GENOMICA
Next Generation Genetics

Non-invasive maternal blood test for identifying chromosomal causes of miscarriage

Analysis of circulating cell-free fetal DNA by Next Generation Sequencing

POCADVANCE
cfDNA Karyo

www.pocadvance.it

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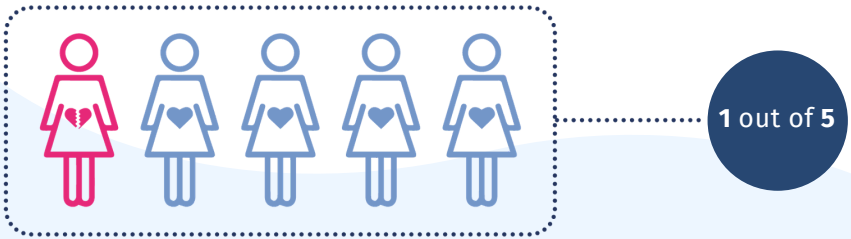
CLINICAL BACKGROUND: MISCARRIAGE AND DIAGNOSTIC RATIONALE

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 the etiological factors are multiple, approximately **60% of cases** are attributable to **chromosomal abnormalities** of the product of conception (POC).[1–2]

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.[3]



1. D'Ippolito S. et al., 2017
2. Rosenfeld J.A. et al., 2015
3. Committee Opinion No. 581, 2016

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THE POCADVANCE cfDNA KARYO TEST

POCADVANCE cfDNA Karyo is a non-invasive genetic test that, through the analysis of **circulating cell-free fetal DNA** (**cfDNA**) obtained from a maternal peripheral blood sample, allows determination of the **molecular karyotype** of the product of conception. The test uses high-sensitivity, high-resolution **massively parallel sequencing technologies** (***Next Generation Sequencing* – NGS**) to detect fetal chromosomal abnormalities that may be responsible for the miscarriage event.

Advantages of cfDNA analysis

- **Non-invasive**: performed with a simple maternal blood draw
- **Clinically useful timing**: can be performed before any uterine evacuation procedure
- **Applicable early**: potentially feasible in the **early stages** of pregnancy (≥ 5 weeks)

CIRCULATING CELL-FREE FETAL DNA (cfDNA): BIOLOGICAL RATIONALE

During pregnancy, the placenta physiologically releases DNA fragments into the maternal circulation (apoptosis), starting from approximately the **5th week of gestation**. This material is defined as circulating **cell-free fetal DNA** (**cfDNA**).

Close to a miscarriage event, placental tissue may continue to release cfDNA into the maternal blood, making it possible to perform **non-invasive** investigations immediately after the ultrasound diagnosis of spontaneous pregnancy loss, with the aim of obtaining information useful on the potential genetic contribution.

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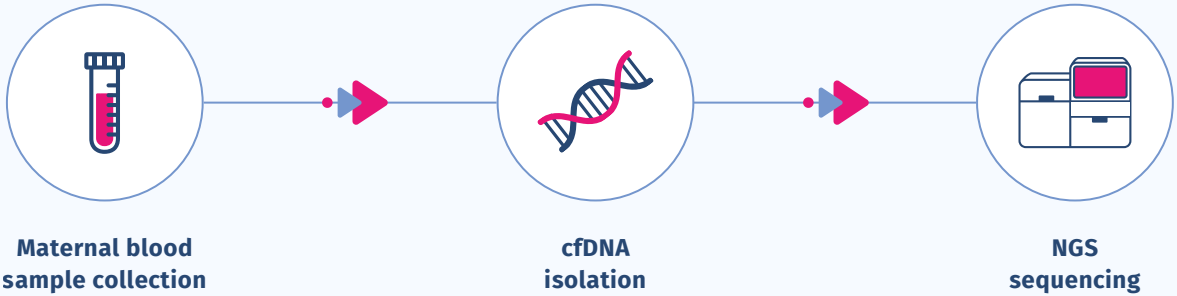
TECHNOLOGY: HIGH RESOLUTION AND ADVANCED BIOINFORMATICS



Maternal blood sample collection

cfDNA isolation

NGS sequencing

Bioinformatic analysis using a proprietary algorithm

Reporting of test result

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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 cfDNA Karyo is particularly indicated in the following clinical settings:

- ✓ **spontaneous miscarriage** (anembryonic gestational sac, absence of cardiac activity, miscarriage in progress) when a timely, non-invasive investigation is desired;
- ✓ history of **recurrent miscarriage**
- ✓ **repeated pregnancy losses** in the absence of an exhaustive clinical explanation.

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HOW TO ORDER THE TEST

Kit request → **Documentation completion** → **Sample collection** → **Sample shipment** → **Report delivery**

Blood draw timing

The blood sample should be collected **as soon as possible** after ultrasound confirmation of pregnancy loss and **before** any uterine evacuation procedure; alternatively, **within 24 hours** of expulsion of the products of conception.

The test can be performed in **singleton or twin pregnancies** (monozygotic or dizygotic) at **≥ 5 weeks** of gestation.

Turnaround time **2-3 working days**

GENOMICA
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Leading diagnostic laboratory and research center of excellence specializing in genetics and molecular diagnostics.

GENOMICA is a highly innovative company with extensive technical and scientific expertise, active in both clinical applications and research. Supported by a team with over 20 years of experience in molecular diagnostics, GENOMICA combines cutting-edge technology with a strong commitment to innovation, delivering increasingly accurate and accessible diagnostic services.

Over **100.000 genetic tests/year**

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

Dedicated **R&D Team**

International **Partnerships**

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

20+ years experience in prenatal molecular diagnostics

POCAdvance cfDNA Karyo ENG rev.00

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Visit the website dedicated to the test