

A close-up photograph of a young girl with dark brown hair and bangs. She is looking directly at the camera with a neutral expression. The background is dark, and there is a horizontal light streak across the bottom of the image.

Rare Diseases

**Three benefits
from international cooperation**


October 2014



Rare Diseases

Three benefits from international cooperation

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About Istituto Superiore di Sanità

The Istituto Superiore di Sanità is the leading technical and scientific body of the Italian National Health Service. Its activities include research, clinical trials, control and training in public health; it also serves as a major national clearing-house for technical and scientific information on public health issues.

The Institute is involved in collaboration and consultation with other institutions responsible for public health, including the Ministry of Health, regional health authorities, local health agencies and hospitals. It cooperates with those responsible for the design and implementation of health and scientific programmes at local and national level and also plays a leading role in several major international research projects.

The Institute provides scientific advice and assessments in the framework of international organizations, such as EFSA, EU, IARC, OECD, UNEP and WHO.

About National Centre for Rare Diseases of Istituto Superiore di Sanità

The National Centre for Rare Diseases (CNMR - www.iss.it/cnmr), directed by Dr. Domenica Taruscio, is the result of a strategic approach, which the Istituto Superiore di Sanità (ISS - Italian National Institute of Health) has been developing for over 10 years, to deal with the public health challenges associated with rare diseases.



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PREFACE

Rare diseases figure among the priorities in European healthcare not only because they are many and they affect millions of people and their whole families, but also for the challenge they raise in terms of establishing fair and sustainable healthcare and research networks.

In this sense Europe demands we chart a common course and during the semester of the Italian Presidency of the Council of the European Union, the National Centre for Rare Diseases at the Italian National Institute of Health, Italy (Istituto Superiore di Sanità) could not but deal with this issue, considering Italy's remarkable commitment in the coordination of many important European projects such as EUROPLAN aimed at developing and implementing national plans/strategies, EPIRARE, a European platform for rare disease registries, and RARE-Best-practices to provide updated and evidence-based clinical practice guidelines on rare diseases. I am especially pleased to speak about this important topic in this context, together with the Italian Parliament, where having set up an Inter-Parliamentary Group on rare diseases shows that the commitment of civil society as a whole is required.

Research on rare diseases often provides the opportunity to study pathogenic mechanisms that are shared with more common diseases. A fascinating challenge for science but also fruitful for the development of new therapies.

We cannot ignore the many patients who, although suffering from severe and disabling symptoms, have yet to receive a diagnosis for their diseases due to the current limits of medical knowledge.

In this context, the National Centre for Rare Diseases at ISS and the US National Institutes of Health (NIH) are working to identify new strategies and effective approaches to diagnosis and treatment by promoting the International Undiagnosed Diseases Network.

The complex clinical and care needs of patients with rare diseases drive us to search for integrated health care models but they are also the opportunity to bring medicine back to a global view of the patient, putting people at the centre and listening to their stories.

Rare disease patients ask for visibility and not to remain hidden behind unpronounceable names or syndromes that are absent in medical handbooks. They ask to be considered by researchers and health professionals in spite of an epidemiology often hard even to establish.

Their condition belongs to the history of medicine with full rights. A history where, time and again, the construction of scientific knowledge and that required to protect rare disease patients need to travel at the same speed.

Walter Ricciardi

Commissioner, Istituto Superiore di Sanità

FOREWORD

The European Community's role in the area of health under Article 152 of the Treaty is to encourage cooperation between the Member States and if necessary to lend support to their action.

The specificities of rare diseases - limited number of patients for each disease and scarcity of relevant knowledge and expertise - single them out as a unique domain of very high European added-value. They are key priorities in research and public health. Indeed, despite their rarity, there are so many different diseases that, overall, millions of people are affected in Europe. Although each rare disease has a low prevalence, the total number of affected EU citizens is estimated between 27 and 36 million. Many of them suffer from less frequently occurring diseases affecting one in 100,000 people or less; these patients are particularly isolated and vulnerable.

In these years, European countries have been involved in international projects and activities on rare diseases aiming to accelerate scientific research and attain the overarching goal of improving the health outcomes of rare disease care. We are proud that Italy has established the National Network for Rare Diseases since 2001 as well as Regional Networks in the following years. Moreover, the National Centre for Rare Diseases of the Istituto Superiore di Sanità has a main role in this field, coordinating key projects, such as EUROPLAN, EPIRARE, RARE-Bestpractices; of course, many other Italian stakeholders have important roles in developing initiatives including projects and Orphanet. An important and active role is played by Patient Associations.

A landmark in the EU and Member States activities is the **Council Recommendation of 8 June 2009 on an action in the field of rare diseases** (2009/C 151/02) [COM(2014) 548 final], which witnesses the commitment of national health authorities in developing effective actions, mainly through dedicated European cooperation, to improve rare disease care.

Indeed, European and international collaboration and cooperation are playing a key role in the future of rare disease patients. From the patients' right to access to healthcare to a more fair and equal world, the international cooperation in the rare diseases field offers a range of benefits, some well-known, others not so obvious.

