STATI GENERALI DELLE MALATTIE RARE:

MALATTIE RARE E SVILUPPO ECONOMICO

Martedì, 18 settembre 2012 ore 9.30 – 13.00

AUDITORIUM MINISTERO DELLA SALUTE

Domenica Taruscio
Direttore Centro Nazionale Malattie Rare
Istituto Superiore di Sanità
Roma

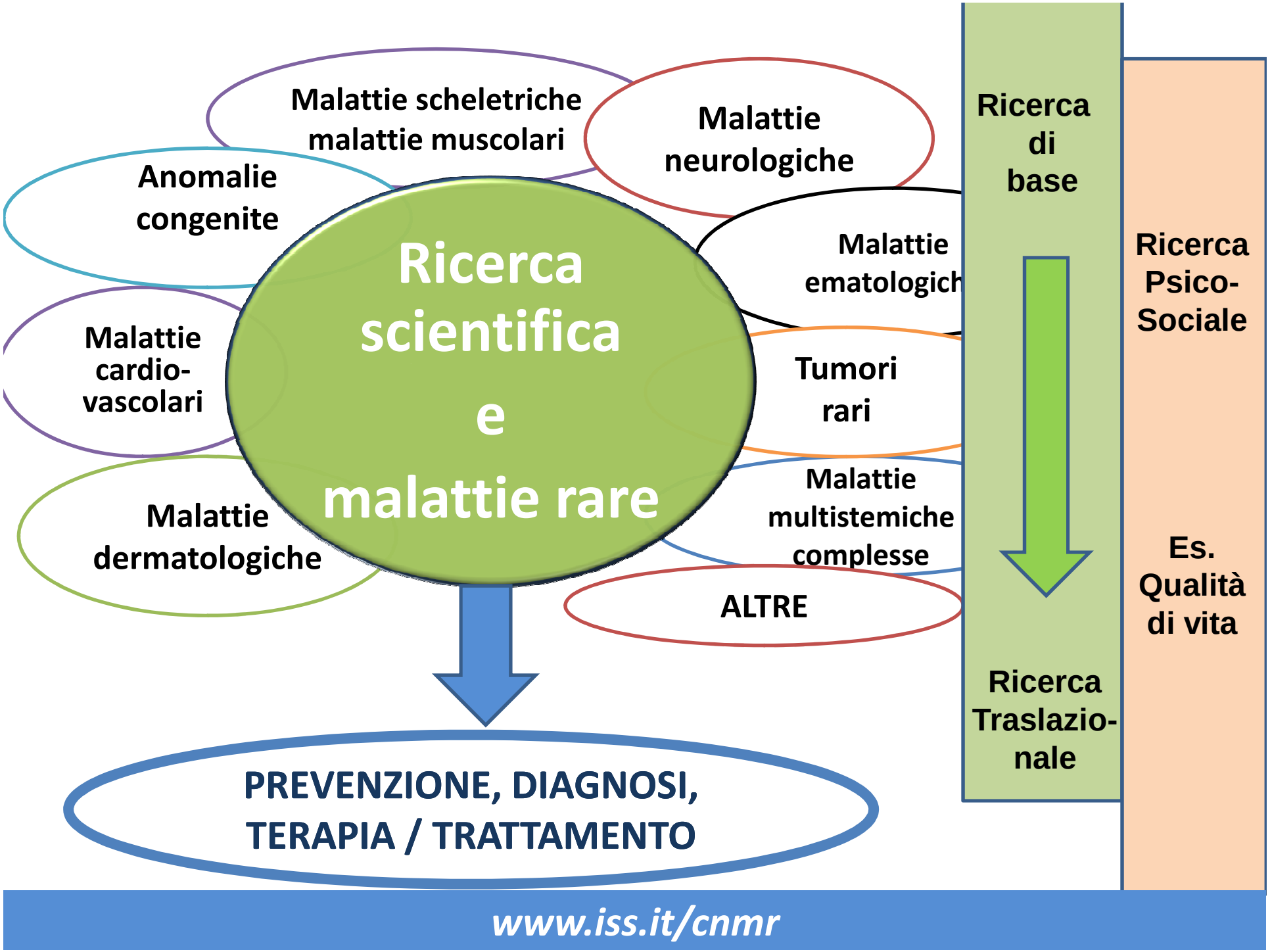
FATTORI CHE INFLUENZANO LA RICERCA SCIENTIFICA NELLE MALATTIE RARE

- **Presenza di registri e biobanche**
  condivisione di campioni biologici e superamento di problemi legati ai piccoli numeri

- **Collaborazioni internazionali**
  progetti europei e internazionali

- Presenza di **Associazioni di Pazienti**

- Disponibilità di **finanziamenti** dedicati


Malattie scheletriche
malattie muscolari
Malattie
neurologiche
Anomalie
congenite
Malattie
ematologich
Ricerca
scientifica
e
malattie rare
Malattie
cardio-
vascolari
Tumori
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complesse
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Es.
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di vita
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Traslazio-
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3.7.2009 EN Official Journal of the European Union C 151/7

COUNCIL RECOMMENDATION

of 8 June 2009

on an action in the field of rare diseases

(2009/C 151/02)

… **RECOMMENDS THAT MEMBER STATES:**

Elaborate and adopt a plan or strategy as soon as possible, *preferably by the end of 2013 at the latest*

Take action to integrate current and future initiatives at local, regional and national levels into their plans or strategies **for a comprehensive approach**

Define a limited number of priority actions within their plans or strategy, **with objectivs and follow-up mechanisms**

Take note of the development of guidelines and recommendations of the ongoing european project EUROPLAN (www.europlanproject.eu)

EUROPEAN COUNCIL RECOMMENDATION



Council Recommendation of June 2009 on an action in the field of rare diseases (2009/C 151/02)

Recommends that member States:

1. PLANS AND STRATEGIES IN THE FIELD OF RARE DISEASES
2. ADEQUATE DEFINITION, CODIFICATION AND INVENTORYING
3. RESEARCH ON RARE DISEASES
4. CENTRES OF EXPERTISE AND EUROPEAN REFERENCE NETWORKS FOR RARE DISEASES
5. GATHERING THE EXPERTISE ON RARE DISEASES AT EUROPEAN LEVEL
6. EMPOWERMENT OF PATIENT ORGANISATIONS
7. SUSTAINABILITY

National Centre for Rare Diseases
CNMR

EUROPLAN

European Project for Rare Diseases National Plans Development

A three year (2008-2011) project, funded by DG SANCO

Coordinated by the Italian National Centre for Rare Diseases

Main goals

- **Elaborate recommendations as "tools" to facilitate the development of a national plan or strategy for rare diseases**
- **Elaborate indicators for monitoring national plans/strategies**
- **Discuss the recommendations with stakeholders**
- **Disseminate the recommendations**

EUROPLAN: *(2008-2011; 2012-2015) project, funded by the EU Commission*





57 **Partners**

Associated & Collaborating:
clinicians, scientist, health authorities, patients' Groups

from:

34 **Countries**

+

EURORDIS
Rare Diseases Europe

www.europlanproject.eu

RESULTS


European Union Committee of Experts on Rare Diseases

2009 REPORT
ON INITIATIVES AND INCENTIVES
IN THE FIELD OF RARE DISEASES

OF THE EUROPEAN UNION COMMITTEE OF EXPERTS ON RARE DISEASES

Joint Action to Support the Scientific Secretariat of the Rare Diseases Task Force/European Union Committee of Experts on Rare Diseases (N° 2008 22 91)

European Project for Rare Diseases National Plans Development - EUROPLAN (N° 2007 119)

➢**Report on EU Member States <u>initiatives</u>**, previous and current experiences on rare diseases (Joint document RTFD/EUCERD-EUROPLAN)


Public Health

EUROPLAN

EUROPLAN
EUROPEAN PROJECT FOR RARE DISEASES NATIONAL PLANS DEVELOPMENT

FUNDED:	EUROPEAN COMMISSION PUBLIC HEALTH PROGRAM 2003-08
PRIORITY AREA:	1. HEALTH INFORMATION (HI 2007).
ACTION:	1.4 DEVELOPING STRATEGIES FOR INFORMATION EXCHANGE AND RESPONDING TO NON-COMMUNICABLE HEALTH THREATS
CONTRACT NUMBER:	101120 - 2007119
PROJECT COORDINATOR:	DOMENICA TARUSCIO (ISS)
MAIN PARTNER:	ISTITUTO SUPERIORE DI SANITÀ (ITALY)
ASSOCIATED PARTNERS:	10
PROJECT PERIOD:	1 APRIL 2008 – 31 MARCH 2011
REFERRING PERIOD:	1 JUNE 2010 – 31 DECEMBER 2010

RECOMMENDATIONS
FOR THE DEVELOPMENT OF NATIONAL PLANS FOR RARE DISEASES
GUIDANCE DOCUMENT
Guidance_Doc_EUROPLAN_20100601

DOCUMENT AUTHORS	ALL EUROPLAN PARTNERS
REVISION DATE	8 JULY 2010
DIFFUSION LEVEL	PUBLIC DOCUMENT
INTENDED TARGET	PARTICIPANTS OF THE 16 NATIONAL CONFERENCES
KEY WORDS	RECOMMENDATIONS, NATIONAL PLANS, RARE DISEASES

➢**EUROPLAN <u>recommendations</u>** for the elaboration of National Plans or Strategies for rare diseases


PLAN Draft version. Selecting indicators to evaluate the achievements of RD initiatives 1

EUROPLAN
European Project for Rare Diseases National Plans Development

European Project for Rare Diseases National Plans Development (EUROPLAN)

Selecting indicators to evaluate the achievements of RD initiatives

By WP5: Manuel Posada, Manuel Hens, María José Carroquino and EUROPLAN Working Group on Indicators

Rare Diseases Research Institute
Instituto de Salud Carlos III

Madrid March, 29th, 2010

➢**Report on <u>indicators for monitoring</u> the implementation and evaluating** the impact of National Plan or Strategy for rare diseases

EUROPLAN DEFINITION OF A NATIONAL PLAN OR STRATEGY

a set of integrated and comprehensive health and social policy actions for rare diseases, to be developed and implemented at national level, characterized by identified objectives to be achieved within a specified timeframe.

The allocation of appropriate **resources** (human, financial, infrastructural)and its **monitoring and evaluation** are of special value to ensure the efficacy of the plan or strategy.

