Transcript Details
This is a transcript of a continuing medical education (CME) activity. Additional media formats for the activity and full activity details (including sponsor and supporter, disclosures, and instructions for claiming credit) are available by visiting:
https://reachmd.com/programs/cme/nipt-counseling-strategies-considering-test-reporting-results/11688/

Released: 06/30/2020
Valid until: 01/31/2022
Time needed to complete: 15 mintues

ReachMD
www.reachmd.com
info@reachmd.com
(866) 423-7849

NIPT Counseling Strategies: Considering the Test, Reporting the Results

Announcer:
Welcome to CME on ReachMD. This activity, entitled “NIPT Counseling Strategies: Considering the Test, Reporting the Results” is provided by Omnia Education and is supported by an independent educational grant from Illumina.

Prior to beginning the activity, please be sure to review the faculty and commercial support disclosure statements as well as the Learning Objectives.

Dr. Dugoff:
Community OB/GYNS have an obligation to discuss and offer prenatal genetic screening and diagnostic testing options to all pregnant patients regardless of their age or their underlying risk of a chromosomal abnormality. The majority of women initially opt for genetic screening tests. Prenatal genetic screening tests include cell-free DNA screening which is also referred to as noninvasive prenatal screening or NIPS or noninvasive prenatal testing NIPT and serum screening is another option with or without ultrasound measurement of fetal nuchal translucency.

Cell-free DNA is the most sensitive and specific screening test for trisomy 21 and trisomies 13 and 18. Cell-free DNA screening can be performed as early as 10 weeks gestation. It can detect more than 99% of cases of trisomy 21 and 13, 98% of cases of trisomy 18, with a false positive rate of 0.13% when test results are obtained. Cell-free DNA is the only test that can screen for fetal sex and sex chromosome aneuploidies.

First-trimester screening which is performed between 10 and 13 weeks and 6 days gestation includes measurement of serum analytes and the fetal nuchal translucency. The detection rate for trisomy 21 is 85% with a 5% false-positive rate. The quadruple marker screen (quad screen) which can be performed between 15 weeks and 22 weeks and 6 days gestation has a detection rate of 80% for trisomy 21 with a 5% false positive rate. Sequential and contingent screens involve a combination of first and second trimester screening. The detection rates range from approximately 90-95% with a 5% false positive rate.

Patients should have pretest counseling so that they can make informed choices regarding screening and diagnostic testing. Patients should also have posttest counseling. In this activity we will be demonstrating counseling strategies for discussing NIPT with our patients. The first segment will focus on a discussion with a woman who is considering whether or not to undergo NIPT. The second will address means to discuss with your patient the outcomes from an NIPT, whether it is negative, positive, or a “no-call” result.

VIGNETTE 1

Dr. Dugoff:
Hi, Jen.

Jen:
Hi, Dr. Dugoff. How are you today?

Dr. Dugoff:
I’m doing great. How are you doing?

Jen:
Good, thanks.

Dr. Dugoff:
Good. Well, I know you're coming in later this week for an ultrasound, and so I think today we were planning on talking about different prenatal diagnosis screening and testing options so that you'll have a better idea maybe what you'd like to do when you come in and have your ultrasound.

Jen:
Yeah, you know, I've heard about a couple of different blood tests and maybe some other tests that are a bit more involved, and I just need to figure out what we want to do.

Dr. Dugoff:
Great, okay. Well, first of all, maybe it would help to start just talking about the difference between screening and diagnostic testing. So the screening tests that are available to you, really one consists of simply purely just of a blood test, and the other one is a blood test and an ultrasound, and then whereas with diagnostic tests, that would... Have you heard of CVS, chorionic villus sampling?

Jen:
Yeah, I've heard of that and the other one with the amnio or something along those lines.

Dr. Dugoff:
Yes. And so, with both of those, we're not drawing a blood sample, but with those we're actually sampling fetal—or with CVS, in that case we're sampling placental tissue, so we are actually getting tissue that is reflective of the baby's chromosome makeup and sending that tissue to the lab, so we're actually getting results that tell us about the baby's genetic makeup, whereas with the screening tests, they are giving us an idea what the likelihood is that a baby could have Down syndrome or another chromosomal abnormality. And then, in general, if a screening test is positive, then we would recommend that a patient have a diagnostic test to see if a baby truly has a chromosome problem or not.

Jen:
Okay.

Dr. Dugoff:
Does that make sense?

Jen:
Yeah, so you can do one and then the other if that's what we choose to do, like the blood test first?

