

# “Personalized medicine and digital health: achievements for rare disease patients”

**Anthem - Mario Negri Institute partnership**  
**29th June 2026, Centro Anna Maria Astori, Bergamo**

11:00 - 11:05 Welcome – *Prof. Giuseppe Remuzzi*

11:05 – 11:20 Update on Anthem Consortium - *Prof. Guido Cavaletti*

## **Genomic diagnosis of rare diseases towards precision medicine**

11:20 - 11:35 Integrated genetic diagnosis of patients with rare diseases by whole exome sequencing (NextSeq2000), copy number variation (QuantStudio 7Pro) and Sanger validation (SeqStudio 24 Flex)  
*Caterina Mele, Matteo Breno*

11:35 – 11:50 Uncovering the molecular etiology of undiagnosed rare diseases following negative exome sequencing  
*Annamaria Morotti, Alessia Lorenzi*

11:50 – 12:05 Mapping the whole transcriptome in genetic rare diseases (GeoMx profiler)  
*Annamaria Morotti, Matteo Breno*

Lunch

## **Innovative tools for functional studies in rare diseases**

13:15 – 13:30 Functional characterization of genetic abnormalities in patients with rare diseases by recombinant proteins purified through ÄKTA pure™  
*Sofia Padoa*

13:30 – 13:45 Automated analysis using the Jess™ capillary-based western blot system of autoimmune abnormalities in patients with rare diseases  
*Francesca Penati*

13:45 – 14:00 Real-time assessment of platelet aggregation and thrombus formation under flow conditions in patients with rare diseases  
*Sistiana Aiello, Valeria Guaschino*

## Advanced diagnostics, imaging and AI-based tools

- 14:00 – 14:15      Cardiovascular ultrasound imaging: understanding the role of hemodynamics to improve patient care  
*Michela Bozzetto*
- 14:15 – 14:30      AI-based tools for the diagnosis, monitoring and prediction of disease progression  
*Alberto Arrigoni*
- 14:30 – 14:45      Imaging-based AI tools supporting surgical planning and treatment response evaluation  
*Andrea Mangili*
- 14:45                Conclusions  
*Prof. Stefano Paleari, Prof. Giuseppe Remuzzi*