

# Down Syndrome

The following examinations are available:

First trimester screening – weeks 11+0 to 13+6

Triple test – 14+0 to 18+6

Quadruple test – 14+0 to 18+6

Integrated test – 1<sup>st</sup> screening 11+0 to 13+6, 2<sup>nd</sup> screening 14+0 to 18+6

General:

Risk calculations cannot provide 100% guarantee but will help in making decisions regarding amniocentesis based on scientifically evaluated risk assessments. Also other fetal malformations cannot be excluded. Please note that FML, Synlab and collaborating laboratories are certified according to the Fetal Medicine Foundation Guidelines (FMF).

For the optimal risk calculations we require the following data:

<b>Required details</b>	
<b>Date of birth (mother)</b>	<b>Date of blood collection</b>
<b>Weight (mother)</b>	<b>Single fetus / twins</b>
<b>Week + day of gestation</b>	<b>Ethnicity</b>
<b>CRL in mm (for 1<sup>st</sup> trimester)</b>	<b>Smoker status</b>
<b>NT in mm (for 1<sup>st</sup> trimester)</b>	<b>Diabetes mellitus status</b>
<b>Date of the ultrasound examination</b>	<b>Previous chromosomally abnormal child/fetus (NTD or trisomy)</b>

- **First Trimester Screening (PAPP-A, free beta-HCG, NT)**

General:

The screening involves a combination of ultrasonographic measurement of nuchal translucency (NT) and biochemical analysis of maternal serum levels of two pregnancy-related proteins: free beta-hCG (beta-human chorionic gonadotropin) and PAPP-A (Pregnancy Associated Plasma Protein-A). Please note that numerous studies have revealed that the first trimester screening is far more sensitive for detecting Down syndrome than the second trimester screening (Triple test). It is currently the most effective non-invasive screening for Down Syndrome risk calculation available. Nicolaidis et al. described the standardization of sonographic examinations and in particular, the nuchal translucency measurement.

Together with statistical evaluation of biochemical characteristics, it permits an early recognition of approx. 90% of the cases with Down syndrome with a false positive rate of approx. 5% for the 1st trimester screening. This accuracy is achieved if ultrasonic examiners and laboratories work with the required quality standards. We calculate the risk of the 1st-trimester screening with the FMF certified software - "Astraia". Due to the algorithm, a calculation is possible only from the week of gestation 11+0 to 13+6 and a CRL of 45-84 mm. By combining the results of the ultrasound and blood test along with the age, ethnic origin and weight of the mother, a statistic evaluation of the biochemical parameters gives the NT-adjusted risk calculation which allows an early recognition of **approx. 90% of Down syndrome cases** as well as trisomies 13 + 18 at a false-positive rate of approx. 5%.

Freiburg Medical Laboratory calculates the risk of the FTS with FMF-certified software. A calculation is possible starting from weeks of gestation 11+0 to 13+6 and a crown-rump length of 45-84 mm. In our results, we indicate the adjusted (combined) risk for trisomy 21 apart from the risk based on the mother's age. For a risk greater than **1:150** further clarification is recommended. Similarly, a negative result does not completely rule out these abnormalities but instead decreases the specific risks for Down syndrome and trisomies 13 + 18.

**Material:** 2 ml serum, and please contact us for the special request form. This form must be used to fill in all required patient data. Click [here](#) for download.

**TAT:** 2-3 days, FML

**Preanalytics:** If transportation time is >24h, the sample must be frozen for dispatch!

**Parameters:** PAPP-A, free beta-HCG

**Comment:** In cases of a calculated risk higher than 1:150 the patient should be considered for amniocentesis. Normal results obtained with these tests point to a normal constellation regarding trisomies 13, 18 and 21; however, they cannot be excluded 100%. Other genetic defects are not included in the risk calculations.

**Note:** Chromosomal analysis or FISH for confirmation!

- **Second Trimester: Triple Test (AFP, beta-HCG, free estriol)**

General:

The Triple Test has a detection rate >70 %, false-positive rate about 5% depending on the examined age group. Additionally, neural tube defects are recognized by the AFP determination. Time period: week of gestation: 14+0 to 18+6.

Material: 2 ml **frozen** serum and please contact us for the special request form. This form must be used to fill in all required patient data. Click [here](#) for download.

TAT: 7-10 days\*

Parameters: AFP, beta-HCG, free estriol

Comment: In cases of a calculated risk higher than 1:150 the patient should be considered for amniocentesis. Normal results obtained with these tests point to a normal constellation regarding neural tube defects and trisomies 18 and 21; however, they cannot be excluded 100%. Other genetic defects are not included in the risk calculations.

**Note: Chromosomal analysis or FISH for confirmation!**

- **Second Trimester: Quadruple Test (AFP, beta-HCG, free estriol, Inhibin A)**

General:

The Quadruple Test has a detection rate >80 %, a false-positive rate about 5% depending on the examined age group. Additionally, neural tube defects are recognized by the AFP determination. Time period: week of gestation: 14+0 to 18+6.

Material: 2 ml **frozen** serum and please contact us for the special request form. This form must be used to fill in all required patient data. Click [here](#) for download.

TAT: 7-10 days\*

Parameters: AFP, beta-HCG, free estriol, Inhibin A

Comment: In cases of a calculated risk higher than 1:150 the patient should be considered for amniocentesis. Normal results obtained with these tests point to a normal constellation regarding neural tube defects and trisomies 18 and 21; however, they cannot be excluded 100%. Other genetic defects are not included in the risk calculations.

**Note: Chromosomal analysis or FISH for confirmation!**

- **Second Trimester: AFP as part of the Triple/Quadruple Test**

General:

The AFP concentration in the 2<sup>nd</sup> trimester is used in the detection of neural tube and abdominal wall defects (gastroschisis). AFP-MoM (multiple of median) is the basis for the assessment also in combination with other parameters (triple test). Time period: week of gestation: 14+0 to 18+6.

Material: 2 ml serum

TAT: 7-10 days\*

Comments: AFP-MOM < 2.0	without pathological findings
AFP-MOM > 2.0 < 2.5	borderline constellation, ultrasound clarification is recommended
AFP-MOM > 2.5	increased MoM, ultrasound clarification required
AFP-MOM < 0.5	decreased MoM, triple test is recommended

- **Integrated Test (First and Second Trimester)**

General:

The Integrated Test has a detection rate >90 %, false-positive rate about 5% depending on the examined age group. Serum is collected twice, during the 1st and then 2nd trimester. Ultrasound data at the time of blood collection during the 1st trimester are also required (CRL, NT).

Time period, 1st collection: week of gestation: 11+0 to 13+6.

Time period, 2nd collection: week of gestation: 14+0 to 18+6.

Material: 2 ml frozen serum and please contact us for the special request form. This form must be used to fill in all required patient data. Click here for download.

TAT: 7-10 days\*

Parameters: PAPP-A in weeks 11+0 to 13+6  
AFP, beta-HCG, free estriol in weeks 14+0 to 18+6

Comment: In cases of a calculated risk higher than 1:150 the patient should be considered for amniocentesis. Normal results obtained with these tests point to a normal constellation regarding neural tube defects and trisomies 18 and 21; however, they cannot be excluded 100%. Other genetic defects are not included in the risk calculations.

Note: **Chromosomal analysis or FISH for confirmation!**

For complete list of laboratory test offered at Freiburg Medical Laboratory, please visit <http://www.fml-dubai.com/parameter-listings/>