> ***If we have a rare disease plan, in addition to our cancer plan, we will show that, in the face of these common challenges, Europe is there. A concrete, approachable, efficient, and human Europe.***

Clément Beaune, Secretary of State
for European Affairs, France

A EURORDIS-Rare Diseases Europe report from the

High-level Ministerial Conference: Care and innovation pathways for a European rare diseases policy

This report has been prepared by EURORDIS-Rare Diseases Europe to shine a light on what was said at the Ministerial Conference. It is not an official report of the conference.

Organised by the French Presidency of the Council of the European Union

Paris, 28 February 2022

EURORDIS
RARE DISEASES EUROPE

A EURORDIS-Rare Diseases Europe report from the

High-level Ministerial Conference: Care and innovation pathways for a European rare diseases policy

Executive summary

Photo credit: Frank Vandenbroucke

"We can only change this situation together, and that is why I fully support the idea of a European plan for rare patients"

Frank Vandenbroucke, Minister of Social Affairs and Health, Belgium

On 28 February 2022, the **Ministerial Conference on Innovation and Care Pathways: For a European policy on rare diseases** took place in Paris at the French Ministry of Solidarity and Health. This was an official event of the French Presidency of the Council of the European Union.

The conference marked a major milestone in the proposal for a European health union for rare diseases, as the first occasion for multi-stakeholder discussions – including the recorded or physical presence of representatives from 12 Member State Ministries of Health.

Rare diseases affect over 30 million people in Europe, and are largely incurable, complex conditions. They are often accompanied by a scarcity of knowledge and expertise making them an area where cross-sector and cross-border collaboration is critical.

The conference demonstrated that there is a clear appetite to build on the progress from over 20 years of EU policies on rare diseases, as reflected in the majority of speeches by Member States, Members of the European Parliament and civil society. France and Czech Republic, through their concrete commitments during their Presidencies of the EU Council, offered the launchpad for a clear strategic plan, with Health Ministers such as the Belgian assuring their full support.

Photo credit: Ministères sociaux / DICOM / Jeanne ACCORSINI / SIPA

A coordinated strategy is the only way to ensure that actions are considered and federated in a comprehensive manner to reach meaningful goals for patients, families and for society at large.

Participants at the conference recognised how the work towards this had been done in the Rare 2030 Foresight Study recommendations: they need to be taken forward as "a battle plan" as set out by Belgian MEP Frédérique Ries.

The Ministerial Conference showed the strong voice and will of policy makers and wider community for a comprehensive strategy on rare diseases. It's now time for Europe to act.

Photo credit: Ministères sociaux / DICOM / Jeanne ACCORSINI / SIPA

Implementing the Rare 2030 Foresight Study Recommendations

It's been a year since the Rare 2030 Foresight Study published its widely supported recommendations. This project was initiated by the European Parliament and funded by the European Commission to bring together experts and patients from around Europe to propose policy solutions for measurable change by 2030.

The main recommendation was for a coordinated European policy framework for rare diseases, to bring Europe in line to work towards important measurable goals on rare diseases. These recommendations are rooted in extensive consultation and promoted by all stakeholders. Since then, it has received wide support from the European Parliament and individual Member States, and been recognised by the European Commission. However we are yet to see concrete action to implement this framework.

An initiative of the French Presidency of the European Union Council

Key facts and figures

30 Speakers

150 Participants

4 French Ministers

12 Member States represented with senior officials from Health Ministries

9 Ministers of Health

3 Senior representatives from health ministeries

France, as the first EU country to adopt a national plan for rare diseases, has been a pioneer in driving rare disease legislation at the European level during their EU Council Presidency in 2008, leading the adoption of the 2009 Council Recommendations on rare diseases, with the strong support of the ensuing Czech Presidency. Along with the Commission Communication that preceded it, the Council Recommendation marked the first and last European framework for rare diseases.

Photo credit: Jérémy Barande / Ecole polytechnique Université Paris-Saclay

"These actions, which follow on from a commitment of almost two decades, make France one of the European leaders in rare disease research. However, this leadership cannot be understood as a solitary exercise. It only makes sense if it plays a driving role within the European Community. The French Presidency of the Council of the European Union is a unique opportunity to intensify this European dynamic to the service of patients."

Frédérique Vidal, Minister of Higher Education, Research and Innovation, France

Photo credit: Marie-Pierre Bichet

"In France, we are very happy to have had three national plans for rare diseases, it is a success. But the victories of tomorrow will be obtained at European level, that is obvious and for us it is essential."

