



2° International Summer School “Rare Disease and Orphan Drug Registries”

Domenica Taruscio

Director

National Centre for Rare Diseases

domenica.taruscio@iss.it



II International Summer School RARE DISEASE AND ORPHAN DRUG REGISTRIES

September 15-19, 2014

Venue:
Casa dell'Aviatore
Viale dell'Università, 20
00185 Rome (Italy)

Organised by the
National Centre for Rare Diseases
Istituto Superiore di Sanità (ISS)



Course Director

Domenica Taruscio
Director
National Centre for Rare Diseases
Istituto Superiore di Sanità (ISS)
Viale Regina Elena, 299
Rome (Italy)

Secretariat and Contacts

Scientific Secretariat
Claudio Carta, Sabina Gainotti, Yllka Kodra, Luciano Vittozzi

Didactic Secretariat
Marta De Santis, Rosa Giuseppa Frazzica, Amalia Egle Gentile

Technical Secretariat
Linda Agresta, Norina Serpa, Giorgio Vincenti

e-mail: rareregistries-school@iss.it

National Centre for Rare Diseases
Istituto Superiore di Sanità (ISS)
Viale Regina Elena, 299
Rome (Italy)



ISTITUTO SUPERIORE DI SANITÀ' NATIONAL INSTITUTE OF HEALTH



**it is the leading scientific and technical public body of the
Italian National Health Service**

> 1800 persons : 7 Department , 8 National Centres

Research

Control

Training

Consultation

IN THE INTEREST OF PUBLIC HEALTH



National Centre for Rare Diseases

CNMR

Director: Dr. Domenica TARUSCIO

Centro Nazionale Malattie Rare

Responsabile: Domenica Taruscio

Le malattie rare

Cosa sono le malattie rare? Malattie che colpiscono 1 ogni 2000 abitanti

Malattie rare esentate in Italia

Elenco ordinato delle patologie e ricerca on line

Ho una malattia rara? Cosa fare? Guide all'esenzione

Dal sospetto di malattia ai cambi di diagnosi e cura

Il disabile e i suoi diritti

Una guida completa per orientarsi tra agevolazioni, detrazioni, sconti fiscali e altre opportunità

Le malattie rare e l'Europa

Le informazioni e le iniziative della Commissione Europea.

Ulteriori informazioni sul portale ad esse dedicato.

Rete Nazionale Malattie Rare

Telefono Verde Malattie Rare

Il servizio anonimo e gratuito fornisce informazioni su esenzioni, centri di cura, associazioni, sperimentazioni, ricerca, non formulando diagnosi cliniche. Il numero è attivo dal lunedì ai venerdì dalle ore 9,00 alle ore 13,00.

FAQ

Le domande più frequenti rivolte al Centro Nazionale Malattie Rare per ricevere informazioni sulle patologie esenti ticket, i presidi accreditati, le associazioni di pazienti, ecc.

Il Centro Nazionale Malattie Rare

Ricerca scientifica finalizzata a prevenzione, sorveglianza, diagnosi e trattamento delle malattie rare, è la principale attività del CNMR.

CentroNazionale-MalattieRare

CNMR

In rilievo

HALF OF A SCORE - Video scritto e diretto da Gianni del Corral versione in lingua inglese versione sottotitolata versione lunga con intervista(eng)

RARE DISEASES AND ORPHAN DRUGS - An International Journal of Public Health Call for paper submission

11 dicembre 2013 Apertura ISCRIZIONI - Convegno: "Prevenzione primaria di malformazioni congenite e screening neonatale esteso"

21-22 ottobre 2013 2nd International Workshop RARE DISEASES AND ORPHAN DRUG REGISTRIES.

