L'essentielle
Maladies Mitochondriales

1er trimestre 2025

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Sommaire

Ataxie de Friedreich - *Friedreich ataxia*

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Atrophie optique autosomique dominante – *ADOA*

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Encéphalopathie myo-neuro-gastrointestinale - *MNGIE*

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

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

Pas de résultat ce trimestre

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*

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MERFF

Pas de résultat ce trimestre

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

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