European Economic and Social Committee

Univerzita Karlova

Call to Action in the field of rare diseases

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National Coordination Centre for Rare Diseases
Czechia

Session 1: Rare diseases: organisational models and good practices in health and social care

Conference on Rare Diseases and European Reference Networks.
How to guarantee solidarity for patients?

Bilbao, 11 October 2023

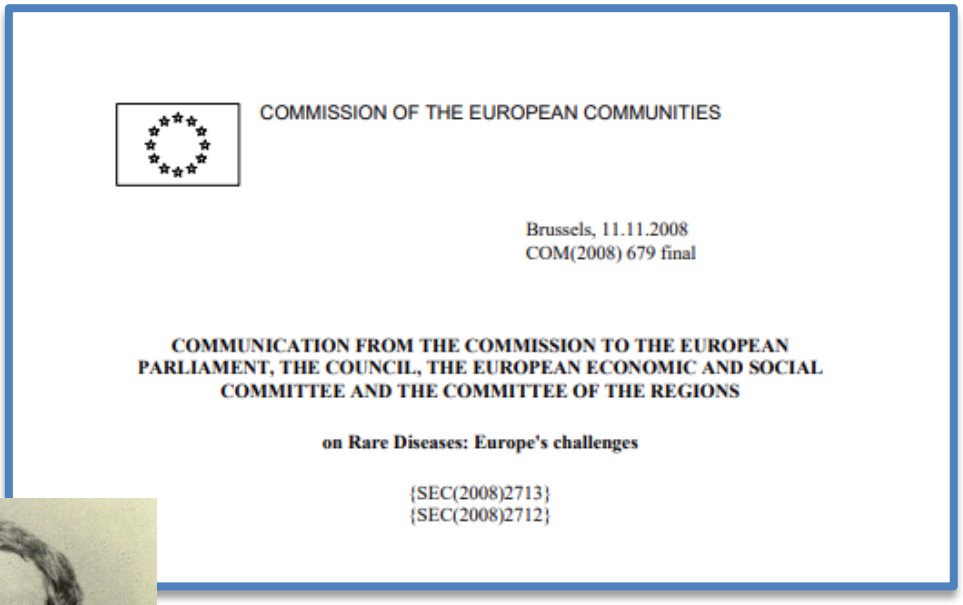
Where are we coming from?

A long standing leadership of Czech Republic in the field of rare „Mendelian“ diseases: the Council Recommendation on Rare Diseases adopted in **2009** under the Czech Presidency of the EU Council

National Strategies / National Action Plans
Empowering patient associations
Orphanet, European Reference Networks

We have made major progress since the 2008/2009 EU (soft) Legislations on Rare Diseases

High unmet needs and inequalities across Europe remain.



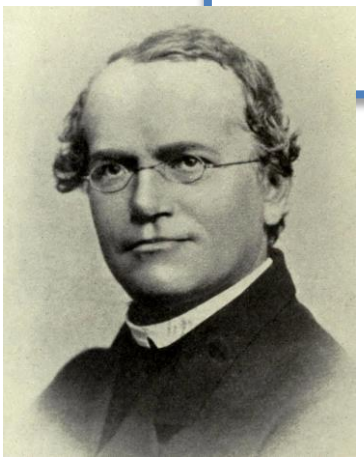
COMMISSION OF THE EUROPEAN COMMUNITIES

Brussels, 11.11.2008
COM(2008) 679 final

COMMUNICATION FROM THE COMMISSION TO THE EUROPEAN PARLIAMENT, THE COUNCIL, THE EUROPEAN ECONOMIC AND SOCIAL COMMITTEE AND THE COMMITTEE OF THE REGIONS

on Rare Diseases: Europe's challenges

{SEC(2008)2713}
{SEC(2008)2712}



European Commission | English

Public Health

Home > Latest updates > National plans or strategies for rare diseases: page updated

NEWS ANNOUNCEMENT | 1 January 1970 | Directorate-General for Health and Food Safety

National plans or strategies for rare diseases: page updated

National plans or strategies for rare diseases: page updated

The Council adopted in June 2009 the Recommendation on an action in the field of rare diseases.

The Recommendation supports adoption of national plans and strategies for responding to rare diseases (RD) before 2013.

The Council recommends that Member States should:

- Establish and implement plans or strategies for rare diseases or explore measures for rare diseases in other public health strategies, to ensure that patients have access to high quality care, including diagnostics, treatments, habilitation for those living with the disease and, if possible, effective orphan drugs.

In particular, the Member States should:

(a) elaborate and adopt a plan or strategy **by the end of 2013 at the latest** to guide and structure actions in rare diseases within their health and social systems;

https://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF;
https://health.ec.europa.eu/latest-updates/national-plans-or-strategies-rare-diseases-page-updated-1970-01-01_en

European court of auditors report - 2019

EN 2019 NO 07

Special Report

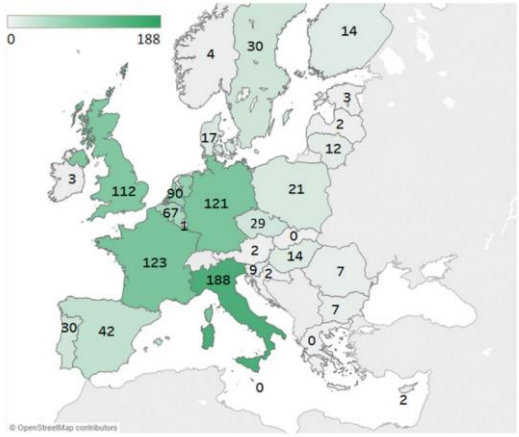
EU actions for cross-border healthcare: significant ambitions but improved management required

(pursuant to Article 287(4), second subparagraph, TFEU)