1-2 Novembre 2013 5th International Conference on Rare Diseases and Orphan Drugs - St. Petersburg

ICORD International Conference on Rare Diseases & Orphan Drugs

Appuntamenti

16 Dicembre - STATE OF THE ART FOR A GLOBAL ALLIANCE

30 Gennaio - XVII CONVEGNO PATOLOGIA IMMUNE E MALATTIE ORFANE

27 Febbraio - VI INTERNATIONAL MEETING ON PULMONARY RARE

Tematiche

- Acido folico e folati prima della gravidanza
- Associazione Pazienti
- Farmaci Orfani
- Linee Guida
- Medicina Narrativa
- Progetti europei
- Test genetici

Registri

Registro Nazionale Malattie Rare

Registro Nazionale Farmaci Orfani

Registro Italiano Fibrosi Cistica (RIFC)

Concorso il volo di Pegaso

"Il Volo di Pegaso - Raccontare le malattie rare: parole e immagini": il concorso artistico letterario nato per garantire spazi espressivi e visibilità alle persone con malattie rare.

Notiziario CNMR e altre pubblicazioni

Notiziario

Il Notiziario del CNMR (Supplemento al Notiziario ISS), i notiziari ISS e i rapporti ISTISAL.

Ultimo Notiziario CNMR pubblicato

Are riservate

Accesso alle aree riservate per gli autorizzati in possesso delle credenziali di autenticazione

Link

Istituzioni internazionali

Istituzioni nazionali

Newsletter sulle malattie rare

Regioni

Siti e portali sulle malattie rare

Mission:

research, information, education, surveillance and epidemiological studies for prevention, diagnosis, treatment of rare diseases

Official Bulletin N. 157, 07.07.2008

NATIONAL REGISTRIES
RARE DISEASES
ORPHAN DRUGS



NATIONAL NETWORK ON
RARE DISEASES



ORPHAN DRUGS



EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

ITALIAN NETWORK
FOLIC ACID



efsa
European Food Safety Authority



PATIENTS' ASSOCIATIONS



RESEARCH



**Research
activities**

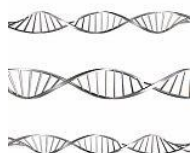
**Public health
activities**

**Educational
activities**

EDUCATION



GENETIC TESTS



NARRATIVE MEDICINE



European
projects!

European Union Committee of Experts
on Rare Diseases

EURO
PLAN



EPIRARE

GUIDELINES



EPIRARE
European Platform for Rare Disease Registries



37 partners in 16 countries in 3 continents




EUROPLAN
European Project for Rare Diseases National Plans Development



EUROPLAN
European Project for Rare Diseases National Plans Development

For more information:
www.europlanproject.eu
or contact:
euroman@iias.it

Domenica Tancredi
National Centre for Rare Diseases
Istituto Superiore di Sanità
Via Regina Elena, 299
00161 Roma - Italy
Tel. +37 06 4992 4016
www.iass.it/cnrmr

A three-year project co-funded by the European Commission within the framework of the EU program of Community Action in the field of Public Health



Rare Best Practices




EU NETWORK OF EXPERTS ON NEWBORN SCREENING



ISTITUTO SUPERIORE DI SANITA'



