Pilot Study Recommendation

One Case Requirement

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Advisory Committee on Heritable Disorders in Newborns and Children
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Background

Pilot Study Workgroup

- Recognize and support current efforts regarding pilot studies and evaluation
- Identify other resources that could support pilot studies and evaluation
- Identify the information required by the Committee to move a nominated condition into the evidence review process (i.e., define the minimum pilot study data required for a condition to be accepted for evidence review)

What are the minimal necessary data to move a nominated condition to the evidence review process, NOT what evidence is necessary to approve a condition for the RUSP

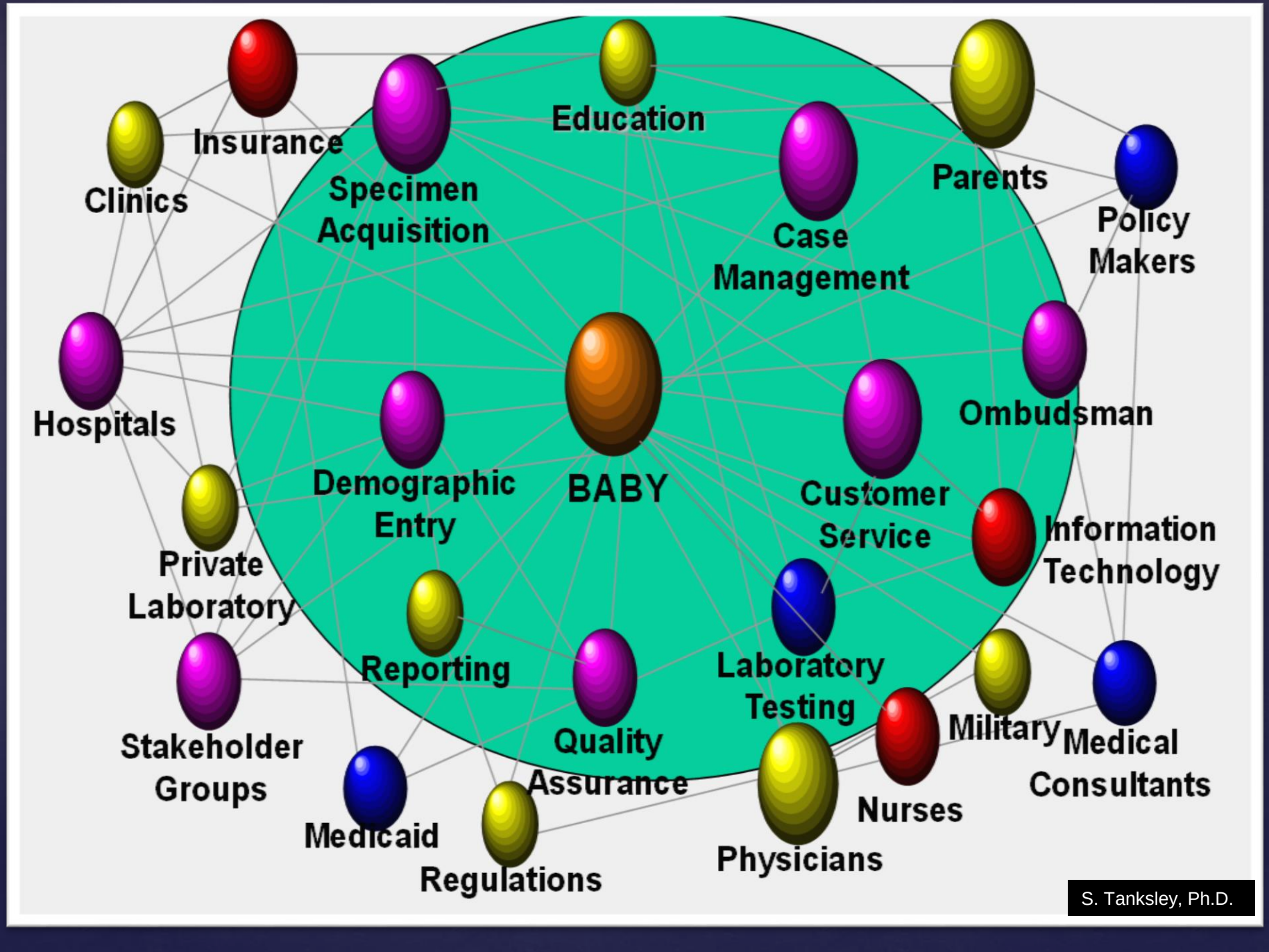
Recommendation 3

Data should be available from pilot studies involving population-based screening of identifiable newborns.

