NBS Molecular Network Website

2011 NBS and Genetic Testing Symposium

Laura Hancock, M.S.
Molecular Quality Improvement Program
Newborn Screening and Molecular Biology Branch,
Division of Laboratory Sciences
NCEH, CDC

National Center for Environmental Health
Centers for Disease Control and Prevention

APHL | Association of Public Health Laboratories

PROGRAMS | PROFESSIONAL DEVELOPMENT | POLICY | MEMBERSHIP | ABOUT APHL | MEMBER RESOU

- Environmental Health
- Food Safety
- Global Health
- Infectious Diseases
- Informatics
- Laboratory Systems and Standards
- Newborn Screening and Genetics
  - Assuring Laboratory Quality
  - Genetic Testing
  - Policy and Positions
- Public Health Preparedness and Response
- Research

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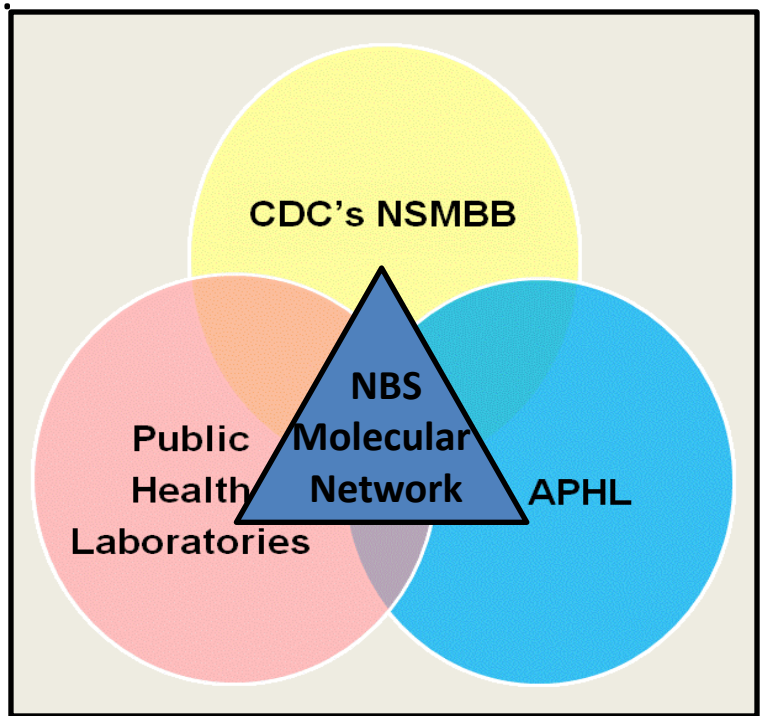
Newborn Screening Molecular Network

A forum to exchange molecular practices, quality improvements and educational resources to enhance laboratory performance.

Mission: Synergistically work with public health partners to provide a NBS specific molecular public health forum to exchange laboratory methods, quality improvements and educational resources to enhance laboratory performance.

Vision: Enhance newborn screening disorder detection with molecular tests.



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Newborn Screening Molecular Network

A LOOK AT PROCEDURES
View Laboratory Assays

LEARN ABOUT AND REQUEST A MAP VISIT
Molecular Assessment Program (MAP)

LOCATE STORED MATERIALS YOU NEED
Archived Molecular Training Materials

FIND WHAT YOU'RE LOOKING FOR
Resources and Laboratory Templates

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Newborn Screening Laboratory Molecular Assays

Please select disorder of interest:

- Cystic Fibrosis
- Galactosemia
- Hemoglobinopathies
- Medium Chain Acyl CoA Dehydrogenase (MCAD)
- Maple Syrup Urine Disease (MSUD)
- Severe Combined Immunodeficiency Disorder (SCID)

Fill out new Assay Description Form for molecular assays ongoing in your laboratory

Newborn Screening Laboratory Molecular Assays – Cystic Fibrosis

Laboratory	Hologic	Luminex	Fluorescent Probe Mutation Detection	DNA sequencing
California Dept of Public Health Genetic Disease Screening Program				
Michigan Dept of Community Health Newborn Screening Program	●			
Minnesota Dept of Health Newborn Screening Program				
New York State Dept of Health Newborn Screening Program	●			●
Texas Dept of State Health Services Newborn Screening Program	●			
Washington State Dept of Health Newborn Screening Laboratory			●	
Wisconsin State Laboratory of Hygiene Newborn Screening Laboratory	●			

DNA sequencing: The process of determining the exact sequence of the bases in a region of DNA. Fluorescent dyes specific for each class of nucleotides are incorporated by PCR and the resulting fragments are separated on a capillary electrophoresis instrument.

Newborn Screening Laboratory Molecular Assays
California Cystic Fibrosis Screening Assay: Luminex Method

Contact:	Marty Kharrazi or George Helmer
Assay duration:	3 - 10 days
Mutations or Target Screened (i.e. CF): Please provide a list of mutations that this assay detects	CFTR panel of 41 mutations designed to detect 90% of known cystic fibrosis mutations in California. Mutations screened: 1288insTA, 1717-1G>A, 1812-1G>A, 2055del9>A, 2105-2117del13insAGAAA, 2307insA, 296+2T>A, 3120+1G>A, 3272-26A>G, 3791delC, 3849+10kbC>T, 3876delA, 406-1G>A, 621+1G>T, 663delT, 711+1G>T, 935delA, A559T, CFTRdele2,3(21kb), delF311, delF508, delI507, G330X, G542X, G551D, G85E, H199Y, N1303K, P205S, Q98R, R1066C, R1162X, R334W, R553X, R75X, S492F, S549N, W1089X, W1204X(3743G>A) , W1204X(3744G>A), W1282X
Assay Limitations:	Assay does not have ability to detect unknown mutations and unknown large deletions
Population Frequency: Please provide citation or link to data (i.e. OMIM or gene review), if possible	1:7000 based on CA NBS data. Our births are 52% hispanic origin
Other information:	Specimens with only one mutation undergo direct DNA Sanger sequencing

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Molecular Assessment Program (MAP)

Request a MAP visit

Representative Name:	
Phone number:	
Email Address:	
Laboratory name:	
Lab Director:	
City:	
State:	
Molecular assays that lab currently analyzes:	
Molecular assays expected in next year:	

View MAP guidance and checklist

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Next Steps

- Look for an email announcing official website launch with access instructions
- Fill out Molecular Assay Description Forms to describe all molecular assays that your lab runs for each disorder
- Visit CDC's Newborn Screening and Molecular Biology Branch table for more information
- All ideas and suggestions are encouraged! (LHancock@cdc.gov)

NBS Molecular Network Website

NBS Molecular Network Steering Committee:

Fred Lorey (CA)
Stan Berberich (IA)
Kevin Cavanagh (MI)
Mark McCann (MN)
Michele Caggana (NY)
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Matthias Martin

CDC:
Suzanne Cordovado
Christopher Greene
Laura Hancock
Carla Cuthbert

For more information please contact Centers for Disease Control and Prevention

1600 Clifton Road NE, Atlanta, GA 30333
Telephone, 1-800-CDC-INFO (232-4636)/TTY: 1-888-232-6348
E-mail: cdcinfo@cdc.gov Web: www.cdc.gov