

**Course Title: MOD1-SOC102-Genetic Screening****Credits:** 1.00

**Course Description:** Covers the identification of mothers for whom risk screening is indicated, and the types of risk screening available in the U.S., along with their appropriate timing, reliability, and risks. Common genetic defects are covered, as are defects that cannot be identified by screening.

**Learning Objectives:**

A: Students will be able to answer the Study Questions (below).

B: Student will be able to demonstrate thorough knowledge of the Clinical Skills required for this course (below).

C: Student will demonstrate thorough knowledge of the MANA Core Competencies for Midwives required for this course (below).

D: Students will be able to demonstrate knowledge of any new information in the area of study.

**Learning Activities:**

I. Student Reads required texts.

II. Student Completes study questions.

III. Preceptor elaborates on study questions.

IV. Clinical Skills and Core Competencies training consists of the following (may take place at clinical visits):

1. Preceptor Explanation of	Safe, evidence-based midwifery care for the individual Clinical Skills and Core Competencies including etiology, sequelae, appropriate management and follow-up for the individual patient, appropriate times and reasons for consult and referral, access to relevant resources and information, complete, thorough and timely record keeping, appropriate, professional, and compassionate management of every task involved, receptiveness and responsiveness to patient's concerns. The Explanation will include a discussion of midwifery decisions and actions as they relate to possible outcomes and their wider impact, based on the Midwives Model of Care®.
2. Preceptor Demonstration of	
3. Student Practice of	
4. Student Demonstration of	

V. Student researches and presents to the preceptor relevant latest developments in academic and clinical midwifery.

VI. Student/Preceptor discussion.

**Learning Materials / Resources:**

(Please use textbooks less than 5 years old, or most recent edition)

1. Dudek, Ronald W. High-Yield Embryology. Lippincott Williams & Wilkins. 2009.
2. Weaver, Pam and Evans, Sharon K. Practical Skills Guide for Midwifery, 4<sup>th</sup> Edition. Morningstar Publishing Co. Wasilla. 2007.
3. MEAC Abbreviated NARM Skills Form.
4. MANA Core Competencies for Midwives
5. Midwives Model of Care®.
6. Internet links as needed for latest developments in midwifery care:  
[The Cochrane Collaboration](#)

[EBSCO](#)  
[National Library of Medicine](#)  
[PubMed](#)  
[Medline](#)  
[SCIRUS](#)  
[Medscape](#)  
[World Health Organization](#)

### **Evaluation Tools / Methods:**

1. Answers to study questions: Student must achieve at least 80% correct to pass. The preceptor evaluates each answer for correctness and explains the questions that were incorrect. This counts for 85% of the final grade.
2. Clinical Skills: Student must demonstrate thorough knowledge of each skill. This counts for 5% of the final grade. *Academic courses CAN be completed without the student achieving "mastery" of each skill, however the skills on the MEAC Abbreviated NARM Skills Form (which is a separate requirement) are not filled in until the student achieves Mastery\* of the skill.*
3. MANA Core Competencies: Student's ability to apply MANA Core Competencies for Midwives in discussion to simulated and real-life situations. This counts for 5% of the final grade.

Evaluation of NARM Skills and MANA Core Competencies: The student demonstrates thorough knowledge to the satisfaction of the preceptor in the following areas:

*The student will be able to, in accordance with safe, evidence-based midwifery care, explain the condition, verbalize etiology and sequelae, verbalize appropriate management for the individual patient, follow up appropriately, consult and refer appropriately, access resources and information, accomplish complete, thorough and timely record keeping, appropriately manage every task involved correctly, professionally, and compassionately, while being receptive and responsive to patient's concerns. She/he will be able to explain her decisions and actions as they relate to possible outcomes and their wider impact.*

4. Student presentation of new information in area of study. The preceptor evaluates the correctness of the information presented. This counts for 5% of the grade.

