30th Workshop of the EURORDIS Round Table of Companies (ERTC)

Newborn Screening: Shaping the future

15-16 October 2020 (13:00 to 18:30 each day)
Online event

CONCEPT PAPER

Introduction

Newborn screening (NBS) is a comprehensive system that includes multidimensional components, including testing, diagnosis, communication of information to parents, follow up care and storage of samples for secondary use. Recent and continued scientific and technological advancements have opened up the discussion on the expansion of NBS programmes to include rare diseases that could be screened using new sequencing techniques.

For approximately 50% of rare diseases, the onset occurs during childhood but for many diseases, clinical signs of symptoms do not appear in the first days or months following birth. Some rare diseases fully qualify for inclusion in NBS programmes where early intervention can prevent the onset of disease symptoms or delay disease progression, improving the quality of life of the newborn, deriving a benefit for the patients, their families and the society. On the other hand, inclusion of actionable diseases for which there is no treatment in NBS programmes raises ethical issues that need to be addressed since although there is no available treatment for these types of diseases, knowing about the disease could affect families' reproductive choices.

The number of conditions included in national newborn screening programmes varies per country (i.e. newborn screening in Ireland includes 8 conditions[1], in UK 11 conditions[2,*], in Germany 17 conditions[3,*] and in the Netherlands 22 conditions[4,*]). Although in some Member

*including hearing screening

States (MS), the new and expanded NBS programmes include screening for a wider range of conditions, there is little agreement among MS on which diseases should be included in

national screening programs. Variations in the newborn screening practices among European countries[5], not only on the diseases included but also on access and availability, highlight the need for collaboration and common uptake of NBS criteria across the MS. Having a uniformed approach among all Member States is of particular importance to improve the lives of babies born with a rare disease and their families.

We would like to explore the potentials and limits of current newborn screening approaches, implications of expanding newborn screening programmes, as well as to foster collaboration among all stakeholders in order to thoroughly discuss the current issues surrounding tests used to detect rare diseases in NBS programmes and their implications.

Key Objectives

This workshop will enable its participants to:

- Understand the newborn screening decision-making processes and gain insight about diverse national approaches;
- Discussing the ethical, social and economic ramifications of the advancing landscape of Newborn Screening in light of new technologies;
- Gain insight into the impact on patients and families of current NBS programmes;
- Discuss and refine the draft position statements from the EURORDIS Working Group on Newborn screening;
- Learn how rare disease community can support the advocacy of Newborn Screening

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[1]Health Service Executive Ireland- A Practical Guide toNewborn Bloodspot Screening In Ireland (2018)
https://www.hse.ie/eng/health/child/newbornscreening/newbornbloodspotscreening/information-for-professionals/a-practical-guide-to-newborn-bloodspot-screening-in-ireland.pdf
[2]United Kingdom National Health Service -Newborn blood spot test (2020)
https://www.nhs.uk/conditions/pregnancy-and-baby/newborn-blood-spot-test/

[3]Deutsche Gesellschaft für Neugeborenenscreening eV- Richtlinie des Gemeinsamen Bundesausschusses über die Früherkennung von Krankheiten bei Kindern (2019) https://www.screening-dgns.de/Pdf/RichtlinienGesetze/RL_Screening2019-08-09.pdf

[4]Rijksinstituut voor Volksgezondheid en Milieu(2018) Heel prick - Clinical Picture https://www.rivm.nl/en/heel-prick/clinical-picture

**This information is provided by the International Society for Newborn Screening (ISNS)* http://isns-neoscreening.org/

[5]EURORDIS Rare Diseases Europe(2020) A knowledge-base summary:Rare Disease Diagnostics http://download2.eurordis.org.s3.amazonaws.com/rare2030/Knowledge%20Based%20Summaries/Knowledge%20Base%20Summary%20-%20Rare%20Disease%20Diagnostics%20%281%29.pdf