

# SHORT-TERM FOLLOW-UP FOR POMPE

NEW DISORDERS MEETING  
JUNE 22, 2017

MISSOURI DEPARTMENT OF HEALTH AND  
SENIOR SERVICES



# KEY DECISION POINTS



- ❖ Establish Lysosomal Task Force
- ❖ Determine funding and staff
- ❖ Screening and implementation algorithm
- ❖ Education

# LYSOSOMAL TASK FORCE

- ❖ Geneticists
- ❖ Genetic Counselors
- ❖ Nurse Practitioners
- ❖ Nurses
- ❖ Laboratory staff
- ❖ Follow-up staff
- ❖ Affected Individual



# FUNDING AND STAFF

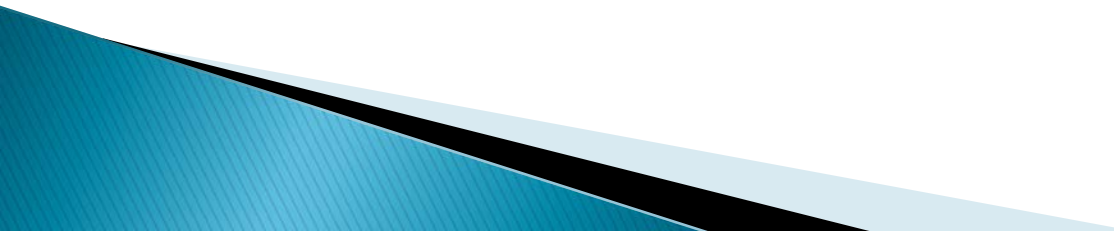


- ❖ Total Cost for short-term follow-up
  - Contracts with Genetic Tertiary Centers
  - Additional staff within the program
  - Printing costs
  - Data system changes

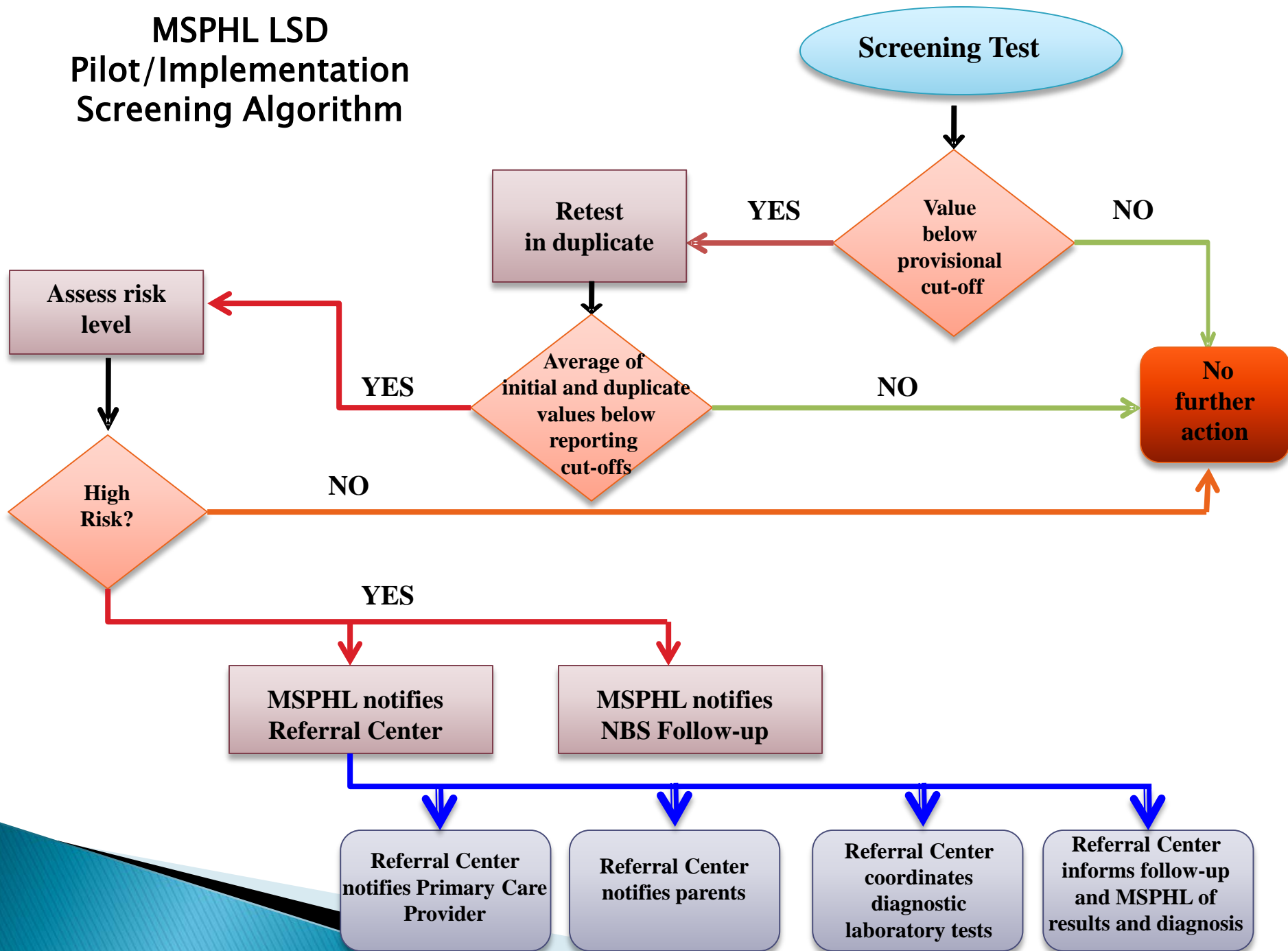
# SCREENING AND IMPLEMENTATION ALGORITHM

