



Update on Newborn Screening

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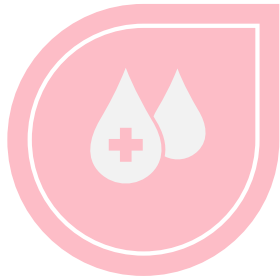
27 Nov 2020

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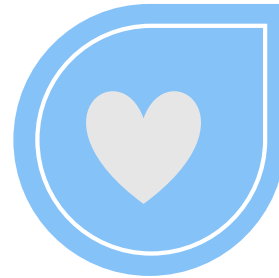


Newborn screening

Heel prick



Pulse oximetry



Hearing test



Newborn screening identifies specific disorders in the following groups:

- Metabolic Disorders (Phenylketonuria)
- Endocrine Disorders (Primary Congenital Hypothyroidism)
- Hemoglobin Disorders (β -Thalassemia)
- Genetic Disorders (Cystic Fibrosis - CF)
- Immune Deficiency Disorders (Severe Combined Immunodeficiency-SCID)



Why is Newborn Screening a priority for EURORDIS?

- Most of the screened diseases are rare diseases.
- 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.
- Currently there is no consensus on equal access and availability of screening programmes in Europe

“ If you have a rare disease, **your chances of being diagnosed often depend on where you are born**. This is why I am part of the EURORDIS Newborn Screening Working Group. I want to contribute to improving newborn screening programmes and earlier, accurate diagnosis of rare diseases. ”

Eduardo Lopez - President of Lysosomal Acid Lipase Deficiency Patient Organization (AELALD)

EURORDIS Newborn Screening Working Group

Scope

The Newborn Screening Working Group **reviews current policy and practice in the field of NBS**, in order to **develop principles** for harmonious uptake of the NBS programs **across the MS** with a view to delivering maximum benefit and improving outcomes for babies born with rare diseases

Activities

- **Collect contributions** from specific disease communities/national disease communities on challenges and potential solutions
- Contribute to the development of a series of **principles** for decision making bodies of NBS in the MS.
- Advise on the **endorsement and the dissemination strategy** of the outcomes of the WG

EURORDIS Newborn Screening Working Group

Members of the NBS-WG

Jayne Spink, Genetic Alliance UK

Simona Bellagambi, UNIAMO, IT

Domenica Taruscio, ISS, IT

Cor Oosterwijk, VSOP, NL

Maurizio Scarpa, MetabERN, IT

Laetitia Ouillade, SMA, FR

Mark Turner, University of Liverpool, UK

Martina Cornel, VUMC, NL

Johan Prevot, IPOPI, PT (observer)

