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Genetics & Genomics in Nursing

(Previously known as Human Genetics in Nursing)

Suresh K Sharma

3rd Edition

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Chapter

2 Emerging Paradigm of Genetics in Nursing

Learning Objectives

This chapter is designed to enable the reader to:

- List out the milestones of genetics nursing practices
- Describe the role of nurses in genetics
- Appraise the importance of genetics in nursing curriculum
- Recognize the barriers and approaches for implementation of genetics in nursing
- Discuss the assumptions regarding genetics and health care in future.

Key Terms

- Genetics Nursing
- Genetics Nurses
- Genetics Nursing Network
- Genetics Advanced Practice Nurses
- International Society of Nurses in Genetics
- Genetics Nursing Credentialing Commission

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INTRODUCTION

In the last few years, genetic nursing practice has been transformed from a nearly hidden specialty to a recognized specialty practice with formal recognition, publication of scope and standards of practice, and most recently, the availability of credentialing for genetic nurses. All areas of nursing practice have been impacted by recent advances in genetic knowledge and technology. Nearly all diseases are now recognized to have a genetic component.

Nurses now provide education to patients about hereditary risk for developing disease, counsel about the benefits and risks associated with genetic testing, and manage disease risk based on genetic information.

The recent development of commercial testing for susceptibility genes (such as the predisposition genes for breast ovarian cancer syndrome and colon cancer) have had a great impact on nursing's role in the identification and management of individuals at risk for developing many diseases. These developments have led to tremendous changes in genetic nursing practice. Nurses have been involved in managing genetic information since the 1960s, when nurses provided services to children with genetic disorders and their families. Although in some respects the nurse's role today in managing genetic information and caring for individuals and families at risk for or diagnosed with genetic diseases or conditions is similar to this traditional role, the scope of practice is much broader and more encompassing. What has also changed, according to Forsman, is the amount of genetic information available and the population to which this information may be applied. We now know that genetic changes contribute to most, if not all, diseases. Consequently, the scope of genetic knowledge application in nursing is limitless. 'Nursing can ignore genetics no longer. The time for meaningful action is now'.

There is an opportunity for nursing to practice health promotion tailored to highly specific and uniquely personal predispositions; in other words, for individualized health care, nurses can offer care that protects patients and families from the risks associated with genetic information, including addressing family issues. Nurses are also needed to refer patients to genetic specialists and assist patients in making choices as a genetic health care. Every nurse is needed; do not miss the revolution. All nurses have a role in the delivery of genetics services and the management of genetic information. Nurses require genetic knowledge to identify, refer, support, and care for persons affected by or at risk for genetic conditions.

'Genetic nursing is a holistic practice that includes assessing, planning, implementing, and evaluating the physical, spiritual, ethical, and psychosocial aspects of patients and families who have genetic concerns'. Genetics nurses practice in many different environments. Genetic nursing practice includes client and family

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assessment to identify genetic risk factors and intervention, information, service, and referral needs, take a detailed family history and construct a pedigree, analyze the assessment data, provide genetic education, and develop and carry out a plan of care to address genetic concerns. In addition, nurses in genetics provide genetic counseling, order and interpret genetic tests if within their scope of practice, and provide surveillance and management of persons affected by or at risk for genetic conditions.

In oncology, for example, areas of responsibility begin with risk assessments for cancer in patients and family members. This assessment includes a detailed family history, construction of a pedigree, and assessment of hereditary, environmental and lifestyle cancer risk factors. Once these risk factors are determined, the information is interpreted to the patient. Recommendations for cancer prevention and early detection can be tailored to the specific risks identified. If genetic testing for a cancer susceptibility gene is appropriate, the individual/family should be informed about the risks and benefits associated with the test, and the potential risks and benefits associated with the screening and prevention measures. The nurse also needs to explore possible psychosocial responses to the potential outcomes of a genetic test. This education may be done individually or with several family members. If testing is pursued, results are disclosed in person. At that point, a tailored plan for prevention and early detection is written and given to the family or individual. Long-term follow-up includes assessment of adjustment to the test results and compliance and follow-through with the plan for cancer prevention and early detection.

GENETICS NURSING PRACTICE MILESTONES

Genetics nursing practice has seen several memorable milestones in less than five years. In the past, there has been minimal recognition of the important role that nurses can play in genetics. The International Society of Nurses in Genetics (ISONG) has clearly been the leader in working with nursing leaders to promote genetic nursing practice and develop a credentialing process for genetic nurses. Through these efforts, only five years ago, the American Nurses Association established genetics nursing as an official specialty of nursing practice.

