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Decoding Risk: Genetic Testing in High-Risk Colon Cancer Patients

Dr. Takemoto:
You're listening to *GI Insights* on ReachMD. I'm your host, Dr. Jody Takemoto. And today I'm speaking with Dr. Bryson Katona about his presentation at the 2024 Digestive Disease Week Annual Meeting, which focused on genetic testing for patients with high-risk colon cancer. Not only is Dr. Katona an Assistant Professor of Medicine at the Hospital of the University of Pennsylvania, but he's also the Director of Gastrointestinal Cancer Genetics Program and Risk Evaluation Program and the Lynch Syndrome Program at Penn Medicine. Dr. Katona, thanks for being here today. Dr. Katona, thanks for being here today.

Dr. Katona:
Well, thank you so much for the invitation.

Dr. Takemoto:
So to get us started, Dr. Katona, can you tell us about the prevalence of colon cancer?

Dr. Katona:
Yeah. So colon cancer, of course, we know is quite common. We think that it's about the third most common cause of cancer in both men and women. Amongst colon cancer, we think that probably about 1/4 of all colon cancers have some sort of familial component to them. Now it's a smaller slice of that pie where we know that they're due to known hereditary cancer predisposition syndromes, but we think probably it's about 5 percent; maybe even a little bit higher of all colon cancers are due to known hereditary GI cancer syndromes.

There are multiple different syndromes that are out there. The most common one that we run into is something called Lynch syndrome. Lynch syndrome increases risk of colorectal cancer and uterine cancer, but then also many other cancers, including gastric cancer, pancreatic cancer, and urothelial cancer, among others. And Lynch syndrome is actually incredibly common. We estimate that it affects about one in 270 people, which translates into about 1.1-1.2 million Americans. So when you think about hereditary colorectal cancer syndrome, that's a lot of people.

Dr. Takemoto:
Now with that being said, when do you send somebody for genetic testing for a hereditary colorectal cancer syndrome?

Dr. Katona:
There are multiple different personal and family characteristics that you can take into account. For example, if somebody does have a colorectal cancer or if they're very young with colon cancer, then they should be sent really regardless of any of their other history or any of their family history. And so typically, we say anybody under age 50 should have genetic testing for colorectal cancer predisposition syndrome.

Not everybody, of course, that we do testing on has cancer, and so sometimes we can identify appropriate people based on the number of polyps that they've had. So if you think about colon polyps, and especially colonic adenomas, which are, of course, the type of polyps that we're finding most commonly and we think are the most important to remove, the threshold at which a genetic evaluation should be triggered has actually dropped, and so now we'd say that anybody with more than 10 cumulative colonic adenomas should have genetic testing. Most gastroenterologists, I think, are actually surprised by that number because it's not that many. It used to be 20, but we think that really the yield having 10 or more is still acceptable.

Other types of polyps that should trigger alarm is if somebody has multiple hamartomatous polyps of the colon or the rest of the digestive tract. And there are upper digestive tract polyps too, such as duodenal adenomas, or if there is kind of advanced fundic gland,

polyposis, especially if there's dysplasia in these polyps, and especially if there are no other risk factors for fundic gland polyps, that could prompt evaluation for a colon cancer predisposition syndrome.

One or two others that I'll just briefly mention is of course, family history; it's incredibly important. So you may have a patient who's totally unaffected and who hasn't had any polyps themselves, but if they have a strong family history of colorectal cancer or other hereditary GI cancer risk syndromes like Lynch syndrome, then your patient likely is a testing candidate themselves.

And the last thing I'll mention about how to identify people who need genetic testing is you may have a patient who had genetic testing maybe 10 or 15 years ago, but in fact, the number of genes that we test for to look for colon cancer risk syndromes has increased dramatically over the last 5 to 10 years. If you had somebody that had genetic testing 10 or 15 years ago, that genetic testing may not be comprehensive, and so that individual may actually be able to come in to get updated genetic testing.

Dr. Takemoto:
So for those of you just tuning in, you're listening to *GI Insights* on ReachMD. I'm Dr. Jody Takemoto, and I'm speaking with Dr. Bryson Katona about his research on genetic testing in colon cancer. So, Dr. Katona, if you send a patient for genetic testing, how frequently do we find a positive result?

