390/muscles3040033](https://doi.org/10.3390/muscles3040033).

Meldau, S., McCormick, E. M., George-Sankoh, I., Riordan, G. T., Khan, K., MacMullen, L. E., Dawlat, S., Blackhurst, D., Falk, M. J. et Elson, J. L. (2025). **Mitochondrial DNA Pathogenic Variant Prevalence in Primary Mitochondrial Disease Patients With African (L) Mitochondrial Genome Haplogroups.** JIMD Rep 66, e70036, doi:[10.1002/jmd2.70036](https://doi.org/10.1002/jmd2.70036).

Miwa, T., Hashimoto, K., Seto, T. et Sakamoto, H. (2025). **Mitochondrial Hearing Loss: Genetic Variants and Clinical Progression.** Cureus 17, e85825, doi:[10.7759/cureus.85825](https://doi.org/10.7759/cureus.85825).

Nakashima, D., Tosaki, T., Sasaki, T., Honda, Y., Yokote, S., Mori, T., Sohara, E., Uchida, S., Tsuboi, N. et Yokoo, T. (2025). **Combined Alport Syndrome Type 3A and Mitochondrial Disease Presenting with a Thin Base Membrane and Overt Albuminuria: A Case Report.** Intern Med, doi:[10.2169/internalmedicine.5838-25](https://doi.org/10.2169/internalmedicine.5838-25).

Pisano, A., Belloli, S., Pignataro, M. G., Rainone, P., Valtorta, S., Coliva, A., de Turreis, V., Mosca, L., Di Micco, P., Moresco, R. M., d'Amati, G. et Morea, V. (2025). **Peptide-mimetics derived from leucyl-tRNA synthetase are potential agents for the therapy of mt-tRNA related diseases.** Front Pharmacol 16, 1607343, doi:[10.3389/fphar.2025.1607343](https://doi.org/10.3389/fphar.2025.1607343).

Mitochondrial disorders : The Essential

MMITO-2025-3 from july 1st to september 30th, 2025

Preston, G., Jacob, N., Elsharkawi, I., Morava, E. et Kozicz, T. (2025). **Phosphodiesterase type 5 inhibition as a therapeutic strategy in primary mitochondrial disease: Evidence from patient fibroblasts and clinical observations.** *Mol Genet Metab* 146, 109197, doi:[10.1016/j.ymgme.2025.109197](https://doi.org/10.1016/j.ymgme.2025.109197).

Purandare, N., Pasupathi, V., Xi, Y., Rajan, V., Vegh, C., Firestine, S., Kozicz, T., Fribley, A. M., Grossman, L. I. et Aras, S. (2025). **Pseudohypoxia-Stabilized HIF2 α Transcriptionally Inhibits MNRR1, a Druggable Target in MELAS.** *Cells* 14, 1078, doi:[10.3390/cells14141078](https://doi.org/10.3390/cells14141078).

Sane, I. A. et Gupte, J. R. (2025). **Diagnosing Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-Like Episodes (MELAS) Syndrome in a Young Adult Female Patient With Seizures and Lactic Acidosis.** *Cureus* 17, e88031, doi:[10.7759/cureus.88031](https://doi.org/10.7759/cureus.88031).

Sasaki, T., Nakashima, D. et Tosaki, T. (2025). **Response to « Diagnose autosomal recessive Alport syndrome in an m.3243A>G carrier only if a pathogenic COL4A3 variant has been detected ».** *Intern Med*, doi:[10.2169/internalmedicine.6277-25](https://doi.org/10.2169/internalmedicine.6277-25).

Scarabello, A., Mason, F., Ferri, L., Baccari, F., Maresca, A., Morgia, C. L., Belotti, L. M. B., Licchetta, L., Bisulli, F., Carelli, V. et Vito, L. D. (2025). **Progressive encephalopathy in m.3243A > G/MT-TL1 mutation carriers: a quantitative EEG analysis.** *Clin Neurophysiol* 177, 2110822, doi:[10.1016/j.clinph.2025.2110822](https://doi.org/10.1016/j.clinph.2025.2110822).

Sporn, K., Kumar, R., Marla, K., Ravi, P., Vaja, S., Paladugu, P., Zaman, N. et Tavakkoli, A. (2025). **Epigenetic Profiling of Cell-Free DNA in Cerebrospinal Fluid: A Novel Biomarker Approach for Metabolic Brain Diseases.** *Life (Basel)* 15, 1181, doi:[10.3390/life15081181](https://doi.org/10.3390/life15081181).

Tiwari, S., Gunasekaran, P. K. et Saini, L. (2025). **Neuroimaging in Mitochondrial Encephalomyopathy with Lactic Acidosis and Stroke-Like Episodes (MELAS): Authors' Reply.** *Indian J Pediatr*, doi:[10.1007/s12098-025-05743-z](https://doi.org/10.1007/s12098-025-05743-z).

Ueda, M., Tanaka, N., Momota, Y. et Kawaguchi, M. (2025). **Anesthetic Management With Remimazolam for Adolescent Mitochondrial Encephalomyopathy With Lactic Acidosis and Stroke-like Episodes (MELAS): A Case Report.** *Anesth Prog* 72, 46-48, doi:[10.2344/23-0051](https://doi.org/10.2344/23-0051).

Wang, L., Zhou, T., Bu, X., Pan, D. et Liu, X. (2025). **Hypoparathyroidism in a Child with MELAS Syndrome: A Case Report of Severe Lactic Acidosis and Symmetrical Bilateral Basal Ganglia Calcification.** *Int J Endocrinol Metab* 23, e161585, doi:[10.5812/ijem-161585](https://doi.org/10.5812/ijem-161585).

Mitochondrial disorders : The Essential

MMITO-2025-3 from july 1st to september 30th, 2025

Winqvist, S., Kärppä, M., Moilanen, J. S. et Majamaa, K. (2025). **Cognitive impairment profile in patients with the m.3243A> G variant in mitochondrial DNA**. BMC Neurol 25, 316, doi:[10.1186/s12883-025-04325-y](https://doi.org/10.1186/s12883-025-04325-y).

Zhao, Y., Chen, D., Chen, H., Fan, R., Xu, W. et Jiang, H. (2025). **A review of the link between the lactate-GPR81 axis and mitochondrial angiopathy in MELAS based on imaging characteristics**. J Neurol 272, 572, doi:[10.1007/s00415-025-13318-3](https://doi.org/10.1007/s00415-025-13318-3).

MERFF

Pisano, A., Belloli, S., Pignataro, M. G., Rainone, P., Valtorta, S., Coliva, A., de Turris, V., Mosca, L., Di Micco, P., Moresco, R. M., d'Amati, G. et Morea, V. (2025). **Peptide-mimetics derived from leucyl-tRNA synthetase are potential agents for the therapy of mt-tRNA related diseases**. Front Pharmacol 16, 1607343, doi:[10.3389/fphar.2025.1607343](https://doi.org/10.3389/fphar.2025.1607343).

Watson-Fargie, T., Anderson, D. G., Stewart, W., Longman, C., Hopton, S., Ng, Y. S., Blakely, E. L., Taylor, R. W. et Farrugia, M. E. (2025). **Selective muscle MRI changes in a patient with a rare mitochondrial DNA variant causing myoclonic epilepsy with ragged red fibres**. Neuromuscul Disord 54, 106204, doi:[10.1016/j.nmd.2025.106204](https://doi.org/10.1016/j.nmd.2025.106204).

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

Ali, L., Hazzard, I., Tehrani, N. S., Ansari, U., Ali, A., Kumar, P., Ali, N., Gakhal, G. et Lui, F. (2025). **Extraocular features of Leber hereditary optic neuropathy: A scoping review**. J Biol Methods 12, e99010055, doi:[10.14440/jbm.2024.0113](https://doi.org/10.14440/jbm.2024.0113).

Amore, G., Carbonelli, M., D'Angeli, D., Bonan, L., Faustini-Fustini, M., Maresca, A., Carelli, V. et La Morgia, C. (2025). **Late-onset Leber's hereditary optic neuropathy and antiandrogens for prostate cancer: is there a causative link?** Front Neurol 16, 1616992, doi:[10.3389/fneur.2025.1616992](https://doi.org/10.3389/fneur.2025.1616992).

Aoshima, K., Takagi, Y., Funato, M., Kuse, Y., Nakamura, S. et Shimazawa, M. (2025). **Establishment of human Leber's hereditary optic neuropathy model using iPSC-derived retinal organoids**. Front Cell Neurosci 19, 1635775, doi:[10.3389/fncel.2025.1635775](https://doi.org/10.3389/fncel.2025.1635775).

Ashour, A. M., Harbi, M. H., Alshehri, F. S., Wali, S. M., Aldurdunji, M. M. et Alorfi, N. M. (2025). **Clinical Research for Inherited Retinal Disease Related Pediatric Blindness: A Preliminary Descriptive Analysis Based on ClinicalTrials.gov**. J Multidiscip Healthc 18, 5865-5874, doi:[10.2147/JMDH.S539699](https://doi.org/10.2147/JMDH.S539699).

Mitochondrial disorders : The Essential
MMITO-2025-3 from July 1st to September 30th, 2025

Britton, J. O. T., Yu-Wai-Man, P. et Chen, B. S. (2025). **Technological advances in the diagnosis and management of inherited optic neuropathies**. Front Neurol 16, 1609033, doi:[10.3389/fneur.2025.1609033](https://doi.org/10.3389/fneur.2025.1609033).

Cayenne, S. A., Naser, M., Jaiswal, S., Arogundade, E., Mortensen, P. et Lee, A. G. (2025). **An Additional Case of Leber Hereditary Optic Neuropathy With G9804A Mutation**. J Neuroophthalmol, doi:[10.1097/WNO.0000000000002397](https://doi.org/10.1097/WNO.0000000000002397).

Chapelle, A.-C. (2025). **Anterograde degeneration along the visual pathway following optic nerve injury: a review**. Front Neurol 16, 1623798, doi:[10.3389/fneur.2025.1623798](https://doi.org/10.3389/fneur.2025.1623798).

Chen, B. S., Perot, S., Taiel, M., Yu-Wai-Man, P., Horton, M., et LHON Study Group (2025a). **Rasch analysis of the NEI-VFQ-25: vision-related quality of life in Leber hereditary optic neuropathy after lenadogene nolparvovec gene therapy**. BMJ Open Ophthalmol 10, e002164, doi:[10.1136/bmjophth-2025-002164](https://doi.org/10.1136/bmjophth-2025-002164).

Chen, B. S., Seikus, C., Ferguson, J., Tadić, V., Horton, M., Yu-Wai-Man, P. et Archer, S. (2025b). « **Adrift From the World** »: Exploring the Lived Experiences of Individuals Affected by an Inherited Optic Neuropathy in the United Kingdom-A Qualitative Study. Value Health S1098-3015(25)02490-8, doi:[10.1016/j.jval.2025.07.023](https://doi.org/10.1016/j.jval.2025.07.023).

Chiang, M.-R., Chen, C.-Y., Chang, Y.-H., Yang, Y.-P., Chien, Y., Hu, J.-L., Pan, W.-C., Lao, H. M., Lien, H.-W., Lin, T.-C., Chen, S.-J., Tsai, S., Hu, S.-H. et Chiou, S.-H. (2025). **In Vivo Reprogramming Dysfunctional Retinal Ganglion Cells and Visual-phototransduction via Wireless Charging Nanogold for Leber's Hereditary Optic Neuropathy**. Adv Mater e04509, doi:[10.1002/adma.202504509](https://doi.org/10.1002/adma.202504509).

Elmaseh, S. A., Gauthier, D. A., Golmohammadi, M., Pargalava, N., Carelli, V. et Sadun, A. A. (2025). **Metformin may alter the course of Leber's hereditary optic neuropathy: a case report**. Front Med (Lausanne) 12, 1609941, doi:[10.3389/fmed.2025.1609941](https://doi.org/10.3389/fmed.2025.1609941).

García Gómez, E., San-Juan, D., Sandoval Luna, L. et Morales Morales, M. A. (2025). **Leber Hereditary Optic Neuropathy and Epilepsy in a Mexican Patient**. Cureus 17, e86663, doi:[10.7759/cureus.86663](https://doi.org/10.7759/cureus.86663).

Ha, D. H. et Kim, U. S. (2025). **Large language models in neuro-ophthalmology diseases: ChatGPT vs Bard vs Bing**. Int J Ophthalmol 18, 1231-1236, doi:[10.18240/ijo.2025.07.05](https://doi.org/10.18240/ijo.2025.07.05).



Mitochondrial disorders : The Essential

MMITO-2025-3 from july 1st to september 30th, 2025

Hedges, T. R., Dombrovsky, D., Onwuka, O., Vuong, L. N., Martin-Paez, Y. et Mendoza-Santiesteban, C. (2025). **Initial Macular Ganglion Cell Changes During Conversion of Leber Hereditary Optic Neuropathy.** J Neuroophthalmol, doi:[10.1097/WNO.0000000000002384](https://doi.org/10.1097/WNO.0000000000002384).

Hilali, Z., El Korno, O., Laarif, Y., Boutimzine, N. et Cherkaoui, L. O. (2025). **Acute Bilateral Vision Loss in a Young Male: A Case of Leber's Hereditary Optic Neuropathy.** Cureus 17, e86607, doi:[10.7759/cureus.86607](https://doi.org/10.7759/cureus.86607).

Hu, C., Su, T., Zhang, Y., Yang, J., Su, X., Zhang, Z., Wang, X., Zou, W., Liang, D., Liu, Y., Ji, D. et Cao, Y. (2025). **Estradiol alleviates disease phenotypes caused by m.3635G > a mutations by activating mitochondrial biogenesis and PINK1-Parkin mediated mitophagy in iPSC-derived retinal pigment epithelium cells.** Cell Signal 136, 112121, doi:[10.1016/j.cellsig.2025.112121](https://doi.org/10.1016/j.cellsig.2025.112121).

Huang, X., Pan, C.-H., Yin, F., Peng, J. et Yang, L. (2025). **The Role of Estrogen in Mitochondrial Disease.** Cell Mol Neurobiol 45, 68, doi:[10.1007/s10571-025-01592-8](https://doi.org/10.1007/s10571-025-01592-8).

Idebenone for treating visual impairment in Leber's hereditary optic neuropathy in people 12 years and over (2025). National Institute for Health and Care Excellence (NICE), London, URL: <http://www.ncbi.nlm.nih.gov/books/NBK618262/>.

Iorga, R. E., Moraru, A. D., Munteanu-Dănulescu, R. S., Urdea, D. et Danielescu, C. (2025). **Evaluation of Visual and Optical Coherence Tomography Outcomes in Patients with Leber's Hereditary Optic Neuropathy Treated with Idebenone.** Life (Basel) 15, 1172, doi:[10.3390/life15081172](https://doi.org/10.3390/life15081172).

