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

Domenica Taruscio
Director
National Centre for Rare Diseases
domenica.taruscio@iss.it

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, Ylka 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)

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)

National Centre for Rare Diseases
CNMR

ISTITUTO SUPERIORE DI SANITA'
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

Training

Control

Consultation

IN THE INTEREST OF PUBLIC HEALTH

National Centre for Rare Diseases

CNMR

Diretctor: Dr. Domenica TARUSCIO

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

National Centre for Rare Diseases
CNMR

ITALIAN NETWORK
FOLIC ACID

efsa
European Food Safety Authority

PATIENTS' ASSOCIATIONS

Research activities

Public health activities

Educational activities

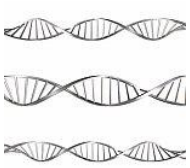
RESEARCH

ISTITUTO SUPERIORE DI SANITÀ

NATIONAL INSTITUTES OF HEALTH

EDUCATION

GENETIC TESTS



NARRATIVE MEDICINE

European projectsl

European Union Committee of Experts on Rare Diseases

EURO PLAN

E-Rare

EPIRARE

GUIDELINES


EPIRARE
European Platform for Rare Disease Registries
37 partners in 16 countries in 3 continents
EURORDIS
eurocat
HD


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:
europlan@iss.it
Domenica Taruscio
National Centre for Rare Diseases
Istituto Superiore di Sanità
Viale Regina Elena, 299
00161 Roma - Italy
Tel. +39 06 4990 4016
www.iss.it/cnmr
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
CNMR


EU NETWORK
OF EXPERTS ON
NEWBORN SCREENING


ISTITUTO SUPERIORE DI SANITÀ
National Centre for Rare Diseases
CNMR


EUCERD
European Union Committee
of Experts on Rare Diseases


RD Connect


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

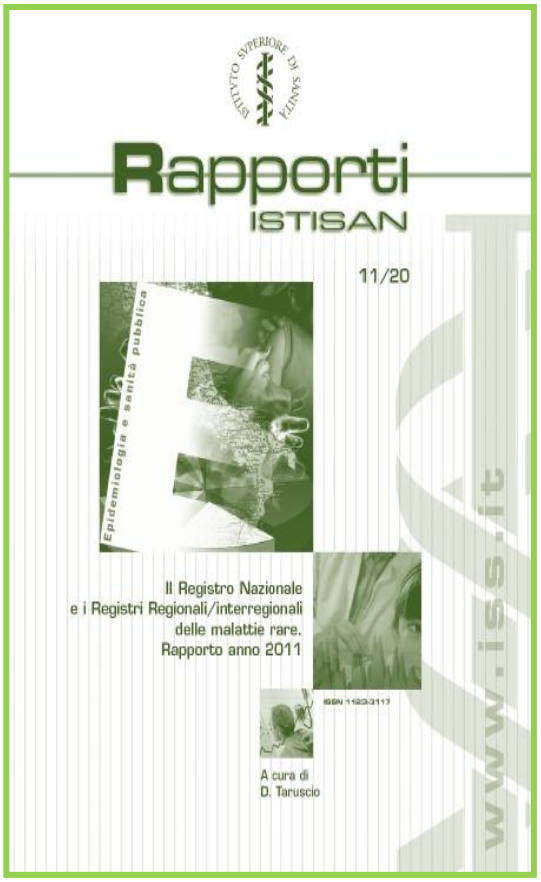

E-Rare


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
University of
ULSTER

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

National Registry of rare diseases



Centres of "Expertise" → Regional registries → Common data set

www.epirare.eu

EPIRARE
European Platform for Rare Disease Registries

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

Home | The project | Workpackages | Partners | Join us | The EPIRARE Survey | Deliverables | Meetings | News | Contact

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.

RD-CONNECT

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

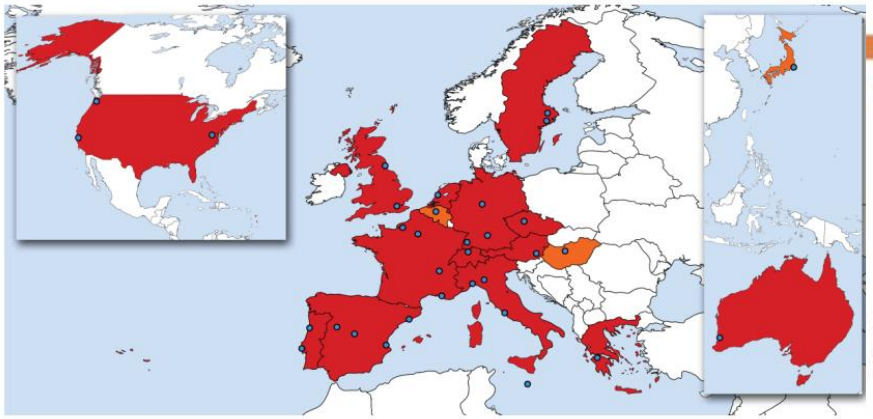
Proposal submitted under:

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

http://www.rd-connect.eu


EUROPLAN
European Project for Rare Diseases National Plans Development
THE PROJECT
PARTNERS
EVENTS
CAPACITY BUILDING
NATIONAL PLANS
DOCUMENTS
LINKS
15 - 19 SEPTEMBER 2014
Rare diseases and orphan drug registries
II International Summer School
IN ON-LINE STREAMING
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 Schünemann
Professor and Chair
Department of Clinical
Epidemiology and Biostatistics
McMaster University
Hamilton, Canada

www.rarejournal.org

Publisher
Istituto Superiore di Sanità

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!

