Mitochondrial disorders : The Essential

4th 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.

Summary

Ataxie de Friedreich - *Friedreich ataxia*

Ariello, L. E., Rastall, D. P. W. et Rosenthal, L. S. (2025). **Treatment of primary adult-onset neurodegenerative cerebellar ataxias**. Neurotherapeutics e00805, doi:10.1016/j.neurot.2025.e00805.

Asunis, E., Vitulli, F., Nicotra, R., Rubino, A., Cicala, D., Cinalli, G. et Colella, G. (2025). **Progressive scoliosis in a pediatric patient with cerebellar ataxia: surgical challenges and literature review**. Childs Nerv Syst *41*, 418, doi:10.1007/s00381-025-07091-x.

Bailly, C. (2026). **A new horizon for the anesthetic drug dyclonine**. Biochem Pharmacol *244*, 117569, doi:10.1016/j.bcp.2025.117569.

Casey, H. L., Shah, V. V., Muzyka, D., McNames, J., El-Gohary, M., Sowalsky, K., Safarpour, D., Carlson-Kuhta, P., Schmahmann, J. D., Rosenthal, L. S., Perlman, S., Rummey, C., Horak, F. B. et Gomez, C. M. (2025). **Digital Gait Measures Discriminate People with Friedreich's Ataxia from Healthy Controls**. Mov Disord Clin Pract, doi:10.1002/mdc3.70465.

Cordaro, M., Neri, G., Ansari, S. A. M. K., Buccheri, R., Scala, A. et Piperno, A. (2025). **Synthesis and Biological Profile of Omaveloxolone: The Cornerstone for Friedreich Ataxia Treatment**. Int J Mol Sci *26*, 9747, doi:10.3390/ijms26199747.

Das, S., Patel, M., Khandelwal, S., Rawat, R., Shukla, S., Kumari, A. P., Singh, K. et Kumar, A. (2025). **From Mutations to Microbes: Investigating the Impact of the Gut Microbiome on Repeat Expansion Disorders**. J Neurochem *169*, e70278, doi:10.1111/jnc.70278.

Devore, M. C., Lam, C., Wiley, G., Park, C. C., Lynch, D. R. et Bidichandani, S. I. (2025). **Unrecognized high prevalence of expanded composite repeats in Friedreich ataxia**. Hum Mol Genet ddaf190, doi:10.1093/hmg/ddaf190.

Kneer, K., Stahl, J.-H., Winter, N., Wittlinger, J., Männlin, S., Gasimli, T., Schöls, L., Fleszar, Z., Hayer, S. et Grimm, A. (2025). **Nerve Ultrasound in Patients With Friedreich Ataxia**. Muscle Nerve, doi:10.1002/mus.70091.

Kovacevic, G., Todorovic, S., Novakovic, I., Dobricic, V., Savic-Pavicevic, D., Milic Rasic, V., Svetel, M., Brkusanin, M., Vukomanovic, V., Vucinic, D., Ostojic, S., Putnik, J. et Kosac, A. (2025). **Multi-Center National Study of Genotype-Phenotype Correlation and Clinical Characteristics in Children and Young Adults with Friedreich's Ataxia from Serbia**. Biomedicines *13*, 2646, doi:10.3390/biomedicines13112646.

Kuo, S. H., Tian, C., McKay, J., England, S. M., Simon, M., Graf, M., Brewer, I. P., Land, N., Chou, J. W. et Lawson, R. (2025). **Delphi study to elicit expert consensus around decision-making in the treatment of Friedreich ataxia**. Front Neurol *16*, 1669059, doi:10.3389/fneur.2025.1669059.

Lima, S. M., Caltagirone, M., Messina, C., Quartetti, U., Rini, N., D'Amico, F., Brighina, F. et Di Stefano, V. (2025). **Early experience on omaveloxolone in adult patients with Friedreich's ataxia: a real-world observational study**. J Neurol *272*, 742, doi:10.1007/s00415-025-13487-1.

Lischewski, S. A., Dogan, I., Giunti, P., Parkinson, M. H., Mariotti, C., Durr, A., Ewenczyk, C., Boesch, S., Nachbauer, W., Klopstock, T., Stendel, C., de Rivera Garrido, F. J. R., Schöls, L., Fleszar, Z., Klockgether, T., Grobe-Einsler, M., Giordano, I., Rai, M., Pandolfo, M., Jacobi, H., Hilgers, R.-D., Schulz, J. B., Reetz, K., et EFACTS Study Group (2025). **Analysis of a Modified Version of the Inventory of Non-Ataxia Signs Over 12 Years in Patients with Friedreich's Ataxia in the EFACTS Study**. Mov Disord, doi:10.1002/mds.70084.

Machado, D. S., Silveira, C., Vinagre, A. M., Rezende, T. J. R., Dogini, D., Martinez, A. R. M. et França, M. J. C. (2025). **Survival in Brazilian Patients with Friedreich´s Ataxia**. Cerebellum *24*, 182, doi:10.1007/s12311-025-01936-6.

Meisel, J. D., Joshi, P. R., Spelbring, A. N., Wang, H., Wellner, S. M., Wiesenthal, P. P., Miranda, M., McCoy, J. G., Barondeau, D. P., Ruvkun, G. et Mootha, V. K. (2026). **Mutations in mitochondrial ferredoxin FDX2 suppress frataxin deficiency**. Nature *649*, 713-720, doi:10.1038/s41586-025-09821-2.

Mercado-Ayón, E., Lazaropoulos, M. P., Mercado-Ayón, Y. et Lynch, D. R. (2025). **Neuropathology of Friedreich ataxia and its links to metabolic pathways**. Neurodegener Dis Manag 1-12, doi:10.1080/17582024.2025.2607957.

Omaveloxolone (Skyclarys): Therapeutic area: Friedreich's ataxia: Reimbursement Review (2025). Canadian Agency for Drugs and Technologies in Health, Ottawa (ON), URL: http://www.ncbi.nlm.nih.gov/books/NBK619995/ [Consulté le janvier 2026].

Payne, R. M., O'Connell, T. M., Pride, P. M., Wagner, G. R., Eckert, G. J., Johnson, T. R., Shou, W. et Hutchins, G. D. (2025). **Positron emission tomography reveals increased myocardial glucose uptake in a subset of Friedreich ataxia patients**. Sci Rep *15*, 37247, doi:10.1038/s41598-025-21330-w.

Pernaci, C., Johnson, A., Gillette, S., Warden, A. S., McCormick, C., Weiser-Novak, S., Ramirez, G., Broersma, E. H., Mishra, P., Sivakumar, A., Cherqui, S. et Coufal, N. G. (2025). **Microgliopathy as a primary mediator of neuronal death in models of Friedreich's Ataxia**. Nat Commun *17*, 81, doi:10.1038/s41467-025-66710-y.

Ranaweera, K., Nguyen, B. A., Pathirana, P. N., Milne, S. C., Horne, M., Delatycki, M. B. et Corben, L. A. (2025a). **Enhancing the Objective Assessment of Friedreich Ataxia Severity: A Multiview IMU-Based Approach**. Annu Int Conf IEEE Eng Med Biol Soc *2025*, 1-6, doi:10.1109/EMBC58623.2025.11253596.

Ranaweera, K., Randeniya, M., Pathirana, P. N., Milne, S. C., Horne, M., Delatycki, M. B. et Corben, L. A. (2025b). **Reliable Objective Assessment of Friedreich Ataxia Through Isolation Forest-Based Anomaly Detection**. Annu Int Conf IEEE Eng Med Biol Soc *2025*, 1-6, doi:10.1109/EMBC58623.2025.11253335.

Ranaweera, K., Tran, M., Nguyen, B. A., Ngo, T., Pathirana, P. N., Milne, S. C., Horne, M., Delatycki, M. B. et Corben, L. A. (2025c). **Objective Assessment of Friedreich Ataxia in Children: Accounting for Developmental Deficits**. Annu Int Conf IEEE Eng Med Biol Soc *2025*, 1-7, doi:10.1109/EMBC58623.2025.11253250.

Reetz, K., Lischewski, S. A., Schulz, J. B., Praster, M., Pishnamaz, M., FACROSS study group, Dogan, I., Romanzetti, S., Dadsena, R., Konrad, K., Clavel, T., Jankowski, V., Jankowski, J., Pabst, O., Marx, N., Moellmann, J., Jacobsen, M., Marx-Schütt, K., Dukart, J., Eickhoff, S. et Hilgers, R.-D. (2026). **Scoliosis Surgery in a Patient With Advanced Friedreich's Ataxia-It Is Not Too Late**. Ann Clin Transl Neurol *13*, 200-203, doi:10.1002/acn3.70219.

Rummey, C., Farmer, J. M. et Lynch, D. R. (2025). **Domain Specific Placebo Response in the Modified Friedreich's Ataxia Rating Scale**. Ann Clin Transl Neurol, doi:10.1002/acn3.70239.

Sanz-Alcázar, A., Britti, E., Delaspre, F., Medina-Carbonero, M., Pazos-Gil, M., Tamarit, J., Ros, J. et Cabiscol, E. (2025). **Correction: Mitochondrial impairment, decreased Sirtuin activity and protein acetylation in dorsal root ganglia in Friedreich ataxia models**. Cell Mol Life Sci *82*, 428, doi:10.1007/s00018-025-05948-7.

Shafiei, A., Baldir, N., Na, J., Kim, J. H. et DeMirci, H. (2025). **Comparative Structural Analysis of Escherichia Coli Cyay at Room and Cryogenic Temperatures Using Macromolecular and Serial Crystallography**. Chembiochem *26*, e202500442, doi:10.1002/cbic.202500442.

Subramony, S. H. et Lynch, D. R. (2026). **Friedreich Ataxia**. Pediatr Neurol *174*, 148-154, doi:10.1016/j.pediatrneurol.2025.10.020.

Tang, H., Gupte, S., Xu, E., Calabro, K. R., Friend, H., Crosson, S. M., Fajardo, D., Kostamo, Z., Zhang, H., Peterson, J. J., Lin, F., Kozmik, Z., Lutz, C. M., Boye, S. L. et Boye, S. E. (2025). **Development of an AAV-based gene therapy for the ocular phenotype of Friedreich's ataxia**. Mol Ther S1525-0016(25)00869-X, doi:10.1016/j.ymthe.2025.10.048.

Viana, C. F., Jaques, C. S., Bezerra, M. L. E., Barsottini, O. G. P. et Pedroso, J. L. (2025). **Respiratory Function in Friedreich's Ataxia**. Mov Disord, doi:10.1002/mds.70162.

Want, K., Gorny, H., Turki, E., Noiray, M., Monfort, B., Mor-Gautier, R., Tubiana, T., Jullian, E., Monnier, V. et D'Autréaux, B. (2026). **Cross-regulation of [2Fe-2S] cluster synthesis by ferredoxin-2 and frataxin**. Nature *649*, 721-728, doi:10.1038/s41586-025-09822-1.

Wong, J., Kwok, J. et Kim, K. (2025). **Neuropsychiatric Challenges of Friedreich Ataxia in a Patient Residing in a Long-Term Care Facility**. Prim Care Companion CNS Disord *27*, 25cr03965, doi:10.4088/PCC.25cr03965.

Zhang, N. et Xu, Y. (2025). **The Impact of Calcitriol on Frataxin Levels in Friedreich's Ataxia Patients: Insights and Future Directions**. Mov Disord, doi:10.1002/mds.29933.

Zhong, R., Yang, H., Li, X., Wang, F., Zhai, L. et Gao, J. (2025). **Mitochondria-Mediated Mechanisms of Ferroptosis in Neurological Diseases**. Neurochem Res *50*, 354, doi:10.1007/s11064-025-04605-6.

Zhou, B. Y., Ren, N., Zhang, Y. Y. et Geng, J. (2025). **[A case of Friedreich Ataxia and left ventricular hypertrophy induced by FXN gene mutation]**. Zhonghua Xin Xue Guan Bing Za Zhi *53*, 1271-1274, doi:10.3760/cma.j.cn112148-20250917-00659.

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

Ding, X., Jiang, Y., Wang, X., Li, C., Kong, Z., Fu, L., Lei, Z., Cai, H., Dong, Y., Shi, C., Su, X., Kunath, T., Wang, Z., Buratto, D., Lin, A., Shao, J., Zhang, D., Liu, Z., Liang, P., Zhou, R. et Yan, Q. (2025). **Novel DNJ Derivative Ameliorates Cardiac Hypertrophy by Targeting OPA1 and Restoring Mitochondrial Health**. Circ Res, doi:10.1161/CIRCRESAHA.125.327407.

Edelmayer, M. V., Strasser, T., Jung, R., Sonntag, A., Jendritza, R., Tonagel, F., Peters, T., Wilhelm, H., Wilhelm, B. et Kelbsch, C. (2025). **Chromatic pupil campimetry as objective diagnostic tool for progressive optic neuropathies**. Doc Ophthalmol, doi:10.1007/s10633-025-10054-x.

Han, Y., Zhao, L., Ma, T. et Zheng, Q. (2025). **Visual Acuity Alone Is Not Enough: The Need for Multimodal Biomarkers in Dominant Optic Atrophy**. Clin Exp Ophthalmol, doi:10.1111/ceo.70028.

Jauregui, R., Diaz Curbelo, C., Galetta, S. L. et Grossman, S. N. (2025). **The Neuro-Ophthalmologic Manifestations of SPG7-Associated Disease**. J Pers Med *15*, 495, doi:10.3390/jpm15100495.

