Mitochondrial disorders : The Essentials

4th quarter 2024

Quarterly literature follow-up
carried out by the AFM-Téléthon documentation department

This literature follow-up, 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*

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Maladie liée au gène GFM1 – GFM1-related disorder

No results

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

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

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

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

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

MELAS

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

MERFF

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Karaa, A., Bertini, E., Carelli, V., Cohen, B., Ennes, G. M., Falk, M. J., Goldstein, A., Gorman, G., Haas, R., Hirano, M., Klopstock, T., Koenig, M. K., Kornblum, C., Lamperti, C., Lehman, A., Longo, N., Molnar, M. J., Parikh, S., Phan, H., Pitceathly, R. D. S., Saneto, R., Scaglia, F., Servidei, S., Tarnopolsky, M., Toscano, A., Van Hove, J. L. K., Vissing, J., Vockley, J., Finman, J. S., Abbruscato, A., Brown, D. A., Sullivan, A., Shiffer, J. A., Mancuso, M., et MMPOWER-3 Trial Investigators (2024). **Genotype-specific effects of elamipretide in**

patients with primary mitochondrial myopathy: a post hoc analysis of the MMPOWER-3 trial. Orphanet J Rare Dis *19*, 431, doi:10.1186/s13023-024-03421-5.

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

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

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

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

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

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

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

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

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

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

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

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

Syndrome de Leigh - *Leigh syndrome*

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Goolab, S., Terburgh, K., du Plessis, C., Scholefield, J. et Louw, R. (2025). **CRISPR-Cas9 mediated knockout of NDUFS4 in human iPSCs: A model for mitochondrial complex I deficiency**. Biochim Biophys Acta Mol Basis Dis *1871*, 167569, doi:10.1016/j.bbadis.2024.167569.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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