Undiagnosed Rare Diseases: a joint Italy – USA project

- **Bilateral project** granted by

  the Ministry of Foreign Affairs and Cooperation (Italy)

- **Duration:** 2 years (January 2016 – December 2017)

- **Coordination:**

National Center for Rare Diseases , Istituto Superiore di Sanità. Dr. **Domenica Taruscio** & Team

National Human Genome Research Institute, NIH. Dr. **William A. Gahl** & Team

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Centro Nazionale Malattie Rare
CNMR

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Objectives and main results

to gather UDN Italian patients (starting from 5 clinical /research centres) :

work flow and patients characterization as agreed with UDN USA partners (white paper)

to establish and implement the National UDN database containg information on patients

using common standards , terminology & classification

interoperable at International level (UDN International database)

to develop knowledge management strategies (also through site visits)

to identify and characterize new rare diseases by sharing data

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to foster disseminatation of scientific results and knowledge exchange (meetings, workshops, scientific publications)

among partners of the bilateral project

among partners of the UDN International

To strengthen international collaborations and to facilitate interactions among countries **worldwide in the field of UDN**

to implement the UDN International website & **the national and International database**

to provide better and faster means of high quality and clinical usefulness for **the correct diagnosis** of UDN patients for which there is no, or unsatisfactory, diagnosis available for Italian patients trough US and UDN International experts

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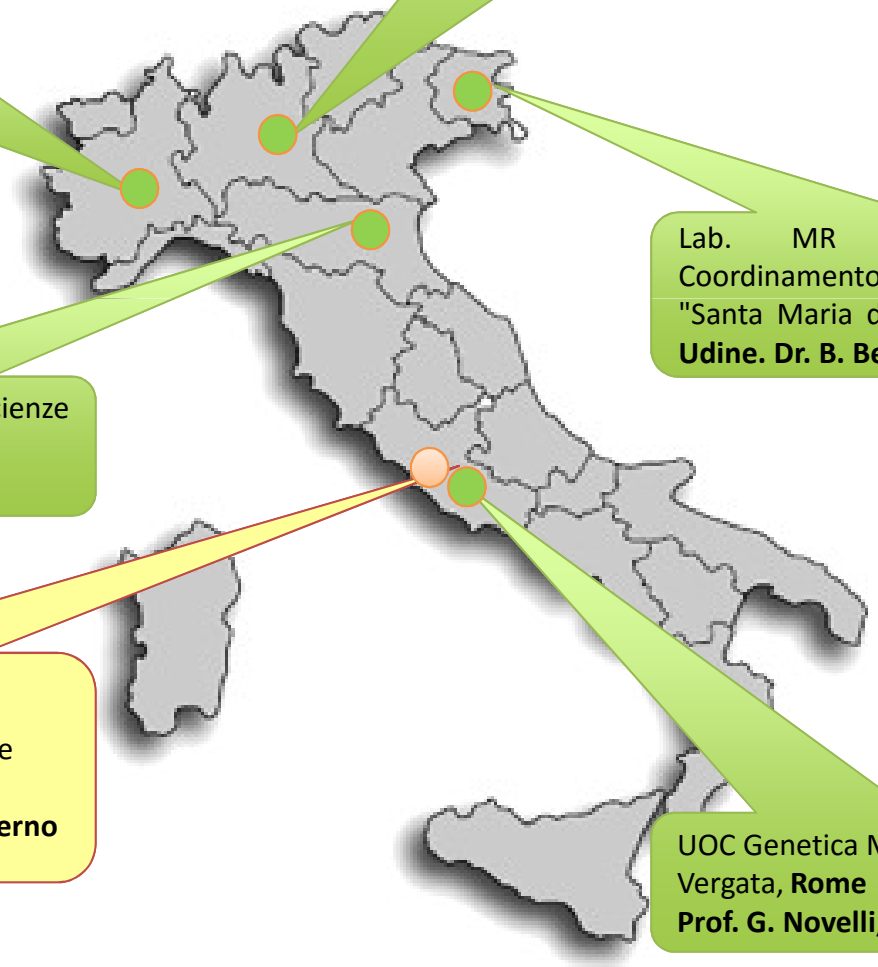
Involvement of 5 Clinical/Research Italian Centres.

