

Poster 14: Identification of hereditary cancer risk in a population-based genetic testing program**Presenting Author:** Christine Walsh, MD, University of Colorado - Anschutz Medical Campus

Topic

Genetics

Objectives

Most hereditary cancer risk carriers are unaware until after a cancer diagnosis, representing a missed opportunity for cancer prevention. Individuals with Jewish ancestry are estimated to carry a 2.5% risk for carrying a BRCA mutation, a ten-fold higher rate compared to the general population. Our objective was to provide education and genetic testing to individuals with Jewish ancestry and to describe the pathogenic variant (PV) rate in a population-based genetic testing initiative.

Methods

We launched an IRB-approved University/Community partnership in our state to provide education and access to fully reimbursed, low-barrier germline genetic testing to individuals at least 25 years of age with at least 1 grandparent of Jewish ancestry. After signing informed consent, individuals underwent multi-gene panel testing and completed on-line surveys collecting information about baseline demographics, personal and family cancer history, and satisfaction with a fully virtual genetic testing process. The Multidimensional Impact of Cancer Risk Assessment (MICRA) tool was utilized to measure the psychological impact of genetic testing (score range 0-95 with higher scores demonstrating greater distress, greater uncertainty, more negative experience).

Results

Among 130 individuals who have thus-far completed the study processes, the median age was 58 (range 25-96), the majority were female (n=106, 81.5%) and the majority had 4 Jewish grandparents (n=106, 81.5%). There were 4 individuals (3.1%) with BRCA1/BRCA2 Jewish founder mutations including 1 with a second PV in CHEK2. PVs of other genes were identified in 10 (7.7%) individuals, including 9 in APC and 1 in NTHL1. Among the 4 participants with Jewish founder mutations, 3 (75%) had no personal or family history of cancer in a first degree relative. MICRA scores ranged from 0 to 54 with a median score of 8.

Conclusions

A population-based genetic testing initiative in Jewish individuals is feasible and is associated with low levels of negative psychological impact. We identified a greater than expected number of individuals with pathogenic variants in cancer-predisposing genes and the majority with BRCA1/BRCA2 Jewish founder mutations had no personal or family history of cancer.