Study guide

Chapter 6: Student activity 6-A

ANSWER KEY

Preconception planning and fetal health

1. A genetic counselor works with a person or family that may be at risk for inherited disease or an abnormal pregnancy outcome and discusses their chances of having children who are affected.
2. The genetic counselor is experienced in helping families understand birth defects and how inheritance works.
3. The purpose of a screening test is to identify pregnancies at increased risk for genetic disorders or birth defects, and may be given even when there are no risk factors.
4. MSAFP and ultrasound.

   MSAFP measures the amount of alpha-fetoprotein in the mother's system. High levels of AFP are linked to a few major defects like neural tube defects. Low levels are linked to certain chromosome disorders such as Down syndrome. Most cases of high or low levels are false alarms.

   MSAFP Plus screening is a noninvasive way to screen the pregnancy for Down syndrome, an open neural tube defect, and trisomy 18. MSAFP Plus screening analyzes three substances in the mother's bloodstream, combined with the mother's age, race, weight, diabetic status, and gestational age to determine a risk for the above three conditions. The screening test is offered between 15 to 20 weeks during pregnancy. It is standard of care for a doctor to offer the test to the woman.

   Ultrasound uses sound waves to show the fetus. It can detect birth defects, the age of the fetus, and identify twins. Not all defects are detectable. Ultrasound is a procedure performed on most pregnancies in the United States. It assesses the growth and physical development of the fetus. In most pregnancies, two ultrasounds are done. The first is to date the pregnancy and the second is to assess fetal growth and physical development. There are no known risks to the mother or baby.
5. Diagnostic testing: Diagnoses a specific condition. Usually invasive. Used in pregnancies known to be at increased risk.

   Screening tests: Used to identify pregnancies in which there may be a problem. Used on low-risk populations. Noninvasive.
6. Amniocentesis: Can be done beginning at 15 weeks. Upper risk of 1/200. Withdraws amniotic fluid that contains fetal cells. Tests for chromosome disorders, some genetic disorders, and open neural tube defects.

   Chorionic villus sampling: Performed between 10 to 12 weeks. Risk of about 0.5 to 1 percent. Withdraws a piece of the developing placenta (chorion). Tests for chromosome disorders and some genetic conditions. Targeted ultrasound and MSAFP screening recommended at appropriate times following CVS.
7. Benefits of screening tests: Good for low-risk populations of women; noninvasive; lower costs; identify pregnancies that need further attention; may affect medical management of pregnancy.

   Disadvantages of screening tests: False negatives and false positives; if all women were routinely screened (and individual choice was not considered) then many women/couples would be led to diagnostic testing and possibly to making decisions that they do not want to make and having information that they did not want to have.
8. Benefits of diagnostic testing: Usually provides definitive answers about the pregnancy so that decisions can be made and parents prepared.

   Disadvantages of diagnostic testing: Invasive and therefore has a risk to the pregnancy; expensive; not appropriate for all women to have, but appropriate if there is an indication for testing.