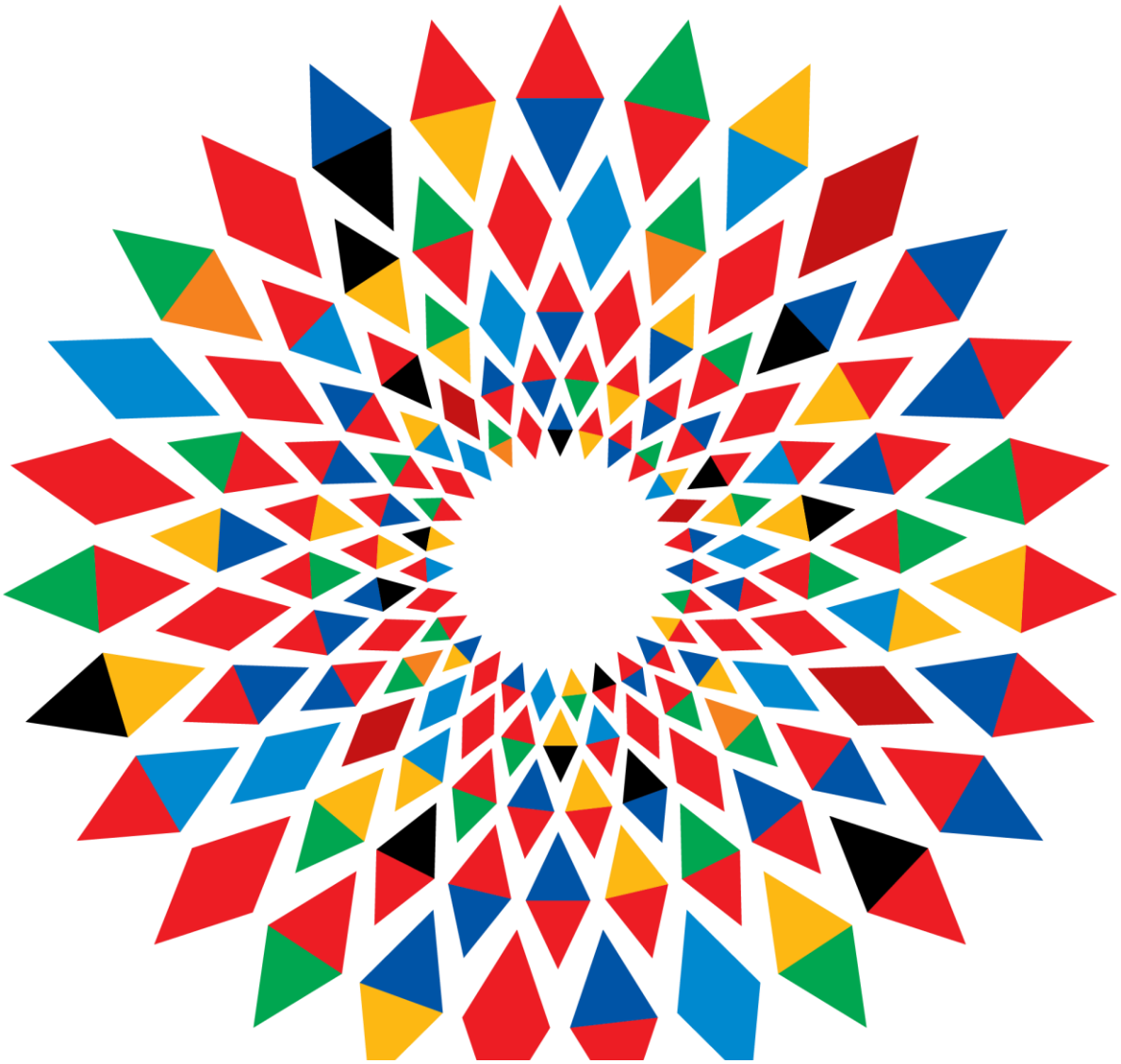

Background document

Technical meeting under the auspices of the Presidency of the Czech Republic in the Council of the EU.

