

Pre-implantation genetic diagnosis

For couples wanting to avoid the birth of a baby with an inherited genetic disorder, prenatal diagnosis is sometimes possible, using procedures such as amniocentesis and chorionic villus sampling. These tests both involve taking samples for testing from an already established pregnancy. If the result is unfavorable, couples then have to decide whether or not to continue with the pregnancy or to have a termination. Pre-implantation genetic diagnosis may offer an alternative approach for some couples.

Pre-implantation Genetic Diagnosis

Pre-implantation Genetic Diagnosis is a relatively new procedure that can be used to prevent a couple having a child with a serious genetic disorder. It has been developed in an attempt to avoid termination of pregnancy. It involves creating embryos outside the mother's body and checking them for specific genetic conditions before implanting them in (transferring them to) the mother's uterus (womb).

Pre-implantation Genetic Diagnosis combines techniques which were developed for In Vitro Fertilisation, to create "test tube babies", together with techniques of genetic testing. It is hoped that unaffected embryos can be selected by using these two procedures, and that the resulting pregnancy will be healthy.

Pre-implantation Genetic Diagnosis is currently available only to couples at risk of having children with certain serious genetic disorders.

What is involved?

In order to create embryos for genetic testing, the ovaries (parts of the body that produce a woman's eggs) have to be stimulated artificially, using hormones, to produce several eggs at one time.

These eggs are then collected, usually under local anaesthetic. The eggs are fertilised in a laboratory using the partner's sperm sample.

Any resulting embryos are allowed to develop to a stage (approximately 8 cells) at which a small part of each embryo (one or two cells) can be removed for genetic testing. Although

experience is still limited, there is no indication at present that removing a small part of the embryo at this stage causes abnormalities in the baby.

Following this procedure, if any unaffected embryos are identified, they can then be transferred to the mother's uterus (womb). The number to be transferred will be discussed at that time, and is usually not more than two. There is an increased chance of multiple pregnancy with Pre-implantation Genetic Diagnosis.

Sadly, the outcome can sometimes be that no embryos are produced, or that none are of good enough quality for transfer to take place.

How accurate is Pre-implantation Genetic Diagnosis?

Because this is a new and technically demanding process, there is no guarantee that a correct diagnosis will be made, despite the highest standards of practice being employed. For this reason, if a pregnancy is successfully achieved, conventional prenatal diagnosis by amniocentesis or chorionic villus sampling will be offered to check the baby does not have the particular genetic disorder.

Is Pre-implantation Genetic Diagnosis available on the National Health Service?

Pre-implantation genetic diagnosis is sometimes fully or partly funded by the National Health Service (NHS), depending upon individual circumstances and local health authority policy. It is not provided routinely across all geographical areas at present. However it is hoped that this may be achieved in the future. Help and advice in obtaining funding is often given by the individual centres providing this service.

Where can I get more information?

This is a very brief summary of Pre-implantation Genetic Diagnosis and much more information is provided by the centres offering this procedure.

In the UK each centre offering this service has to be licensed and is regulated by the Human Fertilisation and Embryology Authority. It provides advice and information to people seeking fertility treatment and Pre-implantation Genetic Diagnosis.

Human Fertilisation and Embryology Authority

Paxton House,

30, Artillery Lane,

London E1 7LS

Telephone: 020 7377 5077 FAX: 020 7377 1871

Website: <http://www.hfea.gov.uk>

You can get further information through your regional genetics center and from the following addresses:-

The Genetic Interest Group

Unit 4D, Leroy House

436 Essex Rd

London

N1 3QP

Telephone: 020 7704 3141

Email: mail@gig.org.uk

Web: www.gig.org.uk

Contact a Family

209-211 City Rd

London EC1V 1JN

Telephone: 020 7608 8700 FAX: 020 7608 8701

Helpline: 0808 808 3555

e-mail: info@cafamily.org.uk

Website: www.cafamily.org.uk

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Glossary (difficult words and their meanings): Pre-implantation Genetic Diagnosis