Murati Calderon, R. A., Landestoy, G. et Izquierdo, N. (2025). **Clinical and Genetic Findings in an Autosomal Dominant Optic Atrophy-Compatible Phenotype Harboring an OPA1 Variant: A Case Report**. Cureus *17*, e95622, doi:10.7759/cureus.95622.

Rufa, A., Plantone, D., Bargagli, A., Righi, D., Bacci, T., Serchi, V., Primiano, G., Gallus, G. N., Lopergolo, D., Pretegiani, E., Rosini, F., Locci, S., Tosi, G. M. et De Stefano, N. (2025). **Serum neuronal, glial and mitochondrial markers in autosomal dominant optic atrophy and Leber hereditary optic neuropathy**. Brain Commun *7*, fcaf446, doi:10.1093/braincomms/fcaf446.

Sampige, R., Seaborn, L. E. A., Pluenneke, M., Jyothi, A., Saland, S., Chinedu-Obi, C. M., Keehn, C. et Lee, A. G. (2025). **IT TAKES TWO TO TANGO: potential novel therapies for autosomal dominant optic atrophy**. Front Ophthalmol (Lausanne) *5*, 1688232, doi:10.3389/fopht.2025.1688232.

Valentin, K., Kustermann, M., Schneider, M. R., Aminfar, H., Vollnhofer, K., Wedrich, A., Stapf, C., Bertich, M., Ritter, M., Mendrina, T., Valcanover, D., Berger, W., Eckhard, M., Sombke, A., Lilja, S. V., Paquay, A., Rosensteiner, B., Schmidt, I., Bittner, R. E., Georgi, T. P., Pemp, B. et Schmidt, W. M. (2026). **A recurrent missense variant in the PPIB gene encoding peptidylprolyl isomerase B underlies adult-onset autosomal dominant optic atrophy**. Genet Med *28*, 101595, doi:10.1016/j.gim.2025.101595.

Wang, H., Shi, T., Wang, W., Li, J., Chen, Z., Lan, L., Xie, L., Xiong, F., Wang, D., Guan, J. et Wang, Q. (2025). **Distinct genetic patterns and natural history of OPA1-related auditory neuropathy in Chinese population**. Orphanet J Rare Dis *20*, 520, doi:10.1186/s13023-025-04040-4.

Wong, D. C. S., Makam, R. et Yu-Wai-Man, P. (2025). **Advanced therapies for inherited optic neuropathies**. Eye (Lond), doi:10.1038/s41433-025-04109-1.

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

Gilio, D., Pelosini, C., Magno, S., Venanzi, J. M., Daniotti, M., Paoli, M., Palladino, L., Sessa, M. R., Ricci, F., Procopio, E., Ceccarini, G. et Santini, F. (2025). **Expansion of the Phenotypic Spectrum of MNGIE: Lipodystrophy and Metabolic Alterations Associated with a p.Arg393_Val400dup TYMP Variant**. Int J Mol Sci *26*, 9751, doi:10.3390/ijms26199751.

Zeng, G., Liu, R., Li, X., Lv, Z., Cui, L., Zhang, X. et Wang, J. (2025). **Sequential development of three syndromes in a patient with m.3243A>G mutation: a case report**. Front Med, doi:10.1007/s11684-025-1186-7.

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

Ando, M., Higuchi, Y., Yuan, J.-H., Yoshimura, A., Yano, C., Hobara, T., Kojima, F., Hiramatsu, Y., Nozuma, S., Nakamura, T., Sakiyama, Y., Mitsui, J., Tsuji, S. et Takashima, H. (2026). **Nationwide Characterization of MFN2-Related CMT in 176 Japanese Patients: Clinical and Genetic Insights**. Ann Clin Transl Neurol *13*, 170-179, doi:10.1002/acn3.70218.

Bird, T. D. (2025). **Charcot-Marie-Tooth Hereditary Neuropathy Overview**. In: **GeneReviews®**, Éd. Adam, M. P., Bick, S., Mirzaa, G. M., Pagon, R. A., Wallace, S. E., et Amemiya, A. University of Washington, Seattle, Seattle (WA), URL: http://www.ncbi.nlm.nih.gov/books/NBK1358/ [Consulté le janvier 2026].

Bora, P., Zaman, M., Oviedo, S., Kutseikin, S., Madrazo, N., Mathur, P., Pannikkat, M., Krasny, S., Aldakhlallah, R., 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**. Proc Natl Acad Sci U S A *122*, e2517552122, doi:10.1073/pnas.2517552122.

Borisova, N. R., Emelyanova, A. A., Solovjeva, O. N., Balashova, N. V., Sidorova, O. P. et Bunik, V. I. (2025). **Relationships of GDAP1 Mutations to Disease Phenotype and Mechanisms of Therapeutic Action of Oxidative Metabolism Activators in a Patient with Charcot-Marie-Tooth Neuropathy Type 2K**. Biochemistry (Mosc) *90*, 1678-1697, doi:10.1134/S0006297925601911.

Cai, Q., Wang, H., Luo, R., Qian, X. et Zhou, Z. (2026). **A novel homozygous COX6A1 variant causes axonal charcot-marie-tooth disease, developmental delays and mitochondrial dysfunction**. J Hum Genet *71*, 91-96, doi:10.1038/s10038-025-01411-4.

Kavoosi, S., Karimi, Z., Moghaddam, N. A. et Ashtari, F. (2025). **Clinical and Genetic Variant Profile of Asian Charcot-Marie Tooth Patients: A Systematic Review**. Iran J Public Health *54*, 2199-2211, doi:10.18502/ijph.v54i10.20138.

Molaei, N., Alagha, P., Khanbazi, A., Beheshtian, M., Ahangari, F., Dehdahsi, S., Fadaee, M., Ashki, M., Ghaderi, Z., Elahi, Z., Vazehan, R., Parsimehr, E., Nouri, M. M., Saei, P., Noudehi, K., Fatehi, Fatemeh, Najafabadi, S. Z., Abolhassani, A., Afroozan, F., Yazdan, H., Kelishomi, M. A., Azad, M., Parvini, F., Kassaee, S. M., Ramezani, M., Zemorshidi, F., Salimipour, H., Abdi, S., Bakhshandeh, M., Fayyazi, A., Zamani, G., Ashrafi, M. R., Jamali, P., Sarraf, P., Okhovat, A. A., Ashtiani, B. H., Fatehi, Farzad, Karimzadeh, P., Nafissi, S., Kahrizi, K., Kariminejad, A. et Najmabadi, H. (2025). **Genetic spectrum among 2009 Iranian individuals with neuromuscular disorders using next generation sequencing and multiple ligation dependent probe amplification methods**. Sci Rep *15*, 42736, doi:10.1038/s41598-025-26836-x.

Raga, S. V., Kandawasvika, G. Q., Ndondo, A., Hanna, M. G., Reilly, M. M., Record, C. J., Krause, A., Essop, F., Esterhuizen, A. et Wilmshurst, J. M. (2025). **Expression of genetic peripheral neuropathies in South African children**. Neuromuscul Disord *59*, 106283, doi:10.1016/j.nmd.2025.106283.

Rizzuti, M., Pagliari, E., D'Agostino, M., Ottoboni, L., Parente, V., Comi, G. P., Corti, S., Rizzo, F. et Abati, E. (2025). **Advances and challenges in modeling Charcot-Marie-Tooth type 2A using iPSC-derived models**. Stem Cell Reports *20*, 102711, doi:10.1016/j.stemcr.2025.102711.

van Paassen, B. W., Verhamme, C., van Ruissen, F., Haagmans, M. A., Dijkgraaf, M. G. W., De Visser, M., Baas, F., Van Der Kooi, A. J. et Weterman, M. A. J. (2025). **Search for Additional Pathogenic Variants to Explain Variation in PMP22-Related Neuropathies**. Neurol Genet *11*, e200315, doi:10.1212/NXG.0000000000200315.

Yano, C., Nakamura, T., Ando, M., Higuchi, Y., Yuan, J.-H., Yoshimura, A., Hobara, T., Kojima, F., Hiramatsu, Y., Nozuma, S., Sakiyama, Y., Hashiguchi, A., Rosales, R. L., Arimura, K. et Takashima, H. (2025). **Tibial to ulnar nerve amplitude ratio as a marker of length-dependent neuropathy**. Clin Neurophysiol Pract *10*, 499-506, doi:10.1016/j.cnp.2025.10.006.

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

No results

Maladie liée au gène GFM1 – GFM1-related disorder

No results

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

Casas, R. B., Risato, G., Zuppardo, A., Viscomi, C., Argenton, F., Doimo, M., Facchinello, N. et Tiso, N. (2025). **Model organisms in POLG-related disorders: insights from yeast to multicellular systems**. Cell Death Dis, doi:10.1038/s41419-025-08366-6.

Kalampokini, S., Keramidiotis, I., Ravanidis, S., Lochner, P., Kimiskidis, V. K. et Hadjigeorgiou, G. M. (2025). **A Scoping Review of POLG-Related Cerebellar Ataxia: Insights and Clinical Perspectives**. Tremor Other Hyperkinet Mov (N Y) *15*, 55, doi:10.5334/tohm.1027.

Kristensen, E., Naess, K., Engvall, M., Klingenberg, C., Rasmussen, M., Brodtkorb, E., Ostergaard, E., de Coo, I., Pias-Peleteiro, L., Isohanni, P., Uusimaa, J., Majamaa, K., Kärppä, M., Martikainen, M. H., Ortigoza-Escobar, J. D., Tangeraas, T., Berland, S., Sue, C. M., Walker, J. S., Harrison, E., Biggs, H., Horvath, R., Darin, N., Rahman, S. et Hikmat, O. (2025). **Liver Involvement in POLG Disease-a Multicentre Cohort Study of 202 Patients**. J Inherit Metab Dis *48*, e70112, doi:10.1002/jimd.70112.

Kuznetsova, N. A., Kochegarova, K. K., Baikova, I. P., Korshunov, E. N., Ilchuk, L. A., Kubekina, M. V., Bruter, A. V., Silaeva, Y. Y. et Varlamova, E. A. (2025). **Ubiquitous Expression of mPolgmut Leads to the Accumulation of Cytotoxic CD8+ T Lymphocytes in Young Mice**. Life (Basel) *15*, 1863, doi:10.3390/life15121863.

Mathavan, Akshay, Rathi, K., Mundhra, L. J. et Mathavan, Akash (2025). **Juvenile/adult-onset POLG-related disease unmasked by valproate-associated fulminant hepatic failure**. BMJ Case Rep *18*, e269373, doi:10.1136/bcr-2025-269373.

Meyrick, J., Stefanetti, R. J., Errington, L., McFarland, R., Gorman, G. S. et Lax, N. Z. (2023). **Model systems informing mechanisms and drug discovery: a review of POLG-related disease models**. Wellcome Open Res *8*, 33, doi:10.12688/wellcomeopenres.18637.2.

Molla, G. K., Uzun, Ö. Ü., Agakisili, H. B., Genç, E., Gülten, Z. A., Yıldırım, T., Ersoy, A. S., Ak, B., Gülbahçe, A., Yıldız, S., Çakar, N. E., Karaca, M., Zübarioğlu, T., Hişmi, B. Ö., Erdöl, Ş., Önal, H., Kara, B. et Gökçay, G. F. (2025). **Profiles of paediatric patients experiencing stroke-like episodes associated with mitochondrial disease**. Front Neurol *16*, 1657852, doi:10.3389/fneur.2025.1657852.

Wang, J., Tan, C., Deng, F., Liu, X. et Chen, L. (2025). **A case of POLG-related mitochondrial DNA maintenance defect**. Acta Neurol Belg, doi:10.1007/s13760-025-02922-9.

Zhang, Z., Yangzom, T., Lu, N., Deng, S., Xiao, X., Yang, G. et Liang, K. X. (2026). **Metformin Restores Mitochondrial Function and Neurogenesis in POLG Patient-Derived Brain Organoids**. Adv Sci (Weinh) *13*, e17721, doi:10.1002/advs.202417721.

Zhou, F., Chen, J., Zhang, T., Niu, F., Hu, J., Xu, Y. et Zhang, M. (2025). **Characterization of Novel POLG Mutations in Mitochondrial Encephalomyopathy: Pathogenic Validation and Comprehensive Genetic Profiling**. Brain Behav *15*, e71045, doi:10.1002/brb3.71045.

MELAS

Chu, K.-Y. (2025). **Mitochondrial DNA A3243G variant: Current perspectives and clinical implications**. Intractable Rare Dis Res *14*, 249-257, doi:10.5582/irdr.2025.01051.

Finsterer, J. (2025). **Fatal Methaemoglobin Intoxication Following Ketamine Infusion in a Depressed MELAS Patient: A Possible Association?** Case Rep Crit Care *2025*, 5200754, doi:10.1155/crcc/5200754.

Finsterer, J. (2026). **MELAS should only be diagnosed if the Hirano or Japanese criteria are met**. Radiol Case Rep *21*, 863-864, doi:10.1016/j.radcr.2025.10.089.

Goel, K., Sinha, N., Singh, D., Jambagi, S. A., Bhargav, R. et Kansagara, H. (2025). **Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-like Episodes: A Masquerader of Young-onset Stroke**. J Assoc Physicians India *73*, 24-26, doi:10.59556/japi.73.1235.

González Escobar, A. B. (2025). **Retinopathy associated with MELAS syndrome. A case report**. Arch Soc Esp Oftalmol (Engl Ed) *100*, 810-811, doi:10.1016/j.oftale.2025.10.008.

Janiak, J., Piątkowska, W., Podlejska, P., Zakrzewski, D., Zawadzka, M., Sawicka, A. et Mazurkiewicz-Bełdzińska, M. (2025). **Atypical clinical manifestation of MT-TL1 mutation in 6 months old patient**. Acta Neurol Belg, doi:10.1007/s13760-025-02921-w.

