

Rare opportunity for co-operation

“ 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. ”

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 mount the large multidisciplinary projects that are needed to make serious progress in combating this major social problem. E-Rare is a preparatory project bringing together three Member States which have already established national rare disease research programmes.

But they hope to recruit additional partners from other European countries – and have already embarked on an ambitious programme of practical activities which will quickly improve the speed, efficiency and impact of European rare disease research.

This is a preparatory project. The consortium will use it to look for additional partners and to define the workplan for a full Coordination Action under the ERA-NET scheme.

By definition, individual rare diseases affect no more than one person in every 2,000. But there are between 6,000 and 7,000 different ones, mainly of genetic origin. Taken together, 5% of the population – some 20 million European citizens – are affected. Most of these diseases lead to chronic illness, and they represent a significant burden on health care systems. Yet the specific causes of the vast majority – in particular the wide range of auto-immune disorders – 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 is fragmentary. And since most public funding is devoted to common diseases, the complex research projects that are needed – which normally require interdisciplinary co-operation – are rarely mounted.

Recognising that special funding mechanisms are necessary to address these serious

difficulties, three EU Member States – France, Germany and Spain – have very recently launched national research programmes specifically devoted to rare diseases. In the United States, the November 2002 Rare Disease Act supported regional centres of excellence for clinical research, training, and the development of new methods for the prevention, control and treatment of rare diseases. The lack of a Europe-wide platform that could mobilise funding, scientists and patients on an equivalent scale may place Europe at a considerable disadvantage.

Immediate benefits

Now the bodies responsible for the French, German and Spanish rare disease programmes, each of which has an annual budget of around €5 million, have joined forces under the ERA-NET scheme in the E-Rare Specific support action. Although there are differences between the three programmes, all are designed to increase networking among scientists working in the field of rare diseases, and to pool knowledge and resources in co-operative networks.

The partners' aim in E-Rare is to explore the potential for leveraging their national resources through synergistic transnational co-operation. As soon as possible, they plan to take the first steps towards practical coordination – first, between their own research programmes, and later with similar programmes, or parts of programmes, in other European countries. In the course of the Specific support action they plan to build a larger consortium with the critical



Specific support action



“ We hope that E-Rare will convince EU Member States to strengthen their own national programmes on rare diseases, and to participate in our joint activities. ”

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: GIS-Institut des maladies rares, Inserm
- Germany: Projekttraeger im DLR – Gesundheitsforschung (PT-DLR)
- Spain: Centro de Investigación SAT y Enfermedades Raras (CISATER), Instituto de salud Carlos III

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mass necessary to launch a full, five-year ERA-NET project. Additional eligible partners will be invited to participate in E-Rare immediately.

A European response

The difficulty of assembling the expertise and resources to tackle these diseases effectively, even in a country the size of France or Germany, makes this field one of those in which an ERA-NET is particularly appropriate and is likely to deliver significant added value. The E-Rare partners are confident that there is considerable scope to improve the speed, efficiency and impact of the very large research effort required by linking research databases and by regrouping scientists, research infrastructures and platforms, and patient cohorts at European level so as to avoid duplication.

The partners are convinced that open calls, covering all disease types, will help to overcome the problems of fragmentation by fostering collaboration, networking and integration. They view the Specific support action as an opportunity to prepare for a subsequent ERA-NET project in which the design, funding, evaluation and management of selected programmes or programme elements would

be fully coordinated. But many of the activities needed to achieve this will begin at once. Exchanges, both of information and of programme managers, will be followed by more formal benchmarking, paving the way for the harmonisation of strategies and administrative practice, where this is necessary. A common database of experts is also being built for the scientific review of research proposals.

E-Rare will establish a steering committee, and the partners are preparing to set up an advisory board to develop long-term strategies for optimising the use of existing infrastructures, for defining common programmes and for increasing the impacts of research. They do not expect that they will be able simply to merge existing national programmes. Instead, they hope to start by exploiting obvious overlaps and potential synergies – for example, through common calls, which will be promoted and evaluated transnationally while the resulting projects continue to be funded at national level. The partners hope that the transnational framework provided by E-Rare will encourage other European countries to establish rare disease research programmes which can then join the project.