

# **NYMAC Newborn Screening Screen-Positive Follow-up Protocols**

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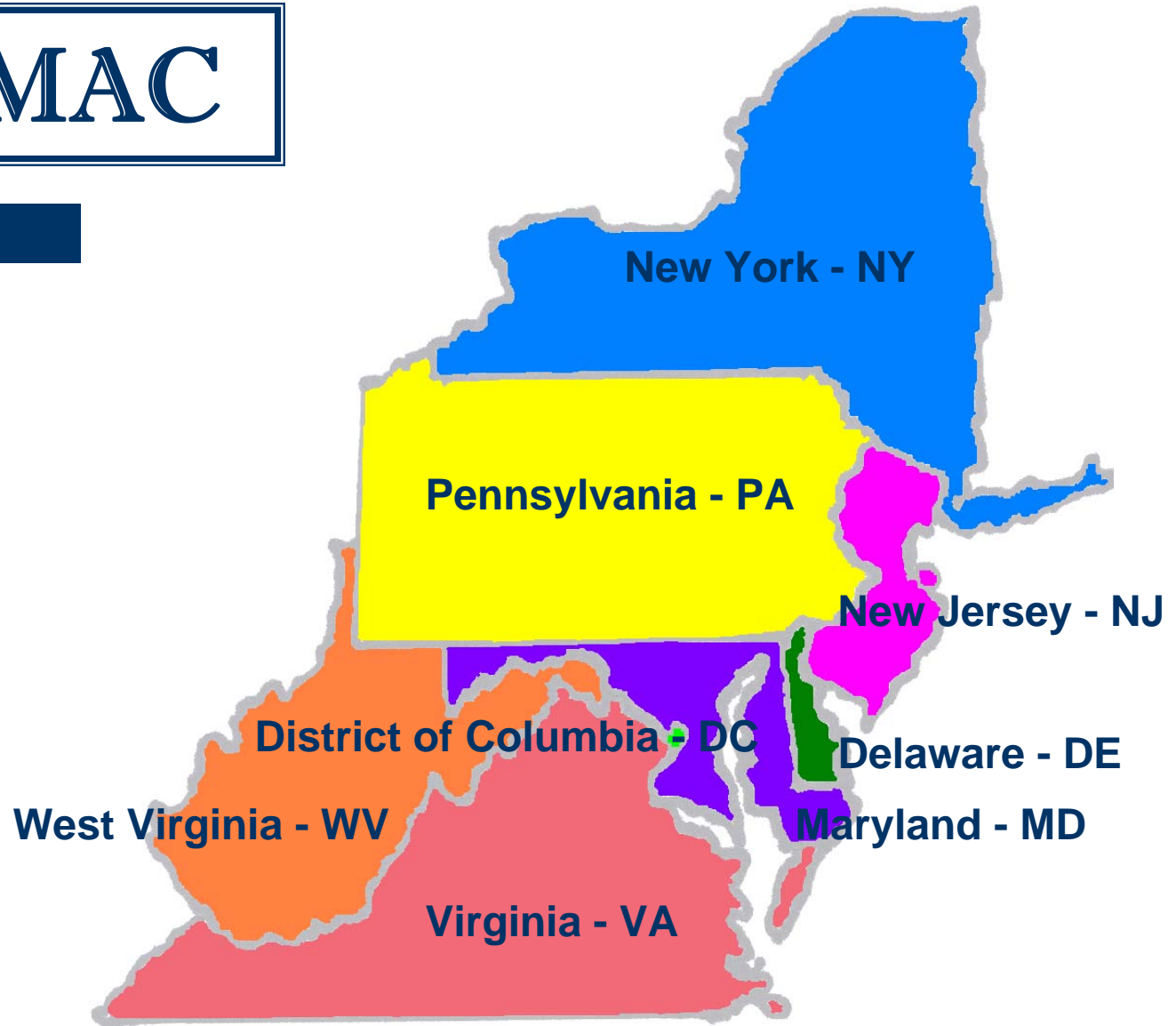
# Acknowledgements

- NYMAC (New York – Mid-Atlantic Consortium for Genetic and Newborn Screening Services) is one of 7 Genetics Collaborative Regions funded through grants from HRSA/MCHB.
  - U22 MC 03956
- The survey data was collected and summarized by Bonnie Fredrick, MS, NYMAC Project Coordinator.
- The information is from the state Newborn Screening Follow-up Coordinators.

# Limitations

- The NBS FU coordinators reviewed their own information for accuracy, but did not see that from other coordinators.
  - Perhaps additional information would have been added or clarified with such a step.
  - There are a lot of questions that can be generated from this information – to clarify, enhance and provide greater depth.
- Those steps are for a further survey.