CURIA RATIONUM

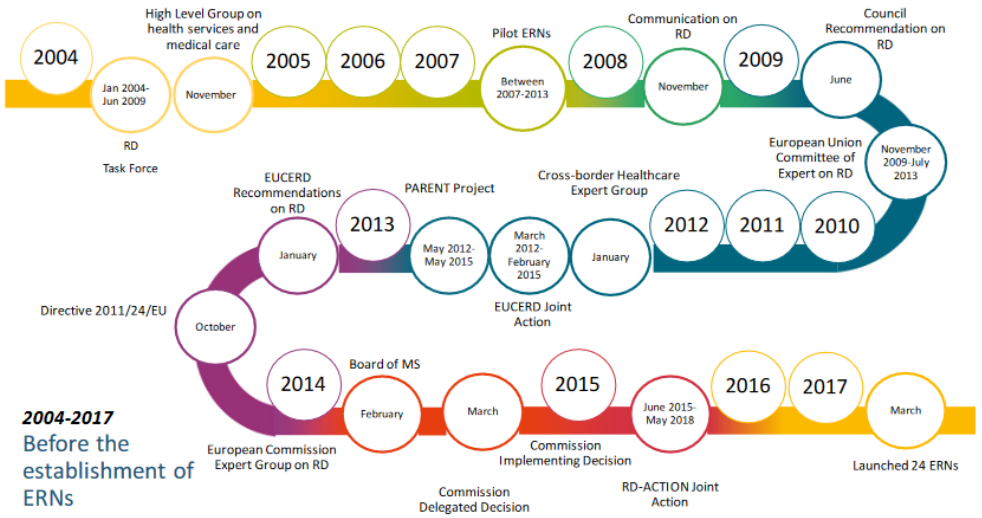
EUROPEAN COURT OF AUDITORS

Figure 3 – Distribution of healthcare provider members of European Reference Networks across the EU



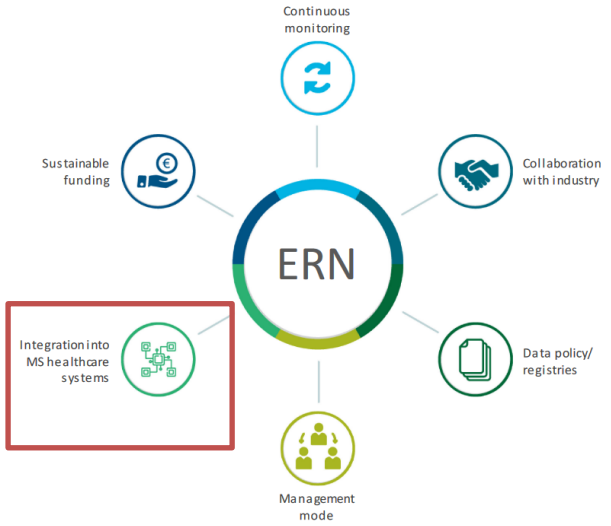
Source: ECA based on data on health care provider members to European Reference Network per Member State provided by the Commission, February 2019.

Figure 2 – Successive policy developments leading to the establishment of the European Reference Networks



Source: ECA.

Figure 5 – Challenges to the European Reference Networks development



Source: ECA, based on Board of Member States for European Reference Networks minutes.

https://www.eca.europa.eu/Lists/ECADocuments/SR19_07/SR_HEALTH_CARE_EN.pdf

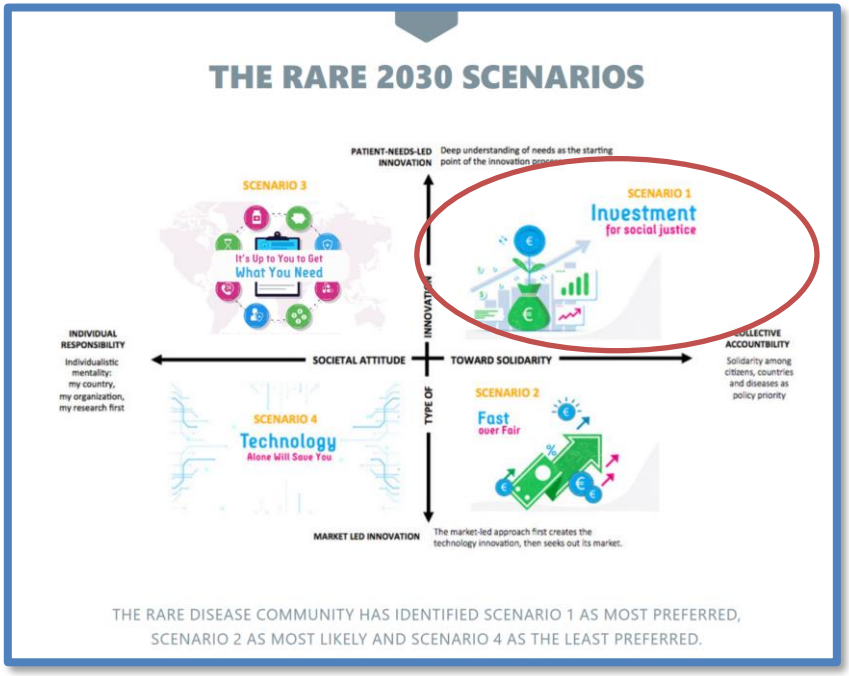
A broad renewed support from the rare disease community and EU institutions (1)

Rare 2030: a Foresight to look long term and a Roadmap for rare disease policy

- In **2021** after two years of multistakeholder collaborative foresight study**, 8 policy recommendations were adopted**

- In the field of diagnosis, access to healthcare, integrated care, partnerships with patients, research, data and treatments

	OVERARCHING TRENDS IN RARE DISEASES	RELEVANT BROADER TRENDS IN HEALTH AND HEALTHCARE	TYPE OF TREND
1	Rise of pan-European multi-stakeholder networks to advance diagnostics, treatment and care for rare diseases	Multi-stakeholders governance	POLITICAL
2	Strains on the health care budget and the emergence of new care delivery models	New healthcare delivery models	ECONOMIC
3	Greater variation in access to treatments and care resulting in more inequality across Europe for people with rare diseases	Access to medical products	ECONOMIC
4	Demographic change of RD patients introducing new challenges	Ageing population in a changing family structure	SOCIO-CULTURAL
5	Threats to solidarity equity, and the prioritization of rare diseases	Increase inequality and threats to solidarity	SOCIAL AND ECONOMIC
6	Increasingly empowered rare disease patient and the patient advocacy evolution	Advocacy evolution and patient empowerment	SOCIO-CULTURAL AND POLITICAL
7	Rise in innovation-oriented, multi-stakeholder, needs-led (patient-led) research	Innovation in Healthcare Research	SOCIO-CULTURAL AND POLITICAL
8	Facilitation of knowledge exchange and local care delivery through digital health	Digitization of healthcare	TECHNOLOGICAL
9	Increased potential for large sets of standardised and interoperable data	Big Data	TECHNOLOGICAL
10	Rise in the use of AI for diagnostics, treatment and care, opening-up the potential of 'big data'	Big Data and Artificial Intelligence	TECHNOLOGICAL
11	New technologies and advanced therapeutics	Innovation in Medical knowledge	TECHNOLOGICAL
12	Application of Whole Genome Sequencing from the research to the clinical sphere	Genomics	TECHNOLOGICAL, ETHICAL AND LEGAL



http://download2.eurordis.org/rare2030/Rare2030_recommendations.pdf

A broad renewed support from the rare disease community and EU institutions (2)

