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Information to Combined test

- Most children are born healthy.
- In every pregnancy and in every woman there is a small risk that the child will be born with a bodily or a mental handicap. In some cases the reason for this handicap is a chromosomal anomaly called trisomy 21 (earlier name: Down syndrome). This is just one of many known anomalies.
- The risk of a child being born with trisomy 21 increases with maternal age.
- A Combined Test can correctly identify about 93% of fetuses (=babies) with trisomy 21.
- The Combined Test assesses the risk for the fetus to be affected by any one of the most common trisomies (trisomy 21, trisomy 18 and trisomy 13) by combining ultrasound observations with special maternal blood examination.
- ***The test does not give you 100% certainty. It is not a diagnostic test.*** **Only an invasive test (needle test) can give a diagnosis (100% certainty).**
- The **combined test** measures the size of the fetus, also the fluid under the skin at the back of the neck (**nuchal translucency**), further **first trimester ultrasound markers** for chromosomal anomalies. Also a **small blood sample** will be taken from the mother to test for possible signs not visible in ultrasound.
- It can also detect many (but not all) of the major developmental anomalies of the fetus.
- Not all fetuses with an increased risk are truly affected by a chromosomal anomaly (false positive result ca. 2,5%)
- **Many fetuses are healthy despite a screen-positive result (=increased risk) after Combined Test.**
- Most common reasons for a screen positive result in chromosomally normal fetuses are cardiac defects, genetic syndromes and infections in the pregnancy.
- The only way to have **100% certainty** about the chromosomes of your baby is to opt for an **invasive test** (needle test) like **chorionic villous sampling** (CVS or placenta test) or an **amniocentesis** (AC=amniotic fluid test).
- **A combined test does not have a miscarriage risk!** The invasive tests (CVS and AC) have **miscarriage rate** of about 0,2% (1:500) or lower – this is called procedure related risk.
- An invasive test will not be performed today!
- **It is best to combine a combined test with a screening test for preeclampsia.**

Combined Test – this is how it is done:

You receive a detailed counselling about your options for first trimester screening (this includes written and verbal information about Combined Test, NIPT (non invasive prenatal test a.k.a. cell free fetal DNA, preeclampsia screening and invasive testing).
The scan is usually performed through your abdomen, only seldom it is necessary to perform a vaginal ultrasound.
At the time of the scan you will learn about the **results of the ultrasound assessment**. **These are preliminary results.**
The final results of the first trimester risk assessment (Combined Test w/wo Preeclampsia screening) are usually very similar to the preliminary result you receive at your visit and it **will be sent to your gynaecologist**. Routinely you will be **informed by SMS**, rarely it is necessary to explain the results in more detail – in this case you will be called and sometimes even offered an appointment for further counselling.

Results of 1:1000 are considered as good.
Results between 1:50 – 1:1000 are called intermediate risk – you should consider going for an NIPT.
In case of results of 1:50 or worse you will be offered an invasive test (CVS or amniocentesis).

Important to know: A screen-negative result unfortunately **does not guarantee a healthy fetus** since the Combined Test is a screening and not a diagnostic test.
Even **in case of a screen-positive result** most fetuses are not affected by a chromosomal anomaly. An **NIPT** (**N**on-**I**nvaseve **P**renatal **T**est also known as **C**ell-**f**ree **f**etal DNA Test) can give you more information, but ultimately only an invasive test like a CVS or an amniocentesis can prove or exclude a chromosomal anomaly or a genetic disorder. This is your decision and you do not have to have it done if you do not want to. Your doctor cannot make this decision for you since this is a very personal decision.

Irrespective of the results of the Combined Test it is recommended that a detailed ultrasound scan (anomaly scan = organscreening) will be performed at 20-22 weeks to detect fetal developmental anomalies.

If you have more questions please address them at your appointment.

Consent to *Combined Test*

I have read and understood the above information and I have all my questions answered by Dr Csapó.
I wish to have a Combined Test and with my signature I consent to the examination:

Date _____ Signature _____