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 500¹ 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.

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.



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.

NIPT How to arrange screening

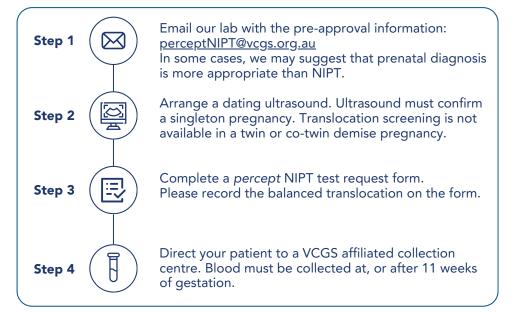
Pre-approval:

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.



Our team is here to support you and your patients through screening. **P**) 03 9936 6402

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