European Reference Network
for rare or low prevalence complex diseases

Network Neuromuscular Diseases (ERN EURO-NMD)

7th ERN EURO-NMD
ANNUAL MEETING

NBS – Patients perspective

21st – 23rd February 2024

Marie-Christine Ouillade

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Funded by the European Union

Marie-Christine Ouillade

- Volunteer at AFM Telethon for more than 20 years
- Mum of a 30 years old lady affected by SMA (Spinal Muscular Atrophy)
- Member of AFM TELETHON and GENETHON Board
- Member of SMA Europe board
- Chair of the SMA NBS Alliance

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Agenda

- Patient / parent point of view
  - Paediatric disaeses
  - Adult disaeses
- Current situation of NBS in Europe
  - Regulatory
  - Access to NBS
  - Health economy
- Patient organisation proposal

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Parent of young patients point of view

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Diagnosis in Paediatric neuromuscular diseases

- SMA, Duchenne, LGMD …
- Mother or father discovers some things is wrong
  - Too protective
  - Mother imagination
  - “Don’t worry each child has i progression”
- Child regression
  - Identify the ri cer of expertise
  - Series of
- Geneti ver
- Implementation of standard of care and treatment if available
- Genetic counselling

Between a few weeks to a few years

- SMA NBS 2 or 3 copies of SMN2
- 10 days
- Treatment
- Genetic counselling

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SMA : today
70% of European babies are screened at birth

- 30% of babies are not screened

In France

- 30% of type1 babies died in 20[illegible] and have no access to in[illegible]ve treatment just pa[illegible] care due to the time[illegible]gnosis
- Al[illegible] and 2 have permanent disabilities

Severe psychologic impact on parents

- 70% of babies screen

- In Germany
  - Less than 3% of 2 SMN2 copies babies died before the age of 2 years
  - 50% of 2 SMN2 copies babies shows light symptoms at birth
  - No disabilities at the age of 3 years old for 3 SMN2 copies babies
  - 4 SMN2 copies babies are treated at birth

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Diagnosis in the neuromuscular domain
Adult neuromuscular diseases

- Steinert, Myastenia …
- No need of NBS

- Important need to shorten and secure the diagnosis pathway

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NBS : current situation in Europe

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Regulatory situation : NBS is state relevant

- Lack of clear process to apply for a new disease in many countries
- Limited capacity for a PAG to advocate
  - Access to key arguments
  - No identified pathway
- Genetic testing
  - Ethical
  - Technics
- Procedural delays and efficiency
- In some area as Spain it’s region relevant
  - Huge difference and inequity through Europe

SMA NBS Alliance

Whitepaper Version 2, 25 November 2021

European Alliance for Newborn Screening in SMA

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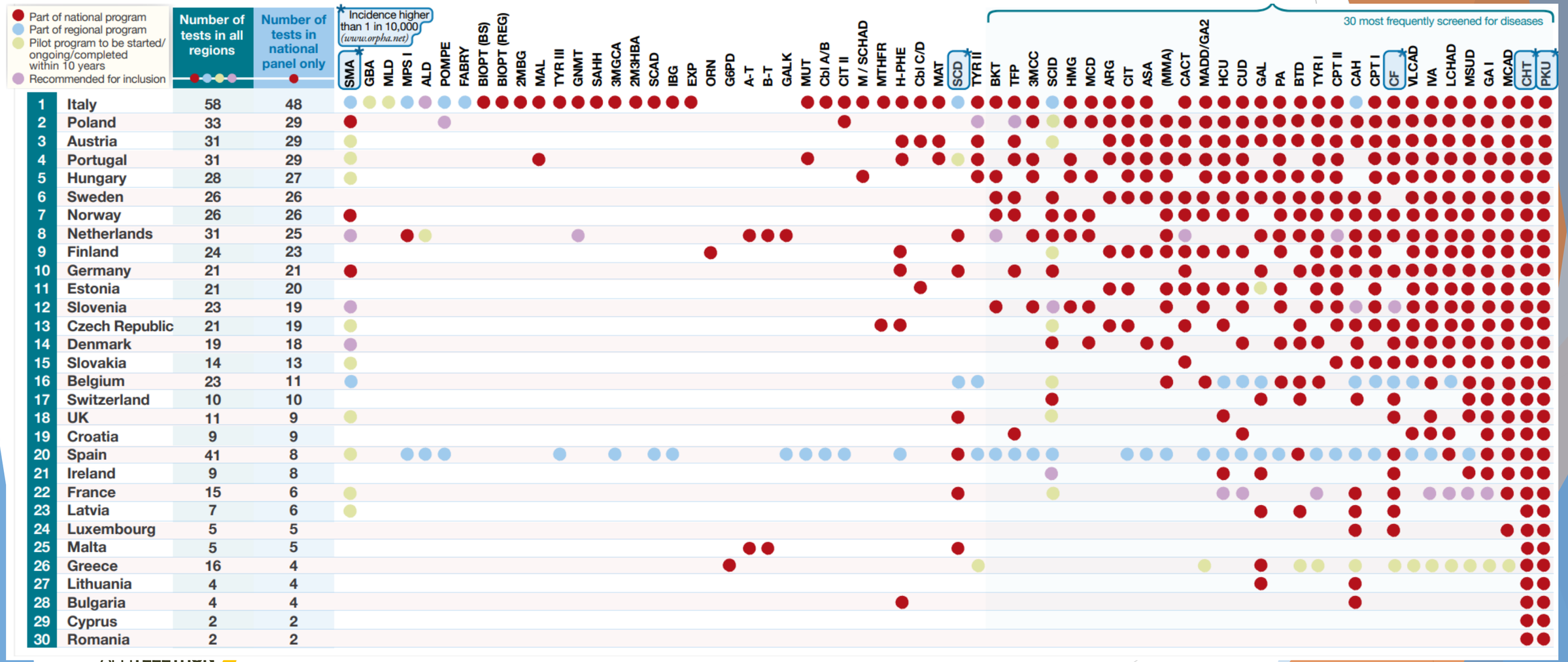
Access in dec 2022



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Health economic SMA NBS studies

Efficiency is country dependant

All studies shows a positive balance to NBS at 3 to 5 years

Without NBS

- Time for diagnosis
- Cost of treatment
- Long life follow up
- Permanent disability
- Impact on family life
  - Caregiver work
  - House and car equipment
- Access to school, work

With NBS

- Cost of NBS
- Cost of treatment
- Long term follow up
- No or low disabilities

For SMA : Time to diagnose is disabilities

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NM Patient organisation proposals

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Recommandation

At EU/international level

- International recommendation for a panel of diseases to EU states
- Revision of Wilson and Jungner´s principles
- Address also actionable pediatric genetic diseases
- Multi diseases testing technics

At national level

- Unique panel in the country
- Clear pathway to apply for a new disease
- PO must be include in the decision pathway

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Thanks to :

SMA EUROPE

EURORDIS
RARE DISEASES EUROPE

WORLD
DUCHENNE
ORGANIZATION
UPPMD

IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
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Questions

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