

## What are genes?

The human body is made up of billions of cells, all of which originated from the fertilised egg at conception. While the baby is developing in the uterus, groups of cells begin to change and specialise to become different tissues and organs. Even though the cells within the heart are very different from the cells with the kidneys or certain cells in the blood, they have one common link - they all contain Deoxyribo Nucleic Acid (DNA).

DNA carries all the information needed to make a human and differs from one person to the next. It is often referred to as the blue print of life. DNA is organised into 'building blocks' known as genes which, in turn, are packaged into microscopic structures called chromosomes. The DNA within cells is responsible for creating each different person. A person's DNA blueprint is a mixture of the DNA inherited from their mother and father, which is why people commonly resemble their parents. The re-mixing of genetic information with each new generation is vital for a healthy population, but can also create problems, if the chromosomes become altered or combined in a way that doesn't work properly. Genetic diseases can run in a family from one generation to the next.

Some genetic re-arrangements may not cause serious disease but can affect fertility. If a patient is having problems achieving a pregnancy or has suffered recurrent miscarriages then it may be useful to have their genes and chromosomes checked via a blood sample. MFS offers three types of genetic screening:

- Cystic Fibrosis (CF) (see MFS infosheet: Genetic Screening 1)
- chromosome analysis (karyotyping) (see MFS infosheet: Genetic Screening 2)
- Y chromosome deletion

## What is Y chromosome deletion?

The Y chromosome is carried only by men and is important because it contains all the genes responsible for sperm production. The absence of a particular gene is referred to as a 'deletion'. Geneticists have identified regions within the Y chromosome which, if absent, cause azoospermia (no sperm in the ejaculate) or oligozoospermia (very few sperm in the ejaculate).

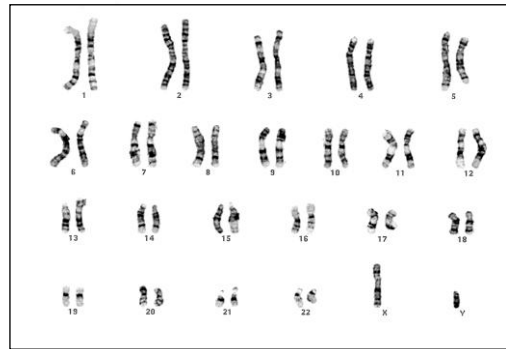


Figure 1: Male karyotype showing Y chromosome

## How is the test carried out?

A sample of blood is taken and sent to a specialist laboratory where the sample is analysed to detect the Y chromosome and any genes which are missing in the sperm producing regions.

## Who should be tested?

Men with no or very few sperm (fewer than 1 million /ml) should consider this test as they may have problems caused by a Y chromosome deletion. Sperm which carry a deleted Y chromosome are capable of producing a healthy baby using ICSI treatment, but are likely to pass on the deletion and its problems to any boys born as a result of the treatment.

## What does a positive test result mean?

Several research groups around the world are studying the effects of the Y chromosome deletions. Before assisted conception techniques became available, deletions resulting in infertility were never passed from father to son, but may do so when using sperm known to have a Y deletion in treatment. Ongoing research will establish how this may affect any offspring, and at this time, no definitive answer is available. However, it is fairly certain that boys born following such treatment may also be infertile.

## What happens if the test is positive?

A positive test result can have consequences for anyone trying to have a baby and also for their everyday life. It may even affect their close relatives. A patient will have the chance to discuss the results and any possible future fertility treatment with an MFS nurse, doctor or embryologist. It is important that the risks of continuing with treatment are explained and understood.

and any alternatives, if appropriate, can be discussed with an MFS specialist. MFS can also arrange for a patient to see a genetic counsellor, as appropriate.

**What is included in the cost?**

- Y chromosome deletion screening

**What is not included in the cost?**

- screening for any other genetic conditions
- any other fertility investigations, treatments or preservation services for either partner

**Costs**

Please refer to the current List of Charges in either the Patient Finance Information leaflet or via [midlandfertility.com/fees](http://midlandfertility.com/fees).

**Further information**

Please visit [midlandfertility.com](http://midlandfertility.com) and search for 'Investigations' or read the following MFS infosheets:

- Genetic Screening 1: Cystic Fibrosis
- Genetic Screening 2: Karyotyping
- Counselling
- Reproductive Organs
- Surgical Sperm Retrieval
- ICSI

(downloadable from [midlandfertility.com](http://midlandfertility.com) by searching for 'MFS Treatment Literature', or in hardcopy from MFS).