FRAUENPRAXIS
WEINFELDEN
im Hausarztzentrum

PRENATAL SCREENING

Screening tests and diagnostic examinations for fetal abnormalities

PRENATAL SCREENING

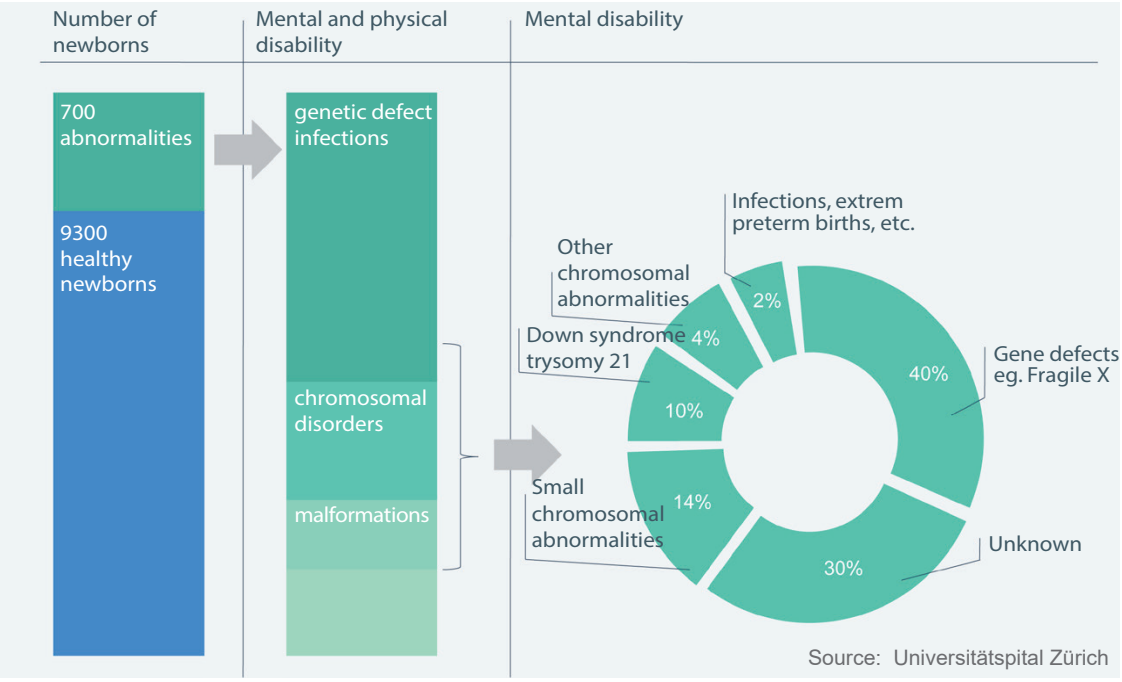
Chromosomal disorders, genetic diseases and malformations

Human cells normally contain 23 pairs of chromosomes, for a total of 46 chromosomes in each cell. A change in the number of chromosomes can cause problems with growth, development, and function of the body's systems. One of the most common chromosomal anomalies is trisomy 21, also called down syndrome, where affected people have three copies of chromosome 21 in each cell (approx. 1 in 600 births). The risk of trisomy increases with the mother's age and is also increased if trisomy 21 has already occurred with earlier pregnancies. Trisomies 13 and 18 occur less frequently and are characterized by the presence of numerous malformations with a limited survival rate for most children.

In the case of genetic diseases, one or more genes of the human genome are altered.

Please let us know if a genetic disorder is diagnosed in your family. Parents can be healthy carriers of a genetic disease where only the child is affected. Sometimes genetic mutations occur, and genetic disorders might not be inherited from the parents.

Malformations are a structural defect in the body due to abnormal embryonic or fetal development. There are many types of malformations (ie heart disorders, cleft lip, cleft palate or spina bifida).



PRENATAL RISK ASSESSMENT FOR TRISOMIES

Prenatal screening and diagnostic tests are designed to detect disorders in a fetus before birth. Some conditions can be treated after birth. However, chromosome abnormalities and some other genetic disorders cannot be reversed, which may have serious consequences for the baby. This test is not compulsory and is only carried out with your consent. According to the risk assessed, more extensive tests may be discussed. No prenatal test can give a full guarantee that the baby will be normal in every way.