Jia, J., Wang, H., Wang, W., Xie, P., Li, P., Jiang, P., Xie, W., Liu, L. et Wang, G. (2025). **Developing Intravenous Delivery of Water-Soluble Prodrugs of Idebenone for the Treatment of Acute Ischemic Stroke.** ACS Chem Neurosci 16, 3541-3553, doi:[10.1021/acscchemneuro.5c00340](https://doi.org/10.1021/acscchemneuro.5c00340).

Jiang, L., Guo, S., Li, B., Xiao, S., Wang, F. et Wei, W. (2025). **Efficacy and safety of intravitreal rAAV2-ND4 therapy for Leber's hereditary optic neuropathy.** Eye (Lond), doi:[10.1038/s41433-025-03972-2](https://doi.org/10.1038/s41433-025-03972-2).

Kim, H., Yang, H. K., Seong, M.-W. et Hwang, J.-M. (2025). **Clinical features of Leber's hereditary optic neuropathy carrying a rare m.13051G>A mtDNA mutation.** Korean J Ophthalmol, doi:[10.3341/kjo.2025.0086](https://doi.org/10.3341/kjo.2025.0086).

Mitochondrial disorders : The Essential
MMITO-2025-3 from july 1st to september 30th, 2025

Li, Z., Liu, H., Wu, Q., Huang, Z., Jiang, L. et Chen, F. K. (2025). **Clinical Characteristics of Myelin Oligodendrocyte Glycoprotein Antibody-Associated Optic Neuritis in Carriers of a Leber Hereditary Optic Neuropathy Variant.** J Neuroophthalmol, doi:[10.1097/WNO.0000000000002366](https://doi.org/10.1097/WNO.0000000000002366).

Meldau, S., McCormick, E. M., George-Sankoh, I., Riordan, G. T., Khan, K., MacMullen, L. E., Dawlat, S., Blackhurst, D., Falk, M. J. et Elson, J. L. (2025). **Mitochondrial DNA Pathogenic Variant Prevalence in Primary Mitochondrial Disease Patients With African (L) Mitochondrial Genome Haplogroups.** JIMD Rep 66, e70036, doi:[10.1002/jmd2.70036](https://doi.org/10.1002/jmd2.70036).

Orssaud, C. et Reynier, P. (2025). **MetabOCT: a clinical trial looking for a metabolomic signature predicting the onset of Leber's hereditary optic neuropathy in healthy MtDNA mutations carriers.** Metabolomics 21, 134, doi:[10.1007/s11306-025-02328-x](https://doi.org/10.1007/s11306-025-02328-x).

Popovic-Beganovic, A. et Dzinic, V. (2025). **Leber's Hereditary Optic Neuropathy.** Med Arch 79, 241-248, doi:[10.5455/medarh.2025.79.241-248](https://doi.org/10.5455/medarh.2025.79.241-248).

Portaro, G., Cavallieri, F., Amore, G., Carbonelli, M., Pelloni, S., Cavallieri, G., Carelli, V., Valzania, F. et Morgia, C. L. (2025). **A Case of Late-Onset Leber's Hereditary Optic Neuropathy in Association with Heteroplasmic m.11778G>A/ND4 Mutation.** Neuroophthalmology 49, 397-401, doi:[10.1080/01658107.2024.2440398](https://doi.org/10.1080/01658107.2024.2440398).

Ribeiro, P. V. Z., Pari Mitre, L., Gauza, M. M., Furukawa, P. H., Ferraz, V. E. D. F. et Gomy, I. (2025). **Therapeutic benefit of idebenone in Leber hereditary optic neuropathy: a systematic review and meta-analysis.** Ophthalmic Genet 1-6, doi:[10.1080/13816810.2025.2521647](https://doi.org/10.1080/13816810.2025.2521647).

Sanders, F. W. B. et Votruba, M. (2025). **Outcomes of idebenone therapy for Leber hereditary optic neuropathy in a cohort of patients from Wales.** Eye (Lond), doi:[10.1038/s41433-025-03993-x](https://doi.org/10.1038/s41433-025-03993-x).

Sereikaite, L., Vilkeviciute, A., Glebauskiene, B., Traberg, R., Gelzinis, A., Piskiniene, R., Zemaitiene, R., Ugenskiene, R. et Liutkeviciene, R. (2025). **First Case in Lithuania of an Autosomal Recessive Mutation in the DNAJC30 Gene as a Cause of Leber's Hereditary Optic Neuropathy.** Genes (Basel) 16, 993, doi:[10.3390/genes16090993](https://doi.org/10.3390/genes16090993).

Sergott, R. C., Carelli, V., Newman, N. J., Biousse, V., Yu-Wai-Man, P., Vignal-Clermont, C., Josse, C., Taiel, M., Sahel, J.-A., Barboni, P., et LHON Study Group (2025). **Predictors of Final Visual Outcome in Patients With Leber Hereditary Optic Neuropathy Treated With Lenadogene Nolpharvovec Gene Therapy.** Invest Ophthalmol Vis Sci 66, 42, doi:[10.1167/iovs.66.9.42](https://doi.org/10.1167/iovs.66.9.42).

Mitochondrial disorders : The Essential
MMITO-2025-3 from July 1st to September 30th, 2025

Sherratt-Mayhew, S., Page, C., Berman, G. et Mollan, S. P. (2025a). **Infographic: landmark trials in neuro-ophthalmology - results of the REVERSE trial for unilateral gene therapy rAAV2/2-ND4 in Leber hereditary optic neuropathy.** Eye (Lond), doi:[10.1038/s41433-025-03932-w](https://doi.org/10.1038/s41433-025-03932-w).

Sherratt-Mayhew, S., Page, C., Lowe, M., Labella Alvarez, F., Mollan, S. P. et Berman, G. (2025b). **Infographic: therapeutic benefit of idebenone in patients with Leber hereditary optic neuropathy: the LEROS nonrandomized controlled trial.** Eye (Lond), doi:[10.1038/s41433-025-03991-z](https://doi.org/10.1038/s41433-025-03991-z).

Sherratt-Mayhew, S., Page, C., Lowe, M., Mollan, S. P. et Berman, G. (2025c). **Infographic: Efficacy and safety of intravitreal gene therapy for Leber hereditary optic neuropathy treated within 6 months of disease onset (RESCUE Trial).** Eye (Lond), doi:[10.1038/s41433-025-03950-8](https://doi.org/10.1038/s41433-025-03950-8).

Srilekha, S., Ambika, S., Hemavathy, N., Vidhya, D. et Yu-Wai-Man, P. (2025). **Whole mitochondrial genome sequencing in individuals with Leber hereditary optic neuropathy negative for the common pathogenic mitochondrial DNA variants.** Front Neurol 16, 1584748, doi:[10.3389/fneur.2025.1584748](https://doi.org/10.3389/fneur.2025.1584748).

Takai, Y., Yamagami, A., Iwasa, M., Inoue, K., Yasumoto, R., Ishikawa, H. et Wakakura, M. (2025a). **Age-Associated Differences in Optic Disc Findings of Leber's Hereditary Optic Neuropathy.** Neuroophthalmology 49, 359-365, doi:[10.1080/01658107.2025.2501053](https://doi.org/10.1080/01658107.2025.2501053).

Takai, Y., Yamagami, A., Iwasa, M., Inoue, K., Yasumoto, R., Ishikawa, H. et Wakakura, M. (2025b). **Optic nerve MRI findings in Leber's hereditary optic neuropathy.** Jpn J Ophthalmol, doi:[10.1007/s10384-025-01246-8](https://doi.org/10.1007/s10384-025-01246-8).

Trinquet, L., Ajasse, S., Chavane, F., Legras, R., Matonti, F., Sahel, J.-A., Vignal-Clermont, C. et Lorenceau, J. (2025). **Uncovering the Characteristics of Pupil Cycle Time (PCT) in Neuropathies and Retinopathies.** Vision (Basel) 9, 51, doi:[10.3390/vision9030051](https://doi.org/10.3390/vision9030051).

Xhuti, D., Chiarot, A., Minhas, M., Tobia, S., de Maat, N., Manta, K., Ng, S. Y., Tarnopolsky, M. A. et Nederveen, J. P. (2025). **Combination treatment with antioxidants and creatine alleviates common and variant-specific mitochondrial impairments in Leber's hereditary optic neuropathy patient-derived fibroblasts.** Hum Mol Genet 34, 1780-1795, doi:[10.1093/hmg/ddaf125](https://doi.org/10.1093/hmg/ddaf125).

Xiang, J., Xu, W., Wu, J., Luo, Y., Liu, C., Hou, Y., Chen, J. et Yang, B. (2025). **Structural insights into DdCBE in action enable high-precision mitochondrial DNA editing.** Mol Cell 85, 3357-3372.e9, doi:[10.1016/j.molcel.2025.08.016](https://doi.org/10.1016/j.molcel.2025.08.016).

Mitochondrial disorders : The Essential
MMITO-2025-3 from july 1st to september 30th, 2025

Yasheng, M., Ji, Y., He, Y., Yi, Q., Zhang, H., Shan, W., Wang, K., Zhang, J., Li, Y., Meng, F., Zhang, M., Mo, J. Q., Wei, S. et Guan, M.-X. (2025). **Leber's hereditary optic neuropathy-associated ND1 3733G>C mutation ameliorates the mitochondrial quality control and cellular homeostasis.** J Biol Chem 301, 110464, doi:[10.1016/j.jbc.2025.110464](https://doi.org/10.1016/j.jbc.2025.110464).

Zhao, Q., Ma, Y., Zhou, X., Wang, R., Yang, W., Peng, X. et Wei, W. (2025). **Quantitative assessment of retinal microvasculature using optical coherence tomography angiography and correlation with visual acuity in leber's hereditary optic neuropathy.** Int Ophthalmol 45, 376, doi:[10.1007/s10792-025-03619-x](https://doi.org/10.1007/s10792-025-03619-x).

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

Erdinest, N., Shemesh, N., London, N., Landau, D. et Lavy, I. (2025). **Chronic Progressive External Ophthalmoplegia (CPEO): Rehabilitation utilizing scleral contact lenses.** Am J Ophthalmol Case Rep 39, 102411, doi:[10.1016/j.ajoc.2025.102411](https://doi.org/10.1016/j.ajoc.2025.102411).

Karaa, A., Goldstein, A., Cohen, B. H., Haas, R. H., Vockley, J., Gorman, G. S., Mancuso, M., et RePOWER, MMPOWER-3, and MOTOR investigators (2025). **RePOWER: An International, Prospective, Non-Interventional Registry of Patients With Primary Mitochondrial Myopathy.** Clin Genet, doi:[10.1111/cge.70026](https://doi.org/10.1111/cge.70026).

Lopergolo, D., Berti, G., Gallus, G. N., Bianchi, S., Santorelli, F. M., Malandrini, A. et De Stefano, N. (2025). **The First Heterozygous TWNK Nonsense Mutation Associated with Progressive External Ophthalmoplegia: Evidence for a New Piece in the Puzzle of Mitochondrial Diseases.** Biomolecules 15, 1337, doi:[10.3390/biom15091337](https://doi.org/10.3390/biom15091337).

MacMullen, L. E., Reynolds, E., Weis, M., George-Sankoh, I., Nguyen, S., Stanley, K. D., Redko, M., Poblete, M., Goldstein, A. C. et Ganetzky, R. D. (2025). **Single large-scale mitochondrial DNA deletion syndromes: scientific and family conference optimizes the collection of rare disease research outcomes.** Orphanet J Rare Dis 20, 399, doi:[10.1186/s13023-025-03632-4](https://doi.org/10.1186/s13023-025-03632-4).

Manini, A., Brusati, A., Grassano, M., Scacciatella, G., Peverelli, S., Spagliardi, J., Pensato, V., Doretto, A., Vasta, R., Manera, U., Canosa, A., Brunetti, M., Gentilini, D., Messina, S., Verde, F., Moglia, C., Morelli, C., Dalla Bella, E., Keagle, P. J., Landers, J. E., Gellera, C., Lauria Pinter, G., Chiò, A., Ratti, A., Calvo, A., Silani, V. et Ticozzi, N. (2025). **Whole genome sequencing analysis in primary lateral sclerosis (PLS) patients reveals mutations in neurological diseases-causing genes.** J Neurol 272, 587, doi:[10.1007/s00415-025-13328-1](https://doi.org/10.1007/s00415-025-13328-1).



Mitochondrial disorders : The Essential

MMITO-2025-3 from july 1st to september 30th, 2025

Mendel, R. M., Matsuno, S., Lu, A., Falk, M. J. et Haroon, S. (2024). **Mitophagy modulation rescues single large-scale mitochondrial DNA deletion (SLSMD) disease symptoms in the C. elegans uaDf5 animal model.** bioRxiv 2024.10.27.620333, doi:[10.1101/2024.10.27.620333](https://doi.org/10.1101/2024.10.27.620333).

Pradhan, S., Mito, T., Khan, N. A., Olander, S., Zhaivoron, A., McWilliams, T. G. et Suomalainen, A. (2025). **PDK4 and nutrient responses explain muscle specific manifestation in mitochondrial disease.** Clin Transl Med 15, e70404, doi:[10.1002/ctm2.70404](https://doi.org/10.1002/ctm2.70404).

Sobeh, T., Granek, T., Bar-Yosef, O., Jacoby, E., Hoffmann, C. et Shrot, S. (2025). **Neuroimaging characteristics of single Large-Scale mitochondrial DNA deletion syndromes.** Neuroradiology, doi:[10.1007/s00234-025-03689-9](https://doi.org/10.1007/s00234-025-03689-9).

Ueda, N. K., Mimaki, M., Ito, S., Murakami, A., Yokoi, S., Nishino, I., Katsuno, M. et Goto, Y.-I. (2025). **A novel m.14677 T > C variant in mitochondrial tRNA^{Glu} gene causes chronic progressive external ophthalmoplegia.** J Hum Genet 70, 537-540, doi:[10.1038/s10038-025-01381-7](https://doi.org/10.1038/s10038-025-01381-7).

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

Pas de résultat ce trimestre

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

Chiang, B. K.-C., Tsang, S. H., Aycinena, A. R. P. et Sharma, T. (2025). **Mitochondrial Disorder: Kearns-Sayre Syndrome.** Adv Exp Med Biol 1467, 173-175, doi:[10.1007/978-3-031-72230-1_30](https://doi.org/10.1007/978-3-031-72230-1_30).

Finsterer, J. (2025). **Comprehensive basic and clinical studies are needed to improve the treatment and outcome of patients with Kearns-Sayre syndrome.** Transl Pediatr 14, 2075-2077, doi:[10.21037/tp-2025-425](https://doi.org/10.21037/tp-2025-425).

Finsterer, J. et Zarrouk, S. (2025). **Diagnosing Kearns-Sayre syndrome requires documentation of the underlying genetic defect.** Pan Afr Med J 50, 92, doi:[10.11604/pamj.2025.50.92.37174](https://doi.org/10.11604/pamj.2025.50.92.37174).

Krupka, D., Rakoczy, K., Chetmoński, A., Zakliczyński, M., Przybylski, R. et Sokolski, M. (2025). **Crucial Role of Early Detection in Managing Heart Failure in Kearns-Sayre Syndrome: A Case Report.** Am J Case Rep 26, e947439, doi:[10.12659/AJCR.947439](https://doi.org/10.12659/AJCR.947439).

