

Guidelines for health professionals about family issues in genetics in Europe

SUMMARY OF GENDER AND FAMILY ISSUES:

- ***Knowledge of genetics may be limited amongst your patients and the area in which you practice. Therefore patients may not understand genetic diseases, how they are transmitted, what the risks are or what help that is available to them***
- ***Men and women sometimes have different attitudes to seeking medical treatment. Studies show that men are often more reluctant to do so, and may require encouragement and support***
- ***Women are often responsible for much of a family's health. Where this is the case they may need help, as this is a significant responsibility and can be a stressful experience***
- ***Not all diseases affect both genders, and the impact that they have can differ between men and women. Please contact your national genetics society to find out more about different conditions, by following this link: http://www.eurogenguide.org.uk/european_societies.htm***

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Information for Men

Men often delay seeking medical advice until later than women. This can mean delays in the diagnosis and treatment of diseases

This means that sometimes diseases such as cancer in men can be more advanced by the time a test is done and a diagnosis is made

Male patients may be reluctant to ask for a test for a disease that they are concerned about

Male patients may need reassurance that a test may be able to *eliminate* the possibility of disease as well as confirm it

Likewise, it is important patients know that the longer a medical condition is left undiagnosed, the more difficult it may be to treat or cure

- The impact on a family is likely to be worse the later it is that a condition is diagnosed
- Some diseases, for example prostate cancer, kill large numbers of men every year. You should encourage men to be aware of any unusual signs or symptoms, particularly if there is a history of it in their family. There may a diagnostic test available that you can inform them of, depending on the suspected condition

Information for Women

Women may occupy a number of roles within their families

Gender is an issue which can have a significant impact upon a family's healthcare decisions

Women in families are often responsible for the health of their families, and in particular for reproductive decisions

- In many places, for socio-historical reasons, reproductive decisions are still discriminatory towards woman. The familial role of women is often assumed on the basis of tradition. This assumption may not correspond with the changing family structures of today

- Given that many reproductive decisions still falls to women, if a particular disease exists in the family of a female patient, you should direct them towards genetic counseling services if they are pregnant or considering having a child. Genetic counsellors will be able to explain any risks and give advice about options for genetic testing
- Genetic counselling services may or may not be available in the country in which you practice. If you have patients who could benefit from genetic counseling, please visit the European Society of Human Genetics website for further details:
<https://www.eshg.org/>

Men, Women and Disease

There are differences in the prevalence and severity of a broad range of diseases, disorders and conditions between the sexes.

Many general differences in the basic biology of males and females can affect health. Apart from purely anatomical differences (e.g. prostate cancer occurs only in men), better knowledge of sex differences in diseases is needed for better diagnosis, prevention and treatment

- Some conditions affect one sex but not the other. For example, only men are affected by some forms of muscular dystrophy. On the other hand, only a woman will be faced with making decisions about her own body when faced with reproductive choices
- Relevant information for men may differ from information for women, and male and female patients may have different concerns when considering their treatment options

Inheritance

All individuals share genetic information, i.e. DNA, with their parents, children and siblings

This is how family members are genetically related to each other

Abnormalities in parts of the DNA - known as 'mutations' can sometimes cause diseases

Each person inherits half of the genetic information from each of their parents. Therefore a mutation that causes a disease in a parent, can sometimes be passed by the parent to a child

- Whether or a mutation is passed on depends on many factors, for instance which half of each parent's DNA the child receives, the exact nature of the particular mutation, and which genes have been affected
- Some people's genetic make-up can make them susceptible to developing certain condition through exposure to environmental factors. For example, some people's genetic make-up puts them at a higher risk of developing lung cancer from exposure to cigarette smoke than others
- Just because a patient has or may develop a genetic condition does *not necessarily* mean that their children or other family members will develop it too. Genetic testing can give patients more detailed information about the likelihood of their passing on a disease to their children
- Being able to talk to a genetic counsellor is often a good way for patients to understand how inheritance works and how diseases may or may not affect any children that they have. You can find out about the genetic counselling services in your country by visiting the website for the European Society of Human Genetics: <https://www.eshg.org/>
- To become a parent is a significant responsibility, and the situation is still more weighty where there is the possibility of having a child with a serious disease or disability. The implications of bearing a child with an impairment of this kind are serious for both the parents and the children, but particularly for the mother, as the responsibility for reproductive decision still falls primarily to women

Regional differences throughout Europe

There are differences in prosperity across Europe at every level, from the local to the international. This can have an impact on the quality of, and distribution of access to, genetic services

- Genetics is not recognised as a medical specialty in its own right in every country within Europe, and this may be the case for the country in which you are working
- This means that genetic services and treatment cannot be provided equally in all places. However, they might be provided in a region or a country close to and accessible by your patients

Upholding patients' rights

Your patients have a right to seek the best treatment available to them, whatever their circumstances

- You can help your patients by finding out about genetic testing for a condition that they might have, or which is prevalent in their family. You can find out what is available and where by contacting your national genetics society, or via the EuroGenTest website: <http://www.eurogentest.org>
- You should explain to your patient what is involved in a genetic test, and what the implications of the possible outcomes might be. Having understood this information your patient may or may not decide to take the test. It is your patient's right to decide their own treatment, and without the relevant information they will be unable to do this
- If a patient has a test and discovers that they have, or are a carrier of, a genetic condition, other family members may also be affected. Patients may be happy for you to contact other family members to discuss their taking a test as well, but you should obtain your patient's permission before you do so. It is their right to give or refuse permission as they wish
- Some people may be aware that one of their relatives has a condition but do not seek out a test, possibly because they are anxious about what the outcome might mean. Therefore it can be beneficial for them if your affected patient allows you to contact them. Once again however it is necessary to obtain your patient's permission before you do so

Improving services and information about genetics

- Genetic research and genetic counselling services have the potential to bring about widespread benefits to health. By providing information, testing, and treatment, the impact of the effects of genetic diseases on families and individuals can be reduced
- By being willing to take a genetic test, or prepared to participate in research, patients contribute to the longer term achievement of helping an increasing number of people affected by genetic disorders in the future

- Genetic testing and counselling is done by specialists in genetics. However, it is possible that tests for genetic susceptibilities to common conditions - for example heart disease or cancer - could become incorporated into general practice to aid in disease prevention. Contact your national genetics society if you would like further information about this

Taking advantage of new technologies

Europe is becoming increasingly closely integrated in many different respects, both culturally and technology. Increasing technological integration means it is becoming easier to establish contact with other people across the continent with a clinical interest in a particular disease or condition

- This is particularly advantageous for people with the rarest disorders, where only a very few people over a wide geographical area will be affected. Very rare conditions are known as 'Orphan' conditions, and an example one of these is Gaucher's disease (<http://www.gaucher.org.uk/>), where only one person in between 50 000 and 100 000 will be affected.
- If you have a patient who has a disease that is very rare, they may not know anybody else who has the disease, and you may have no other patients also affected. This might make it difficult for your patients to find the help and support that they need easily within their local region. There are resources online which patients can use to find others with the same condition in Europe.
- In the UK there is the national alliance for rare disease – Rare Disease UK. You can contact them via their website, here: <http://www.raredisease.org.uk/>. For help outside the UK, the worldwide portal for information about rare diseases is the website for Orphanet: <http://www.orpha.net/consor/cgi-bin/index.php>