Validation of a guidelines-based digital tool to assess the need for cancer genetic testing

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1. GeneMatters. 2. Strata Oncology

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Introduction

- Most people with a hereditary predisposition to cancer are unaware of their increased risk.
- New scalable, efficient, and reliable approaches are needed to identify these patients.
- The RISE Hereditary Cancer Risk Assessment Tool is a patient-administered digital tool that assesses the need for cancer genetic testing, based on criteria included in NCCN Guidelines®.
- To ensure reliability of the tool, clinical validation is needed.

Aim

To assess the clinical validity of the tool by comparing its assessment of need for genetic testing to the same assessment made by a cancer genetic counselor (GC).

Methods

- Pre-test oncology cases from our telehealth genetic counseling organization were used for validation.
- History was extracted via chart review, then entered into the tool by study staff.
- The tool's assessment was compared to the assessment made by the GC who saw the patient.
- Patients who met criteria (n=100) were selected based on how they met criteria, in order to validate the rules that are most frequently invoked.
- Consecutive cases were used in selecting patients who didn't meet criteria (n=47).

Results

Figure 1: Characteristics of patients

- 48.1 years (standard deviation 15.3)

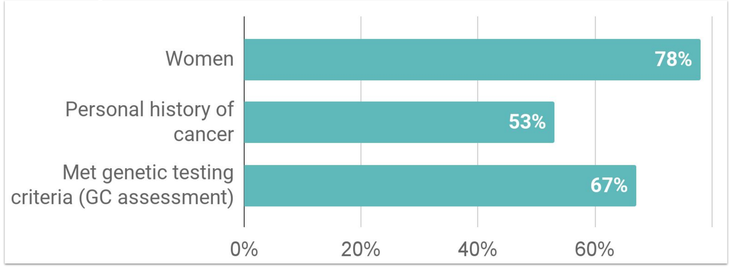


Figure 2: Personal & family history of cancer

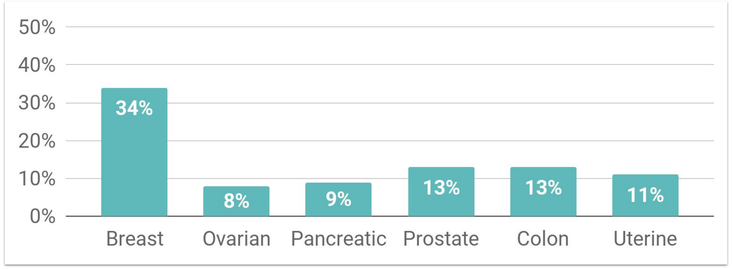
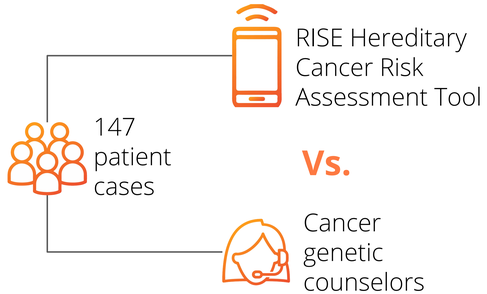


Figure 3: High level of agreement



97% agreement
on meeting genetic testing criteria

Results

- 3% disagreement between tool & GC was due to genetic testing criteria intentionally left out of tool because patients likely can't answer the necessary history questions. Specifically, MSI/IHC (3 cases) and increased model-predicted risk for Lynch (1 case).

3.0 minutes
Mean time to fill out the tool.
(Standard deviation 1.0 minutes)

Discussion

- High degree of agreement between tool and GC suggests tool is a clinically valid method of identifying individuals in need of cancer genetic testing.
- The tool takes little time to complete, which is key to scalability and feasibility.
- Patient-administered tools like this are limited by the history questions patients can answer.
- Further validation with a larger prospective cohort of consecutive cases would help to confirm these findings and to also estimate sensitivity and specificity.

Limitations

- Patients may respond to history questions in the tool differently than study staff did when entering history based on chart review. As a result the level of agreement may differ when patients use the tool.