EUROPLAN Recommendations

reflect the EU Council Recommendation structure:

AREA 1. plans and strategies in the field of rare diseases

AREA 2. adequate definition, codification and inventorying

AREA 3. research on rare diseases

AREA 4. centres of expertise and european reference networks for rare diseases

AREA 5. gathering the expertise on rare diseases at european level

AREA 6. empowerment of patient organisations

AREA 7. sustainability

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EUCERD JOINT ACTION - Workpackage 4

"Support for the implementation
of plans or strategies at MS level (continuation of EUROPLAN)"

EUROPLAN

European Project for Rare Diseases National Plans Development

Project coordinated by the Italian National Centre for Rare Diseases
Italian National Institute of Health

2012-2015

A capacity building network with policy makers of all MS

for the implementation of Rare Diseases National Plans/Strategies

We will provide technical and scientific support for developing and implementing National Plans/Strategies on Rare Diseases

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EUROPLAN

European Project for Rare Diseases National Plans Development

Coordinated by the Italian National Centre for Rare Diseases
Italian National Institute of Health

2012-2015

EUROPLAN 2012-2015

provides technical and scientific
support to Countries
in order to develop and implement
National Plans and Strategies
for Rare Diseases





EUROPLAN 2012-2015

a three-year project, embedded in the EUCERD Joint Action as Work Package 4, it is coordinated by the Italian National Institute of Health - Italian National Centre for Rare Diseases.

Its main goal is to establish an international and interactive network of stakeholders (mainly policy makers) to speed up the elaboration and the implementation of Rare Diseases National Plans/Strategies, trough scientific and technical assistance, Workshops and the active participation of patients Groups (EURORDIS and National Alliances). *[read more]*

News

RARE DISEASE AND ORPHAN DRUG REGISTRIES
International Workshop
8-9 October 2012
ISS - Rome, Italy
Website - Agenda

The Czech government adopted on August 28/2012 in its Decree 633
The Czech National Plan for Rare Diseases 2012-2014
[see the document]

It's available the DRAFT (english version) of the
Strategy of The Netherlands in the field of Rare Diseases
[see the document]



EUROPLAN

European Project for Rare Diseases National Plans Development

Coordinated by the Italian National Centre for Rare Diseases
Italian National Institute of Health

2012-2015

Objectives

To provide technical and scientific support to develop and implement National Plans and Strategies for Rare Diseases.
This will be achieved by:

1. establishing an interactive Network of public health policy markers from participating Countries

2. producing a comprehensive, consistent and feasible operational proposal for National Plan / Strategy

3. supporting EURORDIS National Conferences





EUROPLAN 2012-2015

a three-year project, embedded in the EUCERD Joint Action as Work Package 4, it is coordinated by the Italian National Institute of Health - Italian National Centre for Rare Diseases.
Its main goal is to establish an international and interactive network of stakeholders (mainly policy makers) to speed up the elaboration and the implementation of Rare Diseases National Plans/Strategies, trough scientific and technical assistance, Workshops and the active participation of patients Groups (EURORDIS and National Alliances). *[read more]*

News

RARE DISEASE AND ORPHAN DRUG REGISTRIES
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8-9 October 2012
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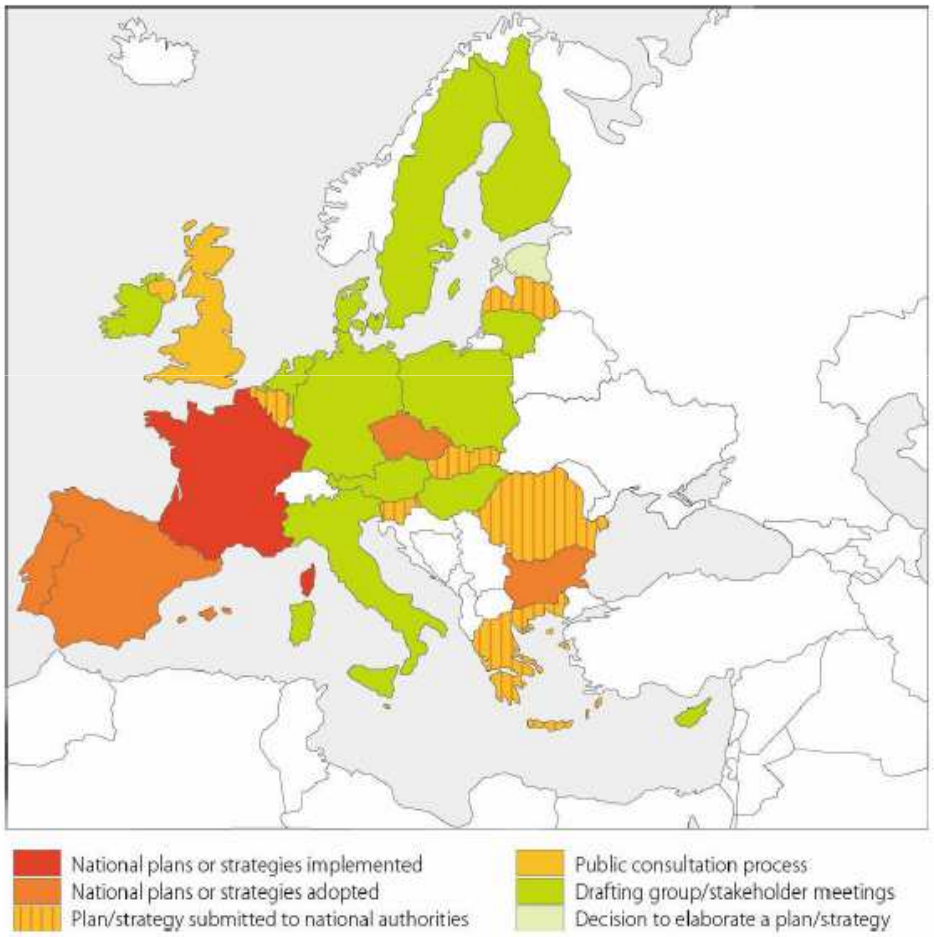
The Czech government adopted on August 28/2012 in its Decree 633 **The Czech National Plan for Rare Diseases 2012-2014**
[see the document]

It's available the DRAFT (english version) of the **Strategy of The Netherlands in the field of Rare Diseases**
[see the document]



Download the EUROPLAN flyer

DEVELOPMENT OF NP/NS IN EU MS



www.eucerd.eu



Rare Disease registries

- There are many registries (more than 500) on rare diseases
  with different aims
  with different coverage (regional, national, international)
  on a specific rare disease or on groups of them

- ***The EU Council Recommendation on Rare Diseases (adopted in June 2009)*** recommends **the implementation of registers and databases** for epidemiological purposes (point 15).
  Therefore, many new initiatives are expected in the coming years.

BUT:

- **There are several difficulties for EU data collection and exchange**
  deriving from rules on data protection, quality issues, ownership of data.

"Today, even though many rare disease registries exist, the fact that they all operate in different ways, hinders the effective systematic data collection and sharing necessary for a positive effect on the health of patients"
(www. eurordis.org)

EPIRARE

European Platform for Rare Disease Registries

Building Consensus and Synergies

European Platform for Rare Disease Registries

EPIRARE is a project co- funded by the EU Commission (DG SANCO)

Starting date: 15 April 2011

Duration: 30 months (Aprile 2011 – October 2013)

Coordinator of the project: National Centre for Rare Diesases, Italian National Institute of Health – Rome, Italy

www.epirare.eu

EPIRARE

European Platform for Rare Disease Registries

To define the needs of registries and databases on rare diseases

To agree on the Platform Scope, Governance and long-term sustainability

To agree on a Common data set, disease-specific data collection and data validation

To identify key issues and

To prepare a legal basis for the EU platform, taking into account privacy and ethical issues

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





RARE DISEASE AND ORPHAN DRUG REGISTRIES
International Workshop

October 8-9, 2012
Italian National Institute of Health – Viale Regina Elena, 299 - Rome (Italy)

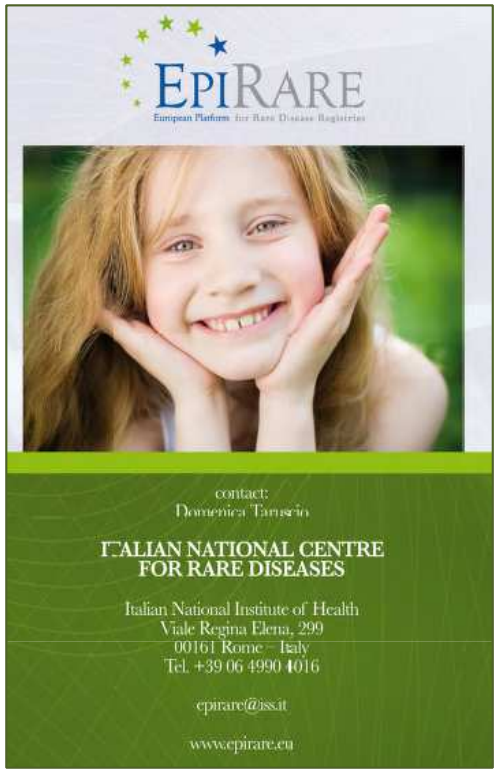
Agenda

The International Workshop "Rare Disease and Orphan Drug Registries" is organized in the framework of the EPIRARE project ("European Platform for Rare Disease Registries", co-funded by EU Commission, DG SANCO).
It is open to scientists, clinicians, patients' and parents' Associations, policy makers and enterprises. The main aims of the workshop are:

- (i) to share different experiences in the registration of rare diseases, including rare cancers and malformations, in Europe and beyond;
- (ii) to highlight the strengths and opportunities of linking rare disease registration activities, orphan drug post-

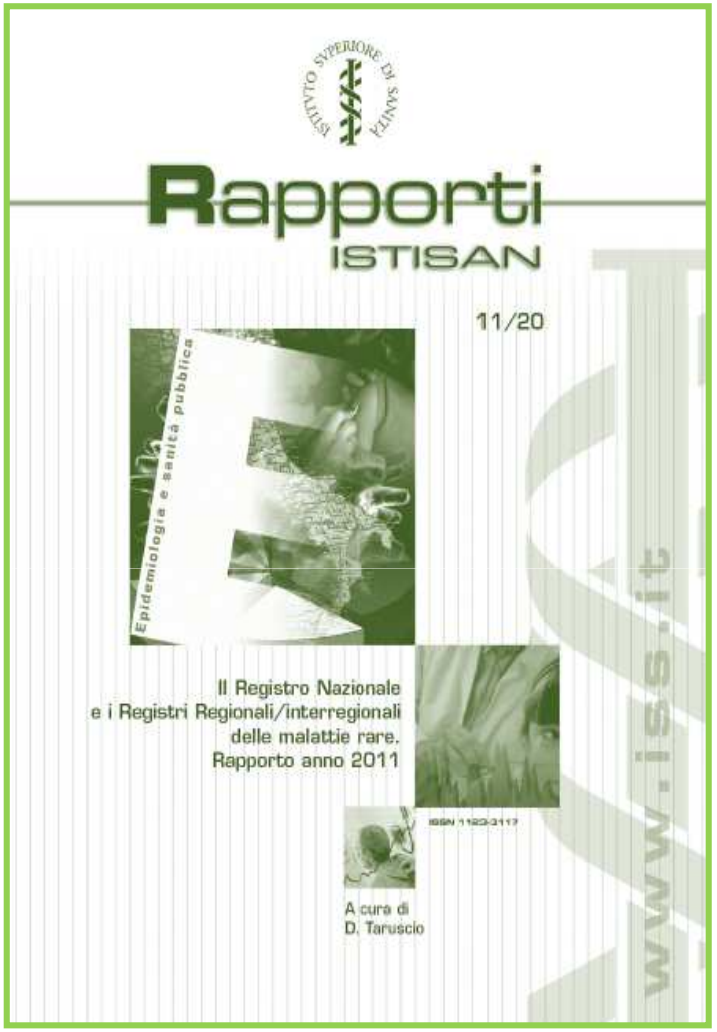






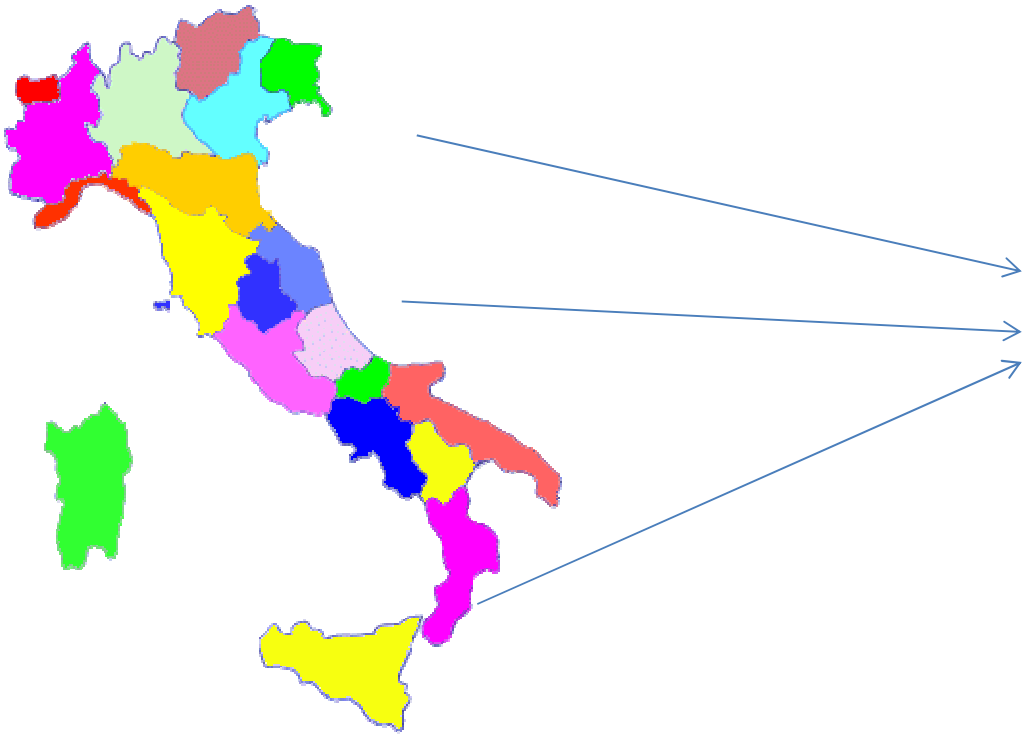


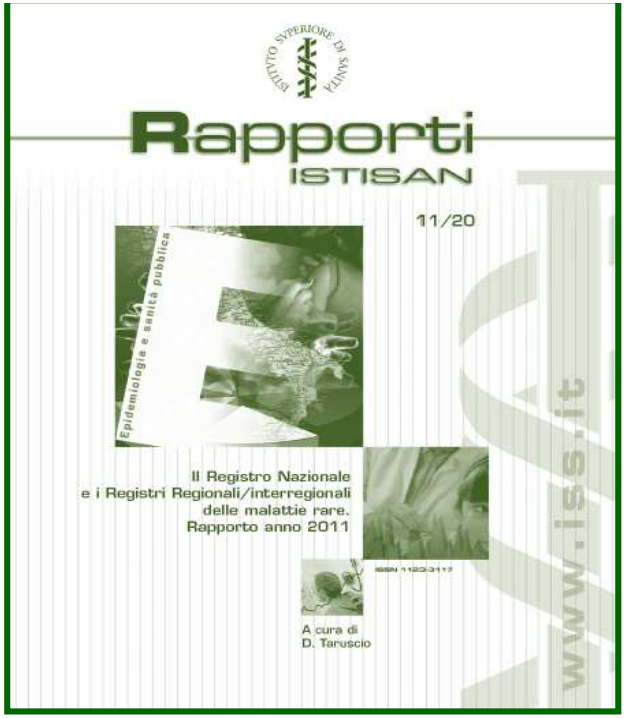
www.epirare.eu



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Flusso epidemiologico:
dai registri regionali al Registro nazionale





NATIONAL REGISTRY OF RARE DISEASES

EACH AUTHORIZED HOSPITAL IN A GIVEN REGION → COORDINATOR CENTRE OF THE REGIONAL REGISTRY → AGREED DATA SET (EVERY 6 MONTHS)



ACTIVITY/AREA

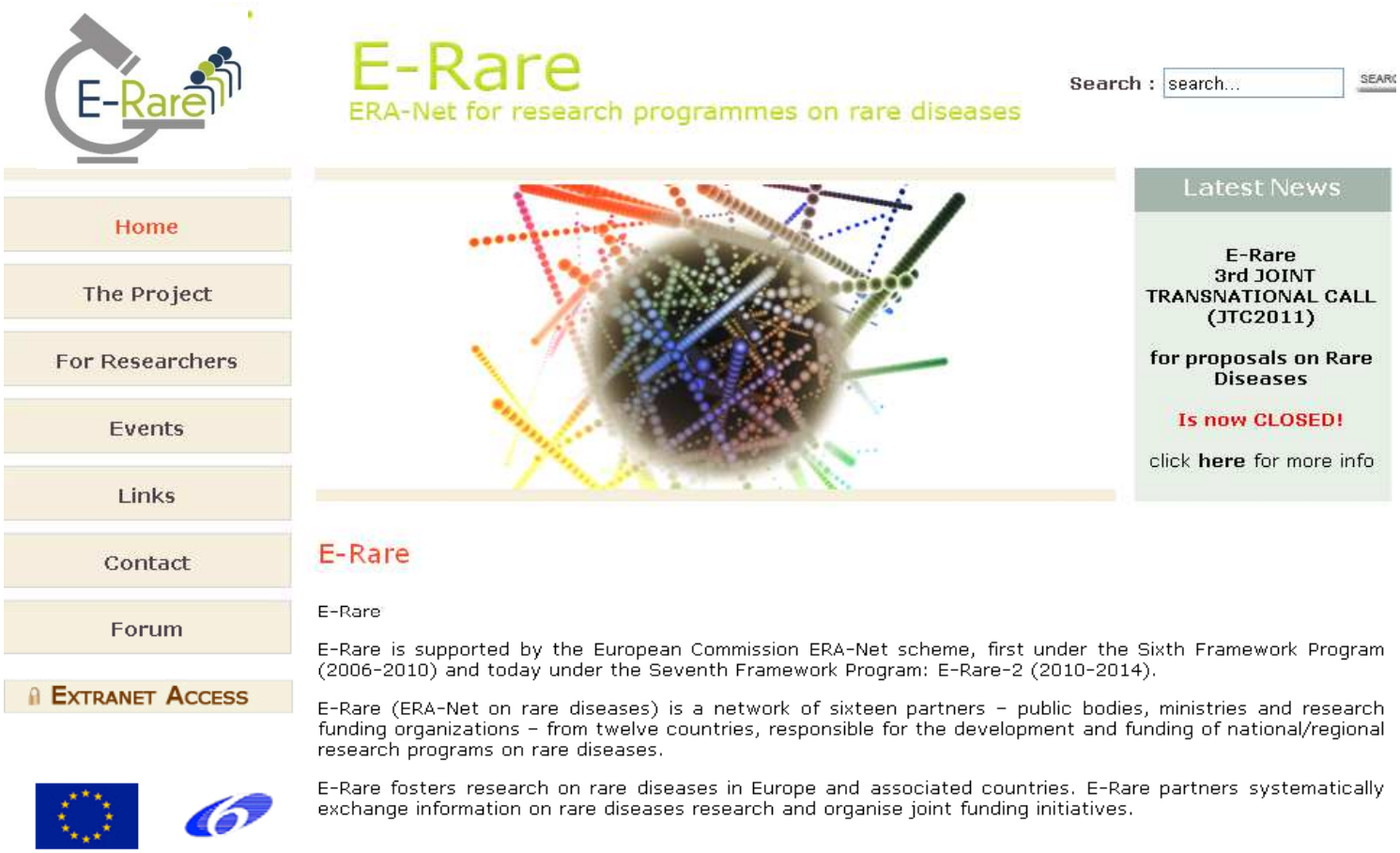
HEALTH.2012.2.1.1-1 (A-C) – OMICS FOR RARE DISEASES: INTEGRATING OMICS, BIOBANKS, REGISTRIES

HEALTH.2012.2.4.4 RARE DISEASES

RD-Connect

HEALTH.2013.4.2-3: New methodologies for clinical trials for small populations.

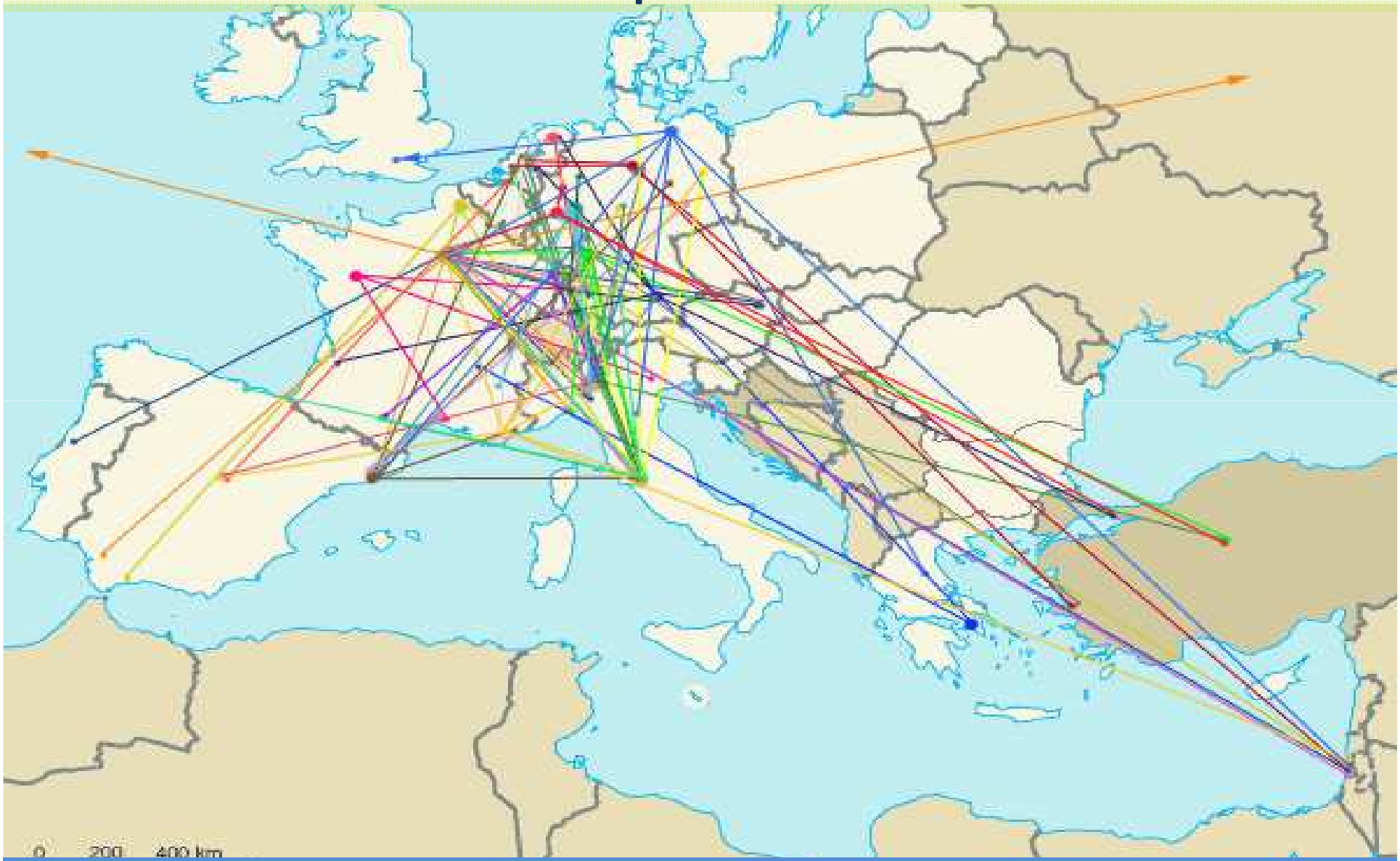
HEALTH.2013.1.2-1. Development of imaging technologies for therapeutic interventions in rare diseases



