# NMNEC Concept: **Genomics**

**Mega Concept:** Health and Illness

**Concept Category:** Homeostasis & Regulation

**Concept Name:** Genomics

**Concept Definition:**
Genomics is the "study of all the genes in the human genome together, including their interactions with each other, the environment, and the influence of other psychosocial and cultural factors" and genetics is the "study of individual genes and their impact on relatively rare single gene disorders" according to the World Health Organization (American Nurses' Association, 2008, p. 8-9). Another useful definition is epigenetics, which "refers to the variations in phenotype that occur due to the influence of the environment and our lifestyle on genetics" (Perry, et al, 2018, p. 123).

**Scope and Categories**:
All diseases and conditions have a genetic or genomic component. Health care options for all persons will increasingly include genetic and genomic information for prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness (American Nurses' Association, 2008).
Genomics includes conditions that may develop during conception and pregnancy, i.e., Down syndrome, cystic fibrosis, or sickle cell disease. Also, the concept includes the presence of or susceptibility to adult onset conditions, i.e., hereditary colorectal cancer or Huntington's disease (Perry, et al, 2018).

**Populations at Risk and Risk Factors:**
All individuals are at risk for spontaneous genetic changes. Those at higher risk for passing on or developing a genetic condition are pregnant women of advanced age or those with a family history of a genetic condition (Perry, et al, 2018). An individual may have known or unknown genetic alterations or conditions.

Non-modifiable Risk Factors:

- Family history of a genetic condition
- History of genetic alterations or conditions

Modifiable Risk Factors:

- Family history of a genetic condition may increase risk of developing a genetic condition but does mean that an individual will get the condition. Lifestyle changes, such as exercising, healthy nutrition, or not ingesting nicotine, may lower risk for developing the condition (Green, 2014).
- Individual with genetic alterations or conditions may have interventions, such as medication, surgery (e.g., mastectomy for individuals with BRCA1 mutation) or earlier or more frequent screenings, can lower risk for developing or minimizing impact from the genetic alteration or condition (Green, 2014).

**Physiological Processes and Consequences:**

The process begins with meiosis, which produces gametes in male and females. During conception, the egg and sperm unite to form 23 pairs of chromosomes (Perry, et al, 2018). A unique individual develops, who passes their genetic material on to the next generation. Environment influences, i.e., radiation, may alter an individual's genes or genetic changes may occur spontaneously. Patterns of genetic transmission include unifactorial inheritance, which is controlled by a single gene. These conditions may result from autosomal dominant, autosomal recessive, or X-linked inheritance. Multifactorial inheritance occurs as a result of two or more genes on different chromosomes. Most common congenital conditions, such as cleft lip, result from a combination of genetic and environmental factors (Perry, et al, 2018).

**Assessment:**

History – The nurse obtains a family history of at least three generations and develops a pedigree (genogram). This may illustrate the patterns of inheritance for those at high risk for developing genetic-related disorders such as heart disease, stroke, diabetes, or cancers (Lewis, Bucher, Heitkemper, and Harding, 2018).

Physical Examination – The nurse assesses for dysmorphic facial features as well as cognitive or behavioral characteristics, such as an unusual cry of a newborn (Perry, et al, 2018).

**Diagnostic Tests:**

There are tests for single-gene disorders in patients with clinical symptoms or for those who have a family history of a genetic disease. A karyotype is a pictorial analysis to illustrate the number, form, and size of an individual's chromosomes. Pharmacogenomic testing can be used to guide a patient's drug therapy, i.e., warfarin, or to target therapies, such as drug therapy for a genetic variant of breast cancer (Perry, et al, 2018).

**Planning:**

The Healthy People 2020 goals for genomics are to: "Improve health and prevent harm through valid and useful genomic tools in clinical and public health practices." The two objectives are based on two recommendations for independent panels on genetic testing. The recommendations are: "Women with certain high-risk family health history patterns for breast, ovarian, tubal, or peritoneal cancer could benefit from receiving genetic counseling to learn about genetic testing for BRCA1/2." and "All people who are newly diagnosed with colorectal cancer should be offered testing for a hereditary form of colorectal cancer called Lynch syndrome."(Healthy People 2020, 2019a). Related maternal, infant, and child health objectives are to: "Increase the proportion of pregnant women who receive early and adequate prenatal care" and "Increase appropriate newborn bloodspot screening and follow-up testing" (Healthy People 2020, 2019b).

**Nursing Diagnoses:**

Possible nursing diagnoses are deficient knowledge or compromised family coping (Ladewig, Ackley, and Makic, 2017).

**Clinical Management: (Interdisciplinary)**

**Primary:** Prevention

Nurses can teach all women of reproductive age how to avoid teratogens. Also, nurses can teach all individuals to avoid environmental exposures, such as excessive sunlight, which could cause genetic changes.

