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TRANSCRIPT

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ReachMD

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

NIPT: Tips for Counseling Your Patient

Announcer: Welcome to ReachMD. This medical industry feature, titled "NIPT: Tips for Counseling your Patient" is sponsored by Roche. This program is intended for healthcare providers. Here's your guest, Ms. Kelly Chen, Senior Genetic Counselor, Roche Sequencing Solutions, San Jose, California.

Kelly Chen: The subject of non-invasive prenatal testing, or NIPT, is coming up more and more often in conversations with patients, which creates a high demand for those in clinical practice to provide timely, accurate, and practical information. Today I'm going to share some counseling tips I hope you'll find useful to provide patients more clarity and reassurance.

A positive way to start the conversation is to encourage patient decision-making by exploring values and reproductive goals for each person. Understanding these goals can help clarify which testing is most appropriate. Giving patients the choice to accept or decline genetic testing can reduce stress and empower them early in their pregnancy journey.

Let's focus on the Harmony prenatal test, which screens for Down syndrome, trisomy 18, and trisomy 13. This test was developed to be an accurate prenatal screening test for Down syndrome compared to traditional screening. It's important to recognize that no screening test is designed to detect all possible conditions or to provide a diagnostic result. When counseling patients, including the words "probability" or "chance" and naming the specific conditions screened for when discussing NIPT can help set their expectations for results.

There are three potential results for this test that can be reviewed with patients BEFORE testing is done.

The first is a low probability result, meaning there's a low chance for the conditions screened.

The second is a high probability result, meaning there's a higher chance than expected for a certain condition. Additional testing during the pregnancy and/or evaluation after delivery may be recommended for confirmation if the patient desires.

And third, there may be no result reported. Occasionally, a result can't be obtained for one or more conditions tested. One common reason is not having enough DNA from the pregnancy in the blood sample to obtain an accurate result.

So let's focus on factors to consider, when reviewing an NIPT result. Two numbers to note are the fetal fraction and the probability score.
(1) The fetal fraction is an estimate of the percentage of DNA from the pregnancy in a mother's blood sample. The minimum fetal fraction needed for analysis is 4 percent. Fetal fraction increases with gestation and decreases with higher maternal weight.
(2) The probability score is the probability that the cell-free DNA sample analyzed is consistent with trisomy, and SHOULD NOT be interpreted as the probability that the *fetus* is affected. When explaining a low probability score to patients, this could be stated as "The chance of trisomy 21 in your sample is low."

These are all considerations to keep in mind when counseling patients about NIPT. I hope they'll be helpful to enrich the patient conversations in your practice.

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