Marie-Pierre Bichet, President of Alliances Maladies Rares

A strong message has been sent through the French EU Presidency by making rare diseases a priority, within a wider context of a stronger European Health Union. By holding this conference on 28 February, Rare Disease Day, they once again have shown leadership in taking steps towards a stronger European collaboration on rare diseases.

Photo credit: Ministères sociaux / DICOM / Jeanne ACCORSINI / SIPA

Objectives

The aim of this in-person Ministerial conference, held in accord with the Presidency Trio of France, Czech Republic and Sweden, was to put forward recommendations to further develop European policy and legislation concerning rare diseases.

By looking at levers to improve care pathways and innovation - including European Reference Networks and a stronger data ecosystem - it provided the occasion to define the EU roadmap on rare diseases and to collectively prepare to implement a European plan for rare diseases.

Stakeholders from across the rare disease community and European policy makers, came together to highlight the added value of strengthening EU action to improve the health and quality of life of its citizens with rare diseases.

Photo credit: Revelli Beaumont

"Together, we will achieve our commitment to leave no one behind by 2030 so that every EU citizen facing rare diseases can be guaranteed the same opportunities wherever they live in the EU."

Oliver Véran, Minister for Solidarity and Health, France

Photo credit: EURORDIS-Rare Diseases Europe

Putting rare diseases in a new political context: COVID-19, Ukraine and the European Health Union

While the focus of the conference was on renewed, concerted EU action on rare diseases, this conference sat with the backdrop of Russia's invasion of Ukraine, only five days before, with speakers demonstrating their solidarity with Ukraine. European Health Ministers – from France to Belgium to Czech Republic to Latvia – showed their support for and readiness to help some of the most vulnerable affected by the war in Ukraine – those living with a rare disease.

Photo credit: Jakub Dvořáček

"We have thousands of people who are coming to us as refugees from Ukraine. A number of them will be patients with rare diseases and again we have to ensure that those people will get the treatment, that they will get the care that they need. And I believe this is also one of the things that we jointly have to work together on."

Jakub Dvořáček, Deputy Minister for Health, Czech Republic

As the situation escalates rapidly, EURORDIS and the whole rare disease community commits to ensuring that people living with a rare disease, whose conditions are often debilitating or need frequent medical attention, are not left behind.

This means supporting Ukrainians living with a rare disease who stay in the country as well as assisting neighbouring countries to ensure that those fleeing Ukraine receive the treatment and care needed.

As the majority of patients are arriving in neighbouring countries, whose capacities are limited, this emergency calls into question the resilience of the European health system, in parallel with the acute pressures created by COVID-19.

"With today's broad range of challenges, European action must draw on and coordinate the work of the Member States, European Reference Networks and patient organisations – in Ukraine, in neighbouring countries and across Europe. The EU has a chance to change the narrative that it cannot afford to miss by leveraging its resources and increasing investment in cross-border healthcare infrastructures to address the needs of the rare disease community. Now, more than ever, we need a coordinated, holistic approach to rare diseases to meet the challenges of today and the years to come,"

Yann Le Cam, Chief Executive Officer, EURORDIS-Rare Diseases Europe

Photo credit: Ministères sociaux / DICOM / Jeanne ACCORSINI / SIPA

A call for a new, goal-orientated, comprehensive, integrated strategy for rare diseases: a European Action Plan

In response to the the outdated nature of the current strategy and the continually siloed approach to managing rare diseases in Europe, a comprehensive and cohesive strategy across the European Commission's policy areas and programmes, integrating EU and national level actions is the only way to address the significant remaining unmet needs of the rare disease community and achieve meaningful goals for patients, families and for society at large.

Photo credit: Ministères sociaux / DICOM / Jeanne ACCORSINI / SIPA

Photo credit: Frédérique Ries

*"**Everywhere, this global approach, this concerted approach to create synergies, to infuse and disseminate European best practice and strengthen each stage of the care pathway is always necessary.** It is not uncommon for someone, as we have heard many times, to wait 10 years for a diagnosis. And even then, it does not mean that because they have received a diagnosis and the disease finally has a name, that a treatment is available. So I am here, to say again and hammer home the message of the European Parliament that it is an action plan on rare diseases that we need today, a real action plan, as a battle plan that makes it possible to regain this momentum of 2009."*

Frédérique Ries, Member of the European Parliament

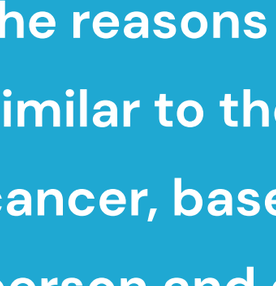
Avril Daly, Vice President of EURORDIS-Rare Diseases Europe, stressed that "as patients it begins and ends with an action plan", calling on Europe to deliver on the consensus reached through the Rare 2030 Foresight Study for its main recommendation for a European framework on rare diseases.

