

Validation of patient-facing digital tool for risk stratification of those at risk for hereditary cancer syndrome: The CARE program™ accurately identifies high risk individuals



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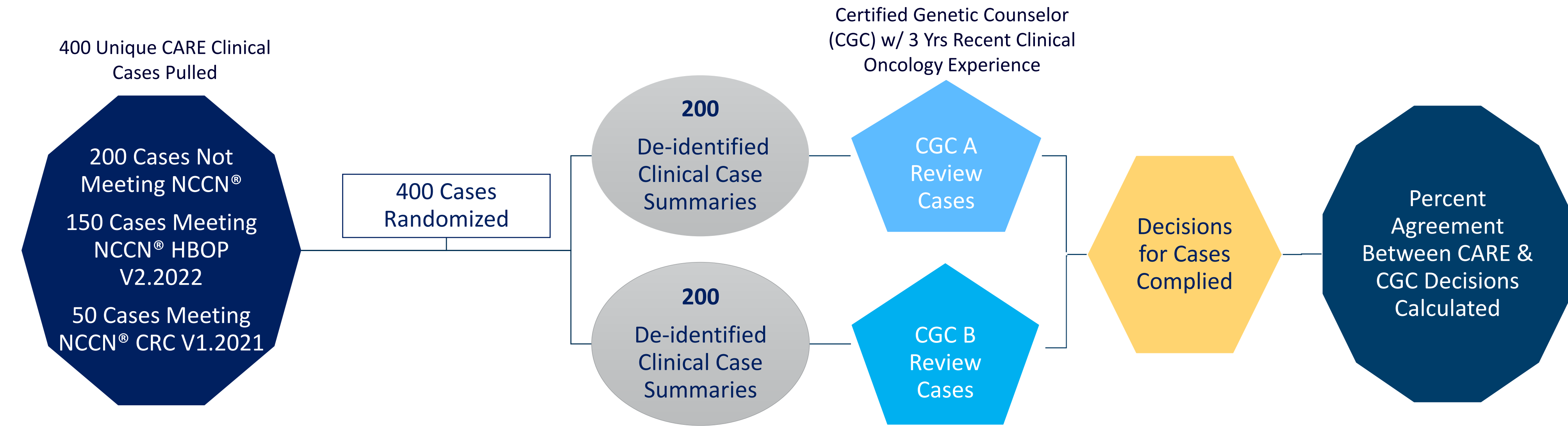
BACKGROUND

- National Comprehensive Cancer Network (NCCN[®]) publishes curated genetic testing criteria based on personal and family cancer history (P/FHx).
- Clinicians may not have a consistent, systematic way to collect and evaluate P/FHx information
- A HIPPA-compliant digital tool (CARE) was developed to collect P/FHx information and risk stratify those who meet NCCN[®] criteria for hereditary breast, ovarian, pancreatic, and prostate cancer (HBOP); and Lynch syndrome and familial adenomatous polyposis

AIMS

To assess the analytic validity for the CARE platform by determining the accuracy of the digital tool's execution and interpretation of NCCN guidelines[®] as compared to certified genetic counselor's (CGC) interpretation of the same guidelines.

