It is becoming increasingly difficult to assign a single ethnicity to individuals. It is reasonable, therefore, to offer CF carrier screening to all patients.

—ACOG, 2011

The CFTR gene encodes the cystic fibrosis transmembrane conductance regulator protein.

**The CPT code provided is based on AMA guidelines and is for informational purposes only. CPT coding is the sole responsibility of the billing party. Please direct any questions regarding coding to the payer being billed.**
See the evidence: clinically validated Causal variants

CFvantage mutations beyond the 23 common CFTR variants were largely derived from an analysis by Sosnay, et al., of the Clinical and Functional Translation of CFTR (CFTR2) database from the US CF Foundation, as well as from published data.1

- Representing 39,696 genomes of patients diagnosed with CF
- Data gathered from 24 countries

Trust the test: identifies more at-risk couples

Sun, et al., observed that CFvantage identified one additional carrier for every 190 patients tested when compared to the ACMG/ACOG panel.2

- Study compared CFvantage performance in the first series of 11,568 clinical samples tested with how the ACMG/ACOG panel alone would have performed
- Corresponding carrier detection rate (DR) was 1 in 34 for the CFvantage panel and would have been 1 in 42 if limited to the ACMG/ACOG panel
- 61 of the mutations in CFvantage that are not part of the ACMG/ACOG recommended variants were detected at greater frequency than were more than half of the mutations in the guidelines-based variants
- Findings support use of an expanded panel that also accounts for multiple ethnicities

<table>
<thead>
<tr>
<th>Racial or Ethnic Group</th>
<th>Carrier Risk</th>
<th>Detection Rate (%) ACMG-ACOG</th>
<th>Detection Rate (%) CFvantage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ashkenazi Jewish</td>
<td>1/24</td>
<td>94</td>
<td>95</td>
</tr>
<tr>
<td>Non-Hispanic White</td>
<td>1/25</td>
<td>88</td>
<td>90</td>
</tr>
<tr>
<td>Hispanic White</td>
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<td>72</td>
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</tr>
<tr>
<td>African American</td>
<td>1/61</td>
<td>64</td>
<td>78</td>
</tr>
<tr>
<td>Asian American</td>
<td>1/94</td>
<td>49</td>
<td>53</td>
</tr>
</tbody>
</table>

Compared to the ACMG/ACOG panel, the CFvantage Cystic Fibrosis Expanded Screen detects a higher percentage of CF-causing mutations across ethnicities.3-13

1 in 34 DR vs. 1 in 42 DR
= 19% increase in detection
vs. 23-mutation panel
= 1 additional carrier per 190 patients2

References
2. Sun W, et al. Increased identification of CFTR mutations using an expanded panel of validated pathogenic mutations. 65th Annual ASHG Meeting; October 6-10, 2015; Baltimore, MD. Abstract 2012T.