



Interview



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Q1 How did you enjoy the 11th European Conference on Rare Diseases (ECRD) and what were your highlights from this event?

The conference happens every 2 years and the purpose is to formulate strategies at a national, international, and European level. The conference also provides an opportunity to look at legislative and non-legislative policies and learn about the latest research. The scope is broad, covering the entire spectrum of rare diseases.

Essentially, ECRD is a policy conference. We want to promote certain areas of policy with the help of the Programme Committee, speakers, and stakeholders. The virtual format worked well. We had a great studio in Paris, France, for the plenary sessions and most of the presentations that took place online were live. In addition, professionals and patients had the opportunity to record testimonials in advance of the conference. Being online definitely allowed for seamless attendance.

Q2 What were the recommendations from the Rare2030 Foresight Study?

Firstly, let me state the issue. There are approximately 6,000 different rare diseases, which are currently estimated to affect 20 million people throughout

the 27 European Union (EU) Member States and 30 million individuals in the World Health Organization (WHO) European Region. Seventy percent of these rare diseases appear in childhood. Furthermore, 94–95% have no specific treatments approved. That does not necessarily mean that patients are not taking treatment because there is the option to use off-label medications in some instances. Clearly, the unmet needs are still extremely high and not yet satisfied. In terms of diagnostics, the average time to diagnosis is still 5 years across Europe and across all rare diseases. Obviously, it has improved in the last 10–20 years for some rare diseases; however, the average remains 5 years, which is not acceptable.

It was for the above-mentioned reasons that the Rare2030 Foresight Study was conducted. We have 20–30 years of action to reflect on in Europe, the USA, and internationally. Based on this knowledge base, can we identify big trends in terms of science, healthcare, product development, and social services over the next 10–20 years? Answering this question was the purpose of Rare2030. Next, a multistakeholder platform developed four scenarios based on the results of the analysis. At the 2020 conference, the community then voted on one of the scenarios. Based on that scenario, the

multistakeholder platform synthesised eight recommendations. The main recommendation is that we need a new policy framework in Europe for the next 10–15 years. We also need to put an end to policy silos.

The study concluded by emphasising that we need a strategy for rare diseases, which is based on our current level of knowledge, maturity, and opportunities in science. We also need to set goals. This represents a substantial transformation because the focus is no longer on creating centres of expertise, establishing registries, or product production. Ten years from now, we are hoping to achieve diagnosis within 6 months instead of 5 years; we should have 1,000 new products, of which 30–40% are curative for rare diseases; we should reduce premature mortality in children aged less than 5 years by one-third; and, finally, we should reduce the socioeconomic burden faced by families by one-third. We believe that these goals can be reached with the appropriate strategy. So, this was the main outcome of Rare2030. The seven other recommendations detail how these goals can be achieved.

Q3 How important was the ECRD conference in delivering the messages of Rare2030?

Thanks to our speakers, it was very important. The conference was an official event of the French Presidency of the Council of the EU. It took place after the high-level conference on rare diseases we held on 28th February in Paris at the Ministry of Health, which involved EU Member States. This time, it was a community and stakeholder event but with the label of the French EU Presidency. The ongoing Czech Presidency will organise an expert conference on 25th–26th October in Prague, Czechia, in order to continue moving the needle towards a new European action on rare diseases.

The conference helped to deliver the message of commitment from Member States such as France, Czechia, and Sweden. The Belgian and Spanish health ministers were also in attendance. These countries were calling for greater collaboration in different areas of healthcare and the implementation of actions to increase access to medicines and diagnostics.

The second strong message was from the European Parliament. Several members of the European Parliament participated in the ECRD conference, and they referred to their previous plenary session organised on 24th November 2021 in Strasbourg, France. This plenary session was dedicated to rare diseases and involved 19 members of the European Parliament from different parties, countries, and committees. They were united in their call for a new European action plan on rare diseases. During the ECRD conference, they argued that action had to be taken in 2022–2023. The European Court of Auditors also had a representative. Two years ago, they published a report on cross-border healthcare and concluded that there was a need to revise the policy on rare diseases in order to take into account new developments. The European Commission (EC) agreed to this and, therefore, it should be delivered. The above messages were also echoed by industry representatives as well as clinicians and researchers. Clearly, the community knows what they want and there is no lack of concrete solutions.

"We wish Europe to be a leader in the development of innovative therapies, not just a follower of the USA"

Q4 Was there an active decision by the Programme Committee to include topics such as health data ecosystem, General Data Protection Regulation, sustainability, infrastructure? How are these decisions made?

The Programme Committee wanted to concentrate on the question of data because it is important in diagnostics, healthcare management, and the development of and access to medicines. Regarding diagnosis, in the context of the European Health Data Space, it is agreed as one of the standards that in a patient's electronic record, the rare disease will be mentioned. Not only are there International Classification of Diseases (ICD) codes but there are also ORPHA codes (codes of the rare diseases). In the future, this will enable a better and more harmonised collection of quality data. Clearly, this is very important in the context of research. Data is also important at the time of diagnosis. There is a common standard for recording the genotype and phenotype and this means we know the exact profile of an individual. Again, this is crucial for future research. We are able to quickly identify the number of patients with a particular geno-clinical profile and then conduct research, especially clinical trials.