Mitochondrial disorders : The Essential
MMITO-2025-3 from july 1st to september 30th, 2025

MacMullen, L. E., Reynolds, E., Weis, M., George-Sankoh, I., Nguyen, S., Stanley, K. D., Redko, M., Poblete, M., Goldstein, A. C. et Ganetzky, R. D. (2025). **Single large-scale mitochondrial DNA deletion syndromes: scientific and family conference optimizes the collection of rare disease research outcomes.** Orphanet J Rare Dis 20, 399, doi:[10.1186/s13023-025-03632-4](https://doi.org/10.1186/s13023-025-03632-4).

McCully, K. K., Bossie, H. M. et Kendall, F. D. (2024). **Noninvasive Assessments of Mitochondrial Capacity in People with Mitochondrial Myopathies.** Muscles 3, 393-403, doi:[10.3390/muscles3040033](https://doi.org/10.3390/muscles3040033).

Mendel, R. M., Matsuno, S., Lu, A., Falk, M. J. et Haroon, S. (2024). **Mitophagy modulation rescues single large-scale mitochondrial DNA deletion (SLSMD) disease symptoms in the C. elegans uaDf5 animal model.** bioRxiv 2024.10.27.620333, doi:[10.1101/2024.10.27.620333](https://doi.org/10.1101/2024.10.27.620333).

Ramakumar, V., Akkineni, K. P., Gawalkar, A. et Agstam, S. (2025). **Recurrent Syncope and Drooping Eyes in a Young Woman: Kearns-Sayre Syndrome.** JACC Clin Electrophysiol S2405-500X(25)00554-7, doi:[10.1016/j.jacep.2025.06.034](https://doi.org/10.1016/j.jacep.2025.06.034).

Shafiee, Amirmohammad, Akhlaghi, A. A., Ellstrom, A., Mohammadi, S. O., Shafiee, Alimohammad et Sathyamoorthy, M. (2025). **Molecular Aspects of Mitochondrial Dysfunction in Diabetes, Pearson and Kearns-Sayre Syndromes, and Neurodegenerative Disorders.** Int J Gen Med 18, 5355-5366, doi:[10.2147/IJGM.S539967](https://doi.org/10.2147/IJGM.S539967).

Sobeh, T., Granek, T., Bar-Yosef, O., Jacoby, E., Hoffmann, C. et Shrot, S. (2025). **Neuroimaging characteristics of single Large-Scale mitochondrial DNA deletion syndromes.** Neuroradiology, doi:[10.1007/s00234-025-03689-9](https://doi.org/10.1007/s00234-025-03689-9).

Zhang, J., Song, Z., Zhou, H., Zhou, Y. et Wang, S. (2025). **Fatal pneumonia in a patient with Kearns-Sayre syndrome case report and literature review.** Front Med (Lausanne) 12, 1575384, doi:[10.3389/fmed.2025.1575384](https://doi.org/10.3389/fmed.2025.1575384).

Syndrome de Leigh - Leigh syndrome

Abe, Yuichi, Hamasaki, T., Natsume, J., Mogami, Y., Murayama, K., Shiraishi, H., Abe, Yuki, Kumada, S., Tanaka, R., Ihara, K., Sakakibara, T., Okazaki, Y., Nakagawa, H., Takahashi, K., Yamauchi, M., Nakajima, M. et Ohtake, A. (2025). **Efficacy and Safety of 5-Aminolevulinic Acid Hydrochloride Combined with Sodium Ferrous Citrate in Pediatric Patients with Leigh Syndrome and Central Nervous System Disorders: An Initial Exploratory Trial with a Double-Blind Placebo-Controlled Period, Followed by an Open-Label Period and a Subsequent Long-Term Administration Study.** Life (Basel) 15, 1168, doi:[10.3390/life15081168](https://doi.org/10.3390/life15081168).

Ahmadi, Z. A. et Brandt, U. (2025). **The genotype/phenotype conundrum of inherited mitochondrial disorders: Insights from a survey of mtDNA mutations associated with Leigh syndrome in complex I.** Biochim Biophys Acta Mol Basis Dis 1871, 167996, doi:[10.1016/j.bbadis.2025.167996](https://doi.org/10.1016/j.bbadis.2025.167996).

AlFaris, H. S., Yoganathan, S., Callister, M. N., Aljouda, L., Pai, V., Krishnan, P., LeBlanc Miller, A., Ganos, C. et Gorodetsky, C. (2025). **Reply: « Leigh Syndrome Due to the Variant c.1019T>C in COX15 ».** Mov Disord Clin Pract, doi:[10.1002/mdc3.70376](https://doi.org/10.1002/mdc3.70376).

Anderson, E. D., Cronkite, C. A., Baldwin, P. R., Abella, C. P., Duman, J. G., Simmonds, A. N., Waxham, M. N., Tolias, K. F. et Ludtke, S. J. (2025). **Primary cortical neurons precipitate and extrude large mitochondria-associated calcium-phosphate sheets with a bone-precursor-like ultrastructure.** bioRxiv 2025.08.04.668590, doi:[10.1101/2025.08.04.668590](https://doi.org/10.1101/2025.08.04.668590).

Ayala-Hernandez, M. G., Torales, A. B., Tan, H. C., Montgomery, C. B., Padavannil, A., Cortopassi, G. et Letts, J. A. (2025). **Respiratory complex III2 assembles complex I via toxic intermediate in mitochondrial disease.** bioRxiv 2025.06.17.660237, doi:[10.1101/2025.06.17.660237](https://doi.org/10.1101/2025.06.17.660237).

Bhérier, C., Grenier, J.-C., Pelletier, J., Boucher, G., Gagnon, G., Goyette, P., Ashton-Beaucage, D., Stevens, C., Battat, R., Bitton, A., Campeau, P. M., Laprise, C., NIDDK IBD Genetics Consortium, Quebec IBD Genetics Consortium, iGenoMed Consortium, Huang, H., Daly, M., Taliun, D., Hussin, J. G., Mooser, V. et Rioux, J. D. (2025). **Frequency enrichment of coding variants in a French-Canadian founder population and its implication for inflammatory bowel diseases.** medRxiv 2025.07.11.25331388, doi:[10.1101/2025.07.11.25331388](https://doi.org/10.1101/2025.07.11.25331388).

Mitochondrial disorders : The Essential
MMITO-2025-3 from july 1st to september 30th, 2025

Burg, L., Yoon, H., Peng, M., Germano, P., Reese Gretzmacher, E., Xiao, R., Anderson, V. E., Nakamaru-Ogiso, E. et Falk, M. J. (2025). **Zagociguat prevented stressor-induced neuromuscular dysfunction, improved mitochondrial physiology, and increased exercise capacity in diverse mitochondrial respiratory chain disease zebrafish models.** Front Pharmacol 16, 1588426, doi:[10.3389/fphar.2025.1588426](https://doi.org/10.3389/fphar.2025.1588426).

Distelmaier, F., Corral-Sarasa, J., Jiménez-Sánchez, L., Díaz-Casado, M. E., Rohmann, M., Seibt, A., Herebian, D., Smits, S. H. J., Münch, J., Mayatepek, E., Breitzkreutz, J., Husain, R. A., López-Herrador, S., González-García, P. et López, L. C. (2025). **Preclinical and first-in-human evidence of 4-hydroxybenzoic acid for mitochondrial COQ2 deficiency.** Brain awaf334, doi:[10.1093/brain/awaf334](https://doi.org/10.1093/brain/awaf334).

Dumbuya, J. S., Tian, C., Deng, L., Ahmad, B., Chen, X. et Lu, J. (2025). **Clinical features, disease burden and impact on quality of life in participants with mitochondrial encephalomyopathy.** Front Neurol 16, 1585906, doi:[10.3389/fneur.2025.1585906](https://doi.org/10.3389/fneur.2025.1585906).

Farsana, M. K., Arunachal, G., Nandeesh, B. N., Kulanthaivelu, K., Mahale, R. R., Padmanabha, H., Mathuranath, P. S. et Mailankody, P. (2025). **Expanding the Clinical, Pathological, and Molecular Phenotypes of Tetratricopeptide 19 (TTC19) Gene Mutations: A Case Report from India.** Neurol India 73, 156-159, doi:[10.4103/neurol-india.Neurol-India-D-24-00143](https://doi.org/10.4103/neurol-india.Neurol-India-D-24-00143).

Finsterer, J. (2025). **Leigh Syndrome Due to the Variant c.1019T>C in COX15.** Mov Disord Clin Pract, doi:[10.1002/mdc3.70377](https://doi.org/10.1002/mdc3.70377).

Fois, A., Deschênes, S., Bourel, C., Beauchamp, C., Lombard-Vadnais, F., Ruiz, M., Charron, G., Coderre, L., LSFC Consortium, Rioux, J. D. et Lesage, S. (2025). **Disruption of Lrpprc affects B cell development and proliferation in a mouse model of Leigh Syndrome French Canadian type.** J Rare Dis (Berlin) 4, 31, doi:[10.1007/s44162-025-00094-x](https://doi.org/10.1007/s44162-025-00094-x).

Fouché, B. R., Lindeque, Z., van der Westhuizen, F., Venter, M. et van der Sluis, R. (2025). **The xenobiotic detoxification pathway - glycine conjugation - is downregulated in a mouse model of Leigh syndrome.** Biochem Biophys Res Commun 777, 152278, doi:[10.1016/j.bbrc.2025.152278](https://doi.org/10.1016/j.bbrc.2025.152278).

Jia, Y., Huang, Z., Chen, C., Chen, F. K. et Jiang, L. (2025). **Genetic and Clinical Investigations of C12orf65 Gene Mutations in Three Chinese Pedigrees.** J Neuroophthalmol, doi:[10.1097/WNO.0000000000002378](https://doi.org/10.1097/WNO.0000000000002378).

Mitochondrial disorders : The Essential

MMITO-2025-3 from july 1st to september 30th, 2025

Jiang, H., Xu, C., Liu, Z., Duan, R., Yao, X., Fu, X., Xu, J., Kang, X., Yu, T., Wang, Y. et Fang, F. (2025). **Case Report: Biallelic variants in MRPS36, encoding a component of the 2-oxoglutarate dehydrogenase complex, cause leigh syndrome.** Front Pediatr 13, 1608840, doi:[10.3389/fped.2025.1608840](https://doi.org/10.3389/fped.2025.1608840).

Kashem, M. S. B., Varnum, S., Lazorik, O., Giwa, R., Iyer, S., Yao, C., Piston, D. W., Zu, C., Brestoff, J. R. et Mukherji, S. (2025). **Multiplexed quantum sensing reveals coordinated thermomagnetic regulation of mitochondria.** bioRxiv 2025.07.30.666664, doi:[10.1101/2025.07.30.666664](https://doi.org/10.1101/2025.07.30.666664).

Ling, Q., Rioux, M., Higgs, H., Hu, Y., Dwyer, S. E. et Gray, S. J. (2025). **Improved AAV9-based gene therapy design for SURF1-related Leigh syndrome with minimal toxicity.** Mol Ther Methods Clin Dev 33, 101554, doi:[10.1016/j.omtm.2025.101554](https://doi.org/10.1016/j.omtm.2025.101554).

Liu, Y., Li, T. Y., Wang, J. L., Xu, C. L., Song, M. H., Xu, M. T., Liu, Z. M. et Fang, F. (2025). **[Clinical characteristics analysis of mitochondrial short-chain enoyl-CoA hydratase 1 deficiency with ECHS1 gene c.489G>A compound heterozygous variants].** Zhonghua Er Ke Za Zhi 63, 1085-1091, doi:[10.3760/cma.j.cn112140-20250718-00662](https://doi.org/10.3760/cma.j.cn112140-20250718-00662).

Luo, J., Chen, L., Zhang, X., Su, Q., Zhou, X. et Lian, Q. (2025). **Pioglitazone Ameliorates Mitochondrial Oxidative Stress and Inflammation via AMPK-Dependent Inhibition of Mitochondrial Fission in Leigh Syndrome.** Cell Prolif e70109, doi:[10.1111/cpr.70109](https://doi.org/10.1111/cpr.70109).

Mandia, D., Benoit, C., Stojkovic, T. et Nadjar, Y. (2025). **Motor Neuropathy in a Patient With Mitochondrial Disease and a Novel TTC19 Variant: An Underrecognized Phenotypic Feature.** J Peripher Nerv Syst 30, e70060, doi:[10.1111/jns.70060](https://doi.org/10.1111/jns.70060).

Mitchell, D. V., Iadarola, D. M., Mathew, N. D., Keith, K., Seiler, C., Yu, S., Kim, M. S., Woodard, N., Anderson, V. E., Nakamaru-Ogiso, E., Taylor, D. M. et Falk, M. J. (2025). **ndufs2^{-/-} zebrafish have impaired survival, neuromuscular activity, morphology, and one-carbon metabolism treatable with folic acid.** bioRxiv 2025.07.16.664929, doi:[10.1101/2025.07.16.664929](https://doi.org/10.1101/2025.07.16.664929).

Mittal, P., Karkhur, S., Verma, V. et Singh, P. (2025). **NDUFV1 mutation presenting as isolated progressive optic neuropathy: a unique manifestation of mitochondrial complex I deficiency.** BMJ Case Rep 18, e266155, doi:[10.1136/bcr-2025-266155](https://doi.org/10.1136/bcr-2025-266155).

Mitochondrial disorders : The Essential

MMITO-2025-3 from July 1st to September 30th, 2025

Roesch, S., O'Sullivan, A., Tschani, S., Baghdasaryan, A., Balasubramaniam, S., Barić, I., de Boer, L., Grünert, S. C., Guzek, A., Janssen, M., Krumina, Z., Koenig, M. K., Lewkowitz, A. M., Mochel, F., Naldi, A. M., Plecko, B., Öztürk, K., O'Grady, L., Riordan, G., Rymen, D., Sahai, I., Santer, R., Schiff, M., Stettner, G. M., Tsiakas, K., Uçar, S. K., Uzun, Ö. Ü., Weigel, C., Witters, P., Merkevicus, K., Mayr, J. A., Wortmann, S. B. et Iwanicka-Pronicka, K. (2025). **Hearing rehabilitation in SERAC1 related MEGD(H)EL syndrome - implications from a multi-center retrospective cohort study.** Mol Genet Metab 146, 109193, doi:[10.1016/j.ymgme.2025.109193](https://doi.org/10.1016/j.ymgme.2025.109193).

Sadek, A. A., Aladawy, M. A., Mansour, T. M., Fouad, M. F., Magdy, R. M., Elmoursy, M. M. et Abdelkreem, E. (2025). **Expanding the Epidemiological and Phenotypic Spectrum of MEGDEL Syndrome: The First Case Report From Egypt.** Clin Med Insights Pediatr 19, 11795565251348345, doi:[10.1177/11795565251348345](https://doi.org/10.1177/11795565251348345).