Such support for a new generation of European rare disease policies was echoed by all speakers at the conference, including specific calls for a new strategy from the French, Czech and Belgian health ministers.

Adrien Taquet, Minister of State for Children and Families, France

> **“Indeed, since the European Union's last global policy on rare diseases in 2009, technological and scientific advances have improved the diagnosis, treatment and management of rare diseases. However, the pace of change is uneven on the European continent and in the absence of a general framework at European level we risk backtracking, especially in this period of health crisis. This is one of the reasons why a holistic approach, similar to the European plan against cancer, based on the needs of the person and on better access to high-quality medical and social care, is now essential.”**

Photo credit: Ministères sociaux / DICOM / Jeanne ACCORSINI / SIPA



“A strong European Health Union is a Union that protects and cares for every citizen, no matter what their disease is or where they live. It is therefore clear that we need to bring EU solutions to this EU-wide issue. This is why, together with the French Presidency, we are working towards a stronger EU response on rare diseases.”

Health Commissioner Stella Kyriakides

Photo credit: European Parliament

Why is European action on rare diseases a priority?

To leave no one behind

Strengthening the cooperation and coordination of Member States on rare diseases through renewed European action will create the ecosystem required to address the unmet needs and persisting inequalities all along the patient journey in accessing a diagnosis, treatments and care.

A structured framework through which to contribute to the United Nations Sustainable Development Goals, ensuring equity in access to diagnosis, treatment and care and ultimately ensuring that no one is left behind was emphasised by many speakers.

Photo credit: Ministères sociaux / DICOM / Jeanne ACCORSINI / SIPA

"Because making a European Health Union means fighting against inequalities. All the testimonies we have heard show the extraordinary disparities, injustices in access to diagnosis and treatment. We must and we can remedy these inequalities at European level, not only from one country to another in the European Union, but also from one region to another, within countries, using all the mechanisms that have been mentioned. And probably even more importantly, through solidarity outside Europe"

Véronique Trillet-Lenoir, Member of the European Parliament

Photo credit: Adrien Taquet Facebook

"This conference should allow us to discuss topics at the heart of the challenges of rare diseases: the question of equity, quality of life, the burden of disease, the fight to break isolation. And this, to bring people together, to unify and to move forward even more together."

Adrien Taquet, Minister of State for Children and Families, France

Photo credit: Frank Vandenbroucke

"It is incomprehensible that a patient with a rare disease in a certain European country has access to an innovative treatment but not a patient with the same disease in another country."

Frank Vandenbroucke, Minister of Social Affairs and Health, Belgium

This will also support Europe's effort in implementing the United Nations Resolution on Addressing the Challenges of Persons Living with a Rare Disease, adopted by the General Assembly in December 2021, as described by Irene Norstedt, Director of the Regional Directorate, DG Research and Innovation at the European Commission, as **"a guiding star in terms of what we need to do - and what we need to do together"**.

Photo credit: EURORDIS-Rare Diseases Europe

Photo credit: Laurence Tiennot-Herment

"It is a fundamental human right, that of access to health for all, as the Resolution adopted by the United Nations National Assembly on 16 December last year has shown."

Laurence Tiennot-Herment, President, AFM-Téléthon

Photo credit: WHO Europe

"This is an important milestone that recognises the critical set of policies to support the more than 300 million people living with a rare disease worldwide."

Natasha Azzopardi, WHO Europe Director of the Division of Country Health Policies and System

To bring high EU added value by connecting the dots between different policy areas

Rare diseases are typically accompanied by a scarcity of knowledge and expertise, and often small patient population sizes in each country, as was emphasised by Michael Hadjipantela, Minister of Health, Cyprus.

These characteristics single out rare diseases as a major area of very high community added value, demanding collaboration in terms of knowledge, data, and research. Such collaboration, whether at the local, national or European and international level, requires clear policies to coordinate efforts in a strategic and concerted manner. As has already been highlighted by Vidal, **"because Europe is the real framework, the real stage of the fight against diseases."**

We can do more by bringing initiatives at EU and national level into one comprehensive strategy - from diagnosis to data to innovation to social care - to ensure we optimise efforts to drive innovation, and for this to reach people living with a rare disease in every corner of Europe.