METHODS



RESULTS

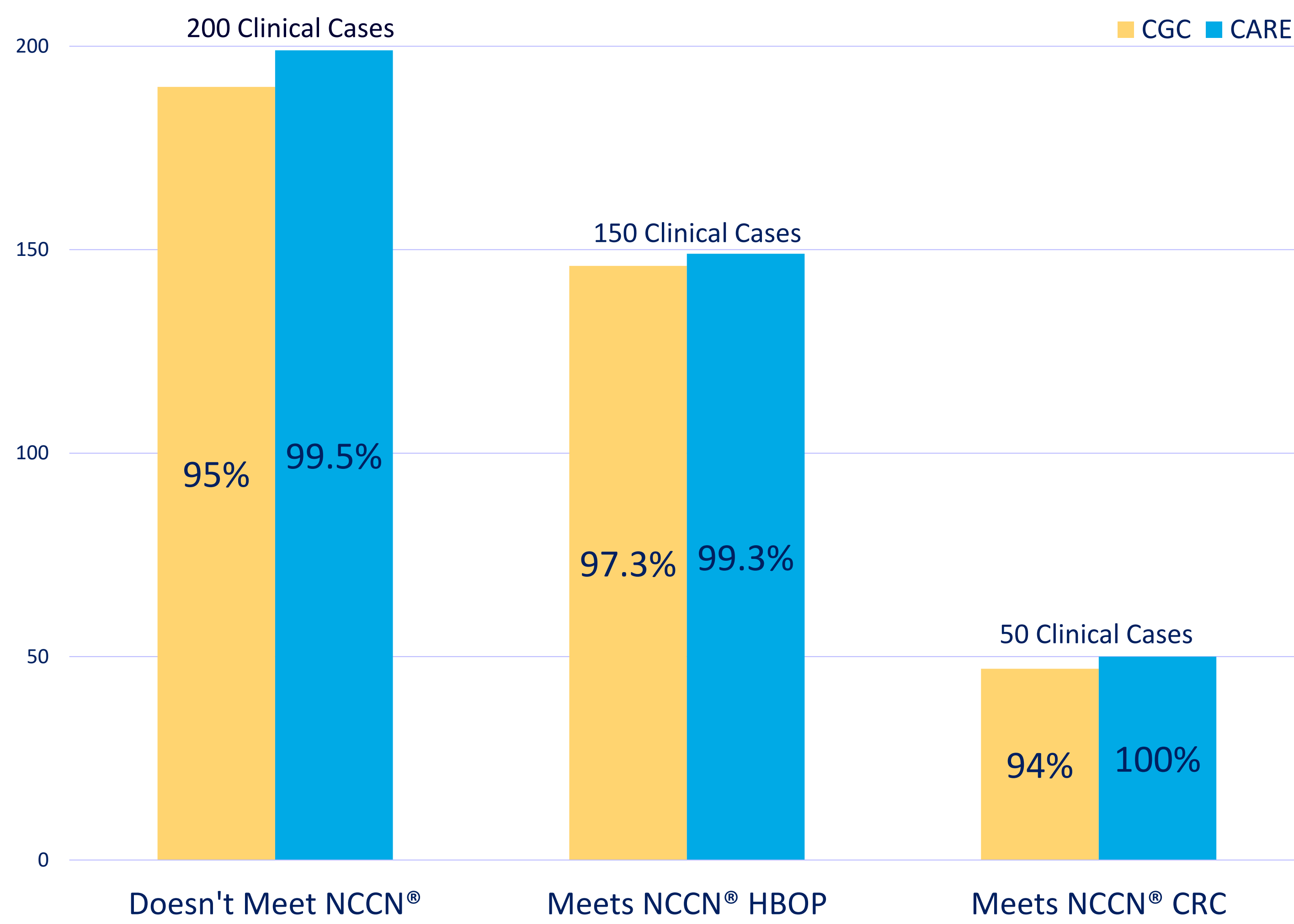
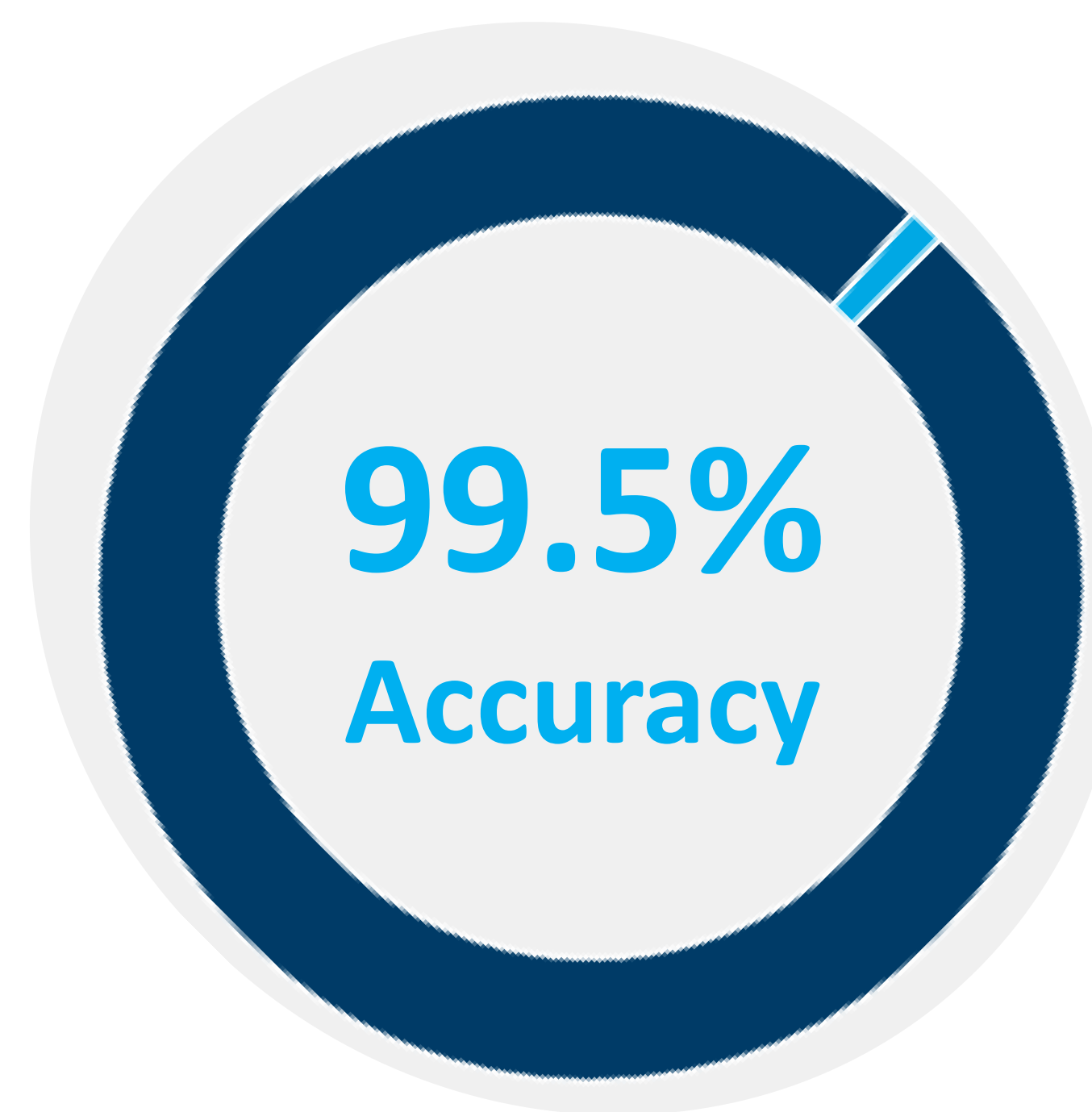


