



*Key Principles of Newborn Screening
&
Roadmap to Implementation*

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EURORDIS.ORG



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



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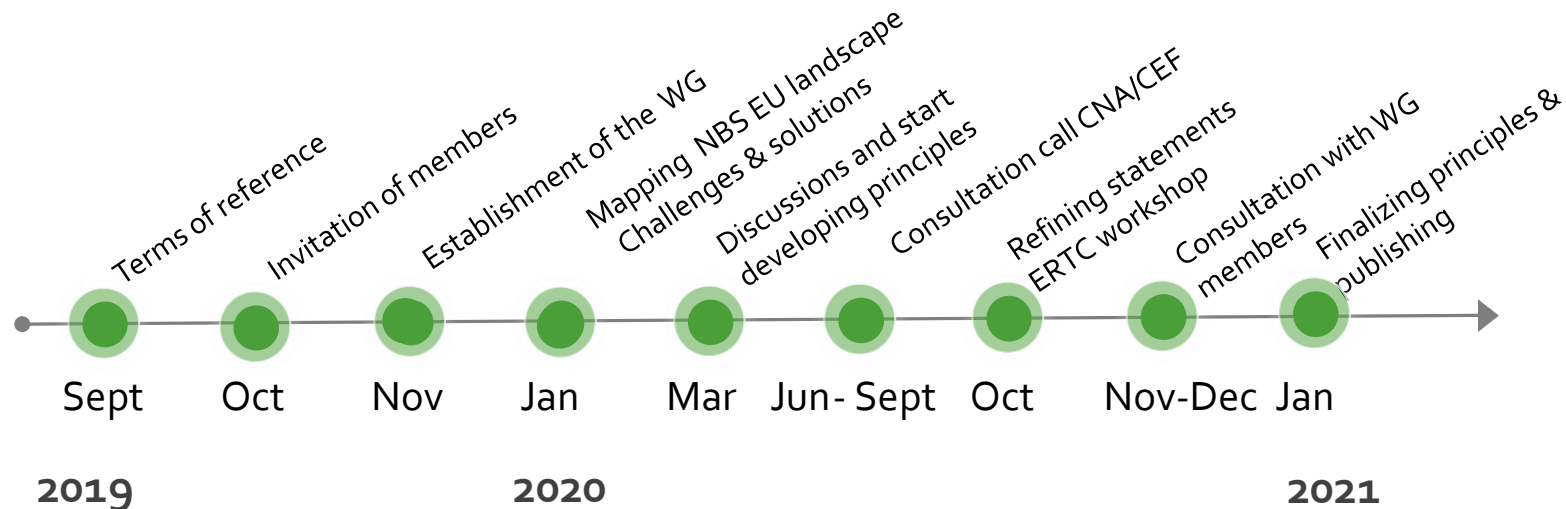
“If you have a rare disease, your chances of being diagnosed often depend on where you are born. This is why I am part of the EURORDIS Newborn Screening Working Group. I want to contribute to improving newborn screening programmes and earlier, accurate diagnosis of rare diseases.”

Eduardo López, President, Spanish Lysosomal Acid Lipase Deficiency Patient Organisation (AELALD)

”

EURORDIS Newborn Screening Working Group

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



CNA/CEF Consultation Call (22 September 2020)

Gathering 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 (15-16 October 2020)

Understanding the landscape and diverse national approaches

Ethical, social and economic ramifications

Refining the Newborn Screening Position Statements

VOTING

AGREE, FINE AS IT IS

AGREE BUT NEEDS EDITING

NO, I COMPLETELY DISAGREE



KEY PRINCIPLES FOR NEWBORN SCREENING

A EURORDIS Position paper

January 2021

eurordis.org/newbornscreening

EURORDIS.ORG



KEY PRINCIPLES FOR NEWBORN SCREENING

1. Screening should identify opportunities to help the newborn and the family as broadly as possible. That is, **screening should identify actionable diseases including treatable diseases.**
2. **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 of actionable conditions as part of the broader care pathway.
3. The family of the newborn who has been diagnosed through NBS should be provided with **psychological, social and economic support** by the competent national health authorities.
4. **All stakeholders should be included** in the different stages of the NBS process.

