Expert Conference on Rare Diseases

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

25 – 26 October 2022, Prague, Czech Republic

MINISTRY OF HEALTH
OF THE CZECH REPUBLIC

EU2022.CZ

Vlastimil Válek
Minister of Health

Dear colleagues,
ladies and gentlemen,

It is my great pleasure and honour that the most esteemed European experts on rare diseases and stakeholders in this area accepted our invitation to Prague for an Expert Conference on Rare Diseases: *"Towards a New European Policy Framework: Building the future together for rare diseases"*.
You are opening a brochure that will guide you through your stay in Prague to make it as pleasant as possible. On the following pages, you will find useful information that will help you to get acquainted with the Conference, its program, and our honoured speakers. It will also provide you with some practical advice.

The conference is taking place at the Vyšehrad quarter of Prague, the capital of the Czech Republic and a jewel of our country. It is also sometimes called the City of Hundred Spires or the Mother of Cities for its unmatched beauty and historical panorama. Vyšehrad quarter is not only the best place to observe this magnificent panorama, but also the historical seat of some of the Bohemian kings. The beautiful historical quarter is now a National Heritage site and lies only minutes from the Conference venue.
The second expert conference of the Czech Presidency of the Council of the EU in the field of health follows the success of the Expert conference on oncology that took place in Brno. Besides oncology, the Czech Presidency sees addressing the challenges of rare diseases in Europe as one of the priorities of today´s healthcare. Every patient in Europe should be helped and even the most vulnerable and those suffering from rare diseases have a right to be heard and treated. As such, we believe the problematic of rare diseases deserves the attention of the Member States just as the European institutions. The following conference brings together some of the most renowned names in the field and allows them to present their views alongside the political representation of the Member States and the EU along with other stakeholders.
Building on a series of technical and ministerial gatherings on proposals for a renewed strategy for rare diseases following the Slovenian and French presidencies, the Conference will try to continue discussions on rare diseases and focus on how the Czech EU Council Presidency can take further steps towards a coordinated strategy for rare diseases that better address current unmet needs by setting meaningful goals for patients, families and society at large integrated at the national and regional levels. Such needs include, among others, an updated framework of EU actions and support for national plans and strategies on rare diseases.
The EU is a key actor in this regard and has a central role in creating an environment conducive to strengthening the cooperation and coordination of Member States concerning rare diseases. Such measures could be outlined in a European Action Plan for Rare Diseases. The ambition of the Czech Presidency is to further reinforce EU efforts to create such an Action Plan and support non-legislative policy actions and initiatives dealing with rare diseases. However, these ambitions can only be turned into reality if they are shared by all the stakeholders who contribute to a general discussion and act with unity and determination 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 only allows for partial coverage of all the important topics we have to consider when dealing with rare diseases. Early diagnosis remains one of the most effective ways of fighting rare diseases, and the better use and

accessibility of currently effective and available diagnostic tools and technologies, best practices and programmes should be considered as part of a continuum of health strategies along the life course of any person living with a rare disease in Europe. Improvements and accelerated patients' access to safe, effective, and affordable medicines and support innovation in the EU pharmaceutical sector is another priority with regard to rare diseases. Solving these issues will have a remarkable impact on the lives of those who suffer from rare diseases and this Conference will hopefully help to further the discussion and allow all of you to move forward in the right direction.

Ladies and gentlemen, I am sure that this Conference will elucidate the paths experts and stakeholders need to take to reach the desired goal as well as pinpoint measures to tackle the problems of effective treatment of Rare Diseases.

I wish you a fruitful Conference and discussion as we believe the future of the Rare Diseases policy is being set here, with your help. You are the frontrunners of the discussion, and your expert voices have the power to shape this Conference into a resounding success, all for the benefit of European patients.

Scope and purpose

This expert meeting Towards a New European Policy Framework: Building the future together for rare diseases bridges a series of technical and ministerial gatherings on proposals for a renewed strategy for rare diseases following the Slovenian and French presidencies.

In the spirit of continuity stakeholders in the rare disease community are particularly keen to continue discussions on rare diseases and focus on how the Czech EU Council Presidency can 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.

Despite tremendous progress demonstrated by the measures already implemented and 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[1] which aimed to "encourage cooperation between the Member States and set out an overall Community strategy for support to Member States" was a cornerstone legislation for today's progress, but drafted in an era during which scientific breakthroughs, technological potential and Community crisis and values were not the same as today.

Today's strategy must strengthen the cooperation and coordination of Member States with regards to rare diseases by combining the critical building blocks discussed during this conference that make up the rare disease ecosystem along the patient journey (diagnosis, centres of expertise, treatment, and social care) and the life cycle of innovative solutions from "bench to bedside".

Strengthened cooperation and coordination of Member States could be outlined in an European Action Plan for Rare Diseases that addresses remaining gaps by combining and supplementing current legislative actions (e.g. revisions of legislation of Orphan Medicinal Products Regulation, Paediatric Use of Medicines and adoption of the Pharmaceutical Strategy)[2]; non-legislative policy actions; and new Commission initiatives such as the European Health Data Space[3]

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[1] https://ec.europa.eu/health/non-communicable-diseases/steering-group/rare-diseases_cs

[2] https://ec.europa.eu/health/medicinal-products/medicines-children/evaluation-medicines-rare-diseases-and-children-legislation_en

[3] https://www.europarl.europa.eu/legislative-train/theme-promoting-our-european-way-of-life/file-european-health-data-space

and the European Pillar of Social Rights Action Plan[4].

A European Action Plan for Rare Diseases would provide a road map for all European countries to work towards common measurable goals that respond directly to unmet needs and ensure that inequalities are not exacerbated by a person's country of residence:

1. **improved health outcomes (via reduced delays in diagnosis)**
2. **reduced inequalities and**
3. **increased innovation**

This conference will mark a major 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[5], the cross-sector consensus from over 250 stakeholders in the Rare 2030 Foresight Study[6], the 43 cosignatory members of the European Parliament in their letter of support for Europe's Action Plan[7], the recognition of the "undeniable benefit" of stronger cooperation during the Informal meeting of Ministers of Health in Grenoble[8] and the support of thousands of patients and key opinion leaders and policy makers presented at the High Level Ministerial Conference: Care and innovation pathways for an EU policy on rare diseases[9] in support of the proposal for a European Action Plan on Rare Diseases.

Early diagnosis (via preconception carrier testing and newborn screening) can significantly decrease mortality and morbidity caused by rare diseases and improve quality of life by expediting access to the most effective care and treatments. Yet the quest for diagnosis often remains an odyssey. Better use and accessibility of current effective and available diagnostic tools and technologies, best practices and programmes should be considered as part of a continuum of health strategies along the life course of any person living with a rare disease in Europe.

Despite the significant advances in the care of rare disease patients afforded by the Orphan and Paediatric Drug Regulations, many people living with a rare disease still do not benefit from timely access to medicines. The European Commission's revision to these legislations as a part of the EU pharmaceutical strategy will introduce improvements and accelerate patients' access to safe, effective, and affordable medicines and to support innovation in the EU pharmaceutical sector.

