Screening & Risk Reduction Options for Carriers of BRCA Mutations

Female Carriers:

Breast Cancer:

1. **Screening:**
   - Beginning by age 25 or individualized based on family history if the youngest diagnosis in the family is under 30
     - Age 25-29, yearly breast MRI screening (preferred) or mammography if MRI is unavailable and clinical breast exams every 6-12 months by a breast specialist.
     - Age 30-75:
       - Yearly mammogram
       - Yearly Breast MRI
     - Age >75, management should be considered on an individual basis.
     - Clinical breast exam every 6-12 months (consider one by a breast specialist and one by a gynecologist or primary care physician)

2. **Risk Reduction:**
   - **Tamoxifen or Evista (raloxifene):**
     - Medications that have been shown to reduce the risk of future breast cancer in women at increased risk of the disease.
     - We do not have a lot of data on how these medications will affect risk in BRCA carriers, but expect more data to be available in the future.

   *Note: Taking tamoxifen following a diagnosis of an estrogen-receptor negative tumor is somewhat controversial; please speak to your physicians about the pros and cons of taking tamoxifen.*

   - **Prophylactic Surgery:**
     - Bilateral prophylactic mastectomy
     - Studies have shown that BRCA carriers who had this surgery reduced their risk of a future breast cancer by >90%.
- **Ovarian Cancer:**

  1. **Screening:**
     - Note: For women who have chosen not to pursue preventative removal of the ovaries and fallopian tubes, data do not support routine ovarian screening. The current screening techniques for ovarian cancer have not been shown to detect ovarian cancer at an early, more treatable stage; therefore, women should consider risk reduction options and discuss these options with their physicians.

     - However, physicians and patients may find screening helpful under certain circumstances and can consider and discuss the pros and cons of the available screening options beginning at age 30-35 including transvaginal Doppler ultrasound (preferably day 1-10 of menstrual cycle in premenopausal women) and CA125 blood marker screening (preferably after day 5 of menstrual cycle in premenopausal women) every 6 months.

  2. **Risk Reduction:**
     - **Oral Contraceptives (Birth Control Pills):**
       - Use of oral contraceptives can reduce a woman's lifetime risk of ovarian cancer.
       - Several studies now suggest that this reduction in risk holds true in carriers of BRCA mutations.

       *Note: Taking oral contraceptives following a diagnosis of breast cancer is often contraindicated; if you have had breast cancer, please speak to your physicians about the appropriateness of taking oral contraceptives.*

     - **Prophylactic Surgery:**
       - Due to difficulties detecting ovarian cancer at an early, treatable stage with current screening techniques, it is recommended that women who carry BRCA mutations consider bilateral salpingo-oophorectomy (removal of the ovaries AND fallopian tubes) typically between ages 35-40, once they are finished having children, or individualized based on the earliest ovarian cancer in the family (whichever is youngest).

       - This surgery has been shown to reduce the risk of ovarian and fallopian-tube cancers in BRCA carriers by >90%.
- This surgery has also been shown to significantly reduce the future risk of breast cancer, particularly in young women who have surgery prior to menopause.

- Some data suggest that many ‘ovarian’ cancers may originate in the fallopian tubes. This has led some researchers to question whether prophylactic salpingectomy (removal of the fallopian tubes only) with delayed removal of the ovaries closer to menopause may be an option to delay or prevent potential side effects from early, surgical menopause, while reducing the risk of ‘ovarian’ cancers that begin in the fallopian tubes. **However, this option is still an area of active research and data are limited regarding its effectiveness. We expect more answers about this option to be evolving over the next few years. Until more information is available, this option should be considered research (not ready for routine clinical use) and the pros and cons should be carefully weighed.**

- **Pancreatic cancer and Melanoma:**

  - Based on the possible increased risk for melanoma, individuals with a BRCA2 mutation should consider a yearly full-body skin exam with a dermatologist and eye examinations by a specialist for eye (ocular) melanoma.

  - Although BRCA2 carriers are at increased risk for pancreatic cancer, the majority of individuals with a BRCA2 mutation will not develop pancreatic cancer. No specific guidelines for pancreatic cancer screening in BRCA2 carriers exist and the risks, benefits, and limitations of pancreatic cancer surveillance are unknown. Individuals with a BRCA2 mutation may wish to speak to a specialist in high risk pancreatic cancer screening to further discuss the available screening options, particularly if they also have a family history of pancreatic cancer. In the New Haven area, please contact 203-200-5083 to schedule an appointment with Dr. James Farrell.
References:


http://www.ncbi.nlm.nih.gov/books/NBK1247/