Importance of Coalition Building to Support Newborn Screening

EveryLife Foundation
Newborn Screening Bootcamp

April 6, 2019

Allison May Rosen, 3D Communications for the Firefly Fund

Beryl Elizabeth Andrews, 2010

Abigail Mae Andrews, 2014

With Mom and Dad, 2018

Clinical, Science-Based Criteria Used to Evaluate New Conditions for Newborn Screening Lists

- Demonstrate condition poses a public health problem, without easily identified symptoms at birth
  - Significant risk of illness, disability, death if babies not treated promptly
- Validated screening test available with acceptable sensitivity, specificity
  - Public health laboratories' ability to perform test
- Effective treatment available, more beneficial to treat sooner than later
  - Resources and access to treatment, counseling widely available
- Benefits outweigh risks and burdens of screening and treatment

Public and Patient Voices Matter Too… At Federal Level

The Advisory Committee on Heritable Disorders in Newborns and Children

HRSA Headquarters - 5600 Fishers Lane, Rockville, MD 20852
February 8, 2018

TIME	TOPIC	PRESENTER
8:30 – 8:50 AM	• Welcome • Roll Call • Opening Remarks • November 2017 Minutes	**Joseph Bocchini, MD** Committee Chair Professor and Chairman, Department of Pediatrics, Louisiana State University **Catharine Riley, PhD, MPH** Designated Federal Official Health Resources and Services Administration
8:50 – 9:10 AM	An Overview of Cutoff Determinations and Risk Assessment Methods Used in Dried Blood Spot Newborn Screening	**Joe Orsini, PhD** Wadsworth Center, New York State Department of Health Co-Chair, APHL Newborn Screening Quality Assurance Quality Control Subcommittee
9:10 – 9:25 AM	Laboratory Standards and Procedures Workgroup: Review of the Overview of Cutoff Determinations Document	**Kellie Kelm, PhD** Ex-Officio Committee Member Chair, Laboratory Standards and Procedures Workgroup
9:25 – 9:50 AM	Committee Discussion and Vote	**Joseph Bocchini, MD** Committee Chair
9:50–10:30 AM	**Public Comment**	

TIME	TOPIC	PRESENTER
10:45 – 11:45 AM	Newborn Screening for Spinal Muscular Atrophy (SMA): A Systematic Review of Evidence (Part 1)	**Alex R. Kemper, MD, MPH, MS** Lead, Evidence-Based Review Group **Jelili Ojodu, MPH** Member, Evidence-Based Review Group **Lisa A. Prosser, Ph.D.** Member, Evidence-Based Review Group
11:45 – 12:30 PM	Lunch	
12:30 – 1:30 PM	Newborn Screening for Spinal Muscular Atrophy (SMA): A Systematic Review of Evidence (Part 2)	
1:30 – 2:00 PM	Committee Report: Newborn Screening for Spinal Muscular Atrophy (SMA)	**Dietrich Matern, MD, PhD** Committee Member **Beth Tarini, MD, MS, FAAP** Committee Member
2:00 – 2:30 PM	Committee Discussion and Vote on SMA	**Joseph Bocchini, MD** Committee Chair
2:30 – 2:40 PM	Follow-Up and Treatment Workgroup Report: The Role of Quality Measures to Promote Long-Term Follow-Up of Children Identified by Newborn Screening Programs	**Jeffrey P. Brosco, MD, PhD** Committee Member Chair, Follow-Up & Treatment Workgroup
2:40 – 2:55 PM	Committee Discussion	**Joseph Bocchini, MD** Committee Chair
2:55 – 3:00 PM	New Business	**Joseph Bocchini, MD** Committee Chair
3:00 PM	Adjourn	