Antoni Montserrat, ALAN, LU

Francesca Forzano, NHS, UK

Manuela Vaccarotto, AISMME/UNIAMO, IT

Luc Zimmerman, EFCNI, DE

Eduardo Lopez, AE LALD, ES

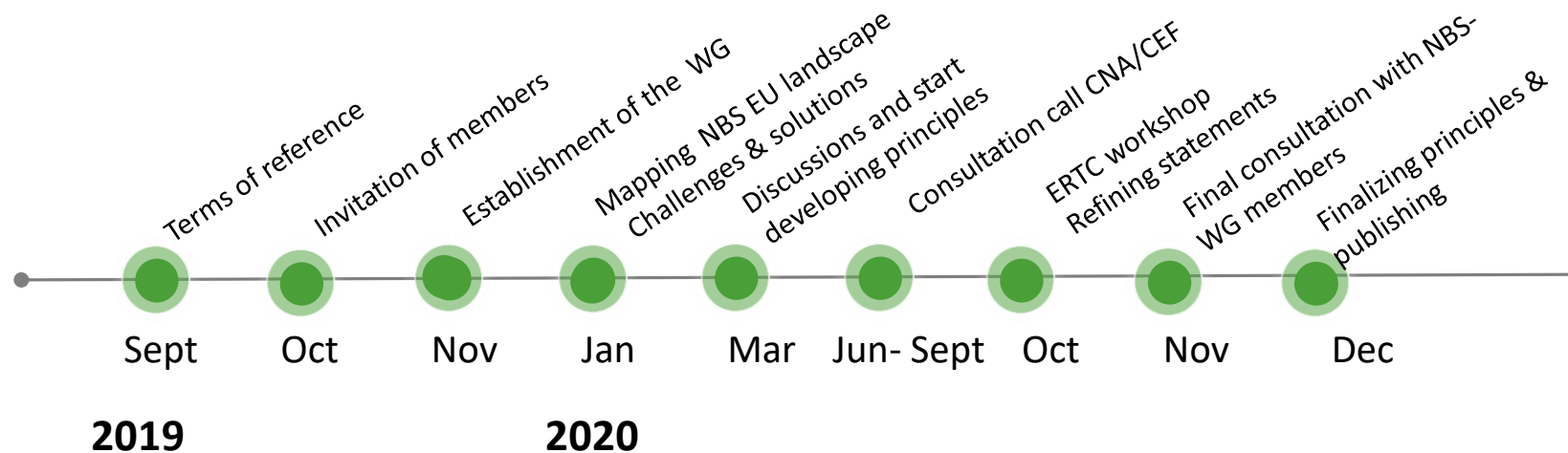
Gulcin Gumus, EURORDIS, SP

Virginie Bros-Facer, EURORDIS, FR

Valentina Bottarelli, EURORDIS, BE

Clara Hervas, EURORDIS, BE

Timeline



Also rounds of consultation with ...

- 1-1 calls with Eurordis NBS-WG members
- EURORDIS Advocacy Strategy meeting
- EURORDIS Board of Directors

KEY PRINCIPLES FOR NEWBORN SCREENING

1. **Newborn Screening (NBS) should be organised as a system** with clearly defined roles, responsibilities, accountability and communication pathways that are embedded into the national health care system and recognised as a mechanism for earlier diagnosis as part of the broader care pathway.
2. Taking into account that more possibilities for treatment and testing are becoming available, **transparent and robust governance for expanding NBS programmes** is needed. Every country/region should have a clearly defined process for deciding which conditions are covered by the NBS programme that is accessible to all stakeholders
3. Screening should identify opportunities to help the newborn and the family as broadly as possible, that is **screening should include actionable diseases as well as treatable diseases.**

KEY PRINCIPLES FOR NEWBORN SCREENING

4. **All stakeholders** (i.e patients, patient advocates, the general public, hospitals, healthcare professionals, researchers, ERN, biobanks, national authorities and EU States) **should be included** in the different stages of the NBS process.
5. The evaluation process on the inclusion/exclusion of diseases in NBS programmes needs to be **based on the best available evidence**, reflecting health economic evidence but not determined only by health economics.
6. **Education and information** of all stakeholders on rare diseases and the whole NBS process is essential for a broad and fair implementation of NBS programmes.
7. **European wide standards** addressing the timing, sample collection methods, follow-up, and information shared with parents are needed to guarantee uniformity and quality throughout the process.

KEY PRINCIPLES FOR NEWBORN SCREENING

8. **Blood spot samples** should be stored in national biobanks during a minimum time period of 5 years for research purposes while ensuring appropriate safeguards for **data protection and data access** are in place.
9. **Governance of newborn screening programmes** should be **explicit, comprehensive, transparent** and accountable to national authorities.
10. **ERN affiliated centres should be integrated in the care pathways** of the different Healthcare systems and should be considered as preferential partners in providing recommendations on NBS policies.
11. The family of the newborn who has been diagnosed through NBS should be provided with **psychological, social and economic support** by the National Healthcare System and partner agencies.

