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The role of the maternal-fetal medicine specialist

ABSTRACT

The maternal-fetal medicine specialist is trained to manage high-risk pregnancies and obstetric complications. This paper describes the role of the maternal-fetal medicine specialist on the obstetric health care team and conditions in which he or she may enhance the outcome of pregnancy.

OBSTETRICS has become more complicated in recent years. Not only have we seen an explosion of new technology and discoveries about the physiology and pathology of pregnancy, but other areas of medicine have been advancing as well. These developments now make having a baby a possibility for women who might not have had this option a few years ago owing to the risk of complications.

As knowledge increases, medicine specializes. The specialty of maternal-fetal medicine was created to provide expertise for women who may require special skills and resources during pregnancy.

WHAT IS A MATERNAL-FETAL MEDICINE SPECIALIST?

A maternal-fetal medicine specialist is an obstetrician-gynecologist with additional years of education, experience, and training in the medical and surgical complications of pregnancy: a 4-year residency and a 3-year fellowship program leading to board certification.

Maternal-fetal medicine specialists act as consultants for the obstetrician, family practitioner, and nurse-midwife; they care for patients directly; they provide preconception counseling, antenatal supervision or surveillance; and they direct intrapartum or postpartum management.

WHEN SHOULD A PATIENT SEE A MATERNAL-FETAL MEDICINE SPECIALIST?

According to the Society of Perinatal Obstetricians (now the Society of Maternal-Fetal Medicine),[1] a maternal-fetal medicine specialist may enhance the outcome of pregnancy:

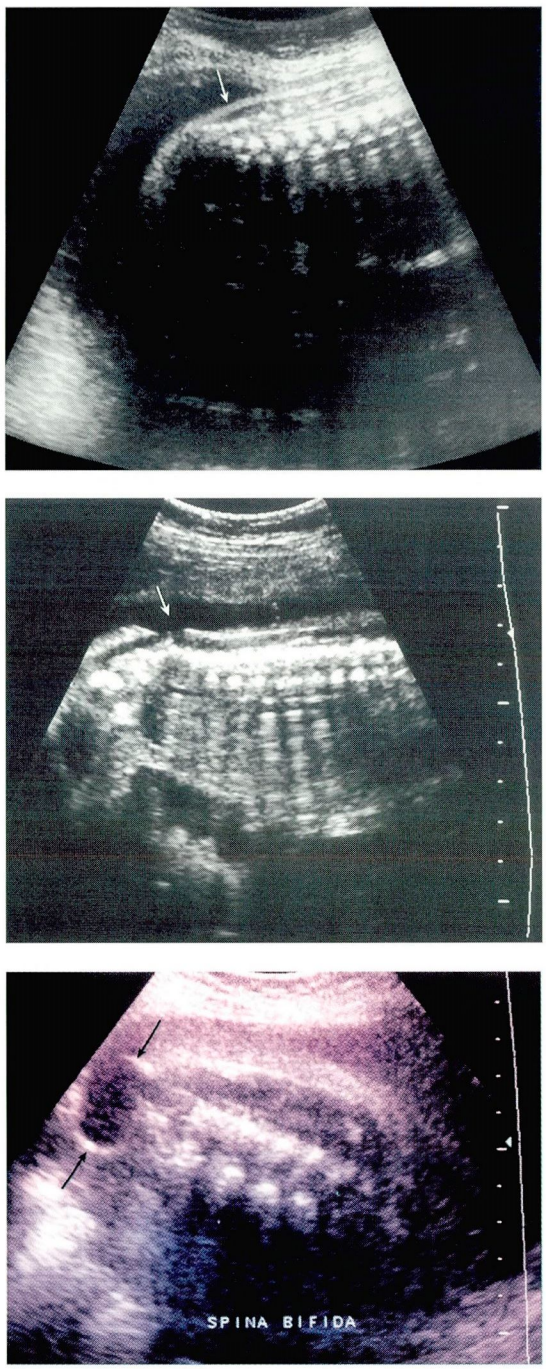
- When a diagnostic or therapeutic procedure is required because of the risk of fetal chromosomal abnormalities
- When the mother is healthy but the fetus is at high risk, ie, in cases of multiple pregnancy, incompetent cervix, or a history of complications during a previous delivery
- When an antepartum patient is admitted for a reason other than delivery, or when a postpartum patient has complications
- When an expectant mother has a known medical or surgical condition.

Although the impact of maternal-fetal medicine specialists on outcome for mother and fetus has yet to be proven in clinical trials, I believe that convincing data will be forthcoming.