The rare disease world is a paradigm of complexity. This booklet reflects the complex and dynamic nature of this field. It highlights some of the benefits of the international cooperation. That is one of the most important reasons for having collaborations in projects and activities: they are opportunities for countries to thrash out their differences on rare disease issues.

The principles and overarching values of universality, access to good quality care, equity and solidarity, as endorsed in the Council conclusions on common values and principles in EU health systems of June 2nd 2006, are of paramount importance for patients with rare diseases.

Domenica Taruscio

Director, National Centre for Rare Diseases, Istituto Superiore di Sanità

Message from the Inter-Parliamentary Group for Rare Diseases

The Inter-Parliamentary Group on rare diseases was established on 29 February 2012, on the occasion of the national Rare Disease Day, in order to draw constant attention to an issue anything but “rare”. There are many and very different rare diseases affecting no more than 5 in 10,000 people. Rare but numerous. The Intergroup is a necessary step in maintaining a virtuous interaction among the different political parties and between politics and healthcare. The aim is to keep up a relationship with scientific and patient associations in order to solicit the Government to take, as early as possible, effective decisions on Rare Diseases. In 2012 the Senate of the Republic approved a cross-party Order of the Day for the treatment of rare diseases and the Chamber of Deputies approved a unanimous motion that committed the Government to adopt tangible initiatives and programs for research in the field of rare diseases and their treatment. On both occasions, the Ministry of Health announced the intention “to establish a moment for coordination within the Cabinet”.

Our target is to turn good intentions into practice. Caring for people with rare diseases is an act of justice consistent with our Constitution that sets forth the universal right to health: nobody should be left alone. Raising attention towards rare diseases strongly encourages advanced scientific research. An Inter-Parliamentary Group on rare diseases means solidarity among everyone involved. This has led to the birth of Patient associations to support people with rare diseases and their families, to the creation of scholarships for young researchers and, above all, to the protection of patients’ rights.

It is a duty of politics to work out those legislative measures which can combine justice and solidarity, research and care, health and social policies. Since 2012 we have been working in

close contact with the National Centre for Rare Diseases of the Italian National Institute of Health, with UNIAMO FIMR Onlus and with the people involved from both associations and pharmaceutical industries; an important work which is yielding significant results.

Paola Binetti

Coordinator, Italian Inter-Parliamentary Group for Rare Diseases

Activities of Italian Inter-Parliamentary Group for Rare Diseases during the XVI and XVII Legislatures

The Inter-Parliamentary Group for Rare Diseases was instituted in February 2012 in order to set up a debate which would include scientific, clinical, economic and political aspects. One of its most important objectives is to achieve the approval of a bill concerning rare diseases, worked out and shared with all the patient associations that fight for the rights of people with rare diseases.

President of the Inter-Parliamentary Group: MP (Member of Parliament) Paola Binetti.

29 February 2012. Rare but numerous: an Inter-Parliamentary Group for Rare Diseases was instituted. A press conference was held in the press room of Palazzo Montecitorio to announce the new Intergroup.

Since then, the Intergroup has promoted several national and international meetings, press conferences and events in the Senate, Chamber of Deputies and other institutional locations. For instance, to highlight the patients' experiences and needs, the Intergroup organized the projection of the movie "The dark side of the Sun" in the Globe's Hall of Montecitorio.

Main Parliamentary initiatives

15 March 2013. President Binetti was the first signer of the changes to law n. 123 of 4 July 2005, in the matter of the diagnostic and follow-up tests for children aged 6-10 years ("Regulations for the protection of subjects suffering from Celiac Disease") and proposed a motion (1/00094) in the same field in June 2013.

15 March 2013. The Intergroup (MP Paola Binetti first proponent) introduced a bill on mandatory newborn screening for inherited metabolic disorders.

June 2013. The Intergroup introduced a bill (n. 1,168) before the House "Regulation on rare disease research, prevention and treatment and institution of a national Agency for rare diseases".

8 July 2014. The Intergroup proposed a Regulation on rare cancers and their prevention and treatment (Paola Binetti first proponent).

INTRODUCTION

Rare diseases are life-threatening or chronically debilitating diseases with a low prevalence and a high level of complexity. The European Union considers diseases to be rare when they affect not more than 5 in 10,000 people in the European Union. Due to their high number - between 5,000 and 8,000 different diseases - this nevertheless means that rare diseases affect an estimated 29 million people in the European Union.

Due to the high number and variety of diseases, research on individual rare diseases is scarce; it is also scattered in different laboratories throughout the EU.

The scarcity of expertise translates into delayed diagnosis and difficult access to care. This results in additional physical, psychological and intellectual impairments, inadequate or even harmful treatments and loss of confidence in the health care system, despite the fact that some rare diseases are compatible with a normal life if diagnosed on time and properly managed. Misdiagnosis and non-diagnosis are the main obstacles to improving life quality for thousands of rare disease patients.

Because of their low prevalence, their specificity and the high total number of people affected, rare diseases call for a **global approach** based on special and combined efforts to prevent significant morbidity or avoidable premature mortality, and to improve the quality of life and socioeconomic potential of affected persons.