The revision of the Orphan drug and paediatric drug regulations will only address part of the unmet medical needs faced by the 30 million people living with a rare disease in Europe. A new ecosystem in Europe that ensures sustainability for healthcare systems could include additional solutions to address a persisting lack of treatments and inequalities between countries in Europe, such as a common European fund to support the generation of evidence across the whole life cycle of products and to ensure attractiveness of the European R&D ecosystem for rare diseases and consolidated and structured cooperation in Europe on pricing and negotiations.

With the majority of rare disease patients not having a dedicated treatment for their disease on the market, non-pharmaceutical care (both clinical and social) become central to a patient's survival and quality of life. Accessing this care quickly no matter where one lives in Europe is essential to a comprehensive strategy on rare diseases.

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[4] https://ec.europa.eu/info/strategy/priorities-2019-2024/economy-works-people/jobs-growth-and-investment/european-pillar-social-rights/european-pillar-social-rights-action-plan_en

[5] https://op.europa.eu/webpub/eca/special-reports/cross-border-health-care-7-2019/en/

[6] https://www.rare2030.eu/

[7] https://www.europarl.europa.eu/doceo/document/O-9-2021-000069_EN.html

[8] https://presidence-francaise.consilium.europa.eu/media/44cf1u01/ven-epsco-sante-dp.pdf

[9] https://presidence-francaise.consilium.europa.eu/en/news/ministerial-conference-care-and-innovation-pathways-for-an-eu-policy-on-rare-diseases/

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

Time	Programme	Speaker
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

Time	Programme	Speaker
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**	

Time	Programme	Speaker
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**	

Time	Programme	Speaker
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

Time	Programme	Speaker
14:20 – 14:45	**CONCLUSION – DAY 2**	
14:20	Summary of the discussion from Session III. to V.	Mr. Yann Le Cam CEO, EURORDIS-Rare Diseases Europe
14:30	Call to Action – Invitation to support	Mr. Jakub Dvořáček Deputy Minister, Ministry of Health of the Czech Republic
14:35	Closing remarks	Mr. Vlastimil Válek Deputy Prime Minister and Minister of Health of the Czech Republic

Keynote speakers and panelists (in alphabetical order)

Ms. Anna Arellanesová

(Session I.)

is the chairperson of Rare Diseases Czech Republic, a Czech patient-led umbrella association for rare diseases. She is also a member of the board and former chairperson of the Czech Cystic Fibrosis Association and board member of Eurordis – Rare Diseases Europe. Being in the role of a vice-chair of the Patient Council of the Ministry of Health, where she represents rare diseases, she was able to participate in the creation of new legislation for an orphan drug reimbursement approval system, one which counts on the active participation of patients as decision makers. She is also a member of the working group for rare diseases at the Czech Ministry of Health. Thanks to a long-term cooperation with experts, she prepared and fulfilled the National Strategy for Rare Diseases as well as National Action Plans for Rare Diseases.

She has a bachelor's degree in humanities from Charles University and lives in Prague.

Mr. Alexis Arzimanoglou

(Session V.)

is the Coordinator of the European Reference Network for Rare and Complex Epilepsies ERN EpiCARE, currently also chairing the 24 ERN Coordinators Group. He is a Child Neurologist, Director of the Paediatric Clinical Epileptology, Sleep Disorders and Functional Neurology Dpt., University Hospitals of Lyon, France and Scientific Coordinator of the Epilepsy Unit at San Juan de Déu Hospital, Universitat de Barcelona, Spain. Elected member of the current ILAE-Europe Commission, Editor-in-Chief Emeritus of the ILAE journal *Epileptic Disorders* and Associate Editor of the *European Journal of Paediatric Neurology*. He received the EPNS Aicardi Award for Excellence in Paediatric Neurology, the ILAE Ambassador award and the European Epilepsy Education award and is an Honorary member of the Academia Iberoamericana de Neurologia Pédiatrica.

Keynote speakers and panelists (in alphabetical order)

Mr. Michael Berntgen

(Session III.)

Michael Berntgen is Head of the Scientific Evidence Generation Department at the European Medicines Agency (EMA), Amsterdam.

Michael is a pharmacist by training and holds a PhD as well as a Master of Regulatory Affairs. From 1999 to 2006, Michael worked in various positions in regulatory affairs in the pharmaceutical industry in Germany and in the UK. In 2006 he joined the German national competent authority BfArM as Scientific Administrator in the Scientific Advice unit. Following this assignment, he moved to the European Medicines Agency in 2007 where he initially took up a position as Scientific Administrator in the Therapeutic Group "Anti-infectives" of the Safety and Efficacy sector, followed in September 2009 by the assignment as Head of Rheumatology, Respiratory, Gastroenterology and Immunology in this sector. From September 2013 he was heading the Scientific and Regulatory Management Department and from September 2016 the Product Development Scientific Support Department. In March 2020 he took over the current position as Head of the Scientific Evidence Generation Department.

Ms. Anna Bucsics

(Session IV.)

has an MD from the Karl-Franzens-University of Graz, Austria, where she did postgraduate research at the Dept. of Experimental & Clinical Pharmacology. In 1991 she moved to Vienna where she audited pharmaceutical expenditures at the Viennese Social Health Insurance and worked at the Main Association of Austrian Social Insurance Institutions, assessing pharmaceuticals for reimbursement. She was Head of the Department of Pharmaceutical Affairs from 2010 until 2014. She was an instructor at the Department of Finance, University of Vienna and a member of the European Commission Experts Group on Rare Diseases, and has participated in European projects (EUnetHTA, the Pharmaceutical Forum, and the Platform on Access to Medicines in Europe).

Currently, she is advisor to the MoCA project, (Mechanism of Coordinated Access to Orphan Medicinal Products, www.eurordis.org/content/moca) and Judicial Advisor at the Federal Administrative Court of Austria.

Keynote speakers and panelists (in alphabetical order)

Ms. Vittoria Carraro

(Session III.)

is the Government Affairs Director at the European Confederation of Pharmaceutical Entrepreneurs (EUCOPE). She leads EUCOPE's work on pharmaceutical incentives and rare diseases. Previously, she was External Relations Director for the European Pain Federation, a European non-profit healthcare professionals organisation focused on pain management, and Account Manager (Health Policy and Market Access) for Edelman, a global public affairs consultancy. Ms Carraro has been working on innovation and health policies through her accumulated professional experience including with the European Commission (DG Sante), national and regional governmental institutions. She was a Member of the Committee on Data for Healthy Societies of the European Health Parliament from 2018–2019 and currently, she is part of the multistakeholder group the European Expert Group on Orphan Drugs Incentives.

