



Propuesta de Trabajo Fin de Máster

Año académico 2024-2025

MÁSTER EN MÉTODOS COMPUTACIONALES EN CIENCIAS

Proyecto Nº 17

Título: Benchmarking of tools that infer gene expression from genetic data.

Departamento/ Laboratorio: Biología Computacional

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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 benchmark different methods that predict gene expression from genetic data. The student will consider different dimensions, such as the number of tissues predicted or the number of genes inferred. The student will use public datasets having genetic and gene expression data, to compare the inferred gene expression with the measured gene expression.

OPTATIVAS RECOMENDADAS

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