Newborn Bloodspot Cystic Fibrosis Communication to Primary Care Providers

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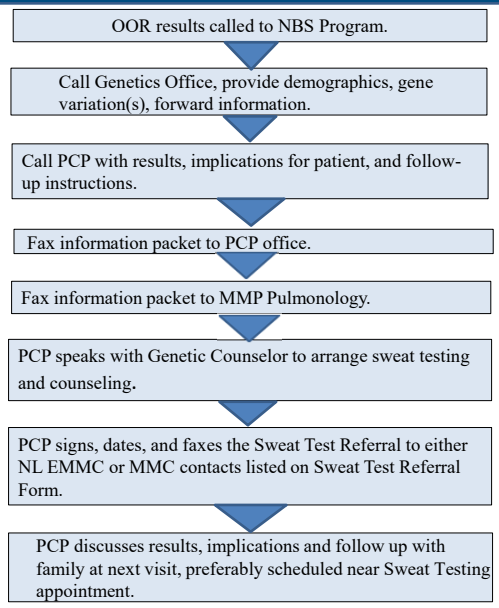
FLOW CHART OF NEWBORN SCREENING PROCESS-CYSTIC FIBROSIS



Maine Center for Disease Control and Prevention

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CF INFORMATION SHEET FOR PRIMAY 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.**

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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
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- Fax Cover Sheet MMP Pulmonology

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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

Targeted Disorder/Disorder Group	Result	Ref. Range	Interpretation
* Acylcarnitines Disorders			In Range
* Aminoacidopathies Group 1			In Range
* Aminoacidopathies Group 2			In Range
* Tyrosinemia I - SUAC			In Range
* Biotinidase Deficiency			In Range
Congenital Adrenal Hyperplasia (CAH)			In Range
Congenital Hypothyroidism			In Range
Cystic Fibrosis	1 CFTR Variant Detected †	None of Tag-It 39+4 †	Out Of Range
Cystic Fibrosis			Out Of Range
IRT	97.29%	≤ 95.0%	Prompted CFTR Variant
CFTR Variant 1	DF508 †	None of Tag-It 39+4 †	Out Of Range
CFTR Variant 2	None of Tag-It 39+4 †	None of Tag-It 39+4 †	In Range

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PRIMARY CARE PROVIDER
CF ACTION SHEET-CAT B

Patient Name:
DOB:
DOS:

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 Foutch, 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 <u>in person.</u>**

For questions call Maine Newborn Bloodspot Screening Program 207-287-5357

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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)

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SWEAT TEST REFERRAL FORM

Maine Newborn Screening Program Referral Form for Sweat Testing

Instructions: Complete this form and fax it to the number indicated at the bottom of the form.

Maine Newborn Screening Program
11 State House Station
Augusta, ME 04333
207-287-5357

Ordered Procedure: Sweat Test

Procedure Date: _____ **

Procedure Location: MMC-NorDx

Date of Referral: _____

**To (re)schedule sweat test and counseling call Genetics at MMC 662-5522 opt 8 , EMMC 973-7520

PATIENT INFORMATION

Name ☐ Male DOB _____
Mom: ☐ Female
Address Weight _____ G
City Primary Phone Number _____
State _____ Zip code Screening Result Category B

INSURANCE INFORMATION

PRIMARY INSURANCE SECONDARY INSURANCE
Insurance Name Insurance Name
Policy # Policy #
Policy Holder Birth Date Policy Holder Birth Date
Policy Holder name Policy Holder name

PHYSICIAN INFORMATION

Ordering Physician Name: Phone: Fax:
Primary Care Physician Name
Ordering Physician Signature

ORDERING DIAGNOSIS INFORMATION (ICD 10 CODES REQUIRED)

[X] P09 ABNORMAL FINDINGS NEONATAL SCREEN

FAX COMPLETED REFERRAL FORMS TO SWEAT LAB LOCATION

[X] NorDx (Maine Medical Center) Fax: 1-855-372-9695

[ ] Eastern Maine Medical Center Lab Fax: 207-973-7988

Attention Sweat Lab: Fax a copy of results to :

[X] Maine Newborn Screening Program Fax: 207-287-4743
[X] MMP-Pedi Genetics Fax: 207-774-1814
[ ] EMMC-Pedi Genetics Fax: 207-973-7674
[X] CF Center @ MMC Fax: 207-662-5527

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RELATIVE RISK FORM-
CAT B CF GENE VARIATION 1:100 RISK

Name: DOB:
Lab ID:
DOS:
Mother:
Birth Hospital:
Provider:
Report Date:

This baby 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**	ng/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.

*Immunoreactive Trypsinogen (IRT) is a biomarker used in NBS CF screening because it is measurable on bloodspot and elevated in most babies with cystic fibrosis and some CF carriers in the first week of life. Studies have shown that based on the IRT level the relative risk of cystic fibrosis can be calculated in Category B infant.

** CFTR assay utilizes ASR (Analyte Specific Reagent) CFTR 39+4: 39 mutation panel with reflex analysis for I506V, I507V, F508C, 5, 7.9T as appropriate. The 39 mutation panel included ΔF508, R117H,G551D, G542X, W1282X, N1303K, R334W, 621+1G>T, R553X, ΔI507,1717-1G>A, R347P, R560T, 3849+10kbC>T, A455E, 3120+1G>A, 3659delC, R1162X, 711+1G>T, 2789+5G>A, G85E, 1898+1G>A, 2184delA, 1078delT, 394delTT, Y122X, R347H, V520F, A559T, S549N, S54(T>G),1898+5G>T, 2183AA>G, 3207insA, Y1092X, M1101K, S1255X, 3876delA, 3905insT. This test has not been cleared or approved by the FDA. However, the New England Newborn Screening Program determined the performance characteristics of the test and the FDA has determined that its clearance and approval are not required for the NENSP-specific uses.

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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

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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

Maine Center for Disease Control and Prevention

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Thank You

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

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

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