



Rare Disease Day



Major outcomes from the

European Workshop Bridging Patients and Researchers to Build the Future Agenda for Rare Disease Research in Europe

*Monday, March 1st, 2010
Centre de Presse International (IPC)
Brussels, Belgium*

As part of this year's Rare Disease Day 2010 awareness-raising campaign "Researchers & Patients: Partners for Life!", EURORDIS organised a Workshop to bring together the main players to discuss the future of rare disease research in Europe. The meeting, entitled "Bridging Patients and Researchers to Build the Future Agenda for Rare Disease Research in Europe", was held on March 1st, 2010 at the Residence Palace, Centre de Presse International in Brussels.

The European Workshop, was co-organised with E-RARE in partnership with the European Commission, [ORPHANET](#) and [EUROPLAN](#). European and national research authorities, researchers, policy-makers and representatives from patients' organisations and industry met to identify the future priorities in rare disease research and define concrete steps to ensure better collaboration of all interested parties.

The Workshop offered the opportunity to outline a detailed picture of the rare disease research landscape in Europe and to start shaping the future agenda for rare disease research both at

the national and EU level. This European Workshop “Bridging Patients and researchers to Build the Future Agenda for Rare Disease Research in Europe” was organised at the moment when all 27 EU Member States are developing their national plans on rare diseases before the December 2013 deadline – as a result of the Council Recommendation on Rare Diseases adopted in June 2009- and when the Commission and Parliament are starting to develop the strategy for the 8th Research Framework programme for the period 2014-2020. All attendees had the opportunity to contribute to the ultimate goal of this meeting: to analyse how to increase and secure long-term resources for real patient-centred rare disease research for the upcoming years, research that is expected to improve the health of 30 million patients affected by one of the 5 000 (possibly 8 000) different rare diseases in Europe.

I. A clear picture of the current situation

The **surveys** conducted in parallel by EURORDIS, ORPHANET and the E-RARE network provide a clear picture of the ongoing rare disease research in Europe.

The **study** performed by **ORPHANET**, describes trends and determinants of rare disease research in Europe and it shows that:

- 1- There is a growing activity in rare disease research: approximately 5 000 research projects ongoing covering 2 000 different rare diseases, plus over 650 clinical trials for more than 300 diseases.
- 2- There are only about 500 rare diseases for which there are more than 5 research projects ongoing and less than 1 000 rare diseases for which 1 or 2 research projects are ongoing. What about the 3500 rare diseases for which there is no research at all?
- 3- Strikingly, research activities such as research projects, clinical trials, registries and orphan drug development are focused on relatively few RDs, such as Cystic Fibrosis, Duchenne Muscular Dystrophy.
- 4- The two major achievements at either end of the research pathway so far are :
 - a. The identification of disease responsible genes and their mutations, that could translate into the development of diagnostic tests;
 - b. The arrival on the European market of about 100 medicines specific for rare indications (around 10 new medicines each year), either having received an orphan drug designation or not.
- 5- The development of basic research activities is independent of the prevalence of the single rare disease, whereas therapeutic research projects are mainly conducted in diseases with a prevalence between 1/10 000 and 5/10 000. For the majority of rare diseases affecting less than 1 person in 10 000, therapeutic research is absent or very limited.
- 6- The three main determinants for reaching a significant research activity level in a given rare disease are the existence of

- a. patient organisations;
 - b. patient registries;
 - c. a European network (of centres of expertise or of research) where all actors are already involved.
- 7- The quality of rare disease research projects is very high and they successfully compete with projects in other health research areas. Rare disease research is an area of excellence and innovation.
- 8- More than in other medical fields, the discontinuity of funding and the lack of sustainable funding for networking, research infrastructures and tools like registries, are reasons of major concerns.

The **E-RARE** network presented the results of a **survey** that consulted mainly researchers to identify priorities and bottlenecks in rare disease research. The main recommendations to the European Commission that came out of this study are:

- increase funding;
- promote rare diseases as a model for common diseases;
- facilitate mobility for clinicians;
- fund proof of concept studies;
- fund gaps in translational research;
- address the case of neglected diseases (the very rare ones) and promote EU-funded networks.

