Newborn Bloodspot Cystic Fibrosis Communication to Primary Care Providers

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Innovations in Pediatric Healthcare
Maine Chapter of American Academy of Pediatrics
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FLOW CHART OF NEWBORN SCREENING PROCESS-CYSTIC FIBROSIS

1. OOR results called to NBS Program.
2. Call Genetics Office, provide demographics, gene variation(s), forward information.
3. Call PCP with results, implications for patient, and follow-up instructions.
4. Fax information packet to PCP office.
5. Fax information packet to MMP Pulmonology.
6. PCP speaks with Genetic Counselor to arrange sweat testing and counseling.
7. PCP signs, dates, and faxes the Sweat Test Referral to either NL EMMC or MMC contacts listed on Sweat Test Referral Form.
8. PCP discusses results, implications and follow up with family at next visit, preferably scheduled near Sweat Testing appointment.

Maine Center for Disease Control and Prevention
**CF INFORMATION SHEET FOR PRIMARY CARE PROVIDERS**

**Category C: Two gene mutations detected.** The infant most likely has CF.

**Category B: One gene mutation detected.** The relative risk of CF is based on the Immunoreactive Trypsinogen (IRT) concentration measured in the bloodspot. See reporting letter for relative risk.

- Assess for symptoms of Malabsorption and Respiratory problems.
- Newborn bloodspot screens positive for CF Gene variation need Sweat Testing.
- Most Children who have a screen positive for CF (CAT B) will not have CF.
- Babies need to weigh at least 2kg
- Contact NL EMMC Genetics 275-4251 or MMP Pediatric Specialty Care Genetics 662-5522, option 8.
- Infants with Category B results will often be identified as carriers of CF. Infants with Category B results who have a positive sweat test are determined to have CF. These infants have a second mutation that is not included in the newborn DNA assay.

**INFORMATION/FORMS PROVIDED**

**NL EMMC DOCUMENTS:**
- Lab Report from UMMS
- Primary Care Provider Action Sheet
- Primary Care Provider Information Sheet
- Sweat Test Referral Form
- Relative Risk Form
- Fax Cover Sheet NL EMMC Genetics
- Fax Cover Sheet MMP Pulmonology

**MMC DOCUMENTS:**
- Lab Report from UMMS
- Primary Care Provider Action Sheet
- Primary Care Provider Information Sheet
- Sweat Test Referral Form
- Relative Risk form
- Fax Cover Sheet MMP Pulmonology
### CF GENE VARIATION IDENTIFIED
NBS LAB REPORT

**NEWBORN SCREENING SUMMARY RESULTS (Initial Blood Filter Paper Specimen)**

**Action Required: Please see Result-Specific Fact Sheets Attached**

<table>
<thead>
<tr>
<th>Targeted Disorder/Disorder Group</th>
<th>Result</th>
<th>Ref. Range</th>
<th>Interpretation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Acylcarnitines Disorders</td>
<td></td>
<td></td>
<td>In Range</td>
</tr>
<tr>
<td>Aminoacidopathies Group 1</td>
<td></td>
<td></td>
<td>In Range</td>
</tr>
<tr>
<td>Aminoacidopathies Group 2</td>
<td></td>
<td></td>
<td>In Range</td>
</tr>
<tr>
<td>Tyrosinemia I - SUAC</td>
<td></td>
<td></td>
<td>In Range</td>
</tr>
<tr>
<td>Biotinidase Deficiency</td>
<td></td>
<td></td>
<td>In Range</td>
</tr>
<tr>
<td>Congenital Adrenal Hyperplasia (CAH)</td>
<td></td>
<td></td>
<td>In Range</td>
</tr>
<tr>
<td>Congenital Hypothyroidism</td>
<td></td>
<td></td>
<td>In Range</td>
</tr>
<tr>
<td>Cystic Fibrosis</td>
<td></td>
<td></td>
<td>Out Of Range</td>
</tr>
<tr>
<td>Cystic Fibrosis</td>
<td></td>
<td></td>
<td>Out Of Range</td>
</tr>
<tr>
<td>1 CFTR Variant Detected↑</td>
<td></td>
<td>None of Tag H 394↑</td>
<td>Out Of Range</td>
</tr>
<tr>
<td>IRT</td>
<td>97.29%</td>
<td>≤ 95.0%</td>
<td>Prompted CFTR Variant</td>
</tr>
<tr>
<td>CFTR Variant 1</td>
<td>DF508↑</td>
<td>None of Tag H 394↑</td>
<td>Out Of Range</td>
</tr>
<tr>
<td>CFTR Variant 2</td>
<td>None of Tag H 394↑</td>
<td>None of Tag H 394↑</td>
<td>In Range</td>
</tr>
</tbody>
</table>

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### PRIMARY CARE PROVIDER
CF ACTION SHEET-CAT B

**Patient Name:**
DOB:
DOB:

Your patient was found to have one gene mutation for Cystic Fibrosis in the newborn bloodspot screening. See attached report.

