Evidence-based newborn screening strategy

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European Reference Network | rita

The European Reference Network that aims at improving the care of patients with Rare Immunological Disorders

Newborn screening for rare diseases: state of play

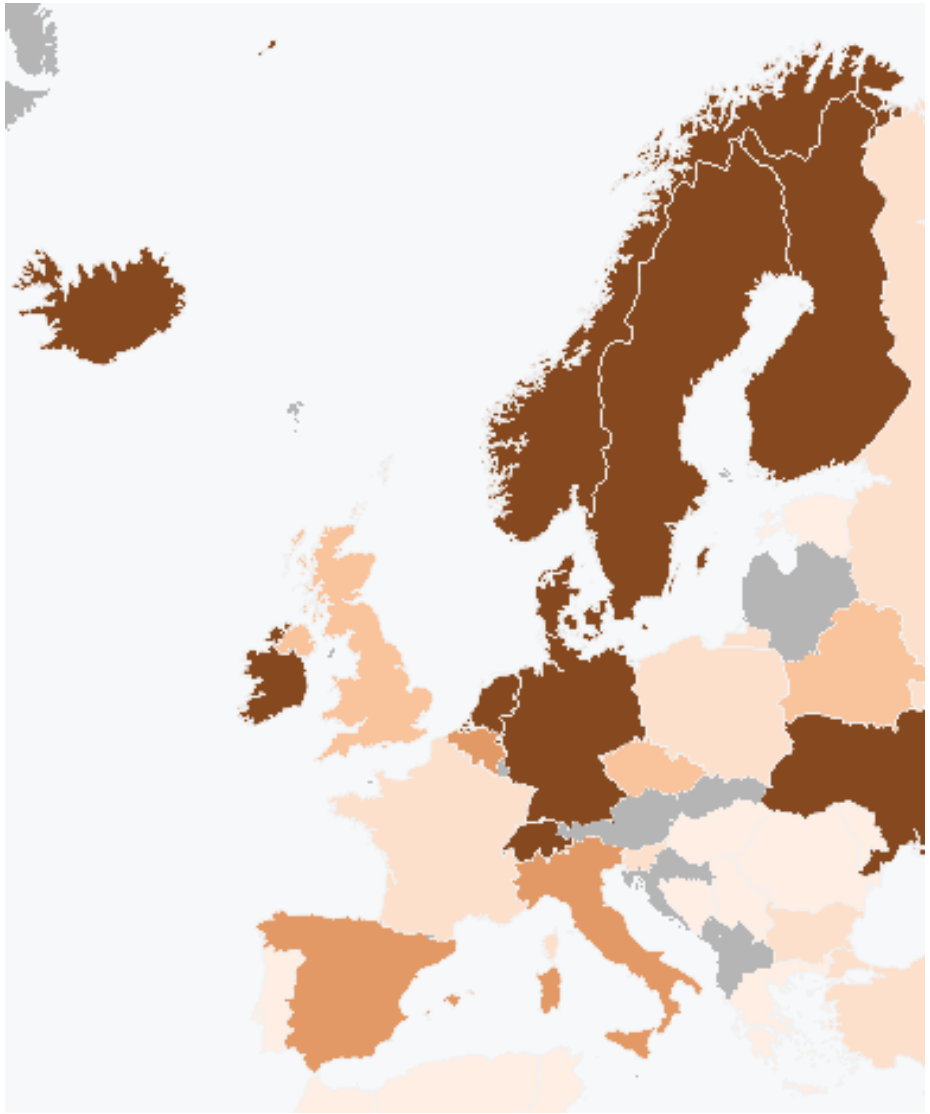
- Newborn screening (NBS) for rare diseases (RD): facilitates early detection and treatment of RD by testing newborns for a range of conditions.
- ‘One of the major Public Health Advances of the 20th Century’.
- Each year, 3.7 million children are tested in Europe.
- Number of conditions 2 - 40 conditions screened in different countries in Europe.

Newborn screening for rare diseases: state of play

- Recognition of the Wilson and Jungner criteria when making decisions about which disorders to include in the panel.

- So why do we see significant variation in the range of conditions included?

- Largely because countries apply the W&J very differently:
  - Level of evidence required;
  - Cost effectiveness studies and the acceptable cost/ QUALY / similar;
  - The make-up of the decision-making bodies;
  - Level of direct political involvement.



Status of NBS for severe combined immunodeficiency (SCID)

- No
- Pilot without decision
- Current pilot screening
- Regional implementation only
- National implementation
- No answer

Launch of Screen4Rare

Creation of Screen4Rare, partnership between ESID, IPOPI and ISNS, with the following objectives:

- Work to ensure that all babies have equitable access to newborn screening.
- Exchange knowledge and best practices on NBS for treatable RD.
- Unbiased information and evidence, so as to help ensuring the best decisions are made.

Partnership with ERNs was considered key:

- ERN RITA
- MetabERN

screen4rare

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Paediatric Rheumatology / Primary Immunodeficiency
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Launch of the ERN NBS expert platform

- Launch of the ERN NBS Expert Platform (Feb 2021):
  - Meeting organised by the EU Commission, Screen4Rare, ERN reps.
  - Objective of meeting: review of Screen4Rare campaign and the development of an ERN initiative on NBS for RD
  - Agreement: creation of an ERN Task Force on NBS for RD, coordinated by MetabERN and ERN RITA.
  - 70 participants including representatives from 18 ERNs.

ERN NBS Expert Platform: work packages

- WS 1: NBS blueprint on differences between NBS programmes in the EU Member States – led by prof Maurizio Scarpa (MetabERN) and Prof Peter Schielen (ISNS)

- WS 2: Case definitions for NBS and approaches to confirmatory testing - led by Prof Mirjam van der Burg (ERN RITA)

- WS 3: Registries and data interoperability – led by Prof Stefan Koelker (MetabERN) and Prof Jim Bonham (ISNS)

Key take aways

- ERN NBS Expert Platform: free from bias or national interests, trusted high-quality information to support decision making at a national level.
- The ERNs have the expertise available and are key to foster equity on NBS for RD in the EU.
- Need of continued support from EU Presidencies & Commission to make of NBS a key component of future EU rare disease policy.

Thank you for your attention

Gracias por su atención

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