The maternal-fetal medicine specialist is a care provider, consultant, and counsellor

DETECTING FETAL CHROMOSOMAL ABNORMALITIES

The risk of chromosomal abnormalities increases as the mother gets older. In this country, a mother who is 35 years or older is offered genetic counseling due to this



A level 2 ultrasound includes a detailed survey of fetal cardiac, neurologic and genitourinary systems

FIGURE 1. Top, level 2 ultrasonogram showing a normal fetal spine. Note the white line (arrow) representing the skin covering the spine. Middle, spina bifida (note the break in the white line indicated by the arrow). Bottom, another spina bifida in the lower aspect of the fetal spine (between arrows). Both of these cases were identified by an abnormal maternal serum triple test obtained at 15 or 16 weeks.

increased risk. Counseling is also offered if the father is older than 50 or 55 years, or if either of the parents has or carries the trait for a genetically inherited disease.

Techniques to assess fetal chromosomal abnormalities can be noninvasive (eg, ultrasound examination, triple-screen marker blood testing) or invasive (eg, chorionic villus sampling, amniocentesis, and percutaneous umbilical cord blood sampling). Invasive techniques tend to be more accurate than the noninvasive techniques but entail greater risks. A maternal-fetal medicine specialist can perform both the testing and the counseling and follow-up.

Ultrasound examination

A screening or basic (level 1) ultrasound examination can ascertain the gestational age, fetal position, volume of amniotic fluid, and location of the placenta. A variety of health-care professionals can perform this procedure.

A comprehensive (level 2) ultrasound examination includes all of the level 1 data plus a detailed anatomical survey of the fetus, particularly the fetal cardiac, neurologic, and genitourinary systems. It is usually done between 16 and 24 weeks of gestation, and generally for a specific reason, such as if the level 1 examination has disclosed an abnormality or if the fetus is otherwise at risk.

Either a radiologist or fetal-maternal medicine specialist can perform a level 2 examination, but a fetal-maternal medicine specialist has the advantage of being able to follow up any abnormal results. For example, if an ultrasound examination to determine the gestational age of the fetus reveals a gastroschisis (a large anterior abdominal wall defect), a maternal-fetal medicine specialist could discuss the significance of these findings with the patient and family, make recommendations about whether to deliver the baby by cesarean section, and coordinate care with other members of the health care team such as a pediatric surgeon.

The triple-screen marker blood test

Offered to all patients as a screening test at 15 to 20 weeks of gestation, the triple-screen blood test consists of measurements of alpha-fetoprotein, beta human chorionic gonadotropin, and estriol in the mother's blood.

Abnormalities in these substances can indicate a neural tube defect or other abnormality in the development of the fetal brain, spinal cord, or other tissues of the central nervous system (FIGURE 1), or another developmental or genetic disorder.

This test often gives the first indication that a problem exists, but it also often gives false-positive results. Therefore, skilled interpretation and evaluation of abnormal results is essential.

Chorionic villus sampling

Chorionic villus sampling can diagnose chromosomal disorders such as Down's syndrome (FIGURE 2), cystic fibrosis, Tay-Sachs disease, and sickle cell disease. Unlike maternal serum triple-screen testing, it cannot detect open neural tube defects.

Chorionic villus sampling is performed between 10 and 12 weeks of gestation. Depending on the location of the placenta, the procedure is performed transvaginally or transabdominally. A small sample of cells (chorionic villi) is taken from the placenta where it is attached to the wall of the uterus. Results are generally available within 7 to 10 days.

This procedure carries approximately a 1% risk of miscarriage. Other complications may include bleeding, cramping, and rarely infection.

Before the procedure, patients should receive genetic counseling and information about the risks and possible benefits of the procedure. Afterward, they should receive counseling to help them interpret the results.

Amniocentesis

Of the invasive tests, amniocentesis is the safest, most reliable, and most often used. It is usually done between 15 and 20 weeks of gestation. Early amniocentesis (at 13 to 15 weeks) has been promoted by some, but recently the risks associated with the procedure have been reported to be increased when it is performed at this earlier gestational age.

Amniocentesis can detect certain neural tube defects, as well as Down's syndrome and other chromosomal abnormalities. In families with genetic risks, amniocentesis can be used to diagnose sickle cell disease, cystic fibrosis, muscular dystrophy, and Tay-Sachs disease.

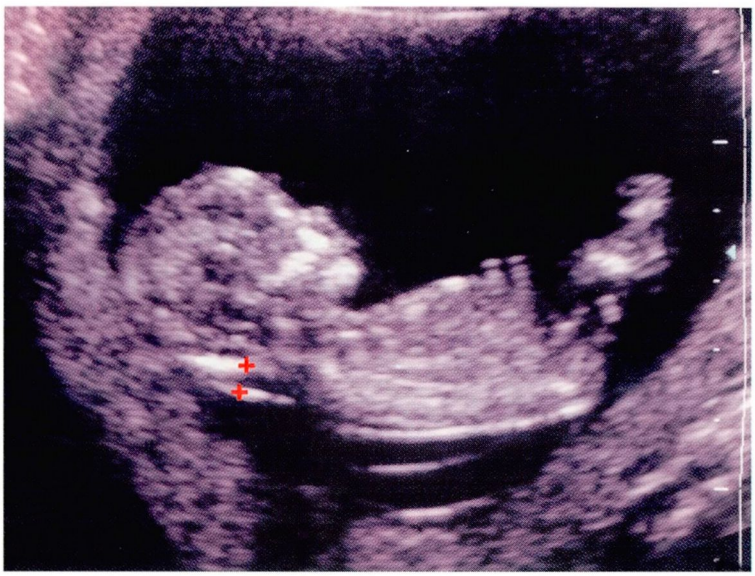


FIGURE 2. Transvaginal ultrasonogram performed at approximately 10 weeks of gestation because of advanced maternal age. The nuchal thickness or translucency (indicated by the cross-hairs) measured 0.4 cm (upper limit of normal 0.2 cm). Because this finding has been associated with an increased risk of trisomy 21 (Down's syndrome), a transvaginal chorionic villus sampling was performed, indicating that this fetus did have the 47, XY+21 karyotype ie, Down's syndrome.

Percutaneous umbilical cord blood sampling

Percutaneous umbilical cord blood sampling, performed while the fetus is in utero, is often used for conditions in which rapid diagnosis or management decisions are crucial. Indications include Rh disease with red blood cell isoimmunization, fetal hemolytic disease, hemoglobinopathies, coagulation defects, platelet abnormalities, evaluation of fetal acid-base status, rapid karyotyping, and intrauterine infection such as rubella, toxoplasmosis, and cytomegalovirus. Generally, the umbilical cord is accessible after 20 weeks of gestation.

WHEN A MOTHER HAS A MEDICAL OR SURGICAL CONDITION

Hypertension, diabetes mellitus, heart disease, and other conditions increase the risk of complications for both mother and fetus during pregnancy. Other conditions include anything from infectious disease to renal disease to eating disorders.

TABLE 1

Some drugs and substances to avoid during pregnancy

ACE inhibitors
Alcohol
Diethylstilbestrol
Iodine
Isotretinoin (Vitamin A derivative)
Organic mercury
Phenytoin
Tetracycline
Thalidomide
Trimethadione
Valproic acid
Warfarin

Diabetes mellitus

The risk of congenital malformations of the fetus due to abnormal glucose metabolism is well established,[2,3] and the risk is higher when the father also has diabetes. Euglycemic control in diabetic women before pregnancy may reduce the risk of congenital malformations. Preconception counseling with a maternal-fetal medicine specialist can help mothers achieve good glucose control, although many women with diabetes mellitus do not receive this type of counseling.

Systemic lupus erythematosus

A key factor in pregnancy outcome for women with systemic lupus erythematosus appears to be the state of activity of the disease at the time of conception. Outcome is optimal when the maternal condition is in remission for at least 6 months and when renal function remains good.[4] Again, preconception counseling by the maternal-fetal medicine specialist would be beneficial.

History of pulmonary embolism or deep venous thrombosis

The hypercoagulable state of pregnancy and venous compression of the major vessels by the pregnant uterus on the extremities may necessitate use of anticoagulant drugs throughout pregnancy.

Pregnancy's impact on chronic disease

The maternal-fetal medicine specialist also focuses on the effects of pregnancy on the mother's chronic medical condition. For example, diabetic mothers may experience worsening of eye function during pregnancy, while renal function may be less affected or even unaltered.[5,6] Certain cardiac conditions—ie, pulmonary hypertension, coarctation of the aorta with valvular involvement, and Marfan syndrome with aortic root dilation—are associated with an extremely high (approximately 50%) maternal mortality rate.[7]

Use of medications during pregnancy

A maternal-fetal medicine specialist may be able to provide useful information regarding the use and choice of medications during pregnancy, including which drugs and substances to avoid (TABLE 1).[8–10] For example, methyldopa has been the drug of choice for pregnant women with hypertension for many years, but new algorithms now involve other agents. On the other hand, women should avoid angiotensin-converting enzyme (ACE) inhibitors during pregnancy, as they have been associated with a decrease in amniotic fluid, renal failure, and sudden intrauterine fetal demise.[8]

All medications are categorized according to the risk they pose in pregnancy.[9,10]

Category A and B drugs pose little risk to pregnancy.

Category C drugs should be given if the potential benefit justifies the potential risk to the fetus: early studies may have revealed an adverse effect, but no controlled studies have been done in pregnant women.

Category D drugs are those with positive evidence of fetal risk in human pregnancy, but in which the benefits for use in pregnancy may be acceptable despite the risk.

Category X drugs are those shown to pose a definite risk to the fetus.

LINKING PREPARTUM, PERIPARTUM, AND POSTPARTUM CARE

After giving birth, women tend to return to their family physicians or nurse-practitioners. The maternal-fetal medicine specialist helps

Postpartum patients should be referred back to the family doctor with complete documentation

coordinate postpartum care and may provide valuable insights into a patient's postpartum management through contact with the family physician. In addition, the maternal-fetal medicine specialist refers the patient back to her medical provider with complete documentation of the obstetrical and medical course, so that the entire system of referral remains beneficial and intact.

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