National Centre for Rare Diseases

CNMR

EUCERD
European Union Committee of Experts on Rare Diseases



RDConnect



BURQOL RD

SOCIAL ECONOMIC BURDEN AND HEALTH-RELATED QUALITY OF LIFE IN PATIENTS WITH RARE DISEASES IN EUROPE



www.burqol-rd.com

NORD
National Organization for Rare Disorders

CORD Canadian Organization for Rare Disorders

EURORDIS
Rare Diseases Europe

ICORD
International Conference on Rare Diseases & Orphan Drugs



E-Rare



IRDIRC
INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

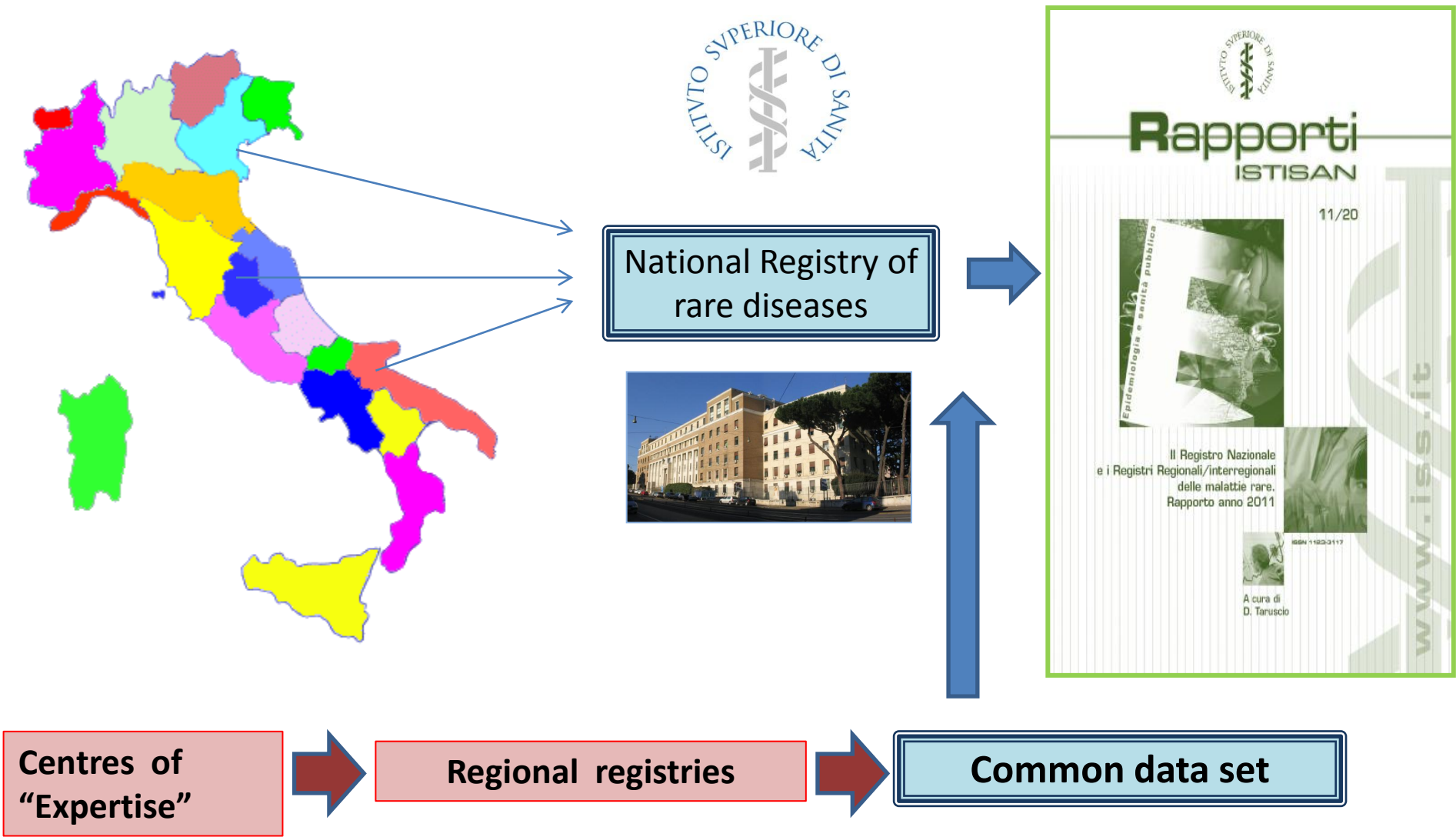


euRoCAT
European surveillance of congenital anomalies

EUROCAT Joint Action 2011-2013
Funded by the Public Health Programme 2008-2013 of the European Commission
WHO Collaborating Centre for the Surveillance of Congenital Anomalies




National Registry of Rare Diseases (D.M.279/2001)





A three year project co-funded by the European Commission within the framework of the EU program of Community Action in the field of Public Health



