

## Genetics and Prenatal Screening

### **Learning Objectives**

Review the following Learning Objectives as an organized beginning to your study of this module. As you read the Learning Objectives, note key words which will aid you in finding the information in the texts. When you complete the module, revisit this list and check for areas that require further investigation.

- Review the actions and properties of DNA and RNA.
- Review the Embryology and Fetal Development module.
- Identify family and health history issues that indicate genetic screening may be appropriate for your clients.
- Become familiar with counseling techniques and informed choice issues around choosing genetic screening during pregnancy.
- Examine your own bias and belief system about prenatal testing.
- Understand what disorders can be detected by the MsAFP, screen.
- Identify the accuracy of the MsAFP screen.
- Identify the components of the Triple Marker Screen and its accuracy and compare to the more recent Quad Screen.
- Understand the chorionic villi sampling procedure, and iatrogenic risks.
- Understand the amniocentesis procedure and iatrogenic risks.
- Identify the methods of pregnancy termination in each trimester.
- Identify community resources for genetic counseling, CVS and amniocentesis.
- Review Grief module.
- Demonstrate your ability to inform and discuss genetic and prenatal screening with clients, including the use of informed consent.

## Genetics and Prenatal Screening, continued

### **Study Sources**

The following texts are recommended for completion of this module. Use them to cross reference and build a more comprehensive understanding.

Using key words from the Learning Objectives, search the index. Read those pages listed, and read the chapter in which they are found. Establish a context for the information so that you understand how other topics are related. In addition, read the chapter headings in the Table of Contents, and flip through each text to familiarize yourself with the content of chapters. As you work through Study Group modules, you will eventually read each text in its entirety.

*Human Anatomy and Physiology, Marieb*

*Varney's Midwifery*

*Myles Textbook for Midwives*

*Holistic Midwifery, Vol. I, Frye*

*Understanding Diagnostic Tests in the Childbearing Year, Frye*

*The Tentative Pregnancy, Katz Rothman*

### **Related Topics**

- ◇ Prenatal Lab Work and Assessment
- ◇ Informed choice/informed consent
- ◇ Fertility and conception
- ◇ Embryology and Fetal Development
- ◇ Biology of cells
- ◇ IUGR
- ◇ Miscarriage and Stillbirth
- ◇ Grief

## Genetics and Prenatal Screening Questions

1. What is the difference between a screening procedure and a test?
2. Which screens and tests are invasive procedures, and which are non-invasive procedures?
3. Describe the sonogram procedure. When are sonograms utilized during pregnancy?
4. What does the abbreviation MsAFP represent?
5. What does the MsAFP screen for?
6. Describe the collection procedure for the MsAFP.
7. Describe the amniocentesis procedure.
8. What does the acronym CVS represent?
9. Describe the CVS procedure.
10. Why might a woman choose CVS over amniocentesis?
11. If a woman chooses CVS testing is it also necessary for her to have the MsAFP?
12. What is the most likely reason for a low MsAFP result?
13. List the components of the Triple Marker test.
14. When is the Triple Marker test performed?
15. What is the Triple Marker test designed to detect? How does it compare to the Quad Screen?
16. What is the difference between recessive and dominant genetic conditions?

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17. Name four recessive disorders.
18. Name three dominant disorders.
19. Where do the X-linked inherited disorders originate?
20. Are male and female offspring at equal risk of being afflicted with an X-linked disorder?
21. Can a female be afflicted with hemophilia?
22. List some conditions that are elusive to testing.
23. What might family and health history reveal that would indicate genetic screening may be appropriate by your clients?
24. Who is at risk for the following disorders?
  - Tay-Sachs Disease
  - open neural tube defects
  - Cystic Fibrosis
  - thalasemias
  - Sickle Cell
  - trisomy disorders
  - phenylketonuria
  - galactosemia
  - maple syrup urine disease
25. What is trisomy 21 commonly called?
26. What is the risk in any pregnancy that a baby will be born with some type of disorder?
27. What is the risk of having a baby with a disorder that can not be detected by amniocentesis?