Dr. Katona:
That's a great question, and one that I think there's a lot of misperceptions about as well. So a lot of times when we're sending tests, the thought about how often we're going to find a positive result can kind of be skewed by the rest of our approach in medicine. In genetic testing as a field, we really think that if we're finding a positive result in 5 percent or more of individuals who get tested, we actually consider that a success and consider that a victory. So 5 percent is a very low number, and what that means is that the vast majority of people that you should be sending for genetic testing should actually be coming back with negative results or no actionable gene mutations. It's a very different mindset from a lot of the other tests that we send in medicine.

There have actually been several studies that have looked at this in patients with colorectal cancer. And if you look at large cohort studies that have looked at thousands of patients who had colorectal cancer, what we find is that somewhere between 10 to 15 percent of these patients will have an actionable gene mutation found on genetic testing. So 10 to 15 percent is not high, but you're still talking about one out of seven to one out of eight people with colorectal cancer has an actionable gene mutation, which, in the genetic testing world, makes testing from our perspective worth it.

But I think that is an important number to keep in the back of your mind, especially when you're counseling patients as well, because if you're trying to sell patients on genetic testing, you can say, "Well, you know, more than likely your testing is going to come back normal." That actually may get more people to kind of be more accepting of moving forward with genetic testing compared to if they thought that there was a very high risk, you know, over 50 percent chance that they were going to come back with an identifiable actionable gene mutation.

Dr. Takemoto:
And are we at the point where genetic tests inform our approaches to managing patients with colon cancer?

Dr. Katona:
Yeah, certainly. I mean, genetic testing is important for the patients, but incredibly also important for the families as well, and so I think when you're sending somebody and when you identify one of these hereditary gene mutations, you always have to think about the fact that, yes, it has impacts for your patient themselves, but it also has major impacts for the family members. And so if you have somebody with colon cancer who tests positive for Lynch syndrome, that then allows you to go and find out which of their kids have Lynch syndrome or which of their brothers and sisters have Lynch syndrome because those who are identified to have the genetic predisposition can then undergo the appropriate screening and other risk reduction strategies. Whereas, on the alternative side, if you have somebody with colon cancer who tests negative for Lynch syndrome, that's also very helpful because in that situation, you know that Lynch syndrome didn't cause their colon cancer, but that's also important for further informing recommendations for that individual's family members as well.

Dr. Takemoto:
Before we end today, Dr. Katona, what recommendations would you give to other clinicians who not only want to incorporate genetic testing into their practice, but also want to encourage their patients to get tested?

Dr. Katona:
Yes, it's a great question. Genetic testing is complicated, I should say. I will just highlight there are a couple of easy things about genetic testing. So nowadays, we can just do it with saliva. It's very easy to collect that sample. Cost is actually relatively inexpensive.

Even if patients want to forgo and pass up insurance and just want to pay out of pocket, it's usually no more than $250, and it's very quick too. I mean, we're usually getting results back within a couple weeks.

But like I said, genetic testing is complicated, and I think that the complicated side of it is that there are implications of when a genetic finding comes on a patient's chart, and those implications primarily have to do with insurance. So while there are protections in place for medical insurance as well as workplace discrimination, there are actually no universal protections in place for discrimination in the life insurance arena, the disability insurance, or the long-term care insurance arena, and so I think it's important that patients be counseled about that before they undergo genetic testing because if they do undergo genetic testing and they're found to have something, that's really on their record forever. And so that counseling is done by genetic counselors who are an absolutely amazing resource for individuals who are considering genetic testing, and I strongly advise any physician who is considering sending an individual for genetic testing to make sure that that individual has the opportunity to speak with a genetic counselor. They will kind of go through the pros and the cons of testing. The genetic counselors can help the patients determine exactly what type of genetic testing they want to proceed with. Ultimately at the end of the day, after speaking with a genetic counselor, the patient is going to feel very informed about their decision, and they're going to really feel like they're making the appropriate decision for them.

Dr. Takemoto:
Well, with those strategies in mind, I want to thank my guest, Dr. Bryson Katona, for sharing his insights on genetic testing in colon cancer. Dr. Katona, it was great speaking with you today.

Dr. Katona:
Thank you so much for the opportunity and the invitation.

Dr. Takemoto:
For ReachMD, I'm Dr. Jody Takemoto. To access this and other episodes in our series, visit *GI Insights* on ReachMD.com, where you can Be Part of the Knowledge. Thanks for listening.