RAD51D Mutations

What You Should Know About RAD51D Mutations

Individuals with a RAD51D mutation have an increased risk to develop ovarian cancer, and potentially female breast cancer and prostate cancer. There is limited information regarding the lifetime cancer risks for individuals with a RAD51D mutation.

Cancer Risks Associated with a RAD51D Mutation

- **Ovarian Cancer**: Females with a RAD51D mutation have an increased risk to develop ovarian cancer. The specific lifetime risk for ovarian cancer is estimated to be between 7-14% compared to the general population risk of 1-2%.
- **Female Breast Cancer**: There is a potential increased risk for females with a RAD51D mutation to develop triple negative breast cancer, although information is limited, and the specific risk estimates are unknown at this time.
- **Prostate Cancer**: Based on limited information, RAD51D mutations may also be associated with an increased risk to develop prostate cancer.

Risks to Family Members

Mutations in the RAD51D gene are inherited in an autosomal dominant fashion. This means that children, brothers, sisters, and parents of individuals with a RAD51D mutation have a 1 in 2 (50%) chance of having the mutation as well. Individuals with a RAD51D mutation may develop ovarian cancer, female breast cancer, prostate cancer, or none of the above. Both males and females can inherit a familial RAD51D mutation and can pass that on to their children.

Managing Cancer Risks

The following are general management recommendations from the National Comprehensive Cancer Network (v1.2020):

- Consideration of risk reducing salpingo-oophorectomy (RRSO) for women with a RAD51D mutation. A discussion about surgery should occur around age 45-50 (or earlier based on a family history of ovarian cancer).
- For women who have not elected RRSO, transvaginal ultrasound combined with serum CA-125 for ovarian cancer may be considered at their clinician’s discretion. However, the benefit of ovarian cancer surveillance is uncertain at this time.
- RAD51D mutation carriers may be sensitive to specific chemotherapy agents and thus may benefit from therapies such as poly ADP ribose polymerase (PARP) inhibitors.
- Current guidelines suggest that there is insufficient evidence for additional cancer interventions based on a RAD51D mutation alone. An individual’s personal risk factors and family history of cancer should be considered in developing an appropriate screening plan.

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