

INNOVATIVE TECHNOLOGIES AND APPROACHES FOR RARE DISEASE DIAGNOSIS AND TREATMENT

EMERGING OPPORTUNITIES FOR THE REGION OF SARDINIA

Morning

- 9:00** Registration
- 9:30** Institutional Greetings
- 9:45** **Michela Onali, Gli Equilibristi - HIBM, Cagliari**
Patient Engagement and Patient Centricity: a priority and model for Care and Research in Rare Diseases.
- 10:15** **Paolo Moi, Regional Coordination Centre for Rare Diseases, Cagliari**
Diagnostic procedures in Sardinia, indicators and costs.
- 10:45** *Coffee Break*
- 11:00** **Leslie Matalonga, Centro Nacional de Análisis Genómico - Centre for Genomic Regulation (CNAG-CRG), Barcelona**
The RD-Connect Genome-Phenome Analysis platform: data sharing and impact on rare diseases.
- 11:45** **Maddalena Migliavacca, Clinical research Unit - IRCCS Ospedale San Raffaele, Milano**
From diagnosis to treatment: the experience of Istituto San Raffaele in gene therapy.
- 12:30** **Roberto Cusano, CRS4 Biosciences, Pula**
Enabling technologies for diagnostics and biomedical research in Sardinia.
- 13:00** *Lunch*

Afternoon

- 14:00** **Giorgio Fotia, CRS4 Biosciences, Pula**
Enabling infrastructure for biomedical research in Sardinia.
- 14:15** **Laura Crisponi, IRGB CNR, Cagliari**
How exome sequencing is shedding light on the complexity of Mendelian disorders: some examples from Sardinia.
- 14:45** **Tommaso Pippucci, Policlinico S.Orsola-Malpighi, Bologna**
Exome analysis in the clinical practice: diagnostic performance and new disease genes.
- 15:15** **Franco Cappai, Sardegna Ricerche, Cagliari**
I-FAIR and SR initiatives in the Biomedicine sector.
- 15:30** **PANEL DISCUSSION**
Multidisciplinary approach in Rare Diseases: from Diagnosis to Treatment.
Moderator: Valentina Careddu, ATS Sardinia press officer
- Laura Crisponi, Leslie Matalonga, Maddalena Migliavacca, Paolo Moi, Tommaso Pippucci**
- 17:30** Conclusions



RARE DISEASE DAY®

28 FEBRUARY 2020

**SA MANIFATTURA - MANIFATTURA TABACCHI
VIALE REGINA MARGHERITA 33, CAGLIARI**

