



Conference

UNDIAGNOSED RARE DISEASES: A JOINT ITALY-USA PROJECT

ISTITUTO SUPERIORE DI SANITA'
AULA POCCHIARI
Rome, Italy
November 20, 2017

Project funded by Ministry of Foreign Affairs and International Cooperation

8.30-9:00 Registration

9:00-9:10 Welcome address
Walter Ricciardi

Session 1: The International and the ISS Undiagnosed Diseases Network –UDN-
Chair: Giuseppe Novelli

9.10-9.40 The USA Undiagnosed Diseases Network (UDN)
David Adams

9.40-9.55 The Wilhelm Foundation
Helene Cederroth

9.55- 10.15 Undiagnosed Diseases: International Network and the ISS Italy-USA Project
Domenica Taruscio

10.15-10.30 Data sharing in ISS UDN Network: using PhenoTips and PhenomeCentral tools
Maria Chiara de Stefano, Marco Salvatore, Federica Censi, Giovanna Floridia

10.30-10.45 ISS UDN: next steps
Marco Salvatore

10.45-11.00 Discussion

11.00-11.20 Coffee Break

Results of the Centres involved in the USA-Italy Project:

11.20-11.35 Tor Vergata University, L'Aquila University
Francesco Brancati, Giuseppe Novelli

11.35-11.50 Ferrara University
Alessandra Ferlini, Marcella Neri

11.50-12.05 IRCCS - Mario Negri Institute for Pharmacological Research Clinical Research
Center for Rare Diseases "Aldo e Cele Daccò", Ranica
Erica Daina, Paraskevas Iatropoulos, Giuseppe Remuzzi

12.05-12.20 Piedmont and Aosta Valley Inter-Regional Network Coordinating Centre
for Rare Diseases, Turin University
Dario Roccatello, Simone Baldovino, Elisa Menegatti

12.20-12.35 Udine Rare Diseases Centre
Bruno Bembi

12.35-13.00 Discussion

13.00-13.45 Lunch

Session 2: Further Italian experiences on Undiagnosed Diseases

Chairs: Giuseppe Remuzzi and Domenica Taruscio

13.45-14.05 Undiagnosed Diseases: the experience of the Ospedale Pediatrico Bambino Gesù
Marco Tartaglia, Bruno Dallapiccola

14.05-14.25 Telethon Network for Undiagnosed Diseases
Vincenzo Nigro, Giorgio Casari

14.25 -16.00	Round Table “From National Network for Rare diseases, to European Reference Networks and International initiatives: strengths, weaknesses and opportunities” <i>Dario Roccatello, Erica Daina, Bruno Bembi, Maya Di Rocco, Domenico Coviello, Domenica Taruscio, Lucia Guidotti, Olaf Horst Riess, Maurizio Scarpa, Marta Mosca, Luca Sangiorgi, Claudio Ales</i>
16.00-16.20	The experiences of Patient Associations <i>Alessia Massaccesi, Stefano Collatina</i>
16.20 – 17.00	General discussion
17.00	End of Conference

Speakers and Chairs

David Adams, NIH Undiagnosed Diseases Program, National Human Genome Research Institute, National Institutes of Health, United States

Claudio Ales, European Patient Advocay Group representative, Palermo, Italy

Simone Baldovino, Center of Research of Immunopathology and Rare Diseases - Coordinating Center of the Network for Rare Diseases of Piedmont and Aosta Valley, University of Turin, Turin, Italy

Bruno Bembi, Regional Coordinator Center for Rare Diseases, Academic Hospital of Udine, Udine, Italy.

Francesco Brancati, L’Aquila University, L’Aquila, Italy

Giorgio Casari, Telethon Institute of Genetics and Medicine (TIGEM), Pozzuoli, Italy

Helene Cederroth, Wilhelm Foundation, Sweden

Federica Censi, National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy

Stefano Collatina, Hopen Onlus Foundation, Rome, Italy

Domenico Coviello, Diagnostic group, Scientific Committee for Regional Coordination of rare diseases, A.Li.Sa. – Azienda Sanitaria Liguria Region, Genoa, Italy

Erica Daina, IRCCS - Mario Negri Institute for Pharmacological Research Clinical Research Center for Rare Diseases “Aldo e Cele Daccò”, Ranica, Italy

Bruno Dallapiccola, Bambino Gesù Children's Hospital, IRCCS, Rome, Italy.

Maria Chiara de Stefano, National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy

Maya Di Rocco, Clinical group, Scientific Committee for Regional Coordination of rare diseases A.Li.Sa. – Azienda Sanitaria Liguria Region, Genoa, Italy

Alessandra Ferlini, University-Hospital of Ferrara, Italy

Giovanna Florida, Bioethics Unit, Istituto Superiore di Sanità, Rome, Italy

Lucia Guidotti, Direzione Generale della Programmazione Sanitaria- Ministero della Salute

Paraskevas Iatropoulos, IRCCS - Mario Negri Institute for Pharmacological Research Clinical Research Center for Rare Diseases “Aldo e Cele Daccò”, Ranica, Italy

Alessia Massaccesi, Comitato “I Malati Invisibili Onlus”, Genoa, Italy

Elisa Menegatti, Center of Research of Immunopathology and Rare Diseases - Coordinating Center of the Network for Rare Diseases of Piedmont and Aosta Valley, University of Turin, Turin, Italy
Marta Mosca, ERN ReCONNECT, University-Hospital Pisana, Pisa, Italy
Marcella Neri, University-Hospital of Ferrara, Italy
Vincenzo Nigro, Telethon Institute of Genetics and Medicine (TIGEM), Pozzuoli, Italy
Giuseppe Novelli, Tor Vergata University, Rome, Italy
Giuseppe Remuzzi, IRCCS - Mario Negri Institute for Pharmacological Research
Clinical Research Center for Rare Diseases “Aldo e Cele Daccò”, Ranica, Italy
Walter Ricciardi, President of Istituto Superiore di Sanità, Rome, Italy
Olaf Horst Riess, Institute of Human Genetics and Applied Genomics, University of Tübingen, Germany
Dario Roccatello, Center of Research of Immunopathology and Rare Diseases - Coordinating Center of the Network for Rare Diseases of Piedmont and Aosta Valley University of Turin, Turin, Italy
Marco Salvatore, National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy
Luca Sangiorgi, ERN BOND, Istituto Ortopedico Rizzoli, Bologna, Italy
Maurizio Scarpa, Coordinator of ERN Coordinators, Helios Dr Horst Schmidt Kliniken, Germany
Marco Tartaglia, Bambino Gesù Children's Hospital, IRCCS, Rome, Italy.
Domenica Taruscio, National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy

Scientific Director

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GENERAL INFORMATION

Meeting Venue: Istituto Superiore di Sanità, Aula Pocchiari, Viale Regina Elena, 299 – Rome

Target audience: scientists, clinicians, patients and policy makers

Official language of the Conference: English

Predicted number of participants: 180

ECM accreditations: NO

Participation is free of charge. The organizers will provide refreshments and lunch but will not pay for travel and accommodation expenses of attendees.

Conference registration: The application form, available at https://it.surveymonkey.com/r/Conf_Nov20-2017 must be duly completed and sent via web by 13/11/2017.

The application is properly sent only when the system displays the final "thank you" page. Submissions will be automatically accepted in chronological order of registration, until reaching the maximum meeting room capacity.

Certificate of attendance: Participants will receive a certificate of attendance at the end of Conference. For every relevant information related to the event, please contact the Scientific Secretariat at the numbers listed above.