

## **Testimonial delivered by Yann Le Cam**

At : « European Actions to improve the Life of Patients' Living with Rare Diseases » 25 October 2011, Luxembourg

On : « Testimonial by patients and family members on concrete impact of EU funding / policies in every day life »

"It is exceptional for me to accept to present a testimony as in my role it is more important to speak on behalf of all people living with rare diseases than about my family. However, in recognition of and support the EU Public Health Programme, I will try to illustrate how I think the EU has a concrete impact on the rare disease affecting my family and rare diseases in general.

When my first daughter was born in 1990, one month later we received a letter from one Paris' Children Hospital saying that "the interpretation of the blood results were uncertain" and that "the test has to be repeated". It was impersonal, vague and abrupt. Few weeks later, she was diagnosed with cystic fibrosis (CF).

Indeed...We didn't know a neonatal screening blood test had been performed on our baby for this specific disease. We didn't know a pilot neonatal screening programme was going on in the Paris area. We never signed an information consent for such test.

So, here we were with our baby, in a hospital paediatric pulmonary service with dedicated physicians and nurses however not specialised in CF and surely not in rare diseases. They were even more afraid by the diagnosis than we were ourselves. The diagnosis announcement was so badly delivered that my wife couldn't look at her baby for one year without wondering if her life will be worth living. The funny thing is that I didn't even know the name of the disease in French. Never heard about it. When I realised that actually I knew it in English from my living the USA. I understood that this was reflecting more than an "awareness gap" between Europe and North America. So, we decided to get second opinions. Firstly in France, visiting two leading and renowned hospital professors.

Guess what? The three doctors had different opinions about the course of disease, its prognosis, the possible therapeutic strategies and diverging views on how to manage day to day care. We were confused.

**For our families and relatives, this diagnosis of CF meant "ignorance", "fears" and "death".**

Quite hard for us as new parents to cope with that...

The French doctors as well as the French CF patient association said they expected for our daughter a survival of 20, 25 years maybe 30 years....even though in the early 90s, only a small proportion of CF patients were reaching adulthood. As an evidence of it, the group of CF adults in the CF patient association, were composed of only few. We came across the information that CF life expectancy was around 45 years in Denmark and Canada.

Nobody really knew. There was no systematic collection of clinical data at that time, no patient registries to measure health outcomes and compare between countries, so to identify the best possible therapeutic strategies.

One year before my first daughter was born, the CF gene was identified. This had spurred a lot of research projects going on and lot's of hope. Maybe too high expectations: All doctors and support groups were telling us "don't worry, 5 years, at the latest 7 years from now, there will be a cure". As I was working in the field of international research on cancers, HIV and hepatitis, I hardly believe this will come true so fast...

This was enough to put my wife, the baby and me on the road, up to USA, at John Hopkins Hospital where there was a "CF Clinic"... a "CF Clinic"! ... supposed to be the best in the US, maybe in the world. After two days of exams and meetings, we got a real independent and valid second opinion. We left with a clear therapeutic strategy. We came back with hope. We had also been educated to do physical therapy and to manage nutrition of our daughter. We knew what to do; We knew how to take care of our baby.

In the course of this quest for best possible quality care, I became more aware of the importance of the US Orphan Drug Act. Indeed, I knew it because the public affairs company (Capitol Associates) I was hiring for work on cancer research and hiv, was the same that NORD was employing for their advocacy on the US OD Act.

I knew the value of this regulation to approve the first AIDS treatments and for some rare cancers. until then, I never thought I could need it for myself, to attract interest of pharma & biotech companies to stimulate drug development into CF and other orphan diseases.

Today, my first daughter is 21 years old.

She is studying at the University, in her 4th year for a Master at Law School. She has an active social life, a boy friend, and she practices sports - gym, skying, tennis, even trapezes once a week.

She lives with three permanent infections - two bacteriological infections and one fungus infection. Her daily care requires a minimum of one hour, usually 1h30' with the physical therapy and the aerosol therapy.

She takes about 30 pills per day, for different medicines. She is resistant to an increasing number of antibiotics and she suffers from several adverse effects of antibiotics which she manages with additional medicines and treatments.

20 years later... we are still waiting for a cure ...indeed, no significant new treatment have been approved for CF.

Still, she is 21 years and doing well. This is due to 20 years of careful and compliant treatments, of autonomous day to day care management, a supportive family and personal strategy. And her own strengths. **Her health status is good today due to the dedication of healthcare professionals, the**

**creation of Children CF Centres of Expertise, and even now, Adult CF Centres of Expertise, were she is educated to be an autonomous adult making well informed therapeutic decision.**

**In 20 years everything has changed.**

20 years ago, rare diseases was not even a concept. There was no public policy. No public awareness. Each rare disease was considered in isolation. The vast majority of rare diseases were simply ignored.

**What we have in common in rare diseases, is rarity and the commonality of issues we are facing. Rarity of patients...of experts...of knowledge...of resources... Common issues in accessing information, diagnosis, care, in promoting research, drug development and access.**

**So, our diseases are rare...**

**...but all together, patients with rare diseases are many...**

5000 rare diseases. Overall, 27 to 30 million people affected by a rare disease in the course of their 2life.

This represent a critical mass of 30 million people with unmet medical needs in Europe.

Europe is the right level to tackle rarity and address our common issues. A European common approach on rare diseases has a high community added value.