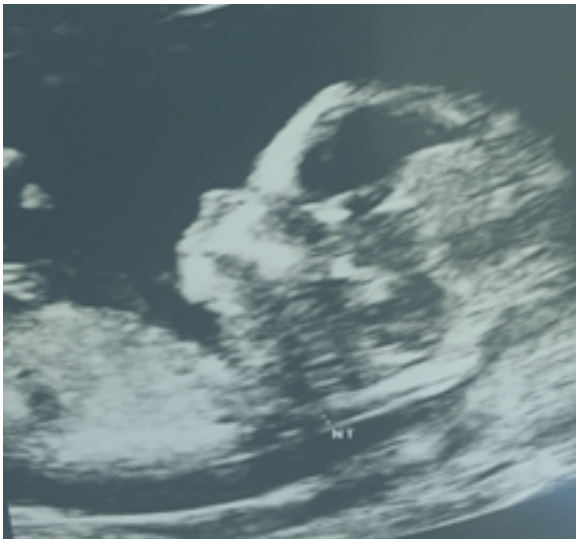
FIRST TRIMESTER TEST (FTT)

The first trimester test consists of an ultrasound, performed at about 12 weeks, and a maternal blood analysis. The maternal blood sample measures a hormone, free beta-HCG, and a protein, PAPP-A. These substances are found in the blood of all pregnant women, but with a different level for a fetus which is a trisomy 21, 18 or 13 carrier.

The blood test, ultrasound scan and age are combined to calculate the risk of the fetus having Down syndrome or other less common chromosome problems. Down syndrome is more likely if the fetus has extra fluid at the back of the neck, if the nasal bone can not be seen and/or the blood test is out of the normal range.

However, a positive result does not mean that the fetus is actually affected by a chromosomal or genetic anomaly. Invasive diagnostics (see below) is necessary for a definitive clarification. The first-trimester test detects about 90% of all trisomy 21 cases, but conversely also misses about 10%. The test is covered by the basic health insurance.

NUCHAL TRANSLUCENCY



The most important element for calculating the risk is the ultrasound measurement of the thickness of the nape of the neck of the embryo which forms a zone called nuchal translucency. The risk increases with the thickness of this. Furthermore, a thick nuchal translucency may be an important index for other fetal abnormalities, but may also be observed in children where the development is totally normal.

NON-INVASIVE PRENATAL TESTING (NIPT)

This screening blood test identifies fetal DNA in the blood stream of the mother, and tests for Down syndrome as well as some other common chromosome problems. If you have a positive test result for Down syndrome, it is more than 99% likely that the fetus has Down syndrome. The costs are covered by the basic health insurance if the FTS shows a risk of 1 in 1000 or higher.

If positive, this test does not replace invasive testing, and the results should be confirmed with either a chorionic villus sampling or amniocentesis. This test may not be possible for all pregnancies, such as triplets or in pregnancies where one twin has died.

The advantage of this method is that only a blood sample from the mother has to be taken which involves no risk of miscarriage. The result is available within about two weeks.

INVASIVE DIAGNOSTIC TESTING
CHORIONIC VILLUS SAMPLING / AMNIOCENTESIS

Chorionic villus sampling is performed from 11 weeks of pregnancy. A sterile needle is guided into the placenta and a small sample is taken for testing.