# NYMAC



## Background

- The Newborn Screening and Standardization Work Group is one of six NYMAC work groups.
- Its Chair is Dr. David Kronn, a biochemical and clinical geneticist at Westchester Medical Center in New York.
- Its NYMAC Coordinator is Bonnie Fredrick.

# NBS Standardization Work Group

- Its vision is to ensure
  - that all newborns receive screening for all recommended analytes and conditions;
  - that infants who screen positive receive appropriate clinical evaluation and diagnosis;
  - that infants who diagnose positive are entered into appropriate clinical management; and
  - that the state newborn screening programs can ensure that the children receive appropriate care over the short-term to diagnosis and over the long-term through adulthood.

# Objectives of the NBS Short-term Follow-up Survey

- To demonstrate some of the similarities and differences among the eight newborn screening programs within the NYMAC Region
- To identify the different technologies used to communicate NBS results with all the parties responsible for an infant's health
- To encourage standardization among the NBS programs, leading, perhaps, to a better understanding about NBS among providers

# Survey Process

- Specific questions were asked.
  - Some responders had more information to share than others.
- All the people surveyed received a summary of their own responses and confirmed that the information was correct.
- The entire summary has been shared with all the NBS Standardization Work Group members, though not specifically among those surveyed.

**Updates and additional information are anticipated and welcome!**

# Criteria for Second Screens and Repeat Specimens\*

- DE – a **second screen** is required on all infants
- DC, NJ, NY, PA<sup>†</sup>, VA – a **repeat specimen** is required only if the first specimen was collected on an infant < 24 hours old
- MD – a **second screen** is required on all infants at 1-2 weeks of age
- PA<sup>†</sup> – a **second screen** is recommended on all infants between 48-72 hours of age
- WV – a **repeat specimen** is required if the first one is collected on an infant < 48 hours old

\*From National Newborn Screening and Genetics Resource Center

# Who is the Pediatrician of Record?

Universally within NYMAC  
the pediatrician is identified on  
the blood collection form

**Unless it's wrong**

## Then – How to Identify the Correct Pediatrician

**Ask the Hospital  
of Birth**

**Ask the Pediatrician listed if  
he knows**

**Match first and  
second screen  
information**

**Send Parents a letter with  
NBS results and ask them to  
call NBS with name**

**If parents can't be found, check with  
neighbors – where did they go?**

**Call Parents**

## Newborn Screening Results

**Screen-Negative**

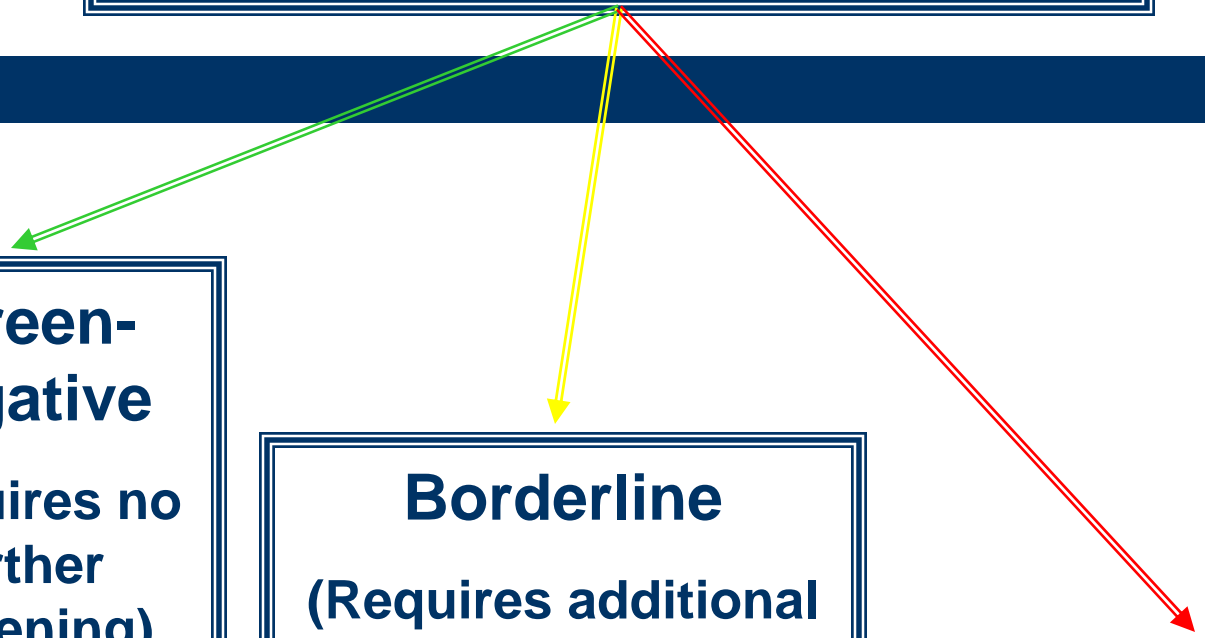
(Requires no further screening)

