Photo credit: Marek Šplíchal

Czech Presidency Expert Conference on Rare Diseases

Towards a New European Policy Framework: Building the future together for rare diseases

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

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

Prague, 25-26 October

EURORDIS
RARE DISEASES EUROPE

Towards a New European Policy Framework: Building the future together for rare diseases

Photo credit: Milan Macek

"We should not work in silos anymore. There are great initiatives that have their own path but we need to put it under one roof, and especially we need to adopt a managerial approach to have goals, indicators, and a clear path."

Milan Macek, National Coordination Centre for Rare Diseases UH Motol and Charles University, Czech Republic

Photo credit: Wikipedie

"Our presidency wants to support joint efforts on rare diseases, to address unmet needs and find sustainable solutions for families and society."

Mr. Vlastimil Válek, Deputy Prime Minister and Minister of Health of the Czech Republic

On 25-26 October the Expert Conference "**Towards a New European Policy Framework: Building the future together for rare diseases**" took place in Prague. This was an official event of the Czech EU Presidency, bridging a series of technical and ministerial gatherings on proposals for a renewed strategy for rare diseases following the French EU Council Presidency.

Crucially, a Call to Action on rare diseases, alongside a specific call to the European Commission for a European Action Plan on rare diseases, was presented by Jakub Dvořáček, Deputy Health Minister of the Czech Republic. This will be shared with all EU Member States to get their support for the document ahead of the Employment, Social Policy, Health and Consumer Affairs Council in December 2022.

Photo credit: Jakub Dvořáček

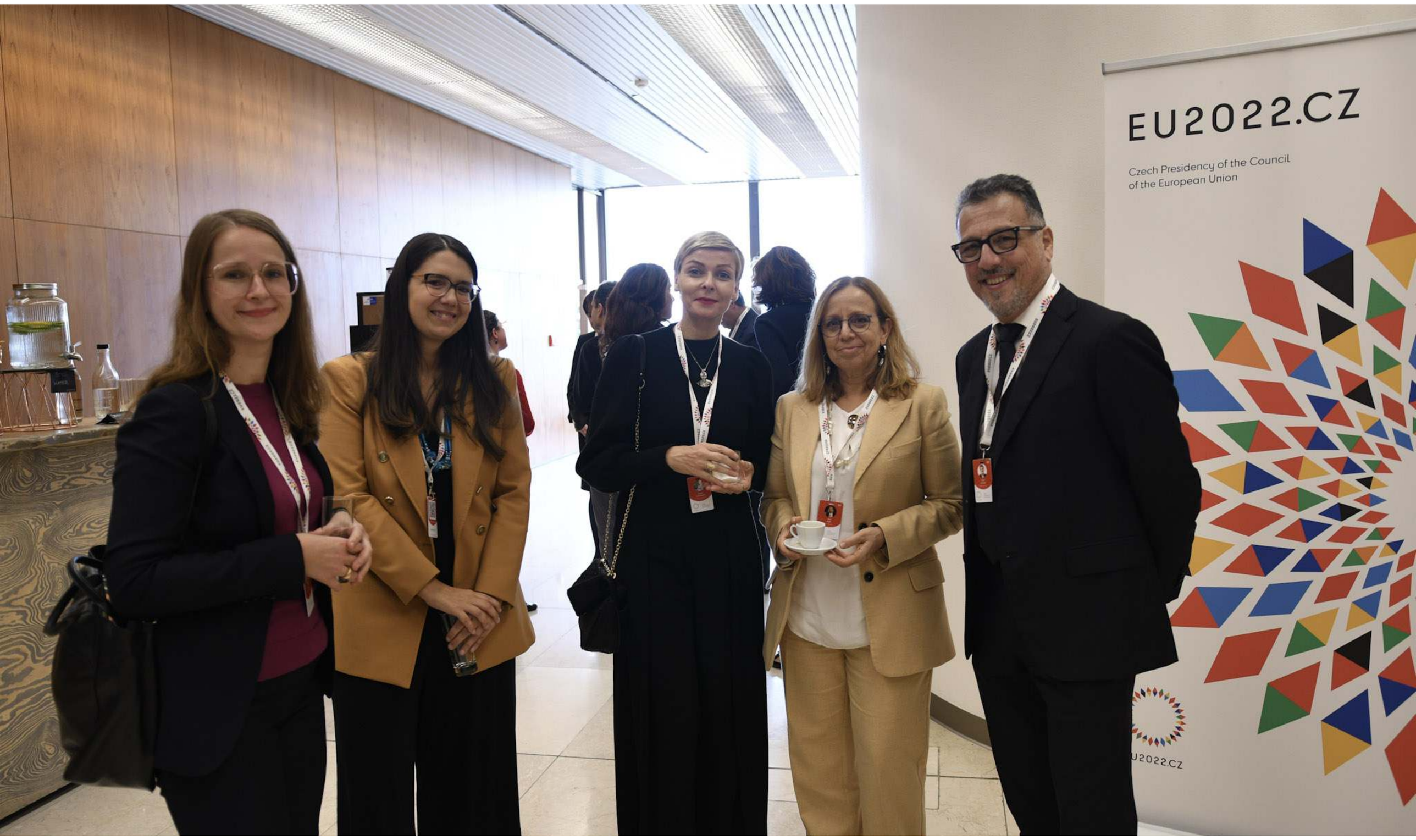
"It's only the Czech Presidency Call to Action for a few more weeks. We invite the support from all Member States."

