METHODS

Objective:
To improve screening specificity for 21OHD by modifying 17OHP cutoff levels based on both collection time and birth weight.

Design:
• Retrospective assessment of de-identified screening results from newborns collected from January-December 2019
• Mean/median values from data used to delineate sub-categories within co-variates of collection time (CT) and birth weight (BW)
• 95th and 99th percentiles calculated based on 17OHP levels for CT and BW
• Percentiles used as cutoffs and applied to a cohort of confirmed cases

RESULTS

Figure 1. 17OHP levels are affected by a combination of collection time and birth weight

Figure 2. Application of modified 95th and 99th percentile 17OHP cutoffs to confirmed cases

CONCLUSIONS

• Both BW and CT are found to impact 17OHP levels
• Application of modified first-tier 17OHP cutoffs as the 99th percentile based on CT and BW correctly identified all confirmed cases
• Utilization of the 95th percentile identified two additional, previously missed cases of 21OHD

NEXT STEPS

• Determine number of samples referred for second-tier testing based on proposed cutoff levels
• Compare false positive rates based on current and proposed 17OHP cutoff levels
• Evaluate the impact of other co-variates such as gestational age on 17OHP levels

REFERENCES


BACKGROUND

• Congenital adrenal hyperplasia is a group of autosomal recessive disorders with most cases associated with 21 hydroxylase deficiency (21OHD)
• Newborn screening for 21OHD is a two-tiered approach; first-tier immunoassay for quantification of 17-hydroxyprogesterone (17OHP) followed by second-tier mass spectrometry steroid profile analysis
• Data varies regarding which co-variates to use for establishing 17OHP cutoff levels when screening for 21OHD