

Utilization of the Newborn Screening Translational Research Network (NBSTRN) in a Pilot of Severe Combined Immune Deficiency (SCID) Newborn Screening



N **B S T R N** Newborn Screening Franslational Research Network

Amy Brower, PhD Newborn Screening Translational Research Network



- Session Objectives
- Severe Combined Immune Deficiency (SCID)
- SCID Newborn Screening
- Newborn Screening Translational Research Network (NBSTRN)



- This session will focus on the efforts of states to implement newborn screening for severe combined immunodeficiency (SCID) and will describe related efforts to support SCID research pilots while creating a model for other candidate conditions.
- Findings from a multi-year pilot in a single state as well as results from a four-state pilot will be presented, and a quality assurance strategy for screening in premature infants will be proposed.





- SCID and related T-cell lymphocyte deficiencies are a group of disorders
- Characterized by lack of functioning immune system



- Known as the "Bubble Boy Disease"
- Babies born with SCID appear healthy
- Classic SCID is universally fatal in the first two years without immune reconstitution*





Severe Combined Immune Deficiency

- Early diagnosis is essential for lifesaving treatment
- Historically the best outcomes in siblings of deceased infant
- Many cases occur in families with no identifiable family history





Importance of Family History









Severe Combined Immune Deficiency





SCID Newborn Screening





Goals of the Hunter Kelly Newborn Screening Program



Identify, develop and test the most promising technologies



Increase the specificity of newborn screening and expand the number of conditions for which screening tests are available



Develop experimental treatments and disease management strategies for additional newborn screening conditions, and other genetic, metabolic, hormonal and or functional conditions that can be detected through newborn screening for which treatment is not yet available







Expansion of SCID Newborn Screening Pilots

- NIH initiated project to enable additional states to pilot screening
 - National SCID Pilot Study
- Key Features
 - Initiates pilots in high number birth states (New York, California)
 - High capacity assay development (New York, California)
 - Regionalization model
 - Puerto Rico \rightarrow Massachusetts
 - Louisiana \rightarrow Wisconsin
 - CDC quality assurance program
 - Utilize NBSTRN



National SCID Pilot Study

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Dr. Michele Caggana,

Deliverables

- Analytical
 - Technology
 - Protocols
 - Analysis Tools
 - Quality Assurance Methods
 - Pilot data set
- Clinical















NBSTRN Role

- Convene Experts
- Facilitate and Host Monthly National Calls
- Develop Analytical Tools
 - R4S
 - LTFU
- Develop VRDBS and Sample Banks
- Disseminate Findings

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SCID Data Portal

- Goal was to collect, aggregate and analyze de-identified screening data generated during the pilot
- Enables real-time laboratory performance quality improvement
- Stores laboratory protocols
- Facilitates tracking of emerging findings
- Provides disease definitions
- Available to any newborn screening program and or researcher

NEWBORN SCREENING COLLABORATIVE PROJECTS



Welcome to the Newborn Screening Domain

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ÓË	LSD Lysosomal Storage Disorders

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DOCUM	ENTATION									



- Screening technology is a robust biomarker for SCID and profound T cell lymphopenia
 - Future investigations of this valuable biomarker will accelerate research in immunology.
- Majority of classic SCID cases have zero TREC
- Molecular etiology of low TREC cases is varied
- Incidence rates different from published findings
- Incidence rates vary by race and ethnic categories







Thank You!





Eunice Kennedy Shriver National Institute of Child Health and Human Development, National Institutes of Health NBSTRN -HHSN27520080001C SCID Trial -HHSN267200603430

