



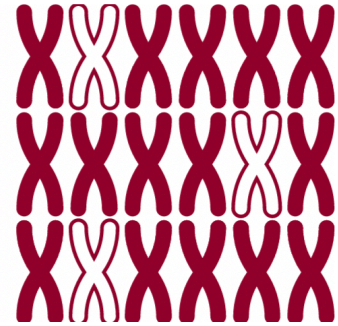
SAPIENZA  
UNIVERSITÀ DI ROMA

International  
WINTER SCHOOL

Rare diseases

Genetics,biomedicine and political challenges

December 12-15 2022 - Aula Monesi-online Zoom link



## International Winter School

Rare diseases

### Genetics, biomedicine and political challenges

December 12-16, 2022 Aula Monesi-Online Zoom

December 12

10-10.30 Opening *Romina Burla, IBPM-CNR Rome*

#### **Genetic and cell bases of progeria**

*Discussant: Romina Burla, IBPM-CNR Rome*

10.30-11.45 **Chiara Lanzuolo**, *ITB-CNR, Istituto Nazionale di Genetica Molecolare "Romeo ed Enrica Invernizzi", Milan, Italy*

Role of DNA conformation in human diseases

11.45-13.00 **Vicente Andres**, *Centro Nacional de Investigaciones Cardiovasculares Carlos III (CNIC), Madrid, Spain*

Hutchinson-Gilford progeria syndrome: from gene discovery to mechanisms and therapies

#### **Genetic and cell bases of Amyotrophic lateral sclerosis**

*Discussant: Alessandro Rosa, Sapienza University of Rome*

14-15.30 **Luc Dupuis**, *University of Strasbourg, Institute for Advanced Studies, Strasbourg, France*

Pathophysiological mechanisms of FUSopathies

15.30-17.00 **Simon Alberti**, *Center for Molecular and Cellular Bioengineering (CMCB), Dresden, Germany* The role of biomolecular condensates in protein aggregation linked disease

December 13

**Genomic studies for rare diseases**

*Discussant: Domenico Raimondo, Sapienza University of Rome*

10.00-11.30 **Alfredo Iacoangeli**, *King's College, London, UK*

Bioinformatics, Interpretation and Data Quality in Genome Analysis of Genetic Disorders

11.30-13.00 **Daniel D'Andrea**, *Cardiff University, UK* Application of bioinformatics in the process of new molecular-targeted therapies

14.00-15.00 Special Guest **Daniela Rhodes** *Fellow of The Royal Society – MRC Cambridge, UK*

**Round table**

**Writing papers and grant proposals**

**Tips and Tricks to be successful**

**Daniela Rhodes**, *Fellow of The Royal Society – MRC Cambridge, UK*

**Antonio Musarò**, *Sapienza University of Rome*

**Alessandro Rosa**, *Sapienza University of Rome*

15.00 coffee break

**15-17.30 International Poster Session**

December 14

**Genetic and cell bases of Spinal Muscular Atrophy (SMA)**

*Discussant: Antonio Musarò, Sapienza University of Rome*

10.00-11.30 **Fred Bernard**, *Jacques Monod Institute, Paris, France*

Nucleus positioning in the *Drosophila* oocyte

11.30-13.00 **Claudio Sette** *Cattolica University, Gemelli Hospital, Rome, Italy* Gene expression regulation in Spinal Muscular Atrophy: from the molecular mechanism to a targeted therapy

14-15.30 **Marika Pane**, *Cattolica University, Gemelli Hospital, Rome, Italy* The incredible impact of RNA therapies on SMA

December 15

**Political challenges for rare diseases**

*Discussants Romina Burla, IBPM-CNR Rome and Alessandro Rosa, Sapienza University of Rome*

10.00-10.15 **Sammy Basso**, *AIProSaB, Vicenza, Italy*

10.25-10.35 **Gloria Antonini**, *Parent Project, Rome, Italy*

10.35-10-50 **Silvia di Angelantonio**, *Sapienza, University of Rome, Italy*  
*Dialogue between patients and scientists: the rare disease Syngap1*

11-13 participatory table with Sammy Basso, Parent Project, Silvia di Angelantonio, Sapienza University

14.00-15.00 Special Guest **Daniela Rhodes** *Fellow of The Royal Society – MRC Cambridge, UK*

**Telomeres: a history of women and science**

15.00 coffee break

15.30-17.30 **Italian Poster Session**

17.30 **Best poster awards**

December 16

10.00-13.00: Exam

14.00-17.00: Course evaluation

**Organizers**

Isabella Saggio, Sapienza University of Rome

Romina Burla, IBPM-CNR Rome

Antonio Musarò, Sapienza University of Rome

Alessandro Rosa, Sapienza University of Rome

Domenico Raimondo, Sapienza University of Rome

Prisca Ornaghi, Sapienza University of Rome