Cystic Fibrosis (CF) is a common inherited disease of children and young adults. The American College of Obstetricians and Gynecologists recognizes that because it is becoming increasingly difficult to assign a single ethnicity, it is reasonable to offer CF screening to all patients.1

Indications for testing include:

- Pregnant and preconception couples
- Individuals with a family history of CF
- Individuals who are negative on a lesser mutation panel when:
  - There is a family history of CF
  - Their partners are positive
- Gamete donors
- Partners of men with Congenital Bilateral Absence of Vas Deferens (CBAVD)

Integrated Genetics also offers:

- Result interpretation provided by board-certified clinical molecular geneticists and access to these genetic professionals to help with any questions
- Full and partial CF sequencing for those patients with a family history of mutations not found on CFplus®
- Rapid 5-8 day turnaround time allowing you to provide your patients with quick answers
- Multiple specimen types to meet your clinic’s and patients’ needs

References:
1) Update on Carrier Screening for Cystic Fibrosis. ACOG Committee Opinion, Number 486, April 2011.
9) Update on Carrier Screening for Cystic Fibrosis. ACOG Committee Opinion, Number 325, December 2005.
CFplus® tests for 97 clinically relevant mutations.

**CFplus® detects more mutations in many ethnic backgrounds.**

- Approximately 1 in 8 carriers overall, and specifically 1 in 4 Hispanic or African American carriers and 1 in 11 Caucasian carriers, would otherwise be missed using the ACMG 23-mutation panel.
- Approximately 1 in 9 carriers overall, and specifically 1 in 5 Hispanic or African American carriers and 1 in 13 Caucasian carriers, would otherwise be missed using a 32-mutation panel.

**CFplus® provides higher detection rates for the pan-ethnic U.S. population.**

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>CF Carrier Risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>Caucasian</td>
<td>1 in 25</td>
</tr>
<tr>
<td>Ashkenazi Jewish</td>
<td>1 in 24</td>
</tr>
<tr>
<td>Hispanic</td>
<td>1 in 58</td>
</tr>
<tr>
<td>African American</td>
<td>1 in 61</td>
</tr>
<tr>
<td>Asian</td>
<td>1 in 94</td>
</tr>
</tbody>
</table>

*CF carrier risk in individuals with no known family history of CF.*

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Integrated Genetics offers over 25 years of experience with CF Testing

The first of its kind molecular genetic testing laboratory.

- Cystic fibrosis transmembrane regulator (CFTR) gene is discovered.
- Testing begins of ∆F508; detection rate (DR) = 72% for Caucasians.
- 4 mutations (added G542X, G551D, and R553X); DR = 77% for Caucasians.
- 12 mutations; DR = 95% for Ashkenazi Jews and 85% for Caucasians.
- 70 mutations; DR = 61% for African Americans. Introduction of new high-throughput technology; improving turnaround time to 7–10 days.
- 86 mutations, additions specific to African Americans, Caucasians and Hispanics; DR = 75% for African Americans.
- D1270N removed after careful review of data shows frequency much higher in carriers than in affected patients, i.e. should be redefined as variant.
- 97 mutations; DR = 78% for Hispanics, turnaround reduced time to 5–8 days.
- ACMG publishes updated Committee Opinion stating "it is reasonable to offer CF carrier screening to all couples regardless of race or ethnicity."
- Cystic Fibrosis Screening in an Ethnically Diverse U.S. Population appears in Clinical Chemistry.

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Integration of CFTR poly T testing added for individuals with CBAVD or pancreatitis, and as a reflex for R117H positive individuals.

- CFTR poly T testing added for individuals with CBAVD or pancreatitis, and as a reflex for R117H positive individuals.
- ACMG/ACOG publish initial guidelines for CF carrier screening. Katherine Klinger was a contributing author.

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**Additional CFplus Detection Rates**

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Detection Rate (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ashkenazi Jewish</td>
<td>97%</td>
</tr>
<tr>
<td>Native American</td>
<td>81%*</td>
</tr>
<tr>
<td>Asian</td>
<td>37–52%**</td>
</tr>
</tbody>
</table>

*Based on 21 self-identified individuals
**Based on 8 self-identified individuals