Villa-Villegas, L., Lira-Jaime, L. G., Farías-Moreno, K. C., González-Ruffino, B. D., Soto-Escageda, A., Mercado-Pimentel, R., Piña-Avilés, C. E. et Zúñiga-Ramírez, C. (2025). **Deep Brain Stimulation in Leigh-Like Syndrome Due to DNM1 Pathogenic Variant.** Tremor Other Hyperkinet Mov (N Y) 15, 32, doi:[10.5334/tohm.1017](https://doi.org/10.5334/tohm.1017).

Warwick, A. M., Yu, L., Klingeborn, M., Travis, A. M., Parmar, V. M., Bomze, H. M., Malek, G. et Gospe Iii, S. M. (2025). **Reduced complex I activity in the retinal pigment epithelium, but not in rod photoreceptors, affects light signaling without impacting cell survival.** J Biol Chem 301, 110586, doi:[10.1016/j.jbc.2025.110586](https://doi.org/10.1016/j.jbc.2025.110586).

Maladies mitochondriales (autres) – Mitochondrial disorders (other)

Abe, Yuichi, Hamasaki, T., Natsume, J., Mogami, Y., Murayama, K., Shiraishi, H., Abe, Yuki, Kumada, S., Tanaka, R., Ihara, K., Sakakibara, T., Okazaki, Y., Nakagawa, H., Takahashi, K., Yamauchi, M., Nakajima, M. et Ohtake, A. (2025). **Efficacy and Safety of 5-Aminolevulinic Acid Hydrochloride Combined with Sodium Ferrous Citrate in Pediatric Patients with Leigh Syndrome and Central Nervous System Disorders: An Initial Exploratory Trial with a Double-Blind Placebo-Controlled Period, Followed by an Open-Label Period and a Subsequent Long-Term Administration Study.** Life (Basel) 15, 1168, doi:[10.3390/life15081168](https://doi.org/10.3390/life15081168).

Adorisio, R., Cantarutti, N., Siri, B., Bellettini, E., Ingrassiotta, G., Mencarelli, E., Graziani, F., Lillo, R., Di Marzio, S., Di Mambro, C., Drago, F., Amodeo, A. et Martinelli, D. (2025). **Mitochondrial cardiomyopathies: navigating through different clinical and management pictures between adult and paediatric forms.** Front Cardiovasc Med 12, 1621096, doi:[10.3389/fcvm.2025.1621096](https://doi.org/10.3389/fcvm.2025.1621096).

Mitochondrial disorders : The Essential
MMITO-2025-3 from july 1st to september 30th, 2025

Ahmadi, Z. A. et Brandt, U. (2025). **The genotype/phenotype conundrum of inherited mitochondrial disorders: Insights from a survey of mtDNA mutations associated with Leigh syndrome in complex I.** Biochim Biophys Acta Mol Basis Dis 1871, 167996, doi:[10.1016/j.bbadis.2025.167996](https://doi.org/10.1016/j.bbadis.2025.167996).

Ahmed, F., Tsang, S. H., Aycinena, A. R. P. et Sharma, T. (2025). **Mitochondrial Disorder: Maternally Inherited Diabetes and Deafness.** Adv Exp Med Biol 1467, 177-180, doi:[10.1007/978-3-031-72230-1_31](https://doi.org/10.1007/978-3-031-72230-1_31).

Al Masseri, Z., Guilder, L., Inbar-Feigenberg, M., Cameron, J., Venkatachalam, S. et Chitayat, D. (2025). **Mitochondrial Complex V Deficiency Caused by a Homozygous Splice Variant in ATP5PO.** Am J Med Genet A e64239, doi:[10.1002/ajmg.a.64239](https://doi.org/10.1002/ajmg.a.64239).

Alatawi, A., Alshehri, O., Alessa, A., Al Mutairi, F., AlSaleh, N., Eyaid, W., Alsamri, A., Fageih, E., Mushiba, A., Saleh, M., Alotaibi, M., Tabarki, B., Aljadhai, Y. I., Katsonis, P., Lichtarge, O., Alkuraya, F. S., Alfadhel, M. et Almannai, M. (2025). **SLC25A42-Related Mitochondrial Disorder: New Cases and Literature Review.** Clin Genet, doi:[10.1111/cge.70021](https://doi.org/10.1111/cge.70021).

AlFaris, H. S., Yoganathan, S., Callister, M. N., Aljouda, L., Pai, V., Krishnan, P., LeBlanc Miller, A., Ganos, C. et Gorodetsky, C. (2025). **Reply: « Leigh Syndrome Due to the Variant c.1019T>C in COX15 ».** Mov Disord Clin Pract, doi:[10.1002/mdc3.70376](https://doi.org/10.1002/mdc3.70376).

Alhamad, A. R., Mushiba, A., Alkhawaja, H., PeerZada, A., Bawazeer, S. et Saleh, M. (2025). **Mutation of NDUFAF2 Linked to Mitochondrial Complex I Deficiency.** Cureus 17, e86670, doi:[10.7759/cureus.86670](https://doi.org/10.7759/cureus.86670).

Ali, L., Hazzard, I., Tehrani, N. S., Ansari, U., Ali, A., Kumar, P., Ali, N., Gakhal, G. et Lui, F. (2025). **Extraocular features of Leber hereditary optic neuropathy: A scoping review.** J Biol Methods 12, e99010055, doi:[10.14440/jbm.2024.0113](https://doi.org/10.14440/jbm.2024.0113).

Almuqbil, M., Binsabbar, N., Alsaif, S., Almasoud, S., Albasry, T., Baarmah, D., Altwaijri, W. et Alrumayyan, A. (2025). **Clinical and Molecular Characterizations of Mitochondrial Disorders: A Tertiary-Care Center Experience.** Children (Basel) 12, 1102, doi:[10.3390/children12081102](https://doi.org/10.3390/children12081102).

Amore, G., Carbonelli, M., D'Angeli, D., Bonan, L., Faustini-Fustini, M., Maresca, A., Carelli, V. et La Morgia, C. (2025). **Late-onset Leber's hereditary optic neuropathy and antiandrogens for prostate cancer: is there a causative link?** Front Neurol 16, 1616992, doi:[10.3389/fneur.2025.1616992](https://doi.org/10.3389/fneur.2025.1616992).

Mitochondrial disorders : The Essential
MMITO-2025-3 from july 1st to september 30th, 2025

Aoshima, K., Takagi, Y., Funato, M., Kuse, Y., Nakamura, S. et Shimazawa, M. (2025). **Establishment of human Leber's hereditary optic neuropathy model using iPSC-derived retinal organoids.** Front Cell Neurosci 19, 1635775, doi:[10.3389/fncel.2025.1635775](https://doi.org/10.3389/fncel.2025.1635775).

Armirola-Ricaurte, C., Morant, L., Adant, I., Hamed, S. A., Pipis, M., Efthymiou, S., Amor-Barris, S., Atkinson, D., Van de Vondel, L., Tomic, A., Seneca, S., de Vriendt, E., Zuchner, S., Ghesquiere, B., Hanna, M., Houlden, H., Lunn, M. P., Reilly, M. M., Rasic, V. M. et Jordanova, A. (2025). **Biallelic variants in COX18 cause a mitochondrial disorder primarily manifesting as peripheral neuropathy.** Brain awaf300, doi:[10.1093/brain/awaf300](https://doi.org/10.1093/brain/awaf300).

ATAD3 duplications bridge mitochondrial diseases and Aicardi-Goutières syndrome (2025). 67, e155, doi:[10.1111/dmcn.16446](https://doi.org/10.1111/dmcn.16446).

Ayala-Hernandez, M. G., Torales, A. B., Tan, H. C., Montgomery, C. B., Padavannil, A., Cortopassi, G. et Letts, J. A. (2025). **Respiratory complex III2 assembles complex I via toxic intermediate in mitochondrial disease.** bioRxiv 2025.06.17.660237, doi:[10.1101/2025.06.17.660237](https://doi.org/10.1101/2025.06.17.660237).

Baheer Abdulwahhab, S., Ahmed, A. et Ben Omran, T. (2025). **Neonatal Presentation of a Case of Carbonic Anhydrase VA Deficiency.** Cureus 17, e90845, doi:[10.7759/cureus.90845](https://doi.org/10.7759/cureus.90845).

Baker, M. J., Yek, K. Q. et Stojanovski, D. (2025). **Quality control at the powerhouse: mitochondrial proteostasis dysfunction and disease.** Biochem Soc Trans 53, 1-13, doi:[10.1042/BST20253044](https://doi.org/10.1042/BST20253044).

Ball, M., van Bergen, N. J., Compton, A. G., Thorburn, D. R., Rahman, S. et Christodoulou, J. (2025). **Therapies for Mitochondrial Disease: Past, Present, and Future.** J Inherit Metab Dis 48, e70065, doi:[10.1002/jimd.70065](https://doi.org/10.1002/jimd.70065).

Barca, E. et Emmanuele, V. (2025). **Updates on Mitochondrial Myopathies.** Curr Neurol Neurosci Rep 25, 55, doi:[10.1007/s11910-025-01444-4](https://doi.org/10.1007/s11910-025-01444-4).

Belousova, V., Ignatko, I., Bogomazova, I., Sosnova, E., Pesegova, S., Samusevich, A., Zarova, E., Kardanova, M., Skorobogatova, O. et Maltseva, A. (2025). **Causes of and Solutions to Mitochondrial Disorders: A Literature Review.** Int J Mol Sci 26, 6645, doi:[10.3390/ijms26146645](https://doi.org/10.3390/ijms26146645).

Benarroch, E. (2025). **What Is the Potential Relevance of Growth and Differentiation Factor-15 in Neurologic Disease?** Neurology 105, e214131, doi:[10.1212/WNL.0000000000214131](https://doi.org/10.1212/WNL.0000000000214131).



Mitochondrial disorders : The Essential

MMITO-2025-3 from july 1st to september 30th, 2025

Bérat, C.-M., Hully, M., Rötig, A., Barcia, G., Assouline, Z., Abi-Warde, M.-T., Barnerias, C., Payen, E., Jaroussie, M., Gaignard, P., Lebigot, E., Roubertie, A., Boddaert, N., Roux, C.-J., de Lonlay, P., Desguerre, I., Munnich, A., Schiff, M. et Gitiaux, C. (2025). **Childhood POLG-related disorders: Focus on polyradiculoneuropathy**. *Mol Genet Metab* 146, 109213, doi:[10.1016/j.ymgme.2025.109213](https://doi.org/10.1016/j.ymgme.2025.109213).

Beutner, G., Huyck, H. L., Deutsch, G., Pryhuber, G. S. et Porter, G. A. (2025). **Dysfunctional Electron Transport Chain Assembly in COXPD8**. *J Cardiovasc Dev Dis* 12, 318, doi:[10.3390/jcdd12080318](https://doi.org/10.3390/jcdd12080318).

Bľandová, G., Murgašová, M., Markocsy, A., Baldovič, M., Krasňanská, G., Eliaš, V., Repiská, V. et Konečný, M. (2025). **Pearson syndrome with atypical presentation of short stature and atypical limb proportions - First reported case in Slovakia**. *Mitochondrion* 85, 102079, doi:[10.1016/j.mito.2025.102079](https://doi.org/10.1016/j.mito.2025.102079).

Bo, K., Kelly, C., Trumpff, C., Hirano, M., Thiebaut de Schotten, M., Picard, M. et Wager, T. D. (2025). **Mitochondrial Energy Transformation Capacity Influences Brain Activation During Sensory, Affective, and Cognitive Tasks**. *bioRxiv* 2025.06.25.661482, doi:[10.1101/2025.06.25.661482](https://doi.org/10.1101/2025.06.25.661482).

Bordi, M., Testa, B., Compagnucci, C., Colasuonno, F., Cipressa, F., Betterini, E., Mancini, A., Carsetti, C., Salvatori, I., Ferraina, C., Yang, M., De Cegli, R., Del Prete, E., Veroni, C., Rizza, S., Mauri, S., Ziviani, E., Macchiaiolo, M., Vecchio, D., Panfili, F. M., Rizza, T., Weber, G., Carrozzo, R., Ferri, A., Campello, S., Ballabio, A., Frezza, C., Cestra, G., Tartaglia, M., Bartuli, A. et Cecconi, F. (2025). **ADSL deficiency is a secondary mitochondrial disease affecting organelle homeostasis and ERK2/AKT signaling in a linear genotype-phenotype relation**. *Cell Rep* 44, 116230, doi:[10.1016/j.celrep.2025.116230](https://doi.org/10.1016/j.celrep.2025.116230).

Burg, L., Yoon, H., Peng, M., Germano, P., Reese, Gretzmacher, E., Xiao, R., Anderson, V. E., Nakamaru-Ogiso, E. et Falk, M. J. (2025). **Zagociguat prevented stressor-induced neuromuscular dysfunction, improved mitochondrial physiology, and increased exercise capacity in diverse mitochondrial respiratory chain disease zebrafish models**. *Front Pharmacol* 16, 1588426, doi:[10.3389/fphar.2025.1588426](https://doi.org/10.3389/fphar.2025.1588426).

Carney, O. S., Harris, K., Santizo, M., Silva, V., Davis, J., Lee, K., Vishwanath, S., Hamacher-Brady, A. et Vernon, H. J. (2025). **A review of disorders of cardiolipin metabolism: Pathophysiology, clinical presentation and future directions**. *Mol Genet Metab* 145, 109184, doi:[10.1016/j.ymgme.2025.109184](https://doi.org/10.1016/j.ymgme.2025.109184).

Chan, J. Z., Tomczewski, M. V., Berdeklis, A. N. et Duncan, R. E. (2025). **The B-lymphoblastoid model in Barth syndrome**. *Biochim Biophys Acta Mol Cell Biol Lipids* 1870, 159691, doi:[10.1016/j.bbalip.2025.159691](https://doi.org/10.1016/j.bbalip.2025.159691).

Mitochondrial disorders : The Essential

MMITO-2025-3 from july 1st to september 30th, 2025

Chen, B. S., Seikus, C., Ferguson, J., Tadić, V., Horton, M., Yu-Wai-Man, P. et Archer, S. (2025). « **Adrift From the World** »: Exploring the Lived Experiences of Individuals Affected by an Inherited Optic Neuropathy in the United Kingdom-A Qualitative Study. Value Health S1098-3015(25)02490-8, doi:[10.1016/j.jval.2025.07.023](https://doi.org/10.1016/j.jval.2025.07.023).

Chiang, B. K.-C., Tsang, S. H., Aycinena, A. R. P. et Sharma, T. (2025). **Mitochondrial Disorder: Kearns-Sayre Syndrome**. Adv Exp Med Biol 1467, 173-175, doi:[10.1007/978-3-031-72230-1_30](https://doi.org/10.1007/978-3-031-72230-1_30).

Cox, S. N., Varvara, A. S. et Pesole, G. (2025). **MitSorter: a standalone tool for accurate discrimination of mtDNA and NuMT ONT reads based on differential methylation**. Bioinform Adv 5, vbaf135, doi:[10.1093/bioadv/vbaf135](https://doi.org/10.1093/bioadv/vbaf135).