**The five key success factors so far have been:**

- To clarify the concepts: Rare Diseases as a Public Health Priority, as a relevant tool for public policy. This concept makes it possible to take action actions all rare diseases in all countries of EU
- To build a Rare Disease Community of Stakeholders - policy makers, patients, healthcare professionals, academia, industry - working together at European level
- To promote professional and public information as well as public awareness
- To build well structured networks such as ORPHANET and EURORDIS
- To create a strategic policy framework.

With this in mind, EURORDIS was initiated.

Our first discussions started in 1993 between few patient advocates and a top civil servant in order to prepare the ground for a proposal of an EU Regulation for orphan & neglected diseases under the upcoming France EU Presidency. In 1996 the first European public conference was held at the French Senate with the support of few people and patient groups, and EURORDIS was created in 1997 thanks to the vision of few people and the determinant generous support of the AFM-Telethon.

I was one of these founding members, writing the by-laws, identifying potential Board members, hiring our first public affairs consultant in Brussels and first staff. We advocated for the adoption of the EU regulation on Orphan Drugs, which happened in Dec 1999 after an intense collaboration between policy makers, industry and patients representatives, as well as an intense public and media mobilisation.

In 2000, I was appointed to the Committee of Orphan Medicinal Products at the European Medicines Agency in London.

This was the first time patient representatives were appointed in an EU decision making body. And the first time patient representatives were full members of any scientific regulatory committee in Europe or in the world.

This appointment was a privilege and a new responsibility. I candidated and was elected Vice Chair of this Committee. This was a tough election. Three turns of votes. And what I was most proud of, was to be re-elected Vice Chair unanimously 3 years later. I had demonstrated the added value of patient participations in such committees.

Meantime, I was gaining vision and confidence that it was possible to create new opportunities to improve the life of million of people with rare diseases in Europe. I decided to dedicate my life to rare diseases. In 2001, I became Chief Executive of EURORDIS.

EURORDIS today is composed of 470 member patients groups representing over 4 000 rare diseases in 45 countries. We have national alliances in 26 countries. Today, we can say that EURORDIS is the Voice of 30 million patients.

EURORDIS is an non-governmental organisation, patient-driven, supported by the European Commission, able to work on common issues on information, diagnosis, care, to promote research, 3drug development and access. We propose policies to address common challenges. We take responsibilities in implementing this policies through projects and public committees, like the EU Committee of Experts on Rare Diseases in which we have four patient representatives and four alternates, along representatives from all 27 Member States, industry and main research or public health projects. I am now the Vice Chair of this EUCERD, representing patients and EURORDIS to implement the EU policy on rare diseases.

In 15 years, EU policy has provided a robust framework on rare diseases:

- The EU Regulation on Orphan Drugs in 1999 This regulation and COMP has delivered over 900 orphan designations and 70 orphan medicines approved. I can see the benefit for my family with over 30 orphan designation in cystic fibrosis and one recently approved for marketing authorisation.
- The EU Regulation on Paediatric Use of Medicines in 2006 and the EU Regulation on Advanced Therapies in 2007. I may need this regulation for a potential gene or cell therapy in cystic fibrosis one day.
- The Commission Communication on Rare Diseases in 2008 and the Council Recommendation for Actions in the field of Rare Diseases in 2009.

In few years, we, as the rare disease community of policy makers, patients and stakeholders, we have put in place a policy framework which is:

- comprehensive: covering from research to drug development and from centres of expertise to registries and information
- integrating EU level with Member States and coordinating actions between Member States in a unique

and consistent way so far

- strategic and long term as it focused in mostly six priority areas such as research, drug development, registries & biobanks, centres of expertise and their network, patient empowerment, information as blocks on which to build future policies to deliver health outcomes.

So, what is already changing?

National plans are the key instrument to turn this vision into reality.

Rare Diseases are a research priority and increasingly so with the International Rare Disease Research Consortium.

Centres of Expertise are currently being identified and progressively supported in all Member States and in the coming years they will be integrated with laboratory diagnosis and common tools such as registries into European Reference Networks for Rare Diseases, covering all rare diseases, to improve access to expertise and care and improve overall the quality of diagnosis and care.

Databases, registries and biobanks on rare diseases will be promoted and linked with common policies, standards, and interoperability so to provide a critical mass of biological material and clinical data to enhance research work.

Web has changed everything with regard to access to information. ORPHANET as the Web servers providing medically validated information and each website of all rare disease patient groups are providing essential information to professionals and patients. Telephone help lines are being developed and networked at the European level. Social media with online patients communities are providing new opportunities to patients and families to create clusters, exchange, support each other and generate new knowledge.

I can already see the benefit of these activities for my family. Centres of Expertise for children and adult with cystic fibrosis exist in some countries and are being increasingly created. CF experts are already integrated into European networks of exchange of medical information and all contribute to a pan-European CF patient registry. These activities are being supported by the EU Public Health Programme and the EU Research Framework. Medical practice in CF is improving thanks to standard of diagnosis and care. In 20 years, my daughter has gained 10 to 15 years of life expectancy, with or without any new important medicines, thanks to the improvement of the offer for care and care management. She now has an expected survival of at least 40 to 45 years. And more progress are expected to come.

**Thanks to EU Policy and funding, people with rare diseases:**

- are not any more isolated in the darkness of society
- they exist in our society with increasing public awareness, public policies, progressively adapted healthcare services
- they live longer and better life.

And there is a tremendous hope that this is only the beginning. We are looking ahead for: - more research - more new treatments to be approved - more healthcare pathways to access best possible **diagnosis and care in Europe - more quality information and support. If there is one health area where EU can make a concrete positive difference in its citizens life, this is it: Rare Diseases!**

Yann Le Cam Father of a young adult living with Cystic Fibrosis

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