PGS Embryo Screening

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Contents

What are chromosomes?

Chromosomes are the packages of genetic information within our cells. The correct number of chromosomes is 46 chromosomes (23 chromosome pairs) per cell. Chromosomes are numbered from 1-22 (these are referred to as the autosomes) and X and Y which are the sex chromosomes.

Usually our cells have 23 pairs of chromosomes, but random errors in the number of chromosomes (aneuploidy) may result in failure of the embryo to implant, lead to a miscarriage or result in the birth of a baby with serious developmental and other health problems (e.g. trisomy 21 – Down Syndrome).

Why should I consider chromosome testing of my embryos?

Many embryos created either via natural conception or IVF have chromosomal abnormalities which means that they will not result in either an ongoing viable pregnancy or a baby.

Preimplantation genetic screening (PGS) gives you a way of reducing this risk.

Through the exclusion of embryos identified with an abnormal chromosome complement, PGS can maximise treatment outcomes per embryo transfer.

Preimplantation genetic screening (PGS) for chromosome abnormalities such as aneuploidy gives you a way of reducing this risk. Aneuploidy testing identifies embryos that have the wrong number of chromosomes, thereby significantly reducing the risks of failed implantations, miscarriages and abnormal live births.

In a standard IVF treatment, a choice must be made of which embryo will be transferred. That choice is determined by scientists and fertility specialists based on information from microscopic inspection of the development and appearance of the embryos, usually over a five day period to identify which embryo is likely to result in a pregnancy. Unfortunately, embryos with a chromosome error may appear normal at this time.

PGS provides another factor on which to base this decision - the chromosomal health of your embryos. PGS help us to exclude embryos that contain an obvious chromosomal abnormality.

So, if you're considering IVF, PGS could be an important part of the equation.

Embryo testing using preimplantation genetic screening (PGS)

An advanced tool for embryo selection, PGS involves testing each suitable embryo for its chromosome status for all of the numbered pairs of chromosomes (chromosome pairs 1-22) and the pair of sex chromosomes (X, Y). On average, approximately half (50%) of embryos which are tested using PGS are shown to have chromosomal problems that reduce the likelihood of a healthy ongoing pregnancy. Maternal age is the most important factor in determining the proportion of aneuploidy embryos.

PGS lets us screen your embryos to find one in which no chromosomal abnormalities are identified. Through the transfer of only these embryos, PGS can maximise treatment outcomes per embryo transfer.

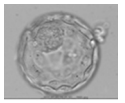
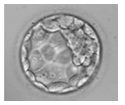
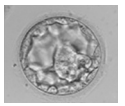
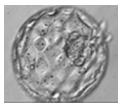
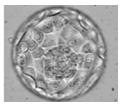
If you decide to go ahead with chromosome testing on your embryos, we will:

1. **Exclude embryos with major chromosomal abnormality**
2. **Choose the best quality embryo**
3. **Reduce your risk of miscarriage from chromosome abnormality**
4. **Maximise treatment outcome following an embryo transfer procedure.**

How does PGS and the chromosome test work?

Consider the following scenario:

If you have five good quality embryos from your IVF cycle, the table on page 5 shows the difference that testing could make. With this more detailed information we would know which embryos would not achieve a viable, ongoing pregnancy and we would also reduce the number of unnecessary embryo transfers that would not lead to pregnancy. The potential treatment outcome from your initial stimulation cycle is not changed because we would only transfer those embryos that are most viable – but you would achieve an ongoing pregnancy faster.

		Chromosome status	Expected outcome	If testing had occurred
Embryo 1		No abnormality	Normal live birth	Suitable for transfer ✓
Embryo 2		Trisomy 16	Miscarriage	Excluded ✗
Embryo 3		No abnormality	Normal live birth	Suitable for transfer ✓
Embryo 4		Trisomy 21	Miscarriage or abnormal live birth	Excluded ✗
Embryo 5		Mosaic Monosomy 2	Risk of an adverse clinical outcome	Last preference ?

What you need to know

1. Testing will reduce the number of embryos available for transfer, but will ensure those that are transferred have the best chance of survival;
2. You may have an embryo that is not suitable for testing
3. Embryos must be frozen pending test results which are usually available in 2-3 weeks;
4. It is possible that all of your embryos may have an abnormality resulting in no embryos being suitable for transfer from your IVF cycle;
5. Due to technical limitations of the testing process, it is possible that some embryos do not achieve a result;
6. An embryo may return a result indicating a mixture of abnormal and normal cells (referred to as mosaicism) and will require additional counseling before it can be considered for transfer
7. The test does not detect all types of genetic abnormalities; and
8. There is no guarantee a tested embryo will result in pregnancy.

What you need to do next

1. Visit our website **genea.com.au** and review the relevant PGS information.

   The information can be found at:

   genea.com.au/Myfertility/PGS under the section called **Next Generation Sequencing (NGS).** This information includes a description of how testing is done.

2. You will be contacted by a Genea Scientist on Day 3 or Day 4 after your egg pick up to discuss the benefits and risks of continuing with embryo screening. If you are a couple, we suggest that both partners are available for a conference call. However, if this is not possible please nominate who will receive the call.

3. Review the following timeline of treatment if you would like to consider opting in to PGS testing of your embryos.

The timeline for PGS

1. Before starting your cycle

- Read the associated information booklet (Your fertility Journey) and refer your questions to your Fertility Specialist; and
- Ensure you complete and return your consent form.

2. Day of egg collection

- Confirm with your Embryologist that you are still considering PGS as an option; and
- Confirm with your Embryologist which phone number should be called on Day 3 or Day 4 for the Science discussion.

3. After your egg collection

- Your embryos will be hatched in preparation for potential testing on Day 5 or Day 6.

4. Day 3 or 4 after your egg collection

- A scientist will contact you to discuss your embryos and the technical aspects you need to consider before proceeding.

5. Day 5 after your egg collection

- A Scientist will call to provide an update on the progress of your embryos and if your embryos are suitable for testing. They will also ask you for your final confirmation on whether to proceed with PGS testing and/or discuss non-testing options with you.

If you make the decision to proceed with testing, your embryos will be biopsied and immediately frozen. If you choose to not proceed with testing your nursing team will liaise with you and your Fertility Specialist to discuss embryo transfer and freezing as part of a standard IVF cycle treatment.

Please note that in some cases embryos that are potentially healthy and could lead to a viable pregnancy may not be robust enough to allow a biopsy. In this instance you might consider an untested embryo transfer or freezing untested embryos (if they are suitable).

6. Approximately 3 weeks later

- A Scientist will call you to discuss the outcomes from your PGS testing. These results will also be made available to your Fertility Specialist who will discuss your treatment options and next steps.

You're in good hands

If you have any questions, please do not hesitate to contact a Genea Scientist on **+61 2 9229 6420** and they will be happy to help.