SPT THE SWISS GROUP OF PHARMACOGENOMICS AND PERSONALISED THERAPY

INFORMED CONSENT FOR DIAGNOSTIC PHARMACOGENETIC TESTING

Pharmacogenetic testing involves the examination of genetic changes, which influence the effectiveness or the risk of adverse effects from drug therapies. This information sheet lists some aspects, which should be considered prior to a pharmacogenetic test, as well as some background information about these tests.

A pharmacogenetic test, like any diagnostic procedure, is voluntary and requires your consent. Because it is a genetic test, it provides very personal information. Therefore, please make sure that you have sufficient time to make an independent decision about the test and to ask all the questions you may have. Your medical care will **not be jeopardized** whatever decision you make.

Below are some points that should be discussed with you in an easily understandable language before you decide for or against the pharmacogenetic test. In particular, **it should be clear to you when you make your decision, how your drug therapy may be changed based on the result of the pharmacogenetic test**.

- Basic information regarding the test itself, including a description of the **tested genes and the specific variants of these genes that will be examined.** This should also include a description of **what test results are possible**, and how commonly a certain test result may occur.
- **Why the test is important for your drug therapy**, including the **consequences** that each test result would have for your drug therapy (for example: how your therapy would be changed based on a given test result).
- Whether or not the test result will also be **relevant for other drug therapies**, which you might receive in the future.
- How accurately the test can predict or explain the outcome of your drug therapy, including the **limitations** of what the pharmacogenetic test can predict or explain, and what it cannot.
- **Alternatives** to the pharmacogenetic test.
- Your right to **refuse** the test and your right to **refuse knowledge** of information about your genetic makeup.
- Information about the **costs** of the test and whether these costs are covered by your health insurance.
- Your decision concerning the **use of your blood sample and DNA** after the test is completed:
    - **Storage** so it can be used for potential future analyses (only upon request and with your consent). This means that for any additional pharmacogenetic tests that may be performed in the future, no new blood sample would need to be collected.
    - Use for **quality testing**. This means that your sample may be used by the laboratory to ensure that quality standards for performing pharmacogenetic tests are met. Your sample will only be used anonymously (without including any information that would identify you such as your name or birth date).
    - Use for **medical research**. This means that researchers are allowed to contact you, if in the future your sample is of interest to them for a research study. If you are contacted by researchers, they will first explain the research study to you. Afterwards, you are still free to decide whether or not your sample should be used for this study or not.
    - **Disposal** of the sample. This means the sample will be destroyed once the test is completed.

BACKGROUND

The human genetic material consists of DNA and is present in 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. Genes are the units of inheritance and are located on the chromosomes. In humans, it is estimated that there are around 20'000 different genes. The total of all genes (genome) form the blueprint for all the structures in our body and its functions. This includes instructions for making proteins that are important for your body so it can react to a drug that you take. Examples of such proteins are proteins that help your body to take up a drug, bring it to different organs or into cells, convert it into other molecules, or eliminate it from your body.

With a pharmacogenetic test, small changes in your genetic material (in the DNA) are studied, which are called "gene variants". The gene variants analyzed with a pharmacogenetic test only concern variants that are known to have an impact on a drug therapy (for example: whether a therapy is effective, or its risk of adverse effects). This means, for example, that a gene variant may influence how fast a medication is taken up, converted, or eliminated from your body, or if there is an increased risk for serious adverse effects. These gene variants are part of the natural genetic variability that make each person a unique individual. They generally have no major effect on your overall health.

Pharmacogenetic tests are carried out using molecular methods. This means that small parts of your DNA are analyzed, which contain the specific gene variants that are important for your drug therapy. The test then detects whether a particular gene variant is present or not. In a pharmacogenetic test, no broad genetic screening (a non-directed search for other changes in the genome) is performed.

IMPLICATIONS

Pharmacogenetic tests allow us to determine whether you carry specific gene variants that may impact how your body reacts to a specific drug therapy. This means that based on the result of this test, your drug therapy can be adjusted so it best fits the specific gene variants that you carry. Such adjustments may include the use of a different drug dose, or the use of an alternative medication that is better suited for you.

Because these genetic variants can be inherited, it is possible that your relatives (for example children, siblings, parents) also carry the same gene variants. This means that in the future, a pharmacogenetic test may also be helpful for them, if specific drug therapies are prescribed to them.

Besides their effect on certain drug therapies, the gene variants analyzed with a pharmacogenetic test generally have no serious negative consequences for your health. In case there are possible consequences of the pharmacogenetic test result other than its effect on a drug therapy, you will be informed about these possible consequences before the test. However, based on current knowledge, this will only happen in very rare cases.

PROCEDURE

A pharmacogenetic test requires a small amount of blood drawn from a vein (about 3 ml are usually sufficient). There is no need for fasting prior to this test.

LEGAL FRAMEWORK

Federal Act on Human Genetic Testing (LAGH – GUMG, SR810.12).

This patient information and consent form is based on the general consent regarding genetic testing developed by the Swiss Society of Medical Genetics and was adapted for the requirements relevant for pharmacogenetic testing.

INFORMED CONSENT FOR PHARMACOGENETIC TESTING

Name: _____ Given name: _____

Birth date: _____

I hereby confirm that I have been informed about the different aspects of pharmacogenetic testing as explained in the information sheet "Information for patients". I have understood the information and had sufficient time for questions and to make my decision.

I give my consent for the following pharmacogenetic test(s):

Molecular testing for (name of drug(s): _____

Genes or gene variants variants tested: _____

Sample material: EDTA blood

Decision regarding the use of my sample after this test is completed:

Storage and use of the remaining biological material and data for further analyses
If possible, please store my sample for future analyses, which will only be performed upon my request and if I provide my consent. ☐ 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

Use of your sample and data for research purposes
In principle, I agree that my biological sample and data could be used for research purposes. ☐ YES ☐ NO
A positive answer is not yet consent for the participation in any actual research study. You may, however, be contacted at a later stage with details concerning a research study.

Signature: _____ Place and date: _____

(parent/legal representative if applicable)

Referring physician:

I confirm that I have provided appropriate consultation to the above-named person regarding the planned test(s) and answered this person's questions.

Full name: _____ Stamp:

Signature: _____ Place and date: _____
(required)