Cacchiarelli

Integrated Omics Graph Medicine  
  
This doctoral project will investigate the application of graph medicine and integrated omics. The core research topic is developing a computational platform to translate high-throughput genomics and epigenomics into actionable insights for rare genetic diseases, precision oncology, and population screening. It will integrate diverse data, including long-read whole-genome and transcriptome, single-cell transcriptomic, and epigenomics, to model complex human biology. The project will investigate how to resolve previously undiagnosed Mendelian disorders by pinpointing pathogenic variants in regulatory regions and how to reclassify solid tumours into transcriptomic subtypes linked to drug-response probabilities, offering a direct path from computational models to clinical application.  
  
A significant focus will be on creating robust and reproducible Nextflow pipelines on our dedicated CPU/GPU high-performance computing cluster, machine learning in the application of variational auto-encoders for compressing multi-omic signatures, and work on the emerging field of graph neural networks, learning to build and train these models to propagate information across an integrated biological graph of genes, regulatory elements, variants, and cell states.  
  
The project will integrate a unique skill set at the intersection of data engineering, machine learning, and clinical genomics to manage and analyze large-scale biological data and, most importantly, translate computational findings into testable hypotheses for precision medicine.