The specificities of rare diseases - limited number of patients and scarcity of relevant knowledge and expertise - single them out as a distinctive domain where European cooperation is of very high added-value. Indeed, national health policies dedicated to rare diseases, sup-

ported by European cooperation, can help to ensure that scarce knowledge and resources can be shared and combined as efficiently as possible, in order to tackle rare diseases effectively across the EU as a whole.

The expected **3 benefits of cooperation and global approach** are:

1. Accelerated knowledge of rare diseases
2. Improved diagnosis and care for rare disease patients
3. Higher equity for rare disease patients.

Section 1.

Accelerated knowledge of rare diseases

Picture: Antonino Sandullo, Pègaso 2013
(www.iss.it/binary/publ/cont/ONLINEpegaso.pdf)



DEFINITION

The key to improving overall strategies for rare diseases is to ensure that they are recognised, so that all the other linked actions can follow appropriately. An appropriate **identification** needs to be accompanied by accurate information, provided and disseminated in formats adapted to the needs of professionals and affected persons. International coding and classification systems are the basis for collection and sharing information for research and care.

REGISTRIES AND DATABASES

Registries and databases constitute key instruments to increase knowledge on rare diseases and develop clinical research. They are the only way to pool data in order to achieve a sufficient sample size for epidemiological and/or clinical research.

Collaborative efforts to establish and maintain data collections, which are open and accessible, should be considered.

RESEARCH

For most rare diseases that would potentially be treatable, there is simply no current specific treatment. The specificities of rare diseases lead to think that international cooperation in the **research** field is necessary. The fragmentation of resources and knowledge among different priorities and groups, as well as the lack of treatment for the majority of them, call for a coordinated European approach to unravel the underlying molecular basis and pathophysiological mechanisms.

RELEVANT EU PROJECTS

ORPHANET

Orphanet is a relational database available in 7 languages. It aims to link together information on over 6,000 diseases and allows for multiple queries. Each country has also its own entry page in its national language.

www.orpha.net

EPIRARE (European Platform for Rare Disease Registries)

The project is a feasibility study of a Platform for RD registries, addressing regulatory, ethical and technical issues associated with the registration of RD patients. The feasibility of a minimum data set common to all RDs designed to inform policy-makers is also being assessed by the project.

www.epirare.eu

RD-Connect

The project aims at developing robust mechanisms and standards for linking detailed clinical information with genetic information, biomaterial availability and research/trial datasets, in particular those generated by the omics research projects EURenOmics and Neuromics.

www.rd-connect.eu

E-RARE 2 (ERA-Net for Research Programmes on Rare Diseases)

The project aims at deepening and extending the cooperation among the E-Rare-1 and the four new partners by systematic exchange of information, yearly launching of joint calls, thorough assessment of the funding mechanisms and results of the funded research projects and, finally, strategic activities aiming at a sustainable development and extension of the network.

www.e-rare.eu

IRDirc (International Rare Diseases Research Consortium)

The project has two main objectives: to deliver 200 new therapies for rare diseases and means to diagnose most rare diseases by the year 2020. This collaboration will also require to harmonise the policies related to research utilisation, standardisation and dissemination. Each organisation will use its own funding mechanism to support rare diseases research.

www.irdirc.org

MEMBER STATES WITH A NATIONAL RARE DISEASE REGISTRY ALREADY ESTABLISHED

- Italy (since 2001)
- France (2005-08)
- Spain (since 2008)
- Belgium (since 2013)

Source: Taruscio D, Vittozzi L, Choquet R, *et al.* National Registries of Rare Diseases in Europe: An Overview of the Current Situation and Experiences. *Public Health Genomics* 2014.

WHAT HAPPENS IN ITALY...

Once a year, since 2013, the National Centre for Rare Diseases of the Italian National Institute of Health organises the **International Summer School on Rare Disease and Orphan Drug Registries**.

The Course takes participants through the main concepts and practical steps that must be undertaken in the establishment and management of a RD registry. The programme builds on the challenges raised by the evolving technological innovations, new data collection and sharing possibilities, legal requirements and security needs.

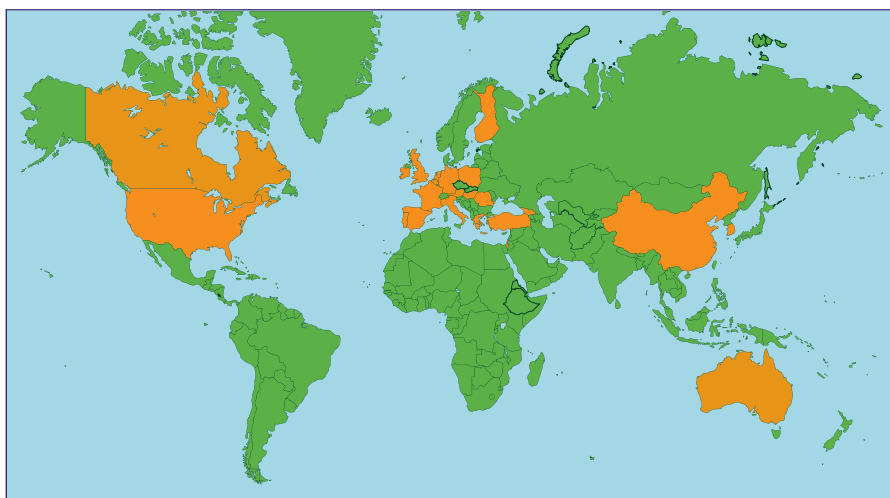
The Course adopts an interactive didactic methodology and is open to researchers, medical specialists, experts in statistics, health authorities, academia, medical students and patient organizations, who are involved or intend to establish a rare disease patient registry.

