Information Protocol for Prenatal Fetal Genetic Testing using Maternal Blood

Name: First name: Date of birth:

This information completes the information sheet “Prenatal Fetal Testing (Prenatal Diagnostics)” and the individual consultation with your doctor. During the consultation, please ask about anything that is unclear or important to you and tell us if you feel sufficiently informed or would like to know more about the upcoming test and its results.

You should only make a decision on prenatal testing for genetic conditions (e.g. trisomy 21, 18 or 13) after careful consideration and sufficient time to decide. This is a personal decision that you should make without being influenced by external parties. Of course, you may decide not to go ahead with the test, as you have a “right not to know”. This can cover all testing, or only part of the technically possible analyses and subsequent tests. You do not have to know all the information about the genetic material, unless there is an immediate physical danger to the baby (Art. 6 and Art. 18(2) of the Federal Act on Human Genetic Testing (Bundesgesetz ueber genetische Untersuchungen beim Menschen; GUMG).

Method: The placenta releases small amounts of DNA into the mother’s blood. From the tenth week of pregnancy, these small amounts can be used to test the unborn child for anomalies in the genetic material (chromosomes). If the baby has an extra chromosome 21, 18 or 13, this is called trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome) or 13 (Patau syndrome). Trisomies lead to physical and mental development disorders to different degrees. This method can also be used to detect other anomalies that affect only part of a chromosome or a single gene.

Result: A normal blood test does not guarantee a healthy child. Trisomy 21 (the most common genetic anomaly) can be detected with a reliability of up to 99%, while the reliability for detecting trisomy 18 and trisomy 13 is somewhat lower. For other, rare genetic conditions, it is not currently possible to state the reliability of the results. In rare cases, such tests can also reveal a condition affecting the mother. In 1% of cases, the result of the test cannot be established as the amount of the baby’s DNA in the mother’s blood is too low. It takes approximately one week to receive the results.

Abnormal or difficult to interpret findings must be confirmed with a subsequent test, such as chorionic villus sampling or amniocentesis, as in rare cases the result can be abnormal although the child is healthy. Unexpected or unfavourable results can mean that you have difficult decisions to make, and be very difficult psychologically. Additional consultations and support from other specialists may be necessary. If a serious incurable condition is detected, you may consider terminating the pregnancy. However, you always have the right to be advised of alternatives to termination and be informed of groups for parents of disabled children and self-help groups (Art. 15 GUMG).

Cost: Testing for trisomies is covered by health insurance if the risk assessment carried out between the 11th and 14th weeks of pregnancy using the first trimester trisomy risk test has given a result of 1:1,000 or more. If this is not the case, but you have supplementary insurance, a voluntary monetary contribution may be made under certain circumstances. Other investigations with this test for other rare conditions are not covered by statutory health insurance.

Your questions:

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This protocol documents genetic counselling as per Art. 18 of the GUMG and the test contract below applies as consent to genetic testing. For further consultation, the relevant Cantonal counselling centres are also available (pursuant to Art. 17).

Explanatory consultation

Interpreter:

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Proposed test: Prenatal Fetal Genetic Testing using Maternal Blood:

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Doctor's notes on explanatory consultation (waiver of explanation stating reason, individual risk factors etc.).

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Alternative treatment possibilities:

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Date: Time: Duration of explanatory consultation:

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Treatment contract

Dr_____ carried out an explanatory consultation with me. I have understood the explanation and could ask any questions I had. I have been given a copy of the consultation protocol.
I have had enough time to decide and consent to the planned test. I know that I can withdraw my consent at any time (Art. 5(3) GUMG).

Place, date: Patient:

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The text on the front page was discussed with the patient, the questions answered, and a copy of this explanation protocol was given to the patient.

Date, time: Doctor: