SMA NBS Alliance

European Alliance for Newborn Screening in SMA

Why did SMA Europe decide to create an Alliance ?

- February 2020 : Evry Congress evidence
  Test at birth, save a life

SMA: Test at birth, save a life.

Identifying children living with SMA when pre-symptomatic through NBS allows for treatment to be started on time, preventing motor neuron death and a life with severe disabilities.

- NBS in SMA prevents family burden
  - Disabilities by reducing time between first symptoms and diagnosis
  - Mother / parent guilt

- Complexity to go to NBS in all the European countries
  - One dossier per country, sometime per region

= > SMA NBS ALLIANCE support national stakeholder to apply for SMA NBS

SMA NBS Alliance

Alliance Members

Founding member

SMA Europe

Members

eamda

EURORDIS

TREAT-NMD Neuromuscular Network

umcg

HEALTH-ECORE

NOVARTIS

Biogen

Roche

university of groningen

PerkinElmer For the Better

LaCAR LIFE AND CARE ADVANCED RESEARCH

Secretariat support by

admedicum Business for Patients

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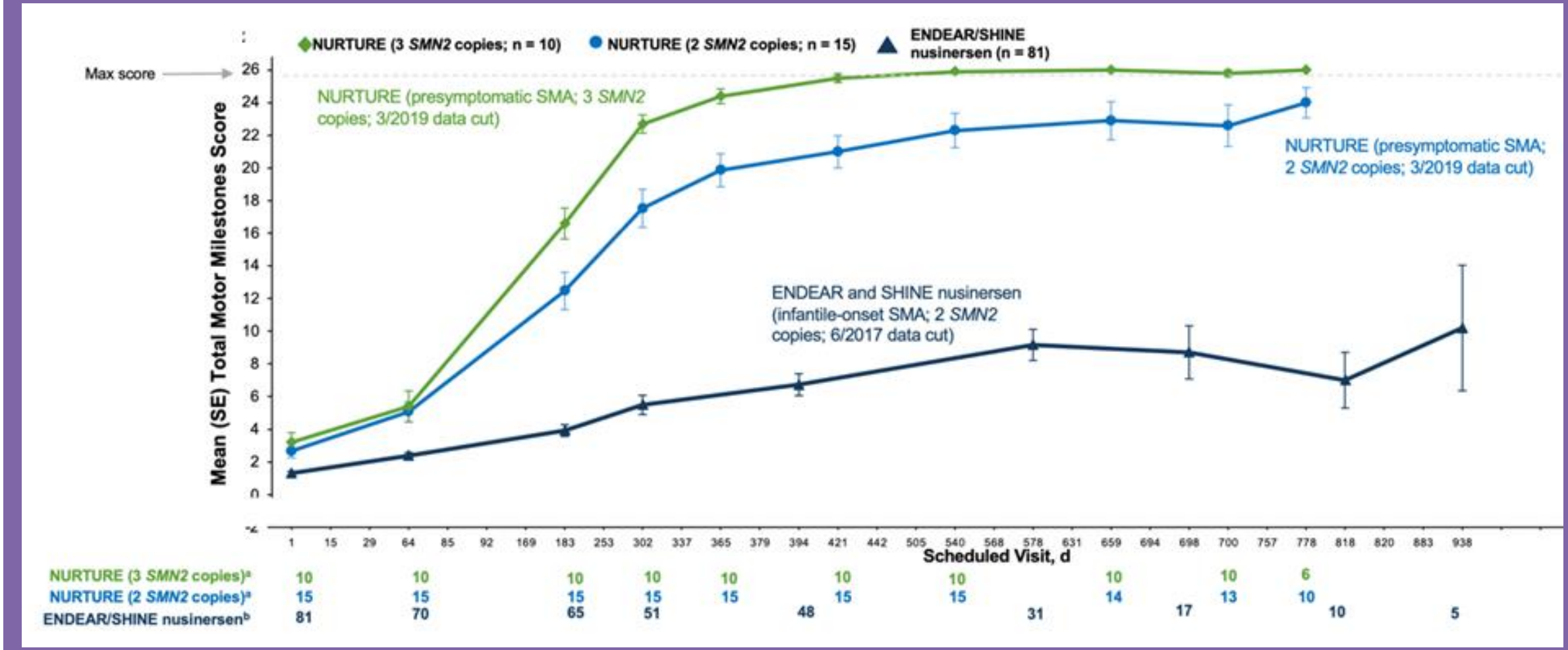
Why do we need NBS?



Alliance first steps

- August 2020 launch of the Alliance
  - Communication support :
    - Policy paper
    - Flyer
- Steering committee
  - Need to collect and gather all the scientific evidence in a unique document
  - Identify best experts
  - Identify and deal with gaps
  - Prevent multiple local pilot trials
- Organising Workshops
  - "Whom to treat?"
- Writing the Whitepaper: ***SMA, Screen at birth, save lives***

Why do we need a Whitepaper?

