

# AMERICAN ACADEMY OF PEDIATRICS

## Committee on Genetics

### $\alpha$ -Fetoprotein Screening

The Committee on Genetics of the American Academy of Pediatrics endorses the following guidelines for maternal  $\alpha$ -fetoprotein screening, developed by the American Society of Human Genetics.<sup>1</sup>

Before maternal serum  $\alpha$ -fetoprotein (MSAFP) screening is started, the following minimal criteria should be met.

1. Adequate physician/health professional education: The objective of this education is to assure that physicians and other health care professionals understand the nature and range of severity of each of the detectable conditions and anomalies, the objectives of screening and testing, the importance of timing and reporting the low predictive value of test results, and the follow-up procedures and counseling.

2. Access to a qualified MSAFP and amniotic fluid  $\alpha$ -fetoprotein laboratory: Criteria for quality control in these laboratories are detailed in the full statement.

3. Adequate facilities and personnel for follow-up of abnormally high or low MSAFP values: Counseling, level I and level II sonography, amniocentesis, and qualitative amniotic fluid acetylcholinesterase assay should be available.

4. Adequate patient education: Information should be provided on the nature of the defects detectable by MSAFP screening, on confirmatory procedures, and on the options available should an abnormal result be confirmed.

The optimal time for MSAFP screening is between 16 and 18 weeks of gestation.

The common MSAFP screening protocol would include: first serum specimen evaluation, second serum specimen evaluation (if indicated), counseling, ultrasonography, amniocentesis, amniotic fluid  $\alpha$ -fetoprotein testing (acetylcholinesterase and fetal blood evaluation), karyotyping.

Once the program is underway, physician's offices, laboratories, and referral centers should have staff available who can answer questions concerning procedures related to MSAFP screening. Educational brochures that are appropriate for the population being served should be made available to patients, ie, accurate clinical and incidence data should be clearly stated in lay language.

Before the specimen for MSAFP is obtained, the patient should have been informed about the procedure and its implications and should indicate her willingness to be tested. For religious and ethical reasons, some couples may not want to be confronted with the dilemmas posed by an abnormal test result. Prenatal MSAFP screening should be voluntary. The provider should indicate its availability, educate the patient about its potential, and allow the patient to make decisions concerning participation in screening and the sequential steps in the management of pregnancies. The patient's decision on whether or not to have MSAFP screening should be recorded by means such as the patient's written signature.

Both high and low MSAFP values may be predictive of a serious birth defect or adverse pregnancy outcome. Screening protocols vary in whether a second sample is requested before proceeding with follow-up. Second samples are most appropriate when (1) the first MSAFP is minimally elevated and (2) there is time for a second specimen. When the result is elevated and sonography that is capable of detecting a fetal defect is available, or when the pregnancy is relatively advanced, some centers dispense with a second sample. Appropriate information and counseling should be made available to the patient during each step of the process.

The designation of elevated (or investigational low) MSAFP values should depend on characteristics of the patient (eg, age and health status) and on the prevalence of the defect in the population being screened. The risk of Down syndrome of a 35-year-old woman (ie, 1/270 in the second trimester)

ter) would be reasonable for defining the cut-off point for low values.

When incorrect gestational age, multiple gestation, and fetal death have been excluded by level I ultrasound as causes for elevated results, there should be prompt consultation or referral to a center where level II ultrasound, amniocentesis, and other confirmatory techniques are available. Both levels of ultrasound should be performed by those experienced in sonography.

Laboratory measurements are obtained in quantitative units (nanograms or international units per milliliter) and are frequently reported as multiples of the median. Such expressions do not address the degree of risk to the individual patient. As information is collected that permits an estimate of risk in subpopulations comparable to the woman being screened, laboratories should include in the laboratory report the risk to the woman on the basis of her test result.

Pregnant women with insulin-dependent diabetes mellitus should be considered as a separate category.  $\alpha$ -Fetoprotein levels are lower, on average, for women with diabetes, and this should be taken into consideration when interpreting results.

COMMITTEE ON GENETICS, 1986-1987

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REFERENCE

1. The American Society of Human Genetics: Policy statement for a maternal serum alpha fetoprotein screening program and quality control for laboratories performing quality control for laboratories performing maternal serum and amniotic fluid alpha fetoprotein assays. *Am J Hum Genet* 1987;40:75-82

**$\alpha$ -Fetoprotein Screening**  
*Pediatrics* 1987;80;444

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OFFICIAL JOURNAL OF THE AMERICAN ACADEMY OF PEDIATRICS

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*Pediatrics* 1987;80;444

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