“A Way You Can Contribute to Taking Care of Your Family”

New genetic testing from Invitae shows how genetic abnormalities in prostate cancer patients can affect both future and family cancer risks.

March 29, 2019 By Justin Birckbichler

If you haven’t deduced by now, I enjoy reading research studies about men’s health and putting my own spin on them. Though that has never made it into my dating profiles (to be fair, I haven’t had a dating profile since I had two testicles, which I’ve said held me back from my best life), it’s truly something I enjoy — learning from these studies and making the results more easily understood. It must be the teacher in me.

Recently, I came across a study entitled “Prevalence of Germline Variants in Prostate Cancer and Implications for Current Genetic Testing Guidelines” by Invitae. While that study name is a mouthful, the main investigation was to discover if genetic abnormalities in prostate cancer patients had effects on later diagnoses or family history. I had the opportunity to connect with one of the authors of the study, Robert Nussbaum, MD, to discuss it more in depth.

Before delving into the inspiration and the results of the study, I wanted to get to know Dr. Nussbaum and gain a stronger understanding of obstacles to genetic testing.

After graduating from Harvard Medical School, Dr. Nussbaum spent 40 years in medicine in a variety of roles — working at cancer clinics; academia, at the Genome Institute; and more — culminating in his current role as Chief Medical Officer, Invitae.

He said he has had a “lifelong commitment to integrating genetics into mainstream medicine,” as he’s felt it’s been underutilized for a variety of reasons. One of these roadblocks are economic issues, as it has traditionally been an expensive practice, but that obstacle is coming down as genetic testing is becoming less expensive and more widely available.

Additionally, medical providers haven’t always seemed comfortable with recommending genetic testing and insurance companies have been resistant to approve testing. However, through showing the benefits of genetic testing and how it can provide better care, these barriers are also slowly being removed.

Now that I was a world-renowned expert on genetics, I was ready to dive into the research study itself.

A few years ago, the New England Journal of Medicine found that there was a strong link between metastatic prostate cancer patients and mutations in genes predisposed to familial cancers, such as BRCA2 and breast cancer. Patients with a Gleason score of over 7 (indicative of metastatic prostate cancer) showed a significant gene mutation likelihood — about 1 in every 6 or 7 men passed on some sort of mutation.

However, this wasn't easy to study in low-grade prostate cancer because of "noise that would obscure this." Additionally, guidelines were too narrow — genetic testing would only be recommended for men with family history of cancer or aggressive forms of cancer. The researchers in this study wanted to study it for all prostate cancers — not just metastatic.

Keep reading on — the findings are easier to understand than the background information or the inspiration.

I know regular readers of ABSOT expect humor when they visit this site rather than genetic research studies, but this study is important and shouldn't be the butt of any jokes!

In layman's terms, the researchers found that many dozens of genes were altered in all forms of prostate cancer — not just aggressive forms — and in men of all backgrounds. What this means is two-fold. First, this increases the risk for future cancers in that man's life. Specifically, if a mutation in BRCA2 is found, it "significantly increased risk for both male breast cancer and pancreatic cancer compared to someone who does not carry."

I found the second point to more interesting, however. If these abnormalities are present, it will increase chances of passing on cancer in the family — but not just prostate cancer. For example, if a man passes on these genetic abnormalities to a female in his family, she can be at a higher risk for breast or ovarian cancer. I thought this to be important to highlight because not many people think to look for cancers across "gender lines" when trying to look for cancer histories.

This genetic testing can also affect men without prostate cancer. If they are tested for BRCA2 abnormality because a relative tested positive and is found to also be positive, risks for prostate cancer developing is also increased over someone of same age without a BRCA2 abnormality.

Now that they've gotten to the bottom of this, genetic testing can help dictate the course of screenings for the future man's life and offer genetic counseling to the rest of the family. Furthermore, the company aims to launch a collaboration with a national urology clinic and will begin offering patient-initiated testing later this summer. On a larger scale, they hope to do research into other cancer types and what genetic abnormalities are present in those.

In addition to sharing these breakthroughs, I also asked Dr. Nussbaum what he felt was important to relay to men about prostate cancer.

First and foremost, he felt that it was important to stress that prostate cancer is very common and also very treatable.

On the topics of treatments, he said that there are new targeted treatments (called checkpoint inhibitors) being developed. Observing tumors has shown that they become surrounded by immune cells, but the cells are ‘blocked’ from attacking the tumor. Drugs have been developed to remove this ‘block’, which means the patient’s own immune system can attack the tumor without needing outside help or other treatments.

One of the co-authors on the study, Oliver Sartor, MD, had a patient that was not responding to traditional treatments. In the course of the patient’s genetic testing, they found a MSH2-defective gene, which makes strange proteins in the prostate. These proteins makes it difficult for the immune cells to attack since they see it as part of the body. Dr. Sartor developed drugs to remove the block and immune cells began to attack the tumor. This option won’t be a miracle, but it is a good first step of getting genetic testing as a more embraced option.

I always like to hear why doctors think men are so reluctant to discuss their health.

While Dr. Nussbaum didn’t have direct contact with the men in this study, he was able to offer his own conjecture. He feels that male culture in America puts a premium on being tough, strong, silent, resilient, not complaining. He said that this is perpetrated as the ideal man, and it spills over into health matters. This is nothing new to me, but it’s always reassuring to hear that we’re all on the same page.

However, this genetic testing, especially paired with how it can help determine family risk, could be a turning point.

Dr. Nussbaum closed out the interview by saying that men are conditioned to be a solid rock and provider. To his patients, he says, “Look — you and I can talk about what this means for you, but you recognize this testing is a gift. By going through this testing, you are being a caregiver and giving this gift to the others in your family. It’s hard but it is worthwhile. As men, we protect our loved ones. This is a way you can contribute to taking care of your family.”

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