CFvantage®
Expanded Cystic Fibrosis Carrier Screen

Sonora Quest Laboratories™
A Subsidiary of Laboratory Sciences of Arizona

Greater clinical evidence so you can screen with greater confidence

Test Name	Test Code	CPT Code**
CFvantage Cystic Fibrosis Expanded Screen	906672	81220

Trust an expanded panel of clinically relevant mutations for true pan-ethnic screening

A more effective screen for today's multi-ethnic population

More than 1,900 mutations of the CFTR* gene have been identified. A growing number have been confirmed to cause classic cystic fibrosis. However, the 23 common CFTR mutations recommended for screening by ACOG/ACMG were based on Ashkenazi Jewish population studies in Europe and North America. CFvantage adds more validated mutations, including those derived from multinational registeries, for higher detection rate across ethnicities.[1]

CFvantage tests for more clinically relevant mutations

- Detects only CFTR mutations that have been validated to cause cystic fibrosis
- Includes the common CFTR mutations recommended by ACOG/ACMG
- Most mutations are based on a database of nearly 90,000 genomes of well-phenotyped patients, all affected with cystic fibrosis[4]

CFvantage tests for more ethnically relevant mutations

- Adds mutations seen in African-, Hispanic- and Asian-American populations for pan-ethnic screening, as recommended by ACOG[3]
- Informed by multinational registries managed by the US CF Foundation, Johns Hopkins University and The Hospital for Sick Children[4]

> **"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*[3]

*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.

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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

CF detection and carrier rates ACMG/ACOG panel vs. CFvantage[a,b]

Racial or Ethnic Group	Carrier Risk	Detection Rate (%) ACOG	Detection Rate (%) CFvantage
Ashkenazi Jewish	1/24	94	95
Non-Hispanic White	1/25	88	90
Hispanic White	1/58	72	88
African American	1/61	64	78
Asian American	1/94	49	53

Compared to the ACMG/ACOG panel, the CFvantage Cystic Fibrosis Expanded Screen detects a higher percentage of CF-causing mutations across ethnicities.[5–13]

1 in 34 DR vs. 1 in 42 DR
= 19% increase in detection

vs. 23-mutation panel
= 1 additional carrier per 190 patients[2]

a. Detection rates and residual risk estimates are based on a subset of 78 mutations detectable by the panel,[5–13] including the 23 ACMG-ACOG-recommended mutations[6]; exact data are currently unavailable for all mutations in the CFvantage Cystic Fibrosis Expanded Screen.

b. Risks are based on the assumption that there is no family history of CF.

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