Jakub Dvořáček, Deputy Health Minister of the Czech Republic

The Call to Action received support from participants in the room, and other EU countries, including France who publicly endorsed the document at the conference. It proposes action across the cross-cutting themes of the conference, from a coordinated goals-based strategy, to early diagnosis, the revision of upcoming EU pharmaceutical legislation, improving access to treatment and holistic care.

The feeling in the room was one of innovation and direction. There was a sense that this was the right time and the right place to take continued steps towards a coordinated strategy for rare diseases that better addresses current unmet needs by setting meaningful goals for patients, families and for society at large, integrated at the national and regional levels.

Photo credit: Marek Šplíchal



The conference benefited from experts from across the rare disease field, leading discussions on improving access to diagnostic services, using data to its maximum potential, improving access to treatments, and on how European Reference Networks can continue to develop solutions for people living with a rare disease.

We hope the conference will leave a clear footprint for decision makers across Europe on the steps that need to be taken to address the unmet needs of people living with a rare disease for the decades to come.

Towards Europe's Action Plan on Rare Diseases

Mar 2017	The **EURORDIS Malta Declaration**, on the occasion of the Conference on Development and Access of Medicines for Rare Diseases, Malta.
2019	**The European Court of Auditors** called to "update, adapt or replace" the European rare disease strategy by 2023, which is fast approaching, in their Special Report n°7/ 2019.
2020	**The European Parliament** included a specific reference for an EU Action Plan on rare diseases in the resolution on the EU's public health strategy post-COVID-19.
Feb 2021	The EU-backed Rare 2030 Foresight Study, developed by 250 experts, concluded that a renewed European rare disease framework was essential to leave no one living with a rare disease behind.
Oct 2021	Slovenia held a conference on "Achieving equity and innovation in newborn screening and in Familial Hypercholesterolemia paediatric screening across Europe".
Nov 2021	**The European Parliament** held a **plenary session debate on 24 November 2021,** with over **40 MEPs adding their signature to this call in a letter.**
Dec 2021	**EU Member States** signed up to the United Nations Resolution on Addressing the Challenges of Persons Living with Rare Diseases. This built on the 2019 UN Political Declaration on Universal Health Coverage (UHC) which included rare diseases and committed all governments to strengthen efforts to address rare diseases in their plans to achieve UHC.
Feb 2022	France hosted a conference on "Innovation and Care Pathways: For a European policy on rare diseases", with Europe's Health Ministers expressing their support for an action plan on rare diseases.
Mar 2022	France included rare diseases as an agenda point over lunch at the EPSCO Council Meeting.
Jun 2022	The European Conference on Rare Diseases and Orphan Products 2022 was designated an official event of the French EU Presidency.
Jul 2022	Czech Republic hosted a technical meeting on Newborn Screening.
Oct 2022	**A Czech EU Presidency Expert Meeting took place on 25-26 October on an EU Action Plan on rare diseases.**

And above all, people living with a rare disease and their families have spoken, explaining in their own words why the EU needs an Action Plan on rare diseases through #30millionreasons for European action campaign.

SESSION I

A New Goal-Based and Coordinated Strategy for Rare Diseases

"Why are we asking for a 'Plan' when there are already all these actions? Simply for one reason: each of these wonderful actions has their own logic. It needs to be federated for the rarest of diseases, with an approach that is comprehensive, integrated and goal-based. And that is the big innovation: goal-based."

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

What does the Czech Presidency Call to Action say?

"The Czech Presidency calls upon the European Commission to adopt a European Action Plan on Rare Diseases to support and complement on-going and future efforts at both the EU and Member State level to reduce unmet needs of the 20 million people living with a rare disease in the EU."

It suggests specific actions including bringing together existing actions, integrating and sustaining national plans on a long term basis, creating an informal working group to establish indicators to monitor the implementation of a new strategy, and introducing measurable goals.

The first session on the call for a new goal-based and coordinated strategy framed the conference in demonstrating how all areas of rare diseases intertwine.

Photo credit: RadioZurnál - Cesky rozhlas

"We recognise rare diseases require significant efforts from policy makers, whether at the national or EU level. A new goal-based approach will facilitate coordinated action by Member States & implement changes more effectively."

Anna Arellanesová, Rare Diseases Czech Republic

It contextualised the **political situation,** understanding the support from the Trio of EU Council Presidencies of France, Czech Republic and Sweden, and especially where Member States want to see collaboration to add value.

Photo credit: RespiFIL

"The Call to Action is really important, and our Cabinet and ministers support it. We need a strategy at European level and not only at national levels."

Anne Sophie Lapointe, Ministry for Solidarity and Health, France

Actions were proposed to strengthen the European rare disease ecosystem, namely by unlocking the potential of data, of securing the future and integration of ERNs into health systems, and improving access to treatments. It was also noted how innovation in rare diseases can pave the way for further progress across other areas of health.

Photo credit: ResearchGate / Thomas Linden

"Rare diseases are the engine of the healthcare of the future."

Thomas Linden, Government Chief Medical Officer, Sweden

The session also saw an overview of the various on-going and planned initiatives in health and research at the European Commission on Rare Diseases.

Photo credit: Governing health futures 2030

"Let's use the potential of Europe, let's use this great family."

Andrzej Rys, Scientific Advisor, DG Sante, European Commission

Importantly, the session put the needs of people living with a rare disease and their families at the centre of the discussion for the conference. It reflected the pace of change in rare diseases - we have seen extensive progress but so many fundamental needs are left unmet.

