How is the PrenaTest® carried out?

- Your physician takes 20 ml blood from you, after you were informed in detail and subjected to genetic counseling by your physician and have signed the declaration of consent for the PrenaTest®. This blood is sent to the LifeCodexx diagnostics laboratory. The analysis generally takes two weeks (i.e. 10 working days, Mon.–Fri.). Your physician then receives a letter with the result and will inform you about the outcome of the analysis. The expenses for the PrenaTest® and the related medical services are not covered by health insurance. Thus, you have to pay these expenses yourself.

What will happen in case of a negative test result?

- A negative test result means that the existence of trisomy 21, 18 and 13 in the unborn child can be excluded with a high degree of certainty. Depending on the medical reason which caused the PrenaTest® to be performed, your responsible physician will nevertheless track the course of your pregnancy with special attention and advise further examinations, if necessary.

What will happen in the event of a positive test result?

- A positive test result means that your child is almost certainly carrying the respective trisomy. Then it is strongly recommended to that you undergo genetic counseling and clarify the result of the PrenaTest® further diagnostically by means of an invasive examination, an amniocentesis in most cases. This further examination is necessary to diagnose a genetic reason for the trisomy.

Please consider the following:

- Most prenatal examinations do not show any abnormalities and this helps to reduce fears and ensures an undisturbed course of pregnancy. However, if one examination shows medical findings, this can often lead in part to great uncertainty and a situation of conflict. Your physician will provide you with comprehensive information and support. In addition, psychosocial counseling is recommended in such situations. Your physician will also inform you about that in detail.
Dear pregnant woman,

You already talked with your gynaecologist about the risk of chromosomal imbalances in your unborn child.

In addition to other methods, the PrenaTest® offers the possibility of testing for the existence of the trisomies 21, 18 or 13 in your child in more detail.

This brochure should provide you with general information on the PrenaTest® and serve as preparation for a further talk with your physician. Whether the PrenaTest® is suitable for you personally or not can only be clarified together with your physician. Do not hesitate to address any questions that may arise about your personal situation or the test to your physician.

What are trisomies and how do they develop?

- The genetic material of all cells is organised in chromosomes. Two of them (the sex chromosomes X and Y) determine the sex of a person, the others (chromosomes 1 to 22) are called ‘autosomes’.

As a first step, the chromosomes double before each cell division before they are evenly distributed to the two daughter cells. Errors can occur in the course of this distribution. If such errors occur during the development of egg and sperm cells and a child is created from such modified cells, then the child will carry the modified genetic information. Many of these imbalances of chromosomes, however, prevent the development of a pregnancy from the respective germ cells or cause early miscarriages.

If the cells of a child carry a given chromosome in triplicate instead of duplicate (as normally) this is called ‘trisomy’. Only very few of such trisomies are compatible with life. The most frequent and best known such imbalance of chromosomes is trisomy 21, in which chromosome 21 is present three times instead of twice in the child. Trisomy 21 is typical for Down’s syndrome.

Trisomy 18, typical for Edwards syndrome, occurs much more rarely. And trisomy 13, typical for Patau syndrome, occurs even less.

The expression of these trisomies in form of a disease in the affected children can be very different. Please take advice and counseling from your doctor regarding your individual risk and what this may mean to you and your unborn child.

Which possibilities exist to examine such imbalances already during pregnancy?

- The possibilities of non-invasive or invasive testing do exist in principle before birth.

Non-invasive methods are ultrasound examinations and blood tests. They do not interfere with the body of the woman (except the blood sampling) and thus are gentler. The nuchal translucency screening, for example, in the course of the first trimester screening (12th to 14th week of pregnancy) is one of the most frequently carried out ultrasound examinations of the unborn child. Using these previous non-invasive methods, your individual risk that your child will carry trisomy 13, 18 or 21 for example can be determined. A definitive diagnosis, however, cannot be provided by these non-invasive methods.

Invasive methods mean an intervention on the body of the pregnant woman. Amniocenteses or puncture of the placenta are such examples. Both methods make it possible to collect cells from the unborn child to carry out a chromosomal analysis. The number, shape and structure of the chromosomes are analysed in this process and thus a definitive diagnosis can be made about the existence or exclusion of trisomy 13, 18 or 21 for example. The risk of losing the child due to this invasive procedure is specified at approximately 0.2–1%.

The PrenaTest®, as a novel non-invasive method, offers the possibility of detecting the most frequent forms of the trisomies 13, 18 and 21, the so-called “standard trisomies” in the unborn child. Blood is taken from a maternal vein for this purpose, a procedure which should be harmless for your unborn child.

When is it sensible to carry out the PrenaTest®?

- Carrying out the PrenaTest® is optional. Doing so makes sense if you belong to a risk group or if one of the mentioned trisomies is suspected in your unborn child based on ultrasound findings or previous blood tests. Please consult your physician about how high your personal risk is that your unborn child carries a trisomy 13, 18 or 21.

Limits of the test

- A healthy child is born in the majority of pregnancies. This is, however, not the case for a minority of pregnancies. Today it is possible to search specifically for many diseases in the course of pregnancy. But under no circumstances is it possible to exclude all conceivable diseases. Nobody can guarantee you a healthy child.

Standard trisomies 21, 18 and 13 can be detected by the PrenaTest® with a high degree of certainty.

In the context of clinical studies to evaluate the PrenaTest®, more than 99% of all blood samples were correctly classified. In only one sample was an existing trisomy 21 not detected (false-negative), in one other sample there was a trisomy 18 indicated although in reality this was not present in the unborn child (false-positive).

No other chromosomal defects are detected by the PrenaTest® so that no statements can be made based on the test results. You should also be aware that certain rare forms of the mentioned trisomies cannot be detected with certainty using the PrenaTest®. This is true for very few cases, in which only parts of the respective chromosome exist in triplicate or if only parts of the child’s body show the respective chromosome in triplicate (called ’mosaic trisomy’). These rare occurrences cannot be detected reliably using the PrenaTest®. In addition the PrenaTest® is currently not suitable for multiple pregnancies. No test result or an unclear test result may occur despite careful implementation in very rare cases. Thus, it may become necessary to repeat the test and also to request a new blood sample if required.