CFRI Cystic Fibrosis Research Institute

RESEARCH · EDUCATION · ADVOCACY · SUPPORT

For More Information:

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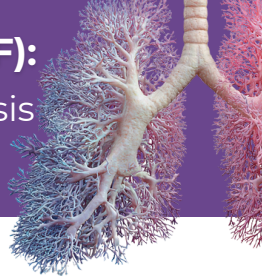
CFRI offers a range of *free* or *low-cost* resources to support patients, families, loved ones & caregivers:

- Support groups & peer connections
- Up-to-date research & educational materials
- Mental & physical wellness classes
- Financial assistance for counseling
- Programs in English & Spanish
- And more!

CYSTIC FIBROSIS (CF):

Detection and Diagnosis in Diverse Populations



Clinical Reasoning Pre-Assessment

Clinical Pre-Assessment Answer

Correct Answer: All of the patients.

All of the cases highlight clinical presentations where a diagnosis of cystic fibrosis is often missed or delayed due to racial and ethnic bias, the presence of extrapulmonary symptoms, prior misdiagnosis, or reduced newborn screening sensitivity in people of color.

Beyond One Color. Beyond One Breath. #ISeeCF

JOIN THE MOVEMENT:

Show your support and learn more about the case studies, clinical insights and resources referenced in the brochure.

SCAN

Which of the following patients may have undiagnosed CF?

- A 2-week-old Hispanic newborn with prolonged jaundice, poor feeding, and weight loss. Newborn screening was normal.
- A 15-year-old white male who recently immigrated from Eastern Europe with chronic cough, wheezing, and a history of recurrent pneumonia.
- A 45-year-old Black woman with a history of irritable bowel syndrome, chronic constipation, and nutritional insufficiency despite stable intake, who was recently diagnosed with colorectal cancer.
- A 21-year-old South Asian American male with chronic sinus congestion, low BMI, and rectal bleeding, who was previously diagnosed with anxiety and hemorrhoids.

Please review the inside of the brochure before finalizing your answer.
The correct response is located on the back panel.

References

1. The Clinical and Functional Translation of CFTR. (n.d.). CFTR2. http://cftr2.org
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3. Cystic Fibrosis Foundation Patient Registry. (2023). 2022 Annual Data Report. Bethesda, MD: Cystic Fibrosis Foundation.
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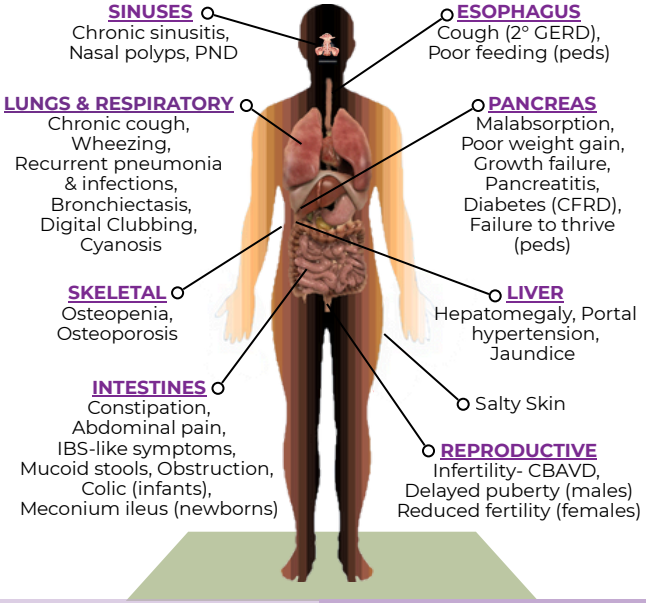
5. Baldwin, C. E., Hayes, D., Jr., & Kopp, B. T. (2023). Severe lung disease in children with cystic fibrosis missed in newborn screening. Pediatric Pulmonology, 58(4), 1046–1053.
6. Cystic Fibrosis Foundation. (n.d.). Sweat test: Clinical care guidelines.
7. Farrell, P. M., White, T. B., Ren, C. L., Hempstead, S. E., Accurso, F., Derichs, N., Howenstine, M., McColley, S. A., Rock, M., Rosenfeld, M., Sermet-Gaudelus, I., Southern, K. W., Marshall, B., & Davis, P. B. (2017). Diagnosis of cystic fibrosis: Consensus guidelines from the Cystic Fibrosis Foundation. The Journal of Pediatrics, 181(Suppl), S4–S15.e1.
8. Currier, R. J., Sciortino, S., Liu, R., Bishop, T., Alikhani Koupaei, R., & Feuchtbaum, L. (2017). Genomic sequencing in cystic fibrosis newborn screening: What works best, two-tier predefined CFTR mutation panels or second-tier CFTR panel followed by third-tier sequencing? Genetics in Medicine, 19(10), 1159–1163.

What is Cystic Fibrosis (CF)?

- Inherited autosomal recessive disease caused by mutations in the CFTR gene, which encodes the CFTR protein responsible for regulating chloride and water transport across cells.
- In CF, dysfunction or absence of the CFTR protein causes accumulation of abnormally thick mucus that affects multiple organ systems.
- While over 1,000 CF-causing CFTR variants have been identified, many non-White patients have variants *not covered* by commonly used genetic testing panels.[1,2]

Detection & Diagnosis for Every Patient



Epidemiology & Demographics

- CF affects patients of all racial and ethnic backgrounds.
- People of color make up a significant and growing proportion of new CF diagnoses.[3,4]
- CF is often underdiagnosed and misdiagnosed in non-White populations.[2]

Newborn Screening

- Most state screening panels do not include CFTR mutations common in newborns of color.[2]
- False-negative screens are more frequent in non-White populations.[2]
- Do not rule out CF in symptomatic patients, even with a negative screen.[5]

Signs & Symptoms

- CF is a multi-system disease that affects more than just the lungs and respiratory system.
- Non-respiratory symptoms are often the first or most prominent signs of CF in people of color.[2,3]

Diagnosis & Testing

- Consider CF in the differential diagnosis for all patients.
- Order a sweat chloride test: ≥ 60 mmol/L confirms CF; < 60 mmol/L warrants further testing if CF is still suspected.[6,7]
- Consider referral to Pulmonology for genetic testing (preferred and only comprehensive method to detect mutations across all racial and ethnic groups).[2,8]