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INCORPORATING GENETICS AND GENOMICS NURSING INTO CLIENT CARE

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ABSTRACT

The burden of genetic diseases cuts across all continuums of care with its attendant economic, social, physical and psychological impacts on patients, families and their caregivers. Nurses remain the most easily accessible of all health care providers to the majority of persons who seek health care information in Nigeria. They are now expected to be aware of and provide information about various inherited disorders, as well as to recognize the numerous concerns associated with genetic testing. In order for people to benefit from widespread genetic/genomic discoveries, nurses must be competent to obtain comprehensive family histories and identify family members at risk of developing a genomic and genetic influenced conditions.

Essentially all diseases and conditions have a genetic or genomic component. Options for care for all persons will therefore increasingly include genetic and genomic information along the pathways of prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness. Hence, it is vital for nurses to possess a fundamental understanding of genetic and genomic information. This paper provides an explanation for incorporating genetics and genomics nursing into client care. It discusses brief overview of genes, history and professional nurses' responsibilities.

INTRODUCTION

Prior to the mapping of the human genome which began in 1988, nurses in most practice settings did not consider providing genetic health care as a routine aspect of nursing practice. However, with the completion of sequencing the human genome in 2003, foundation for a focus on genomics was laid[1]. Hence, during recent years, nurses have been asked increasingly to provide information about genetic testing and its implications. Nurses are now expected to think "genetically," that is, they should incorporate genetic knowledge and skills into their nursing care and they have many opportunities to integrate the latest genetic advances into their practice as direct care providers, consultants, administrators, educators, and researchers.

Genetic and genomic science is redefining the understanding of the continuum of human health and illness [2]. Therefore,

KEY WORDS: *Genetics, Genomics, Nursing, Client care*.

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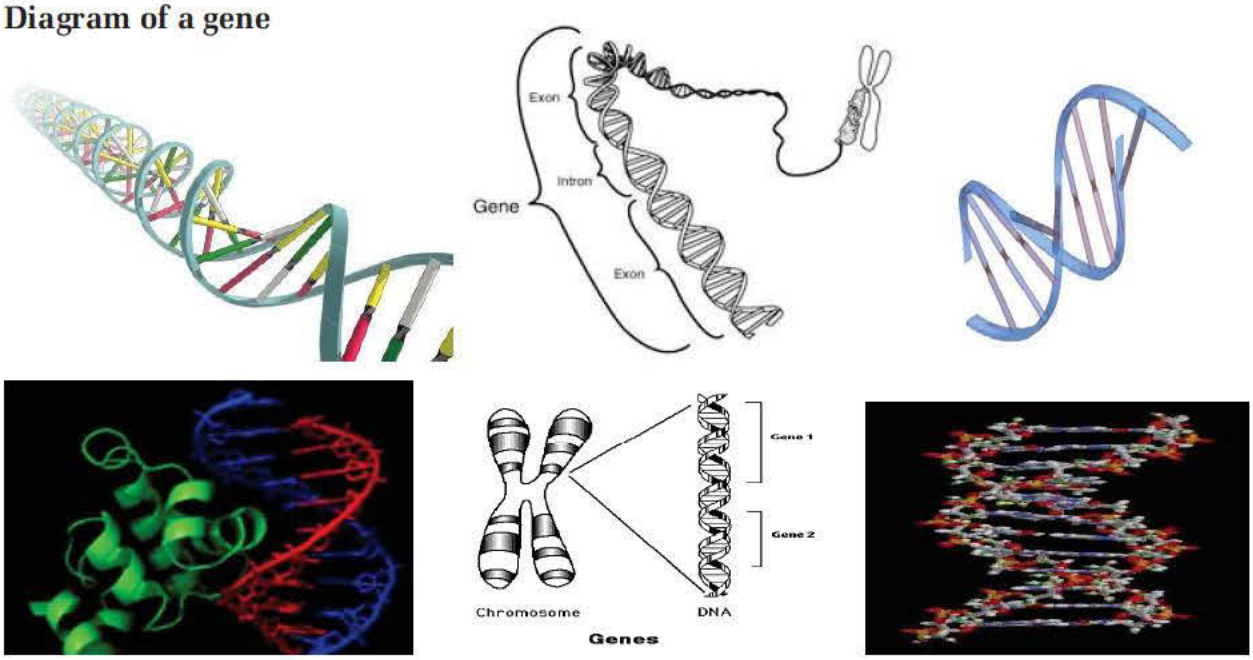
recognition of genomics as a central science for health professional's knowledge is essential because essentially all diseases and conditions have a genetic or genomic component. [2,3] Consequently, options for care for all persons will increasingly include genetic and genomic information along the pathways of prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness. The clinical application of genetic and genomic knowledge has major implications for the entire nursing profession regardless of academic preparation, role, or practice setting. In view of the afore-mentioned, the public will increasingly expect that the professional nurse will use genetic and genomic information when providing care.
The rate of progress for applying a genomic approach throughout the continuum of care depends not only on technologic advances, but also on nursing expertise. In its report on genetics and nursing in 2000, an expert Health Resources and Services Administration (HRSA) panel emphasized the importance of integrating genetics content into nursing curricula in order to provide an adequately prepared nursing workforce now and for the future[4]. To care for persons, families, communities and or populations throughout the life span, nurses will need to demonstrate proficiency with incorporating genetic and genomic information into their practice.

