NIPT
Translocation screening with *percept*

What are chromosome translocations?

A balanced reciprocal translocation is a two-way exchange of genetic material between chromosomes. There is usually no net loss or gain of genetic material. About 1 in 500[1] people are carriers for these translocations.

How do these translocations affect reproductive health?

Carriers of balanced reciprocal translocations may experience reduced fertility and multiple miscarriages as a result of passing on an unbalanced form of the chromosome translocation during conception. Unlike a balanced translocation, an unbalanced translocation leads to a loss and/or gain of genetic material.

A pregnancy carrying an unbalanced translocation may show fetal anomalies. A child born with an unbalanced translocation can have a range of physical and/or learning disabilities.

Can *percept* be used to screen pregnancies of translocation carriers?

Yes, *percept* NIPT offers a rapid, accurate and non-invasive option for balanced translocation carriers. Pregnancies are screened for unbalanced translocations and other chromosome conditions.

Our data shows that *percept* is an accurate way to identify pregnancies with an unbalanced translocation and is valued by carriers as a non-invasive screening option[2,3]. *percept* is the only NIPT with a dedicated service for known translocation carriers.

[1] *Jacobs, Patricia A., et al. Estimates of the frequency of chromosome abnormalities detectable in unselected newborns using moderate levels of banding. J Med Genet. 29.2 (1992): 103-108.*

[2] *Flowers, Nicola Jane, et al. Genome-wide noninvasive prenatal screening for carriers of balanced reciprocal translocations. Genet Med 22.12 (2020): 1944-1955.*

[3] *Cifuentes Ochoa M, et al. 'I Could Trust It': Experiences of Reciprocal Translocation Carriers and Their Partners With Prenatal Cell-Free DNA Screening for Unbalanced Translocations. Prenat Diagn. 2025 Feb;45(2):155-162.*

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NIPT
How to arrange screening

Pre-approval:
Our laboratory needs to review the history of a translocation carrier before we can offer screening.

We need:

- A chromosome report (G-banded karyotype) with balanced translocation details.
- If available, a chromosome microarray report if an unbalanced translocation was found in a previous pregnancy.
- And a description of how the translocation was identified in the family, if known.

***Note**: Carriers of Robertsonian translocations do not need pre-approval.*

Step 1 

Email our lab with the pre-approval information: perceptNIPT@vcgs.org.au
In some cases, we may suggest that prenatal diagnosis is more appropriate than NIPT.

Step 2 

Arrange a dating ultrasound. Ultrasound must confirm a singleton pregnancy. Translocation screening is not available in a twin or co-twin demise pregnancy.

Step 3 

Complete a *percept* NIPT test request form.
Please record the balanced translocation on the form.

Step 4 

Direct your patient to a VCGS affiliated collection centre. Blood must be collected at, or after 11 weeks of gestation.

Our team is here to support you and your patients through screening.
P) 03 9936 6402
E) reproductivegc@vcgs.org.au

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