

OVERVIEW OF GENETIC COUNSELLING IN CANCER CARE

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ABSTRACT

The accelerated discovery of disease and susceptibility genes made possible by the sequencing of the human genome has brought new and exciting challenges to the field of genetic counselling. Genetic counselling has therefore been the topic of many studies and wide discussion because of its importance in providing and interpreting genetic information to patients and their relatives. Besides, as the use of genetics in clinical care has increased, both interest in and concern about the impact of genetic information on the life of individuals have been raised. As the scope of genetic counselling therefore expands and evolves, patient, professional and community education will be imperative. Hence, the importance of cancer genetic counselling cannot be over emphasized and it is hoped that this write up will contribute to this quest.

Various definitions of genetic counselling and roles were examined; a typical structure was described following a model guideline of pre and post-test counselling. An illumination into vital aspect of family history, pedigree development and its relevant to health care providers especially nurses was also provided.

Introduction

According to Globocan¹, 8.2 million people Worldwide died from cancer in 2012, 60% of world's total new annual cases occur in Africa, Asia, Central and South America. Yet, 30% of cancers could be prevented if detected early. Cancer is a renegade system of growth that originates within a patient's biosystem with over 200 different known types that affect humans but with one hallmark characteristic: unchecked growth that

progresses toward limitless expansion according to National Cancer Institute². Many factors are known to increase the risk of cancer, including tobacco use, dietary factors, certain infections, exposure to radiation, lack of physical activity, obesity, and environmental pollutants³. These factors can directly damage genes or combine with existing genetic faults within cells to cause cancerous mutation⁴. Approximately 5–10% of cancers can be traced directly to inherited genetic defects⁵. The focus of this write up is not to explore cancer in details but to provide an overview of a veritable tool for prompt screening and early detection known as cancer genetic counselling which has not been explored in our environment. Cancer can be detected in a number of ways, including the presence of certain signs and symptoms, screening tests, or medical imaging. Genetic counselling enhances all of these and therefore plays a vital role in cancer care and management.

KEYWORDS:

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Cancer results from an accumulation of genetic changes within a cell that allow uncontrolled cell growth. In the vast majority of cancers, these changes are not inherited but occur after birth due to random biological events and exposure to certain environmental agents. Occasionally, families have a very strong cancer history suggesting that a major, inherited cancer predisposition gene is responsible. The most common inherited cancers include breast, ovarian and colon although other types exist. Nonetheless, the vast majority of cancers are non-hereditary (sporadic cancers). Hereditary cancers are primarily caused by an inherited genetic defect. Although, less than 0.3% of the population are carriers of a genetic mutation and these cause less than 3-10% of all cancers, it has a large effect on cancer risk according to Genome-wide association studies⁶. Some of these syndromes include: certain inherited mutations in the genes BRCA1 and BRCA2 with a more than 75% risk of breast cancer and ovarian cancer⁶ and hereditary nonpolyposis colorectal cancer (HNPCC or Lynch syndrome) which is present in about 3% of people with colorectal cancer⁷ among others. As the use of genetics in clinical care has increased, both interest in and concern about the impact of genetic information on the life of individuals have been raised. Hence, the importance of cancer genetic counselling cannot be over emphasized.

A genetic counsellor will listen to the client's concerns and counsel based on his or her risk of developing cancer such as being a first, second or third degree relative of a cancer patient. In this article, A first-degree relative is defined as a close blood relative which includes the individual's parents, full siblings, or children, a second-degree relative is defined as a blood relative which includes the individual's grandparents, grandchildren, aunts, uncles, nephews, nieces or half-siblings and a third-degree relative is defined as a blood relative which includes the individual's first-cousins, great-grandparents or great grandchildren The meeting may last

30-60 minutes or more depending on the individual's need, it may not involve a physical examination, but a joint decision may be reached by both the counsellor and the client to see a doctor or a specialist nurse or to go for regular screening to see if there are any abnormalities where there are no symptoms. For example, it can help find a cancer earlier. About 5-10% of all cancers are inherited. This means that mutations in specific genes are passed from one blood relative to another. Individuals who inherit one of these abnormal genes have a much greater chance of developing cancer within their lifetime and at an earlier age. In general, people at risk for an inherited form of cancer share the following characteristics or have family members who were diagnosed with: Cancer at an early age, the same type of cancer, two or more different cancers in the same person, a rare cancer, such as male breast cancer or sarcoma and a BRCA1 or BRCA2 mutation.

