Severe Combined Immunodeficiency (SCID) In-Person National Meeting

Meeting Summary and Notes

August 8 – 9, 2017
Washington, DC

This meeting and all related activities are supported by a Cooperative Agreement #UG5MC27837 from the Health Resources and Services Administration (HRSA). Meeting presentations are solely the responsibility of the authors and should not be construed as the official position or policy of, nor should any endorsements be inferred by HRSA, HHS, or the US Government.

APHL | NewSTEPs

A Program of the Association of Public Health Laboratories™

TABLE OF CONTENTS

ACKNOWLEDGEMENTS

The Association of Public Health Laboratories would like to recognize the National Severe Combined Immunodeficiency (SCID) In-Person Meeting Planning Committee members who generously donated their time and input for the development of the meeting agenda.

Lynn Albizo, Esq.
Mei Baker, MD, FACMG
Amy Brower, PhD
Carol Johnson
Lisa Kobrynski, MD, MPH
Francis Lee, PhD, MSc
Jelili Ojodu, MPH
Emily Phillips, BSN, RN, CCRC
Jamie Sexton
Ruthanne Sheller, MPH
Sikha Singh, MHS, PMP
Suzanne Skoda-Smith, MD
Marci Sontag, PhD
Jennifer Taylor, PhD
John Thompson, PhD, MP, MPA
Beth Vogel, MS, CGC
Andrea Williams, BA

We also would like to thank our presenters, speakers, and facilitators who shared their experiences.

Carol Johnson
Francis Lee, PhD, MSc
Marci Sontag, PhD
Careema Yusuf, MPH
Jennifer Puck, MD
Lisa Kobrynski, MD, MPH
Mei Baker, MD, FACMG
Carlos A. Saavedra-Matiz, MD
Susan E. Prockop, MD
Christine Seroogy, MD
James, Verbsky, MD, PhD
Suzanne Skoda-Smith, MD
Jaclyn Seisman, MPH
Lynn H. Albizo, Esq.
Jamie Sexton
Charlotte Cunningham-Rundles, MD
Tracey Bishop, BS
Amy Brower, PhD
Joyal Meyer, BSN, RN

BACKGROUND

Severe Combined Immunodeficiency (SCID) was added to the Recommended Uniform Screening Panel (RUSP) in May 2010. As of August 2017, 47 newborn screening (NBS) programs offer universal newborn screening for SCID and the remaining NBS programs continue to work towards full implementation. Supporting widespread adoption of early and accurate laboratory detection of SCID in newborns requires relationships with specialists in immunology and the development of clinical referral networks that can assist with follow-up and treatment.

PURPOSE

The purpose of this meeting was to convene SCID newborn screening stakeholders and to facilitate the strengthening of relationships between the SCID clinical network and the newborn screening community within each state. During this meeting participants shared solutions for SCID newborn screening implementation in order to support ongoing quality practices, quality data collection and interpretation across the United States. Participants included representatives from state newborn screening laboratories, follow-up programs, and clinicians.

OBJECTIVES

- Provide input and offer expert guidance on implementation of SCID
- Share information on progress toward universal SCID newborn screening in the United States and discuss emerging challenges
- Identify quality improvement initiatives in SCID newborn screening
- Develop interactive relationships between clinical and NBS program staff to facilitate collaboration
- Consider case definitions for SCID and the value of data reporting

INTRODUCTION

In 2014, the Association of Public Laboratories was awarded $4 million dollars through a two-year cooperative agreement from HSRA. At that time, less than half of states were screening for SCID. Under this cooperative agreement, APHL, in close partnership with the Colorado School of Public Health, provided funding to support full implementation of SCID newborn screening in 12 states and programs. Through this collaborative initiative, NewSTEPs provided technical and financial assistance to help expand the capacity of existing newborn screening programs to incorporate the addition of SCID screening, education and/or follow-up health care services.

In July 2015, APHL held a national meeting which addressed current challenges faced by states newborn screening programs in implementing SCID screening including the integration of new technology, laboratory staffing to conduct screens, clinical follow-up capacity and resources, funding for personnel, equipment, education and legislative or statutory approval. The audience during that first national APHL SCID meeting was laboratory and follow-up staff from newborn screening programs.

Two years later, the SCID community has come has come so far, with 47 newborn screening programs now offering population screening for SCID. It was an opportune time to hold a second meeting which focused on engaging the newborn screening community and clinicians to strengthen clinical referral networks within each state and region.

