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When you or a member of your family are first told that your child might have a genetic condition you may feel shocked, very upset, or just numb. Perhaps this news confirms what you have suspected for some time. Your doctor may have suggested genetic counselling to you. Many people are not sure what genetic counselling involves, however, or what to expect from a genetics appointment. Furthermore, many people may want to know how a condition arose in the family and whose side of the family it came from. These unanswered questions may cause tensions within a family and generate feelings of anxiety.

What are genes and how do they cause genetic conditions?

If you were to look at your skin under a microscope, then what you would see would be millions of tissue cells. In fact, every part of our body is made up of cells, rather like a house is made up of bricks. In the centre, or nucleus, of most cells in our body are thread-like structures known as chromosomes. Usually, there are 23 pairs of chromosomes (46 in total) in each cell.

Chromosomes carry genes. We can think of genes as 'strung' along chromosomes, in much the same way as beads are strung along a necklace. Genes are the instructions about how to make a new baby from a sperm and an egg - the blueprint from which the body is constructed. Genes contain all the biological information needed for us to grow and develop and remain healthy from the moment of conception to the day we die. Genes give us our physical characteristics including our eye colour, our ability to run fast as well as our susceptibility to disease.

Genes are made up of DNA (Deoxyribose Nucleic Acid). This is the code or language of the body's instruction manual. If there is a change or spelling mistake in this instruction manual, then the body is not able to function properly because it has not received the correct instructions in order to do so. The features of a genetic condition occur, or may occur, therefore, when there is a spelling mistake or change in a person's genetic material.

The functioning of our bodies requires that many thousands of genes work together. Changes or spelling mistakes in different genes have resulted in many different genetic conditions. Some genetic conditions, such as Cystic Fibrosis or Huntington's disease, are caused by changes in single genes. Other conditions, such as spina bifida, are caused by changes in a number of different genes. Chromosomal conditions are due to changes in the number or structure of chromosomes, an example of a condition caused by a change in the number of chromosomes is Down syndrome.

Does the condition run in the family?

Although it is often said that a condition 'runs in the family', not all genetic conditions are inherited or passed on in families. Some genetic conditions occur sporadically. This means that usually other members of the family are not, or will not be, affected by this condition.

Many genetic conditions, however, are inherited or passed on in families. There are a number of ways in which genetic conditions may be inherited. In some families, a condition is inherited when both parents pass on a change in a single gene to their child. In other families, a condition is inherited when a change in a single gene is passed on by only one parent. In yet other families, the likelihood of a genetic condition arising in a child depends upon whether the change in a single gene is passed on by the mother or father.

There are many genetic conditions where scientists have not been able to identify the specific gene changes which cause the features of an individual's condition. In these cases, an estimate of the likelihood that the genetic condition will be passed on or inherited is given to families.

What is genetic counselling?

Genetic counselling involves giving information to individuals and families about genetic conditions and the way these conditions are inherited. Clinical geneticists (doctors) and genetic counsellors (who may have a nursing background) provide information about the likelihood of a genetic condition happening in a family and about the medical management of a condition. Other health professionals (such as paediatricians) also play a role in the diagnosis and communication of genetic information and in the medical management of a condition. Individuals who receive genetic counselling are supported in the choices they face and are helped to make the decisions which are best for them. Genetic counselling helps individuals to deal with the psychosocial issues arising in their situation.

Who is offered genetic counselling?

Your GP or hospital doctor may have suggested genetic counselling to you or alternatively you may have sought genetic counselling for yourself. Some of the reasons for this might include:

You or your partner already have a baby or child who has a physical problem or delay in development. The diagnosis may be uncertain and either you or your doctors are wondering if there may be a genetic cause for your child's problems.

You and your partner have lost a baby during pregnancy or infancy.

You are concerned about a condition which may be genetic and you would like further information.

You or your partner have, or carry, a condition which might be passed on to your children.

There is a known genetic condition in your family or your partner's family.

You and your partner are close blood relatives.

There is a strong history of cancer in the family.

What happens at a genetics appointment?

You and your partner, and possibly other members of your family, will be seen by a clinical geneticist or genetic counsellor or both. You will spend time discussing your concerns and will be asked for information about your personal and family medical histories. A family tree will be drawn up which can give valuable information.

Sometimes you will be asked for medical details about other people in your family, if this is relevant. However, relatives will never be approached without your permission. If you do give permission, relatives may be asked to give consent for access to their medical records. If appropriate, your child or other members of the family will be offered a physical examination. Blood tests and other tests may be discussed and arranged.

If your concern is about a child in the family, he or she will usually have a detailed physical examination and the doctor may ask if photographs can be taken for the child's medical record. This helps the staff to recall the child accurately, without having to rely on memory.

When there is a genetic condition within the family this will be explained to you. The clinical geneticist or genetic counsellor will discuss ways of coping with the condition and will give information about the available medical and social support. During this time, you will be encouraged to ask questions. You might have questions about the chance of either inheriting or passing on a genetic condition. You may want to know what tests are available to confirm a diagnosis or if there are tests that can be offered during a pregnancy.