Healthcare digitalisation was also discussed at the conference. As a result of the COVID-19 pandemic, there have been rapid developments in the fields of teleconsultation and telemedicine, which are welcomed by patients and their families as well as healthcare professionals themselves. At this year's ECRD, there was discussion around the governance aspect and execution of these technologies within EU Member States.

Q5 At the conference, you spoke in the session 'Can Europe be attractive and sustainable at the same time?' What did this session focus on and how was this addressed with regard to therapies for rare diseases?

We wish Europe to be a leader in the development of innovative therapies, not just a follower of the USA. On the one hand, we are seeing more possibilities to create startup companies in Europe and more access to funding. However, when we look at the products approved in Europe, they are mostly derived from USA-based companies. This is why we are calling for a policy that makes Europe more attractive. The regulatory environment is essential to achieving this. This includes incentives in the upcoming revisions of regulations on orphan drugs and paediatrics; the overall regulatory framework, such as conditional marketing approval and co-ordination between the European Medicines Agency (EMA) and Health Technology Assessment (HTA); and measures to accelerate and de-risk therapeutic development within Europe.

Non-legislative policies are also important. For instance, we need European networks not just for care but also for rare diseases and clinical research. Care networks should be focused on natural history study, registries, research on clinical endpoints, and patient-reported outcomes, which are key elements to perform a clinical trial. Such networks exist in the USA and we hope to align the two networks and share the same standards. This will allow us to conduct trials across the continent quickly.

Although we might have more products approved, this alone is not sufficient. They need to be accessible and sustainable for society and payers. This will require competent authorities within EU Member States to collaborate at the European level and negotiate the price. This is particularly true for the most expensive medicines, such as biologics or cell and gene therapies, which are used for rare diseases.

Q6 In her winning speech, the recipient for the 2022 Young Patient Advocate Award, Danielle Drachmann said: "I am an expert by necessity, not an expert by choice." What is your take on this?

I think this is an excellent definition of patient advocacy. It is not something you have chosen. Instead, you are trying to transform your personal story into a collective action, which generates public interest and ultimately creates benefit. Importantly, it is not just to help yourself or the current generation. It extends to the next generation and across all rare diseases. The actions of patient advocates will likely have long-term and far-reaching impact. This is particularly true in the context of clinical trials. Specifically, the motivation for a patient advocate to participate is only 30% for themselves and 70% to help others. Personally, I find this extraordinary. So, your expertise is based on experience but also knowledge. You have to learn and therefore we offer plenty of training opportunities.

Q7 Do you think there are enough patient advocates on boards and in committees for rare diseases? What efforts could be made by organisations to involve patient advocates in scientific advice and protocol assistance?

The short answer is "Yes" for today's needs but "No" for tomorrow's needs. As you mentioned, what we have tried to do over the years is not only promote policies based on the needs and perspectives of the people but also to promote solutions. The other very important part of our strategy is to be part of the execution. That means being part of the management boards, scientific committees, and assessments. For that reason, at the EMA there are patient representatives on the different scientific committees, including orphan drugs, paediatrics, pharmacovigilance, and in many other subgroups. More and more, we have been able to involve patient representatives in scientific advice and protocol assistance at the EMA, which is great.



However, we are not always able to have the participation of patient representatives. Although we can typically identify a patient representative for a particular disease, you also need to find someone who can speak on the disease beyond their own case. This requires several years of experience at the collective level and at the national, European, or international level. It also means that they will need to speak English. All these meetings are in English, which is a big barrier. Ideally, you also need to have them trained in the basics of protocol assistance and scientific advice as well as the essential steps of a clinical trial. In order to have an impact, patient representatives need to bring something specific to the discussion. To do that, we have developed the EURORDIS Open Academy training courses, which is on clinical trials, drug development, and European clinical affairs. We have also developed workshops on HTA as well as pricing and reimbursement. We have schools focusing on various diagnostic aspects, research, and healthcare organisation at the European level. We have also contributed to initiatives such as Patient Engagement

Through Education (EUPATI). However, we need to scale up. We need more patient representatives not only for the EMA but also for HTA, especially now that patients will be involved in the early scientific advice of HTA and the joint clinical assessment. We also need patient representatives to work with industry along the value chain of the medicine. This could encompass the different steps of development, ranging from early research to access. Importantly, the same patients cannot be involved with both industry and the public regulators or HTA. Therefore, we require double the capacity.

Also, you do not want just one patient representative speaking about a specific disease. In each procedure, you want to have at least two from two different countries. You also need to have representatives across a large number of diseases. This is because of the unmet needs and also the rapidly growing pipeline of projects under development. For each of these therapeutic areas, we should have several patient representatives ready to engage with the developers of medicinal products and also with the authorities. So, we need to expand the training of patients advocates. I am confident that we can achieve this because there is a new generation across Europe, who usually speak more English than previous generations. There is also more openness to working online. We need resources to do this but it remains one of our objectives. ●