

Antenatal Care: Routine Labs and Tests During Pregnancy (Aneuploidy Screening)

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Antenatal care plays a vital role in the good health of a pregnant woman and her yet-to-be-born baby. According to the World Health Organization, the goal of antenatal care is to prepare the pregnant woman for childbirth and prevent, diagnose, alleviate and treat health problems arising during the pregnancy. Good antenatal care helps to reduce maternal and neonatal mortality as well as morbidity rates.



Features of Routine Antenatal Care

The essential aspects of routine antenatal care include:

- Identification and follow-up for the pregnant woman and her unborn child with at least four (ideally ten visits for nulliparous and 7 visits for multiparous women) antenatal visits to the healthcare facility. This includes **confirming the pregnancy** with a urine pregnancy test during the first visit, between the 8th and 12th week, along with **routine laboratory tests**.
- Early detection and treatment of **anemia**, **gestational diabetes** and pregnancy-induced **hypertension** (PIH) at the second visit (24-26 weeks)
- Early detection and management of pregnancy-related conditions like **pre-eclampsia** and **gestational diabetes**
- Screening tests to detect **sexually transmitted diseases** and **HIV infection**
- Providing **tetanus toxoid**, **iron**, and **folic acid** supplementation
- Providing information about **healthy lifestyle choices** (avoiding

alcohol/smoking), barrier contraceptive use, etc.

- Evaluating **maternal and fetal well-being** during the third (32 weeks) and fourth (36–38 weeks) visit

Initial Visit

Ideally, the first antenatal visit should be in the **first trimester**. Once the pregnancy is confirmed with history and a urine pregnancy test, screening should be performed, irrespective of the gestational age, in the order described below.

Routine laboratory tests

Complete blood count

A complete blood count includes **hemoglobin level, red cell count, white cell count, platelet count**, and a **peripheral smear**. Low hemoglobin level is indicative of iron deficiency anemia, with red cell morphology indicating the type and severity of the condition. Abnormally high or low platelet levels are a cause for concern, and a hematology consult should be obtained.

Blood group and atypical antibody screen

Identifying **the ABO blood group, rhesus D factor and red cell antibodies** is vital in order to prevent [hemolytic disease in the newborn](#). If the mother is rhesus D negative and the baby is rhesus D positive, then anti-D antibodies can form during a [miscarriage](#), during pregnancy-related bleeding, amniocentesis, external cephalic version or delivery.

These antibodies can affect subsequent pregnancies causing hemolytic disease in the newborn. To prevent this, Rh-negative women should undergo **rescreening** during the second trimester and should be administered **RhoGAM** at 28 weeks. The procedure should be repeated again in the immediate postpartum period if the baby is Rh-positive.

Rubella antibody titer



Transmission electron micrograph of rubella virus.

This should be measured in all pregnancies as it helps to identify women who have either been immunized or are susceptible to contracting [rubella](#). As maternal rubella can cause congenital rubella syndrome, **rubella vaccination** should be given to susceptible women postpartum. Pregnant women should also be advised to avoid contact with individuals suspected to have rubella.

Varicella titer

Varicella titer is required to detect the mother's susceptibility to [varicella](#) and to

vaccinate her in the postpartum period if she is found to be susceptible.

Syphilis serology

Vertical transmission of maternal [syphilis](#) infection is associated with a high incidence of **neonatal mortality and morbidity**, e.g., non-immune hydrops, intrauterine growth retardation as well as severe malformations. Therefore it is important to screen the mother for syphilis with a **Venereal disease laboratory test** (VDRL) or the **Treponema pallidum particle assay** (TPPA).

Urinalysis

Urinalysis between 11 and 16 weeks of gestation is recommended to detect **asymptomatic bacteriuria**.

Also indicated are [gonorrhea](#) and [chlamydia](#) tests to detect infection and apply the appropriate treatment, if present.

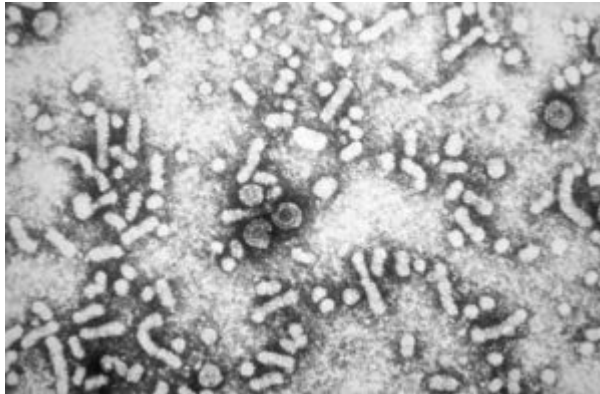


Image: "This electron micrograph reveals the presence of infective hepatitis B virus (HBV) virions also known as "Dane particles." These particles measure 42nm in their overall diameter and contain a DNA-based core that is 27nm in diameter." By CDC. License: Public Domain.

Hepatitis B serology screening

This screening should be offered especially for pregnant women who have had **multiple sexual partners** or have a history of **intravenous drug use**. Vertical transmission of [hepatitis B](#), especially if mothers have an active HBeAg infection, results in the infants getting infected and becoming lifelong carriers and developing **chronic liver disease**. Therefore, in addition to screening mothers, the **hepatitis B vaccine** and **immunoglobulin** should be provided to infants at birth.

