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Chorionic villus sampling / placenta tissue biopsy

Reasons for chorionic villus sampling – indications:

The term “invasive diagnostics” refers to procedures used to take cell’s from the child for further examination. Whilst developmental defects and organ anomalies are detected with the pure ultrasound scan, chorionic villus sampling is used to detect chromosomal abnormalities, meaning changes in the number and structure of the chromosomes. Normally every person has 46 chromosomes in each cell of their body, which carry genetic information.

Around the conception stage, random defects can occur during division. The most familiar of these random errors is trisomy 21, Down’s Syndrome. In every pregnancy there is a certain degree of risk that the child could have a random defect of this kind. This risk increases the older the pregnant woman is (age indication). The chorion is a layer of cells on the outside of the amniotic sac. The chorionic cells develop into so-called chorionic villi, which go on to form the foetal part of the placenta. Although these cells are not part of the unborn child, they are genetically identical as a rule.

As chorionic villus sampling can be carried out as early as the 12th week of pregnancy, it is suitable for couples wanting examination results as early as possible. Other reasons for chorionic villus sampling may be conspicuous findings from the screening in the first trimester (measurement of nuchal translucency and the blood parameters beta hCG / PAPP-A), from the ultrasound scan of the foetus or in case of inherited and frequently occurring disorders.

Although heart defects and other structural anomalies are often found alongside many chromosomal abnormalities (such as Down’s Syndrome), in some cases the organ diagnostics reveal no noticeable abnormalities.

The risk of a chromosomal abnormality is indeed reduced if the ultrasound scan is normal, but this cannot be definitely ruled out with ultrasound. It is only possible to rule out a chromosomal abnormality with certainty by means of invasive diagnostics.

The blood test for trisomy 21, 13 and 18 from the mother's blood, which has been available for a few years, is unable to answer many questions resulting from such situations and therefore does not fully replace invasive diagnostics as a rule This blood test may be helpful in certain situations, however, and we will be happy to advise you about this face to face.

The actual hereditary conditions are based on genetic changes, which have either just emerged or were already present in one or both parents. Examples of these kinds of conditions are cystic fibrosis or other metabolic disorders. With the majority of possible problems, the ways of detecting a condition and also any consequences of it must be clarified during a human genetic consultation before carrying out invasive diagnostics. If no cases of disorders have previously been known in the families, prenatal screening tests for the presence of such conditions are only seldom possible.

Examination process

Chorionic villus sampling is the earliest way of performing invasive diagnostics. It normally takes place between the 12th and 14th week of pregnancy. Before each chorionic villus sampling, a detailed ultrasound scan is performed. You do not have to undergo the procedure without an anaesthetic. Under constant ultrasound guidance and under sterile conditions, following a local

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anaesthetic a thin needle is pushed into the placenta through the abdominal wall and tissue is taken from the placenta. The needle remains outside the amniotic cavity meaning that any injury to the foetus is ruled out. Part of the tissue taken is treated in a direct preparation in such a way that a preliminary result is available after just 1 to 2 days. A culture is set up from the second part of the sample. A final result can be expected after 2 to 3 weeks, which can sometimes differ from the result of the direct preparation. In these cases, which are very rare however, a further invasive procedure may be necessary.

Value of the diagnostics and possible problems

In the case of chorionic villus sampling, a so-called direct preparation is carried out from part of the tissue sample taken. Using special techniques, it is possible to determine the broad structure and number of chromosomes within just 24-48 hours here. A normal result largely rules out the most common chromosomal abnormalities (these are trisomies 21, 18 and 13 and Turner syndrome). The remaining tissue is then used to set up a long-term culture, whose result is available after approx. 2-3 weeks. A study of the cultured cells then forms the final result, from which even minor anomalies (if they are microscopically detectable) can be recorded or ruled out.

In seldom cases, no clear diagnosis can be formulated from the chromosomal examination. This may be due to the fact that one normal and one abnormal cell line are located next to each other (so-called mosaicism). For further clarification, it may be necessary to examine other cells from the child (amniotic fluid) or the chromosomes from the parents' blood. It cannot be ruled out with absolute certainty that the mother's cells are growing in preference to the child's cells. In case of twins, the results may only apply to one twin under unfavourable conditions.

In seldom cases, growth may progress very slowly or not at all, meaning that we need more than the usual time to obtain a result or – very rarely – we get no result.

For further clarification, in these cases it may be necessary to examine other cells from the child (e.g. by examining the amniotic fluid) or the chromosomes from the parents' blood.
If there are known genetic disorders in the family (where mostly not a whole chromosome but only small sections on the chromosome – the so-called genes – are changed), in some cases it is possible to check these too (molecular genetic examination).

Changes to very small chromosome segments or to single genes cannot be detected under the microscope. The array CGH diagnosis provides an approx. 100 times higher resolution compared to conventional methods and is thus able to detect minor abnormalities (e.g. microdeletion syndromes) at submicroscopic level. This technology is not paid for by the statutory health insurance schemes in routine cases, however. Additionally, there are specific gene panels (clinical exome sequencing, CES) which can also be a valuable tool in the setting of sonographic abnormalities.

In the event of abnormalities or unclear findings, we recommend a consultation from a specialist in human genetics, who will discuss these with you face to face. On request or in case of specific issues, a specialist genetic consultation may also be useful before chorionic villus sampling.

Limits of the examination

As no amniotic fluid can be taken during chorionic villus sampling to determine the alpha-fetoprotein (AFP) to provide evidence of open gaps in the spine, an ultrasound scan should be done in the 20th22nd week of pregnancy in order to rule out defects of this kind (and also other physical malformations).

The possibility that the expected child will reveal other physical or mental abnormalities, despite the diagnosis of a normal set of chromosomes, cannot be ruled out. Although these organ malformations can be shown with high probability by differential organ diagnostics (organ ultrasound) in the 20th - 22nd week of pregnancy, an unremarkable examination can never fully guarantee a healthy child