EURORDIS
RARE DISEASES EUROPE

2017

20 Years of Achievements in the Rare Disease Community

First version published 12/01/17, revised 26/01/17

20 Years of Achievements in the Rare Disease Community

The rare disease community, including all rare disease patient organisations and their partners, can be deservedly proud of its achievements over the last 20 years.

The landscape has changed drastically during this time. We have gone from near ignorance to the recognition of rare diseases as a public health priority in Europe. EU law now incentivises companies to develop orphan medicines.

European expertise and innovative technologies that can potentially benefit people living with a rare disease have been developed. Nearly all EU Member States now have national rare disease strategies and specialised centres of care. Rare disease research is stronger and recognised for its excellence.

The rare disease patient community is bigger, better connected and highly skilled with great leaders at its helm. Its spirit is collaborative across very different diseases, countries and stakeholders in Europe. Patients are now being taken seriously and listened to by policy makers, researchers and companies. Patient centricity has been the buzz term of the past decade.

As EURORDIS-Rare Diseases Europe celebrates its 20th anniversary in 2017, we take stock of the achievements from the last two decades:

1. **From ignorance to increased awareness**
2. **Rare diseases now recognised as a public health priority**
3. **An empowered rare disease community**
4. **A strong, cohesive patient voice**
5. **Increased patient engagement in rare disease policy, research, healthcare and development of medicines**
6. **Less isolated rare disease patients and families**
7. **An escalation of new medicines for rare diseases**
8. **Translation of scientific advancement into therapeutic innovation of therapies**
9. **Expert centres and a European network providing the best possible care**
10. **Diagnosis of more rare diseases**

EURORDIS
RARE DISEASES EUROPE

1. From ignorance to increased awareness

20 years ago, society was living in ignorance of rare diseases. A special Eurobarometer survey on European awareness of rare diseases published in 2001 found that Europeans had a relatively accurate understanding of what rare diseases are but detailed knowledge and awareness remained low.

Over the last two decades, we have raised awareness so that people better understand rare diseases and their impact through initiatives such as Rare Disease Day.

Since Rare Disease Day started in 2008, tens of thousands of events have been held by thousands of patient organisations in over 100 countries. Rare Disease Day brings together millions of patients, families, carers, medical professionals, policy makers and members of the general public in solidarity to raise awareness.

Photo: Rare Disease Day 2016 event

Photos included in this document were submitted to the EURORDIS Photo Contest or rarediseaseday.org

2. Rare diseases now recognised as a public health priority

EURORDIS, national rare disease alliances and patient organisations have campaigned tirelessly to ensure that rare diseases are at the top of European and national policy makers' agendas. **Since 1999, the European Union has recognised rare diseases as a public health priority.**

EURORDIS actively contributed to the adoption of the 2008 Commission Communication on Rare Diseases, the 2009 Council Recommendation on an action in the field of rare diseases (and has promoted their implementation), and the 2011 EU Directive on patients' rights in cross-border healthcare.

In particular, EURORDIS has supported national alliances with the **development of national rare disease plans in most Member States**. For the next decade, we must act with authenticity and determination to promote bold strategies to ensure that rare diseases continue to be recognised as a public health priority across Europe and around the world. This will ensure that the huge unmet needs of rare disease patients are addressed, and that all patients benefit from equal access to the new opportunities stemming from scientific, technological and organisational innovation in a sustainable and fair way.

