

Review of NBS Implementation for Added RUSP Conditions

Presented to the Advisory Committee on Heritable Disorders in
Newborns and Children

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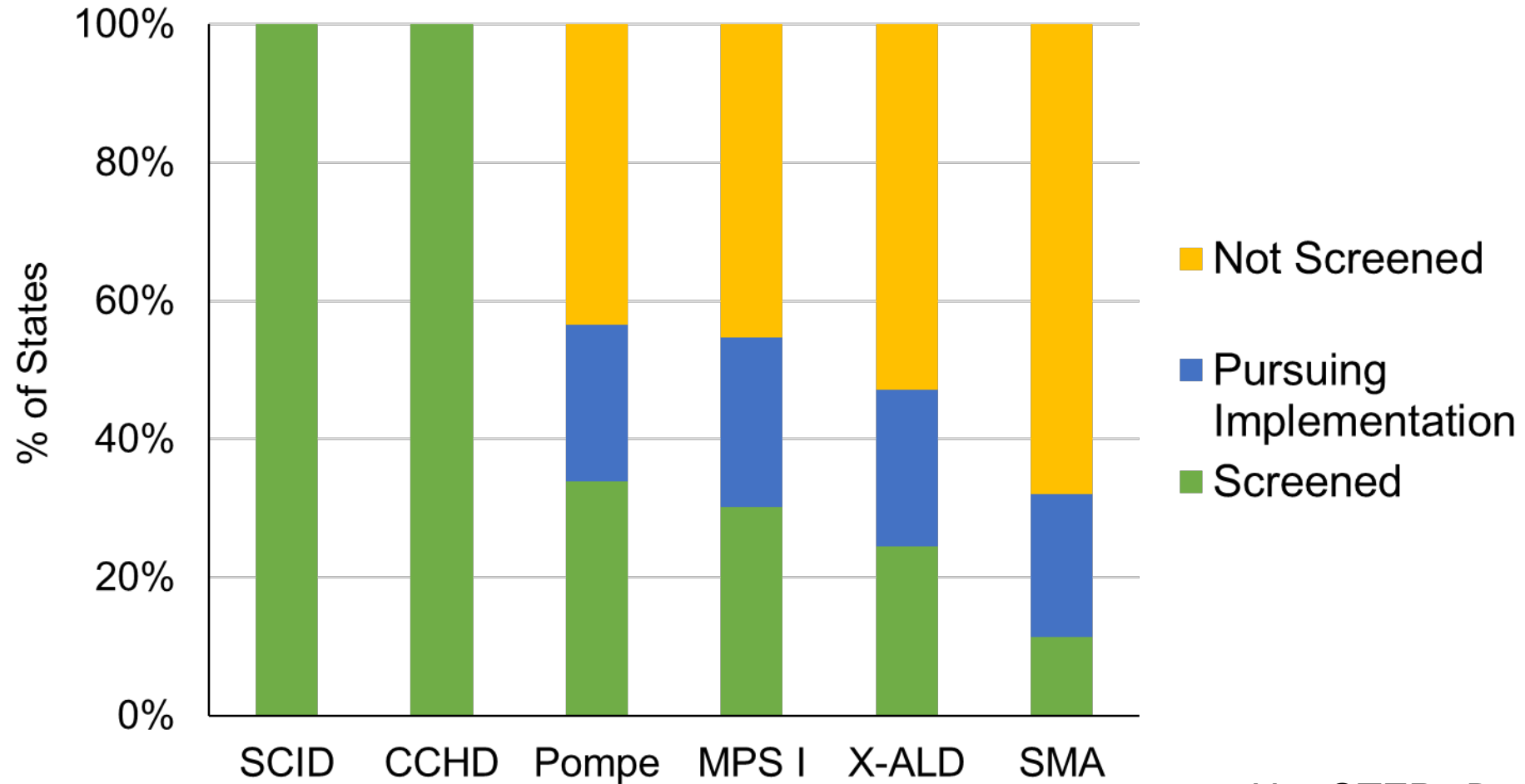
Background

- Review of SCID, CCHD, Pompe Disease, MPS I, X-ALD
 - Primary Goal
 - Request from HRSA/ACHDNC to review implementation of newborn screening for conditions added to the RUSP between 2010-2017
 - Secondary Goal
 - Opportunity to develop standard review methods to address screening implementation and outcomes after addition the RUSP
- Review of SMA for the Report to the Secretary

Historical Reminder

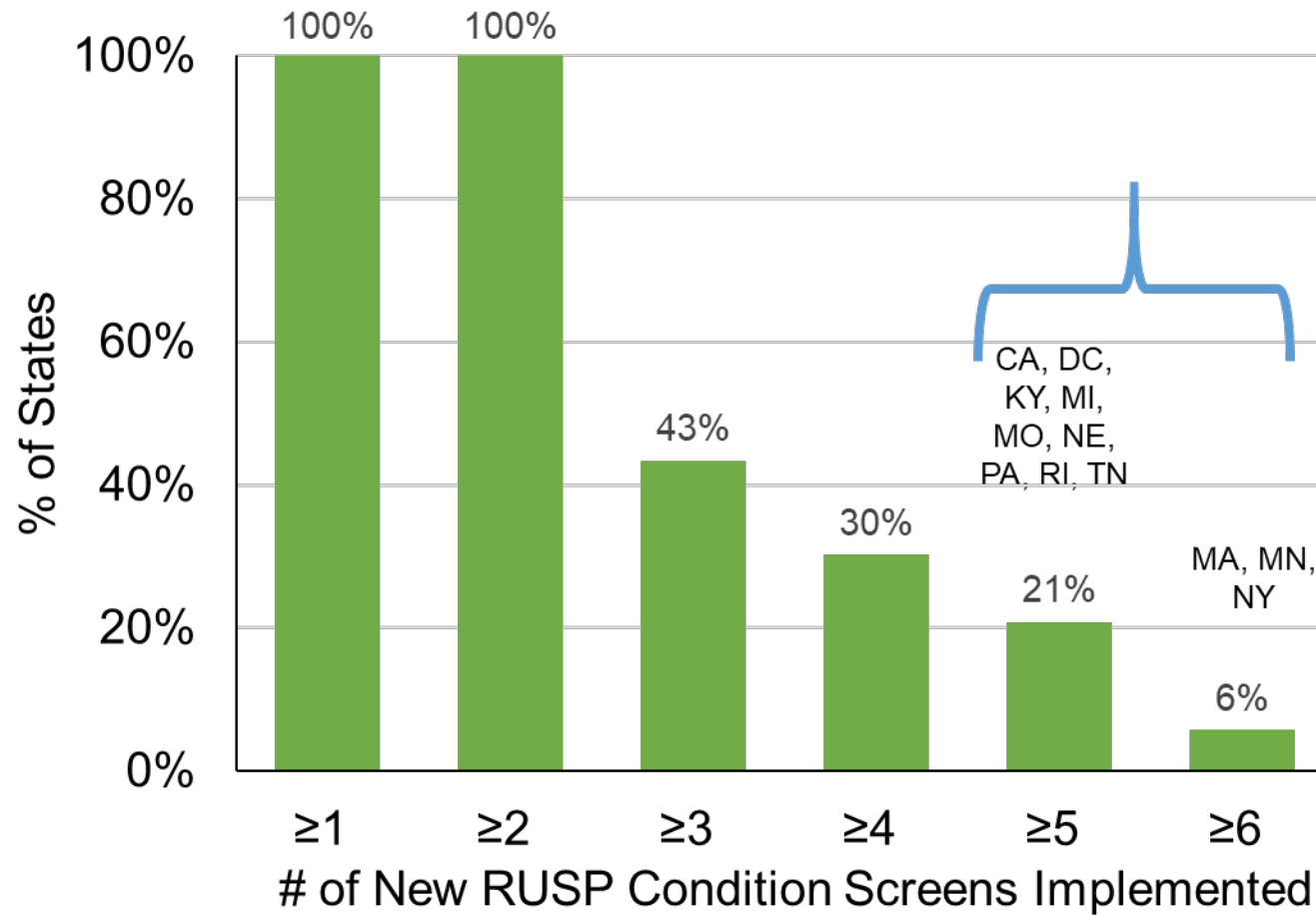
Condition	Year Added to the RUSP
SCID	2010
CCHD	2011
Pompe Disease	2015
MPS I	2016
X-ALD	2016
SMA	2018 (SMA will be reviewed in a slightly different report)

