

Information for patients and the public about genetics and vulnerable groups

This section covers some of the concerns that both able-bodied and disabled men and women from different backgrounds and ethnic groups might have when they

- ***Try to access medical advice about genetic conditions***
- ***Consider taking a genetic test***
- ***Are considering donating biological samples or becoming involved in research***

All of us feel vulnerable when faced with the prospect of illness and disease. Some groups are even more vulnerable.

These groups not only have to deal with the physical and psychological consequences of their condition, but also with other factors, such as the availability of funded genetic services where they live, their age, ethnic background, mental health or a learning disability.

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Availability of funded genetic services

Healthcare systems can have different priorities when they decide on which services to fund.

Studies show that there is a mismatch in Europe between medical need and the genetic services needed to meet those needs.

So it may be that, in your country, lack of funding for genetics could affect the range of genetic services available to you, especially if you or a member of your family is affected by a very rare disorder.

This does not mean that there is no help available. There may be a patient group or support organisation that can help you elsewhere in Europe.

You can use the EuroGenTest patient section to find a genetic test, a support group, or to find out your rights as a European patient, by following this link:

<http://www.eurogentest.org/patient/>

You can also use the EuroGenGuide web forum to try and find other people and groups around Europe who are affected by the same condition.

Genetic services for rare disorders

- If you have a rare disorder, significant funds may not have been spent on providing tests and treatments for it, and at first it may be difficult to find where you can get the help you need.
- Also, if genetic services are limited in your country, your doctors may not know where you can find the information and support you need. Patients with rare disorders have the same rights as any other patient.
- Orphanet is a European organisation which provides comprehensive information about rare diseases. In Europe, a disease defined as rare when it affects one in 2000 people. Their website can be found here: <http://www.orpha.net/consor/cgi-bin/index.php>

- You can use the EuroGenTest patient section to find a genetic test, a support group, or to find out your rights as a European patient, by following this link: <http://www.eurogentest.org/patient/>

Genetic research into rare disorders

- Families affected by rare disorders are small in number and only a few researchers may be working on this condition.
- This can mean that the families all know one another or the researchers through patient support groups or direct links with the researchers. This may mean that anonymity within this circle is difficult.
- It is also important for people with rare disorders not to feel pressured into participating in research.
- Experience shows that families with rare disorders usually want to help research proceed as quickly as possible. They are the one who have most to gain and most to lose. An in-depth discussion of the issues can be found on the Genetic Interest Group website, here: <http://www.gig.org.uk>

Age

Your age may be a factor that will affect your ability to make decisions about complex issues such as whether or not to take a genetic test or become involved in research.

Normally, adult people are able to make rational decisions about their own lives and their own bodies.

However, there are some life stages in which that may be difficult or not possible. For example, children are often not mature enough to make fully rational decisions.

- Adolescents, even those who are nearing adulthood, are vulnerable: they may appear more able to make serious decisions about their health than they actually are.

- In old age people sometimes suffer from dementia or Alzheimer's Disease, and because of that may be unable to make decisions for themselves.
- When the consent of a legal representative is required, the person (whether child or adult) must be involved in the decision to the fullest extent possible for them.
- In the case of an adult who is unable to consent or make an informed choice about having a genetic test, the test must be in their interest, for example, to improve treatment and care.
- If you are looking after a relative who, because of their age, is unable to give consent or make an informed choice about genetic testing or involvement in research, you may find it difficult to work out what is in their best interest and the interest of their family.
- You might feel a conflict of interest, because you want to support them by agreeing to a genetic test or involvement in research, but you may also want to protect them by not releasing their personal information to other people.
- The health professionals involved should answer any questions you have so that you understand what a genetic test result will mean for the person, and the nature of any risks involved in the treatment or research.

Genetic testing and children

There are special issues involved in testing children for genetic conditions.

There is a disagreement among health professionals about whether to test children for genetic conditions which have adult onset and for which there are no treatments.

This decision can face parents and families with a very difficult dilemma.

It is usually recommended that genetic testing in children, including carrier testing, is postponed until they can understand what is involved and give consent.

The exception to this is when there is a specific medical benefit in carrying out predictive tests on a child.

- An in-depth discussion of the issues and good practice recommendations can be found in the European Journal of Human Genetics, by following this link:
<http://www.spgh.net/docs/ESHG%202009%20EJGH%20recommendations%20minors.pdf>
- The support and advice of a genetic counsellor in these circumstances can be a great help.

Ethnic background

Some ethnic groups are more likely to have particular genetic conditions than the rest of the population.

- If you are from an ethnic group that has a high risk incidence of a particular disease and you're worried that you are at risk, you need to speak to your doctor or health professional and discuss your concerns.
- If you are from a particular ethnic group that has a high known incidence of a particular condition, then your doctor may offer you a test for the disease, in recognition of your ethnicity and the factors that are particularly relevant to it.
- If you live in a country where your ethnic group is large or dominant and there are many others affected by a particular disorder, for example sickle-cell disease or thalassaemia, there may already be screening and testing services in place. Your doctor will be able to tell you if this is the case and, if so, where you can use these services.
- You may live in an area of Europe where knowledge of genetics is limited and genetic information is not comprehensive.

