Newborn Screening

1963: Phenylketonuria (PKU) on filter paper blood samples 1974: First screening for congenital hypothyroidism

1960s-2000s: Testing techniques improved; individual screening programs pilot screening for other conditions

2006: Expert panel convened by American College of Medical Genetics and Genomics recommends conditions for *Recommended Uniform Screening Panel (RUSP)*

2010: RUSP endorsed by DHHS

2011-present:

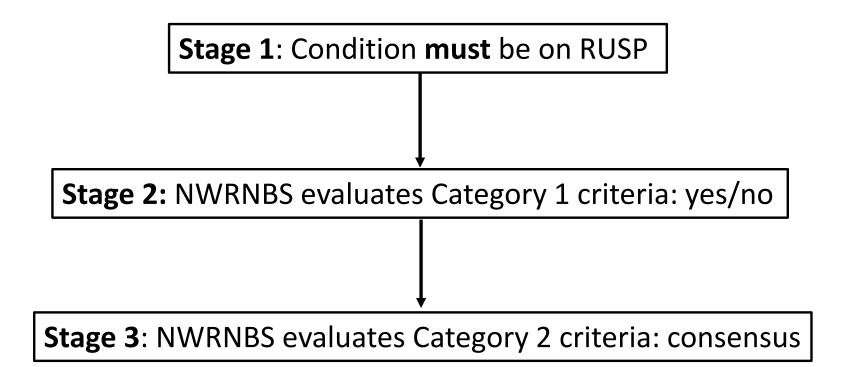
For each new condition considered, independent group evaluates published and unpublished data and makes recommendations to Advisory Committee on Heritable Disorders in Newborns and Children

2019: Oregon Legislature creates NWRNBS advisory board





How is a Condition Added to NWRNBS Program?





Why Be Guided By Science?

- Rare conditions managed by experts
- > Often wide range of severity
- Testing very specialized and must be accurate: must identify all affected babies but not identify too many normal babies
- Screening tests and follow-up confirmatory tests change frequently
- Treatments are being studied for many conditions in newborns where previously there was no treatment



Stage 1:

How Are Conditions Added to RUSP?

Scientific evidence based recommendations from Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC)

- Panel of experts : academic pediatricians and geneticists, directors of newborn screening labs, experts in translational research, members from CDC, NIH and FDA
- Evaluates scientific data for each condition
 - Independent group evaluates *published* and *unpublished* data and makes recommendations to *ACHDNC*
 - Benefits vs risk of screening
 - Readiness of screening programs to do testing



Stage 1: RUSP Core Conditions

- Metabolic diseases
 - Organic acid (9)
 - Fatty acid oxidation (5)
 - Amino acid (6)
- Hemoglobin disorders
- Endocrine disorders

- Other
 - Biotinidase deficiency
 - Cystic fibrosis
 - Galactosemia
 - Pompe
 - SCID
 - Mucopolysaccharidosis
 - xALD
 - SMA
 - Critical heart disease
 - Hearing loss



Stage 2:

NWRNBS Category 1 Criteria

Technical/scientific/objective answer: Yes/No

- Condition well defined in newborns
- Population prevalence known
- Treatment is available
- Early intervention improves outcomes
- FDA-approved dried blood spot testing method available
- Confirmatory diagnostic and specialty testing available
- Appropriate specialized medical consultation available
- Specific condition in funded region of prioritized list of Oregon Health Evidence Review Commission
- Sufficient information for NWRNBS to perform fiscal analysis
- Impact to NWRNBS partners has been assessed



Stage 3: NWRNBS Category 2 Criteria

- Evaluate public health, societal, practical and other issues in adding a specific condition to screen
- > Not "objective": requires judgement
- Relies on consensus of advisory board members



Stage 3:

NWRNBS Category 2 Criteria Consensus Process

After discussion, members indicate their level of approval:

- 1. I enthusiastically agree with the proposal/recommendation
- 2. I agree with the proposal/recommendation
- 3. I am on the fence, have questions, or am neutral but can live with the proposal
- 4. I have serious questions or concerns, but am not willing to block the proposal
- 5. I object and will block the proposal

All members must be able to "live with" recommendation



Stage 3:

NWRNBS Advisory Board Category 2

- 1. Public health benefits of screening outweigh risks and harms
- 2. Population level incidence/prevalence/disease burden significant enough to merit screening
- 3. Diagnostic and specialty testing available and accessible
- 4. Effective treatment is available and accessible. There is equitable care and treatment for disorder.
- 5. Adequate capacity/expertise in NWRNBS to implement testing, reporting, follow-up, and education for providers and parents
- 6. NWRNBS has adequate fiscal resources
- 7. Addition of disorder not prohibitive to NWRNBS contracted partners.

