

Information for patients and the public about informed consent

INFORMED CONSENT SUMMARY:

- ***'Consent' means permission. Health professionals must have your permission to carry out any procedure.***
- ***You don't have to agree to anything that you don't want to or are not comfortable with.***
- ***You are always entitled to ask for more information if there is anything you don't understand about your treatment.***
- ***In emergencies decisions may have to be made quickly, but in other procedures, such as genetic tests, it is usually ok to take as long as you need to make a decision.***
- ***Where the person undergoing a procedure is a minor, or has a mental impairment, a responsible adult or carer may need to make a decision on the person's behalf.***

Informed Consent

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What is Informed Consent?

Informed consent is a process where you decide whether to:

- ***Undergo a medical procedure or treatment***
- ***Take part in an experiment or research programme***

You will want to understand the relevant medical facts and any risks involved in any procedure you are considering. Your doctor, nurse or a researcher can provide you with information, and help you to understand it.

A successful informed consent process is:

- ***A collaboration between health professional or researcher and patient or research participant***

- ***One which will ensure that the donor or research participant is informed and can make decisions that they are happy with, and enter into freely.***
- ***One which ensures that the donor or research participant and his or her genetic material is protected.***

When you give your consent to a procedure, you will be asked to sign a form to show that you have given your permission to the procedure in question. Visit the following URL for an example of a generic consent form:

http://www.p3gobservatory.org/download/Modelconsentform_Finalnov6.doc

Why is informed consent important?

Medical knowledge is specialist in nature. This means some of the terminology involved in procedures may be difficult to understand.

So that you can decide freely whether or not to take a genetic testing or become part of a research study, and what the implications for you will be if you do.

Genetic testing aims to:

- ***Establish or diagnose what kind of disease you may be affected by. A correct diagnosis and knowing what disease you have is the first step towards treatment.***
- ***Find out if you are a 'carrier' of a genetic condition, i.e. you are not affected yourself but could pass the defective gene that causes it on to your children. For more information about how this happens, see the section explaining inheritance.***

Genetic research aims to:

- ***Cure and prevent diseases, and improve the lives of those affected by them.***
- ***Improve the way health services are provided to patients.***

Continued research will help thousands of people affected by genetic disorders all over Europe. Research depends on people like you participating in a range of ways, all involving taking personal data of different kinds, depending on the aims of the particular study.

- Information on genetic testing in the UK and what is involved can be found at the website for the United Kingdom Genetic Texting Network:
<http://www.ukgtn.nhs.uk/gtn/Home>
- Contact details for regional genetics centres in the UK can be found at:
http://www.bshg.org.uk/genetic_centres/uk_genetic_centres.htm
- Information on genetic testing around the rest of Europe can be found at the EuroGenTest website: <http://www.eurogentest.org/>

How does it affect me?

Whether you are already a patient, or are considering taking part in a research program, the aim of the informed consent process is the same. Some of your concerns may be different depending on which group you are from however.

As a patient:

If you are a patient about to take a test, then you will already be considering the possibility that you or your family will be affected by a genetic condition.

You should be told exactly what the test involves, and what your result will mean. Not all tests will always give a very clear answer, and this should be made clear to you if this is the case in your situation.

The informed consent process is important as the test result may have a big impact upon you or your family. You may, after having been informed of the risks and the probable outcomes, decide that you would prefer not to take a genetic test.

Taking part in research:

If you are considering becoming part of a research program, you are not necessarily considering the possibility of discovering that you will be found to be a carrier of a genetic disorder. You may have questions about how your data is stored and what it is used for though.

- This means that what you ask of the researcher may be different from if you were patient.
- Even so, the informed consent process is similar in both cases.

You might only want to take part in a research program after being satisfied:

- ***That you have all the relevant information from those conducting the research.***
- ***That this has all been clearly explained to you.***
- ***That the researchers have sought to answer fully any questions that you have.***
- ***That you know whether you will be able to find out the results of the research.***

It is the responsibility of the researchers looking after you to be sensitive to your particular situation and needs.

As a test you might ask yourself questions such as 'Am I happy with the amount of information I have been provided with?', or 'Can I explain back to my doctor the risks that have been explained to me?'.

Risks and your rights in research

Two issues to think about when considering whether or not to take part in research:

- ***What will happen to your DNA sample after it has been taken?***
- ***Will it be used for a different study in the future?***

Some research projects are done by groups of researchers in different countries. Different countries have different rules about how your personal data can be used, so you may wish to find out who else will be carrying out the research and in which countries.