More information: rareregistries-school@iss.it

COUNTRIES WITH FUNDING AGENCIES COMMITTED TO THE IRDiRC

(www.irdirc.org)

IRDiRC Members are funding bodies/organisations investing a minimum of \$10 million US over 5 years in research projects/programmes contributing towards IRDiRC objectives, and invited patient advocacy groups. Participants include the European Commission and E-Rare 2 Consortium.

**Relevant points from the COUNCIL RECOMMENDATION (2009/C 151/02):**

- Adequate Definition, Codification and Inventorying of Rare Diseases
- Research on rare diseases.

Section 2.

**Improved diagnosis
and care for rare disease patients**

Picture: Vera Puoti, from the videotale "Con gli occhi tuoi"
(www.congliocchitui.salute.gov.it/portale/congliocchitui/homeTrailer.html)



CENTRES OF EXPERTISE AND EUROPEAN REFERENCE NETWORKS

It is necessary to facilitate improvements in access to diagnosis, treatment and provision of high-quality, accessible and cost-effective healthcare for rare disease patients. They have medical conditions requiring a particular concentration of expertise and/or resources.

Due to their features, rare diseases have been selected for the first implementation of the Directive 2011/24/EU on the application of patients' rights in cross-border healthcare, setting the rules for access to safe and good quality treatment across EU borders and for reimbursement. This directive also defines the rules for the establishment of Centres of Expertise and European Reference Networks, paving the way to improved diagnosis and care of rare disease patients through cooperation among the most specialised centres in the EU.

GATHERING THE EXPERTISE ON RARE DISEASES AT EUROPEAN LEVEL

For an effective and equitable care of rare disease patients, it is essential to gather national and international expertise on rare diseases and support its pooling. Rare disease patients may require different types of treatment (medical, pharmacological, psychological, rehabilitation, etc.) and an integrated approach to care is often necessary.

The majority of Member States support the pooling of expertise with European counterparts in order to foster the sharing of best practices on diagnostic tools and medical care as well as education and social care in the field of rare diseases. Several of them have organized education and training for health professionals to make them aware about existing sources available.

RELEVANT EU PROJECTS

EJA (EUCERD Joint Action: Working for Rare Diseases) - Work Package 8

The project is mandated to assist the EC in formulating and implementing the Community's activities in the field of rare diseases, to foster exchanges of relevant experience, policies and practices between Member States and stakeholders.

WP8 aims at identifying actions which could improve access to higher-quality healthcare in rare diseases (RD), enhancing patients' quality of life.

www.eucerd.eu/?page_id=54

RARE Best Practices

The project aims to develop a sustainable networking platform which supports the collection of standardised and validated data and the efficient exchange of knowledge and reliable information on RD.

www.rarebestpractices.eu

STORK

The aim of the STORK project is to establish an European eID Interoperability Platform that will allow citizens to establish new e-relations across borders, just by presenting their national eID.

www.eid-stork.eu

epSOS

epSOS aimed at designing, building and evaluating a service infrastructure that demonstrates cross-border interoperability between electronic health record systems in Europe.

www.epsos.eu

S.T.o.Re. (Story Telling on Record)

The partnership project aims at designing an action-research regarding Medical Records integrated with Narrative Based Medicine (patient narration), starting from existing experiences and considering e-health solutions.

www.storeproject.eu



EUROPEAN REFERENCE NETWORKS

(http://ec.europa.eu/health/ern/policy/index_en.htm)

European Reference Networks bring together highly specialised health-care providers from different Member States. They help provide affordable, high-quality and cost-effective healthcare to patients with conditions requiring a particular concentration of resources or expertise.

Expected benefits to patients and healthcare systems are improvements in: services delivery; working systems; patient pathways; clinical tools; earlier adoption of scientific evidence.

WHAT HAPPENS IN ITALY...

Regulatory Framework for Rare Diseases

The Ministerial Decree 18 May 2001, n. 279 “Regulation for the institution of the National Network for the prevention, surveillance, diagnosis and therapy of rare diseases and the exemption from patients’ participation in the costs of the relevant healthcare”:

