PGT

Pre-implantation Genetic Testing

Bridge Clinic
Fertility Centre

www.thebridgeclinic.com

Preimplantation Genetic Testing - Everything you need to know

Having a healthy baby is something all parents hope for. But, what if you could go even further than that? What if you could help ensure the health of not only your child, but your child's children and many generations to follow?

At Bridge Clinic we offer couples who are undergoing IVF treatment for fertility reasons, or couples who prefer to undergo IVF treatment for the option of having their embryos thoroughly examined for specific gene mutations or chromosomal abnormalities, the option of Preimplantation Genetic Testing (PGT) formerly know as Preimplantation Genetic Diagnosis (PGD) and Preimplantation Genetic Screening (PGS).

Known in short as PGT, tests are performed on early (day 3 – 5) embryos prior to implantation and pregnancy. Offering specialists the opportunity to examine far beyond the superficial appearance of an embryo, PGT is increasingly being used at fertility institutes around the globe to help couples have healthy babies, no matter what challenges they face.

The method has also been found to dramatically improve the chances of a successful IVF pregnancy. Since PGT is newly available in Nigeria, many couples undergoing IVF may not be familiar with the procedure and its benefits.

Over the next few pages we'll aim to answer a few of your most pressing questions.

What is PGT?

PGT is a sophisticated reproductive technology used in conjunction with IVF treatment to test early embryos for specific genetic conditions. Many diseases, such as sickle cell anemia, cystic fibrosis and Tay-Sachs, are caused by a specific gene mutation. This is where PGT comes in to determine which embryos are carriers for such diseases and which are not.

It was first introduced to the medical world in the early 1990s by Dr Alan Handyside and colleagues. It was targeted at couples who had a high risk of transmitting genetic diseases to their offspring.

The process requires In Vitro Fertilisation (IVF) to obtain a number of oocytes or embryos for evaluation. Studies have shown that PGT cycles are most successful when implemented on 8 or more embryos, with at least 5 graded good quality by the embryology lab.

The process involves removing cells (trophectoderm) not destined to form the baby from a day 5 (blastocyst) (trophectoderm biopsy) and the cells which is a representative of the embryo is sent for genetic analysis.

Apart from selecting embryos without a genetic disorder, PGT also increases the chances of successful pregnancy.

PGT Definition:

A process which allows parents to have the option of detecting potential defects in an embryo before transfer.

It's important to note that the new term PGT doesn't change the way embryos have been tested with PGD or PGS, it is just a terminology now used to describe all genetic tests.

> “On the day of embryo transfer, I was nervous as usual but with the team of doctors I was privileged and lucky to have, it went without a hitch. Two weeks later I had a positive pregnancy test and I couldn’t believe it. The most amazing thing was seeing my baby’s heart beat on the first scan. It’s an indescribable feeling.”

What is PGT?

At Bridge Clinic, PGT is an important treatment option for couples who are undergoing IVF treatment for fertility reasons or couples who prefer to undergo IVF treatment for the option of having their embryos thoroughly examined for specific gene mutations or chromosomal abnormalities.

There are 3 types of Preimplantation Genetic Testing (PGT);

- PGT-M
- PGT-A
- PGT-SR

What is PGT-M?

PGT-M (PGT- Monogenic/Single Gene Disorders) formerly known as Preimplantation Genetic Diagnosis (PGD) is a sophisticated reproductive technology used in conjunction with IVF treatment to test early embryos for specific or single gene disorders. This treatment is important for couples that have a history of genetic disease which can be passed down to the child e.g., Sickle cell aneamia, cystic fibrosis, beta-thalassemia, etc.

What is PGT-A?

PGT-A (PGT-“Aneuploidy”) formerly known as PGS is now used to describe screening embryos for sporadic chromosome abnormalities. The process is used to test embryos by counting the 46 chromosomes in order to detect an additional or missing chromosome(s) (“aneuploidy”). Normal embryos are usually the most likely to implant and result in a successful pregnancy. The screening helps to reduce the chance of having a child with extra or missing chromosomes, such as Down syndrome, Turners syndrome, Patau’s syndrome, Edward’s syndrome, abnormal sperm parameters etc.

What is PGT-SR?

