Maine Center for Disease Control and Prevention
An Office of the Department of Health and Human Services
Paul R. LePage, Governor Mary C. Mayhew, Commissioner

Department of Health and Human Services
Maine Center for Disease Control and Prevention
286 Water Street
11 State House Station
Augusta, Maine 04333-0011
Tel.: (207) 287-8016; Fax: (207) 287-9058
TTY Users: Dial 711 (Maine Relay)

Direct Line Tel. (207)287-5351 Fax. (207)287-4743

Maine Newborn Bloodspot Screening Program

ACTION SHEET
For Primary Care Provider

YOUR PATIENT HAS A POSITIVE CYSTIC FIBROSIS (CF) NEWBORN SCREENING RESULT.

RECOMMENDED NEXT STEPS FOR ALL BABIES WITH POSITIVE CF SCREENS BY PRIMARY CARE PROVDIER

The Newborn screening program will guide you in scheduling of the sweat test and genetic counseling with specific instructions depending on infants result category

1. To schedule sweat testing at MMC or CMMC, contact the Genetics Center at MMC (662-5522, Option 8). For testing at EMMC, call the CF clinic at 973-7520. **The genetic counselor will coordinate scheduling of the sweat test at an accredited CF sweat lab and genetic counseling.** She will call you back with the appointment time/date for you to communicate to the family. If the sweat test needs to be rescheduled, contact the appropriate genetic counselor above.
2. Physician order is required for the sweat test. Fax the signed form provided to the indicated laboratory.
3. **Please discuss the results with the family in person at the two week check up**. Please give them the three page handout provided and the sweat test/counseling time and location.

*Assess the baby for malabsorption and respiratory problems.
*For any medical questions regarding the care of a child with CF, call the CF Center at MMC (662-5522 option 3).
***NICU: Consult CF Center and order sweat test when >2kg >2wks and stable.**

The specific risk is dependent on the category of positive screen as indicated below.

- **Category C**: Two gene mutations detected. **The infant has CF**.
- **Category B**: One gene mutation detected. **The relative risk of CF is based on the IRT concentration. See reporting letter for relative risk.**
  - Infants with Category B results who have a positive sweat test are confirmed to have CF. These infants have a second mutation that is not included in the newborn screen DNA assay **OR**
  - Infants with Category B results may also have a negative sweat test identifying them to be unaffected carrier.

Maine Center for Disease Control and Prevention
An Office of the Department of Health and Human Services
Paul R. LePage, Governor Mary C. Mayhew, Commissioner

Department of Health and Human Services
Maine Center for Disease Control and Prevention
286 Water Street
11 State House Station
Augusta, Maine 04333-0011
Tel.: (207) 287-8016; Fax: (207) 287-9058
TTY Users: Dial 711 (Maine Relay)

Direct Line Tel. (207)287-5351 Fax. (207)287-4743

Maine Newborn Bloodspot Screening Program

Important Points and Screening Interpretation For PCP's

- Any baby who has a positive CF newborn screen result should be assessed for symptoms of malabsorption and respiratory problems and must be referred for a sweat test, even if the parents had prenatal CF carrier testing.
- Of our 14,000 births annually in ME, about 50 babies will screen positive for CF. Of these, 5-10 babies will be diagnosed with CF.
- Face to face encounters are recommended when you tell a family that their baby screened positive for CF.
- It is strongly recommended that sweat tests be performed at a CF Foundation accredited sweat test lab. Labs that are not accredited often have a high level of error - both false positive, false negative and quantity not sufficient results. The three CF Foundation accredited sweat labs in Maine are at Central Maine Medical Center in Lewiston, Eastern Maine Medical Center in Bangor and Maine Medical Center in Portland.
- While at the sweat test appointment the family will meet with a genetic counselor to discuss the inheritance pattern for CF. Families whose babies have their sweat test at EMMC will be provided genetic counseling by phone. If the sweat test is negative and the baby is found to be a carrier of a CF mutation, further genetic counseling will be offered to the family.
- Babies with a Category C result will be referred to the MMC CF Center for confirmatory sweat test, genetic counseling (Update based on discussions about timing of counseling for Cat C) and clinical evaluation. If the family is from northern Maine, they can receive ongoing CF management at EMMC.
- The goal is to schedule the test and counseling so that no more than one week pass between the family knowing about the positive result and actually getting the sweat test. The literature notes that this will help minimize the family's anxiety and angst as much as possible.
- Remind families that the majority of children who have a screen positive with a Category B (1 gene mutation) result will not have CF. The specific risk will be discussed when results are reported to you.
- A sweat test cannot be done until a baby is at least 2 weeks old, term or weighs at least 2 kg. Premature, younger and smaller babies do not produce enough sweat to give a reliable result.
- Maine (Should we add the NBS Program?) CF Center or Genetic Center staff will be available to talk to you and/or the family at any time to clarify information or answer questions, EMMC CF/Genetics: 973-7559 or MMC CF Center: 622-5522 option 3 or Genetics 662-5522 (Option 8).
- Newborn screening for CF is recommended by the CF Foundation (www.cff.org). The Centers for Disease Control (CDC) has found a favorable risk benefit after evidence-based review (MMWR, Oct 15, 2004/53 (RR13), 1-36).

www.mainepublichealth.gov\bloodspot