

Laboratory Standards and Procedures Subcommittee Report

September 20, 2013

Kellie Kelm, Ph.D. – Chair
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Priorities for Lab Subcommittee

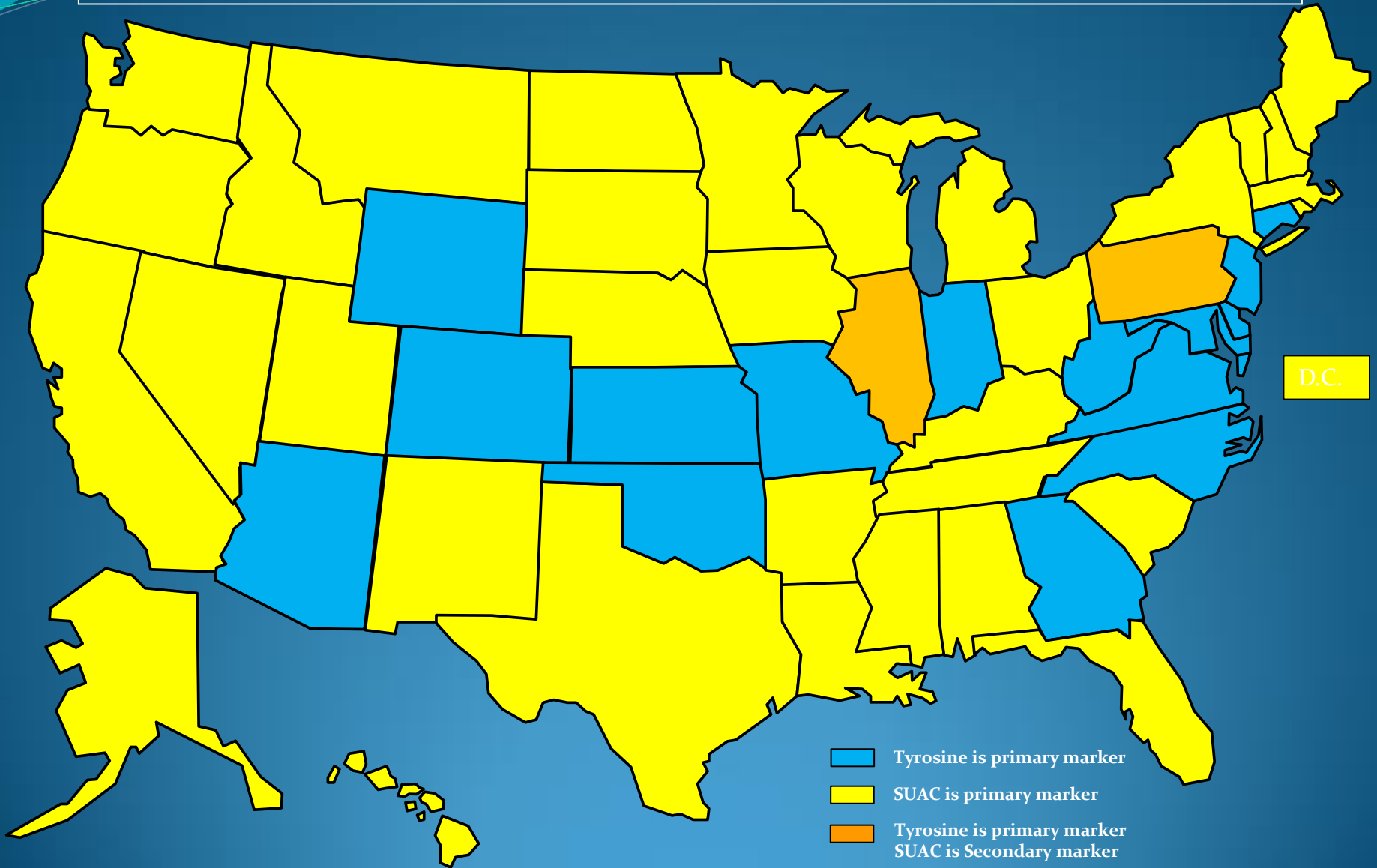
- **Priority A:** Review new enabling/disruptive technologies
 - SUAC Implementation Survey
- **Priority B:** Provide guidance for state NBS programs in making decisions about lab implementation, integration, follow-up (FU), and quality assurance (QA)
 - SCID Slide Deck
 - APHL Updates
- **Priority C:** Establish process for regular review and revision of the Recommended Uniform Screening Panel (RUSP)
 - No update at this time

SUAC Implementation Survey Results

SUAC Implementation Survey Results

- Tyrosine is not a specific marker for TYR I, but also elevated in:
 - Tyrosinemia type II
 - Tyrosinemia type III
 - Transient benign hypertyrosinemia
 - Liver disease
- Succinylacetone is a specific marker for TYR I, but not detectable by routine newborn screening.

NBS for TYR I in the USA



Survey: All U.S. Newborn Screening Laboratories

- Response: 31/38 laboratories (82% response rate)
- Only domestic NBS programs participated in survey
- Does your laboratory measure succinylacetone (SUAC) in dried blood spots?
 - Yes – 16
 - No – 15
- Survey tailored next questions based on this answer

Conclusions

- TYR I should remain in RUSP
- 51 of 51 US NBS programs screen for TYR I
- CDC provides QA/QC/PT for TYR I (incl. SUAC)
- Survey revealed limitations of NBS for TYR I using SUAC in the USA
- Several NBS labs indicated strong pushback for SUAC testing using a specific kit/method
- Potential barriers: Funding, infrastructure, staff, technical expertise
- 8 out of 15 states surveyed stated that a recommendation from DACHDNC/HHS would not influence their NBS programs to adopt SUAC as primary screen for TYR I

Updates

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- SCID slide deck: Monthly calls ongoing
- APHL
 - 50 year NBS celebration and activities
 - NewSTEPS
 - Website updates to include case definitions, quality indicators, and limited state profiles
 - MOUs on data collection
 - MPS 1 public health impact review
 - Survey states to access implementation/uses of recommendations for biochemical genetic testing

Consensus Committee on Immunology and Ligand Assay

Standing
Subcommittee on
Newborn Screening

Working
Group(s)

Document Development
Committee(s)



Newborn Screening Document Library and New Numberings

NBS01 – A6 (LA04) – Blood Collection on Filter Paper for Newborn Screening Programs

NBS02 – A2 (I/LA27) – Newborn Screening Follow-up

NBS03 – A (I/LA31) – Newborn Screening for Preterm, Low Birth Weight, and Sick Newborns

NBS04 – A (I/LA32) – Newborn Screening by Tandem Mass Spectrometry

NBS05 – A (I/LA35) – Newborn Screening for Cystic Fibrosis

NBS06 – A (I/LA36) – Newborn Blood Spot Screening for Severe Combined Immunodeficiency by Measurement of T-cell Receptor Excision Circles

NBS07 – A *New* – Newborn Blood Spot Screening for Pompe Disease by Lysosomal Acid Alpha-Glucosidase Activity Assays

Coming soon – Terminology harmonization

Questions?