Mitochondrial disorders : The Essentials

1st 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*

No results

Maladie liée au gène GFM1 – GFM1-related disorder

Ahmad, B., Dumbuya, J. S., Li, W., Tang, J.-X., Chen, X. et Lu, J. (2025). **Evaluation of GFM1 mutations pathogenicity through in silico tools, RNA sequencing and mitophagy pahtway in GFM1 knockout cells**. Int J Biol Macromol *304*, 140970, doi:10.1016/j.ijbiomac.2025.140970.

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

Bjerknes, T. L., Rubiolo, A., Shadad, O., Tysnes, O.-B. et Tzoulis, C. (2025). **Chromogen-based double immunohistochemical detection of mitochondrial respiratory chain deficiencies in human brain tissue**. Acta Neuropathol Commun *13*, 63, doi:10.1186/s40478-025-01980-7.

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MELAS

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MERFF

No results

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

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

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

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

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