



Informed Consent for Diagnostic Genetic Testing

Fact form for patients

"Genetic testing consists of medical examinations aimed at detecting or ruling out the presence of hereditary illnesses or predisposition to such illnesses in a person by directly or indirectly analyzing their genetic heritage (chromosomes, genes)."

Counsel of Europe: Convention on Human Rights and Biomedicine, Oviedo, 04 VI 1997.

The human genetic heritage is present in the nucleus of every cell of the body. This material consists of 22 pairs of chromosomes plus one pair of sex chromosomes: XX in women, XY in men. Sperm and egg cells, however, carry but one element of each pair allowing the pairs to form again by fertilization. Genes are the units of heredity and consist of DNA, they are lined up to form the chromosomes and their number is estimated at around 30'000. A few genes are located in the cell but outside of the cell nucleus in so called mitochondria present in a varying number of copies. Genes form the blueprint of our physical existence by coding the protein structure of tissues and enzymes.

There are basically two levels of genetic changes:

Chromosome changes

Changes in the number of chromosomes or their structure are detectable under a microscope and usually involving hundreds or thousands of genes. Chromosomes are studied on a "karyotype", where chromosomes are arranged pair-wise from an image obtained by a photographic or digital camera connected to the microscope.

Molecular changes

Small changes at the level of single or few genes called " gene or DNA mutations ". Numerous molecular methods allow the study of changes in the DNA structure of a single gene. The choice of test depends on the specific disease. They are not always informative and sometimes no result can be obtained. Molecular genetic testing is always related to a specific question, no screening (non directed search for changes) of the genome can be offered.

Implications

Genetic tests may provide information of a highly private nature and require your consent. They allow determining whether a person is a carrier of a mutation, which may be inherited, and cause disease in his or her offspring. Genetic tests may show that a person is affected by a specific genetic disease or at increased risk of being affected by it someday in the future. They may also reveal an increased risk for other family members or for the child of an ongoing pregnancy. In order to be aware of the aims, consequences and limitations of genetic testing adequate genetic counseling by a trained professional is highly recommended in genetic clinics worldwide.



Genetic counseling

Genetic testing like any other diagnostic procedure is voluntary and you have to formally consent. You should take the time to ask all the questions you might have as well as to make an independent personal decision. It may be appropriate to reconsider your decision and ask for a second appointment.

Below you will find some aspects that **must be discussed** in language easily understandable to you **before** you decide for or against genetic testing.

1. The major medical facts including the diagnosis, the prognosis and ways of treatment of the disorder tested.
2. The genetic facts involved including risks for other family members.
3. The chance that the test will give a correct prediction as well as indeterminate or unexpected findings.
4. The risk of receiving an unfavorable test result and the possible consequences for yourself and your family. In case of a prenatal diagnosis this may include the risk of facing a decision about abortion.
5. Alternative options including your right to refuse the test.
6. Potential benefits and disadvantages, including unsettled questions of privacy protection dealing with insurances, banks or employers.
7. Your care will not be jeopardized whatever decisions you and your family make.
8. Possible use of the your tissue sample after testing: destroyed - kept for reanalysis upon request - DNA banking.

Procedure

Genetic tests require a small amount of blood drawn from a vein (about 3 teaspoons) Genetic material can also be extracted from a biopsy (skin, muscle etc.). These procedures are considered harmless. In case of a prenatal diagnosis a small amount of fetal cells obtained by amniocentesis, chorionic villus sampling or fetal blood sampling are required. These procedures carry a small risk (about 1 %) for a loss of the pregnancy.

Legal frame of genetic testing

Counsel of Europe: Convention on Human Rights and Biomedicine, Oviedo, 04 VI 1997.

