

Laboratory Procedures and Standards Subcommittee

January 26, 2012

Second Screen study

- Stuart Shapira from CDC presented the preliminary data from the retrospective study
 - Higher incidence of CH in 2 screen states
 - Gender difference – 2:1 female to male incidence for CH
 - Birth weight / feeding method difference in CH incidence

Second Screen study

- CAH
 - Reported incidence of CAH in 2 screen states is higher
 - Nonclassical in particular
 - Salt wasting- about 2x as many cases picked up in 2 screen states
 - Gender not different, but significant difference by birth weight-more picked up in normal birth weight range in 2 screen states
 - No difference in types of cases picked up on first screen between groups
 - Second screen did pick up more simple virilizers and nonclassics
- Higher proportion of Hispanics picked up on second screen

Second Screen Study

- Still cleaning data
- Going to do some modeling for the cases
- Going to evaluate the clinical significance of those detected on the second screen
- Limits due to lack of long term follow-up, screening algorithms and retrospective analysis with missing data
- Will follow-up in subcommittee at May meeting

NLM standing items

- CF LOINC mutation reporting
 - Working to standardize the lists and order
 - Have 116 LOINC codes developed
 - cDNA, protein or traditional name searchable
 - Reports will need to be very clear for reporting purposes in order to not be confusing in the report given wide variety in reporting terminology and methodology

NLM standing item

- Hemoglobinopathy reporting
 - New code for conditions suspected
 - Will accommodate for those that confirm diagnosis as well as suspected diagnoses
- Using CLSI guidelines to develop results reporting terminology
- Reason for lab test- to include mandatory second screen.