



NEWBORN SCREENING ADVISORY PANEL RECOMMENDATIONS: SPINAL MUSCULAR ATROPHY

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Presenters



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NEWBORN SCREENING ADVISORY PANEL RECOMMENDATIONS



Background & Process

Advisory Panel Formation

- SBOH approached in 2017 to consider adding Spinal Muscular Atrophy (SMA) to the newborn screening (NBS) panel
- Federal advisory board added SMA to the Recommended Uniform Screening Panel (RUSP) in early 2018
- Internal DOH workgroup formed
- Technical Advisory Committee requested by SBOH
- Technical Advisory Committee met in April 2019

SBOH Criteria for NBS Conditions

- Available screening technology
- Diagnostic testing and treatment available
- Prevention potential and medical rationale
- Public health rationale
- Cost benefit/cost effectiveness

SMA

- Progressive, neuromuscular disorder characterized by muscle weakness, cardiac and respiratory failure
- Autosomal recessive, affects males and females equally
- Spectrum disorder from severe infantile onset to slowly progressing adult onset
- Prevalence 1:15,000
- Recently FDA-approved drug improves survival and physical milestone achievement

Available Screening Technology

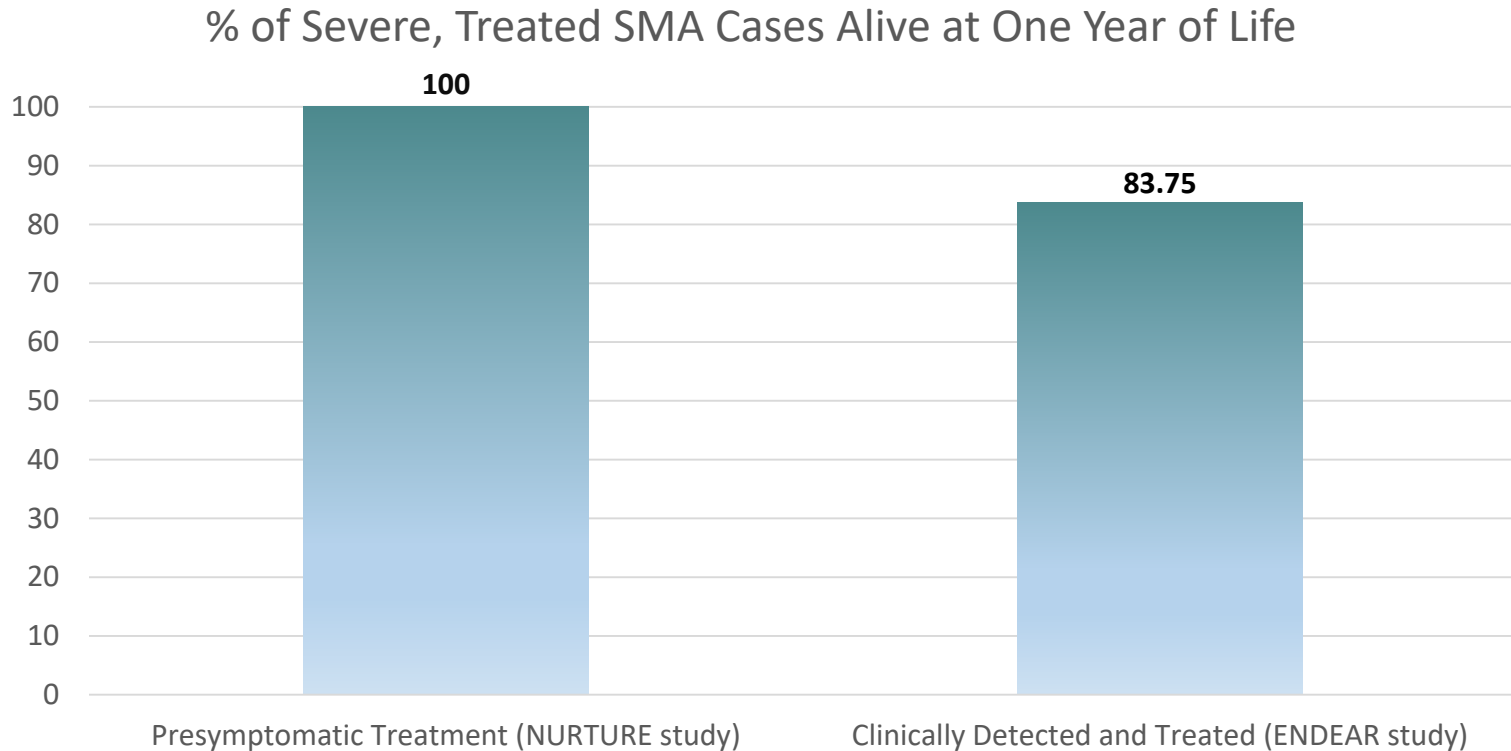
- A sensitive, specific and timely screening test on dried bloodspots exists
 - Test piggybacks on current screen for SCID (bubble boy disease)
 - Known false(-) rate of 5%
- Five states are currently screening (MN, MO, NY, PA, UT)
 - 234,000 babies screened