This glossary is intended only to explain terms used in the information: Pre-implantation Genetic Diagnosis. Words shown in **bold** are defined elsewhere in the glossary

amniocentesis. A test on an unborn baby's **genes** or **chromosomes**. The baby is surrounded by fluid in the **womb**. The fluid contains a few **cells**. A small sample of the fluid is taken with a thin needle, through the skin of the mother's abdomen (tummy or belly). The fluid is sent to a laboratory for testing.

anaesthetic. Treatment given to reduce sensation for a short time. Local anaesthetic acts on a particular part of the patient's body to stop discomfort, which might otherwise be caused by a medical or surgical procedure. The patient remains awake under local anaesthetic.

cell. The human body is made up of millions of cells, which are like building blocks. There are many specialised types of cells. These include skin cells, brain cells, and blood cells. Cells in different parts of the body look different and do different things. Every cell (except for **eggs** in women and **sperm** in men) contains all the body's **genes**.

chorionic villus sampling, CVS. A test on an unborn baby's **genes** or **chromosomes**. This test can be done earlier than **amniocentesis**. A small number of **cells** is taken from the developing **placenta** and sent to a laboratory for testing.

chromosomes. Thread-like structures which can be seen under the microscope and contain the **genes**. Usually people have 46 chromosomes in every **cell**. There are two sex chromosomes and 22 other pairs of chromosomes. Twenty-three chromosomes come from the mother, and twenty-three come from the father. One chromosome of each pair comes from each parent. (As an analogy: a chromosome is like a book; a **gene** is like a story in the book).

egg. The mother's contribution to the **cell** which will grow to make a new baby. The egg contains 23 **chromosomes**; one from each pair in the mother. A **sperm** also contains 23 **chromosomes** one from each of the father's pairs. The egg joins with a **sperm** to make a complete **cell**. A baby develops from this first **cell**.

embryo. Earliest stage of human development. The embryo develops from the first **cell** in the very early stages of pregnancy. It arises after **fertilisation** of an **egg** by a **sperm**. It does not look like a baby yet, but is made up of the **cells** that will develop into a baby. It is possible for very early embryos to grow outside the mother's **womb**.

fertilisation. The union of **egg** and **sperm** to make the first **cell** that can develop into a baby. Fertilisation may take place inside the mother. **In vitro fertilisation** allows the **egg** and **sperm** to join together in a tube, in a laboratory.

gene. Information needed for the body to work, stored in a chemical form on **chromosomes**. Changes in genes alter the information and this can change how the body works. Most genes are in pairs, one from the mother and one from the father. (As an analogy: a gene is like a story in a book, changes in genes are like changes in a story).

genetic. Caused by **genes**, concerning **genes**.

hormones. Chemicals made by the body to control particular tasks such as making **eggs** or **sperm**. Hormones may also be given as medicines.

in vitro fertilisation. Joining of the **egg** and **sperm** outside the body, in a tube in a laboratory. In pre-implantation **genetic** diagnosis, this is done to allow very early **genetic** testing of an **embryo**. Unaffected **embryos** can be transferred into the mother's **womb**.

multiple pregnancy. Pregnancy with more than one baby.

ovaries. Parts of a woman's body that produce **eggs**.

placenta (or after-birth). The placenta lies against the wall of the **womb** in a pregnant woman. The baby gets its nourishment from the placenta. The placenta has the same **genes** as the baby.

pre-implantation. Before the **embryo** is transferred to the **womb**.

prenatal diagnosis. Test during a pregnancy for the presence or absence of a **genetic** disorder in the baby.

sperm. The father's contribution to the **cell** which will grow to form a new baby. Each sperm contains 23 **chromosomes**; one from each pair in the father. An **egg** contains one **chromosome** from each of the mother's 23 pairs. The sperm joins with an **egg** to make a complete **cell**. A baby develops from this first **cell**.

termination. Early ending of a pregnancy by doctors.

test tube babies. Babies resulting from pregnancies started by **in vitro fertilisation**.

uterus. Medical term for the **womb**.

womb. The part of a woman's body in which a baby grows during pregnancy.

This glossary is intended only for use by patients and families with the genetic information to which it refers.