- Progetto co-finanziato dalla Commissione Europea (FP6; FP7)
- Network di **16 Partners** (public bodies, ministries and research funding organizations) da 12 Paesi (**Italia: Istituto Superiore di Sanità**)
- **Sviluppo e finanziamento** di programmi di ricerca scientifica a livello nazionale

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E-RARE
creazione di una rete europea di collaborazioni scientifiche



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IN FOCUS NEWS

BIOMEDICAL RESEARCH

Rare-disease project has global ambitions

Consortium aims for hundreds of new therapies by 2020.

BY ALISON ABBOTT

Prader–Willi syndrome. Fabry renal disease. Spinocerebellar ataxia. Few people have heard of these and the other 'rare diseases', some of which affect only hundreds of patients worldwide. Drug companies searching for the next blockbuster pay them little attention. But the diseases are usually incurable — and there are thousands of them.

This week, the US National Institutes of Health (NIH) and the European Commission launch a joint assault on these conditions, whose small numbers of patients make it difficult to test new treatments and develop diagnostic methods. The International Rare Disease Research Consortium being formed under the auspices of the two bodies has the ambitious goal of developing a diagnostic tool for every known rare disease by 2020, along with new therapies to treat 200 of them. "The number of individuals with a particular rare disease is so small that we need to be able to pool information from patients in as many countries as possible," says Ruxandra Draghia-Akli, the commission's director of health research.

At the launch meeting in Bethesda, Maryland, on 6–8 April, prospective partners will map out research strategies to identify diagnostic biomarkers, design clinical trials and coordinate genome sequencing in these diseases. Nearly all the rare diseases, of which there are an estimated 6,000–8,000, are the result of small genetic changes.

The meeting will also discuss the governance of the project, which is most likely to be modelled on the pioneering Human Genome Project. As such, the consortium is open to research agencies and organizations from all over the world. Representatives from countries including Canada, Japan and some individual European nations are all attending the meeting, and may join the consortium. Those wishing to participate will have to pledge a minimum financial contribution, which has not yet been agreed, and share all relevant data. Indeed, the project will have to overcome numerous obstacles to information sharing, such as the fact that physicians in different countries often use entirely different words to describe the same disease.

Draghia-Akli points out that the project could yield major benefits for the emerging field of personalized medicine — another political priority for the NIH and the commission — which also faces the challenge of small populations of patients.

"We need to be able to pool information from patients from as many countries as possible."

Regulatory agencies such as the US Food and Drug Administration and the European Medicines Agency rely on large, randomized and controlled clinical trials when deciding whether to approve new medicines, and one of the aims of the consortium will be to develop alternative clinical-trial methods for diseases that affect few people.

These methods are becoming ever more important now that genome analysis is helping to break down common diseases into ever smaller subclasses. "Soon there will be no disease called breast cancer," says Draghia-Akli. Instead, the catch-all term will be replaced by "a large number of rare diseases, each of which causes malignant growth in breast tissue and requires individual treatment", she says.

The commission will launch a €100-million (US$140-million) call for research proposals in July, which will support the consortium's scientific goals by focusing heavily on developing appropriate clinical trials. ■



http://ec.europa.eu/research/health/medical-research/rare-diseases/irdirc_en.html

Nature **472**, 17 (2011)

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International Rare Diseases Research Consortium (IRDiRC)

In the Spotlight

Members of IRDiRC Scientific Committes appointed, 29/02/2012.



Read News Alert: Top experts guide international effort in rare diseases research. .

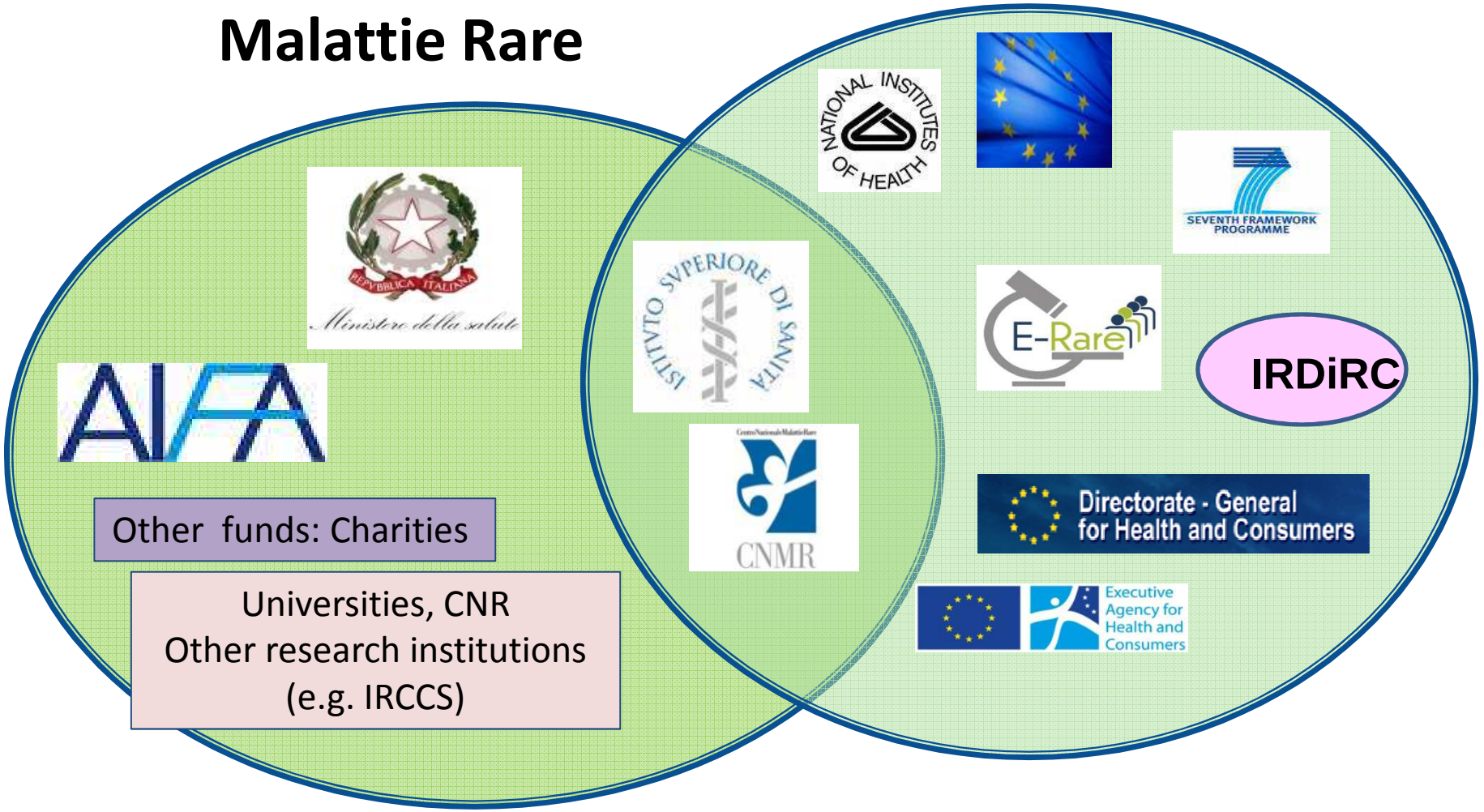
Introduction

Maximising scarce resources and coordinating research efforts are key elements for success in the rare diseases field. Worldwide sharing of information, data and samples to boost research is currently hampered by the absence of an exhaustive rare disease classification, standard terms of reference and common ontologies, as well as harmonised regulatory requirements.

The International Rare Diseases Research Consortium (IRDiRC) **was launched in April 2011** to foster international collaboration in rare diseases research. IRDiRC will team up researchers and organisations investing in rare diseases research in order to achieve two main objectives, namely to deliver **200 new therapies for rare diseases and means to diagnose most rare diseases by the year 2020.**

For any questions or queries with regard to IRDiRC, please use the following e-mail address: RTD-IRDIRC [at] ec.europa.eu

Ricerca scientifica
e
Malattie Rare





BURQOL RD

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

BURQOL-RD is a 3 year project under the 2nd Programme of Community Action in the Field of Public Health, that commenced in April 2010 and is promoted by the DG Sanco.

The main aim of BURQOL-RD is to generate a model to quantify the socio-economic costs and Health Related Quality of Life (HRQOL) , of both patients and caregivers, for up to 10 rare diseases in different European countries.

This model will be adaptable and sufficiently sensitive to capture the differences in the distinct Health and Social Care Systems in the EU Member States. Our hope is that, the information generated by the BURQOL-RD consortium will help to:

- Design future policies in the area of rare diseases, which will ultimately have positive benefits for EU citizens health, both that of patients and of their caregivers. Readily transfer the protocols established to other RD and to other countries.
  Compare the availability and access to specific health resources for specific RD in each country.
- Explore the potential relationships between HRQL and access to healthcare resources.

