Genetic Laboratory

Innovators in Reproductive Genetics

iMGE Test™
Genetic diagnosis of miscarriages

Innovators in Reproductive Genetics
Identification of the causes of miscarriage

**Over 60% of spontaneously miscarried embryos/fetuses have chromosomal defect**.

Conducted by INVICTA chromosome analysis identified causes of pregnancy loss in 62% of all miscarriages which in other cases would remain unexplained.

997

of 1000 pregnancies with chromosomal defects are miscarried

Genetic errors in a developing embryo may cause early pregnancy loss, death of the embryo/fetus or premature delivery occur in the vast majority of cases. Therefore, the incidence of live births of children with chromosomal defects amounts only to 3 of 1000 births.

*Information on the page refers to the analysis of the situation of women aged 35 and older who have experienced recurrent miscarriages. The majority of chromosomal defects in such cases are aneuploidies, i.e. chromosomal number abnormalities (Marquard et al. FertilSteril. 2010 Sep;94(4):1473–7).*
INVICTA Genetic Laboratory offers:

**Miscarriage Genetic Evaluation Test (Genetic Diagnosis of Miscarriages)**

- iMGE Test™ is a group of analyses of the miscarriage material performed with molecular biology techniques which allow to obtain information on the presence of chromosomal defects and about the sex of the fetus.

  - In contrast to classical cytogenetic methods:
    - the tests do not require in vitro cultivation prior to the analysis,
    - the results are obtained from over 95% of tests conducted,
    - the technique allows to avoid false positive results due to contamination with mother cell material.

**Why is it worth to perform iMGE Test™?**

- It allows to detect the most frequent chromosomal defects which may cause miscarriages
- It allows quick and reliable determination of the fetus’ sex
- NGS method allows simultaneous analysis of numerical changes in all chromosomes which are present in human body

**Indications for iMGE Test™**

- In the case of miscarriage, testing for genetic conditions should be routinely conducted in order to determine the probable cause of pregnancy loss.

  - iMGE Tests is recommended in particular if miscarriage occurs in:
    - women who decides to get pregnant after 35 years of age
    - couples who were diagnosed as the carriers of chromosomal defects
    - couples whose family presented with genetic defects (so-called positive genetic history)
    - women who have history of recurrent miscarriages
    - couples whose in vitro programmes failed despite transferring embryos with normal morphology
    - couples who have been treated for idiopathic infertility for a long time

**What next?**

- If the genetic causes of miscarriage are identified, the following actions are recommended:
  - genetic consultation
  - undertaking appropriate diagnosis and therapeutic steps
  - Pre-implantation Diagnosis

**What is iMGE Test™?**

- Specialist collection kit
- Free transport
- Online access to the results

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Cooperation step by step

1. Sample collection & preparation
   - Sample collection
     - Targeted biopsy during the procedure typically performed by the physician after the miscarriage
   - Preparation of the sample
     - Place in the solution of physiological saline
   - Filling the referral note
   - Preparation of the material for transport
     - Transport at temperature of 2–8°C
   - Preparation of shipping document
   - Shipment to the INVICTA Genetic Laboratory
     - To be delivered within 48 hours since the collection
     - Until it is sent, store at temperature of 2–8°C

2. Transport
   - Transport at room temperature
   - Preparation of shipping document
   - Shipment to the INVICTA Genetic Laboratory
     - To be delivered ANY time since the collection

3. Result
   - Results available within 7-14 days

Shipping address:
INVICTA Genetics Laboratory
Gdańsk Science and Technology Park
ul. Trzy Lipy 3, 80–172 Gdańsk
E-mail: genetyka@invicta.pl • Tel.: +48 784 373 593

NGS methodology
- Sample preparation
- Libraries preparation with barcoding
- Sequence preparation
- Information read-out
- Data analysis

Limitations
This test was designed to detect aneuploidies and imbalance resulting from Robertsonian translocations. It does not detect segmental aneuploidies, germline mosaicism related to aneuploidy, structural chromosome defects (e.g., deletion of the chromosome part, inversion, duplication), uniparental disomy, triploidy, tetraploidy.
First in the world use of NGS (Next Generation Sequencing) in the genetic diagnosis of miscarriages.

INVICTA Genetics Laboratory, July 2014.

Options INVICTA’s genetic diagnosis of miscarriages

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<tr>
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<th>iMGE XY Test™</th>
<th>iMGE+ Test™</th>
<th>iMGE 24 Test™</th>
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<td>X, Y chromosomes</td>
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<td></td>
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**NGS / Next Generation Sequencing**

is currently the most up-to-date method of analysing DNA information in the world. It ensures exceptionally precise, reliable and comprehensive result to determine the causes of miscarriage.

99,999%

Performing iMGE 24 Test™ ensures accuracy of 99,999% (quality assessment of Q50 acc. to Phred Quality Scores – the most common metric used to assess accuracy of a sequencing platform).
Literature