[Home](#) [The project](#) [Workpackages](#) [Partners](#) [Join us](#) [The EPIRARE Survey](#) [Deliverables](#) [Meetings](#) [News](#) [Contact](#)



In Europe a disease is considered rare when it affects no more than 5 individuals among 10,000 persons

News

- September 15-19, 2014

II International Summer School
RARE DISEASE AND ORPHAN DRUG
REGISTRIES
also in on-line streaming

[\[website\]](#)

[\[program\]](#)

[\[streaming on line\]](#)

EPIRARE (European Platform for Rare Disease Registries) is a three-year project co-funded by the European Commission within the EU Program of Community Action in the field of Public Health. EPIRARE started officially on April 15, 2011.

Final EPIRARE Deliverables

The EPIRARE deliverables are available.

The issues raised by the legislation on Personal Data Protection and the actions taken by EPIRARE are dealt with in the Deliverables 2.1, 2.2, 2.3, 2.4.

The EPIRARE Deliverables D3, D4, D5; D9.3 describe the basic features of a European Platform for Rare Disease Registries.

These documents have been elaborated on the basis of surveys and consultations involving the many potential stakeholders of the Platform.

The results of the surveys and consultations are reported in the other EPIRARE Deliverables.



PETITION TO THE EUROPEAN PARLIAMENT

Special Provisions for rare diseases
in the current proposal for a General
Data Protection Regulation

RD-CONNECT

An integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research



Proposal submitted under:

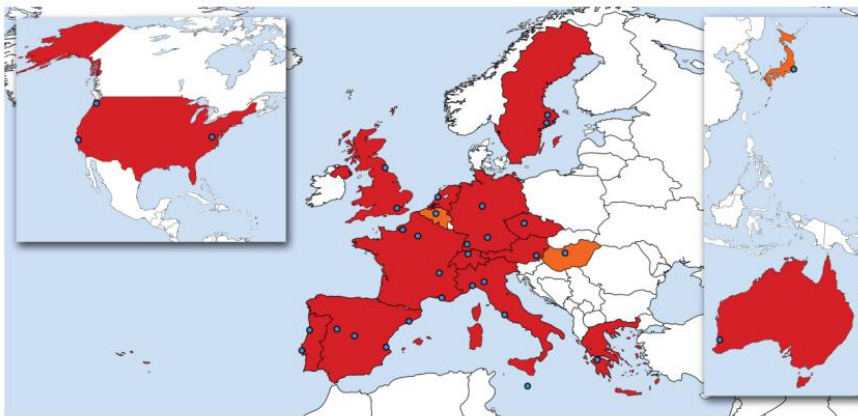
FP7 HEALTH.2012.2.1.1-1-C:

"Databases, biobanks and clinical bio-informatics hub for rare diseases"

Coordinator: [Hanns Lochmüller](#)



Global partner distribution



Workpackage leaders

WP1: Coordination



Hanns Lochmüller
Newcastle and TREAT-NMD

WP2: Patient registries



Domenica Taruscio
ISS and EPIRARE

WP3: Biobanks



Lucia Monaco
Fondaz. Telethon & EuroBioBank

WP4: Bioinformatics



Christophe Bérout
INSERM Montpellier

WP5: Unified platform



Ivo Gut
CNAG Barcelona

WP6 Ethical/legal/social



Mats Hansson
Uppsala

WP7: Impact and innovation



Kate Bushby
Newcastle and EUCERD/ EJARO

<http://www.rd-connect.eu>



[Indicators for National Plans](#)

The development and use of indicators is an integral part of planning and designing health and social services for Rare Diseases [»](#)

[Reports and Publications](#)

Visit this section and download all the EUROPLAN documents reports and publications [»](#)

[News](#)

II International Summer School - Rare disease and orphan drug registries in on-line streaming [\[Streaming on line\]](#) - [\[website\]](#)



IRDiRC

INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

Co-operation at international level
to stimulate, better coordinate & maximise output
of rare disease research efforts around the world

www.irdic.org

IRDiRC vision and 2020 goals in rare diseases research



200 New Therapies



Means to Diagnose Most Rare Diseases

www.irdic.org

MAIN FACTORS INFLUENCING SCIENTIFIC RESEARCH IN RARE DISEASES

- Presence of **registries and biobanks:**
sharing epidemiological and biological samples
- **International collaboration** *trans-national call*
- Presence of **Patient's Associations/Groups**
- Dedicated **resources** (funds, etc.)