RARE 2030 RECOMMENDATION 1

LONG-TERM, INTEGRATED EUROPEAN AND NATIONAL PLANS AND STRATEGIES

A European policy framework for rare diseases defined by societal responsibility, equity and driven by the needs of people living with a rare disease should guide the implementation of consistent national plans and strategies, secure major investments at both the European level and by governments that are fairly shared across Europe in order to pool scarce resources, share expertise and information, scale-up good practices and provide access to timely and accurate diagnosis and the highest available quality of treatment and care for people living with a rare disease, no matter where they live in Europe. Both EU and national policies are supported by measurable outcomes that are monitored and assessed by a multistakeholder body on a regular basis.

RARE 2030 RECOMMENDATION 2

EARLIER, FASTER, MORE ACCURATE DIAGNOSIS

The time to diagnosis should be shortened - whilst avoiding erroneous and subsequent negative consequences - which should be achieved by better use and accessibility of currently effective and available diagnostic testing technologies, best practices and programmes. New technologies and innovative approaches must be driven by patient-needs and applied rapidly and strategically. Inequalities in access to diagnosis and ensuing care must be eradicated through the harmonisation of standards and programmes across Europe (and beyond). A particular focus is necessary for patients with undiagnosed rare diseases, which demands greater and more strategic global collaboration via data-sharing and diagnostic platforms and infrastructures.

RARE 2030 RECOMMENDATION 3

ACCESS TO HIGH QUALITY HEALTHCARE

Political, financial, operational and technical support at European, national and regional levels should be provided to establish a mature highly specialized healthcare ecosystem that, in collaboration with patient organizations and all relevant stakeholders, leaves no person living with a rare disease with uncertainty regarding their diagnosis, care and treatment.

RARE 2030 RECOMMENDATION 4

INTEGRATED AND PERSON-CENTRED CARE

Implement EU-wide and national actions by all stakeholders that guarantee the integration of people living with a rare disease in societies and economies, enabling them to live life to their full potential, by implementing innovative solutions and approaches to integrated and person-centred care along the full lifespan of people living with a rare disease.

EU-wide and national actions must be undertaken by all stakeholders to guarantee equal opportunities and access to the labour market, active support for employment, fair working conditions, social protection and inclusion and integrated and person-centred, long-term care for people living with a rare disease and their families.

RARE 2030 RECOMMENDATION 5

PARTNERSHIP WITH PATIENTS

An overall culture, reflected in policies and funding, that encourage the meaningful participation, engagement, involvement and leadership of people living with a rare disease in the research, care and development of diagnostic tools, treatments and innovative solutions to improve the health and social status, healthcare delivery, autonomy, quality of life and well-being of people living with a rare disease in Europe. Patient partnership should be encouraged in both the public and private sectors and people living with a rare disease and their representatives may often serve as a partnering link between the two.

RARE 2030 RECOMMENDATION 6

INNOVATIVE AND NEEDS-LED RESEARCH AND DEVELOPMENT

Maintain basic, clinical, social and translational research on rare diseases as a priority by increasing the funds for competitive and pre-competitive research, establishing greater incentives in more neglected areas (or in areas of high unmet needs), and supporting infrastructures required to expedite discovery and knowledge acquisition. Research in public health, social sciences, healthcare organisation, health economics and health policy research must also be promoted, to ensure that research outputs are applied for the benefit of people living with a rare disease.

RARE 2030 RECOMMENDATION 7

OPTIMISING DATA FOR PATIENT AND SOCIETAL BENEFIT

All European data sources of relevance to addressing the challenges faced by people living with a rare disease should be federated in a continuum encompassing epidemiological, healthcare, research, quality of life and treatment-related data, and should be linked at the global level where possible. Sharing of data for care and research should be optimised across infrastructures and countries, relying upon commonly adopted codification systems (Orphanet nomenclature), harmonised standards and interoperability requirements. Cohesive data ecosystems should be developed at national level, linking seamlessly via Findable, Accessible, Interoperable and Reusable (FAIR) data approaches to an integrated European ecosystem, positioned within the European Health Data Space and centred on robust European Reference Networks (ERNs), the European Platform on Rare Disease Registration, and other key infrastructures. Legal and ethical guidelines and regulations should incentivise practices that best lead to addressing these challenges while respecting international, national and regional laws and conventions – particularly the preferences and privacy of people living with a rare disease and their families.

RARE 2030 RECOMMENDATION 8

AVAILABLE, ACCESSIBLE AND AFFORDABLE TREATMENTS

Establish streamlined regulatory, pricing and reimbursement policies. These policies should encourage a continuum of evidence generation along the full life cycle of a product or technology as well as the patient journey from diagnosis to treatment access. A European ecosystem able to attract investment in areas of unmet need, foster innovation, and address the challenges of healthcare system sustainability.

http://download2.eurordis.org/rare2030/Rare2030_recommendations.pdf

A wide renewed support from the rare disease community and EU institutions (3)

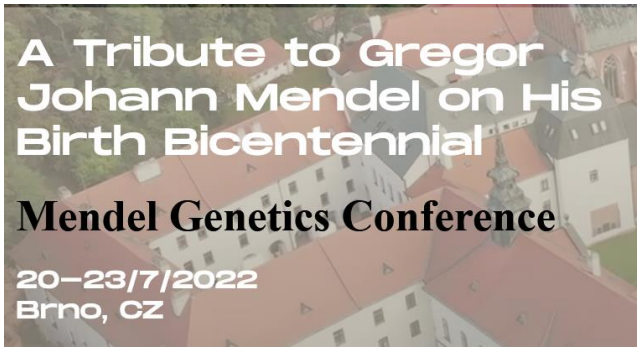
France EU Presidency High-Level Conference on Rare Diseases (28/2/2022)