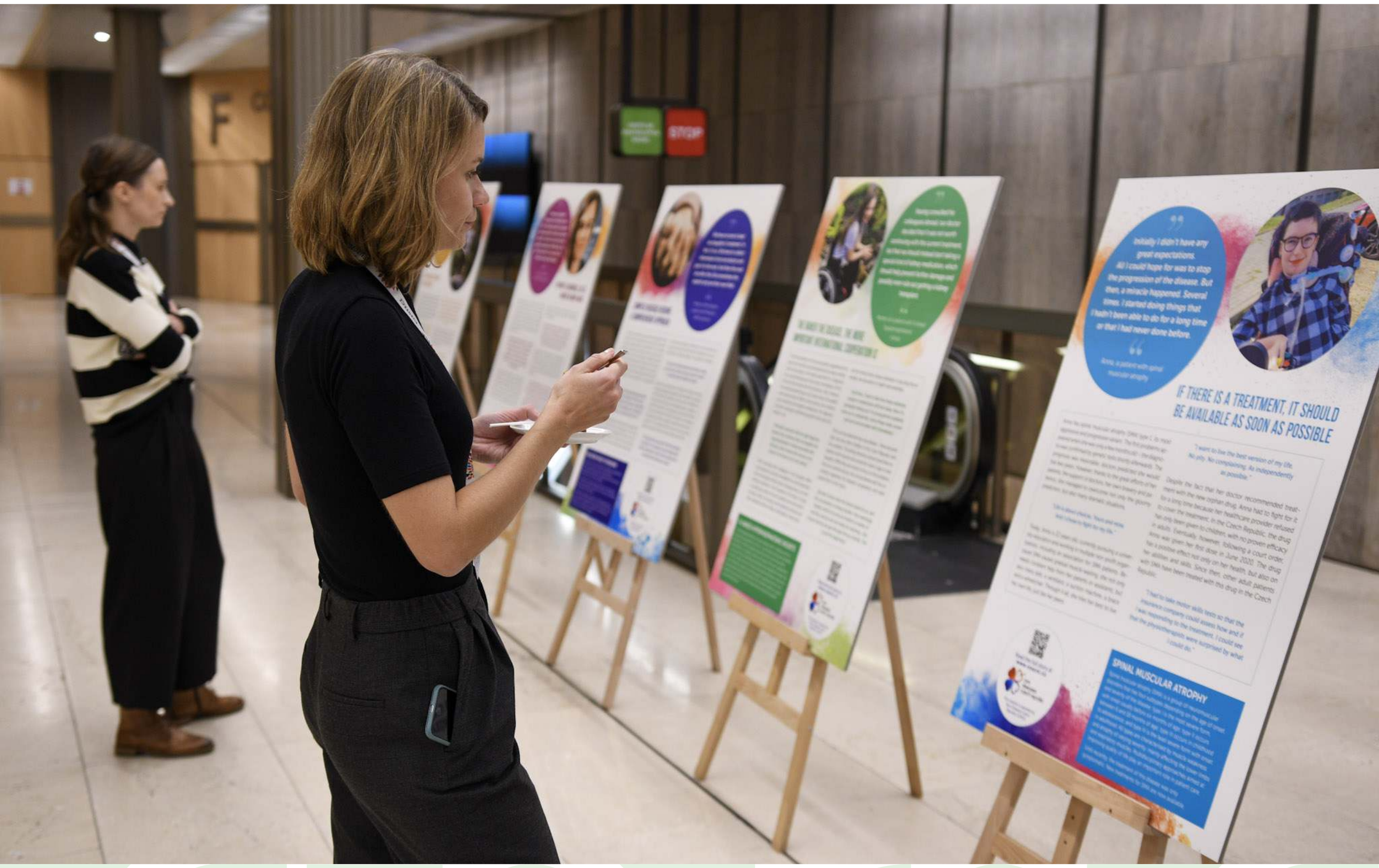
Examples of best practice to provide holistic, person-centred care, such as Resource Centres, were presented to demonstrate how they can help address these needs. It showed how a European approach can help address the needs of the 30 million people living with a rare disease in Europe.

Photo credit: Marek Šplíchal

"As a young patient advocate I see even more the importance of the "now" and not "in 2 years". I had to search for answers myself. I had to discover my diagnoses myself. It took me over 12 years, I have always had to fight for my most basic rights related to my healthcare and still nowadays, I have non-existent support and no specialist that I can go to who would be able to help me navigate in my care. I don't want other young people, other potential rare disease patients to go through what I've been through, to have the same traumatising experience."

Adéla Odrihocká, Rare Diseases Czech Republic

Photo credit: Marek Šplíchal



SESSION II

Early Diagnosis for Rare Diseases

Photo credit: Spot the Early Signs of Mucopolysaccharidosis

"You have seen the different ways of conducting newborn screening, and this is really difficult to accept in 2022. The test is not just a test: it's a system."

Maurizio Scarpa, Coordinator MetabERN and representative of Screen4Rare, Italy

What does the Czech Presidency Call to Action say?

"The Czech Presidency calls on the Commission and EU Member States:

- to support initiatives that aim at promoting the best Newborn Screening (NBS) practice to ensure availability and equity of access to well-structured NBS programmes for all EU citizens and that may benefit from a coordinated EU-wide approach.

- to support such an approach to an expanded number of disease areas and countries across Europe to better diagnose currently "unsolvable" cases."

How can Europe better use the tools at its fingertips to speed up and improve the experience of diagnosis for people living with a rare disease - and how can we improve what happens next?

The striking variation in newborn screening across Europe set out the need for a more harmonised approach across Europe, as an investment in health systems. The number of diseases screened differs between two and 48, sampling practices are widely inconsistent, with timings between sampling and analysis between one day to 30 days, as well as the differing costs of the intervention.

