Prenatal care: Initial assessment

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INTRODUCTION — This topic will discuss the initial prenatal assessment (which may require more than one visit) in the United States. Most of these issues are common to pregnancies worldwide. Preconception care and ongoing prenatal care after the initial prenatal visit are discussed separately. (See "The preconception office visit" and "Prenatal care: Second and third trimesters".)

GOALS — The major goal of prenatal care is to help ensure the birth of a healthy baby while minimizing risk to the mother. There are several components involved in achieving this objective:

- Early, accurate estimation of gestational age
- Identification of pregnancies at increased risk for maternal or fetal morbidity and mortality
- Ongoing evaluation of maternal and fetal health status
- Anticipation of problems with intervention, if possible, to prevent or minimize morbidity
- Health promotion, education, support, and shared decision-making

Healthy women have described the following factors as those that matter most to them for a positive pregnancy experience [1]:

- Maintaining physical and sociocultural normality
- Maintaining a healthy pregnancy (preventing and treating risks, illness, and death)
- Having an effective transition to positive labor and birth
- Achieving positive motherhood (maternal self-esteem, competence, and autonomy)

TIMING — Prenatal care should be initiated in the first trimester, ideally by 10 weeks of gestation since some prenatal screening and diagnostic tests can be performed at this gestational age. Early initiation of care is also useful to establish early baseline measurements (eg, weight, blood pressure, laboratory evaluation in women with chronic diseases) and provide early social service support and intervention, when warranted.

The percentage of pregnant women who initiate prenatal care in the first trimester is one of the standard clinical performance measures used to assess the quality of maternal health care. In the past decade, approximately 75 percent of pregnant women in the United States obtained prenatal care in the first trimester [2].

CARE PROVIDER

Standard one-on-one care — Prenatal care is generally provided by midwives, obstetrician-gynecologists, family medicine clinicians, and maternal-fetal medicine (MFM) subspecialists. Midwives and family medicine clinicians generally provide prenatal care for women with pregnancies where major complications are not anticipated. Obstetrician-gynecologists are specialists who provide prenatal care for uncomplicated and some complicated pregnancies. MFM clinicians are subspecialist obstetrician-gynecologists with expertise for managing high-risk, complicated pregnancies. (See 'Subspecialty obstetrical care' below.)

Collaboration between clinicians and midwives is common and influenced by the medical/obstetric needs and personal preferences of the pregnant woman as well as local licensing regulations. In a systematic review of trials in which pregnant women were randomly assigned to midwife-led continuity models of care or other models of care (15 trials, over 17,000 women), women in midwife-led continuity models of care were less likely to experience intrapartum intervention, more likely to be satisfied with their care, and had similar rates of adverse outcomes as women who received other models of care [3]. The trials included women at low risk of complications and women at increased risk, but not currently experiencing problems.

Group prenatal care — Group prenatal care is an alternative means of providing prenatal care in which participants with the same month of expected delivery receive the majority of their care in a group setting. The only private time between patient and clinician is during the initial prenatal assessment, when health concerns involving need for privacy arise, and during cervical assessment late in pregnancy. The majority of the visit, which may last two hours, involves facilitated group discussion, education, and skills building to address explicit learning objectives in prenatal care, childbirth preparation, and postpartum and parenting roles. Group prenatal care appears to result in at least equivalent obstetric outcomes and high levels of patient satisfaction compared with traditional prenatal care. (See "Group prenatal care".)

Subspecialty obstetrical care — MFM subspecialists are obstetrician-gynecologists with additional training in the area of high-risk, complicated pregnancies. Referral to a MFM subspecialist is appropriate for women with chronic health conditions, women who have experienced pregnancy complications in the past, and women who develop complications during their current pregnancy; however, this decision depends on the obstetrician-gynecologist's level of expertise with the specific problem.

Subspecialist care for high-risk pregnancies has not been studied extensively, except for specialty clinics that provide a prenatal care package for women at high risk of preterm birth. A 2017 systematic review that analyzed randomized trials of this issue found no significant difference in preterm birth, very preterm birth, or stillbirth between the group that received care at a clinic to prevent the onset of preterm labor and facilitate its early identification and treatment and the usual care group [4]. There were many limitations to these data, including heterogeneity in outcome focus, target populations, study design, and specific intervention components.

Multidisciplinary care — Women with medical comorbidities benefit from collaborative multidisciplinary care by a team that includes their obstetric provider and appropriate medical or surgical subspecialists, and possibly specialists in genetics, anesthesia, and pediatrics.

COMPONENTS OF THE INITIAL PRENATAL VISIT — Appropriate history, physical examination, and laboratory studies can help identify pregnant women at increased risk of medical complications, pregnancy complications, or fetal abnormalities. Early identification of these women gives the provider an opportunity to discuss these issues and their management with the patient and, in some cases, offer interventions to prevent or minimize the risk of an adverse outcome.

History

Medical/obstetrical history — At or prior to the first prenatal visit, it is efficient for the patient to complete a questionnaire detailing her psychosocial, medical, obstetrical, and family history. This information can be used to start an obstetrical record that will document her prenatal, intrapartum, and postpartum course. Several paper and computerized obstetrical record forms are available for this purpose (eg, Federal Government Prenatal Record form SF533). They help to ensure complete and systematic documentation of the pregnancy and often may be used for risk-assessment planning.

The major elements of the patient history include:

- Personal and demographic information
- Past obstetrical history
- Personal and family medical history, genetics screening
- Past surgical history
- Menstrual and gynecological history
- Current pregnancy history
- Travel to areas endemic for malaria, tuberculosis, Zika virus
- Psychosocial information

If the patient has risk factors for ectopic pregnancy (eg, history of ectopic pregnancy or pelvic infection), early identification of the location of the pregnancy is important. (See "Ectopic pregnancy: Incidence, risk factors, and pathology", section on 'Risk factors' and "Ectopic pregnancy: Clinical manifestations and diagnosis", section on 'Diagnostic evaluation'.)

Although rare, patients should be queried about a history of phenylketonuria, which is detected on newborn screening. If the patient does not volunteer the diagnosis because she is no longer on a restricted diet, asking her about dietary restrictions during childhood might reveal the diagnosis. Elevated serum phenylalanine concentration during early pregnancy in a mother with phenylketonuria or hyperphenylalaninemia can result in phenylalanine embryopathy, which can be prevented by dietary restriction of phenylalanine intake. (See "Overview of phenylketonuria".)

Psychosocial history — Psychosocial issues of potential concern that should be identified and discussed with the patient include whether the pregnancy was planned or unintended, potential barriers to care (eg, communication, transportation, child care issues, economic constraints, work schedule), whether the patient has stable housing, and her mental health and level of stress (including depression screening) [5]. This is an opportunity to provide information and, if indicated, make suggestions for possible changes or referral. (See "Unipolar major depression during pregnancy: Epidemiology, clinical features, assessment, and diagnosis".)

American College of Obstetricians and Gynecologists (ACOG) guidelines on intimate partner violence and reproductive and sexual coercion recommend that clinicians routinely assess all pregnant women for past or current exposure to these behaviors [6-8]. The clinician should be aware of markers and characteristics of abuse, such as bruising, improbable injury, depression, late prenatal care (presentation in the late second or the third trimester), missed prenatal visits, and appointments cancelled on short notice. Exposure to intimate partner violence is associated with an increased risk of low birth weight newborns and preterm birth [9]. Counseling and intervention can reduce intimate partner violence and improve pregnancy outcome [10]. (See "Intimate partner violence: Epidemiology and health consequences", section on 'Pregnancy' and

"Intimate partner violence: Diagnosis and screening" and "Intimate partner violence: Intervention and patient management".)