The EU's Beating Cancer Plan was referenced as a good example of Europe addressing public health issues by MEP Véronique Trillet-Lenoir and Clément Beaune, French Minister of State for European Affairs, who said **"it is this logic, which was followed for the cancer plan, that we must adapt to, and that we must think quickly and concretely about the subject of rare diseases."**

Photo credit: Philippe Berta

"In this case, it is the European Union that will be the force around rare diseases."

Philippe Berta, French National Assembly deputy and chair of the rare disease group

Photo credit: Frank Vandenbroucke

"Let us take our cooperation forward by creating a framework that facilitates a common and supportive approach at the EU level in this area."

Frank Vandenbroucke, Minister of Social Affairs and Health, Belgium

Photo credit: Clément Beaune

"Developing a Europe wide health policy is no longer an option, but a necessity"

Clément Beaune, Secretary of State for European Affairs, France

To integrate national and European initiatives on rare diseases

The latest research as part of the Rare 2030 Foresight Study from October 2020 shows that three EU Member States had never introduced a national rare disease plan, and several had technically expired policies. The National Plans of nine more countries were due to expire at the end of 2020.

Many speakers, especially health ministries, put the emphasis on how a renewed framework on rare diseases should act as a catalyst for a new round of National Rare Disease Plans or Strategies, which have lost momentum and are no longer integrated nor even coordinated at European level.

> **A European Action Plan for Rare Diseases would provide a road map for all European countries to work towards common measurable goals of improved health outcomes, reduced inequalities and increased innovation, in response to unmet needs and ensures that inequalities in addressing the challenges faced by people living with a rare disease are not exacerbated by their country of residence.**

Photo credit: Por Fonte

"By recognising the need to continue to provide specific responses to these vulnerable groups, it is extremely important to extend the implementation periods of the Portuguese strategy created for diseases until 2026. The focus will be on developing improved participation in the European project such as Orphanet, the European Joint Programme for Rare Diseases, the European Reference Networks for Rare Diseases"

Antonio Lacerda, Deputy Minister of Health, Portugal

Photo credit: Republic of Latvia

"I invite us to search for more synergies between national and regional strategies, ***only partnerships on the European level will fully allow us to take advantage of digital solutions and innovations and promote Europe as a global leader in rare disease research.*** *Only together can we improve recognition and visibility of great diseases.* ***Only together we can improve the health and well-being of our people so that rare does not mean alone.****"*

Daniel Pavluts, Minister of Health, Latvia

To make Europe a world leader in rare disease innovation

A renewed focus on rare diseases as a public health priority through strengthened cooperation and coordination of Member States will ensure that scientific, technological and therapeutic advances result in the greatest impact for the 30 million people living with a rare disease in Europe.

Franck Mouthon, President of EuropaBio/ FranceBiotech, stressed the importance of early dialogue between innovators and health authorities at the European level **"because this allows us to do two things. It allows health systems to express their needs and it allows this entrepreneurial dynamic to position itself to be able to propose solutions."**

As EURORDIS Chief Executive Officer Yann Le Cam said during the conference, **"How do we bring innovation to patients?"**. Speakers insisted on the ecosystem built on competition and collaboration in science and innovation to ensure that progress across all areas makes it to patients.

Photo credit: Véronique Trillet-Lenoir

"There is, we know, a specific medico-economic model to be invented to amplify innovation on rare diseases."

Frédérique Vidal, Minister of Higher Education, Research and Innovation, France

Photo credit: Laurence Tiennot-Herment

"The medicine we deploy today is innovative and disruptive, therefore our actions should be innovative and disruptive"

Laurence Tiennot-Herment, President, AFM-Téléthon

Photo credit: Katia Julienne

"There is a need for an ecosystem that encourages investment and innovation, placing the unmet needs of patients at the centre of all activities. To break down barriers to the scarcity of these diseases, we need to imagine resilient infrastructure, promote inclusive and sustainable industrialisation by encouraging innovation."

Katia Julienne, French Ministry for Solidarity and Health , DG Directorate of Health Care Supply

To ensure rare disease data sits within a strong ecosystem

Photo credit: EURORDIS-Rare Diseases Europe

There are a number of types of data important to advancing the field of rare diseases: quality of life, health economics, public health, natural history, genomics and much more.

Collecting and sharing all of these data sources on rare diseases emerged as a strong need as part of any rare disease strategy in order to achieve a critical mass of information, given the scarcity of knowledge and expertise and the geographically dispersed patient populations. This was noted especially in the context of the upcoming European Health Data Space legislation.

“I can tell you that patients want their information, their data to be utilised and utilised appropriately for research purposes and to improve the lives of others, and that is absolutely true.”