Dr. Dugoff:
You can, yeah; you definitely can. A number of patients choose to have a blood test first as opposed to having a diagnostic test first just because with diagnostic tests there is—it's very small, but there is a chance that you could lose a pregnancy as a result of a complication of having the diagnostic test.

Jen:
Sure, okay. So, what are the best blood tests for us to think about if that's what we wanted to do first?

Dr. Dugoff:
So there are 2 different options. One of them is called—and some people call it NIPT or NIPS or cell-free DNA screening. I don't know if you're familiar with any of those terms.

Jen:
It sounds a little bit familiar, but I don't really know what they mean.

Dr. Dugoff:
Okay, yeah, so NIPS is noninvasive prenatal screening. NIPT is noninvasive prenatal testing. I tend to call it cell-free DNA screening just because the lab is actually isolating cell-free DNA from your blood. It's placental tissue, basically. And by analyzing that they can screen for Down syndrome as well as some other chromosomal abnormalities. Well, I'll start with that one. So, with cell-free DNA screening, again, it's one blood draw, and we do it any time after 10 weeks gestation. So you're going to be, I think, 12 weeks gestation tomorrow, or later this week, so you're within the range. With cell-free DNA, it screens for Down syndrome, which is trisomy 21, as well as a couple other serious chromosomal abnormalities called trisomy 13 and trisomy 18. Have you heard anything about the detection or how accurate this test is?

Jen:
I've heard from a few friends that have had it that they were told it was a very accurate test, but I haven't heard any actual numbers.

Dr. Dugoff:
Yeah, so it detects—actually detects over 99% of cases of Down syndrome, so it's really an excellent screening test.

Jen:
And the other conditions, is it... I can't remember what you called them, but is it the same? Is it as good for those other things it tests for?

Dr. Dugoff:
It's not quite as good, but it's still really, really pretty good, over 90% of cases of trisomy 18 and a very high proportion of cases of trisomy 13 as well, yeah. And then also, depending if you're interested, with cell-free DNA we can also screen for disorders called sex chromosome aneuploidy or something called Turner syndrome where there's an X chromosome missing in a female or other conditions where there's either an additional X or Y chromosome present.

I know you were really interested in talking about especially screening for Down syndrome, and so I wanted to talk to you. The other test we have is called a first-trimester screen. You can also have a sequential screen where we draw two blood draws, one in the first trimester and one in the second trimester, and with a sequential screen, on the ultrasound we would measure something called a nuchal translucency, which is the thickness of the tissue in the back of the baby's neck, and it turns out that that area is thickened in babies that have Down syndrome as well as other chromosomal abnormalities. And so we would send the measurement, that number of the measurement, to the lab as well as your blood. With that test, with the first trimester part of that test, it picks up about 80–85% of cases of Down syndrome.

Jen:
Okay, so that blood test is different from the other one.

Dr. Dugoff:
It is. So the detection rate is a little bit lower, and then the false-positive rate, which is the chance that if the test comes back positive, it's the chance that your baby really doesn't have Down syndrome, it's 5% of that test, whereas it's 0.2% with cell-free DNA screening.

Dr. Dugoff:
With the second test we would measure the nuchal translucency on ultrasound. Since you're coming for an ultrasound anyway, we wouldn't need any of the ultrasound information for the cell-free DNA test; although, if we saw a known abnormality on that test, we would offer you a CVS. If it looked like there was a large nuchal translucency or another structural abnormality on ultrasound, we would talk to you more about doing chorionic villus sampling, which is a diagnostic test.

Jen:
Okay. Well, thank you. I think that answers a lot of the questions that we had before this appointment coming up.

Dr. Dugoff:
Okay, great. And then the last thing I wanted to mention is—just so you know that this is something that could happen if you chose to have the cell-free DNA screening—is that there is a small chance that the test would come back and you wouldn't get a result with it. With both tests you can get a screen negative result, a screen positive result, meaning there's an increased chance the baby could have Down syndrome or another chromosome problem. And with the cell-free DNA test though, there is a small chance that we would not get a result, and then if that were the case, we would counsel you a little bit more about your options.

Jen:
Okay. All right. Well, thank you.

Dr. Dugoff:
Okay, that's great.

VIGNETTE 2

Announcer:
Calling your patient after NIPT screening. Positive test results.

Dr. Dugoff:
Hi, Jen.

Jen:
Hi, Dr. Dugoff. How are you today?

Dr. Dugoff:
I'm calling today because I wanted to give you the results of your cell-free DNA test.