After a thorough selection process, a set of 10 rare diseases to be targeted was decided:

- Cystic fibrosis
- Prader-Willi Syndrome
- Haemophilia
- Duchenne Muscular Dystrophy
- Epidermolysis Bullosa
- Fragile X Syndrome
- Scleroderma
- Mucopolysaccharidosis
- Juvenile idiopathic arthritis
- Histiocytosis

http://burqol-rd.com/

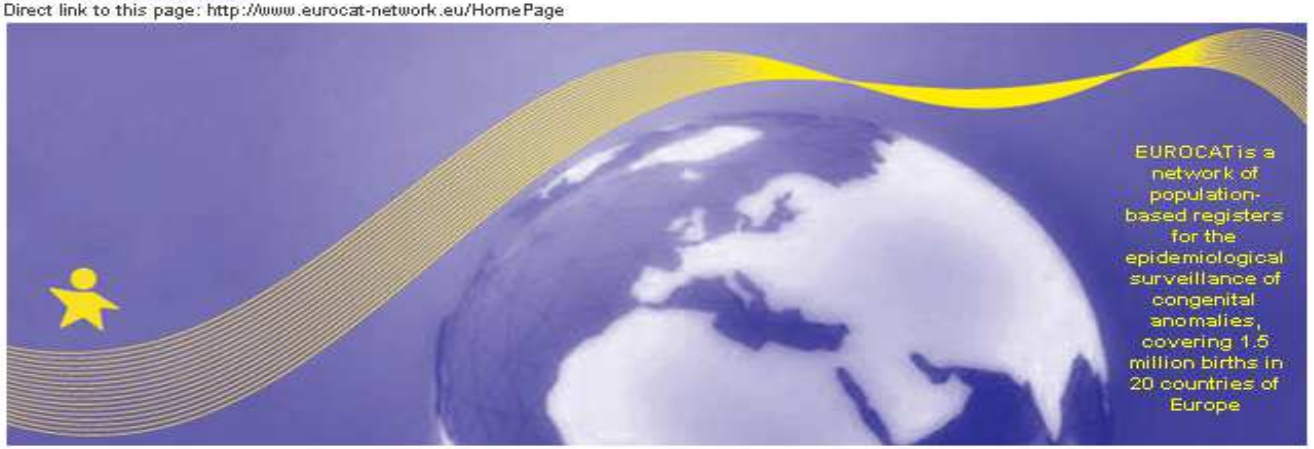
eurocat
european surveillance of congenital anomalies

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  - EU Rare Diseases Policy
  - Joint Action EUROCAT
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- ACCESS PREVALENCE DATA
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  - Interpretation Guide
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Where Am I? -> Home Page
Direct link to this page: http://www.eurocat-network.eu/HomePage



Announcements [Archive Announcements]

EUROCAT as a WHO Collaborating Centre
The Centre for Maternal, Fetal and Infant Research, School of Health Sciences, University of Ulster – home of EUROCAT's Central Registry has been re-designated on 19 May 2011 for a further period of 4 years as a WHO Collaborating Centre for the Surveillance of Congenital Anomalies. In this capacity EUROCAT will assist the WHO in implementing the resolution WHA63.17 of the 63rd World Health Assembly (2010) on birth defects at both a European and global level. EUROCAT will also assist the WHO in implementing its strategy for the prevention and control of non-communicable diseases (NCDs action plan 2008-2013).

"EUROCAT receives funding from the European Union, in the framework of the Public Health Programme"
"EUROCAT is a WHO Collaborating Centre for the Epidemiology Surveillance of Congenital Anomalies"
"EUROCAT Central Registry is based at the University of Ulster"

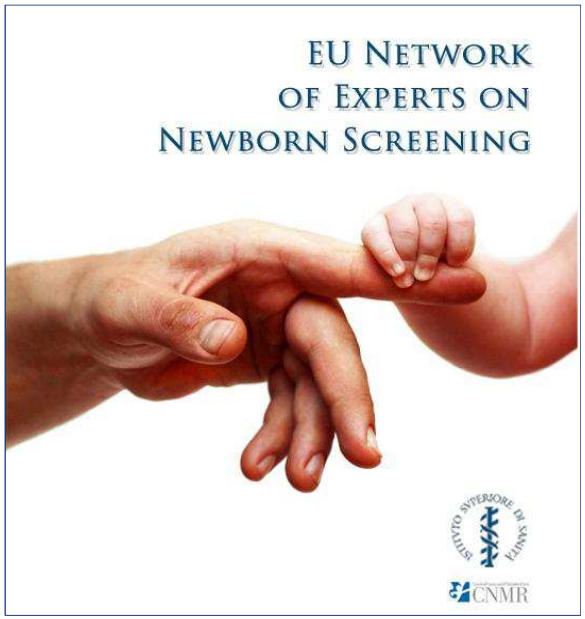
Website created and hosted by BioMedical

University of ULSTER | Executive Agency for Health and Consumers

www.eurocat-network.eu/

NEWBORN SCREENING

Evaluation of population newborn screening practices for rare disorders in Member States of the European Union



www.iss.it/cnmr

Tender funded by EU Commission (DG SANCO)

coordinated by the Italian Centre for rare Diseases

(Italian Public Health Institute, Rome), partners: all MS

df - Adobe Reader

Documento Strumenti Finestra ?

1 / 2 70,4% Trova



For more information visit the website
www.iss.it/cnmr



Course Director

Domenica Taruscio
Director
Italian National Centre for Rare Diseases
Italian National Institute of Health
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Email: cristina.morciano@iss.it
Tel. 0039 6 49904422

ISTITUTO SUPERIORE DI SANITÀ

First announcement

Summer School
Clinical practice guidelines on rare diseases

July 9-11, 2012

Italian National Institute of Health
Viale Regina Elena, 299
Rome (Italy)

Organised by the
Italian National Centre for Rare Diseases
Italian National Institute of Health

CentroNazionaleMalattieRare
CNMR

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IRDiRC

SEVENTH FRAMEWORK PROGRAMME

ICORD
International Conference on Rare Diseases & Orphan Drugs

CentroNazionaleMalattieRare
CNMR



E-Rare

EU NETWORK
OF EXPERTS ON
NEWBORN SCREENING



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



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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? Guida all'esenzione
Dal sospetto di malattia ai centri di diagnosi e cura

Il disabile e i suoi diritti
Una guida concreta 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 e centri di diagnosi e cura

Il sito per **cercare i centri di diagnosi e cura** e **conoscere la Rete Nazionale**

Associazioni di pazienti

CentroNazionaleMalattieRare
CNMR

In rilievo

8-9 ottobre 2012
International Workshop RARE DISEASE AND ORPHAN DRUG REGISTRIES.

5 novembre 2012
Convegno: "PREVENZIONE PRIMARIA DELLE MALFORMAZIONI CONGENITE"
CALL FOR ABSTRACT - Deadline **15 settembre 2012 -(prorogata al 1 ottobre 2012).**



ACIDO FOLICO E FOLATI PRIMA DI UNA GRAVIDANZA
On line il nuovo sito

10-11 settembre 2012
EUROPLAN 2012 - 2015 - INCEPTION WORKSHOP ON

Tematiche

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

Registri

Registro Nazionale Farmaci Orfani

Registro Nazionale Malattie Rare

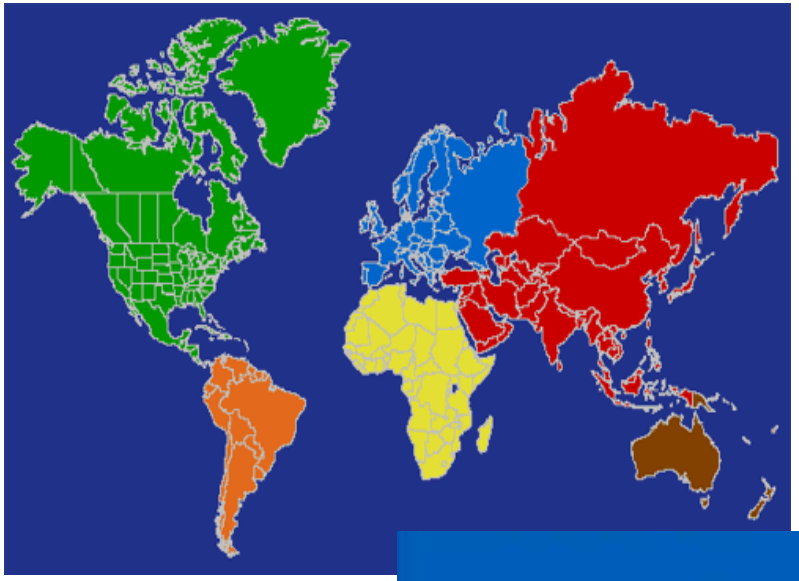
Registri Malformazioni Congenite

Il sito dedicato al concorso Il Volo di Pegaso



"Il Volo di Pègaso - 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



Grazie per l'attenzione





National Helpline on Rare Diseases and Orphan Drugs (since 2008)

Telefono Verde Malattie Rare

800.89.69.49



A multidisciplinary team (psychologists, sociologists and medical doctors) trained on telephone counselling

inform patients, health and social operators on:

- rare diseases and orphan drugs
- **centres for prevention, diagnosis**, **treatment and surveillance** (present at regional, national and international level)









Welcome to Rome

EUROPLAN

European Project for Rare Diseases National Plans Development

Coordinated by the Italian National Centre for Rare Diseases
Italian National Institute of Health

2012-2015

Considering Rare Diseases as a whole

public health issue helps highlighting and recognizing a series of healthcare problems and planning tailored pubblic health actions





EUROPLAN 2012-2015

a three-year project, embedded in the EUCERD Joint Action as Work Package 4, it is coordinated by the Italian National Institute of Health - Italian National Centre for Rare Diseases.

Its main goal is to establish an international and interactive network of stakeholders (mainly policy makers) to speed up the elaboration and the implementation of Rare Diseases National Plans/Strategies, trough scientific and technical assistance, Workshops and the active participation of patients Groups (EURORDIS and National Alliances). *[read more]*

News

RARE DISEASE AND ORPHAN DRUG REGISTRIES
International Workshop
8-9 October 2012
ISS - Rome, Italy
Website - Agenda

The Czech government adopted on August 28/2012 in its Decree 633
The Czech National Plan for Rare Diseases 2012-2014
[see the document]

It's available the DRAFT (english version) of the
Strategy of The Netherlands in the field of Rare Diseases
[see the document]



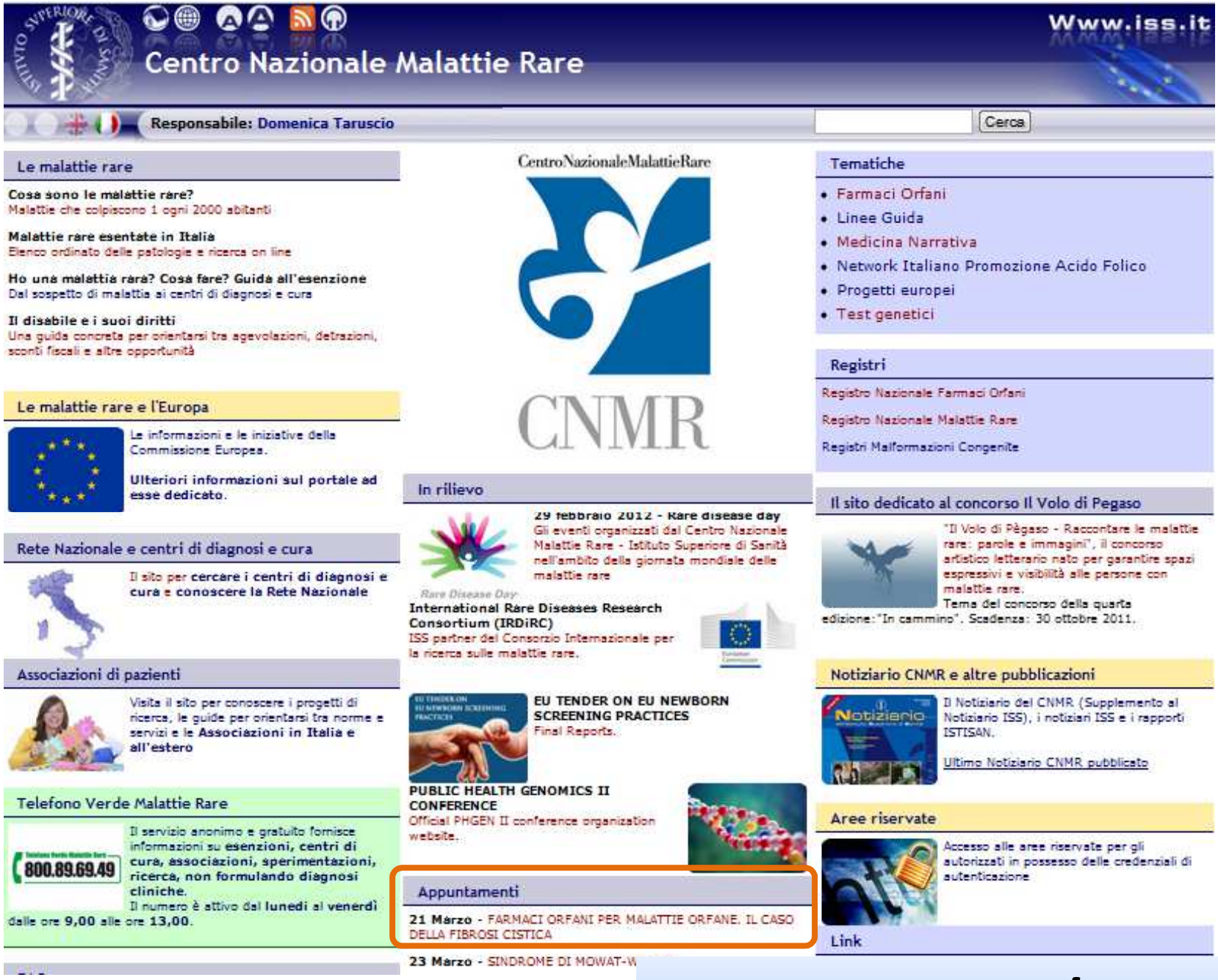
Download the EUROPLAN flyer

CentroNazionaleMalattieRare
CNMR

Direttore Dr.ssa Domenica TARUSCIO

"Il Centro svolge attività di **ricerca**, **sorveglianza**, **consulenza** e **documentazione** finalizzate alla prevenzione, diagnosi, trattamento, valutazione e controllo nel campo delle malattie rare e farmaci orfani".

Gazzetta Ufficiale N. 157 del 07.07.2008



www.iss.it/cnmr

ACCORDO BILATERALE ITALIA (ISS) – USA (NIH) PER LA RICERCA SULLE MALATTIE RARE (2002-2013)

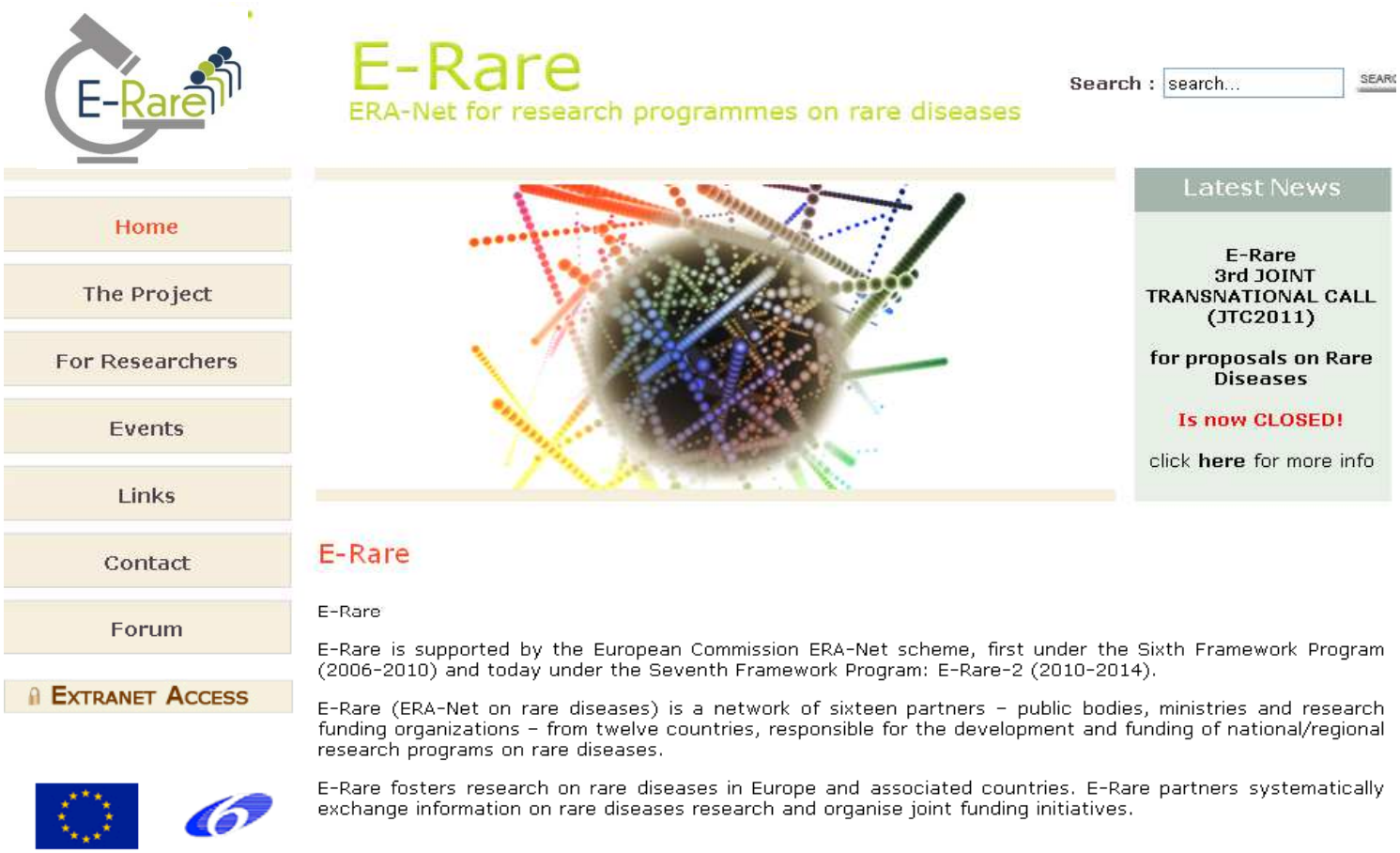
❖ 2 calls nazionali per progetti di ricerca:

1. 2005-7: **54** progetti finanziati (5 ML Euro)
2. 2007-11: **82** progetti finanziati (8 ML Euro)

RESULTATI: progetti principalmente focalizzati sulle seguenti aree tematiche

- prevenzione
- patogenesi
- sviluppo di nuovi strumenti diagnostici, prognostici (inclusa identificazione di nuovi biomarcatori) e terapeutici.
- Più di **200 publicazioni** su riviste peer reviewed (PubMed)

❖ Call nazionale per progetti scientifici: collaborazione Italia - USA (2011- 2013)



- Progetto co-finanziato dalla Commissione Europea (FP6; FP7)
- Network di **16 Partners** (public bodies, ministries and research funding organizations) da 12 Paesi (**Italia: Istituto Superiore di Sanità**)
- **Sviluppo e finanziamento** di programmi di ricerca scientifica a livello nazionale

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International Rare Diseases Research Consortium (IRDiRC)

In the Spotlight

Members of IRDiRC Scientific Committes appointed, 29/02/2012.



Read News Alert: Top experts guide international effort in rare diseases research. .

Introduction

Maximising scarce resources and coordinating research efforts are key elements for success in the rare diseases field. Worldwide sharing of information, data and samples to boost research is currently hampered by the absence of an exhaustive rare disease classification, standard terms of reference and common ontologies, as well as harmonised regulatory requirements.

The International Rare Diseases Research Consortium (IRDiRC) **was launched in April 2011** to foster international collaboration in rare diseases research. IRDiRC will team up researchers and organisations investing in rare diseases research in order to achieve two main objectives, namely to deliver **200 new therapies for rare diseases and means to diagnose most rare diseases by the year 2020.**

For any questions or queries with regard to IRDiRC, please use the following e-mail address: RTD-IRDIRC [at] ec.europa.eu

IN FOCUS NEWS

BIOMEDICAL RESEARCH

Rare-disease project has global ambitions

Consortium aims for hundreds of new therapies by 2020.

BY ALISON ABBOTT

Prader–Willi syndrome. Fabry renal disease. Spinocerebellar ataxia. Few people have heard of these and the other 'rare diseases', some of which affect only hundreds of patients worldwide. Drug companies searching for the next blockbuster pay them little attention. But the diseases are usually incurable — and there are thousands of them.

This week, the US National Institutes of Health (NIH) and the European Commission launch a joint assault on these conditions, whose small numbers of patients make it difficult to test new treatments and develop diagnostic methods. The International Rare Disease Research Consortium being formed under the auspices of the two bodies has the ambitious goal of developing a diagnostic tool for every known rare disease by 2020, along with new therapies to treat 200 of them. "The number of individuals with a particular rare disease is so small that we need to be able to pool information from patients in as many countries as possible," says Ruxandra Draghia-Akli, the commission's director of health research.

At the launch meeting in Bethesda, Maryland, on 6–8 April, prospective partners will map out research strategies to identify diagnostic biomarkers, design clinical trials and coordinate genome sequencing in these diseases. Nearly all the rare diseases, of which there are an estimated 6,000–8,000, are the result of small genetic changes.

The meeting will also discuss the governance of the project, which is most likely to be modelled on the pioneering Human Genome Project. As such, the consortium is open to research agencies and organizations from all over the world. Representatives from countries including Canada, Japan and some individual European nations are all attending the meeting, and may join the consortium. Those wishing to participate will have to pledge a minimum financial contribution, which has not yet been agreed, and share all relevant data. Indeed, the project will have to overcome numerous obstacles to information sharing, such as the fact that physicians in different countries often use entirely different words to describe the same disease.

Draghia-Akli points out that the project could yield major benefits for the emerging field of personalized medicine — another political priority for the NIH and the commission — which also faces the challenge of small populations of patients.

"We need to be able to pool information from patients from as many countries as possible."

