Who should take the NaTALIA prenatal test?

The test is suitable for all pregnant women, who are fully informed about its informative value and possible consequences. For pregnant women with atypical first trimester screening (FTS) results, the NaTALIA prenatal test can provide valuable information about the tested chromosomal changes. At present however, the test does not analyse all chromosomal changes that can lead to atypical FTS results. Therefore, it should be done only after detailed consultation with your doctor. The same considerations apply to pregnant women with an abnormal foetal ultrasound finding.

What are the limitations of the NaTALIA prenatal test?

The test only examines the mentioned changes, which are only a part of the prenatal chromosomal changes (approx. 70%). It does not replace regular check-ups or an obstetric ultrasonography of the unborn baby.

NaTALIA Prenatal tests without result

Among all NIPT procedures, it is possible in few examined cases that an accurate result cannot be obtained. The most common reason for this is a very low level of foetal DNA in the mother's blood. In such cases, the examined abnormalities can neither be confirmed nor excluded. With the NaTALIA prenatal test, the failure rate of approx. 1–2 % is remarkably low. In such cases, a free repeat test should be performed with a new blood sample. In consultation with your doctor, invasive diagnostics may also be considered, depending on other previous findings.

The costs for the NaTALIA prenatal test for trisomies 13, 18 and 21 are covered by health insurance in justified cases. Optional testing for foetal sex (€29.14) and sex chromosome maldistribution (€58.28) is to be paid for by the patient.

For more information and an informative video on the NaTALIA prenatal test, visit www.LADR.de/nipt

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NaTALIA Pränataltest

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Information for patients

NaTALIA Pränataltest

Non-invasive blood test for chromosomal defects in the unborn baby - Information for pregnant women

Why take the NaTALIA prenatal test?

The NaTALIA prenatal test provides an early, highly reliable assessment of the risk of the most common chromosomal disorders in your unborn child.

Which chromosomal disorders are investigated?

The test determines the risk of trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome) and trisomy 13 (Patau syndrome). These changes are caused because the number of copies of the respective chromosomes deviate from the norm and this is associated with physical abnormalities and/or developmental delays.

The occurrence of these trisomies increases with the mother's age. For example, the risk of trisomy 21 in a 20-year-old pregnant woman is about 1:2000, while that in a 40-year-old pregnant woman is already 1:100. Optionally, in singleton pregnancies, the NaTALIA prenatal test also allows testing for sex chromosome maldistribution. These are generally associated with relatively less developmental disruption compared to trisomies 21, 13 and 18.

Which material is used for the test?

The test only requires a blood sample from you, because your blood also contains fragments of the genetic material of your unborn baby. Therefore, this test is called a non-invasive prenatal test (NIPT). Unlike invasive procedures (e.g., amniocentesis), there are no procedure-related risks for you or your unborn baby.

From when can the test be taken?

The test can be taken from the 10th week of pregnancy. The NaTALIA prenatal test can also be used for twin pregnancies and after fertility treatments (e.g., IVF, ICSI).

Is the child's sex also examined?

The NaTALIA prenatal test can also determine the sex of your unborn child. If you choose this option, the result will be sent to you in accordance with the requirements of the Genetic Diagnostics Act after the 14th week of pregnancy.

What is the procedure for this NaTALIA prenatal test?

First, your doctor or a human genetics specialist will explain the possibilities and limitations of this test. A blood sample (10 ml) can be taken after your written consent. The blood sample will be sent to our human genetics laboratory in Germany. The result of the test will be available after approx. 7 working days and will then be sent to your doctor, who will discuss any unresolved questions with you.

What are the possible results of the NaTALIA prenatal test - and what do they mean?

Your doctor will receive a written report of the results, indicating a low or high risk of the chromosomal disorders tested.

- A **low risk** means that the chromosomal disorders tested can almost certainly be excluded.
- A **high risk** means that there is a high probability that the particular chromosomal disorder is present.

However, this result can also occur for a pregnancy with a healthy unborn child. Hence in the case of each abnormal result, **further tests are required for confirmation** (usually invasive tests such as amniocentesis) before irreversible decisions about the pregnancy are made. Your doctor will advise you on the next steps.

Investigated chromosomal disorders

	Prevalence	Symptoms
Trisomy 21 (Down syndrome)	1:700	mental and physical developmental disorders, congenital heart defects
Trisomy 13 (Patau syndrome)	1:5000	severe mental developmental disorders, varied physical abnormalities, life expectancy mostly less than 1 year
Trisomy 18 (Edwards syndrome)	1:3000	

Investigated chromosomal disorders of the sex chromosomes

	Prevalence	Symptoms
Monosomy X (Ullrich-Turner syndrome; X0)	1:3000^{+}	Infertility, microsomia, heart defects
Klinefelter syndrome (XXY)	1:1000*	Often undetected, infertility, risk of slight learning difficulties
Jacobs syndrome (XYY)	1:1000*	Mostly unobtrusive
Trisomy X (XXX)	1:1000^{+}	Mostly unobtrusive

$^{+}$among female new-borns, *among male new-borns