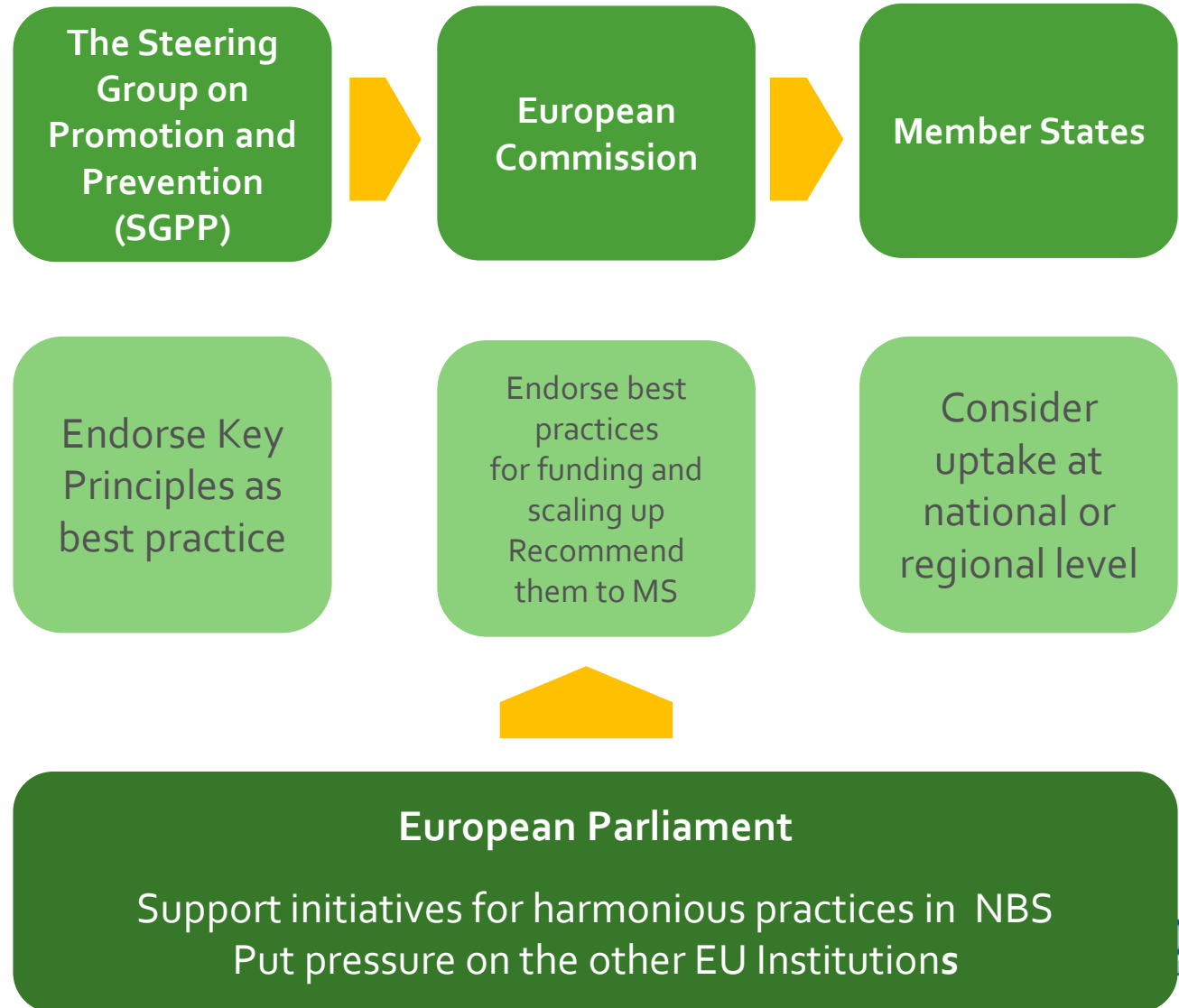
Call to Action

Expert Working Group

Experts + Stakeholders
MS representatives

Actions with clear EU added value
(respecting subsidiarity)

Hosted by JRC ?



Call to Action

The EURORDIS WG on NBS calls for:

- 1) the creation of a EU-level expert working group composed of Member States representatives, relevant experts and stakeholders to enact EU-level collaboration in areas where it would have an added value, while respecting the principle of subsidiarity e.g.:
 - exchange knowledge and best practices on NBS for rare diseases;
 - gathering evidence and data on efficacy from pilot studies or field trials in view of including specific diseases in NBS programmes;
 - entrusting assessment of new proposals for NBS, when new technologies allow for such a consideration, to EU level HTA joint assessment;
 - identifying and agreeing upon criteria and mechanisms for expanding disease panels;
 - defining guidelines on for example education and training of professionals, on communication with families and citizens at large, etc.; and
 - other areas for collaboration as already identified in the 2013 Opinion of the EU Committee of Experts on Rare Diseases.

