**Prenatal Genetic Screening**

National Midwifery Institute, Inc.

Study Group Coursework

*Syllabus*

Description:

This module explores genetic screens and genetic test during the perinatal period. It includes recommended reading materials in print and online, and asks students to complete short answer questions for assessment, long answer questions for deeper reflection, and learning activities/projects to deepen your hands-on direct application of key concepts.

Learning Objectives:

* 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 prenatal genetic screening.
* Examine your own bias and belief system about prenatal genetic testing.
* Understand what disorders can be detected by prenatal genetic screening.
* Identify the accuracy of prenatal genetic screening.
* Identify the different screenings and tests available to your clients for prenatal genetic screening.
* Understand the risks and benefits of each prenatal genetic screening/test.
* Identify the methods of pregnancy termination in each trimester.
* Identify community resources for genetic counseling, and advanced investigations.
* Review Grief module.
* Reflect on the tension between the disability justice movement and prenatal genetic screening

Learning Activities:

* Research and read appropriate study sources, seeking out additional study sources where needed
* Complete short answer questions in attached module document for assessment
* Complete long answer questions for deeper reflection in attached module document for assessment
* Complete learning activities listed in attached module document for assessment
  + Create information sheet to provide to clients regarding options
  + Identify local state/province/jurisdiction resources for families
  + Identify local labs and hospitals offering genetic counseling and labwork
  + Create a client information sheet about continuing to carrying a pregnancy with known anomolies
  + Reflect on personal stories about genetic screens, test and decisions.
  + Create Practice Guidelines
* Submit work to Study Group Course Coordinator
* Reflect on feedback from Study Group Course Coordinator and re-submit work as needed

Study Sources (print):

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

Using keywords 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.

* Review the Embryology and Fetal Development module.
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Study Sources (online):

See NMI website Prenatal Genetic Screening module web resources section for current online study sources for this module.

Related Modules:

* 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

Submitting Module for Assessment:

Study Group modules are accepted electronically in PDF format *only*. We encourage you to submit modules as you complete them throughout each quarter of enrollment.

Please e-mail your completed Study Group module to:

Study Group Course Work Instructor nmistudygroup@nationalmidwiferyinstitute.com

Once your module has been emailed to us, you will receive an email confirmation that we have received it. Study Group modules are reviewed and returned in digital format as PDF documents. Modules can take up to 1 month from submission to be reviewed and returned to you. We will return your module as an e-mail attachment. Each module includes an Evaluation Sheet at the end of the pdf. The module’s page on the student portal also includes a link to a fillable online module evaluation sheet. Please take the time to fill out the module evaluation sheet and return it to us for each module, it helps us to improve our course work.

Please follow these formatting guidelines when submitting modules:

* Your first initial and last name in title of PDF, along with name of module. Example: “ERyanFirstStage.pdf”
* Title of module on the document’s front page
* Your name on the document’s front page
* Provide the text of each question, followed by a blank line and then your thoughtful answer (without the question, you have commentary without context)
* Blank line between the answer for a question and the next question: question, blank line, answer, blank line, question, blank line, answer…
* Please leave margin space for our comments!
* Don’t use script or cursive writing style text
* Font size not smaller than 12
* Credit sources of direct quotes

Completion Requirements and Feedback:

In order to complete this module for graduation purposes from National Midwifery Institute you must review all resources, complete the attached short answer questions for assessment, long answer questions for deeper reflection, and learning activities/projects, and submit them as detailed above. Upon return to you, your coursework may have feedback or ask for additional information or exploration on certain topics. Your work will be evaluated n the following Rubric (pasted below). You must achieve a minimum score of **7.5** in order to move on to your next module, though we encourage all students to strive for a **10.**