Di Donfrancesco, A., Adelizzi, A., Giri, A., Duchi, R., Boito, S., Barandalla, M., Massaro, G., Santanatoglia, C., Cappellosa, E., Perota, A., Di Meo, I., Tiranti, V., Bottani, E., Galli, C., Persico, N. et Brunetti, D. (2025). **Transabdominal ultrasound guided AAV9-GFP delivery in fetal pigs: a translational and minimally invasive model for in utero fetal gene therapy**. Gene Ther 32, 487-496, doi:[10.1038/s41434-025-00551-8](https://doi.org/10.1038/s41434-025-00551-8).

Distelmaier, F., Corral-Sarasa, J., Jiménez-Sánchez, L., Díaz-Casado, M. E., Rohmann, M., Seibt, A., Herebian, D., Smits, S. H. J., Münch, J., Mayatepek, E., Breitzkreutz, J., Husain, R. A., López-Herrador, S., González-García, P. et López, L. C. (2025). **Preclinical and first-in-human evidence of 4-hydroxybenzoic acid for mitochondrial COQ2 deficiency**. Brain awaf334, doi:[10.1093/brain/awaf334](https://doi.org/10.1093/brain/awaf334).

Djalalvandi, A., Takeda, K., Grespi, F., Fan, H., Fonseca, T. B., Nogara, L., Sharifi, S., Barison, C., Semenzato, M., Omori, A., Cerutti, R., Steffan, D., Tsansizi, L., Balmaceda, V., Alan, L., Bean, C., Blaauw, B., Viscomi, C. et Scorrano, L. (2025). **Opantimirs: A class of antagonizing microRNAs that upregulate Opa1 and improve mitochondrial and disuse myopathies**. Cell Rep Med 6, 102248, doi:[10.1016/j.xcrm.2025.102248](https://doi.org/10.1016/j.xcrm.2025.102248).

Domínguez-González, C., Chiang, C., Colson, A.-O., Rebollo Mesa, I., Baixauli, E., Quan, J., VanMeter, S. et Hirano, M. (2025). **Pyrimidine Nucleos(t)ide Therapy in Patients With Thymidine Kinase 2 Deficiency: A Multicenter Retrospective Chart Review Study**. Neurology 105, e213908, doi:[10.1212/WNL.0000000000213908](https://doi.org/10.1212/WNL.0000000000213908).

Donis, R., Wakeling, M. N., Jeffery, N., Govier, M., Johnson, M. B., Hassan, S. S., Abdullah, M. A., Eltonbary, K. Y. E., Parvaneh, N., Amoli, M. M., Abbasi, F., Elmaogullari, S., Çetinkaya, S., Gunes, K., Tayfun, M., Yaghootkar, H., Hattersley, A. T., Flanagan, S. E. et De Franco, E. (2025). **Neonatal diabetes mellitus is a significant feature of COXPD-24 caused by recessive NARS2 variants**. Diabet Med 42, e70129, doi:[10.1111/dme.70129](https://doi.org/10.1111/dme.70129).



Mitochondrial disorders : The Essential

MMITO-2025-3 from july 1st to september 30th, 2025

du Preez, M. J., Schoonen, M., Williams, M. E., Bisschoff, M. et van der Westhuizen, F. H. (2025). **OXPHOS complex deficiency in congenital myopathy: A systematic review.** Eur J Clin Invest 55, e70114, doi:[10.1111/eci.70114](https://doi.org/10.1111/eci.70114).

Eisenkölbl, A., Lochmüller, H., Carter, M. T., Hamilton, L. E. et McMillan, H. J. (2025). **Congenital-onset MLASA2 from a novel YARS2 variant: A literature review.** J Neuromuscul Dis 22143602251369227, doi:[10.1177/22143602251369227](https://doi.org/10.1177/22143602251369227).

Elmaseh, S. A., Gauthier, D. A., Golmohammadi, M., Pargalava, N., Carelli, V. et Sadun, A. A. (2025). **Metformin may alter the course of Leber's hereditary optic neuropathy: a case report.** Front Med (Lausanne) 12, 1609941, doi:[10.3389/fmed.2025.1609941](https://doi.org/10.3389/fmed.2025.1609941).

Ferasin, V., Raichich, A., Ancora, C., Bonemazzi, I., Di Paola, A., D'Errico, I., Nosadini, M., Ancona, C., Pelizza, M. F., Cassina, M. et Toldo, I. (2025). **Hyperkinetic Movement Disorder in KARS1-Related Disease: An Illustrative Video-Recorded Case and Narrative Literature Review.** Neurol Int 17, 143, doi:[10.3390/neurolint17090143](https://doi.org/10.3390/neurolint17090143).

Finsterer, J. (2025a). **Before diagnosing stroke-like episodes in OPA1-related mitochondrial disorder, its imaging criteria must be met.** Epilepsy Behav Rep 31, 100772, doi:[10.1016/j.ebr.2025.100772](https://doi.org/10.1016/j.ebr.2025.100772).

Finsterer, J. (2025b). **Leigh Syndrome Due to the Variant c.1019T>C in COX15.** Mov Disord Clin Pract, doi:[10.1002/mdc3.70377](https://doi.org/10.1002/mdc3.70377).

Finsterer, J. (2025c). **Letter to the Editor From Finsterer « Diverse Phenotypes of Mitochondrial Disease With Varying Levels of Heteroplasmy ».** JCEM Case Rep 3, luaf127, doi:[10.1210/jcemcr/luaf127](https://doi.org/10.1210/jcemcr/luaf127).

Finsterer, J. et Zarrouk, S. (2025). **Diagnosing Kearns-Sayre syndrome requires documentation of the underlying genetic defect.** Pan Afr Med J 50, 92, doi:[10.11604/pamj.2025.50.92.37174](https://doi.org/10.11604/pamj.2025.50.92.37174).

Fouché, B. R., Lindeque, Z., van der Westhuizen, F., Venter, M. et van der Sluis, R. (2025). **The xenobiotic detoxification pathway - glycine conjugation - is downregulated in a mouse model of Leigh syndrome.** Biochem Biophys Res Commun 777, 152278, doi:[10.1016/j.bbrc.2025.152278](https://doi.org/10.1016/j.bbrc.2025.152278).

Fu, Q., Qiu, R., Li, S., Qin, Y., Lu, Z., Liyao, S., Yang, Z., Cheng, X., Chen, Y., Xu, H. et Cheng, Y. (2025). **ECHS1: pathogenic mechanisms, experimental models, and emerging therapeutic strategies.** Orphanet J Rare Dis 20, 430, doi:[10.1186/s13023-025-03959-y](https://doi.org/10.1186/s13023-025-03959-y).

Mitochondrial disorders : The Essential
MMITO-2025-3 from july 1st to september 30th, 2025

Gannon, J. L. (2025). **Nucleos(t)ide Therapy for TK2-Deficient Mitochondrial Myopathy: Small Molecules With Big Potential.** *Neurology* 105, e214187, doi:[10.1212/WNL.00000000000214187](https://doi.org/10.1212/WNL.00000000000214187).

García Gómez, E., San-Juan, D., Sandoval Luna, L. et Morales Morales, M. A. (2025). **Leber Hereditary Optic Neuropathy and Epilepsy in a Mexican Patient.** *Cureus* 17, e86663, doi:[10.7759/cureus.86663](https://doi.org/10.7759/cureus.86663).

Garone, C., Sabeni, S. et Carli, S. (2025). **Gene therapy and mRNA drugs approach for mitochondrial OXPHOS deficiencies.** *Mol Ther* S1525-0016(25)00765-8, doi:[10.1016/j.ymthe.2025.09.036](https://doi.org/10.1016/j.ymthe.2025.09.036).

German, H. M., Zaki, M. S., Usmani, M. A., Karagoz, I., Efthymiou, S., Abdel-Hamid, M. S., Arabiyat, H. A., Ghaffar, A., Shahzad, M., van Bokhoven, H., Ahmed, Z. M., Yaghini, O., Hosseini, N., Majidinezhad, M., Alavi, S., Bosma, M., Broeks, M. H., Türdoğan, D., Suri, M., Laura de Godoy, L., Verhoeven-Duif, N. M., Riazuddin, Sheikh, Gleeson, J. G., Alves, C., Jans, J. J. M., Riazuddin, Saima, Houlden, H. et Maroofian, R. (2025). **Comprehensive Genotypic, Phenotypic, and Biochemical Characterization of GOT2 Deficiency: A Progressive Neurodevelopmental Disorder with Epilepsy and Abnormal Movements.** *Genet Med* 101587, doi:[10.1016/j.gim.2025.101587](https://doi.org/10.1016/j.gim.2025.101587).

Ghazal, N., Huang, B., Shoemaker, L. J., Faundez, V. et Kwong, J. Q. (2025). **Meclizine rescues cardiac function and mitochondrial ultrastructure by ATP- and glycolysis-independent mechanisms in a genetic model of mitochondrial energy dysfunction.** *bioRxiv* 2025.09.03.674000, doi:[10.1101/2025.09.03.674000](https://doi.org/10.1101/2025.09.03.674000).

Ghosh, S., Jett, K. A., Baker, Z. N., Boulet, A., Hossain, A., Moore, S. A., Ralle, M., Ling, B., Cobine, P. A. et Leary, S. C. (2025). **Heart is the most susceptible organ in an isogenic background to loss of function mutations in the mitochondrial metallochaperone SCO1.** *Hum Mol Genet* 34, 1599-1609, doi:[10.1093/hmg/ddaf123](https://doi.org/10.1093/hmg/ddaf123).

Gillespie, H., Ng, Y. S., Wood, K. M., Hopton, S., Alston, C. L., Blakely, E. L., Thompson, N., Taylor, R. W., Browning, A. C., McFarland, R. et Sayer, J. A. (2025). **Severe clinical manifestation of mitochondrial disease due to the m.3243A>T variant: a case report of early-onset, multi-organ involvement and premature death.** *J Rare Dis (Berlin)* 4, 47, doi:[10.1007/s44162-025-00110-0](https://doi.org/10.1007/s44162-025-00110-0).

Girgenti, A., Palumbo, L., Kubat, G. B., Turkel, I., Picone, P. et Nuzzo, D. (2025). **Vesicle-guided mitochondria: A new perspective for brain mitochondrial transplantation.** *Ageing Res Rev* 112, 102881, doi:[10.1016/j.arr.2025.102881](https://doi.org/10.1016/j.arr.2025.102881).

Mitochondrial disorders : The Essential

MMITO-2025-3 from july 1st to september 30th, 2025

González-García, P., Jiménez-Sánchez, L., Corral-Sarasa, J., López-Herrador, S., Torres-Rusillo, S., Díaz-Casado, M. E. et López, L. C. (2025). **The treatment of primary CoQ deficiency requires the targeting of multiple pathogenic mechanisms.** Commun Med (Lond) 5, 286, doi:[10.1038/s43856-025-01000-8](https://doi.org/10.1038/s43856-025-01000-8).

Guo, X., Geng, X., Zhang, H., Liu, R., Li, Q., Li, Z., Zhang, Y., Zhang, L., Fu, Z., Wang, L., You, H., Xue, J. et Luo, D. (2025). **Calsequestrin-1 Deficiency Induced Malignant Hyperthermia-Like Skeletal Injury through Mitochondrial Disorder.** Biol Pharm Bull 48, 1079-1088, doi:[10.1248/bpb.b24-00829](https://doi.org/10.1248/bpb.b24-00829).

Hakimjavadi, H., Eom, E., Christodoulou, E., Hjelm, B. E., Omidshar, A. A., Ostrow, D., Biegel, J. A. et Gai, X. (2025). **Distinct Mitochondrial DNA Deletion Profiles in Pediatric B- and T-ALL During Diagnosis, Remission, and Relapse.** Int J Mol Sci 26, 7117, doi:[10.3390/ijms26157117](https://doi.org/10.3390/ijms26157117).

Harhai, M., Foged, M. M., Zarges, C., Landoni, J. C., Chollet, S., Simonelli, M., Reczens, E., Lisci, M., Laban, N., Manley, S., Riemer, J., Lopez-Escamez, J. A., Lysakowski, A. et Jourdain, A. A. (2025). **An updated inventory of genes essential for oxidative phosphorylation identifies a mitochondrial origin in familial Ménière's disease.** Cell Rep 44, 116069, doi:[10.1016/j.celrep.2025.116069](https://doi.org/10.1016/j.celrep.2025.116069).

He, H., Zhang, X., Xiong, L. et Qin, X. (2025). **A Review of FUN14 Domain-Containing 1 Involvement in Mitochondrial Biological Processes and Mechanisms Across Various Systems.** Cell Biochem Funct 43, e70125, doi:[10.1002/cbf.70125](https://doi.org/10.1002/cbf.70125).

Helali, H. A. et Almutaser, S. (2025). **Expanding the Phenotype: A Case Report of a Novel Alanyl-tRNA Synthetase 2 (AARS2) Homozygous Mutation in a 17-Month-Old Child.** Cureus 17, e86330, doi:[10.7759/cureus.86330](https://doi.org/10.7759/cureus.86330).

Hemachudha, P., Anukoolwittaya, P., Pongpitakmetha, T., Joyjinda, Y., Ruchisrisarod, C., Saraya, A. W., Rattanawong, W., Thanapornsungsuth, P. et Hemachudha, T. (2025). **Understanding the genetics and neurology: an overview of adult neurogenetics.** Asian Biomed (Res Rev News) 19, 196-208, doi:[10.2478/abm-2025-0022](https://doi.org/10.2478/abm-2025-0022).

Hong, S., Kim, S. P., Kim, S., Kang, S. K., Jung, S., Oh, Y., Choi, S. H., Lee, S. B., Cha, H., Kim, J., Bae, J., Park, J., Kim, K., Choi, C. G., Park, S.-J., Kim, D. H., Kim, L. K., Seong, J. K. et Lee, H. (2025). **Alterations in mitochondrial base editors enhance targeted editing efficiency for mouse model generation.** Mol Ther Nucleic Acids 36, 102678, doi:[10.1016/j.omtn.2025.102678](https://doi.org/10.1016/j.omtn.2025.102678).

Mitochondrial disorders : The Essential

MMITO-2025-3 from july 1st to september 30th, 2025

Hu, C., Su, T., Zhang, Y., Yang, J., Su, X., Zhang, Z., Wang, X., Zou, W., Liang, D., Liu, Y., Ji, D. et Cao, Y. (2025). **Estradiol alleviates disease phenotypes caused by m.3635G > a mutations by activating mitochondrial biogenesis and PINK1-Parkin mediated mitophagy in iPSC-derived retinal pigment epithelium cells.** Cell Signal 136, 112121, doi:[10.1016/j.cellsig.2025.112121](https://doi.org/10.1016/j.cellsig.2025.112121).

