

Friday
April 29, 2022
15.00 (GMT+1)

Computational and Quantitative Biology Lecture Series



Nicole Soranzo
Human Technopole

The seminar will be held on line. Please register at <https://bit.ly/3Mt46wj>

You will receive an invite with the link to the seminar.
Visit <https://cqb.dieti.unina.it/index.php/events> for the event series.

Population and medical genomics applications to human traits and diseases

Characterizing the multifaceted contribution of genetic and epigenetic factors to disease phenotypes is a major challenge in human genetics and medicine. In our team, we apply large-scale genomic analyses to multi-omic phenotypes to investigate the contribution of genetic variants associated with complex human traits. Here I will describe the value of genome sequencing and imputation to extend genomic analyses to low frequency and rare variants and locus fine- mapping. I will describe how these expansive, high-resolution atlases of multi-omics changes inform understanding of mechanisms of disease. Furthermore, I will describe first efforts to assess quantitatively the contribution of cis-genetic effects to transcriptional variance, to characterise coordination of genetic effects on gene expression, methylation and histone variation and characterise genetic and epigenetic influences at human disease loci and key immune pathways. These results pave the way for a better understanding of genetic and molecular events underpinning cardiometabolic and immune function in humans.

Nicole Soranzo is Head of the Genomics Research Centre at Human Technopole (Milan, IT) since 2021, Senior Group Leader at the Wellcome Sanger Institute (Hinxton, UK) since 2017, Professor of Human Genetics, School of Clinical Medicine, at the University of Cambridge, since 2015.

After completing her degree in Biological Sciences at the University of Milan in 1994, she obtained her PhD in Genetics and Biotechnology at the University of Dundee, UK. From 1999 to 2002, she carried out post-doctoral research at the University of Milan, and from 2002 to 2005 at University College in London, where she applied human genetics to study human evolution. From 2005 to 2007 she worked as Senior Scientist at the Pharmacogenomics Department of Johnson and Johnson Pharmaceutical applying human genetics to improve drugs discovery and pharmacogenomics. She returned to the UK at the Sanger Institute, where she started her group in 2009. In 2013 she became adjunct faculty at the University of Cambridge School of Clinical Medicine, and in 2015 she was awarded a personal chair in Human Genetics. For her outstanding performance, she received many awards and honours as the Italian Female Researcher and Scientist of Impact, National Observatory for Women's Health, in 2016. Nicole Soranzo studies how the human genome influences the risk of common diseases in the general UK population.

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