|  | **Level 1**  **(0 Points)**  **Not Adequate** | **Level 2**  **(1 Point) Developing Adequacy** | **Level 3**  **(1.5 points)**  **Meets Basic Expectations** | **Level 3**  **(2 points) Exceeds Expectations** | **Student Score** |
| --- | --- | --- | --- | --- | --- |
| **Completion of module prompts and elements** | -Module not completed | -Major Elements of module are missing | -All aspects of module elements present, with some minor questions unanswered or missing | -All aspects of module elements present and answered completely |  |
| **Demonstrates Comprehension of module content and concepts** | - Lack of comprehension | - Responses are unclear and do not reflect basic comprehension of module concepts | - Responses are clear and reflect basic comprehension of module content and concepts | - Responses are clear, well written, and reflect in-depth comprehension of module content and concepts. Added subpoints and additional reflections demonstrate a deeper knowledge and curiosity. |  |
| **Analysis** | - Key terms not defined | -Inaccurate definitions of key items  -Limited connections made between evidence, subtopics and clinical experience | -Accurate definitions of key items  -Connections made between evidence, subtopics and clinical experience -Incorporation of original ideas and incorporates some clinical experience  in responses where possible | - Accurate definitions of key items  -Strong connections made between evidence, subtopics and clinical experience |  |
| **Evidence** | - No research evidence used | -Research not used  -Research not clearly connected to questions asked in module | -Research is present but limited -Research presented is weak or not relevant to communities served by midwives | -Research is abundant -Research is compelling and relevant to communities served by midwives |  |
| **Engagement with Learning Resources** | -Evident study sources were not utilized | -Evident study sources were partially utilized | -Evident that study sources were fully utilized | -Evident that study sources were fully utilized and independent research was undertaken -Full incorporation of original ideas, personal analysis and incorporates relevant clinical experience in all areas possible |  |

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 forms *Form 52 - Assessment of Student’s Midwifery Skills* and *Form 53 - 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 ongoing 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 postpartum

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

3. Maternal Health Assessment:

K. Recognizes and responds to potential prenatal complications by:

8. Identifying and dealing with preterm 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

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*Short Answer Questions*

Short Answer Questions:

1. What is the difference between a screening procedure and a diagnostic test?

2. Consider the following 9 prenatal procedures.

1. Early Ultrasound
2. Nuchal Translucency Screen
3. Fetal Anatomy Scan
4. Cell-free DNA Testing / Maternal Serum Blood Test
5. Ancestry-Based Carrier Screening
6. First Trimester Screen or Integrated Screen
7. Triple Marker / Quad Screen
8. Chorionic Villus Sampling
9. Amniocentesis

For each of the above procedures please answer the following:

1. What is it?
2. Is it a screen or a diagnostic test?
3. What does it measure, or what are they looking for?
4. What is the timing window for the procedure? why?
5. What may the results indicate?
6. What are the next steps if results are abnormal to offer to parents?
7. Any other notes you have about these procedures

3. Which screens and tests are invasive procedures, and which are non-invasive procedures?

4. Describe the ultrasound procedure in the first, second, and third trimester. What preparations must clients make?

5. Describe transvaginal ultrasound and abdominal ultrasound. How do the procedures differ?

6. What are the risks associated with ultrasound?

7. What are the risks associated with CVS?

8. What are the risks associated with amniocentesis?

9. What is a false positive? What is a false negative? What is sensitivity of a test? What is specificity of a test?

10. For each of the procedures listed in Question 2, what are their rates of false positives and false negative?

11. What might family and health history reveal that would indicate prenatal genetic screening may be appropriate or recommended for certain clients?

12. What is an inherited disorder? How is it passed down between generations?

13. What is the difference between recessive and dominant genetic disorders?

14. Why might pregnant individuals and their partners (or the people who gave the genetic makeup for the embryo) choose to pursue their own genetic screening?

15. Why might pregnant individuals and their partners (or the people who gave the genetic makeup for the embryo) choose to pursue ancestry-based carrier screening?

