European Reference Network
for rare or low prevalence complex diseases

Network Neuromuscular Diseases (ERN EURO-NMD)

Newborn Screening in

Neuromuscular Diseases

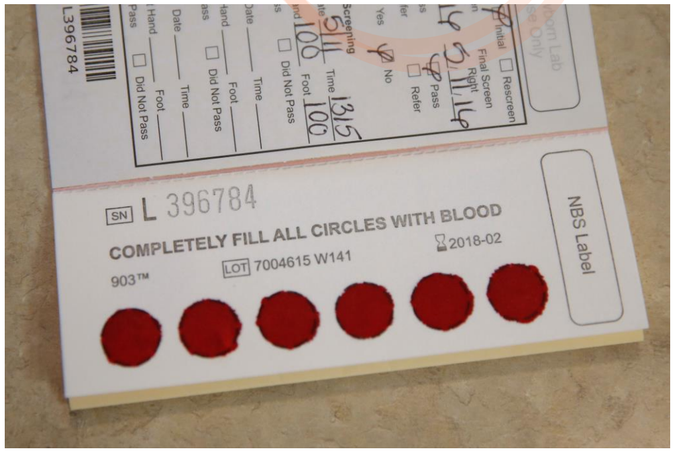
Satellite Scientific Symposium organized by ERN EURO-NMD

March 6th, 2025

Teresinha Evangelista

Funded by the European Union

Overview of Newborn Screening



Newborn screening programs are public health actions aimed at the early detection of genetic, metabolic, and congenital disorders

Newborn screening programs across Europe **are quite different in the number of diseases they cover**

Newborn screening **policies are determined by the member states**

Wilson and Jungner's Principles of Screening (1968)

Wilson and Jungner's Principles of Screening (1968)

Box 1. Wilson and Jungner's Principles of Screening[1]

- Wilson and Jungner's 1968 report remains a reference in balancing the benefits and risks of population screening

- It was well defined that effective screening governance requires:

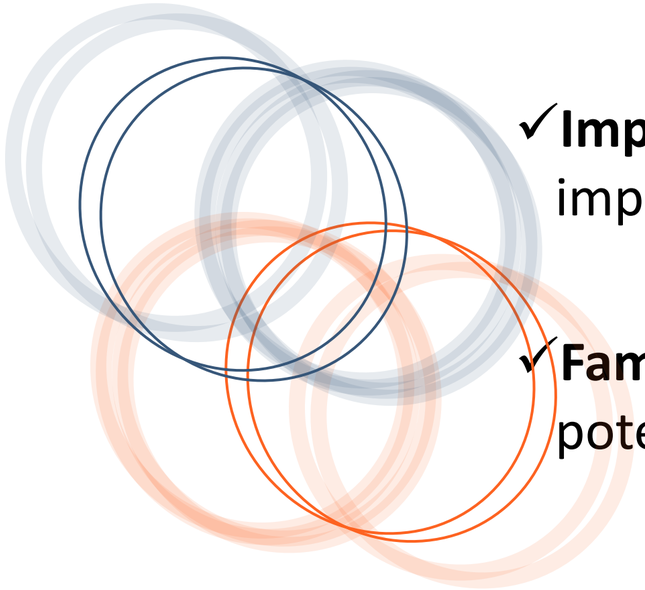
clear criteria,
robust evidence review,
public engagement,
defined target populations,
integrated care pathways,
strong data systems,
and long-term quality assurance

Importance of Newborn Screening

- Newborn screening is a critical component of public health allowing:

✓**Early Intervention:** Early intervention can prevent or minimize the progression of the disease and its associated complications.

✓**Prevention of Serious Health Issues and long-term disabilities:** such as developmental delays, intellectual disabilities, or organ damage.



✓**Improved Quality of Life:** Timely diagnosis and treatment can significantly improve the quality of life

✓**Family Planning:** make informed decisions about future pregnancies and potential genetic risks.

Received: 27 February 2022 | Accepted: 5 March 2022
DOI: 10.1111/ene.15320

european journal of neurology

POSITION PAPER

How to approach a neurogenetics diagnosis in different European countries: The European Academy of Neurology Neurogenetics Panel survey

Michelangelo Mancuso[1] | Henry Houlden[2] | Maria Judit Molnar[3] | Alessandro Filla[4] | Marianthi Breza[2] | Holm Graessner[5] | Claudio L. A. Bassetti[6] | Sylvia Boesch[7]

Answers: 239 neurologists
40 European member states affiliated to EAN

FIGURE 1 European countries (in red) affiliated with the European Academy of Neurology where we have obtained responses to the survey. Numbers in brackets indicate the number of responders per country [Colour figure can be viewed at wileyonlinelibrary.com]

Results:

Lack of information about new-born screening

47.3% of the responders did not know which new-born screening programs were available in their countries

European Reference Network
for rare or low prevalence complex diseases
Network Neuromuscular Diseases (ERN EURO-NMD)

Orphanet Journal of Rare Diseases

Orphanet J Rare Dis. 2021; 16: 75.
Published online 2021 Feb 10. doi: 10.1186/s13023-020-01670-8

PMCID: PMC7874448
PMID: 33568176

Survey on patients' organisations' knowledge and position paper on screening for inherited neuromuscular diseases in Europe

F. Lamy,[1] A. Ferlini,[2] ERN EURO-NMD Patient Advisory Board, and Teresinha Evangelista[3,4]

POs from 18 countries (17 are part of the EU) with ethnically and genetically heterogeneous populations and different economic backgrounds.

Independently of the ethnic and cultural heterogeneity, most POs (28 out of 30) were in favour of screening irrespectively of the existence of a disease-modifying medical treatment.

Only few mitochondrial diseases and more recently SMA are included in national screening programs this study reveals that **screening for NMD is a largely unmet need**

Symposium Objectives

INTERNATIONAL NEONATAL SCREENING DAY
JUNE 28

- **Raise Awareness**
- **Share Scientific Advances**
- **Discuss Best Practices**
- **Explore Policy and Guidelines**
- **Foster Collaboration** – Encourage partnerships among healthcare professionals, researchers, policymakers, and industry stakeholders to enhance screening programs.
- **Showcase Case Studies** – Provide real-world insights into the impact of newborn screening on patient outcomes.
- **Identify Future Directions** – Discuss emerging technologies and potential expansions in newborn screening programs.

Acknowledgments