

CYSTIC FIBROSIS REFERRAL GUIDELINE

MAINE MEDICAL PARTNERS - PEDIATRIC SPECIALTY CARE (DIV. PULMONARY MEDICINE) • 887 CONGRESS ST, SUITE 320, PORTLAND, ME • (207) 662-5522

HIGH RISK

SUGGESTED EMERGENT CONSULTATION

SYMPTOMS AND LABS

Abnormal CF newborn screen
Category C
2 DNA mutations identified

NBS Program will call and fax Urgent CF results to the PCP and the CF Center

SUGGESTED PREVISIT WORKUP

PCP should discuss results with family in person and give family the handout "My baby has a positive newborn screen-two CF gene changes"

A CF provider will call the PCP and arrange for an urgent Pulmonary appointment.

The CF Clinic will arrange sweat testing when baby is 2 weeks old and 2 kg in weight (small babies do not give enough sweat)

Genetic counseling will be arranged by the CF Center

MODERATE RISK

SUGGESTED CONSULTATION OR CO-MANAGEMENT

SYMPTOMS AND LABS

Non-urgent abnormal CF newborn screen
Category B

High IRT and 1 DNA mutation identified

NBS Program will call and fax NON-URGENT results to the PCP, the CF Center and Genetics

A sweat test is required

SUGGESTED WORKUP

The PCP should discuss results with family in person and give them the handout "My baby has a positive newborn screen-one CF gene change"

The PCP should then contact the MMP genetic counselor (207-662-5522 option 8) to schedule a sweat test and genetic counseling at MMC/CMMC lab

The PCP must complete the sweat test order form and fax it to the lab

Sweat test results are reported to the PCP who should inform the family

*QNS sweat tests must be repeated

** Indeterminate sweat tests must be repeated at 8 weeks of age

LOW RISK

SUGGESTED ROUTINE CARE

SYMPTOMS AND LABS

Negative Newborn Screen
Referral not needed

The CF newborn screen is only a screen. It does not completely exclude a diagnosis of CF

If a patient displays signs or symptoms such as failure to thrive, steatorrhea, recurrent pulmonary symptoms or rectal prolapse, CF should be considered

CF should still be considered if the parents had normal prenatal CF carrier screening

SUGGESTED MANAGEMENT

Patients should be referred to Pulmonary for consultation if CF is being considered

Only the pediatric pulmonologists can order a sweat test on patients outside the newborn period

CLINICAL PEARLS

- The sweat test is still required for a diagnosis in all cases.
- Sweat tests must be performed at an accredited lab either MMC Lab (Fax: 207-662-6176) or CMMC Lab (Fax: 207-795-2336).
- The initial sweat test should be performed within a week of disclosure of results to the family.
- IF THE INITIAL SWEAT TEST APPOINTMENT NEEDS TO BE CHANGED PLEASE CONTACT THE GENETIC COUNSELORS (662-5522 OPTION 8) so that they can be present to provide genetic counseling.
- The PCP should order the repeat sweat test in a newborn if the test is indeterminate (values of 30-60 mmols/L) when the baby is 8 weeks old.
- The PCP should order the repeat sweat test in a newborn if the test is QNS (quantity insufficient).

Maine Medical
PARTNERS

These clinical practice guidelines describe generally recommended evidence-based interventions for the evaluation, diagnosis and treatment of specific diseases or conditions. The guidelines are: (i) not considered to be entirely inclusive or exclusive of all methods of reasonable care that can obtain or produce the same results, and are not a statement of the standard of medical care; (ii) based on information available at the time and may not reflect the most current evidenced-based literature available at subsequent times; and (iii) not intended to substitute for the independent professional judgment of the responsible clinician(s). No set of guidelines can address the individual variation among patients or their unique needs, nor the combination of resources available to a particular community, provider or healthcare professional. Deviations from clinical practice guidelines thus may be appropriate based upon the specific patient circumstances.