

SACHDNC Laboratory Standards and Procedures Subcommittee – Projects for 2013

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CLSI Guideline

- ILA-36, " Newborn Blood Spot Screening for Severe Combined Immunodeficiency by Measurement of T-cell Receptor Excision Circles" received final approval (unanimous vote to approve) in March and is currently being prepared for printing.
- It will be available at the APHL meeting in May.

Newborn Screening Case Definitions

- Over 150 records from 16 actively participating states are available as of today for Endo and CF cases.
- Confirming metabolic and hemoglobinopathy case definitions – in progress
- Consulting with HRSA, NLM, NBSTRN
 - Develop appropriate definitions, codes for new disorders, new data elements

Quality Indicators

- Pilot tested final list of 8 quality indicator measures
- All components of the indicators are undergoing careful scrutiny by stakeholders to ensure agreement on definitions of indicators.
- Will begin collecting data in May
 - Data will be used to provide longitudinal comparisons within a program and comparisons to aggregate data across programs.

SUAC Implementation Survey

- Succinylacetone (SUAC), specific marker: Tyrosinemia Type I
- Survey laboratories
 - Labs measuring SUAC
 - Only as second-tier test
 - Using SUAC and Tyrosine
 - Budgetary restrictions? Additional staff or instruments?
 - Other major challenges
 - Not measuring SUAC
 - Considering SUAC testing
 - Requesting resources for pilot with SUAC
 - Obstacles to implementation, most major challenge
 - Impact if SUAC recommended as a primary marker
- Survey Status
 - Data gathering completed
 - CDC presently analyzing data

Guidance to State Laboratories

- Conditions newly added to RUSP
- Severe Combined Immunodeficiency (SCID)
 - In progress

Questions?