For example: nurses need to understand the genetic and genomic basis of health and/or an illness for which the person is seeking care and the variables that impact his or her response. They need to recognize a newborn at risk of morbidity or mortality resulting from genetic metabolism errors, identify an asymptomatic adolescent who is at high risk of hereditary colon cancer, identify a couple at risk of having a child with a genetic condition, guide interventions for the prevention of cardiovascular disease in young adults, promote informed consent that includes the risks, benefits, and limitations of participation in genetic research and assist anyone having questions about genetic and genomic information or services. The goal of clinical genetics and genomics is to improve the quality of health care for patients and families. Nurses knowledgeable about genetics/genomics and skilled at obtaining and assessing risk in a family history have the potential to help people avert adult onset disorders and consequent morbidity and mortality [5]. This paper provides a review of literature on incorporating genetics and genomics into client care. It discusses overview of genes, history and professional nurses' responsibilities in genetic and genomic health .

What is a Gene?
A gene is the basic unit of heredity in a living organism. All living things depend on genes. Genes hold the information to build and maintain their cells and pass genetic traits to offspring. A modern working definition of a gene is "a locatable region of genomic sequence, corresponding to a unit of inheritance, which is associated with regulatory regions, transcribed regions, and or other functional sequence regions ".
The notion of a gene is evolving with the science of genetics, which began when Gregory Mendel noticed that biological variations are inherited from parent organisms as specific, discrete traits. The biological entity responsible for defining traits was termed a gene, but the biological basis for inheritance remained unknown until DNA was identified as the genetic material in the 1940s. All organisms have many genes corresponding to many

different biological traits, some of which are immediately visible, such as eye color or number of limbs, and some of which are not, such as blood type or increased risk for specific diseases, or the thousands of basic biochemical processes that comprise life. Mendel demonstrated that inheritance occurs through genes, unit of heredity that maintains their structural identity from one generation to another. As a rule, genes are in pairs because they are aligned along chromosomes (strands of genes), which also come in pairs. (As an exception to this rule, a male's X and Y chromosome are unpaired, having different genes.)

Diagram of a gene



Source: National Health Museum [6]

Genetics is another foundation of human behavioral pattern. The extent to which personality, traits, or characteristics is determined by our genetic composition is termed hereditary. Many aspects of behavior are influenced by genetics [7, 8, 9] Reactions and cognitive abilities are all significantly influenced by genetic inheritance [10, 11]. This explains why some people are naturally friendly or aggressive. Some cope better with stress, others give in to stress; some socialize easily, others are withdrawn. The striking similarities we share with either or both of our parents result from genetic transmissions. The genes carry the attributes which the child inherits from the parents.