**Secondary:** Screening

Genetic testing includes procedures to analyze chromosomes, genes, or gene products that can determine whether a mutation or predisposition to a condition exists. This may be done for carriers, preimplantation, prenatal, newborn, or parental testing (Lewis, et al, 2017). Prenatal screening could include maternal serum screening, ultrasound, or invasive procedures, such as amniocentesis. All states require that newborns are screened for inborn errors of metabolism, such as phenylketonuria (Perry, et al, 2018). In addition, presymptomatic, diagnostic, predictive, pharmacogenonmic, or forensic genetic testing may be done (Lewis, et al, 2017). Nurses provide general information about genetic testing and screening, the risk of developing a disease, and passing that risk to offspring (Carranti, 2017, p. 419). Nurses can assist families with the many psychologic, emotional, and ethical issues associated with genetic testing (Lewis, et al, 2017).

**Tertiary:** Treatment

"Genetic counseling is a professional service that provides genetic information, education, and support to individuals and families with ongoing or potential genetic concerns" (Perry, et al, 2018, p. 130). The genetic counseling team may discuss options for reproductive technologies or clinical trials for gene therapies (Perry, et al, 2018).

Pharmacogenetic and pharmacogenomic studies can potentially lead to drugs that can be adapted to each person's genetic makeup. This makes it possible to have personalized medicine by choosing the right drug and the right dose for the right person. Also, gene therapy and stem cell therapy may use used by the health care team in treatments, such bone marrow transplants (Lewis, et al, 2017). The interdisciplinary health care team would be involved in the rehabilitation of those who need ongoing multiple therapies, i.e., individuals with cystic fibrosis.

**Interrelated Concepts:**

- Cellular Regulation
- Development
- Ethics
- Health Promotion
- Patient Education
- Reproduction

**Exemplars:**

**New Mexico Nursing Education Consortium (NMNEC) Required Exemplars:**

- Cystic Fibrosis (CF):
  CF affects about 30,000 children and adults in the United States. It is one of the most common genetic conditions in this country. About 1 in 3,500 babies is born with CF. (March of Dimes. 2013)

**Optional Exemplars:**

- Newborn Screening for Inborn Errors of Metabolism (or Inborn Errors of Metabolism, phenylketonuria (PKU), or Sickle Cell Disease):
  In the United States, about 1 in 10,000 to 15,000 babies is born with PKU each year. SCD affects about 1 in 500 black births and about 1 in 36,000 Hispanic births in this country. (March of Dimes, 2016)

**References:**

American Nurses Association. (2008). *Essentials of genetic and genomic nursing: Competencies and curricula guidelines, and outcome indicators* (2nd ed.). Silver Spring, Maryland: Author.

Carranti B. (2017). Patient Education. In J. F. Giddens, J. F. (Ed.). *Concepts for nursing practice* (2nd ed., pp. 414-419). St. Louis, MO: Elsevier.

Green, R. F. (2014, November 13). Family health history is a non-modifiable risk factor—or is it? [Web log post]. Retrieved from https://blogs.cdc.gov/genomics/2014/11/13/family-health-history/

Healthy People 2020 (2019a). *Genomics*. Retrieved from Whttps://www.healthypeople.gov/2020/topics-objectives/topic/genomics

Healthy People 2020 (2019b) Maternal, Infant, and Child Health Objectives. Retrieved from https://www.healthypeople.gov/2020/topics-objectives/topic/maternal-infant-and-child-health/objectives

Ladewig, G. B., Ackley, B. J., and Makic, M. F. (2017). *Mosby's guide to nursing diagnosis* (5th ed.). St. Louis, MO: Elsevier.

Lewis S. (2017). Genetics and Genomics. In Lewis, S. L., Bucher, L., Heitkemper, M. M., and Harding, M., M. *Medical-surgical nursing*, (10th ed., pp. 178 - 190). St. Louis, MO: Elsevier.

March of Dimes. (2013). *Health topics; Cystic fibrosis and your baby.* Retrieved from https://www.marchofdimes.org/complications/cystic-fibrosis-and-your-baby.aspx

March of Dimes. (2016). *Health topics; Newborn screening tests for your baby.* Retrieved from https://www.marchofdimes.org/baby/newborn-screening-tests-for-your-baby.aspx

Perry, S. E., Lowdermilk, D. L., Cashion, K., Alden, K. R., Olshanky, E. F., Hockenberry, M. J., Wilson, D., and Rodgers, C. C., (2018). *Maternal Child Nursing Care* (6th ed.). St. Louis, MO: Elsevier.

**Resources:**

American Association of Colleges of Nursing. (2008). *The essentials of baccalaureate education for professional nursing practice*. Retrieved from www.aacnnursing.org/Education-Resources/AACN-Essentials>

Daack-Hirsch, S., Dieter, C. D., and Griffin, M. T. (2011). Integrating genomics into undergraduate nursing education. *Journal of Nursing Scholarshi*p. DOI: org/10.111/j.1547-5069.2011.01400.x

National Institutes of Health. (2017). *National human genome research institute; An overview of the human genome project*. Retrieved from https://www.genome.gov/12011238/an-overview-of-the-human-genome-project/

March of Dimes. (2013). *Health topics; Cystic fibrosis and pregnancy.* Retrieved from https://www.marchofdimes.org/complications/cystic-fibrosis-and-pregnancy.aspx

**Websites:**

Genetics/Genomics Competency Center at https://genomicseducation.net/

International Society of Nurses in Genetic (ISONG) at https://www.isong.org/