### **Study Questions**

173. Explain:
  - a. Triploidy
  - b. Tetraploidy
174. Describe Aneuploidy. Give the characteristics of the following:
  - a. Trisomy 13 (Patau's syndrome)
  - b. Trisomy 18 (Edward's syndrome)
  - c. Trisomy 21 (Down syndrome)
  - d. Klinefelter's syndrome
  - e. Turner's syndrome

175. Describe what is meant by a structural chromosomal abnormality. Give one example of each of the following: (Include the causes and the characteristics of each).
- Deletions
  - Microdeletions
  - Translocations
  - Fragile Sites
  - Isochromosomes
  - Inversions
  - Breakage
  - Photomicrograph
176. Describe Autosomal Dominant Inheritance, and give an example.
177. Describe Autosomal Recessive inheritance and give an example.
178. Describe X-Linked Recessive inheritance and give an example.
179. Describe Mitochondrial inheritance and give an example.
180. Describe what is meant by family pedigree and give draw a graphic representation of one.
181. Explain what is meant by Multifactorial inherited diseases, and give an example.
182. Explain Teratogen. Describe the resistant period, the maximum susceptibility period, and the lowered susceptibility period.
183. Describe the effects of the following diseases on the fetus:
- Rubella virus (German measles)
  - Cytomegalovirus (CMV)
  - Herpes virus type 2 (HSV-2)
  - Varicella zoster virus (VZV; chickenpox)
  - Himan immunodeficiency virus (HIV)
  - Non-viral infections:
    - Toxoplasma gondii (a protozoan parasite)
    - Treponema pallidum (a spirochete)
184. Describe what is meant by a Category X drug.
185. Give the uses for and the effects on the fetus of the following drugs:
- Thalidomide
  - Aminopterin and methotrexate

- c. Busulfan (Myleran), chlorambucil (leukeran) and cyclophosphamide (Cytoxan)
  - d. Phenytoin (Dilantin)
  - e. Triazolam (Halcion) and estazolam (ProSom)
  - f. Warfarin (Coumadin)
  - g. Isotretinoin (Accutane)
  - h. Clomiphene (Clomid)
  - i. Diethylstilbestrol
  - j. Ethisterone, norethisterone, megestrol (Megace)
  - k. Ovcon, Levlén, Norinyl
  - l. Nicotine
  - m. Alcohol
186. Describe what is meant by a Category D drug.
187. Give the uses for and the effects on the fetus of the following drugs:
- a. Tetracycline
  - b. Streptomycin
  - c. Penobarbital (Donnatal), pentobarbital (Nembutal)
  - d. Valproic acid (Depakene)
  - e. Diazepam (Valium), chlordiazepoxide (Librium), alprazolam (Xanax), lorazepam (Ativan)
  - f. Lithium
  - g. Chlorothiazide (Diuril)
188. Describe the effects on the fetus of the following chemical agents:
- a. Organic Mercury
  - b. Lead
  - c. Polychlorinated biphenyls (PCBs)
  - d. Potassium
189. Describe the effects on the fetus of the following recreational drugs:
- a. Lysergic acid diethylamide (LSD)
  - b. Marijuana
  - c. Caffeine
  - d. Cocaine
  - e. Heroin
  - f. Methadone
190. Describe the effects to the fetus of the following ionizing radiation:
- a. Acute high dose radiation (>250rads)
  - b. Diagnostic radiation
191. Explain which mothers are generally identified as needing counseling for genetic screening and give the rationale for each.

192. Describe 3 types of genetic screening available in the USA. Tell how they are done, why timing of each is important, when each is generally done, give the reliability of each test, and the risks of each test.
193. Name 4 common genetic defects and describe their effects on the fetus.
194. Explain which kinds of birth defects will not be detected by genetic screening.

### Clinical Skills (NARM Skills)

#### II. General Healthcare Skills

(31)-II D 8. Gestation calculation wheel/calendar

#### III. Maternal Health Assessment

(71)-III D. Estimates due date based upon a variety of methods

### Core Competencies (MANA Core Competencies for Midwives)

#### III. Care During Pregnancy

The midwife provides health care, support, and information to women throughout pregnancy. She determines the need for consultation or referral as appropriate.

The midwife uses a foundation of knowledge and/or skill which includes the following:

3 H. Basic understanding of genetic factors, which may indicate the need for counseling, testing, or referral.

3 J. Indications for, risks, and benefits of bio-technical screening methods and diagnostic tests used during pregnancy.