Rare Best Practices

The RARE-Bestpractices project is funded by the European Union Seventh Framework Programme. Project Ref.: nº 305690. 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	*Adjourn*

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	*Adjourn*

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	*Adjourn*

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	*Adjourn*

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.

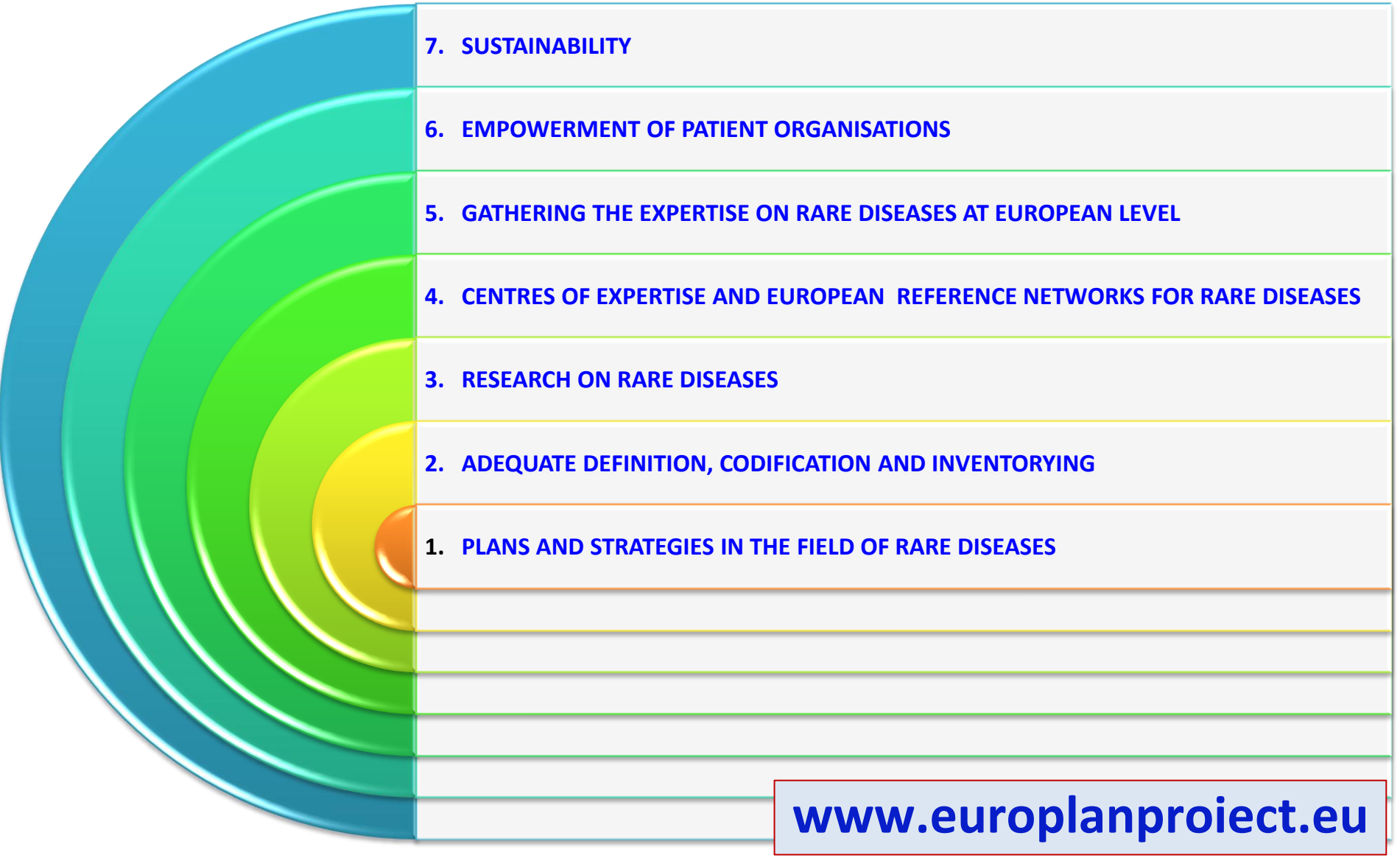
Speakers

Fabrizio Bianchi *- National Council of Research, Pisa, Italy*
Elena Bravo *- Dep. of Cell Biology & Neurioscience, 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*
Yllka 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



www.europlanproiect.eu