Kawano, S., Saegusa, C., Masano, Y., Becker, F., Nakamura, M., Shiozawa, S., Fujikura, J., Toyohara, T., Abe, T., Okano, H. et Fujioka, M. (2025). **Modeling Mitochondrial Disease Using Brain Organoids: A Focus on Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-like Episodes**. J Vis Exp, doi:10.3791/69303.

Kim, C.-H., Lee, J. K., Park, H.-R. et Choi, B. Y. (2026). **Personalized Anesthesia Strategies for Cochlear Implantation: Insights on Local Anesthesia From a Single-institution Experience**. Otol Neurotol *47*, e19-e22, doi:10.1097/MAO.0000000000004655.

Mascarella, D., Zhuang, Z. A., Gliga, O., Santis, F. D., Rosignoli, C., Daputke, A., Njohjam, M. N., Mazzotta, E., Adewinle, F. E., Caronna, E., Pozo-Rosich, P., et International Headache Academy of the International Headache Society (IHS-iHEAD) (2025). **Migraine in monogenic disorders: Shedding light on new therapeutic targets**. Cephalalgia *45*, 3331024251389241, doi:10.1177/03331024251389241.

Medrano, P., Banderas, B., Brimmer, M., Settel, L., Berger, S., Shields, A., Goldstein, A., Karaa, A., Larson, A., Parikh, S., Scaglia, F., Harrington, K. D., Edgar, C. J., Ventola, P., Webster, M., Chickering, J., Gwaltney, C., Wilson, P. et Glasser, C. (2025). **Signs, symptoms, and health-related quality of life in MELAS: measuring what's important from the patient and clinician perspectives**. J Patient Rep Outcomes *9*, 127, doi:10.1186/s41687-025-00962-6.

Molla, G. K., Uzun, Ö. Ü., Agakisili, H. B., Genç, E., Gülten, Z. A., Yıldırım, T., Ersoy, A. S., Ak, B., Gülbahçe, A., Yıldız, S., Çakar, N. E., Karaca, M., Zübarioğlu, T., Hişmi, B. Ö., Erdöl, Ş., Önal, H., Kara, B. et Gökçay, G. F. (2025). **Profiles of paediatric patients experiencing stroke-like episodes associated with mitochondrial disease**. Front Neurol *16*, 1657852, doi:10.3389/fneur.2025.1657852.

Othmani, N. B. et Mathew, S. (2025). **Clinical insights into mitochondrial retinopathy: A case report on m.3243A>G mutation and macular dystrophy**. Saudi J Ophthalmol *39*, 416-418, doi:10.4103/sjopt.sjopt_314_24.

Santhosh, T., Ramamurthy, P. et Simha, V. (2025). **Safe Use of Metformin in Mitochondrial Diabetes in Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-Like Episodes: Insights From a Patient With m.3243A>G Mutation**. AACE Endocrinol Diabetes *12*, 150-152, doi:10.1016/j.aed.2025.07.001.

Sevencan, Y., Rode, S., Park, A., Levin, I., Masri, D. et Derman, A. (2025). **From stroke workup to mitochondrial disease: A case report of MELAS**. Radiol Case Rep *20*, 6248-6252, doi:10.1016/j.radcr.2025.09.031.

Stefanetti, R. J., Charman, S. J., Newman, J., Hallsworth, K., Blain, A. P., Ng, Y. S. et Gorman, G. S. (2025). **Outcomes misaligned in mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes (MELAS): implications for trial design**. Brain Commun *7*, fcaf342, doi:10.1093/braincomms/fcaf342.

Uehara, N., Fujita, T., Uehara, K., Shimoda, H., Yamashita, D., Yokoi, J., Katsunuma, S., Kakigi, A. et Nibu, K.-I. (2025). **Clinical characteristics and hearing impairment in carriers of the m.3243 A > G variant**. J Hum Genet, doi:10.1038/s10038-025-01412-3.

Wakigawa, T., Mito, M., Ando, Y., Yamashiro, H., Tomuro, K., Tani, H., Tomizawa, K., Chujo, T., Nagao, A., Suzuki, Takeo, Nureki, O., Wei, F.-Y., Shichino, Y., Itoh, Y., Suzuki, Tsutomu et Iwasaki, S. (2025). **Monitoring the complexity and dynamics of mitochondrial translation**. Mol Cell *85*, 4279-4297.e8, doi:10.1016/j.molcel.2025.10.022.

Wang, R., Yu, Q., Sun, C., Liu, X., Hu, B., Yang, L., Geng, D., Lin, J. et Li, Y. (2025). **Altered connectome gradient and its association with gene expression profiles in MELAS patients with stroke-like episodes**. AJNR Am J Neuroradiol ajnr.A9154, doi:10.3174/ajnr.A9154.

Yang, X., Xu, Y., Zhang, S., Zhang, H., Suo, X., Tan, Q. et Yue, Q. (2025). **Multiparametric MRI detection of cerebral metabolism: a tool for early differentiation between MELAS and ischemic cerebral infarction**. Quant Imaging Med Surg *15*, 9466-9478, doi:10.21037/qims-24-2396.

Yu, Q., Wang, R., Sun, C., Hu, B., Liu, X., Yang, L., Lin, J., Geng, D. et Li, Y. (2025). **Dynamic reconfiguration and transition of whole-brain networks in patients with MELAS revealed by a hidden Markov model**. Front Neurol *16*, 1625888, doi:10.3389/fneur.2025.1625888.

Zeng, G., Liu, R., Li, X., Lv, Z., Cui, L., Zhang, X. et Wang, J. (2025). **Sequential development of three syndromes in a patient with m.3243A>G mutation: a case report**. Front Med, doi:10.1007/s11684-025-1186-7.

MERFF

Kalaria, T., Wanninayake, S. et Geberhiwot, T. (2025). **Lipomatosis in Myoclonic Epilepsy with Ragged Red Fibers Syndrome**. Ann Indian Acad Neurol *28*, 886-887, doi:10.4103/aian.aian_198_25.

Mi, L., Li, Y.-X., Lv, X., Wan, Z.-L., Liu, X., Zhang, K., Li, H., Yao, Y., Zhang, L., Xu, Z., Zhuang, X., Ji, K., Jiang, M., Wang, Y. et Lu, P. (2025). **Computational design of a high-precision mitochondrial DNA cytosine base editor**. Nat Struct Mol Biol *32*, 2575-2586, doi:10.1038/s41594-025-01714-2.

Rahman, A., Faiz, M., Khalid, M., Talha, M. et Waafira, A. (2025). **Gut instinct: microbiome as a modifiable target in the management of neurologic symptoms in myoclonic epilepsy with ragged-red fibers (MERRF)**. Ann Med Surg (Lond) *87*, 6904-6905, doi:10.1097/MS9.0000000000003777.

Zeng, G., Liu, R., Li, X., Lv, Z., Cui, L., Zhang, X. et Wang, J. (2025). **Sequential development of three syndromes in a patient with m.3243A>G mutation: a case report**. Front Med, doi:10.1007/s11684-025-1186-7.

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

Alnajashi, H. et Eltantawi, W. (2025). **Mitochondrial-Immune Overlap in Leber Hereditary Optic Neuropathy: A Case Report and Lessons Learned**. Reports (MDPI) *8*, 258, doi:10.3390/reports8040258.

Cao, S., Liu, Y., Sun, M., Zhang, Y., Sun, Y., Xu, Q., Wei, S. et Zhou, H. (2025). **Exploring rare mitochondrial DNA in Leber hereditary optic neuropathy**. Adv Ophthalmol Pract Res *5*, 278-284, doi:10.1016/j.aopr.2025.08.001.

Dong, Y., Hao, Z., Liu, Y., Chang, A., Porciatti, V., Lam, B. L., Jin, X. et Yu, H. (2025). **Preclinical Assessment of Mitochondrial-Targeted ND4 Gene Therapy for Leber Hereditary Optic Neuropathy**. Invest Ophthalmol Vis Sci *66*, 58, doi:10.1167/iovs.66.14.58.

Edelmayer, M. V., Strasser, T., Jung, R., Sonntag, A., Jendritza, R., Tonagel, F., Peters, T., Wilhelm, H., Wilhelm, B. et Kelbsch, C. (2025). **Chromatic pupil campimetry as objective diagnostic tool for progressive optic neuropathies**. Doc Ophthalmol, doi:10.1007/s10633-025-10054-x.

Finsterer, J. (2025). **Hearing Impairments Are Not Uncommon in LHON Patients**. Ear Nose Throat J 1455613251389432, doi:10.1177/01455613251389432.

Fiorini, C., Jurkute, N., Torraco, A., La Morgia, C., Ghezzi, D., Tioli, G., Rigobello, L., Ormanbekova, D., Berghella, A., Pietro Pasti, A., Palombo, F., Barboni, P., Lucia Cascavilla, M., Sadun, F., De Negri, A., Bertini, E., Musumeci, O., Ardissone, A., Rizza, T., Iarossi, G., Silvestri, G., Rossi, S., Altobelli, A., Moore, A. T., Cullup, T., Webster, A. R., Davagnanam, I., Michaelides, M., Malka, S., Ptackova, H., Stufkova, H., Tesarova, M., Liskova, P., Zeng, L., Klopstock, T., Kopajtich, R., Neuhofer, C., Prokisch, H., Lamperti, C., Sadun, A. A., Yu-Wai-Man, P., Carelli, V., Musiani, F., Iommarini, L., Carrozzo, R., Arno, G. et Caporali, L. (2025). **Recessive variants in mitochondrial Complex I nuclear subunits are an underrated cause of optic atrophy**. Brain awaf422, doi:10.1093/brain/awaf422.

Ha, D. H. et Kim, U. S. (2025). **The Pedigree Study of Leber's Hereditary Optic Neuropathy in Korean Population**. Semin Ophthalmol 1-5, doi:10.1080/08820538.2025.2588702.

Janigian, N. G. et Bakaeva, T. V. (2025). **Another One: A Novel Variant Associated With Leber Hereditary Optic Neuropathy**. J Neuroophthalmol, doi:10.1097/WNO.0000000000002430.

Kim, S., Kim, J., Cha, S., Ju, S., Lim, C. J., Hong, S., Bae, J., Oh, Y., Jung, S., Kim, S. P., Shin, H.-S., Yoon, J. H., Park, Jeeyoon, Ryou, S., Lim, S.-Y., Lee, S. B., Choi, S. H., Park, S.-J., Choi, C. G., Choi, M., Kim, L. K., Park, Jiyoon, Lee, S., Seo, K. Y., Seong, J. K., Kim, K., Kim, J.-S. et Lee, H. (2025). **In vivo mitochondrial base editing restores genotype and visual function in a mouse model of LHON**. Nat Commun *16*, 10923, doi:10.1038/s41467-025-66600-3.

Koch, A., Misiewicz, R., Marsh, M. et Poincenot, L. (2025). **Navigating Health When Sudden Blindness Occurs: The Experience of Leber Hereditary Optic Neuropathy**. Creat Nurs *31*, 422-432, doi:10.1177/10784535251388675.

Li, X., Yuan, J., Chen, Z., Zhang, Yue, Liu, Y. et Zhang, Yong (2025). **Development of Two In Vitro ND1-LHON Models for Evaluating Gene Therapy Efficacy**. Invest Ophthalmol Vis Sci *66*, 34, doi:10.1167/iovs.66.13.34.

Panusatid, C., Soiyangsuk, R., Tanadjindarat, M. et Peerapittayamongkol, C. (2025). **Metformin promotes mitochondrial integrity through AMPK-signaling in Leber's hereditary optic neuropathy**. FEBS Open Bio, doi:10.1002/2211-5463.70165.

Paranjpe, R., Yadav, H., Abraham, P. et Jakhalu, K. (2025). **Visual Recovery in Leber's Hereditary Optic Neuropathy Plus: A Case Report and Literature Insight**. Case Rep Ophthalmol *16*, 686-692, doi:10.1159/000547946.

Rufa, A., Plantone, D., Bargagli, A., Righi, D., Bacci, T., Serchi, V., Primiano, G., Gallus, G. N., Lopergolo, D., Pretegiani, E., Rosini, F., Locci, S., Tosi, G. M. et De Stefano, N. (2025). **Serum neuronal, glial and mitochondrial markers in autosomal dominant optic atrophy and Leber hereditary optic neuropathy**. Brain Commun *7*, fcaf446, doi:10.1093/braincomms/fcaf446.

Sadun, A. A., Gauthier, D. A. et Golmohammadi, M. (2025). **From Castro to Quantum Mechanics: Marching Through Tunnels With Reactive Oxygen Species**. J Neuroophthalmol *45*, 504-514, doi:10.1097/WNO.0000000000002400.

Shah, M. P., Chen, A. et Rizzo, J. F. (2025). **Reply: Leber's Hereditary Optic Neuropathy in a Nonagenarian**. J Neuroophthalmol *45*, 538-539, doi:10.1097/WNO.0000000000002399.

Vegda, M., Pandit, L., D'Souza, N., Agasti, N., Alva, S. et Alexander, L. (2025). **Differentiating Leber's Hereditary Optic Neuropathy from Optic Neuritis in a Clinical Setting**. Ann Indian Acad Neurol *28*, 920-922, doi:10.4103/aian.aian_451_25.

Wang, Y., Yu, H.-Y., Yi, Z.-J., Qi, L.-Y., Yang, J.-S., Xie, H.-X., Zhao, M., Liu, N.-H., Chen, J.-Q., Zhou, T.-J., Xing, L., Cheng, X.-W. et Jiang, H.-L. (2025a). **Super mitochondria-enriched extracellular vesicles enable enhanced mitochondria transfer**. Nat Commun *16*, 9448, doi:10.1038/s41467-025-64486-9.

