

Screening for Birth Defects: Pros & Cons

The risk of birth defects is one of the few concerns capable of dampening expectant parents' euphoria. In recognition of this concern, screening blood tests for birth defects in younger pregnant women (under age 35) are available through the California Expanded AFP Screening Program. This article describes the tests and their goals, and then discusses the major issues regarding their use. While state law requires that eligible patients be offered the tests, every clinician should consider the following issues when counseling individual patients.

What Screening Tests are Offered, When and To Whom? A single blood sample is drawn for measurement of AFP (Alpha Feto-Protein), HCG (Human Chorionic Gonadotropin) and UE (Unconjugated Estriol) levels in maternal serum. The sample must be drawn during weeks 15-20, and ideally during weeks 16-17 in the second trimester. State-approved laboratories measure serum levels, returning results only as "screen positive" or "screen negative." A pregnant woman who tests "screen positive" is referred on for follow-up; i.e., genetic counseling, ultrasound and amniocentesis. (Older women are already candidates for genetic counseling, and may choose to bypass the screening test.)

Screening with follow-up can detect the following conditions:

- 97 percent of the cases of anencephaly
- 80 percent of the cases of open spina bifida
- 85 percent of the cases of abdominal wall defects
- 50 percent of the cases of Trisomy 18
- 40-66 percent of the cases of Down syndrome

"Screen positive" results are most commonly associated with non-genetic causes, such as incorrect gestational age, multiple pregnancies or normal variations in serum levels. Therefore, follow-up of "screen positive" results is necessary to properly evaluate the patient's condition and to recommend appropriate care.

What Factors Influence Use of the Screening Blood Test? Expanded AFP Screening is effective in detecting several types of birth defects. Thus, the test is indicated for pregnant women in their late 20s and early 30s, particularly those with a family history of inherited conditions or other "high risk" pregnancy factors. Despite their obvious utility, pregnant women retain the right to decline blood tests.

The window for reliable testing is relatively brief (i.e., weeks 15-20) and dating the pregnancy is often imprecise. False positives require follow-up, including sonography, to determine the correct gestational age.

The informational booklets, provided by the Department of Health Services, include a two-part Consent/Refusal form with a permanent copy for the patient's medical record. Booklets are available from the Genetic Disease Branch of the California Department of Health Services (510) 540-2534. However, no fail-safe procedure ensures that the blood sample will actually arrive at the lab for testing, nor that results are reported back to the ordering clinician. Therefore, it is up to the clinician to devise follow-up procedures to guard against mishaps. By drawing the blood sample or sending the patient to a lab, the clinician assumes the responsibility to evaluate the results and to initiate follow-up for "screen positives."

Finally, the limited reach of the Expanded AFP Screening Program must be thoroughly understood. The screening blood test cannot detect all birth defects and a "screen negative" result is no assurance that a fetus will be born healthy. Even with timely screening and conscientious follow-up, only some defects can be identified. Given the litigious climate, a patient may claim that a "screen negative" result provided her with a false sense of security. That is, a

negative blood test served to discourage further genetic testing that could have revealed the abnormality. Parents of infants born with unexpected anomalies are quick to seize upon any express or implied assurance from their doctor that their new baby would be born healthy. As always, a frank discussion of the risk and benefits, coupled with thorough documentation of informed consent, are necessary risk management precautions.