Women with past histories of sexual trauma may have psychological distress triggered by the normal process of labor and delivery; discussion of these issues with the patient prior to delivery and modifications in some aspects of care may alleviate some of this distress (table 1) [11]. (See "Evaluation and management of adult and adolescent sexual assault victims".)

Calculation of estimated date of delivery — Calculators are available for determining estimated date of delivery (EDD) and gestational age (calculator 1 and calculator 2) from the date of the last menstrual period. Accurate dating is crucial for managing the pregnancy, especially with regard to timing interventions and monitoring fetal growth. Sonographic estimation of the EDD before 20 weeks of gestation is desirable in all pregnancies. (See 'Ultrasound examination' below.)

Physical examination — Baseline blood pressure, weight, and height should be recorded as part of the examination. Calculating body mass index (BMI) facilitates counseling about the appropriate amount of weight gain over the course of pregnancy. Underweight and obese women are counseled about their specific risks in pregnancy. (See "Weight gain and loss in pregnancy" and "Obesity in pregnancy: Complications and maternal management".)

If the baseline blood pressure is borderline high or elevated, the clinician should attempt to find records of prepregnancy blood pressures to document whether the patient has chronic (pre-existing) hypertension. This information can be important in establishing the correct diagnosis (pre-existing hypertension versus preeclampsia) if blood pressure increases in the second half of pregnancy (see "Preeclampsia: Clinical features and diagnosis", section on 'Pre-existing hypertension versus preeclampsia'). Management of pre-existing hypertension in pregnancy is discussed separately. (See "Management of hypertension in pregnant and postpartum women".)

A complete physical examination should be performed, with special attention to uterine size and shape and evaluation of the adnexa. As discussed above, when the uterine size on physical examination differs from that predicted by menstrual dating, sonographic assessment is indicated. Causes for a discrepancy between the actual uterine size and that predicted by the last menstrual period include uterine fibroids, uterine malposition (eg, retroverted uterus), multiple gestation, and incorrect menstrual date. (See "Prenatal assessment of gestational age and estimated date of delivery".)

When fetal cardiac activity is present, the fetal heart can usually be heard by 12 weeks of gestation using a hand-held Doppler ultrasound device. Transvaginal ultrasound scanners can identify fetal cardiac motion as early as 5.5 weeks.

Ultrasound examination — Routine early (before 20 weeks of gestation) ultrasound examination is beneficial because of better estimation of gestational age than menstrual dates, resulting in significantly reduced frequency of labor induction for post-term pregnancy and use of tocolysis for suspected preterm labor. Ultrasound examination in the first trimester, if possible, is particularly important when menses are irregular, the last menstrual period is unknown or uncertain, in patients who conceive while taking oral contraceptive pills, and when the uterine size is discordant with menstrual dates. These data are reviewed separately. (See "Prenatal assessment of gestational age and estimated date of delivery".)

First-trimester ultrasound examination can lead to earlier detection of clinically unsuspected fetal malformations (including aneuploidies) and earlier detection of multiple pregnancy. This

information has not been proven to improve overall fetal outcome, although studies have lacked power to assess for secondary outcomes. (See "Routine prenatal ultrasonography as a screening tool".)

Discussion of screening and testing for genetic abnormalities in offspring — Patients undergoing any screening test should understand the difference between a screening test and diagnostic test. This is particularly important in genetic screening, where couples need to understand what is and is not being screened for, the interpretation of screen positive and negative results, the possibility of false positive and negative results, possible follow-up invasive or noninvasive testing, and possible reproductive choices.

Aneuploidy — ACOG recommends that (1) all women should be offered aneuploidy screening before 20 weeks of gestation and (2) all women should have the option of having a diagnostic invasive procedure (genetic studies on samples obtained by chorionic villus biopsy or amniocentesis) instead of screening, regardless of maternal age [12,13].

Multiple screening tests are available and fall into two major categories:

- Assessment of maternal serum levels of specific biochemical markers associated with Down syndrome (trisomy 21) and trisomy 18, with or without assessment of specific ultrasound markers, is one option (table 2). This approach may lead to detection of fetal conditions beyond the primary targets (trisomies 21 and 18) (See "Down syndrome: Overview of prenatal screening".)

- Assessment of cell-free DNA in the maternal circulation to screen for trisomy 21, trisomy 18, trisomy 13, and sex chromosome aneuploidies is the other option. (See "Prenatal screening for common aneuploidies using cell-free DNA".)

Either approach is acceptable as long as the patient receives appropriate pretest genetic counseling to make an informed choice. The cost to the patient can differ substantially depending on the approach and is another factor for the patient to consider.

Carrier screening — The ACOG position on carrier screening is that "Ethnic-specific, panethnic, and expanded carrier screening are acceptable strategies for prepregnancy and prenatal carrier screening (table 3). Each obstetrician-gynecologist or other health care provider or practice should establish a standard approach that is consistently offered to and discussed with each patient, ideally before pregnancy. After counseling, a patient may decline any or all carrier screening" [14].

In addition, "All patients who are considering pregnancy or are already pregnant, regardless of screening strategy and ethnicity, should be offered carrier screening for cystic fibrosis and spinal muscular atrophy, as well as a complete blood count and screening for thalassemias and hemoglobinopathies" [14].

Fragile X premutation carrier screening is recommended for women with a family history of intellectual disability, developmental delay, or autism; a family history of fragile X syndrome (confirmed premutation or full mutation of *FMR1* gene); known elevated levels of follicle-stimulating hormone or a family history of premature ovarian insufficiency; or a late-onset intention tremor or ataxia, especially with a family history of movement disorders.

Consanguineous couples should be offered genetic counseling to discuss the increased risk of recessive conditions in their offspring.

If the patient is found to be a carrier for a specific condition, her reproductive partner should be offered screening to determine the couple's risk of having an affected child. Referral to a genetic counselor is useful to discuss the specific disorder, residual risk, options for prenatal diagnosis, and the woman's reproductive options.

Information on specific disorders is available separately:

- Cystic fibrosis. (See "Cystic fibrosis: Carrier screening".)
- Spinal muscular atrophy. (See "Spinal muscular atrophy", section on 'Genetics'.)
- Fragile X. (See "Prenatal screening and diagnosis for fragile X syndrome", section on 'Candidates for screening'.)
- Eastern European (Ashkenazi) Jews (and Pennsylvania Dutch, Southern Louisiana Cajun, and Eastern Quebec French Canadian descent). (See "Carrier screening for genetic disease in the Ashkenazi Jewish population".)
- Hemoglobinopathies – Hemoglobinopathy screening based upon ethnic origin is not considered reliable by some authorities because of the increasingly diverse ethnic and geographic distribution of hemoglobinopathy genotypes in the United States [15]. Instead, their preferred approach is to take a comprehensive personal and family medical history, with emphasis on history of anemia and other hematological disorders. Individuals who are found to have risk factors for hemoglobinopathy should be informed of their risk and offered screening; however, screening is also offered to any individual who is concerned about possible hemoglobinopathy.

  A simple screening test that should be performed on all pregnant women as part of the baseline complete blood count (CBC) is evaluation of red cell indices: Mean corpuscular volume (MCV) less than 80 femtoliters (fL) in the absence of iron deficiency denotes patients at increased risk for alpha or beta thalassemia. A detailed description of patients and increased risk, indications for hemoglobin electrophoresis, and interpretation and management of test results can be found separately. (See "Prenatal screening and testing for hemoglobinopathy".)

  However, a CBC and MCV may not detect carriers of hemoglobin S, C, or E. Maternal hemoglobin analysis can be performed either by high-performance liquid chromatography (HPLC) or isoelectric focusing (IEF) to identify these abnormal hemoglobins. The sickle (S) hemoglobin and hemoglobin C mutations are most common in individuals with ancestries from Africa including North Africa and the Caribbean, Central and South America, Greece, Southern Italy, Turkey, Arab countries, and India [16]. Hemoglobin E is common among individuals with Southeast Asian ancestry.

