



Registration  
by 5 Mar 2023



31/03/2023  
h. 09:30 - 17:15



Human Technopole  
Milan (IT)

# SYMPOSIUM

## *Single-molecule Sequencing Technologies & Applications*



# CONCEPT

## COURSE OVERVIEW

The aim of this Symposium, co-organised by Human Technopole (HT) and the Italian Institute of Technology (IIT), is to present and discuss single molecule sequencing techniques, one of those being Nanopore sequencing, an advanced and relatively new technique that is revolutionising the field of DNA/RNA sequencing. The technology allows sequencing of ultra-long DNA/RNA strands. It is fast, has very high accuracy and is easy to use.

The Symposium can be of interest for scientists at different levels, from PhD students who are approaching these technologies to more senior researchers.

Lectures by international researchers will address scientific questions in the fields of RNA discovery/modifications and long-reads genome assembly. Dedicated Q&A sessions on these topics will also highlight potential applications of Nanopore sequencing, promoting fruitful discussions between speakers and participants.

## TARGET AUDIENCE

Open to the national and international community of scientists working in the field of genomics.

# FACULTY

## SCIENTIFIC ORGANISERS

### **PIERO CARNINCI**

(Head, HT Genomics Research Centre - Functional Genomics)

### **CLELIA PEANO**

(Senior Manager, HT Genomics Facility)

### **FRANCESCO NICASSIO**

(Center Coordinator, IIT Center for Genomic Science)

## INVITED SPEAKERS

### **MATT LOOSE**

Professor of Developmental and Computational Biology, University of Nottingham, UK

### **GIOVANNI PASCARELLA**

Research Scientist, RIKEN Center for Integrative Medical Sciences, Japan

### **PHILIPP RESCHENEDER**

Director, Genomic Applications Bioinformatics, Oxford Nanopore Technologies, UK

### **HAZUKI TAKAHASHI**

Research Scientist, RIKEN Center for Integrative Medical Sciences, Japan

### **WINSTON TIMP**

Associate Professor of Biomedical Engineering, Johns Hopkins University School of Medicine, US

## HT & IIT SPEAKERS

### **DAVIDE BOLOGNINI**

Senior Bioinformatician, HT Genomics Research Centre, Italy

### **LOGAN MULRONEY**

Postdoctoral Fellow, IIT-CGS & EMBL, Italy

(time in CET, UTC +1)

**9:30 - 10:30** **REGISTRATION AND WELCOME COFFEE**

**10:30 - 10:45** SESSION 1, chaired by Piero CARNINCI  
"Greeting and Welcome to Human Technopole"  
**Piero Carninci** (HT)

**10:45 - 11:15** "Everything everywhere all at once"  
**Philipp Rescheneder**  
Introduced by Angelica Vittori (Oxford Nanopore Technologies)  
Oxford Nanopore Technologies has created scalable DNA and RNA sequencing platforms which aim to facilitate the sequencing of any living thing, by anyone, anywhere. Nanopore devices can sequence all lengths of fragment, from very short to very long. Longer reads provide access to genomic and transcriptomic features such as long-range phasing, complex structural variants and isoforms.

**11:15 - 11:45** "Full length cap-trap long read sequencing provides discovery of novel transcriptome"  
**Hazuki Takahashi** (RIKEN)  
Long read sequencing technology promises to discover true structure of human transcriptome. The discovery of novel conventional and unconventional transcripts from 5' cap-trapped full length Nanopore sequencing technologies will be introduced, which may serve a roadmap of novel gene catalogue.

**11:45 - 12:15** "Detecting RNA modifications and full-length reads using direct RNA nanopore sequencing"  
**Logan Mulrone**y (IIT-CGS & EMBL)  
Nanocompore and NRSeq methods will be presented. Nanocompore software detects RNA modifications from direct RNA sequencing and has the potential to detect several modifications simultaneously. NRSeq detects the native RNA 5' ends due to the 3' end bias of direct RNA sequencing.

**12:15 - 12:45** Discussion: RNA discovery and modifications

**12:45 - 14:00** LUNCH BREAK

(time in CET, UTC +1)

- 14:00 - 14:30** SESSION 2, chaired by Clelia PEANO  
"Recombination of repeat elements generates somatic complexity in human genomes"  
**Giovanni Pascarella** (RIKEN)  
The main findings in the investigation of the somatic recombination of repeat elements in the human genome will be highlighted and discussed.
- 14:30 - 15:00** "Profiling single-cell transcript isoforms with nanopore sequencing"  
**Davide Bolognini** (HT)  
scRNA-seq profiles transcriptome in single cells but short reads can only yield information from a small region close to one end of the transcripts, precluding the identification of their isoforms. With long reads it is now possible to span transcripts entirely, enabling isoform-level gene expression analysis.
- 15:00 - 15:30** COFFEE BREAK
- 15:30 - 16:00** SESSION 3, chaired by Francesco NICASSIO  
"Beyond assembly: the increasing flexibility of single-molecule sequencing technology"  
**Winston Timp** (Johns Hopkins University School of Medicine)  
Over the past five years, single molecule sequencing has matured rapidly, improving in accuracy, throughput, and ease of use. It will be described how it has emerged as an essential tool in deciphering genome structure and function, measuring the epigenome, and characterizing the transcriptome.
- 16:00 - 16:30** "Nanopore Adaptive Sampling applications in the clinic and field"  
**Matt Loose** (University of Nottingham)
- 16:30 - 17:00** Discussion: genome assembly with long-reads
- 17:00 - 17:15** **CLOSING REMARKS**  
**Clelia Peano & Francesco Nicassio**