Huang, Q., Trumpff, C., Monzel, A. S., Rausser, S., Devine, J., Liu, C. C., Kelly, C., Kurade, M., Li, S., Engelstad, K., Tanji, K., Lauriola, V., Wang, T., Wang, S., Sloan, R., Juster, R.-P., Hirano, M. et Picard, M. (2025). **The mitochondrial disease biomarker GDF15 is dynamic, quantifiable in saliva, and correlates with disease severity.** Mol Genet Metab 145, 109179, doi:[10.1016/j.ymgme.2025.109179](https://doi.org/10.1016/j.ymgme.2025.109179).

Huang, X., Pan, C.-H., Yin, F., Peng, J. et Yang, L. (2025). **The Role of Estrogen in Mitochondrial Disease.** Cell Mol Neurobiol 45, 68, doi:[10.1007/s10571-025-01592-8](https://doi.org/10.1007/s10571-025-01592-8).

Ito, M., Huang, Z., Nomura, K., Teranishi, M., Zhao, F., Mino, H., Yoneda, M., Tanaka, M. et Ohno, K. (2025). **Extremely low-frequency electromagnetic field (ELF-EMF) enhances mitochondrial energy production in NARP cybrids.** Sci Rep 15, 24369, doi:[10.1038/s41598-025-10536-7](https://doi.org/10.1038/s41598-025-10536-7).

Jacob, N., Schechter, D., Marshall, M., Bansal, N., Lamour, J., Vernon, H., Morava, E. et Elsharkawi, I. (2025). **Elamipretide in the Management of Barth Syndrome: Current Evidence and a Case Report.** Mol Genet Metab 146, 109220, doi:[10.1016/j.ymgme.2025.109220](https://doi.org/10.1016/j.ymgme.2025.109220).

Jana, A., Chen, Y.-H. et Zhao, L. (2025). **Mitochondria-Targeting Abasic Site-Reactive Probe (mTAP) Enables the Manipulation of Mitochondrial DNA Levels.** Angew Chem Int Ed Engl 64, e202502470, doi:[10.1002/anie.202502470](https://doi.org/10.1002/anie.202502470).

John, R., Chapla, A., Chacko, G., Yoganathan, S., Thomas, M. M. et Thomas, N. (2025). **Response to Letter to the Editor from Finsterer « Diverse Phenotypes of Mitochondrial Disease With Varying Levels of Heteroplasmy ».** JCEM Case Rep 3, luaf128, doi:[10.1210/jcemcr/luaf128](https://doi.org/10.1210/jcemcr/luaf128).

Kamble, N., Arunachal, G., Harish, S., Rao, V. B., Holla, V. V. et Pal, P. K. (2025). **Acute refractory chorea as a novel phenotype of NARS2 gene mutation-related mitochondrial disorder.** Parkinsonism Relat Disord 140, 108025, doi:[10.1016/j.parkreldis.2025.108025](https://doi.org/10.1016/j.parkreldis.2025.108025).

Kankanam Gamage, S. U. et Morimoto, Y. (2025). **Significance of Mitochondrial Dynamics in Reproductive Physiology: Current and Emerging Horizons in Mitochondrial Therapy for Assisted Reproductive Technologies.** Reprod Med Biol 24, e12672, doi:[10.1002/rmb2.12672](https://doi.org/10.1002/rmb2.12672).

Mitochondrial disorders : The Essential
MMITO-2025-3 from july 1st to september 30th, 2025

Karaa, A., Goldstein, A., Cohen, B. H., Haas, R. H., Vockley, J., Gorman, G. S., Mancuso, M., et RePOWER, MMPOWER-3, and MOTOR investigators (2025). **RePOWER: An International, Prospective, Non-Interventional Registry of Patients With Primary Mitochondrial Myopathy**. Clin Genet, doi:[10.1111/cge.70026](https://doi.org/10.1111/cge.70026).

Kashem, M. S. B., Varnum, S., Lazorik, O., Giwa, R., Iyer, S., Yao, C., Piston, D. W., Zu, C., Brestoff, J. R. et Mukherji, S. (2025). **Multiplexed quantum sensing reveals coordinated thermomagnetic regulation of mitochondria**. bioRxiv 2025.07.30.666664, doi:[10.1101/2025.07.30.666664](https://doi.org/10.1101/2025.07.30.666664).

Khetrapal, S., Tuli, A., Pulipati, Y., Farah, V. et Poornima, I. (2025). **PPA2 deficiency-a rare cause of genetic cardiomyopathy: a case report**. Eur Heart J Case Rep 9, ytaf307, doi:[10.1093/ehjcr/ytaf307](https://doi.org/10.1093/ehjcr/ytaf307).

Krupka, D., Rakoczy, K., Chetmoński, A., Zakliczyński, M., Przybylski, R. et Sokolski, M. (2025). **Crucial Role of Early Detection in Managing Heart Failure in Kearns-Sayre Syndrome: A Case Report**. Am J Case Rep 26, e947439, doi:[10.12659/AJCR.947439](https://doi.org/10.12659/AJCR.947439).

Kumar, N. et Mangla, M. (2025). **The Mitochondrial Deoxyribonucleic Acid Puzzle: Controversies, Challenges, and Critical Perspectives - A Narrative Review**. Curr Gene Ther, doi:[10.2174/0115665232358476250708091107](https://doi.org/10.2174/0115665232358476250708091107).

Langeveld, M., Brouwers, M. C. G. J., van Dussen, L., Koens, L. H., Langendonk, J. G. L. et Janssen, M. C. H. (2025). **[Inherited metabolic disorders in adults: growing patient numbers, advanced diagnostic tools and new treatment modalities]**. Ned Tijdschr Geneesk 169, D8476.

Lawal, T. A., Riekhof, W., Groom, L., Varma, P., Chrismer, I. C., Kokkinis, A., Grunseich, C., Witherspoon, J. W., Razaqyar, M. S., Sinaii, N., Meilleur, K. G., Xiang, L., Buzkova, J., Euro, L., Mohassel, P., Dirksen, R. T. et Todd, J. J. (2025). **NAD⁺ dyshomeostasis in RYR1-related myopathies**. Skelet Muscle 15, 22, doi:[10.1186/s13395-025-00390-6](https://doi.org/10.1186/s13395-025-00390-6).

Li, Wensheng, Gui, Y., Guo, C., Huang, Y., Liu, Y., Yu, X., Zhang, H., Wang, J., Liu, R., Mahaman, Y. A. R., Duan, Q. et Wang, X. (2025). **Molecular mechanisms of mitochondrial quality control**. Transl Neurodegener 14, 45, doi:[10.1186/s40035-025-00505-5](https://doi.org/10.1186/s40035-025-00505-5).

Li, Weini, Shen, J., Zhuang, A., Wang, R., Li, Q., Rabata, A., Zhang, Y. et Cao, D. (2025). **Palmitoylation: an emerging therapeutic target bridging physiology and disease**. Cell Mol Biol Lett 30, 98, doi:[10.1186/s11658-025-00776-w](https://doi.org/10.1186/s11658-025-00776-w).

Mitochondrial disorders : The Essential
MMITO-2025-3 from july 1st to september 30th, 2025

Liao, Y., Xu, D. et Li, L. (2025). **A case of mitochondrial encephalomyopathy remarkable presenting with refractory shock.** Zhong Nan Da Xue Xue Bao Yi Xue Ban 50, 511-516, doi:[10.11817/j.issn.1672-7347.2025.240250](https://doi.org/10.11817/j.issn.1672-7347.2025.240250).

Lickfett, S., Menacho, C., Cambridge, S. et Prigione, A. (2025). **Neuronal branching in stem cell models of mitochondrial and neurological diseases.** Stem Cells 43, sxaf050, doi:[10.1093/stmcls/sxaf050](https://doi.org/10.1093/stmcls/sxaf050).

Lin, T.-Y., Stone, Y. et Glatt, S. (2025). **Mechanistic insight into the pseudouridylation of RNA.** RNA Biol 22, 1-25, doi:[10.1080/15476286.2025.2541421](https://doi.org/10.1080/15476286.2025.2541421).

Ling, Q., Rioux, M., Higgs, H., Hu, Y., Dwyer, S. E. et Gray, S. J. (2025). **Improved AAV9-based gene therapy design for SURF1-related Leigh syndrome with minimal toxicity.** Mol Ther Methods Clin Dev 33, 101554, doi:[10.1016/j.omtm.2025.101554](https://doi.org/10.1016/j.omtm.2025.101554).

Liu, J. Y., Koenig, M. K., Riggs-Harpur, K. et Numan, M. T. (2025). **Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-Like Episodes (MELAS) Patients with m.3243A > G have High Prevalence of Wolff-Parkinson-White Syndrome.** Pediatr Cardiol, doi:[10.1007/s00246-025-03985-4](https://doi.org/10.1007/s00246-025-03985-4).

Liu, X.-H., Shen, X. et Zhong, Y.-S. (2025). **Retinal multimodal-imaging and functional tests in a mitochondrial disease with focal and segmental glomerulosclerosis.** Int J Ophthalmol 18, 1770-1776, doi:[10.18240/ijo.2025.09.19](https://doi.org/10.18240/ijo.2025.09.19).

Lopergolo, D., Berti, G., Gallus, G. N., Bianchi, S., Santorelli, F. M., Malandrini, A. et De Stefano, N. (2025). **The First Heterozygous TWNK Nonsense Mutation Associated with Progressive External Ophthalmoplegia: Evidence for a New Piece in the Puzzle of Mitochondrial Diseases.** Biomolecules 15, 1337, doi:[10.3390/biom15091337](https://doi.org/10.3390/biom15091337).

Lovell-Badge, R. (2025). **Reducing the Risks of Mitochondrial Disease in Children.** N Engl J Med 393, 500-503, doi:[10.1056/NEJMe2507753](https://doi.org/10.1056/NEJMe2507753).

Luce, J. (2025). **Mitochondrial replacement techniques: The perspectives of mitochondrial disease specialists in Germany.** Soc Sci Med 382, 118177, doi:[10.1016/j.socscimed.2025.118177](https://doi.org/10.1016/j.socscimed.2025.118177).

MacVicar, T. (2025a). **High tide or low tide: the transport and metabolism of mitochondrial nucleotides.** Biochem J 482, 1105-1122, doi:[10.1042/BCJ20253237](https://doi.org/10.1042/BCJ20253237).

MacVicar, T. (2025b). **High tide or low tide: the transport and metabolism of mitochondrial nucleotides.** Biochem J 482, BCI20253237, doi:[10.1042/10.1042/BCJ20253237](https://doi.org/10.1042/10.1042/BCJ20253237).

Mitochondrial disorders : The Essential
MMITO-2025-3 from july 1st to september 30th, 2025

Makowski, M., Almendro-Vedia, V. G. et López-Montero, I. (2025). **Cardiolipin acyl chain composition tailors the conformation of mammalian ATP synthase dimers**. Commun Chem 8, 220, doi:[10.1038/s42004-025-01611-1](https://doi.org/10.1038/s42004-025-01611-1).

Makridou, A., Sintou, E., Chatzianagnosti, S., Dermitzakis, I., Gargani, S., Manthou, M. E. et Theotokis, P. (2025). **Mapping Disorders with Neurological Features Through Mitochondrial Impairment Pathways: Insights from Genetic Evidence**. Curr Issues Mol Biol 47, 504, doi:[10.3390/cimb47070504](https://doi.org/10.3390/cimb47070504).

Mandia, D., Benoit, C., Stojkovic, T. et Nadjar, Y. (2025). **Motor Neuropathy in a Patient With Mitochondrial Disease and a Novel TTC19 Variant: An Underrecognized Phenotypic Feature**. J Peripher Nerv Syst 30, e70060, doi:[10.1111/jns.70060](https://doi.org/10.1111/jns.70060).

Manini, A., Brusati, A., Grassano, M., Scacciatella, G., Peverelli, S., Spagliardi, J., Pensato, V., Doretti, A., Vasta, R., Manera, U., Canosa, A., Brunetti, M., Gentilini, D., Messina, S., Verde, F., Moglia, C., Morelli, C., Dalla Bella, E., Keagle, P. J., Landers, J. E., Gellera, C., Lauria Pinter, G., Chiò, A., Ratti, A., Calvo, A., Silani, V. et Ticozzi, N. (2025). **Whole genome sequencing analysis in primary lateral sclerosis (PLS) patients reveals mutations in neurological diseases-causing genes**. J Neurol 272, 587, doi:[10.1007/s00415-025-13328-1](https://doi.org/10.1007/s00415-025-13328-1).

McCormick, E. M., Peterson, J. T., Santos, J. D. D., Flickinger, J., Xiao, R., Haas, R. et Zolkipli-Cunningham, Z. (2025). **The profound implications of mitochondrial myopathy on activities of daily living: an observational qualitative study of standardized structured and semi-structured patient interviews**. Ther Adv Chronic Dis 16, 20406223251344763, doi:[10.1177/20406223251344763](https://doi.org/10.1177/20406223251344763).

McCully, K. K., Bossie, H. M. et Kendall, F. D. (2024). **Noninvasive Assessments of Mitochondrial Capacity in People with Mitochondrial Myopathies**. Muscles 3, 393-403, doi:[10.3390/muscles3040033](https://doi.org/10.3390/muscles3040033).

McKeague, I. W., Engelstad, K., Ge, Y., Tucker, A., Li, S., Uddin, J., Zhao, Z., Thompson, J. L. P. et Hirano, M. (2025). **Assessing a Mitochondrial Disease Treatment via a Novel Statistical Technique for Accelerometer Data**. Ann Clin Transl Neurol, doi:[10.1002/acn3.70180](https://doi.org/10.1002/acn3.70180).

Mei, J., Ding, P., Gao, C., Zhou, J., Li, Z., Zhang, C. et Gao, J. (2025). **Mitochondrial Diseases: Molecular Pathogenesis and Therapeutic Advances**. MedComm (2020) 6, e70385, doi:[10.1002/mco2.70385](https://doi.org/10.1002/mco2.70385).

Mitochondrial disorders : The Essential
MMITO-2025-3 from july 1st to september 30th, 2025

Meldau, S., McCormick, E. M., George-Sankoh, I., Riordan, G. T., Khan, K., MacMullen, L. E., Dawlat, S., Blackhurst, D., Falk, M. J. et Elson, J. L. (2025). **Mitochondrial DNA Pathogenic Variant Prevalence in Primary Mitochondrial Disease Patients With African (L) Mitochondrial Genome Haplogroups**. JIMD Rep 66, e70036, doi:[10.1002/jmd2.70036](https://doi.org/10.1002/jmd2.70036).

Mendel, R. M., Matsuno, S., Lu, A., Falk, M. J. et Haroon, S. (2024). **Mitophagy modulation rescues single large-scale mitochondrial DNA deletion (SLSMD) disease symptoms in the C. elegans uaDf5 animal model**. bioRxiv 2024.10.27.620333, doi:[10.1101/2024.10.27.620333](https://doi.org/10.1101/2024.10.27.620333).

Mickelsson, N., Hirvonen, J. et Martikainen, M. H. (2025). **Widespread and progressive brain atrophy is a common feature in patients with mitochondrial disease**. J Neurol 272, 648, doi:[10.1007/s00415-025-13354-z](https://doi.org/10.1007/s00415-025-13354-z).

