PGD TRS™ preimplantation diagnosis allows examining the cells collected from embryos for the presence of abnormalities resulting from balanced reciprocal translocations in parents. This testing technique is adapted to the needs of a given patient.

Chances for pregnancy with the use of PGD TRS™

Translocation means the movement of a fragment of genetic material from one chromosome to another. Most often, the altered arrangement of the genetic material on chromosomes is inherited from one of the parents.

In the case of balanced translocation, the genetic material is not lost or in excess, but only has altered location, e.g. fragments of two or more chromosomes change places, or chromosomes join each other. A carrier of a balanced translocation produces abnormal reproductive cells, thus preventing pregnancy, causing miscarriages or disease syndromes with chromosome abnormalities as the underlying cause.

One of 500 individuals carries chromosomal translocations

Chances of healthy pregnancy in a female carrier without PGD TRS™: 5-30%

Chances of healthy pregnancy in a female carrier with PGD TRS™: 60.5%

There are also situations where a chromosome fragment is missing (deletion) or is duplicated (duplication) or changes location (insertion).

There can also be cases of “de novo” translocations, i.e. the occurrence of altered chromosomes in the child whose parents have normal karyotype.

1 www.ncbi.nlm.nih.gov/pubmed/25624194 [23 June 2016]
What is PGD TRS™?

PGD diagnosis for translocations consists in the examination of the embryo’s genetic material for the presence of abnormal fragments or entire chromosomes involved in translocation.

It has to be stressed that carriers of chromosomal translocation are also exposed to higher risk of disorders in the number of other chromosomes, unrelated to chromosomes involved in translocation. It is recommended to complement the PGD TRS™ targeting the chromosomes involved in translocation with the comprehensive diagnosis for aneuploidies in all chromosomes – PGS NGS 360°™.

Why is it worth to perform PGD TRS™?

- Most reproductive cells of the translocation carrier are abnormal, which causes poorer prognosis for fertilisation and development of healthy children. PGD TRS™ allows selecting normal embryos, thus significantly reducing the risk of reproductive failure.
- PGD TRS™ allows checking for the occurrence of a “de novo” abnormality of the genetic material number in the embryo.
- Preimplantation diagnosis reduces the risk of miscarriages and increases chances for giving birth to a healthy child.

Indications for PGD TRS™

- Confirmed presence of a translocation in an individual trying for a child.

INVICTA Genetic Laboratory provides:

- Specialist Collection Kit INVICTA PGD BIOPSY KIT™
- Free transport
- Online access to the results
The test provides information on the status of chromosomes involved in translocation. The test does not determine the status of other chromosomes. The absence of lost or extra fragments in chromosomes involved in translocations does not exclude the presence of changes in the number or structure of other chromosomes. The probability of detecting derivative regions of translocations depends on the location of the points of breakage on chromosomes and on the size of regions being subject to translocation. The type of technique used in the test is determined depending on the individual situation of a given patient with confirmed translocation. Techniques other than NGS may be applied in the test if the patients’ situation so requires. The NGS technique for this test has been validated for detecting genetic material losses or gains involving a fragment larger than 5 Mpb in the material constituting blastocyst cells. The reliability of this technique is lower in case of translocations where breakage regions are located near telomeres or centromeres.

**Limitations**

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The first in the world use of NGS (Next Generation Sequencing) technology in the clinical practice in preimplantation diagnosis for translocations.

INVICTA Genetic Laboratory: August 2013

32 possibilities of the genetic material distribution in the reproductive cells produced by the balanced translocation carrier.

Legend:
- Gametes with the potential for healthy children
- Gametes without the potential for healthy children

NGS / Next Generation Sequencing

is at present the most innovative technique of DNA analysis available in the world. It ensures extremely accurate, reliable and comprehensive result which allows examining cells collected from embryos for the presence of translocations of various types.


References