Definitions

Genetics – Study of individual genes and their impact on relatively rare single gene disorders.

Genomics – 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.

Clients – Recipients of health care may include persons, families, communities, and/or populations from any race, ethnicity/ ancestry, culture, or religious background.

Professional Responsibilities

All nurses are expected to engage in professional role activities that are consistent with the American Nurses Association. (ANA, 2004) In addition, competent nursing practice now requires the incorporation of genetic and genomic knowledge and skills in order to: recognize when one's own attitudes and values related to genetic and genomic science may affect care provided to clients. The registered nurse according to ANA[12,13] should be able to do the following: Demonstrates an understanding of the relationship of genetics and genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness. Demonstrates ability to elicit a minimum of three-generation family health history information. Constructs a pedigree from collected family history information using standardized symbols and terminology. Collects personal, health, and developmental histories that consider genetic, environmental, and genomic influences and risks. Conducts comprehensive health and physical assessments which incorporate knowledge about genetic, environmental, and genomic influences and risk factors. Critically analyzes the history and physical assessment findings for genetic, environmental, and genomic influences and risk factors. Assesses clients' knowledge, perceptions, and responses to genetic and genomic information. Develops a plan of care that incorporates genetic and genomic assessment information.

Identification

The registered nurse: Identifies clients who may benefit from specific genetic and genomic information and/or services based on assessment data. Identifies credible, accurate, appropriate, and current genetic and genomic information, resources, services, nd/or technologies specific to given clients. Identifies ethical, ethnic/ancestral, cultural, religious, legal, fiscal, and societal issues related to genetic and genomic information and technologies. Defines issues that undermine the rights of all clients for autonomous, informed genetic- and genomic-related decision- making and voluntary action.

Referral Activities

The registered nurse: Facilitates referrals for specialized genetic and genomic services for clients as needed.

Provision of Education, Care, and Support

The professional nurse provides clients with interpretation of selective genetic and genomic information or services. Provides clients with credible, accurate, appropriate, and current genetic and genomic information, resources, services, and/or technologies that facilitate decision- making. Uses health promotion/disease prevention practices that: Consider genetic and genomic influences on personal and environmental risk factors. Incorporate knowledge of genetic and/or genomic risk factors (e.g., a client with a genetic predisposition for high cholesterol who can benefit from a change in lifestyle that will decrease the likelihood that the genetic risk will be expressed). Uses genetic- and genomic-based interventions and information to improve clients' outcomes. Collaborates with healthcare providers in providing genetic and genomic health care. Performs interventions/treatments appropriate to clients' genetic and genomic healthcare needs. Evaluates impact and effectiveness of genetic and genomic technology, information, interventions, and treatments on clients' outcome.

Implementation Strategies

Practice and curriculum change requires the commitment of nursing leaders and academic faculty to develop a long-term plan to incorporate genetic and genomic information into practice and educational curriculum in order to improve the public's health. Faculty and practicing nurses must be supported by their institutions to attend continuing education or academic courses to update their genetic and genomic knowledge. Collaboration with other disciplines is necessary to provide a strong foundation of knowledge of basic human genetics and current applications to practice. Once content gaps are identified, many curriculum change strategies can be used to add genetic and genomic content to instructional resources. Potential solutions include incorporating genetics and genomics as a central science including the following: Add genetic and genomic content to existing lectures; Integrate assignments and test questions incorporating genetic and genomic knowledge into existing courses; Include genetic- and genomic-focused objectives in all nursing courses; Create a curriculum thread focused on genetics and genomics; Develop an elective genetics and genomics nursing course that can be transitioned into a required course; and Collaborate with interdisciplinary colleagues to design courses and curricula. Outcomes associated with some of the curriculum options listed above have already been published.

