

# SACHDNC Laboratory Standards and Procedures Subcommittee

## *Subcommittee Report*

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Division of Laboratory Sciences

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National Center for Environmental Health  
Division of Laboratory Sciences



# Welcome to New Members

*Great to see interest from various parties  
and programs*

- ❑ Stan Berberich
- ❑ Joann Bodurtha
- ❑ George Dizikes
- ❑ Koon Lai
- ❑ Bill Slimak
- ❑ Robert Zori
- ❑ Ed McCabe
- ❑ Susan Tanksley

# Updates from the Newborn Screening and Genetics in Public Health Program

*Presentation by Jelili Ojodu, APHL*

- **Update on 50 Years of Newborn Screening Celebration**
  - Newborn Screening Exhibits to 13 States for viewing at State Houses, NBS labs or Health Departments
  - Exhibits for Professional Meetings (including AMCHP, ACMG, ACOG, AAP, APHA, ASH)
  - Coffee Table Book and e-Book
  - Social Media Outreach and Media Coverage
  - Washington DC Reception and Awards Ceremony in September (Elected Officials and leadership)

# Updates from the Newborn Screening and Genetics in Public Health Program

*Presentation by Jelili Ojodu, APHL*

- **Update on Meeting of the Newborn Screening and Genetic Testing Symposium and the International Society for Neonatal Screening**
  - May 5<sup>th</sup> - 10<sup>th</sup>, 2013 in Atlanta, GA
  - Co-sponsored by APHL and CDC
  - Theme: “50 Years of NBS: Celebrating the Past and Preparing for the Future”
  - Laboratory tour of CDC and the Georgia Public Health Labs
  - Registration open at [www.aphl.org](http://www.aphl.org)

# Updates from the Newborn Screening and Genetics in Public Health Program

*Presentation by Jelili Ojodu, APHL*

## □ **Molecular Assessment Program – MAP**

- Address specific concerns as states implement molecular testing into routine workflow
- Goal: Provide feedback (CDC report) to NBS laboratories on their molecular testing capabilities and suggest improvements using peer-review process
- Friendly site visit – not regulatory
- CDC, State Laboratory and APHL representative
- Visits: NY, WI, WA (pilots) then TX and FL ... MN in Feb 2013
- Requests for MAP visits can be made through APHL website

# Updates from the Newborn Screening and Genetics in Public Health Program

*Presentation by Jelili Ojodu, APHL*

- **Newborn Screening Technical assistance and Evaluation Program workflow**
  - Mission, Vision, Program Overview, Goals
  - Accomplishments: Building Team, Networking, Creating national presence, Steering Committee
  - Incorporation of quality indicators and case definitions all aimed at continuous quality improvement
  - Steering committee guided activities

# Newborn Screening Health IT Update

*Presentation by Swapna Abhyankar, NLM*

- **Creation of LOINC Terms for Critical Congenital Heart Disease (CCHD)**
  - Terms created based on input from CNMC, HRSA CCHD Technical Assistance committee and grantees, and Oz Systems
  - Align with the HL7 Implementation Guide
  - Cover the screening results only, Submitted to Regenstrief Institute, waiting for codes
  - Work is ongoing ... Number of CCHD terms being requested

# Newborn Screening Health IT Update

*Presentation by Swapna Abhyankar, NLM*

## □ Mutation Nomenclature

- Have created a LOINC answer list for cystic fibrosis genetic mutation results that includes both HGVS and traditional mutation names
- Working with Regenstrief Institute to figure out best way to incorporate the mutation synonyms and related information into the LOINC answer list
- **Need information from states about:**
  1. Which conditions include mutation testing as part of NBS
  2. What specific mutations are screened for; and
  3. How the results are reported (nomenclature)

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# Evaluation of Current Markers

## *Newborn Screening for Tyrosinemia Type 1*

*Presentation by Dieter Matern, Mayo Clinic*

### □ Overview of Tyrosinemia Type 1

- Incidence ~ 1:100,000 live births (~1:17,000 in Quebec)
- Acute: hepatic failure and death in infancy
- Chronic: hepato-renal symptoms

### □ Concern with NBS for Tyrosinemia Type 1

- Tyrosine is not a specific marker for TYR I (also elevated in TYR II, TYR III, Transient TYR and Liver Disease)
- Succinylacetone is a specific marker for TYR I, but not detectable by routine newborn screening

