

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)
- Y chromosome deletion (see MFS infosheet: Genetic Screening 3)

What is a karyotype?

Chromosomes arranged in pairs according to size and shape are known as a karyotype. Most people possess 46 chromosomes (23 pairs) in each cell. Forty four of these are the same in both men and women. The remaining two are the sex chromosomes and are responsible for the differences between men and women. A woman has two identical X chromosomes while a man has one X chromosome and one Y chromosome.

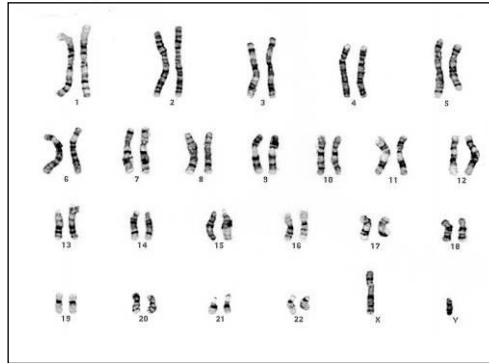


Figure 1: Male karyotype showing Y chromosome

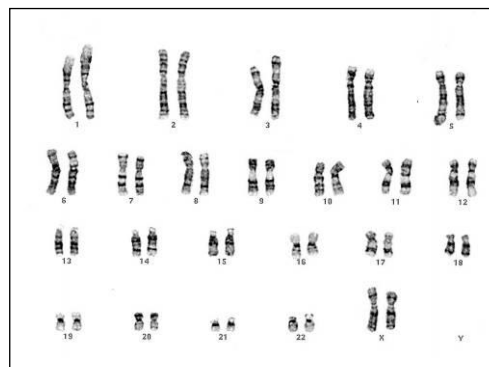


Figure 2: Female karyotype showing 2 X chromosomes

What does an abnormal karyotype mean?

Many alterations to the number or structure of the chromosomes will result in some form of physical or mental disability, often diagnosed at birth. Other rearrangements may be present in people with no disabilities and only become apparent when tested for later in life. The sex chromosomes are important as they control a person's sexual characteristics and fertility; alterations in these frequently result in a failure to conceive. For example, Klinefelter syndrome is a common condition in infertile men, characterised by small testes and azoospermia (no sperm) and is caused by the presence of an extra X chromosome.

Although many chromosomal rearrangements can occur, they are uncommon as most embryos with abnormal karyotypes either fail to implant or miscarry. Around 14 patients from every 1,000 tested will be affected by a chromosomal abnormality.

How is the karyotyping test carried out?

The patient's blood sample will be taken at MFS and sent to a specialist laboratory where it is prepared so that the chromosomes can be observed under a microscope. They are then examined by a geneticist who will arrange them into a karyotype and look for any alterations to either the number or shape of the chromosomes.

Who should be karyotype tested?

Any man with severe oligozoospermia (very few sperm in the ejaculate) or azoospermia (no sperm in the ejaculate) should have the karyotyping test. About 12% of such men, whose infertility is not caused by a blockage of the tube carrying the sperm (the vas deferens), will have a chromosomal abnormality which can be detected with a Karyotype. Some men having surgical sperm retrieval are also recommended to have the test.

A woman who has suffered from recurrent miscarriages may also be advised to have a karyotype test. The cause of miscarriage may sometimes be unidentified, although some may be due to a genetic defect. Performing karyotyping on both partners may detect the reason for the miscarriages.

Many chromosomal disorders can be inherited and karyotyping can help identify the specific gene or chromosome responsible for any genetic condition of a patient's close relatives.

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?

- chromosome analysis (karyotyping)

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.

Further information

Please visit midlandfertility.com and search for 'Investigations' or read the following MFS infosheets:

- Genetic Screening 1: Cystic Fibrosis
- Genetic Screening 3: Y Deletions
- Counselling
- Reproductive Organs
- Surgical Sperm Retrieval
- ICSI

(downloadable from midlandfertility.com by searching for 'MFS Treatment Literature', or in hardcopy from MFS).