Laboratory tests — In the absence of diagnostic physical findings of pregnancy (ie, an ultrasound image of the gestational sac/fetus or auscultation of the fetal heart beat by a hand-held Doppler device), suspected pregnancy should be confirmed by detection of the beta-subunit of human chorionic gonadotropin (hCG) in blood or urine. (See "Clinical manifestations and diagnosis of early pregnancy", section on 'Diagnosis'.)

Standard panel — A standard panel of laboratory tests is obtained on every pregnant woman at the first prenatal visit, augmented by additional tests in women at risk for specific conditions. (See 'Selective screening' below.). Repetition of tests performed preconceptionally is unnecessary.

We perform the following assessments, which are generally consistent with recommendations from ACOG [17]. The rationale for each test and implications of findings are also addressed.

Rhesus type and antibody screen — Rh(D)-negative women without alloantibodies should receive prophylactic anti(D)-immune globulin at 28 weeks and when clinically indicated to prevent alloimmunization. (See "Prevention of Rhesus (D) alloimmunization in pregnancy", section on 'Guidelines for use of anti-D immunoglobulin (United States)'.)

Rh(D)-positive or -negative women who have a positive antibody screen may be at risk for hemolytic disease of the fetus and newborn in offspring. Further evaluation and management of these pregnancies is reviewed separately. (See "Overview of Rhesus D alloimmunization in pregnancy" and "Management of pregnancy complicated by Rhesus (D) alloimmunization" and "Management of non-Rhesus (D) red blood cell alloantibodies during pregnancy".)

Hematocrit or hemoglobin and mean corpuscular volume — Anemia is defined by a hemoglobin level <11 g/dL (hematocrit <33 percent) in the first trimester, and is commonly related to iron deficiency. (See "Hematologic changes in pregnancy", section on 'Anemia'.)

An MCV <80 fL in the absence of iron deficiency suggests thalassemia; further testing with hemoglobin electrophoresis is indicated. (See "Prenatal screening and testing for hemoglobinopathy".)

Documentation of rubella immunity — Serologic screening for rubella immunity is performed unless the woman is known to be immune by previous serologic testing [18]. Once documentation of immunity to rubella as a result of infection or immunization has been obtained, repeat testing is unnecessary. If nonimmune, she should be counseled to avoid exposure to individuals with rubella and receive postpartum immunization. The rubella vaccine is a live vaccine and thus contraindicated during pregnancy. (See "Immunizations during pregnancy", section on 'Measles, mumps, rubella'.)

Documentation of varicella immunity — Immunity to varicella is based on a health care provider's diagnosis of varicella or verification of history of varicella disease, documented vaccination, or laboratory evidence of immunity. Women who do not have evidence of immunity to varicella should be counseled to avoid exposure to individuals with varicella, may be candidates for passive immunization during pregnancy if exposed to varicella, and are candidates for varicella vaccination postpartum [19]. The varicella vaccine is a live vaccine and thus contraindicated during pregnancy. (See "Immunizations during pregnancy", section on 'Varicella'.)

Urine protein — Screening for proteinuria, such as with a dipstick, is useful as a baseline for comparison with testing performed later in pregnancy. (See "Proteinuria in pregnancy: Evaluation and management".)

Urine culture — Routine urine culture is recommended because pregnant women with untreated asymptomatic bacteriuria are at high risk of developing pyelonephritis, modest risk for preterm birth, and rapid tests for bacteriuria do not have adequate sensitivity and specificity. (See "Urinary tract infections and asymptomatic bacteriuria in pregnancy", section on 'Diagnosis'.)

Treatment of a positive culture is per standard guidelines; however, some clinicians treat group B streptococcal (GBS) bacteriuria regardless of colony count and presence of GBS is an indication for GBS prophylaxis in labor and delivery to prevent early-onset neonatal infection. (See "Urinary tract infections and asymptomatic bacteriuria in pregnancy", section on 'Management' and "Group B streptococcal infection in pregnant women", section on 'Asymptomatic bacteriuria'.)

The optimum frequency of retesting is unclear. We retest women with asymptomatic bacteriuria monthly until delivery and give suppressive therapy for the remainder of pregnancy if they have recurrent or persistent bacteriuria. (See "Urinary tract infections and asymptomatic bacteriuria in pregnancy", section on 'Management of persistent bacteriuria'.)

Cervical cancer screening — The frequency of cervical cancer screening is not influenced by pregnancy, but management of an abnormal test is different for pregnant women. (See "Screening for cervical cancer" and "Cervical intraepithelial neoplasia: Management of low-grade and high-grade lesions", section on 'Pregnant women' and "Cervical cancer in pregnancy".)

Human immunodeficiency virus — ACOG supports universal human immunodeficiency virus (HIV) testing of pregnant women early in each pregnancy using an "opt-out" approach [20]. Other national organizations have also taken this approach [21-23]. Advantages of universal testing include:

- An informed decision can be made about continuing the pregnancy
- Appropriate maternal medical management can be initiated
- Women can be counseled about prevention of transmission to or identification of infected partners
- Perinatal transmission can be substantially reduced with appropriate intervention (eg, antiretroviral therapy antepartum and intrapartum, cesarean delivery, avoidance of breastfeeding) (see "Prenatal evaluation of the HIV-infected woman in resource-rich settings" and "Antiretroviral and intrapartum management of pregnant HIV-infected women and their infants in resource-rich settings")

In some Canadian provinces, an opt-out approach achieved high rates of testing, 95 to 100 percent, whereas an "opt-in" policy had testing rates of only 50 to 60 percent due to patient refusal and clinician failure to offer the test [24]. Similar success with the opt-out approach has been described in the United States [25].

Local law may require patient notification as well as a signed consent form indicating permission for HIV testing. The medical record should document the patient's decision to accept or decline testing. Reasons for refusal should be explored and testing offered at another time.

Risk factors for acquiring HIV and serologic testing are discussed in detail separately. (See "Screening for sexually transmitted infections".)

Syphilis — Serologic testing to diagnose syphilis should include the use of both nontreponemal and treponemal tests. Either test can be used as the initial screening test depending on the preference of the laboratory doing the test. Confirmatory testing is necessary due to the potential for a false-positive screening test result. The diagnostic approach is the same as in nonpregnant individuals. The cost and morbidity associated with testing for syphilis are low and the benefits of detecting and treating the disease during pregnancy are high for both mother and child [21]. (See "Syphilis in pregnancy".)

Hepatitis B virus — All pregnant women are tested for the hepatitis B surface antigen (HBsAg), regardless of previous vaccination status, because prevaccination screening to exclude acute or chronic hepatitis B virus (HBV) infection is not commonly performed. Women who carry the HBsAg can transmit HBV to the fetus, typically during delivery. Passive and active

immunization of the newborn within 12 hours of delivery can reduce the risk of HBV transmission by more than 95 percent. Management of screen-positive women is shown in the algorithm (algorithm 1) and discussed in detail separately. (See "Epidemiology, transmission, and prevention of hepatitis B virus infection", section on 'Mother-to-child transmission' and "Hepatitis B and pregnancy".)

Women who are HBsAg-negative and are at high risk for HBV infection (eg, injection drug user, sexual partner or household contact has chronic HBV) should be tested for hepatitis B surface antibody (anti-HBs) and hepatitis B core antibody (anti-HBc). Mothers without evidence of prior HBV infection or exposure (negative for anti-HBs and anti-HBc) should be vaccinated.