Wang, Y., Zhao, M., Xie, H.-X., Yu, H.-Y., Yang, J.-S., Qie, L.-X., Liu, N.-H., Chen, J.-Q., Yi, Z.-J., Zhou, T.-J., Xing, L., Cheng, X. W. et Jiang, H.-L. (2025b). **Mitochondria-targeted gene delivery using fluorinated lipid nanoparticles to alleviate Leber's hereditary optic neuropathy**. Nat Commun *16*, 10891, doi:10.1038/s41467-025-65874-x.

Williams, J. J., Swan, R. T., Naum, A. M. et Mejico, L. J. (2025). **Comment: Leber's Hereditary Optic Neuropathy in a Nonagenarian**. J Neuroophthalmol *45*, 537-538, doi:10.1097/WNO.0000000000002398.

Wong, D. C. S., Makam, R. et Yu-Wai-Man, P. (2025). **Advanced therapies for inherited optic neuropathies**. Eye (Lond), doi:10.1038/s41433-025-04109-1.

Xie, Q., Wu, Haibo, Qiu, J., Liu, J., Jiang, X., Wu, Huihui, Lyu, Q., Long, H., Li, W., Zhang, S., Zhou, Y., Gao, Y., Hsueh, A. J. W., Kuang, Y. et Suo, L. (2025). **Trans-generational maintenance of mitochondrial DNA integrity in oocytes during early folliculogenesis**. PLoS Genet *21*, e1011562, doi:10.1371/journal.pgen.1011562.

Zhang, H., Shan, W., Yasheng, M. et Guan, M. (2025). **[Pathogenic mechanisms of Leber hereditary optic neuropathy caused by m.3472T>C mutation]**. Zhejiang Da Xue Xue Bao Yi Xue Ban *54*, 830-840, doi:10.3724/zdxbyxb-2025-0241.

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

Cicchinelli, M., Primiano, G., Canu, F., Gervasoni, J., Primiano, A., Santucci, L., Percio, A., Greco, V., Leoni, C., Sabino, A., Ardito, M., Zampino, G., Servidei, S., Urbani, A. et Iavarone, F. (2025). **Urinary Multi-Omics Profiling Reveals Systemic Molecular Alterations in Progressive External Ophthalmoplegia**. Int J Mol Sci *26*, 11257, doi:10.3390/ijms262311257.

Jauregui, R., Diaz Curbelo, C., Galetta, S. L. et Grossman, S. N. (2025). **The Neuro-Ophthalmologic Manifestations of SPG7-Associated Disease**. J Pers Med *15*, 495, doi:10.3390/jpm15100495.

Li, B. Y., Greenberg, J. H., Steigerwald, C., Bai, R., Tanji, K., Zakin, E. et Abreu, N. J. (2025). **Pearls and Oy-sters: Chronic Progressive External Ophthalmoplegia With Electrical Myotonia and Negative Initial Genetic Testing**. Neurology *105*, e214313, doi:10.1212/WNL.0000000000214313.

Pan, X., Wang, Y., Liu, N., Luo, X., Sutton, V. R., Craigen, W. J. et Sun, Q. (2025). **Detecting mitochondrial electron transport chain enzyme defects in low-heteroplasmy single large-scale mtDNA deletion syndromes (SLSMDSs)**. Mol Genet Metab *146*, 109260, doi:10.1016/j.ymgme.2025.109260.

Savino, G., Giannuzzi, F., Cima, V., Brando, D., Fossataro, C., Servidei, S., Savastano, M. C. et Primiano, G. (2025). **Ophthalmological signs and sensorimotor evaluation in mitochondrial chronic progressive external ophthalmoplegia: a multidisciplinary prospective study**. BMC Ophthalmol *25*, 520, doi:10.1186/s12886-025-04194-6.

Wang, J., Tan, C., Deng, F., Liu, X. et Chen, L. (2025). **A case of POLG-related mitochondrial DNA maintenance defect**. Acta Neurol Belg, doi:10.1007/s13760-025-02922-9.

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

No results

Syndrome de Barth – *Barth syndrome*

Abedin, Z. U., Waggan, A. I., Khan, E., Suleman, M. U. et Tabassum, S. N. (2025). **Letter to the Editor: CRISPR-based gene editing for cardiac protection in Barth syndrome**. Ann Med Surg (Lond) *87*, 9163-9164, doi:10.1097/MS9.0000000000004188.

Carlein, C., Hoffmann, M. D. A., Bickelmann, C., Amaral, A. G., Lotfinia, A., de Selliers, L.-A., Audouze-Chaud, J., Wrublewsky, S., Lauterbach, M. A., von der Malsburg, K., van der Laan, M., Bozem, M., Hoth, M., Gilon, P., Ravier, M. A., Morgan, B., Ampofo, E., Higuchi, T., Bertero, E., Dudek, J., Maack, C. et Roma, L. P. (2026). **Adaptive mechanisms in pancreatic islets counteract mitochondrial dysfunction in Barth syndrome**. Diabetologia *69*, 173-197, doi:10.1007/s00125-025-06575-4.

Dong, C., Finigan, A., Figg, N., Jenkins, B., Koulman, A., Chowdhury, S. R., Tricolici, A.-M., Lambert, J., Oc, S., Jørgensen, H. F., Prudent, J., Murphy, M. P., Bennett, M. et Yu, E. (2025). **Defective vascular smooth muscle cell tafazzin impairs mitochondrial function and promotes atherosclerosis in preclinical models**. Nat Commun *16*, 10909, doi:10.1038/s41467-025-65873-y.

Marjoram, L., Huang, Y., Koenig, M. K., Cohen, B. H. et Anderson, E. (2025). **Real-world disease burden and health care resource utilization for patients with Barth syndrome**. J Med Econ *28*, 2010-2026, doi:10.1080/13696998.2025.2588729.

Matias, C., Snider, P. L., Sierra Potchanant, E. A., Huot, J. R., Raghav, R., Chin, M. T., Conway, S. J. et Brault, J. J. (2025). **Deficient Cardiolipin Remodeling Alters Muscle Fiber Composition and Neuromuscular Connectivity in Barth Syndrome**. bioRxiv 2025.10.31.685856, doi:10.1101/2025.10.31.685856.

Schomakers, B. V., Passadouro, A. S., Trętowicz, M. M., Simpson, P. J., Jaspers, Y. R. J., van Weeghel, M., Hu, I. M., Lamboo, C. M. E., Cloutier, D., Byrne, B. J., van Klinken, J. B., Janssen, P. M. L., Piersma, S. R., Jimenez, C. R., Vaz, F. M., Salomons, G. S., van der Velden, J., Houtkooper, R. H. et Mosegaard, S. (2025). **Integrated multi-omics mapping of mitochondrial dysfunction and substrate preference in Barth syndrome cardiac tissue**. EMBO Mol Med *17*, 3227-3246, doi:10.1038/s44321-025-00320-5.

Shafaati, T., Greenwell, A. A., Saed, C. T., Tabatabaei Dakhili, S. A., Chan, J. S. F., Dong, L., Stenlund, M. J., Ferrari, S. R., Han, R., Kruger, J., Eaton, F., Gopal, K., Davidge, S. T., Oudit, G. Y. et Ussher, J. R. (2025). **Pharmacological increases in circulating ketones fail to alleviate the hypertrophic cardiomyopathy present in the Tafazzin knockdown mouse model of Barth syndrome**. J Pharm Pharm Sci *28*, 15688, doi:10.3389/jpps.2025.15688.

Shirley, M. (2025). **Elamipretide: First Approval**. Drugs, doi:10.1007/s40265-025-02269-8.

Vishwanath, S., Brockmeyer, A., Hornby, B., Thompson, W. R. et Vernon, H. J. (2025). **Barth syndrome: Natural history in infants and young children**. Mol Genet Metab *146*, 109270, doi:10.1016/j.ymgme.2025.109270.

Zhao, C., Zhuang, X. et Gao, J. (2026). **Elamipretide: The first cardiolipin-directed mitochondrial therapeutic for Barth syndrome approved under accelerated approval**. Drug Discov Ther *19*, 435-436, doi:10.5582/ddt.2025.01111.

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

Ehrenberg, M., Dotan, A., Gal-Or, O., Dotan, G., Ehrlich, R. et Sternfeld, A. (2025). **Choroidal neovascularization in a teenager with Kearns Sayre syndrome**. Ophthalmic Genet 1-2, doi:10.1080/13816810.2025.2572711.

Pan, X., Wang, Y., Liu, N., Luo, X., Sutton, V. R., Craigen, W. J. et Sun, Q. (2025). **Detecting mitochondrial electron transport chain enzyme defects in low-heteroplasmy single large-scale mtDNA deletion syndromes (SLSMDSs)**. Mol Genet Metab *146*, 109260, doi:10.1016/j.ymgme.2025.109260.

Wang, J., Song, M., Liu, Z., Xu, C., Zou, Y., Duan, X., Liu, Y., Zhang, W., Li, J. et Fang, F. (2025). **Genotype-Phenotype Correlations in Chinese Pediatric Patients With Single Large-Scale Mitochondrial DNA Deletion Disorders**. Clin Genet, doi:10.1111/cge.70089.

Syndrome de Leigh - *Leigh syndrome*

Ahmadi, Z. A., Cabrera-Orefice, A., Zaninello, M., Barth, E., Veronese, M., Rodenburg, R., Rugarli, E. I., Brodesser, S., Arnold, S. et Brandt, U. (2026). **Pathogenesis of mtDNA point mutation m.10191T>C affecting complex I function is a multifactorial process leading to metabolic remodeling of mitochondria**. Biochim Biophys Acta Mol Basis Dis *1872*, 168070, doi:10.1016/j.bbadis.2025.168070.

Biswas, S. R., Tomsick, P. L., Kelly, C., Lester, B. A., Milner, J. P., Henry, S. N., Soto, Y., Brindley, S., DeFoor, N., Morton, P. D. et Pickrell, A. M. (2025). **Impaired Complex I dysregulates neural/glial precursors and corpus callosum development revealing postnatal defects in Leigh syndrome mice**. EMBO Mol Med, doi:10.1038/s44321-025-00367-4.

Corbaz, S., Pibernus, D. A., Loos, M. A., Pérez, M. M., Buompadre, M. C., García, F. M., Rodriguez, M. P., Rugilo, C., Armeno, M., Seo, G. H., Khang, R., Alonso, C. N. et Aráoz, H. V. (2025). **From Severe Neonatal Encephalopathy to Slowly Neurologic Progressive Disease: Pyruvate Dehydrogenase Deficiency Related to PDHA1 Variants**. J Child Neurol 8830738251404115, doi:10.1177/08830738251404115.

de Mendonça, R. S., Arruda Santana, A. B., Azzoni, A. B. M., de Almeida, A. L. V., Castro, M. A. A., Lucato, L. T., Kok, F. et de Gusmao, C. M. (2025). **VPS13D-Related Disorders: Description of New Variant and Phenotypic Spectrum Based on Age of Onset**. Cerebellum *24*, 183, doi:10.1007/s12311-025-01938-4.

Finsterer, J. (2025a). **Knowledge of the underlying genetic defect and detailed phenotype can prevent complications from general anaesthesia in Leigh syndrome**. Indian J Anaesth *69*, 1432-1433, doi:10.4103/ija.ija_711_25.

Finsterer, J. (2025b). **Progressive spinal cord involvement in Leigh syndrome due to an NDUFV1 variant**. Radiol Case Rep *20*, 5929-5933, doi:10.1016/j.radcr.2025.08.031.

Fiorini, C., Jurkute, N., Torraco, A., La Morgia, C., Ghezzi, D., Tioli, G., Rigobello, L., Ormanbekova, D., Berghella, A., Pietro Pasti, A., Palombo, F., Barboni, P., Lucia Cascavilla, M., Sadun, F., De Negri, A., Bertini, E., Musumeci, O., Ardissone, A., Rizza, T., Iarossi, G., Silvestri, G., Rossi, S., Altobelli, A., Moore, A. T., Cullup, T., Webster, A. R., Davagnanam, I., Michaelides, M., Malka, S., Ptackova, H., Stufkova, H., Tesarova, M., Liskova, P., Zeng, L., Klopstock, T., Kopajtich, R., Neuhofer, C., Prokisch, H., Lamperti, C., Sadun, A. A., Yu-Wai-Man, P., Carelli, V., Musiani, F., Iommarini, L., Carrozzo, R., Arno, G. et Caporali, L. (2025). **Recessive variants in mitochondrial Complex I nuclear subunits are an underrated cause of optic atrophy**. Brain awaf422, doi:10.1093/brain/awaf422.

Granberry, M. T., Severance, T., Biedermann, G., Quinsey, C., Mazuru, D. G. et Bach, A. (2025). **Severe Chemoradiotherapy Toxicity in a Pediatric Patient with Leigh Syndrome and Grade IV Isocitrate Dehydrogenase-Mutant Astrocytoma: A Case Report**. Am J Case Rep *26*, e949191, doi:10.12659/AJCR.949191.

Jeon, M., Yang, S.-S., Lee, S. et Choi, J. Y. (2025). **From Congenital Torticollis to Leigh Syndrome: A Case Report of Diagnostic Evolution in an Infant**. Children (Basel) *12*, 1522, doi:10.3390/children12111522.

