

# ELEVATE GENETICS VERIFY

Independent assessment of clinical genetic or genomic test claims through expert review of the test provider’s validation documentation. – For Research Use Only. Not for clinical use.

*The test and lab details in this mock report are fictional yet are modeled after real clinical lab scenarios.*

**NPI #####**

**Test Name: Risk Score for Colon Cancer**

**Laboratory:** CANCER CARE LABS, INC

**Date:** 04 January 2023

## Test Description

A polygenic risk score estimating an 8-year risk for primary colon cancer. It is applicable only for those of Western European descent who have not yet had a primary cancer of any type and who are 60 years or under in age.

CGI’s independent review of validation documentation for *Risk Score for Colon Cancer* concludes that the **one major claim made by Cancer Care Labs, Inc is fully substantiated by the laboratory’s own validation documentation.** (See page 2 for details).

## EVIDENCE

Unsubstantiated

Partially Substantiated

Substantiated

**CLAIM #1 IS SUBSTANTIATED**

### **Claim #1 Made by Test Provider:**

*“For patients of Western European ancestry, this Polygenic Risk Score-based test identifies patients with a high lifetime-risk of developing colon cancer (Hazard Ratio = 13).”*

## METHODOLOGY & RESULT DETAILS

### General Methods

The validation data for the test and claims under investigation were obtained by CGI through contacting the laboratory. CGI scientists were provided with documentation and publications corresponding to the claims assessed on Page 1 of this report. This data was scrutinized for rigorous validation practices, including appropriateness of chosen samples and sample sizes, statistical analyses, and clinical/analytical study design. A summary of ratings provided to each claim are provided below - details can be found in the attached Appendix A.

### Claim 1 Methods and Findings

Three internal validation documents were reviewed in reference to this claim. Three peer-reviewed publications related to the claim were also reviewed (see Appendix). The claim is synthesized from aggregate data from a subset of the internal validation documents and published clinical studies. The samples used for the supporting studies were appropriate for the clinical accuracy claims and came from the appropriate intended use population. Additionally, the combined data from the studies were sufficiently powered to give a reasonable approximation of how the Polygenic Risk Score (PRS)-based test would behave in the clinical setting. Given that there are no concerns with the study design, the stated Hazard Ratio (HR) is consistent with the observed increased risk of developing colon cancer for the high PRS patients in the study data provided. CGI rates this claim as “Fully Substantiated” by the validation documents and clinical studies in support of the claim.

### Disclaimer

Results are for research purposes only. Ratings are based on information provided to CGI by the laboratory test provider. CGI is not responsible for assessments that may be inaccurate due to laboratories withholding documentation, or providing inaccurate or other information that may validate or invalidate the claims addressed in this report. The identification of fraudulent data or studies is not within the scope of ELEVATEGENETICS VERIFY. The assessments presented in this report consist of expert opinions from CGI scientists and/or statisticians, who are not affiliated with the laboratory or any of the laboratory's competitors for a third-party, independent review.

## Appendix A

### Details of the Ratings

[This is a demonstration sample document. However, in a real analysis, details of how CGI arrived at their ratings would be provided here].

SAMPLE