Mitochondrial disorders : The Essentials

2nd 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. Alemany-Perna, B., Tamarit, J., Cabiscol, E., Delaspre, F., Miguela, A., Huertas-Pons, J. M., Quiroga-Varela, A., Merchan Ruiz, M., López Domínguez, D., Ramió I Torrentà, L., Genís, D., & Ros, J. (2024). Calcitriol Treatment Is Safe and Increases Frataxin Levels in Friedreich Ataxia Patients. *Movement Disorders: Official Journal of the Movement Disorder Society*, *39*(7), 1099-1108. https://doi.org/10.1002/mds.29808

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4. Cantrell, A. C., Besanson, J., Williams, Q., Hoang, N., Edwards, K., Bishop, G. R., Chen, Y., Zeng, H., & Chen, J.-X. (2024). Ferrostatin-1 specifically targets mitochondrial iron-sulfur clusters and aconitase to improve cardiac function in Sirtuin 3 cardiomyocyte knockout mice. *Journal of Molecular and Cellular Cardiology*, *192*, 36-47. https://doi.org/10.1016/j.yjmcc.2024.05.003

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11. Grander, M., Haschka, D., Indelicato, E., Kremser, C., Amprosi, M., Nachbauer, W., Henninger, B., Stefani, A., Högl, B., Fischer, C., Seifert, M., Kiechl, S., Weiss, G., & Boesch, S. (2024). Genetic Determined Iron Starvation Signature in Friedreich's Ataxia. *Movement Disorders: Official Journal of the Movement Disorder Society*, *39*(7), 1088-1098. https://doi.org/10.1002/mds.29819

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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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65. Hikmat, O., Naess, K., Engvall, M., Klingenberg, C., Rasmussen, M., Brodtkorb, E., Ostergaard, E., de Coo, I., Pias-Peleteiro, L., Isohanni, P., Uusimaa, J., Majamaa, K., Kärppä, M., Ortigoza-Escobar, J. D., Tangeraas, T., Berland, S., Harrison, E., Biggs, H., Horvath, R., … Bindoff, L. A. (2024). Status epilepticus in POLG disease : A large multinational study. *Journal of Neurology*, *271*(8), 5156-5164. https://doi.org/10.1007/s00415-024-12463-5

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