Diagnostic Testing & Treatment Available

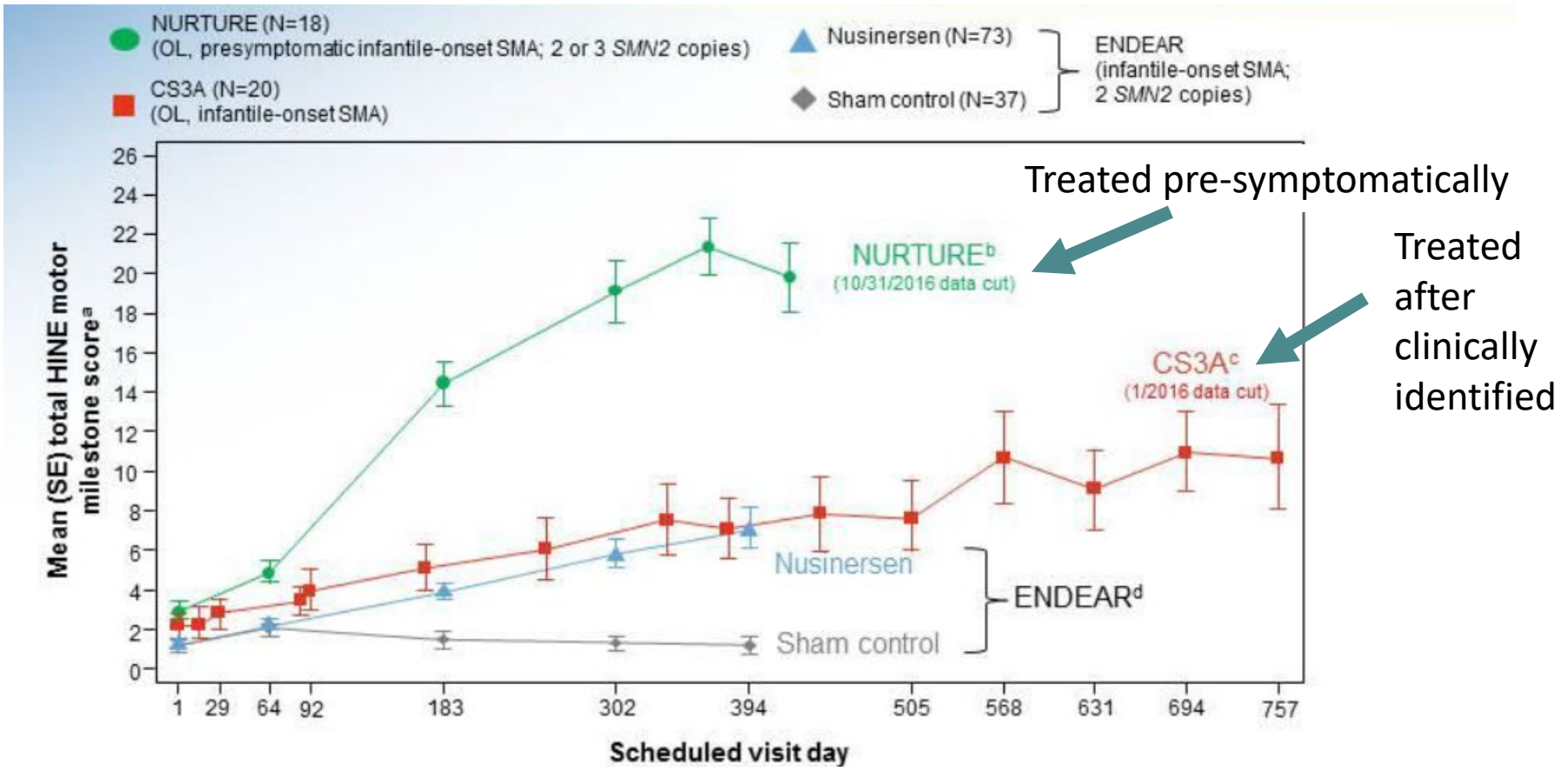
- Testing and treatment is available for affected individuals
 - Laboratory diagnosis - Seattle Children's Hospital
 - Treatment with nusinersen (Spinraza) approved by the FDA in 2016
 - Delivered via lumbar puncture
 - Six doses in first year
 - Three doses per year thereafter
 - Currently only administered at Seattle Children's Hospital
 - Goal to create a network of treatment sites in WA

Prevention Potential & Medical Rationale

- Studies showed treatment reduced mortality and need for mechanical ventilation, with increased milestone achievement