Ms. Elisabeth Dequeker

(Session II.)

is Master in Chemistry (M.S.) and Doctor (Ph.D.) in Science (Organic Chemistry) from the University of Leuven (Belgium). Since her postdoctoral research, she specialized in Quality Assessment aspects of medical diagnostic laboratories. She is currently full professor and Head of the Biomedical Quality Assurance Research Unit (University of Leuven), Head of Quality Assurance and Regulatory Affairs Officer for Medical Diagnostics at the University Hospitals Leuven. She has been active for more than 25 years in the field of quality management in medical laboratories, where she is an internationally recognized expert and key opinion leader. She built a strong reputation as initiator and organizer of large international external quality assessments (EQA). Her current research largely emphasizes on observational studies of various quality aspects of molecular oncology and pathology applied in diagnostic laboratories. Her work at the University of Leuven was at the basis of the design of clinically important quality assessment programs for genetic diseases and oncology biomarker tests. In her role as Head of Quality Assurance at the University Hospitals Leuven she oversees the implementation of the quality management system as well as the validation and verification of new tests and instruments used, including in vitro diagnostic (IVD) tests.

Keynote speakers and panelists (in alphabetical order)

Ms. Pavla Doležalová

(Session V.)

is a professor of paediatrics at the General University Hospital and 1st Faculty of Medicine, Charles University in Prague, where she has been leading a Centre for paediatric rheumatology and autoinflammatory diseases, member of ERN RITA. After completing her specialist training in general paediatrics in 1992 she became attracted by paediatric rheumatology. She received her subspecialty clinical training at the University of Birmingham and Birmingham Children's Hospital NHS. Her clinical and academic career has been linked to the paediatric rheumatology units of the General University Hospital in Prague and Great Ormond Street Hospital in London. She has been a Czech national coordinator for the Paediatric Rheumatology International Trials Organisation (PRINTO). As a chair of ERN RITA IT/eHealth working group she has been participating in CEF TELECOM and other national and international interoperability and rare disease projects. As a co-representative of the Czech Republic in Board of Member States for ERNs she has been coordinating Czech participation in the Joint Action devoted to the integration of ERNs into the national healthcare systems.

Mr. Jakub Dvořáček

(Session I., II. and IV.)

joined the Ministry of Health and took up the role of Deputy Minister for CZPRES in February 2022.

He has been the Executive Director of Association of the Innovative Pharmaceutical Industry since 2011.

From 2009 to 2011 he served as Director of the Investment Division in CzechInvest, investment promotion agency at the Ministry of Industry and Trade, supporting the inflow of foreign direct investments to the Czech Republic and managing the network of foreign offices. Concurrently he served as an advisor to the minister at the Czech Ministry of Education, Youth and Sport.

Previously he held number of management positions in Caritas Czech Republic managing large health awareness programs, development and humanitarian projects around the world.

He graduated from Charles University (Prague) and later accomplished postgraduate program Master of Healthcare Administration (MHA). In 2020, he completed his Master of Laws studies (LL.M.).

Keynote speakers and panelists (in alphabetical order)

Ms. Sarah Garner

(Session IV.)

is currently the Senior Policy Advisor, Access to Medicines and Health Products, WHO Regional Office for Europe. Sarah is a pharmacist specializing in global access issues and she is responsible for the strategic planning and delivery of policy dialogue and technical support to improve patient access. This includes pharmaceutical systems strengthening, regulation, selection, HTA, pricing and procurement.

Her previous roles have included the Coordinator for the 'Innovation, Access and Use' Team in the Essential Medicines and Health Products Department at WHO HQ and the Associate Director for Science Policy and Research at the UK's National Institute for Health and Care Excellence (NICE), and Pharmacist Lead for the UK Government's Special Advisory Committee on Antimicrobial Resistance.

Sarah is the technical lead for the Oslo Medicines Initiative and has led policy work-packages of public private research partnerships funded by the EU Innovative Medicines Initiative (GetReal, ADAPTSMART and Big Data for Better Outcomes).

Mr. Holm Graessner

(Session IV. and V.)

has graduated in Biomedical Engineering, Electrical Engineering, German Language and Literature, Philosophy as well as Business Administration. He received his PhD "Summa cum laude" in 2004 and, then, he obtained his MBA degree in 2008.

He has been Managing Director of the Rare Disease Centre, since 2010, at the University and University Hospital Tübingen, Germany. www.zse-tuebingen.de He is Coordinator of the European Reference Network for Rare Neurological Diseases (ERN-RND). www.ern-rnd.eu. Together with Olaf Riess, he coordinates the H2020 Solve-RD project on "Solving the unsolved rare diseases". www.solve-rd.eu

He has been co-leading one of the four working groups of the German Action Plan for Rare Diseases from 2010 until 2013. Since 2020, as a fellow of the European Academy of Neurology (EAN) he is a member of the management teams of the Neurogenetics Panel and the Rare Neurological Disease Coordinating Panel of the EAN.

Keynote speakers and panelists (in alphabetical order)

Ms. Virginie Hivert

(Session III.)

joined EURORDIS in June 2014. Between June 2014 and January 2022, she has served as Observer on the EMA Committee for Orphan Medicinal Products (COMP). During this period, she was also chairing the Therapeutic Action Group (TAG) put in place by EURORDIS to give a platform for RD patient representatives who are members of the EMA Scientific Committees to exchange and reflect on transversal topics in an environment where confidentiality is ensured. From March 2019 to February 2022, she has been PRAC Alternate member representing patient organizations. Since June 2022, she has been appointed as one of the Civil Society representatives on the EMA Management Board.

At global level, she is involved in the International Rare Diseases Research Consortium (IRDiRC) since its inception in 2011, first on the side of the Scientific Secretariat (prior to joining EURORDIS), then as Member of the Therapies Scientific Committee and later as its vice-Chair (March 2017 -February 2021). She is now representing EURORDIS in the IRDiRC Consortium Assembly/Patient Advocacy Constituent Committee. During these years, she has been contributing to and/or leading on several taskforces (Repurposing, Orphan Drug Development Guidebook, etc).

Ms. Daria Julkowska

(Session II.)

has over 15 years of experience in research and management. She is the Scientific Coordinator of the European Joint Programme on Rare Diseases that brings together over 130 institutions representing different type of stakeholders from 35 countries from Europe and beyond and is also responsible for the coordination of the IRDiRC Scientific Secretariat. She is involved in the rare diseases field since 2010. She developed and put into action a set of collaborations facilitating research. She has an extensive knowledge and understanding of European funding schemes and programmes and serves as expert on the Expert Group on support for the strategic coordinating process for European partnerships of the European Commission.

She obtained her international PhD in molecular biology at the University of Paris XI, France and University of Gdansk, Poland in 2005. She pursued her scientific vocation by the post-doctoral experience in cellular biology, at Institut Pasteur, Paris and extensive training in communication and European Union counselling. She also holds MSc in Management of Research from the University of Paris Dauphine. In 2020 she received EURORDIS Back Pearls European Rare Diseases Leadership Award.

