

An Updated Look at Newborn Screening

Objectives

- Summarize the rationale that supported development of the current screening programs in the United States and describe the process for adding a screen to the Recommended Uniform Screening Panel (RUSP)
- Describe the screening procedure for disorders on the RUSP and outline the appropriate timing of collection and the approach to be used in obtaining and handling DBS specimens
- Discuss the disorders included in the RUSP and specific issues that arise in regard to primary and second-tier testing including factors that can potentially impact results causing false-positives and missed cases

Content Outline

1. Newborn Screening (NBS)
 - 1.1 Purpose and evolution to standardized screening panels in the US
 - 1.1.1 Recommended Uniform Screening Panel (RUSP) process
 - 1.2 Description of core disorders in RUSP including CCHD and hearing loss
 - 1.3 Description of secondary disorders in RUSP
 - 1.4 Screening procedure
 - 1.4.1 Specimen and specimen collection procedure
 - 1.4.2 Timing of collection
 - 1.4.3 Screening tests
 - 1.4.4 Secondary tests
 - 1.4.5 Management of abnormal test results
 - 1.5 Wilson and Jungner original criteria for screening
 - 1.6 False-positive results and Missed Cases
 - 1.7 Potential future use and issues related to primary or second-tier NGS

Reading Material Resources

Module WB2578: An Updated Look at Newborn Screening is based on the resource listed below. A copy of the resource is included with the module.

Chapter 18 Newborn Screening, Sahai I, in *Avery's Diseases of the Newborn, Eleventh Edition* (2024), Elsevier, 199-215