Regulatory agencies such as the US Food and Drug Administration and the European Medicines Agency rely on large, randomized and controlled clinical trials when deciding whether to approve new medicines, and one of the aims of the consortium will be to develop alternative clinical-trial methods for diseases that affect few people.

These methods are becoming ever more important now that genome analysis is helping to break down common diseases into ever smaller subclasses. "Soon there will be no disease called breast cancer," says Draghia-Akli. Instead, the catch-all term will be replaced by "a large number of rare diseases, each of which causes malignant growth in breast tissue and requires individual treatment", she says.

The commission will launch a €100-million (US$140-million) call for research proposals in July, which will support the consortium's scientific goals by focusing heavily on developing appropriate clinical trials. ■

Nature **472**, 17 (2011)



International Rare Disease Research Consortium (IRDiRC)

Stabilire collaborazioni finalizzate a massimizzare le scarse risorse disponibili e coordinare le ricerche allo scopo di evitare dispersione di risorse e sforzi per raggiungere entro il 2020 la messa a punto di 200 nuove terapie per le malattie rare.

http://ec.europa.eu/research/health/medical-research/rare-diseases/irdirc_en.html

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Committed members

The European Commission (EC) and the US National Institutes of Health (NIH) initiated the discussions that led to the launch of IRDiRC. Since the launch, the interest to participate has been growing rapidly. For a current list of committed members. please see the list below

Country	Organisation
Australia	Western Australian Department of Health
Canada	Canadian Institutes for Health Research
Canada	Genome Canada
Italy	Italian National Institute of Health
Italy	Telethon Foundation
European Union	European Commission
France	French Association against Myopathies
France	French National Research Agency
France	Lysogene
Germany	Federal Ministry of Education and research
the Netherlands	The Netherlands Organisation for Health Research and Development

ICORD WEBSITE

ICORD
International Conference on Rare Diseases & Orphan Drugs



- **Welcome to ICORD!**
- Preamble
- Aims
- Conferences
- Society & Membership
- Board
- Secretariat
- Bank Information

Events

- 7th ICORD Tokyo
- Past Events

Welcome to ICORD!

International Conferences for Rare Diseases and Orphan Drugs

Welcome to the 7th ICORD meeting in Tokyo 4-6 February, 2012. Please go to the link in the left menu to find more information.

Due to the consequences of the earthquake in Japan early in 2011, the ICORD meeting planned to be held 21-23 May, 2011 in Tokyo was rescheduled to 4-6 Feb 2012.

The 6th international conference was held in March
ICORD 2010 in Buenos Aires.

Search

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Mission

The ICORD mission is to improve the welfare of patients with rare diseases and their families world-wide through better knowledge, research, care, information, education and awareness.

ICORD is an International Society for all individuals active in rare disease orphan drugs, including health care, research, academic, industry, patien organizations, regulatory authorities, health authorities, and public policy

http://icord.se/

http://icord.se





IRDiRC

SEVENTH FRAMEWORK PROGRAMME

ICORD
International Conference on Rare Diseases & Orphan Drugs

CentroNazionaleMalattieRare
CNMR







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



RARE DISEASES AND LIFE EXPECTANCY (Eurordis study)

323 rare diseases studied:

EURORDIS
Rare Diseases Europe

- **37.5%** of affected people have a normal life expectancy

- **25.7%** have a **disorder lethal at birth or within the first 5 yrs of life**

- **36.8% have a consistent reduction of life expectancy**, secondary to the severity of clinical manifestations and to the age of onset (i.e. infantile, juvenile, adult) of the disease

CRITICAL POINTS - RARE DISEASES

- **Diagnostic delay:** information / education medical doctors

  new diagnostic tools (e.g. genetic tests: validation, clinical utility, ..
- Needs for new orphan treatments and drugs
- clinical research: small numbers of patients
- Difficulties in coding systems of diseases (WHO-TAG rare diseases)
- Still poor understanding of the pathogenesis of many rare diseases

Several initiatives :
EU level, national and regional
International level

RARE DISEASES IN EUROPE: Programs of Community action



EU Reg. 141/2000 Orphan Medical Products (incentives, Scientific advices, Market exclusivity, etc.)

Decision No 1786/2002/EC of the European Parliament and of the Council of 23 September 2002 adopting a Programme of Community action in the field of public health (2003-2008)

General objectives of the programme

(a) **to improve information and knowledge for the development of public health;**

(b) to enhance the capability of responding rapidly and in a coordinated fashion to threats to health;

(c) to promote health and prevent disease through addressing health determinants across all policies and activities.

Decision No 1350/2007/EC of the European Parliament and of the Council of 23 October 2007 establishing a second Programme of Community action in the field of health (2008-13)

Objectives:

- **to improve citizens' health security**
- **to promote health, including the reduction of health inequalities**
- **to generate and disseminate health information and knowledge**

EUCERD

European Union Committee of Experts on Rare Diseases

The European Union Committee of Experts on Rare Diseases was formally established via the European Commission Decision of 30 November 2009 (2009/872/EC)

The European Union Committee of Experts on Rare Diseases replaces the European Commission's Rare Diseases Task Force (RDTF) established via Commission Decision 2004/192/EC of 25 February 2004 on the programme of Community action in the field of public health (2003 to 2008).

RARE DISEASES IN EUROPE: EUROPE'S CHALLENGES



November, 11 2008

Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases - Europe's challenges

OBJECTIVIES:

- **Improving recognition and visibility of rare diseases**
- **Supporting policies on rare diseases in the Member States**
- **Developing European cooperation, coordination, and regulation for rare diseases**


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
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The EPIRARE Survey
Deliverables & meetings
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In Europe a disease is considered rare when it affects no more than 5 individuals among 10,000 persons
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Public Health Genomics II
Three Conferences
April 17-20, 2012
Radisson Blu, Rome
The Epirare Project
The interest of researchers in the establishment of disease registries, is shown by the increasing number of RD registers in EU Member States as national or local initiatives mainly located at universities (Orphanet, 2009). Applications for funding RD networks, promoting data sharing and collaboration among researchers, are also submitted regularly to the European Commission.
The need for shared quality data collections for different health and healthcare purposes and the awareness of difficulties posed by their maintenance is witnessed by a number of recent documents (EPPOSI, 2009; Pharmaceutical Forum Reports, 2008).
The recent adoption of the Council Recommendation on RDs, which recommends the development of registers and databases for epidemiological purposes, is expected to result in a burst of initiatives for RD registration.
While a wide population base is especially needed for epidemiological and clinical research in RDs, the Regulation on
Second EPIRARE Meeting
23 May 2012
Brussels
Would you like to join Epirare?
Download Epirare flyer
Rome first Epirare meeting
11 - 12 July 2011
ISS
The meeting was held on July, 11-12 2011 in Rome (Italy) - Sanità – Viale Regina Elena, 299
Agenda
Participants
Accomodations
Proceedings
Meeting Secretariat
How to get to the meeting site
www.epirare.eu
Copyright 2011 Epirare. All right reserved.
EPIRARE
European Platform for Rare Disease Registries
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> Specific objectives
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The Epirare project
EPIRARE (European Platform for Rare Disease Registries) is a three-year project co-
within the EU Program of Community Action in the field of Public Health.
EPIRARE started officially on April 15, 2011.







Project

- Overview of the Project
- Partners
- Deliverables and Meetings
- Database including MS official documents
- Latest Documents
- Contact us
- Links
- Access restricted to partners
- National Conferences
- Final Conference

DOWNLOAD EUROPLAN BROCHURE

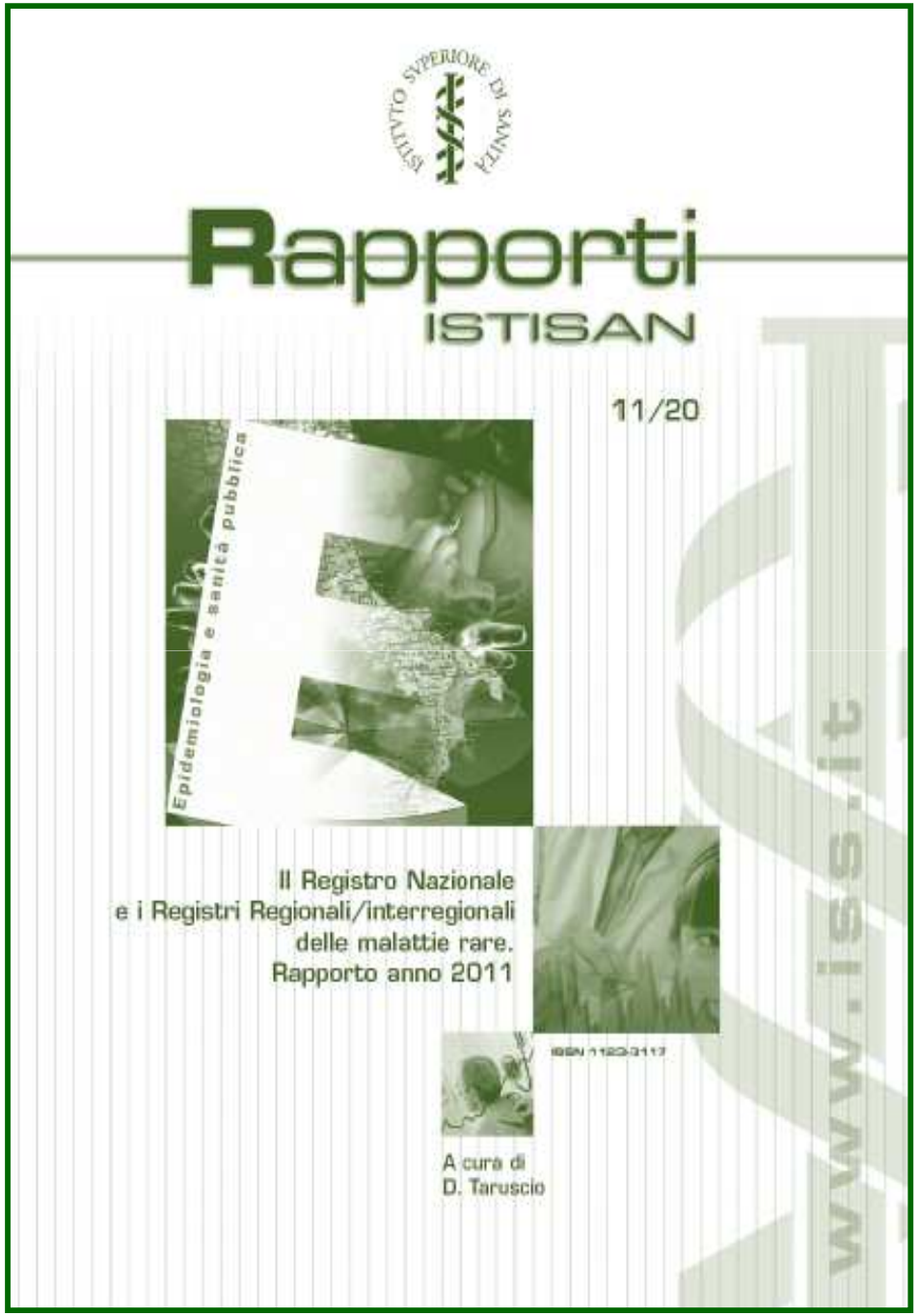
The European Project for Rare Diseases National Plans Development (EUROPLAN) is a three-year project of the Programme of Community action in the field of Public Health (2003 - 2008), which began in April 2008.