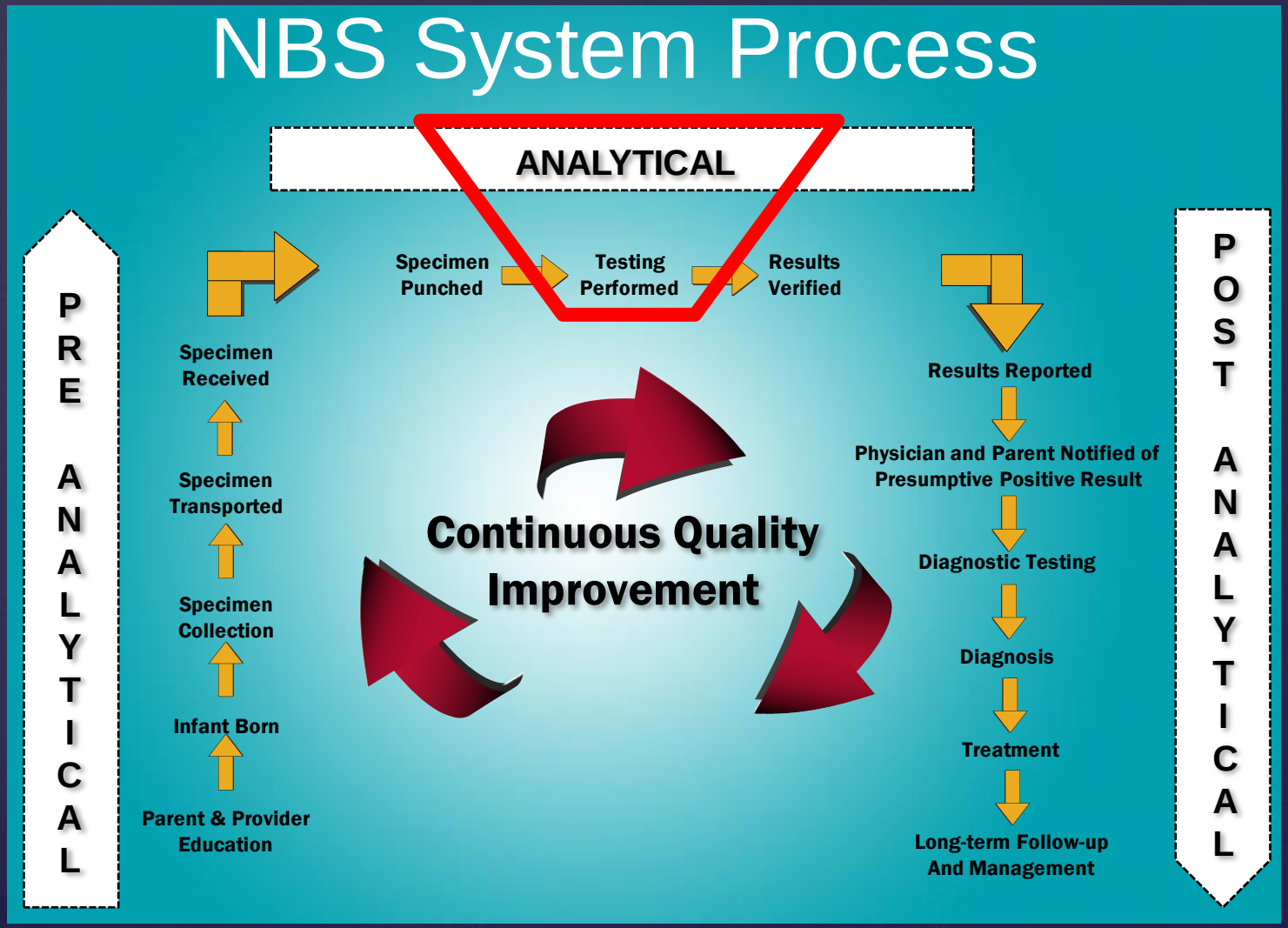
3A) The study should be sufficiently large to identify at least one true positive newborn for the condition under consideration

3B) The population included in the pilot study, and the screening protocol used, should be similar to the US population and to state NBS programs with respect to known prevalence of the condition, the timing and approach to screening, and the screening modality used.