Li, Y., Wang, D., Zhou, M., Sun, H., Hong, S., Jiang, L. et Guo, Y. (2025). **Hypocitrullinemia as an Early Diagnostic Biomarker for MT-ATP6 Mitochondrial Diseases**. J Mol Neurosci *75*, 154, doi:10.1007/s12031-025-02440-6.

Magrinelli, F., Taylor, L. S., Sedighzadeh, S., Moualek, D., Severino, M., Grba, D. N., Alston, C. L., Champion, M., Tavasoli, A. R., Lascelles, K., Gowda, V. K., Srinivasan, V. M., Fateh, S. T., Kordi-Tamandani, M., Khajeh, A., Yaghoubi, S., Dominik, N., Babaei, M., Javadzadeh, M., Varaghchi, J. R., Miryounesi, M., Ghayoor Karimiani, E., Tazir, M., Ali Pacha, L., Bhatia, K. P., Taylor, R. W., Houlden, H. et Maroofian, R. (2025). **Biallelic NDUFA9 variants cause a progressive neurodevelopmental disorder with prominent dystonia and mitochondrial complex I deficiency**. Brain Commun *7*, fcaf369, doi:10.1093/braincomms/fcaf369.

Nakamura, K., Kishita, Y., Sugiura, A., Ozaki, K., Yatsuka, Y., Matsumoto, N., Okazaki, A., Prokisch, H., Maruyama, K., Iwasa, H., Murayama, K., Matsumoto, H., Ohtake, A., Shiraishi, Y. et Okazaki, Y. (2026). **Identification of Intronic Variants in NDUFA3 as a Cause of Leigh Syndrome by Whole Genome Sequencing and RNA Sequencing**. Neurol Genet *12*, e200330, doi:10.1212/NXG.0000000000200330.

Pieper, A. A. et Paul, B. D. (2025). **Hydrogen Sulfide Signaling in Neurodegenerative Movement Disorders**. Handb Exp Pharmacol, doi:10.1007/164_2025_757.

Pizzoccheri, R., Falchi, F. A., Alloni, A., Caldarulo, M., Camboni, T., Zambelli, F., Pavesi, G., Visentin, C., Camilloni, C., Sertic, S. et Briani, F. (2025). **Pathological PNPase variants with altered RNA binding and degradation activity affect the phenotype of bacterial and human cell models**. NAR Mol Med *2*, ugae028, doi:10.1093/narmme/ugae028.

Radder, S. et Radder, N. (2025). **Fatal Presentation of Leigh Syndrome in a Neonate: Comprehensive Neuroimaging Findings With MT-ND5 Mutation**. Cureus *17*, e96098, doi:10.7759/cureus.96098.

Sánchez-Benito, L., González-Torres, M., Fernández-González, I., Cutando, L., Royo, M., Compte, J., Vila, M., Jurado, S., Sanz, E. et Quintana, A. (2025). **Dysfunctional LHX6 pallido-subthalamic projections mediate epileptic events in a mouse model of Leigh syndrome**. J Clin Invest *135*, e187571, doi:10.1172/JCI187571.

Sun, Y., Wei, X., Xiao, B., Luo, Y., Wang, Y., Liu, R., Zhan, Y., Ye, X., Cai, X., Xu, S., Lyu, J., Fang, H. et Yu, Y. (2025). **Identification of novel NDUFA3 variants in a patient with mitochondrial disorders**. Pediatr Res, doi:10.1038/s41390-025-04403-4.

Toutain, G., Hoebeke, C., Gastaldi, M., Milh, M. et Chabrol, B. (2025). **Mitochondrial Leigh syndrome: the state of the art**. Arch Pediatr *32*, 509-516, doi:10.1016/j.arcped.2025.04.007.

Ugarteburu, O., Farré-Tarrats, L., Muñoz-Pujol, G., Unceta, M., Las Heras, J. D., Garcia-Ribes, A., Arza-Ruesga, A., de la Morena, B., Arauz-Garofalo, G., Gay, M., Garrabou, G., Corral, J., Vilaseca, M., Ribes, A., García-Villoria, J., Gort, L. et Tort, F. (2026). **Complex IV deficiency due to COX4I1 deep intronic and de novo variants results in progressive motor impairment and Leigh syndrome**. Mitochondrion *86*, 102095, doi:10.1016/j.mito.2025.102095.

Yasser, R., Imran, F., Khalid, M., Talha, M. et Waafira, A. (2025). **AI-powered neuroimaging markers: a new era in paediatric Leigh syndrome diagnosis**. Ann Med Surg (Lond) *87*, 7842-7843, doi:10.1097/MS9.0000000000003999.

Zilber, S., Burnworth, M., Afolabi, T., Brestoff, J. R., Minczuk, M., Luis, A. R., Ling, Q., Prigione, A., Tolle, I., Elsharkawi, I., Perlstein, E., Boyce, D., Johnson, S. et Woleben, K. (2025). **Expanding research and care for Leigh syndrome: efforts of a patient-led advocacy organization**. Res Involv Engagem *11*, 137, doi:10.1186/s40900-025-00808-x.

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

Abrosimov, R., Borlepawar, A., Hajieva, P. et Moosmann, B. (2025). **An Alternative Metabolic Pathway of Glucose Oxidation Induced by Mitochondrial Complex I Inhibition: Serinogenesis and Folate Cycling**. Int J Mol Sci *26*, 11349, doi:10.3390/ijms262311349.

Adnan, A., Rakha, A., Tao, L., Shen, Q., Chen, H. D., He, H. F., Ullah, M. F., Nazir, S., Zafar, A. A., Wang, C.-C. et Wang, H. (2025). **Maternal lineage diversity and health-related haplogroups in the Gilgiti and Kohistani populations of northern Pakistan**. BMC Genomics, doi:10.1186/s12864-025-12373-4.

Ahmadi, Z. A., Cabrera-Orefice, A., Zaninello, M., Barth, E., Veronese, M., Rodenburg, R., Rugarli, E. I., Brodesser, S., Arnold, S. et Brandt, U. (2026). **Pathogenesis of mtDNA point mutation m.10191T>C affecting complex I function is a multifactorial process leading to metabolic remodeling of mitochondria**. Biochim Biophys Acta Mol Basis Dis *1872*, 168070, doi:10.1016/j.bbadis.2025.168070.

Alshami, Y., Hroub, O., Hroub, M., Abouodeh, S., Makhamre, Z., Hammouri, A. G., Alzatari, I. et Atawneh, O. (2025). **Mitochondrial HMG-CoA Synthase Deficiency Presenting as Pediatric Metabolic Stroke: A Case Report of a Novel Homozygous HMGCS2 (p.Ile56Asn) Variant**. Clin Case Rep *13*, e71622, doi:10.1002/ccr3.71622.

Angelin, A., Keller, K., Lu, P., Guarnieri, J. W., Widjaja, G. A., Sule, R., Janssen, K., Olali, A., Cen, Z., Carosi, V., Beauplan, M., Murdock, D. G., Potluri, P., Pei, L. et Wallace, D. C. (2025). **Partial restoration of mitochondrial dysfunction by AAV-Ant1 protects from dilated cardiomyopathy in Ant1-/- plus mtDNA mutant mice**. Nat Commun *17*, 447, doi:10.1038/s41467-025-67134-4.

Bacman, S. R., Shoop, W., Henley-Beasley, C. S., Thompson, R., Barrera-Paez, J. D., Theard, L.-M., Rodriguez-Silva, M., Gorsuch, C., Barton, E. R. et Moraes, C. T. (2025). **Transient muscle expression of mitoARCUS in mice leads to sustained reductions in pathogenic mtDNA and reduces fatigability**. Mol Ther S1525-0016(25)01064-0, doi:10.1016/j.ymthe.2025.12.041.

Bannwarth, S. et Paquis-Flucklinger, V. (2026). **Sporadic amyotrophic lateral sclerosis: what is the impact of the mitochondrial genome?** J Neurol Neurosurg Psychiatry *97*, 199-200, doi:10.1136/jnnp-2025-337087.

Bernardino Gomes, T. M., Childs, J. B., Di Leo, V., Warren, C., Hudson, G., Turnbull, D. M., Lawless, C. et Vincent, A. E. (2025). **Quantifying variability of mitochondrial markers in m3243A > G myopathy**. Sci Rep, doi:10.1038/s41598-025-33106-3.

Bottino, M., Boyer, M., Steenari, M. R., Barrick, R. et Abdenur, J. E. (2025). **Lactic acidosis, rhabdomyolysis, and hyperammonemia: Atypical presentation in a new patient with PDE-ALDH7A1 defect**. Mol Genet Metab Rep *45*, 101260, doi:10.1016/j.ymgmr.2025.101260.

Campbell, T., Slone, J., Vu, J., Liu, W., Yang, L., Dourson, A., Queme, L. F., Jankowski, M. P. et Huang, T. (2025). **Ferroptosis is a novel pathogenic mechanism of FDXR-related disease via disruption of the NRF2 pathway**. Cell Death Discov *11*, 563, doi:10.1038/s41420-025-02840-y.

Campos-Ribeiro, M. A., Donnarumma, E., Nolte, H., Cobine, P., Vimont, E., Milenkovic, D., Hernandez-Camacho, J. D., Langa-Vives, F., Kornobis, E., Pénard, E., Yde, S., Langer, T., Paquis-Flucklinger, V. et Wai, T. (2025). **Mutant CHCHD10 disrupts cytochrome c oxidation and activates mitochondrial retrograde signaling**. EMBO Mol Med, doi:10.1038/s44321-025-00358-5.

Cao, J., Zhang, J., Wu, Z., Luan, W., Zhou, Y., Huang, H., Li, L. et Hu, W. (2025). **Slirp2 modulates oogenesis via regulating mitochondrial protein translation**. J Mol Cell Biol mjaf047, doi:10.1093/jmcb/mjaf047.

Cavalcanti, J. F. B., Haydar, A., Keller, G., Araujo Martins Moreno, C., Macedo-Silva, A. et Zanoteli, E. (2025). **Systemic lipomatosis as a clue to mitochondrial myopathy**. Pract Neurol pn-2025-004808, doi:10.1136/pn-2025-004808.

Chaudhry, A., Thompson, D. M. et Chanoine, J.-P. (2026). **Diabetes management in maternally inherited diabetes and deafness (MIDD): A review and a proposed treatment algorithm**. Diabetes Obes Metab *28*, 826-839, doi:10.1111/dom.70240.

Chavez-Guevara, I. A., Vázquez-Lorente, H., Herrera-Quintana, L., Rubio-Valles, M., López, L. C., Plaza-Díaz, J. et Amaro-Gahete, F. J. (2026). **Noncoding RNA molecules mediating skeletal muscle mitochondrial function and their potential applications in exercise molecular physiology: a systematic review**. Am J Physiol Cell Physiol *330*, C201-C210, doi:10.1152/ajpcell.00313.2025.

Chen, C. X., Venkatesh, V. et Allen, M. G. (2025). **Lactate Sensors for Early Detection of Metabolic Disorders in Dynamic Muscle Tissue Models**. Annu Int Conf IEEE Eng Med Biol Soc *2025*, 1-5, doi:10.1109/EMBC58623.2025.11252833.

Chen, Y. H., Esparza Sanchez, M., Hung, T. I., Tang, J., Xu, W., Yin, J., Wang, Y., Chang, C.-E. A., Zhang, H., Chen, J. et Zhao, L. (2025). **Glutathionylated DNA adducts accumulate in mitochondrial DNA and are regulated by AP endonuclease 1 and tyrosyl-DNA phosphodiesterase 1**. Proc Natl Acad Sci U S A *122*, e2509312122, doi:10.1073/pnas.2509312122.

Cheshkov, S., Krishnamurthy, L. C., Chang, A., Baek, H.-M., Ganji, S., Babcock, E., Spence, J. S., Briggs, R. W. et Haley, R. W. (2025). **Decadelong low basal ganglia NAA/tCr from elevated tCr supports ATP depletion from mitochondrial dysfunction and neuroinflammation in Gulf War illness**. Sci Rep *15*, 39741, doi:10.1038/s41598-025-24099-0.

Corbaz, S., Pibernus, D. A., Loos, M. A., Pérez, M. M., Buompadre, M. C., García, F. M., Rodriguez, M. P., Rugilo, C., Armeno, M., Seo, G. H., Khang, R., Alonso, C. N. et Aráoz, H. V. (2025). **From Severe Neonatal Encephalopathy to Slowly Neurologic Progressive Disease: Pyruvate Dehydrogenase Deficiency Related to PDHA1 Variants**. J Child Neurol 8830738251404115, doi:10.1177/08830738251404115.

Dalili, S., Rostampour, N., Mousavi, S. T., Joni, S. S., Mahdieh, N., Rad, A. H., Hakemzadeh, S. T., Nikpour, S., Talea, A. et Moravvej, H. (2025). **Comprehensive Iranian guidelines for the diagnosis and management of mitochondrial disorders: an evidence- and consensus-based approach**. Orphanet J Rare Dis *20*, 623, doi:10.1186/s13023-025-04127-y.

Denova, J. R., Flanagan, B. A., Jah, M., Applebaum, S. L. et Edmands, S. (2025). **Molecular consequences of mitochondrial replacement may be masked from organismal traits in Tigriopus californicus**. PLoS One *20*, e0335181, doi:10.1371/journal.pone.0335181.

Ding, X., Jiang, Y., Wang, X., Li, C., Kong, Z., Fu, L., Lei, Z., Cai, H., Dong, Y., Shi, C., Su, X., Kunath, T., Wang, Z., Buratto, D., Lin, A., Shao, J., Zhang, D., Liu, Z., Liang, P., Zhou, R. et Yan, Q. (2025). **Novel DNJ Derivative Ameliorates Cardiac Hypertrophy by Targeting OPA1 and Restoring Mitochondrial Health**. Circ Res, doi:10.1161/CIRCRESAHA.125.327407.