Pedigrees

A pedigree is a diagram of a family history that shows the family members and their relationship to the proband – the family member who has been identified as having a genetic disorder. Pedigrees are helpful because they provide a visual display of how disorders and characteristics occur in a family and across generations of the family. Typically three-generation pedigrees are used by genetic nurses, genetic counselors, and geneticists to identify risk for a disorder and plan a program to help prevent chronic conditions. Pedigrees of autosomal dominant, autosomal recessive, X-linked dominant, and X-linked recessive disorders are shown in Figures 1-4

Figure 1. Autosomal Dominant Inheritance

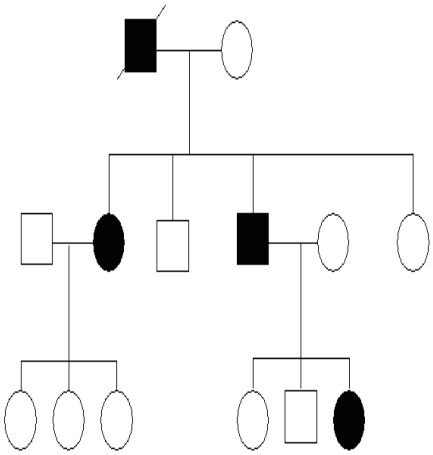


Figure 2. Autosomal Recessive Inheritance

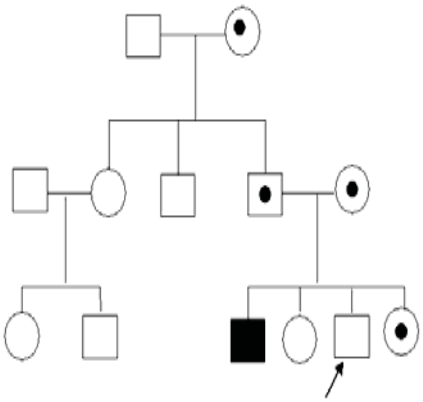


Figure 3. X-Linked Dominant Inheritance

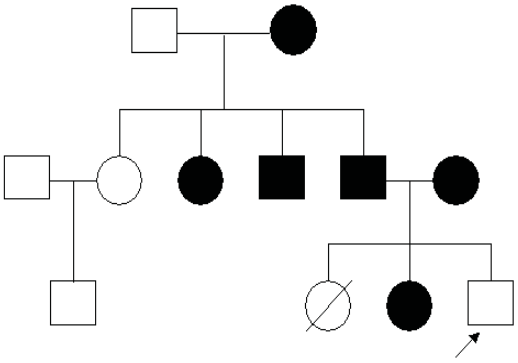
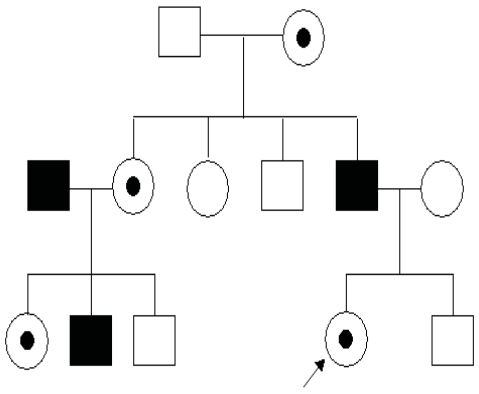
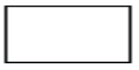


Figure 4. X-Linked Recessive Inheritance



Key for Figures 1-4.



Unaffected Male

Unaffected Female

Affected Female

Deceased Affected Male

Consultand (seeking counseling)

Female Carrier

Genetic Testing

Genetic tests are conducted by examining a person's blood, other body fluid, or tissue for biochemical, chromosomal, or genetic markers. The terms genetic testing and genetic screening may be used interchangeably. In some areas of practice, genetic screening refers to testing that identifies persons who are at higher risk of having or developing a genetic disorder, while genetic testing refers to those tests that are conducted for purposes of diagnosis. In this paper, the term genetic testing refers to testing that determines a person's risk for a disorder, carrier status, diagnosis, and prognosis. Tests may include molecular analysis of DNA and RNA, cytogenetics on chromosomes and biochemical analysis of proteins or certain metabolites. Prenatal genetic testing is used to identify genetic disorders such as Down syndrome through cytogenetic testing. Newborn screening programs in the U.S. are established on a state-by-state basis and typically include various genetic disorders. The National Newborn Screening and Genetic Resource Center (http://genes-r-us.uthscsa.edu/) provides up-to-date information on tests required in each state. Carrier genetic tests are conducted to determine whether an unaffected person is carrying a gene that causes a certain disorder that could be transmitted to offspring. Diagnostic genetic testing is carried out on a person who has symptoms for the purpose of identifying the disorder and providing a prognosis. Predictive genetic testing is offered to a person who is asymptomatic yet is at-risk for a genetic disorder. Presymptomatic testing for prediction is conducted on a person who will eventually develop the symptoms if the gene mutation is present. Susceptibility testing for prediction is conducted on a person who is at-risk for developing the genetic disorder but for whom there is no certainty that the disorder will develop if the mutation is present [12].