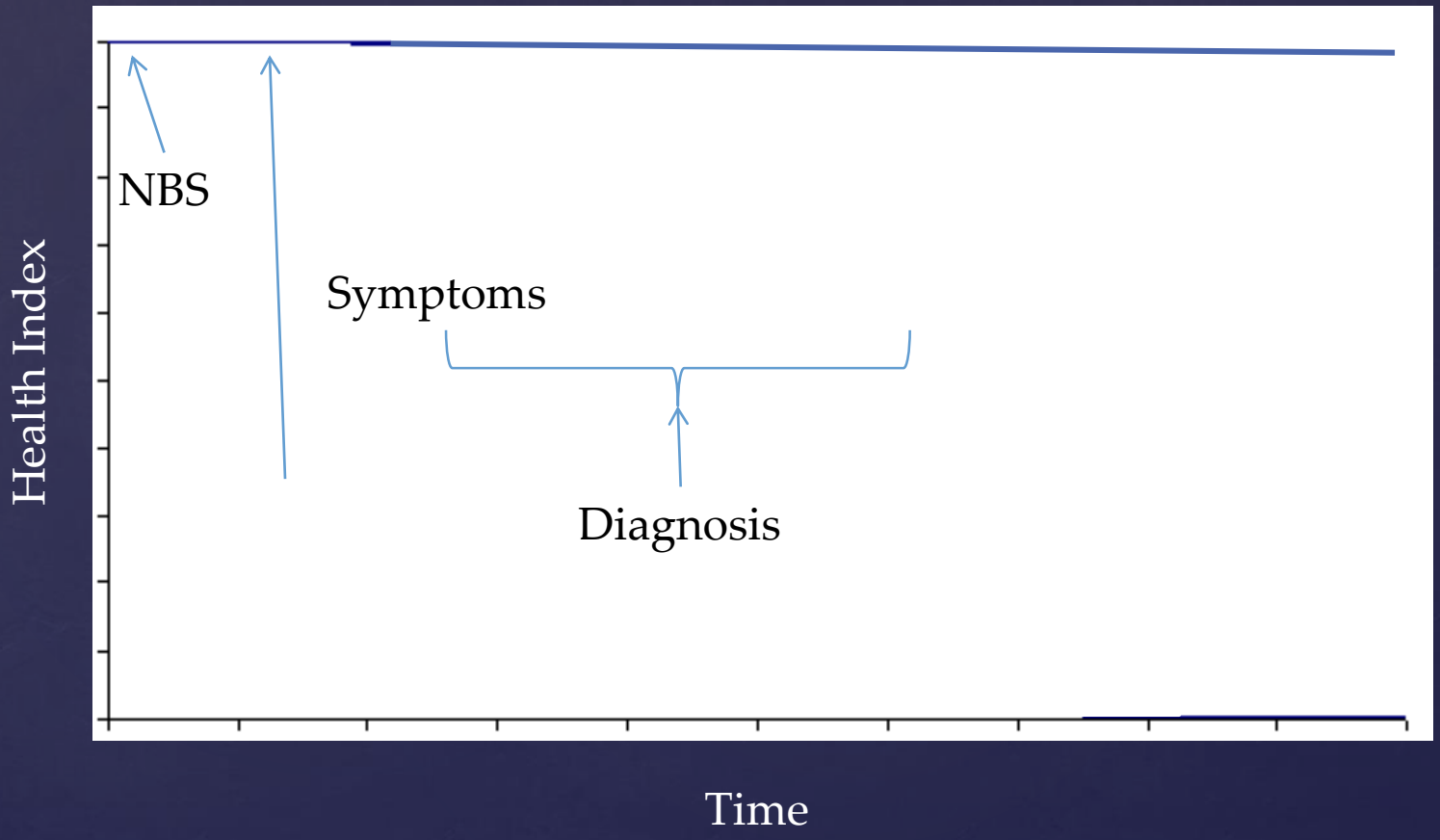

Nomination
Evidence Review
9 months
RUSP
Are sufficient data available to allow for a thorough evaluation of the nominated condition in evidence review?
Are the data robust enough to warrant recommending 4 million babies be screened for a condition every year?

