Mitochondrial disorders : The Essentials

2nd quarter 2025

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*

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

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

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

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

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Maladie liée au gène GFM1 – GFM1-related disorder

Ahmad, B., Dumbuya, J. S., Li, W., Tang, J.-X., Chen, X. et Lu, J. (2025). **Corrigendum to « Evaluation of GFM1 mutations pathogenicity through in silico tools, RNA sequencing and mitophagy pathway in GFM1 knockout cells » [Int. J. Biol. Macromol. Volume 304, Part 2, April 2025, 140970]**. Int J Biol Macromol 145547, doi:10.1016/j.ijbiomac.2025.145547.

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

Berrahmoune, S., Dassi, C., Pekeles, H., Cheung, A. C. T., Gagnon, T., Waters, P. J., Buhas, D. et Myers, K. A. (2025). **Investigating the safety and efficacy of deoxycytidine/deoxythymidine in mitochondrial DNA depletion disorders: phase 2 open-label trial**. J Neurol *272*, 307, doi:10.1007/s00415-025-13060-w.

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MELAS

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