Keynote speakers and panelists (in alphabetical order)

Ms. Kateřina Kopečková

(Session III.)

graduated with an M.D. from the Charles University Prague. She is trained in internal medicine and clinical oncology. She is currently a senior physician and vice-chair for education and research at the Department of Oncology at Motol University Hospital and 2nd Faculty of Medicine, Charles University in Prague. She obtained her PhD. in clinical pharmacology. Her area of expertise and research interest includes tumour immunology, targeted therapy in oncology and rare cancer. She was appointed for 10 years Czech representative at the EMA in COMP (Committee for Orphan Medicinal Product) and was a member of the EUCERD (European Union Committee of Experts on Rare disease). She is involved in the research of novel targeted anticancer drugds. She teaches at the Medical faculty of Charles university of Prague. She is a member of the Board of EURACAN (European Reference Network for Rare Cancer) and a member of the Working Group for Rare Diseases at the Czech Ministry of Health and represents Czech Republic in Commission SGPP subgroup on Cancer.

Mr. Viktor Kožich

(Session II.)

is a Professor of Medical Genetics at the Department of Pediatrics and Inherited Metabolic Disorders, General University Hospital in Prague and Charles University-First Faculty of Medicine, Prague, Czech Republic.
Viktor Kožich graduated from the School of General Medicine, Charles University in Prague in 1985. Since his graduation he has been working in the Institute of Inherited Metabolic Diseases and he specialized in clinical biochemistry and medical genetics, in 2012 he became the Full Professor of Medical Genetics at Charles University. His main interests are genetic, biochemical, clinical, epidemiological and ethical aspects of inherited metabolic disorders (with a focus on metabolism of B-vitamins and sulfur amino acids), and newborn screening for these disorders. Professor Kožich has been a tutor of graduate and postgraduate students, he is an author of over 140 publications in international peer reviewed journals, several chapters in books, and of articles and chapters in Czech medical literature. He has been a member of councils of several international learned societies (SSIEM, ERNDIM, and ESHG) and he is a member of the Executive Board of E-HOD since 2013. Since 2009 Prof. Kožich has been a Chairman of the Czech Coordination Center on Neonatal Screening.

Keynote speakers and panelists (in alphabetical order)

Ms. Anne-Sophie Lapointe

(Session V.)

has been directly concerned by rare diseases with two of her children suffering from a lysosomal disease. For 15 years, she was a member of the board of directors of national and European rare disease associations. She was appointed as member of the INSERM ethics committee for nearly 6 years.

She has a thesis and a master's degree in health ethics. She worked as a project manager for the rare disease health network AnDDI-Rares and was able to build with Orphanet the axis 4 of the national plan for rare diseases 3 (PNMR3) dealing with information, training and e-health. Since October 2018 she joined the Ministry of Health as project manager for the rare diseases mission. She sits on the European Commission's board of Member States for the European reference networks for rare diseases.

In 2022, Anne-Sophie Lapointe was involved in the high-level conference of the French Presidency in the rare diseases field.

Mr. Yann Le Cam

(Session I., IV. and V.)

is a patient advocate who has dedicated over 30 years of professional and personal commitment to health and medical research NGOs in France, Europe and the United States in the fields of cancer, HIV/AIDS and rare diseases. He holds an MBA from HEC Paris. He has three daughters, the eldest of whom is living with cystic fibrosis. He was one of the founders of EURORDIS-Rare Diseases Europe in 1997 and has been the organisation's Chief Executive Officer since 2000. Yann initiated Rare Diseases International in 2009, for which he is an elected member of the Council and Chair of the RDI Advocacy Committee. He was a founding member of the NGO Committee for Rare Diseases (United Nations, New York) in 2014 and is its vice-chair. Yann is a co-chair of the Global Commission to End the Diagnostic Odyssey of Children with Rare Diseases since its launch in 2018. He promoted the Commission Communication on Rare Diseases (2008) and the Council Recommendation on Actions on Rare Diseases (2009). He led the Foresight Project Rare 2030 (2019-2021) laying the ground toward a Europe's Action Plan for Rare Diseases (2022–2023). From 2016–2019, he served as a member of the EMA Management Board. He was also one of the first patient representatives appointed.

Keynote speakers and panelists (in alphabetical order)

Mr. Thomas Linden

is the Director of the Swedish National Board of Health and Welfare's Department for Knowledge-Based Policy of Health Care and is also the Swedish Government's Chief Medical Officer. He is an associate professor in neurology at Gothenburg University and visiting professor to the Florey Institute of Neuroscience and Mental Health in Melbourne, Australia.
His background is a medical doctor specialized in neurology and psychiatry, PhD in neurology and MSc in Epidemiology. He is a former chair of the Swedish Medical Association's Ethics Board and an official advisor to the World Medical Association in Medical Ethics and Socio-Medical Affairs and has had several leadership positions within hospitals and health care.

Mr. Milan Macek

(Session II.)
is the chairman of the Department of Biology and Medical Genetics of Charles University Prague-2nd School of Medicine and Motol University Hospital, and of the National Coordination Centre for Rare Diseases. He is a Czech National coordinator of Orpha.net, a past President of the European Society of Human Genetics and a current ESHG liaison for European National Human Genetics. He served at the European Commission Expert Group on Rare Diseases and is currently involved in the European Board of Member States for European Reference Networks for Rare Diseases, including the newly formed EU Advisory Board on ERN sustainability. He is currently the president of the Czech Society of Medical Genetics and Genomics.
He did his postdoctoral studies at the Department of Medical and Human Genetics at Humboldt University Berlin (1989-1992) followed by McKusick-Nathans Centre for Genetic Medicine, Johns Hopkins University in Baltimore (1992-1996). In 1992 he was also a fellow at Harvard School of Medicine in the field of non-invasive prenatal diagnosis of rare diseases.
His citation index is over 11,000x with H -index of 33.

Keynote speakers and panelists (in alphabetical order)

Mr. Ondřej Májek

(Session II.)

is the Head of Department of International Affairs at the Institute of Health Information and Statistics of the Czech Republic and the scientific lead of its National Screening Centre, where he is involved in monitoring and evaluation of screening programmes and introduction of new disease early detection projects. He is also an assistant professor and the head analyst of cancer screening programmes at the Institute of Biostatistics and Analyses, Faculty of Medicine, Masaryk University in Brno, Czech Republic. His research interests include monitoring and evaluation of disease detection programmes, cancer epidemiology, and medical biostatistics. He is a co-author of over 40 articles in international peer-reviewed journals.

Mr. Tomáš Mlčoch

(Session III.)

currently leads a team of health economics in Value Outcomes/iHETA which are responsible of approximately 30-40 submissions per year. He is an ISPOR member, current member of Czech pharmacoeconomic society board (Czech ISPOR chapter) and future president (2023-2024) of Czech pharmacoeconomic society. His primary field of expertise is in health economics and outcomes research (HEOR), and preparation of cost-effectiveness, budget impact and comparative efficacy/safety analyses which are submitted to health technology assessment (HTA) agencies.

In particular, his field also covers rare diseases such as cystic fibrosis, phenylketonuria, multiple myeloma, Hodgkin lymphoma or many others.

His educational background is economics, economic modelling and behavioural economics. He published many studies in impact factor journal mainly covering cost-of-illness analyses (cystic fibrosis, phenylketonuria, rheumatoid arthritis), cost-effectiveness analyses (Crohn's disease, pulmonary arterial hypertension), utility mapping studies (psoriatic arthritis, ankylosing spondylitis) or outcomes research/modelling studies (propensity score or new outcomes in rheumatoid arthritis).