Chlamydia — Chlamydia prevalence is highly related to age and sexual behavior. We screen all pregnant women for chlamydia, consistent with guidelines from ACOG [17]. Other major organizations in the United States have taken a more selective approach based on young age and presence of risk factors. The Centers for Disease Control and Prevention (CDC) and US Preventive Services Task Force (USPSTF) recommend screening all pregnant women <25 years of age and those pregnant women ≥age 25 with risk factors for sexually transmitted infection (table 4) [21,26].

Nucleic acid amplification tests (NAAT) have high sensitivity and excellent specificity for detection of *Chlamydia trachomatis*, and are superior to culture. During prenatal care, the preferred approach is to test a specimen obtained from a swab of the endocervix or vagina, although urine testing appears to be as sensitive [21,27-31]. Some NAATs have been cleared by the US Food and Drug Administration for use on liquid-based cytology specimens.

Women with positive test results should be treated. In pregnancy, women with a positive test then undergo a test-of-cure three to four weeks after treatment and are retested three to four months later [21]. (See "Clinical manifestations and diagnosis of Chlamydia trachomatis infections", section on 'Nucleic acid amplification'.)

Selective screening

Thyroid function — Both hyper- and hypothyroidism during pregnancy can have adverse effects on the mother and child. (See "Hyperthyroidism during pregnancy: Clinical manifestations, diagnosis, and causes", section on 'Pregnancy complications' and "Hypothyroidism during pregnancy: Clinical manifestations, diagnosis, and treatment", section on 'Pregnancy complications'.)

Women with signs or symptoms of thyroid disease should have their thyroid-stimulating hormone (TSH) level measured. The TSH should be interpreted using population and trimester-specific TSH reference ranges for pregnant women, when available. If the TSH is abnormal, free or total T4 should be measured.

- The diagnosis of overt hyperthyroidism during pregnancy is based primarily upon a suppressed (<0.1 milli-units/L) or undetectable (<0.01 milli-units/L) serum TSH value and a free T4 and/or free T3 (or total T4 and/or total T3) measurement that exceeds the normal range for pregnancy (see "Hyperthyroidism during pregnancy: Clinical manifestations, diagnosis, and causes", section on 'Diagnosis').

- The diagnosis of overt primary hypothyroidism during pregnancy is based upon a TSH above the population and trimester-specific upper limit of normal, or above 4.0 mU/L when local reference ranges are not available, in conjunction with a decreased free T4 concentration

(below assay normal using reference range for pregnant women). (See "Hypothyroidism during pregnancy: Clinical manifestations, diagnosis, and treatment", section on 'Diagnosis'.)

- The diagnosis of subclinical hypothyroidism during pregnancy is based upon an elevated trimester-specific serum TSH concentration and a normal free T4 concentration.

  Screening for asymptomatic hypothyroidism is controversial. Professional societies (eg, ACOG [32], the Endocrine Society [33], the American Thyroid Association [34]) recommend targeted rather than universal screening in pregnant women, which is our approach. However, a targeted approach will miss as many as one-third of women with subclinical hypothyroidism [35-38]. For this reason and concern that not treating these women may be associated with adverse pregnancy outcomes, some authorities have advocated universal screening for thyroid dysfunction in pregnant women or those attempting to become pregnant [39]. Criteria for selecting candidates for targeted TSH screening, diagnostic evaluation (free T4, TPO antibodies) of women with an elevated trimester-specific serum TSH concentration, and decision-making regarding treatment of these women are reviewed separately. (See "Hypothyroidism during pregnancy: Clinical manifestations, diagnosis, and treatment", section on 'Screening' and "Hypothyroidism during pregnancy: Clinical manifestations, diagnosis, and treatment", section on 'Effect of thyroid hormone replacement' and "Hypothyroidism during pregnancy: Clinical manifestations, diagnosis, and treatment", section on 'Indications for treatment'.)

Type 2 diabetes — Both the American Diabetes Association (ADA) [40] and ACOG suggest early pregnancy testing for undiagnosed type 2 diabetes in women with risk factors [41]. In contrast, a 2014 USPSTF guideline concluded available evidence was insufficient to assess the balance of benefits and harms of screening asymptomatic pregnant women for glucose intolerance before 24 weeks of gestation [42]. (See "Diabetes mellitus in pregnancy: Screening and diagnosis", section on 'Identification of overt diabetes in early pregnancy'.)

The ADA defines women at increased risk of overt diabetes based on body mass index (BMI) ≥25 kg/m^2 (≥23 kg/m^2 in Asian Americans) plus one or more of the following [40]:

- Gestational diabetes mellitus in a previous pregnancy
- A1C ≥5.7 percent (39 mmol/mol), impaired glucose tolerance, or impaired fasting glucose on previous testing
- First-degree relative with diabetes
- High-risk race/ethnicity (eg, African American, Latino, Native American, Asian American, Pacific Islander)
- History of cardiovascular disease
- Hypertension (≥140/90 mmHg or on therapy for hypertension)
- HDL cholesterol level <35 mg/dL (0.90 mmol/L) and/or a triglyceride level >250 mg/dL (2.82 mmol/L)
- Polycystic ovary syndrome
- Physical inactivity
- Other clinical condition associated with insulin resistance (eg, severe obesity, acanthosis nigricans)

For women at increased risk of diabetes, a diagnosis of overt diabetes can be made at the initial prenatal visit if:

- Fasting plasma glucose ≥126 mg/dL (7.0 mmol/L), or
- A1C ≥6.5 percent using a standardized assay, or
- Random plasma glucose ≥200 mg/dL (11.1 mmol/L) that is subsequently confirmed by elevated fasting plasma glucose or A1C

Infection — Symptomatic women should be tested for the suspected infection. The following discussion applies to selection of asymptomatic women for screening.

Gonorrhea — Pregnant women with risk factors for gonorrhea or living in an area where the prevalence of *Neisseria gonorrhoeae* is high are screened for gonorrhea, in agreement with ACOG and CDC guidelines [17,21]. Risk factors for sexually transmitted infection are listed in the table (table 4). Clinicians should consult local public health authorities for information on disease prevalence in their specific population.

NAAT is the preferred test for the microbiologic diagnosis of *N. gonorrhoeae* because of its superior accuracy; a swab is used to collect a vaginal or endocervical specimen for testing.

Pregnant women who test positive are treated immediately and retested in three months. Diagnosis and management of gonococcal infections are discussed in detail separately. (See "Clinical manifestations and diagnosis of Neisseria gonorrhoeae infection in adults and adolescents" and "Treatment of uncomplicated gonococcal infections".)

Hepatitis C — Pregnant women at high risk for hepatitis C infection are screened for hepatitis C antibodies [21]. Several organizations have provided guidelines that describe criteria for considering an individual high risk. Despite having reviewed similar data, the various guidelines do not agree, except that pregnancy alone is not an indication for testing. A summary of recommendations from the major organizations can be found separately. (See "Screening for chronic hepatitis C virus infection", section on 'Patient selection' and "Screening for chronic hepatitis C virus infection", section on 'Screening method'.)

Tuberculosis — Asymptomatic women are screened for latent tuberculosis (TB) infection (LTBI) during pregnancy when the woman has one of the following significant risk factors for progression to active disease during pregnancy that would justify prompt treatment for LTBI [43]:

- Suspicion for recent TB infection based on epidemiologic exposure (see "Diagnosis of latent tuberculosis infection (tuberculosis screening) in HIV-uninfected adults", section on 'Indications for testing')
- Significant immunocompromise, such as HIV infection or profound immunosuppressive therapy

Tools for diagnosis of latent tuberculosis include tuberculin skin tests (TST) and interferon-gamma release assays (IGRAs). An IGRA is preferred for patients with history of Bacillus Calmette-Guerin vaccination, and for individuals from groups that historically have poor rates of return for skin test reading. The procedure for and interpretation of these tests and management of women with positive test results are described separately. (See "Diagnosis of latent tuberculosis infection (tuberculosis screening) in HIV-uninfected adults", section on 'Tools for LTBI testing' and "Tuberculosis in pregnancy", section on 'Treatment'.)

