



Propuesta de Trabajo Fin de Máster

Año académico 2024-2025

MÁSTER EN MÉTODOS COMPUTACIONALES EN CIENCIAS

Proyecto Nº 03

Título: Artificial Intelligence (AI) to define actionable genetic risk predictors

Departamento/ Laboratorio: Biología Computacional

Director: Carlos Ruiz Arenas

Correo electrónico: cruzarenas@unav.es

Codirector: Mikel Hernáez

Correo electrónico: mhernaez@unav.es

Resumen:

Although an individual's genetic variants affect their predisposition to different common diseases, their clinical utility for prevention and treatment is currently very limited. The main reason is that Polygenic Risk Scores (PRS), the best predictors of an individual's genetic risk, are constrained to a reference population (limiting their application) and provide a global measure of risk without specifying which regulatory pathways may be affected and in which tissues to propose specific treatment. The shortcomings of PRS can be addressed by predicting the effect of variants on gene expression and combining them into Gene Set Activity Scores (GSAS). GSAS provide a measure of how the gene expression of different molecular processes is affected in various tissues, offering a much more specific and actionable measure of genetic risk than current PRS.

In this project, we propose to develop tissue-specific genetic risk predictors based on GSAS and molecular functions. To achieve this, the predicted expression by different variants of an individual will be combined into GSAS using NetActivity, a tool developed by our group. In this way, GSAS will be obtained for each tissue and molecular pathway. These GSAS will be correlated with the risk of breast cancer in public datasets, in addition to comparing their prediction with established PRS. Thus, specific markers of risk for molecular pathways and tissues will be generated. These new markers may enhance the genetic prediction of breast cancer and provide actionable genetic markers for common variants.

Presupuesto solicitado (máximo 1500€) y breve justificación

Se solicita 1500 euros para la validación biológica de los resultados obtenidos a través de paneles genómicos de variantes, así como para la gestión y almacenamiento de los datos.

OPTATIVAS RECOMENDADAS

1. Machine Learning I
2. Machine Learning II
3. Tecnologías de alto rendimiento
- 4.