PHENOMAPPING mtDNA. NEW PERSPECTIVES IN PERSONALISED MEDICINE.

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Maternally inherited polymorphic variants of mitochondrial DNA (mtDNA) alter the risk of developing common late-onset human diseases, but the underlying mechanisms are not understood. To date, *in vitro* experiments in cell lines carrying mtDNA population variants have shown variation in the mitochondrial function. However, the differences in mitochondrial bioenergetics and biogenesis do not fully explain their underlying mechanisms in disease. Here, by using analysis of open databases such as GTEx and Interval in combination with transcriptomic and metabolomics analysis, we show major changes between mtDNA haplogroups involving key cellular prosteotatic pathways such as mTORC1 and EIF2A. Chemical and genetic modification of these pathways showed great differences between mtDNA backgrounds with impact on a myriad of functions including mitochondrial function and biogenesis, morphology, metabolism rewiring and maintenance of cytoplasmic reactive oxygen species levels (ROS). Our findings provide a new mechanism linking mtDNA variation with proteostatic metabolism regulation that does not directly involve oxidative phosphorylation, highlighting the importance of considering the mtDNA genetic background in personalized medicine.

Aula 1, Hospital Materno-Infantil

21 enero 2020

12:30 h

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FUNDACIÓN PROFESOR NOVOA SANTOS