- All countries in the European Union have laws governing the privacy of personal data, and these can be found via the information commissioners for each country, via the EU website: <http://europa.eu/>
- In 2003, UNESCO made a declaration on the use of human genetic data, and protecting the privacy of the donor. Countries will have to observe and follow these laws and guidelines if they want to be able to participate in genetic research. The declaration can be found here: <http://unesdoc.unesco.org/images/0013/001312/131204e.pdf#page=27>

Types of consent

When you are asked by your doctor, nurse, or other health professional if you will agree to give over DNA samples for research or testing, they may ask you for consent at two different levels:

- **Consent for your samples to be used in a certain, specific test or tests, or research program.**

OR

- **For 'blanket' permission to use your samples in future research projects.**

Once you have allowed your samples to be taken, it may be difficult to withdraw them if you decide that you do not feel comfortable. So, you might wish to give some thought to the kinds of tests or research that you would and would not be happy for your samples to be used in.

A good informed consent procedure should eliminate the risk of your samples being used in a way that you are not happy with.

The advantages of the 'blanket' consent option has several advantages for research:

Uncontactability

If you move to live elsewhere it may be difficult or impossible to re-establish contact with you. Agreeing in advance to give consent for further research studies will help to ensure that publicly valuable research can still be carried out in these circumstances.

Efficient use of resources

It may be very costly and time consuming to try and trace previous donors to re-establish consent, and there is no guarantee that all donors will be traceable.

Attempting to contact all previous donors may also use resources that could be used more directly for research and development into treatments for disease.

So it is more efficient if research subjects are prepared to give overall consent for the further use of their samples, in the future, and maintain the pace of research towards bringing about benefits to public health.

Consent 'fatigue'

You may not want to be asked repeatedly each time somebody wishes to carry out research using your DNA sample.

- Often the work is highly technical and may take some time to understand. You might decide that to avoid this you are happy to give consent to future, and as yet unknown research studies by giving blanket consent.
- All countries in the European Union have strict laws governing the privacy of personal data. They have specific recommendations and guidelines about the best way to deal with personal data samples.
- Countries will have to observe and follow these laws and guidelines outlined in the 2003 UNESCO declaration on the use of human genetic data and protecting the privacy of the donor if they want to be able to participate in genetic research.
- Donor privacy can also be achieved by the anonymisation or encryption of the DNA samples. See the EuroGenGuide section on Biobanking.

Anonymisation, Encryption and Consent

This is dealt with fully in the section dedicated to biobanking, however here is a brief explanation:

- There are different kinds of genetic studies, which use samples from different types of biobank, depending on what is being investigated.

- DNA samples can be 'un-linked' from information about who they came from in several ways. There are a number of different levels of this, where at the most private extent, it is impossible to re-identify donors from their samples.
- You may be uncomfortable with taking part in research where you can be identified from the samples you donate, whereas you may be happy to be involved if you can be sure that your identity is being protected.
- However for some studies it may be necessary for you to be identifiable as there may be outcomes of arising from the research that you could benefit from
- It may depend on the type of research that is being done whether or not samples are anonymised or encrypted, so if you have concerns about this, it is something that you may want to raise with the researchers dealing with you. and this particular study.
- You are entitled to ask him or her as much about this as you wish to in the interests of being able to make a decision with which you are completely happy.

Those unable to give consent, e.g. children, mentally incapacitated adults

Guidelines for helping people who may not be competent to make fully rational decisions about their own treatment, such as children, the mentally handicapped, or the very old are also dealt with in their own section.

Laws governing genetic testing of children differs throughout Europe differ from country to country, but a breakdown of the various legal positions can be found via this link: http://www.bionetonline.org/English/content/gh_leg2.htm

Different types of genetic tests

- **Diagnostic testing:** Diagnostic testing is used to diagnose or rule out a specific genetic condition. In many cases, genetic testing is used to confirm a diagnosis when a genetic condition is suspected based on physical mutations and symptoms. Diagnostic testing can be done at any time in a person's life, but is not available for all genetic conditions.
- **Carrier testing:** This is done to identify people who carry one copy of a gene mutation that, when present in two copies, causes a genetic disorder. This is often offered to individuals who have a family history of a genetic disorder, or to people in ethnic groups with an increased risk of specific genetic conditions. If both parents are tested, the test can provide information about a couple's risk of having a child with a genetic condition.
- **Predictive testing:** Predictive tests are used to detect gene mutations associated with disorders that appear after birth, often later in life. These tests can be helpful to people who have a family member with a genetic disorder, but who have no features of the disorder themselves at the time of testing. Predictive testing can identify mutations that increase a person's chances of developing disorders with a genetic basis, such as certain types of cancer.
- **Newborn screening:** This is the most routine form of testing and is done to infants just after birth to identify genetic disorders that can be treated early in life, for example phenylketonuria (a genetic disorder that causes mental retardation if left untreated) and congenital hypothyroidism (a disorder of the thyroid gland).
- **Prenatal testing:** This is done to detect changes in a foetus's genes or chromosomes before birth, and is offered to couples with an increased risk of having a baby with a genetic or chromosomal disorder. Sometimes, prenatal testing can lessen a couple's uncertainty or help them decide whether to abort the pregnancy. However, prenatal testing cannot identify all possible inherited disorders and birth defects.