The question of diagnosis was triangulated in this session by data and research. By allowing better access to data through the Rare Disease Partnership for clinicians, ERNs and researchers we can begin to provide more answers. Orphanet provides a pillar for an integrated knowledge base of data, expertise and research, to reach most people living with a rare disease who are not yet reaching the European Reference Networks.

As we are in a new era with genomics giving us even more information, this requires an ever-more integrated multidisciplinary team. Diagnostic devices developed by healthcare institutes are also necessary to be able to conduct these tests. Under the In Vitro Diagnostic Medical Devices Regulation, these will currently need to be reevaluated, risking not being able to be used and slowing down services.

Photo credit: UNSED

"By giving a name to rare diseases - through Orphacodes - it allows to provide, across time, more precision in a diagnosis to a patient."

Ana Rath, Director, Orphanet, France

Photo credit: EOSC Secretariat

"This is a unique opportunity that we have through Rare Disease Partnership: instead of working top down from the European level and working out how this could be translated to national level, we are calling for the national competencies to come with everything you are already doing, so that we can take advantage of this and fill gaps at national level by joining forces at the European level."

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

Photo credit: Ondřej Májek

"Cancer screening recommendations give us a good example of possible pathways towards common key indicators across Europe."

Ondřej Májek, Representative of the National Screening Center of the Institute of Health Information and Statistics, Czech Republic

Photo credit: Monika Glauch

"If the IVDR Regulation kicks in it will be very difficult to develop innovative in-house diagnostic tests and bioinformatics pipelines - then we will have a real problem."

Holm Graessner ERN-RND, Center for Rare Diseases, Tübingen, Germany

SESSION III

Revision of the Orphan Drug and Paediatric Drug Regulations

Photo credit: Rare Disease and Orphan Drugs Journal

"Bringing more innovative treatments is possible: we need to evolve the incentives framework, drive investments in underserved areas and transform the R&D ecosystem and EU pathway from development to access, to ensure innovation is coupled with affordability."

Virginie Hivert, EURORDIS-Rare Diseases Europe

What does the Czech Presidency Call to Action say?

"The Czech Presidency encourages the Commission: To use the opportunity of the upcoming revision of the Orphan Medicinal Products and Paediatric Regulation, together with the planned revision of General Pharmaceutical Legislation, to evolve the incentives framework to maintain predictability for sponsors while enhancing Europe's competitiveness. This needs to be the main focus of the European Action Plan on Rare Diseases."

The Revision of the Orphan Medicinal Products and Paediatrics Regulations offers an unprecedented opportunity to create a robust regulatory framework that will drive innovation and improve access to transformative treatments for rare disease populations in the years to come.

The session brought together all perspectives, including industry, patients and policy makers representing where science meets regulation. Looking at the unique problems and challenges in rare diseases, such as insufficient development in areas of unmet needs, significant variation in access across Member States and scientific advances that are not integrated into regulatory frameworks.

It was stressed to pay close attention to not reduce incentives that could drive away investments from the European market. Different thinking is needed to address innovative therapies such as ATMPs, using existing tools such as the provisions of patients rights in cross border healthcare was encouraged.

Photo credit: DIA/Michael Berntgen

"Connecting the life cycle vision of evidence generation is of paramount importance."

Michael Berntgen, Head of Scientific Evidence Generation Department, European Medicines Agency

Photo credit: EFPIA

"It is extremely important to get the regulation right to allow science to flourish from one area to the next."

Nathalie Moll, Director General, EFPIA

Photo credit: EUCOPE

"We should not be afraid to evolve something that works - and could work better."

Vittoria Carraro, Director Government Affairs, EUCOPE

Photo credit:

