

# Rare opportunity for cooperation

“ Coordination of scientific platforms and centres will open the way to networking and to the creation of a real European research programme on rare diseases. ”

Please note that this fact sheet replaces the previous one, known as E-RARE and included in Series 1 of this publication.

The field of rare diseases offers an outstanding opportunity for the coordination of national research programmes.

No single European country can assemble the critical mass of funding, expertise, facilities or patients needed to launch the large multidisciplinary projects needed, to make serious progress in combating this major social problem. E-Rare will establish internal and external communication infrastructures, and will develop specific communication with the scientific community, EU Commission, external funding organisations, all relevant public stakeholders and patient organisations. The results will be passed on to research policymakers with the aim of applying them to the healthcare system.

E-Rare aims at maximizing the efficiency and impact of research on rare diseases by developing a common research policy, as well as promoting actions to foster multidisciplinary projects, facilitate transnational access to research infrastructures, and gather patient cohorts and related biological material on a European scale

By definition, individual rare diseases affect no more than 1 person in every 2 000. But there are between 6 000 and 7 000 different types of diseases, mainly of genetic origin. Taken together, they affect some 20 million European citizens. Most of these diseases lead to chronic illness, and they represent a significant burden on healthcare systems. Yet for the vast majority, particularly the auto-immune disorders, the specific causes remain unknown. Very few are well-enough understood for effective treatment to be available.

Their rarity itself constitutes a real obstacle to research. The thin distribution of patients makes it hard to gather enough subjects for proper studies. Specialist research groups are isolated, and clinical data are fragmentary. Furthermore, since most public funding is devoted to research on common diseases, the complex research projects that are needed for rare diseases, and which normally require interdisciplinary cooperation, are seldom set up.

Recognising that special funding mechanisms and coordination are necessary to address these serious difficulties, 9 partners from 8 different countries decided to join their forces in this coordination action. E-Rare aims at maximizing the efficiency and impact of research on rare diseases, by developing a common research policy, as well as promoting actions to foster multidisciplinary projects, facilitate transnational access to research infrastructures and gather patient cohorts and related biological material, on a European scale.

## Developing common research policy

Mutual learning and exchange of information, identification of gaps and overlaps among national research programmes and activities on rare diseases, will create a knowledge base for the development of joint and transnational activities. Interaction with scientists and stakeholders involved in the study of rare diseases will allow for the definition of strategic priorities to be included in the research policy agenda on rare diseases, and also in future research programmes and activities at national, transnational and European level.

## Realising synergies

Overcoming the inherent fragmentation

**Full title:**

ERA-NET for research  
programmes on rare diseases

**Research field:**

Rare diseases

**Coordinator:**

France: GIS-Institut des maladies  
rares, Institut National de la Santé  
et de la Recherche Médicale  
(Inserm)

**Partners:**

- France: Inserm/GIS-Institut des  
maladies rares
- Germany: Der Projektträger  
im Deutschen Zentrum für Luft-  
und Raumfahrt;  
Bundesministerium für Bildung  
und Forschung
- Spain: Instituto de Salud Carlos III  
Fundación para la Cooperación y  
la Salud Internacional
- Belgium: Fonds National de la  
Recherche Scientifique
- Israel: Chief Scientist Office,  
Ministry of Health
- Italy: Istituto Superiore di Sanità
- The Netherlands: De  
Nederlandse Organisatie  
voor Gezondheidsonderzoek en  
Zorginnovatie
- Turkey: Türkiye Bilimsel ve  
Teknolojik Araştırma Kurumu

**Affiliated partners:**

- Russia : Ministry of Education of  
the Russian Federation
- Italy: Direzione Generale Sanità,  
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“ We hope that E-Rare will convince more EU Member  
States to strengthen their own national programmes on rare  
diseases, and to participate in our joint activities. ”

of research in the field of rare diseases, and promoting multidisciplinary approaches are certainly two of the principal challenges in this research area. To reach this goal, the first objective of E-Rare is the opening and support of rotational positions, allowing clinicians to get involved in basic research in the field of rare diseases. A second objective is to facilitate the links between scientists working in the field of rare diseases, and platforms or centres that could provide them with the resources and/or expertise they might need. This objective can be achieved through a transnational opening of platforms, with more collaboration and easier access for rare diseases researchers, both at national and at European level.

**Promoting  
common understanding**

The setting up of common procedures, evaluation approaches and guidelines for the use of data management systems and biobanks, will lead to common understanding and unity within programmes. This, together with the preparation of specific cooperation agreements, will prepare the ground for further transnational research activities.

E-Rare partners will launch transnational calls for research on rare diseases. These calls will enable scientists in different countries to establish effective collaboration on a common research project, based on complementarities and sharing of expertise. The results of the first call (launched in 2007, for some 8 million euro) will provide a basis for the

development of future research policies and other transnational actions.

E-Rare actions should trigger a leverage effect, resulting in the extension and development of human resources and technological and logistical possibilities. This will eventually provide a basis from which to overcome the fragmentation and effort multiplication in rare diseases research.

The E-Rare project will certainly have considerable impact on the structuring of research in rare diseases, basic and clinical, in the participating countries (and possibly throughout Europe). In addition, it could help its partners and the European Commission in defining their strategic objectives for rare diseases research. Altogether, E-Rare should eventually provide for the development of better diagnostic tools, and new therapies for the benefit of patients suffering from rare diseases.