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 200th Anniversary of G. J. Mendel's birth

July 20-23, 2022 (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)

Format of conference: hybrid – visit the 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., LLM, 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 Institute of Health Information and Statistics of the Czech Republic

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
**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 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, EURORDIS.org

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.org

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

Prof. Jim Bonham, United Kingdom