Comparison and concordance of reported cancer family history between an automated patient assessment tool and genetic counselor recorded pedigrees: findings from a pilot study

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Introduction

Methods to collect family history and recommend genetic counseling and/or testing for hereditary cancer syndromes vary. Counsyl developed an automated patient assessment (APA) tool to collect family history in 5-10 minutes, align such history to National Comprehensive Cancer Network (NCCN) guidelines, and offer education on hereditary cancer syndromes.

Studies show family history collection methods are used interchangeably and accurately, yet few include APA tools. We analyzed concordance of family history between APA and 3-generation genetic counselor recorded pedigrees ("pedigrees") in a pilot study conducted at a large health system.

Methods

- Counsyl and Intermountain Healthcare (IH) piloted Counsyl's APA for 30 patients across hereditary cancer indications.
- Consented patients received unique APA links by text/email prior to their IH genetic counseling sessions.
- Information entered into the APA generated an individualized report that included all family history reported.
- APA reports and IH genetic counselor-recorded pedigrees were compared for concordance across enrolled patients.

Concordance data points

- Reported cancer type and age at diagnosis by decade were abstracted from APA reports and pedigrees by each degree of relation [first-, second- and third-degree relatives] (FDR, SDR, TDR).
- Specific data points were assessed by degree of relation to determine concordance between tools (Table 1).

Table 1. Concordance data point definitions

Data Point	Definition
Pedigree Affected	Number of total relatives affected reported on GC pedigree, recorded by degree
Cancer Type Reported by APA	Number of total relatives affected reported from APA, recorded by degree
Cancer Type NOT Reported by APA	Number of affected relatives NOT reported in APA but recorded on GC pedigree, recorded by degree
Correct Type on APA	Number of cancer types that match between APA and GC Pedigree
Incorrect Type on APA	Number of cancer types that do not match between APA and GC Pedigree
Similar Ages within 1 Decade	Ages of diagnoses within the same decade (40s, 50s, 60s) when analyzing cancers reported on both APA and GC pedigrees

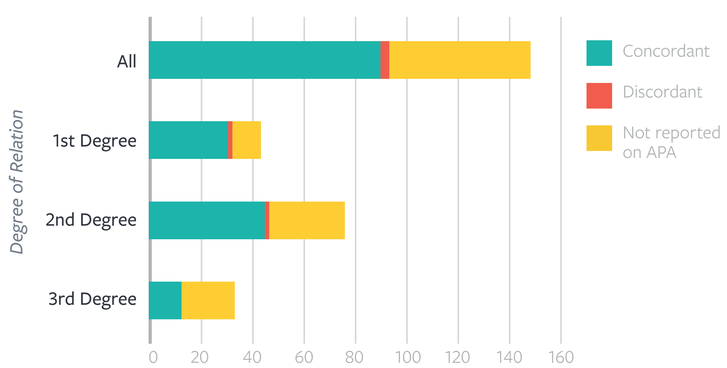
Results

Concordance of reporting across tools

29/30 probands had family history recorded on both APA and pedigrees with reported histories of 149 cancers from 143 cases. 97% of probands reported Caucasian ethnicity and 79% were female.

Over 60% of cases (92/143) and cancers (93/149) were reported first through APA. When cancers were reported on both APA and pedigrees, 96% of cancers were reported concordantly (89/93, Figure 1). Similar trends were seen for age of diagnosis reporting for cases, with 93% (86/92) reported concordantly on both tools.

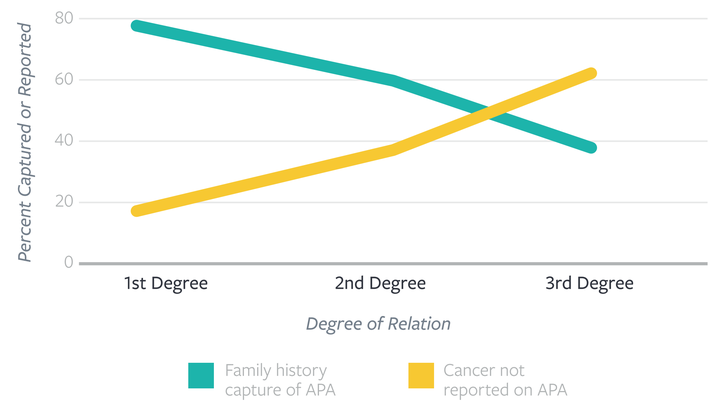
Figure 1. Comparison of APA-reported cancers to pedigree-reported cancers



Degree of relation effect

The rate at which cancers were reported first on APA were impacted by degree of relation (Figure 2, yellow). Degree of relation also impacted the "Family history capture of APA" (teal), where APA cancer concordance was evaluated relative to GC pedigree histories (including cancers not reported). Updates to Counsyl's APA have been made to highlight the importance of cancer history in distant relatives to increase frequency and accuracy of reporting.

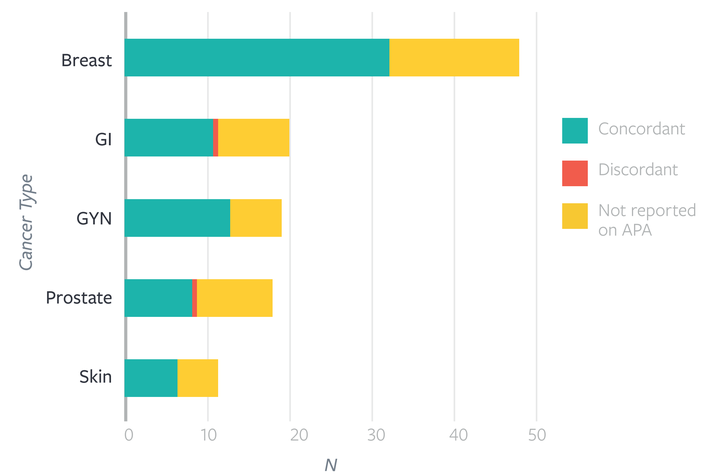
Figure 2. Degree of relation impact on rates of family history capture and frequency of cancer reporting



Trends by cancer type

Common cancers (n>10) were analyzed by subgroup to determine trends in reporting and concordance between pedigrees and APA. When cancers were reported on both tools, favorable concordance rates were seen (Figure 3).

Figure 3. Distribution of reported cancers on APA and pedigrees



Notable trends

- Prostate cancer had the highest rates of cancers not reported on APA; breast and GYN cancers had the lowest.
- 90% (40/44) of common cancers not reported on APA yet found on pedigrees were in second- or third-degree relatives.
- Discordant cases (n=4) included incorrect APA reports of 'Ureter' and 'Other Cancer NOS' in first-degree relatives and incorrect APA reports for Stomach and Prostate cancers in second-degree relatives. There were no TDR cancers reported incorrectly on APA when compared to pedigrees.

Conclusions

In this pilot study, family history reported first through Counsyl's APA is highly concordant for cancer type and age of diagnosis when compared to genetic counselor recorded pedigrees. APA may be able to complement family history collection, as families may come to clinic ready to discuss their family history (per IH genetic counselor experience).

Degree of relation impacts APA reporting rate and accuracy of cancers reported when compared to pedigrees. Education of patients and adjustments to APA tools, such as help text on the value of extended family history, may improve family reporting and accuracy. (Note: Such updates have been implemented in Counsyl's APA as a result of this pilot study.)

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