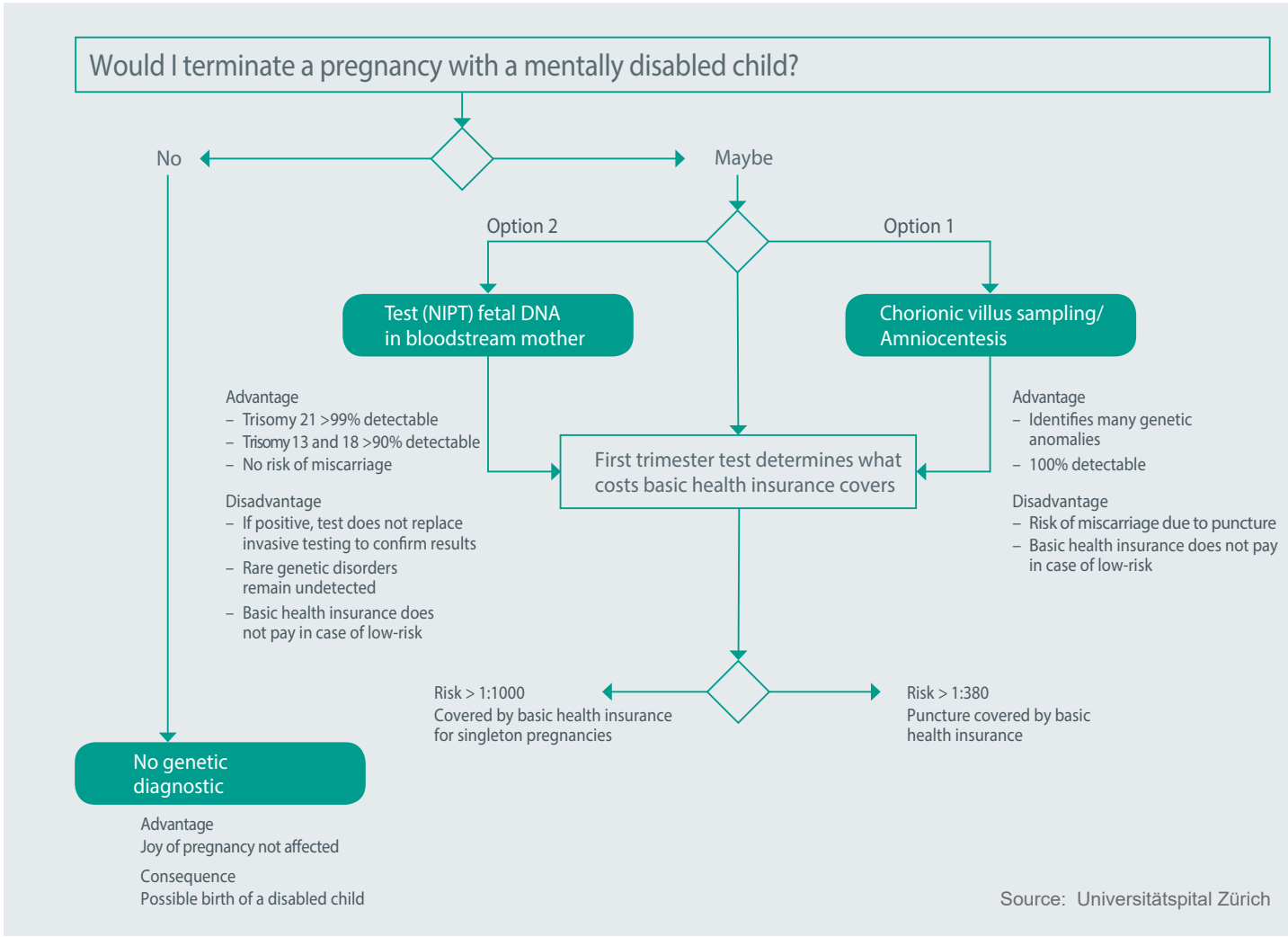
Amniocentesis is performed from 14 weeks of pregnancy. Under ultrasound guidance, a needle is inserted through the abdomen into the amniotic sac around the fetus and fluid is taken for testing.

Chorionic villus sampling and amniocentesis test the fetal chromosomes.Other genetic testing can occur where indicated. These are invasive procedures which may, in rare cases (0.5 to 1%), trigger a miscarriage.

A rapid chromosome screening test takes 1-2 days. This only assesses for a handful of chromosomal disorders including Down syndrome, Edward syndrome, and Patau syndrome, amongst others.

Normal rapid screening results are very reassuring; however it is important to wait for the final chromosome results which may take 10–14 days for confirmation.

BEFORE THE EXAMINATION



CONSENT FORM

I hereby declare and confirm that I received a detailed verbal explanation from Kathrin Holderegger on potential prenatal risks, complicatations and possible diagnostic methods and, that all relevant questions were answered and are well understood.

