**MRE11A Mutations**

**Cancer Risks and General Management Recommendations**

<table>
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<th>MRE11A Mutation Carrier Cancer Risks</th>
<th>General Population Lifetime Cancer Risks</th>
<th>Surveillance/Management Recommendations³</th>
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| **Female Breast**¹  
37.2% (3-fold of general population risk) | 12.4% | **Surveillance and Surgery**  
- Insufficient evidence for intervention with breast MRI.  
- Insufficient evidence for intervention with risk-reducing mastectomy (RRM).  
- Family history and other personal factors (e.g. breast density), in conjunction with the presence of a MRE11A mutation, should be considered when evaluating medical management options.  
- These options should be discussed with a physician to determine the most appropriate management. |

| **Ovarian Cancer**²  
Unknown | 1.3% | **Surgery**  
- Insufficient evidence to make any recommendations for risk-reducing salpingo-oophorectomy (RRSO).  
- Risk management for ovarian cancers should be based on an individual’s personal and family history.  
- These options should be discussed with a physician to determine the most appropriate management. |

Some studies have proposed an increased risk for breast cancer in females with a MRE11A mutation (lifetime risk of 37.2%, 3-fold of general population risk).¹ However, others have found no increased risk for breast cancer. It is currently unknown if there is an increased risk for ovarian cancer in individuals with a MRE11A mutation.² Additionally, it is currently unknown if MRE11A mutations cause a predisposition to other cancers. Current NCCN guidelines assert that there is insufficient evidence to make any recommendations for breast MRI, risk-reducing mastectomy (RRM), or risk-reducing salpingo-oophorectomy (RRSO) based on MRE11A mutation status alone.³ An individual’s personal and family history should be considered in developing an appropriate surveillance and management plan.

**Implications for Family Members/Reproductive Considerations**

- First-degree relatives (i.e., parents, siblings, and children) have a 50% chance to have the familial MRE11A mutation. Second-degree relatives (i.e., nieces/nephews, aunts/uncles, and grandparents) have a 25% chance to have the familial mutation.
- Rarely, children inherit MRE11A mutation from both parents. Individuals with two MRE11A mutations have ataxia-telangiectasia-like disorder (ATLD), an autosomal recessive neurodegenerative disorder affecting multiple body systems.⁴ Parents who each carry a MRE11A mutation have a 25% cancer for a child with ATLD with every pregnancy.  
  - MRE11A genetic testing for the partner of an individual with a MRE11A mutation may be appropriate to clarify the risk of having children with ATLD.  
- For carriers of a known mutation, assisted reproduction (with or without egg or sperm donation), pre-implantation genetic testing, and prenatal diagnosis options exist.
- All family members are encouraged to pursue genetic counseling to clarify their risks. Family members can visit www.FindAGeneticCounselor.com to find genetic services near them.
References


