NBSTRN SCID
National Call
March 22, 2013

Call in number: 1-866-671-5988
Conference code: 841-900-2082
Standing Agenda – Led by Dr. Michele Caggana

- Introduction of New Participants
- Implementation Status Review and Stakeholder Reports
- Effort to Report on National Experience
- CDC Update
- IDF Meeting
- Resources and Tools
- Submitted Discussion Items
- Discussion
March 2013 Reports

- Report from States Currently Screening
- Report from States Not Currently Screening
- Report from Partners and Industry
- Other Reports
Purpose – To report on the efforts and findings of the state-based newborn screening programs and the clinicians who diagnose and treat newborns identified with SCID and related T lymphocyte deficiencies.

Scope – This report describes the screening, diagnosis and treatment activities of states that are actively screening for SCID. This is a descriptive report only and will not include statistical analysis of the submitted data.
Effort to Report on National Experience

- **Rationale** – Capitalize on NICHD funded pilot by creating infrastructure and mechanism to collect information regarding natural history of conditions resulting from newborn screening and health outcomes of affected newborns. Established infrastructure available for future grantees and investigators.

- **Participation** – Participation is open to states that are screening for SCID and to individuals within the state and/or program who play a role in the screening, diagnosis and treatment of affected newborns.
Effort to Report on National Experience

- **Authorship** – Individuals who submit data and/or are identified by the state programs as an essential contributor will be asked to review the draft manuscript and included as an author.

- **Mechanism** – Contributors may submit data using the R4S SCID module or by completing a worksheet that lists the R4S data elements. The NBSTRN-CC will compile the data and organize the writing of the report.
Privacy/Security – The requested data does not contain information considered identifiers (also called protected health information, or PHI) for the purpose of HIPAA compliance.

Future Efforts – The submitted data will be maintained for future efforts and made available to researchers, public health team members and clinicians in a transparent and open process.
Effort to Report on National Experience

- Planned Data Elements to Collect
  - R4S SCID Module Items
  - State
  - Date NBS-SCID initiated
  - Total # of infants screened (indicate start and end date of data provided)
  - % Male/Total Infants
  - % of total screened by race/ethnicity
  - Total # of Positive NBS-SCID (meeting analytical criteria for state for True positive) : includes pre-term alone infants
  - % of Positive NBS-SCID by gender (male/total)
  - % of Positive NBS-SCID by race/ethnicity
  - Total # Pre-term alone infants identified by NBS-SCID
  - Total # of non-pre-term alone infants identified by NBS-SCID
  - Total # Term referred for f/u
  - Total # pre-term referred for f/u
- CLSI Document – Bob
- NSQAP TREC PT Program – Joanne
- NSTRI – Bob, Francis
National Conference Highlights

- IDF Advocacy Day on Thursday
- Thursday Orientation Reception
- Stars and Zebra Stripes Gala
- THINK ZEBRA! Silent Auction
- Youth will visit the Maryland Science Center for their off-site trip on Saturday afternoon.
- IDF Family Night at the National Aquarium
Saturday Professional Medical Education Program co-sponsored with Clinical Immunology Society
New & Updated Sessions at 2013 National Conference

- Orientation for First timers
- Families Facing Chronic Illness
- Allergies and Primary Immunodeficiency Disease
- Can I Really Ask that Question? Getting the Answers You Need for Today’s World
- Managing Depression and Anxiety
- Common Variable Immunodeficiency Frequently Asked Questions
Newborn Screening: SCID, related T-Cell Lymphocyte Deficiencies and Beyond

Women’s Issues

Parenting Skills for the Whole Family

Making a Seamless Transition to Adulthood

Living Life to the Fullest as an Adult with Primary Immunodeficiency
Resources and Tools

National SCID Pilot Study Report

- 1. New York Standard Operating Procedure for TREC Analysis
- 2. New York SCID Validation
- 3. LA/WI Validation and Standard Operating Procedure for Receipt for Specimens and Transmission of Results
- 4. PR/MA Validation and Standard Operating Procedure for Receipt for Specimens and Transmission of Results
- 5. Assay Elements Spreadsheet
- 6. NY Laboratory Algorithm
- 7. CA Laboratory Algorithm
- 8. NY Clinical Algorithm
- 9. CA Clinical Algorithm
- 10. LA Information for Providers
- 11. CA Educational Materials for Families and Providers
- 12. NY Educational Materials for Families and Providers
ACMG ACT Sheet

