How to Decide About Prenatal Genetic Testing

There are many prenatal genetic testing options available today that provide information about genetic conditions or birth defects in your baby. Prenatal genetic tests can be put into two categories:

1) **Diagnostic tests** such as amniocentesis and chorionic villus sampling (CVS) can provide definitive “yes” or “no” results. However, these tests involve some risk, including a chance of miscarriage.

2) **Screening tests** include blood tests, such as traditional maternal serum screening (quad screen, triple screen, first trimester screen, integrated screen, and sequential screen) and prenatal cfDNA screening, and ultrasound. In general, screening tests pose no risk to the baby; however, they only tell you if the chance of a genetic condition or birth defect is higher or lower and do not give you “yes” or “no” answers.

Prenatal genetic testing is your choice. Some women would prefer “yes” or “no” answers, and since screening tests cannot provide that they may choose to go straight to a more definitive test, such as CVS or amniocentesis. Or, they may consider a diagnostic test like CVS or amniocentesis after having a screening test that suggests a higher chance of a genetic condition in their baby.
Decisions About Diagnostic Tests Such as Amniocentesis and CVS

Questions to consider regarding diagnostic testing include:

- If your baby had a genetic condition, would you want to know before birth?
  - For example, some women would want to know if their baby had a genetic condition or birth defect because they would want to be able to prepare before delivery. Some women would consider making an adoption plan for their baby, and some would consider ending the pregnancy if they knew their baby had a genetic condition.
- Do you feel like “yes” or “no” answers would help you feel less worried?
- Are you comfortable with the risk of miscarriage associated with these procedures (less than 1%)?
- Have you thought about these two scenarios?
  - You don’t have the diagnostic test and you find out at birth that the baby has a genetic condition, versus
  - You do have the diagnostic test and you miscarry a pregnancy that did not have a detectable genetic or chromosome condition

Both situations would be difficult, but which scenario is more acceptable to you?
Decisions About Screening Tests Such as Maternal Serum Screening and cfDNA Screening

Questions to consider regarding screening tests such as traditional maternal serum screening or cfDNA screening include:

- How would you feel if results indicated a higher chance for a genetic condition or birth defect?
- Would you consider a diagnostic test if a screening test indicated an increased chance for a genetic condition?
  - If not, would you be ok waiting until the baby is born to know for sure if the condition is present?
- Do you think this information would help you feel more prepared?
- Does more information with the possibility of uncertainty make you anxious?

Decisions About Not Having Testing

Some women feel that prenatal genetic testing is not right for them. Consider these questions:

- Would you prefer not to face the decision of whether or not to have a diagnostic test if your screening test came back with a result showing your baby has a higher chance of having a genetic condition?
- Are you confident that, even if the baby did have a genetic condition, it wouldn’t change your pregnancy plans?

Making a Decision That is Right for You...

There are many prenatal tests available, and your healthcare professional will be able to determine which prenatal testing options are the most appropriate to offer you. The decision
to undergo prenatal genetic testing is personal and should reflect your values, personality, beliefs, and needs.

Your decision may be very different than the path your friend or neighbor might take… and that's ok. In most cases, prenatal genetic testing is a choice and the best way to make the decisions that are right for you is to be informed.

You can find more information and videos about prenatal genetic testing options at: