POC Products of Conception

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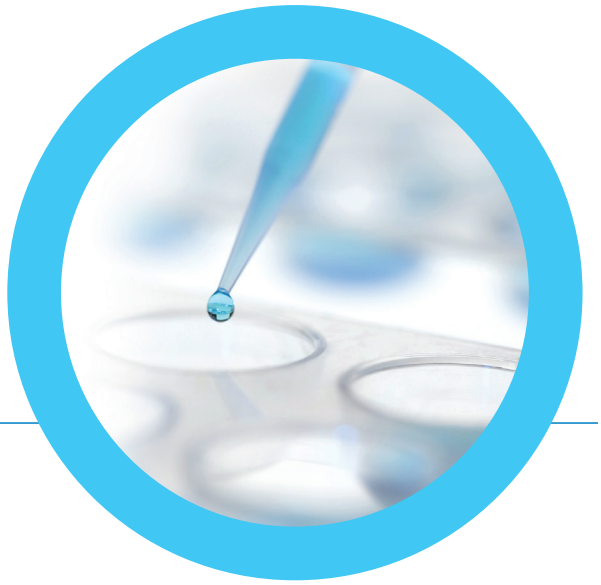
PIONEERS IN REPRODUCTIVE GENETICS

Next-Generation Sequencing (NGS) to analyze fetal tissues

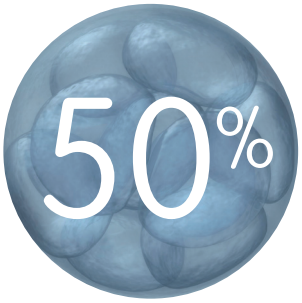
Identifying the cause of pregnancy loss may be of great benefit to couples who have experienced a spontaneous miscarriage.



Reasons

to use Next-Generation Sequencing (NGS) to analyze fetal tissue:

Conventional karyotype	NGS (POC)
Requires in vitro cell culture	1 It does not require in vitro culture
42% of the tests performed are not informative due to tissue degradation	2 Results are obtained in more than 98.6% of the tests performed
33.3% are false negatives due to maternal contamination	3 This technique rules out false negatives caused by maternal cell contamination
Results are provided in 2-4 weeks	4 Results are obtained in 1 week
Low resolution analysis	5 Higher resolution than conventional karyotyping

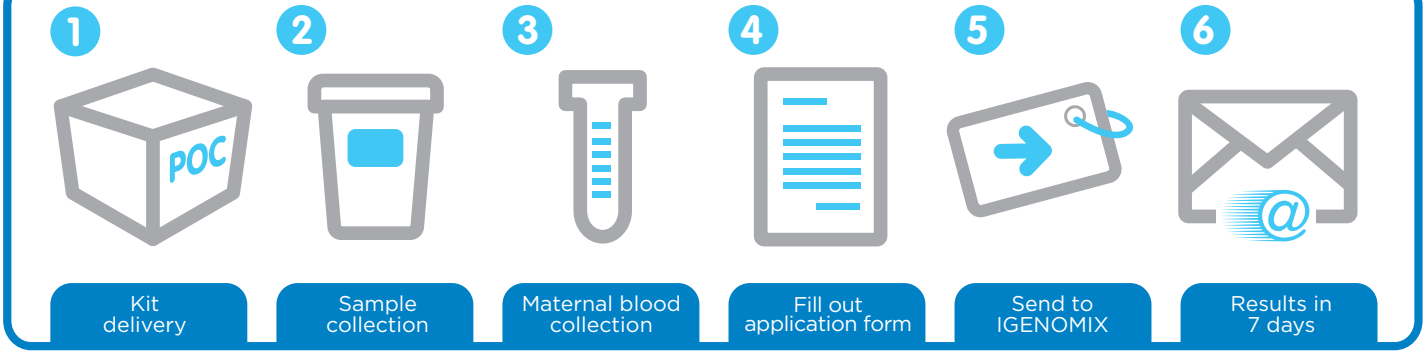


50% of first trimester pregnancy losses are due to

chromosomal abnormalities

In women who undergo assisted reproduction techniques this value exceeds **60%**

SIMPLE AND EASY



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NEXT-GENERATION SEQUENCING (NGS) TO ANALYZE FETAL TISSUES

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1 What is POC?

- NGS is a molecular technique that allows all **24 chromosomes** to be studied in order to rule out aneuploidies in fetal tissue.
- It does not require in vitro culture before the analysis. Therefore, it increases the percentage of cases which obtain results at the same time that it reduces the time required to acquire them.
- **In addition, analysis of microsatellite markers in blood collected from the mother allows any maternal contamination to be detected.**

2 What is POC useful for?

- Aneuploidies are chromosome abnormalities that can lead to spontaneous miscarriage and chromosome disorders in newborns (babies).
- Chromosome abnormalities are responsible for **50%** of first trimester pregnancy losses occurring in both spontaneous conceptions and pregnancies resulting from assisted reproduction treatments.
  (Martinez *et al.* 2010; Campos-Galindo *et al.* 2012).
- Genetic diagnosis of the products of conception is necessary to identify the etiology of a gestation failure and to ensure appropriate counseling is provided to the couple.

3 Who is POC indicated for?

- It is recommended to any couple who have suffered a pregnancy loss, but especially to those who have experienced recurrent miscarriages or are undergoing assisted reproduction treatments.

4 How should the sample be collected?

- The sample is obtained either by directed biopsy after pre-curettage hysteroembryoscopy (which is then collected and transported in a 10 ml conical tube in sterile saline solution), or in conventional curettages samples (as far as possible avoiding the inclusion of maternal tissue) are placed in a urine collection vial containing sterile saline. It is essential to also include 5 ml of maternal blood collected in an EDTA vial.

5 How should the sample be shipped? 4ºC

- The sample is transported at room temperature in a sealed tube or urine collection vial using appropriate protective measures for shipment. The sample must be shipped within 48 hours. The sample should be stored at 4º C at the place of origin before transport.

6 How long does it take to obtain results?

- The laboratory protocol requires between 24 - 48 h. meaning there is a maximum of one week before the results are issued.

one week

7 METHODOLOGY MAIN STEPS OF THE ANALYSIS

FETAL TISSUE › DILACERATION › DNA EXTRACTION › LIBRARY CONSTRUCTION › TEMPLATE PREPARATION › SEQUENCING › **RESULTS** › **A 46XX** RESULT REQUIRES ADDITIONAL MICROSATELLITE ANALYSIS

8. Limitations. This technique does not detect structural chromosome abnormalities and cannot identify: Low degree mosaicism aneuploidies, triploidy/tetraploidy, uniparental disomy, deletions or duplications smaller than 10Mb.

Campos-Galindo I, Martínez-Conejero JA, García-Herrero S, Ayala-Álvarez G, Rubio Lluesa C. Tecnología BACs-on-Beads™ aplicada al diagnóstico prenatal y al estudio citogenético de restos abortivos. Diag Pren. 2012 Volume 23, Issue 2, April-June 2012: 76-82.

Martínez MC, Méndez C, Ferro J, Nicolás M, Serra V, Landeras J. Cytogenetic analysis of early nonviable pregnancies after assisted reproduction treatment. Fertil Steril. 2010 Jan;93(1):289-92.

Ferro, Jaime; Martínez, Ma.Carmen; Lara, Coral; Pellicer, Antonio; Remohí, José; Serra, Vicente. Improved accuracy of hysteroembryoscopic biopsies for karyotyping early missed abortions. Fertility and Sterility vol. 80 issue 5 November, 2003. p. 1260-1264.