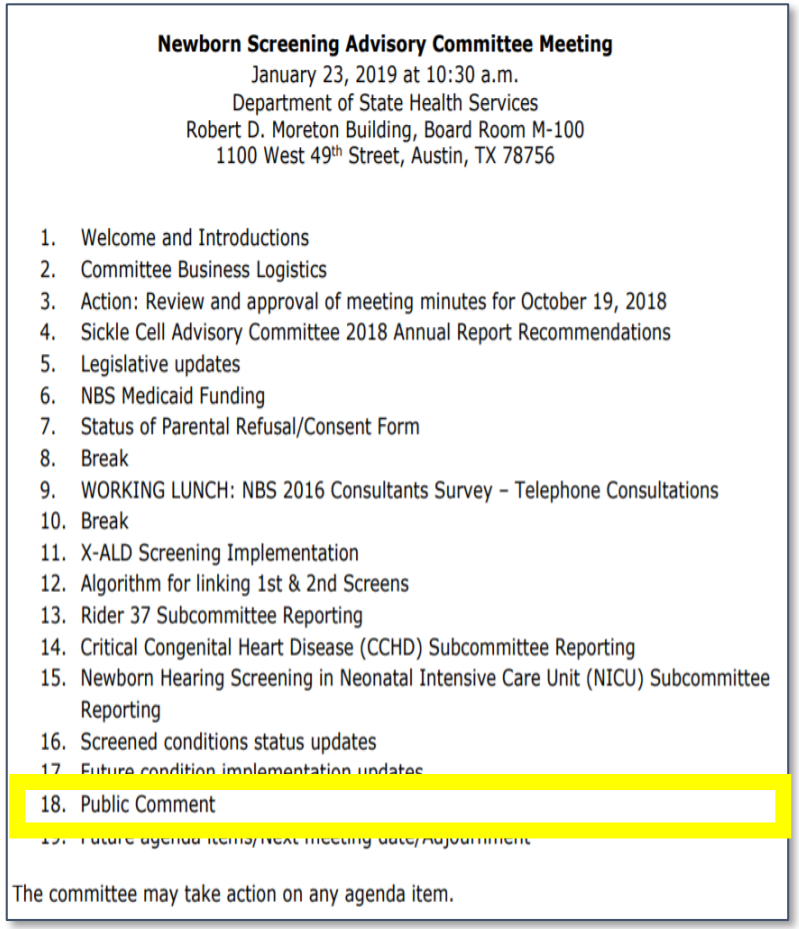
Public and Patient Voices Matter Too… In the States

DEPARTMENT OF HEALTH

AGENDA, Newborn Hearing Screening Advisory Committee

NOVEMBER 7, 2018 1:00-4:00 PM

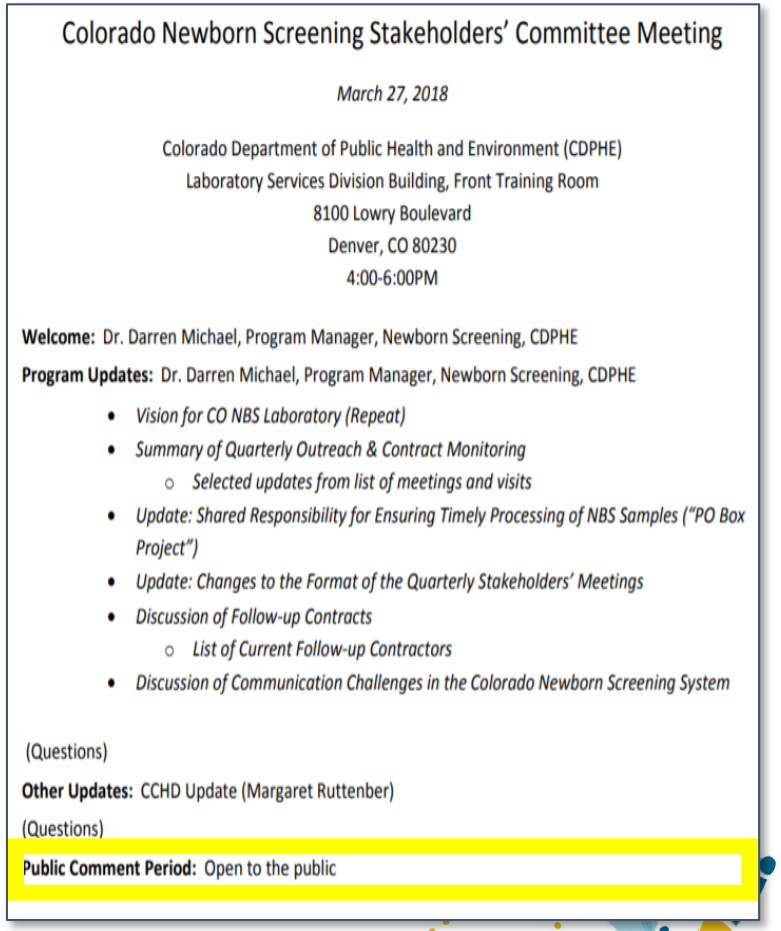
Time frame of meeting	Meeting Agenda
1:00 – 1:20 pm	**Welcome and Announcements** • Newborn Screening Advisory Committee Liaison Update • Open Appointments • Nominations/vote
1:20 -1:30 pm	**Family Story (CMV video)**
1:30– 2:00 pm	**CMV** National Conference Highlights & MN Study Update Maggie Dreon
2:00 – 2:30 pm	**Audiology Guidelines** Assessment and Referrals Discussion and Vote Darcia Dierking and Melanie Wege
2:30 – 2:45 pm	**EHDI Educational Update** Kathy Anderson



