GENOMICA
Next Generation Genetics

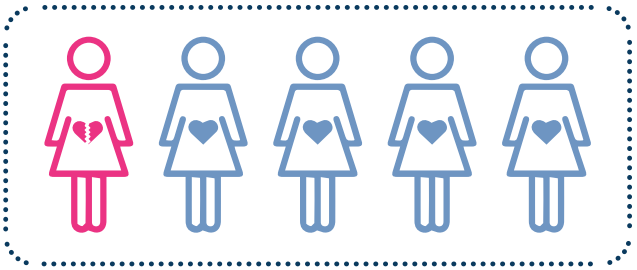
Product of Conception Karyotyping by Cell-Free Fetal DNA Analysis

POCADVANCE cfDNA
cfDNA -BASED POC KARYOTYPING

www.pocadvance.it

POCADVANCE cfDNA

ABOUT MISCARRIAGE



1 out of **5**

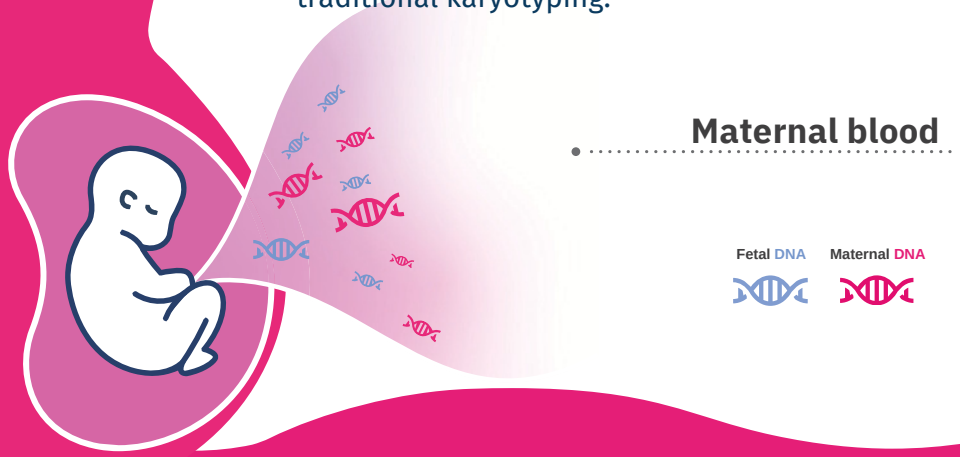
Pregnancies terminated due to miscarriage

Miscarriage is the most common complication during the early stages of pregnancy and occurs in about **15-25%** of clinically diagnosed pregnancies, most of which in the first trimester of gestation.
There are many known causes and risk factors for spontaneous abortion but about 60% of cases originate from chromosomal abnormalities[1-2].
Diagnosing a chromosomal abnormality as a cause of miscarriage **provides important information for predicting the risk of recurrence** and helps identify familial chromosomal rearrangements that can predispose couples to recurrent miscarriages or the birth of babies with congenital abnormalities and / or intellectual disabilities. In 2016, the American College of Obstetricians and Gynecologists (ACOG) and the Society for Maternal-Fetal Medi- cine recommended the search for chromosomal abnormalities on the placenta, amniotic fluid and the **product of conception** (POC) **in cases of intrauterine fetal death** and **perinatal mortality[3]**.

1. D'ippolito et al., 2017
2. JA Rosenfeld et al., 2015
3. Committee Opinion No. 581, 2016

POCADVANCE cfDNA

An advanced molecular test that, using groundbreaking technologies, determine the karyotype of **product of conception** (**POC**). By analyzing the circulating **cell- free fetal DNA** (**cfDNA**) in maternal blood, using the latest fully automated sequencing technologies (**Next Generation Sequencing** - **NGS**), it screens for genome-wide numerical and structural chromosomal abnormalities, providing unparalleled accuracy and detection compared to traditional karyotyping.



During pregnancy, placental trophoblasts, through a physiological process known as apoptosis, releases DNA fragments into the maternal blood, starting from the 5th week of gestation. This DNA is also named circulating **cell-free fetal DNA** (**cfDNA**).

During miscarriage, the placental tissue continues to release cfDNA into the maternal bloodstream, making it possible to perform early non-invasive investigations, thus obtaining valuable information on the genetic causes of the abortion.

POCADVANCE cfDNA

BENEFITS

POCADVANCE **cfDNA** is able to provide information on the possible genetic causes of a spontaneous abortion, with the aim of helping the couple in family planning with specific reproductive genetic counseling.

cfDNA testing

- **Non-invasive** test, performed by a simple **blood sample** collected from 5^ weeks of gestation;
- Test executable even in very **early stages of abortion**, before performing curetage.