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- *1960s:* Nurses published about the nurse's role in genetics and conducting genetics research
- *1984:* Genetics Nursing Network was formed, which later became ISONG
- *1988:* International Society of Nurses in Genetics (ISONG) was incorporated
- *1997:* Genetics Nursing designated an official nursing specialty by American Nurses Association
- *1998:* Statement on the Scope and Standards of Genetics Clinical Nursing Practice published by ISONG and ANA
- *2001:* ISONG approved formation of the Genetic Nursing Credentialing Commission (GNCC)
- *2001:* Credentialing of the first Genetics Advanced Practice Nurses [APNG(c)]
- *2002:* GNCC goes online at [www.geneticnurse.com] , develops a logo and becomes incorporated
- *2002:* GCN credential is first offered by GNCC.

ROLES OF NURSES IN GENETICS

Following are the main roles of nurses in the field of genetics:

- Planning, implementing, administering (managing), and evaluating screening and testing programs.
- Monitoring and evaluating clients with genetic disorders in a similar manner to other disorders working with families under stress caused by problems related to a genetic condition.
- Coordinating care and services for individuals or families affected by genetic conditions, managing home care and therapy for persons with genetic disease.
- Following up on positive newborn screening tests, interviewing clients with possible genetically related conditions.
- Assessing needs and interactions in clients and families affected by genetic disease.
- Taking comprehensive and relevant family histories.
- Assessing the client and family's cultural or ethnic health beliefs and practices as they relate to the genetic problem.
- Assessing the client and family's strengths and weaknesses and family functioning.

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- Providing health teaching and education related to genetics serving as an advocate for a client and family affected by a genetic disorder.
- Participating in public education about genetics.
- Developing an individualized plan of care, including anticipatory guidance for a person with a genetic condition.
- Explaining the purpose, meaning, and implications of genetic tests and results.
- Reinforcing and interpreting genetic counseling and testing information.
- Supporting families when they undergo counseling and make decisions.
- Recognizing the possibility of a genetic component in a disorder and taking appropriate referral action. Appreciating and ameliorating the social impact of genetic problems on the patient, family, and members of the community.

IMPORTANCE OF GENETICS IN NURSING CURRICULUM

Nursing programs traditionally include certain non-nursing courses, like general biology, microbiology, sociology/anthropology, ethics, pharmacology and genetics depending on the design of the curriculum.

Genetic concepts must be in all courses in the nursing program. They must be included in both didactic teaching and clinical applications. They must include practical and relevant examples with applications. Before making decisions about how and where to include genetic content in the nursing curriculum, it is important first to appraise the content to which students are actively exposed. In a nursing program, sometimes there is more genetic content than is realized. For example, when listing genetic content in a survey, one school did not include discussing cystic fibrosis as genetic under chronic illness for children. The first step is to meet with the instructors of courses both within nursing and outside of nursing and find out what content is already present. If a particular topic is not covered that the nursing program believes is important, they can request that it should be added. If the nursing program believes that a course in molecular biology or human genetics or an interdisciplinary course that focuses on genetics is needed,

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then they can work directly and collaboratively with the relevant department(s) to design a course that can be added to the basic nursing curriculum. It is also important that the nursing program examines what knowledge students should have retained from prior learning. It may not be necessary to reteach the complete content, such as meiosis, mitosis, and typical patterns of inheritance, etc.

Some other ideas for including genetic content within nursing curricula for both nursing and non-nursing courses are listed later with examples. Graduate programs can use similar ideas by providing more complexity and depth for topics and by incorporating genetic content at a higher level into the specialty courses. The exact nature depends on the area of specialization.

Important knowledge for the basic nursing graduate in regard to genetics includes the following:

- Basic mechanisms of inheritance and transmission of chromosomes and genes, including the concepts of variation and mutation. Nonmendelian mechanisms should be included. A major issue is how much of this should be retaught as it typically is in maternal child health or obstetrics and pediatrics? Should a separate course on molecular biology required for nursing be taught within the university or should all undergraduate students regardless of major have an interdisciplinary course that relates knowledge from biology, anthropology, philosophy, sociology, and other disciplines to genetically related issues?
- Information about the common inherited genetic disorders, for example, cystic fibrosis, Down's syndrome, phenylketonuria, sickle cell anemia, and hemochromatosis. Disorders that are particularly frequent in persons who reside in the geographic location of school should be included.
- An understanding of genetic contributions to human diversity including concepts, such as discrimination and eugenics.
- Ways of thinking about health and disease in relation to genetics, including how genetics and the environment interact and how genes interact with genes. This should lead to new ways of thinking about health promotion and disease prevention.