- ❖ Guidelines for Follow-up
  - Who receives abnormal screens
  - What confirmatory tests are needed
  - What information will be reported back to the state
- ❖ Tracking and reporting
  - Initially paper reports during the pilot phase
  - Case closure
  - Entry into data system

# GUIDELINES FOR FOLLOW-UP

- ❖ During the implementation phase, the primary care physician was not notified of a negative result.
  - ❖ Designated referral center depending on the location of the infant received a phone and fax screen-positive test result.
  - ❖ Centers contacted the PCP to coordinate care.
  - ❖ Newborn Screening Program Follow-up Manager also received a fax result.
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# MSPHL LSD Pilot/Implementation Screening Algorithm



# EDUCATION

- ❖ Providers

- Letter describing the new disorder to be tested

- ❖ Parents

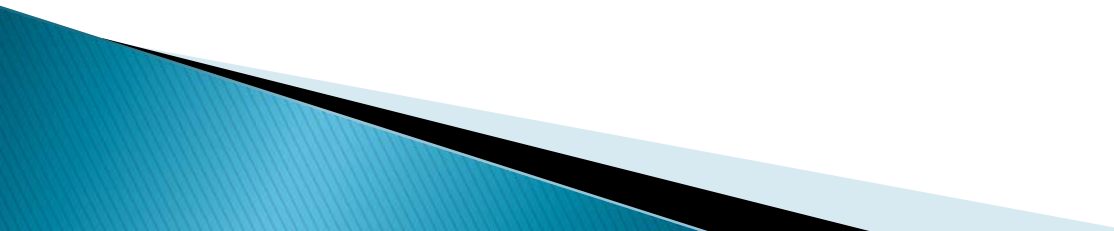
- Revised newborn screening booklet

- ❖ Website

- Links to Baby's First Test



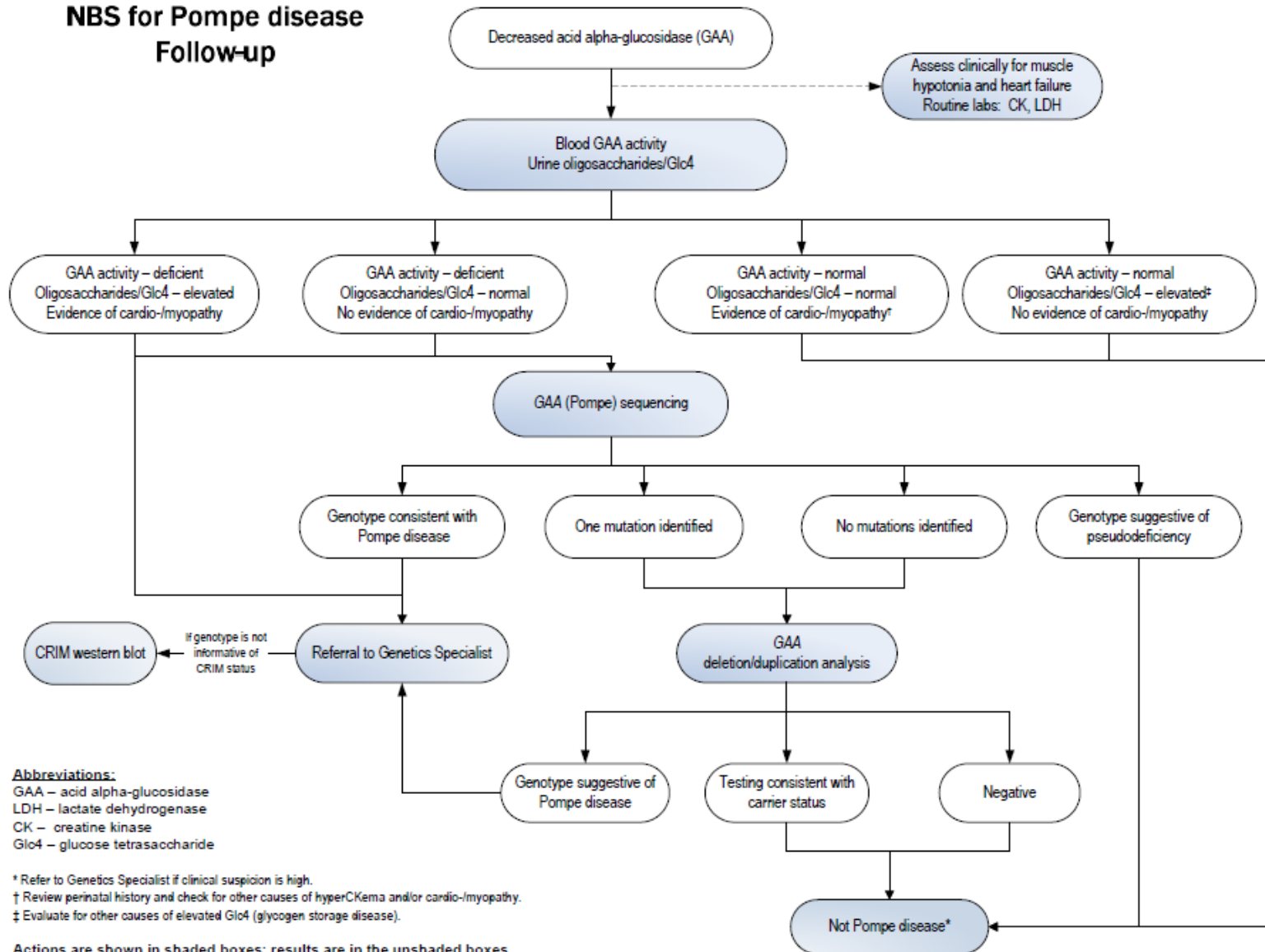
# CHANGES FOR SHORT-TERM FOLLOW-UP

- ❖ Amended contracts to add additional funding to Genetic Centers
  - ❖ Added diagnostic categories to the data system
  - ❖ Hired an additional FTE
  - ❖ Explored the possibility of Long-term follow-up