HIV test

In the United States, mothers have to 'opt in' for an [HIV](#) test, while in several countries the HIV test is a mandatory part of the routine antenatal screening. To reduce the risk of vertical transmission, all HIV-positive pregnant women should be prescribed **antiretroviral therapy**, should be scheduled for an **elective cesarean delivery** and should be advised against [breastfeeding](#).

PAP smear

Guidelines do not recommend PAP screening for women younger than 21 years. However, PAP screening is recommended every 3 years for all women between the ages of 21 and 29 years, and every 5 years for women after the age of 30.

During **prenatal screening** for chromosome abnormalities, genetic diseases and birth defects the healthcare provider should discuss the following:

- Prenatal screening in the first trimester, quad screening and sequential screening
- Alfa-fetoprotein levels for neural tube defects screening
- **Cystic fibrosis** screening is offered to patients with a family history of the disease or to Caucasian patients.
- Hemoglobin electrophoresis for patients of African American, Mediterranean or Asian descent to detect sickle cell trait, thalassemia, and other hemoglobinopathies.
- In women of Jewish or Eastern European descent, screening for Tay-Sachs disease is recommended.
- A tuberculin skin test should be performed if the pregnant woman has recently immigrated from a **tuberculosis**-endemic region or has a history of contact with tuberculosis patients in a prison or a mental health institution.

Urinalysis

Pregnant women should be screened for asymptomatic bacteriuria between 11th and 16th week of gestation.

History and examination

- Assess obstetric risk factors like the previous history of **miscarriages**, preterm labor, pre-**eclampsia**, **gestational diabetes**, cesarean delivery and growth retardation in the baby
- Counseling the cessation of alcohol, smoking and substance use
- Screening for domestic violence
- Screening for depression
- Discuss ideal weight gain and diet during pregnancy

Ultrasonography

- Confirm gestational age with crown-rump length measurement

Routine screening

- Blood type and Rh factor
- Complete blood count
- Infections (rubella, HIV, RPR, hepatitis B)
- Pap smear and cultures for gonorrhea and chlamydia
- Hemoglobinopathies inherited diseases

Subsequent Antenatal Visits

- Assess weight, blood pressure, urine dipstick test for glucose, albumin, and ketones
- Document fetal heart rate with auscultation or ultrasound after 10 weeks
- Document fetal movement and fundal height after 20 weeks

Non-stress test

Fetal heart rate monitoring by EFM:

→ **Reactive** (= two accelerations of greater than 15 ppm lasting at least 15 seconds over a 20 minute time period): fetal well-being assured

→ **Non-reactive**: additional testing is necessary to assure fetal well-being.

Biophysical profile

- Fetal movement
- Fetal tone
- Fetal breathing
- Amniotic fluid volume
- Non-stress test

Contraction stress test

- **Positive**: more than 50 % of contractions are associated with late decelerations.
- **Equivocal**: there are intermittent late decelerations, less than 50 %.
- **Negative**: less than 50 % of contractions are associated with late decelerations.

Umbilical artery Doppler

- Doppler ultrasound is performed when fetal growth restriction is suspected.
- As resistance in the placenta increases, the flow can be absent during diastole or is reversed.

Laboratory tests 24–28 weeks

- Assess **anemia** with hemoglobin and hematocrit levels
- Screening for **gestational diabetes** with a 50 gm 1-hour oral glucose tolerance test, followed by a 3-hour oral glucose tolerance test, if abnormal levels detected. This screening should be performed earlier in women with BMI > 40 or a previous history of gestational diabetes or **neonatal macrosomia**.
- Repeat **Rh screening** in Rh-negative mothers and administer RhoGam

Laboratory tests 32–34 weeks

- Hemoglobin/hematocrit
- Rescreen for **HIV** in high-risk patients

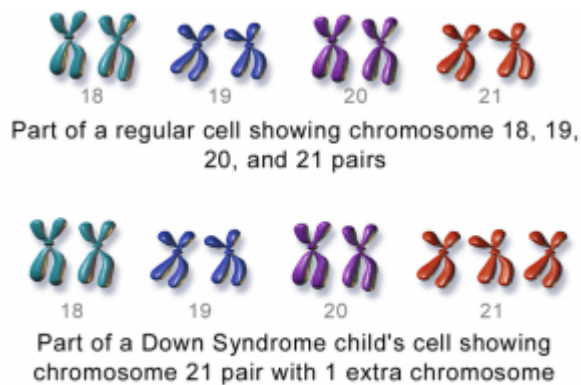
Laboratory tests 35–37 weeks

- **Group B streptococcal culture** (GBS): As 25 % of women are likely to have a GBS colonization in their genital tract, and since GBS is a cause of neonatal sepsis, GBS screening should be performed with an anovaginal culture at 35 to 37 weeks of gestation, except in women who will receive treatment due to GBS bacteriuria in this pregnancy or a history of a previous baby with GBS.

