

HOW TO DISCUSS PRENATAL SCREENING RESULTS

KEY POINTS

- NIPT is a screening test, it is not diagnostic
- NIPT screens for trisomy 21, 18, 13 (+/- sex chromosome differences)
- Invasive diagnostic testing should be considered in the context of a high risk NIPT

High Risk Screening Results

- This typically means the chance for trisomy 21, 18,13 is significantly increased
- The chance that a high risk screen result truly represents an affected pregnancy varies by chromosome and the pregnant person's age
- A referral for genetic counselling should be offered
- NIPT is a screening test - only invasive diagnostic testing (chorionic villus sampling or amniocentesis) can provide a diagnosis

"No call" or Failed Results

- There are different reasons why NIPT fails such as not enough fetal/placental DNA (due to high maternal weight or blood drawn too early), chromosome difference in mother or baby
- Repeating the bloodwork (i.e. redraw) will yield a result in most cases

NON-INVASIVE PRENATAL TESTING (NIPT)

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



Low Risk Screening Results

- This typically means the chance for trisomy 21, 18, 13 is <1:10,000
- A low risk screen result is reassuring, but this depends on the indication for testing
- A low risk screen result does not guarantee the birth of a baby without any health concerns or other genetic conditions
- A low risk screen result would not prompt the offer of invasive diagnostic testing