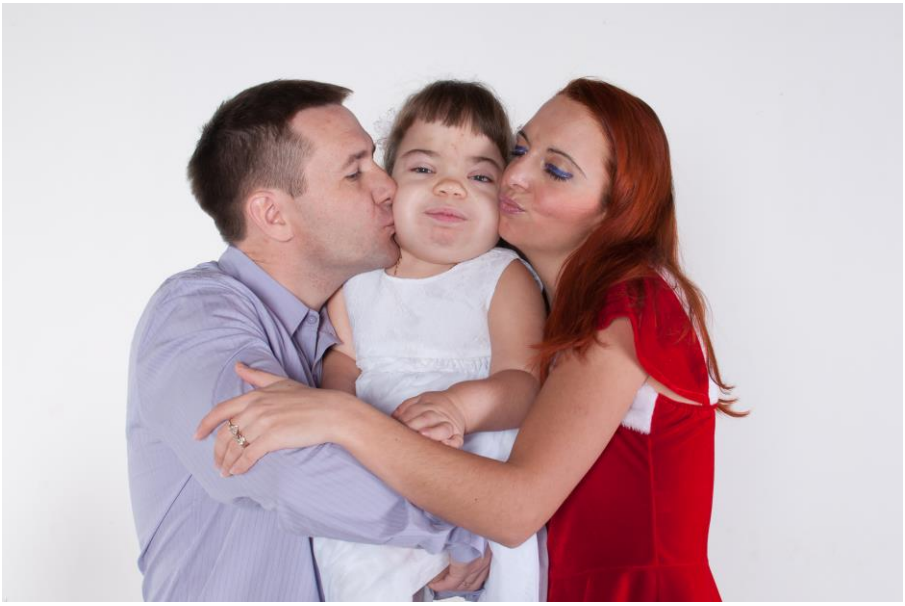

Photo submitted to the EURORDIS Photo Contest

EURORDIS
RARE DISEASES EUROPE

3. An empowered rare disease community

Over the last 20 years, policy makers, researchers and companies have progressively recognised patients as disease experts. EURORDIS and rare disease patient groups have dedicated themselves to empowering patients to effectively engage in research, policy making and the development of therapies:

- Tools and programmes such as the EURORDIS Summer School and online training, as well as the European Patients' Academy (EUPATI), have informed patients and built their capacities so that they are empowered in their roles in important policy-making forums.

  To date, more than 300 participants from over 40 countries and representing more than 75 different diseases have been trained through the EURORDIS Summer School since it began in 2008.

- Sources developed over the last 20 years like Orphanet, the online portal recognised as the primary directory of rare diseases and orphan medicines, are vital to ensure patients are **informed** to empower them in their advocacy activities.

- Capacity building through dissemination of information via: eurordis.org (available in 7 languages), the EURORDIS eNews and Member News, webinars, workshops at the 20 annual EURORDIS Membership Meetings that have taken place all over Europe, and workshops though European Federations and National Alliances.

- Patients are also **empowered by their connections** to the rest of the rare disease community through networks such as RareConnect.