Art 12: "Tests which are predictive of genetic diseases or which serve to identify the subject as a carrier of a gene responsible for a disease or to detect a genetic predisposition or susceptibility to a disease may be performed only for health purposes or for scientific research linked to health purposes, and subject to appropriate genetic counseling. "



Informed consent for genetic testing

Full name: _____

Date of birth (D-M-Y): _____

Code number:

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 Sample number:

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I hereby agree to have the indicated genetic test done:

- Karyotype
- DNA testing for (name of disease): _____

I confirm that the aspects listed in the annexed form of the Macedonian Human DNA Bank have been discussed and that enough time for reflexion was granted. I have been given the possibility of independent informed choice.

My decisions about the storage of samples:

I agree that my samples be stored for possible reanalysis or additional analysis in my interest and on my request only.

I agree that my sample be anonymized and used for scientific research.

Signature: _____ (parent/legal representative when applicable)

Date: _____

Referring physician: I have explained the requested procedure to this person, addressed the limitations outlined above and answered this person's questions.

Signature: _____

Date: _____



Антрополошки податоци / Anthropology Data

(пополнува ИИБХГ) / filled by ИИВБГ)

Бр./No.: _____ Дата/Date: _____ Код/Code: _____ Група/Group: _____ Подгрупа/Subgroup: _____

Примерок/Sample: крв/blood брис од уста/buccal swab друго/others: _____

(пополнува испитаникот/ filled by the subject)

Испитаник/Subject

Име и презиме/Name and Surname: _____

Место на раѓање (со општина)/Birth Place (Municipality): _____

Дата на раѓање/Birth Date: _____ Живее (општина)/Living (Municipality): _____

Националност (етнос)/Nationality (ethnicity): _____ Религија/Religion: _____

Зборува на/Spoken Languages: _____

Адреса на живеење/Living Address: _____

Телефон/Telephone: _____

Пол/Gender: _____

Татко/Father

Име и презиме/Name and Surname: _____

Место на раѓање (со општина)/Birth Place (Municipality): _____

Дата на раѓање/Birth Date: _____ Живее (општина)/Living (Municipality): _____

Националност (етнос)/Nationality (ethnicity): _____ Религија/Religion: _____

Зборува на/Spoken Languages: _____

Дедо од татко/Father's father

Име и презиме/Name and Surname: _____

Место на раѓање (со општина)/Birth Place (Municipality): _____

Дата на раѓање/Birth Date: _____ Живее (општина)/Living (Municipality): _____

Националност (етнос)/Nationality (ethnicity): _____ Религија/Religion: _____

Зборува на/Spoken Languages: _____

Баба од татко/Father's mother

Име и презиме (моминско)/Name and Surname (Maiden): _____

Место на раѓање (со општина)/Birth Place (Municipality): _____

Дата на раѓање/Birth Date: _____ Живее (општина)/Living (Municipality): _____

Националност (етнос)/Nationality (ethnicity): _____ Религија/Religion: _____

Зборува на/Spoken Languages: _____

Мајка/Mother

Име и презиме (моминско)/Name and Surname (Maiden): _____

Место на раѓање (со општина)/Birth Place (Municipality): _____

Дата на раѓање/Birth Date: _____ Живее (општина)/Living (Municipality): _____

Националност (етнос)/Nationality (ethnicity): _____ Религија/Religion: _____

Зборува на/Spoken Languages: _____

Дедо од мајка/Mother's father

Име и презиме/Name and Surname: _____

Место на раѓање (со општина)/Birth Place (Municipality): _____

Дата на раѓање/Birth Date: _____ Живее (општина)/Living (Municipality): _____

Националност (етнос)/Nationality (ethnicity): _____ Религија/Religion: _____

Зборува на/Spoken Languages: _____

Баба од мајка/Mother's mother

Име и презиме (моминско)/Name and Surname (Maiden): _____

Место на раѓање (со општина)/Birth Place (Municipality): _____

Дата на раѓање/Birth Date: _____ Живее (општина)/Living (Municipality): _____

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Зборува на/Spoken Languages: _____