The format for this meeting was not unprecedented. With an expanding field, new screening technologies and more conditions being added to the RUSP, the newborn screening community has had many reasons to convene for joint strategizing. NewSTEPs 360 engaged the newborn screening community with the clinical community at the Cystic Fibrosis Foundation Meeting in June 2016 in Denver, which considered issues of timeliness in cystic fibrosis screening and diagnostic testing. Additionally, NewSTEPs held a meeting in Orlando in October 2016, which brought together short follow-up coordinators with state pulmonologists to brainstorm ways to improve timeliness of state NBS systems and address other issues affecting state system performance.

While this meeting focused on educational presentations, there were many opportunities for interactive engagement and idea sharing. Participants were encouraged to share their experiences, to learn from others, and to tackle existing issues together.

SCID NEWBORN SCREENING STATUS

Figures 1 - 10 depict the evolution of SCID newborn screening from 2008 through August 2017.

Figure 1

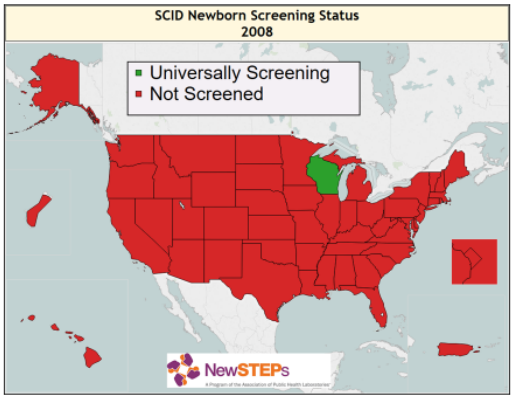


Figure 2

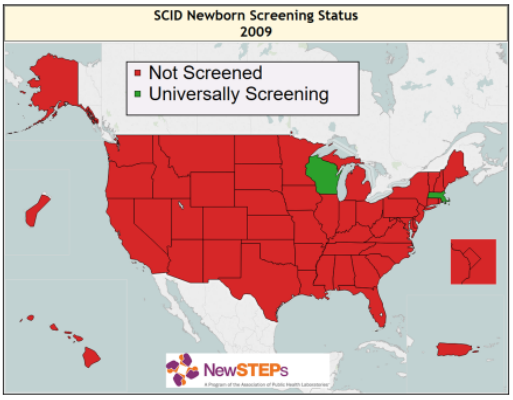


