Genetic counseling perspective of engagement with urology and primary care

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Germline genetic testing for prostate cancer is helping to inform risk stratification and staging of prostate cancer and also screening for men with family history of prostate cancer. Genetic counseling is an important piece of germline genetic testing; however there can be limitations of access to genetic counselors and other genetic professionals. It is important to integrate genetic counseling with urology and primary care practices.

Key Words: genetic counseling, genetic testing, primary care, urology, prostate cancer

Prostate cancer is a common cancer that also has a high hereditary component.[1] Studies have shown 15% to 17% of men with prostate cancer have a germline mutation regardless the stage of prostate cancer.[2,3] Germline mutations are reported in 12% of men with metastatic prostate cancer.[4]

The National Comprehensive Cancer Network (NCCN) recommends that family or personal history of high-risk germline mutations be included in considerations of prostate cancer screening.[5] Given that men with *BRCA1* and *BRCA2* germline mutations have an increased risk of developing prostate cancer at an earlier age with higher mortality rates, it is discussed that PSA screening should start at age 40 with consideration of annual screening intervals.

After an initial prostate cancer diagnosis, germline genetic testing is important in risk stratification and staging.[6] The NCCN also discusses asking about high-risk germline mutations and family history. Germline genetic testing is recommended for individuals with a strong family history, a high or very high-risk prostate cancer, or intraductal histology.

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Active surveillance is often recommended for very low-risk, low-risk, and favorable intermediate-risk prostate cancer depending on other factors such as life expectancy.[6] However, men with prostate cancer and *BRCA1* or *BRCA2* germline mutation may have an increased risk of progression on local therapy and decreased overall survival.

The NCCN also recommends that, prior to genetic testing, an expert in cancer genetics provide pretest genetic counseling.[6,7] Pretest genetic counseling includes, collecting a three generation family history, evaluating who is best to test in the family, determine which genetic testing would be best for the patient and/or their family, educating on possible genetic testing results, and addressing privacy/psychosocial implications. Genetic counselors (GCs) play an important role of ensuring patients are properly consented to genetic testing.

However, there are limitations to access to GCs and genetic professionals. Many men who meet genetic testing criteria will see their urologist and primary care doctor, but do not have access to a GC. Common barriers to this access include hardship for patients to travel to another appointment and long wait times at genetics clinics. Limitations in access to genetic testing can restrict men from obtaining vital information regarding their cancer screening and treatment. Before

a urology group or primary care office considers genetic testing in their office, it is important to consider a few challenges that might occur with genetic testing.

A considerable challenge in the genetic testing process is obtaining accurate family history information.[8] GCs take a comprehensive family history by a medical pedigree, which is a graphic representation of an individual's family history. They ask questions to determine family history of cancer and obtain a three-generation representation of the family. It is important for the pedigree to be as accurate as possible. When possible, family history should be confirmed with a pathology report or doctors' note. The time it takes to gather a complete pedigree varies between patients, however it is a very detailed process. Taking a family history is also very personal and brings up psychosocial concerns. Family history information is used to help guide which genetic test is ordered, screening recommendations for family members, and interpretation of genetic testing results. The amount of time spent with patients is 9 to 16 minutes for urologists and 13 to 24 with family medicine doctors.[9,10] A detailed family history would be difficult to get in that amount of time with other concerns a urologist or primary care physician will need to address. GCs spend around 60 to 90 minutes with a new patient.

Genetic counseling also includes a thorough discussion of the possible results and the implications on cancer risk. Results of genetic testing may not always be clear. Variants of uncertain significance (VUS) are reported in about 30% of men with prostate cancer who have undergone genetic testing.[2] A VUS may not affect recommendations at the time of reporting; however it can cause confusion and a lack of understanding for the patient as well as providers. In addition, testing using a large panel can uncover mutations in genes with unexpected or ambiguous cancer risks. Pretest genetic counseling may aid in addressing the possibility of these ambiguous or unexpected results.

How do we improve access to genetic counseling for patients without overwhelming already busy providers? Offices will need to determine which patients can be consented in the office or which patients need a GC appointment. The key is to ensure patients are fully consented and are prepared for the information it will provide. Providers also need to ensure they have the correct family history information to interpret these results. A collaborative approach with GCs and primary care and urology offices will ensure patients are being properly consented, receiving the right testing and getting this information in an easy and timely way.

Disclosures

Carey McDougall and Jessica Russo have no disclosures. Colette Hyatt is a consultant for GenomeSmart. □

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