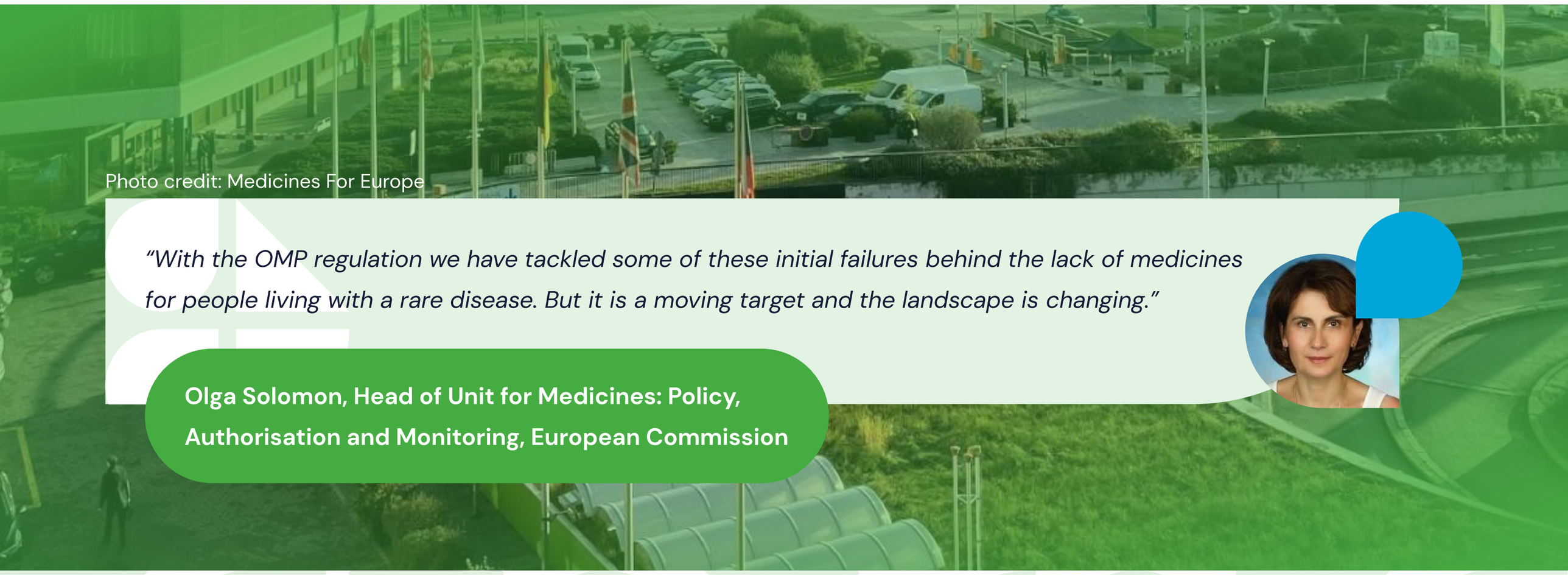


Photo credit: Medicines For Europe

"With the OMP regulation we have tackled some of these initial failures behind the lack of medicines for people living with a rare disease. But it is a moving target and the landscape is changing."

Olga Solomon, Head of Unit for Medicines: Policy, Authorisation and Monitoring, European Commission

SESSION IV

Instruments for Improving Access to Rare Diseases Treatments

Photo credit: EHC

"There is a nonsense expression in English: 'you can't have your cake and eat it'. For people living with a rare disease it seems to have become 'you can't have a therapy for your rare disease and use it'."

Declan Noone, Representative of the European Haemophilia Consortium

What does the Czech Presidency Call to Action say?

"The Czech Presidency finds it necessary to:

A. Explore the feasibility of piloting cross-country mechanisms to improve best practices and information exchanges, value assessments, demand pooling, negotiating and purchasing models, as mentioned by the WHO Europe Statement. [...]

B. Explore opportunities for joint negotiations with producers, and should there be a support of Member States even for opportunities for joint procurement or procurement by the Commission on behalf of the Member States, of complex treatments and treatments for small populations that have the potential to improve accessibility of treatment across the EU providing timely access to patients at an affordable manner, in a way that could possibly be incorporated into the revision of Orphan Drug and Paediatric Drug Regulations as regulatory incentive."

This includes proposals to explore new approaches to affordable pricing, reimbursement and funding, and joint negotiations have the potential to improve accessibility of complex treatments, since they would significantly increase attractivity of even smaller EU markets.

Photo credit: Wikipedie

"Close cooperation between EU countries, between societies and between stakeholders is necessary. The same strategy like we chose for vaccinations, could be a good strategy for orphan products. And then we can offer, as soon as possible, drugs to each European citizen. This is our most important take home message for Europe, Brussels, for the Commission, and for other countries."

Mr. Vlastimil Válek, Deputy Prime Minister and Minister of Health of the Czech Republic

This session was rooted in consensus that people living with a rare disease are not getting access to the treatments they need, with huge inequalities across Europe. The system is complex, with options meeting the "average" needs but not each individual patient's needs.

It was also recognised that collaboration at the EU level is the only way forward. There are reasons to be optimistic: there is a wealth of experience across these areas through MoCA and a new drive through the WHO new mandate on the Oslo Medicines Initiative. This offers a neutral platform to reshape the political discourse: how can the European Union incorporate this?

This hangs on dialogue as early in the process as possible with all relevant stakeholders to be clear on the patient's needs and to reduce uncertainties at the time of regulatory approval. A continuum of evidence generation is critical to this and can offer hope to those living with an ultra-rare disease. We need a roadmap for collaboration of member states in key areas where cooperation doesn't currently exist.

But it was also stressed that this is part of a system. Everything starts with diagnosis, and it does not finish with a treatment. There needs to be a clear line and responsibilities, especially with the increasing number of highly specialised advanced therapies in the pipeline.

“We want to have 3 to 5 times more new therapies approved per year, 3 to 5 times cheaper than today by 2025. And so, we need a structured European cooperation framework for fair prices and sustainable healthcare budgets.”

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

Photo credit: Marek Šplíchal

“Access to medicines is around systems and the way in which healthcare systems have developed, especially around roles and responsibilities for access. Access is as strong as the weakest link. If we don’t have any of these chains in place - from R&D through manufacturing, registration, procurement - we won’t get medicines to patients.”

Sarah Garner, World Health Organization

Photo credit: Monika Glauch

“Access to treatment doesn’t stop there - there is a need to access social care and rehabilitation; also non-pharmaceutical treatment for rare disease.”

Mr. Holm Graesner, ERN-RND, Center for Rare Diseases, Tübingen

SESSION V

Holistic Healthcare Pathways: Integrating Ern into European Health Care and Social Systems

Photo credit: Pavla Dolezalova

"Even if we have the treatments - and they make it to the patient, we need to have the expertise of the clinicians to administer the treatments and follow the drug through the patient lifetime."

Pavla Dolezalova, Head of Paediatric Rheumatology Unit, General University Hospital in Prague

What does the Czech Presidency Call to Action say?

"The Czech Presidency would like to emphasise the need for:

- Stronger integration of the European Reference Networks (ERNs) into national healthcare systems [...]
- Sustainable and proportionate investment from national and EU budgets into strengthening the capacities of ERN centres and enhancing their competencies to better serve patients suffering from a rare disease. [...]
- Leveraging network-based health data, experience and knowledge [...]
- A fully-fledged data strategy for rare diseases [...]
- Implement EU-wide and national policies and programmes to person-centred and integrated care [...]"