- To accelerate access in the European countries

- NBS is country relevant
  => You have to prepare an application in each country even in the European Union

- The questions are always the same
  => we need to collect all useful document and gather them into a unique paper

- Each country wanted their own pilot
  => gather all the result of the different European pilots

  Objective : Accelerate access in the different countries

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Whitepaper structure

- SMA fulfils the Wilson and Juenger criteria
- Organising the main publications on the topic to makes them available for each country
- Acknowledging the different questions where there's no consensus
- Find a way to answer it

Whitepaper: some discussions

- Whom to treat ?
  - Workshop between main clinician of the field

No. of SMN2 copies	No symptoms	Mild symptoms	Severe symptoms
1	DMT	BSC+DMT	BSC only
2	DMT	BSC+DMT	BSC only
3	DMT	BSC+DMT	BSC and revisit genetic findings
≥ 4	DMT (define monitoring and potential start of DMT)	BSC+DMT	BSC and revisit genetic findings (check for modifiers)

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Whitepaper: cost-effectiveness

- Cost of a case finding
  - Easy to include into the current NBS program
  - Additional cost per baby between 3-5 €

- Challenging discussion : price of treatment
  - Additional cost of treatment
    - 2 or 3 copies is mainly additional weeks or month , easily balance by the reduction of disabilities
    - 4 copies ?

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Whitepaper: NBS process

- Six main components :
  - Education
  - Screening
  - Diagnosis
  - Management
  - Follow-up
  - Evaluation

- Each country has its own way to address them

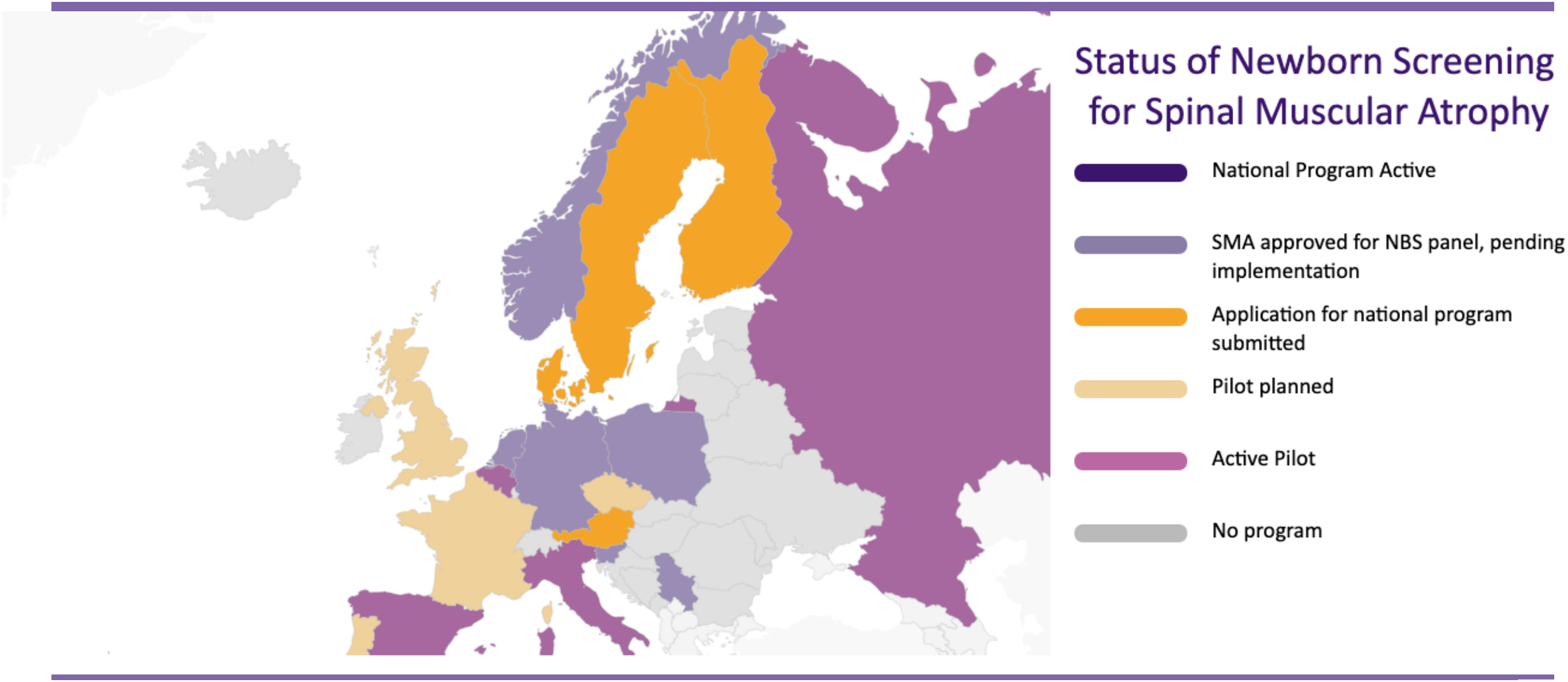
Whitepaper: Ethical issues

- UN : Right of the child to have optimal care
- NBS address babies 2-3 days after birth
- Equal access in the EU
- Parental guilt

- Right not to know
- False positive / false negative

website : www.sma-screening-alliance.org

Current : Status of NBS for SMA in Europe



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What will happen now?

- Dissemination of the Whitepaper
  - Launching
  - Translation in different languages
- State by state support
  - SMA Europe survey
  - Workshops on bottlenecks
- Tool to support EURORDIS' actions
  - NBS shall be EU relevant

=> All the information available on our Website

Can all rare diseases duplicate SMA NBS Alliance?

- We need to simplify the track to include a new disease in NBS
  - EU shall propose a support for national entities to decide
- We need to accelerate the number of diseases screened for at birth
  - More and more effective treatments will be on the market
  - Early treatment will save life
- Adding a specific test each time will become unaffordable for the national insurance
  - We need to think genetic screening more broadly
  - We need to prevent accidental finding
  - We need to preserve if possible the right “not to know” for parents

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Thanks