

# NGS-based molecular fetal karyotype on miscarriage tissue (product of conception)



*Identification of chromosomal  
causes of pregnancy loss*





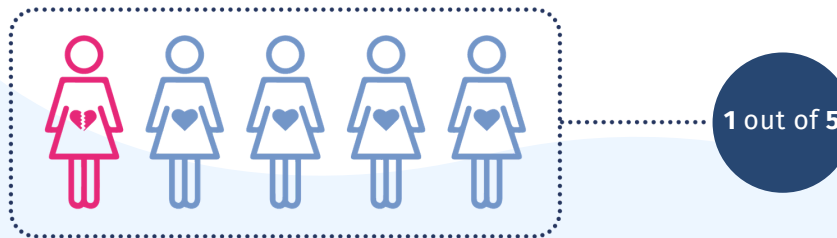
## CLINICAL BACKGROUND: MISCARRIAGE AND DIAGNOSTIC RATIONALE

Miscarriage is the most frequent complication of the first trimester of pregnancy, with an estimated incidence of **15% to 25%** of clinically recognized pregnancies.

Although etiological factors are multiple, approximately **60% of cases** are attributable to **chromosomal abnormalities of the product of conception (POC)**.<sup>1–2</sup>

Identifying a chromosomal abnormality as the cause of pregnancy loss has significant clinical value: it enables **recurrence risk estimation** and may reveal **familial chromosomal rearrangements** that can predispose the couple to recurrent miscarriage or to the birth of offspring affected by congenital malformations and/or intellectual disability.

In this context, since 2016 the **American College of Obstetricians and Gynecologists (ACOG)** and the **Society for Maternal-Fetal Medicine** have recommended chromosomal analysis on placental tissue, amniotic fluid, or product of conception in all cases of intrauterine fetal demise or perinatal mortality.<sup>3</sup>



<sup>1</sup> D'Ippolito S. et al., 2017

<sup>2</sup> Rosenfeld J.A. et al., 2015

<sup>3</sup> Committee Opinion No. 581, 2016



# POCADVANCE

Tissue Karyo

## THE POCADVANCE TISSUE KARYO TEST

**POCADVANCE TISSUE Karyo** is an advanced genetic test that enables determination of the **molecular fetal karyotype directly from miscarriage tissue** (product of conception—POC).

The test uses high-sensitivity, high-resolution **massively parallel sequencing technologies (Next Generation Sequencing—NGS)**, allowing detection of fetal chromosomal alterations that may be responsible for the miscarriage event.



The primary clinical objective is to provide genetically relevant information on the cause of pregnancy loss, supporting reproductive genetic counseling and family planning for the couple.





## ADVANTAGES COMPARED WITH TRADITIONAL METHODS

**POCADVANCE TISSUE Karyo** overcomes key limitations of conventional cytogenetic karyotyping, offering significant technical and clinical benefits:

- ➔ **No cell culture required**  
eliminates the risk of **culture failure** and **non-informative results**, a frequent limitation of conventional techniques.
- ➔ **Maternal cell contamination (MCC) exclusion**  
fetal origin of the DNA is verified by comparing the genetic profile of the POC sample with the maternal profile, ensuring analytical **specificity**.
- ➔ **High resolution**  
NGS-based molecular karyotyping enables detection of microdeletion and **microduplication syndromes >1 Mb**, compared with the ~10–15 Mb resolution threshold of traditional cytogenetic karyotyping.
- ➔ **Rapid turnaround time**  
results available in approximately **5 working days**.



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## TECHNOLOGY: HIGH RESOLUTION AND ADVANCED BIOINFORMATICS



**Collection of fetal POC  
tissue and maternal sample**



**DNA  
extraction**



**NGS  
Sequencing**



**Bioinformatic  
analysis**



**Reporting of  
test result**



## RESULT INTERPRETATION



### POSITIVE

**Chromosomal abnormality detected.**

A numerical and/or structural chromosomal abnormality has been identified in the fetal karyotype.



### NEGATIVE

**No chromosomal abnormality detected.**

No chromosomal abnormalities were identified; the fetal profile is compatible with a euploid karyotype.





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## INDICATIONS FOR TESTING

**POCADVANCE Tissue Karyo** is indicated in the following clinical scenarios:

- ✓ **Miscarriage** in women who wish to obtain a genetic characterization of the product of conception;
- ✓ History of **recurrent miscarriage**;
- ✓ **repeated pregnancy losses** in the absence of an exhaustive clinical explanation.



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Tissue Karyo

## HOW TO ORDER THE TEST



Kit  
request



Documentation  
completion



Sample  
collection



Sample  
shipment



Report  
delivery

### Samples required:

1

POC Tissue

2

Maternal buccal swab

**Sample handling notes:** the tissue must be placed in the provided tube and stored in saline solution. If shipment is delayed, refrigeration at +4°C is recommended.

Turnaround time



5 working days

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## LABORATORIES



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