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Should You Be Looking at NIPT & Carrier Screening Differently?

Welcome to ReachMD. The following program: “Should You Be Looking at NIPT & Carrier Screening Differently?” is sponsored by Roche Diagnostics.

This program is intended for general physicians, OBGYNs, Nurse Practitioners, Midwives, and other prescribers that offer genetic testing for pregnant women.

Dr. Setty: There are numerous options for pregnancy screening, making the decision of what type of genetic testing to offer our patients more complex. On today’s program, we will discuss the most important factors for healthcare professionals to consider when screening their pregnant patients. This is ReachMD and I am Dr. Prathima Setty. Joining me today is Dr. Justin Brewer, practicing OB/GYN and Maternal Fetal Medicine Specialist at North Mississippi Medical Center in Tupelo, Mississippi. Dr. Brewer is a professional member of the American College of Obstetrics and Gynecology, the Society of Maternal Fetal Medicine, the American Institute for Ultrasound and Medicine and the Mississippi State Medical Association. Dr. Brewer thank you so much for being here today.

Dr. Brewer: Oh, thank you so much for having me.

Dr. Setty: Let’s start with a question that many of us in obstetrics need to field. What do you tell your referral base when they ask if they should be ordering expanded panels for noninvasive prenatal testing and carrier screening?

Dr. Brewer: This is an excellent, very common question. You know, any time I have asked this question I always try to stick with the recommendations that our major societies have set out for us such as ACOG and the Society for Maternal Fetal Medicine. In my clinic, I use NIPT as a screening test for trisomy 13, 18 and 21 only. There are going to be some rare instances where I may also use it to look for perhaps a 22q11 microdeletion, but that has only occurred a time or two. In general, if I am concerned enough about testing for some type of microdeletion, then I am really going to be talking to my family about diagnostic testing options such as an amniocentesis with a micro array. I think it is always important to stress the fact that NIPT is a screening test only. It does not replace the diagnostic testing options that we have available such as an amniocentesis or a CBS. We also have to remember that adding some of these very rare conditions can lower our positive predicted value substantially. Also, the key to testing is to be able to take that information and use it in a meaningful way to help the patient. Patients need to have an informed idea about what they are being tested for and how we may or may not be able to use that information. You know, the old saying holds true that just because you can test for something does not always mean that you should.

Dr. Setty: Professional societies don’t endorse adding microdeletions to NIPT due to the lack of data. Is this a concern for you as well and are there any other issues that arise from this?

Dr. Brewer: So, the lack of robust data is a huge concern of mine. You know, without good data we are left wondering about the sensitivity and specificity of these tests and how these tests are going to perform in a real-world clinical situation. That really makes it almost impossible to counsel these families appropriately about these tests when they are trying to figure out what testing scheme is right for them. I also worry about the cost to families. Many of these microdeletions are going to be extremely rare and thus you would have to test a large number of patients to actually find an affected fetus. This can add a good bit of cost of these tests, possibly without really improving or altering the care of the majority of the pregnancies that you are going to test.

Dr. Setty: So, are there instances when you would choose the basic panel over the expanded carrier panel and vice versa?

Dr. Brewer: I can certainly see sending an expanded panel in some cases where I feel like there is a high likelihood of finding a disorder that is not found on the basic panel. Now, when we are speaking about a basic panel, we are really talking about those 22 or 23

disorders that our major societies such as ACOG feel are the most prevalent and ones that we ought to be offering to all of our patients. The situations that I am thinking about or perhaps when you are testing a specific ethnic group or geographic group of patients or perhaps in those cases of consanguinity; that said, when you start to look for extremely rare conditions in a general population, you increase the chances of finding a carrier or patient, however, you have to remember that the chance for the partner to also be a carrier for such a rare condition remains extremely low. This can lead to greatly increased patient anxiety and costs without actually improving patient care in the end. Again, these are all important points that need to be discussed with the family from the beginning so that they are able to make a truly informed decision about what testing scheme is going to be right for them. Also, I feel that this is a place where having access to a genetic counselor is going to be extremely important.

Dr. Setty: That is a very good point Dr. Brewer. If you are just tuning in, this is ReachMD, and I am your host Dr. Prathima Setty. I'm speaking with OB/GYN and Maternal Fetal Medicine Specialist, Dr. Justin Brewer, on the topic of expanding genetic testing panels for pregnant women. What are your priority considerations when deciding what to order for carrier screening and NIPT?

Dr. Brewer: So, I always try to follow those society guidelines that have been outlined in the past, but I also let family wishes help guide me in this decision. This is where in-depth patient counseling is going to be crucial. You know, patients need to have a good understanding about what they are electing to be tested for and how we may use this information or may not be able to use this information to impact their pregnancies. They also need to be counseled about the particular tests that we may be considering, including detection rate and overall performance. Again, I am a strong believer in making use of a genetic counselor if one is available. I feel that a genetic counselor can really help ensure that these patients are able to make a truly informed decision about their case.

Dr. Setty: Dr. Brewer, you just shared some considerations when deciding what to order for carrier screening and NIPT. What factors influence your decision when choosing a particular company or test to offer your patients?

Dr. Brewer: First and foremost is going to be the performance of the test. Obviously, I want to choose a test that has robust outcome data on the large number of pregnancies. Preferably, I would like to have this data generated from independent sources and if possible to have come from prospective trials. The reason this is so important is simple. I want to know how this test is going to perform in everyday clinical situations. When your test has been studied in a large number of pregnancies in different situations, you are really able to have a more accurate idea of how to expect this test to behave under real-world conditions when you are testing "all comers group" under differing scenarios. This gives me more confidence when I am offering a test that can be trusted to perform as expected in my patient population. The cost of the test and the way that test is going to be covered by the patient's insurance plan is also very important. Obviously, there is no reason to offer a patient a test that they simply cannot afford, and then finally a reasonable turnaround time is very important. You know, these patients are going to be extremely nervous until they get the results back, so I want to try to keep that waiting time to a minimum if possible.

Dr. Setty: Is there a situation when you would consider adding additional microdeletions when ordering NIPT?

Dr. Brewer: I view NIPT as an extremely good screening tool for trisomy 13, 18 and 21. Now, that said, there have been some rare cases when I have had mothers who were known to have DiGeorge syndrome and we also knew that their fetus has cardiac defects; however, for various reasons, the patients were not amenable to a diagnostic testing option such as an amniocentesis. You know, in those cases we were actually able to use NIPT to help confirm a suspected diagnosis prenatally that otherwise we would have had to have waited until after delivery to confirm. I still, though, need to stress the idea that when we are dealing with a fetus with a known defect of some type, these patients are going to be best served with diagnostic testing options such as amino as opposed to NIPT in the vast majority of situations.

Dr. Setty: Well, with that I want to thank Dr. Brewer for joining me today to discuss expanded genetic testing panels for pregnant women. Dr. Brewer thanks for joining us.

Dr. Brewer: Thank you so much for having me. These are really important questions that I know we are faced with on a daily basis in our clinics and I hope that we have been able to answer some of these questions for our listeners.

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