More centres will be involved in the future among the Italian Network of Rare Diseases

IRCCS - Istituto di Ricerche Farmacologiche "Mario Negri", Centro di Ricerche Cliniche per le Malattie Rare, **Bergamo**
Dr. E. Daina, Prof G. Remuzzi

Centro Multidisciplinare di immunopatologia e documentazione su malattie rare, **Torino:**
Prof . D. Roccatello, Dr. S. Baldovino



Lab. MR Centro Regionale di Coordinamento per le Malattie Rare A.O.U. "Santa Maria della Misericordia" di Udine, **Udine. Dr. B. Bembi**

U.O. Logistica Genetica Medica, Dip. Scienze Mediche, Università Ferrara, **Ferrara**
Prof. A. Ferlini

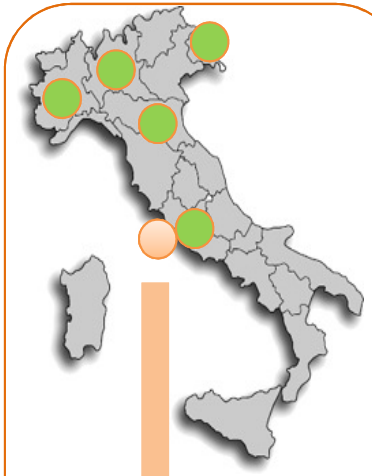
COORDINATOR: National Center for Rare Diseases, Istituto Superiore di Sanità, Rome
Dr. D. Taruscio, dr. G. Floridia, Dr. Marco Salvatore, dr. Claudio Carta , dr. Paolo Salerno

UOC Genetica Medica, Policlinico Tor Vergata, **Rome**
Prof. G. Novelli, Prof F. Sangiuolo

National Network for Rare diseases
(DM 279/2001)

172 Centres

Italian Registry Rare Diseases



UDN Italian Database

Sharing
standards, terminology
core data elements..

UDNI Database
Genetic Characterization
Phenotype Description (Human Phenotype Ontology - HPO)
Standardized high quality clinical data
Other laboratory data

UDNI database

www.undiagnosedinternational.org
(ID & PW)

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Information on methodologies, analytical tools and preliminary data will be shared and discussed among the Italian Group and the NIH Team:

- Call Conferences (monthly)
- 2 Site visits (Italy- USA)
- 1 Operational Meeting (April 2016, in Ranica – Bergamo amog Italian partners only)
- 2 Bilateral Workshops :

  a) Workshop (Turin, January 2017)

  b) Final Workshop (Rome, end of 2017)

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Contents lists available at ScienceDirect

Molecular Genetics and Metabolism

journal homepage: www.elsevier.com/locate/ymgme

Commentary

Undiagnosed Diseases Network International (UDNI): White paper for global actions to meet patient needs

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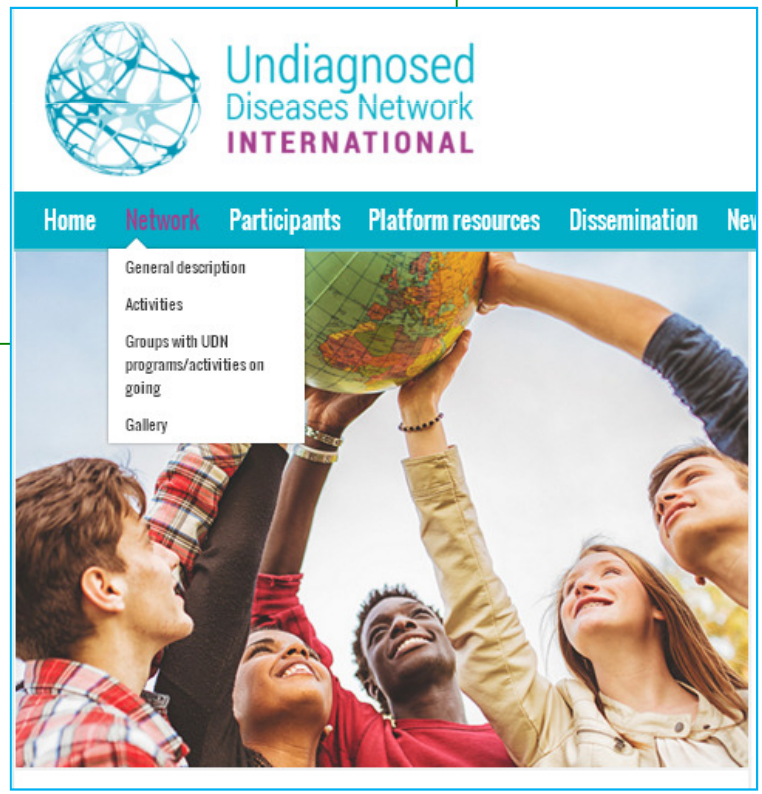
[g] Princess Margaret and King Edward Memorial Hospitals, Perth, Australia
[h] Office of Population Health, Public Health Division, WA Department of Health, Australia
[i] Telethon Kids Institute, Perth, Australia
[j] Western Australian Registry of Developmental Anomalies, Perth, Australia
[k] School of Paediatrics and Child Health, University of Western Australia, Perth, Australia
[l] Institute for Immunology and Infectious Diseases, Murdoch University, Perth, Australia
[m] Center for Biomedical Informatics, Department of Medicine, Harvard Medical School, Boston, Massachusetts, United States
[n] NIH Undiagnosed Diseases Program, National Human Genome Research Institute, National Institutes of Health, United States

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for your attention
&
for sharing…