- If you are a member of a very small ethnic group, local understanding of your condition may be restricted. In this case, your doctor may be able to direct you to a patient group in either your country or elsewhere in Europe.
- Perceived or actual discrimination can be very distressing, and may add to what is already a difficult situation for you and your family.
- If you are from an ethnic group which is not the majority one on the place that you are living, this can make you feel even more vulnerable. For example, people seeking asylum in a country other than their own may have difficulties in communication due to language differences, or they may not know where to seek out treatment.
- Unfortunately, in some cases, they may not even be eligible to receive treatment, according to the laws of that country. If this is the case, remember that you should still try to find out as much information about your situation as possible. There may be a patient group or support organisation that can help you elsewhere in Europe.
- Some ethnic or religious communities may not approve of certain interventions in genetics, such as prenatal testing, and termination of pregnancy.
- If you are a member of a community like this, and are facing the possibility that you or another family member may have a particular genetic disorder, you will need as much information as possible to make the right choice or you and your family.
- Don't be afraid to discuss your concerns, beliefs, culture and priorities with your doctor or health professional: this is the only way they can provide you with all the information you need in order to make the right decision.

People with learning disabilities

The **World Health Organisation** defines learning disability as having '*significant impairment of intellectual functioning*'.

- This means that a learning disability affects the person's ability to learn, understand or remember new things. They may not be able to cope with social tasks, take care of themselves or communicate with others.
- These impairments are there from childhood, and are not acquired as a result of an accident or illness later in life.
- People with learning disabilities are people first: they have the same rights as any other citizen, though they can often be discriminated against in many societies.
- The extent of learning disability can vary from individual to individual: Down Syndrome is a good example of this.
- Sometime the learning disability is labelled mild, moderate or severe. These are only medical labels: each individual will have different support needs and different abilities.

Learning disabilities caused by genetics

Some disabilities are caused by genetics. This is where either new errors occur in the DNA sequence, or they occur as a result of abnormal genes inherited from parents.

The most common types of genetic conditions that cause learning disabilities are listed below:

[Down syndrome](#)

[Klinefelter's syndrome](#)

[Fragile X syndrome](#)

[Neurofibromatosis](#)

[Congenital hypothyroidism](#)

[Williams syndrome](#)

[Phenylketonuria \(PKU\)](#)

[Prader-Willi syndrome](#)

- If you are involved in caring for somebody with a learning disability you will want to act in the person's best interest, but sometimes it can be difficult to know what the right thing is to do.
- For example, someone with a learning disability may be unable to give informed consent <http://www.eurogenguide.org.uk/public-info-informed-consent.htm> to treatment.
- People in this position require protection by everyone involved in their care. However, many people with learning difficulties may have the capacity to consent if time is spent explaining the issues to them in simple language. Their capacity must never be underestimated.
- When the consent of a legal representative is required, the person with a learning disability (whether child or adult) must be involved in the decision to the fullest extent possible for them.
- The test must be in their interest, for example, to improve treatment and care. You may find that talking to a genetic counsellor may be a great help in these circumstances.
- Most people agree that it is important to support research into the care of people with learning disabilities and into the cause of these disabilities.
- It should not be assumed that someone with learning disabilities would be less altruistic than a competent adult in wanting to help other family members by taking part in genetic investigations.

- At the same time you will need to ensure that people with learning disabilities are not exploited in the course of research or their treatment. There may be a request for them to donate DNA samples which contribute to the development of treatments for their condition.
- However, you may also have worries about data privacy, and whether or not the person in question really understands what will happen to the samples that they have donated.
- All these concerns should be discussed with your health professional and the researchers concerned. There should be no pressure on you to proceed until you have all the explanations you need in order to make the right decision.

People with mental health problems

Research studies now show that some mental health problems have a genetic component.

- Some people appear to be genetically more susceptible to the effects of external factors that can cause or worsen mental illness.
- This susceptibility to mental health problems can run in families, but having a parent with mental health problems does not mean that you will have them. This is because the likelihood of developing mental health problems appears to be due to the interaction of many genes with non-genetic factors in a way which 'triggers' the illness.
- This is a complex area of research. As with physical illnesses, we do not know enough about how and why other environmental factors can affect our susceptibility to develop mental health problems.

- Mental health problems, unlike learning disabilities, may be temporary even if the person may have developed the condition because they are genetically susceptible to certain kinds of mental problems.
- For example, individuals affected by schizophrenia, depression, or bi-polar disorder can have periods when they are more seriously affected by their condition than at other times.
- This means that their ability to think rationally and take reasoned decisions can vary. There may be times when they are quite capable of doing so, whilst at other times they require care and need others to be responsible for them.
- Just because somebody has mental health problems and is unable to make rational and healthy choices for themselves at one point in their life, does not mean that they will be unable to do so at all times. The day-to-day mental health of an individual who is affected by this type of condition can be affected by a wide range of factors and events in their lives.
- If you or a relative are considering taking a genetic test or becoming involved in research and have experienced mental health problems, it is important to tell your doctor or the health professionals so that they can advise you appropriately.
- The fact that a research participant has suffered from mental health problems at some point in the past is an important factor for researchers to take into account.
- For example, an individual might agree to take part in research whilst going through a 'better' period but then be asked for permission for their data samples to be used again at a time when the illness is worse and their ability to take decisions is compromised. All this needs to be discussed and a way forward agreed in advance.