praenatalschall.de
Praxis für pränatale Diagnostik München

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General information about prenatal diagnostics

Dear Patient,

with this download information we would like to introduce our range of prenatal diagnostic services as well as some basic terms and principles of prenatal diagnostics. The general term prenatal diagnostics comprises all the examinations and scans that are applied during pregnancy. In this respect, the special ultrasound scans are used to give the most accurate assessment of the unborn child. As a rule we are able to reassure you with inconspicuous findings and show you that the baby is developing normally.

In rare cases an abnormal development is detected: this may involve chromosomal abnormalities, hereditary disorders or developmental defects of the organs as well as growth problems. Chromosomal abnormalities mean that the chromosomes, which carry the genes, show deviations from the norm: the number or structure of the chromosomes may be changed in this case. The most common chromosomal abnormality, and therefore probably the best known one, is Down's Syndrome. Due to an error when dividing, there are three instead of two copies of chromosome 21 in each cell (trisomy 21). The risk of this increases with the mother's age. Changes in the genes that lead to the actual hereditary conditions can only be determined by targeted screening tests as a rule. If you have a genetic illness or there is one in your family, we recommend that you firstly seek a human genetic consultation. Before a planned ultrasound scan, a basic genetic consultation is also carried out by us.

There are also illnesses or malformations, however, which do not accompany a chromosomal abnormality or a familiar genetic modification, such as a heart defect in the child. Sometimes external influences like medicines, the effects of nicotine and alcohol, X-rays or infections in the mother can be partly responsible for these.

Below we would like to give you an overview of the different examinations so that you can take your time getting information and guidance yourself. We are also happy to talk to you personally to help you find out which prenatal examinations make sense for you. You can find more information on our website under "Downloads" (www.praenatalschall.de/Information/info material/).

First trimester screening / nuchal translucency scan
The examination consists of two components: firstly, with the help of various parameters possibly combined with a blood analysis, a risk assessment is made regarding the presence of a chromosomal abnormality. The decision for or against invasive diagnostics can then be made on a much more individual basis than just by the age of the pregnant woman. Secondly, at a very early stage already, the physical development of the child is depicted as comprehensively as possible and an early precise diagnosis (organ ultrasound) is made. In addition, your individual risk of pre-eclampsia (gestosis or "toxaemia of pregnancy") is determined in order to monitor the pregnancy more intensively or arrange for early therapy if need be.

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Chorionic villus sampling (CVS) and amniocentesis (AC)
These belong to the so-called invasive procedures, in other words methods where puncturing is necessary and hence there is a certain intervention risk. They are usually applied when there is an increased risk of chromosomal abnormalities or hereditary conditions. The procedures are distinguished above all by their timing (CVS approx. 12th/13th week of pregnancy, AC approx. 16th/17th week of pregnancy). The adjusted intervention risks are similar for both procedures. We will advise you individually as to which procedure is the most suitable for you.

Cytogenetics / Molecular genetics / Array CGH / CES
Following invasive diagnostics procedure, the material taken is examined in a genetic laboratory. Various methods can be used here. The cytogenetic examination represents the classic chromosomal analysis with assessment of the number and structure of the chromosomes. Individual gene loci can be examined for certain disorders using molecular genetics. Using the array CGH method it is possible to check all chromosomes with high resolution simultaneously for tiny losses (microdeletions) or gains (microduplications). Additionally, there are specific gene panels (clinical exome sequencing, CES) which can also be a valuable tool in the setting of sonographic abnormalities.

Organ ultrasound / Precise diagnostics / Echocardiography / Doppler / 3D/4D
Preferably in the 21st to 22nd week of pregnancy, this major ultrasound scan can be used to examine the anatomical development of your child with the highest resolution and accuracy. In case of defective development, organ malformations such as heart defects in the child or early developmental defects can be detected and the pre and post-natal management can be optimised in these cases. In the advanced stages of pregnancy, a growth ultrasound with blood flow measurements is also used to detect a placenta insufficiency in good time. 3D/4D sonography can provide additional information in case of certain issues and thus improve the diagnostics. Furthermore, we are able to give you a very realistic impression of your child with the three-dimensional representation. In the following table you will find the meaningfulness of the different prenatal examinations offered by our establishment in summary:

Blood test for trisomy 21 / NIPT
In Germany, (three) different so-called “Non-invasive prenatal tests (NIPT)” are currently being offered. The prerequisite for conducting the tests is a comprehensive ultrasound before taking a blood sample. The blood sample is then taken after a human genetic consultation. A result can usually be expected after a few (10) working days. It is important for us to stress that the new tests are not to be valued as diagnostic tests, but as screening tests with the highest reliability. An abnormal test result must therefore be confirmed by an invasive diagnostic procedure (chorionic villus sampling, amniocentesis). A normal test can rule out trisomy 21 with a high degree of certainty.

We hope to have given you sufficient information with this summary. For all further information we refer you to our website www.praenatalschall.de and of course we are at your disposal for a personal consultation