Photo credit: Marek Šplíchal

"ERNs can act as a catalyst of a chain reaction as a revolution for Europe."

Maurizio Scarpa, Coordinator MetabERN and representative of Screen4Rare

Photo credit: Oliver Miller-Aichholz

"We have the structures there with European Reference Networks, it's about how we use them."

Mr. Till Voigtländer, Co-chair of the Board of Member States for ERNs

There is no denying that European Reference Networks have completely changed the rare disease ecosystem since their inception five years ago. Rounding off the conference was one of the most dynamic sessions, drawing on how many Airbus 320s that would be needed to transport the number of people involved in ERNs (153 aircraft, for around 20,000 doctors across 500 hospitals), and how they could be used to do better in addressing inequity.

While geographical inequities remain, the call from the speakers focused on using them correctly, with particular emphasis on ensuring the national component is strong. It is usually at this level that healthcare professionals are interacting with patients. The Joint Action on Integrating ERNs into National Healthcare Systems was described to be "a lifetime opportunity to get things right" to ensure the sustainability of ERNs as well as to more effectively to build national capacities and to strengthen the dynamic between Europe and Member States. And a chance to go further: integrating areas that were not addressed in the first round of national plans, including the social aspects.

Photo credit: Alexis Arzimanoglou

"It is not how to integrate ERNs into national healthcare systems, it is the opposite. So let's use the field experience of ERNs to build into the national networks."

Alexis Arzimanoglou, Coordinator of the European Reference Network for Rare and Complex Epilepsies (EpiCARE), Chair of the ERN Coordinators Working Group

Photo credit: MetabERN/Birutė Tumienė

"National plans and strategies are often predecessors of ERNs, so these need to be rethought."

Birute Tumiene, Head of Unit, Center for Medical Genetics, Vilnius University Hospital, Lithuania

Conclusions

Photo credit: Jakub Dvořáček

"After these two days, everything in the Call to Action makes even more sense. We need more coordination, and to do it together."

Jakub Dvořáček, Deputy Health Minister of the Czech Republic

The need for strengthened coordination between areas and countries could not be more apparent: no session worked without reference to other parts of the rare disease ecosystem. A diagnosis is not just a diagnosis, as Prof Maurizio Scarper highlighted, "When you make a diagnosis the family are not interested in the diagnosis - they care about how a child will be in two, five, ten, twenty years". Likewise, in the case of there being a treatment to be administered, this does not end there.

Photo credit: Marek Šplíchal

"On a personal level, I still need to work in silos - treatments, symptom management, trying to find different specialists, working full time while managing my care. If my doctors worked together, it would be a huge help for me. But we also need to work together with a systematic approach, so nobody is left behind."

Adéla Odrihocká, Young Patient Advocate, Rare Diseases Czech Republic

The call for a coordinated European Action Plan on Rare Diseases was therefore strongly echoed by conference participants. We need continued steps towards a coordinated strategy for rare diseases that better addresses current unmet needs by setting meaningful goals for patients, families and for society at large integrated at the national and regional levels.

Five key conclusions from the Expert Conference on Rare Diseases in Prague

1

Data holds the key to improving diagnosis, research, and care.
Evidence needs to be generated throughout the life of a patient, especially with regard to treatment and care. This will support diagnosis, feed into regulatory systems, and improve access to treatments. ERNs can act as instruments for data collection and sharing.

2

Nothing works in silos.
We need a framework that ensures a consistent thread between all areas and all European countries. The ERNs offer a great opportunity, but still have a primarily medical focus. Measurable goals for all countries to aspire to would allow all countries and areas to align their efforts in a common direction.

3

Innovation, innovation, innovation.
It's already happening and we don't want to put the brakes on it. People living with a rare disease and their families should have access to it: from improved genetics, to the most innovative treatments.

