

Conference
UNDIAGNOSED RARE DISEASES:
A JOINT ITALY-USA PROJECT

ISTITUTO SUPERIORE DI SANITA'
National Centre for Rare Diseases

8.30-9.00 Registration
9.00-9.10 Welcome address
W. 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)
D. Adams
9.40-9.55 The Wilhem Foundation
H. Cederroth
9.55-10.15 Undiagnosed Diseases: International Network and the ISS Italy-USA Project
D. Taruscio
10.15-10.30 Data sharing in ISS UDN Network: using PhenoTips and PhenomeCentral tools
M. C. De Stefano, M. Salvatore, F. Censi G. Floridia
10.30-10.45 ISS UDN: next steps
M. Salvatore
10.45-11.00 Discussion
11.00-11.20 **Coffe Break**

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

11.20-11.35 Tor Vergata University, L'Aquila University
F. Brancati, G. Novelli
11.35-11.50 Ferrara University
A. Ferlini, M. Neri
11.50-12.05 IRCCS - Mario Negri Institute for Pharmacological Research Clinical Research Center for Rare Diseases "Aldo e Cele Daccò", Ranica
E. Daina, P. Iatropoulos, G. Remuzzi
12.05-12.20 Piedmont and Aosta Valley Inter-Regional Network Coordinating Centre for Rare Diseases, Turin University
D. Roccatello, S. Baldovino, E. Menegatti
12.20-12.35 Udine Rare Diseases Centre
B. Bembi

12.35-13.00 Discussion

13.00-13.45 **Lunch**

SESSION 2: Further Italian experiences on Undiagnosed Diseases

Chair: Giuseppe Remuzzi and Domenica Taruscio

13.45-14.05 Undiagnosed Diseases: the experience of the Ospedale Pediatrico Bambino Gesù
M. Tartaglia, B. Dallapiccola
14.05-14.25 Telethon Network for Undiagnosed Diseases
V. Nigro, G. Casari
14.25-16.00 **Round Table** "From National Network for Rare diseases, to European Reference Networks and International initiatives: strengths, weaknesses and opportunities"
D. Roccatello, E. Daina, B. Bembi, M. Di Rocco, D. Coviello, D. Taruscio, L. Guidotti, O. Horst Riess, M. Scarpa, M. Mosca, L. Sangiorgi, C. Ales
16.00-16.20 The experiences of Patient Associations
A. Massaccesi, S. Collatina
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 Advocacy 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
Deborah Capanna, Comitato "I Malati Invisibili", Genoa, 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, Consiglio di Amministrazione della Fondazione Hopen Onlus, 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 Floridia, 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

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Target audience

scientists, clinicians, patients and policy makers

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 .**

**Conference**

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November 20, 2017**

Aula Pocchiari

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