Jen:
Okay.

Dr. Dugoff:
So, it did come back positive for trisomy 21, which is Down syndrome. And as we talked about the other day—I don't know if you remember this—but that does not mean that your baby has Down syndrome.

Dr. Dugoff:
I know this isn't what you were expecting, and I don't know if you remember about our conversation about the positive-predictive value.

Jen:
Not exactly.

Dr. Dugoff:
Right, so the positive-predictive value is the chance when you have a positive test that your baby truly does have Down syndrome, so in your case it's 79% chance, so that also means there's a 21% chance that even though the test result is positive, there's a 21% chance or about a 1:5 chance that your baby does not have Down syndrome.

Jen:
Okay, so about a 79% chance that it is Down syndrome.

Dr. Dugoff:
Yeah, and so I think the next step... I know you had an appointment for later this week, and the next step is to come in, and we can talk a little bit more about different options. The last time we talked a little bit about diagnostic testing.

Dr. Dugoff:
And that is certainly something we'll talk more about—CVS as well as amniocentesis—when you come in later this week.

Jen:
Okay. Okay, that sounds good. And I can bring my husband, and we can just talk through all the pluses and minuses to those tests, I guess.

Dr. Dugoff:
Absolutely, yes.

Jen:
Okay. Okay, well, thank you for—thank you for letting me know, and I guess we'll just kind of take the next step.

Announcer:
Calling your patient after NIPT screening. Negative test results.

Dr. Dugoff:
Hi, Jen, it's Dr. Dugoff.

Jen:
Hi, Dr. Dugoff. How are you doing today?

Dr. Dugoff:
Well, I'm calling to give you your cell-free DNA result. I have good news for you today.

Jen:
Oh, great, I've been waiting for your call.

Dr. Dugoff:
Yeah, so the test came back screen negative.

Dr. Dugoff:

So, as you might remember, so we tested for Down syndrome as well as trisomy 18 and 13, and you also chose to have the test to see if there was an extra or missing X chromosome or an extra Y chromosome, so all of those tests came back negative. What that means is there's a very, very low likelihood that the baby has any of those conditions. It's not zero, but it's very, very low.

Jen:
Oh, great. That's such good news. That's very reassuring.

Dr. Dugoff:
Yeah, it's great news. I know you're coming in for your anatomy scan, so we look forward to seeing you for that. And this is great news. I did want to remind you that with this specifically screening for Down syndrome and trisomy 18 and 13, it doesn't mean the baby doesn't have any chromosomal abnormality or any genetic disorder, but it's very reassuring.

Jen:
Okay, great. Well, I'll take the good news, so thank you so much.

Dr. Dugoff:
Okay. All right, I'll look forward to seeing you when you come in.

Announcer:
Calling your patient after NIPT screening. No-call result.

Dr. Dugoff:
Hi, Jen. It's Dr. Dugoff.

Jen:
Hi, Dr. Dugoff. How are you?

Dr. Dugoff:
Good. Well, I know you've been waiting for your cell-free DNA test result, so that's why I'm calling you today.

Jen:
Oh, good. Great.

Dr. Dugoff:
I don't know if you remember we talked about like in rare cases patients get a no—what I call a no-call result or they don't get a result back with the test.

Jen:
Right.

Dr. Dugoff:
And your test result came back as a no-call result.

Dr. Dugoff:
Yeah, so that definitely doesn't mean your baby has Down syndrome or any other chromosomal abnormality or any other abnormality. And I know you're coming in later this week. We can talk more about it in person, but we certainly want to talk—we'll talk more about prenatal diagnosis options. You may want to end up having chorionic villus sampling or amniocentesis or a diagnostic test, and we can also talk about repeating—the pros and cons about repeating the cell-free DNA test.

Jen:
Oh, okay, so we have a couple of different things we can do to try to get a result?

Dr. Dugoff:
We do, yeah, and we'll talk a little bit about more—about the significance when you do get a no-call result.

Dr. Dugoff:
But it definitely doesn't mean your baby has Down syndrome.

Jen:
Okay. Well, I guess we'll just kind of talk a little bit about what our options are and try again to figure out what's going on.

Dr. Dugoff:
Okay. All right, I'll look forward to seeing you when you come in.

Announcer:
You have been listening to CME on ReachMD. This activity is provided by Omnia Education and is supported by an independent educational grant from Illumina.

To receive your free CME credit, or to download this activity, go to ReachMD.com/Omnia. Thank you for listening.