Though only recently proposed as a subspecialty[1], primary care fetal medicine began in the late 1950s with the development of techniques for the evaluation and subsequently the treatment of fetal hemolytic anemia.[2–4] With several exceptions, further developments in fetal medicine were limited to the diagnosis of abnormalities and the characterization of their natural histories until the early 1980s. These obligatory first steps continue today, since it is impossible to treat without an accurate diagnosis and unwise to treat without knowledge of the likely outcome. Though the subspecialty is still in its early development, the fetus is no longer hidden and unapproachable. The emergence of the fetus as a patient generates the potential for conflict between the autonomous woman and her dependent fetus.

While this paper focuses on one advantage of fetal diagnosis, the potential for therapy, there are a number of other salient advantages which should be acknowledged: (1) Alleviation of anxiety. Ninety-eight percent of tests confirm normality, providing reassurance and perhaps dissuading a woman from terminating an at-risk pregnancy. (2) Improvement in both maternal and neonatal care. Obstetric and neonatal care may be modified specifically to support the ill fetus/neonate. (3) Improved likelihood of family adjustment to a fetal problem. Without question, learning that their future child is not perfect constitutes a traumatic event for the parents. Prenatal diagnosis provides time to activate coping mechanisms and to initiate appropriate medical and psychological counseling of the couple while there is still time for reflection.

**Fetal Diagnosis**

The first step in fetal therapy is diagnosis. Most patients who conceive a fetus with an anomaly or whose fetus experiences a complication during pregnancy, such as a viral infection or uteroplacental insufficiency, were not known to be at risk at the outset of their pregnancy. Screening is thus of the utmost importance. To deny a patient access to effective screening when
available, whatever the modality, is to directly deny the patient and, indi-
rectly, her fetus access to medical care.

Noninvasive Diagnosis

Hormonal Screening
Current hormone screening during pregnancy consists of measuring maternal
α-fetoprotein (MSAFP), estriol, and hCG between 16 and 20 weeks’ gesta-
tion. This “triple screen” is valuable for the identification of (1) a variety of
fetal anatomic defects including open spina bifida and abdominal wall defects,
(2) fetal chromosome abnormalities; (3) fetal infection, and (4) pregnancies
with normal fetuses but an abnormal placenta placing the fetus at risk for
preterm delivery, growth retardation, and stillbirth.[5–10]

MSAFP is usually elevated when the fetus has an open spine or abdomi-
nal wall defect. The measurement of MSAFP permits the identification of
almost all fetuses with abdominal wall defects and approximately 80% of
fetuses with a neural tube defect. The expected outcome of a fetus with an
abdominal wall defect unaccompanied by another malformation is very good
after postnatal repair. In contrast, the long-term prognosis for children with
a neural tube defect depends upon both the location and the extent of the lesion.
Counseling must take these elements into consideration.

Women whose fetuses have one of several chromosome abnormalities have
a low MSAFP. Application of the “triple screen” to the detection of fetal
chromosome abnormalities permits the identification of 60% of all trisomy
21 fetuses compared to only 24% if maternal age alone is used as the criteria
for amniocentesis. Though the information on the detection of chromosome
abnormalities other than trisomy 21 is preliminary, it appears that a similar
number of other aneuploidies will also be detected. The percentage of screened
women who undergo amniocentesis remains constant since the triple screen
allows modification of the predicted risk for the older gravida. A woman
who might have chosen to undergo amniocentesis because of her age may now
decline if the predicted risk after triple screening is similar to that of a much
younger woman. Because the positive yield from amniocentesis is higher
with the triple screen than that achievable using other screening criteria, the
cost per case identified and the number of normal fetuses lost per case
identified are also significantly reduced.

Ultrasound
Primary care fetal medicine could not exist without high-resolution diag-
nostic ultrasound to illuminate this previously dark and forbidden area. Initially,
sonographic resolution was poor and the examinations focused primarily on
the documentation of gestational age. With technologic improvements, it
became possible accurately to identify a wide variety of malformations. High-
resolution ultrasound now permits not just the determination of fetal viability,
number, position, and gestational age, but also the near 100% accurate