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### Hereditary Cancer Syndromes and the Obstetrician/Gynecologist

Dr. Matt Birnholz:

You're listening to Reach MD, and I'm Dr. Matt Birnholz. Today I'm joined by Dr. Andrew Wagner. He's Associate Professor of OB/GYN at the University of Oklahoma Health Science Center, and he's going to be talking about a topic of focus for him which is hereditary cancer syndromes and the obstetrician/gynecologist.

Dr. Andrew Wagner:

Sounds good.

Dr. Matt Birnholz:

So why don't we start with hereditary cancer syndromes and the OB/GYN. Why did you title it that way? Why specifically do we have to highlight the OB/GYN with hereditary cancer syndromes? What's the misconception, or what are we not knowing right now?

Dr. Andrew Wagner:

Well, I think it's more awareness of personal histories and family histories in the patients we're already seeing. We're already so involved in screening for breast cancer, and OB/GYN I think has been on the forefront about asking about family history and asking about certain genetic conditions.

But I guess the complete awareness of what families or what patients may have had, certain types of cancers, which ones go together that could be in a particular cancer syndrome, if that's identified that could have great implications in terms of further screening, or medication management, or potentially prophylactic surgeries to improve the life of that woman or other family members, which may not just include women. Could include male members of the family.

Dr. Matt Birnholz:

I see. And in your opinion, do you think the OB/GYN in just a nationwide point of view is not involved enough in screening, history-taking, et cetera for hereditary cancer syndromes?

Dr. Andrew Wagner:

I think in general we're doing a good job, but it could always be better. I'd say most women come into an OB/GYN practice related to pregnancy. We're already family-focused. It's just I think going beyond what's initially the mom and the baby, but thinking about other family members. I mean, we're trained in medical school to ask a family history, and I remember being taught how to do pedigrees or how to draw a pedigree chart that detailed family history, but not really taught to do that in the typical history-taking.

When I did my genetics training, yes, that's a part of everything, so my brain is now wired that way. But I think thinking more about the family, and how to make connections, and how to put those together I think would greatly benefit at least in terms of what we're talking about here with the hereditary cancer syndromes.

Dr. Matt Birnholz:

Right, it makes sense. Why don't you take us through a little bit about the process of history-taking with this particular bent in mind?

Dr. Andrew Wagner:

So part of it is knowing what may be red flags in a family history, but also I guess before that is knowing what are the right questions to ask. So obviously we're getting detailed histories of the individual's own medical history and what their previous surgical history is, and then we're also using screening methods we may already have, which is breast exam, pelvic exam, or that we're talking about risk for

breast cancer or ovarian cancer, or endometrial cancer.

But then with the family history sometimes it's in a lot of places with electronic medical records there are different forms or click boxes that are done, and whether that's done by the nurse or the medical assistant or whether that's done by us the OB/GYN part of it is working through that system and not just, okay, we'll click that box or check that box if it's actually still on paper, then putting it into context.

And so making sure that we're asking questions about other family members with certain types of cancer. And I mean, we're going to be focusing on the gynecologic cancers, so endometrial ovarian cancer, cervical cancer a major player, but not typically genetic. Also breast cancer, because we're typically on the forefront of breast exam as part of an annual exam. As many OB/GYNs do take on the role as primary care providers we can't forget about colon cancer screening, so that's another important element, to ask whether we're the ones ordering colonoscopies in the appropriate patient or not.

Dr. Matt Birnholz:

And you're given specific attention within your lecture to the hereditary breast and ovarian cancers plus lymph syndrome.

Dr. Andrew Wagner:

Correct.

Dr. Matt Birnholz:

Any specific details that you could impart to our listener audience?

Dr. Andrew Wagner:

So hereditary breast and ovarian cancer syndrome which typically involves mutations in the genes called BRCA1, BRCA2, for the most part when somebody has a mutation in one of those genes they are at a greatly higher risk of developing breast or ovarian cancer. For breast cancer it's up to an 87 percent chance, ovarian cancer 27 to 44 percent chance. So with that knowledge screening or medication prophylaxis, or for some individuals prophylactic surgery, are warranted.

In terms of Lynch syndrome this overlaps with gynecologic cancers, primarily with endometrial cancer, which depending on who you read it's anywhere from about 40 to 70 percent lifetime risk with a mutation in one of four genes called MLH1, MSH2, MSH6, and PMS2. They all have weird names which are hard to remember. BRCA, a lot easier, B-R. breast, C-A, cancer.

Dr. Matt Birnholz:

BRCA, okay, right.

Dr. Andrew Wagner:

Exactly. But with a Lynch syndrome mutation they're saying increased risk for endometrial cancer, lifetime risk of 80 percent for colon cancer, and then what's actually number four on the list if you list all of the different cancers that could be involved with that is ovarian cancer, where there's up to about a 12 percent lifetime risk.

Dr. Matt Birnholz:

And just before we move on to your other topic of investigation, what about cancer panel testing? I know that that's an area that you've focused on specifically.

Dr. Andrew Wagner:

Part of the advances in that have been driven by advances in technology with what's called next generation sequencing, meaning that with a small amount of sample and a lot of bioinformatics we can sequence multiple genes much more rapidly and much more cheaply. So that's one of the drivers. The other driver with that is the previous patent that had been on BRCA1 and 2 testing, which was overturned by the Supreme Court last summer.

So with that all of the other labs besides the one that had the patent were able to test for the other genes that could be associated with for example breast cancer or ovarian cancer, but not BRCA1 and 2. And so as the technology was getting better they were developing panels, basically everything else but BRCA1 and 2. And then when the Supreme Court decision went down then BRCA 1 and 2 were opened up to everybody, so that was able to be part of a panel.