Avril Daly, Vice-President of EURORDIS-Rare Diseases Europe + CEO Retina International

Photo credit: ResearchGate / Thomas Linden

“It's essential that infrastructure, legislation and guidelines are another force of knowledge contributing to the effective, secure, and ethical use of quality registries and health data registries. We for our part want to be an active part in the establishment of the European Health Data Space.”

Dr Thomas Lindén, Government Chief Medical Officer, Sweden

Photo credit: Véronique Trillet-Lenoir

“We need Europe, because we absolutely need data. We need structured data, data from clinical trials, but also data from real life and, of course, with respect for confidentiality, which is a hallmark of European research.”

Véronique Trillet-Lenoir, Member of the European Parliament

And why we need action now

The last rare disease strategy and the only one so far was over a decade ago. Since then, technology, science and indeed legislation have progressed.

People living with a rare disease require urgent action, now.

People living with a rare disease are a vulnerable population continually challenged by premature death, significant inequalities in health and well-being, and a lack of access to effective treatments – most recently exaggerated by the COVID-19 pandemic and geopolitical conflict.

Photo credit: Ministères sociaux / DICOM / Jeanne ACCORSINI / SIPA

Photo credit: Jérémy Barande / Ecole polytechnique Université Paris-Saclay

"Every day that passes, waiting for a diagnosis, is one day abandoned to the progress of the disease. A day lost for management for genetic counselling or for inclusion in a therapeutic trial."

Frédérique Vidal, Minister of Higher Education, Research and Innovation, France

Photo credit: Frédérique Ries

"I would simply like to conclude by giving the floor to a mother, Isabel, the mother of a Belgian child living with a rare disease, who told me that for them, for us, ***there is no plan B, there is no other life. We must not wait for the next Commission, we must not wait for 2024.*** *That is the message of the European Parliament."*

Frédérique Ries, Member of the European Parliament

The flagships of EU collaboration

EU Health Commissioner Stella Kyriakides, in a pre-recorded video, highlighted several policy areas where progress has been made and the actions that are on-going.

This highlights many of the key pillars of work at the European level which are instrumental in driving change, as well as many areas such as the European Health Data Space, the General Pharmaceutical Package, and the Cross-border Healthcare Directive which are currently undergoing review.

It was stressed that with these pieces, we have "the contours of global action", however that these initiatives need to be glued together by a coherent, comprehensive plan.

Photo credit: EURORDIS-Rare Diseases Europe

"We recognise and praise the past, on-going and planned initiatives of the European Union, but this Commission needs to stop the prevailing siloed thinking. The ongoing evaluation of the Cross Border Health Care Directive, the revision of the EU Regulation on Orphan Medicines, the launch of European Health Data Space and more, are not federated by clear public health objectives. They are therefore at risk of missing the point. An urgent strategy to coordinate and learn from each area and each country, and to create the eco-systems for progress is paramount to make measurable change."

Yann Le Cam, Chief Executive Officer of EURORDIS-Rare Diseases Europe

Photo credit: Clément Beaune

"As far as a plan dedicated to rare diseases is concerned, I think that finally we are almost there with a whole series of building blocks that draw the contours of a global action."

Clément Beaune, Secretary of State for European Affairs, France

European Reference Networks: an exemplar area of EU collaboration

European Reference Networks (ERNs) were at the heart of discussions at the conference, standing out as a success story in how to drive collaboration across the European Union, with their networking function praised by several health ministers.

The progress since their initiation five years ago, now with 24 networks connecting over 1500 clinical centres, was outlined by Chair of the ERN coordinators, Professor Hélène Dollfus. With the review of the Cross-border Healthcare Directive and the evaluation of the first five years of life of the ERNs, the next phase of ERNs begins. Speakers referenced the need for stronger integration with national healthcare systems.

Photo credit: EURORDIS-Rare Diseases Europe

Daria Julkowska, Scientific Coordinator of the European Joint Programme on Rare Diseases, also developed on how ERNs as a "major milestone in the approach to equal care for rare disease patients" should be the basis for future European clinical research.

Photo credit: Vrij te gebruiken ovv: Martijn Beekman / D66

"The ERNs have already proven their importance. Our aim is to have more countries represented within the networks and the need to cover more diseases. At the same time, the ERNs need to be cast manageable. The connection of the ends to National Health care systems, for example, is critical."

Ernst Kuipers, Minister of Health, Welfare and Sports, The Netherlands

Securing Orphanet's future

Orphanet, a unique inventory of rare disease data, information and knowledge was praised with calls from speakers to ensure its sustainability.

Ana Rath, Director of Orphanet, said that with political will behind Orphanet to be able to generate and exploit accurate data, it will become possible to answer public health questions at the national and European level.