Newborn Screening Advisory Committee Meeting
January 23, 2019 at 10:30 a.m.
Department of State Health Services
Robert D. Moreton Building, Board Room M-100
1100 West 49th Street, Austin, TX 78756

1. Welcome and Introductions
2. Committee Business Logistics
3. Action: Review and approval of meeting minutes for October 19, 2018
4. Sickle Cell Advisory Committee 2018 Annual Report Recommendations
5. Legislative updates
6. NBS Medicaid Funding
7. Status of Parental Refusal/Consent Form
8. Break
9. WORKING LUNCH: NBS 2016 Consultants Survey – Telephone Consultations
10. Break
11. X-ALD Screening Implementation
12. Algorithm for linking 1st & 2nd Screens
13. Rider 37 Subcommittee Reporting
14. Critical Congenital Heart Disease (CCHD) Subcommittee Reporting
15. Newborn Hearing Screening in Neonatal Intensive Care Unit (NICU) Subcommittee Reporting
16. Screened conditions status updates
17. Future condition implementation updates
18. Public Comment
19. Future agenda items/Next meeting date/Adjournment

The committee may take action on any agenda item.



Colorado Newborn Screening Stakeholders' Committee Meeting

March 27, 2018

Colorado Department of Public Health and Environment (CDPHE)
Laboratory Services Division Building, Front Training Room
8100 Lowry Boulevard
Denver, CO 80230
4:00-6:00PM

Welcome: Dr. Darren Michael, Program Manager, Newborn Screening, CDPHE

Program Updates: Dr. Darren Michael, Program Manager, Newborn Screening, CDPHE

- *Vision for CO NBS Laboratory (Repeat)*
- *Summary of Quarterly Outreach & Contract Monitoring*
  - *Selected updates from list of meetings and visits*
- *Update: Shared Responsibility for Ensuring Timely Processing of NBS Samples ("PO Box Project")*
- *Update: Changes to the Format of the Quarterly Stakeholders' Meetings*
- *Discussion of Follow-up Contracts*
  - *List of Current Follow-up Contractors*
- *Discussion of Communication Challenges in the Colorado Newborn Screening System*

(Questions)

Other Updates: CCHD Update (Margaret Ruttenber)

(Questions)

Public Comment Period: Open to the public

Firefly's Newborn Screening Initiative: A Multi-Stakeholder Coalition Filling an Unmet Need for an Ultra-Rare Disease

- Activities:
  - Establish Working Group
  - Research NBS environment
  - Design pilot NBS studies
  - Develop scientific protocols
  - Host Clinical Roundtable
  - Build stakeholder support
  - Raise community awareness

