

AVAILABLE POSITIONS

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Institute of Affiliation	Human Technopole

PROJECT INFO	
Title of the proposed project:	Advancing molecular and cellular dissection of cardiometabolic diseases
Short description of the project	Cardiovascular disease remains the leading cause of mortality worldwide and arises from a complex interplay between genetic susceptibility, environmental exposures, ageing and immune-mediated processes. While large-scale genomic studies have identified thousands of genetic associations with cardiometabolic traits and disease, understanding how these variants influence cellular function and disease mechanisms remains a major challenge. This PhD project is embedded within the Human Technopole Cardiometabolic Flagship Research Program and will contribute to the integration of population-scale genomics, single-cell biology and functional genomics to uncover the molecular basis of cardiometabolic disease. The student will leverage large-scale multiomics datasets, including genomic, clinical and single-cell transcriptomic data from deeply phenotyped human cohorts, to identify genetic variants, molecular pathways and immune-cell states associated with cardiovascular and metabolic disease risk. The student will be responsible for the analysis of the data and the selection of candidate genes and variants emerging from these analyses, which will then be prioritised for experimental validation using CRISPR/Cas9 genome editing in human pluripotent stem cells and disease-relevant cellular systems using our wet lab capability. We will therefore design functional studies including the differentiation of engineered stem cells into endothelial, cardiac and immune-cell populations, followed by phenotypic characterisation using flow cytometry, single-cell transcriptomics and high-content imaging. Selected targets will also be investigated in advanced multicellular systems, including immune-vascular co-cultures and cardiac organoid models, to establish causal links between genetic variation, immune dysfunction and cardiometabolic disease. This interdisciplinary project will provide training in population genomics, human genetics, genome editing, stem cell biology and single-cell technologies, offering a unique opportunity to bridge large-scale human data with mechanistic experimental biology.
Main research area for the project	Genomic Medicine
Second research area for the project	Molecular & Cellular Biology

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3 key words for the project	Single cell genomics, immunity, cardiovascular diseases
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LAB INFO	
Main topic/s of the lab	Investigating the impact of germline variation on complex phenotypes and diseases
Short description of the lab activity	Our laboratory investigates how inherited and acquired genetic variation influences human health and disease through its effects on molecular, cellular and physiological processes. We combine population-scale genomics, single-cell multi-omics and experimental functional genomics to identify the biological mechanisms underlying complex human traits, with a particular focus on cardiometabolic, immune and blood-related disorders. A major focus of the group is the integration of genomic data with rich clinical and molecular phenotypes to identify genetic determinants of disease risk and progression. Through the Human Technopole Cardiometabolic Flagship Research Program and the CARDINAL project, we are generating large-scale resources that link human genetic variation, immune-cell states, lifestyle exposures and cardiometabolic outcomes across diverse populations. To translate these discoveries into biological insight, we develop experimental systems that enable causal interrogation of candidate genes, variants and pathways. These include CRISPR/Cas9 genome editing, human pluripotent stem-cell models and single-cell transcriptomics. The research group combines cutting-edge computational analyses with state-of-the-art experimental capabilities. Our goal is to bridge population-level observations with mechanistic understanding, ultimately identifying new biomarkers, therapeutic targets and strategies for precision medicine. This is a computational only, or mixed computational-wet lab position. We are seeking a motivated and self-reliant student with prior expertise in one of the following areas: immunology, cardiovascular diseases, or haematology. Computational biology experience is required. Previous experience with core experimental techniques in tissue culture and molecular biology is advantageous but not essential.
Recent bibliography	• Tardaguila M, Von Schiller D, Colombo M, Gori I, [...] Soranzo N. Integrating natural and engineered genetic variations to decode regulatory influence on blood traits. Cell Rep. 2026 Feb 24;45(2):116937. doi: 10.1016/j.celrep.2026.116937. Epub 2026 Feb 3. PMID: 41637188; PMCID: PMC12932927. • Guo J, Walter K, Quiros PM, Gu M, Baxter EJ, Danesh J, et al. Inherited polygenic effects on common hematological traits influence clonal selection on JAK2V617F and the development of myeloproliferative neoplasms. Nature Genetics. 2024;56(2):273-80. • Kundu K, Tardaguila M, Mann AL, Watt S, et al. Genetic associations at regulatory phenotypes improve fine-mapping of

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	causal variants for 12 immune-mediated diseases. Nat Genet. 2022 Mar;54(3):251-262. • Bomba L , Walter K, Guo Q, Surendran P, et al. Whole-exome sequencing identifies rare genetic variants associated with human plasma metabolites. Am J Hum Genet 2022 Jun 2;109(6):1038-1054. • Vuckovic D, Bao EL, Akbari P, Lareau CA, Mousas A, Jiang T, et al. The Polygenic and Monogenic Basis of Blood Traits and Diseases. Cell. 2020;182(5):1214-31.e11.
Group composition	3 PhD students, 3 post docs, 1 senior bioinformatician, 1 research specialist, 1 staff scientist + centre-wide scientific support units in genome analysis, cell screens and biostatistics.
Institutional page link	https://humantechnopole.it/it/gruppi-di-ricerca/soranzo-group/