German Federal Health Minister, Karl Lauterbach, referenced the role of Orphanet in contributing to the diagnosis of rare diseases, and how by mutalising data across the EU will position the Union better in it's fight against rare diseases.

Photo credit: Michael Hadjipantela

"Orphanet as a European inventory for physicians, researchers, and patients is extremely valuable and would like to see it financially secure in a long term plan."

Michael Hadjipantela, Minister of Health, Cyprus

Rare Diseases: a use case in the European Health Data Space

Policy makers from across Europe expressed their interest in supporting the upcoming European Health Data Space, to ensure that rare diseases stand out as an area with high added value within this new legislation.

Photo credit: Revelli Beaumont

"In the perspective of the establishment of the European Health Data Space, I am convinced that we would be well advised to make rare diseases a pilot use case for the primary use of health data, which leads me to suggest that the collection of data on rare diseases be supplemented with a component integrating indicators documenting life expectancy and the waiting time for access to treatment"

Oliver Véran, Minister for Solidarity and Health, France

European Joint Programme on Rare Diseases and the Rare Disease Partnership

The European Joint Programme on Rare Diseases (EJP RD) is a programme aiming to create an effective rare diseases research ecosystem for progress, innovation and for the benefit of everyone with a rare disease. Frédérique Vidal, the French Minister of Higher Education, Research and Innovation, highlighted how this had made it possible to prioritise and coordinate research efforts, and even joint procurement of medicines. The Rare Diseases Partnership aims to build on this to pursue the innovation pathway.

> **"The vision of the rare disease partnership is really to leave no one behind, and to deliver an integrated rare disease multi stakeholder ecosystem and this must be done by supporting robust patient need-led research and utilising the power of health and research data to spearhead the digital transformation change in rare disease research and innovation and to develop new treatments and diagnostic pathways."**

Daria Julkowska, Scientific Coordinator of European Joint Programme on Rare Diseases

Photo credit: EURORDIS-Rare Diseases Europe

Policy initiatives supporting the development and access to treatments for people living with a rare disease

This year, the European Commission is set to adopt a revised general pharmaceutical legislation on medicines for human use, marking a step change in Europe's role in pushing for a future-proof and crisis-resistant regulatory system for medicines, as part of the EU Pharmaceutical Strategy. This includes the revision of the Orphan Medicinal Products regulation, exploring new incentives.

Models of joint purchasing of medicines between countries were recognised as one initiative during the conference to improve access to treatments for people living with a rare disease. Frank Vandenbroucke, the Belgian Minister of Social Affairs and Health, referenced the Benelux initiative to inspire the EU to "take our cooperation to the next level by creating a framework that facilitates a common and supportive approach at EU level in this area".

Only 6-8% of people living with of rare diseases have a treatment, which was cited by many speakers including Adrien Taquet, French Minister of State for Children and Families, who described it a "source of frustration and disencouragement for patients and their families".

Photo credit: Ministères sociaux / DICOM / Jeanne ACCORSINI / SIPA

Photo credit: Carolina Darias

"The pandemic has led us to many adjustments. One of them is that Europe had to come together as Europe. The centralised purchasing response for vaccines is another good example and a necessity for all the treatments to come."