Toxoplasmosis — Whether all pregnant women should undergo serological screening for toxoplasmosis is controversial. It is a routine practice in some areas of relatively high

prevalence, such as France. The most common means of acquisition of toxoplasmosis are via environmental exposure (maternal ingestion of oocysts from consumption of contaminated soil or water or cat litter, which may contaminate fruit or vegetables and other unwashed handheld foods) and ingestion of undercooked or cured meat from infected animals. (See "Toxoplasmosis and pregnancy".)

Bacterial vaginosis — Whether women with a history of prior preterm birth should be screened for bacterial vaginosis and treated, if positive, to lower the risk of recurrent preterm birth is controversial.

Trichomonas vaginalis — Although screening for *Trichomonas vaginalis* is not recommended as a routine component of prenatal care for HIV-negative women, women with HIV infection should be screened at the first prenatal visit and treated with metronidazole if infected [21]. Trichomoniasis in HIV-infected individuals is associated with an increased risk of vertical and horizontal transmission of HIV. Treated women should be retested three months after treatment. (See "Trichomoniasis".)

Herpes simplex virus — Type-specific screening may be reasonable in asymptomatic partners of symptomatic men [44], but this is controversial. Serologic screening has been proposed to (1) identify women without herpes simplex virus (HSV) so they can take precautions to avoid acquiring an HSV infection and (2) identify women with a past history of HSV so they can be offered suppressive antiviral therapy, examined carefully for lesions at the onset of labor, and offered cesarean delivery, if indicated. Although accurate type-specific serologic tests are available to identify these women [45-47] and guide counseling, expert panels recommend against universal screening [21,47-49]. Available evidence indicates that screening for HSV would not meet usual criteria for an effective preventive strategy [50-52], as has been demonstrated in other infections, such as HIV and hepatitis B virus [53]. (See "Genital herpes simplex virus infection and pregnancy", section on 'Screening pregnant women with no HSV history'.)[44,54]

Cytomegalovirus — ACOG [55] and the Society for Maternal-Fetal Medicine [56] recommend against routine serological screening for cytomegalovirus (CMV). Proponents of universal screening argue that knowing that her serology is negative for CMV antibodies and CMV counseling increase some women's motivation to practice good hygiene and thus decrease the risk of seroconversion during pregnancy. (See "Cytomegalovirus infection in pregnancy", section on 'Screening'.)

Testing pregnant women for CMV is indicated as part of the diagnostic evaluation of mononucleosis-like illnesses (see "Infectious mononucleosis in adults and adolescents", section on 'Clinical manifestations'), when a fetal anomaly suggestive of congenital CMV infection is detected on prenatal ultrasound examination (see "Cytomegalovirus infection in pregnancy", section on 'Prenatal (fetal) diagnosis'), or if the woman requests the test.

Zika — In areas with no mosquito-borne Zika virus transmission, health care providers should ask all pregnant women about possible exposure: residence in or travel to an area where mosquito-borne transmission of Zika virus infection has been reported or unprotected sexual contact with a person who meets these criteria. Pregnant women should be tested within 12 weeks of possible exposure, regardless of symptoms. Issues related to diagnostic evaluation of pregnant women with Zika virus exposure are reviewed in detail separately. (See "Zika virus infection: Evaluation and management of pregnant women".)

Chagas disease — Chagas disease is endemic to Latin America; thus, the possibility of this infection should be considered in pregnant women who have lived in this region [57,58]. Infected individuals are often unaware of their infection and the potential seriousness of the condition (eg, cardiovascular/digestive complications, transmission to the fetus, hydrops fetalis). Prenatal screening is performed in some countries [59]. (See "Chagas heart disease: Clinical manifestations and diagnosis" and "Chagas disease: Epidemiology and control", section on 'Vertical transmission' and "Chagas disease: Natural history and diagnosis", section on 'Congenital Chagas disease'.)

Lead level — Selective screening is indicated if the clinician has reason to suspect that the woman has any of the characteristics in the table (table 5), which increase the likelihood of lead exposure and increased blood lead levels [60,61]. If the blood lead level is <5 mcg/dL, no follow-up testing is needed. Otherwise, follow-up testing depends on the initial level (table 6) [60]. At delivery, the pediatric provider should be informed of the mother's blood lead level (see "Childhood lead poisoning: Exposure and prevention", section on 'Prenatal exposure'). The management of pregnant women with elevated blood lead levels is discussed separately. (See "Adult occupational lead poisoning", section on 'Pregnancy and breastfeeding'.)

Slight elevations of blood lead levels in pregnant women are of concern because of the potential for adverse effects on the mother and fetus (spontaneous abortion, gestational hypertension, low birthweight, impaired neurodevelopment). Major organizations in the United States do not recommend universal lead level screening in pregnancy because the prevalence of blood lead levels over 5 mcg/dL in pregnant women is less than 1 percent [60-62].

HEALTH EDUCATION AND HEALTH PROMOTION — (See "Prenatal care: Patient education, health promotion, and safety of commonly used drugs".)

SPECIAL POPULATIONS

Adolescents — (See "Pregnancy in adolescents".)

Advanced maternal age — (See "Effect of advanced age on fertility and pregnancy in women" and "Management of pregnancy in women of advanced age".)

Incarcerated women — (See "Prenatal care for incarcerated women".)

Disabled women — The American College of Obstetricians and Gynecologists provides resources for obstetricians serving women with disabilities [63]. General issues regarding care of adults with disabilities are discussed separately. (See "Primary care of the adult with intellectual and developmental disabilities" and "Disability assessment and determination in the United States".)

Grand multiparity — (See "Grand multiparity".)

Obese women — (See "Obesity in pregnancy: Complications and maternal management".)

Women with chronic medical diseases — Refer to individual topic reviews on the specific disorders.

GUIDELINES — The American College of Obstetricians and Gynecologists and others have published guidelines for prenatal care in the United States [64-66]. The World Health Organization and national organizations in other countries have also published guidelines for routine prenatal care [67-69]. Some of the major differences among guidelines are whether they take a risk-factor-

based approach to screening for some disorders (eg, chlamydia, diabetes) versus universal screening, and the available resources in the country.

INFORMATION FOR PATIENTS — UpToDate offers two types of patient education materials, "The Basics" and "Beyond the Basics." The Basics patient education pieces are written in plain language, at the 5th to 6th grade reading level, and they answer the four or five key questions a patient might have about a given condition. These articles are best for patients who want a general overview and who prefer short, easy-to-read materials. Beyond the Basics patient education pieces are longer, more sophisticated, and more detailed. These articles are written at the 10th to 12th grade reading level and are best for patients who want in-depth information and are comfortable with some medical jargon.

Here are the patient education articles that are relevant to this topic. We encourage you to print or e-mail these topics to your patients. (You can also locate patient education articles on a variety of subjects by searching on "patient info" and the keyword(s) of interest.)

- Basics topics (see "Patient education: Prenatal care (The Basics)" and "Patient education: Activity during pregnancy (The Basics)")

- Beyond the Basics topics (see "Patient education: Avoiding infections in pregnancy (Beyond the Basics)" and "Patient education: Should I have a screening test for Down syndrome during pregnancy? (Beyond the Basics)" and "Patient education: Group B streptococcus and pregnancy (Beyond the Basics)")

SUMMARY AND RECOMMENDATIONS

- Prenatal care should be initiated in the first trimester, ideally by 10 weeks of gestation since some prenatal screening and diagnostic tests can be performed at this gestational age. (See 'Timing' above.)

