

It's all in the genes

Blue eyes, brown eyes. Tall or short. It's all a question of what's in your genes. We look at the part genetics plays in the make up of your baby.



Every cell in our body (with the exception of the sperm and ovum) contains 46 chromosomes. These are grouped in pairs, making 23 pairs. The chromosomes act like a library of information. The chromosomes are in turn made up of long threads of a chemical that has become known as DNA (deoxyribonucleic acid). Each thread has segments called genes. It is thought that humans have around 50,000 genes. It is the genes that determine our physical characteristics such as our skin colour, height and shape of our faces, plus perhaps having some influence on our personalities, intelligence and physical talents.

Genes are also linked to our health as it is becoming clear that faulty genes can cause diseases and some genes may pre-dispose us to some conditions.

Your baby's unique inheritance

Your baby will get half of their chromosomes and genetic information from you and the other half from your partner. This is because while normal human cells have 46 chromosomes, the sperm and the egg only have 23 each. The make up of the 23 chromosomes in the man's sperm and the woman's egg is a random selection and this is why children with the same parents are not the same. At the moment of conception the sperm fuses with the egg and the chromosomes pair off. The genetic lottery has then begun. Everyone, with the exception of identical twins, has their own unique DNA make-up. Identical twins have the same DNA make up because shortly after conception, the fertilised egg splits into two.

Problems with DNA

Like most processes, conception and the copying of genes is not completely problem free. Sometimes chromosomes are damaged or the wrong number are produced. The risk of this happening increases with age as the genetic information in a woman's eggs will be the same age as her. When this happens, a woman may have a miscarriage or a child may be born with some kind of developmental delay. This is one reason why many fertility experts are keen for women who are thinking about starting a family to begin earlier rather than later.

As well as damaged or extra chromosomes, some genes that we inherit are faulty. In many cases, this has no effect on us at all as the genes are 'switched off'. Unfortunately, combined with our partner's genetic information, they can sometimes be switched back on again. This is why certain conditions such as colour blindness, cystic fibrosis and sickle cell anaemia can jump generations. If all this sounds alarming, it is worth remembering that nature tends to get it right more often than wrong and so genetic conditions are reasonably rare.

Miscarriages

A high percentage of miscarriages will be caused because nature's lottery has not worked out. The genetic information in the developing embryo can be faulty and this causes the pregnancy to stop.

Genetic COUNSELLING

Do we need it?

While nature plays a sort of lottery, sometimes the odds of having a baby with a serious hereditary illness can be predicted. For couples worried about passing on a life-threatening

disease or serious birth defect, it is now possible to find out the actual risks by going through genetic counselling.

What does genetic counselling do?

The aim of genetic counselling is to provide couples with information and support so they can make informed choices. Genetic counsellors do not push couples in any particular direction, so the couples decisions are very much their own and depend on the disease and their own feelings. In some cases, couples will decide to let nature take its course, others may ask for IVF treatment, while others may decide to use donor sperm or eggs.

How does genetic counselling work?

Genetic counselling is carried out by specialist counsellors who have a nursing or medical background. It is a very sensitive area so counsellors are chosen very carefully. Couples may be asked about their family history as well as for a blood test or a swab. The information allows the counsellor to calculate the risk of disease being passed on to a child. Alongside this information, couples are then talked through their own feelings about the risk and about the possible options that may be available.

Why do people go for genetic counselling?

Genetic counselling is not about creating designer babies. Couples tend to seek genetic counselling because they want to avoid having a baby with a serious disease or wish to understand the risk of this happening. They may already have a baby with a disorder, know that they are a carrier or have a close member of their family who has a life threatening disease.

Name of disease	Effects	Inheritance pattern	Incidence	What you can do
Sickle cell anemia	Serious and painful blood disorder.	Both parents must pass on a copy of this gene.	Found mainly in the UK among people of African and Caribbean descent. Estimated 1 in 10-40 people will carry the gene.	Consider seeking advice if you are both from this group and if either of you have close relatives with this illness.
Tay Sachs	Life threatening progressive illness.	Both parents must pass on a copy of this gene.	Found mainly in the UK amongst Ashkenazi Jews. Estimated 1 in 25 will carry the gene.	Consider seeking advice if you are both from this group and if either of you have close relatives with this illness.
Cystic fibrosis	Life threatening progressive illness.	Both parents must pass on a copy of this gene.	Found mainly amongst people of white Western European descent. Estimated 1 in 25 will carry the gene.	Consider seeking advice if you are both from this group and if either of you have close relatives with this illness.
Thalassaemia	Life threatening blood disorder.	Both parents must pass on a copy of this gene.	Found mainly in the UK among people from Mediterranean, Middle Eastern or Asian descent.	Consider seeking advice if you are both from this group and if either of you have close relatives with this illness.

The decision on whether to go for genetic counselling is a difficult choice for many couples. Perhaps both partners feel differently about the decision? Here we answer some of the common concerns surrounding this topic.



We answer some of the common concerns on genetic counselling

Q How do we get help?

A First of all, it is important not to panic and to seek the support and advice of your family doctor. She will talk through your concerns with you and will, if required, refer you for genetic counselling. It may be that while there is a member of your family with a disease, there is no risk of this being passed on.

Q I am thirty-six years old and worried about my chance of having a baby with Down's syndrome.

A The risk of having a child with Down's syndrome does increase dramatically with a woman's age. Talk through the statistics and risks with your doctor and also your partner. Tests can be carried out in pregnancy to detect babies who have chromosomal disorders. While some couples decide not to continue with such pregnancies, others do and go on to have and love their children.

Q There is a history of mental illness in my family. I am worried about passing this on.

A It is important that you start by going to your doctor and talking through your concerns. Not all types of mental illness are hereditary and while this was very much a taboo subject a few years ago, attitudes and treatment are changing. You will need to find out as much as you can about your family history, both medical and social. In some cases, people were wrongly diagnosed as having mental illnesses when in fact they had underlying emotional difficulties or medical illnesses.

Q My husband is desperate for a boy. Will we be able to ask for treatment to select only male embryos?

A Genetics is a very sensitive topic and sex selection is very carefully regulated. At present, doctors will only carry out treatment to select the sex of a baby if there is a significant medical reason for doing so. You can however try tipping the odds in your favour through some DIY techniques (see p.19), but note that these are not guaranteed.

Q I am heavily overweight and have dieted all my life. Will I be at risk of having children who are overweight?

A While it is thought that there may be some genetic link to obesity, the food that you eat and your activity level play a more important role. Put simply, your activity level must balance with the calories that you are taking in. Begin by seeking advice and support about your own diet and attitudes towards food. You can make an appointment with your family doctor, practice nurse or consider joining a reputable slimming club. This will help you to make sure that you pass on good eating habits and healthy attitudes towards food to your children.

SHOULD WE GO FOR GENETIC COUNSELLING?

Overall very few couples will need genetic counselling, but it is advisable to talk to your doctor before you get pregnant if you have any concerns.

- Does either of your parents or close relatives have an inherited disease or birth defect?
- Have either of you already had a child with a birth defect or genetic disorder?
- Have you had two or more miscarriages?
- Are you closely related e.g. first cousins or have a closely related family tree?
- You are aware that you may be at risk because of your shared racial or ethnic descent (see table on p.77)