# HOW TO DISCUSS PRENATAL GENETIC SCREENING RESULTS

## KEY POINTS

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

### High Risk Screening Results

- This typically means the chance for trisomy 21, 18,13 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.
- NIPT is a screening test - only invasive diagnostic testing (chorionic villus sampling or amniocentesis) can provide a diagnosis.

### Low Risk Screening Results

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

## NON-INVASIVE PRENATAL TESTING (NIPT)

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

### "No call" or Failed Results

- Clinical factors that increase the chance for a "no call" result include: NIPT blood drawn too early, high maternal weight, and 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 and referral for genetic counselling to include a discussion about invasive testing.