[1] Trisomy

Everyone has 23 pairs of chromosomes, namely 46 individual chromosomes, which contain genetic information (DNA) and of which half come from the mother and half from the father. A trisomy is present if a particular chromosome occurs three times rather than the normal twice. Trisomies occur more frequently with increasing maternal age and effects range from childhood developmental disorders up to reduced life expectancy of the child.

[2] Sex chromosomes

The sex chromosomes X and Y determine the sex of a human being. X and Y chromosomal disorders occur when one sex chromosome is absent, or there is an additional or incomplete copy of a sex chromosome. Syndromes caused by an incorrect distribution of sex chromosomes include Klinefelter syndrome (XXY) and Ulrich-Turner syndrome (XO), also known as monosomy X.

[3] Microdeletions

Loss of a fragment from a chromosome that is so small it cannot be detected by normal chromosome analysis.

Experts do not currently recommended microdeletion screening, and the investigation of sex chromosome aberrations is recommended only after in-depth consultation.

[4] First trimester screening (FTS)

Nuchal translucency measured by ultrasound, together with the determination of two biochemical values from maternal blood, can be used to calculate a risk factor for a fetal chromosomal anomaly between the 10th and 12th week of gestation.

[5] Limitations

In „low risk“ groups (below 35 years old), as with any screening test, frequent “false positive” results are to be expected. This means that the test indicates an abnormality that would not be confirmed when examined by another diagnostic method, e.g., amniocentesis.

[6] Chromosomal mosaic

A chromosome change that is not present in all cells of an organism. A mosaic in the placenta can cause a “false negative” result meaning a chromosomal disorder that is present cannot be detected by NIPT.

Contact

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PATIENT INFORMATION

VERACITY

NON-INVASIVE PRENATAL TEST (NIPT)

INFORMATION FOR PREGNANT WOMEN

As a pregnant woman you wish above all for a healthy baby, but the happiness and expectations of a pregnancy are also accompanied by insecurities.

As well as providing traditional prenatal care, your physician is able to advise you on additional diagnostic options.

There are reliable methods available to determine fetal chromosomal changes. Until recently, these were diagnostic puncture techniques: amniocentesis or chorionic villus sampling. Although the complication rate of these invasive procedures has steadily decreased, they carry a low risk of miscarriage.

A non-invasive prenatal test (NIPT) can determine the risk of the most common fetal chromosomal disorders (trisomy[1)] 21, 18, 13, number of sex chromosomes[2)]) as well as certain microdeletions[3)] and potentially avoids a diagnostic puncture technique indicated by the result of an abnormal first trimester screening (FTS)[4)].

The NIPT VERACITY reliably detects certain fetal chromosome disorders from a maternal blood sample. The test is also suitable for twin pregnancies and after IVF/ICSI. The test can also determine the sex of the unborn child; however, in compliance with the Genetic Diagnostics Act this information can only be disclosed from the 14th week of pregnancy (postmenstrual).

When does the test make sense?

- Maternal age ≥ 35 (trisomy risk)
- Abnormal first trimester screening
- Abnormal ultrasound findings

However, in principle the test is available to every[5)] pregnant woman.

How is maternal blood used to determine chromosomal disorders?

Free DNA (genetic material) fragments of fetal origin that come primarily from the placenta circulate in maternal blood. Following isolation of the fragments from maternal blood, modern analysis techniques can determine numerical deviations from the normal number of 46 chromosomes.

How does the VERACITY test work?

1. Information and genetic counseling

An appropriately qualified medical person must provide a comprehensive explanation and (professional) genetic counseling. The responsible medical person remains your principal contact person throughout the entire VERACITY test procedure. If your physician is unable to perform the genetic counseling, we are happy to help you to find a genetic counseling center in your area.

2. Blood sample collection

Following your written consent, your physician will collect a blood sample (20 ml).

3. Analysis

The blood sample will be sent in a special transport box by courier to the medical laboratory (MVZ) Martinsried, where it will be tested by state-of-the-art analytical methods.

4. Result

After 5-10 working days (Monday to Friday), your physician will receive the result and can discuss the findings and clarify any unresolved issues with you.

As the VERACITY test is considered a genetic analysis in the sense of the Genetic Diagnostics Act, it is performed only after information has been provided by the responsible physician and after genetic counseling and your written informed consent.

Which results are possible?

An unnoticeable or inconspicuous result: normal pregnancy – prenatal care including ultrasound.

A noticeable or conspicuous result: counseling and verification preferably with amniocentesis.

What are the limitations?

Chromosome disorders other than those mentioned, mosaics[6)] and mutations in individual genes cannot be detected.

In rare cases, a result is not possible due to the low amount of fetal DNA (under 3%) and the test must be repeated following collection of a new blood sample. Very rarely, a result cannot be achieved.

What are the advantages of the VERACITY test?

- Early risk estimation (procedure from the 10th week of pregnancy) for trisomy 21, 18 and 13, disturbances of the sex chromosomes and microdeletions if necessary
- No intervention risk (associated with invasive procedures)
- Procedure and analysis exclusively in Germany
- Evaluation of results and consultation in a medical-led laboratory