Figure 3

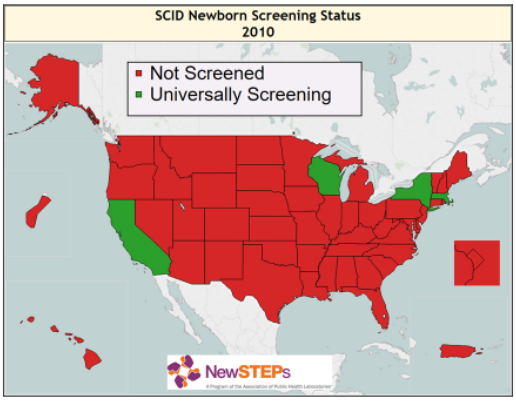


Figure 4

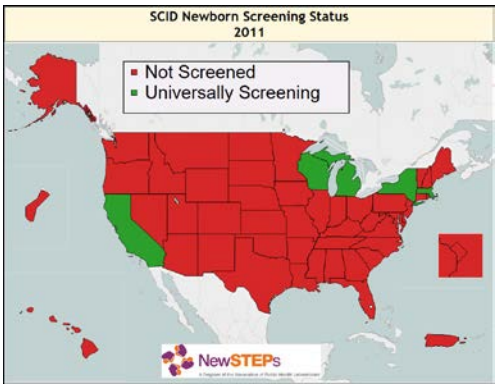


Figure 5

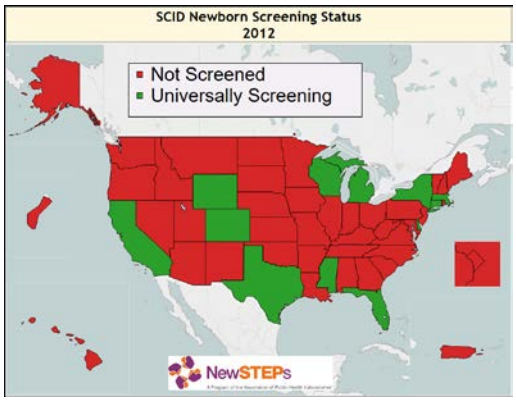


Figure 6

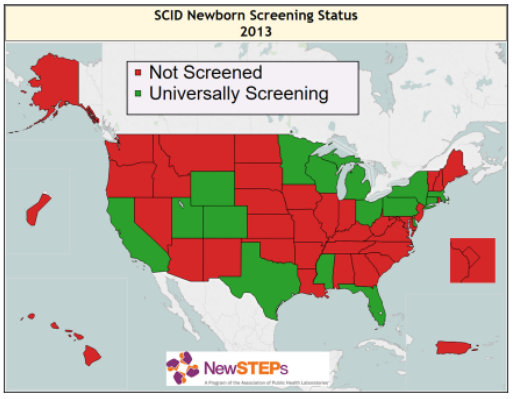


Figure 7

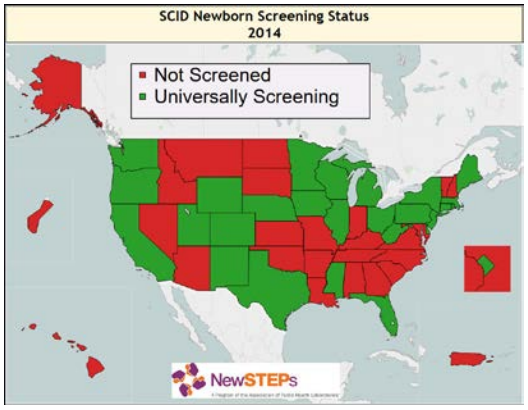


Figure 8

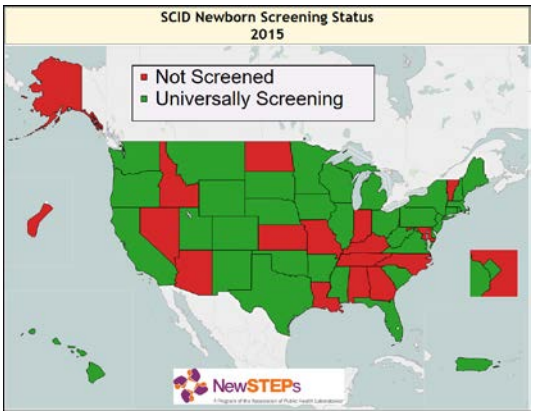


Figure 9

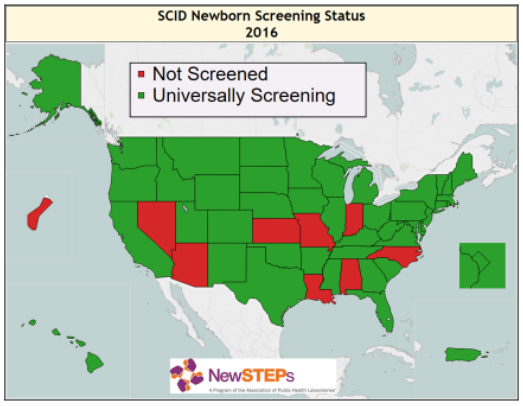
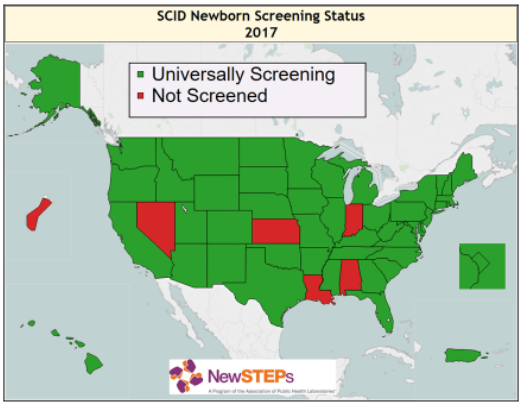


Figure 10



MEETING SUMMARIES

Brief summaries of each of the presentations presented during the in-person National Severe Combined Immunodeficiency (SCID) meeting of August 2017 are found below. The purpose of this meeting was to convene SCID newborn screening stakeholders and to facilitate the strengthening of relationships between the SCID clinical network and the newborn screening community within each state. During this meeting participants shared solutions for SCID newborn screening implementation in order to support ongoing quality practices, quality data collection and interpretation across the United States. Participants included over 100 representatives from state newborn screening laboratories, follow-up programs, and clinicians, representing 38 states.

The presenters are listed in order of appearance during the SCID In-Person Meeting. For additional details, please contact Ruthanne Sheller (ruthanne.sheller@aph.org).

State of SCID Newborn Screening in the United States

Presenter: Ruthanne Sheller, MPH, NewSTEPs, Association of Public Health Laboratories

Ruthanne Sheller, MPH, Specialist for NewSTEPs at the Association of Public Health Laboratories, reviewed the progression of SCID newborn screening in the United States. In 2014, the Association of Public Laboratories was awarded $4 million dollars through a two-year cooperative agreement from HSRA. At that time, less than half of states were screening for SCID. Three years later, as of August 2017, 47 newborn screening programs offer universal SCID newborn screening.

The Newborn Screening Process: Birth to Confirmation

Presenter: Carol Johnson, Iowa Newborn Screening Follow-Up Program

Carol Johnson, Follow-Up Coordinator of the Iowa Newborn Screening Program, provided an overview of the newborn screening process, capturing steps and complexities from pre-analytic through post-analytic stages. Carol highlighted the variability that exists in SCID NBS algorithms across states. Programs are unique, but all working toward the same goal.

Newborn Screening for SCID in the Laboratory

Presenter: Francis Lee, PhD, MSc, Centers for Disease Control and Prevention

Dr. Francis Lee, PhD, MSc, Research Microbiologist at the Centers for Disease Control and Prevention, discussed the different methods of testing, analysis and reporting being used in different state public health NBS labs.

SCID Case Definitions

Presenters: Marci Sontag, PhD, NewSTEPs, Colorado School of Public Health & Careema Yusuf, MPH, NewSTEPs, Association of Public Health Laboratories

Dr. Marci Sontag, PhD, Associate Director of NewSTEPS, stressed the value of entering SCID data in the NewSTEPs National Data Repository for newborn screening. These case definitions are intended to count infants consistently across time, hospitals and states.

Diagnosis of SCID and Disorders with Insufficient T-Cells

Presenter: Jennifer Puck, MD, University of California, San Francisco, School of Medicine

Dr. Jennifer Puck, MD, Professor of Pediatrics at the University of California, San Francisco, highlighted all of the steps that need to happen once a baby screens positive for SCID through diagnosis and up to treatment. Dr. Puck emphasized the need to harmonize diagnosis terminology and discussed current challenges.

SCID Newborn Screening: Building the Clinical Network

Presenter: Lisa Kobrynski, MD, MPH, Emory University School of Medicine

Dr. Lisa Kobrynski, MD, an allergist-immunologist in Atlanta, Georgia, highlighted the importance of establishing and maintaining clinical referral networks. Dr. Kobrynski stressed that strong clinical networks require multiple partners and that communication is key.

Multiplexing Real-Time PCR Assay to Simultaneously Screen for SCID and SMA

Presenter: Mei Baker, MD, FACMG, Wisconsin State Laboratory of Hygiene

Dr. Mei Baker, MD, FACMG, Wynne Mateffy Professor in the Department of Pediatrics and Co-Director of the Newborn Screening Laboratory at the University of Wisconsin School of Medicine and Public Health, shared information on Spinal Muscular Atrophy (SMA), highlighting the similarities in polymerase chain reaction (PCR) based methodology and the ability to harness existing SCID infrastructure for implementing SMA newborn screening.

Multiplexing TREC & KREC in NYS

Presenter: Carlos A. Saavedra-Matiz, MD, NYSDOH Wadsworth Center

Dr. Carlos A. Saavedra-Matiz, Supervisor of the Molecular Laboratory of the New York State Department of Health, highlighted information on a combined TREC/KREC approach that can increase the scope of what is detected by newborn screening.

Next Generation Sequencing in Newborn Screening for SCID in New York State

Presenter: Carlos A. Saavedra-Matiz, MD, NYSDOH Wadsworth Center

Dr. Carlos A. Saavedra-Matiz, Supervisor of the Molecular Laboratory of the New York State Department of Health, provided an overview of next generation sequencing in newborn screening for SCID in New York. Next generation sequencing can be used to detect mutations in genes that can cause SCID. New York is currently developing an assay and pilot test next generation sequencing in a population that screening positive for SCID in New York.

