

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For each new condition considered, independent group evaluates published and unpublished data and makes recommendations to Advisory Committee on Heritable Disorders in Newborns and Children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

**Stage 1:** Condition **must** be on RUSP

**Stage 2:** NWRNBS evaluates Category 1 criteria: yes/no

**Stage 3:** NWRNBS evaluates Category 2 criteria: consensus

# 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  - Independent group evaluates *published* and *unpublished* data and makes recommendations to *ACHDNC*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.