# Evaluation of Current Markers

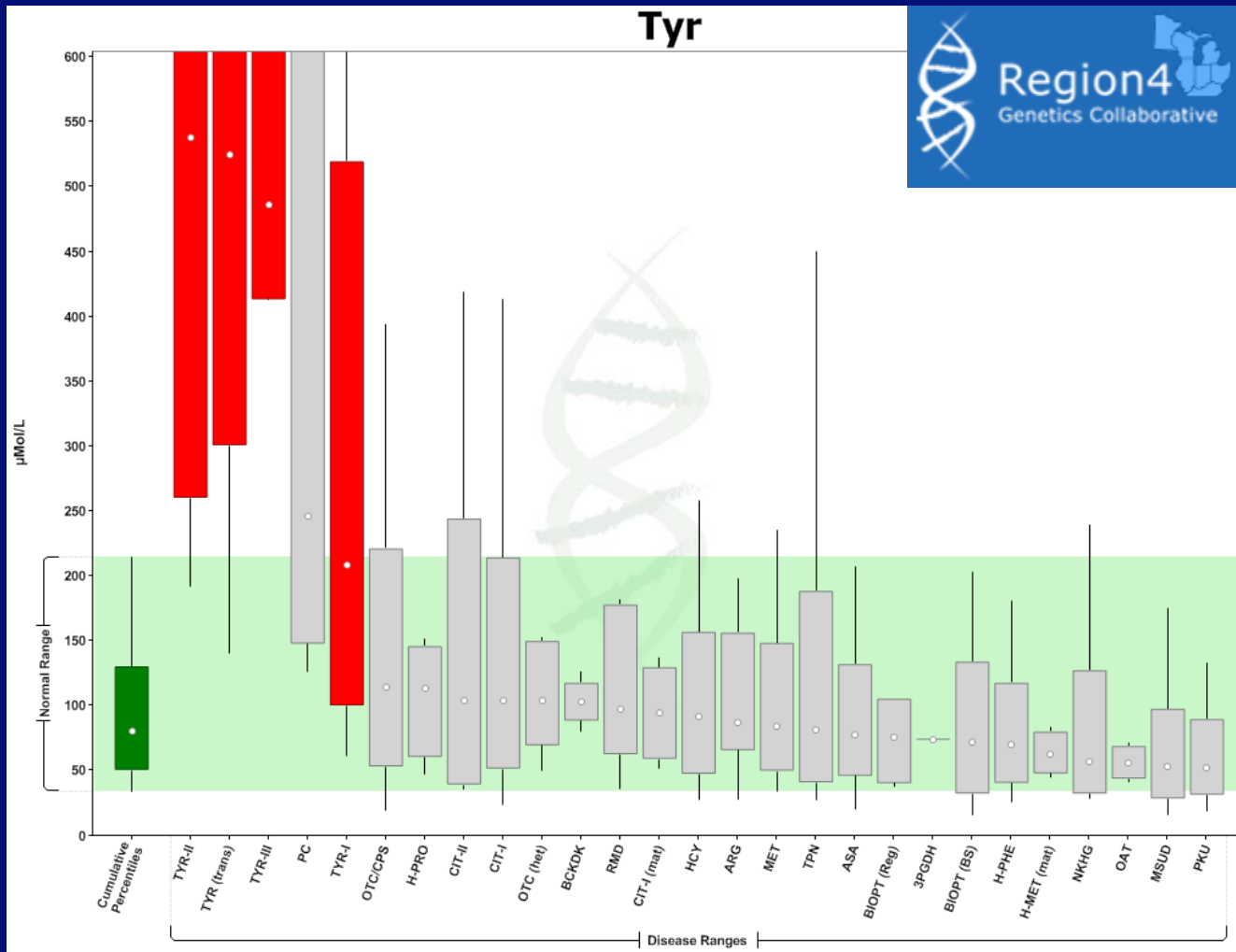
## *Newborn Screening for Tyrosinemia Type 1*