Please mark with a cross where applicable:

I agree to a routine fetal ultrasound

☐ Yes, I would like a first trimester test

☐ No, I do not want a first trimester test

☐ I am not interested in a routine fetal ultrasound

Date: _____ **Signature:** _____

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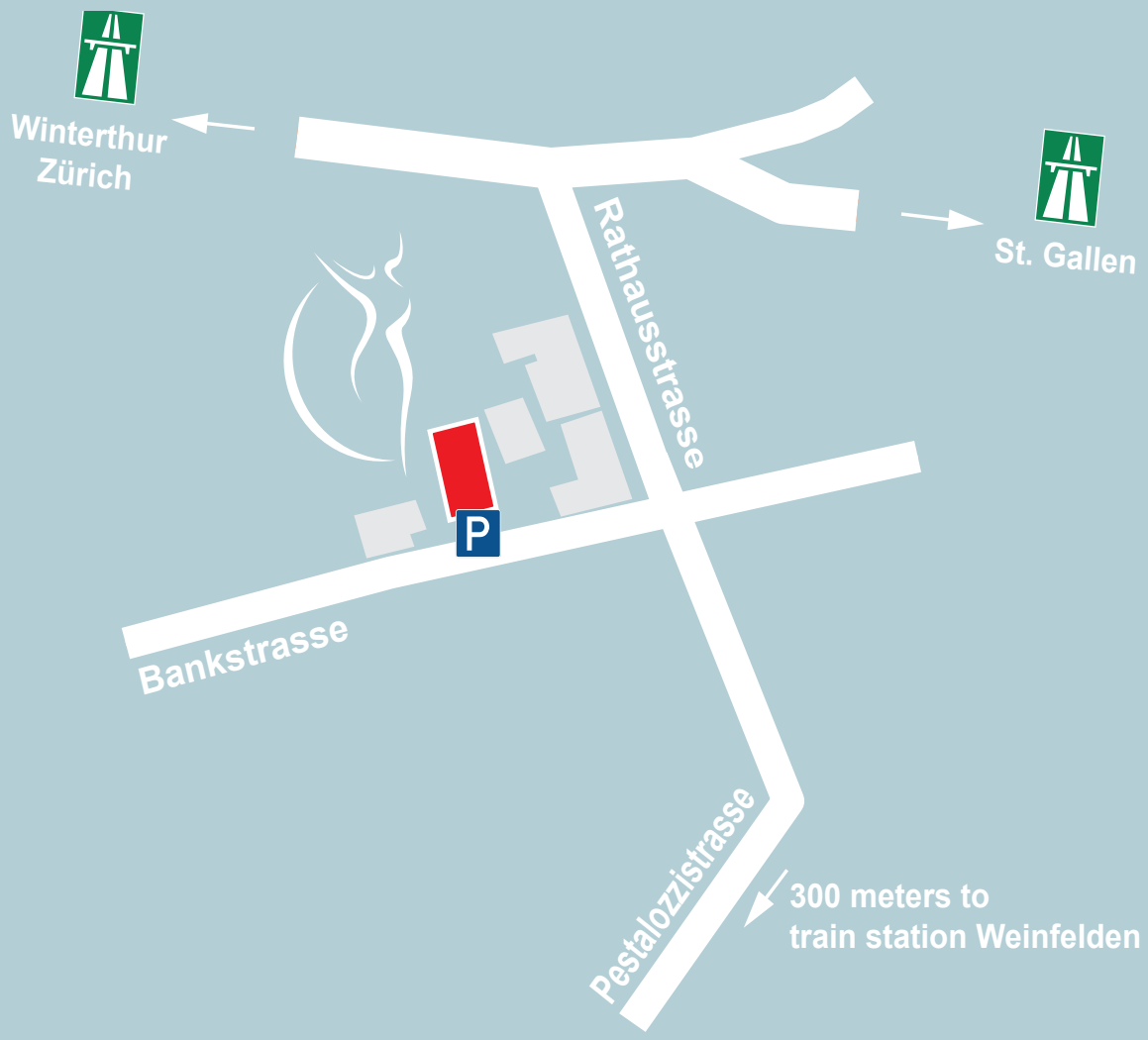

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