Keynote speakers and panelists (in alphabetical order)

Ms. Lisen Julie Mohr

(Session I.)

works as a senior communications advisor at Frambu - Resource Center for Rare Disorders. She has a university degree in medical librarianship and in languages. Before she started working at Frambu; she worked for several years at Oslo University. Since 2009, Lisen has coordinated projects with European countries financed by EEA and Norway Grants. All projects are aimed at rare diseases and Frambu is a partner representing the donor country.

Lisen also has experience of being the mother of a young man, now 40 years old, who has an intellectual disability, probably caused by a rare chromosomal disorder of unknown origin. As a child, her son was first clinically diagnosed with Williams syndrome, but later lost the diagnosis as his chromosome 7 does not have a deletion. Lisen established and ran the Norwegian organization for Williams syndrome for more than 10 years. During these years she also helped start up the Federation of European Williams Syndrome Associations.

Ms. Nathalie Moll

(Session III.)

joined the European Federation of Pharmaceutical Industries and Associations (EFPIA) as Director General in April 2017.

Prior to joining EFPIA, between 2010 and 2017 she was the Secretary General of EuropaBio. She spent over 20 years working for the biotech industry at EU and national level in associations and corporate positions and held the position of Chair and Vice Chair of the International Council of Biotech Associations (ICBA).

In 2013, Nathalie won the Technovisionaries Women Innovation Award organised by Women & Technologies® while in 2009, Nathalie and the Green Biotech Team of EuropaBio were presented with the Leadership and Excellence in Advancing Ag-Biotech and Food Issues Award. Nathalie was also named one of the 15 leading women in biotech in Europe in 2017 and recognised as one of the "20 Women Who Shape Brussels" by POLITICO Europe in 2020.

She holds an Honours Degree in Biochemistry and Biotechnology from St Andrews University, Scotland.

Keynote speakers and panelists (in alphabetical order)

Mr. Declan Noone

(Session IV.)

is the President of the European Haemophilia Consortium (EHC). Declan has extensive experience as a patient advocate at the national, European and international levels. He currently is the patient representative on the EMA's PRAC committee and has been involved in over 25 advice procedures as a patient representative with EMA over the last 6 years. He also sits on the Irish national product selection board for the purchase and monitoring of products for the treatment of haemophilia and other related conditions since 2006. He has worked with agencies such as the UNDP, NICE and ICER US looking at all aspects of drug development from concept to access. He is also an engineer and health economist by background.

Ms. Irene Norstedt

(Session I.)

works at the European Commission where she is the Director responsible for the People Directorate within the DG for Research and Innovation.

The People Directorate works towards the development of a healthy, safe, more equal, free, open and fair society, where the voice of the citizen and different communities are better heard.

Irene has been at the European Commission since 1996, and has worked on various aspects of research in life sciences and particular health research throughout her career in the Commission. Areas of particular interest have been the set up of the public private partnership the Innovative Medicines Initiative (IMI) and the International Rare Diseases Research Consortium (IRDiRC), Prior to joining the European Commission, she worked for the Swedish life science company Biacore AB and at the Swedish embassy in London.

Irene studied at the Royal Institute of Technology in Stockholm and University of Sussex, and holds a Master of Science (MsC) in Chemical Engineering.

Keynote speakers and panelists (in alphabetical order)

Ms. Adéla Odrihocká

(Session I.)

is a translator and interpreter specializing in healthcare, medicine and pharmaceutical area. Adela became involved in patient advocacy after being diagnosed with a rare disease after 12 years of being undiagnosed and searching for a diagnosis. During her studies, she led workshops for academic staff members to help improve study conditions for students with chronic illnesses and to talk about accessible education in university settings. She has been a member of ČAVO (The Czech Association For Rare Diseases) and the only ambassador for her disease in the Czech Republic. In recent years, she has led many chronic disease related workshops for diverse audiences (healthcare professionals, students, etc.) not only in the Czech Republic, but also in France and other European countries.

Last year, she shared her "diagnosis odyssey" story on Czech national television. She has been part of the Eurordis – 30 million reasons for European action on rare diseases campaign and was invited as a speaker and a young advocate to a panel discussion at the 11th European conference on rare diseases and orphan products.

Mr. Anders Olauson

(Session I.)

was involved in the founding of the Agrenska Centre in 1989. He served as director until 2004 and since then been chairman. He is responsible for establishing The Agrenska Virtual International Academy, a research centre for rare disorders. In 2003 The Eesti Agrenska Foundation was inaugurated, and Anders is serving as Chairman of the Council since then.

He is past member of the board of Eurordis and was president from 1999 to 2001.

EURORDIS was during this period instrumental in the establishment of COMP (The Committee for Orphan Medicinal Products) at the European Medicine Agency. Between 2006 and 2012, he was a member of the advisory group for Health Research within DG Research at the European Commission. Since 2013/14 until 2018 Anders was appointed member of the Horizon 2020 Advisory group for Societal Challenge -Configuration 'Health, demographic change and well-being.

He received HM The King of Sweden's Medal for valuable contributions in the field of disability. He received City of Gothenburg's medal for outstanding service for the city. In February 2017, Anders received EURORDIS Lifetime Achievement Award.

Keynote speakers and panelists (in alphabetical order)

MS. Ana Rath

(Session II.)

is the Director of INSERM, US14 – Orpha.net. Ana is a medical doctor and coordinates the global Orphanet network and the production of the Orphanet nomenclature, ontology and database. She coordinates the EU OD4RDI_II projects that aim to promote the improved codification of RD, she leads Pillar 2 of the European Joint Programme Co-Fund on RD, and a work package in the EU Solve-RD.eu project. She has previously coordinated the RD-Action, RD-Code - EU Joint Action bridging data and policies for RD and coordinated the Scientific Secretariat of the International Rare Disease Research Consortium. Ana brings her experience and expertise in the rare disease field, in particular in the area of systematic literature reviews, to the Rare 2030 project, leading the INSERM team's work in WP4. Ana is delighted to contribute Orphanet's expertise and 15 years of experience in delivering literature reviews to the RD community via the OrphaNews newsletter to this project. She hopes that building on the past, we can help shape an inclusive future political scenario for RD that leaves no-one behind.

Mr. Andrzej Rys

is a medical doctor specialised in radiology and public health, graduated from Jagiellonian University, Krakow, Poland. In 1991, he founded the School of Public Health at the Jagiellonian University that he ran as Director until 1997. Thereafter, from 1997 to 1999, he served as Director of the Krakow's City Health Department.

Between 1999 and 2002, he continued his career as Deputy Minister of Health in Poland where he developed a new system of emergency medical service and reformed the education system for the health professionals.

He was a member of the Polish EU accession negotiators team for the harmonisation of the Polish Health Care Law with the EU's Acquis Communautaire.

He served as a Senior Consultant of "Health and Management Ltd" for the World Bank, WHO and EAR in Serbia in 2002.

In 2006, he became Director for Public Health and Risk Assessment at DG SANCO.

