**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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