Carolina Daria San Sebastiàn, Minister of Health, Spain

#30millionreasons for European Action on rare diseases at the conference

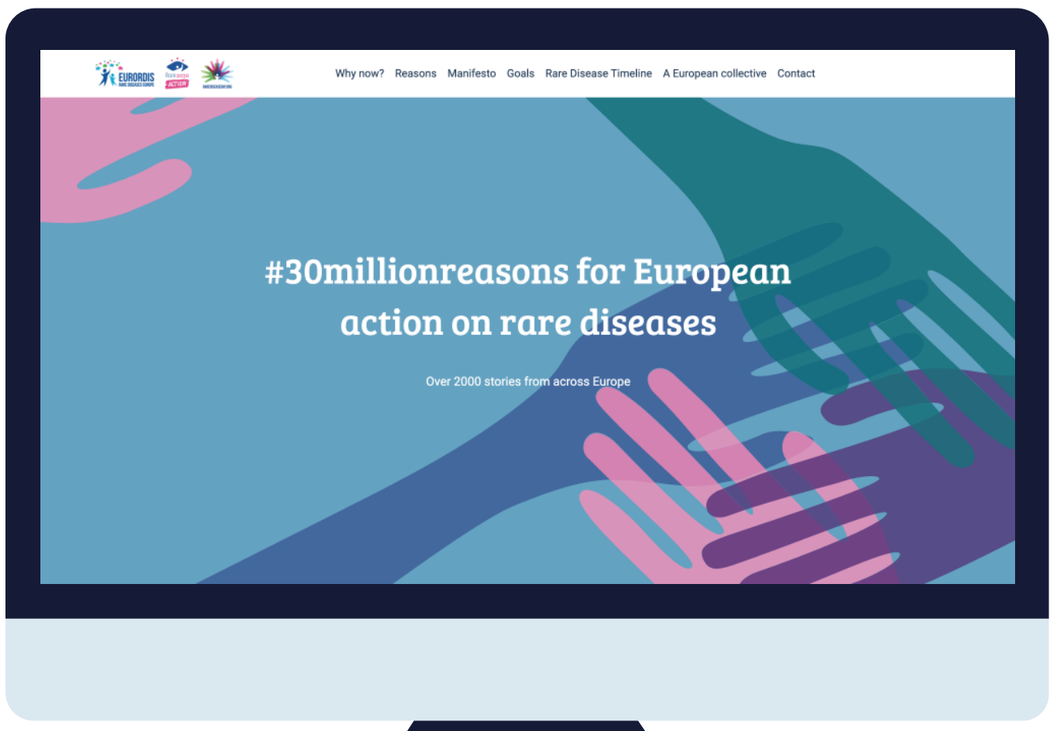
Photo credit: EURORDIS-Rare Diseases Europe

Presentation of the book #30millionreasons for European Action on rare diseases by European patient representatives

Over the past few months, EURORDIS-Rare Diseases Europe asked European Federations, National Alliances for Rare Diseases and its 988 Patient Organisation Members, as well as the wider rare disease community to share their personal reasons for change.

Across Europe, over 2100 people – those living with a rare disease, parents, siblings, friends, advocates, healthcare professionals and researchers – shared their reasons why Europe must act. These are personal experiences, hopes and fears that are moving, motivating, sometimes devastating, impassioned and humbling.

These reasons were brought together in a book and a dedicated website, which were presented to policy makers by patient representatives as part of the conference. All reasons can be read at reasons.eurordis.org



Videos from three case studies were also shown at the conference to bring the patient voice to the three pillars of discussion: ERNs, Data and Innovation.

Prof. Dr. Ildikó Horváth, Secretary of State for Health is presented with the EURORDIS' latest book "#30millionreasons for European action on rare diseases" by Attila Német, Hungarian patient advocate

Photo credit: Jutta

"We really need to make sure that there are harmonised standards in each country; that there is an exchange of information. This is absolutely key to improving the situation and treatment for the patient, and of course also the emotional situation of the family. At the end of the day, despite nurses, doctors, therapists, the burden is carried by the family."

Jutta, patient advocate and mother of Haiko living with alpha mannosidosis

"Only finding out the name of the disease – a diagnosis – when Rocío was already 31 years old, when she was already grown up, brings problems. It brings up things that you might have done differently or better, knowing what we know now."

Rosa, patient advocate and mother of Rosa living with PACS1

"The whole process of drug development and approval was very frustrating because it's a very linear process. You cannot do one thing before having done the next one and there are big gaps between them."

Nick, patient advocate and Founder of AKU Society

Next Steps

The ministerial conference was a platform to bring together high level representatives for the first time since the publication of the Rare 2030 Foresight Study recommendations just one year before. From this milestone event it emerged the shared intention of EU Member States and other stakeholders to strengthen EU-wide cooperation around rare diseases.

We cannot lose momentum now. There will be a series of events under the French EU Council Presidency, including further discussions on a future European Union Health Union, where future collaboration on rare diseases should feature heavily.

Photo credit: Slovenian Government Communication Office

"At an informal EPSCO meeting [of health ministers] in Grenoble, it was pointed out that enhancing coordination and strategic planning at the EU level is of particular importance when it comes to rare diseases."

Janez Poklukar, Minister of Health, Slovenia

An official French EU Presidency event, the European Conference on Rare Diseases, organised by EURORDIS-Rare Diseases Europe and taking place online from 27 June to 1 July, will bring together over 1000 stakeholders with an objective to put rare disease policy into action across three goals - to improve health and well-being, to reduce inequalities and to build resilient infrastructure and innovation - in line with the UN's Sustainable Development Goals.

There is a historic rare disease partnership between France and Czech Republic, who 13 years ago advocated together during their consecutive presidencies for the Council recommendations on rare diseases, marking the first comprehensive EU initiative for rare diseases. It is compelling to see these two nations show their leadership once again in 2022: as France hands over the baton to the Czech Republic in July 2022, they are planning several technical meetings to continue the dedicated discussions on rare diseases at the European level.

