# Prenatal Genetic Screening for Low Risk Women

## Definition
Most pregnancies that are complicated by chromosomal abnormalities and by neural tube defects occur in low risk women. Therefore, in order to identify as many cases as possible, screening tests must be done for all low risk pregnant women. At this time, there are 3 different types of screening tests: the quadrivalent marker screening, serum integrated screening, and full integrated screening. Selection of testing depends upon the gestational age at time of prenatal care initiation, the women’s ability to access (and pay for) the different tests, her personal preferences, and local practice.

## Subjective
Must include pregnant woman not at high risk for a fetus with an anomaly.

May exclude women who would benefit from diagnostic testing (not screening tests) outlined in Identification of Prenatal Candidates for Genetics Referral protocol, such as:
1. Women with personal or family history of an inherited disorder.
2. Women who know that the father of her fetus has a personal or family history of inherited disorders.
3. Women who are using teratogenic medications.
5. Women who will be age 35 or older at delivery.
6. Women with previous NTD-affected pregnancy.

Note: Some local protocols call for all women to be screened, with high risk women also being referred to genetics, even if screening values are normal.

## Objective
Fundal height is compatible with gestational age.

## Laboratory
See Plan.

## Assessment
Routine risk pregnant woman who desires prenatal screening for selective chromosomal and neural tube defects.

## Plan
There are 3 different screening tests; a fourth option (Contingency Screening) has recently been added. Discuss with patient which would best meet her priorities. See Table 1 for details of each option.

## Patient Education
1. Advise patient that the testing is voluntary.
2. Inform her which tests are covered by her insurance or if testing will cost out of pocket expenses for her.
3. Remind the patient that a positive screening test does not mean that her fetus will have a problem. A positive test means only that other tests need to be done to determine if her fetus is affected. Similarly, a negative test does not mean that the fetus is not affected.
4. Remind the patient that if the screening tests show that the fetus has a high probability of having a serious defect, all options will be open to her:
   a. She (and her family) will be able to prepare to care for a severely ill child.
   b. She (and her family) may be prepared to lose the fetus before or shortly after birth.
   c. She may decide to terminate her pregnancy.

## Refer to MD
1. Immediate consultation should be obtained for any questionable prenatal genetic screening test results that require additional interpretation for referral.
2. Patients with questions remaining about pregnancy dating or interpretation of results.

## References

**Table 1: Screening Options**
See below.
Table 1: Screening Options

<table>
<thead>
<tr>
<th>Quadrivalent Marker Screening</th>
<th>Serum Integrated Screening</th>
<th>Full Integrated Testing</th>
<th>Contemporary Screening</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. One blood specimen is drawn between 15 0/7 weeks and 20 0/7 weeks gestational age and 4 markers are tested: *AFP, hCG, uE3, Inhibin-A.</td>
<td>This has 2 blood tests: a. One blood specimen is drawn between 10 and 13 6/7 weeks and 2 markers are tested: PAPP-A and hCG. b. A second blood specimen is drawn between 15 and 20 weeks gestational age and 4 markers are tested: AFP, hCG, uE3, Inhibin-A.</td>
<td>1. This has 2 blood tests and an ultrasound. a. One blood specimen is drawn between 10 and 13 6/7 weeks and 2 marker tests are tested: PAPP-A and hCG. b. An ultrasound is done between 11 2/7 weeks and 14 2/7 weeks to look for nuchal translucency. c. A blood specimen will be drawn between 15 and 20 weeks gestational age and 4 markers are tested: AFP, hCG, uE3, Inhibin-A.</td>
<td>1. Combines the first trimester blood test between 10-13 6/7 weeks with 2 markers (PAPP-A and hCG).</td>
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<tr>
<td>2. Efficacy: this blood test can detect:</td>
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<td>2. Follow-up depends on outcomes of initial testing. a. Those in highest risk (0.1%), refer to CVS b. Those in lowest risk (74.9%), stop testing c. Those in intermediate risk (25%), proceed to 2nd trimester Quadrivalent testing.</td>
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<tr>
<td>80% Down Syndrome</td>
<td>85% Down Syndrome</td>
<td>90% Down Syndrome</td>
<td></td>
</tr>
<tr>
<td>67% Trisomy 18</td>
<td>79% Trisomy 18</td>
<td>81% Trisomy 18</td>
<td></td>
</tr>
<tr>
<td>97% Anencephaly</td>
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<td>97% Anencephaly</td>
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<tr>
<td>80% Open spina bifida</td>
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<tr>
<td>85% Abdominal wall defects</td>
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<td></td>
</tr>
<tr>
<td>60% SLOS (Smith Lemli Optiz syndrome)</td>
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<td>60% SLOS</td>
<td></td>
</tr>
<tr>
<td>3. If the screen is interpreted as being positive, the patient must be referred for more sensitive testing, including an anatomical ultrasound and possible amniocentesis.</td>
<td>There is no result given after the first test. The tests are combined to give a total result of positive or negative after the results of the second test are reviewed. 4. If the screen is interpreted as being positive, the patient must be referred for more sensitive testing including an anatomical ultrasound and possible amniocentesis.</td>
<td>The results of the first blood test will be combined with the ultrasound results to give an early answer. a. If the early tests are interpreted as positive, the woman may be referred for more sensitive testing early, including anatomic ultrasound and possible amniocentesis. The woman may decide to wait until the second blood test results are done before she goes for more sensitive testing. b. If the first set of tests is not interpreted as positive, the results are combined with the results of the second blood test. c. If her combined test results are positive, the woman will be referred for more testing for possible birth defects.</td>
<td></td>
</tr>
</tbody>
</table>

*MS-AFP maternal serum alpha-fetoprotein  
hCG total human chorionic gonadotropina  
uE3 unconjugated estriol

80% Down Syndrome  
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Inhibin-A  
PAPP-A pregnancy associated plasma protein A