

# 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
 <b>High Risk</b>	<ul style="list-style-type: none"><li>• This typically means the chance for trisomy 18, 13, or a sex chromosome difference is significantly increased.</li><li>• 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.</li><li>• A referral for genetic counselling should be offered.</li><li>• Only diagnostic testing (chorionic villus sampling or amniocentesis) can provide a definitive “yes” or “no” answer.</li></ul>
 <b>Low Risk</b>	<ul style="list-style-type: none"><li>• This typically means the chance for the tested conditions is low.</li><li>• A low risk screening result is reassuring, but the residual risk depends on the reason for testing.</li><li>• A low risk screening result does not guarantee that the fetus does not have a chromosome difference or health concern.</li><li>• A low risk screening result would not prompt the offer of diagnostic testing.</li></ul>
<b>“No Call” or Failed Results</b>	<ul style="list-style-type: none"><li>• Clinical factors that increase the chance for a “no call” result include:<ul style="list-style-type: none"><li>◦ NIPT blood drawn too early</li><li>◦ Higher body weight</li><li>◦ Carrying a twin pregnancy</li></ul></li><li>• A “no call” result is associated with a higher chance for a fetal chromosome difference.</li><li>• Repeating the bloodwork (i.e. redraw) will yield a result in most cases.</li><li>• Options that can be considered after failed NIPT include:<ul style="list-style-type: none"><li>◦ Repeat NIPT</li><li>◦ Alternative screening test</li><li>◦ Detailed anatomy ultrasound</li><li>◦ Referral for genetic counselling to include a discussion about diagnostic testing</li></ul></li></ul>