FIGURE 1
Percent of Clinical Cases With Correctly Interpreted NCCN[®] Genetic Testing Criteria by CARE Platform and Certified Genetic Counselors



The CARE program's analytic validity was 99.5% (398/400) for identifying individuals who met NCCN[®] genetic testing criteria*

*Within the parameters of the CARE risk model

TAKE HOME POINTS

The CARE program™ accurately identifies individuals who meet NCCN[®] testing criteria to aid in risk stratification.

Digital tools may be helpful:

- Collecting personal/family history
- Identify individuals for genetic counseling/testing

CASE EXAMPLES OF DISCORDANCE

Example 1: Example of family history where CGC ruled proband didn't meet NCCN[®] testing criteria (but would clinically offer the genetic testing)
CARE Platform: Does meet NCCN[®] HBOP testing criteria; Family hx of breast cancer diagnosed ≥51y in a second-degree relative and ≥1 close relative with breast cancer dx ≤50y on the same side of the family
CGC Reviewer: Does not meet NCCN[®] testing criteria

Example 2: Example of family history where CGC ruled proband didn't meet NCCN[®] testing criteria (but would clinically offer the genetic testing)
CARE Platform: Does meet NCCN[®] CRC testing criteria; Family hx of ≥1 first-degree relative with colorectal and another synchronous LS-related cancer at any age.
CGC Reviewer: Does not meet NCCN[®] testing criteria

Example 3: Example of family histories where CGC ruled proband didn't meet NCCN[®] testing criteria (but would clinically offer the genetic testing)
CARE Platform: Does meet NCCN[®] HBOP testing criteria; Family hx of breast cancer dx at ≥51y in a second-degree relative and ≥2 close relatives with prostate cancer at any age on the same side of the family
CGC Reviewer: Does not meet NCCN[®] testing criteria

FIGURE 2
Examples of CARE Platform Correctly Identifying Individuals Who Meet NCCN[®] Genetic Testing Criteria

Example 1: Example of personal history where CGC used clinical judgement to offer genetic testing when individual had reported 10-19 colon polyps of unknown pathology
CARE Platform: Does not meet NCCN[®] testing criteria; CARE does not assume adenomatous colon polyp unless specifically chosen
CGC Reviewer: Does meet NCCN[®] CRC testing criteria; Personal hx of ≥10 adenomatous colon polyps

Example 2: Example of family history where CGC felt the first-degree relative's cancer history meets NCCN[®] testing criteria
CARE Platform: Does not meet NCCN[®] testing criteria; CARE flags first-degree relative with LS-related cancer <50y with another first-degree or second-degree relative with LS-related cancer
CGC Reviewer: Does meet NCCN[®] CRC testing criteria; Father with gastric ca <50, brain tumor in paternal cousin

Example 3: Example of family history where CGC felt the family history of cancer meets NCCN[®] testing criteria
CARE Platform: Does not meet NCCN[®] testing criteria; CARE only considers first-degree relatives with personal hx of pancreatic cancer
CGC Reviewer: Does meet NCCN[®] HBOP testing criteria; Paternal Uncle/paternal grandmother with pancreatic at any age

FIGURE 3
Examples of CARE Platform Correctly Identifying Individuals Who Do Not Meet NCCN[®] Genetic Testing Criteria

Example 1: Example family history where CGC correctly classified proband as meeting NCCN[®] HBOP
CARE Platform: Does not meet NCCN[®] testing criteria; CARE does not assess "limited family hx"
CGC Reviewer: Does meet NCCN[®] HBOP testing criteria; Mother <50 with limited family hx

Example 2: Example family history where CGC correctly classified proband as not meeting NCCN[®] HBOP
CARE Platform: Does meet NCCN[®] HBOP testing criteria; CARE assumed stomach cancer was diffuse type
CGC Reviewer: Does not meet NCCN[®] testing criteria

FIGURE 4
Examples of CARE Platform Incorrectly Identifying Individuals Who Meet/Don't Meet NCCN[®] Genetic Testing Criteria