Non-Invasive Prenatal Testing (NIPT)

For trisomy 21, 18, 13 (+/- sex chromosome differences)

Key Points

- NIPT is a screening test — it is not diagnostic.
- NIPT can be used to screen for trisomy 21, 18, and 13 (with the option of sex chromosome differences).
- Diagnostic testing can be considered in the context of a high risk result.

Screening Results & Discussion Points

Screening Result	Discussion Points
High Risk	• This typically means the chance for trisomy 18, 13, or a sex chromosome difference is significantly increased. • The chance that a pregnancy with a high risk screening result truly has the chromosome difference varies by chromosome and the risk prior to testing. • A referral for genetic counselling should be offered. • Only diagnostic testing (chorionic villus sampling or amniocentesis) can provide a definitive “yes” or “no” answer.
Low Risk	• This typically means the chance for the tested conditions is low. • A low risk screening result is reassuring, but the residual risk depends on the reason for testing. • A low risk screening result does not guarantee that the fetus does not have a chromosome difference or health concern. • A low risk screening result would not prompt the offer of diagnostic testing.

“No Call” or Failed Results	• Clinical factors that increase the chance for a “no call” result include:
	  ◦ NIPT blood drawn too early
	  ◦ Higher body weight
	  ◦ Carrying a twin pregnancy
	• A “no call” result is associated with a higher chance for a fetal chromosome difference.
	• Repeating the bloodwork (i.e. redraw) will yield a result in most cases.
	• Options that can be considered after failed NIPT include:
	  ◦ Repeat NIPT
	  ◦ Alternative screening test
	  ◦ Detailed anatomy ultrasound
	  ◦ Referral for genetic counselling to include a discussion about diagnostic testing