16. Who is at greater risk for the following inherited disorders?

1. Tay-Sachs Disease
2. open neural tube defects
3. Cystic Fibrosis
4. thalassemias
5. Sickle Cell
6. trisomy disorders
7. phenylketonuria
8. galactosemia
9. maple syrup urine disease

17. What is trisomy 21 commonly called?

18. What are you likely to observe after birth in a baby with Down Syndrome?

19. List some conditions that cannot be detected with prenatal genetic screening.

20. What is the risk in any pregnancy that a baby will be born with some type of disorder or genetic condition?

21. Psychologically, what are some of the benefits and drawbacks of genetic testing?

22. If a client chooses to terminate their pregnancy after genetic screening...

1. describe the procedures available in the first trimester.
2. describe the procedure available in the second trimester
3. What are your local resources for pregnancy termination after prenatal screenings? Can they be done at the local hospital? Separate clinic? Does the client need to go out of state?
4. What does it cost? Is it covered by insurance?

23. How do you support a client who decides to carry a fetus with known lethal fetal anomalies? How might their birth plans and options look different? How might their prenatal and postpartum care look different?

24. How do you support a client who decides to carry a fetus with known non-lethal fetal anomalies? How might their birth plans and options look different? How might their prenatal and postpartum care look different?

25. What does genetic screening cost in your area? Who pays for it? Is it covered by insurance?

26. Which screening tests and procedures listed in Question 2 can you perform as a midwife? Do you know where to refer clients for tests you cannot perform?

27. What is the term “disability justice”? Describe the tension between prenatal genetic screening and the disability justice community.

Continued…..

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*Long Answer Questions for Deeper Reflection*

Questions Requiring Longer, More Thoughtful Answers:

(number continued from previous section).

28. Explain all the options for genetic testing, in order of when they are offered by gestational age, as you would present it to clients.

29. Describe the use of informed choice as it applies to Prenatal Genetic Screening.

30. At what point do you discuss the possibility of terminating a pregnancy with your client? How do you present this information?

31. What is your personal bias on genetic testing? How might that impact your genetic screening counseling?

32. What is your personal bias on pregnancy termination? How might that impact your genetic screening counseling?

33. What is your personal bias on continuing to carry a pregnancy with known lethal or non-lethal fetal anomalies? How might that impact your genetic screening counseling?

Continued………..

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*Projects/Learning Activities*

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

(number continued from previous section).

34. Select or create an information sheet to provide to clients regarding options for

prenatal genetic screening.

35. What resources does your local state/province/jurisdiction provide to families for genetic counseling and testing, including primary information, follow-up testing, financial resources, and more?

36. Identify the regional lab and ultrasound facility that provides supplies, processing and/or services for each procedure each of the following:

1. Early Ultrasound
2. Nuchal Translucency Screen
3. Fetal Anatomy Scan
4. Cell-free DNA Testing
5. Ancestry-Based Carrier Screening
6. First Trimester Screen or Integrated Screen
7. Triple Marker / Quad Screen
8. Chorionic Villus Sampling
9. Amniocentesis

37. Using the online resources listed on the NMI Website, explore more stories from families who have used different kinds of genetic testing. Write about your reactions to the stories. Did your ideas about genetic testing change after reading these personal accounts? Do you feel strongly about genetic testing?

38. Select or create a client information sheet about pregnancy termination after fetal diagnosis. Understand where clients can go for termination procedures, testing, and follow-up care. Include support groups, hotlines, and other emotional support resources.

39. Select or create a client information sheet about continuing to carrying a pregnancy with known fetal anomalies. Provide community resources and support for pregnancy, birth, and postpartum with

1) lethal fetal anomalies, and

2) non-lethal fetal anomalies.

Also include ongoing community resources for parenting differently-abled children.

40. Draft practice guidelines for genetic counseling and screening in your own practice. Include reference to your informed choice practices, and your referral practices to pregnancy termination when requested. Submit this draft along with this module, and include it later in your Practice Guidelines projects (in the Charting and Practice Guidelines Module.)

41. Draft practice Guidelines for prenatal, birth, and postpartum care for a client carrying a fetus with known

1) lethal fetal anomalies, and

2) non-lethal fetal anomalies.

Include reference to your informed choice practices and transport guidelines. Submit this draft along with this module, and include it later in your Practice Guidelines projects (in the Charting and Practice Guidelines Module.