Miller, D. M., Archer, S. L. et Dunham-Snary, K. J. (2025). **Preclinical models of mitochondrial dysfunction: mtDNA and nuclear-encoded regulators in diverse pathologies**. Front Aging 6, 1585508, doi:[10.3389/fragi.2025.1585508](https://doi.org/10.3389/fragi.2025.1585508).

Mitra, K. S., Holmberg, S. R., Mota, M., Sabharwal, A. et Ekker, S. C. (2025a). **Nondestructive Larval Genotyping of Danio rerio for Mitochondrial and Nuclear DNA Genetics**. Zebrafish, doi:[10.1177/15458547251379703](https://doi.org/10.1177/15458547251379703).

Mitra, K. S., Holmberg, S. R., Mota, M., Sabharwal, A. et Ekker, S. C. (2025b). **Non-Destructive Larval Genotyping of Danio rerio for Mitochondrial and Nuclear DNA Genetics**. bioRxiv 2025.04.30.651585, doi:[10.1101/2025.04.30.651585](https://doi.org/10.1101/2025.04.30.651585).

Miwa, T., Hashimoto, K., Seto, T. et Sakamoto, H. (2025). **Mitochondrial Hearing Loss: Genetic Variants and Clinical Progression**. Cureus 17, e85825, doi:[10.7759/cureus.85825](https://doi.org/10.7759/cureus.85825).

Morava, E., Elsharkawi, I. et Kozicz, T. (2025). **Reframing primary mitochondrial disease as a sterile interferonopathy**. Mol Genet Metab 146, 109217, doi:[10.1016/j.ymgme.2025.109217](https://doi.org/10.1016/j.ymgme.2025.109217).

Murdock, D. G., Janssen, K. A., Keller, K., Mitchell, K. L., Beauplan, M., O'Brien, W. T., D'Alessandro, L., Haltom, J. A. et Wallace, D. C. (2025). **A mouse model of MEPAN demonstrates a role for mitochondrial fatty acid synthesis in iron-sulfur cluster and supercomplex formation**. Proc Natl Acad Sci U S A 122, e2506761122, doi:[10.1073/pnas.2506761122](https://doi.org/10.1073/pnas.2506761122).

Mitochondrial disorders : The Essential
MMITO-2025-3 from july 1st to september 30th, 2025

Nagatomo, R., Hiramatsu, Y., Sakiyama, Y., Miyahara, H., Nasu, T., Yamazaki, I., Yoshimura, A., Ando, M., Nozuma, S., Higuchi, Y., Akagi, A., Iwasaki, Y., Yuan, J.-H., Okamoto, Y. et Takashima, H. (2025). **Systemic mitochondrial involvement in mitochondrial myopathy with episodic hyper-creatine kinase-emia: insights from an autopsy case.** J Neurol 272, 576, doi:[10.1007/s00415-025-13326-3](https://doi.org/10.1007/s00415-025-13326-3).

Nakagawa, H., Shima, Y., Sasagawa, Y. et Nikaido, I. (2025). **MitoDelta: identifying mitochondrial DNA deletions at cell-type resolution from single-cell RNA sequencing data.** BMC Genomics 26, 810, doi:[10.1186/s12864-025-11931-0](https://doi.org/10.1186/s12864-025-11931-0).

Nakamura, K., Kishita, Y., Imai-Okazaki, A., Omata, T., Nodera, M., Yatsuka, Y., Sugiura, A., Matsumoto, N., Prokisch, H., Matsumoto, H., Ohtake, A., Murayama, K. et Okazaki, Y. (2025). **Identification of a pathogenic RNU4-2 variant in patients with mitochondrial disease: Broadening the spectrum of non-coding RNA gene variants in mitochondrial dysfunction.** J Hum Genet 70, 541-543, doi:[10.1038/s10038-025-01356-8](https://doi.org/10.1038/s10038-025-01356-8).

Nakashima, D., Tosaki, T., Sasaki, T., Honda, Y., Yokote, S., Mori, T., Sohara, E., Uchida, S., Tsuboi, N. et Yokoo, T. (2025). **Combined Alport Syndrome Type 3A and Mitochondrial Disease Presenting with a Thin Base Membrane and Overt Albuminuria: A Case Report.** Intern Med, doi:[10.2169/internalmedicine.5838-25](https://doi.org/10.2169/internalmedicine.5838-25).

Nasca, A., Catania, A., Legati, A., Izzo, R., D'onofrio, C., Ciavattini, T., Lamantea, E., Lamperti, C. et Ghezzi, D. (2025). **A De Novo DNMT1L Mutation in Twins with Variable Symptoms, Including Paraparesis and Optic Neuropathy.** Biomolecules 15, 1230, doi:[10.3390/biom15091230](https://doi.org/10.3390/biom15091230).

O'Hagan, S., Meldau, S., Rose, P., Van Niekerk, M., Ackermann, C., Riordan, G. et Van Toorn, R. (2025). **Retrospective observational study of the magnetic resonance imaging features of MPV17-related mitochondrial DNA depletion syndrome.** Pediatr Radiol 55, 2102-2114, doi:[10.1007/s00247-025-06341-z](https://doi.org/10.1007/s00247-025-06341-z).

Oikawa, R., Yokota, K., Fujikura, J., Uchimura, T., Miyashita, K., Hayashi, K., Sakurai, H. et Sone, M. (2025). **Impact of Mitochondrial A3243G Mutation on Skeletal Muscle Energy Metabolism: Evidence from Human Induced Pluripotent Stem Cell-Derived Skeletal Muscle Cells.** Stem Cells Dev 34, 333-342, doi:[10.1177/15473287251359330](https://doi.org/10.1177/15473287251359330).

Oláhová, M., Guerra, R. M., Collier, J. J., Heidler, J., Thompson, K., White, C. R., Castañeda-Tamez, P., Cabrera-Orefice, A., Lightowlers, R. N., Chrzanowska-Lightowlers, Z. M. A., Galkin, A., Wittig, I., Pagliarini, D. J. et Taylor, R. W. (2025). **RTN4IP1 is required for the final stages of mitochondrial complex I assembly and CoQ biosynthesis.** EMBO J 44, 5482-5508, doi:[10.1038/s44318-025-00533-x](https://doi.org/10.1038/s44318-025-00533-x).



Mitochondrial disorders : The Essential

MMITO-2025-3 from july 1st to september 30th, 2025

O'Leary, K. (2025). **Three-donor IVF prevents mitochondrial disease**. Nat Med, doi:[10.1038/d41591-025-00047-3](https://doi.org/10.1038/d41591-025-00047-3).

Orbach, R., Maio, N., Butterfield, R. J., Foley, A. R., Silverstein, S., Li, Y., Chao, K., Lehky, T. J., Potticary, A., Rouault, T. A., Donkervoort, S. et Bönnemann, C. G. (2025). **BCS1L-Associated Disease: 5'-UTR Variant Shifts the Phenotype Towards Axonal Neuropathy**. Ann Clin Transl Neurol 12, 1834-1845, doi:[10.1002/acn3.70108](https://doi.org/10.1002/acn3.70108).

Penicaud, R., Ferron, J.-B., Valette, X., Bauduin, P., Faisant, M., Schiff, M., Nguyen, A., Fontaine, F. et Allouche, S. (2025). **Late-Onset Multiple Acyl-CoA Dehydrogenase Deficiency (MADD): A Case Report With a Complex Biochemical Profile**. JIMD Rep 66, e70035, doi:[10.1002/jmd2.70035](https://doi.org/10.1002/jmd2.70035).

Pérez, M. J., Colombo, R. B., Real, S. M., Branham, M. T., Laurito, S. R., Moraes, C. T. et Mayorga, L. (2025). **Rewriting nuclear epigenetic scripts in mitochondrial diseases as a strategy for heteroplasmy control**. EMBO Mol Med 17, 2354-2383, doi:[10.1038/s44321-025-00285-5](https://doi.org/10.1038/s44321-025-00285-5).

Pisano, A., Belloli, S., Pignataro, M. G., Rainone, P., Valtorta, S., Coliva, A., de Turreis, V., Mosca, L., Di Micco, P., Moresco, R. M., d'Amati, G. et Morea, V. (2025). **Peptide-mimetics derived from leucyl-tRNA synthetase are potential agents for the therapy of mt-tRNA related diseases**. Front Pharmacol 16, 1607343, doi:[10.3389/fphar.2025.1607343](https://doi.org/10.3389/fphar.2025.1607343).

Planté-Bordeneuve, P., Bérat, C.-M., Hanein, S., Gitiaux, C., ATAD3 Study Group, Steffann, J., Desguerre, I., Rötig, A., Boddaert, N. et Barcia, G. (2025). **ATAD3 duplications bridge mitochondrial diseases and Aicardi-Goutières syndrome**. Dev Med Child Neurol, doi:[10.1111/dmcn.16414](https://doi.org/10.1111/dmcn.16414).

Plouzenec, S., Chao de la Barca, J. M. et Chevrollier, A. (2025). **The Role of Phospholipids in Mitochondrial Dynamics and Associated Diseases**. Front Biosci (Landmark Ed) 30, 27634, doi:[10.31083/FBL27634](https://doi.org/10.31083/FBL27634).

Pradhan, S., Mito, T., Khan, N. A., Olander, S., Zhaivoron, A., McWilliams, T. G. et Suomalainen, A. (2025). **PDK4 and nutrient responses explain muscle specific manifestation in mitochondrial disease**. Clin Transl Med 15, e70404, doi:[10.1002/ctm2.70404](https://doi.org/10.1002/ctm2.70404).

Prasanpanich, M., Husain, M., Halnon, N. J., Chang, R., Zadeh, N., Knight, J. et Fishbein, G. A. (2025). **Cardiac pathology in a patient with a novel pathogenic variant c.703del (p.Ile235SerfsTer4) of the TFAZZIN gene**. Cardiovasc Pathol 79, 107749, doi:[10.1016/j.carpath.2025.107749](https://doi.org/10.1016/j.carpath.2025.107749).

Mitochondrial disorders : The Essential

MMITO-2025-3 from july 1st to september 30th, 2025

Preston, G., Jacob, N., Elsharkawi, I., Morava, E. et Kozicz, T. (2025). **Corrigendum to « Phosphodiesterase type 5 inhibition as a therapeutic strategy in primary mitochondrial disease: Evidence from patient fibroblasts and clinical observations » [Molecular Genetics and Metabolism Volume 146, Issues 1-2 (2025) Pages 109197].** Mol Genet Metab 146, 109219, doi:[10.1016/j.ymgme.2025.109219](https://doi.org/10.1016/j.ymgme.2025.109219).

Preston, Graeme, Jacob, N., Elsharkawi, I., Morava, E. et Kozicz, T. (2025). **Phosphodiesterase type 5 inhibition as a therapeutic strategy in primary mitochondrial disease: Evidence from patient fibroblasts and clinical observations.** Mol Genet Metab 146, 109197, doi:[10.1016/j.ymgme.2025.109197](https://doi.org/10.1016/j.ymgme.2025.109197).

Qu, Y., Edwards, K., Li, M., Williams, M., Liu, Y., Tsai, P.-Y., Cheng, C., Blum, J., Acor, N., Oshoe, T., Walter, C., Thirumalaikumar, V., Thalacker-Mercer, A., Skirycz, A. et Barrow, J. J. (2025). **Small molecule oxybutynin rescues proliferative capacity of complex III-defective muscle progenitor cells.** Am J Physiol Cell Physiol 329, C911-C923, doi:[10.1152/ajpcell.00141.2025](https://doi.org/10.1152/ajpcell.00141.2025).

Ramakumar, V., Akkineni, K. P., Gawalkar, A. et Agstam, S. (2025). **Recurrent Syncope and Drooping Eyes in a Young Woman: Kearns-Sayre Syndrome.** JACC Clin Electrophysiol S2405-500X(25)00554-7, doi:[10.1016/j.jacep.2025.06.034](https://doi.org/10.1016/j.jacep.2025.06.034).

Ramya, R. P., Megha, K. B., Reshma, S., Krishnan, M. J. A., Amir, S., Sharma, R. et Mohanan, P. V. (2025). **Mitochondrial disease management through phytochemical interventions.** Mol Cell Biochem, doi:[10.1007/s11010-025-05360-6](https://doi.org/10.1007/s11010-025-05360-6).

Reinsch, J. L. et Garcia, D. M. (2025). **Concurrent detection of chemically modified bases in yeast mitochondrial tRNAs by Nanopore direct RNA sequencing.** bioRxiv 2025.05.09.653160, doi:[10.1101/2025.05.09.653160](https://doi.org/10.1101/2025.05.09.653160).

Ribeiro, P. V. Z., Pari Mitre, L., Gauza, M. M., Furukawa, P. H., Ferraz, V. E. D. F. et Gomy, I. (2025). **Therapeutic benefit of idebenone in Leber hereditary optic neuropathy: a systematic review and meta-analysis.** Ophthalmic Genet 1-6, doi:[10.1080/13816810.2025.2521647](https://doi.org/10.1080/13816810.2025.2521647).

Roesch, S., O'Sullivan, A., Tschani, S., Baghdasaryan, A., Balasubramaniam, S., Barić, I., de Boer, L., Grünert, S. C., Guzek, A., Janssen, M., Krumina, Z., Koenig, M. K., Lewkowicz, A. M., Mochel, F., Naldi, A. M., Plecko, B., Öztürk, K., O'Grady, L., Riordan, G., Rymen, D., Sahai, I., Santer, R., Schiff, M., Stettner, G. M., Tsiakas, K., Uçar, S. K., Uzun, Ö. Ü., Weigel, C., Witters, P., Merkevicus, K., Mayr, J. A., Wortmann, S. B. et Iwanicka-Pronicka, K. (2025). **Hearing rehabilitation in SERAC1 related MEGD(H)EL syndrome - implications from a multi-center retrospective cohort study.** Mol Genet Metab 146, 109193, doi:[10.1016/j.ymgme.2025.109193](https://doi.org/10.1016/j.ymgme.2025.109193).

Mitochondrial disorders : The Essential

MMITO-2025-3 from july 1st to september 30th, 2025

Rossi, V., Brooks, D., Dai, H., Mizerik, E., Salazar, K., Davila-Williams, D., Ben-Moshe, Y., Lalani, S. R., Elsea, S. H., Gijavanekar, C., Scott, D. A., Machol, K., Bekheirnia, M. R. et Scaglia, F. (2025). **A Rare Molecular Diagnosis in a Patient With Hepatocerebral Syndrome Contributes to the Expansion of the Phenotypic Spectrum of POLG2 - Related Mitochondrial Disorder.** Am J Med Genet A 197, e64177, doi:[10.1002/ajmg.a.64177](https://doi.org/10.1002/ajmg.a.64177).

Saha, S., Jha, A., Yadaw, M. et Tiwari, B. (2025). **Hypertrophic cardiomyopathy with ataxic gait: a cardiac clue to a neurologic diagnosis.** BMJ Case Rep 18, e265662, doi:[10.1136/bcr-2025-265662](https://doi.org/10.1136/bcr-2025-265662).

