UNIVERSITÀ DEGLI STUDI DI BRESCIA

I SEMINARI DEL DIPARTIMENTO DI MEDICINA MOLECOLARE E TRASLAZIONALE

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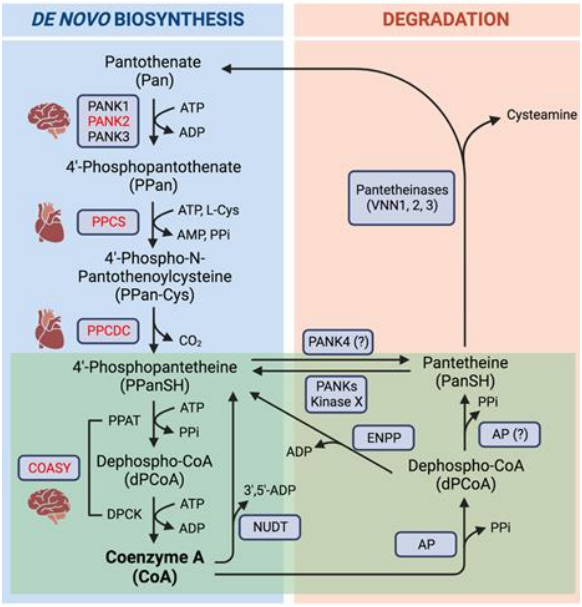
Inborn errors of Coenzyme A biosynthesis: from neurodegeneration to heart failure.

Coenzyme A (CoA) is an essential cofactor required for over a hundred metabolic reactions in the human body. It is synthesised from vitamin B5, through five consecutive enzymatic steps performed by four enzymes: PANK, PPCS, PPCDC, and COASY. Pathogenic variants in PPCS and PPCDC lead to PPCS and PPCDC deficiency disorders, two ultra-rare diseases presenting with early-onset dilated cardiomyopathy, with variable neuromuscular and extra-cardiac manifestation, and no apparent neurodegeneration. In contrast, pathogenic variants in PANK2 and COASY are associated with two rare neurological diseases, PKAN and COPAN respectively, characterized by progressive neurodegeneration with brain iron accumulation and no cardiac symptoms. Dr Iuso will describe the underpinnings of the intricate genotype-phenotype correlations and the search for potential therapeutic approaches for these rare disorders.



Mercoledì 5 giugno 2024, ore 13, aula F

Ospite: Prof Dario Finazzi

Con il supporto dei Fondi per le Attività a Carattere Internazionale