

# Proactive Genetic Screening as an Employee Benefit: Insights into Actionable Conditions

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**Background:** Routine health screenings are a critical component of early detection and disease prevention. Clinical factors, such as age, sex, and lifestyle behaviors influence risk, and have been incorporated into prevention strategies. There is growing recognition that genetic factors also greatly influence risk for cancer, cardiovascular disease and other important conditions, and that preventive strategies and early detection may be improved by incorporation of genetic screening. However, genetic screening is widely underutilized in healthy populations<sup>1</sup>. To address the health of an employee population, a preventive-focused health benefit program has been implemented combining access to genetic screening, genetic counseling, navigation and education. Navigation includes awareness and review of genetic screening results, access to education or genetic counseling as appropriate and personalized follow-up guidance offered by experienced professionals trained in diverse fields such as nursing, oncology and genetics. As a result of this enhanced model, we are assessing how the elements of this preventive program can lead to better patient outcomes through access to genetic counseling and tailored navigation.

**Methods:** Retrospective, deidentified data for a 30-month period (1/2023 - 6/2025) was evaluated under an IRB-approved protocol. Results from genetic testing were grouped by result type for descriptive statistics. Analysis included evaluating by risk level (elevated, high), as defined in a large physician-directed genetic screening study<sup>2</sup>. Additionally, results were assessed by clinical area (cancer, cardiovascular and bleeding disorders, and other actionable disorders) with percentage of findings grouped by category. Individuals identified to have increased risk for a heritable condition received educational outreach from a healthcare navigator and were offered genetic counseling, as appropriate. The navigator educates the individual on healthy lifestyle habits, appropriate screenings, and surveillance measures to empower informed decision-making about health and wellbeing. A navigator facilitates next health actions, such as sharing results with local healthcare provider teams. Access to education and/or genetic counseling regarding genetic testing results were assessed between a self-directed model (2023-2024) and a navigator-directed model (2025). A self-directed model is defined as an individual scheduling their own genetic counseling session regardless of result type. A navigator-directed model is defined as an individual receiving outreach with access to education and/or genetic

counseling based on result type. Other result types (carrier, negative) received digital education or access to a navigator, if desired.

**Results:** 8,534 individuals with this employee benefit requested a genetic screening test and received results. Of these individuals, 59.3% self-identified as female with an average age of 44.9 (range 21-79). Results for the screening panel, which evaluated more than 160 genes related to cancer, cardiovascular disease and other medically actionable disorders, showed a positive rate for a pathogenic, likely pathogenic or increased risk allele variant of 1599/8534 (18.7%), a carrier finding in 3473/8534 (40.7%) or a negative result 33462/8534 (40.6%). Of the positive results, 484 (5.7%) were associated with cancer, 886 (10.3%) were associated with a cardiovascular condition or bleeding disorders, and 229 (2.7%) were associated with an additional actionable disorder. More than one positive finding was identified in 178/1599 individuals (11.1%). Across all positive results, engagement for education/genetic counseling and follow-up increased from 61% to 76% following implementation of tailored navigation.

**Conclusions:** This model utilizes referrals to genetic counseling to provide comprehensive post-test follow-up, when appropriate, based on high-risk results. Navigators can provide test follow-up and education to those with elevated risks. The success of this approach can be seen in the increased uptake of result follow-up (education/genetic counseling) from self-directed to navigator-directed models. Combining access to genetic screening, education/genetic counseling and tailored navigation ensures an individual can make informed decisions that could impact their future health outcomes. Tailored navigation provides personalized guidance that can increase adherence to guideline-recommended care. Further data collection is necessary to assess long-term adherence to recommended screenings and outcomes.

#### References:

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