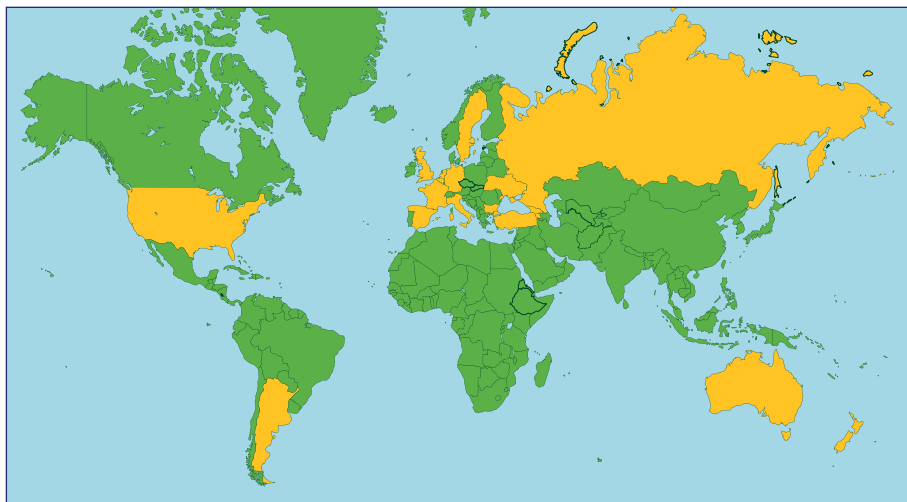
- establishes the National Network for Rare Diseases (www.iss.it/cnrmr, section “Centres in Italy”). Since 2001, the Regional authorities have designated centres for the diagnosis and treatment of RD patients and regional reference centres coordinating the care activities of the centres within and across the regions and keeping regional RD patient registries.
- establishes the National Registry of Rare Diseases (Istituto Superiore di Sanità, Rome) functionally connected with regional RD patient registries;
- regulates the exemption from patients’ participation to the costs for diagnosis and treatment of rare diseases, listed in the Annex of the decree (284 diseases and 47 rare diseases’ groups; 331 codes which include a larger number of rare diseases).

Source: www.salute.gov.it

COUNTRIES INVOLVED IN RARE BEST PRACTICES PROJECT

(www.rarebestpractices.eu)

Argentina, Armenia, Australia, Belgium, Bulgaria, France, Georgia, Germany, Italy, Netherlands, New Zealand, Russia, Spain, Sweden, Turkey, United Kingdom, Ukraine, USA.

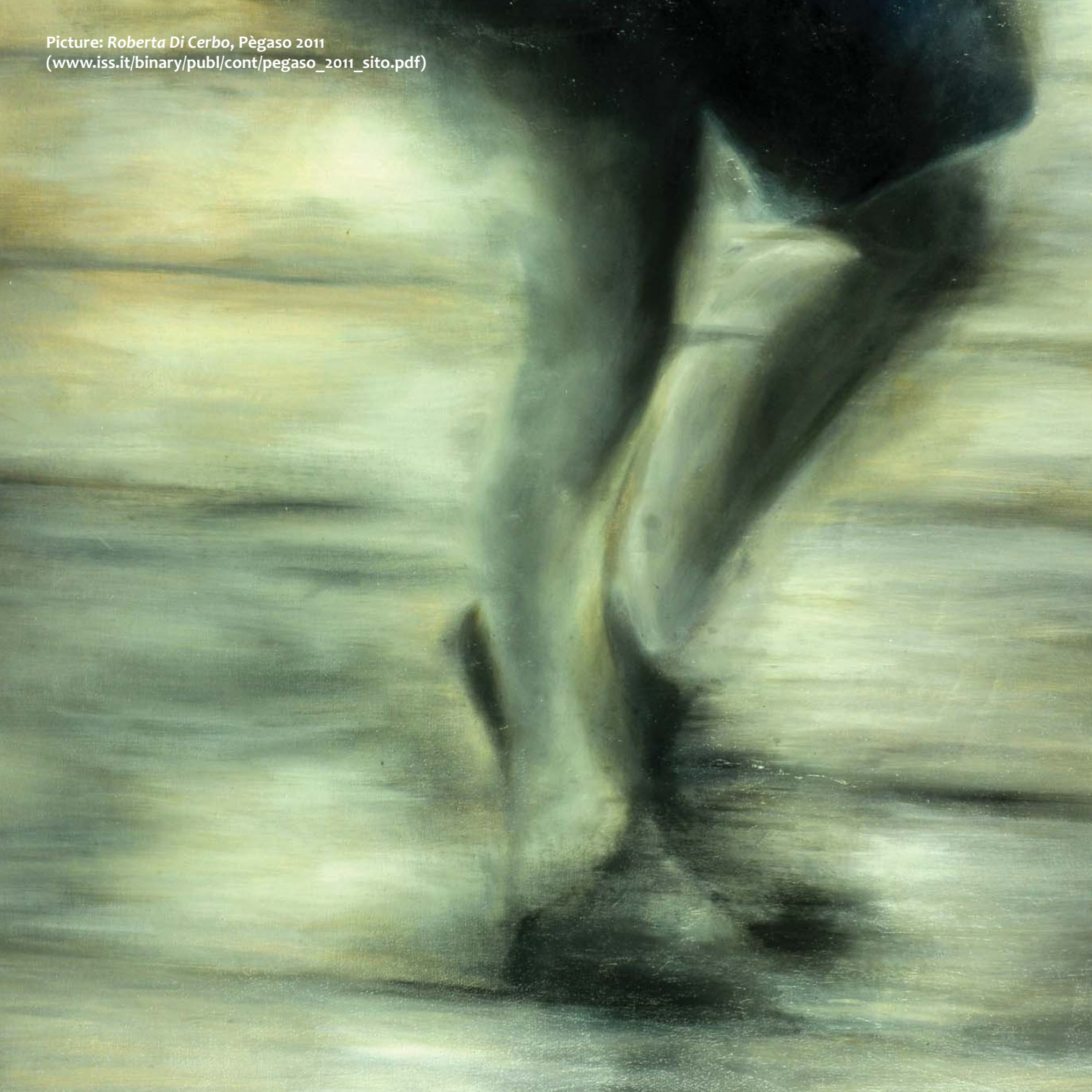
**Relevant points from the COUNCIL RECOMMENDATION (2009/C 151/02):**

- Centres of Expertise and European Reference Networks for rare diseases
- Gathering the expertise on rare diseases at European level.