PGT-SR (PGT- “Structural Rearrangements”) formerly known as PGD for chromosomal translocations (balanced or unbalanced) or inversions is used to test embryos for chromosomal abnormalities and also abnormal chromosomal positions and rearrangements. It is recommended for couples with recurrent pregnancy loss as well as a history of recurrent failed IVF/implantation and also for couples whose female partners are 35 years and above.

Difference between PGT-M and PGT-A?

PGT-M (PGD) is often confused with PGT-A (PGS), however the two processes are implemented to establish entirely different results. PGT-M as described tests for SPECIFIC genetic conditions, whereas PGT-A tests for OVERALL chromosomal normalcy in embryos. In other words, PGT-M is ideal for prospective parents who have a specific genetic condition they do not want to pass to their offspring.

Benefits of PGT-A

- It can be used to test for more than 100 specific genetic conditions
- Miscarriage rates following PGT for chromosomal screening are far less than with standard IVF
- Since the procedure is performed before implantation, couples may feel more comfortable with deciding whether or not to go ahead with the embryo transfer, should results not be favorable
- The procedure enables couples who may be at risk of passing genetic conditions to their children the opportunity of pursuing biological parenthood, which they may not have done otherwise
- Offers the option of family balancing and gender selection
- It is also useful for eliminating sex-linked hereditary diseases
- Women over the age of 35 years
- Women who have experienced recurrent miscarriages
- Severe male factor infertility

Who can benefit from PGT-M (PGD)?

- Carriers of single gene disorders
- Couples with specific chromosomal disorders in their family history
- Couples that are carriers of sex-linked genetic disorders
- Parents who have a child with a serious genetic condition, but want to get pregnant again

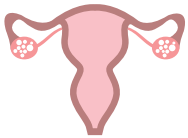
Who can benefit from PGT-SR (PGD for chromosomes translocations)?

- Women over the age of 35 years
- Women who have experienced recurrent pregnancy loss
- Women who have experienced repeated IVF and implantation failures.
- Couples with history of chromosomal balanced and/or unbalanced translocations or inversions

PGT is increasingly being used around the globe to help couples have healthy babies.

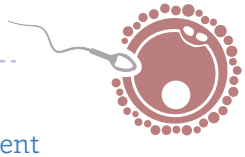
What does the PGT procedure entail?

The process starts off with patients undergoing controlled ovarian stimulation, which is a standard part of any IVF cycle.



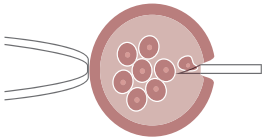
1 Ovarian stimulation and egg collection

Fertility medications stimulate egg production. Eggs are retrieved; sperm from the male is retrieved.



2 Embryo development

After preparation, the sperm and egg are combined (multiple sets). Eggs are observed to ensure fertilisation takes place, resulting in embryos.



3 Embryo biopsy

Cells are removed from day 5 embryos (trophyectoderm biopsy).

4 Amplification and analysis

During the evaluation, embryos free of genetic problems are identified.

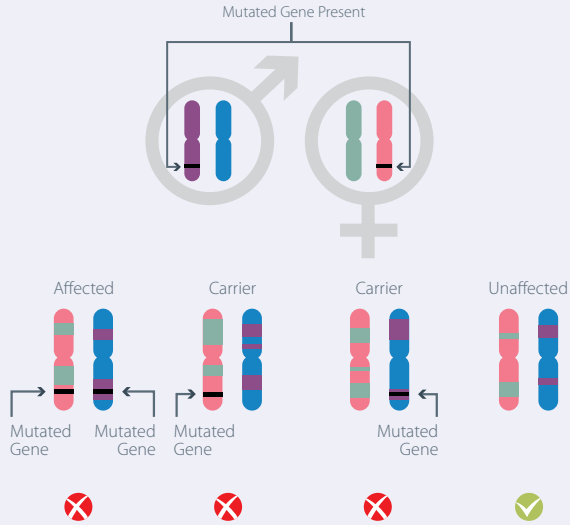
PGT-M (PGT- Monogenic/ Single Gene Disorders)

PGT-A (PGT- Aneuploidy)

PGT-SR (PGT- "Structural Rearrangements")

PGT analysis of chromosomal abnormalities

Cells from the embryo biopsy are carefully analysed to distinguish embryos with problems (to be discarded) from genetically or chromosomally normal embryos (for transfer and implantation). The process helps to achieve more pregnancies, reduce the number of miscarriages and reduce the number of children affected by genetic disorders.