Prevention Potential & Medical Rationale



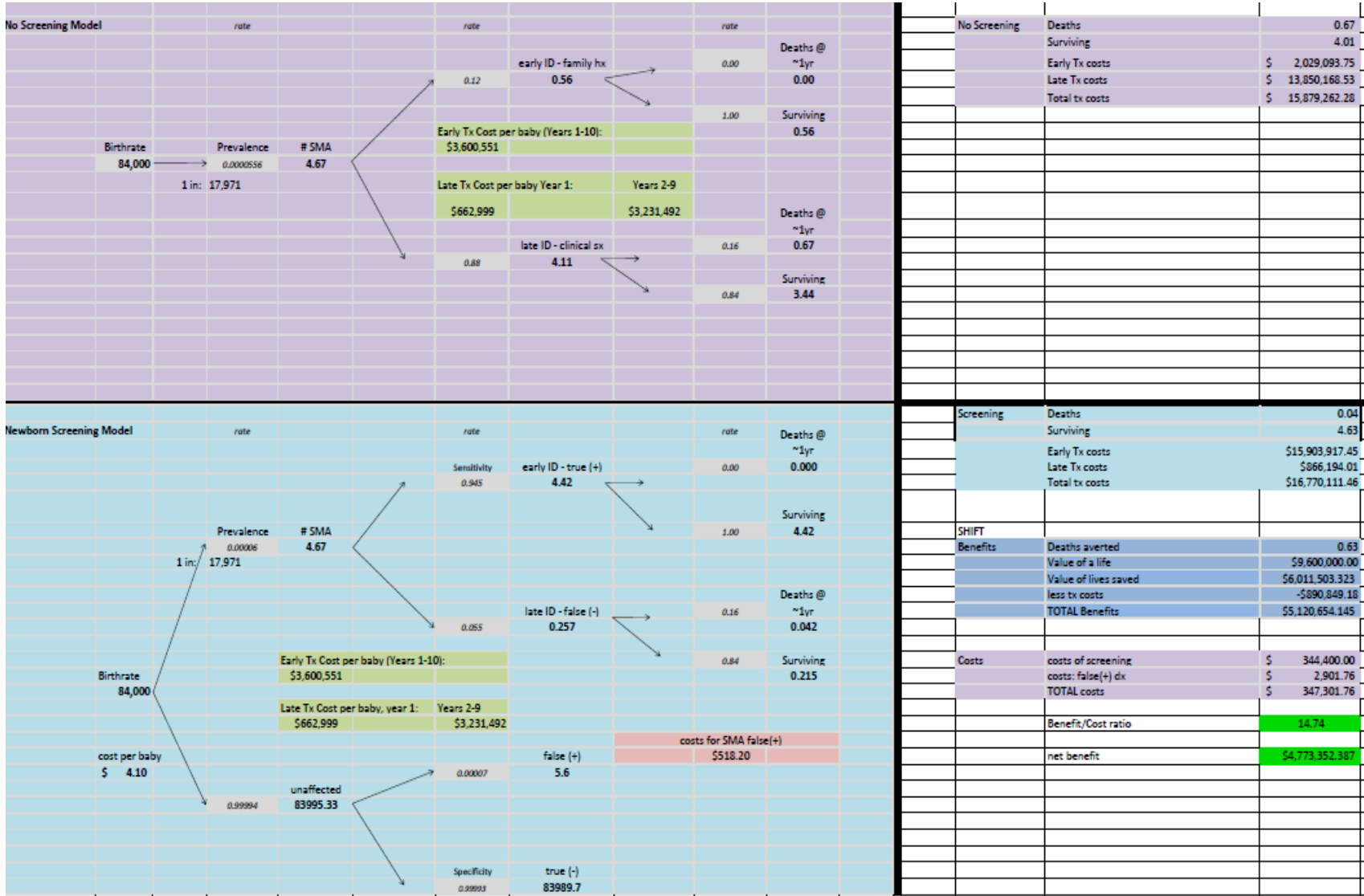
Public Health Rationale

- Most cases have no positive family history – population-based screening is reasonable
 - Estimated to be $\approx 12\%$ based on ENDEAR/NURTURE study demographics

SMA: Cost Benefit Analysis

- Decision tree
 - Compares status quo v. screening model
- Data from
 - Primary literature
 - Reports from MN, MO, NY, UT NBS programs
 - Expert opinion
- Sensitivity analysis – vary assumptions
 - High and low estimates for parameters

SMA: Decision Tree



Cost-Benefit Results

- Lives saved = 0.63 (annual)
 - Value of lives saved \approx \$6 million
 - Shift in treatment costs for early vs late detection = **-\$891,000** (treatment costs increase w/ screening)
 - Cost of screening = \$344,400
 - Costs of false(+) = \$518 per baby
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- Benefit/cost ratio = 14.74
 - Net benefit = \$4,773,352

SMA: Advisory Panel Recommendation

Criteria met?	Yes	No	Unsure
Screening available	15	0	0
Diagnostic testing and treatment available	15	0	0
Prevention potential and medical rationale	15	0	0
Public health rationale	15	0	0
Cost-benefit	14	0	1

SMA: Advisory Panel Recommendation

Criteria met?	Yes	No	Unsure
Screening available	15	0	0
Diagnostic testing and treatment available	15	0	0
Prevention potential and medical rationale	15	0	0
Public health rationale	15	0	0
Cost-benefit	14	0	1
Add to NBS Panel	15	0	0

SMA: Summary Recommendations

- Add to the panel – meets criteria
- Discussion points:
 - Limited long-term data on outcomes
 - About 5% of cases will be missed
 - Detection of late-onset forms
 - Are there enough resources to meet the demands of pre-symptomatic patients needing timely therapy?
 - Cost of drug

Advisory Panel

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Premera

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Regence

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