Title: Use of a patient-facing digital platform to aid in genetic test result delivery and connection to genetic counseling services

Category I,

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Background

Digital tools have been proposed as a method to augment traditional genetic counseling and reduce burden on healthcare professionals for population hereditary cancer risk screening and testing. The Ambry CARE Program™ (CARE) is a patient-facing digital risk stratification tool (DRST) to collect and analyze personal and family history, identify individuals who meet national guidelines for genetic testing, and aid in pre-test and post-test patient care and management. Healthcare sites that use this tool can tailor the extent to which return of results is performed through traditional communication with the clinician versus our DRST. Here we review the use of this DRST to handle the return of genetic test results, and track patient attendance of telegenetic counseling

Methods

This is a retrospective observational study of all female patients from August 2018 through October 2023 who used the DRST to assess their eligibility for hereditary cancer genetic testing, who opted to proceed with genetic testing, and who received return of genetic testing results through the DRST. For this study, 'return of results' consists of two components: results disclosure via the CARE platform and post-test telegenetic counseling. Patients who had both results disclosure and counseling executed without support from the DRST were excluded from the study. Clinics can choose to have patients undergo both disclosure and coordination of counseling performed by the tool or to have disclosure performed by the tool and counseling coordinated by their health care provider. The exception is for positive results, in which the DRST directs the patient to their provider for disclosure and counseling. The patients were grouped by outcomes of genetic testing: negative, variant of uncertain significance (VUS), and positive. Outcome measures included percent of individuals who completed results disclosure and education by the DRST for negative, VUS, and positive results; the number of individuals who had post-test telegenetic counseling with a certified genetic counselor (CGC) coordinated by the DRST; and the number of individuals who completed telegenetic counseling when coordinated by the DRST.

Results

A total of 42,436 females aged 18 years or older had an appointment scheduled at a healthcare site with CARE implemented to aid in the return of genetic test results. Of the patients who had negative genetic test results, 78.93% (22,967/29,095) completed results disclosure and education performed by the DRST. Among 511 negative patients that had telegenetic counseling coordinated by the DRST, 74.36% (380) scheduled and completed their counseling appointment. For patients who received a VUS genetic test result, 79.78% (8,151/10,217) completed results disclosure and education via the DRST, and 77.15% (439/569) who had telegenetic counseling coordinated by DRST scheduled and completed their

counseling appointment. Of the individuals with positive genetic test results (2,617) that had the option for telegenetic results disclosure performed by a CGC, 80.17% (2,098) completed their counseling appointment.

Conclusion

In this study, CARE provided a standardized workflow that enabled healthcare providers to deliver hereditary cancer genetic testing results and offer access to a CGC. Our results indicate that participation in post-test telegenetic counseling following results disclosure and coordination by the DRST was high. Digital interventions may be used to deliver information about genetic concepts and results, and positively impact service engagement.