5 Embryo transfer

One of the healthy embryos will then be placed in the uterus and implantation will be attempted.

Additional embryos free of genetic problems may be frozen for later use, while those with genetic conditions will be discarded.

What are the diseases and conditions that can be detected through PGT?

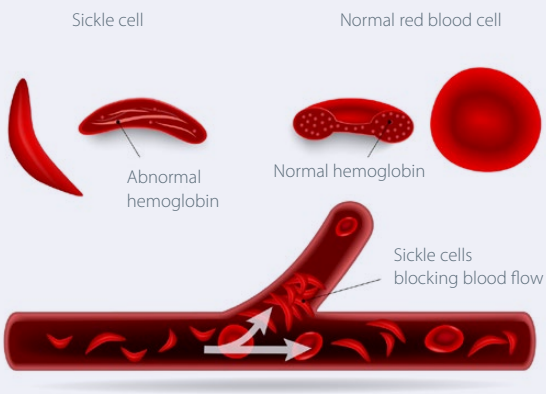
PGT is used to test for more than 100 genetic conditions. At Bridge Clinic, we are currently able to test for the following 13 disorders and chromosomal abnormalities:

Galactosemia

This rare genetic metabolic disorder affects an individual's ability to process the sugar known as galactose properly. There are three different types of galactosemia, with galactosemia type I being the most common and the most severe. Complications of galactosemia include liver damage, premature ovarian insufficiency, speech difficulty and intellectual disability.

Sickle cell anaemia

Sickle cell disease (SCD), as it is also known, is a serious genetic disorder affecting the red blood cells that carry oxygen throughout the body. People who suffer from SCD carry abnormal haemoglobin in their blood, causing the red blood cells to take on the form of a sickle, instead of the usual disc shape. The red cell sickling compromises oxygen delivery causing severe pain known as pain crises. Over time it also results in organ damage, causing harm to a person's spleen, brain, eyes, lungs, liver, heart, kidneys, penis, joints, bones, or skin.



Sickle cell disease prevalence in Nigeria is between 20% and 30% resulting in about 2% of the population directly affected by it. Those individuals who suffer from severe effects of SCD end up dying before they reach reproductive age.

Cystic fibrosis

People affected by cystic fibrosis have a defective gene that causes a thick build-up of mucus in the lungs, pancreas and other organs. They are prone to lung infections and need to work closely with healthcare professionals to steer clear of germs as much as possible. The build-up of mucus in the pancreas can stop the absorption of food and nutrients, causing malnutrition.

Glycogen storage disease

Glucose is a large energy source for the body, stored in the form of glycogen. It is released into the blood as needed with the help of special proteins called enzymes. When a person has glycogen storage disease, they have genetically defective enzymes that cannot successfully perform the tasks of controlling the change in sugar levels. Symptoms may include poor growth, muscle cramps, low blood sugar, a greatly enlarged liver and a swollen belly.

Polycystic kidney disease

Polycystic kidney disease causes numerous fluid-filled cysts to grow in the kidneys. If too many cysts grow or they become too big, the kidneys can become damaged. Most people don't develop symptoms until they are 30 or 40 years old and these include back pain, blood in the urine, frequent bladder or kidney infections and high blood pressure.

Familial hypercholesterolemia

Familial hypercholesterolemia (FH) is a genetic disorder characterised by high cholesterol levels, specifically very high levels of low-density lipoprotein, known as 'bad cholesterol' in the blood. This can cause cardiovascular disease to manifest at a young age.

Haemophilia

This sex-linked recessive genetic disease is more likely to occur in males than in females and impairs the body's ability to control blood clotting. While there is a common perception that those suffering from haemophilia can bleed to death from a small cut or scratch, the truth is that most bleeding actually occurs internally, into muscles and joints. It is not always clear what causes bleeding.

Duchenne muscular dystrophy

This rare x-linked disease affects around 1 in 3,600 boys. It causes muscle degeneration, starting in the legs, then progressing to the arms, neck and other areas, mostly ending in premature death.