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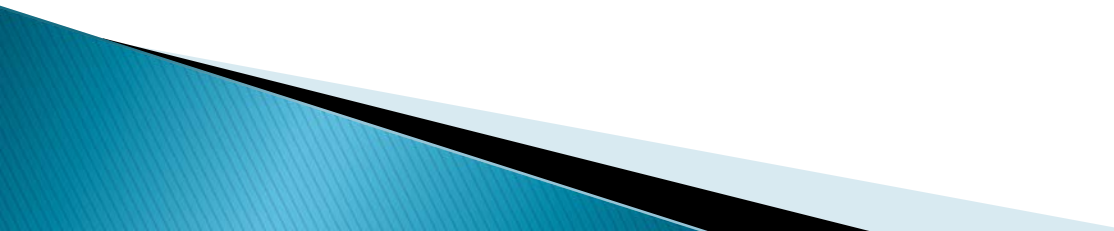
# CHANGES AT THE CENTERS

- ❖ Clinical evaluation needed to occur immediately
  - Dedicated genetic counselor or genetics nurse (practitioner) to manage patients and follow up.
  - Education to clinics who do not have a lot of experience taking care of children with complex medical needs/metabolic disorders.
  - Letters to Primary Care Provider.

## NBS for Pompe disease Follow-up



# ISSUES DURING IMPLEMENTATION

- ❖ More referrals than expected
  - ❖ Parental DNA testing
  - ❖ Prior authorizations
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# SOLUTIONS



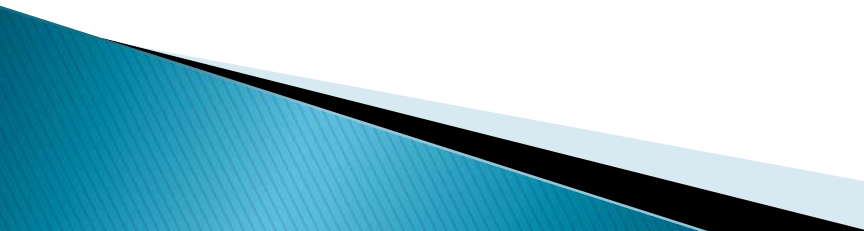
- ❖ Tweaked cutoffs
- ❖ Provided some funding to centers for DNA testing
- ❖ Started early on pre-authorization

# SUPPORT

- ❖ Guidelines from Illinois
- ❖ First in the nation
- ❖ Stayed above water



# ACKNOWLEDGEMENTS

- ❖ Patrick Hopkins, Chief, Newborn Screening State Lab
  - ❖ Jami Kiesling, Newborn Screening Program Manager
  - ❖ LSD Taskforce Members
  - ❖ Follow-up Centers
    - Cardinal Glennon Children's Hospital, St. Louis
    - Children's Hospitals, Columbia
    - Children's Mercy Hospital, Kansas City
    - St. Louis Children's Hospital, St. Louis
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# THANK YOU

## QUESTIONS?