Brno, Czech Republic

July 23, 2022

Early diagnosis of patients with rare disorders in the EU:

crucial role of the newborn screening

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)

Format of conference: hybrid with videorecording and on-line participation by invitation only; the link and password will be distributed after July 18, 2022

SCOPE

Rare diseases. Rare diseases comprise a clinically and etiologically diverse group of conditions defined in the European Union by a prevalence of fewer than 5 per 10,000 in the population. More than 9,300 rare diseases have been described and 83% are of genetic origin (http://www.orphadata.org/). The individual conditions can be rare but collectively they are quite common, affecting about 30 million citizens in the EU. Without well organised systems to identify those affected, the early recognition of patients tends to be poor with a long diagnostic odyssey and consequently less than optimal outcome once the patient enters an appropriate clinical care pathway.

Initiatives to improve quality of life of those with rare diseases. Following the European Council recommendation on rare diseases issued in 2009, effort has concentrated upon ensuring equity of access to health care regardless of disease frequency and rare disease research has benefitted from the interest shown by the pharma industry into the development of new orphan drugs.

Nevertheless, novel approaches such as the use of genetic technologies to identify patients and expensive personalised medicine present new ethical dilemmas for those planning health care policy. On December 16, 2021, the United Nations General Assembly accepted a resolution "Addressing the challenges of persons living with a rare disease and their families" and encouraged UN Member States to support care and research. The EU and the Member States have also promoted clinical care and research for individuals with rare diseases, as recommended by the EU Council and included in the EU Directives, EU Research Plans and national rare disease plans.

Despite this interest and owing to the rarity of individual conditions, expertise tends to be distributed unequally between Member States within and among countries. To help address this and improve standards of care and cohesion across Europe, the European Commission established European Reference Networks in 24 rare disease categories as well as the European Platform for Rare Diseases Registration. The Council of the EU and the former Committee of Experts on rare diseases EUCERD have called for coordinated action at the EU level that, while respecting the subsidiarity principle, competencies and responsibilities of the Member States, supports them in the organization and delivery of health services and medical care, including newborn screening.

Newborn screening as a programme. The early, asymptomatic detection of patients with treatable rare diseases offers the best chance of a good outcome and this can be achieved most effectively by whole population screening shortly after birth. These Newborn Screening Programmes began in the 1970s in many European countries and conditions have been successfully added to the screening panel in EU Member States since that time. Currently around fifty rare diseases can be routinely detected by well-established biomarkers, and as a result patients can be offered effective intervention that will substantially improve the quality of life and

outcome for the individual affected. In practice, the number of these diseases included in national screening panels varies considerably across the EU and worldwide as reviewed recently by *Loeber et al Int J Neonatal Screen. 2021 (7):15*. Despite the need to consider individual Member States' national context and responsibility for defining their health policies, this disparity raises some concern about equitable access to health care for individuals living with rare diseases in the EU.

Variation is not limited only to the number of conditions included but also in the way that screening is conducted including: the information offered to citizens before screening, sample collection and transport arrangements, laboratory analyses and turn-around time, the means of reporting of results and the timeliness of patients entering treatment, the confirmatory testing used to establish a diagnosis, the treatment and follow-up offered and the evaluation of these programs and their governance.

In the interests of the stakeholders of newborn screening programs including the public, patient organizations, experts, policy makers, health care payers and ethicists, it is important to identify good practice and look critically at 'outcomes' so that good practice can become common practice across the EU. Some of this thinking has been explored in a recent publication by *Scarpa et al, Lancet Reg Health Eur. 2022 (13):100311*.

Previous technical meeting. To help improve the equity of provision of newborn screening and ensure that all children can be offered high-quality screening regardless of race, nationality and socio-economic status, a technical meeting, endorsed by the Slovenian Presidency of the Council of the European Union, was held in October 2021. Results of the meeting were published in a paper by *Sikonja et al Int J Neonatal Screen. 2022 (8):31* and they record experiences from individual EU countries, stakeholder initiatives and the meeting's final conclusions, to offer help to countries attempting to establish new screening programs or expand existing provision.