Case Presentations and Discussion

Presenters: Susan E. Prockop, MD, Memorial Sloan Kettering Cancer Center; Christine Seroogy, MD, University of Wisconsin, School of Medicine; James Verbsky, MD, PhD, Medical College of Wisconsin; and Suzanne Skoda-Smith, MD, Seattle Children's Hospital

Clinicians from various states talked through unique cases and challenges they have faced from initial diagnostic testing through post-transplant treatment.

Baby's First Test: An Educational Resource for Healthcare Providers

Presenter: Jackie Seisman, MPH, Baby's First Test

Jackie Seisman, Assistant Director of Baby's First Test, provided an overview of educational resources available for health care providers, including trainings, newborn screening promotional materials, resources for patients, and online forums. More information can be found online.

Immune Deficiency Foundation: Materials for Parents Following Newborn Screening

Presenter: Lynn H. Albizo, Esq. and Jamie Sexton, Immune Deficiency Foundation

Lynn H. Albizo, Esq., Director of Public Policy for the Immune Deficiency Foundation (IDF), and Jamie Sexton, Government Relations Specialist for the IDF, provided an overview of the IDF's SCID initiative to expand public awareness, serve as advocates, fund research efforts, and provide educational events for individuals and families affected by SCID. The following resources have been developed by the IDF to support parents and are available online: A Guide for Parents Following a Diagnosis (flyer); Abnormal Screening (flyer), Rotavirus Vaccine (brochure); and Understanding the Low T Cell Results from Your Baby's Newborn Screening (brochure).

SCID: Long Term Follow-Up: USIDNET and PI Connect

Presenter: Charlotte Cunningham-Rundles, MD, PhD, Mount Sinai School of Medicine

Dr. Charlotte Cunningham-Rundles, David S. Gottesman Professor of Immunology at the Mount Sinai School of Medicine in New York, described the value of linking public health to clinical care. Dr. Cunningham-Rundles provided an overview of USIDNet, a registry that collects clinical and laboratory data on patients with rare diseases and aims to promote recognition of defects, identify optimal therapies, improve research efforts, and investigate what leads to better quality of life. Additionally, PI Connect, a registry which aims to integrate patients, clinicians, and researches, with an interest in research in primary immune deficiencies, was highlighted.

SCID Long Term Follow-Up in California

Presenter: Tracey Bishop, BS, Genetic Disease Screening Program, California Department of Health

Tracey Bishop, Chief of the Newborn Screening Branch with the California Department of Health, provided an overview of California Long-Term Follow-Up process; including data elements collected, how California is working with specialists to collect this information, and current barriers to this process.

Networking Lunch; Identifying Next Steps with State/Regional Teams

Newborn screening programs were asked to identify priorities for the SCID community moving forward. The following priorities were identified:

- Better guidelines for treatment of asymptomatic patients
- Insurance/policy coverage (ex. treatments, insurance coverage for costs associated with NBS)
- Establishment of networks of communications between newborn screeners and pediatric immunology consultants
- Formal recommendations on Long-Term Follow-Up
- Sharing of secondary algorithms
- Education for healthcare providers of newborn screening process through treatment
- Information shared with parents (resources available through IDF)
- Invite legislative representatives from states to conversations/meetings
- Integration with Infectious Disease Specialists
- Peer review/evidenced based and/or published articles/statements concerning transfusion