Definitions of Genetic Counselling

Genetic counselling has been the topic of many studies and wide discussion because of its importance in providing and interpreting genetic information to patients and their relatives. There are several definitions of genetic counselling; probably the most often cited is that published in the American Journal of Human Genetics, in 1974, by Fraser⁸ as "a communication process that deals with the human problems associated with the occurrence, or risk of occurrence, of a genetic disorder in a family." This process involves an attempt by one or more appropriately trained persons to help the individual or family achieve the following:

1. Comprehend the medical facts, including the diagnosis, the probable course of the disorder, and the available management.
2. Appreciate the way that heredity contributes to the disorder, and to the risk of recurrence (occurrence), in specified relatives.

3. Understand the alternatives for dealing with the risk of recurrence (occurrence).
4. Choose a course of action that seems appropriate to them in view of their risk, their family goals, and their ethical and religious standards and act in accordance with that decision.
5. Make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence (occurrence) of that disorder⁸. The core of genetic counselling according to this definition is to present genetic facts to the counselees, and to help them to understand their meaning and choose the course of action most appropriate to them in relation to the genetic problem present in the family. It is an integral part of genetic testing process.

According to Resta, Biesecker and Bennett⁹, the following definition of genetic counselling was approved by the National Society of Genetic Counsellors (NSGC) Board of Directors: Genetic counselling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to diseases. This process integrates the following: Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence, education about inheritance, testing, management, prevention, resources and research, counselling to promote informed choices and adaptation to the risk or condition⁹. Genetic counselling is the process by which patients or relatives, at risk of an inherited disorder, are advised of the consequences and nature of the disorder, the probability of developing or transmitting it, and the options open to them in management and family planning. This complex process can be separated into

diagnostic (the actual estimation of risk) and supportive aspects¹⁰. Genetic counselling is a non-directive process which aims to explain the facts as clearly as possible, giving the person or family accurate information on their options in a way which they can understand, and helping them to make up their own minds¹¹. Genetic Alliance¹² states that counsellors provide supportive counselling to families, serve as patient advocates and refer individuals and families to community or state support services. They serve as educators and resource people for other health care professionals and for the general public.

Counselling session structure

The goals of genetic counselling are to increase understanding of genetic diseases, discuss disease management options, and explain the risks and benefits of testing. Counselling sessions focus on giving vital, unbiased information and non-directive assistance in the patient's decision-making process. Seymour Kessler¹³, in 1979, first categorized sessions in five phases: an intake phase, an initial contact phase, the encounter phase, the summary phase, and a follow-up phase¹⁴. The intake and follow-up phases occur outside of the actual counselling session. The initial contact phase is when the counsellor and families meet and build rapport. The encounter phase includes dialogue between the counsellor and the client about the nature of screening and diagnostic tests. The summary phase provides all the options and decisions available for the next step. If counselees wish to go ahead with testing, an appointment is organized and the genetic counsellor acts as the person to communicate the results.

Counselling the person, couple or family and the role of the counsellor
Counselling should be carried out in a relaxed atmosphere with sufficient time to absorb the initial shock of diagnosis.

