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Improve management of MADD patients – insights and achievements

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Multiple Acyl-CoA dehydrogenase Deficiency (MADD, OMIM #231680), a rare disease from the group of inborn error of metabolism (IEM), is an autosomal recessively inherited disorder of fatty acid, amino acid and choline metabolism. MADD results from defects on electron transfer flavoprotein (ETF), and ETF:ubiquinone oxidoreductase (ETF:QO) proteins. These proteins are responsible for transferring electrons from at least 12 dehydrogenases to the respiratory chain, hence mutations on their genes will cause diminished mitochondria β-oxidation and impaired energy production.

In recent years the development of newborn screening programs worldwide resulted in an increased number of MADD patients being identified. Severe forms of disease result in neonatal death, for milder forms the molecular mechanism that triggers disease symptoms is still unknown, and no effective therapy is established, thus, to make disease prognosis is highly challenging to clinicians.

An enormous gap in the field is the lack of a unifying depository for molecular and clinical data on patients, with the majority of cases found disperse in literature, and many not even reported in international journals with full access to all. In this project we propose to contribute to this societal demand, with tremendous impact in the development of new therapeutic approaches, by organizing a curated database with detailed information on mutations associated to MADD, combining molecular, cellular, and clinical data available in the literature.

In this seminar I will present ongoing experiments on ETF:QO disease variants characterization, and computational analysis of ETF an ETF:QO proteins.

Funded by the European Union (TWIN2PIPSA, GA 101079147). Views and opinions expressed are however those of the author(s) only and do not necessarily reflect those of the European Union or European Research Executive Agency (REA). Neither the European Union nor the granting authority can be held responsible for them.