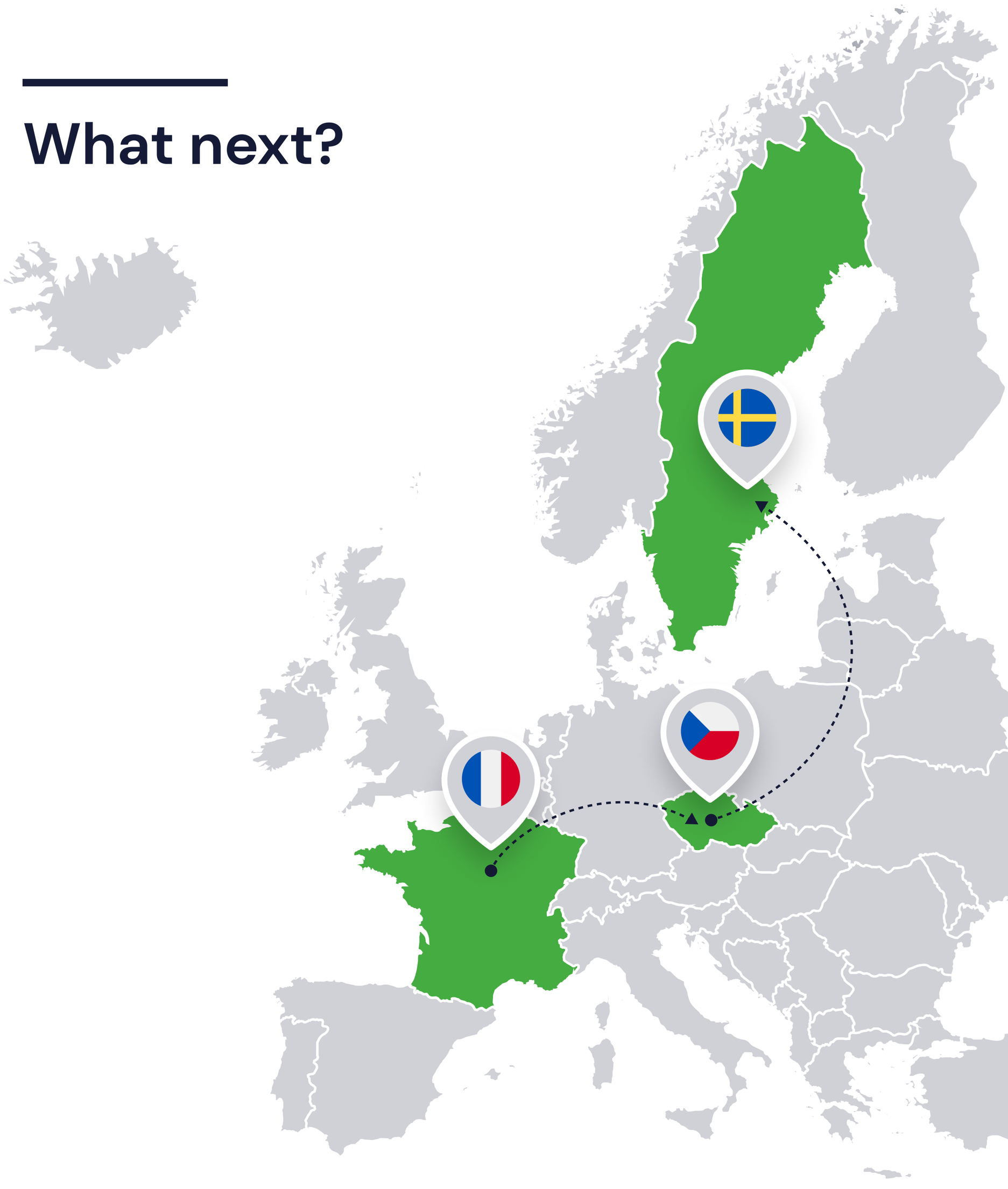
4

Regulatory frameworks matter.
The Revision of the Orphan Medicinal Products and Paediatrics Regulation, and the European Health Data Space offer opportunities to set a clear direction to tackle inequalities while remaining globally competitive.
IVDR regulation should include a clear statement on the possible derogation for medical devices necessary for rare diseases diagnostics.

5

Integrate national and European collaboration.
It is clear that no country can do this alone. However, every Member State has its own competencies in health meaning that they make the decisions on how the health systems operate. There is an appetite for exploring joint working, such as joint negotiations on treatment pricing, or in coordinating care for ultra-rare diseases.
The Joint Action on the Integration of European Reference Networks into national healthcare systems, as well as the Rare Disease Partnerships are upcoming opportunities to ensure a consistent exchange of information between the European Union suited to each Member State. Renewing momentum on National Plans was recognised as a key step.

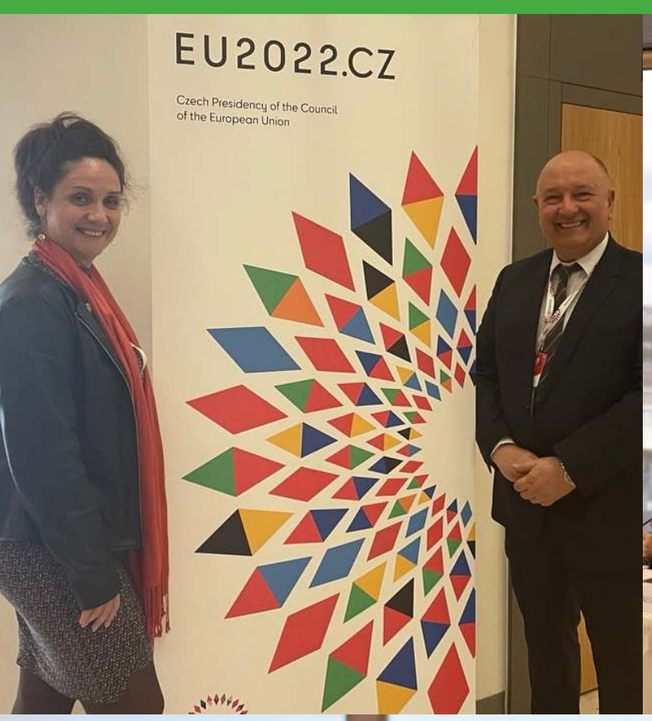
What next?

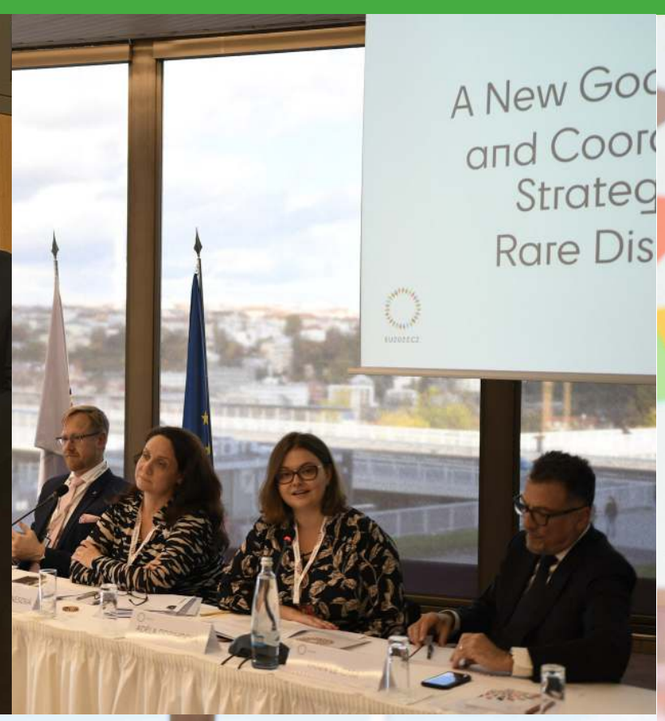


> **We have an army of people and an arsenal of opportunities: let's use them! Deputy Health Minister Jakub commented that he had "never seen such a group".**

The Czech Presidency has shared the Call to Action with all EU Member States for their written endorsement. We hope as many countries as possible will give their support. This document will be shared as part of the Presidency points under the final EPSCO Council Meeting chaired by the Czech Republic in December.