Photo credit: Jakub Dvořáček

"There will be two events during summer and during the autumn and I believe we will continue where we just finished the day here in Paris."

Jakub Dvořáček, Deputy Minister, Czech Republic

Dates for your diary

27 Jun – 1 Jul 2022

THE 11th EUROPEAN CONFERENCE ON RARE DISEASES & ORPHAN PRODUCTS

ONLINE on 27 June - 1 July 2022

European Conference on Rare Diseases and Orphan Products will be the next opportunity for discussion on transforming the proposal for Europe's Action Plan for Rare Diseases into action

20 July 2022

Technical meeting on early diagnosis and Newborn Screening, Brno, Czech Republic

25-26 Oct 2022

High-level conference on the roadmap towards European action for rare diseases, Prague, Czech Republic

At the time of writing, the rare disease community looks with concern at the ongoing developments of the Ukraine conflict, being aware that it may evolve, and its impact is hardly predictable at present.

However, this context could be yet another illustration of why better coordination between Member States and across sectors could translate to concrete support for displaced Ukrainians living with a rare disease and their families.

Photo credit: Ildikó Horváth

*"The fact that we are all together here, so many of us... **We are capable of changing the world for people living with their rare diseases and for their families. We need to be inspired and very, very dedicated and committed to that. And with that it will help us to make a movement that our children and their children can be proud of.**"*

Prof. Dr. Ildikó Horváth, Secretary of State for Health, Hungary

Press coverage

Multiple news sources covered the proposal including the Financial Times, Le Figaro, and Le Parisien. EURORDIS and the official French Presidency of the Council also issue press releases on the event.

Patient representatives at the conference

Photo credit: EURORDIS-Rare Diseases Europe

Rare Diseases: European added value for collaboration and innovation

6% of rare diseases have a treatment

5 years to diagnosis

72% of rare diseases are genetic

70% of genetic rare diseases affect children

84% of people living with a rare disease had their care disrupted during the COVID-19 pandemic

Rare diseases are a heterogeneous group of largely incurable, often complex conditions. Although individually characterised by low prevalence, the sheer number of rare diseases results in a directly-affected community of 30 million people across Europe alone.

Rare diseases are typically accompanied by a scarcity of knowledge and expertise. These characteristics single out rare diseases as a major area of very high community added value, demanding collaboration in terms of knowledge, data, and research.

Out of necessity rare diseases have become pioneers in tomorrow's transformations and creative solutions for research and healthcare. The community continually brings forward innovative concepts and models created at the margins of the healthcare system but with benefits for all.

Rare diseases were nearly invisible in our health and social care systems in previous decades. Today, they are recognised as a public health and human rights priority in large part due to a policy framework largely defined 13 years ago by the European institutions which set the roadmap for a number of legislative acts.

However these policies have not kept pace with new technologies and values. For the 30 million people in Europe living with a rare disease, huge challenges remain: a diagnostic odyssey, lacking treatments, decreased quality of life and isolation, lack of access to the highest available quality of care, invisibility in healthcare and research systems.

Rare disease policies timeline

2000 EU Regulation on Orphan Medicinal Products

2006 EU Regulation on Orphan Medicinal Products

2007 EU Regulation on Advanced Therapies Regulation

2008 Commission Communication on ‘Rare Diseases: Europe’s Challenges’

2009 European Council Recommendation on “An action in the field of rare diseases”

2011 EU Directive on Cross-Border Healthcare

2017 Launch of 24 European Reference Networks

2019 European Court of Auditors Report on EU actions for cross-border healthcare

2021 Rare 2030 Foresight Study Recommendations

2021 United Nations Resolution on Addressing the Challenges of Persons living with a Rare Disease and Their Families

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A report from EURORDIS-Rare Diseases Europe

Thank you

Thank you to the French Presidency of the Council of the European Union for hosting this event and for your commitment to rare diseases.

Thank you to all the speakers for sharing your expertise, and to all the participants for attending. Thank you to the patient advocates who travelled to the event. Thank you to Jutta, Rosa and Nick, who shared their stories in the videos shown at the event. Thank you to everyone who shared their reasons as part of the #30millionreasons for European Action on rare diseases campaign.

This report has been produced by EURORDIS-Rare Diseases Europe based on the video of the conference which can be accessed at tinyurl.com/2x4y9nvs. This is not an official report of the conference. Quotes have been translated where necessary into English. Any pictures without photo credits are copyright of EURORDIS-Rare Diseases Europe.

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