From 2011 to September 2022, he was the Director responsible for Health Systems, Medical Products and Innovation in DG SANTE, where he is now Principal Scientific Adviser.

Keynote speakers and panelists (in alphabetical order)

Mr. Maurizio Scarpa

(Session II. and V.)

is the Director of the Regional Coordinating Centre for Rare Diseases at the University Hospital of Udine, Italy. He is Professor of Paediatrics at the Dept. for the Woman and Child Health, University of Padova, Italy, and the Co-Founder of the Brains For Brain Foundation, together with Prof. David Begley, Kings College of London, London, UK. Prof. Scarpa has extensive expertise as a basic scientist in genetics and biotechnology, as well as a clinician in the diagnosis and treatment of paediatric rare disorders; neurometabolic diseases in particular. Together with dr. Christina Lampe he founded the Center for Rare Diseases at the Helios Dr. Horst Schmidt Kliniken in Wiesbaden, Germany. He is especially interested in developing innovative health approaches for the diagnosis and the treatment of metabolic inherited diseases; to this aim he is also collaborating with major biotech companies as an external independent expert. Prof. Scarpa is the Coordinator of the European Reference Network for Hereditary Metabolic Diseases, MetabERN, formed by 101 healthcare providers in 27 EU countries (www.metab.ern-net.eu).

Ms. Olga Solomon

(Session III.)

studied Chemistry at the Aristotle University of Thessaloniki, Greece and holds an MSc in Food Science from the Gothenburg University, Sweden. Before joining the European Commission she worked for 5 years for a beverage producing company in Greece.

She joined DG SANCO in 2000 and worked for 10 years in the field of Food Safety in particular dealing with legislation on Food Contact Materials, Food Additives and Enzymes. In 2010, she moved to the Directorate 'Health Systems and Products' where she worked in the field of substances of human origin before taking up a post in the pharmaceutical sector in 2011. She is currently the Head of the DG SANTE Unit responsible for Medicines: policy, authorisation and monitoring.

Keynote speakers and panelists (in alphabetical order)

Ms. Birutė Tumienė

(Session V.)

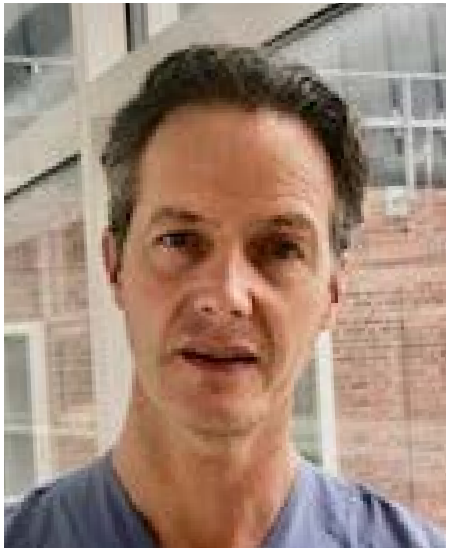
is a clinical geneticist by background. She graduated Faculty of Medicine in Vilnius University, where she also obtained her residency training in clinical genetics and accomplished PhD studies in rare genetic epilepsies; currently, she gives lectures on genetics and rare diseases here. She is the Head of Unit for Genetics and a Coordinator of International Affairs in the Coordination Center for Rare Diseases at Vilnius University Hospital Santaros Klinikos. She is a Vice-Chair of the Diagnostic Scientific Committee in the International Rare Diseases Research Consortium IRDiRC, a member of a coordinating group in the European Joint Program on Rare Diseases EJPRD, National Coordinator of Orphanet Lithuania, Advisory Committee member in the European Rare dIsease research Coordination and support Action ERICA and Lithuanian Representative in the European Reference Network Board of Member States (ERN BoMS). She fosters interests in rare diseases, that combine the perspectives of a health professional, lecturer, policy-maker and an advocate of rare disease patients. In 2021, she was awarded the Black Pearl European Leadership Award for her input into the development of European Reference Networks.

Mr. Till Voigtländer

(Session V.)

is associate professor of neurobiology and neurosciences at the Department of Neurology, Division of Neuropathology and Neurochemistry, Medical University of Vienna. After studying medicine in Heidelberg, Germany, he received his professional training in molecular biology, neuropathology, neurochemistry and neuroimmunology at different universities and institutions in Heidelberg, Berlin, Zurich, and Vienna. Since his board certification as specialist in neurobiology in 2006, he leads a specialised clinical laboratory focussing on the diagnosis of selected rare neurometabolic, neuroimmunological and neurodegenerative diseases. In Austria, he is country coordinator of Orphanet since 2004, was head of the National Coordination Centre for Rare Diseases at the Austrian Healthcare Institute and is now the director of the national office for rare diseases. He was one of the key participants in the elaboration of the national plan of action for rare diseases and is currently involved in its implementation. He is currently for the third time co-chair of the Board of Member States of ERN. He was appointed by all Member States as future coordinator of the intended Joint Action for the Integration of ERNs into national healthcare systems. In 2019, he received the EURORDIS Black Pearl European Rare Disease Leadership Award.

Keynote speakers and panelists (in alphabetical order)



Mr. Rolf Zetterström

(Session II.)

received his MD and PhD at Karolinska Institutet, Stockholm, Sweden. He did his residency in paediatrics and continued in paediatric endocrinology and inborn errors of metabolism. Since 2016 his is manager of the national newborn screening laboratory and works part time as consultant at paediatric endocrinology both at Karolinska University Hospital, Stockholm, Sweden. From 2012 until 2022 he was the chairman of the working group for inborn errors of metabolism in the Swedish paediatric society. Since 2013 he is registry holder for the Swedish national registry for inborn errors of metabolism and in 2019 he was elected member of International Society for neonatal screening (ISNS) council and scientific committee.

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:

1. A call for coordinated European Action Plan on Rare Diseases to address the challenges of people living with rare diseases and their families

CZ PRES 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. Specifically, a European Action Plan on rare diseases should:

- Bring together existing EU strategies and actions in a comprehensive framework (e.g. cancer, data, research, pharmaceuticals, social rights)[10].
- Integrate and sustain EU and national plans and strategies for rare diseases on a long-term basis.
- Update and reinforce the last rare disease strategy from 2008/2009, to prepare better for the next 10-15 years and the continued challenges in terms of genomics, technology and scientific advances.
- Create an informal multistakeholder, multi-country working group to support the European Commission in establishing a scoreboard of indicators to monitor the implementation of the Action plan at the EU and national level, and to identify good practices across disciplines and countries.
- Introduce measurable goals to ensure that all Member States are working to the same objectives to reduce inequalities across the EU.

