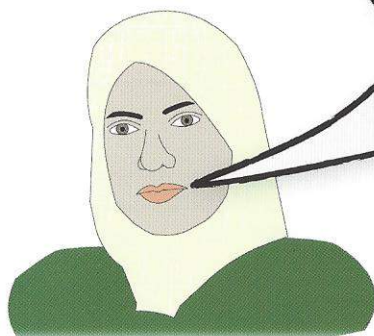


Appendix 8 Leaflets

Birmingham Women's **NHS**
NHS Foundation Trust

Genetic Risk Explained for Parents who are blood relatives (Consanguineous)

Information for patients and families



In this leaflet I will explain
the possible genetic risks linked
to marriage to a blood relative and
provide information, guidance and
advice.

Other leaflets in this series about genetics

Understanding Genetics and Inheritance
Sharing Genetics Information with Relatives

Genes

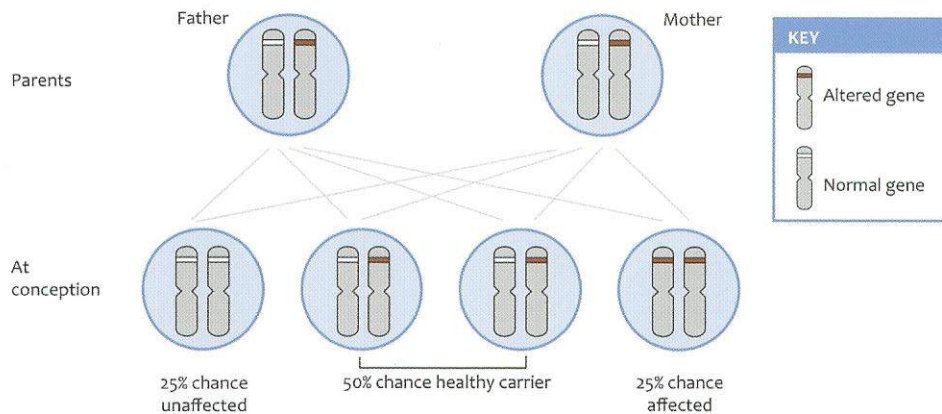
We inherit the 'instructions' which determine how our bodies work and develop from our parents, these instructions are called genes. We have about 30,000 human genes. Each cell in our body contains two copies of most of our genes, one inherited from the father and one from the mother.

Recessive Disorders

Sometimes inheritance of genes that do not work properly can lead to a genetic disorder. Throughout the rest of this information leaflet we refer to these genes as the 'altered gene'. One type of genetic condition is one inherited in a pattern which we call recessive inheritance. In recessive genetic disorders both copies of the gene are altered. Reassuringly although we often have one altered copy of a gene, the second copy is nearly always normal, this normal copy of the gene can compensate for the altered gene and there is no effect on our health. This is why this type of altered gene is called a recessive gene, as it can be passed on for generations with no ill effect. Individuals with just one altered recessive gene are known as carriers. In fact it is estimated that we are all carriers of a handful of recessive genetic conditions.

Having Children with Recessive Genetic Disorders

Healthy carrier parents who both carry the same copy of an altered recessive gene have a risk of having a child with a recessive genetic disorder.



There are four possible outcomes for each pregnancy regardless of the baby's sex

There is a 1 in 4 (25%) chance that the child will have two normal copies of the gene

There is a 2 in 4 (50%) chance that the child will be a healthy carrier of the condition

There is a 1 in 4 (25%) chance that the child will have inherited the altered gene from the mother and father and therefore have the condition