RARE DISEASES AND ORPHAN DRUGS

An International Journal of Public Health

**Announcing
Call for Paper Submissions
First Issue in December 2013**

Co-Editors-in-Chief

Dr. Domenica Taruscio
*Director
National Centre for Rare Diseases
Istituto Superiore di Sanità
Italian National Institute of Health
Rome, Italy*

Prof. Holger Schönemann
*Professor and Chair
Department of Clinical
Epidemiology and Biostatistics
McMaster University
Hamilton, Canada*

RARE Journal (www.rarejournal.org) is a new international open access, online, peer-reviewed journal published three times per year, with **no publishing fees**.

The mission of **RARE Journal** is to provide an **advanced forum** on important aspects of **public health, health policy and clinical research** in ways that will improve health care and outcomes for persons suffering from rare diseases, as well as globally increase rare diseases experience sharing.

RARE Journal publishes original research articles, case reports, systematic reviews and meta-analyses, reports of clinical practice guidelines, HTA reports, epidemiological registry reports, commentaries, letters to Editors, meeting and project reports.

Selected examples of topics covered by the journal are given below:

- ▶ Health policies and services organization on rare diseases and orphan drugs
- ▶ Rare disease epidemiology research and registries
- ▶ Clinical research and methodology
- ▶ Rare disease best practices
- ▶ Comparative effectiveness research
- ▶ Practice guidelines
- ▶ Socio-economical analysis
- ▶ Primary prevention, risk factors and screening
- ▶ Health promotion
- ▶ Quality assurance (genetic testing, etc.)
- ▶ Legal and ethical issues
- ▶ Narrative medicine
- ▶ Patients' needs and health-related quality of life

RARE Journal also anticipates special issues dedicated to a specific topic, such as particular rare diseases or a group of rare diseases. Suggestions for topics are welcome. Accepted papers are published in electronic form and indexed for database reference.

RARE JOURNAL:

- No Publication Fees
- Timely Peer-Review
- Open Access to Articles Online
- Expert Editorial Assistance
- High Scientific Quality

For Further Information Please Visit:
WWW.RAREJOURNAL.ORG

RARE Journal Editorial Secretariat
e-mail: secretariat@rarejournal.org

FIRST ISSUE COMING IN DECEMBER 2013!

www.rarejournal.org

Publisher
Istituto Superiore di Sanità



The RARE-Bestpractices project is funded by the European Union Research Framework Programme. Project Ref.: nr 356890. Sole responsibility lies with the authors and the European Commission is not responsible for any use that may be made of the information contained therein.

WWW.RAREBESTPRACTICES.EU

Monday, 15 September

- 8.30 Registration and pre-test
- 9.15 Welcome and presentation of the course objectives
- 9.30 Introduction of participants and presentation of their activities (D. Taruscio)
- 10.30 Coffee Break
- 11.00 Registry types, Aims, Building a registry, Management, Sustainability (L. Korngut)
- 12.00 Epidemiological study design, data sources of population study variables (Y. Kodra)
- 13.00 Lunch
- 14.00 Cooperative learning on matters dealt with in the morning (CNMR, in collaboration with the speakers)
14:00 - 1st phase: Small groups
15:00 - 2nd phase: Larger groups
16:10 - 3rd phase: Plenary session
- 17.00 Adjournal

