

BroadcastMed | Diagnosing and Managing Patients with BRCA Mutations

SPEAKER 1: My name is Sandia Preethi, and I'm a consultant in the Breast Diagnostic Clinic at the Mayo Clinic in Rochester, Minnesota. And the title of my paper is, "The Identification and Management of Women with a BRCA Mutation or Hereditary Predisposition for Breast and Ovarian Cancer". And this will be published in the Mayo Clinic proceedings in December 2010.

So the most important factor for women who are BRCA mutation carriers is that their lifetime risk of developing an invasive breast cancer can be anywhere from 40 to 85%. And for ovarian cancer, this same BRCA mutations are associated with anywhere from 20 to 65% lifetime risk of ovarian cancer.

So there are some important issues and characteristics that physicians need to think of when they're identifying someone who may be a carrier of the mutation or have a strong hereditary family history. And this includes members in their family who have had both breast and ovarian cancer, a male with breast cancer, young onset multiple relatives with breast cancer or ovarian cancer before age 50, and if somebody has had bi-lateral breast cancer.

So the role of the genetic counselor is to take the time to look at the family history and develop a pedigree, and by using that pedigree they can actually identify if someone has a significant pattern that would be associated with potentially carrying a mutation and can use this then to guide a patient if she needs to proceed or advise her she should proceed with genetic testing.

There's some management options that once a woman is identified as a BRCA carrier or a mutation is learned that she is now carrying a BRCA mutation to help her decide how to manage the situation. And we start with lifestyle modification. We talk about chemo prevention and the role of medications such as tamoxifen and surveillance. How do you follow someone who is now a carrier and is mammography enough or these women should be in addition to mammography get breast MRI screening?

And then, finally, the role of risk reducing surgeries also, more commonly known as prophylactic mastectomy or prophylactic oophorectomy. When is the right time to do this procedure? And what is the benefit of someone pursuing a surgery such as this in terms of reducing your breast cancer mortality or ovarian cancer mortality?

So the takeaway message is that being a woman who's identified as a BRCA mutation carrier or has a strong family history and predisposed to potentially carrying a mutation is a very complicated and challenging situation. And more and more it's becoming individualized approach requiring a multi-disciplinary team of experts.

The goal is not only to provide education but to reduce cancer risk and then improve the woman's survival.

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