UNIVERSITÄTS-
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ZÜRICH

Informed consent for genetic testing

Surname: _____ First name: _____ Date of birth: _____

I confirm that in the context of a genetic counselling session I've been informed about the different aspects of genetic testing. I have understood the information and had sufficient time for decision making.

I give my consent for the following genetic analysis/es:

_____

☐ postnatal
☐ prenatal
☐ Family screening/segregation analyses
☐ predictive/presymptomatic

For the following disorder: _____

Based on the following biological sample: ☐ EDTA-blood other material: _____

Storage and use of the remaining biological material and data for further analyses

- I agree that the remaining biological material and data will be stored for possible further analyses. My informed consent will be necessary should further analyses be requested. ☐ YES ☐ NO
  In case of a negative answer the remaining biological sample will be destroyed after the analysis!
- I agree that my biological sample and data are used anonymously for quality testing ☐ YES ☐ NO

The use of your sample and data for research purposes.

- I agree, that my biological material and the resulting data may be used for research projects and publications in an encoded (pseudonymised) way ☐ YES ☐ NO

Incidental findings: Should the analysis/es reveal results not directly related to the testing requested (so called "incidental findings"), I wish to be informed as follow:

- Carrier of a disorder for which preventive and/or therapeutic measures are available ☐ YES ☐ NO
- Carrier of a severe heritable disorder which could concern the following generation or other family members ☐ YES ☐ NO

The following question is to be answered by persons of legal age capable of judgement only:

- Carrier of a disorder for which no preventive / therapeutic measures are yet available ☐ YES ☐ NO

Should these questions remain unanswered it will be assumed that the patient does NOT want to be informed about incidental findings. Minors and persons incapable of judgement may only be informed about incidental findings relevant for their health and severe hereditary disorder in the family.

Signature: _____ Place and date: _____
(Patient or parent/legal guardian)

Medical counsellor:

I declare that I've informed the above mentioned person/s, according to the law on genetic testing on humans (GUMG), about the planned genetic tests and their limits as well as providing answers to the patient's questions.

Surname: _____ Name: _____

Signature: _____ Place and date: _____

Stamp:

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Informed consent for genetic testing

The purpose of genetic testing is the detection or exclusion of genetic changes which may underlie a given genetic disorder or represent a risk factor for a disorder. This information sheet lists a few aspects which should be considered prior to a genetic test. Furthermore, information is provided regarding different methods used for the genetic investigations and their application.

Before undertaking a genetic investigation

Genetic testing is performed on a voluntary basis and necessitates your formal consent since the analyses may provide sensitive data. It is recommended to consult a specialist in order to discuss the options, pros and cons, consequences and limits of the specific genetic test. You should make sure that you have sufficient time for decision making and that you clarify all questions you may have. For your assistance we have listed several topics which should be discussed in the context of a genetic counselling session and which should provide you with the necessary information to make a personal educated decision whether or not you are willing to perform the genetic test:

- Clinical utility of the genetic test for diagnosis, prognosis, prevention and therapy regarding the disorder to be tested.
- The results of genetic testing may have implications for further family members.
- Meaning of a genetic test result, its clinical validity as well as the possibility of false negative, false positive or inconclusive results as well as incidental findings.
- Probability of an unfavorable result with all the consequences and decision making that may arise (including a possible disclosure of the results to health insurance companies).
- Alternatives to a genetic test.
- Your right to refuse the genetic tests, or to revoke your decision for the analysis without giving any reason.
- Your decision concerning the use of the biological sample after testing: e.g. storage for possible future analyses, archiving, use of the sample for medical research purposes, or disposal of the sample.
- Information regarding costs of the analyses, and whether or not the costs would be covered by the health insurance.
- The possibility of incidental findings and your decision to be informed or not
- Genetic testing may question the declared relationships

A small amount of venous blood is usually sufficient for genetic analyses. Fasting prior to sampling is not necessary.
In Switzerland the analyses are performed according to the law on genetic testing on humans (GUMG, SR 810.12)

Possible application fields for genetic testing

Diagnostic genetic testing aims at establishing or confirming a genetic diagnosis for an affected patient. A clear diagnosis is often possible, with the benefit of enabling appropriate medical care.
Presymptomatic and predictive testing aims at determining whether a healthy individual is at risk or is carrier of a specific genetic disorder.
Prenatal analyses aim at the detection, exclusion or confirmation of genetic defects in a fetus.
Family screenings aim at determining whether family members (with or without symptoms) may be carriers of the genetic defects detected in a close relative and whether they may be at risk of passing the genetic defect to the next generation.
Segregation analyses of variants of unknown significance within family may help to better estimate their relevance.

Background and methods

The human genetic material (which consists of DNA) is located mostly in the nucleus of every cell in the body and in a small part outside the nucleus in so called mitochondria. The nuclear genetic material is subdivided in 23 pairs of chromosomes, one pair of which are the sex chromosomes (XX in women, XY in men). Approximately 20'000 genes are located on the chromosomes. Few genes are situated on the mitochondrial DNA. All genes together build the genetic blueprint for the structure and metabolism of a body. Changes in the genetic blueprint can lead to disorders: n.b. genetic changes involving small as well as large parts of the genome may lead to severe disorders. Some changes will clearly lead to a disorder whilst other changes may only represent a risk factor for a disorder.
Molecular genetic analyses test for changes at the DNA-sequence level of single genes (gene mutations). Gene mutations may be detected with a range of different molecular methods. The genetic test may be restricted to the analysis of a single gene, to several genes or can be extended to the entire genome (high throughput sequencing). The interpretation of results obtained from the analysis of the entire genome can be quite complex. The analysis of a high number of genes increases the risk of detecting genetic changes not directly associated with a patient's disorder (i.e. incidental findings) or in genes with as yet unclear association with a disorder.
Even though high throughput sequencing allows for the analysis of many genes, these analyses can remain inconclusive, and may not always help to rule out the suspected disorder.