**Borderline**

(Requires additional blood-spot screening)

**Screen-Positive**

(Requires clinical evaluation)



# Follow-up – Screen-Negative Results

## Hospital of Birth

- NBS faxes results – MD
- NBS e-mails results – MD
- NBS mails results – DE, MD, NJ, NY, PA, VA, WV
- No results sent – DC

# Follow-up – Borderline Results

All states require at least a repeat blood spot for borderline results

## Hospital of birth

- NBS phones results – PA
- NBS faxes results – PA, WV
- NBS mails results – DE, DC, MD, NJ, NY, VA

# Follow-up – Screen-Positive Results

## Hospital of Birth

- NBS mails results – DE, MD, NJ, NY, VA
- NBS calls results – NY, PA
- NBS faxes results – NY, PA,
- NBS calls results if infant is still hospitalized – VA, WV
- NBS faxes results if infant is still hospitalized – VA
- No results sent – DC

## Follow-up – Screen-Negative Results

### Pediatrician

- Results on-line – DC, MD(?), NY(beta-testing)
- Results on voice response system – NJ, NY
- NBS e-mails results – MD
- NBS faxes results – DE, MD, NY
- NBS mails results– DE, MD, PA, VA, WV
- Pediatrician can call NBS – DC, NJ, NY
- Hospital of birth notifies pediatrician – NJ, NY
- No results sent – DC

# Follow-up – Borderline Results

## Pediatrician

- Results on-line – DC, MD(?), NY(beta-testing)
- Results on voice response system – NJ, NY
- NBS phones results – DC (with permission of parents), MD, PA
- NBS faxes results – DE (with fact sheets), DC (with permission of parents), MD, PA (with fact sheets), WV
- NBS e-mails results – MD (with fact sheets)
- NBS mails results – NJ, NY, VA

# Follow-up – Screen-Positive Results

## Pediatrician

- Results on-line – DC, MD(?), NY(beta-testing)
- Results on voice response system – NJ, NY
- NBS calls results – DE, DC, MD, NJ, NY, PA, VA, WV
- NBS faxes results – DE, DC, MD (with fact sheets), NJ, PA, VA, WV
- NBS e-mails results - MD (with fact sheets)
- NBS mails results – DE, MD (with fact sheets), NJ, NY, VA

# Follow-up – Screen-Negative Results

## Specialist/Specialty Center

- No policies to involve a Specialist in the care of infants with screen-negative results

# Follow-up – Borderline Results

## Specialist/Specialty Center

- Results on-line – DC, MD (2008), NY (beta-testing)
- Results on voice response system – NJ, NY
- NBS mails results – DE, NJ
- NBS calls results – DC (with permission of parents), MD, NJ (if repeat is also borderline)
- NBS faxes results – DC (with permission of parents)
- No results sent – NY, PA, VA
- NBS sends CF results only – WV

# Follow-up – Screen-Positive Results

## Specialist/Specialty Center

- Results on-line – DC, MD(1/2009), NY(beta-testing)
- Results on voice response system – NJ, NY
- NBS calls results – DE, DC (with permission of parents), MD, NJ, NY, PA, VA
- NBS faxes results – DE , DC (with permission of parents), MD, NJ, NY, PA, VA
- NBS e-mails results – MD, WV
- NBS mails results – NY

## Follow-up – Screen-Negative Results

### Parents

- Parents can call NBS for results – DE, DC, MD
- Results provided by pediatrician – DE, MD, NJ, NY, PA, VA, WV
- Parents can call NBS with questions – NJ

# Follow-up – Borderline Results

## Parents

- NBS mails alert about possible borderline result – DE, NJ, PA
- NBS calls for permission to call pediatrician; no results given at that time – DC
- NBS sends letter – DC (Sickle cell disease, G6PD)
- Results from pediatrician – MD, NJ, NY, PA, VA, WV
- Results from hospital of birth – NY, PA

# Follow-up – Screen-Positive Results

## Parents

- NBS mails results – DE
- NBS calls for permission to call pediatrician; no results given at that time – DC
- Pediatrician or specialist calls results – MD, NY, PA, VA, WV
- NBS mails results – MD (hemoglobinopathies), NJ

## Next Steps

- Share among the NYMAC NBS Programs
- Post on NYMAC web-site
- Periodic updates
- Further questions