Screening status of each added condition



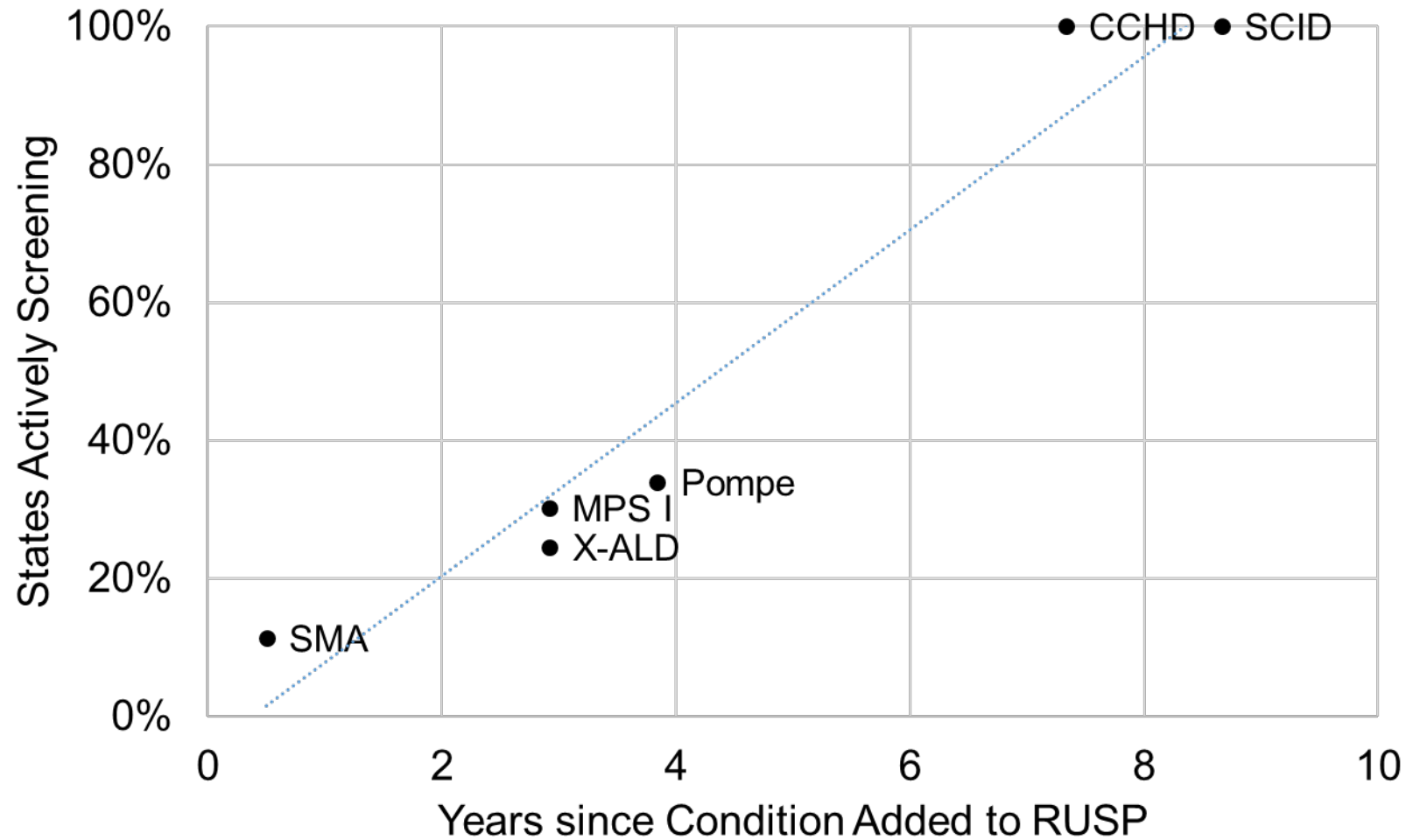
*NewSTEPs Data Repository
(2/20/2019)*

State implementation trends



*NewSTEPs Data Repository
as of 2/20/2019*

Percentage of States Screening related to time on RUSP



Project Scope

- State Implementation
- Public Health Implications
- Clinical outcomes and impact

Guiding issues

- Knowledge about the condition and gaps in understanding when the condition was added to the RUSP
- Status of newborn screening implementation and related long-term follow-up services over time
- Factors related to the condition (natural history, screening, diagnosis, treatment) that has impacted the decision (positively or negatively) to add the condition to newborn screening
- Factors within newborn screening programs that has impacted the decision (positively or negatively) to add the condition to newborn screening
- Contextual factors that have served as barriers or facilitators of adoption of screening for the condition (e.g., issues related to public health activities overall, grant support, advocacy activities, involvement of payers, involvement of clinicians)
- Availability and accessibility of healthcare services after diagnosis
- Changes in understanding of the condition (case definition, phenotypic distribution, diagnosis, treatment) after the condition has been added to the RUSP

Additional Key Questions for SMA

- What activities have states undertaken to implement screening?
- What is known about the clinical outcomes of infants who were treated early?
- What is known about potential harms for infants diagnosed with SMA?

Approach

- For SMA
 - Technical Expert Panel
- Data Sources
 - Previous Report and Advisory Committee Deliberations
 - Targeted interviews
 - Updated systematic literature review
 - Grey Literature Search

Questions?