

# **Course Objectives and Outline**

## **NBS Molecular Training Workshop**

**February 24 – February 28, 2020**

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# Course Objectives

After completion of the NBS Molecular Workshop, participants will be able to:

- ❑ Identify scenarios when a molecular test is appropriate in the newborn screening laboratory
- ❑ Describe the benefits of using a molecular test for different newborn screening disorders
- ❑ Name at least two applications of molecular testing in newborn screening
- ❑ Describe the common protocols for DNA extraction from dried blood spots
- ❑ Describe a PCR-based assay to detect a single or small number of mutations

# Course Objectives – cont.

After completion of the NBS Molecular Workshop, participants will be able to:

- ❑ Conduct DNA extraction, PCR amplification, variant detection method and Sanger sequencing
- ❑ Understand quality control procedures and requirements specific for molecular testing
- ❑ Understand data reporting and interpretation as it relates to molecular testing
- ❑ Describe the follow up process and type of information that physicians consider useful and understandable

# 2020 NBS Molecular Training Workshop Discussions

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Day 1	Pretest Overview Molecular NBS Overview & Utility of Molecular Biology Techniques	Molecular Lab Design, QA/QC Considerations Molarity Future of Molecular NBS
Day 2	Developing a Multiplex Genotyping Assay Fundamental Concepts of Sequencing	Laboratory Automation Demo Utility of Sequencing in NBS How to Validate a Molecular Assay Primer Prep & Dilutions
Day 3	DNA Sequencing & Variant Interpretation Practical Application of Variant Interpretation	
Day 4	Overview of SCID/SMA and real time qPCR Triplex SCID/SMA/RPP30 Assay	Positive Screen Follow-up Process NSQAP Molecular Services Review for test
Day 5	Test Molecular Assessment Program Molecular Resources Website	Q/A of lab issues

# 2020 NBS Molecular Training Workshop Laboratory Workflow

## DNA Extraction

## Variant Detection

## Real Time PCR

Day 1 DNA Extraction (Qiagen Generations S2)  
3 samples with GALT variants  
8 SCID/SMA samples

GALT TaqMan Allelic Discrimination  
Using Qiagen Generations GALT variants

Day 2

GALT TaqMan Allelic Discrimination Analysis  
DNA Sanger Sequencing  
Using pre-extracted DNA from DBS

Day 3

DNA Sanger Sequencing

Day 4

DNA Sanger Sequencing Analysis

SCID/SMA  
Using Qiagen Generations SCID/SMA samples  
SCID/SMA Analysis