PGS-NGS 360°™
Preimplantation Genetic Screening

Innovators in Reproductive Genetics
Primplantation genetic screening for aneuploidy using Next Generation Sequencing (NGS) is currently the most effective method for selecting genetically normal embryos for transfer.

Studies conducted by a team led by Prof. Krzysztof Łukaszuk¹ have shown that using NGS to identify embryos free of any chromosomal changes can significantly increase success rates of IVF procedures. In a study of 45 patients, PGS-NGS 360°™ was carried out on biopsies from embryos in the 3rd day of development, and then transferred in a fresh cycle. The control sample consisted of 53 female patients selected according to age, AMH level, number of antral follicles and the duration of their infertility. After the PGS-NGS 360°™ was performed, a significant increase in the pregnancy rate (PR) and implantation rate (IR) was seen.

Results of IVF programs after PGS-NGS 360°™ using Next Generation Sequencing.


PGS-NGS 360°™ is a test for aneuploidy screening of all 24 chromosomes.
PGS-NGS 360°™ (preimplantation genetic diagnosis using Next Generation Sequencing) employs state of the art human genome sequencing techniques (direct reading of genetic information) to examine embryos. It opens up new diagnostic possibilities.

In the past, embryos were selected mainly based on the basis of their morphology. The FISH and microarray methods were used for chromosome analysis. However, both techniques were limited in terms of range and accuracy. But with the PGS-NGS 360°™, all 24 chromosomes are analysed with unprecedented precision. This provides doctors with a unique opportunity to help couples exposed to increased risk of genetic abnormalities in their children.

Chromosomal aneuploidy (an abnormal number of chromosomes) is one of the main causes for the failure of IVF programs. Most embryos with aneuploidies are unable to nest in the uterus, and those that are implanted often miscarry in the first three months of the pregnancy. Preimplantation genetic diagnosis used to provide optimal selection of embryos for transfer, can greatly increases the chances of success of the procedure and the birth of a healthy child.

What is PGS-NGS 360°™?

- Analyses all autosomal and sex chromosomes
- Allows for diagnosis of the most common genetic defects, including: Downs, Edwards, Patau, Turner, and Klinefelter’s syndrome, while still at the embryo stage
- Increases the embryo implantation rate
- Reduces the risk of miscarriage
- Increases the number of healthy births
- Increases the efficiency of single embryo transfer by reducing the number of multiple pregnancies

Why is it worth doing the PGS-NGS 360°™?

- Age of woman over 35
- Failure of IVF programs (although embryos of normal morphology were transferred)
- Recurrent miscarriages
- Genetic defects diagnosed in previous pregnancies / birth of a child with genetic abnormalities in the past
- A history of genetic defects in the family
- An elective single embryo transfer during in vitro program
- The intention of ruling out the presence of genetic defects in previously frozen embryos
- Need to increase the chances of pregnancy in an IVF cycle with donor cells
- Fear of childbirth with chromosomal defects

What are the indications for a PGS-NGS 360°™?

- Analyses all autosomal and sex chromosomes
- Allows for diagnosis of the most common genetic defects, including: Downs, Edwards, Patau, Turner, and Klinefelter’s syndrome, while still at the embryo stage
- Increases the embryo implantation rate
- Reduces the risk of miscarriage
- Increases the number of healthy births
- Increases the efficiency of single embryo transfer by reducing the number of multiple pregnancies

Age of woman over 35
- Failure of IVF programs (although embryos of normal morphology were transferred)
- Recurrent miscarriages
- Genetic defects diagnosed in previous pregnancies / birth of a child with genetic abnormalities in the past
- A history of genetic defects in the family
- An elective single embryo transfer during in vitro program
- The intention of ruling out the presence of genetic defects in previously frozen embryos
- Need to increase the chances of pregnancy in an IVF cycle with donor cells
- Fear of childbirth with chromosomal defects
Cooperation step by step

1. Biopsy
   - Embryo biopsy
   - Completing biopsy report

2. Transport
   - Preparation of material for transport
   - Preparation of shipping document
   - Dispatch to INVICTA Genetic Laboratory

3. Result
   - Results available within 7–14 days

Shipping address:
INVICTA Genetic Laboratory
Gdańsk Science and Technology Park
Trzy Lipy 3, 80–172 Gdańsk
E-mail: info@invictagenetics.com
Tel.: +48 784 373 593

Ready-to-use universal
INVICTA PGD BIOPSY KIT™

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

Limitations
This test is designed to detect aneuploidy and/or irregularities from Roberstonian translocation. This test does not detect partial aneuploidy (i.e. fragments of chromosomes) or chromosome mosaicism and structural chromosomal abnormalities (e.g. fragments of chromosome deletions, inversions, duplications) or uniparental disomy, triploidy and tetraploidy.
The world’s first application in clinical use of NGS (Next Generation Sequencing) technology in preimplantation diagnosis.

INVICTA Genetic Laboratory: August 2013

Table 1. The results of efficacy studies of IVF programs in patients with Repeated Implantation Failure (RIF) using the PGS-NGS 360™.

<table>
<thead>
<tr>
<th>Specification</th>
<th>RIF patients with PGS-NGS 360™</th>
<th>RIF patients without PGS-NGS 360™</th>
</tr>
</thead>
<tbody>
<tr>
<td>Number of IVF cycles with transfer</td>
<td>45</td>
<td>53</td>
</tr>
<tr>
<td>Number of embryo’s biopsies</td>
<td>252</td>
<td>N/A</td>
</tr>
<tr>
<td>Number of transferred embryos</td>
<td>65</td>
<td>89</td>
</tr>
<tr>
<td>Average number of transferred embryos</td>
<td>14</td>
<td>1.7</td>
</tr>
<tr>
<td>Average female patient’s age</td>
<td>34.0</td>
<td>34.4</td>
</tr>
<tr>
<td>Clinical pregnancy rate</td>
<td>84.4% (38/45)</td>
<td>41.5% (22/53)</td>
</tr>
<tr>
<td>Implantation rate</td>
<td>61.5%</td>
<td>34.8%</td>
</tr>
</tbody>
</table>

* Clinical pregnancy rate provided for both the IVF cycle and for transfer (transfers were made for all female patients) and defined as the detection of a heartbeat at the 6th week and the 1st–3rd day using ultrasound.

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%

NGS provides an accuracy of 99,999% (Q50 quality assessment by Phred Quality Scores – an indicator developed for evaluation of DNA sequence analysis methods).
Literature


Team

Prof. Krzysztof Łukaszuk
MD, Ph. D.
Medical Director
of INVICTA Fertility Clinics

Bozena Maj M. Sc.
Director of INVICTA
Medical Laboratories

Sebastian Pukszta Ph.D.
INVICTA Genetic Laboratory
Deputy Laboratory Manager
for Molecular Biology

Joanna Liss Ph.D
Director of INVICTA
IVF Laboratory