Cell culture is not required

- Exclusion of the risk of achieving **no results** due to lack of cell growth;
- Significant reduction in **turnaround time (3-5 days)**, instead of 15-20 days required with “traditional" karyotyping

High reliability of results

- Exclusion of the risk deriving from the growth in culture of **maternal cells instead of fetal**
- **ones**; Quality derived from NGS-based molecular karyotyping.

Greater resolution and accuracy compared to traditional karyotyping

- **Much higher resolution: 5 Mb** instead of 10-15 Mb achieved with traditional karyotyping.

4.Yaron et al., 2020; **5**. Colley et al., 2020; **6**. Hartwig et al. 2023

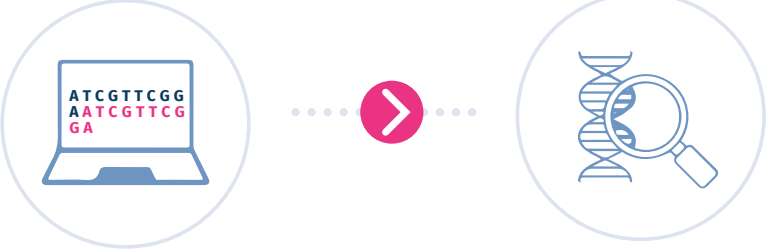
POCADVANCE cfDNA

A GROUNDBREAKING TECHNOLOGY THAT MAKES THE DIFFERENCE

Blood sample collection

cfDNA extraction
LoD: 1% FF

Next Generation Sequencing (NGS)



Advanced bioinformatic analysis

cfDNA screening for detection of genome-wide numerical and structural chromosomal abnormalities

POCADVANCE cfDNA

TEST RESULTS

Chromosomal abnormality detected: this result shows that the test detected a chromosomal abnormality in the fetus.

Chromosomal abnormality NOT detected: this result shows the test has not detected any chromosomal abnormality. This result is suggestive of euploidy (normal fetal karyotype).

This test is unable to detect balanced chromosomal rearrangements (reciprocal translocations, inversions), chromosomal mosaicisms (i.e. the presence of two cell lines with different karyotype) with a poorly represented cell line, microdeletion/ microduplication syndromes, structural chromosomal anomalies with a size below **5** Mb, point mutations, methylation defects, polyploidies.

POCADVANCE cfDNA

INDICATION FOR TESTING

✓ SUGGESTED

* Women who have had a miscarriage

✓ STRONGLY RECOMMENDED

** Women who have had recurrent miscarriages

*** Women who have had miscarriages following IVF treatments

THE TESTING PROCESS

1. **Request the test kit**
2. **Collect the sample**
3. **Ship the sample to Genomica Lab**
4. **Receive results**

Blood draw must be performed immediately after confirming the miscarriage, and in any case before performing curetage, or **within 24h** from POC expulsion. The test can be performed in case of spontaneous abortions of single or twin pregnancies, monozygotic or dizygotic, with at least 5 weeks of gestation.

GENOMICA
Next Generation Genetics

Advanced molecular diagnostics solutions using state-of-the art technologies

GENOMICA is recognized as one of the most advanced molecular diagnostics laboratory in Europe, both for the state-of-the-art instruments and technologies, as well as for its high quality standards.

With a **comprehensive portfolio of over 10.000 genetic tests,** GENOMICA is able to satisfy increasingly specialised requests in the field of molecular genetics, providing physicians and their patients with innovative and highly specialised diagnostic solutions for any clinical need.

Over **100.000** genetic tests/year

Fast TAT

Laboratories with **groundbreaking technologies** and high quality standards

Test performed in Italy
(Rome or Milan)

Dedicated R&D team

International **Partnerships**

Personalized genetic counseling with genetic counselors experts in discussing genetic test results and familial risks

Professionals with 20+ years expe- rience in the field of genetics and prenatal molecular diagnostics

LABORATORIES

Roma: Via Arduino 38 - 00162 - Tel.: 06.21115020
Milano: Viale L. Bodio 29-37 (Bodio 3) - 20158 -
Tel.: 02.21115330
E-mail: info@genomicalab.it -
www.genomicalab.it

REGISTERED OFFICE

Roma: Via Arduino 38 - 00162
Pec: info@pec.genomicalab.it
VAT no.:14554101007 - REA: RM - 1530210