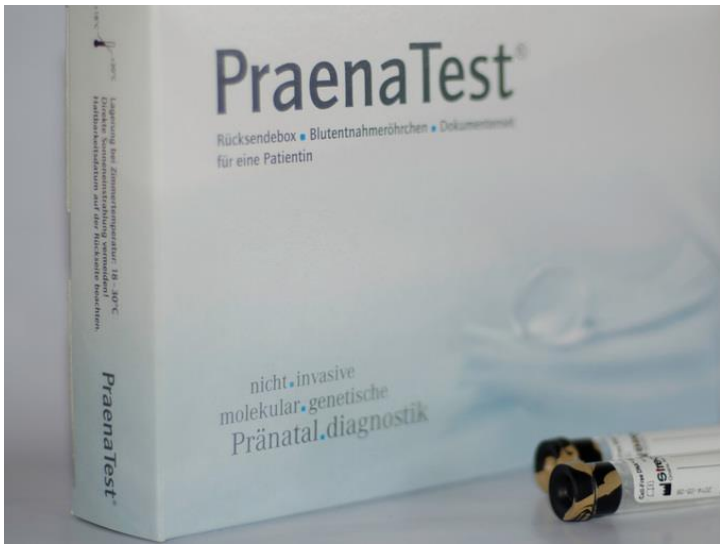
Is 20 ml of maternal blood enough to diagnose Down syndrome?



As you may know, Down syndrome, or trisomy 21, occurs when there is an extra copy of chromosome 21. Trisomy 13, also known as Patau syndrome, results from a genome mutation and is characterised by a third copy of chromosome 13. Children with Patau syndrome suffer from severe mental deficiency. Trisomy 18, also known as Edwards syndrome, is a severe developmental disorder that is caused by the presence of all or part of an extra 18th chromosome.

LifeCodexx AG has developed PrenaTest®, an innovative diagnostic test that is able to reliably confirm or exclude trisomy 21, 18 and 13 just from a simple blood sample (!) of a pregnant woman. The non-invasive test appeared recently on the market as an alternative to current invasive tests (CVS and amniocentesis) which may result in miscarriage.

This test now is available in UAE!!

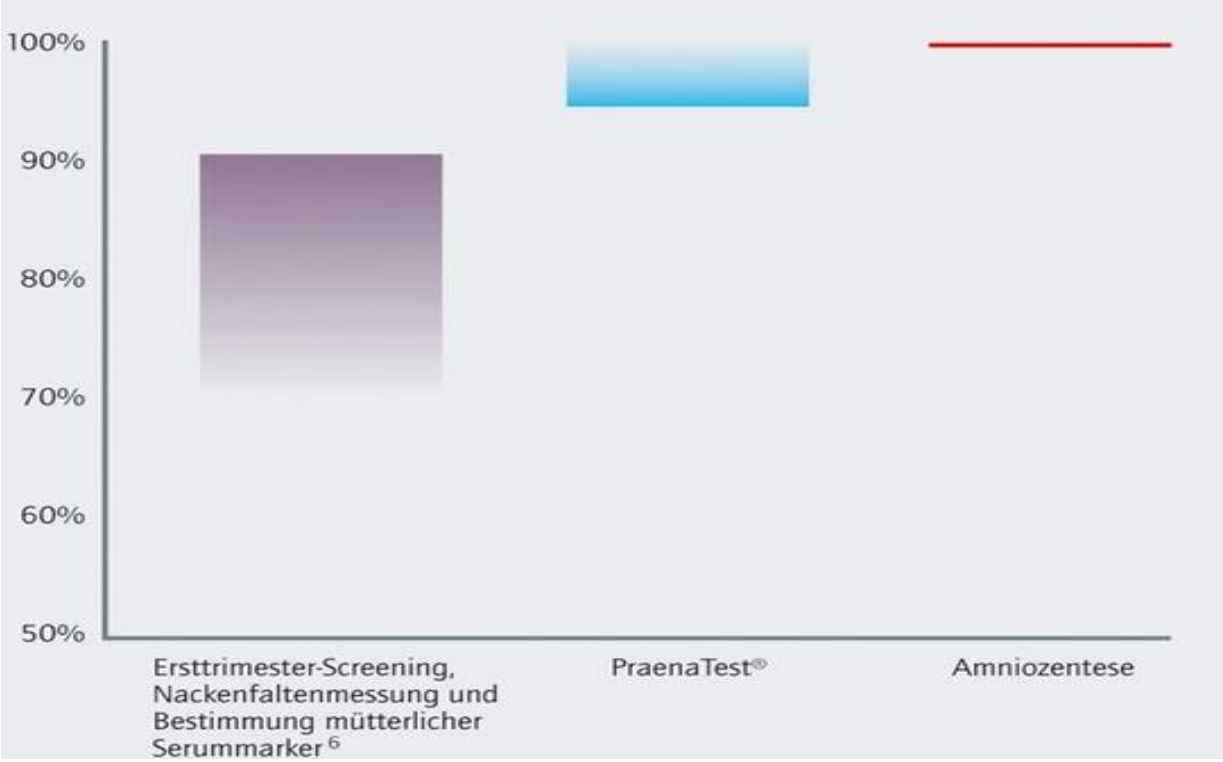
Down syndrome, also known as trisomy 21, is a chromosomal condition caused by the presence of a third copy of chromosome 21 instead of the usual two in each cell in the body. Current tests to confirm or exclude trisomy 21 involve invasive methods, including chorionic villus sampling(CVS) from the placenta, or amniocentesis. Both methods provide reliable results; amniocentesis has a reliability rate of up to 99.9 percent.