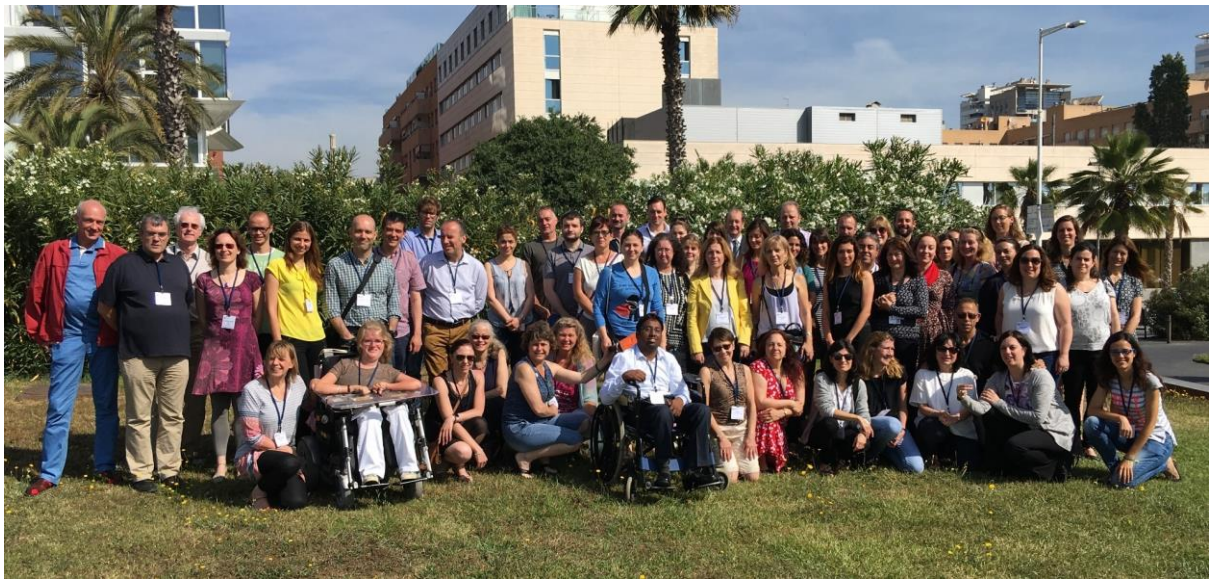

Photo: EURORDIS Summer School 2016 in Barcelona

EURORDIS
RARE DISEASES EUROPE

4. A strong, cohesive patient voice

We have progressed from only two existing National Alliances in 1997 when EURORDIS was created to a network of nearly 50 national rare disease alliances around the world, including the 34 alliances that are members of the Council of National Alliances. Similarly, from 1997 when there were less than 20 existing European Federations to today, when there are 58 European Federations that are all members of the Council of European Federations. By connecting patients and organisations through this network, their voices become stronger and cohesive.

Over the last 20 years and through this network, animated by groups such as EURORDIS, the patient voice has grown in size and strength. **EURORDIS membership has gone from 187 patient organisations in 2000 to over 700 in 2017.**

During the last two decades, empowered and informed patients across the world have used their voice to advocate for improved research, development of therapies and policy making to decrease the impact rare diseases have on their lives.

This voice has been taken to the international level through Rare Diseases International and the NGO Committee for Rare Diseases.

Building on surveys conducted via the EurordisCare Programme, the Rare Barometer Survey Programme is also collecting the opinions of thousands of rare disease patients and family members to ensure that the rare disease patient voice is presented to and heard by policy makers. EURORDIS will continue to act as a knowledge broker providing and encouraging the flow of rare disease patients' perspectives within and outside the rare disease community.

EURORDIS represents the rare disease patient voice in larger coalitions including the European Patients' Forum and International Alliance of Patients' Organizations.

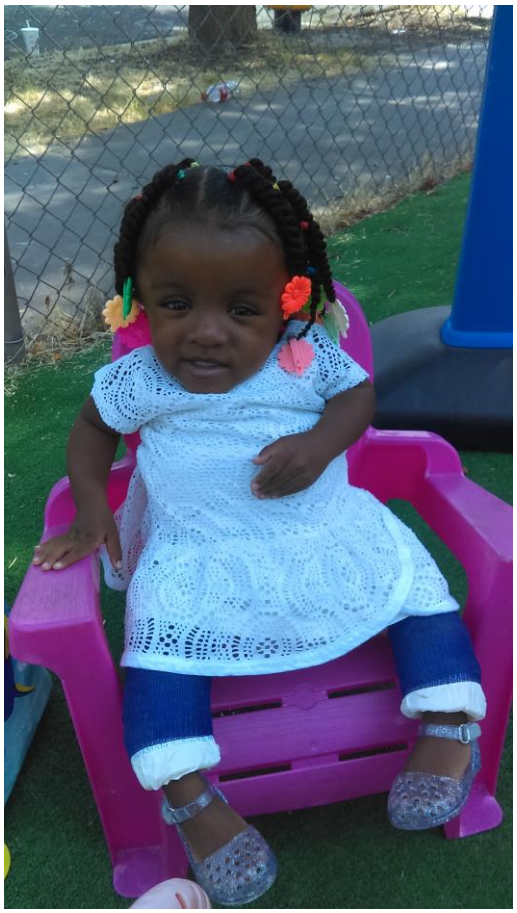

Photo submitted to the EURORDIS Photo Contest

5. Increased patient engagement in rare disease policy, research, healthcare and development of medicines

Over the last 20 years, patients have become further engaged in research and the entire process of developing and accessing care and therapies:

- EURORDIS has developed committees and advisory groups that bring together patients to connect and integrate them into the governance and decision-making structures of bodies that produce or influence rare disease research, policy and develop therapies.

- Patients have become an integral part of the medicines development decision-making process at the European Medicines Agency (EMA) through committees including: the Committee for Orphan Medicinal Products (where EURORDIS Chief Executive Officer Yann Le Cam was one of the first patient representatives to be appointed and also serve as vice-chair); the Committee for Advanced Therapies; the Paediatric Committee; and the Scientific Advice Working Party. **From 2007-2015, there were 743 patients and consumers involved in EMA activities. Since 2008, EURORDIS has facilitated the involvement of 178 patient experts in protocol assistance at the EMA.**

- Patient involvement in the entire lifecycle of medicines development has become mainstream in the last years, from research and development all the way through to post-marketing assessment processes. EURORDIS has supported the involvement of patients in the area of health technology assessment.

