

# GENETICS ROTATION OBJECTIVES

## MATERNAL-FETAL MEDICINE FELLOWSHIP

University of New Mexico

1. **General Description:** UNM MFM fellows rotate through genetics during their PGY5 and PGY7 years. The PGY5 fellow works closely with our genetic counsellors during the clinic/wards rotation, and learn to take genetic histories, construct pedigrees, and offer screening and diagnostic tests. In addition, fellows participate in invasive prenatal testing, if undertaken, and counsel patients before and after these tests. During the PGY7 year, the fellows' continuity clinic is a genetics, prenatal diagnosis, and ultrasound clinic, designed to maximize fellows' experiences in counseling, invasive and non-invasive prenatal diagnosis technologies, and ultrasound correlates.
2. **Goals:**
  - a. To enhance the experience and education of the fellow on the medical knowledge and clinical skills that will enable the fellow to appropriately order, perform, and interpret genetic studies.
  - b. To gain an understanding of patterns of inheritance
  - c. To understand dysmorphology
  - d. To develop clinical competence in obtaining and analyzing pedigrees
  - e. To offer non-directive counseling.
  - f. E. To understand and perform non-invasive and invasive prenatal diagnostic technologies.
3. Learning **Objectives** upon completion of the rotation.

PGY5

A. **Medical Knowledge:** Upon completion of this rotation, fellows will be able to:

1. Understand and describe patterns of inheritance.
  - a. To be able to describe chromosomal duplication and deletion syndromes
  - b. Define: late manifestation, incomplete penetrance, variable expression, phenocopy, genetic heterogeneity, and gene-linkage in human disease
  - c. Discuss significance of new mutations
  - d. Define the Hardy-Weinberg law
2. Understand and describe the following cytogenetic principles
  - a. Cell cycle
    - i. Meiosis
    - ii. Mitosis
  - b. Understand and describe full karyotype analysis
    - i. Derivation and significance of X and Y chromatin
    - ii. Normal karyotype

- iii. Chromosome nomenclature
  - c. Understand and describe FISH
  - d. Understand and describe principles of cell free DNA analysis and limitations
  - e. Understand and describe advantages and limitations of microarray technology
- 3. Describe the following chromosomal abnormalities
  - a. Phenotypes associated with trisomies 13, 18, and 21
  - b. Deletion syndromes – Di George, etc
  - c. Significance of translocations and translocation carrier status
- 4. Discuss chromosomal anomalies prevalent in spontaneous abortions
- 5. Discuss evaluation and management of couples with recurrent spontaneous abortions
- 6. Discuss recurrence risk for couples with a history of chromosomal anomalies

**B. Patient Care:** Upon completion of this rotation, fellows will

1. Perform and/or interpret genetic tests: amniocentesis, karyotype analysis, microarray analysis, cell free DNA analysis, first trimester screening, sequential screening, second trimester screening. Perform and or interpret the genetic sonogram.
2. Counsel patients on screening and diagnostic testing options for chromosomal and single gene disorders.
  - a. Cite published test characteristics (sensitivity, specificity, positive and negative predictive values.
  - b. Explain strengths and limitations of all testing modalities including risk estimates for invasive procedures.
3. Evaluate the level of evidence for test characteristics and (for invasive procedures) complication rates
4. List costs of testing options
5. Interpret diagnostic tests and demonstrate knowledge of the following for each test:
6. Standard terminology, normal values, and test reliability

**C. Practice-based Learning:**

1. Demonstrate ability to perform self-assessment and incorporate feedback into improving clinical practice
2. Critically analyze and understand the appropriate use of the diagnostic studies and clinical management plans formulated and identify areas for improvement
3. Use information technology to locate scientific studies from literature on the various diagnostic tests and management strategies for medical complications of pregnancy and pregnancy-specific conditions and apply these to improve practice and patient care

**D. Interpersonal and Communication Skills:**

1. Demonstrate ability to communicate test results to patients and families
2. Use effective listening skills to elicit and then provide information to patients and families
3. Work effectively, interact, and communicate appropriately with referring providers.