2. Early Diagnosis of Rare Diseases

Early diagnosis (notably via preconception carrier testing and newborn screening) can significantly decrease mortality and morbidity caused by selected rare diseases and improve quality of life by expediting access to the most effective care and treatments. Yet the quest for diagnosis often remains an odyssey. Better use and accessibility of current effective and available diagnostic tools and technologies, best practices and programmes should be considered as part of a continuum of health strategies along the life course of any person living with a rare disease in Europe. As recognised during the Expert Meeting on rare diseases, as well as at the technical meeting also held under the Czech Presidency of the EU specifically on newborn screening (23 July 2022, Brno), newborn screening (NBS) has the potential to detect several dozens of rare conditions, providing the possibility of early treatment and a significantly improved long-term outcome while minimizing harms of NBS programmes. Despite these clear benefits, the availability and conduct of NBS programmes varies considerably across the EU and, with the increasing potential of the newly developed genomic testing, it is likely that these differences may become even more pronounced. Therefore, the CZ PRES calls on the European Commission and the EU Member States

A. to support initiatives that aim at promoting the best NBS practice to ensure availability and equity of access to well-structured NBS programmes for all EU citizens and that may benefit from coordinated EU-wide approach. Activities that will help support this aim include:
    - The need to document and identify good practice in existing national NBS programmes so that these lessons may be promoted and adopted more widely.
    - The need to develop, collect and collate key performance indicators for all stages of the NBS programmes that

[10] This should include: revisions of legislation of Orphan Medicinal Products Regulation, Paediatric Use of Medicines and adoption of the 'general pharmaceutical legislation), Europe's Beating Cancer Plan; non-legislative policy actions; and recent Commission initiatives such as the European Health Data Space, the European Pillar of Social Rights Action Plan and the European Care Strategy

will help to evaluate, monitor and improve the quality of NBS programmes.

- The need for outputs from national pilot programmes in NBS to be shared more effectively to shorten the time needed to introduce screening programmes for new candidate conditions or cease their inclusion.

- The development of agreed case definitions.

- The development of interoperable outcome studies for conditions included within existing screening programmes so that the impact and effectiveness of the current programmes can be assessed and improved.

- The formation of 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.

Special consideration should also be given to evaluate the rapid development of genomics that may improve the efficacy of diagnosis shortly after birth. The novel techniques provide exciting opportunities to bring significant health and economic benefits to society and particularly to EU citizens with rare disorders, but they also raise important technical, logistic, ethical and economic issues that need to be addressed before they can be recommended as a routine part of public health policy offered on a whole population basis to asymptomatic newborns.
As demonstrated by the SOLVE-RD project, a combination of -omics technologies can also contribute to solving the pathogenicity in different patient cohorts and confirming a diagnosis for rare disease patients who did not receive one with other tools (e.g., molecular diagnosis).
The CZ PRES calls on the EU and its Member States

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

Accessibility of medical devices necessary for diagnostics of rare diseases is of crucial importance. Regulation (EU) 2017/746 on in vitro diagnostic medical devices sets several ways for derogation from the generally applicable rules for safety and performance requirements when placing the medical devices on the market. Nevertheless, further specification that would ensure that in vitro medical devices necessary for proper diagnostics of rare diseases remain available on the market is needed. The CZ PRES: Appreciates the ongoing work of the Medical Device Coordination Group MDCG guidance document concerning the art. 54 of the Regulation that will include clear statement when it comes to possible derogation from the Regulation for medical devices necessary for rare diseases diagnostics.

Calls upon the MDCG and the European Commission to prepare without any further delay guidance document on art. 5.5 of the Regulation that would in detail define based on practical experience and including practical ways for their implementation.

3. Revision of the Orphan Drug and Paediatric Drug Regulations

In many ways, the Regulation on Orphan Medicinal Products implemented in 2000 proved to be a great success, leading to progress of care in many overlooked conditions. Despite this progress, concerns about remaining unmet needs, patient access, affordability, and sustainability of pharmaceutical spending have risen in the past few years. In particular, there are concerns related to the appropriateness of the current regulatory framework to attain the societal goal of reducing unmet needs while ensuring value-for-money.
The CZ PRES encourages the European Commission:

A. 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.

In particular, the CZ PRES believes there is a unique opportunity to:

B. Define 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;

C. Transform the European Research & Development for the rare disease ecosystem building upon advances of the past 20 years, for the next 20 years. This must reflect and connect developments across science, technology and policy;

D. Situate Europe as a global leader in research, development and access to diagnostics, treatment and care, through a regulation that is attractive and competitive globally. Reflections should be made in aligning with and maintaining competitiveness with the USA's FDA system;

E. 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;

F. Ensure convergence and coherence of relevant existing as well as currently negotiated legislation.

4. Instruments for improving access to rare disease treatments

The revision of the Orphan Medicinal Product and Paediatric Medicines regulations can only address part of the unmet medical needs. Fragmented access to healthcare across the EU precludes many patients from timely access. Desired system should ensure sustainability for healthcare systems and include additional solutions to address a persisting lack of treatments and inequalities across the Member States.

A new approach that balances the incentives needed for innovation, with financial and fiscal sustainability of health care systems, is required. The approach should consider clinical need, proven therapeutic value and cost-effectiveness, volumes of medicines produced and budget impact, while requiring robust evidence generation to reduce uncertainties about benefits. Public and private initiatives developed in response to the ongoing COVID-19 pandemic demonstrate that greater collaboration is possible and reflects shared responsibility between the national health authorities and the pharmaceutical industry, with inherent price transparency[11].

Further strengthening European cooperation in pricing and negotiations is desirable, while respecting current division of competences.

CZ PRES fids 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[12] Statement. Such a collaboration, supported by a dedicated platform, should be able to explore new approaches to affordable pricing, reimbursement and funding (for example external reference pricing, price regulation, equity-based tiered pricing, value-informed pricing, and staggered, performance-based or subscription payment models). While joint purchasing has shown to be a rather cumbersome process, joint negotiations have the potential to improve accessibility of complex treatments, since they would significantly increase attractivity of even smaller EU markets.

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

[11] Oslo Medicines Initiative - Statement by WHO/Europe (September 2022)

[12] Ibid.

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.

As in very small populations and/or complex treatments, such as Advanced Therapeutic Medicinal Products, evidence at time of pricing and reimbursement is often immature[13], there is a high level of uncertainties at time of Marketing Authorisation which makes clinical value assessment very challenging for all EU Member States. This leads to delays in the HTA processes and results in delayed and incomplete access. CZ PRES believes it is necessary to:

C. Support the generation of evidence across the whole life cycle of products. The generation of additional real-world evidence data in the years following marketing authorisation for selected, innovative and transformative medicines for complex and low prevalence diseases, with true cross-border value, would ensure attractiveness of the European R&D ecosystem for rare diseases and drive consolidation and structured cooperation. This streamlined approach could be included as a new incentive in the forthcoming revision of the Regulation on Orphan Medicinal Products.

5. Holistic healthcare pathways: Integrating European Reference Networks into European health care and social systems

The Expert Conference recognised that European Reference Networks (ERNs) stood out as a success story in how to drive collaboration across the EU. There are now 24 Networks connecting over 1500 clinical centres. Experts at the Conference expressed how crucial the next few years are and the critical importance of European Reference Networks as a pillar within a European Action Plan on Rare Diseases.
CZ PRES would like to emphasise the need for:

A. Stronger integration of ERNs into national healthcare systems, to enable EU countries to strengthen the resilience of their national health system and improve accessibility of highly specialised expertise.

