Before, Between & Beyond Pregnancy
The National Preconception Curriculum and Resources Guide for Clinicians

Guidance for Preconception Cystic Fibrosis Carrier Screening

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This guidance should not substitute for clinical judgments or expert consultation

Cystic Fibrosis Carrier Screening Care and Counseling Guidelines:

- Cystic fibrosis (CF) affects approximately 30,000 people in the United States. It is the most common life-shortening autosomal recessive condition in the Caucasian population. It is seen in all racial and ethnic populations, albeit at lower frequencies (See Table 1 Below).
- Classic CF is characterized by progressive lung disease with or without pancreatic insufficiency. Milder related disorders, including congenital bilateral absence of the vas deferens (CBAVD), are caused by mutations in the same gene.
- More than 1600 disease causing mutations have been reported in the cystic fibrosis transmembrane conductance regulator (CFTR) gene.
    - Significant variations in frequency among individuals of varying geographic backgrounds and ethnicities exist (Table 1).
    - Variations can be associated with classic cystic fibrosis or milder CTFR-related disorders.
- ACOG recommends offering CF carrier screening before conception or early in pregnancy when both partners are Caucasian or of European or Ashkenazi Jewish ancestry.
    - It is reasonable to make CF carrier screening available to all couples, regardless of race or ethnicity.
    - The decision to have cystic fibrosis carrier screening should be by informed choice. Appropriate pre- and post-test counseling by a genetic counselor or other appropriately trained healthcare provider is critical.
- Targeted mutation analysis is the most commonly used method of carrier screening
    - ACOG recommends that 23 specific mutations are included on any CF screening panel.
    - Larger panels or gene sequencing may be appropriate for individuals who have cystic fibrosis, a reproductive partner with cystic fibrosis, or CFTR-

related symptoms such as congenital bilateral absence of the vas deferens (CBAVD).
    - For individuals with a family history of cystic fibrosis, targeted analysis of the known familial mutation is recommended. If possible, records documenting the familial mutation should be obtained before proceeding with carrier screening.
- A negative carrier screening result can reduce, but not eliminate, the risk of being a carrier for cystic fibrosis. Residual risks are shown in Table 1 below and should be available to patients.
- Genetic counseling is warranted when both partners are found to carry a CF mutation and can be considered when a single partner is known to be a carrier. Prenatal diagnosis is available if both parental mutations are known.
- While preconception is the ideal time to proceed with CF carrier screening, it may not be covered by all insurance plans. As with any genetic test, it is recommended patients confirm coverage of testing with their individual insurer before proceeding.

Table 1: Cystic Fibrosis Carrier Screening

Race or Ethnicity	Prevalence of CF	Carrier Frequency	Carrier Testing Detection Rate	Carrier Risk After Negative Result
Northern European	1/2500	1/25-1/29	85-90%	1/240
Southern European	1/2500	1/25-1/29	70%	1/80
Ashkenazi Jewish	1/2800	1/26-1/29	97%	1/833
Hispanic	1/8100	1/46	57%	1/113
African American	1/14,500	1/60-1/65	72%	1/198
Asian	1/32,000	1/90	30%	1/128

Preconception Recommendations of CGC Select Panel on Preconception Care Clinical Committee (Solomon BD, Jack BW, Feero G. The clinical content of preconception care: Genetics and genomics.)

- Couples who are at risk for any ethnicity-based conditions should be offered preconception counseling about the risks of that condition to future pregnancies. Screening and/or testing should be offered based on the couples preferences. This may require referral to a genetic counselor or clinical geneticist.
- *Strength of recommendation: B; quality of evidence II-3.*

References:

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