Collectively, we need to continue to strongly voice the call for a European Action Plan for Rare Diseases. We call on the Swedish Presidency to continue the good work paved by France and Czech Republic.





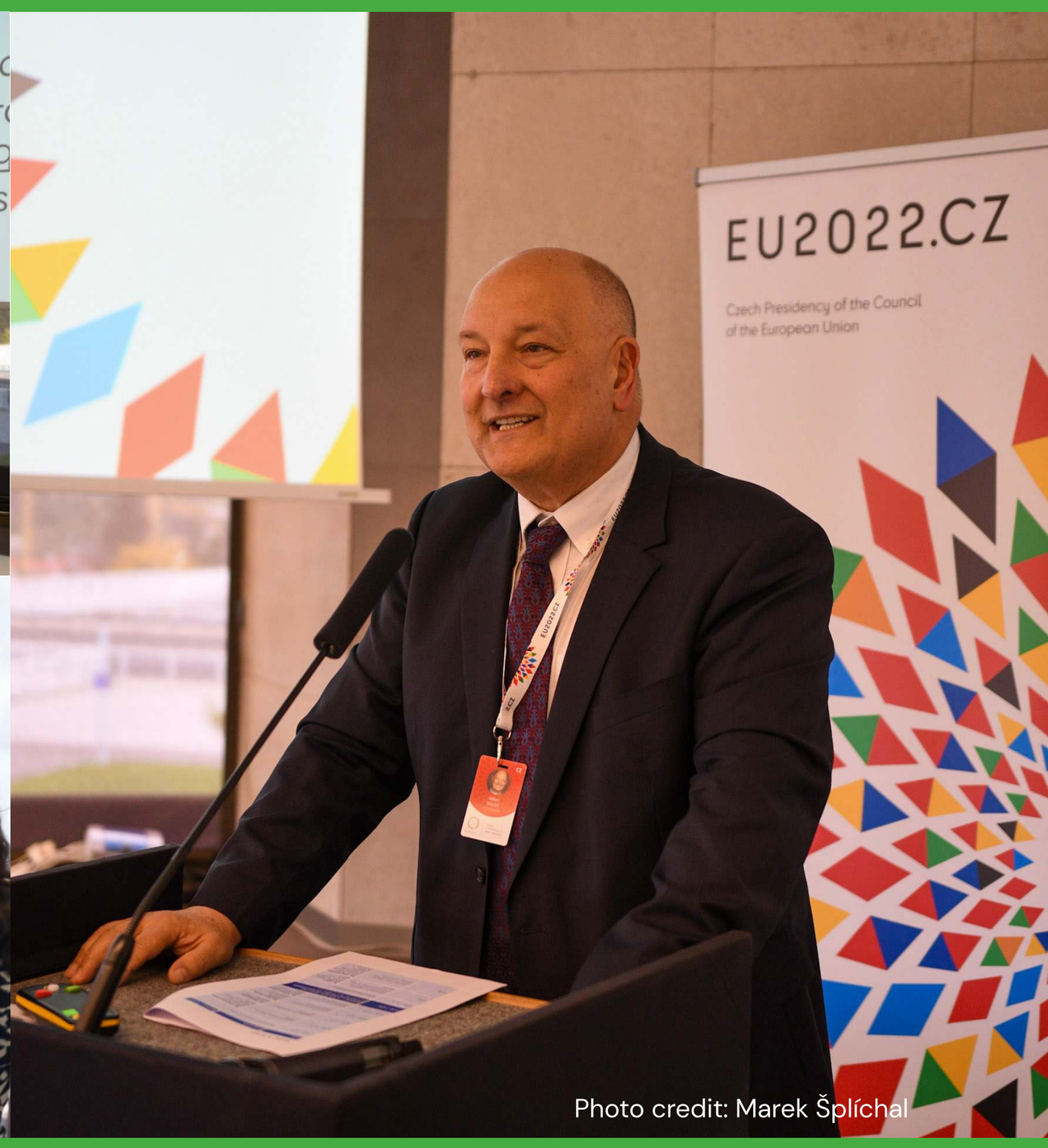


Photo credit: Marek Šplíchal

A report from EURORDIS-Rare Diseases Europe

Thank you

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

Thank you to Professor Milan Macek for your endless drive and to Anna Arrellanesova and all of Rare Diseases Czech Republic (Ceska ascociace pro vzacna onemocneni) for your efforts in preparing the conference and ensuring that the Call to Action receives wide endorsement.

Thank you to all the speakers for sharing your expertise, and to all the participants for attending. 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 from attending the event and based on the recording of the conference which can be accessed here. The Call to Action has been paraphrased in places, made clear by "[...]". This is not an official report of the conference. Any pictures without photo credits are copyright of EURORDIS-Rare Diseases Europe.

EURORDIS-Rare Diseases Europe, December 2022