- Prenatal care is generally provided to individual women by midwives, obstetrician-gynecologists, or family medicine clinicians. Group prenatal care is an appealing alternative for some women. Referral to a Maternal-Fetal Medicine specialist is appropriate for women with chronic health conditions, women who have experienced pregnancy complications in the past, and women who develop complications during their current pregnancy. (See 'Care provider' above.)

- One goal of prenatal care is identification of women at increased risk of medical complications, pregnancy complications, or fetal abnormalities. This is achieved, in part, by taking a comprehensive medical, obstetrical, psychosocial, and family history; establishing an accurate estimated date of delivery (calculator 1 and calculator 2); and appropriate laboratory testing. Forms can be helpful for this purpose (eg, Federal Government Prenatal Record form SF533). (See 'Components of the initial prenatal visit' above.)

- If the woman has risk factors for ectopic pregnancy (eg, history of ectopic pregnancy or pelvic infection), early identification of the location of the pregnancy is important. (See "Ectopic pregnancy: Incidence, risk factors, and pathology", section on 'Risk factors' and "Ectopic pregnancy: Clinical manifestations and diagnosis", section on 'Diagnostic evaluation'.)

- Routine early (before 20 weeks of gestation) ultrasound examination provides better estimation of gestational age than menstrual dates, resulting in significantly reduced frequency of labor induction for postterm pregnancy and use of tocolysis for suspected

preterm labor. Early ultrasound examination can lead to earlier detection of clinically unsuspected fetal malformations and multiple pregnancy. (See "Prenatal assessment of gestational age and estimated date of delivery".)

- The American College of Obstetricians and Gynecologists (ACOG) recommends offering all women aneuploidy screening before 20 weeks of gestation and giving all women the option of having a diagnostic invasive procedure (genetic studies on samples obtained by chorionic villus biopsy or amniocentesis) instead of screening, regardless of maternal age. Screening tests fall into two categories: (1) assessment of maternal serum levels of specific biochemical markers associated with Down syndrome, with or without assessment of specific ultrasound markers and (2) assessment of cell-free DNA in the maternal circulation. Either approach is acceptable as long as the patient receives appropriate pretest genetic counseling to make an informed choice. The cost to the patient can differ substantially depending on the approach and is another factor for the patient to consider. (See 'Aneuploidy' above.)

- ACOG considers ethnic-specific, panethnic, and expanded carrier screening acceptable strategies for offering prenatal carrier screening. For all women, they recommend offering carrier screening for cystic fibrosis and spinal muscular atrophy, and a complete blood count (CBC) for screening for thalassemias and hemoglobinopathies.

  A mean corpuscular volume (MCV) less than 80 femtoliters (fL) in the absence of iron deficiency denotes patients at increased risk for alpha or beta thalassemia. A CBC and MCV may not detect carriers of hemoglobin S, C, or E; maternal hemoglobin analysis by either high-performance liquid chromatography (HPLC) or isoelectric focusing (IEF) is recommended to identify these abnormal hemoglobins. Hemoglobinopathy screening based upon ethnic origin is not considered reliable by some authorities because of the increasingly diverse ethnic and geographic distribution of hemoglobinopathy genotypes in the United States. (See 'Carrier screening' above.)

  Fragile X premutation carrier screening is recommended for women with personal or family risk factors for the disease. Consanguineous couples should be offered genetic counseling to discuss the increased risk of recessive conditions in their offspring.

- The following tests are performed on all pregnant women (see 'Laboratory tests' above):

  - Rhesus type and red cell antibody screen
  - Hematocrit/hemoglobin and mean corpuscular volume
  - Documentation of immunity to rubella and varicella
  - Qualitative assessment of urine protein
  - Assessment for asymptomatic bacteriuria. We suggest urine culture.
  - Cervical cancer screening according to standard guidelines
  - Testing for syphilis, hepatitis B antigen, and chlamydia
  - Opt-out approach to human immunodeficiency virus testing

- Women at increased risk of hypothyroidism or unrecognized type 2 diabetes mellitus are screened for these disorders. (See 'Thyroid function' above and 'Type 2 diabetes' above.)

- Women at increased risk of specific infectious diseases are screened for these disorders. (See 'Infection' above.)

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Topic 446 Version 125.0

GRAPHICS

Strategies for responding to disclosure of abuse or assault in pregnant women

Obstetrically focused response to disclosure of past sexual abuse or assault

1. Offer mental health referral.
2. Create obstetric care plan (trigger avoidance and preparation for unavoidable stressors at delivery).

Preparation for delivery stressors

Generate plan to address intrapartum triggers

- Involve multidisciplinary team for birth planning and counseling early in pregnancy (including but not limited to trauma-trained nurse educators, certified nurse midwives, psychologists, social workers, and clergy when appropriate).
- If team not available, consider nurse educator trained in trauma-informed birth planning to assist the woman in identifying triggers and generating a feasible birth plan.
- Final coordination and collaboration between the obstetrician, the woman, and the trauma-trained multidisciplinary team or educator in preparation for delivery.

A few common triggers and possible modifications

Triggers identified antepartum	Intrapartum modifications	Postpartum modifications
▪ Undressing, genital exposure.	▪ Drape adequately during examinations and at delivery. ▪ Limit spectator cheering/sensitive photos at delivery. ▪ Avoid shining bright light directly at perineum.	▪ Cover breasts adequately when examining or assisting with breast feeding.
▪ Feeling of fluid escaping from vagina, loss of bowel/bladder control.	▪ Attend to keeping perineum clean during labor and at delivery.	▪ Encourage staff to warn a woman before touching her and explain why they are doing so. ▪ Pay close attention to perineal care.
▪ Intrusive touch.	▪ Always ask before touching. ▪ Limit vaginal examinations and caregivers performing internal examination when possible. ▪ Avoid rushing. ▪ Encourage woman to bring calming music and/or support persons.	▪ Avoid pressure to breastfeed if triggers unwanted memories.
▪ Feeling powerless, limited mobility.	▪ Avoid overpowering words/behaviors (eg, command to relax). ▪ Avoid leaning over patient for vaginal examinations.	▪ Encourage postpartum social work or multidisciplinary team involvement. ▪ Consider home visitation program. ▪ Ask about stressors at postpartum visit.

	▪ Consider intermittent monitoring to allow greater mobility. ▪ Discuss epidural timing with emphasis on the loss of mobility and improved pain control.	

Special issues to consider

Timing of abuse disclosure

- Disclosure of past abuse may occur at any time, often later in pregnancy when trust is established. Allow the woman to control the timing of her own disclosure.
- Consider repeating abuse inquiry later in pregnancy if anxiety behaviors or comments lead you to suspect an abuse history, but respect her boundaries if she denies such a history.

Patient privacy, perception of safety, and control

- Seek permission before sharing the woman's past abuse history with the obstetrical care team, other providers, and especially family members who may be unaware of this history.
- Avoid unrealistic promises (eg, "Don't worry, you'll be fine"), which can undermine trust.
- Emphasize realistic goals (eg, "We'll try to follow the delivery plan as much as possible, and if we have to change the plan because of concerns for you or the baby, we will discuss it with you").

Suspected ongoing abuse

- Goal of safety planning takes precedence.
- Offer crisis hotline number, safety card, or educational material (National Domestic Violence Hotline 1-800-799-SAFE).
- Offer community resources (shelters, law enforcement contacts, mental health services, referral to social worker or multidisciplinary team if available to assist with safety planning).

Special safety issues when screening adolescents

- Disclosure of childhood abuse for adolescent patients may suggest ongoing abuse. If ongoing abuse is suspected, state-specific mandatory reporting requirements apply.
- An immediate and coordinated response (including law enforcement involvement) is needed to assist the young woman in securing a safe environment.