Sometimes it is not possible to make an exact diagnosis, because of the current level of scientific knowledge. However, the doctors may be able to say whether they think the condition is genetic or not, even if the diagnosis is not certain.

Often people who attend a genetics appointment will leave feeling reassured. Others will face difficult decisions such as whether or not to have a particular test or what to tell their children or other relatives about a genetic condition. Relationships within the family may be brought into sharp focus at this time. Feelings of guilt may arise

when a child is diagnosed with a genetic condition or if parents have taken the painful decision to terminate a much wanted pregnancy. Healthy members of a family may feel guilty if they have not inherited a condition. Blame may occur where only one member of a couple carries a genetic risk. Although this may be an understandable response, individuals have no control over the genes they inherit or pass on. In addition, couples aware of an increased genetic risk to their children must decide whether or not this knowledge will affect their plans for a family. Family support can be very important for individuals coping with the impact of a genetic disorder.

If you would like to know more about genetic counselling or about a genetics centre near to you, see the useful contacts section at the end of this factsheet.

Is genetic counselling the right choice?

You may like to consider the following points before committing yourself to a genetics appointment.

Why have you been referred for genetic counselling?

Could you meet or talk with a professional, such as a genetic counsellor, to discuss any concerns you may have about your appointment? If not, is there someone else you could talk with (Antenatal Results and Choices, Contact a Family)?

What are your reasons for attending an appointment? What are your reasons for not attending an appointment?

What do you hope to achieve from your appointment?

What questions are you going to ask the geneticist or genetic counsellor?

How will you deal with the information you are given? Who are you going to tell afterwards?

The Genetic Interest Group (GIG) has suggested some 'helpful hints' prior to attending a genetics appointment in a leaflet entitled '*Has your child a genetic disorder? What do you need to know from whom?*' These include:

Take a pen and paper with you to write down the answers.

You could take a tape recorder with you if you want to record the meeting.

Take a friend; they will remember more and can write down information.

Don't be afraid to ask what you really want to know.

It is common practice for the genetic staff to send you a letter after the appointment, summarising the main points of discussion. A copy may be sent to your GP or the doctor who referred you. You could ask at the start of the appointment if this is to be done, as this may enable you to listen carefully without having to take notes.

Common beliefs about genetic counselling

The following statements reflect some commonly held views about genetic counselling:

At the appointment I can find out if a condition is genetic by having a blood test done to look at my genes.

People often think that there is a test for every condition. This is not true. Genetic tests are available but for only a certain number of conditions. If a test is available, then the doctor or counsellor will spend time discussing the reason for the test, its accuracy and the implications of the result. The extent of counselling and the issues to be addressed will depend upon a person's situation and the type of test being offered. Individuals are required to give informed consent before a blood test can be done.

The doctor or nurse is going to tell me that I shouldn't have any more children.

Genetic counselling aims to be non-directive and non-judgemental and this means that doctors and counsellors refrain from telling individuals what they should or should not do. By discussing all the options available in a given situation, doctors and counsellors enable individuals to make their own decisions and offer support for the decisions individuals make.

Genetic counselling is the same as psychological counselling.

Often individuals are reluctant to attend a genetics appointment because they do not consider themselves to have a 'psychological' problem. Genetic counselling differs, however, from psychological counselling. Genetic counselling is a process of communication between doctor or counsellor and individual or family in which the medical and psychosocial issues associated with the occurrence or risk of occurrence of a genetic disorder in a family are discussed.

The Internet

Many people use the Internet to search for genetic information. However, not all information available on the Internet is relevant to each individual or family situation, and furthermore not all the information will be reliable. Importantly, therefore:

Any information you obtain from the Internet should be discussed with your doctor or counsellor.

The website should be from a reputable institution such as a hospital, university or government body. Furthermore, information about a condition should be authored and dated.

Useful contacts

Antenatal Results and Choices (ARC)

73 Charlotte Street, London W1T 4PN

Tel: 020 7631 0285 Helpline

Tel: 020 7631 0280 Admin e-mail: info@arc-uk.org

Web: <http://www.arc-uk.org>

Information and support to parents at the time of antenatal testing and when fetal abnormality is detected.

Genetic Interest Group (GIG)

Unit 4D, Leroy House, 436 Essex Road

London N1 3QP Tel: 020 7704 3141

e-mail: mail@gig.org.uk Web: <http://www.gig.org.uk>

A national alliance of organisations which promotes awareness and understanding of genetic disorders.

Unique - Rare Chromosome Disorder Support Group

PO Box 2189, Caterham CR3 5GN

Tel: 01883 330766 e-mail: info@rarechromo.org

Web: <http://www.rarechromo.org>

Information and support to families of children with any rare chromosome disorder.

Additionally, the Contact a Family website, Web: <http://www.cafamily.org.uk> has a glossary of genetics terms, information on patterns of inheritance and addresses of regional genetics centres. Our freephone Helpline, Tel: 0808 808 3555, can also give information and support.

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