From **EURORDIS' survey** the important role played by the rare disease patient organisations as catalysts of research clearly emerges:

- 1- Not only is the existence of a patient organisation a starting point to raise awareness about a given rare disease, for social and medical interest, it also stimulates the development of research activities on that disease.
- 2- Patient organisations provide two types of support to research:
 - a. Non-financial support such as, for example, their involvement in shaping the research agenda for their own disease, or in facilitating the conduction of clinical trials (contribution to their design, to the recruitment of patients, to the information to patients);
 - b. Financial support that fills the gaps and seeds money to start up research where public money not invested: basic research, epidemiology, research in social and human sciences.
- 3- Patient organisations have a limited budget and are calling on public authorities to invest more in rare disease research and more specifically in therapeutic research (clinical trials and research on management of care).

- 4- Patient organisations already have a robust experience of collaboration with researchers as well as with public and private research institution. The quality of this dialogue grows with the age and size of the patient organisations.
- 5- Patient organisations fully understand that research is a long-term process and that both basic research (molecular mechanisms, natural history, pathophysiology,...) and therapeutic research are needed for the prevention, care and cure of their diseases.

II. Moving forward

What can we do starting from the situation just described?

- There is still a need to raise more awareness about rare diseases and the necessity to increase research on these pathologies in the following communities: research (both academic and industry), health and research policy-makers and society at large.
- Basic research and therapeutic research should be supported concurrently as progress in one contributes to the other and patients benefit from cross-fertilisation of research.
- The key priorities highlighted by the E-RARE's survey on the scientific community and by the EURORDIS' survey on patient groups should be addressed by decision makers both at national and European level: to continue gene identification, support translational research, support existing and future consortia and networks, support the development of new therapeutic tools, including the clinical phase.
- Both studies have also shown that we should invest in rare disease research at two levels:
 - a. Specific rare disease budget lines to fund rare disease networks (national and EU level) and infrastructures like rare disease biobanks and rare disease registries;
 - b. Participation of rare disease projects for competitive allocation of funds under general health research budget lines, where projects are selected not on the basis of rarity, but according to the criteria of excellence, innovative ideas, concepts and technologies.
- The issue of continuity and sustainability of infrastructures and long-term projects should be addressed with new, adapted policy and financial instruments at national and European level.
- The collaboration between scientists, clinicians, patient organisations and industry in the framework of specific rare disease networks should be promoted, at national and, even more appropriately, at the EU level. Rare disease research is one of the health areas where new types of research governance and collaboration for co-production of knowledge are emerging. Research on the social innovation of such positive examples should be supported and adapted good practices should be promoted.

- Today the European Commission advises all stakeholders to become involved in the preparation of the 8th Framework Programme for Research. European research has enormous potential in the rare disease area. In this context it is worth reiterating:
 - a. The exceptionally high community-added value of collaborative research on rare diseases (because of their unique situation of rarity) at the European level;
 - b. The high-quality and high-level of innovation of rare disease research advances European competitiveness in a worldwide competition and contributes to reach the EU political objectives (innovation, competitive knowledge-based society).
- Stakeholders have to be consulted to provide their point of view and express their needs, but the rare disease community should speak with one voice. To make the most of the talents, strengths, financial resources, time invested in research, and to translate research investment into medical benefits perceived by 30 million other patients, we must reach a Common or at least a Coordinated Research Agenda. Each stakeholder needs to strengthen its own efforts for additional data collection and analysis, agree to be involved in the reflection process and be ready to make proposals.
- Recommendations for national strategies on rare disease research policy have been developed in the context of the EC-funded EUROPLAN project. A better coordination between E-RARE, EUROPLAN is needed in view of the development of National Plans that would follow these Recommendations. These policy recommendations for national strategies on rare disease research also need to be integrated in an EU coordinated agenda in order to maximise their impact and optimise the allocation of resources.