European Parliament – debate and letter to the EC ((2021/2940(RSP))

European and Economic Social Committee - Opinion on Rare Diseases (2022)

- Recognition of **Rare 2030 Recommendations** and Ministerial level attendance
- Key messages clearly supported by all stakeholders
- Follow up actions to strengthen French launching of a **call for an Action Plan**

https://eur-lex.europa.eu/legal-content/EN/TXT/PDF/?uri=CELEX:52022IE2323&from=EN
https://www.europarl.europa.eu/doceo/document/CRE-9-2021-11-24-ITM-016_EN.html



novorozenecký screening

SESSION I | SESSION II | PANEL DISCUSION | GENERAL DISCUSSION | SUMMARY |

EARLY DIAGNOSIS OF PATIENTS WITH RARE DISORDERS IN THE EU: CRUCIAL ROLE OF THE NEWBORN SCREENING

Skip to videos

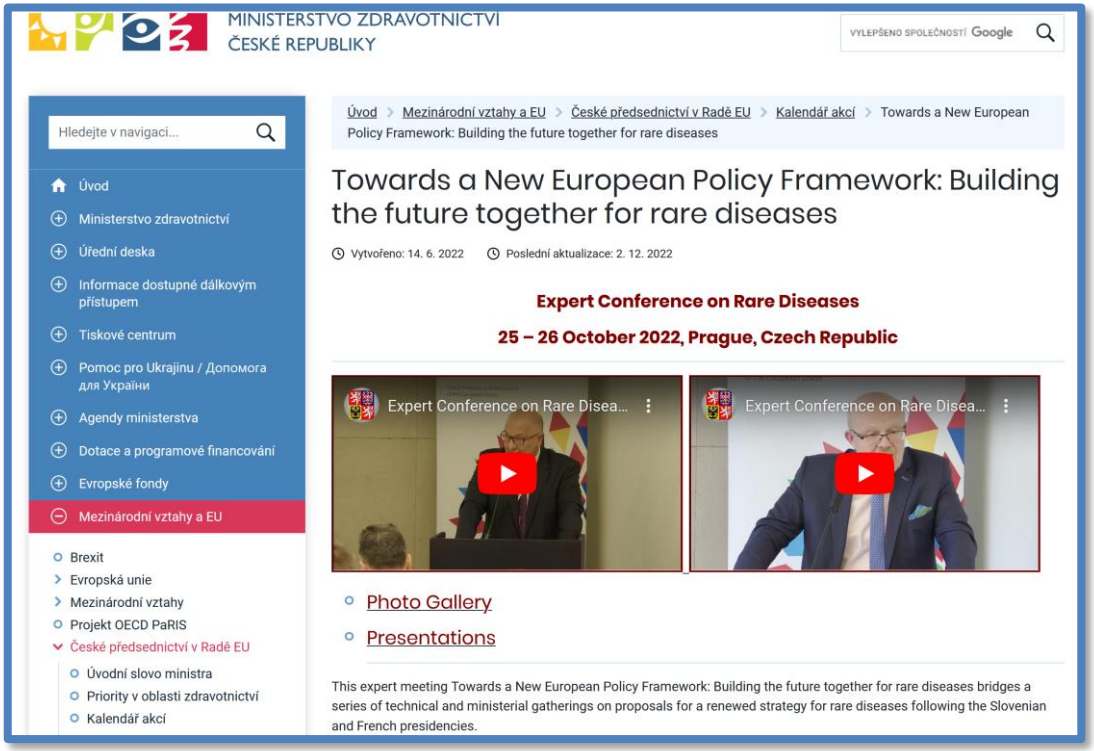
Technical meeting under the auspices of the Presidency of the Czech Republic in the Council of the EU. Brno, Czech Republic, July 23, 2022.

Satellite meeting to the Celebrations of 200th Anniversary of G. J. Mendel‘ s birth July 20-23, 2022 (www.mendel22.cz)
Date of satellite meeting: Saturday July 23, 2022, 13:15- 18:00, Brno, Czech Republic
Venue of satellite meeting: Mendel Museum at the Augustinian Abbey, Brno (mendelmuseum.muni.cz/en)

https://novorozeneckyscreening.vzacna-onemocneni.cz/

Expert conference on rare diseases Prague, October 25-26 / 2022

- Czech EU Council Presidency event
- 110 delegates, including 3 invitees from each MS
- Speakers incl. EC representatives, ERN representatives, Patient Advocates, Industry
- Agenda split into **five blocks**:
  - A new goal-based and coordinated strategy for rare diseases
  - Early Diagnosis of Rare Diseases
  - Revision of the Orphan Drug and Paediatric Drug Regulations
  - Instruments for improving access to treatments for rare diseases
  - **Holistic healthcare pathways**: Integrating European Reference Networks into European healthcare and social systems



https://www.mzcr.cz/towards-a-new-european-policy-framework-building-the-future-together-for-rare-diseases/

What did the conference cover?

- Improving access to treatments
- Early Diagnosis
- Coordinated European Strategy on Rare Diseases
- Revision of the OMP & Paediatric Regulations
- Holistic care through ERNs