Tuesday, 16 September

- 8.30 Reference Standards and Catalogues (P. Landais)
- 10.30 Coffee break
- 11.00 Coding of rare disease (P. Landais)
- 12.00 Omics and links with biobanks and registries (M. Calissano)
- 13.00 Lunch
- 14.00 Cooperative learning on matters dealt with in the morning (CNMR, in collaboration with the speakers)
14:00 - 1st phase: Small groups
15:00 - 2nd phase: Larger groups
16:10 - 3rd phase: Plenary session
- 17.00 Adjournal

Wednesday, 17 September

- 8.30 Epidemiologic analyses, confounders, sample stratification (F. Bianchi, M. Santoro)
- 10.30 Coffee break
- 11.00 Personal Data Protection; Informed Consent; Registry termination (S. Gainotti)
- 13.00 Lunch
- 14.00 Cooperative learning on matters dealt with in the morning (CNMR, in collaboration with the speakers)
14:00 - 1st phase: Small groups
15:00 - 2nd phase: Larger groups
16:10 - 3rd phase: Plenary session
- 17.00 Adjournal

Thursday, 18 September

- 8.30 Patient unique identifier (M. Posada)
- 10.30 Coffee break
- 11.00 Quality assurance (M. Posada)
- 13.00 Lunch
- 14.00 Cooperative learning on matters dealt with in the morning (CNMR, in collaboration with the speakers)
14:00 - 1st phase: Small groups
15:00 - 2nd phase: Larger groups
16:10 - 3rd phase: Plenary session
- 17.00 Adjournal

Friday, 19 September

- 8.30 The application of the Human Phenotype Ontology (M. Haendel)
- 10.00 Coffee break
- 10.15 Data reporting and dissemination (E. Bravo, P. De Castro)
- 11.45 Discussion, evaluation questionnaire and post-test
- 12.30 Certificate delivery and greetings
- 13.00 Lunch and Course closure

Objectives and Methods

The II International Summer School on Rare Disease and Orphan Drug Registries will take participants through the main concepts and practical steps that must be undertaken in the establishment and management of a rare disease registry. The programme builds on the challenges posed by the evolving technological innovations, new data collection and sharing possibilities, legal requirements and security needs.

The School will consist of frontal presentations followed by Cooperative Learning (CL) sessions, where participants work together to maximize their own and each other's learning potential. Basic elements of CL are positive interdependence, individual accountability, face-to-face interaction, group processing and exercise of small-group interpersonal skills.

Speakers

Fabrizio Bianchi - National Council of Research, Pisa, Italy

Elena Bravo - Dep. of Cell Biology & Neuroscience, ISS, Rome, Italy

Mattia Calissano - MRC Centre for Neuromuscular Diseases at Newcastle, UK

Paola De Castro - Publishing Unit, ISS, Rome, Italy

Sabina Gainotti - National Centre for Rare Diseases, ISS, Rome, Italy

Melissa Haendel - Oregon Health & Science University, USA

Ylika Kodra - National Centre for Rare Diseases, ISS, Rome, Italy

Lawrence Korngut - University of Calgary, Canada

Paul Landais - Université de Montpellier, France

Manuel Posada - Institute of Health Carlos III, Madrid, Spain

Michele Santoro - National Council of Research, Pisa, Italy

Domenica Taruscio - National Centre for Rare Diseases, ISS, Rome, Italy

Raphael (Vatican Museum) - The school of Athens (1509-1511)

Plato, Aristotle, Pythagoras, Diogenes, Heraclitus, Euclides, etc.



AREAS OF THE COUNCIL RECOMMENDATION



7. SUSTAINABILITY

6. EMPOWERMENT OF PATIENT ORGANISATIONS

5. GATHERING THE EXPERTISE ON RARE DISEASES AT EUROPEAN LEVEL

4. CENTRES OF EXPERTISE AND EUROPEAN REFERENCE NETWORKS FOR RARE DISEASES

3. RESEARCH ON RARE DISEASES

2. ADEQUATE DEFINITION, CODIFICATION AND INVENTORING

1. PLANS AND STRATEGIES IN THE FIELD OF RARE DISEASES