MEETING PARTICIPANTS

First Name	Affiliation	Specialization	Email Address
Prescott Atkinson	Alabama	Clinical	PAtkinson@peds.uab.edu
Rachael Montgomery	Alabama	Follow-Up	Rachael.Montgomery@adph.state.al.us
Sabra Anckner	Alaska	Follow-Up	sabra.anckner@alaska.gov
Jeffrey Demain	Alaska	Clinical	jdemain@allergyalaska.com
Constantino Aznar	California	Laboratory	Constantino.Aznar@cdph.ca.gov
Tracey Bishop	California	Follow-Up	Tracey.Bishop@cdph.ca.gov
Manish Butte	California	Clinical	MButte@mednet.ucla.edu
Jennifer Puck	California	Clinical	Jennifer.Puck@ucsf.edu
Erwin Gelfand	Colorado/Wyoming	Clinical	GelfandE@NJHealth.org
Darren Michael	Colorado	Laboratory	darren.michael@state.co.us
Corina Bello	Connecticut	Laboratory	Corina.Bello@ct.gov
Debra Studwell	Connecticut	Laboratory	Debra.Studwell@ct.gov
Megan Ford	Delaware	Clinical	Megan.Ford@nemours.org
Dusty Edwards	Florida	Follow-Up	Dusty.Edwards@flhealth.gov
Gary Kleiner	Florida	Clinical	gary.kleiner@med.miami.edu
Emily Paynter	Georgia	Laboratory	Emily.Paynter@dph.ga.gov
Lisa Kobrynski	Georgia	Clinical	lkobryn@emory.edu
Gwen Palmer	Hawaii	Follow-Up	Gwen.Palmer@doh.hawaii.gov
Marian Melish	Hawaii	Clinical	MarianM@kapiolani.org
Jennifer Crew	Illinois	Laboratory	Jennifer.Crew@Illinois.gov
Megan Griffie	Indiana	Laboratory	MGriffie@isdh.IN.gov
Barb Lesko	Indiana	Laboratory	bglesko@iupui.edu
Alison Hendrick	Indiana	Follow-Up	AHendrick@isdh.in.gov
Mary Beth Fasano	Iowa	Clinical	Mary-fasano@uiowa.edu
Carol Johnson	Iowa	Follow-Up	carol-johnson@uiowa.edu
Valerie Van Zee	Iowa	Laboratory	Valerie-vanzee@uiowa.edu
Kelley Hale	Kansas	Laboratory	Kelley.hale@ks.gov
Shawn Manos	Kansas	Follow-Up	shawn.m.manos@ks.gov
Angela Brown	Kentucky	Follow-Up	angelak.brown2@ky.gov
Darrin Sevier	Kentucky	Laboratory	Darrin.Sevier@ky.gov
Jantz Malbrue	Louisiana	Follow-Up	Jantz.Malbrue@LA.GOV
Ricardo Sorensen	Louisiana	Clinical	RSoren@lsuhsc.edu
Adam Coleman	Maryland	Laboratory	adam.coleman@maryland.gov
Fizza Gulamali-Majid	Maryland	Laboratory	Fizza.Majid@maryland.gov
Howard Lederman	Maryland	Clinical	hlederm1@jhmi.edu
Elizabeth Younger	Maryland	Clinical	myounge2@jhmi.edu
Elizabeth Secord	Michigan	Clinical	esecord@med.wayne.edu
Lois Turbett	Michigan	Follow-Up	turbettl@michigan.gov
Diana Vilkama	Minnesota	Clinical	diana.vilkama@childrensmn.org

Carrie Wolf	Minnesota	Laboratory	carrie.wolf@state.mn.us
Amber Bryant	Missouri	Follow-Up	Amber.Bryant@health.mo.gov
Alan Knutsen	Missouri/Illinois	Clinical	knutsenm@slu.edu
Maleewan Kitcharoensakkul	Missouri	Clinical	mkitcharoensakkul@wustl.edu
Nikita Raje	Missouri/Kansas	Clinical	nraje@cmh.edu
Angela Dusko	Montana	Laboratory	ADusko@mt.gov
Jay Patel	Washington/Montana	Clinical	jpatel1@uw.edu
Hana Niebur	Nebraska	Clinical	hniebur@childrensomaha.org
Bonifacio Dy	Nevada	Laboratory	bdy@med.unr.edu
Nevin Wilson	Nevada	Clinical	nwilson@med.unr.edu
Linda Kincaid	New Hampshire	Follow-Up	Linda.Kincaid@dhhs.nh.gov
Alyssa MacMillan	New Jersey	Laboratory	alyssa.macmillan@doh.nj.gov
Charlotte Cunningham-Rundles	New York	Clinical	charlotte.cunningham-rundles@mssm.edu
Susan Prockop	New York	Clinical	prockops@MSKCC.ORG
Carlos Saavedra-Matiz	New York	Laboratory	carlos.saavedra@health.ny.gov
Beth Vogel	New York	Follow-Up	beth.vogel@health.ny.gov
Candace Leslie	North Carolina	Laboratory	candace.leslie@dhhs.nc.gov
Jennifer Taylor	North Carolina	Laboratory	jltaylor@rti.org
Eve Wu	North Carolina	Clinical	eywu@email.unc.edu
Chris Cleveland	North Dakota	Clinical	Chris.Cleveland@SanfordHealth.org
Joyal Meyer	North Dakota	Follow-Up	jbmeyer@nd.gov
James Love	Oklahoma	Clinical	jlovemd@aol.com
Tonya McCallister	Oklahoma	Laboratory	TonyaJ@health.ok.gov
Sara Denniston	Oregon	Follow-Up	Sara.denniston@state.or.us
Sonia Ramírez	Puerto Rico	Laboratory	sonia.ramirez@upr.edu
Rafael Zaragoza	Puerto Rico	Clinical	zaragoza.urdaz@gmail.com
Ona Adair	South Carolina	Laboratory	adairoo@dhec.sc.gov
Kelli Williams	South Carolina	Clinical	williamske@musc.edu
Christine Dorley	Tennessee	Laboratory	M.Christine.Dorley@tn.gov
Amy Neeley	Tennessee	Follow-Up	Amy.J.Neeley@tn.gov
Rachel Lee	Texas	Laboratory	Rachel.Lee@dshs.texas.gov
Howard Rosenblatt	Texas	Clinical	HMRosenblatt@seton.org
Julianna Ybarbo	Texas	Follow-Up	Julianna.YBarbo@dshs.texas.gov
David Jones	Utah	Laboratory	dejones@utah.gov
Cindy Ingham	Vermont	Follow-Up	Cindy.Ingham@vermont.gov
Kristen Barbieri	Virginia/Maryland/ Washington, DC	Clinical	KBarbier@childrensnational.org
Blachy Davila	Virginia/Maryland/ Washington, DC	Clinical	BJDAVILAS1@childrensnational.org