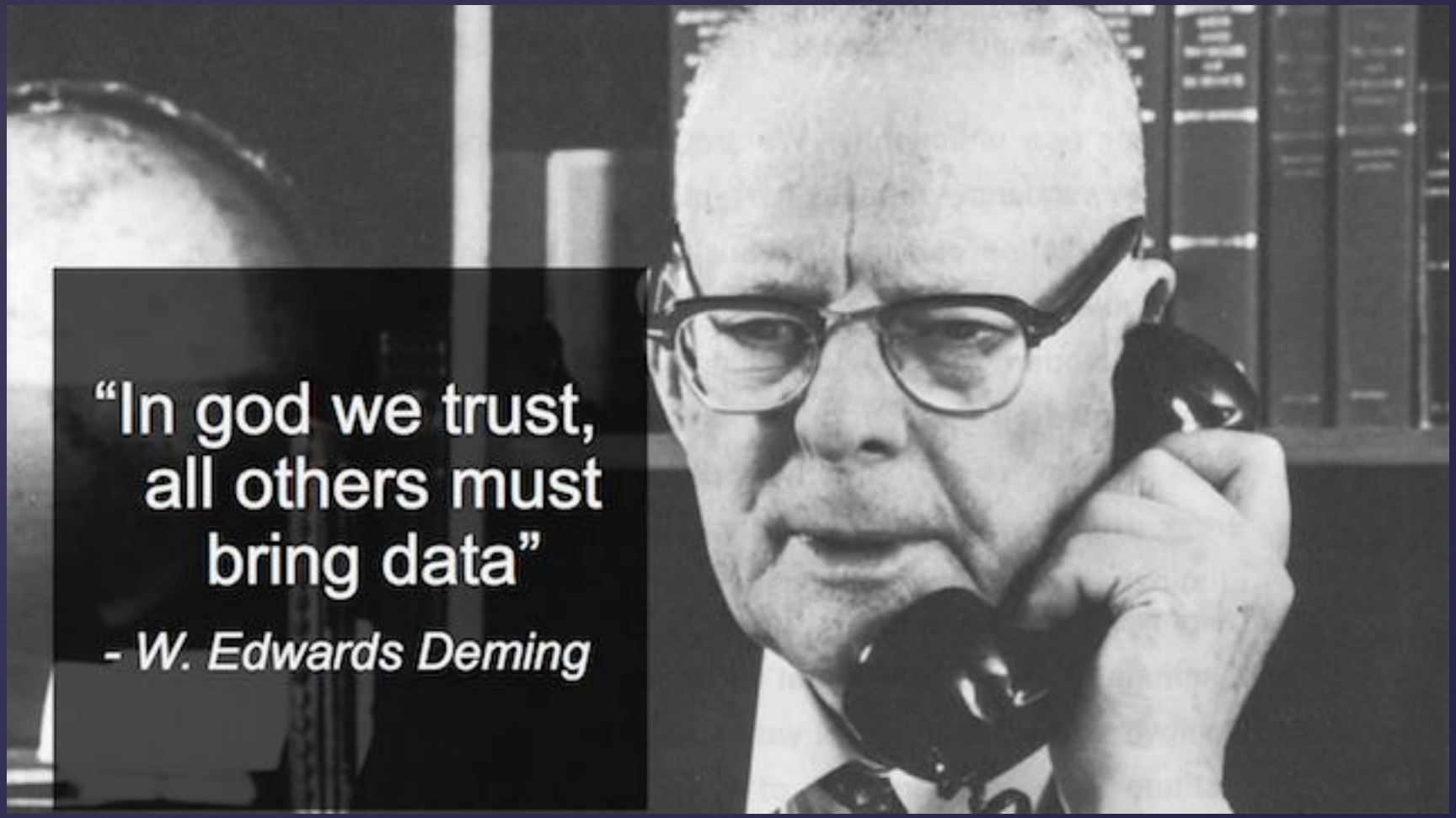

Insurance
Education
Clinics
Specimen Acquisition
Parents
Case Management
Policy Makers
Hospitals
Ombudsman
Demographic Entry
BABY
Customer Service
Information Technology
Private Laboratory
Reporting
Laboratory Testing
Stakeholder Groups
Quality Assurance
Military
Medical Consultants
Medicaid
Nurses
Regulations
Physicians
S. Tanksley, Ph.D.