Consanguinity and Recessive Inheritance

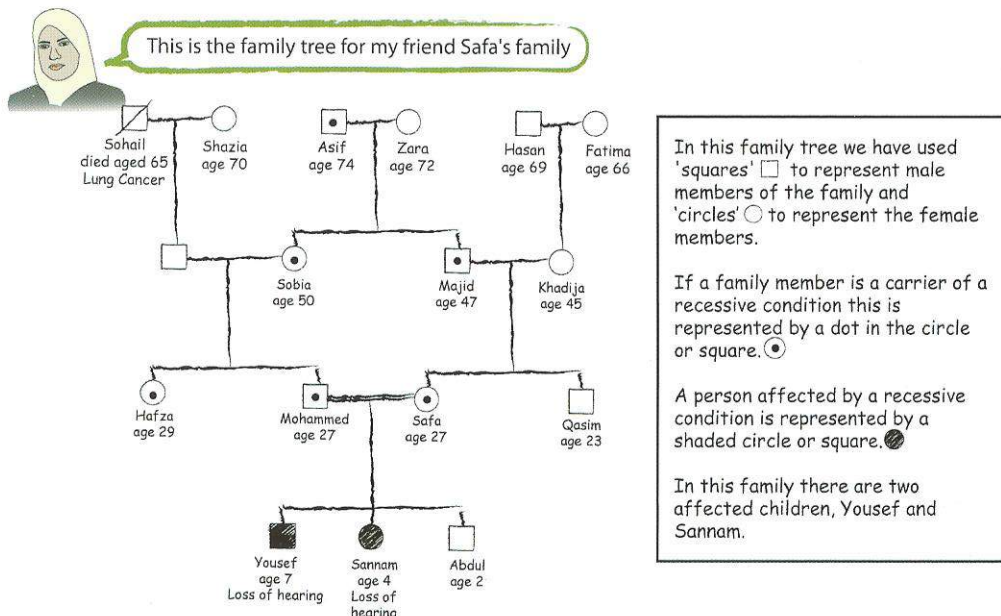
Marriage between close blood relatives (e.g. first cousins) is known as a consanguineous marriage. Consanguineous marriages are common in many parts of the world and are an integral part of many cultures often with many benefits.

People from the same family will share some of their genes as they have inherited them from common ancestors. This is why if two people from the same family get married there is an increased chance that they may both carry the same altered recessive gene.

Children of unrelated parents have on average a 2-3% chance of being born with a genetic disorder, this risk is doubled to 4-6% for children of consanguineous parents. This small additional risk is because these recessive genetic disorders are more common in consanguineous marriage. But it is important to remember that 90% of children will be born healthy and free from genetic disease.

Carrier testing

Healthy carriers of a recessive disorder may or may not be aware they carry an altered gene. In some families where the alteration in a specific gene has been identified, there may be a simple test that can be offered to possible carriers of the genetic condition. The genetics services will be able to help families and provide further guidance and advice.



Key information for families

- If you know of someone with a genetic disorder, then their parents and possibly other close relatives may be carriers of the disorder.
- Families may be able to seek advice and information and have carrier testing if the gene alteration is known.
- Those who wish to obtain genetics advice should contact their GP who can refer to the clinical genetics services.

Genetics Information

For further genetics information please see our other leaflets and DVD:

- Genetics and Inheritance
- Sharing Genetics information with Relatives
- Understanding Genetics DVD

Or if you wish to speak to someone you can contact the Birmingham Women's Hospital Clinical Genetics Unit either by telephone on 0121 6272630 or by e-mail at egsp@bwhct.nhs.uk

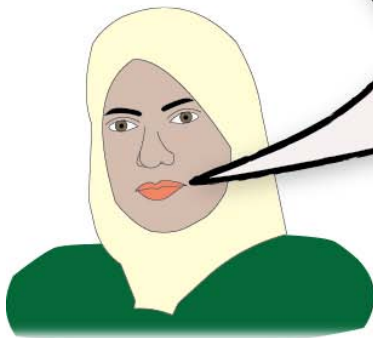
This information leaflet has been produced as part of a Health Initiative. This health initiative is the Enhanced Genetics Services project funded by the Heart of Birmingham PCT and the aims of the project are to raise awareness of genetics amongst the community so that they can make informed choices for themselves and their families.

Clinical Genetics Unit
Birmingham Women's NHS Foundation Trust
Mindelsohn Way
Edgbaston
Birmingham
B15 2TG



Sharing Genetics Information with the Family

Information for patients and families



In this leaflet I will offer
guidance to those people who
need to discuss genetics information
with their families.

Other leaflets in this series about genetics

Understanding Genetics and Inheritance
Genetic Risk Explained for Parents who are Consanguinous

Key information for families

Families with a child that has a recessive genetic condition should seek further information on ways to share this information with the wider family.

Individuals and or families who wish to seek genetic information about carrier testing should contact their GP who will be able to arrange an appointment with a genetics specialist .

Genetics Information

For further genetics information please see our other leaflets and DVD:

- Genetics and inheritance
- Sharing genetics information with relatives
- Understanding genetics DVD

Or if you wish to speak to someone you can contact the Birmingham Women's Hospital Clinical Genetics Unit either by telephone on **0121 627 2630** or you can go to the website

www.talkinggenetics.co.uk

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Recessive Genetic Disorders

The diagnosis of an inherited genetic disorder in a family often means that brothers and sisters may also be at risk of the same condition or may be carriers of the condition, as well as other close family members such as aunts, uncles, cousins. This is something they will all need to think about when they come to have children of their own.