- It should include the clinical presentations of the disease, treatment, natural history, prognosis, complications, and a clear explanation of the genetics. The risk to the individual of developing symptoms, the risk to future offspring, and the way in which the disease is transmitted.
- All information must be given in simple, easy to understand language.
- Carefully assess the understanding of the client about the problem: have they any misconceptions, which need rectifying, or any misplaced guilt?
- Explaining the mechanism of inheritance of a disease to the patients.
- Dispel unnecessary fears in patients without significantly increased risk of disease.
- Advise on lifestyle changes for patients who may have inherited an increased susceptibility to a particular cancer disease.
- Provide support to patients with a clear family history of high levels of certain forms of cancer occurring at an early age – e.g. breast, colorectal, ovarian, endometrial and familial adenomatosis polyposis.
- Genetic counselling has to be provided or supervised by a healthcare professional appropriately trained for genetic counselling.
- Non-genetics healthcare professionals have a responsibility to recognise their abilities and limitations with regard to provision of genetic services.
- Healthcare professionals should not agree to test without pre-test counselling in circumstances where doing so would go against their professional judgement.
- Predictive tests for future severe illnesses with no options for treatment or prevention should not be performed without pre- and post-test genetic counselling, psychosocial evaluation and follow-up.
- Before actual testing takes place, there should be free and informed consent.
- In situations where testing children or other persons who are not able to give informed consent is considered, those individuals should be involved in genetic counselling and in the decision-making process, according to their capacities.
- Testing for adult-onset conditions in children should only be considered when treatment or surveillance would begin in childhood.

Pre-test genetic counselling

Individuals are informed about the purpose of the test, including:

- Up-to-date, reliable description about symptoms and natural history of the disease.
- Prospects of prevention or early diagnosis and treatment.
- Inheritance pattern.
- The risk of disease, available reproductive choices, reliability and limitations of the test concerned, and possible psychological impact and other consequences of the test result to the person and their relatives.

Genetic counselling is not primarily 'counselling' in the psychological sense. Genetic counselling is non-directive and aims to explain the facts as clearly as possible, giving the person or family accurate information on their options in a way which they can understand, and helping them to make up their own minds. Guidelines for genetic counseling¹⁰

- Privacy and confidentiality of the results, as well as possible consequences related to its disclosure to third parties, such as insurance companies and employers, are discussed, when appropriate.
- Pre-test counselling includes discussion about the rights to know and to decide including the right not to know.
- Possible uncertainties due to present lack of knowledge are declared.
- Discussions about the need to inform relatives about the test result, as well as the best ways to do this, are initiated, especially in conditions where early diagnosis may improve the prognosis.
- Written materials and/or reliable Internet addresses related to the subject should be offered when available.
- A written summary of the discussion should be offered.
- European guidelines on genetic counselling for pre symptomatic testing have recently been developed. These include general principles governing the offer of testing (e.g., autonomous choice of the patient), objectives of genetic counselling in this context (e.g., facilitation of decision making), logistical considerations (e.g., use of trained staff) and topics to be included during counselling discussion with the patient (e.g., consequences of both positive and negative outcomes)¹¹.

Post-test genetic counselling

- After disclosure of test results, the first focus is on the emotional impact on the person and others involved.
- If necessary, follow-up contacts with the genetic counselling unit should be offered, and/or a consultation with a psychologist.
- The possibility to contact a social worker and patient support organisations should also be offered.

- A written summary of the test result and issues discussed should be given.
- Implications to the individual (including a follow-up plan, when relevant) and their near relatives should be discussed
- A strategy to inform relatives should also be discussed. Written material to help the counselee to spread the information in the family should also be offered.

Common Indications for referral to a cancer genetic counsellor

Common indicators for referral to a cancer genetic counsellor according to Marchina, Fontana, Speziani¹⁵, include: A person with a known genetic condition in the family, wanting to know their own risks to cancer based on the condition. A person with a strong family history of cancer, wanting to know if they are at increased risk to such or other related cancers and, if they are, what options they have. A person with a known genetic condition wanting specialist advice about the condition. A person with a possible genetic condition in the family wanting to know if a diagnosis can be made and, if so, their risks and options. A pregnant couple told that a test has given an abnormal result, wanting to talk about what the result means, and what options are available.