Despite the reliability of the tests, they nevertheless represent a risk for the unborn babies. The risk of miscarriage from amniocentesis and chorionic villus sampling is around 0.2 to 1 percent. The innovative PrenaTest® offers the possibility of significantly reducing the number of miscarriages resulting from amniocentesis and chorionic villus sampling.

Cell-free DNA is used for testing

The innovative test is based on the analysis of cell-free DNA (cfDNA) (1) in the blood of pregnant women. cfDNA is not enclosed by cells, is present in small fragments and circulates freely in the blood of pregnant woman. Besides maternal DNA, the blood of pregnant woman contains between two and 30 % (on average around 10 %) foetal DNA (cell-free foetal DNA, cffDNA). This DNA is derived from dead placental cells that are released into the women's blood (2). The individual fragments have a lifespan of under two hours; cffDNA can thus no longer be detected in the blood of a baby's mother even just a few hours after birth (3).

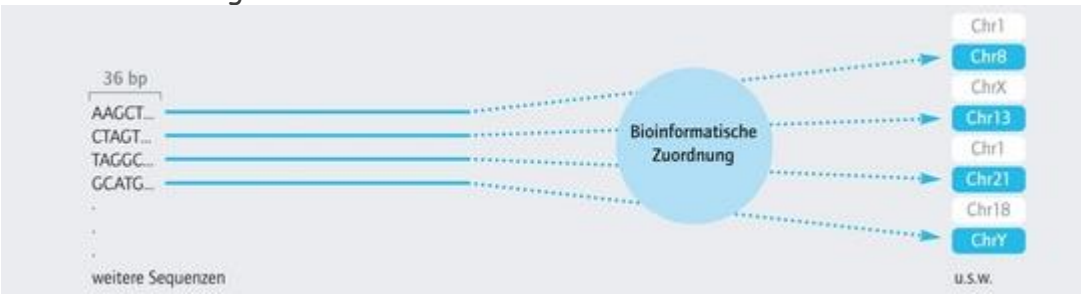
The non-invasive PrenaTest® is based on molecular biology and bioinformatic methods and designed to examine whether there is an elevated amount of chromosome 21-related cffDNA and hence confirm or exclude the presence of trisomy 21 (Down syndrome) in the unborn baby. The examination only requires 20 ml of venous blood from the pregnant woman.



Comparison of invasive and non-invasive prenatal diagnostics methods with regard to their ability to detect foetal trisomy 21. (© LifeCodexx AG)

Novel technology

The test is based on innovative sequencing methods (next-generation sequencing) applied to the DNA isolated from the blood plasma of pregnant women. These methods enable millions of DNA fragments to be sequenced in one single run. A specifically developed software programme determines the quantity of chromosome 21 DNA. A validated algorithm then calculates whether the quantity of chromosome 21 DNA detected is within the standard range or not.



The PraenaTest® is based on state-of-the-art high-throughput DNA sequencing methods, amongst other things. The DNA sequences are analysed using bioinformatic tools, including the assignment of the DNA to the chromosomes. (© LifeCodexx AG)

Reliability of the test

PrenaTest® is only available to pregnant women in the 12th week of pregnancy or later who have a higher risk of chromosomal alterations in the unborn child. The result of the examination is available two to three weeks after blood has been taken from the pregnant women. The efficiency in diagnosing trisomy 21, 18 and 13 in the blood of pregnant women was examined in clinical studies and rated as relatively high. Five prenatal centres and university hospitals in Germany and Switzerland collected blood samples of more than 500 pregnant women who had a higher risk of chromosomal alterations in the unborn child.

- *First results from the prospective blinded multi-center study demonstrate highest clinical accuracy for the planned commercial PraenaTest® test design based on Cell-Free DNA™ blood collection tubes (BCT), detecting all positive cases of fetal trisomy 21* ***(100% sensitivity)*** *with no false positive calls* ***(100% specificity)****.*