For some genetic disorders it may be possible to have a genetic test to confirm if you are a carrier for the disorder.

Sharing Information—what are the Issues

If there is a genetic test available for the genetic disorder in your family then it is possible to have a simple blood test that will tell you whether you are a carrier of the condition. You may choose to have a genetic test so that you can share this information with family members on whom it may have an impact.

It is often very difficult to discuss a genetic disorder that affects your child or yourself with other family members. But do remember that a genetic disorder in a family is no one's fault. Many people prefer to have a genetic test as it confirms their status, and they can use this information to make informed decisions about their own life.

Ideas for sharing genetic Information with the family

- Discuss sharing genetic information with your genetic counsellor and close family members.
- We can offer information leaflets or write information letters for relatives
- Maybe take a family member along to the genetics session where you discuss the condition and its implications for you and your family. Then if you wish this family member can help you make contact with other family members to share the information.
- How you choose to discuss test results will depend on your relationship with individual family members

Consanguineous Marriages

It is particularly important for individuals and families who choose to marry a blood relative (consanguineous) to obtain information and guidance from a genetics specialist about genetic disorders and inheritance.

Relatives who are at a risk of being carriers for an inherited genetic condition are encouraged to talk to their doctor who can arrange for you to see a genetics consultant and/or a counsellor.

This is a quote from a recent study¹: 'patient groups commented that although no one wants to have a diagnosis of a genetic condition, that an accurate diagnosis can open doors to effective and appropriate support, disease management, and treatment, equally the lack of diagnosis can mean that the families may not get the best care'

Information and Support for families

The issues that affect people with a genetic disorder do not exist in isolation and have an impact on all the members of a family, parents siblings and the wider family.

An understanding of the facts and issues related to a particular genetic condition can make living with the condition a little easier. Knowledge can empower you and help you access the right services. Talking about concerns and sharing them can make things easier to deal with.

Patient support groups can be a great source of information and can provide details of services that you may need as well as emotional support.

Support groups:

Genetics alliance UK

Talking genetics

CLIMB (children living with metabolic disease)

Contact –a-a Family

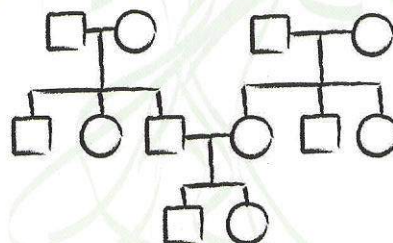
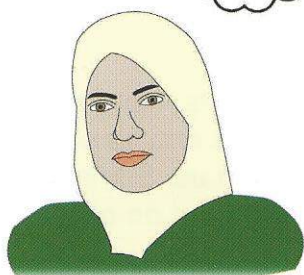
UNIQUE (rare chromosome disorder group)

UK Genetic Testing Group

Understanding Genetics and Inheritance

Information for patients and families

Have you ever wondered why members of the same family look alike or perhaps why you look like a brother or sister?

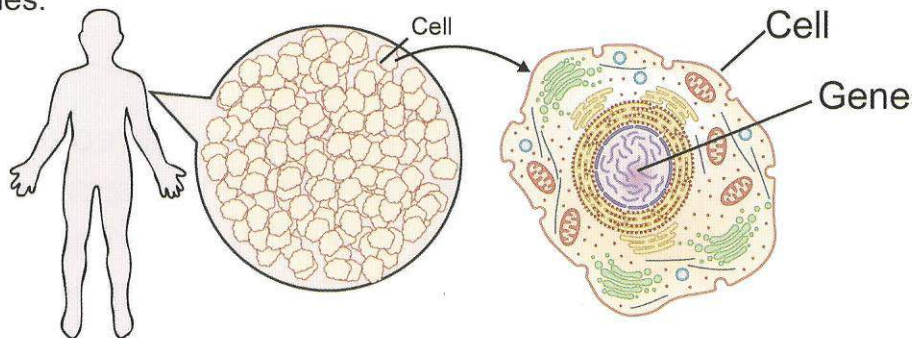


What is Genetics and Inheritance?

Genetics is the study of how characteristics such as features and appearance are inherited (passed on) from our parents to us. These traits are passed on from our parents in our genes. Genes contribute to the way we look and how our body is put together. We have two copies of most genes:

- One copy from our mother
- One copy from our father

Our body is made up of millions of cells and each cell contains our genes.