**Recommendations and Next Steps:**
- **Sign and fax the referral/order form (attached) to the accredited sweat test laboratory at EMMC fax: 973-7988 (referral/order form to be signed by PCP).**
- **Garrett Fouch, NL EMMC Genetics will call your office with an appointment for the sweat test and free genetic counseling. Garrett may be contacted at 275-4259.**

- **Please inform family of results and sweat test appointment in person.**

For questions call Maine Newborn Bloodspot Screening Program 207-287-5357
PRIMARY CARE PROVIDER
CF ACTION SHEET-CAT C

Patient Name:
DOB:
DOS:

Your patient was found to have 2-gene mutations for Cystic Fibrosis (CF) in the newborn bloodspot screening. Your patient most likely has CF. See attached report.

Recommendations:
☐ A Pediatric Pulmonology Practitioner at MMP Pediatric Specialty Care Pulmonology will be contacting you to discuss results and schedule initial CF Clinic appointment prior to informing the family. Each gene mutation has a different clinical expression; some may be milder and others more concerning. MMP Pediatric Specialty Care Pulmonology: 207-662-5522, press option 3
☐ Inform Family of results in person
☐ Review Cystic Fibrosis (CF) Newborn Bloodspot Screening Information Sheet for Primary Care Providers (attached)

SWEAT TEST REFERRAL FORM

Maine Newborn Screening Program Referral Form for Sweat Testing

Date of Referral:
Preparer Location: MHC, Norridgewock

Date:
Preparer Name:

Insurance Information:

Primary Payor:

Policy Number:

Policy Holder:

Policy Effective Date:

Policy Expiration Date:

Physician Information:

Physician Name:

Physician Signature:

MHC Norridgewock

11 State House Street
Augusta, ME 04333

PH: 207-622-5000

Fax: 207-622-5001

Maine Center for Disease Control and Prevention
**RELATIVE RISK FORM**
**CAT B CF GENE VARIATION 1:100 RISK**

<table>
<thead>
<tr>
<th>Name:</th>
<th>DOB:</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mother:</td>
<td>Birth Hospital:</td>
</tr>
<tr>
<td>Provider:</td>
<td>Report Date:</td>
</tr>
</tbody>
</table>

This infant has a positive Cystic Fibrosis Newborn Bloodspot Screen (Category B)

Cystic Fibrosis is a genetic condition that causes malabsorption, failure to thrive and chronic pulmonary disease.

**Cystic Fibrosis newborn screening results for this infant**

Immunoreactive Trypsinogen* 72.1 μg/ml 98.94 % (Reference ≤95%)

ONE MUTATION detected by screen** W1282X/+  

This infant may have cystic fibrosis with a second mutation that is not on the panel or this baby may be an unaffected carrier. The risk of this infant having CF based on the IRT is 1:100.

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**MAINE NEWBORN CYSTIC FIBROSIS SCREENING ALGORITHM:**
**CATEGORY B: ONE GENE VARIATION**

- **Call Genetics Monday-Friday**
  - Provide demographics
  - Report gene variations identified
  - Report (IRT) Concentration/Percentile
  - Fax or email information packet

- **Fax MMP Pulmonology Monday-Friday**
  - Fax Newborn bloodspot lab report and Relative Risk Form to Ann Ladner, RN

- **Call PCP Office Monday-Friday**
  - Report gene variation identified
  - Explain need for Sweat Testing
  - Ask when patient is scheduled to be seen
  - Fax information packet to office
MAINE NEWBORN CYSTIC FIBROSIS SCREENING ALGORITHM:
CATEGORY C: TWO GENE VARIATIONS

Call Genetics Monday-Thursday
- Provide demographics
- Report gene variations identified
- Report (IRT) Concentration/Percentile
- Fax or email information packet

Fax MMP Pulmonology Monday-Thursday
- Report gene variations, IRT Concentration/Percentile to Ann Ladner, RN
- Provide demographics

Call PCP Office Monday-Thursday
- Report gene variations identified
- Report IRT Concentration/Percentile
- Request that patient be seen ASAP
- Fax information packet to office

Thank You

Jodi Philippon, RN, BSN
Newborn Bloodspot Screening and Follow Up Nurse

Contact Information
(207)287-5351
Jodi.Philippon@maine.gov