A case of male breast cancer with BRCA2 mutation

Personal history: 56-year-old male presents with a "bump" on his chest, which he attributes to a prior car accident. Pathology reveals invasive ductal carcinoma of the breast.

Family history: Sister, breast cancer at 60; paternal grandmother, breast cancer at 55.

Genetic testing: Multiple professional society guidelines recommend BRCA1/2 testing for males with breast cancer at any age, even in the absence of family history. The patient was found to have a BRCA2 deleterious mutation.

Management and Surveillance Recommendations:

Breast cancer: Treatment would follow standard protocols based on the specific diagnosis. The lifetime risk for male breast cancer is 7-8% for BRCA2 mutations and 1-2% for BRCA1 mutations. This compares to a risk of 0.1% (1 in 1000) in the general population.

Prostate cancer: BRCA mutation carriers have up to a 20% risk for prostate cancer by age 80, compared to 14% in the general population. Surveillance includes prostate specific antigen (PSA) testing and digital rectal exams beginning at 40. Some reports suggest that prostate cancer among BRCA positive men may occur at younger ages and follow a more aggressive course, so medical management may need to be adjusted accordingly.

Other cancers: In a study of BRCA2 families, the cumulative risk of all BRCA-associated cancers in men was 32% by age 70, compared with 90% for women. Both men and women have an elevated risk of melanoma and pancreatic cancer, up to 4% and 7% respectively. However, the pancreatic cancer risk may be even higher in BRCA2 mutation carriers with a family history of pancreatic cancer.

NCCN recommendations for management of males with BRCA1/2 mutations include:

- Breast self-exam training and education starting at age 35 y
- Clinical breast exam, every 6-12 months, starting at age 35 y
- Consider baseline mammogram at age 40 y; annual mammography if gynecomastia or parenchymal/glandular breast density is present
- Adhere to screening guidelines for prostate cancer
- Educate regarding signs and symptoms of cancer
- Consider full body skin exams for melanoma and investigational protocols for pancreatic cancer

Implications for family members: "Single site" testing for the patient's specific BRCA2 mutation is available for his relatives. Each of his two adult children and four sisters has a 50% chance of carrying the mutation. Numerous professional societies provide medical management guidelines for any relative that tests positive. If a relative tests negative, then his/her cancer risks (in the absence of additional risk factors) would be the same as those for the general population.

Key points:

- Men who are positive for BRCA mutations have a substantially increased risk for cancer, including breast and prostate cancer.
- The National Comprehensive Cancer Network (NCCN) practice guidelines outline management/surveillance recommendations for BRCA positive males.
References:


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