

# Madigan Army Medical Center

## Referral Guidelines

### Cystic Fibrosis

#### Diagnosis/Definition

Cystic Fibrosis (CF) is a genetic disease characterized by chronic lower respiratory infections, poor weight gain, steatorrhea, and abnormally high sweat electrolyte concentrations. Multiple CF gene mutations account for a spectrum of clinical manifestations and disease severity.

#### Initial Diagnosis and Management

- Classic Cystic Fibrosis is manifested by the clinical findings of recurrent lower respiratory symptoms, bronchiectasis, fat malabsorption syndrome, meconium ileus (at birth), or failure to thrive, either as isolated findings or in combination.
- Atypical CF may present with a milder symptom complex to include chronic productive cough, chronic or recurring sinusitis, recurrent idiopathic pancreatitis, and normal pancreatic function. The sweat chloride is commonly in the borderline range (40-60 mEq/L).
- An abnormally elevated sweat chloride value (> 60 mEq/L) is usually diagnostic of CF.
- An abnormal CF newborn screen is an indication for a sweat chloride test and/or CF mutation analysis, and a prompt clinical evaluation by a physician.
- Other clinical findings which may prompt the clinician to order a sweat chloride include digital clubbing, rectal prolapse, persistent sinusitis (especially with nasal polyps), *Pseudomonas aeruginosa* cultured from either throat swab, sputum, or sinus culture, bronchiectasis, unexplained hepatic dysfunction, bowel obstruction, salt depletion syndrome, and azoospermia in the young adult male.
- When CF is suspected then a sweat chloride test and/or CF mutation analysis should be ordered. The DNA analysis for CFTR mutations identifies approximately 95% of the CF gene mutations, so a negative test does not completely rule out C.
- When CF is considered a possible diagnosis, a sweat test should be ordered early in the evaluation. If a sweat test cannot be performed easily (i.e., due to the patient being unable to produce enough sweat for the test or the patient residing a long distance away from a lab which can perform the sweat test) then the DNA/CF mutation blood test should be ordered (as miscellaneous shipping in CHCS).

#### Ongoing Management and Objectives

- Objectives of primary care management of CF following the initial specialty referral include evaluation and treatment of acute minor illnesses and injuries, and periodic well child care and immunization
- All patients under 23 years of age with proven or suspected CF should be referred to the Division of Pediatric Pulmonology for specialty care at the time the diagnosis or tentative diagnosis is made by DNA analysis for CFTR mutations or positive sweat chloride test.

#### Indications for Specialty Care Referral

- CF patients 23 years of age and older should be referred to the adult pulmonary medicine service
- All infants with a positive newborn screen for cystic fibrosis must have a sweat chloride test performed and have a referral to pediatric pulmonology placed.

- All patients with a positive (chloride value >60 mEq/L) or borderline (chloride: 40-60 mEq/L) sweat chloride should be referred for specialty care. Patients with positive DNA/CF mutation blood tests should also be referred once the test results are known.
- Patients with persistent symptoms suggestive of CF but with normal sweat tests and/or negative DNA/CF mutation blood tests should also be referred for evaluation.

### **Criteria for Return to Primary Care**

- Ongoing management of the child, adolescent, or young adult with CF is the responsibility of the pediatric pulmonary specialist.
- Specialty care follow-up will not require primary care referral and will be established with the patient by the specialist.
- Following initial diagnosis and establishment of an adequate treatment plan by the specialist, the patient may return to the primary caregiver for care of acute minor illnesses and injuries and for routine well child care.
- Management of acute illnesses involving increased lower respiratory symptoms, abdominal pain, or increased malabsorption symptoms should be discussed with the subspecialist.
- Adult patients with typical or atypical CF should be seen at least once by the adult pulmonary specialist, and advised where to seek primary health care.

Last Review for this Guideline: **February 2012**

Referral Guidelines require review every three years.

Maintained by the Madigan Army Medical Center - Quality Services Division  
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