We have about 30,000 genes and they act as instructions for our cells, and help determine:

- Our appearance like our eye colour, hair colour, features, etc
- Our development
- Overall health of our body

Similarly, our parents' genes come from our grandparents – and our grandparents' genes come from our great grandparents and so on. A great long line of people have passed on their genes to us. Sometimes we can have changes in our genes that can have an effect on our health.

Why is Genetics important to you?

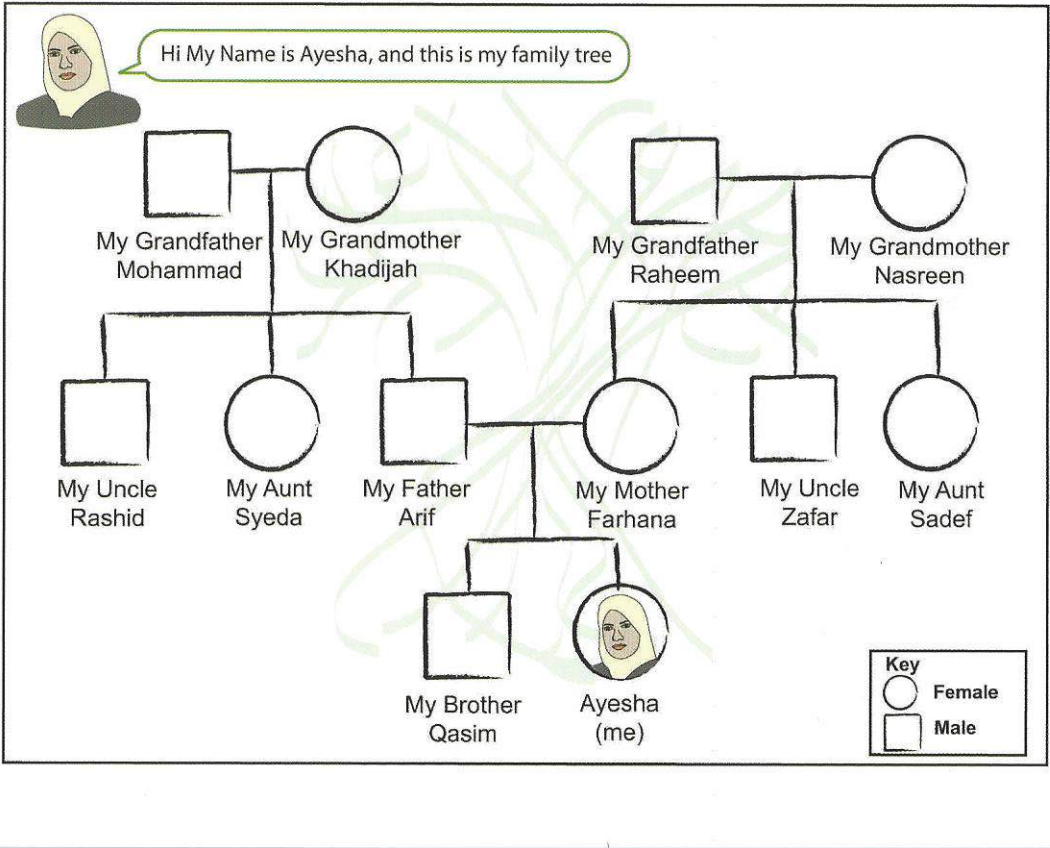
There are different types of illnesses, some of which are a result of our environment and some we experience as our body changes as we get older.

In addition some health concerns are genetic. There are many different types of genetic conditions and these are inherited in different ways. Some genetic conditions occur due to changes with certain genes. Some common examples include:

- Thalassaemia
- Sickle cell disease

Inheritance can be complicated, and its effects vary depending on the genetic disorder involved

Family Tree



Families that are well informed about genetic conditions:

- Are in a stronger position to get services they need and are entitled to.
- Have knowledge to communicate their needs to with family members, friends and schools/colleges.
- Will hopefully deal better with the genetic condition and any associated disability.

Who can Help?

If you wish to speak to a specialist in genetics then please ask your GP to refer you to the West Midlands Clinical Genetics Service.

Genetics Information

If you wish to get more information on genetics and related resources or wish to speak to someone please contact the Clinical Genetics Unit at the Birmingham Women's NHS Foundation Trust either by telephone on **0121 627 2630** or by e-mail at egsp@bwhct.nhs.uk.

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