Reimbursement and office time concerns

- Consider using CPT codes for extended counseling.
- Involve nurse educator or experienced certified nurse midwives if appropriately trained to address trauma-related issues. This may alleviate office time concerns for birth planning and counseling.

Coordinating care with labor and delivery staff and postpartum staff

- Obtain the patient's permission first before discussing her unique birth plan with labor and delivery staff and explain the reasons for sharing the plan.
- The delivery team will need to be aware of specific triggers and planned modifications upfront.
- Postpartum nurses will also need to be aware of specific triggers and planned modifications.

Obstetrical emergencies

- Discuss the possibility of obstetrical emergencies during birth planning.
- Encourage the presence of support persons during labor, especially a trained labor coach or doula to facilitate adaptive stress coping.
- When unplanned obstetrical interventions are necessary to expedite delivery, discuss the labor course with the woman after delivery and encourage her to describe her feelings about the birth experience.
- Consider referral for postpartum birth counseling for abuse survivors who describe an unsettling birth experience or desire such counseling.

While the strategies presented in the table are for pregnant women with a history of prior sexual abuse, the information regarding disclosure, privacy, and safety can be applied to any adult patient.

CPT: current procedural terminology.

From: White A. Responding to prenatal disclosure of past sexual abuse. Obstet Gynecol 2014; 123:1344. DOI: 10.1097/AOG.0000000000000266.

Graphic 111021 Version 1.0

Maternal serum marker pattern in selected fetal syndromes

	Second trimester markers				First trimester markers		
Genetic disorder	**AFP**	**uE3**	**hCG**	**Inh A**	**PAPP-A**	**beta hCG**	**Nuchal translucency**
Down syndrome	↓	↓	↑	↑	↓	↑	↑↑
Trisomy 18	↓	↓↓	↓↓	↔	↓↓	↓↓	↑↑
Trisomy 13	↔	↔	↔	↔	↓↓	↓	↑
Turner syndrome with hydrops	↓	↓	↑	↑	↓↑	↓↑	↑
Turner syndrome without hydrops	↓	↓	↓	↓	↓↑	↓↑	↑
Triploidy (paternal)	↔	↓	↑	↑	↓↑	↑↑	↑
Triploidy (maternal)	↔	↓	↓	↓	↓↑	↓↓	↑
Smith-Lemli-Opitz syndrome	↓	↓↓	↓	NR	NR	NR	NR

Second trimester markers: AFP (alpha-fetoprotein); uE3 (unconjugated estriol); hCG (human chorionic gonadotropin); inh A (inhibin A).
First trimester marker: PAPP-A (pregnancy-associate plasma protein A); beta hCG (beta human chorionic gonadotropin); nuchal translucency.

↑: increased; ↓: decreased; ↔: unchanged; ↓↑: variable; NR: not reported.

Graphic 71552 Version 5.0

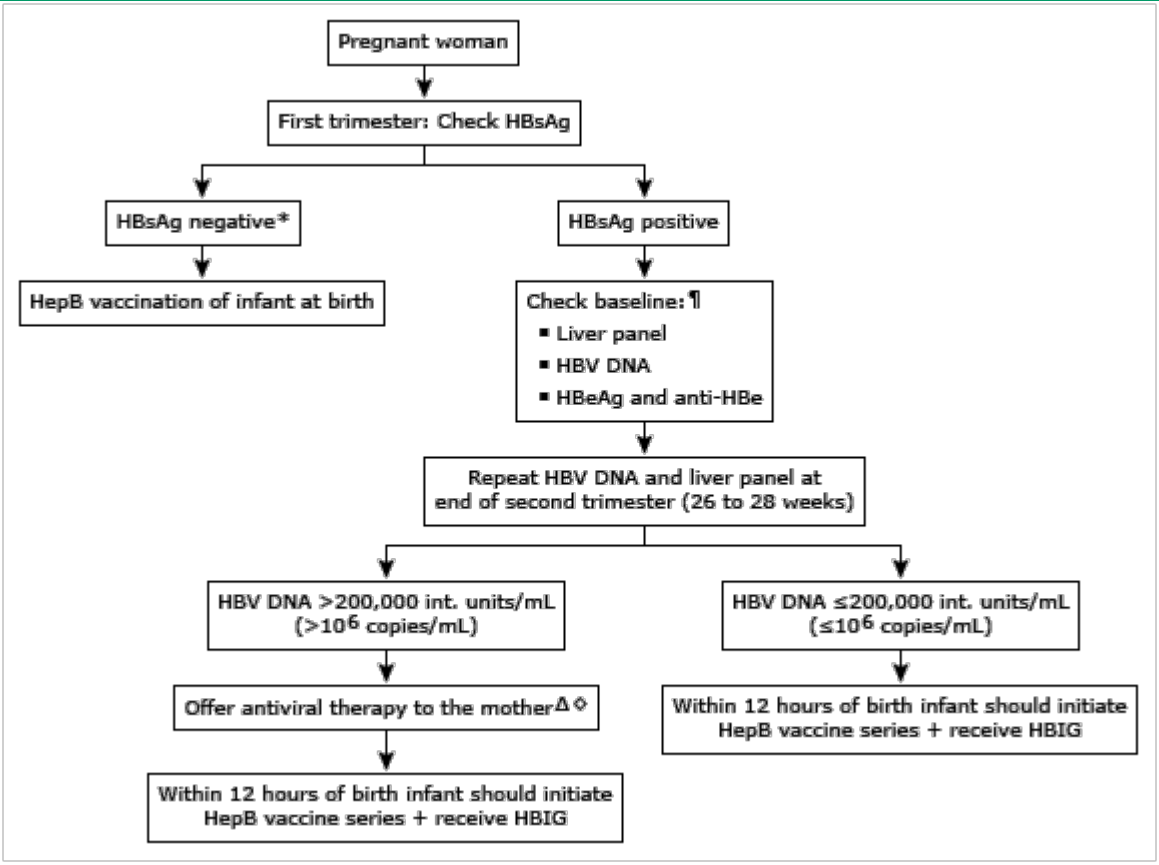
Example of an expanded carrier screening panel*

Condition	Carrier frequency in general population	Carrier frequency in specific ethnic groups
alpha-thalassemia	Unknown	African (particularly sub-Saharan): 1 in 3. Mediterranean: 1 in 30. Southeast Asian and Middle Eastern: 1 in 20.
beta-thalassemia	Unknown	African American: <1 in 8. Ashkenazi Jewish: Varied. Asian: 1 in 20. Mediterranean: 1 in 7.
Bloom syndrome	<1 in 500	Ashkenazi Jewish: 1 in 100.
Canavan disease	<1 in 150	Ashkenazi Jewish: 1 in 41.
Cystic fibrosis	Unknown	African American: 1 in 61. Asian: 1 in 94. Ashkenazi Jewish: 1 in 24. Caucasian: 1 in 25. Hispanic: 1 in 58.
Familial dysautonomia	<1 in 500	Ashkenazi Jewish: 1 in 31.
Familial hyperinsulinism	<1 in 150	Ashkenazi Jewish: 1 in 52.
Fanconi anemia C	<1 in 790	Ashkenazi Jewish: 1 in 89.
Fragile X syndrome¶	1 in 259	
Galactosemia	1 in 87	Ashkenazi Jewish: 1 in 127.
Gaucher disease	<1 in 100	Ashkenazi Jewish: 1 in 15.
Glycogen storage disease type 1A	<1 in 150	Ashkenazi Jewish: 1 in 71.
Joubert syndrome	<1 in 500	Ashkenazi Jewish: 1 in 92.
Medium-chain acyl-CoA dehydrogenase deficiency	Unknown	Caucasian: 1 in 50.
Maple syrup urine disease types 1A and 1B	<1 in 240	Ashkenazi Jewish: 1 in 81 (type 1B). Mennonite: 1 in 10 (type 1A-BCKDHA p.Y438N).
Mucolipidosis IV	<1 in 500	Ashkenazi Jewish: 1 in 96.
Niemann-Pick disease type A	<1 in 500	Ashkenazi Jewish: 1 in 90.
Phenylketonuria	Unknown	Caucasian: 1 in 50. Irish: 1 in 34.
Sickle cell anemia	Unknown	African American: 1 in 10.
Smith-Lemli-Opitz syndrome	Unknown	Caucasian: 1 in 70.
Spinal muscular atrophy	Unknown	African American: 1 in 66. Asian: 1 in 53. Ashkenazi Jewish: 1 in 41. Caucasian: 1 in 35. Hispanic: 1 in 117.
Tay-Sachs disease$^{\Delta}$	<1 in 300	Ashkenazi Jewish: 1 in 30. French Canadian and Cajun: 1 in 30.