*Presentation by Dieter Matern, Mayo Clinic*

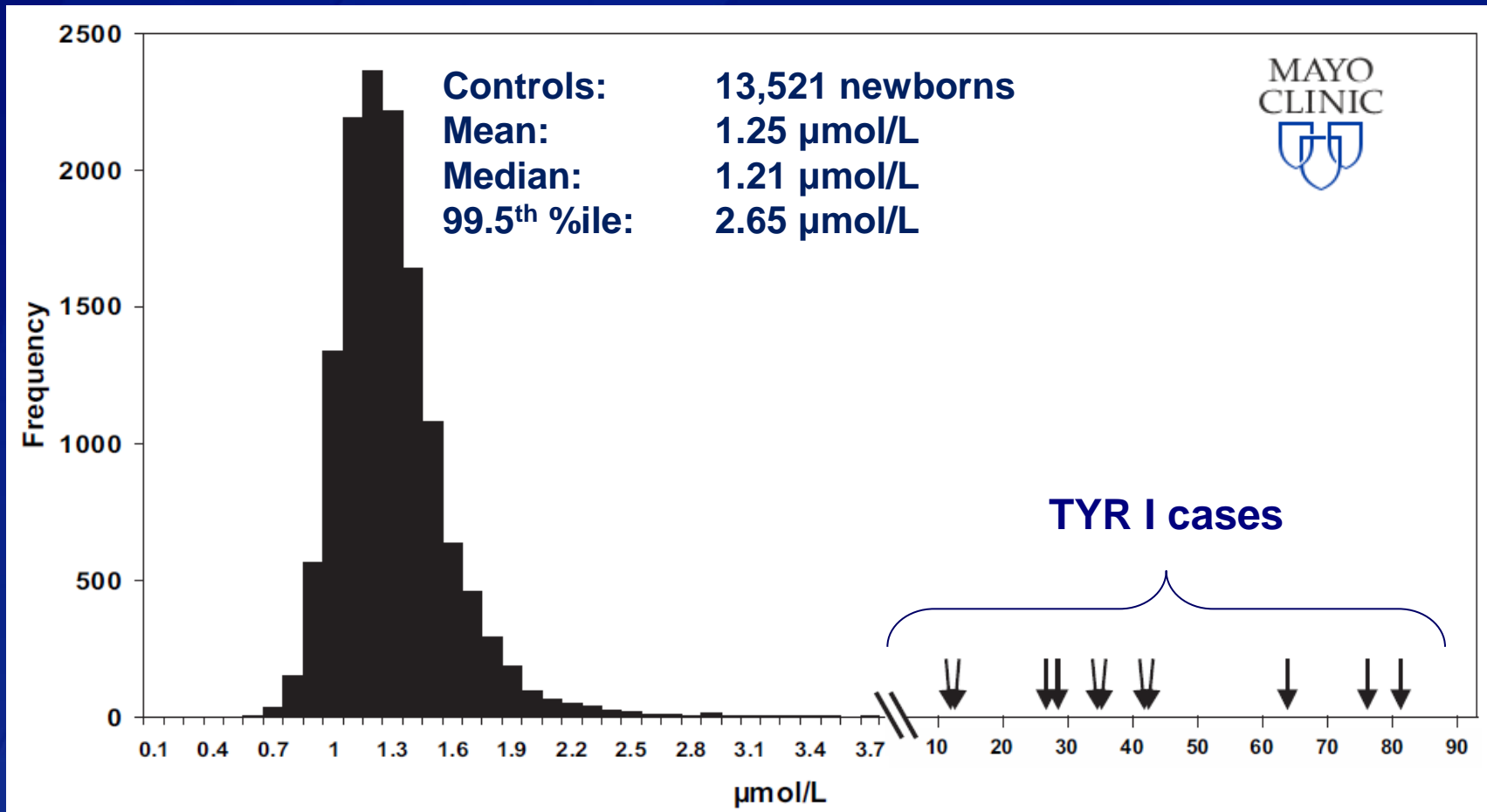
- **Options for screening for TYR I**
  - Removing Tyrosinemia type 1 from state panel
  - Lower cutoff and accept poor sensitivity of Tyrosine
  - Implement an alternative marker
    - 2nd tier test (succinylacetone) when tyrosine is above a clinically significant cut-off
    - Succinylacetone as a primary marker

# Evaluation of Current Markers

## Newborn Screening for Tyrosinemia Type 1



# SUAC Reference Range



\*Turgeon C et al. *Clin Chem* 2008; 54: 657



# **State Survey Investigating the Limitations of SUAC Implementation**

- **CDC interviewed 14 NBS labs**
  - 7 labs measuring SUAC
  - 7 labs not currently measuring SUAC
  - Findings will be reported at the May Meeting
  
- **CDC also received OMB approval on 29 Jan 2013 to survey all US NBS laboratories**
  - Survey questions are listed in the next few slides
  - Findings will be reported at the May Meeting

# Survey: Labs measuring SUAC

**Do you perform SUAC analysis only in response to an elevated tyrosine? (i.e. SUAC is only performed as a second-tier test)**

**Do you use both SUAC and tyrosine as markers to identify infants with Tyrosinemia Type 1?**

**Were budgetary restrictions an obstacle to SUAC implementation in your laboratory?**

**Were technical resources and personnel issues considered obstacles to SUAC implementation in your laboratory? (e.g. were additional staff or instruments required for testing)**

**What were the major challenges to implementing SUAC testing in your laboratory?**



# **Survey: Labs not measuring SUAC**

**Is the laboratory considering the adoption of SUAC testing?**

**Is lab requesting necessary funding, infrastructure, staff, and technical expertise to pilot SUAC testing?**

**Obstacles to implementation: budget, technical resources, personnel, availability of reliable test**

**Impact of formal recommendation by SACHDNC or HHS Secretary to use SUAC as primary marker for TYR I**

**What is labs largest challenge to implementation of SUAC testing?**

# Evaluation of Current Markers

## *Newborn Screening for Tyrosinemia Type 1*

- ❑ TYR I should remain in panel
- ❑ Survey underway to determine limitations and challenges of NBS for TYR I using SUAC in the USA
- ❑ Results from Survey and final report will be presented to SACHDNC at next meeting

# Thank you for your attention!



## *Newborn Screening*

*Saving Lives.  
Promoting Healthier Babies.  
Protecting our Future.*



**For more information please contact Centers for Disease Control and Prevention**

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The findings and conclusions in this report are those of the authors and do not necessarily represent the official position of the Centers for Disease Control and Prevention.

National Center for Environmental Health

Division of Laboratory Sciences

