UT Southwestern Harold C. Simmons Comprehensive Cancer Center | NCI Designated Comprehensive Cancer Center

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 it 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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