

Title: Time to testing for hereditary cancer: a comparison of age at cancer diagnosis and genetic testing

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Introduction:

Hereditary cancer genetic testing referrals can be influenced by clinical experience, expert guidelines, access to care, insurance coverage, and other factors. Despite existing guidelines, many patients who meet criteria experience delays in testing, which can lead to missed therapeutic options or opportunities for early intervention. For example, studies have shown that less than 50% of patients with ovarian cancer complete genetic testing despite multiple expert groups recommending testing for all epithelial ovarian cancers. This study aims to evaluate testing delays by comparing age at prior cancer diagnosis and age at testing across diverse cancer types and ethnicities.

Methods:

Select individuals with a personal history of breast, colorectal, endometrial, ovarian, pancreatic, prostate, or renal cancer who underwent pan-cancer multigene panel testing (49 to 77 genes) at a single genetic testing laboratory between 2016-2024 were included. Cases with unknown ethnicity or age at diagnosis were excluded from the study group. Age at diagnosis and age at testing were compared between cancer types as well as by self-reported ethnicity and age within each cancer type. Comparisons by ethnicity were limited to subgroups of ≥ 5 individuals.

Results;

Preliminary results showed an overall difference between average age at testing and cancer diagnosis of 3.2 years for females and 2.4 years for males, with breast and colorectal cancer accounting for most of the sex-specific variability. The overall difference was 4.0 years for ovarian cancer (n=248; range 3-5 years), 3.9 years for kidney cancer (n=238; range 3-6 years), 3.2 years for breast cancer (n=4403; range 2-5 years), 3.1 years for endometrial cancer (n=261; range 2-4 years), 3.0 years for prostate cancer (n=559; range 1-4 years), 2.5 years for colorectal cancer (n=583; range 2-4 years), and 0.8 years for pancreatic cancer (n=317; range 1-3 years). Across the individual cancer types, the difference between age at diagnosis and age at testing was similar between most ethnicities and differed by ≤ 1 year. For most cancers, the difference between age at diagnosis and testing increased by patient age, with individuals in their 20s and 30s receiving testing within the first year of diagnosis.

However, the greatest difference for pancreatic cancer was between ages 30-39 (3.6 years) and between ages 40-49 for ovarian cancer (7.6 years).

Discussion

While the difference in average age at cancer diagnosis and genetic testing did not appear to differ by reported ethnicity, it did vary among cancer types and between sexes. Of potential concern is the fact that the greatest lag in genetic testing was for ovarian cancer, as germline results may impact treatment for these cancers.