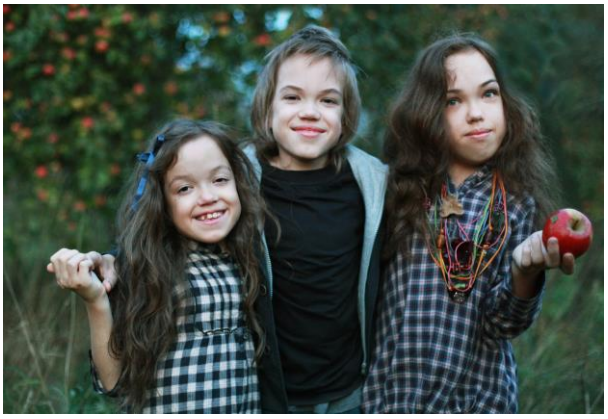

Photo submitted to the EURORDIS Photo Contest

- EURORDIS has empowered patients with the skills they need to participate in, for example, the former EU Committee of Experts on Rare Diseases, the European Commission Expert Group on Rare Diseases, the European Joint Actions on rare diseases (former EUCERD and current RD-Action) and the European Joint Action on Rare Cancers.

- Patients have become more engaged in policy on healthcare pathways. For example, through the establishment of 23 European Reference Networks and the recently launched European Patient Advocacy Groups, which will enhance collaboration between patients, clinicians and policy makers.

- Finally, patients have been integrated into the governance of important research projects and infrastructures that support rare disease research such as the International Rare Diseases Research Consortium (IRDiRC), E-Rare and RD-Connect.

6. Less isolated rare disease patients and families

Because of a lack of information, small patient populations and limited medical expertise, living with a rare disease can be a very isolating experience.

As the rare disease community has grown in size and strength over the last 20 years, people living with or affected by a rare disease have become less isolated.

Rare disease patients and their families are now connected across borders and languages.

Initiatives such as RareConnect and Rare Disease Day help to establish and develop these connections and to reassure patients that they are not alone, that they are part of a growing rare disease community. **There are now 23,000 RareConnect members while there have been 2.5 million visitors to rareconnect.org since April 2012.**

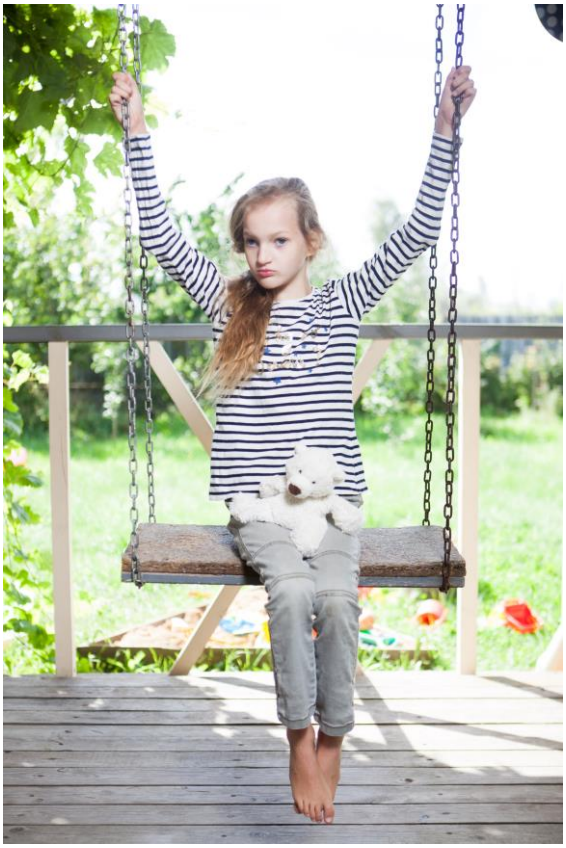

Photo submitted to the EURORDIS Photo Contest

Rare disease helplines offer information and support to people affected by rare diseases. **There are over 50 helplines in Europe.**

In addition, over the past two decades, an increasing number of patient organisations have been created to provide information and create communities so that patients and their families are not isolated.