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- Information about genetic contributions to common and complex conditions, such as breast cancer, colorectal cancer, heart disease and hypercholesterolemia, mental illness, certain behavioral traits, and Alzheimer disease.
- Care management of adults with childhood genetic disorders.
- Care management of persons with adult genetic disorders, such as Huntington disease.
- An understanding of genetic approaches to the therapy of genetic and complex diseases.
- Interpretation of genetic risks (i.e. how to explain the meaning of a 1-in-4 risk for having another child with Tay-Sachs disease).
- What constitutes a proper family history specifically, what key information should be obtained and how to construct and read pedigrees.
- Information about genetic risk, genetic testing and screening, and the implications both positive and negative.
- How to interpret the results of genetic tests.
- Awareness of the possibility of an inherited or genetic component for a client's condition and knowledge of cardinal features of familial predisposition, such as early age of disease onset, multiple family members with the same diagnosis, predisposing risk factors.
- An awareness of social, legal, and ethical issues related to genetics, including effects on individuals, groups, and societies, some of which are unique to genetic conditions.
- An understanding of the social and ethical ramifications of possessing a particular genotype or genetic disorder in terms of societal issues, confidentiality, freedom of choice, and risks in terms of insurance and disclosure.
- Application of traditional nursing skills, such as patient education, confidentiality, and counseling to genetic information. The concept of nondirective counseling can be included.
- Ways in which genetic knowledge is used in diagnosis and treatment applications.
- Development of nonjudgmental attitudes about genetics and related disorders.

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- Ways to access resources relating to genetics for patient and self-education and the need to keep current.
- Knowledge about referral possibilities knowing not only who should be referred to but also how and to whom it should be done.
- The knowledge that responses to drugs used in therapy are at least in part genetically determined.
- Advanced practice nurses may play direct roles in genetic counseling and in advanced assessment. They also may work within a particular specialty in which genetics plays a role, such as an oncology or cardiology clinic, as well as in long-term management of specific genetic disorders depending on the specialty area in which they are trained. An example of the latter is the nurse who works in a sickle cell clinic. The graduate curriculum should include a higher level of the concepts discussed previously as well as advanced material related specifically to the specialization.

BARRIERS AND APPROACHES FOR IMPLEMENTATION OF GENETICS IN NURSING

Numerous barriers including genetic content in basic nursing education have been identified. The most commonly encountered barrier is lack of time. Knowledge continues to expand but, until recently, nursing programs have continued with essentially the same program length. The intensity of the curriculum has increased and has become more dense at a time when many students have less time to spend on learning or are less qualified than previously. Faculty barriers include perceived lack of knowledge, insecurity in their knowledge, not perceiving genetic content as important, and not recognizing the broad scope of genetics as it influences health care but treating the genetic approach as a collection of rare disorders to be memorized.

Potential approaches to addressing these barriers can include the following:

- *Educating faculty:* As the nursing faculty is aging, education in genetics may have taken place sometime ago and must be refreshed.

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- *Instilling confidence in faculty:* Faculty must be confident that they can understand material and are interpreting it correctly. This can be addressed through continuing education and workshops. Faculty must feel secure with content to teach it.
- Providing rewards and incentives for faculty.
- Expanding on what is already in the curriculum after determining what already exists.
- Using genetic examples that may be central or incidental. For example, a person who has a particular genetic condition could be the focus of a surgical case study in which the genetic condition is not central but the genetic information is included.
- Emphasizing a lifespan approach for genetic disorders.
- Emphasizing the relevance of genetic examples and using contemporary examples; for example, cloning or DNA testing as used in the OJ.
- Offering continuing education/workshops for faculty that may include hands on demonstrations.
- Seeking consultation/advice to get started with concrete examples.
- Recognizing need for frequent updating.
- Identifying an advocate or point person in school. This person may be educated to become a resident expert.
- Working with course instructors and the curriculum committee to achieve goals.
- Identifying resources for information about instruction and online information, courses available for faculty updating, core knowledge and curriculum developed for other disciplines and for teaching tools, such as genetic games and deconstructions that are not only informative but also fun. Many scientific supply houses have these products.

ASSUMPTIONS REGARDING GENETICS AND HEALTH CARE IN FUTURE

Some of the guiding principles or basic assumptions in relation to genetics inform that health care must be weighed before considering what core genetic knowledge is needed by the practicing nurses. These assumptions include the following:

