Expert Conference on Rare Diseases

Session II Early Diagnosis for Rare Diseases

Panel discussion

Early diagnosis: from newborn screening to personalized patient care

Proposed urgent initiatives (2021 Slovenian EU PRES)

- **Formation of a time-limited NBS expert advisory committee:** free from bias or national interests, trusted high-quality information to support decision making at a national level
- **Work streams:** sharing of good practice in existing national NBS programmes (including through the Non-Communicable Diseases advising the European Commission)
  - **key performance indicators** : to ensure quality of newborn screening programmes
  - **case definitions:** disorders currently screened and under consideration
  - **interoperable disease registries**: to evaluate long-term clinical outcome
  - **national pilot programmes in NBS**: experience should be shared
- **Consolidation of a NBS group within the existing European Rare Disease Reference Networks**
  - **Expert Platform on NBS formed and supported by Screen4Rare, several ERNs currently involved in NBS: e.g. MetabERN and ERN-RITA**
  - other ERNs: caring for patients that may benefit from the early asymptomatic detection in near future
- **Special consideration- rapid development of genomics** to greatly alter the potential for diagnosis at birth and the ethical challenges and clinical opportunities that this brings

Sikonja et al, Towards Achieving Equity and Innovation in Newborn Screening across Europe, Int. J. Neonatal Screen. 2022, 8(2)

Call to action

EU2022.CZ

Call to Action

from the Expert Conference on Rare Diseases

Towards a new European policy framework on rare diseases:

"Building the future together for rare diseases"

On 25 and 26 October 2022, in Prague

Questions from audience and possible topics for discussion

Overarching themes

- Regulatory environment and governance
- Codification and use of standards for data sharing among national and disease-specific registries

Newborn screening

- Inequality across EU
  - Diagnosis
  - Therapy
- Quality cycle
- Genomic screening and ELSI
- General public perspective
  - Providing information to prospective parents
- HTA

Shortening the path in non-NBS screened RD

- Education of professionals
- Awareness of patients
- Signals of RD in health care systems and in patient records
- Availability of specialized assays
- Genomic diagnosis
- Concerns of public regarding genomic data