



# Prenatal Scanning & Testing

*Information  
for  
Patients*

Dear Patient,

The following information will give you an idea of tests during pregnancy (prenatal tests) offered by the University Hospital of Cologne or - to some extent - at your gynaecologist's surgery.

This information is supplemental to what you will learn in a genetic counselling session but it is not intended to replace such counselling. If you have any questions on prenatal care, please contact us or the prenatal care / ultrasound surgery at the Gynaecology & Obstetrics Department, University Hospital of Cologne; Phone: +49/221/478-4975.

In prenatal care, screening and diagnostic tests are distinguished.

**Screenings** aim to contribute to a better *risk estimation* of a fetal malformation / chromosome abnormality / metabolic disorder.

**Diagnostic tests** aim to *confirm/rule out with certainty* a malformation / chromosome abnormality / metabolic disorder that has been suggested by family history / previous screening.

Prenatal Screening & Testing - Schedule	
non-invasive screening	invasive diagnostic test
no risk	varying risk (0.3-1%)
1st trimester: Maternal serum markers: PAPP-A (10 weeks) β-HCG (13 weeks) + Fetal ultrasound (11-13 weeks) (nuchal translucency, "NT" other measurements, if appropriate)	10-13 weeks of pregnancy  Chorionic villi sampling (11-13 weeks) (CVS)
2nd trimester  Fetal ultrasound (21 weeks or more) (less distinctive malformations of unborn baby)	15-22 weeks of pregnancy Amniocentesis (>15 weeks) (AC)

The above table shows Optimum timing for first trimester screening. In many cases, several screening measurements are performed on the same day, resulting in a slight reduction in accuracy.

## Explanatory information:

- 1.) With a combination of **serum screening** and **fetal ultrasound**, specialists are usually able to calculate relatively reliable probabilities:
  - ◆ approx. 95% of all children with Down's syndrome are detected.
  - ◆ approx. 5% of the women screened get a false alarm (i. e. the results are abnormal but the child does not have Down's syndrome)
- 2.) Screenings are usually not covered by the German statutory health insurance system but must be paid by the patients. Approximate costs:
  - ◆ Maternal serum screening: about 50 €
  - ◆ 1st trimester fetal ultrasound: 200-300 €
- 3.) During **chorionic villi sampling (CVS)**, a small piece of the placenta is taken.
  - ◆ In one out of 100-200 cases, pregnancy complications may occur as a result of the test (bleeding, infection, loss); i.e. additional risk due to test: 0.5-1%.
  - ◆ Final chromosome results take about 10 days.

Failure to obtain a chromosome result occurs slightly more often in CVS compared to amniocentesis. Therefore, chorionic villi sampling is usually performed in situations where a known serious genetic defect in the family should be investigated as early as possible in pregnancy. In all other cases, amniocentesis is generally preferred.

- 4.) During **amniocentesis (AC)** an amount of 10-20 ml of amniotic fluid is taken, containing fetal tissue.
  - ◆ In one out of 200-300 cases, pregnancy complications may occur as a result of the test; i.e. additional risk: 0.3-0.5%.
  - ◆ Final chromosome results within 10 to 14 days.
  - ◆ Additional information: measuring alpha-fetoprotein (AFP) in the amniotic fluid allows to detect possibly existing open spina bifida.

For most, although not all chromosome results, we will be able to tell you whether or not they will cause a disorder in the unborn baby.

- 5.) The costs for CVS and AC are borne by the German health insurance companies. However, additional investigations as for example a rapid test (specific test for Trisomy 13, 18, 21 within 48 hours) must be paid by patients.

If you have any further questions on prenatal diagnosis, we will be happy to help you.

Please contact:

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