Family History

In genetic counselling process, accurate and complete family history is vital. What does this mean? Family members share genes, behaviours, lifestyles, and environments where they grow together but that also together may influence their health and their risk of complex diseases such as cancer. Most people have a family health history of some chronic diseases (e.g., cancer, coronary heart disease, and diabetes) and health conditions (e.g., high blood pressure and hypercholesterolemia). People who have a close family member with a chronic disease may have a higher risk of developing that disease than those without such a family member.

Family health history is a written or graphic record of the diseases and health conditions present in one's family. A useful family health history shows three generations of a biological relatives, the age at diagnosis, the age and cause of death of deceased family members. Family health history is therefore a useful tool for understanding health risks and preventing disease in individuals and their close relatives. Some people may know a lot about their family health history or only a little. It is helpful to talk with family members about their health history and document findings for updating from time to time. This way, family members will have organized and accurate information ready to share with their health care provider. Family health history information may help health care providers determine which tests and screenings are recommended to help family members know their health risk.

Family History Basics

A family history should be obtained from all the clients seeking genetic counselling. This includes the construction of standard three-generation pedigree containing information about the client, the client's first-degree relatives (children, siblings, and parents), second-degree relatives (half-siblings, aunts, uncles, nieces, nephews, grandparents, and grandchildren), and third degree relatives (first cousins). The first consideration when gathering family history information are Who, What, Where, When, and How?

Who: In general, the pedigree begins with the individual for whom genetic counselling is being provided. The consultand (or client) is the individual seeking genetic evaluation, counselling or testing and may or may not be affected while the "Proband" is the family member who brings the family to medical attention¹⁶ and they may be one person.

What: The nature of the referral or reason for the visit such as referral for genetic counselling because of a family history of a

particular disorder should be clarified.

Where: The family history should be obtained in an environment that is comfortable and free of distractions. It should also be obtained in a setting that preserves confidentiality.

When: The pedigree is usually drawn in the presence of the client (s) and a family history questionnaire can be sent to patients in advance of their appointment or telephone interview.

How? A pedigree is part of a client's medical record. All medical documentation, including the pedigree should be recorded in black pen but should be taken in pencil at first to give room for any other additional information that the patient might remember after the form has been filled.

Relevance of Family History to Healthcare Providers

Obtaining a family history is an established and familiar screening activity used by nurses and other healthcare providers across many healthcare settings. Knowledge of the illness and cause of death of biologically related family members gives the nurse important information about shared genes, environment and lifestyle behaviours that may increase a person's risk for the same disease. Knowledge of a family health history also contributes to the decisions regarding who may benefit from genetic testing for common rare conditions, and for factors influencing treatment choices¹⁷. Identification of conditions that pose a serious health risk is an important goal of a genetic family history. For example, the recognition by a nurse that a child's parent died at a young age from colon cancer may lead to further evaluation and recognition of a familial adenomatous polyp, a familial cancer syndrome for which the child and sibling are at risk. The discussion that takes place during a nursing review of family history is therefore an important opportunity for evaluation of risk reduction activities

when there may be genetic or genomic factors increasing risk for disease. In a study findings for example, nurses who interviewed white and black women with family history of breast cancer found that many of the women were unaware of associations between lifestyle behaviours and risk for breast cancer. About one-third of these women who had one or more sisters with breast cancer reported making lifestyle changes¹⁸.

In a recent National Institutes of Health (NIH)¹⁹ report, researchers noted that among studied individuals in United States, being female, having health insurance, and having a moderate to high socioeconomic status increased the likelihood that a family history could be provided. This report also concluded that people are more likely to accurately report the absence of a disease rather than the presence of a disease in family members, and the ability to report family health history information was better for first-degree relatives than for second-degree relatives. There is a less accuracy for reporting a family history of mental illness, which may reflect both the difficulty people have in reporting this information about them, as well as limited sharing of this information within a family¹⁷.

