SCHWEIZERISCHE GESELLSCHAFT FÜR MEDIZINISCHE GENETIK
SOCIÉTÉ SUISSE DE GÉNÉTIQUE MÉDICALE
SOCIETÀ SVIZZERA DI GENETICA MEDICA

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INFORMED CHOICE IN DIAGNOSTIC GENETIC TESTING

Background

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 analysing their genetic heritage (chromosomes, genes).

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 only 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 nucleus in the mitochondria, present in a varying number of copies. Genes form the blueprint of our physical existence **(genome),** for example by coding the protein structure of tissues and enzymes.

There are basically two levels of genetic changes :

- **Chromosomal changes**

  Changes in the number or structure of the chromosomes are detected on a "karyotype" (image/photograph of the chromosomes arranged in a standard order). Some changes can be too small to be detected by this method.

- **Molecular changes**

  Small changes at the level of single or few genes called "gene or DNA mutations" are studied by molecular methods. 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 (disease), 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 to determine whether a person is a carrier of a mutation which may be inherited and associated with 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 counselling by a trained professional is highly recommended in genetic clinics worldwide.

Genetic counselling

Genetic testing, like any other diagnostic procedure, is voluntary and requires formal consent.

You are invited to 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 should be discussed in language easily understandable to you before you decide for or against a genetic test.

- The **major medical facts** including the diagnosis, the prognosis and ways of prevention and treatment of the disorder tested.
- The **genetic facts** involved including risks for other family members.
- The probability that the test will give a correct result or prediction, or indeterminate or unexpected findings.
- The risk of receiving an **unfavourable 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 termination of pregnancy. The decision about abortion is totally independent of the decision about the test.
- **Alternatives** to genetic testing.
- Your right to **refuse** the test.
- Potential benefits and disadvantages, including **unsettled questions** of privacy protection dealing with insurances, banks or employers.
- Your care will **not be jeopardized** whatever decision you and your family make.
- Possible use of your tissue sample after testing : kept for reanalysis upon request - DNA banking (storage) – use for medical research – destroyed.
- Information about the **costs** and whether covered by health insurance or not.

Procedure

Genetic tests require a small amount of blood drawn from a vein (about 3 ml). There is no need to be on an empty stomach. Genetic material can also be extracted from another source (skin, muscle, amniotic fluid, etc.).

Legal framework

Federal law on human genetic analyses (LAGH – GUMG).

Informed consent before genetic testing

Family name : _____ Personal name : _____

Date of birth : _____

"I confirm that I have received genetic counselling according to the federal law on human genetic analyses and that enough time for questions and reflection has been provided".

I hereby agree to have the indicated genetic test(s) done:

☐ Karyotype (chromosome analysis): ☐ prenatal ☐ postnatal

☐ Molecular testing for (name of disease) : _____

On a sample of the following tissue: _____

My decision for the sample after the test is completed :

☐ If possible, my sample(s) should be stored for future analysis in my interest, only on my request.

☐ My sample(s) can also be used for medical research.

☐ with my name

☐ without my name (anonymous). This means that I can't be informed about eventual results.

☐ My samples must be discarded.

☐ Other: _____

_____

Signature _____ Place and date: _____

(parent/legal representative when applicable)

Referring physician :

"I have given an appropriate explanation of the test to this person, addressed the limitations outlined above and answered this person's questions".

Full name: _____

Signature (required) _____

Place and date: _____

Physician's stamp: