Genetic Insights into Hereditary Cancer Risk in the Global Population

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Introduction

Current hereditary cancer risk data is mostly based on genetic testing performed in Caucasian and Ashkenazi Jewish populations. As a result, the distribution of mutated genes and their associated cancer risk in other ethnicities is not well understood. Asian, Hispanic, and African populations are significantly under-represented in studies and databases of hereditary cancer mutations, despite the clear value this information can provide to populations around the globe. Interestingly, in some countries, more than 1 in 4 breast cancer patients were reported to have a BRCA1/2 mutation, indicating that the proportion of hereditary breast cancers in other populations is even higher than previously reported. Current hereditary cancer risk data is mostly based on genetic testing performed in Caucasian and Ashkenazi Jewish populations. Unlike the proportion of breast cancer patients with a BRCA1/2 mutation in the USA (8.3%), the proportion of breast cancer patients in this cohort was 9.9%, which is similar to previously reported rates in Caucasian and Ashkenazi Jewish populations. The 30-gene panel assesses the following genes associated with hereditary breast, ovarian, colorectal, melanoma, hereditary cancer test to assess their risk for hereditary cancer.

Methods

We describe the demographics and genetic results of 7952 international hereditary cancer high risk individuals from 17 countries across multiple continents for breast cancer as well as ovarian, colorectal, melanoma, pancreatic, prostate, uterine and stomach cancers.

Results

Conclusions

• Here, we present the results of testing high-risk individuals from 17 countries with a 30-gene panel for hereditary cancer risk and a 19-gene panel for breast and ovarian cancer risk. The overall pathogenic mutation rate was 15.6%.

• Pathogenic variants were identified in 26 different genes on the panel, with proportions varying widely by country, highlighting the utility of broader panel testing in global populations.

• Nearly half of this high-risk cohort reported a personal history of breast cancer. The BRCA1 and BRCA2 mutation carrier rate for breast cancer patients in this cohort was 9.9%, which is similar to previously reported rates in Caucasian and Ashkenazi Jewish populations.

References

Barycki et al. (2017), Bentrem et al. (2015), Gurusamy et al. (2015), King et al. (2015), and Fall et al. (2020).