Retrospective Specimens

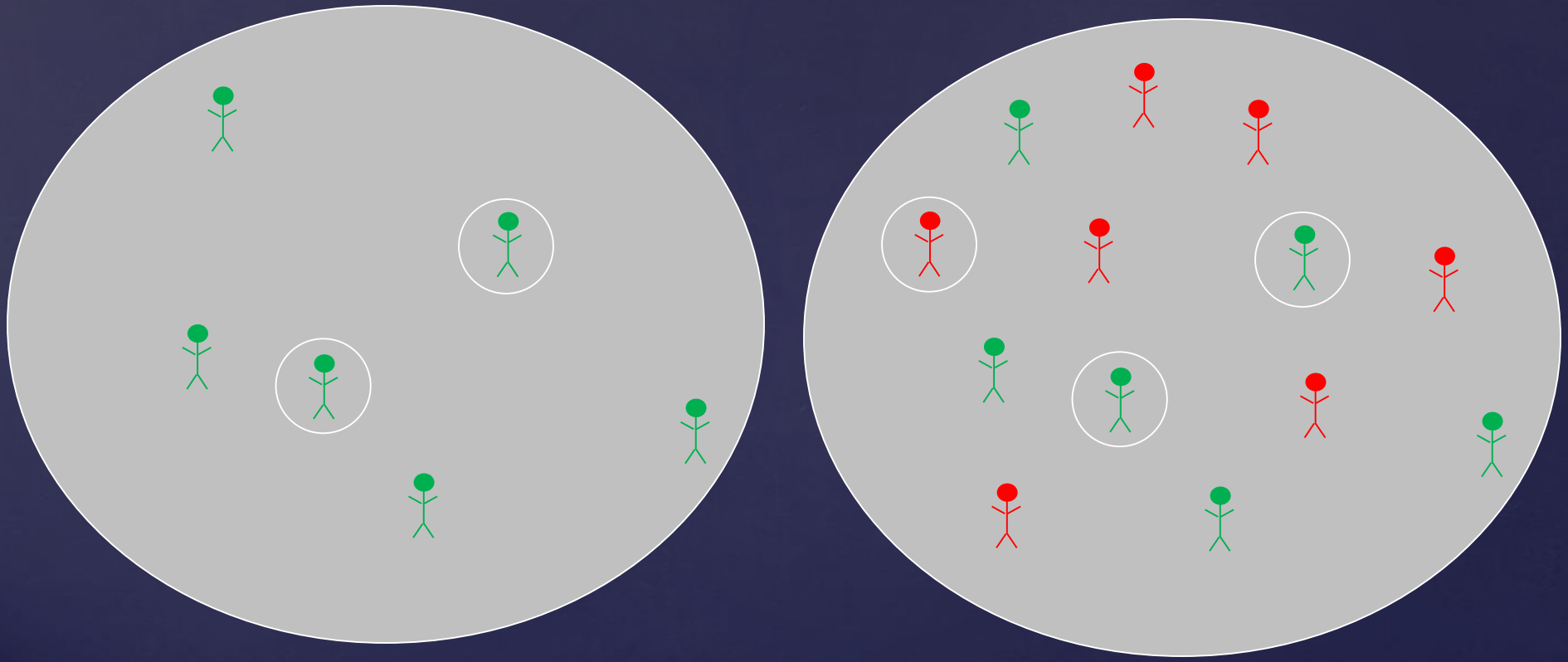


Natural History




"In god we trust,
all others must
bring data"
- W. Edwards Deming

Diagnosis



Diagnosis

tory was not contributory. Routine neonatal screening for guanidinoacetate methyltransferase (GAMT) deficiency was done in this child, who was found to have a mildly elevated guanidinoacetate (GAA) level in the dry blood filter card on the fifth day of life. When recalled, his urinary GAA levels in a first sample were marginally elevated but were within normal limits in a second specimen. The likelihood of GAMT deficiency was considered to be low, although it is well known that false negative results may occur in neonatal screening programs.

Bodamer et. al. Neurology. March 3, 2009

Uniform Process

A lack of uniformity endangers the validity of the process

So What?

Nomination

RUSP

What is 1?

- **Data (permits analysis)**
- **Shows the entire NBS process could work to identify affected newborns (vs. retrospective specimens)**
- **Supports post-NBS incidence review (natural history)**
- **Demonstrates the diagnostic process can actually identify a true case from asymptomatic screen positives (system works, not just NBS test)**
- **Creates uniformity for the review of nominations (standard procedure)**

What is 1?

One is the minimum number of true positive newborns identified in a prospective pilot study needed to demonstrate that data exists from the NBS system to support moving a nominated condition to evidence review

- Pilot Study Workgroup
- Colleagues in Other State Programs
- Committee & Workgroup Members

Many Thanks