NBS Molecular Network Website

2011 NBS and Genetic Testing Symposium

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

www.aphl.org
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
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

<table>
<thead>
<tr>
<th>Laboratory</th>
<th>Hologic</th>
<th>Luminex</th>
<th>Fluorescent Probe Mutation Detection</th>
<th>DNA sequencing</th>
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</thead>
<tbody>
<tr>
<td>California Dept of Public Health Genetic Disease Screening Program</td>
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<td>Michigan Dept of Community Health Newborn Screening Program</td>
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<td>Minnesota Dept of Health Newborn Screening Program</td>
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<td>New York State Dept of Health Newborn Screening Program</td>
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<td>Texas Dept of State Health Services Newborn Screening Program</td>
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<td>Washington State Dept of Health Newborn Screening Laboratory</td>
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<td>Wisconsin State Laboratory of Hygiene Newborn Screening Laboratory</td>
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**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

<table>
<thead>
<tr>
<th>Contact:</th>
<th>Marty Kharrazi or George Helmer</th>
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<tbody>
<tr>
<td>Assay duration:</td>
<td>3 - 10 days</td>
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</table>

| 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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Request a MAP visit

<table>
<thead>
<tr>
<th>Representative Name:</th>
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<tbody>
<tr>
<td>Phone number:</td>
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<tr>
<td>Email Address:</td>
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<tr>
<td>Laboratory name:</td>
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<tr>
<td>Lab Director:</td>
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<td>City:</td>
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<tr>
<td>State:</td>
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<tr>
<td>Molecular assays that lab currently analyzes:</td>
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<tr>
<td>Molecular assays expected in next year:</td>
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View MAP guidance and checklist
Newborn Screening Molecular Network

A LOOK AT PROCEDURES
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Resources and Laboratory Templates
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)
Rachel Lee (TX)
Mike Glass (WA)
Mei Baker (WI)

APHL:
Elizabeth Jones
Jelili Ojodu
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

The findings and conclusions in this report are those of the authors and do not necessarily represent the official position of the Centers for Disease Control and Prevention.