# Mitochondrial disorders : The Essentials

## 1er 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*

1. Ahmad, I., Kamai, A., Zahra, S., Kapoor, H., Kumar Srivastava, A., & Faruq, M. (2024). Generation and characterization of iPSC lines from Friedreich's ataxia patient (FRDA) with GAA.TTC repeat expansion in the Frataxin (FXN) gene's first intron (IGIBi016-A) and a non-FRDA healthy control individual (IGIBi017-A). *Stem Cell Research*, *77*, 103382. https://doi.org/10.1016/j.scr.2024.103382

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11. Doni, D., Cavion, F., Bortolus, M., Baschiera, E., Muccioli, S., Tombesi, G., d'Ettorre, F., Ottaviani, D., Marchesan, E., Leanza, L., Greggio, E., Ziviani, E., Russo, A., Bellin, M., Sartori, G., Carbonera, D., Salviati, L., & Costantini, P. (2024). Correction : Human frataxin, the Friedreich ataxia deficient protein, interacts with mitochondrial respiratory chain. *Cell Death & Disease*, *15*(1), 93. https://doi.org/10.1038/s41419-024-06459-2

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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

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

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