Programme

Tuesday, 25. 10. 2022

Time	Programme	Speaker
13:00 – 14:00	**Registration and welcome coffee**	
14:00 – 14:10	Welcome remarks from the CZPRES	Mr. Vlastimil Válek Deputy Prime Minister and Minister of Health of the Czech Republic
14:10 – 14:20	Remarks from the European Commission	Mr. Andrzej Rys Principal Scientific Adviser, EC
14:20 – 14:30	Remarks from the previous French Presidency	Ms. Anne Sophie Lapointe Ministry for Solidarity and Health, France
14:30 – 14:40	Remarks from the upcoming Swedish Presidency	Mr. Thomas Linden Government Chief Medical Officer, Sweden
14:40 – 16:00	**SESSION I.** **A NEW GOAL-BASED AND COORDINATED STRATEGY FOR RARE DISEASES** (Moderator Ms. Anna Arellanesová)	
14:40 – 15:15	**Series of keynote presentations**	
14:40	Presentation of the Call to Action and proposal for the European Action Plan for Rare Diseases	Mr. Jakub Dvořáček Deputy Minister, Ministry of Health, Czech Republic
14:50	Investing in Rare Diseases Research: a European long-standing commitment	Ms. Irene Norstedt Director "People", Directorate-General for Research and Innovation, European Commission
14:55	Why Europe's Action Plan for Rare Diseases is critical for our community	Mr. Yann le Cam CEO, Eurordis-Rare Diseases Europe
15:15 – 16:00	**Moderated panel discussion:** **Working together with rare disease patient representatives** (Moderator Ms. Anna Arellanesová)	
	Patient life-journey, need for holistic view	Ms. Adéla Odrihocká Rare Diseases Czech Republic
	Patient life-journey, need for holistic view	Mr. Anders Olauson President of Agrenska, and Chairman of "RareResourceNet" a European Network of Rare Diseases Resource Centers
	Patient advocacy – in the field of rare disorders in Norway	Ms. Lisen J. Mohr Representative of the Norwegian rare disease resource centre, Frambu.no
16:00 – 16:30	**Coffee break**	
16:30 – 18:00	**SESSION II.** **EARLY DIAGNOSIS FOR RARE DISEASES** (Moderator Mr. Milan Macek)	
16:30 – 17:15	**Series of keynote presentations**	
16:30	Importance of neonatal Screening for the early diagnosis of rare diseases	Mr. Maurizio Scarpa Coordinator MetabERN and Representative of Screen4Rare
16:45	The Rare Diseases Partnership – improving R&I potential and accelerating clinical trial readiness of the rare diseases	Ms. Daria Julkowska Scientific Coordinator of the European Joint Programme on Rare Diseases
17:00	Orphanet contribution to improving rare disease diagnostic path	Ms. Ana Rath Orphanet Director, France
17:15 – 18:00	**Moderated panel discussion:** **Early diagnosis: from newborn screening to personalized patient care** (Moderator: Mr. Viktor Kožich)	
	IVDR 2017/746: quite a challenge for new tests for rare diseases to preserve the final purpose of the regulation. Call for embedding an incubation period	Ms. Elisabeth Dequeker Representative of European Society of Human Genetics
	Key indicators of newborn screening: International context and future perspectives for cooperation	Mr. Ondřej Májek Representative of the National Screening Center of the Institute of Health Information and Statistics
	Newborn dried blood spot screening and follow up in Sweden	Mr. Rolf Zetterström Representative of Karolinska University Hospital
18:00 – 18:15	**CONCLUSION – DAY 1**	
18:00	Summary – Session I. and II.	Mr. Milan Macek National Coordination Centre for Rare Diseases UH Motol and Charles University
18:10	Closing remarks – Day 1	Mr. Jakub Dvořáček Deputy Minister, Ministry of Health of the Czech Republic
19:30	**Evening reception (Corinthia Hotel Prague, Bellevue Hall, 24th floor))**	

Wednesday, 26. 10. 2022

Time	Programme	Speaker
09:00 – 10:30	**SESSION III.** **REVISION OF THE ORPHAN DRUG AND PAEDIATRIC DRUG REGULATIONS** (Moderator: Mr. Tomáš Mlčoch)	
09:00 – 09:20	**Series of keynote presentations**	
09:00		Ms. Olga Solomon Head of Unit Medicines: Policy, Authorisation and Monitoring, DG SANTÉ
09:10	Evidence for orphan and paediatric medicines – challenges and opportunities	Mr. Michael Berntgen Head of Scientific Evidence Generation Department European Medicines Agency
09:20 – 10:30	**Moderated panel discussion:** **Improved provision of innovative medicines to rare diseases patients** (Moderator: Ms. Kateřina Kopečková)	
	How to make treatments for rare diseases less rare	Ms. Nathalie Moll Representative of the EFPIA
	The OMP Regulation review – developers perspective and reflections on the way ahead	Ms. Vittoria Carraro Representative of Orphan Drug Incentives / EUCOPE
	The revision of the OMP regulation – perspectives from the Rare Disease Community	Ms. Virginie Hivert Representative of the EURORDIS-Rare Diseases Europe
10:30 – 11:00	**Coffee break**	
11:00 – 12:00	**SESSION IV.** **INSTRUMENTS FOR IMPROVING ACCESS TO RARE DISEASES TREATMENTS** (Moderator: Mr. Jakub Dvořáček)	
11:00 – 11:30	**Series of keynote presentations**	
11:05	Access to therapies: how to address the systemic failures with innovative tools	Mr. Yann Le Cam Chief Executive Officer EURORDIS-Rare Diseases Europe
11:15	United Action for Better Health; leave no-one behind	Ms. Sarah Garner World Health Organization
11:30 – 12:00	**Moderated panel discussion:** **Improving access to diagnostics and treatment in rare diseases** (Moderator: Mr. Jakub Dvořáček)	
	Pan-continental ERN based data sharing for solving the unsolved RD in Europe	Mr. Holm Graessner ERN-RND, Center for Rare Diseases, Tübingen
		Mr. Declan Noone Representative of the European Haemophilia Consortium
	Improving access to diagnostics and treatment in rare diseases	Ms. Anna Bucsics Representative of the Mechanism of Coordinated Access to Orphan Medicinal Products
12:00 – 13:00	**Buffet lunch**	
13:00 – 14:20	**SESSION V.** **HOLISTIC HEALTHCARE PATHWAYS: INTEGRATING ERN INTO EUROPEAN HEALTH CARE AND SOCIAL SYSTEMS** (Moderator: Ms. Pavla Doležalová)	
13:00 – 13:30	**Series of keynote presentations**	
13:00	The European Reference Networks at the service of national EU health networks for Rare Diseases	Mr. Alexis Arzimanoglou Coordinator of the European Reference Network for Rare and Complex Epilepsies (EpiCARE), Chair of the ERN Coordinators Working Group
13:10	European Reference Networks: towards equity in rare diseases	Ms. Birutė Tumienė Head of Unit, Center for Medical Genetics, Vilnius University Hospital
13:20	The future Joint Action on Integration: one key stimulus towards multifaceted and holistic healthcare pathways for Rare Diseases in Europe	Mr. Till Voigtländer Co-chair of the Board of Member States for ERNs
13:30 – 14:20	**Moderated panel discussion:** **European Reference Networks for rare diseases as key hubs of research and medical / social care for rare diseases** (Moderator: Ms. Pavla Doležalová)	
	Improving diagnosis for Rare Diseases in Europe – impact of European Reference Networks	Mr. Holm Graessner ERN-RND, Center for Rare Diseases, Tübingen
		Mr. Maurizio Scarpa Coordinator MetabERN and Representative of Screen4Rare
		Ms. Anne Sophie Lapointe Ministry for Solidarity and Health, France

Key outcome: A Call to Action

To support the **early diagnosis** of people living with a rare disease

To adopt a **European Action Plan on Rare Diseases** to support and complement ongoing and future efforts at both the EU and Member State level

To evolve the incentives framework to maintain predictability for sponsors while enhancing Europe's competitiveness through the upcoming **revision of the OMP and Paediatrics Regulations**.