Mona Elmacken	Virginia/Maryland/ Washington, DC	Clinical	MMELMACKEN@childrensnational.org
Richard Haughton	Virginia	Laboratory	richard.haughton@dgs.virginia.gov
Mike Kellar	Virginia/Maryland/ Washington, DC	Clinical	MKeller@childrensnational.org
Christine Nguyen	Washington	Follow-Up	christine.nguyen@doh.wa.gov
Suzanne Skoda-Smith	Washington	Clinical	suzanne.skoda-smith@seattlechildrens.org
Mei Baker	Wisconsin	Laboratory	mei.baker@slh.wisc.edu
Christine Seroogy	Wisconsin	Clinical	cmseroogy@pediatrics.wisc.edu
James Verbsky	Wisconsin	Clinical	jverbsky@mcw.edu
Amy Brower	American College of Medical Genetics and Genomics, Newborn Screening Translational Research Network		abrower@acmg.net
Funke Akinsola	Association of Public Health Laboratories, NewSTEPs		Oluwafunke.Akinsola@aphl.org
Scott Becker	Association of Public Health Laboratories		Scott.Becker@aphl.org
Cierra Bryant	Association of Public Health Laboratories		Cierra.Bryant@aphl.org
Erin Darby	Association of Public Health Laboratories, NewSTEPs		Erin.Darby@aphl.org
Sari Edelman	Association of Public Health Laboratories, NewSTEPs		Sari.Edelman@aphl.org
Jelili Ojodu	Association of Public Health Laboratories, NewSTEPs		Jelili.Ojodu@aphl.org
Ruthanne Sheller	Association of Public Health Laboratories, NewSTEPs		Ruthanne.Sheller@aphl.org
Sikha Singh	Association of Public Health Laboratories, NewSTEPs		Sikha.Singh@aphl.org

Careema Yusuf	Association of Public Health Laboratories, NewSTEPs		Careema.Yusuf@aphl.org
Guisou Zarbalian	Association of Public Health Laboratories, NewSTEPs		Guisou.Zarbalian@aphl.org
Francis Lee	Centers for Disease Control and Prevention		icr0@cdc.gov
Golriz Yazdanpanah	Centers for Disease Control and Prevention		czu1@cdc.gov
Marci Sontag	Colorado School of Public Health, NewSTEPs		marci.sontag@ucdenver.edu
Amelia Mulford	Genetic Alliance, Baby's First Test		amulford@geneticalliance.org
Jaclyn Seisman	Genetic Alliance, Baby's First Test		jseisman@geneticalliance.org
Alaina Harris	Health Resources and Services Administration		aharris@hrsa.gov
Catharine Riley	Health Resources and Services Administration		CRiley@hrsa.gov
Debi Sarkar	Health Resources and Services Administration		dsarkar@hrsa.gov
Jill Shuger	Health Resources and Services Administration		JShuger@hrsa.gov
Lynn Albizo	Immune Deficiency Foundation		lalbizo@primaryimmune.org
Jamie Sexton	Immune Deficiency Foundation		jsexton@primaryimmune.org