* Conditions reasonable for inclusion in an expanded carrier screening panel as of 2016. After counseling regarding screening options, obstetrician-gynecologists or other health care providers and patients may elect to screen for fewer or more conditions than those listed here. The availability of expanded carrier screening does not preclude the appropriateness of ethnic-based screening or screening based on family history. Conditions were selected on the basis of the benefits of detection, the accuracy of current screening methods, and the following consensus-determined criteria: Have a carrier frequency of 1 in 100 or greater, have a well-defined phenotype, have a detrimental effect on quality of life, cause cognitive or physical impairment, require surgical or medical intervention, or have an onset early in life. Additionally, screened conditions should be able to be diagnosed prenatally and may afford opportunities for antenatal intervention to improve perinatal outcomes, changes to delivery management to optimize newborn and infant outcomes, and education of the parents about special care needs after birth.
¶ Included on this list despite a carrier frequency lower than 1 in 100 because, as an X-linked syndrome, fragile X syndrome is more prevalent than other conditions.
Δ DNA testing alone will miss up to 10% of carriers of Tay-Sachs disease, especially in lower-risk groups and, therefore, enzyme-based testing may be a more appropriate choice for some patients.

Graphic 112582 Version 1.0

Algorithm for hepatitis B virus during pregnancy



Anti-HBc: hepatitis B core antibody; anti-HBe: hepatitis B e antibody; anti-HBs: hepatitis B surface antibody; HBeAg: hepatitis B e antigen; HBIG: hepatitis B immune globulin; HBsAg: hepatitis B surface antigen; HBV: hepatitis B virus.

* Check anti-HBs and anti-HBc if mother is at high risk for HBV infection (eg, injection drug user, sexual partner or household contact has chronic HBV). Mothers with no evidence of prior HBV infection (ie, negative for HBsAg, anti-HBs, and anti-HBc) should be vaccinated.

¶ Women who have a high HBV DNA (>200,000 int. units/mL), elevated aminotransferase levels, and/or a positive HBeAg should be referred to a hepatologist to see if early initiation of antiviral medications is needed.

Δ Start at 28 to 30 weeks gestation. We prefer tenofovir disoproxil fumarate rather than other antiviral agents. Refer to the topic on Hepatitis B and pregnancy for a more detailed discussion of treatment.

◇ For those who continue antiviral therapy after delivery, the pros and cons of breastfeeding must be discussed with the mother. Refer to the topic on Hepatitis B and pregnancy for more detailed discussions of breastfeeding.

Graphic 87231 Version 7.0

Women at increased risk of having a sexually transmitted infection

- Personal history of a prior sexually transmitted infection
- Age <25 years
- New sex partner in past 60 days
- More than one sex partner or sex partner with multiple concurrent sex partners
- Sex partner diagnosed with a sexually transmitted infection
- No or inconsistent condom use outside a mutually monogamous sexual partnership
- Trading sex for money or drugs
- Sexual contact with sex workers
- Meeting anonymous partners on the internet
- Unmarried status
- Lower socioeconomic status or high school education or less
- Admission to correctional facility or juvenile detention center
- Use of illicit drugs
- Living in a community with a high prevalence of sexually transmitted infections

Graphic 112388 Version 1.0

Risk factors for lead exposure in pregnancy and during lactation

Recent emigration from or residency in areas where ambient lead contamination is high
Women from countries where leaded gasoline is still being used (or was recently phased out) or where industrial emissions are not well controlled
Living near a point source of lead
Examples include lead mines, smelters, or battery recycling plants (even if the establishment is closed)
Working with lead or living with someone who does
Women who work in or who have family members who work in an industry that uses lead (eg, lead production, battery manufacturing, paint manufacturing, ship building, ammunition production, or plastic manufacturing)
Using lead-glazed ceramic pottery
Women who cook, store, or serve food in lead-glazed ceramic pottery made in a traditional process and usually imported by individuals outside the normal commercial channels
Eating nonfood substances (pica)
Women who eat or mouth nonfood items that may be contaminated with lead, such as soil or lead-glazed ceramic pottery
Using alternative or complementary substances, herbs, or therapies
Women who use imported home remedies or certain therapeutic herbs traditionally used by East Indian, Indian, Middle Eastern, West Asian, and Hispanic cultures that may be contaminated with lead
Using imported cosmetics or certain food products
Women who use imported cosmetics, such as kohl or surma or certain imported foods or spices that may be contaminated with lead
Engaging in certain high-risk hobbies or recreational activities
Women who engage in high-risk activities (eg, stained glass production or pottery making with certain leaded glazes and paints) or have family members who do
Renovating or remodeling older homes without lead hazard controls in place
Women who have been disturbing lead paint, creating lead dust, or both or have been spending time in such a home environment
Consumption of lead-contaminated drinking water
Women whose homes have leaded pipes or source lines with lead
Having a history of previous lead exposure or evidence of elevated body burden of lead
Women who may have high body burdens of lead from past exposure, particularly those who have deficiencies in certain key nutrients (calcium or iron)
Living with someone identified with an elevated lead level
Women who may have exposure in common with a child, close friend, or other relative living in the same environment

Reproduced from: Centers for Disease Control and Prevention. Guidelines for the identification and management of lead exposure in pregnant and lactating women. Atlanta 2010. Available at: http://www.cdc.gov/nceh/lead/publications/leadandpregnancy2010.pdf.

Graphic 86038 Version 3.0

Frequency of maternal blood lead follow-up testing during pregnancy

Venous blood lead level (BLL; mcg/dL)	Perform follow-up test(s)
<5	None (no follow-up testing is indicated).
5 to 14	Within one month. Obtain a maternal BLL* or cord BLL at delivery.
15 to 24	Within one month and then every two to three months. Obtain a maternal BLL* or cord BLL at delivery. More frequent testing may be indicated based on risk factor history.
25 to 44	Within one to four weeks and then every month. Obtain a maternal BLL* or cord BLL at delivery.
≥45	Within 24 hours and then at frequent intervals depending on clinical interventions and trend in BLLs. Consultation with a clinician experienced in the management of pregnant women with BLLs in this range is strongly advised. Obtain a maternal BLL* or cord BLL at delivery.

BLL: blood lead level.
* If possible, obtain a maternal BLL prior to delivery since BLLs tend to rise over the course of pregnancy.

Reproduced from: Ettinger AS, Wengrovitz AG. Guidelines for the Identification and Management of Lead Exposure in Pregnant and Lactating Women. Centers for Disease Control and Prevention. November 2010.

Graphic 81402 Version 5.0

Contributor Disclosures

Charles J Lockwood, MD, MHCM Consultant/Advisory Boards: Celula [Aneuploidy screening (No current products or drugs in the US)]. **Urania Magriples, MD** Nothing to disclose **Susan M Ramin, MD** Nothing to disclose **Vanessa A Barss, MD, FACOG** Nothing to disclose

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