Hereditary breast and ovarian cancer syndrome (HBOC)

Hereditary breast and ovarian cancer (HBOC) is a genetic syndrome that increases the chances to develop certain types of cancer, including breast (female and male), ovarian, prostate, and pancreatic cancers, as well as melanoma in some families. Individuals with HBOC can also be diagnosed with cancer at younger ages than would normally be expected, with some people being diagnosed with cancer in their 20s.

Prostate cancer in men is usually not a fast-growing or rapidly spreading type of cancer, and many men with prostate cancer may not have to undergo extensive treatment. However, prostate cancer in men with HBOC is more likely to occur earlier than it does in men in the general population, and is more likely to metastasize (spread throughout the body) and pose a more significant threat to one’s health.

Approximately 1 in 500 to 1 in 1000 people in the general population have HBOC. It is estimated that HBOC contributes to approximately 5-10% of all breast cancers and 10-15% of all ovarian cancers. Approximately 6% of men with prostate cancer will have HBOC. HBOC is more common in some ethnic backgrounds, particularly the Ashkenazi Jewish population (where approximately 1 in 40 people of Ashkenazi Jewish ancestry are thought to have HBOC). The number of individuals who have HBOC may be higher than this because many people who have HBOC will never go on to develop cancer.

**Causes**

HBOC is caused by harmful changes (called pathogenic variants) in the \textit{BRCA1} and \textit{BRCA2} genes.

**Diagnosing HBOC**

A definitive diagnosis of HBOC can only be made by performing genetic testing to look for pathogenic variants in the \textit{BRCA1} and \textit{BRCA2} genes. The National Comprehensive Cancer Network (NCCN) is a group of medical professionals that regularly meet to look over any updates in research studies and determine recommendations for who should be considered at a higher risk for HBOC. Some patterns or red flags in a personal or family history that make it more likely that a family could have HBOC, including:

- A family history of ovarian cancer
- A family history of metastatic (cancer that has spread to other parts of the body) prostate cancer
- A family history of breast cancer diagnosed before age 50
- A family history of triple-negative (a specific type) breast cancer diagnosed before age
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- A family history of three or more relatives (on the same side of the family), that have had breast, pancreatic, or prostate cancer
- Ashkenazi Jewish ancestry and a family history of breast, ovarian, pancreatic, or prostate cancer

If you or your family have a history of any of these types of cancer, it is recommended that you meet with a specialist, such as a genetic counselor, who can gather more information to help determine if you have a higher chance to have HBOC. It is important to note that not all people who have the above types of cancer in their family will have HBOC.

Medical Management for HBOC

If found to have HBOC, it is recommended to discuss your management plan with your healthcare team, and if available, to seek consultation through a specialized high-risk clinic. General recommendations are included here based on the updated guidelines of the NCCN, but may be tailored to your specific medical and family history.

Breast cancer screening for women with HBOC includes:

- Beginning at age 18: Breast self exam to facilitate awareness and familiarity with breast tissue
- Ages 25-29: Clinical breast exam every 6-12 months, and breast MRI every 12 months
- Ages 30-75: Clinical breast exam every 6-12 months, breast MRI every 12 months, and mammogram every 12 months
- Age 75+: Individualized management

Bilateral prophylactic (preventive) mastectomies or preventive medications are also available options for breast cancer risk reduction, and are a personal decision.

Women at increased risk for ovarian cancer due to HBOC are encouraged to consider prophylactic (preventative) removal of the ovaries and fallopian tubes (called a salpingo-oophorectomy) between age 35-40, or once they are finished having children. For women who do not elect preventive surgery, current ovarian cancer screening includes CA-125 blood levels and transvaginal ultrasound beginning at age 30-35, however this screening is not proven to be effective and is not recommended by many medical organizations.

It is recommended that men with HBOC talk with their doctor about screening options for prostate cancer at the age of 40. Currently, the most effective way to screen for prostate
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cancer is by doing a blood test to look for something called the prostate-specific antigen (PSA), which is a blood test that can be done yearly. It is important for men with HBOC to have an in-depth conversation with their medical provider about the risks, benefits, and limitations for PSA screening because there is the possibility for false positives (positive PSA result when someone doesn’t have prostate cancer) and false negatives (negative PSA result when someone actually has prostate cancer). Because of the possibility of false positives and negatives, some men may opt not to pursue PSA screening.

Additional Resources:

Face Our Risk of Cancer Empowered (FORCE)

Bright Pink

Sharsheret

National Ovarian Cancer Coalition (NOCC)