**E. Professionalism:** Fellows are expected to:

1. Demonstrate a commitment to carrying out professional responsibilities, adherence to ethical principles, and sensitivity to a diverse patient population

2. Demonstrate respect, compassion, and integrity in interactions with patients, families, and other health care professionals
3. Demonstrate sensitivity and responsiveness to patient's culture, age, gender, and disabilities
4. Understand and perform non-directive genetic counseling

**F. Systems-Based Practice:**

1. Practice cost-effective healthcare and demonstrate knowledge of resource allocation that does not compromise quality of care, especially in the use genetic tests
2. Coordinate patient care among different health care providers, including medical students, residents, nurse practitioners, certified midwives and referring family medicine and general obstetrics and gynecology physicians.

PGY7

**A. Medical Knowledge:** Upon completion of this rotation, fellows will be able to:

1. Understand and discuss invasive prenatal diagnosis methods:
  - a. Techniques
  - b. Risks
  - c. Limitations
2. Discuss and describe indications for invasive prenatal diagnosis
3. Understand and discuss use of cell free DNA
  - a. Advantages, limitations, indications
4. Discuss molecular genetic analysis techniques for mutation detections
5. Discuss ethical considerations in prenatal diagnosis
6. Discuss strengths and limitations of the following modalities in prenatal diagnosis
  - a. Ultrasound
  - b. MRI
7. Discuss strengths and limitations of advanced fetal therapeutic techniques
  - a. Fetoscopy
  - b. Fetal blood sampling
  - c. Fetal surgery
8. Discuss technique and expected efficacy of preimplantation genetic diagnosis
9. Discuss screening and counseling of at risk populations for : neural tube defects, chromosomal anomalies, cystic fibrosis, Canavan disease, Tay-Sachs disease, Hemoglobinopathies, and Fragile X
10. Discuss components of preconception counseling

**B. Patient Care:** and procedures will

1. Perform and/or interpret genetic tests: amniocentesis, CVS, karyotype analysis, microarray analysis, cell free DNA analysis, first trimester screening, sequential screening, second trimester screening. Perform and or interpret the genetic sonogram.
2. Counsel patients on screening and diagnostic testing options for chromosomal and single gene disorders.

- a. Cite published test characteristics (sensitivity, specificity, positive and negative predictive values).
- b. Explain strengths and limitations of all testing modalities including risk estimates for invasive procedures.
- c. Evaluate the level of evidence for test characteristics and complication rates
- d. Understand indications and contraindications for advanced fetal therapeutic techniques and evaluate the level of evidence.
- e. List costs of testing options
- f. Interpret diagnostic tests and demonstrate knowledge of the following for each test:
- g. Standard terminology, normal values, and test reliability
- h. Evaluate cost-effectiveness for fetal therapeutic techniques

**C. Practice-based Learning:** Fellows are expected to:

1. Demonstrate ability to perform self-assessment and incorporate feedback into improving clinical practice
2. Critically analyze and understand the appropriate use of the diagnostic studies, invasive fetal therapeutics, and clinical management plans formulated and identify areas for improvement
3. Use information technology to locate scientific studies from literature on the various diagnostic tests, fetal therapeutic techniques and management strategies for medical complications of pregnancy and pregnancy-specific conditions and apply these to improve practice and patient care

**D. Interpersonal and Communication Skills:** Fellows are expected to:

1. Demonstrate ability to communicate test results to patients and families
2. Use effective listening skills to elicit and then provide information to patients and families
3. Work effectively, interact, and communicate appropriately with referring providers.

**E. Professionalism:** Fellows are expected to:

1. Demonstrate a commitment to carrying out professional responsibilities, adherence to ethical principles, and sensitivity to a diverse patient population
2. Demonstrate respect, compassion, and integrity in interactions with patients, families, and other health care professionals
3. Demonstrate sensitivity and responsiveness to patient's culture, age, gender, and disabilities
4. Understand and perform non-directive genetic counseling

**F. Systems-Based Practice:** Fellows are expected to:

1. Practice cost-effective healthcare and demonstrate knowledge of resource allocation that does not compromise quality of care, especially in the use genetic tests
2. Coordinate patient care among different health care providers, including genetic counselors, neonatologists, pediatric subspecialists, pediatric surgery

subspecialists, pediatric cardiologists, etc. Organize care conferences when indicated for mother: fetal pairs with complex congenital malformations.