BACKGROUND

- Scalable and reliable approaches are needed to assist non-genetics clinicians in assessing patients' need for germline genetic testing
- Commercial insurance coverage of genetic testing often depends on this assessment
- A brief patient-administered hereditary cancer risk assessment digital tool was designed to assess the need for germline genetic testing based on history and national guidelines

AIM

To assess the clinical validity of this digital tool by evaluating its agreement with assessments made by genetic counselors

METHODS

Retrospective chart review of patients with a personal and/or family history of cancer from a telehealth genetic counseling practice.

Extracted history entered into the digital tool to model how a patient would complete the tool

Compared the assessments of whether the patient met criteria for germline genetic testing made by the tool and the genetic counselor who saw the patient

Further validation was performed on a subset of cases by analyzing the agreement between the tool and the research team on which specific history-based rules in the tool's algorithm applied for a given case

A digital tool and genetic counselors have high agreement (96%) on assessing need for germline genetic testing.

Hereditary Cancer Risk Assessment Tool

152 patient cases

VS.

Genetic Counselors

96% agreement

on meeting genetic testing criteria

RESULTS

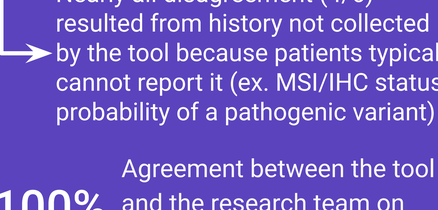
PATIENT CHARACTERISTICS

152 Patients with a personal and/or family history of cancer seen from July to October 2020

81% female

52.3 Mean Age

56% Personal history of cancer



66% 100/152 cases met criteria for genetic testing*

*Cases meeting criteria were sampled to ensure the dataset included the criteria most commonly met in clinical practice.

VALIDATION

Completion of the tool took a mean of 3.3 minutes

96% Agreement between the digital tool and genetic counselors (146/152 cases)

Nearly all disagreement (4/6) resulted from history not collected by the tool because patients typically cannot report it (ex. MSI/IHC status, probability of a pathogenic variant)

100% Agreement between the tool and the research team on which aspects of the patient's history met criteria in subset of cases (62/62)

Validation of a guidelines-based brief patient-administered digital tool to assess the need for germline genetic testing

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