The WG will explore the possibility of entrusting the Joint Research Centre of the European Commission or other EU level bodies or agencies (incl. the ECDC, European Centre for Disease Prevention and Control) with the coordination and the hosting of the committee.

Call to Action

2) The **WG** calls upon relevant **EU bodies and Institutions and EU Member States to promote the uptake of the Key Principles enclosed to this Call to Action** and in [particular](#) :

- **Steering Group on Health Promotion and Prevention (SGPP)** to endorse the Key Principles and recommend their uptake by EU MS as support of the development and implementation of their NBS programmes. These principles would be adopted as European best practices existing at national level;
- **European Commission** - in light of the recommendations of the SGPP- to support this effort by means of endorsing the Key Principles as best practice supported by extended evidence, and recommending their uptake at the national and European level;
- **Member States' governments** to consider the uptake of these Principles for the development and implementation of their NBS programmes and policies at the national/regional level.

3) The WG also calls on the **European Parliament** to endorse initiatives aimed at harmonious NBS programmes across Europe and filling the existing gaps in NBS practice in Europe; and, to this aim, to continue to put pressure on other EU Institutions by means, for example, of Parliamentary questions, in line with the [call for action](#) promoted by Screen4Rare.

CNA/CEF Consultation Call (22 September 2020)

To gather feedback from CNA/CEF members on the draft Newborn Screening position statements that have been developed by the EURORDIS Newborn Screening Working Group.

30th EURORDIS Round Table of Companies Workshop (Online) Newborn Screening: Shaping the future (15-16 October 2020)

Understanding the landscape and diverse national approaches

Ethical, social and economic ramifications

Refining the Newborn Screening Position Statements

Newborn Screening Principles Document

CNA CEF Consultation call
22 September 2020
30 participants

ERTC Workshop Breakouts
16 October 2020
100 participants
4 parallel sessions

VOTING

AGREE, FINE AS IT IS

AGREE BUT NEEDS EDITING

NO, I COMPLETELY DISAGREE

11 Principles

One sentence principles, 2-3 paragraphs explanation

Call to action

Specific requests to each relevant body

Feedback from stakeholders

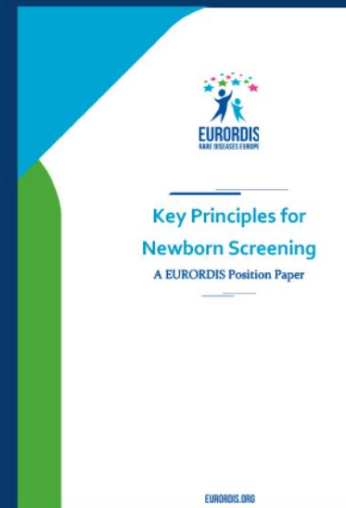
- Need for **consensus driven decisions** and alignment to contribute to an informed decision
- Focus on **the added value of increasing the importance of patient organizations** as a driving force, without creating unnecessary burden (financial, resources)
- The document should **widen the definitions of “treatment”** for other options such as physical therapy, nutrition therapy
- **Collaboration** during clinical development to be ready with a NBS programme for those identified early, to avoid the years long wait to screen

Feedback from stakeholders

- **Mention all three domains of stakeholders** (beneficiaries, actors and owners) not only on implanation but also from evidence generation to delivery of care, quality control and audit.
- The level of detail and specificity
- Separate discussions around **informed consent**
- Better explain the added value of ERNs as a network
- Explore what does require standardization at **EU level** and what can be dealt with at **member state level**

Next steps

- Continue activity in 2021
- Consultation with the BoD of EURORDIS
- Consultation with EURORDIS Members
- Publishing the position statement
- Dissemination



Key Principles for Newborn Screening

A position paper with 11 principles that advocates for harmonised criteria and adequate policies for newborn screening to be uniformly applied across Europe.

Coming soon!

Key messages

- Application of adequate policies for newborn screening requires a **collective effort from all stakeholders**
- **Dialogue** between patients, parents, and treatment developers, together with clinicians with academic experts on newborn screening on the technology, on ethical and economic aspects
- **Collaboration between patient groups and industry** is also of great importance, such as horizon scanning for timely decision making
- **The impact of early diagnosis can be life changing for patients**, this is what we should focus on when we are advocating for newborn screening.



Thank you for your attention

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