Section 3.

**Higher equity
for rare disease patients**

Picture: *Roberta Di Cerbo, Pègaso 2011*
(www.iss.it/binary/publ/cont/pegaso_2011_sito.pdf)



NATIONAL PLANS OR STRATEGIES

Rare disease patients must have the same rights as all other Patients: projects and activities should be oriented towards this objective.

The EC recommended Member States to put in place National Plans/Strategies (NP/S) for rare diseases by the end of 2013. NP/S set out an inter-sectoral framework guiding and structuring relevant actions in the field of rare diseases within national health and social systems. All EU Member States are currently preparing their plans or strategies.

SUSTAINABILITY

NP/S for rare diseases are the common denominator of modern day rare disease public health policies. They harmonise the European common objective to ensure equal access and availability of prevention, diagnosis, treatment and rehabilitation for people with rare diseases and the National Authorities right to choose which specific measures to approve and implement. Member States should ensure, through appropriate funding and cooperation mechanisms, the long-term sustainability of infrastructures developed in the field of information, research and healthcare for rare diseases.

EMPOWERMENT OF PATIENTS' ORGANISATIONS

Empowerment of patients can be considered a **pre-requisite for health**. The participation of patient organisations in all aspects of the development of rare diseases policies is very important to identify patients' needs and implement effective actions. Patient empowerment can be encouraged and supported in various ways, i.e. through specialised social services, which are instrumental to the empowerment of people living with rare diseases and are essential to the

improvement of their well-being and health, and **dedicated helplines**, which can facilitate patient empowerment by enabling patients to self care and to access health advice and services; they are also seen to offer the prerequisites for empowerment perceived to be lacking in the wider national health systems, including time, respect, listening, support, and information.

RELEVANT EU PROJECTS

EUROPLAN (European Project for Rare Diseases National Plans Development)

Since 2008, EUROPLAN has contributed to the implementation of national plans and strategies to tackle rare diseases throughout the EU, to share relevant experiences within Countries, linking national efforts with a common strategy at European level. In particular, the project has elaborated Recommendations for designing National Plans or Strategies for rare diseases and Indicators for monitoring their implementation and evaluating their impact. The cooperation with several stakeholders (Ministry of Health representatives, clinicians, researchers, patient organisations) improves effectiveness. Since 2012, EUROPLAN is embedded in EJA as WP4. www.europlanproject.eu

EU TENDER ON EU NEWBORN SCREENING PRACTICES (Evaluation of population newborn screening practices for rare disorders in Member States of the European Union)

The project indicated the actions to be carried out to increase the equity and to improve the practice of the neonatal screening process for rare diseases in the EU Member States.

www.iss.it/cnmr/prog/cont.php?id=2262&lang=1&tipo=64

EJA (EUCERD Joint Action: Working for Rare Diseases) - Work Package 6

WP6 aims at mapping the social services available in Europe and promoting awareness-raising activities for the purpose of highlighting the need for these services. It will also address the issues concerning the training of staff/volunteers working in these services. www.eucerd.eu/?page_id=54

BURQOL-RD (Social economic BURden and health-related Quality of Life in patients with Rare Diseases in Europe)

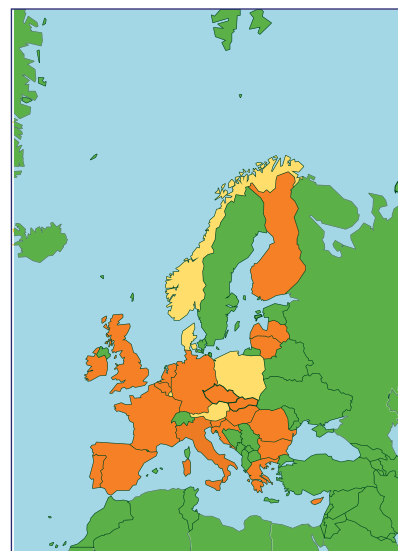
The main aim of BURQOL-RD was to generate a model to quantify the socio-economic costs and HRQOL, of both patients and caregivers, for ten RDs in eight different European countries. www.burqol-rd.com

By October 2014:

- 21 Member States with adopted rare disease National Plans/Strategies: Belgium, Bulgaria, Croatia, Cyprus, Czech Republic, Finland, France, Germany, Greece, Hungary, Ireland, Italy, Latvia, Lithuania, Netherlands, Portugal, Romania, Slovak Republic, Slovenia, Spain, United Kingdom.
- 5 Countries in advanced stage of preparation of their rare disease National Plans/Strategies: 4 Member States (Austria, Denmark, Luxembourg, Poland) and Norway.

