MISSION Children's Hospital | MISSION HEALTH PARTNERS

REFERRAL GUIDELINE

Pulmonology	Abnormal Newborn Screen for Cystic Fibrosis

Background	Cystic Fibrosis (CF) is included in the standard newborn screen. CF screening involves two stages: 1. immunoreactive trypsinogen (IRT) assay 2. follow-up DNA testing (CFTR mutations) for elevated IRT Confirmatory testing for CF is achieved by pulmonologist administered sweat test, and is indicated if both IRT and DNA Testing are positive. Most babies who are screened positive on the initial stage of IRT do not have CF (explanation below). PCP's should be well equipped to educate and answer questions as it pertains to positive (and negative) CF screens.	
Initial Evaluation	The top 5% of the pooled IRT results from NC that day, or any sample > 100 are automatically submitted for the second stage testing which includes DNA testing @ Wisconsin State Lab. Screening results interpretation: • Negative: IRT < 100, and not in the top 5%. No further testing is indicated. • Negative: IRT > 100 or in top 5% but DNA Testing does not identify any CF mutations. • Positive: If one or two mutations are detected then the state will forward the data to pediatric pulmonology in order to schedule a sweat test.	
Initial Management	IRT TESTING IS NOT PERFECT OR ABSOLUTE. IRT values are often elevated in certain disease states such as GI disease in the postnatal period. Recommendations: • IRT Negative: Reassure parents that CF is highly unlikely. DNA testing and sweat test is not indicated. • IRT Positive / DNA Testing Negative (no mutations are identified): Reassure parents that CF is highly unlikely. Sweat test is not indicated. • IRT Positive / DNA Testing Positive: If one or two mutations are identified then the State Agency will refer the patient for a sweat test. Inform family that the state will arrange a sweat test and formal consultation same day with a pediatric pulmonologist. Presence of a mutation suggests patient is either a carrier or may have CF. Some patients with only one mutation and borderline sweat test will be diagnosed with "CFTR metabolic syndrome." Please defer specific disease counseling until patient completes sweat test and pulmonary consultation.	
When to Refer	IRT Positive / DNA Testing Positive: (minimum of 1 mutation identified) (Referral is often initiated by the State.)	
Pre-Visit Work Up	• Fax any paperwork from newborn screen (IRT and DNA) to pediatric pulmonary (828.213.1814) • Neonatal history of meconium ileus, failure to thrive, respiratory concerns • Known family history regarding CF	
Co-management Strategy (as appropriate)	**Specialist scope of care** Coordinate follow up and further testing as indicated.	**Primary care scope of care**
Return to Primary Care Endpoint	Negative sweat test and minimal concerns for presence of CF Please note: Patients with a borderline sweat test or other concerns will continued to be followed but pediatric pulmonology.	
Guidelines Referenced	Description of newborn screening from the CF Foundation. https://www.cff.org/What-is-CF/Testing/Newborn-Screening-for-CF/	

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