Turner's syndrome

This genetic disease affects females exclusively and is caused by a missing or partially missing X chromosome. Signs and symptoms of the disease vary, but in most cases persons affected will have a short, webbed neck, a low hairline, short stature and swollen hands and feet at birth. Women who suffer from Turner's syndrome are typically without menstrual cycles, do not develop breasts and are unable to have children.

Klinefelter Syndrome

Klinefelter syndrome is a genetic condition that results from the presence of an extra copy of the X chromosome in males. One of the main effects of the syndrome is stunted testicular growth and low sperm production. It is often not diagnosed until adulthood.

Hypophosphatemic rickets

This disorder is related to low levels of phosphate in the blood. Phosphate is essential to the formation of bones and teeth. The condition is often picked up in early childhood and signs and symptoms vary greatly from one person to the next. In the most severe cases, growth is stunted and bone abnormalities occur. The most noticeable of these abnormalities would be bowlegs or knock-knees.

Down syndrome

Perhaps the most recognisable chromosomal abnormality, Down syndrome, is caused by the presence of a third copy or partial third copy of chromosome 21. Symptoms and signs of the condition include small stature, an upward slant to the eyes, low muscle tone and mild intellectual disability. It is not hereditary and the extra chromosome occurs by random chance.

Edwards' syndrome

This condition is caused by the presence of an extra copy of the 18th chromosome, or part thereof. It is considered a very serious condition disrupting a baby's development in the womb, with many cases ending in miscarriage or stillbirth. Babies who are born with the condition rarely survive longer than one year of age.

PGT and sex selection

Before conception, preimplantation genetic testing (as part of IVF) can not only help to identify genetic defects within embryos, but also to determine the sex of the embryo, giving parents the option to choose the sex of their child.

Some people find this disconcerting as it approaches the subject of designer babies, that is, when embryos are genetically modified, not for medical reasons, but purely for desirable traits.

Morally, individuals are quite divided on the topic of interfering with embryos and ethically the world is conflicted in terms of allowing or prohibiting procedures which modify embryos for numerous reasons, however potentially beneficial for parents and unborn children.

Nevertheless, in the medical field, PGT and prenatal sex discernment is predominantly associated with preventing sex-linked genetic disorders (among many other diseases) as opposed to choosing one sex over the other.

Are there any medical drawbacks to PGT?

The risks of PGT treatment are similar to those of conventional IVF. These include ovarian hyperstimulation syndrome, multiple pregnancies and increased birth defects not related to genetics.

There is a very slight chance that embryos could be traumatised by the biopsy procedure – particular for day 3 embryos that only have a total of 8 cells in their makeup. As technology advances, however, the risk of this happening decreases.

As with any new technique or technology, there is still much to learn, therefore testing may not be 100% reliable and conclusive.

PGT is also not able to test for ALL possible genetic diseases or disorders, which means you are not guaranteed of having a healthy baby.

PGT at Bridge Clinic

In an effort to stay abreast of international medical innovation, Bridge Clinic offers all the types of PGT as a specialized service forming part of our Advanced Care.

Our affiliation with IVF Centers Prof. Zech in Austria and reputable genetic institutions ensures that our physicians are fully trained internationally and remain at the cutting edge of new technology and procedures in the field of genetics

It also ensures that our patients don't need to search for help beyond the borders of Nigeria in their pursuit of providing a healthier life for their future family.

PGT is currently available at the Bridge Clinic Fertility Centre in Oduduwa Way, Ikeja, Lagos.

Genetically or chromosomally normal embryos are selected for transfer, reducing the risk of conceiving a child affected with a known genetic disease and may increase the chance of a successful implantation and pregnancy.

More information

To find out more about Preimplantation Genetic Testing or any of our other fertility related services at Bridge Clinic Fertility Centre, please get in touch with us.

Book a consultation with any of our renowned fertility specialist online at: www.thebridgeclinic.com

Phone our Call Centre: 01 631 0092
WhatsApp: 0810 460 7791

Chat to one of our friendly personnel on our website by opening an Online Chat window

Email us at enquiries@thebridgeclinic.com

Walk in at any of our clinics in Lagos, Port Harcourt or Abuja

“Life is at the core of everything we value.”

Contact us:

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