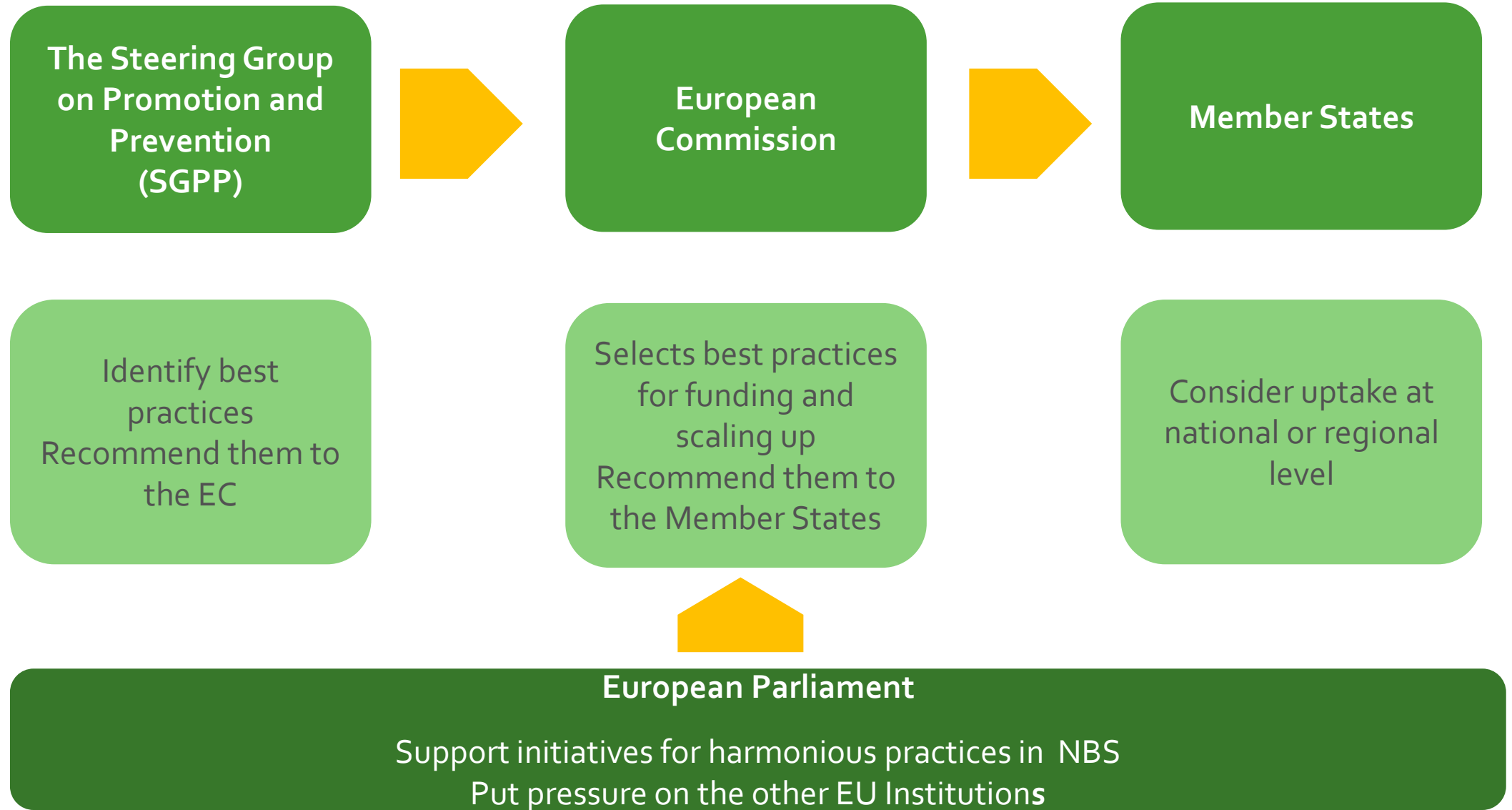
KEY PRINCIPLES FOR NEWBORN SCREENING

5. Transparent and robust governance for expanding NBS programmes is needed. Every country/region should have a **clearly defined transparent, independent, impartial and evidence-based process** for deciding which conditions are covered by the NBS programme that includes all stakeholders.
6. Governance of NBS programmes should be explicit, comprehensive, transparent and accountable to national authorities.
7. 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.
8. **Information and education** of all stakeholders on rare diseases and the whole NBS process is essential for a broad and fair implementation of NBS programmes.

