



PhD student:

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Research project:

Integrated Omics for Precision Oncogenetics in the Sardinian Population

Abstract:

This project aims to integrate genomic, transcriptomic, and epigenomic data to develop innovative strategies for cancer prevention and early diagnosis, with a particular focus on hereditary tumours. The emphasis is on the Sardinian population, which is characterised by high genetic homogeneity and a significant prevalence of neoplasms, making it an ideal context for such studies. The multi-omics approach, enhanced by long-read sequencing technologies, will be applied to the analysis of blood samples, liquid biopsies, and tumour tissues.

The objective is to identify mutations, gene expression profiles, and epigenetic modifications relevant for diagnostic and prognostic purposes. The project is structured into three phases: a) collection and characterisation of biological samples, generating omic data; b) bioinformatic analysis and clinical-molecular correlation to identify patterns associated with specific tumour phenotypes; c) functional validation and development of standardised protocols aimed at the clinical implementation of the findings.

Among the project's primary objectives is the creation of a dedicated database of genetic variants specific to the Sardinian population, encompassing both rare and common variants. For those more frequent in the population, specific Polygenic Risk Scores (PRS) will be developed to estimate the individual risk of developing certain types of cancer.

Liquid biopsies will act as a crucial tool for non-invasive and dynamic monitoring of disease progression, offering valuable insights into treatment efficacy and the potential development of drug resistance.

The anticipated impact is twofold: on one hand, it aims to accelerate the transition towards precision oncology through the identification of novel biomarkers and personalised predictive models; on the other, it seeks to promote equitable access to genomic innovations by introducing sustainable, clinically applicable predictive tools within local healthcare systems. This initiative represents a concrete step towards integrating genomics into public healthcare, with potential national benefits in terms of preventing, diagnosing, and treating familial cancers.