III. **“Why and how to invest in rare disease research?”**

We should give clear messages to the European and national research orientation committees, as well as to the European Commissioner for Research and to national Ministers in charge of research.

In particular, a common clear and explicit answer to the basic and recurring question: **“Why invest in rare disease research?”** is necessary.

A Discussion Paper drafted by EURORDIS and distributed during the Workshop is aimed at answering this question. It is not enough to ask for more budget for rare disease research and to give it priority: we must explain why rare disease research has to be supported and what is the place and role of rare disease research in the general health research agenda, so as to provide for a robust base for budget negotiations and for budget increase. In order to answer the other broad question **“How to invest in rare disease research”**, it is important to bear in mind:

- What should be done by Member States and what should be done by the European Commission (DG Research).
- In which areas patient organisations are most needed to advance research: Involvement in policy-shaping and budget allocation; clinical trial design, patient recruitment and patient information; funding of basic research, some epidemiological and social research.
- In which areas public funding is most needed to advance research: For large research projects; to stimulate the collection of a critical mass of knowledge in different areas and specific rare diseases; for a major financial effort in therapeutic research in particular for clinical trials for innovative approaches which cannot rely only on patient organisations and industry; for research on therapeutic strategies and management of care.
- At which level we need to connect public health policies with the rare disease research agenda: At the level of the Centres of Excellence (CoE) and of the European Research Networks of CoE. These are the central infrastructures apt to coordinate and stimulate collaborations; to develop patients registries so to increase knowledge on the natural history of the diseases; to offer the best framework for clinical trials; to develop the standards of care which are in turn useful to identify on which aspect of the disease more research should be performed; or to compare new therapeutic interventions with available ones.

IV. Conclusions

This European Workshop confirmed that rare disease research is dynamic, high-quality and highly competitive. Rare disease research contributes to the medical advancement of both rare and frequent diseases.

The European Workshop also confirmed that there exists a huge gap between the level of ongoing research and patients needs. For 3 500 of 5 000 diseases, there is no research ongoing and for 1000 there are only one or two research projects ongoing.

The concentration of research activities on few rare diseases provides a showcase to demonstrate that research on rare diseases is doable and productive, and that these models can be transposed to other rare diseases or group of rare diseases.

The studies presented at the Workshop confirm that the existence of patients' organisations sparks off rare disease research. All areas of rare disease research that are successful have a register, a network, a critical mass of publications and an active patient organisation behind them: there is a dynamic between all of these elements and they all need to be in place for research to thrive.

However, these determinants of research are alone not sufficient to face the issue of continuity and sustainability of research projects. Public authorities are necessary to support rare disease

research with new, adapted policy and financial instruments at both national and European levels.

The additional effort required in times of pressure on public budget are justified by the improvement of health of EU citizens and the generation of benefits that go well beyond the constituency of rare diseases, affecting the broader health and research agendas. The health of European citizens is a value and a wealth generator. Research on rare diseases: promotes health of patients in EU, has a unique and exceptional high community added value and, contributes to the EU political objectives of innovation and competitive knowledge-based society.

As pointed out by the Commission's Director General Robert Madelin, the new Barroso's Commission agenda has three main "baskets": economy, innovation and people issues. Research on rare diseases fits all three, as it promotes health of European people, generates innovation and produces positive effects towards a more competitive EU economy.

In light of the appointment of the European Committee for Rare Diseases (EU CERD) in May 2010, EURORDIS proposes to create a EU CERD Working Group on Research Policy, that includes representatives from DG Research, E-RARE, ORPHANET, EUROPLAN, EURORDIS, the pharmaceutical industry, the EMA and the COMP.

Vision, creativity and activism are required from the whole rare disease community. The EU CERD Working Group, which EURORDIS offers to support and facilitate, would allow the integration of the recommendations under development for national strategies with an EU comprehensive research policy agenda taking into account the other aspect of rare disease EU policy, preparing for the 8th EU Research Framework Programme and to coordinate actions between all stakeholders: E-RARE ORPHANET and EUROPLAN.



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