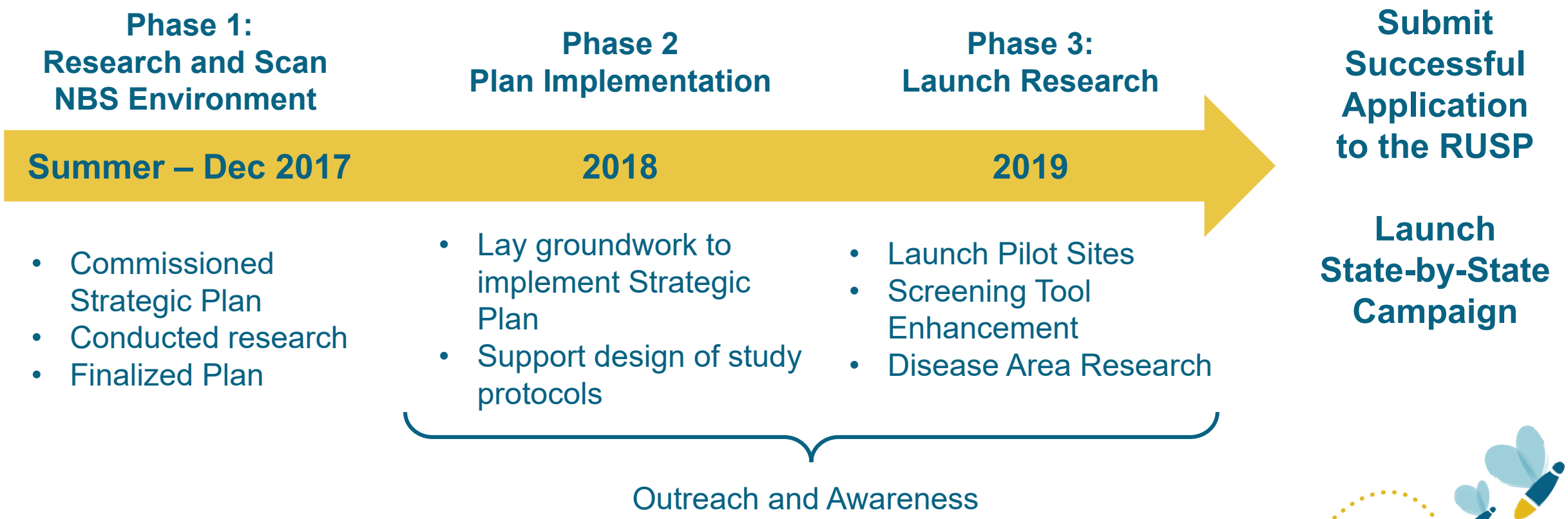
Needed Stakeholder Group with Deep, Diverse Expertise to Satisfy Criteria, Achieve Common Vision

PAM ANDREWS, Firefly Fund
ELIZABETH BERRY-KRAVIS, Rush University Medical Center
JOSLYN CROWE, National Niemann-Pick Foundation
XUNTIAN JIANG, Washington University in St. Louis
SEAN KASSEN, Parseghian Medical Research Fund
BEN MACHIELSE, Drug development
PHIL MARELLA, DART
DEREK NATEN, Mallinckrodt
DAN ORY, Washington University in St. Louis
CINDY PARSEGHIAN, Parseghian Medical Research Fund
DENNY PORTER, National Institutes of Health
REGAN SHERMAN, Orphazyme
MELISSA WASSERTEIN, Children's Hospital at Montefiore
ALLISON MAY ROSEN, *Coordinator, 3D Communications*

- Researchers, clinicians from academia and government
- Practicing physicians
- NPC family foundations
- Industry partners
- NPC advocates
- Laboratorians

Common Vision:
All newborns will be screened for NPC, enabling a diagnosis at birth

Pathway to Achieve Goal – A Marathon Not a Sprint



FIREFLY.FUND

Research Newborn Screening Environment: Interviewed Key Opinion Leaders and Experts, Expanded Network

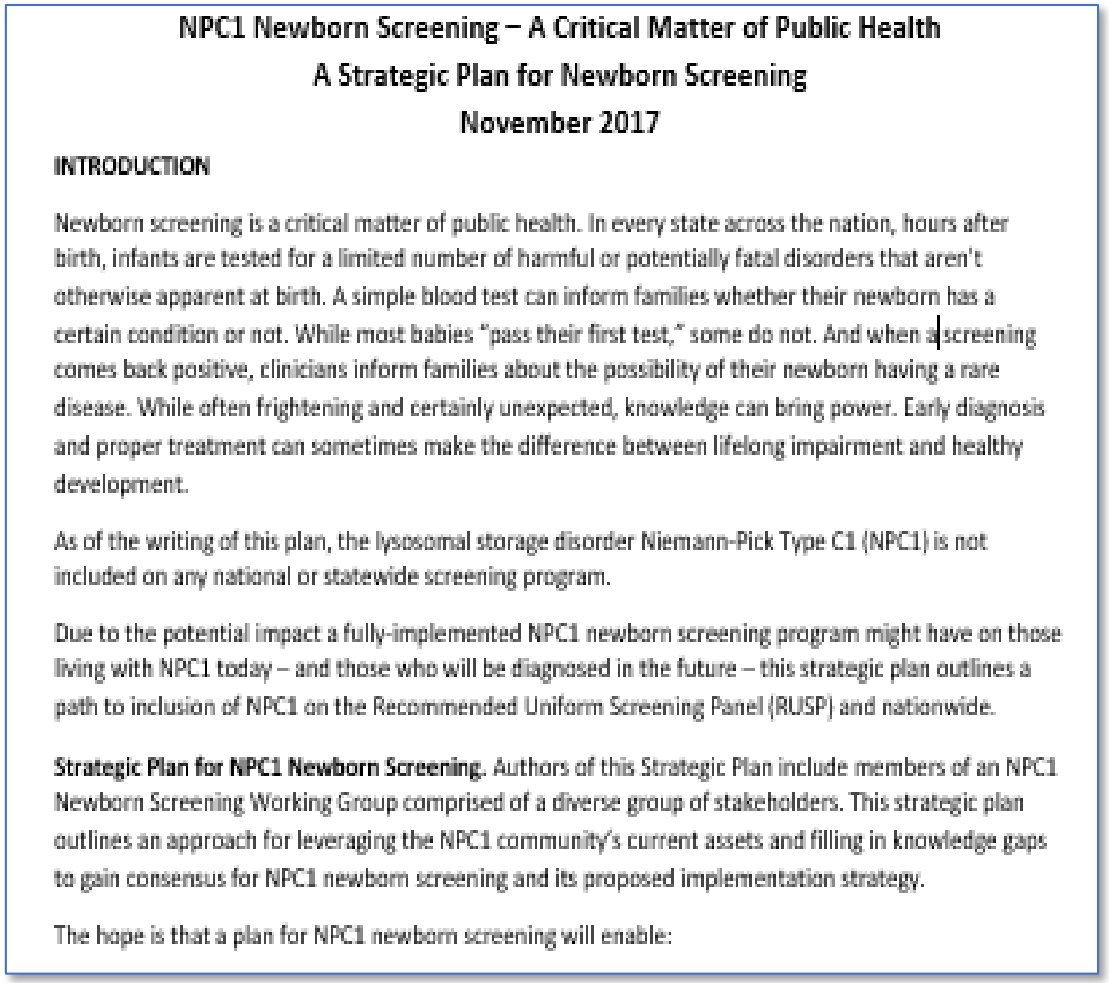
EXISTING MODELS

- Learn from experience of leaders behind recent RUSP additions
  - Pompe Disease, SMA, MPS1
- Investigated
  - How many live births needed
  - Necessary stakeholders

SCREENING TOOL VALIDATION

- Identify gaps, opportunities with screening tool
- Investigated
  - Assay sensitivity and specificity
  - Potential partners

Strategic Plan: Target RUSP, Gather Evidence, Enhance Screening Tools, Advance Knowledge



NPC1 Newborn Screening – A Critical Matter of Public Health

A Strategic Plan for Newborn Screening

November 2017

INTRODUCTION

Newborn screening is a critical matter of public health. In every state across the nation, hours after birth, infants are tested for a limited number of harmful or potentially fatal disorders that aren't otherwise apparent at birth. A simple blood test can inform families whether their newborn has a certain condition or not. While most babies "pass their first test," some do not. And when a screening comes back positive, clinicians inform families about the possibility of their newborn having a rare disease. While often frightening and certainly unexpected, knowledge can bring power. Early diagnosis and proper treatment can sometimes make the difference between lifelong impairment and healthy development.

As of the writing of this plan, the lysosomal storage disorder Niemann-Pick Type C1 (NPC1) is not included on any national or statewide screening program.

Due to the potential impact a fully-implemented NPC1 newborn screening program might have on those living with NPC1 today – and those who will be diagnosed in the future – this strategic plan outlines a path to inclusion of NPC1 on the Recommended Uniform Screening Panel (RUSP) and nationwide.

Strategic Plan for NPC1 Newborn Screening. Authors of this Strategic Plan include members of an NPC1 Newborn Screening Working Group comprised of a diverse group of stakeholders. This strategic plan outlines an approach for leveraging the NPC1 community's current assets and filling in knowledge gaps to gain consensus for NPC1 newborn screening and its proposed implementation strategy.

The hope is that a plan for NPC1 newborn screening will enable:

1. Prioritize submission to RUSP
2. Launch pilot sites and screen 150,000-250,000 newborns
   - → Identify at least one patient
3. Use validated NPC assay for pilot
4. Conduct additional disease-area research
   - → eg, Clinical consensus on pre-symptomatic intervention

Supporting Launch of Newborn Screening Pilot Study

- New York Screen+: Lysosomal Storage Diseases Pilot NBS Program
  - Select New York hospitals with on-site recruitment
- Estimated to screen 30,000 neonates per year
  - Helps with accumulating necessary data for a RUSP submission
  - At least one positive, confirmed test for an NPC-affected newborn
  - Ability to demonstrate clinical care follow up

Develop Scientific Protocols for Pilot Programs

BILE ACID NEWBORN SCREENING PROCEDURE MANUAL

- Step-by-step laboratory procedure manual
- Measure NPC-associated bile acid biomarker in DBS
- Validated and published prototype
- Discriminate NPC carriers from NPC disease

DIAGNOSTIC PROTOCOL FLOWSHEET

- Positive Newborn Screen
- Confirmatory testing:
  - Biochemical
  - Molecular

DEVELOPMENT FOLLOW UP PROTOCOL

- Infants with significant liver dysfunction referred to NIH
- Seek to treat before neurological symptom onset
- Local or NIH/Rush Medical Center follow up
- Neurological exam, Mullen Scales of Early Learning, Vineland

NPC Clinical Roundtable – Consensus Building Regarding Timing for Treatment Initiation Following NBS

- Convened by Working Group PI, Elizabeth Berry Kravis, MD PhD
- 12 experts, treating physicians to attend, provide input and guidance
- Successful RUSP applications require additional disease area research to support the need for newborn screening
  - Why is earlier intervention a clinical benefit for patients?
  - How soon after birth must treatment be initiated to be effective?

Building Community Awareness, Long-Term Support

- Requires "all hands on deck" approach
  - Patient/family nonprofit organizations
  - Academic centers and providers
  - Government
  - Industry

- Shared responsibility funding model
  - Divided among stakeholders: Family foundations/associations/industry
- Stakeholder outreach and awareness *(sample)*
  - ACHDNC
  - American Academy of Pediatrics
  - Early Check
  - EveryLife Foundation
  - Notre Dame Rare Disease Day
  - Niemann-Pick UK
  - Parseghian Annual Scientific Conference
  - Mallinckrodt, Amicus, eScape Bio, etc

Importance of Coalition Building to Support Newborn Screening

EveryLife Foundation
Newborn Screening Bootcamp

April 6, 2019

Allison May Rosen, 3D Communications for the Firefly Fund