Screening for Down Syndrome (Aneuploidy)

Screening tests for aneuploidies can identify fetuses that are likely to be at risk for **Down's syndrome** or **other trisomies**. This can be followed by a diagnostic procedure to confirm the condition.

Performing **screening tests** helps to reduce the number of invasive diagnostic procedures. However, it is important to remember that screening tests do not identify all cases of aneuploidies whereas **invasive diagnostic tests** can identify trisomies as well as sex chromosomal anomalies, deletions, and chromosomal duplications and mosaicisms.



Down Syndrome

Image: "Down Syndrom," by Bruce Blaus. License: [CC BY 3.0](https://creativecommons.org/licenses/by/3.0/)

Choosing a screening test depends on the gestational age, obstetric history of the patient, the number of fetuses, availability of the test and its sensitivity, risk of invasive procedures, limitations of the test and options for termination of pregnancy, in case aneuploidy is diagnosed.

As women above the age of 35 are considered to be at a higher risk for having babies with trisomies, they are recommended to undergo **chorionic villus sampling (CVS)** with genetic counseling and **amniocentesis** as part of the screening tests.

Women under the age of 35 are recommended screening with either **triple markers** (human chorionic gonadotropin (hCG), unconjugated estriol and maternal alfa-fetoprotein levels), which have a 70 % detection rate, or **quadruple markers** (human chorionic gonadotropin (hCG), unconjugated estriol, maternal alfa-fetoprotein levels and inhibin A levels), which have an 80 % detection rate of Down's syndrome. Combined with **ultrasonography**, these screening tests provide an accurate diagnosis of the condition.

Screening tests should not be offered on the basis of the patient's age alone. The healthcare provider should offer the tests on an individual basis after considering the patient's requirements, age and past obstetric and family history.

Free fetal DNA

- Performed after 10 weeks
- Accuracy rate: 99 %

Integrated, sequential, contingency screening

- Performed between 11 weeks and 13 weeks 6 days

- Detection rate of 96 %
- Hormones: PAPP-A, hCG

Quad screen

- Performed between 15 weeks and 22 weeks 6 days
- Detection rate of 81 %
- Hormones: hCG, estriol, inhibin A, α -fetal protein

These discussed tests are meant for screening purposes. If they are abnormal, invasive testing is recommended.

The screening tests depend on the gestational age of the patient and are described in the following section.

First-trimester screening tests

Nuchal translucency measurement, PAPP-A, free or total beta-hCG

An association has been reported between risk of trisomy 21 and the amount of **fluid at the posterior aspect of the fetal neck**. This is called nuchal translucency (NT) and can be measured on ultrasonography as early as the 6th week of gestation.

NT, combined with levels of free hCG and pregnancy-associated plasma protein A (PAPP-A), are used to screen for Down's syndrome in the first trimester, and their **combined detection rate is around 80 %**.

First-trimester screening is advantageous as a woman diagnosed to have a baby with aneuploidy can be provided the information in the early gestation period and can be offered **CV** and, **genetic counseling** as well as **second-trimester amniocentesis**, if she is at high risk.

Targeted ultrasonography and **fetal echocardiography** are offered to women when fetal NT is at least 3.5mm (and aneuploidy screen is negative and no chromosomal abnormalities have been detected) as there is still a high risk of **congenital cardiac/abdominal wall defects** and other genetic syndromes.

While women of Advanced Maternal Age (AMA) are at increased risk of having an infant with Down's syndrome, all women should be offered to screen in the first trimester.

Second-trimester screening tests

Triple markers (human chorionic gonadotropin (hCG), unconjugated estriol and maternal alpha-fetoprotein levels) can detect Down's syndrome in 70 % of the cases.

Quadruple markers (human chorionic gonadotropin (hCG), unconjugated estriol, maternal alpha-fetoprotein levels, and inhibin A levels) can detect Down's syndrome in approximately 80 % of the cases.

First and second-trimester screening tests

- **Integrated (NT, PAPP-A, quadruple) screening** includes markers of the first and second trimester with a 95 % aneuploidy detection rate and has very few false positives. The screening test results are reported after both trimester tests are done and adjusted for the patient's age-related risk. This approach has its limitations, however, as the patient is unable to consider CVS in the first

trimester and has to delay a decision until the second-trimester results become available.

- **Serum Integrated (PAPP-A, quadruple)** screening has an 85 % detection rate.
- **Sequential screening** is of two types: stepwise and contingent. In **stepwise screening**, a high-risk patient can opt out of further screening and receive genetic counseling with diagnostic screening, while a low-risk patient can continue with second-trimester screening. In **contingent screening**, the patient is first classified as high, intermediate or low-risk based on the results of the first-trimester screening. Women in the high-risk category are offered CVS; women at intermediate risk are offered second-trimester screening, while no further screening is offered to low-risk women.

References

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Zolotor, A. J., & Carlough, M. C. (2014). Update on Prenatal Care. *American Family Physician Journal*, 89(3), 199-208.
[ACOG Releases Guidelines on Screening for Fetal Chromosomal Abnormalities](#) via aafp.org

[Screening for Fetal Aneuploidy](#) via healthsciences.utah.edu

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