- Clinical Description
- Recommended Actions
- Clinical Considerations
- Diagnostic Evaluations
- Referrals
- Additional Information

Newborn Screening ACT Sheet
Severe Combined Immunodeficiency (SCID) and Conditions Associated with T Cell Lymphopenia

Condition Description: Severe Combined Immunodeficiency (SCID) includes a group of rare but serious, and potentially fatal, inherited immune disorders in which T lymphocytes fail to develop and B lymphocytes are either absent or compromised. Impairment of both B and T cells leads to the term “combined.” Untreated patients develop life-threatening infections due to bacteria, viruses, and fungi. The screening test for T cell receptor excision circles (TRECs), a byproduct of normal T cell development, identifies SCID, as well as certain related conditions with low T cells. For example, DiGeorge Syndrome with impaired thymus development may cause low T cells and low TRECs.

**YOU SHOULD TAKE THE FOLLOWING ACTIONS:**
- Contact the family to inform them of the newborn screening result. Point out that additional tests are required to determine whether the baby actually has an immune deficiency.
- Avoid exposing patient to illness pending completion of testing.
- If the infant has any signs of illness, refer to a pediatric hospital right away for evaluation, administration of immunoglobulin, and antibodies.
- If the infant requires transfusion of any blood product, be sure that only leukoreduced, irradiated products that are negative for cytomegalovirus (CMV) are used.
- DO NOT give live attenuated viral vaccines, which could cause serious disease in a baby with SCID. This vaccine is to be given only after an immunology specialist confirms that the baby’s immune system is normal.
- Consult with a specialist in pediatric immunodeficiency disease (consult with a pediatric allergy/immunology...

American College of Medical Genetics ACT SHEET
Resources and Tools

- **R4S SCID Module**
  - Diagnosis Categories
  - Laboratory Protocols

- **REDCap Case Report Form**
  - Electronic data entry
  - Paper data entry
  - Import templates
### Submitted Discussion Items

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<th>Item</th>
<th>Submitted By</th>
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<tbody>
<tr>
<td>Reporting of clinical findings, natural history, and health outcomes in individuals identified by newborn screening</td>
<td>Marcia Boyle, President and Founder Immune Deficiency Foundation</td>
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Source of AAAAI specialist list referenced on ACMG ACT Sheets

- AAAAI provided the list to ACMG – Drs. Elena Perez and Lisa Kobrynski compiled the lists.
- ACMG contact for ACT sheets is Alisha Keehn and she suggests that individuals interested in being added contact Dr. Perez and copy ACMG.
  *Elena Perez eperez2@health.usf.edu*
  *Alisha Keehn akeehn@acmg.net*
Running List of Discussion Items (Continued)

- **Premature Infants**
  - Rescreen protocol varies by States
    - DE – 38 weeks along with mandatory second screen
    - NY – follow transfusion protocol
  - Most resolve to negative as gain weight

- **Biggest Hurdle**
  - NY – High-throughput DNA extraction, supplies, programming were biggest hurdles.
Running List of Discussion Items (Continued)

- CA – NICU kids were biggest hurdle.
- DE – Establishing analytical standards but CDC training helped.

- Unanticipated Benefits
  - NY – Buy-in and support from immunologists.
  - DE – Opportunity to set up a molecular laboratory and combine multiple functions under one roof.
  - CA – Updated March 2013 - 1-5 TREC with normal b-Actin are a cause for "Urgent referral to flow cytometry" since these are quite likely to have real SCID
Running List of Discussion Items (Continued)

- **Protocols, Educational Brochures**
  - Posted on NBSTRN website
  - Dr. Caggana’s National SCID Pilot Report
  - Educational Brochures provided by NY and CA
  - Other information provided by WI and MA

- **Cost**
  - DE - $3 screen only not FU
  - NY – Does not charge a NBS fee, estimate $3 for screen.
  - CA - $3 to $5 screen only, raised NBS fee $10 – flow, follow-up, IT changes, etc.
Treatment out of State

- NY – Estimate half of cases on Medicaid and Medicaid pays for treatment. Private insurance also pays.
- CA – Not an issue.
<table>
<thead>
<tr>
<th>Responsible Party</th>
<th>Action</th>
<th>Timeline</th>
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<tbody>
<tr>
<td>All</td>
<td>Review and update status of implementation for your state</td>
<td>Before next call March</td>
</tr>
<tr>
<td>All</td>
<td>Submit questions and/or agenda items</td>
<td>Before next call March</td>
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