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28. What must you discuss with a woman concerning her decision making process around genetic testing?
29. What is the risk of miscarriage after an amniocentesis procedure?
30. How do the risks for amniocentesis compare with those of chorionic villi sampling?
31. What are you likely to observe in a baby with Down Syndrome?

**Essay**

1. Explain the order of genetic testing, as you would present it to clients.
2. Describe the use of informed consent/informed choice as it applies to prenatal screening and genetic testing.
3. At what point do you discuss the possibility of a client having to make a decision about terminating her pregnancy?

**Projects** (send completed projects with the rest of your course work for this module)

1. Select or create an information sheet to provide to clients regarding genetic and prenatal screening.
2. What resources does your state provide to families for genetic counseling and testing?
3. Identify the regional lab that provides supplies and processing of the MsAFP screen.
4. Research in your own community where amniocentesis, CVS and genetic counseling services are available. Create a referral list for your clients.
5. Read *The Tentative Pregnancy*. Write about your reactions to the stories in this book. Did your ideas about genetic testing change after reading this book?



## Genetics and Prenatal Screening, continued

1. Draft practice guidelines for genetic counseling and screening in your own practice. Include reference to your informed consent procedure. Submit this draft and include it later in your Practice Guidelines projects (in the Charting and Practice Guidelines Module.)

### **Skills**

Following are excerpts from the NMI forms for assessment of midwifery skills, which include all skills identified and required by NARM. Review the following skills and consider how they each relate to the content of this module. If you are currently working with a preceptor, take this opportunity to focus on these areas. During Supervised Primary Care you will formally evaluate these skills together using the NMI form *Preceptor Evaluation/Student Self-Assessment of Midwifery Skills*.

#### 1. Midwifery Counseling, Education and Communication:

- A. Provides interactive support and counseling and/or referral services to the mother regarding her relationships with her significant others and other health care providers
- B. Provides education, support, counseling and/or referral for the possibility of less-than-optimal pregnancy outcomes
- C. Provides education and counseling based on maternal health/reproductive/family history and on-going risk assessment
- D. Facilitates the mother's decision of where to give birth
  1. The advantages and the risks of different birth sites
  2. The requirements of the birth site
  3. How to prepare, equip and supply birth site
- E. Educates the mother and her family/support unit to share responsibility for optimal pregnancy outcome
- F. Educates the mother concerning the natural physical and emotional processes of pregnancy, labor, birth and post partum
- G. Applies the principles of informed consent
- H. Provides individualized care
- I. Advocates for the mother during pregnancy, birth and postpartum
- J. Provides education, counseling and/or referral, where appropriate for:
  1. Genetic counseling for at-risk mothers
  3. Prenatal testing
  8. Complications
  9. Environmental risk factors
- G. Evaluates laboratory and medical records from other practitioners
- H. Obtains assistance evaluating laboratory and medical records from other practitioners

*Continued...*

## Genetics and Prenatal Screening Skills continued

### 3. Maternal Health Assessment:

K. Recognizes and responds to potential prenatal complications by:

8. Identifying and dealing with pre-term labor with:

B) Consultation and/or treatment including:

- 1) Increase of fluids,
- 2) Non-allopathic remedies,
- 3) Discussion of the mother's fears,
- 4) Food to be eaten at least every two hours,
- 5) consumption of alcoholic beverage,
- 6) Evaluation of urinary tract infection,
- 7) Evaluation of maternal infection

9. Assessing and evaluating a post-date pregnancy by monitoring /assessing:

- a) The need for consultation,
- b) Fetal movement, growth, and heart tone variability,
- c) Estimated due date calculation,
- d) Previous birth patterns,
- e) Amniotic fluid volume,
- f) Maternal tracking of fetal movements ,
- g) Referral for ultrasound,
- h) Referral for non-stress test
- i) Referral for contraction stress test,
- j) Referral and collaboration for biophysical profile