

High risk of colorectal, endometrial and Lynch syndrome cancers for MSH6 mutation carriers

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People carrying the germ-line MSH6 mutation are at high risk by age 80 years for colorectal and endometrial cancers and any cancer associated with Lynch syndrome, according to a new study published online December 22 in the *Journal of the National Cancer Institute*.

It is known that mutations in MSH6 account for 10%-20% of Lynch syndrome, which causes colorectal and other cancers, but less is known about the cumulative [cancer](#) risks for mutation carriers.

In this study, members of the [Colon Cancer](#) Family Registry, a National Institutes of Health-funded consortium, and colleagues identified 113 families of MSH6 mutation carriers from five countries who were identified through family cancer clinics and population-based cancer registries. Mutation status, sex, age, and histories of cancer, polypectomy, and [hysterectomy](#) were sought from 3,104 of their relatives.

The researchers determined precise estimates of both absolute and relative cancer risks for people who carry the mutation. The estimated cumulative risks to ages 70 and 80 years, respectively, were 22% and 44% for colorectal cancer for men and 10% and 20% for women. For endometrial cancer, risks were 26% and 44% for women. And for any cancer associated with Lynch syndrome, the risks were 24% and 47% for men and 40% and 65% for women. Compared with incidence for the

general population, MSH6 mutation carriers had an eight-fold increased incidence of colorectal cancer. [Women](#) who were MSH6 mutation carriers had a 26-fold increased incidence of endometrial cancer and a six-fold increased incidence of other cancers associated with Lynch syndrome.

"These results demonstrate that the elevated risks for cancers in MSH6 mutation carriers differ by sex of the carrier and continue into older age," the authors write.

Provided by Journal of the National Cancer Institute

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