Doni, D., Grifagni, D., Cavion, F., Buchignani, B., Battini, R., Baschiera, E., Desbats, M. A., Pasquariello, R., Covello, G., De Pascale, E., Boarolo, A., Cestonaro, I., Badocco, D., Pastore, P., Sartori, G., Stehling, O., Lill, R., Santorelli, F. M., Salviati, L., Ciofi-Baffoni, S. et Costantini, P. (2025). **A novel mutation in FDX2 provides insights into the pathogenesis of MEOAL mitochondrial neuromuscular disease**. Cell Death Dis, doi:10.1038/s41419-025-08323-3.

Elsharkawi, I., Goldstein, A., Ganetzky, R. D. et Morava, E. (2025). **Counseling and Prognostic Challenges in Survivorship and Mortality in Primary Mitochondrial Disease: Reshaping a Once Bleak Landscape**. Pediatr Neurol *175*, 223-228, doi:10.1016/j.pediatrneurol.2025.11.019.

Faroud Lopez, R., Huayta, J., Williams, G. D. Z., Seay, S. A., Lalwani, P. D., Bacot, S. N., Vengosh, A. et Meyer, J. N. (2025). **Lithium nickel manganese cobalt oxide particles cause developmental neurotoxicity in Caenorhabditis elegans**. Env Sci Adv *4*, 1767-1781, doi:10.1039/d5va00103j.

Feng, Z., Guo, L., Gao, Y., Zhang, M., Liu, X., Kang, B., Tan, M., Liao, S. et Wang, H. (2025). **Prenatal Diagnosis of COQ2 Variants in Suspected Coenzyme Q10 Deficiency**. Kidney Int Rep *10*, 3945-3957, doi:10.1016/j.ekir.2025.08.023.

Ferrera, G., Segre, G., Lamantea, E., Ghezzi, D., Rivelli, M. et Ardissone, A. (2025). **Amino acid supplementation in mitochondrial aminoacyl-tRNA synthetase defects: two case reports of tyrosine supplementation in YARS2-associated disease and a review of the literature**. Front Pediatr *13*, 1699348, doi:10.3389/fped.2025.1699348.

Fetterman, J. L., Chinnery, P. F., McClellan, R., Wallace, D. C., Suomalainen, A., Ojala, T., Lewis, S. C., Ballinger, S. W., et American Heart Association Council on Genomic and Precision Medicine Council on Cardiopulmonary, Critical Care, Perioperative and Resuscitation Council on Cardiovascular and Stroke Nursing Council on Peripheral Vascular Disease (2025). **Mitochondrial Genetics in Cardiovascular Health and Disease: A Scientific Statement From the American Heart Association**. Circulation, doi:10.1161/CIR.0000000000001393.

Frison, M., Lockey, B. S., Nie, Y., Golder, Z., Theiaspra, E., Ryall, C. D., Lyons, C., Burr, S. P., Prater, M., Bozhilova, L. V., Glynos, A., Stewart, J. B., Jones, N. S., Chiaratti, M. R. et Chinnery, P. F. (2025). **Ubiquitin-mediated mitophagy regulates the inheritance of mitochondrial DNA mutations**. Science *390*, 156-163, doi:10.1126/science.adr5438.

Ganesh, J., Donnelly, C., Ligezka, A., Preston, G., Morava, E., Breilyn, M., Marin-Valencia, I., Raynes, H., Bansal, N., Lamour, J., Mintz, C. et Kozicz, T. (2025). **Cardiac transplant outcomes in a pediatric patient with novel homozygous variants in TOP3A causing mitochondrial dysfunction**. Mol Genet Metab *146*, 109236, doi:10.1016/j.ymgme.2025.109236.

García-Parra, B., Guiu, J. M., Povedano, M. et Modamio, P. (2025). **Real-world evidence supporting orphan drugs approvals for rare neuromuscular disorders in the European Union and the United States: Review of public assessment reports (2015-2025)**. Curr Opin Pharmacol *86*, 102586, doi:10.1016/j.coph.2025.102586.

Gaur, P., Alves, C., Yildiz, H., Mankad, K., Sudhakar, S., Rahman, S. et Biswas, A. (2025). **Imaging patterns of paediatric CNS mitochondrial disorders**. Neuroradiology, doi:10.1007/s00234-025-03805-9.

Gray, R., Kantartzı, V., Lopes, L. R., Savvatis, K. et Westwood, M. (2025). **Cardiac magnetic resonance findings in mitochondrial disease: a guide for clinicians**. Eur Heart J Imaging Methods Pract *3*, qyaf134, doi:10.1093/ehjimp/qyaf134.

Guertin, A., Khanna, R. K., Tardieu, M., François, M., Blasco, H., de Bretagne, I. B., Benoist, J.-F., Bigot, A., Tressel, N., Maillot, F. et Labarthe, F. (2025). **Full recovery of vision following early and intensive hemodialysis in an 18-year-old woman with methylmalonic acidemia-related optic neuropathy**. Mol Genet Metab Rep *45*, 101279, doi:10.1016/j.ymgmr.2025.101279.

Habib, A. A. et Punga, A. R. (2025). **Diagnosis of MG and differential diagnoses**. Int Rev Neurobiol *183*, 21-30, doi:10.1016/bs.irn.2025.04.034.

Honda, K., Hitora, Y., Sadahiro, Y., Wato, M., Nagano, Y. et Tsukamoto, S. (2026). **Natural products that suppress 2-deoxy-d-glucose-induced cell death as potential drug seeds for mitochondrial diseases**. Bioorg Med Chem *132*, 118478, doi:10.1016/j.bmc.2025.118478.

Hu, Z., Yu, J., Liu, Z., Jiang, M. et Zeng, A.-P. (2025). **Targeting multiple genetic defects of mitochondrial diseases with a single bacterial lipoate protein ligase**. Sci Adv *11*, eaea8481, doi:10.1126/sciadv.aea8481.

Ikeda, M., Imasawa, T., Akanuma, T., Goto, Y.-I., Okazaki, Y., Murayama, K., Ohara, M. et Suzuki, T. (2025). **Mitochondrial Nephropathy With m.5538G>A Mutation Within the tRNA-Trp Region Assessed by Mitochondrial Function Analysis: A Case Report**. Nephrology (Carlton) *30*, e70153, doi:10.1111/nep.70153.

Janiak, J., Piątkowska, W., Podlejska, P., Zakrzewski, D., Zawadzka, M., Sawicka, A. et Mazurkiewicz-Bełdzińska, M. (2025). **Atypical clinical manifestation of MT-TL1 mutation in 6 months old patient**. Acta Neurol Belg, doi:10.1007/s13760-025-02921-w.

Jiang, F., Sugiura, A., Kishita, Y., Nakamura, K., Qi, X., Liu, Y., Murayama, K. et Okazaki, Y. (2026). **Functional Screening of NDUFAF6 Variants in Knockout Cells and Complementary Computational Analysis**. J Clin Lab Anal *40*, e70147, doi:10.1002/jcla.70147.

Jouda, H., Browning, A. C., Aftab, A. M., Mahmoud, I., Bibi, S., McFarland, R., Pickett, S. J. et Devine, H. (2025). **Macular OCT inner retinal changes reflect CNS involvement in m.3243A>G disease**. BMJ Neurol Open *7*, e001232, doi:10.1136/bmjno-2025-001232.

Kadiša, A., Vereskuns, R., Tarasovs, M., Mičule, I. et Inashkina, I. (2025). **Mitochondrial cardiomyopathy with skeletal muscle myopathy caused by m.3260A > G mutation in MT-TL1 gene: a case report**. J Med Case Rep *19*, 573, doi:10.1186/s13256-025-05633-0.

Kawano, S., Saegusa, C., Masano, Y., Becker, F., Nakamura, M., Shiozawa, S., Fujikura, J., Toyohara, T., Abe, T., Okano, H. et Fujioka, M. (2025). **Modeling Mitochondrial Disease Using Brain Organoids: A Focus on Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-like Episodes**. J Vis Exp, doi:10.3791/69303.

Kim, C.-H., Lee, J. K., Park, H.-R. et Choi, B. Y. (2026). **Personalized Anesthesia Strategies for Cochlear Implantation: Insights on Local Anesthesia From a Single-institution Experience**. Otol Neurotol *47*, e19-e22, doi:10.1097/MAO.0000000000004655.

Kim, D., Lee, S.-E., Cha, J., Lee, J. H., Kang, Y. C. et Lee, S.-Y. (2025). **Mitochondrial transplantation restores mitochondrial content and function in SSBP1-related mitochondrial DNA depletion syndrome**. BMB Rep 6418.

Kleinveld, V. E. A., Wanschitz, J., Hotter, A., Mayr, J. A., Höftberger, R., Löscher, W. N. et Horlings, C. G. C. (2025). **Diagnostic Value of Muscle Biopsy for the Evaluation of Adult Myopathy in Daily Clinical Practice**. Diagnostics (Basel) *15*, 3102, doi:10.3390/diagnostics15243102.

Kochan, Z. et Karbowska, J. (2025). **How Genes Meet Diet in LCHAD Deficiency: Nutrigenomics of Fatty Acid Oxidation Disorder**. Int J Mol Sci *26*, 10140, doi:10.3390/ijms262010140.

Kubo, R., Iwamoto, Y., Ajihara, S., Murayama, K., Ohtake, A., Fujinuma, S., Sugiyama, K., Ishido, H., Korematsu, S. et Masutani, S. (2025). **A Case of 17β-Hydroxysteroid Dehydrogenase Type 10 (HSD10) Disease Caused by a Novel Variant Presenting With Rapidly Progressive Cardiomyopathy Triggered by Viral Infection**. Cureus *17*, e93552, doi:10.7759/cureus.93552.

Kuikka, P., Nikkinen, H., Majamaa, K. et Martikainen, M. H. (2025). **Obstetric history of women with m.3243A>G: an observational cohort study**. J Med Genet jmg-2025-110875, doi:10.1136/jmg-2025-110875.

Kunz, W. S. (2025). **Turnover and Quality Control of Mitochondrial DNA**. Biochemistry (Mosc) *90*, 1849-1861, doi:10.1134/S0006297925602485.

Kurade, M., Bobba-Alves, N., Kelly, C., Behnke, A., Conklin, Q., Juster, R.-P., Hirano, M., Trumpff, C. et Picard, M. (2025). **Mitochondrial and psychosocial stress-related regulation of FGF21 in humans**. Nat Metab *7*, 2212-2220, doi:10.1038/s42255-025-01388-6.

Kuznetsova, N. A., Kochegarova, K. K., Baikova, I. P., Korshunov, E. N., Ilchuk, L. A., Kubekina, M. V., Bruter, A. V., Silaeva, Y. Y. et Varlamova, E. A. (2025). **Ubiquitous Expression of mPolgmut Leads to the Accumulation of Cytotoxic CD8+ T Lymphocytes in Young Mice**. Life (Basel) *15*, 1863, doi:10.3390/life15121863.

Kwan, J. Y., Lantz, C. I., Korobeynikov, V. A., Snyder, A., Huang, X., Haselhuhn, T., Dore, K. N., Madruga, A., Danielian, L. E., Schindler, A. B., Chia, R., Rasheed, M., Crook, J., Szabo, M., Portley, M., Sherer, C. M., King, M. C., Huang, T.-H., Kosa, P., Bielekova, B., Ward, M. E., Grunseich, C., Shneider, N. A., Traynor, B. J. et Narendra, D. P. (2025). **Clinical, neuropathological, and biochemical characterization of ALS in a large CHCHD10 R15L family**. medRxiv 2025.09.22.25335938, doi:10.1101/2025.09.22.25335938.

Labory, J., Boulaimen, Y., Singh, J., Ait-El-Mkadem Saadi, S., Paquis-Flucklinger, V., Bannwarth, S. et Bottini, S. (2025). **Accelerate the discovery of genetic variants in mitochondrial diseases with Variant prIOritization using Latent spAce**. Brief Bioinform *26*, bbaf612, doi:10.1093/bib/bbaf612.

Laranjeira, M., Oliveira, J. M. A., Santorelli, F. M., Marchese, M. et Nogueira, C. (2025). **Morpholino Knockdown in Zebrafish: A Tool to Investigate the Functional Impact of Variants of Unknown Significance in Mitochondrial Diseases**. Neuromolecular Med *27*, 69, doi:10.1007/s12017-025-08890-w.

Lee, A., Bliksrud, Y. T., Onali, M., Neugebauer, J., Eyskens, F., Haas, D., Mossler, K., Enekwe, A., Kiec-Wilk, B., Diep, L. M., Bellettato, C. M., Zernikow, B., Scarpa, M., Rahman, S., Tangeraas, T., et MetabERN collaboration group on palliative care (2025). **Palliative Care for Children and Adults With Inherited Metabolic Disease in Europe: An Underutilised Service for Supportive Treatment and Care**. J Inherit Metab Dis *48*, e70095, doi:10.1002/jimd.70095.

Lee, Y.-S., Kim, H. S., Nguyen, P. L., Lee, Junhyeong, Kim, D.-I., Lee, Jeongmin, Moon, C., Cho, K.-O., Kim, B.-E., Ahn, J., Osborne, T. F., Duysak, T., Kim, J.-S., Jung, C. H. et Jeon, T.-I. (2025). **Copper deficiency disrupts OXPHOS and mitochondrial dynamics through MTCH2-dependent copper trafficking in skeletal muscle**. bioRxiv 2025.11.19.688750, doi:10.1101/2025.11.19.688750.