PURPOSE

The present technical meeting under the auspices of the Presidency of the Czech Republic in the Council of the EU aims to bring to the table key stakeholders and discuss progress in three workstreams of the Screen4Rare initiative:

- Achieving consistency in the case definitions used in diseases included within national screening panels
- Developing interoperable registries to chart the outcome for individuals identified by newborn screening
- Establishing a blueprint for the 'newborn screening pathway' to optimize practice in the EU in this important area

OUTPUT

It is expected that the meeting will support the development of key performance indicators to identify good practice in NBS programs with recommendations for NBS data collection and governance in EU Member States. It is also anticipated that that it will progress the discussion of the importance of developing consistent case definitions for conditions included within newborn screening programmes in Europe and the value of assessing the long-term outcome of patients with these conditions.

PROGRAMME JULY 23, 2022

Time	Programme	Speaker
13:15 – 13:30	WELCOME	
	Prof. Vlastimil Válek, MD, PhD., MBA, EBIR (TBC) Minister of Health of the Czech Republic **Jakub Dvořáček MSc., LLM** Deputy Minister of Health of the Czech Republic **Lumír Kantor, M.D** (video presentation) Senate of the Parliament of the Czech Republic **Prof. Milan Macek Jr., MD., DSc.** National Coordination Center for Rare Disease **Prof. Viktor Kožich, M.D, CSc.** Coordination Center for Neonatal Screening **Ondřej Májek, RNDr. PhD** National Screening Centre, Institute of Health Information and Statistics of the Czech Republic	
13:30 – 15:30	**SESSION I. – NEWBORN SCREENING (NBS): A GATEWAY TO EARLY DIAGNOSIS** (CHAIRS: Dr. Gulcin Gulmus and Prof. Viktor Kožich)	
13:30-13:50	Overview of European NBS activities-synergies and overlaps	**Prof. Jim Bonham,** United Kingdom
13:50-14:05	Role of European Reference Networks for rare diseases in NBS	**Prof. Maurizio Scarpa,** Italy
14:05-14:20	Developing a blueprint of NBS in Europe: overview of workstreams	**Dr. Peter Schielen,** The Netherlands
14:20-14:35	Key indicators for planning, monitoring and evaluation of newborn screening: international context and future perspectives for cooperation	**Dr. Ondřej Májek,** Czech Republic

14:35-14:50	The tower of Babel: why do we need case definitions?	**Dr. Rolf Zetterstroem,** Sweden
14:50-15:05	The key role of registries in assessing clinical outcome	**Prof. Stefan Koelker,** Germany
15:05-15:20	Experience with expanding NBS in Czechia	**Ms. Anna Arellanesová,** Czech Association for Rare Diseases, Czech Republic
15:20 – 15:45	**Coffee break**	
15:45 – 16:30	**SESSION II. – CURRENT EXPERIENCE AND FUTURE DEVELOPMENTS IN NBS** (CHAIRS: Ms. Anna Arellanesová and Prof. Maurizio Scarpa)	
15:45-16:05	Newborn screening: the perspective of people with RD and future potential	**Dr. Antoni Montserrat,** EURORDIS
16:05-16:20	The use of a patient management system to improve long-term outcome	**Dr. Rolf Zetterstroem,** Sweden
16:20-16:35	Screen4care EU IMI project	**Prof. Alessandra Ferlini,** Italy
16:35-17:10	Panel discussion (CHAIRS: Dr. Antoni Montserrat and Dr. Peter Schielen)	
	Ms. Martine Pergent IPOPI President/Screen4rare, France **Mr. Stelios Kympouropoulos** MEP (TBC) **Dr. Jose Valverde** European Commission, DG SANTE Unit B3 (TBC)	

	Ms. Dorica Dan Specifics of care in the Romanian and minority populations living in Romania, NoRo Romania **Prof. Gunnar Douzgos Houge** IVD-R: beneficial or just an expensive strait jacket? Norway **Dr. Sofia Douzgos Houge** IVD-R: Case example of a rare recessive disorder, Norway **Dr. Gulcin Gulmus** EURORDIS
17:10-17:40	**GENERAL DISCUSSION** (Speakers, panelists and on-line guests (on-line attendance by invitation only)
17:40-18:00	**SUMMARY AND CLOSE OF MEETING**
	Prof. Jim Bonham, United Kingdom **Dr. Rolf Zetterstroem,** Sweden **Dr. Antoni Montserrat,** EURORDIS and ALAN Maladies Rares, Luxembourg