



Research Project Proposal
Academic year 2018-2019

Project Nº 10

Title:

Understanding the Molecular Insights of Primary Hyperoxaluria: Disease Models, Regulators and Modifiers.

Department/ Laboratory

Laboratory of stem cells and reprogramming, Lab 1.01, Regenerative Medicine Program, CIMA.

Director:

JUAN ROBERTO RODRIGUEZ MADOZ

Contact:

jrrodriguez@unav.es
+34948194700 ext 1023

Summary

Primary hyperoxaluria (PH) is a group of rare autosomal recessive metabolic disorders characterized by defects in enzymes involved in glyoxylate metabolism where combined liver-kidney transplantation is the only curative treatment approach. Although there is a considerable interest in understanding the molecular mechanisms of the disease, multidisciplinary approaches aiming to address the regulation of the glyoxylate pathway are very limited with a fundamental lack of data integration. Thus, we think that combining and integrating multi-omics approaches we would contribute to give the first insight into the regulation of glyoxylate metabolism both in health and disease. In particular we aim to combine epigenomics, transcriptomics and metabolomics to characterize patient-specific disease models, decipher molecular regulators and identify disease modifiers.

Overall, our findings could represent an important step to the validation of in vitro patient specific disease models, as well as to understand the regulation of the hydroxyproline/glyoxylate metabolism in animal and cellular models that would help to the identification of new molecular targets that would be of high prognostic and potentially therapeutic relevance, allowing a direct transfer of basic research into clinical applications. The candidate will be directly involved in the characterization of patient-specific disease models at genomic, epigenomic and functional levels. Thus, she/he will gain knowledge and expertise in cellular biology and genomics.

Table with 2 columns and 2 rows: yes, X; no,

Does the project include the possibility of supervised animal manipulation to complete the training for animal manipulator?