B. 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. Sustainable funding mechanisms to accelerate the development and uptake of treatment options for rare diseases within ERNs, as they integrate European-wide clinical research and care settings, supported by registries.

C. Leveraging network-based health data, experience and knowledge, powered by digital tools, where Centres of Expertise act as a trusted universal source and curators of global knowledge and integrate it to daily clinical practice.

D. A fully-fledged data strategy for rare diseases, supported by interoperable infrastructures, to collect and exploit the full value and potential of health-related data in alignment with and contributing to the European Health Data Space and the European data strategy.

E. Implement EU-wide and national policies and programmes to person-centred and integrated care, both in terms of integration across medical disciplines but also bridging the medical and social spheres, with the aim to enable holistic wellbeing of people living with rare diseases and their families.

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[13] Rare diseases are not uniformly spread across prevalence: only 4% of diseases sit in the 1-5 in 10,000 prevalence bracket, while 84.5% affect fewer than 1 in 1,000,000 patients. While the more prevalent diseases are less frequent, the size of the populations suffering from each of these diseases is significantly higher, meaning that 80% of all rare disease patients fall in the 150 diseases with the highest prevalence (Nguengang Wakap, S., Lambert, D.M., Olry, A. et al. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. Eur J Hum Genet 28, 165–173 (2020).

Practical information

Practical information
This Practical Information Note provides general information on logistical and organisational aspects to help you prepare for the meeting.
Please note that all information may be subject to change, in which case it will be communicated by the Czech Presidency in due time.

Accreditation

Please note that attendance at the Conference is by invitation only and only accredited delegates will have access to the main venue and evening reception.
Delegates were kindly requested to register via the online accreditation system of the Czech Presidency of the Council of the European Union, through the designated Delegation Accreditation Officer (DAO) of the respective delegation. Your DAO should have received more detailed instructions.
In case you have not appointed your DAO yet, please proceed at your earliest convenience to register the DAO through an e-mail at accreditation.czpres@mzcr.cz and please indicate the name of the event in the subject of the email. The DAO will receive the link to the accreditation system in due time.
Please contact the email address mentioned above for further information on registration process.
We kindly ask you to register at your earliest convenience via the accreditation system. The online accreditation system will be open until 20 October 2022.
Please note that only accredited delegates will have access to the programme venues.
Please note that the online accreditation must be completed 5 days prior the event at the latest.
If you have any questions regarding the invitation or registration, please send your enquiry to:
accreditation.czpres@mzcr.cz.

All personal information provided during the accreditation will be processed only in accordance with General Data Protection Regulation (GDPR) obligations mentioned in the CZ PRES GDPR memorandum, which can be found on presidency website and accreditation system itself.

Conference venue and accommodation

The Conference will take place at the Prague Congress Centre in the Vyšehrad quarter of Prague (Prague Congress Centre (Kongresové centrum Praha, a. s.) - Prague.eu). The Conference venue address is 5. května 1640/65, 140 21 Prague 4.
Regarding the accommodation, we recommend booking your room at the Corinthia Hotel Prague (www.corinthia.com/prague/) which is located just across the street from the Prague Congress Centre.
If you have any questions regarding the accommodation, please send your enquiry to:
accreditation.czpres@mzcr.cz.

Transport

The city of Prague has an excellent transport accessibility. The Prague airport offers a whole range of possible flights and once you arrive you can use either the Prague bus, tram or metro systems. Below you will find the instructions on how to get to the conference venue:

Arriving by plane

You can find additional information on https://www.prg.aero/en/public-transport-buses

- Take bus nr. 119. Disembark at the stop “Nádraží Veleslavín”.
- On “Nádraží Veleslavín” switch on the metro line A (green, direction to “Skalka” or “Depo Hostivař”.
- Take the metro line A to the stop “Muzeum”.
- Switch to metro line C (red) on the station “Muzeum”. Take the C line in the direction “Háje” to the station “Vyšehrad”.
- Exit the “Vyšehrad” station with the exit in direction of either the “Prague Congress Center” or “Corinthia Hotel Prague”.
- The transfer should take max 50 minutes.
- Taxis are available at the entrance to the airport.

Arriving by train

- Take the metro line C (red) form the station “Hlavní nádraží” (main train station) in the direction “Háje” to the station “Vyšehrad”.
- Exit the “Vyšehrad” station with the exit in direction of either the “Prague Congress Center” or “Corinthia Hotel Prague”.
- The transfer should take max 20 minutes.

Tickets

- The ticket is valid for all means of public transport in Prague.
- Ideal solution is a ticket for 90 minutes.
- Tickets are sold in machines directly on the bus stop (airport) or near the metro entrance (main train station). Most ticket machines accept contactless credit card payments. Tickets can also be purchased via SMS or the Lítačka mobile app, which can be also used to search for the ideal public transport connection. The application is available for Android and iOS.

Prague Congress Centre Map

1	ENTRANCE 1 Vchod 1	6	ENTRANCE 6 Vchod 6	11	PERSONAL ENTRANCE 11 Personální vchod 11
2	ENTRANCE 2 Vchod 2	7	ENTRANCE 7 Vchod 7	12	HOTEL HOLIDAY INN PRAGUE CONGRESS CENTRE
3	ENTRANCE 3 Vchod 3	8	ENTRANCE 8 Vchod 8	13	BUSINESS CENTRUM VYŠEHRAD
4	ENTRANCE 4 Vchod 4	9	ENTRANCE 9 Vchod 9	14	CORINTHIA HOTEL PRAGUE
5	ENTRANCE 5 Vchod 5	10	ENTRANCE 10 Vchod 10		

For the entrance into the Prague Congress Centre, please use the entrance No. 1. The registration desk located by this entrance will be open from 13:00. You will receive your badge there. Please make sure to wear your badges visibly at the meeting venue and evening reception. If you lose your badge, please contact the registration desk at the meeting venue without any delay.
The Czech Presidency will provide all necessary services, such as a cloakroom, internet connection, and catering at the conference venue.

Evening Reception

The evening reception will take place at the Corinthia Hotel Prague from 19:30 on 25 October. All participants of the Conference are invited. The Corinthia Hotel Prague is located only a minute´s walk away from the venue. The reception will take place in the dining space of the Hotel.
The Corinthia Hotel Prague offers extraordinary views of Prague and the surrounding Vyšehrad area.

For more details regarding the hotel and the venue of the evening reception, we recommend you look at this page:
https://www.corinthia.com/prague/

Please note that the Conference and evening reception will be conducted in English only, no interpretation will be provided.

Prague

We cordially invite you to discover the city of Prague. To do this, please check the tourist website at:
https://www.prague.eu/en

Other Information

Emergency number:
112 (fire brigade, medical assistance)

Electricity:
The voltage in Czech is 230V, 50 Hz.

Local time:
Central European Summer Time Zone (CEST) – GMT +2:00

Country code:
Czech Republic +420

Currency:
Czech crown, for the current rate please consult
https://www.cnb.cz/en/

Tap water:
Czech water is regularly tested for quality and is safe to drink.

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