Disease-specific events organised by patient groups and international events including the European Conference on Rare Diseases & Orphan Products also play an important role in connecting patients and also providing them with the opportunity to meet researchers, clinicians and policy makers.

7. An escalation in new medicines for rare diseases

- In 1999, the landmark EU Regulation on Orphan Medicinal Products came into place, introducing for the first time incentives for companies looking to develop orphan medicines. This is just one of numerous regulations and policy documents that EURORDIS has contributed to that directly contribute to the development of therapies for rare diseases. It followed the US Orphan Drug Act of 1983.

  Since then, 126 medicines have been approved for rare diseases and over 1800 medicines have received an orphan designation, meaning the medicine benefits from market exclusivity.

- One of the overarching goals of the International Rare Diseases Research Consortium (IRDiRC) is to contribute to the development of 200 new rare disease treatments by 2020. This goal will be easily met this year, well ahead of time, thanks to the steady increase of orphan medicines that have received market authorisation in the EU and US. This is great progress but only meets the needs of a small proportion of the rare disease community. There remains a large unmet medical need to provide medicines for the rest of the over 6,000 identified rare diseases. Natural history studies should be carried out to increase knowledge of more diseases so that treatments can be developed.

- By being actively involved in the development of EU regulation around orphan medicines and in the designation procedure of orphan medicines at the EMA, EURORDIS has developed strong expertise in these areas over the last 20 years. The EURORDIS Round Table of Companies (ERTC) was set up in 2004 to bring together companies with a common interest in rare diseases and orphan drug development. Through the ERTC, this expertise has been shared with companies and gone towards reaching the end goal of more and innovative rare disease medicines available to patients. Over the last 13 years, ERTC members have benefitted from taking part in constructive dialogue, facilitated by EURORDIS, involving patients and regulators.

- The availability of more medicines is a great success. The real challenge now is to improve access to those medicines. Initiatives such as the EURORDIS Access Campaign and Mechanism of Coordinated Access to Orphan Medicinal Products have already contributed to ensuring patients' access to new, innovative therapies. Improving access to medicines will be a focal point of EURORDIS' advocacy work for the next 10 years.

8. Translation of scientific advancement into therapeutic innovation

Over the last 20 years, the rare disease community has contributed to creating a hospitable environment for the advancement of science that leads to more, innovative therapies and advanced therapies (a medicine for human use that is based on genes, cells or tissue engineering).

Photo submitted to the EURORDIS Photo Contest

Patient groups all around the world including EURORDIS have influenced all stages of the process and relevant legislations to create this environment: from research, to development of medicines, through the regulatory process, and finally to ensuring patients' access to the new medicines:

20 years ago there were no funds allocated to rare disease research. Through the EU Framework **Programmes for research, funds dedicated to rare disease research have exponentially increased from 64m€** (framework programme 5, 1998 – 2002), to **230m€** (framework programme 6, 2002 – 2006) and **530m€** (framework programme 7, 2007 – 2013). Over **100m€** has already been allocated to rare disease research from 2014 – 2015 under **Horizon 2020**, the EU Framework Programme for Research and Innovation 2014 – 2020. These funds allow for research and create the environment needed to advance science and develop therapies.

New challenges and opportunities are arising around new technologies such as genome sequencing and CRISPR. As well as the landmark 1999 EU Regulation on Orphan Medicinal Products, the 2007 EU regulation on advanced therapies has created ground-breaking opportunities for the treatment of diseases. In addition, the 2006 EU regulation on paediatric medicines facilitates the development and availability of medicines for children.

9. Expert centres and a European network providing the best possible care

People living with a rare disease require specialised care and a high level of expertise from their healthcare providers. Rare disease patient populations are small and medical expertise is limited and scattered across borders. Rare disease patients can become lost and subject to marginalisation in classic healthcare systems designed for common diseases.

The rare disease community has advocated tirelessly over the last 20 years to bring about policies and regulations that have improved healthcare for rare disease patients across Europe:

- Centres of Expertise (CEs) have been recognised as a solution to the challenge of providing effective healthcare to rare disease patients. The Council of the EU's Recommendation on an action in the field of rare diseases (2009) recommended that Member States adopt a national rare disease plan, including a series of measures to improve care and research, and within this identify/ support the creation of centres of expertise.