Leeming, P. J., Mercuri-Svik, J. et Stojanovski, D. (2026). **From Biogenesis to Breakdown: How Protein Biogenesis and Quality Control Failures Drive Mitochondrial Disease**. Mol Cell Biol *46*, 21-47, doi:10.1080/10985549.2025.2566671.

Leslie, M. (2025). **First approved drug for mitochondrial disease raises hopes for more**. Science *390*, 114-115, doi:10.1126/science.aec9018.

Lessard, L. E., Bourque, D. K., Bourque, P. J., Lochmüller, H., Machado, J., Del Gobbo, G. F., Marshall, A. E., Smith, I. C., Care4Rare Canada Consortium, Boycott, K. M., Warman-Chardon, J. et Breiner, A. (2025). **MT-ATP6 variant as a cause of adult-onset hereditary spastic paraparesis: A case report and literature review**. J Neuromuscul Dis 22143602251391068, doi:10.1177/22143602251391068.

Levergood, N. R., Ko, M. W., Payne, K. K. et Mackay, D. D. (2025). **Optic Atrophy Predominant WFS1 Disorder-A Case-Control Study**. J Neuroophthalmol, doi:10.1097/WNO.0000000000002428.

Li, Y., Wang, D., Zhou, M., Sun, H., Hong, S., Jiang, L. et Guo, Y. (2025). **Hypocitrullinemia as an Early Diagnostic Biomarker for MT-ATP6 Mitochondrial Diseases**. J Mol Neurosci *75*, 154, doi:10.1007/s12031-025-02440-6.

Liu, C.-N., Yan, H.-Y., Liao, S. C.-K., Tsai, C.-Y., Tseng, L.-H., Lin, P.-H., Lee, N.-C. et Wu, C.-C. (2025). **Bilateral Sudden on Chronic Hearing Loss as an Auditory Manifestation of Mitochondrial DNA Mutations**. Laryngoscope, doi:10.1002/lary.70208.

Lyu, L., Qie, B., He, Y., Chen, F. et Liu, B. (2025). **Advances in gene therapy for mitochondrial genetic disorders: current status and clinical implementation challenges**. J Transl Med *23*, 1415, doi:10.1186/s12967-025-07420-3.

Mancuso, M., Lamperti, C. et Musumeci, O. (2025). **National diagnostic gaps for TK2 Deficiency in Italy: insights from the AIM Multicenter Survey**. Acta Myol *44*, 93-95, doi:10.36185/2532-1900-1424.

Manjunath, U., R, V., Kulkarni, S. S., Reddy, H. N., Anil, A., Mishra, R. K. et Iyer, G. R. (2025). **NMPhenogen: a comprehensive database for genotype-phenotype correlation in neuromuscular genetic disorders**. Front Neurosci *19*, 1696899, doi:10.3389/fnins.2025.1696899.

Marin, J., Fuchs, A., Tarasenko, T., Warren, E., Kirby, M., Anderson, S., Gordon-Lipkin, E. M., Kruk, S., West, A. P. et McGuire, P. J. (2025). **Mapping metabolic dependences and capacities using ATP as a biomarker**. Res Sq rs.3.rs-4836421, doi:10.21203/rs.3.rs-4836421/v2.

Mazzucato, S., Lopriore, P., Daddoveri, F., Lamperti, C., Carelli, V., Musumeci, O., Servidei, S., Micera, S., Mancuso, M. et Bandini, A. (2025). **[Not Available]**. Recenti Prog Med *116*, 613-614, doi:10.1701/4573.45801.

McCain, R. S., Piroli, G. G., Smith, H. H., Sumal, A. S., Grimsrud, P. A., Cotham, W. E., Walla, M. D. et Frizzell, N. (2025). **Region-Specific Quantification of 2-Hydroxyglutarate Enantiomers in Murine Brain during Mitochondrial Complex I Deficiency**. ACS Chem Neurosci *16*, 4172-4179, doi:10.1021/acschemneuro.5c00628.

McNally, M., Thakkar, E. et Hutton, A. (2025). **Safe General Anesthesia in a Pediatric Patient With 1p36.33 Duplication Syndrome: A Rare Case Report**. Clin Case Rep *13*, e71476, doi:10.1002/ccr3.71476.

Merkevicius, K., Smirnov, D., Schlieben, L. D., Ganetzky, R., Feichtinger, R. G., Jiang, H., Fang, F., Ebihara, T., Murayama, K., Ferrera, G., Ardissone, A., Rokicki, D., Wesol-Kucharska, D., Schröder, S., Bauer, P., Bertoli-Avella, A., Østergaard, E., Freisinger, P., Janssen, M. C. H., Wagner, M., Abouyousef, O., Alhaddad, B., AlAbdi, L., Alkuraya, F., Alston, C. L., Baghdasaryan, A., Barca, D., Barić, I., Bellusci, M., Bevot, A., Boltshauser, E., Borggraefe, I., Bouchereau, J., Bruno, C., Burnyte, B., Calhoun, A., Casas, K., Coker, M., Crushell, E., De Lonlay, P., Dionisi-Vici, C., Distelmaier, F., Falk, M. J., Ferreira, A. C., Ferreira, C. R., Ficicioglu, C., Fatma Gokçay, G., Häberle, J., Heath, O., Hellenschmidt, A., Hoefele, J., Hoffmann, G. F., Honzik, T., Huemer, M., Janeiro, P., Karaa, A., Seher Kasapkara, Ç., Kern, I., Klepper, J., Klopstock, T., Knerr, I., Koch, J., Krumina, Z., Lamperti, C., Lebigot, E., Liu, Z., Maier, E. M., Martinelli, D., McFarland, R., Mendelsohn, B., Molnar, M. J., Mundy, H., Nassogne, M.-C., Oliveira, A., Õunap, K., Panicucci, C., Parikh, S., Peters, H., Pichard, S., Plecko, B., Ramadža, D. P., Repetto, G. M., Rivera, I., Rodenburg, R. J., Rossi, A., Schiff, M., Seidemann, K., Smith, W. E., Soares, S., Siri, B., Steinbrucker, K., Striano, P., Sykut-Cegielska, J., Tal, G., Taylor, R. W., Tsiakas, K., Kalkan Ucar, S., van Konijnenburg, E. H., Woidy, M., Yaplito-Lee, J., Yildiz, Y., Zenker, M., Zsidegh, P., Westphal, D., Sperl, W., Meitinger, T., Brown, G. K., Prokisch, H., Mayr, J. A. et Wortmann, S. B. (2025). **The genotypic and phenotypic landscape of PDHA1-related pyruvate dehydrogenase complex deficiency**. Brain awaf430, doi:10.1093/brain/awaf430.

Mi, L., Li, Y.-X., Lv, X., Wan, Z.-L., Liu, X., Zhang, K., Li, H., Yao, Y., Zhang, L., Xu, Z., Zhuang, X., Ji, K., Jiang, M., Wang, Y. et Lu, P. (2025). **Computational design of a high-precision mitochondrial DNA cytosine base editor**. Nat Struct Mol Biol *32*, 2575-2586, doi:10.1038/s41594-025-01714-2.

Molla, G. K., Uzun, Ö. Ü., Agakisili, H. B., Genç, E., Gülten, Z. A., Yıldırım, T., Ersoy, A. S., Ak, B., Gülbahçe, A., Yıldız, S., Çakar, N. E., Karaca, M., Zübarioğlu, T., Hişmi, B. Ö., Erdöl, Ş., Önal, H., Kara, B. et Gökçay, G. F. (2025). **Profiles of paediatric patients experiencing stroke-like episodes associated with mitochondrial disease**. Front Neurol *16*, 1657852, doi:10.3389/fneur.2025.1657852.

Mondal, M., Scifo, E., Ciliberti, R. E., Wischhof, L., Abbariki, T. N., Jackson, J., Menegatou, I.-M., Zeisler-Diehl, V., Riemer, J., Jussila, B., Hopkins, C. E., Kierszniowska, S., Schreiber, L., Nicotera, P., Ehninger, D. et Bano, D. (2025). **Dietary lipid content modifies wah-1/AIFM1-associated phenotypes via LRK-1 and DRP-1 expression in C. elegans**. Nat Commun *16*, 10817, doi:10.1038/s41467-025-66900-8.

Monteuuis, G., Awadhpersad, R., van der Kolk, D., Singh, S. K., Nyman, T. A., Malyutina, A., Zamboni, N., Moisio, K., Juutila, J., Hietakangas, V., Seneca, S., Carroll, C. J. et Jackson, C. B. (2025). **Vacuolar-type H+-ATPase-mediated extra-organellar buffering resolves mitochondrial dysfunction**. Nat Commun *17*, 67, doi:10.1038/s41467-025-66656-1.

Moreno, C. A. M., Gontijo, C. C., Fonseca, A. T. Q. S. M., Horvath, R., Schlesinger, D. et Zanoteli, E. (2025). **A Novel Variant in the TAMM41-Associated Mitochondrial Myopathy**. Am J Med Genet A, doi:10.1002/ajmga.70034.

Nascimento, A., Ortez, C., Expósito, J., Carrera, L., Cerezo, S., Lotz, S., Zschaeck, I., Lujan, A., Gatnau, C., Estévez, B., Tizzano, E. et Natera De Benito, D. (2025). **[Neuromuscular diseases in pediatrics with specific treatments]**. Medicina (B Aires) *85 Suppl 4*, 34-40.

Nielsen, S. R., Gjela, M., Stoico, M. P., Drewes, A. M., Olesen, S. S., Højlund, K., Pedersen, I. S., Brock, C., Frøkjær, J. B. et Frederiksen, A. L. (2025). **Mild pancreatic fibrosis with preserved exocrine function and increased visceral adipose tissue in m.3243A>G carriers: A magnetic resonance imaging study**. Mol Genet Metab *146*, 109282, doi:10.1016/j.ymgme.2025.109282.

Nikitchina, N., Heckel, A.-M., Shebanov, N., Mazunin, I., Tarassov, I. et Entelis, N. (2025). **Targeted deletions in human mitochondrial DNA engineered by Type V CRISPR-Cas12a system**. NAR Mol Med *2*, ugaf021, doi:10.1093/narmme/ugaf021.

Olakunle, I. C. et Tobias, J. D. (2025). **Perioperative Care of a Child With Combined Oxidative Phosphorylation Deficiency 6: Total Intravenous Anesthesia With Remimazolam**. J Med Cases *16*, 434-439, doi:10.14740/jmc5179.

Othmani, N. B. et Mathew, S. (2025). **Clinical insights into mitochondrial retinopathy: A case report on m.3243A>G mutation and macular dystrophy**. Saudi J Ophthalmol *39*, 416-418, doi:10.4103/sjopt.sjopt_314_24.

Pal, S., Ishihara, T., Setoyama, D., Chen, C.-L., Onoue, K., Yonemura, S., Ogasawara, E. et Ishihara, N. (2026). **Non-canonical role of natural quinones in mitochondrial nucleoid organization for maintaining respiration and protecting cardiac function**. J Biochem *179*, 5-20, doi:10.1093/jb/mvaf062.

Petropoulou, A., Kypraios, N., Rizopoulou, D., Kouvela, A., Maniatis, A., Anastasopoulou, K., Anastogianni, A., Korfiatis, T., Grafanaki, K., Stamatopoulou, V. et Stathopoulos, C. (2025). **Mitochondrial tRNA-Derived Diseases**. Int J Mol Sci *26*, 12023, doi:10.3390/ijms262412023.

Pieper, A. A. et Paul, B. D. (2025). **Hydrogen Sulfide Signaling in Neurodegenerative Movement Disorders**. Handb Exp Pharmacol, doi:10.1007/164_2025_757.

Pu, Z., Wang, P., Wang, M., Chen, Y., Zhu, Y., Jiao, Y., Zou, W., Yu, B., Cheng, H., Liu, Y., Cao, Y., Ji, D. et Liang, D. (2025). **Melatonin alleviates oxidative stress induced damage in human iPSCs harboring mtDNA 3243A>G mutation via MAPK pathway**. Stem Cell Res Ther *16*, 554, doi:10.1186/s13287-025-04666-y.

Qin, N., Liu, R., Deng, R., Jia, W., Yang, Y., Sun, J., Gao, S. et Zhu, T. (2026). **Lobetyolin alleviates microglial inflammation by activating CK2α/Opa1-mediated mitochondrial fusion in ischemic stroke**. Phytomedicine *150*, 157651, doi:10.1016/j.phymed.2025.157651.

Qu, J., Zhang, M., Hu, Y., Yang, G., Zhang, X., Zhang, W. et Qu, Y. (2025). **Identifying the Key Mitochondria-Related Genes in COPD by Integrating Machine Learning and Bioinformatics Analyses**. Int J Genomics *2025*, 7060748, doi:10.1155/ijog/7060748.

Ramachandran, H., Becker, A., Dobner, J., Hildebrandt, B., Distelmaier, F., Rossi, A. et Anand, R. (2025). **CRISPR/Cas9-mediated editing of MIC13 in human induced pluripotent stem cells: A model for mitochondrial hepato-encephalopathy**. Stem Cell Res *89*, 103870, doi:10.1016/j.scr.2025.103870.

Ratnaike, T. E., Kule, M. E., Paramonov, I., Matalonga, L., Polavarapu, K., Olimpio, C. et Horváth, R. (2025). **A systematic analysis of mitochondrial aminoacyl tRNA synthetase variants in a rare disease cohort**. Eur J Hum Genet, doi:10.1038/s41431-025-01990-y.