To **improve access to treatments**, including further strengthening European cooperation in pricing and negotiations, while respecting current division of competences.

Call to Action

The Czech Presidency of the EU Council organised the Expert Conference on Rare Diseases in Prague on 25-26 October 2022 to explore how the European Union can take continued steps towards a coordinated strategy for rare diseases to better addresses current unmet needs by setting meaningful goals for patients, families and for society at large, integrated at the national and regional levels.

Rare diseases, including rare cancers, are a heterogeneous group of largely incurable, complex conditions. There are over 6000 rare diseases, and more than 70% have a genetic origin. Although individually characterised by low prevalence, the sheer number of rare diseases results in a directly affected community of 20 million people across the EU. Rare diseases are chronic, progressive, degenerative, disabling and frequently life threatening. They are typically accompanied by a scarcity of knowledge and expertise.

In 2021, an average of 5 years is still needed to obtain a diagnosis, and only 6% of rare diseases can benefit from a specialized treatment. People living with a rare disease experience a high psychosocial, emotional and financial burden and are often excluded from society. The COVID-19 pandemic has exacerbated their vulnerabilities, with 84% of people living with a rare disease in Europe having experienced disruptions to their care during this period. Scarcity and scattered nature of data and expertise single out rare diseases as an area of very high added community value, demanding interdisciplinary as well as cross-border collaboration in terms of sharing knowledge, data, and research.

Despite tremendous progress demonstrated by the measures already implemented, the ongoing commitments and major investments in addressing the challenges of rare diseases from the side of the European Commission, the need for an updated framework of EU actions and support for national plans and strategies on rare diseases remains. The 2008 Communication on *Rare Diseases: Europe's challenge*, which aimed to "encourage cooperation between the Member States and set out an overall strategy for support to Member States", was a cornerstone policy for today's progress, but drafted in an era during which scientific breakthroughs, technological potential and crisis and values were not the same as today.

A Conference focused on strengthened European collaboration on rare diseases

Participants, patient advocates, healthcare professionals, researchers, government representatives and industry set out how strengthened cooperation and coordination of Member States could be outlined in a European Action Plan for Rare Diseases. By bringing together current initiatives under one framework that would provide a roadmap leading towards common measurable goals that respond directly to unmet needs and ensure that inequalities are not exacerbated by a person's country of residence.

The Conference marked another significant milestone in the proposal for a policy framework for rare diseases following the conclusions of the European Court of Auditor's report n°7/2019, the cross-sector consensus from over 250 stakeholders in the EU spearheaded Rare 2030 Foresight Study, the 43 cosignatory members of the European Parliament in their letter of support for Europe's Action Plan, the recognition of the "undeniable benefit" of stronger cooperation during the Informal meeting of Ministers of Health in Grenoble earlier this year and the support of patients, key opinion leaders and policy makers presented at the High Level Ministerial Conference: 'Care and Innovation pathways for an EU policy on rare diseases' (28 February 2022) in support of the proposal for a European Action Plan on Rare Diseases.

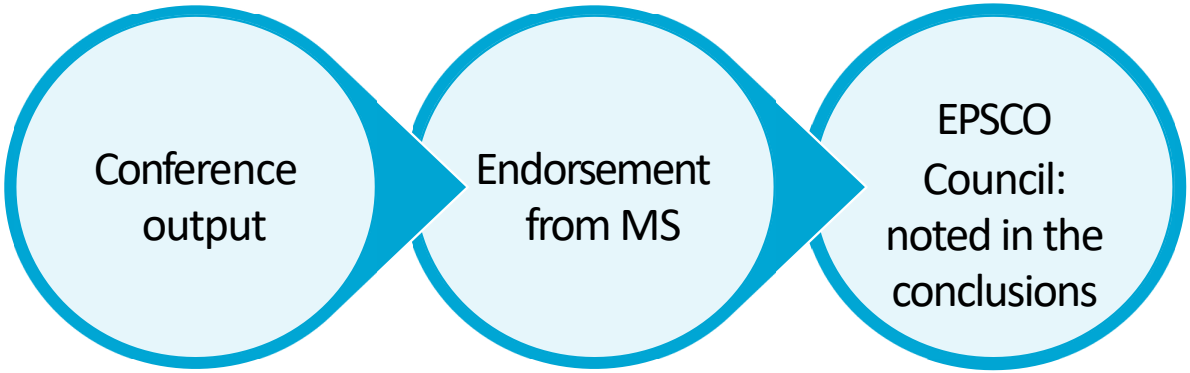
In line with the political support and increased momentum for a stronger European approach to rare diseases, the Conference participants explored what meaningful steps could be taken by the European institutions and Member States to improve the lives of people living with a rare disease.

