Hereditary Breast and Ovarian Cancer

Genetics:

Hereditary cancer is typically suspected in families when several members of more than one generation develop cancer, and these cancers are diagnosed at a relatively young age (less than 50). Hereditary cancer is also suspected if a particular type of cancer, or a recognizable pattern of cancers, cluster in a family. About 5-10% of all cancer cases are thought to be due to an inherited gene mutation. Therefore, most cancer is not hereditary.

Hereditary breast and ovarian cancer syndromes are associated with mutations in one of two genes, BRCA1 and BRCA2. Approximately 20-30% of families that are suggestive of a hereditary breast and ovarian cancer syndrome will test positive for a mutation in BRCA1 or BRCA2. However, there may be other genes that predispose to breast cancer.

***Inheritance of BRCA1 or BRCA2 Mutations**:*

We have two copies of every gene in our body, one from our mother and the other from our father. Having one non-working copy of a BRCA1 or BRCA2 gene is enough to increase one's predisposition to cancer. When an individual (male or female) has a BRCA1 or BRCA2 mutation, he/she has a 50% chance of passing it on to each child. This is known as autosomal dominant inheritance.

Genetic Testing:

If your family and/or personal history of cancer suggests that an inherited mutation may be involved, the possibility of genetic testing will be discussed during your genetic counseling appointment. Due to the complexities of genetic testing, it is most useful to begin testing on a family member who has been diagnosed with breast or ovarian cancer. Genetic test results can take many months to obtain. There are three possible results from initial testing for a BRCA1 or BRCA2 mutation:

Mutation Detected: A gene change (mutation) known to increase cancer risk has been identified. This likely explains the family history of cancer. Genetic testing for this mutation would be available to other family members.

No Mutation Detected: A gene change in BRCA1 or BRCA2 has not been identified. This does not rule out a hereditary cancer predisposition. It is still possible that the individual carries a mutation that the testing cannot detect or a mutation in another, as yet undiscovered, cancer predisposition gene. Screening and management decisions should be based on family history and other personal risk factors.

Uncertain Variant: A gene change that has not been well described. Additional studies would be required to determine whether this gene change is part of normal gene variation or is a mutation which increases cancer risk.

Screening:

All women can consider monthly breast self-exams and have clinical breast exams annually by their physician. Annual mammograms beginning at age 40, or 10 years before the earliest diagnosis of breast cancer in the family, are recommended. Women with ≥ 25% lifetime risk for developing breast cancer are also eligible for MRI (where available). Screening recommendations may change over time as our knowledge of breast cancer increases. Personalized screening recommendations will be provided as part of your genetic counselling appointment.

Ovarian cancer screening is not routinely recommended. The available screening methods have not been proven to reliably detect ovarian cancer at an early stage. All women should be aware of the symptoms of ovarian cancer and should approach their doctor promptly if they have any unusual abdominal, gynecological or urinary symptoms.

*Lifetime Risks Associated with BRCA1 and BRCA2 Mutations**

Type of Cancer	BRCA1	BRCA2	General population risk
Women*			
Breast Cancer	50-85%	50-85%	**11%**
Ovarian Cancer	20-60%	10-30%	**1.5%**
Men*			
Prostate Cancer	Increased risk	Increased risk	**10-15%**
Breast Cancer	Increased risk	6%	**0.1%**

**Men and women who carry a BRCA1 or BRCA2 mutation may have a slightly increased risk for other cancers as well, such as melanoma, gallbladder/bile duct, pancreatic, gastrointestinal and hematologic cancers. The exact risk figures have not yet been calculated.*