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

RAD50 Mutations

What you should know about *RAD50* mutations

Individuals with a *RAD50* mutation have an increased chance to develop female breast cancer and possibly ovarian cancer. There is limited information regarding the specific lifetime cancer risks, but more information may become available with ongoing research. Individuals with two *RAD50* mutations can have a disease known as Nijmegen breakage syndrome-like disorder (NBSLD), which is associated with childhood cancer risks.

Cancer risks associated with a *RAD50* mutation

Females with a *RAD50* mutation may have an increased risk to develop breast cancer. Some studies have proposed a lifetime risk for breast cancer of 24-36% (compared to the average population risk of 12%). Other studies have found no increased risk for breast cancer.

Some studies have proposed that *RAD50* mutations may be associated with an increased risk to develop ovarian cancer. However, these studies are small and data remains limited.

Risks to family members

Mutations in the *RAD50* gene are inherited in an autosomal dominant manner. This means that children, brothers, sisters, and parents of individuals with a *RAD50* mutation have a 1 in 2 (50%) chance of having the mutation. Individuals with a *RAD50* mutation may develop one cancer, more than one cancer, or none at all.

Individuals with two *RAD50* mutations (one from each parent) have Nijmegen breakage syndrome-like disorder. Couples may wish to undergo prenatal genetic counseling before or during pregnancies to better understand their child's risks to have NBSLD.

Managing cancer risks

Currently, there are no formal management guidelines for individuals with *RAD50* mutations. Individuals with a *RAD50* mutation are encouraged to discuss cancer screening options with their physician. An individual's personal and family history should be considered when developing an appropriate surveillance plan.

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