The Expert Conference on Rare Diseases focused on five blocks that make up key pillars of a European strategy on rare diseases and led to five key recommendations:

On **holistic care & integrating European Reference Networks** into national health systems

https://www.mzcr.cz/wp-content/uploads/2022/12/CZPRES_Expert_Conference_on_Rare_Diseases_brochure.pdf

22 EU Member States expressed support to Czech Presidency Call to Action (2022)



25 – 26 October — **9 December**

Czechia
Austria
Croatia
Cyprus
Denmark
Estonia
Finland
France
Germany
Greece
Ireland
Italy
Latvia
Lithuania
Luxembourg
Malta
Poland
Portugal
Romania
Slovakia
Slovenia
Spain

A call for a European Action Plan on rare diseases

- Early diagnosis
- Revision of OMP Legislation
- Instruments for improving access to treatments for rare diseases
- Integrating European Reference Networks into European health care and social systems

> *This Call to Action reinforces the urgency and will of Member States to strengthen collaboration on rare diseases at the European level. It is a major step forward, with Czechia establishing a clear stand on how a European Action Plan on Rare Diseases will lead our health union.*
>
> YANN LE CAM, EURORDIS CHIEF EXECUTIVE OFFICER

https://www.eurordis.org/ms-endorse-czech-cta-rare-diseases/

Qualified majority supports Call to Action (2022)

Voting calculator

Countries participating	Votes
Austria 1.99% of population	✓
Belgium 2.58% of population	✓
Bulgaria 1.55% of population	✗
Croatia 0.90% of population	✓
Cyprus 0.20% of population	✓
Czech Republic 2.36% of population	✓
Denmark 1.30% of population	✓
Estonia 0.30% of population	✓
Finland 1.24% of population	✓
France 15.07% of population	✓
Germany 18.57% of population	✓
Greece 2.39% of population	✓
Hungary 2.17% of population	✗
Ireland 1.12% of population	✓
Italy 13.38% of population	✓
Latvia 0.42% of population	✓
Lithuania 0.62% of population	✓
Luxembourg 0.14% of population	✓
Malta 0.12% of population	✓
Netherlands 3.94% of population	✗
Poland 8.45% of population	✗
Portugal 2.30% of population	✓
Romania 4.29% of population	✓
Slovakia 1.22% of population	✓
Slovenia 0.47% of population	✓
Spain 10.59% of population	✓
Sweden 2.32% of population	✗

Voting rule: Qualified majority

Final result

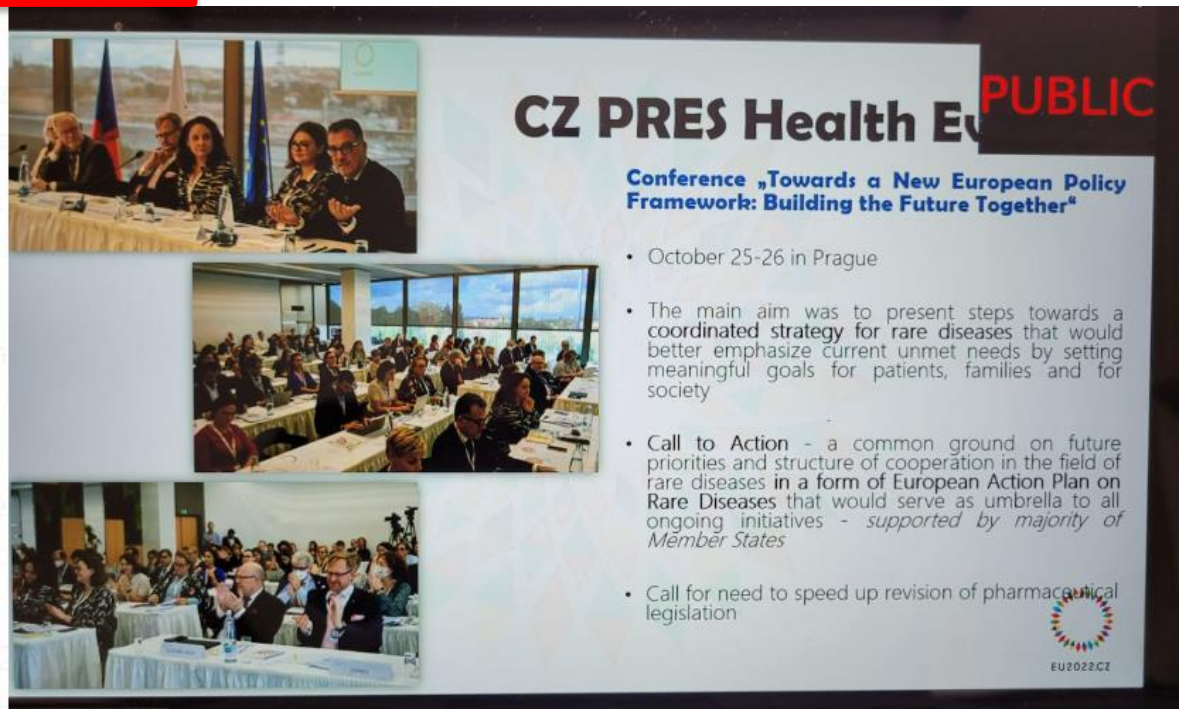
Approved

27 **member states**
Minimum "Yes" required for adoption: (55%) 15

22	5	0
Yes	No	Abstain

% **Population**
Minimum "Yes" required for adoption: 65%

81.57	18.43	0.00
Yes	No	Abstain



I) A CALL FOR A EUROPEAN ACTION PLAN

- Call upon the European Commission to adopt a **coordinated EU Strategy on rare diseases** to support and complement ongoing and future efforts at both the EU and Member State levels.
- This should take the shape of a Commission Communication on addressing the challenges of persons living with rare diseases.
- Bringing together existing legislation towards common goals
- Integrating and sustaining European and national plans and strategies for rare diseases on a long-term basis;
- Measurable and time-bound goals
- Space to innovate

II) EARLY DIAGNOSIS

- Share best practice and lessons from national **newborn screening (NBS) programmes**;
- Collect, collate and develop key performance indicators to improve the quality of NBS programmes;
- Create an **EU-level NBS Expert Advisory Committee**, free from bias or national interests, to provide trusted, high-quality information to support decision-making at a national level.
- Accessibility of medical devices necessary for diagnostics of rare diseases is of crucial importance. **Regulation (EU) 2017/746 on *in vitro* diagnostic medical devices (IVD-R)** sets several ways for derogation from the generally applicable rules for safety and performance requirements when placing medical devices on the market. Nevertheless, the further specification that would ensure that in vitro medical devices necessary for proper diagnosis of rare diseases remain available on the market is needed.