INCORPORATING GENETICS INTO CLIENT CARE : AN EXAMPLE

Pharmacogenomics is a rapidly growing field of research into the ways in which genetic variation affects drug response. Its objective is to develop precisely targeted, optimal drug therapy. One area of pharmacogenomics focuses on identifying genetic markers for differences in the way people metabolize drugs; another concentrates on developing genetic tests that predict how specific patients will respond to agents such as cancer therapies. Nurses are now called upon to interpret such information or services in order to provide appropriate patient teaching regarding drug selection.

Imagine yourself to be a nurse working in a day-surgery recovery room. You frequently administer tramadol, a synthetic analogue of codeine commonly prescribed for postoperative pain, at a standard dosage of 50 to 100 mg by mouth every four to six hours. Although the medication works well in most patients, you have found that in some patients it has almost no effect and in others produces symptoms of apparent overdose (extreme dizziness, drowsiness, slowed respiration, and decreased pupil size). You are concerned for both the patients who appear to be overdosed and those whose pain is unrelieved. You suspect that the varied response is related to individual differences in drug metabolism. Is there a way to predict which people have the type of metabolism that might produce signs of overdose or render the drug ineffective?

These two examples highlight a vital problem in health care: adverse drug reactions cause about 100,000 deaths per

year, according to one 1998 analysis. Clinicians' understanding of the interaction between specific drugs and a person's genetic makeup not only has serious clinical implications, it could also help in the development of tests that reduce the trial and error involved in drug selection. Although the field of pharmacogenomics–pharmacogenetics is relatively new, it is generating an enormous amount of information that nurses can put into practice at the bedside. This creates new opportunities for patient teaching regarding drug selection, which nurses must understand in order to provide patients with an adequate "interpretation of selective genetic and genomic information or services," There is little doubt that genetic and genomic research will further clarify how and why patients differ in their responses to medications. In the meantime, it can be useful to identify the areas of drug research that have expanded most dramatically from such discoveries and have already filtered into clinical nursing practice.

Conclusion

Translating knowledge of genetics and genomics into benefits through improved health was one of the major themes proposed by the National Human Genome Research Institute as the genomic era emerged and the Human Genome Project ended [14,15,16] Nurses are increasingly expected to use genetic and genomic information and technology to improve the health of persons receiving their care. Since essentially all diseases and conditions have a genetic or genomic component (Lewis, Calzone and Jenkins, 2006) [17], nurses providing care to many persons who have a known or sometimes unidentified genetic disorder need to understand that such persons may be predisposed to a health problem that is related to the genetic disorder but could be minimized by lifestyle changes, such as changes in diet or activity level as seen in cardiovascular and cancer genetics.

As persons with chronic diseases live longer and genetic and genomic information and technology continue to increase, nurses have a responsibility to learn about these advances and integrate genetic and genomic information and technology into their every day practices. Adapting to the changes and advances in science, nurses can use their passion to be at the forefront of translating genetic and genomic information and technology into improved care and health outcomes for patients and their families. If Speech-Language-Hearing practitioners do not want to be left out then nurses must not[18].

The lack of a genetically informed nursing workforce along with health care systems that are not prepared to implement genetic/genomic information into holistic care delivery are major obstacles to transforming health care and it is hoped that this article in its bid to review relevant literature provides a call to action for nurses in Nigeria.

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