Numerous factors may influence the accuracy and usefulness of genetic family history for identifying risk for disease or selection of treatment options, when nurses obtain and evaluate a family health history; there are several points to keep in mind. Factors such as limited knowledge of family members' medical information or reluctance to reveal sensitive information may limit the accuracy of family health history. The context in which the history is being discussed can also limit full disclosure. For example, parents may not wish to discuss information that they consider to be private in front of their minor age or adult children. The definition of family may differ when viewed from the perspective of a nurse or health care provider, and from the perspective of members of some ethnic or cultural groups according to Berg¹⁷. In addition to recognising and adapting the family history process to individual and

family circumstances, nurses can use the process of obtaining a family history as an opportunity to determine how family members use this information in making decisions about their own health habits for risk reduction of disease. Family history can identify at risk members who may benefit from genetic testing.

As outlined by American College of Obstetrician and Gynecologists²⁰, collection of a patient's family history, in conjunction with his or her traditional medical history information can do the following:

Inform diagnosis: Whether a patient presents with a relatively uncommon single-gene disorder such as hemophilia, or a common complex condition such as cancer, knowledge of family history can promote more rapid diagnosis, streamlined testing, and better long-term management.

Promote risk assessment: Medical family history information, in combination with other risk factors, can be used to estimate a patient's risk of developing the same or similar condition as a relative, and to stratify that risk into higher or lower categories. Information such as the number of affected and unaffected relatives, age at onset of disease, severity of disease, and degree of relationship comes into play in this assessment. This assessment can also determine if a patient is an appropriate candidate for genetic testing and can identify other relatives who might also be at high risk to develop a medical condition.

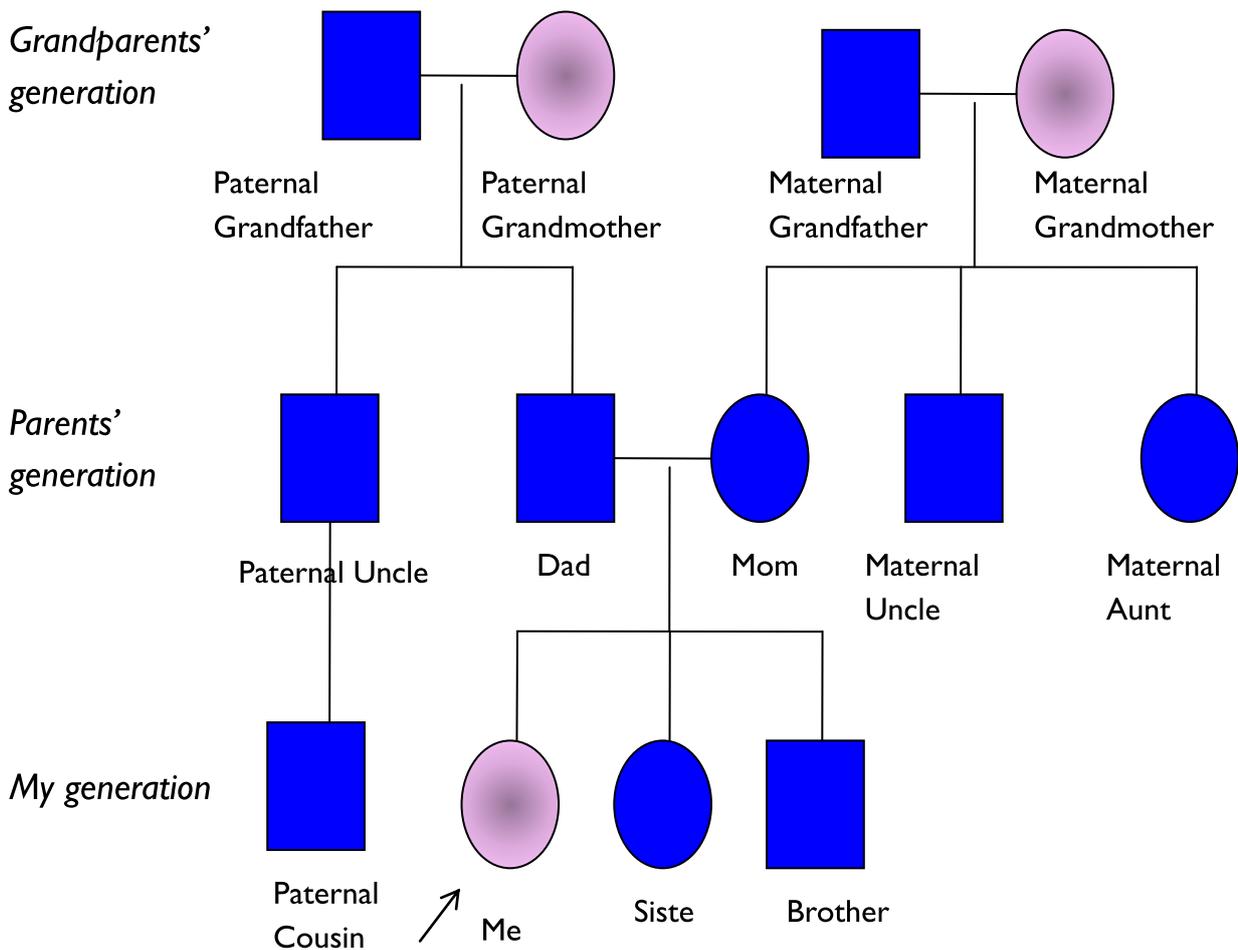
Guide Management: Once a patient's risk has been determined, the provider can suggest or advise appropriate interventions to improve the patient's outcome. A positive family history may lead to a diagnosis in a person with disease. If family history reveals that a patient is at increased risk, the provider can implement screening strategies to detect disease early, when it is most treatable. The emphasis on disease prevention and management based on the family history may