KEY PRINCIPLES FOR NEWBORN SCREENING

9. **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.
10. Blood spot samples should be stored in national biobanks for research purposes while ensuring **appropriate safeguards for data protection and data access** are in place.
11. 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.

Call to Action – a role for everyone



Publication & Dissemination & Social Media Outreach

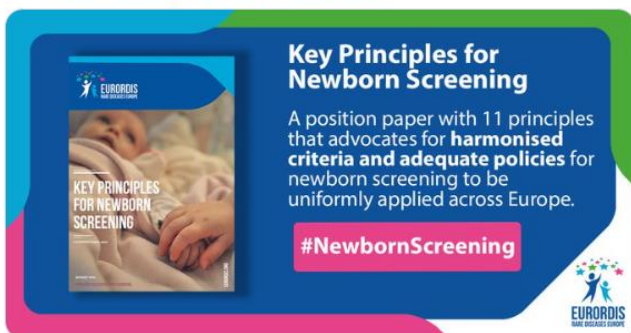


Rare Diseases Europe
@eurordis

For the first time EURORDIS, alongside its members, have set out 11 Key Principles for [#NewbornScreening](#) 🦶

These principles will support an harmonised European approach to NBS that will help to reduce vast inequalities across Europe.

👉 eurordis.org/newbornscreeni...



👤 Gulcin Gumus

2:04 PM · Jan 20, 2021 · Twitter Web App

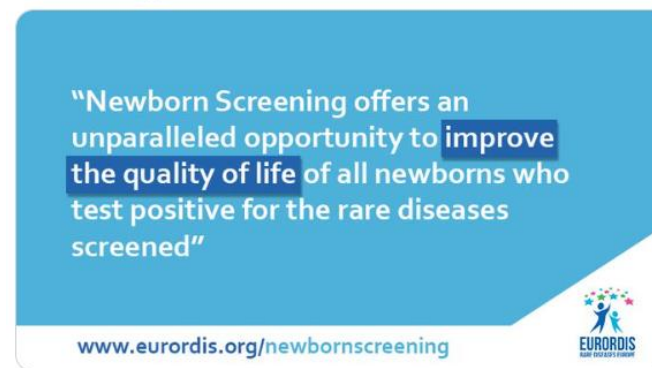


Rare Diseases Europe
@eurordis

[#NewbornScreening](#) 🦶 holds one of the keys to a better future for people born with a [#raredisease](#) in the years to come

It is up to the European Union, and its Member States, to ensure this opportunity is not missed 🇪🇺

💡 Share our 11 Key principles!
eurordis.org/newbornscreeni...



GLOBAL COMMISSION

to End the Diagnostic Odyssey for Children with a Rare Disease

February 26, 2021

11 Key Principles of Newborn Screening to Reduce the Diagnostic Odyssey



[EURORDIS-Rare Diseases Europe](#), in collaboration with its Council of National Alliances, Council of European Federations and its members, has addressed the diagnostic odyssey of children with rare diseases in the recently published "[Key Principles for Newborn Screening](#)".



Total:
Reach: 30.600
Reactions: 666



Reactions

Dravet Syndrome Foundation Spain @DSFeu · 22 Oca
Does it make sense that different regions of Spain screen babies for different diseases?

Read EURORDIS' 11 Key Principles for Newborn Screening and let's make health equity and harmonized newborn screening policy a priority for 2021!

eurordis.org/

Matt @MattwithIH · 22 Nis
@eurordis adlı kişiye yanıt olarak
This isn't just children; people of all age groups face long journeys for a diagnosis, along with ridicule and disbelief. With some adults it can be in the decades as they live with an illness they consider to be part of life.