III) REVISION OF THE ORPHAN DRUG AND PAEDIATRIC DRUG REGULATIONS

- A model that is centred on the unmet needs of people living with a rare disease, and includes patient participation in its establishment and implementation;
- Transform the **European Research & Development for the rare disease ecosystem building upon advances of the past 20 years,** for the next 20 years;
- Situate Europe as a global leader in research, development and access, through a regulation that is attractive and competitive globally;
- Establish a European pathway, from development to access, to ensure innovation coupled with affordability and to gain that crucial strategic autonomy in research and development;
- Ensure convergence and coherence between different relevant legislation.

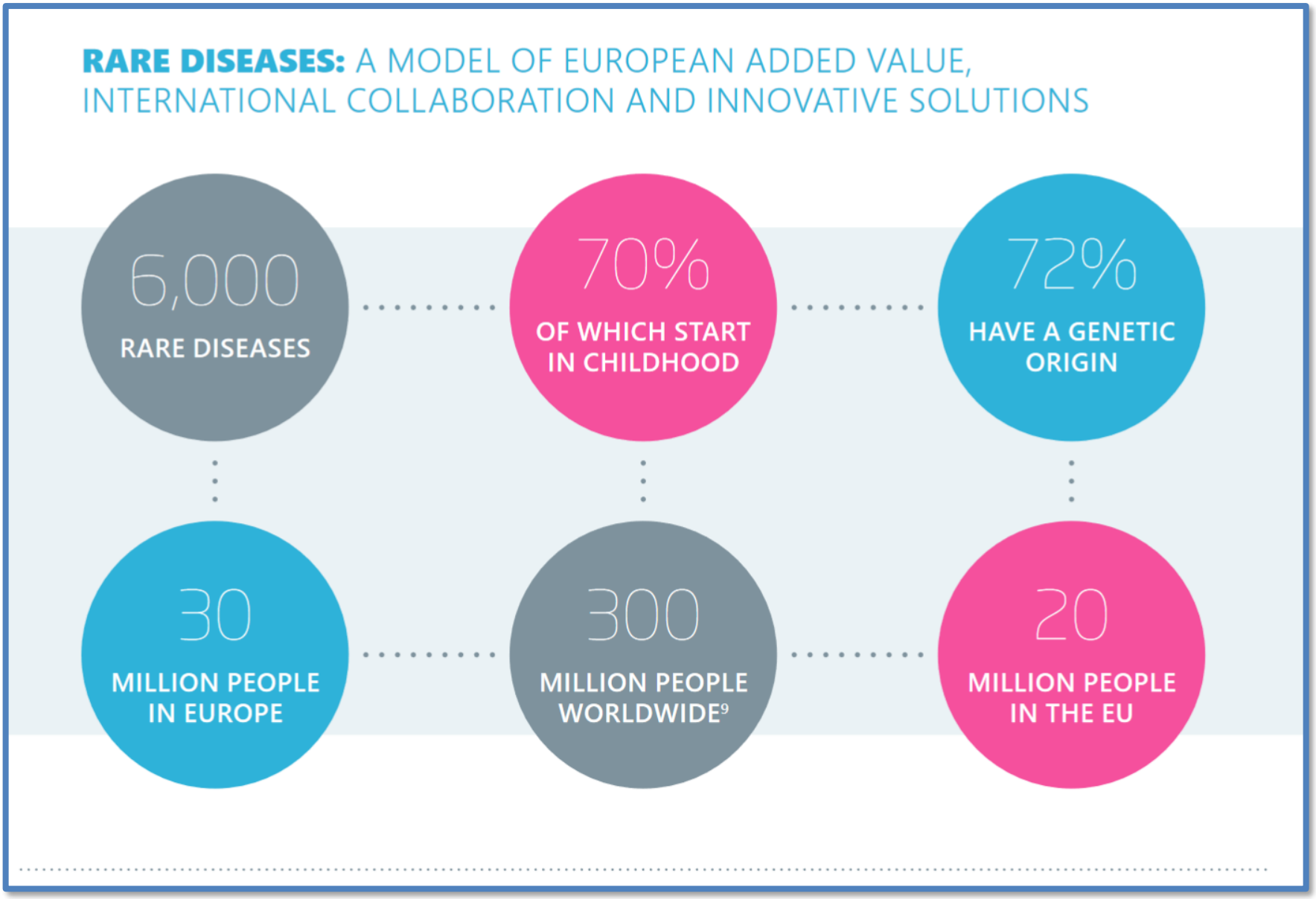
V) INSTRUMENTS FOR IMPROVING ACCESS TO TREATMENTS FOR RARE DISEASES

- **Strengthening European cooperation in pricing and negotiations: mechanisms of voluntary cooperation between EU Member States**
- European pathway, from development to access, to ensure innovation is coupled with affordability and to gain that crucial strategic autonomy in research and development, starting with very low prevalence diseases and complex treatments.
- Explore the feasibility of **piloting cross-country mechanisms to improve best practices and information exchanges, value assessments, demand pooling and negotiating and purchasing models, as stated by WHO Europe**
- Consider the initiation of action towards the creation of a "European Fund" to support the generation of evidence across the whole life cycle of products, focusing on products for very small populations and/or complex treatments, such as **Advanced Therapeutic Medicinal Products (ATMPs)**, for which evidence at time of pricing and reimbursement (P&R) is often immature.
- Explore opportunities for joint negotiations with producers of complex treatments and treatments for small populations that have the potential to improve accessibility of treatment across the EU, in a way that could possibly be incorporated into the revision **of Orphan Drug and Paediatric Drug Regulations as regulatory incentive.**

Why a European Action Plan for Rare Diseases

- ✓ There is a need for a **comprehensive policy framework to connect all policies and initiatives** affecting people living with a rare disease at European and national levels
- ✓ This would **bring areas such as digital to data to research to diagnosis to treatments to social care to ERNs under one umbrella framework**
- ✓ It would **drive innovation** to make the EU a competitive world leader on innovative therapies and technologies
- ✓ It would promote a **European model of care** for persons living with a rare disease. For the first time this would be a strategy that looks beyond health, by addressing the whole spectrum of challenges faced by people living with a rare disease in Europe, in line with the *UN Resolution Addressing the Challenges of Persons Living with a Rare Disease*
- ✓ This would also **introduce measurable goals**, similar to those in obesity or cancer and aligned with the SDGs for all countries to work towards

Rare diseases are forerunners of modern – personalised (stratified/precision) medicine



http://download2.eurordis.org/rare2030/Rare2030_recommendations.pdf

Thank you for your attention!

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