## Genetic testing can be used to find out whether a person is carrying a specific altered gene (genetic mutation) that causes a particular medical condition.

It may be carried out for a number of reasons, including to:

- diagnose a person with a genetic condition
- help work out the chances of a person developing a particular condition

• determine whether a person is a carrier of a certain genetic mutation that could be inherited by any children they have

You'll usually need a referral from your GP or a specialist doctor for genetic testing to be carried out. Speak to your doctor about the possibility of testing if you think you may need it.

## What does genetic testing involve?

Genetic testing usually involves having a sample of your blood or tissue taken. The sample will contain cells containing your DNA.

It can be tested to find out whether you're carrying a certain mutation and are at risk of developing a particular genetic condition.

In some cases, genetic testing can be carried out to find out whether a baby is likely to be born with a certain genetic condition.

This is done by testing samples of the fluid that surrounds the foetus in the womb (amniotic fluid) or cells that develop into the placenta (chorionic villi cells), which are extracted from the mother's womb using a needle.

Depending on the condition(s) being checked for, the fluid or cell samples will be examined and tested in a genetics laboratory to look for a specific gene, a certain mutation on a specific gene, or any mutation on a specific gene.

In some cases, it may be necessary to check an entire gene for mutations using a process called gene sequencing. This has to be done very carefully, and can take a long time compared with most other hospital laboratory tests.

Depending on the specific mutation being tested for, it can take weeks or even months for the results of genetic tests to become available. This is because the laboratory may have to gather information to help them interpret what's been found.

It isn't always possible to give definite answers after genetic testing. Sometimes it's necessary to wait to see if the person being tested, or their relatives, do or don't develop a condition. Other tests may need to be performed.

## Genetic counselling

If your doctor thinks genetic testing may be appropriate for you, you'll usually be referred for genetic counselling as well.

Genetic counselling is a service that provides support, information and advice about genetic conditions.

It's conducted by healthcare professionals who've received training in the science of human genetics (a genetic counsellor or a clinical geneticist).

What happens during genetic counselling will depend on exactly why you've been referred.

It may involve:

- learning about a health condition that runs in your family, how it's inherited, and which family members may be affected
- an assessment of the risk of you and your partner passing an inherited condition on to your child
- a look at the medical history of your family or your partner's family and drawing up a family tree
- support and advice if you have a child affected by an inherited condition and you want to have another child
- a discussion about genetic tests, which can be arranged if appropriate, including the risks, benefits and limitations of genetic testing
- help understanding the results of genetic tests and what they mean
- information about relevant patient support groups

You'll be given clear, accurate information so you can decide what's best for you.

Your appointment will usually take place at your nearest NHS regional genetics centre. The British Society for Genetic Medicine has details for each of the <u>genetics centres</u> in the UK.

The following information is from Genetic Alliance UK regarding access via the NHS

Genetic Alliance UK Level 3, Barclay House 37 Queen Square London WC1N 3BH

# PHONE +44 (0) 20 7831 0883

EMAIL contactus@geneticalliance.org.uk

## SOUTH THAMES

## **S E Thames Regional Genetics Service**

The Genetics Department, 7th Floor, New Guy's House, Guy's Hospital, LONDON SE1 9RT Tel: 020 7188 1364 Fax: 020 7188 1369 Catchment areas: Kent, E Sussex, Bromley, Greenwich, Bexley, Lambeth, Southwark, Lewisham

## **S W Thames Regional Genetics Service**

Department of Medical Genetics, St George's University of London, Cranmer Terrace, LONDON, SW17 0RE Tel: 020 8725 2038 Fax: 020 8725 3444 Catchment areas: Ashford New River, SW London, Surrey, W Sussex

## OXFORD

## **Oxford Regional Genetics Service**

Department of Clinical Genetics The Churchill Hospital, Old Road, Headington, OXFORD OX3 7LJ Tel: 01865 226009 or 01865 226028 Fax: 01865 223572 Catchment areas: Oxfordshire, Northamptonshire, Buckinghamshire, Berkshire, Swindon.

## NORTH THAMES

## **N E Thames Regional Genetics Services**

Clinical Genetics Department, Great Ormond Street Hospital for Children NHS Trust, Great Ormond Street, LONDON, WC1N 3JH Tel: 020 7762 6831/6856/6786/6845 Fax: 020 7813 8141 Catchment areas: North and East London and Essex.

## **N W Thames Regional Genetics Service**

Kennedy Galton Centre, Level 8V Northwick Park & St Marks NHS Trust, Watford Road HARROW, Middlesex HA1 3UJ Tel: 020 8869 2795 Fax: 020 8869 3106 Catchment areas: Hertfordshire, Bedfordshire, Barnet, Hillingdon, Brent & Harrow, Ealing, Hammersmith, Hounslow, Kensington, Chelsea, Westminster, Edgware,

## **N W Thames Regional Genetics Service**

#### Services we offer

We offer genetic counselling and genetic diagnosis to individuals and families living in the area. We are based at Northwick Park Hospital and offer outreach clinics in different hospitals across the region.

There is close co-operation between the laboratory and clinical services and the centre prides itself on providing an integrated clinical genetics service. Please <u>click here for detailed information about the laboratory services</u> for professionals.

#### Information about the genetics clinic for individuals and families

#### How can I get an appointment in the genetics clinic?

If you are concerned about your health, the health of a family member, or your family history, you should ask your family doctor (GP) in the first instance. They may answer your question or refer you on to our service or another service that can help.

#### What happens at a genetics appointment?

Our leaflet for people with a genetics appointment is available here.

We hold clinics at several different hospitals across the region and will usually arrange to see people in the clinic closest to their home. Your appointment letter will state where the clinic will be held.

#### **Information about the clinical genetics service for professionals** Referrals for all genetics clinics should be made to:

Consultant Geneticist North West Thames Regional Genetics Service Level 8V, Northwick Park and St Mark's Hospitals Watford Road Harrow HA1 3UJ

Tel: **020 8869 2795** Fax: **020 8869 3106**