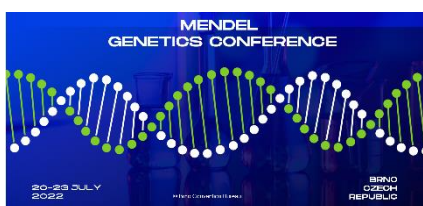


# Technical meeting under the auspices of the Presidency of the Czech Republic in the Council of the EU



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

Satellite meeting to the Celebrations of 200<sup>th</sup> Anniversary of G. J. Mendel' s birth

July 20-23, 2022 ([www.mendel22.cz](http://www.mendel22.cz))

**Date:** Saturday July 23, 2022, 13:15- 18:00, Brno, Czech Republic

**Venue:** Mendel Museum at the Augustinian Abbey, Brno

([mendelmuseum.muni.cz/en](http://mendelmuseum.muni.cz/en))

**Format of conference:** hybrid – visit the [www.mendel22.cz](http://www.mendel22.cz) website for further information

**Program version 11 (July 8, 2022).**

### 13:15-13:30 WELCOME

Prof. MUDr. Vlastimil Válek, CSc., MBA, EBIR, Minister of Health and/or Jakub Dvořáček MSc., LL.M., Deputy Minister of Health

Lumír Kantor, M.D. – Senator

Senate of the Parliament of the Czech Republic

Prof. Milan Macek Jr., MD., DSc. -

Chair of the Coordination Center for Rare Disease

Prof. Viktor Kožich, M.D, PhD.

Chair of the Coordination Center for Neonatal Screening

Prof. Ladislav Dušek, PhD  
Republic

Institute of Health Information and Statistics of the Czech

Dr. Antoni Monserrat

EURORDIS.org - ALAN Maladies Rares Luxembourg

## **13:30-15:30 SESSION 1**

### **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, National Screening Centre, Institute of Health Information and Statistics of the Czech Republic, Brno, Czech Republic

**14:35-14:50 The tower of Babel: why 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 2**

### **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, ALAN Maladies Rares Luxembourg - EURORDIS.org

**16:05-16:20 The use of patient management system to improve long-term outcome**

Dr. Rolf Zetterstroem, Sweden

**16:20-16:35 Screen4care EU IMI project**

Prof. Alessandra Ferlini, Italy

**16:35-17:45 PANEL AND GENERAL DISCUSSION**

**16:30-17:10 PANEL DISCUSSION**

**CHAIRS: Dr. Antoni Montserrat and Dr. Peter Schielen**

**Ms. Martine Pergent**, IPOPI President/Screen4rare, United Kingdom

**Mr. Stelios Kypouropoulos**, 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, EURORDIS.org

**Prof. Gunnar Douzgos Hougé**: IVD-R: beneficial or just an expensive strait jacket? Norway

**Dr. Sofia Douzgos Hougé**: IVD-R: Case example of a rare recessive disorder, Norway

**Dr. Gulcin Gulmus**, EURORDIS.org

**17:10-17:45 GENERAL DISCUSSION**

Speakers, panelists and guests (on-site and on-line)

**17:45-18:00 SUMMARY AND CLOSE OF MEETING**

**Prof. Jim Bonham**, United Kingdom