Salongo, L., Nguyen, B. et Crawford, J. R. (2025). **Neuroimaging features of familial MT-TK related mitochondrial disease in a child.** BMJ Case Rep 18, e265230, doi:[10.1136/bcr-2025-265230](https://doi.org/10.1136/bcr-2025-265230).

Sane, I. A. et Gupte, J. R. (2025). **Diagnosing Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-Like Episodes (MELAS) Syndrome in a Young Adult Female Patient With Seizures and Lactic Acidosis.** Cureus 17, e88031, doi:[10.7759/cureus.88031](https://doi.org/10.7759/cureus.88031).

Sangeeth, T. A., Viswanathan, L. G., Dv, S., Rao, S., Nagappa, M., Saini, J. et Sinha, S. (2025). **Bilateral epilepsy partialis continua in POLG related mitochondrial disease.** Seizure 131, 200-202, doi:[10.1016/j.seizure.2025.07.004](https://doi.org/10.1016/j.seizure.2025.07.004).

Sasaki, T., Nakashima, D. et Tosaki, T. (2025). **Response to « Diagnose autosomal recessive Alport syndrome in an m.3243A>G carrier only if a pathogenic COL4A3 variant has been detected ».** Intern Med, doi:[10.2169/internalmedicine.6277-25](https://doi.org/10.2169/internalmedicine.6277-25).

Schofield, D., Kraindler, J., Lim, K., Tan, O., Haque, S., Crawley, K., West, S., Percival, A., Parmar, J., Shrestha, R. et Sue, C. (2025). **Health-related quality of life in patients with mitochondrial disease and their carers.** J Med Genet jmg-2025-110896, doi:[10.1136/jmg-2025-110896](https://doi.org/10.1136/jmg-2025-110896).

Seethashankar, S., Hariharan, S. et Parvathi, V. D. (2025). **Implications of mtDNA in human health and diseases.** BioTechnologia (Pozn) 106, 209-222, doi:[10.5114/bta/204532](https://doi.org/10.5114/bta/204532).

Selamioglu, A., Altun, M. A., Bliven, K., Ünverengil, G., Güneş, D., Karaca, M., Balci, M. C., Gedikbaşı, A., Atalar, F. et Gökçay, G. (2025). **Expanding the Clinical Spectrum of Mitochondrial Phosphate Carrier Deficiency: A Case Report With Literature Review.** Am J Med Genet A e64259, doi:[10.1002/ajmg.a.64259](https://doi.org/10.1002/ajmg.a.64259).



Mitochondrial disorders : The Essential

MMITO-2025-3 from july 1st to september 30th, 2025

Sereikaite, L., Vilkeviciute, A., Glebauskiene, B., Traberg, R., Gelzinis, A., Piskiniene, R., Zemaitiene, R., Ugenskiene, R. et Liutkeviciene, R. (2025). **First Case in Lithuania of an Autosomal Recessive Mutation in the DNAJC30 Gene as a Cause of Leber's Hereditary Optic Neuropathy.** Genes (Basel) 16, 993, doi:[10.3390/genes16090993](https://doi.org/10.3390/genes16090993).

Sgobbi, P., Farias, I. B., Serrano, P. de L., Badia, B. de M. L., Oliveira, H. B. de, Barbosa, A. S., Pereira, C. A., Moreira, V. de F., Chieia, M. A. T., Barbosa, A. R., Fraiman, P. H. A., Braga, V. L., Machado, R. I. L., Calegaretti, S. L., Fernandes, I. D., Ribeiro, R. C., Orsini Neves, M. A., Pinto, W. B. V. de R. et Oliveira, A. S. B. (2024). **PNPT1 Spectrum Disorders: An Underrecognized and Complex Group of Neurometabolic Disorders.** Muscles 3, 4-15, doi:[10.3390/muscles3010002](https://doi.org/10.3390/muscles3010002).

Sharma, S., Peterson, J., Alves, C. A., Xiao, R. et Goldstein, A. (2025). **Neuroimaging patterns in patients with mitochondrial leukoencephalopathies.** J Neurol Sci 476, 123605, doi:[10.1016/j.jns.2025.123605](https://doi.org/10.1016/j.jns.2025.123605).

Shibata, K.-I., Mukai, T., Nakagaki, H. et Nagano, S. (2025). **[A case of a spinal cord involvement in an adult mitochondrial disease with an m.3243A>G].** Rinsho Shinkeigaku 65, 578-581, doi:[10.5692/clinicalneuro.002071](https://doi.org/10.5692/clinicalneuro.002071).

Skocy, H., McFerren, A., Cairns, D., Kenney, H. M., Tallis, E., Dhamoon, A. S., Garbade, E. et Mackenzie, S. J. (2025). **TANGO2 deficiency disorder in a 61-year-old male with episodic weakness, rhabdomyolysis, myotonia, and a novel missense variant.** Mol Genet Metab Rep 44, 101241, doi:[10.1016/j.ymgmr.2025.101241](https://doi.org/10.1016/j.ymgmr.2025.101241).

Sobeh, T., Granek, T., Bar-Yosef, O., Jacoby, E., Hoffmann, C. et Shrot, S. (2025). **Neuroimaging characteristics of single Large-Scale mitochondrial DNA deletion syndromes.** Neuroradiology, doi:[10.1007/s00234-025-03689-9](https://doi.org/10.1007/s00234-025-03689-9).

Spilsbury, K., Wu, J., Reidy, M. et Kropp, P. A. (2025). **The mitochondrial trans-2-enoyl-coA reductase is necessary for mitochondrial homeostasis in C. elegans.** Genetics iyaf166, doi:[10.1093/genetics/iyaf166](https://doi.org/10.1093/genetics/iyaf166).

Staedler, K., Nectoux, J., Metay, C., Lermine, A., Villar-Quiles, R.-N., Evangelista, T., Labasse, C., Lacène, E. et Stojkovic, T. (2025). **A novel XPNPEP3 gene variant manifesting as rhabdomyolysis and exercise intolerance.** J Neuromuscul Dis 22143602251352986, doi:[10.1177/22143602251352986](https://doi.org/10.1177/22143602251352986).

Théberge, E., Code, J., Khoo, J. K., Lai, C., Ignaszewski, A., Virani, S., Toma, M., Roston, T. M., Sedlak, T. et Lehman, A. (2025). **Nonischemic Cardiomyopathy in Adult-Onset PPA2-Deficient Mitochondrial Disease.** JACC Case Rep 30, 104676, doi:[10.1016/j.jaccas.2025.104676](https://doi.org/10.1016/j.jaccas.2025.104676).

Mitochondrial disorders : The Essential
MMITO-2025-3 from july 1st to september 30th, 2025

Timón-Gómez, A., Doerrier, C., Sumbalová, Z., Garcia-Souza, L. F., Baglivo, E., Cardoso, L. H. D. et Gnaiger, E. (2025). **Bioenergetic profiles and respiratory control in mitochondrial physiology: Precision analysis of oxidative phosphorylation.** Exp Physiol, doi:[10.1113/EP092792](https://doi.org/10.1113/EP092792).

Ueda, M., Tanaka, N., Momota, Y. et Kawaguchi, M. (2025). **Anesthetic Management With Remimazolam for Adolescent Mitochondrial Encephalomyopathy With Lactic Acidosis and Stroke-like Episodes (MELAS): A Case Report.** Anesth Prog 72, 46-48, doi:[10.2344/23-0051](https://doi.org/10.2344/23-0051).

Ueda, N. K., Mimaki, M., Ito, S., Murakami, A., Yokoi, S., Nishino, I., Katsuno, M. et Goto, Y.-I. (2025). **A novel m.14677 T > C variant in mitochondrial tRNAGlu gene causes chronic progressive external ophthalmoplegia.** J Hum Genet 70, 537-540, doi:[10.1038/s10038-025-01381-7](https://doi.org/10.1038/s10038-025-01381-7).

van Tienen, F. H. J., Hoeijmakers, J. G. J., van der Leij, C., Timmer, E., Wanders, N., Lindsey, P. J., Yi, F., Lin, F., Kortekaas, S. P. M., Roelofs, H., Westra, I. M., Meij, P., Wijnen, L. A. C. M., de Coo, I. F. M. et Smeets, H. J. M. (2025). **Intra-arterial transplantation of autologous mesoangioblasts in m.3243A>G mutation carriers is safe: First phase 1/2 human clinical study.** Mol Ther 33, 5061-5072, doi:[10.1016/j.ymthe.2025.07.005](https://doi.org/10.1016/j.ymthe.2025.07.005).

Villa-Villegas, L., Lira-Jaime, L. G., Farías-Moreno, K. C., González-Ruffino, B. D., Soto-Escageda, A., Mercado-Pimentel, R., Piña-Avilés, C. E. et Zúñiga-Ramírez, C. (2025). **Deep Brain Stimulation in Leigh-Like Syndrome Due to DNMT1 Pathogenic Variant.** Tremor Other Hyperkinet Mov (N Y) 15, 32, doi:[10.5334/tohm.1017](https://doi.org/10.5334/tohm.1017).

Wanagat, J., Musci, R., Herbst, A. et Aiken, J. M. (2025). **Mitochondrial DNA Deletion Mutations: A Molecular Cause of Age-Induced Skeletal Muscle Fiber Dysfunction and Fiber Death Contributing to Sarcopenia.** Adv Exp Med Biol 1478, 343-363, doi:[10.1007/978-3-031-88361-3_14](https://doi.org/10.1007/978-3-031-88361-3_14).

Wang, J., Li, H., Lv, N., Wang, M. et Zhao, H. (2025). **Successful treatment of congenital sideroblastic anemia with low-dose decitabine in a patient with NDUFB11 gene mutation (c.276_278del): A case report.** Curr Res Transl Med 73, 103542, doi:[10.1016/j.retram.2025.103542](https://doi.org/10.1016/j.retram.2025.103542).

Wang, L., Zhou, T., Bu, X., Pan, D. et Liu, X. (2025). **Hypoparathyroidism in a Child with MELAS Syndrome: A Case Report of Severe Lactic Acidosis and Symmetrical Bilateral Basal Ganglia Calcification.** Int J Endocrinol Metab 23, e161585, doi:[10.5812/ijem-161585](https://doi.org/10.5812/ijem-161585).

Mitochondrial disorders : The Essential

MMITO-2025-3 from july 1st to september 30th, 2025

Wang, Q., You, C., Qu, X., Zhang, Y., Bai, Y., Lin, X., Wang, Z., Fang, H., Lyu, J., Jiang, M. et Wang, Y. (2025). **Mitochondrial DNA genotypes modify m.3243A>G-associated mitochondrial disease via the 15-HETE/Akt/FoxO1 pathway.** Biochim Biophys Acta Mol Cell Res 1872, 120012, doi:[10.1016/j.bbamcr.2025.120012](https://doi.org/10.1016/j.bbamcr.2025.120012).

Ward, A. S., Kamath, V. G., Hsiung, C.-H., Lizenby, Z. J., Gillish, A. G., Kohtz, D. S. et McKee, E. E. (2025). **Compartmentalized thymidine phosphorylation by mitochondrial nucleotide kinases TK2 and CMPK2.** J Biol Chem 301, 110733, doi:[10.1016/j.jbc.2025.110733](https://doi.org/10.1016/j.jbc.2025.110733).

Warwick, A. M., Yu, L., Klingeborn, M., Travis, A. M., Parmar, V. M., Bomze, H. M., Malek, G. et Gospe lii, S. M. (2025). **Reduced complex I activity in the retinal pigment epithelium, but not in rod photoreceptors, affects light signaling without impacting cell survival.** J Biol Chem 301, 110586, doi:[10.1016/j.jbc.2025.110586](https://doi.org/10.1016/j.jbc.2025.110586).

Wasilewski, M., Mohanraj, K., Zakrzewski, M., Serwa, R. A. et Chacinska, A. (2025). **MitoRUSH as a tool to study the efficiency of mitochondrial import in complex I-deficient cells.** J Cell Sci 138, jcs263701, doi:[10.1242/jcs.263701](https://doi.org/10.1242/jcs.263701).

Watson-Fargie, T., Anderson, D. G., Stewart, W., Longman, C., Hopton, S., Ng, Y. S., Blakely, E. L., Taylor, R. W. et Farrugia, M. E. (2025). **Selective muscle MRI changes in a patient with a rare mitochondrial DNA variant causing myoclonic epilepsy with ragged red fibres.** Neuromuscul Disord 54, 106204, doi:[10.1016/j.nmd.2025.106204](https://doi.org/10.1016/j.nmd.2025.106204).

Xiang, J., Xu, W., Wu, J., Luo, Y., Liu, C., Hou, Y., Chen, J. et Yang, B. (2025). **Structural insights into DdCBE in action enable high-precision mitochondrial DNA editing.** Mol Cell 85, 3357-3372.e9, doi:[10.1016/j.molcel.2025.08.016](https://doi.org/10.1016/j.molcel.2025.08.016).

Zhang, X.-Y., Zhu, Q.-L., Ruan, G.-C. et Li, W.-B. (2025). **[Clinical and Intestinal Ultrasound Findings in Mitochondrial Neurogastrointestinal Encephalomyopathy: Report of One Case].** Zhongguo Yi Xue Ke Xue Yuan Xue Bao, doi:[10.3881/j.issn.1000-503X.16801](https://doi.org/10.3881/j.issn.1000-503X.16801).

Ziemian, M., Szmydtka, J., Snoch, W., Milner, S., Wojciechowski, S., Dłuszcakowska, A., Chojnowski, J. W., Pallach, Z., Żamojda, K., Węgrzyn, G. et Rintz, E. (2025). **Integrative Approaches to Myopathies and Muscular Dystrophies: Molecular Mechanisms, Diagnostics, and Future Therapies.** Int J Mol Sci 26, 7972, doi:[10.3390/ijms26167972](https://doi.org/10.3390/ijms26167972).

Zioudi, A., Gouiza, I., Galai, S., Hechmi, M., Klaa, H., Miladi, Z., Ben Younes, T., Benrhouma, H., Ben Youssef-Turki, I., Omar, S., Lenaers, G., Kefi, R., Gouider-Khouja, N. et Kraoua, I. (2025). **A Tunisian POLG mutation expands the clinical spectrum of POLG-related disorders.** Mitochondrion 85, 102071, doi:[10.1016/j.mito.2025.102071](https://doi.org/10.1016/j.mito.2025.102071).

Mitochondrial disorders : The Essential

MMITO-2025-3 from july 1st to september 30th, 2025

Zupin, L., Capaci, V., Bonati, M. T., Lamantea, E., Suleman, M., Marsala, A., Celsi, F., Spedicati, B., Crovella, S., Gortani, G., Girotto, G., Bruno, I. et Zeviani, M. (2025). **The homoplasmic MT-TK m.8357T>C mtDNA variant as a cause of multiorgan mitochondrial disease.** Mitochondrion 85, 102080, doi:[10.1016/j.mito.2025.102080](https://doi.org/10.1016/j.mito.2025.102080).