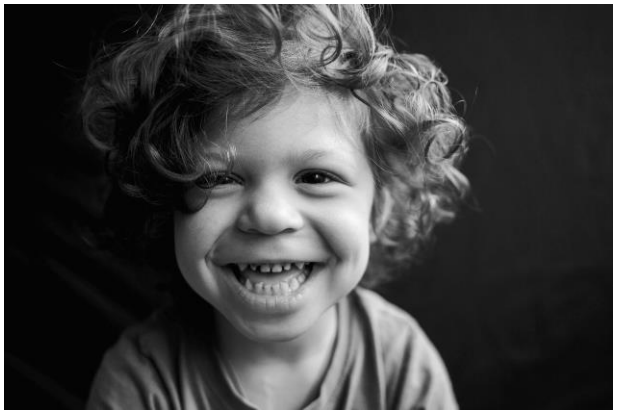

Photo submitted to the EURORDIS Photo Contest

- **Since 2008, nearly all EU Member States have created national rare disease plans and all have established centres of expertise.** EURORDIS and national alliances played a crucial role through EUROPLAN to promote patient-centred national rare disease plans.

- The 2011 EU Directive on Patients' Rights in Cross-border Healthcare states that the European Commission should support the continued development of European Reference Networks (ERNs). The ERNs are networks of the aforementioned centres of expertise, healthcare providers and laboratories that are organised across the EU.

  At the end of 2016, the European Commission announced the first 23 European Reference Networks for rare diseases (ERNs). This momentous step came after years of collaboration and efforts between rare disease patients, clinical experts, and policy makers in EU Member States, at the European Commission and the European Parliament to bring the ERNs to fruition.

  EURORDIS played an integral role in the development of ERNs and in ensuring that patients are at the centre of the new networks. Through the creation of European Patient Advocacy Groups (ePAGs), EURORDIS has structured the rare disease community to ensure that patients are represented at the core of governance and development of ERNs.

10. Diagnosis of more rare diseases

The difficulty in obtaining a correct diagnosis is the first dramatic hurdle for rare disease patients and may take years or even decades to overcome. Some patients are never diagnosed. Late diagnoses delay the beginning of adapted treatments and can have severe irreversible, debilitating and life-threatening consequences.

Over the last 20 years, significant progress has been made in improving diagnosis of rare diseases:

- One of the overarching goals of the International Rare Diseases Research Consortium (IRDiRC) is to make possible the diagnosis of as many rare diseases as possible by 2020. Progress in this field requires the characterisation of new rare diseases and the identification of their cause, mostly through discovery of new genes, and then developing clinical tests.

  IRDiRC therefore monitors (i) the number of new rare diseases identified since 2010 (ii) the number of genes identified as linked to rare diseases and (iii) the number of diseases for which there is a clinical test available.

  More research and funding from targeted efforts by bodies like IRDiRC have contributed to improved diagnostic tools and the data from Orphanet shows that these three indicators are all steadily increasing.

- In 2009, 6,000 patients responded to a survey conducted via the EurordisCare Programme to share their experiences of diagnosis. Analysis of the results of the survey informed EURORDIS' active participation in European projects aimed at improving diagnosis, for example RD-Connect.

- Through membership of the Commission Expert Group on Rare Diseases, EURORDIS contributed directly to the Recommendation on Cross Border Genetic Testing of Rare Diseases in the EU, which recommended that "*obtaining an accurate and timely diagnosis is a priority for all people with a potentially genetic RD; therefore, access to genetic testing […] should be ensured …*".

- There has also been an increase in the development of specific projects across Europe and beyond to improve diagnosis for undiagnosed and rare disease patients (for example, the Undiagnosed Diseases Network International, SWAN UK and the Wilhelm Foundation), while national alliances including the US National Organization for Rare Disorders, the Canadian Organization for Rare Disorders, ASrid Japan and Rare Voices Australia have made great efforts to advocate for the needs of undiagnosed patients. Through these projects, we have seen improved access to next generation sequencing.

  More recently, these groups and EURORDIS came together to publish International Joint Recommendations to Address Specific Needs of Undiagnosed Rare Disease Patients.