



## Propuesta de Trabajo Fin de Máster

Año académico 2020-2021

### *Máster en Métodos Computacionales en Ciencias*

#### Project Nº 50

**Título:** Analysis of somatic variant calls common across different tumor samples

**Departamento/ Laboratorio:** *TECNUN/CIMA*

**Director 1** *Idoia Ochoa and Ibon Tamayo*

**Contacto:** [idoia@unav.es](mailto:idoia@unav.es), [itamayo@unav.es](mailto:itamayo@unav.es)

#### Resumen

Somatic variant calling is the process of identifying variants that are inherent to the tumor sample. In other words, somatic variants correspond to the differences (variants) between the genome of a healthy cell and that of a tumor cell. As such, accuracy in somatic variant calling is paramount, as the downstream analyses assume the identified somatic variants are correct. These analyses can be used for personalized medicine, for example to decide treatment.

In this project the student will have access to the identified somatic variant calls of several tumor samples (patients). The goal of the project is to analyze those somatic variants that are common across different samples/patients. Somatic variants in tumor samples tend to appear randomly, and hence identical somatic variants across different samples are not expected. These may nevertheless appear due to sequencing mechanisms, like sequencing errors, or due to mistakes of the analysis tools that generate them. In this project we will therefore characterize these variants to better understand them, and see if they have a biological meaning or they are caused by an external mechanism. We will apply machine learning techniques to characterize these variants and to potentially identify them directly without having to compare them against the variants on other samples. This will allow to filter them prior to downstream analyses, hence improving the accuracy of the subsequent analyses.

The student is expected to be fluent in English, and to be familiar with topics of machine learning and statistical learning. The student will develop methods in Python or R, and hence familiarity with these tools is needed.