Rare Disease Workshop: Analysis and collection of rare disease data: a joint effort of biomedical and bioinformatics research communities

Sala lettura A (Biblioteca) CNR
P. le Aldo Moro 7 - 00185 Rome

Tuesday, 28th November 2023 - Sala lettura A (Biblioteca) CNR, P.le Aldo Moro 7	
14:30 - 15:00	**Elixir IT**: Rita Casadio UNIBO, Emidio Capriotti UNIBO, Claudio Carta ISS: The Italian Rare Disease Community
15:00 - 15:30	**Elixir NL**: European RD Communities. Marco Roos (ONLINE)
15:30 -16:00	**Elixir UK**: Research Data Management. Munazah Andrabi (ONLINE)
16:00 - 16:30	Coffee Break
16:30 - 17:00	**Rare Disease Patient Representatives** Annalisa Scopinaro, President of UNIAMO, Federazione Italiana Malattie Rare, Rome, Italy (ONLINE)
17:00 - 17:30	**Istituto Superiore di Sanità (ISS): Rare disease activities.** Domenica Taruscio (ONLINE), Former Director National Centre for Rare Diseases
17:30 - 18:00	**Ospedale Bambin Gesù: Orphanet Italia.** Michele Nutini
Wednesday, 29th November 2023 - Sala lettura A (Biblioteca) CNR, P.le Aldo Moro 7	
9:30 - 11:00	**Presentations by young researchers** • Giulia Babbi. UNIBO. Reactome pathways and Rare Diseases. • Damiano Parrone. UNIROMA1. A resource to explore drug repurposing opportunities for rare conformational diseases. • Cesare Rollo. UNITO. Deciphering Myelodysplastic Syndrome: A Deep Learning Approach for Prognosis Prediction and genomic Characterization. • Giulia Sassi. UNIPR. Illuminating Pharos TDarks: a coevoluzionary approach to rare diseases. • Bernardina Scafuri. UNISA. Identification of two possible pharmacochaperones for GALTp.Q188R enzyme by a computational strategy.
11:00 - 11:30	Coffee Break
11:30 - 13:00	**Rare disease diagnosis: Use case presentations** • Michele Pinelli, Universita' Federico II, Napoli Genomic annotation for the interpretation of DNA sequencing in rare diseases. • Tania Giangregorio, Ospedale Sant'Orsola, Bologna Workflows for the analysis of CNVs from microarray and NGS data. • Maria Cerminara, Istituto Gaslini, Genova Complex cases with Autism Spectrum Disorder (ASD), developmental delay, hyperactivity and sleep disturbance explained by possible oligogenic mechanisms. • Alfredo Brusco, Università di Torino. Disentangling uncommon genetic causes of neurodevelopmental disorders. **Perspectives and closing**