aismme
January 21 at 5:41 PM ·

LO "SCREENING NEONATALE ESTESO" ANCHE IN TUTTI GLI STATI EUROPEI: LE INDICAZIONI DELLE ASSOCIAZIONI DEI PAZIENTI
11 Principi chiave sullo Screening neonatale per una attuazione comune in tutti i paesi EU. Documento elaborato dal gruppo di lavoro #EURORDIS in cui Aismme ha partecipato con Manuela Vac Simona Bellagambi in rappresentanza di #uniamofim e con Taruscio dell'ISS . Call to action alle istituzioni europee e agli Membri

Beata Ferencz @Beata_... · 2...
Viktigt med diagnos från dag 1 🙌

HDANI @HDAssocNI · 16 Nis
@eurordis adlı kişiye yanıt olarak
Fantastic idea and hopefully one which will be accessible across the globe

Neuroblues @neuroblues7 · 20 Oca
El desarrollo no se mide solamente en ingresos por habitante o en índices de pobreza, NBI o desnutrición.
Ay, que lejos estamos. Cada vez más.

Dmitriy Zavgorodniy
January 20 at 8:00 PM ·
"масштабное неравенство во всей Европе"
А вытекают из этого масштабного неравенства человеческие трагедии больных людей, которые даже при явно выраженной клинике игнорируют практически все медицинские специалисты на Украине!
#HCU

Latvijas Reto slimību alianse
January 20 at 2:26 PM ·

11 basic principles for newborn screening in Europe 🙌

Vesna Aleksovka
We have already translated it in Macedonian. It is very important document. Already advocating 🙌
<http://challenges.mk/.../%D0%9D%D0%B0%D1%87%D0%B5%D0%BB...>

ACURARE @ACURARE1 · 4 Şub
Yeni doğan tarama için 11 temel ilke @eurordis tarafından yayımlandı. 🙌🙌
#yenidoğantarama #new

Shirlene Badger @shirlenebadger · 20 Oca
Thrilled to see the publication of this paper - the result of wonderfully inclusive and in-depth collaboration across @eurordis membership, calling for a harmonised Newborn Screening approach that leaves no families, regardless of country, in uncertainty.

Dr Suja Somanadhan @suja15 · 20 Oca
Thank you for sharing harmonising approaches to #Newbornscreening @eurordis I note your call for action
"Responsibility for Newborn Screening programmes falls on individual countries in Europe". @DonnellyStephen @Lesmart11659095

MarenT @M_arenT_P · 20 Şub 2020
#Neugeborenencreening rettet Leben!
Ein kleiner Fersenpik in den ersten Lebenstagen des Babys und das Blut kann auf viele verschiedene #SelteneErkrankungen #rareDisease, die behandelbar sind, untersucht werden.
Leider noch nicht in allen (europäischen) Ländern flächendeckend...

ALAN - Maladies Rares Luxembourg
January 20 at 3:57 PM ·

Eng fréi Diagnos ka bei enger rarer Krankheet ee groussen Impakt op d'Liewensqualität hunn.

Den Dépistage néonatal, also d'Teste vu neigebuerene Puppelcher 🙌 fir verschidde vun dëse Krankheeten ze diagnostizéieren, ass dobäi immens wichteg.

An Europa ginn et beim Dépistage néonatal grous Ënnerscheeder tëscht de Länner.

EURORDIS - European Rare Diseases Organisation plädéiert fir eng gemeinsam europäesch Approche fir dass all Puppelchen & Famill dee selwechten Accès op fréi Diagnosetester huet.

Dofir hunn EURORDIS an déi national Allianzen elo een Dokument verëffentlecht mat 11 Prinzippien a konkreten Fuerderunge fir den Dépistage néonatal ze verbesseren & harmoniséieren.

FH Europe @fhpatienteurope · 21 Oca
11 Key Principles for #NewbornScreening



Please write in the chat

Would you be willing to translate the Newborn Screening Principles into your language?





Rare Diseases Europe
@eurordis

...

The Newborn Screening principles are already available in:

- English
- Italian
- Slovenian
- Macedonian

Translate it into your native language to bring about change in your community!

! Get in touch with gulcin.gumus@eurordis.org ([@gulcingumus1](https://twitter.com/gulcingumus1))

Tweeti Çevir



EURORDIS.



Share this with your local communities!



Next Steps

- Translating the position paper
- Second stage with the Newborn Screening Working Group
- Developing a roadmap
- Implementing the call to action

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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