Advantage for the panel could be in those individuals where there is a very suspicious family history, meaning suspicious for some sort of hereditary predisposition. This is as opposed to a sporadic cancer, which is the majority of the time, and in which there is not one mutation in one gene getting passed on generation to generation. Also excluding it from familial types, which is multifactorial, a combination of genes and environmental factors influencing these higher numbers of cancers in a family.

So the hereditary type, basically if you're still thinking that and you've either previously done the just, let's say, BRCA1 and 2 testing, but

something else has got to be going on. This type of testing can look out for other genes or other hereditary cancer syndromes that you didn't test for already.

And for some families when they have wide-ranging types of cancers in a family that don't exactly follow a certain pattern, this could be a reasonable first step, and the way the price point has been right now depending on the lab about the same as BRCA1 and 2 testing alone. But also a thing with the panel testing and there being more companies available to do this, the price point is going down, which definitely benefits the patient, benefits the insurance companies.

Dr. Matt Birnholz:

Well, when you talk about insurance companies, price points, the immediate assumption that people are going to have is that those among other things represent some barriers to be able to enable more testing in practice, enabling more access for patients. Prices are coming down, as you say. But are there any other barriers outside of cost that obstruct clinicians' ability to be able to have more hereditary cancer screening and testing?

Dr. Andrew Wagner:

I think a big thing is the time to explain all of this and to take the detailed family history. How I was trained in my genetics fellowship is, we had specific sit-down appointments for this. I still have that in my practice. But to take a detailed family history and to do the appropriate genetic counseling, I don't know if appropriate's the right word, detailed, to explain everything fully is very difficult when you got a waiting room full of patients and you're not devoting an hour-long appointment for this. So that's one barrier.

Different insurance companies are covering this differently, Medicare has specific rules. For example, BRCA 1 and 2 you have to have had breast or ovarian cancer, and sometimes plus other family history criteria. But the Affordable Care Act actually has a point in there to make sure that BRCA testing is available for women who meet certain criteria, which I believe would be those as set up through a national comprehensive cancer network.

Dr. Matt Birnholz:

And I'm sticking on that point that you made about needing more time to be able to go through these tests. Obviously in general practice the first thing that is thought with that is that of course I don't have more time to do that. I have my, we like to say 15 minutes, but in fact it goes down to eight, nine, seven minutes per patient per visit. How does the OB/GYN incorporate this in practice when they have the same time pressures as a general practitioner in internal medicine or pediatrics?

Dr. Andrew Wagner:

Obviously education of the patient. This might mean that if there is some initial interest scheduling them back solely to discuss that, especially if maybe the initial entry point was an annual exam and if there aren't other chief complaints then you could use that as a focus for a follow-up visit. There are various handouts or websites that families can be forwarded so that they could read up on things.

But the thing you have to keep in mind, yes, it is a simple blood test, or for some labs swish and spit some mouthwash and send that off. But it's not so simple in terms of the implications. But I think another thing is also figuring that out in terms of lifelong care and making the family history more of a focus of how that woman's care will be.

Dr. Matt Birnholz:

Let's take us through the course of working with a patient on this. You've established a diagnosis, and we'll keep that open as to what the diagnosis might be. You had a separate appointment for educating the patient, and your practice is organized to the point where you can do that. What are the next steps for the OB/GYN's role in being able to follow up and keep up with this, as opposed to seeing that patient just float off to someone else?

Dr. Andrew Wagner:

I guess a way to not lose track of that patient and making sure you have the appropriate follow-up, okay, they decide to do the testing, it's positive, or negative. Okay, if it's positive then there are certain guidelines that are already put into place in terms of how to manage in terms of screening. So basically it's getting those ordered, following up with those. They're not usually just a one-time deal. We're talking about yearly mammograms or breast MRIs, at least in the case of BRCA1 and 2. Screening for ovarian cancer is controversial, but some individuals choose to do that.

So there's following up on that and what would be regularly-scheduled screens if somebody is, let's say, found to be BRCA or Lynch positive, and were concerned about risk for ovarian cancer, and they're not done having kids or they're not ready to even have kids, being on oral contraceptives could decrease the risk of ovarian cancer, so following that is an option.

If let's say that a woman is over 35 or 40, done having kids, maybe she already had tubal ligation done, then recommendations are quite strong for risk-reducing salpingo-oophorectomy in the BRCA positive individual and the Lynch syndrome positive individual with the risk

of endometrial cancer, then you're talking about hysterectomy as well as bilateral salpingo-oophorectomy. So in one sense that's your one intervention.

Then there's still the other organ systems that you need to follow up with, and hopefully that's something that the provider is able to continue with. But I would say some OB/GYNs might not be as comfortable with follow-up of ordering of colonoscopy, so that's where they would work with the woman's primary care provider, or with GI, or however the system may work.

Dr. Matt Birnholz:

Any wrap-up thoughts before we finish up with this topic?

Dr. Andrew Wagner:

I think it's just more knowledge and being aware of family history. And I guess one other point is, if somebody has a negative result you have to put that back into context of the family history and be able to use that so that then that individual doesn't get lost, because sometimes negative isn't always a negative. You still have to take into account the family history. Maybe somebody else it would be more appropriate for testing and figuring out how to have your patient get their family member who may live far away in to their doctor or figuring out other screening modalities.

Dr. Matt Birnholz:

I'm going to have to quote you on that sometime. Sometimes a negative isn't always a negative.

Dr. Andrew Wagner:

Sure.

Dr. Matt Birnholz:

That's a great anecdote. And with that in mind I'd like to thank Dr. Andrew Wagner, Associate Professor of OB/GYN at the University of Oklahoma Health Science Center. Again Dr. Wagner, thanks for your time.

Dr. Andrew Wagner:

You're very welcome.

Dr. Matt Birnholz:

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