LADR Ihr Labor vor Ort

Information for Patients

Early Diagnostics during Pregnancy: Assessing the Risk of Congenital Disorders

Thorough advice for a free decision

Many parents would like their doctors to tell them whether everything is all right, even before the baby is born. The risk that the child may be suffering from birth defects can be assessed using modern diagnostic methods. Speak with your doctor. Receiving thorough advice is important so that you can make your own free decision whether to undergo testing and which analyses you would like. A little information now might give you a few pointers.

Risk factors

Age and genetic predisposition play an important role in chromosome abnormalities.

Age

The risk of chromosomal abnormalities rises among older mothers. The most familiar of them is trisomy 21, also known as Down syndrome. The risk of a 35-year-old woman giving birth to a child with trisomy 21 is 1:350. In other words: of 350 women who become pregnant at the age 35, one will give birth to a child with Down syndrome. It is important to bear in mind that some of the foetuses will die after the 12th week of pregnancy, so the probability of trisomy 21 in the 13th week of pregnancy is higher than 1:350. Other chromosomal abnormalities are trisomy 18 (Edwards syndrome) and trisomy 13 (Patau syndrome), although few of these children survive the full term.

Genetic predisposition

Among the other risk factors are a family history of neural tube defects or Down syndrome, hydrocephalus – also known as water on the brain – among close relatives, malformation of an earlier child, as well as a history of miscarriage or stillbirth.

Would you like clarity?

If so, there are two laboratory tests: first trimester screening or the non-invasive prenatal test. These tests help your doctor to provide better and more precise advice on the risks and possible consequences.

First trimester screening

11th–14th week of pregnancy

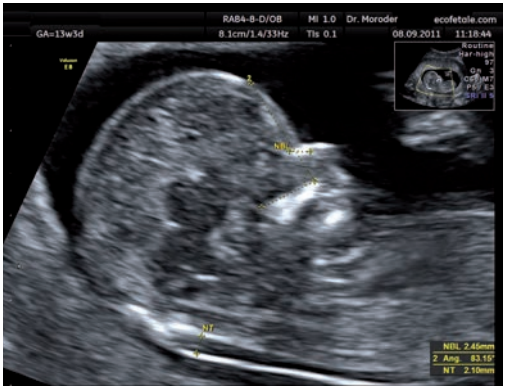
Laboratory testing is performed on two substances from the mother's blood serum, namely PAPP-A and free β-HCG. The doctor also uses ultrasound to measure the child's so-called nuchal translucency. The combination of these findings allows your doctor to state the probability that your child may be affected by trisomy 21, 18 or 13. If the findings exceed certain thresholds, your doctor can perform additional tests in order to make an even more precise statement.

Non-invasive prenatal test

Possible from the 10th week of pregnancy

A non-invasive pre-natal test (NIPT) can be sensible in cases of particular genetic risk, or if the ultrasound or blood screening reveal abnormalities. The LADR laboratories cooperate with the Centre for Human Genetics and Laboratory Diagnostics (MVZ) Dr. Klein, Dr. Rost and Colleagues to perform a so-called Prenatalis® Test. A state-of-the-art procedure, this test examines the child's DNA directly for trisomy 13, 18 and 21. It only requires a normal blood sample from the vein, as the mother's blood contains the child's free genetic material. This test is possible from the 10th week of pregnancy. The findings are available after around 7 workdays. The procedure is very precise and can add to the risk assessment.

Laboratory tests help the doctor to provide good advice on the risks and possible consequences.



Measurement of nuchal translucency by ultrasound

Other chromosomal analyses

Additional, more sophisticated methods to analyse the chromosomes are only appropriate if the risk assessment from first trimester screening is very high. Analysis of placental tissue (chorion villus sampling), an amniotic fluid test (amniocentesis) or cordocentesis may be possible in these cases. However, obtaining the samples for testing in these methods is not entirely free of risk.

Top image: amniocentesis

Individual health services

Examinations to assess the risks of chromosome abnormalities are not included in the prenatal screening that is listed in the maternal health passport. If you are insured by a statutory provider, you can have the tests done as optional services. You will receive an invoice, which you will be required to settle.

If your doctor believes there is a medical necessity, the statutory health insurance may cover all or some of the costs in individual cases. We will then send the laboratory findings to your doctor.

Ask your doctor to explain the costs for the requested analyses and enquire with your health insurance provider which costs it will cover!

For further patient information on many other health issues, go to:

www.LADR.de/patienteninformation

Informationen zu den regionalen Facharztlaboren im deutschlandweiten LADR Laborverbund Dr. Kramer & Kollegen unter www.LADR.de

LADR Der Laborverbund Dr. Kramer & Kollegen GbR
Lauenburger Str. 67, 21502 Geesthacht, T: 04152 803-0, F: 04152 803-369, interesse@LADR.de

Stand 06/2019