Source: www.euoplanproject.eu

Source: European Commission. Report from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions. *Implementation report on the Commission Communication on Rare Diseases: Europe's challenges [COM (2008) 679 final] and Council Recommendation of 8 June 2009 on an action in the field of rare diseases (2009/C 151/02)*. Brussels, 5 September 2014 COM (2014) 548 final.



WHAT HAPPENS IN ITALY...

Italian National Plan for Rare Diseases: highlights

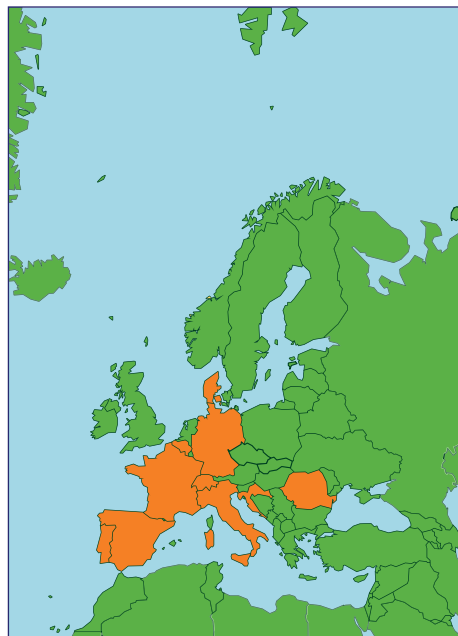
- Areas of intervention with specific actions for a 3-year period.
- Focus on:
 - general organisation and networking of Centres of expertise and coordination of regional activities;
 - early diagnosis and healthcare;
 - treatments and tools for therapeutic innovation;
 - role of Patient Associations;
 - education and training of health professionals;
 - information for health professionals, patients and their families.

Source: www.salute.gov.it

COUNTRIES PARTICIPATING IN THE EUROPEAN NETWORK OF RARE DISEASE HELP LINES, COORDINATED BY EURORDIS

Belgium, Bulgaria, Croatia, Denmark, France, Italy,
Portugal, Romania, Spain, Switzerland.

Source: Houyez F, Sanchez de Vega R, Brignol TN, et al. A European Network of Email and Telephone Help Lines Providing Information and Support on Rare Diseases: Results From a 1-Month Activity Survey. *Interact J Med Res* 2014;3(2):2-16.



Relevant points from the COUNCIL RECOMMENDATION (2009/C 151/02):

- Plans or Strategies in the field of rare diseases
- Empowerment of patient organisations
- Sustainability.



Rare Disease Day

RARE DISEASE DAY

Rare Disease Day is an annual, awareness-raising event co-ordinated by EURORDIS at international level and by National Alliances and Patient Organisations at the national level.

EURORDIS (www.eurordis.org) is a non-governmental patient-driven alliance of patient organisations and individuals active in the field of rare diseases, dedicated to improving the quality of life of all people living with rare diseases in Europe.

The main objective of the Rare Disease Day is to raise awareness amongst the general public and decision-makers about rare diseases and their impact on patients' lives.

The campaign targets primarily the general public but it is also designed for patients and patient representatives, as well as politicians, public authorities, policy-makers, industry representatives, researchers, health professionals and anyone who has a genuine interest in rare diseases.

Since the Rare Disease Day was first launched by EURORDIS and its Council of National Alliances in 2008, more than 1000 events have taken place throughout the world reaching hundreds of thousands of people and resulting in a great deal of media coverage.

The political momentum resulting from the Day has also served for advocacy purposes. It has notably contributed to the advancement of national plans and policies for rare diseases in a number of countries.

At the European level, support for the Rare Disease Day has been demonstrated in different ways, such as the 28 February 2011 release of the European Awareness of Rare Diseases Report, presenting the results of a Eurobarometer survey requested by the Directorate - General

for Health and Consumers (DG Sanco) and coordinated by the Directorate-General for Communication. The Directive 2011/24/EU on Patients' Rights in Cross-border Healthcare was also adopted on 28 February 2011. Even though the campaign started as a European event, it has progressively become a world event, with over 80 countries participating in 2014. Each year, EURORDIS organises a special event at the European level for the Rare Disease Day, inviting various EU level policymakers to participate.

EURORDIS objective is that the WHO recognizes the last day of February as the official Rare Disease Day and to increase awareness for Rare Diseases worldwide.



DIOCESE OF ROME, CENTRE FOR HEALTH PASTORAL CARE

(www.vicariatusurbis.org)

The Pastoral Healthcare Centre of the Vicariate of Rome actively promotes support, care, listening and social integration of patients with rare diseases. The commitment of the Centre contributed to establishing effective coordination aimed at creating a network of research and care institutions, educational organisations and patient associations. The network aims to meet the special social, health and spiritual needs deriving from rare pathologies (that are often invisible) and to make rare diseases a priority in the civil and religious society and in health policies.

On the occasion of the seventh Rare Disease Day in Italy, the Centre collaborated with the Italian National Institute of Health and with UNIAMO Fimr Onlus in order to create and promote the video-tale *Con gli occhi tuoi* (Through your eyes). The Roman Vicariate

patronised this original project in order to make primary school children aware of the importance of welcoming children with a rare disease and to encourage integration. On behalf of UNIAMO, the Pastoral Healthcare Centre of the Vicariate of Rome promoted the meeting with Pope Francis on the Rare Disease Day 2014. Thanks to the Centre's effort the Pope met over 400 Italian patients in preparation for the Event. During the audience, the Pope asked the institutions "to adequately support patients on their difficult path from both a medical and legislative point of view". By request of EURORDIS, the Centre is currently trying to extend the meeting with the Pope to include a European delegation of patients for the Rare Disease Day 2015.

PATIENT ASSOCIATIONS A GROWING REALITY IN THE WORLD!



EURORDIS (www.eurordis.org) is an international non-profit, non-governmental patient-driven alliance of rare disease patient organisations representing an estimated 30 million individuals in Europe.

Our mission

- To build a strong pan-European community of patient organisations and people living with rare diseases
- To be their voice at the European level
- To directly or indirectly fight against the impact of rare diseases on their lives.

Who we are

- 634 members patient organisations in 58 countries (26 EU countries)
- Council of 33 National Alliances of rare disease Patients Organisations (including Canada, USA & LA): Common Goals & Mutual Commitments, National Plans, Rare Disease Day
- Council of 44 European Federations of specific rare diseases and Networks: Cross border health care, accelerated procedure for multi-centred clinical trials for rare diseases, European Reference Networks of Centres of Expertise, Registries, access to medicines
- Organisation of annual Membership Meetings: Barcelona 2002, Namur 2003, Cork 2004, Venice 2005, Berlin 2006, Paris 2007, Copenhagen 2008, Athens 2009, Krakow 2010, Amsterdam 2011, Brussels 2012, Dubrovnik 2013, Berlin 2014, Madrid 2015
- Organisation of the European Conference on Rare Diseases: Copenhagen 2001, Paris 2003, Luxembourg 2005, Lisbon 2007, Krakow 2010, Brussels 2012, Berlin 2014, Edinburgh 2016
- Outreach to over 1800 patient groups
- Over 4,000 rare diseases represented
- 30 staff members, offices in Paris, Brussels, London, Barcelona
- 260 Volunteers: 55 volunteer patient advocates and 205 volunteer moderators of online RD patient communities.

Major advocacy achievements

- Contribution to EU drug regulations
 - Contribution to EU Directives/Regulations
 - Shaping EU Rare Disease Policy
-

- Involvement in EMA Committees: Committee for Orphan Medicinal Products, Pediatric Committee, Committee for Advanced Therapies, Patients' and Consumers' Working Party, Scientific Advice & Protocol Assistance, Committee for Human Medicinal Products
- Involvement in IRDiRC - International Rare Disease Research Consortium
- Involvement in Commission Expert Group on Rare Diseases (former EUCERD) with 8 patients' representatives
- Contribution to the 5 EUCERD Recommendations: Quality Criteria for Centres of Expertise for Rare Diseases in Member States, October 2011; Rare Disease European Reference Networks, January 2013; Clinical Added Value of Orphan Medicinal Products Information Flow, September 2012; Recommendations on Rare Disease Patient Registration and Data Collection, June 2013; Core Indicators for Rare Disease National Plans/Strategies, June 2013
- EURORDIS is partner in the EUCERD Joint Action "Working for Rare Diseases": WP8 Support for the implementation of Rare Disease Plans/Strategies and WP6 Provision of Specialized Social Services and Integration of RD into Social Policies and Services
- EURORDIS is playing an important role as catalysts to foster Research and Research Policies on Rare Diseases. Since 2000, it has advocated for rare diseases to be included in research and health programmes of the European Commission.



UNIAMO FIMR onlus (www.uniamo.org) is the Italian National Alliance of EURORDIS and is represented in its Board of Directors as well as in its Council of Alliances for the achievement of the same goals.

Its main objective is to improve the patients' quality of life through the promotion and the safeguard of their fundamental rights as to research, bioethics, welfare and health policies. It gathers more than 100 patient associations representing over 600 rare diseases. UNIAMO brings patients and their families' perspectives into the decisional process regarding national and regional interventions on the health care system. The Federation participates, as patients' representative, in the institutional tables dedicated to rare diseases in the regions of Lazio, Puglia, Marche, Piemonte and Campania.

The empowerment of patients on different topics, the training of doctors on rare diseases, in collaboration with all stakeholders involved in the field of rare diseases, the Ministry of Health, ISS, AIFA, AgeNaS, the scientific societies and associations of doctors, are some of the most important assets of UNIAMO's action.

Institutional activities are directed to achieve the Council Recommendations of 8 June 2009 (2009/C 151/02) on the development and implementation of national plans or strategies for rare diseases. UNIAMO FIMR with the support of EURORDIS, has organised the two Italian EUROPLAN Conferences in November 2010 and January 2014 to foster the debate on the Italian National Plan among the main national stakeholders.

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