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- Nurses in all areas of practice will encounter clients with traditional genetic disorders and common disorders with a genetic component.
- Most, if not all, disorders have a genetic basis to a greater or lesser extent, even conditions such as fractures. To a degree, a mutant gene can be thought of as an etiologic agent.
- Traditional genetic disorders are seen across the lifespan. Many inherited biochemical disorders have infant, childhood, and adult forms. Other disorders typically appear or are noticed in adulthood, such as Huntington disease and hemochromatosis.
- The previously existing genetic condition of a particular client influences the choice of care and treatment for any new health problem for which he or she is seeking consultation.
- The so-called common or complex diseases, such as cancer and heart disease have a genetic component evident in origin, diagnosis, treatment, management, and preventive approaches.
- Humans are diverse not only because of cultural and social factors but also because of genetic variation. This is not inherently bad or good but a fact that influences how we all respond to foods, drugs, medications, and chemicals in the environment and profoundly affects how people respond to health care interventions because all people have different degrees of possible health.
- Assessment for genetic risk for specific conditions preceding options of testing followed by counseling will become more prevalent and include persons at risk for common diseases, such as heart disease, various types of cancers, and Alzheimer disease, in addition to those already in common use, such as prenatal diagnosis.
- Technologic advances are such that persons with genetic disorders that appear in infancy or childhood and formerly caused early death are living not only to early adulthood but later. For example, some adults with sickle cell anemia have lived into their eighties. Health care providers must deal with how the genetic disorder impacts the appearance and management of common health problems and aging and vice versa. For example, how will co-illness with type 2 diabetes mellitus influence the course of disease and response in an

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adult with cystic fibrosis? How will common conditions such as hypertension be managed against the background of a previously existing genetic disease?

- Ways of thinking about health promotion and disease prevention will change.
- Many diseases will be diagnosed and treated using knowledge from genetics.
- Genetic information that influences health care through the lifespan will be available prenatally.

Genetic knowledge makes it possible to individualize aspects of health care such as prevention and treatment in new ways. In the not-too-distant future, it will be possible to predict the diseases for which a fetus or newborn will have a genetic predisposition, and a fetus or newborn will have a predicted genetic future that may be subject to modulation.

Genetics often has been associated with tertiary health care and rare genetic disorders in the minds of many nurses. Genetics plays and will continue to play a greater role in the common and complex disorders, such as various cancers, infectious diseases, Alzheimer disease, heart disease, and chronic obstructive pulmonary disease, and in relatively common genetic conditions that have significant organ effects, such as hemochromatosis. Clients who see primary care providers such as advanced practice nurses will seek genetically related information and may require assessment for genetic risk at multiple points in the lifespan from prenatal to the older age group. On the basis of such risk assessment, they may choose genetic testing. Studies have demonstrated that a significant percentage of health care professionals misinterpret the results of genetic testing. Often they may not know it is available for a condition relevant to their client or may not recognize that their client is at risk. The curriculum in nursing and other health care professions is deficient in genetic content.

REVIEW QUESTIONS

Short-note Questions

1. Mention genetic nursing practice milestones.
2. What is the role of nurses in genetics?

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3. Describe the importance of genetics in nursing curriculum.
4. Discuss the approaches to barriers in implementation of genetics in nursing.
5. Enumerate the assumptions regarding genetics and health care in future.

Short-answer Questions

1. List four recent important genetics nursing practice milestones.
2. List four essential roles of nurses in genetics.
3. List two reasons why genetics should be included in the nursing curriculum.
4. List two barriers of implementation of genetics in nursing.

Multiple Choice Questions (MCQs)

1. When did the first-time nurses publish about the nurse's role in genetics and conducting genetics research?
a. 1970 b. 1960
c. 1980 d. 1990

2. In which year was the International Society of Nurses in Genetics incorporated?
a. 1977 b. 1988
c. 1999 d. 2009

3. Following are the roles of nurses in genetic practices, except:
a. Planning and implementing the genetic screening tests.
b. Monitoring and evaluating the patients with genetics disorders.
c. Conducting the gene therapy on a patient with genetic condition.
d. Providing genetic counseling services to the patient and family.

4. Which of the following may be considered as the barrier in the implementation of genetics in nursing?
a. Lack of availability of genetics textbook.
b. Lack of available time in nursing program.
c. Lack of scope for genetic nurses in health care.
d. Lack of funds for support of genetics in nursing.

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Genetics & Genomics in Nursing

(Previously known as Human Genetics in Nursing)
Third Edition

The third edition of the book 'Genetics and Genomics in Nursing' is precisely written as per the Indian Nursing Council syllabus for BSc Nursing students. This textbook is an excellent attempt towards presentation of comprehensive, lucid, illustrative and example-oriented contents of genetics and genomics in nursing.

Salient Features

- ***Easy to follow:*** This is an applied, user-friendly and self-explanatory textbook with simple language and presentation for the students.
- ***An example oriented book:*** This is an example-oriented book, where numerous nursing examples are citied throughout the book for enhancing the knowledge about the applicability of genetics and genomics in nursing.
- ***An applied textbook:*** This book will assist the students to prepare for their exams as well as independently applying the knowledge of genetics and genomics in their nursing practices.
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- ***Authentic content:*** The content of this textbook is reviewed by a panel of experts in the field of genetics and genomics.



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