The main goal is to provide National Health Authorities with a supporting tools for the development and implementation of National Plans and Strategies for rare diseases (RDs) following the recently agreed European Council Recommendation on an action in the field of RDs (2009/C 151/02). This supporting tools will be composed of three documents focused on defined priority areas: the Guidance document on recommendations for the definition and implementation of National Plans and Strategies for rare diseases; the report on current practices and relevant cases in the field of rare diseases; and the document on the recommended set of indicators for monitoring and evaluating the implementation of national initiatives.

The National Centre for Rare Diseases (Italian Institute of Health - Istituto Superiore di Sanità, Italy) is the leading partner that organize the contributions from 34 countries and Eurordis (the European Organisation for rare diseases) ensuring a broad representation of different EU contexts and experiences and patients' point of view. In addition, the project ensures an inclusive and wide engagement of stakeholders - Ministries, regional and local authorities, health care planners, programme managers, health care professionals, researchers and patients.

www.europlanproject.eu

ITALIAN NATIONAL REGISTRY FOR RARE DISEASES



Italian National Registry of Rare Diseases:
Ministerial Decree 279/2001

- Planning and evaluating health care programmes (services utilization, patients' mobility…)

- Diseases surveillance (estimation of prevalence and incidence, geographical and temporal distribution of RD)

Agreement between Central State and Regions: (2002, 2007)

AGREED DATA SET

TO BE SENT BY REGIONAL REGISTRIES TO THE NATIONAL REGISTRY AT THE ISS (EVERY 6 MONTHS)

- ❑ **IDENTIFICATION CODE OF PATIENT**
- ❑ **LIVE – DEADH (DATE)**
- ❑ **DIAGNOSIS OF RARE DISEASE**
- ❑ **REGION, HOSPITAL WHICH MADE THE DIAGNOSIS**
- ❑ **DATE OF THE DIAGNOSIS**
- ❑ **ORPHAN DRUG USED**

RD collected in NRRD (30/06/2007- 30/06/2010)

- **485 RD are collected in NRRD**

ICD Groups	Frequency	%
Diseases of the nervous system	19535	21,05
Diseases of the blood and blood-forming organs	19126	20,60
Endocrine, nutritional and metabolic diseases	17594	18,95
Congenital malformations, deformations and chromosomal abnormalities	13963	15,04
Diseases of the musculoskeletal system and connective tissue	8798	9,48
Neoplasms (Cod. ICD-9-CM da 239)	4082	4,40
Diseases of the circulatory system	3976	4,28
Diseases of the skin and subcutaneous tissue	3183	3,43
Diseases of the digestive system	1229	1,32
Certain infectious and parasitic diseases	558	0,60
Diseases of the genitourinary system	552	0,59
Certain conditions originating in the perinatal period	219	0,24
Symptoms, signs and abnormal clinical and laboratory findings, not elsewhere classified	9	0,01
Total	**92824**	**100,0**

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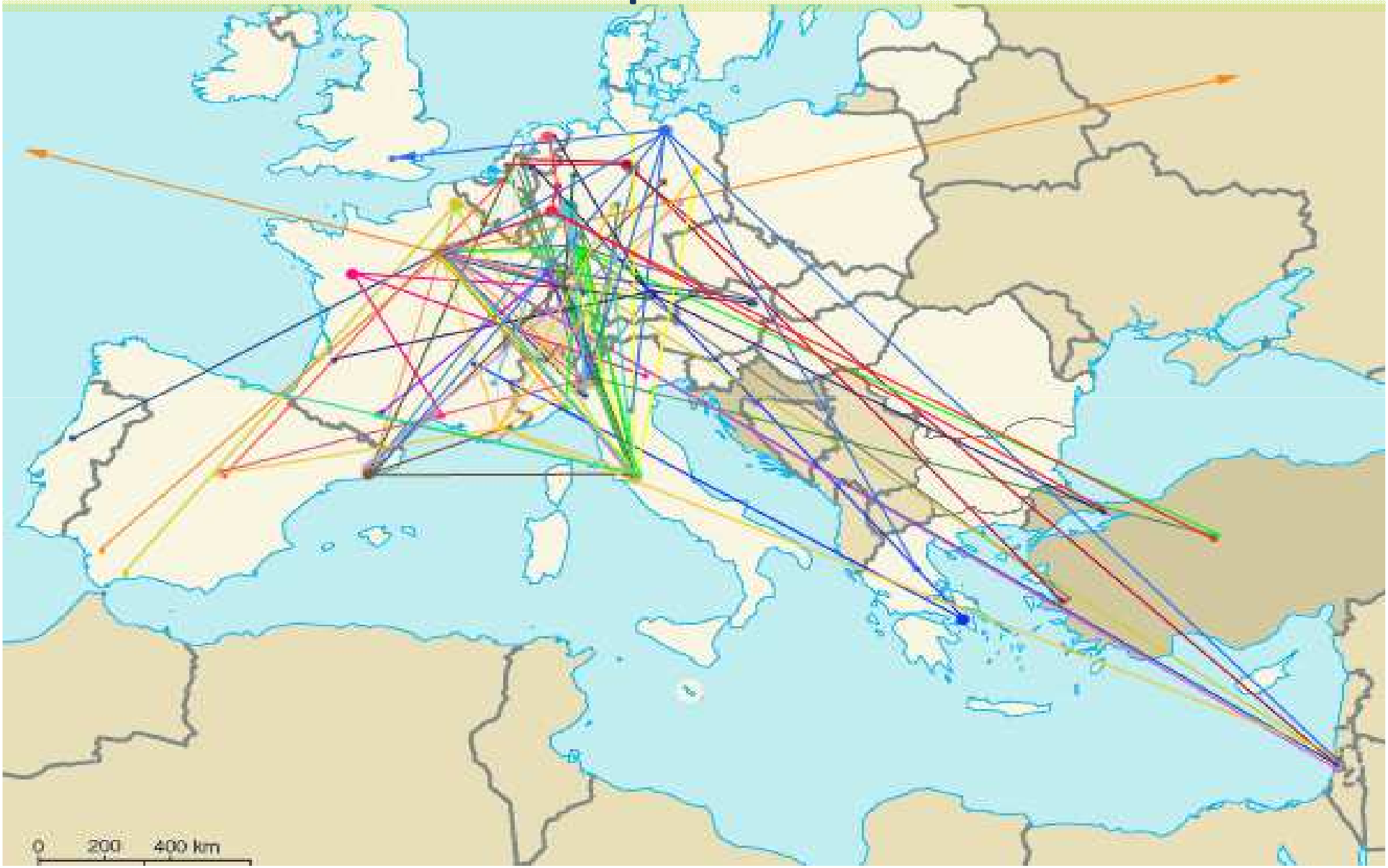


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E-RARE
creazione di una rete europea di collaborazioni scientifiche



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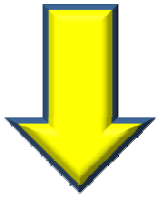
❖ Call nazionale per progetti scientifici: collaborazione Italia - USA (2011- 2013)

ICORD

International Conference on Rare Diseases & Orphan Drugs

ICORD is an International Society for all **individuals active in rare diseases and/or orphan drugs**, including health care, research, academic, industry, patient organizations, regulatory authorities, health authorities, and public policy professionals

MISSION



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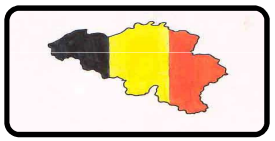
www.icord.se

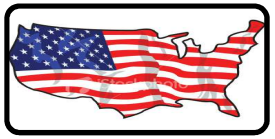
CONFERENCES

PAST EVENTS


ICORD 2005 in Stockholm


ICORD 2006 in Madrid


ICORD 2007 in Brussels


ICORD 2008 in Washington DC


ICORD 2009 in Rome


ICORD 2010 in Buenos Aires

ICORD
International Conference on Rare Diseases & Orphan Drugs

ICORD CONFERENCE 2012



ICORD 2012 in Tokyo, Japan

ICORD
International Conference on Rare Diseases & Orphan Drugs



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Tokyo 2011

Next conference will be held in Tokyo, Japan.

Please click the link for the first announcement: ICORD 2011 First announcement

VII International Conference on Rare Diseases and Orphan Drugs (ICORD 2011)

VENUE: Tokyo, Japan

Www.iss.it

Centro Nazionale Malattie Rare

Responsabile: Domenica Taruscio

Cerca

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? Guida all'esenzione
Dal sospetto di malattia ai centri di diagnosi e cura

Il disabile e i suoi diritti
Una guida concreta 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 e centri di diagnosi e cura

Il sito per **cercare i centri di diagnosi e cura** e **conoscere la Rete Nazionale**

CentroNazionaleMalattieRare

CNMR

In rilievo



Acido folico e folati prima di una gravidanza
On line il nuovo sito sull'Acido Folico a cura del CNMR

17-20 Aprile 2012
"From Population Health to Personal Health"
Triple Event Public Health Genomics II





9 -11 luglio 2012 - Summer School Clinical practice guidelines on rare diseases
Summer School Clinical practice guidelines on rare diseases

Tematiche

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

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Registro Nazionale Malattie Rare

Registri Malformazioni Congenite

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Tema del concorso della quarta edizione:"In cammino". Scadenza: 30 ottobre 2011.

WWW.ISS.IT/CNMR

National Centre for Rare Diseases

CNMR

Director: Domenica Taruscio

MISSION

Research, surveillance, training and information

on rare diseases and orphan drugs

aiming to prevent, diagnose, treat, cure and control RD

5 UNITS

- Genetic Tests
- Molecular Markers Biological Models
- Prevention Surveillance Education Training Information
- Orphan Drugs
- European / International Networks

www.iss.it/cnmr

Official Bulletin, N°157 del 07.07.2008

NATIONAL REGISTRY
RARE DISEASES
AND ORPHAN DRUGS

NATIONAL
NETWORK

ORPHAN DRUGS

AIFA

EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

FOLIC ACID
ITALIAN NETWORK

efsa
European Food Safety Authority

PATIENT'S
ASSOCIATIONS



RESEARCH



ISTITUTO SUPERIORE DI SANITÀ

NATIONAL INSTITUTES OF HEALTH

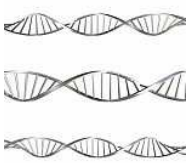
TRAINING and
INFORMATION



CNMR

activities

GENETIC TESTS



NARRATIVE MEDICINE



EUROPEAN
PROJECTS

EURO PLAN

E-Rare

European Union Committee of Experts
on Rare Diseases

GUIDELINES



WWW.ISS.IT/CNMR