UN 11/11 2016 – Terkel Andersen

Rare diseases was coined as a specific issue of societal interest less than 35 years ago in different parts of the world, in the USA as Abbey Meyers just presented, in the Province of Taiwan as Professor Tseng will present in a moment, and in Europe as well. Less well known is the fact that in the Scandinavian countries a similar interest emerged in the late 1970's. The focus here was less on therapies and research and more on combining resources to provide improved counselling and information regarding diseases that would be little known to the decentralized levels of health care and social services.

Representatives of small patient organisations came together with pioneering doctors and geneticists to define and map the issues that would call for a focused societal effort to reduce the extra difficulties linked to the rarity of a disease. At that time only few professionals had sufficient knowledge of rare diseases and the impact on individuals and their family leading to significant often multiannual delays in receiving a proper diagnosis. This quite frequently preceded by misdiagnosis as well as irrelevant and sometimes harmful interventions. In particular many rare diseases were met with misunderstanding and societal discrimination adding to the burden of living with the disease. Support, when provided, would normally be uncoordinated, leaving families with the extra task to make many different systems work together.

I make this sound as a history of the past. For most Europe Union member states it is still happening. And the truth is unfortunately that this is very much still the facts of everyday life of people with rare diseases around the world. But today we have little excuse not to do better. For them as much as for us. The rare disease community at large should benefit from a powerful combination of advances made possible through new knowledge, new technologies, higher professional commitment and the mobilization of patient networks making optimal use of new communication platforms..

Thanks to Internet it is possible to make encyclopedic information and many medical resources available through Orpha-Net and other web services from institutions or patients organisations. A proper diagnosis can be made for most rare diseases thanks to genetic tests or biochemical tests, and sometimes just by using the experienced doctors' eye available in clinical networks. Genetics and information technology have enabled experts to improve classification and codification so to make rare diseases more visible in the healthcare systems. More diseases will have access to a therapy due to large investments in research and drug development. And we benefit from the growing awareness that rare diseases may be rare, but rare disease patients are many and counted in millions on every continent of the planet.

We would not have come this far without the solidarity and vision set out by patients and families themselves working hard to achieve breakthroughs to save their children or beloved ones.

The French patient organization, the AFM-Telethon, has worked for more than three decades to make the dream come true that cures will be found for still more rare diseases. The AFM as well as the Italian Telethon and many others in Europe and the USA, have mobilized public goodwill and raised funding making it possible to invest heavily in banks of biological samples, leading to the first human genome mapping and now exome sequencing as well as funding research which has led to new therapies, including one of the first cures for an extremely rare disease, the ADA-SCID, an immune disease killing babies in their months or years of life.

EURORDIS-Rare Diseases Europe was initiated in 1993 and created in 1997 by 4 French associations AFM-Telethon, Cystic Fibrosis Association, Cancer League and AIDS /HIV Association, joined by patient's organisations from Scandinavia, United Kingdom, Italy, Belgium, Netherlands and Germany. 20 years later

EURORDIS is strong of 800 members from almost all 48 countries from Europe, far beyond EU. 33 national alliances exist now in Europe. 55 European Federations or networks disease specific have developed. Our vision is to improve the quality of life, the duration of life and cure for people living with are diseases. All rare diseases, genetic or not, children or adults, including rare cancers, rare allergies, diseases resulting from environmental impact.

Along these years, our strategy has been to put patients first, and based on patients' & family needs to propose possible strategies in research, in diagnostics, treatment development, in healthcare organisation, in social integration. We build on the US experience to promote an EU Regulation on Orphan Medicinal Products, Pediatric Medicines and Advanced Therapies, but our difference compared to the US was to promote rare diseases as a public health priority so to make it a European priority. And because it was a European priority and because some countries were already engaged, then, the dynamic of rare disease national strategies spread over all EU member states, the European continent at large and disseminated to other countries in the world, picking up on this new set of experience.

The EURORDIS strategy is to empower the patients through information and capacity building and networking and to engage actively in all relevant expertise bodies and decision making committees. And success comes, because everything we do, we do it in partnership with all stakeholders.

A key and unique challenge for EURORDIS, was that from the beginning we didn't develop our action in one country but across a continent, with 48 countries, many different languages and cultures, different national systems, variation in values and priorities. We have overcome these barriers and turned the complexity of this diversity into an advantage to create unity and from unity, impactful actions.

This is why EURORDIS has been taken into international activities.

This international initiatives of EURORDIS are now robust and diverse:

- First with the dissemination of its information produced in 7 languages – English, French, German, Spanish, Italian, Portuguese, Russian -,
- then the Rare Disease Day from 2008 which rapidly became international with 85 countries participation,
- also the development of virtual patient & family networks with the On Line Patients Community of RareConnect bringing 700 patient organisations from around the world and thousand of patients into global conversations
- and more recently the initiative of Rare Diseases International, the global alliance of rare disease patient groups to speak with one voice

All these initiatives are embedded in a spirit of collaboration to push for innovation, information sharing and actions without borders.

The Rare Disease Community is global, and it is rooted in civil society.

With the creation of this new platform within the UN system our hope is that the common effort to source, co-create and co-design intelligent solutions for people living with rare diseases will be integrated much more swiftly into the global community and the thinking and operations of UN Programmes in the future.