also motivate changes in behavior that forestall disease or reduce its adverse affects.

Build rapport with patients: The act of taking a family history is an excellent opportunity to build a relationship with the patient. Through this interaction, the healthcare provider may also become aware of the patient's motivations and concerns, as well as family dynamics. All such information can be beneficial as the provider helps the patient make health-related decisions. While building rapport and taking the history the information obtained is represented diagrammatically as a pedigree.

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, genetic counsellors use three-generation pedigrees to identify risk for a disorder and plan a program to help prevent chronic conditions. See diagram below and commonly used pedigree symbols, definitions, and abbreviations:



The family history is therefore very important in working out the chance of there being a genetic susceptibility to cancer in the clients' family. Hence, it is a good idea for the nurse to find out as much as possible about the family history of cancer. Then, draw up a simple family tree, based on the information obtained from the history showing all close biological relatives and their respective illnesses.

SUMMARY AND CONCLUSION

The focus of this paper is on genetic counselling. It examined various definitions of genetic counselling and brought to fore the relevance of family history and its 3-generation diagrammatic illustration called pedigree. Professional change lies ahead in genetic counselling and anticipating this change offers an opportunity to be prepared. I therefore call for widespread gains in nurses' professional expertise in new genetics and genomics nursing vis a vis an avenue for improved competencies in genetic counselling generally for all chronic conditions and specifically for cancers. And then, some of the improvements in health promised by human genome research may be realized, if not maximized.

NURSING IMPLICATION

At present, there are no professional groups in Nigeria referred to as Cancer genetic counsellors. It is either done by clinicians who have no understanding of cancer genetics or not done at all. While colleagues abroad can boast of this luxury in developed countries, Nigeria may not, hence the dire need to use what we have to get what we need. This can be achieved by expanding nurses' role whose functioning already includes obtaining patient information for nursing care to add the new information needed for the 3 generation pedigree development. Nonetheless, it should be noted that oncology nursing does not exist as a stand alone specialty in any formal university nursing

educational program in Nigeria. This is desperately needed. Also, there is need for the establishment of comprehensive cancer centers to include cancer risk clinics at least at the University teaching hospitals for genetic counselling where nurses with appropriate training can undertake cancer genetic counselling to for increase uptake of cancer screening, early detection, prompt treatment and referral.

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