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Clinical
Genomics Lab

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INFORMATION FOR PATIENTS

Informed choice in diagnostic 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. A consultation with a specialistis recommended 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 points 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, informed 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 unfavourable 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.
- 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.

Procedure

A small amount of venous blood is usually sufficient for genetic analyses. Fasting prior to sampling is not necessary. At times the analyses are performed using other tissues (for example skin and muscle biopsies, or amniotic fluid for prenatal analyses).

Legal framework

In Switzerland the analyses are performed according to the law on genetic testing on humans (GUMG, SR 810.12)

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Possible application fields for genetic testing

Genetic investigations provide important information in many medical fields. The applications can be roughly subdivided into the following groups:

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. Prenatal analyses include non-invasive analyses (e.g. non-invasive prenatal test: NIPT performed on maternal peripheral blood) and invasive procedures (e.g. amniotic fluid sampling).

Screenings in families 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.

Background and methods

The human genetic material (which consists of DNA) is located mostly in the nucleus of every cell in the body and with a small amount 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. A small number of genes are situated on the mitochondrial DNA. All genes together build the genetic blueprint for the structure and metabolism of the 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. Genetic changes are usually analysed at two levels:

At the level of chromosomes: Changes in the number or structure of the chromosomes (chromosomal abnormalities). Large chromosome abnormalities are usually detected by microscopic analysis of the chromosomes. Changes involving small parts of a chromosome are detectable with high resolution molecular chromosome analyses.

At the gene level: 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 is extremely complex. The simultaneous analysis of a number of genes increases the risk of detecting genetic changes in genes not directly associated with a patient's disorder (i.e. incidental findings) or genes with as yet unclear association with a disorder (VUS). Particular conditions apply for the reporting of incidental findings in minors. Occasionally, unexpected results concerning the descendance are found. In general, such results are not reported.

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Patient

INFORMED CONSENT BEFORE GENETIC TESTING

I confirm that I have received genetic counselling and that I have had a sufficient amount of time for questions and reflection. I have been informed by my physician that the advice I received conforms to federal law on human genetic testing (GUMG).

With this signature I give my permission for the following investigations:

..... ☐ prenatal ☐ postnatal ☐ predictive / presymptomatic

For the following **disorder**: .....

Copies of the results should be sent to the following persons (include address):

.....

.....

I wish the testing to be carried out…

- even if it is not under obligatory cover and in the absence of a confirmation of coverage ☐ YES ☐ NO
  (If necessary I will cover the costs up to …… CHF myself)

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 follows:

- disorder for which preventive and/or therapeutic measures are available ☐ YES ☐ NO
- 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.

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

- I agree that the remaining biological material and data will be stored for possible further analyses. ☐ 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
- I agree that my genetic data and / or my clinical data are used anonymously in scientific publications / databases ☐ YES ☐ NO
- In principle, I agree that my biological sample and data could be used for research purposes ☐ YES ☐ NO
  (should this be the case you would be contacted at a later stage)

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

Referring physician:
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 :

Attention: Please also send an order form (see homepage CGL)