UTSouthwestern

Harold C. Simmons Comprehensive Cancer Center



BRIP1 Mutations

What You Should Know About BRIP1 Mutations

Individuals with one *BRIP1* mutation have an increased risk for ovarian cancer. There is some evidence suggesting that females with one *BRIP1* mutation may also have an increased risk for breast cancer; however, this data is less clear at this time. When an individual inherits two *BRIP1* gene mutations (one from each parent), this causes a syndrome called Fanconi anemia (FA).

Cancer Risks Associated with a BRIP1 Mutation

- Ovarian cancer: Women with a BRIP1 mutation have a 5.8-18% lifetime risk of ovarian cancer.
- <u>Breast cancer</u>: There is a potential increased risk for female breast cancer (including triple negative breast cancer) in women with a *BRIP1* mutation. However, the specific lifetime risk is not known at this time.
- As *BRIP1* is a more recently discovered gene, other cancer risks and cancer risks in males with a *BRIP1* gene mutation are unknown at this time.

Risks to Family Members

Mutations in the *BRIP1* gene are inherited in an autosomal dominant fashion. This means that children, brothers, sisters, and parents of individuals with a *BRIP1* mutation have a 1 in 2 (50%) chance of having the mutation as well. Individuals with a single *BRIP1* mutation may or may not develop cancer. Both males and females can inherit a familial *BRIP1* mutation and can pass it on to their children.

When an individual inherits two *BRIP1* gene mutations (one from each parent), this causes a syndrome called Fanconi anemia (FA). FA is associated with physical abnormalities, bone marrow failure, childhood leukemia and other cancers.

Managing Cancer Risks

The National Comprehensive Cancer Network (NCCN v1.2020) recommends the following management for individuals with a *BRIP1* mutation.

- <u>Ovarian cancer</u>: Consider risk-reducing salpingo-oophorectomy (RRSO) at age 45-50 years or earlier based on ovarian cancer family history. Insufficient evidence exists to recommend an optimal age for RRSO.
- <u>Breast cancer</u>: Insufficient evidence to recommend modified breast cancer risk management based on *BRIP1* mutation status alone. An individual's personal and family history should be considered in developing an appropriate surveillance plan.

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