Rehan Ahmad, S., Nasir, N., Kumari, A. et Ul Hassan, A. (2025). **Loss-of-function variant of SLC27A3 causes mitochondrial dysfunction and a metabolic neurodevelopmental disorder via impaired fatty acid transport**. J Hum Genet, doi:10.1038/s10038-025-01435-w.

Riko, M., Kawamoto, D., Hirayama, K., Tsuchihashi, T., Suzuki, T., Sugimoto, T., Okazaki, Y., Murayama, K. et Tokuhara, D. (2025). **Fontaine progeroid syndrome with neonatal mitochondrial disease**. Hum Genome Var *12*, 26, doi:10.1038/s41439-025-00331-1.

Rotuski, G., Paczwa, K., Mędrzycka, J., Różycki, R. et Gołębiewska, J. (2025). **Mitochondrial Macular Dystrophy-A Case Report and Mini Review of Retinal Dystrophies**. J Clin Med *14*, 8236, doi:10.3390/jcm14228236.

Sato, I., Shiraishi, M., Norota, K., Shiraki, K. et Yamada, Y. (2025). **Mitochondrial Activation and Therapeutics: Innovations in Cell- and Organelle-Specific Medicine**. Biol Pharm Bull *48*, 1652-1666, doi:10.1248/bpb.b25-00218.

Shelke, P., Tribhuvan, S., Agrahari, A. K. et Saxena, R. (2026). **The evolving landscape of mitochondrial base editing: advances in precision, modeling, and therapeutic potential**. Mitochondrion *86*, 102093, doi:10.1016/j.mito.2025.102093.

Somai, S., Aloh, C. H., King, D. E. et Copeland, W. C. (2025). **Mitochondrial DNA Replication and Disease: A Historical Perspective on Molecular Insights and Therapeutic Advances**. Int J Mol Sci *26*, 10275, doi:10.3390/ijms262110275.

Son, J.-W., Nam, J.-H., Choi, K.-U. et Hong, G.-R. (2025). **The Evolving Landscape of Hypertrophic Cardiomyopathy: Phenocopies and Diagnostic Pathways in Hypertrophic Cardiomyopathy**. Heart Fail Clin *21*, 533-547, doi:10.1016/j.hfc.2025.06.004.

Spolador, G. M., Polastrini, R. T. V., Zoboli, I., Henrique, A. C., Freitas, E., do Nascimento, A. G., Pugliese, C., Kok, F., Bueno, C. et de Macedo Barbosa, S. M. (2025). **Trigger points of palliative care assessment in inherited metabolic diseases**. Orphanet J Rare Dis *20*, 627, doi:10.1186/s13023-025-04128-x.

Stefano, G. B., Büttiker, P., Weissenberger, S., Raboch, J. et Anders, M. (2025). **Semaglutide and the pathogenesis of progressive neurodegenerative disease: the central role of mitochondria**. Front Neuroendocrinol *79*, 101217, doi:10.1016/j.yfrne.2025.101217.

Stein, B., Simons, R., Sahhar, H., Rishmawi, S. et Shaw, A. K. (2025). **Hypoglycemic Encephalopathy With Multisystem Organ Dysfunction in an Infant With MEGD(H)EL Syndrome**. Cureus *17*, e97867, doi:10.7759/cureus.97867.

Su, S., Hu, W., Liu, Y., Lang, Q. et Zhang, H. (2025). **Stroke-like lesion and status epilepticus in a child with NARS2-related combined oxidative phosphorylation deficiency 24**. Front Neurol *16*, 1731858, doi:10.3389/fneur.2025.1731858.

Sun, Y., Wei, X., Xiao, B., Luo, Y., Wang, Y., Liu, R., Zhan, Y., Ye, X., Cai, X., Xu, S., Lyu, J., Fang, H. et Yu, Y. (2025). **Identification of novel NDUFA3 variants in a patient with mitochondrial disorders**. Pediatr Res, doi:10.1038/s41390-025-04403-4.

Tagliavini, S., Filippini, M., Anderlini, A., Caraffi, R., Baraldi, C., Ruozi, B., Tosi, G., Ottonelli, I. et Duskey, J. T. (2025). **Drug delivery systems for mitochondrial targeting**. Nanoscale *17*, 27085-27110, doi:10.1039/d5nr02199e.

Tai, Y., Eura, N., Miyasaka, T., Nakano, R., Ochi, T., Marugami, N., Tanaka, T. et Sugie, K. (2025). **Myopathies: Radiologist's Essential Tips for Clinical, Pathologic, and Imaging Findings**. Radiographics *45*, e240190, doi:10.1148/rg.240190.

Tekin Neijmann, S., Gunes, D., Karaca, M., Karaman, V., Balci, M. C., Gokcay, G. F. et Gedikbasi, A. (2025). **Evaluation of Serum FGF21 Levels in Patients with Mitochondrial Aminoacyl-tRNA Synthetase Deficiency**. Int J Mol Sci *26*, 9525, doi:10.3390/ijms26199525.

Tekmenuray-Unal, A., Tas, M., Kangin, M. et Bozaci, A. E. (2025). **Arg339Gln Is a Recurrent Variant in Rare Combined Oxidative Phosphorylation Deficiency 4: A New Patient with Biallelic TUFM Gene Variant**. Mol Syndromol, doi:10.1159/000549573.

Tinker, R. J., Jacob, N., Syed, M. G., Kelkar, J., Donnelly, C., Elsharkawi, I., Ganesh, J., Gelb, B., Pejaver, V., Kozicz, T. et Morava, E. (2025). **Drivers of Diagnostic Delay in Mitochondrial Disease: Missed Recognition of Canonical Features**. medRxiv 2025.10.09.25337582, doi:10.1101/2025.10.09.25337582.

Uehara, N., Fujita, T., Uehara, K., Shimoda, H., Yamashita, D., Yokoi, J., Katsunuma, S., Kakigi, A. et Nibu, K.-I. (2025). **Clinical characteristics and hearing impairment in carriers of the m.3243 A > G variant**. J Hum Genet, doi:10.1038/s10038-025-01412-3.

Ugarteburu, O., Farré-Tarrats, L., Muñoz-Pujol, G., Unceta, M., Las Heras, J. D., Garcia-Ribes, A., Arza-Ruesga, A., de la Morena, B., Arauz-Garofalo, G., Gay, M., Garrabou, G., Corral, J., Vilaseca, M., Ribes, A., García-Villoria, J., Gort, L. et Tort, F. (2026). **Complex IV deficiency due to COX4I1 deep intronic and de novo variants results in progressive motor impairment and Leigh syndrome**. Mitochondrion *86*, 102095, doi:10.1016/j.mito.2025.102095.

Villafán-Bernal, J. R., Rosas-Hernández, J., García-Ortiz, H., Martínez-Hernández, A., Contreras-Cubas, C., Guerrero-Contreras, I., Lee, H., Seo, G. H., Carnevale, A., Barajas-Olmos, F. et Orozco, L. (2025). **First Latin American Case of MLASA2 Caused by a Pathogenic Variant in the Anticodon-Binding Domain of YARS2**. Int J Mol Sci *26*, 12039, doi:10.3390/ijms262412039.

Wang, J., Song, M., Liu, Z., Xu, C., Zou, Y., Duan, X., Liu, Y., Zhang, W., Li, J. et Fang, F. (2025). **Genotype-Phenotype Correlations in Chinese Pediatric Patients With Single Large-Scale Mitochondrial DNA Deletion Disorders**. Clin Genet, doi:10.1111/cge.70089.

Wang, K., Wang, H., Zhang, Y., Zhang, Z., Wang, L., Yang, J., Man, J. et Yang, L. (2026). **Reprogramming mitochondrial homeostasis in renal ischemia-reperfusion injury**. Cell Signal *139*, 112294, doi:10.1016/j.cellsig.2025.112294.

Wang, Y., Yu, H.-Y., Yi, Z.-J., Qi, L.-Y., Yang, J.-S., Xie, H.-X., Zhao, M., Liu, N.-H., Chen, J.-Q., Zhou, T.-J., Xing, L., Cheng, X.-W. et Jiang, H.-L. (2025). **Super mitochondria-enriched extracellular vesicles enable enhanced mitochondria transfer**. Nat Commun *16*, 9448, doi:10.1038/s41467-025-64486-9.

Watanabe, C., Miyauchi, A., Aoki, S., Watanabe, M., Jimbo, E. F., Miyama, Y., Kitayama, H., Uno, Y., Watanabe, K., Hattori, Y., Yotsumoto, Y., Onuki, T., Sugiyama, Y., Ichimoto, K., Yatsuka, Y., Okazaki, Y., Imasawa, T., Murayama, K., Ohtake, A., Yamagata, T. et Osaka, H. (2025). **Ferroptosis susceptibility in primary coenzyme Q10 deficiency: Cellular insights from patient fibroblasts and clinical course of six individuals**. Brain Dev *48*, 104497, doi:10.1016/j.braindev.2025.104497.

Watanabe, T., Ishikawa, G. et Tabata, T. (2025). **Mitochondrial Disease Diagnosed Following Preterm Birth at 29 Weeks of Gestation and Postpartum Heart Failure: A Case Report and Literature Review**. Clin Case Rep *13*, e71532, doi:10.1002/ccr3.71532.

Wibom, R., Alsina, D., Naess, K., Engvall, M., Freyer, C., Wedell, A. et Wredenberg, A. (2026). **Advancing a sensitive method for measuring mitochondrial ATP production in small muscle biopsy samples**. Anal Biochem *709*, 116004, doi:10.1016/j.ab.2025.116004.

Wong, S. W., Fung, C. W., Tomlin, F. et Stojanovic, J. (2025). **Kidney transplantation in mitochondrial diseases: a systematic review**. Pediatr Nephrol, doi:10.1007/s00467-025-07034-3.

Xu, B., Chai, J., Bian, P. et Guo, Y. (2025). **Genetics of Mitochondrial Aminoacyl-tRNA Synthetases Associated with Sensorineural Hearing Loss**. Mol Syndromol *16*, 449-460, doi:10.1159/000542981.

Xu, C. L., Fang, F., Zhou, J., Wang, H., Zhang, W. H., Gong, S., Jiang, H. F., Liu, Z. M. et Li, J. W. (2025). **[Long-term efficacy observation of nicotinamide in the treatment of early-onset progressive encephalopathy with brain edema and (or) leukoencephalopathy-2 caused by NAXD gene variation]**. Zhonghua Er Ke Za Zhi *63*, 1246-1249, doi:10.3760/cma.j.cn112140-20250327-00260.

Yang, R.-Z., Wang, Y., Wu, Z., Luo, Y., Wang, D.-D., Zou, Y., Liang, Y., Li, J.-K., Zhang, S., Huang, C.-P., Zeng, W.-R., Chang, S.-Y., Li, S.-M., Meng, X.-Y., Sun, H.-F., Liu, P.-P., Lei, J., Xiang, Y., Gu, Y., Yan, B., Cai, S.-Q., Zhang, J. et Kang, J.-S. (2025). **Ambient light alleviates retinal neurodegeneration in mice by powering mitochondria via the engineered optoenergetic rhodopsin**. Signal Transduct Target Ther *10*, 355, doi:10.1038/s41392-025-02450-1.

Yao, T., Wen, G. A., Wu, Z. et Shen, N. (2025). **Lac-Phe: a central metabolic regulator and biomarker**. Med Rev (2021) *5*, 429-431, doi:10.1515/mr-2025-0030.

Yuan, J.-H., Higuchi, Y., Ando, M., Hashiguchi, A., Okamoto, Y., Hiramatsu, Y., Yoshimura, A., Kodama, K., Sakiyama, Y., Mitsui, J., Tsuji, S. et Takashima, H. (2025). **Respiratory complex I deficiency caused by a novel multi-exonic PUS1 deletion**. J Hum Genet, doi:10.1038/s10038-025-01437-8.

Zeng, G., Liu, R., Li, X., Lv, Z., Cui, L., Zhang, X. et Wang, J. (2025). **Sequential development of three syndromes in a patient with m.3243A>G mutation: a case report**. Front Med, doi:10.1007/s11684-025-1186-7.

Zhang, C., Zheng, X., Xiang, L. et Su, Z. (2025). **Exercise-mediated regulation of mitochondrial dynamics in aging muscle: implications for mitochondrial diseases**. Mol Cell Biochem, doi:10.1007/s11010-025-05441-6.

Zhang, Z., Chen, Z., Bie, X., Xie, Z., Li, X., Liu, J., Xiao, M., Zhang, Q., Zhang, Y., Yang, Y. et Li, D. (2025). **Functional identification of two variants in unrelated Chinese patients with DNM1L-related mitochondrial disorders**. BMC Pediatr *25*, 984, doi:10.1186/s12887-025-06299-9.

Zhao, J., Xu, B., Huang, T., Honfo, S. H., Trumpff, C., Picard, M., Cohen, A. A. et Liu, M. (2025). **Examining saliva proteomic dynamics in mitochondrial diseases from a perspective of intrinsic health**. Sci Rep *15*, 43031, doi:10.1038/s41598-025-23879-y.

Zhao, Ying, Xu, Z., Zhuang, X., Zhao, Yuying, Yan, C. et Ji, K. (2026). **The Case of a 25-Year-Old Woman With Isolated Head Tremor**. Ann Clin Transl Neurol *13*, 209-211, doi:10.1002/acn3.70221.

Zhou, X., Xu, X., He, S., Liu, Q. et Lu, T. (2025). **Case Report: Aveir implantation in an 8.7-year-old, 25-kg pediatric patient with mitochondrial disease via internal jugular vein**. Front Cardiovasc Med *12*, 1593438, doi:10.3389/fcvm.2025.1593438.