



## Non-Invasive Prenatal Screening (NIPS) Comparison of NIPS and Amniocentesis

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NIPS is a blood test for the detection of Down syndrome (trisomy 21), trisomy 18, and trisomy 13 based on testing the cell-free fetal DNA circulating in maternal blood. **NIPS is a funded benefit for women at high risk for trisomies based on one of the following:** a positive screen for Down syndrome or trisomy 18; a previous pregnancy affected with trisomy 13, 18 or 21; or specific ultrasound findings indicating an increased risk of Down syndrome or trisomy 18. For additional Information about funded NIPS.

Some pregnant women may quality for both NIPS and amniocentesis. This table compares both options and was created to facilitate counselling of these women.

Comparison of NIPS and Amniocentesis	NIPS Blood Test	Amniocentesis
<b>Detection Rate</b> Down syndrome (Trisomy 21)	~ 98%*	100%
Trisomy 18	~ 85%*	100%
Trisomy 13	~ 92%*	100%
Sex aneuploidy	96% for 45,X (Turner syndrome)**	100%
Other Chromosome Abnormalities	0	Depends on test done <sup>†</sup>
False Positive Rate (FPR)	<0.1% for T21, T18, T13*** 1% for sex aneuploidy	0
Positive Predictive Value	>95% in high-risk pregnancies (20-80% in low-risk depending on chromosome involved and maternal age)	100%
Risk to Pregnancy	0	1 in 200 pregnancy loss
Failure Rate	~ 2% on first blood draw	< 1 in 1000
Result Turn-Around Time	Approx.8 business days	3-14 days depending on test done <sup>†#</sup>

<sup>\*</sup> Based on PGSP data that includes 229 cases of Trisomy 21, 45 cases of Trisomy 18 and 12 cases of Trisomy 13.

<sup>\*\*</sup> Based on published data for 45,X; Limited data available on detection rates for 47,XXY, 47,XXX, and 47,XYY.

<sup>\*\*\*</sup> Given FPR, women with positive NIPS results are advised to have amniocentesis for diagnostic confirmation.

<sup>†</sup> When amniocentesis is done for a positive screen, only rapid aneuploidy testing is done (BCCH, RCH). If procedure done for a fetal abnormality, a microarray analysis will be done.

<sup>#</sup> If amniocentesis in Victoria, women <20 wks GA and risk <5%, full karyotype done with results ~ 14 days. If GA >20 wks or risk >5%, rapid FISH done with results ~ 3 days.