



Theory Development of Genetic Counseling among Patient with Genetic Diseases

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Abstract. Genetic counseling is a communication process undertaken to address the issue of genetic diseases affected by the family. The basic concept of Selfcare is active awareness, while genetic counseling is passive awareness. The nursing theory is based upon the philosophy that all "patients wish to care for themselves." This study's objective is to sustain the theory development of genetic counseling related to Orem's Selfcare theory following on from three relationship construction theories, which include self-care theory, self-care deficit theory, and nursing system theory. Genetic counseling is relevant to Orem's self-care theory as a gold standard to improve self-care awareness, quality of life and reduce stress levels among patients and families with genetic diseases. This theory's development can be carried out in follow-up studies that focus on implementing public policies in improving health care for patients with genetic disorders. The government needs to construct an adequate education curriculum policy, training management, competency testing, genetic counselor certification, and formal oversight institutions. All of these prerequisites will be one of the initial efforts for stable genetic counseling intervention. Also, the protocol study of genetic counseling still needs to be studied more deeply.

Keyword: Genetic Counseling, Selfcare, Theory Development



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INTRODUCTION

Genetic Counseling Practices in Indonesia currently do not have an operational standards procedure accredited by the World Health Organization (WHO). Research on genetic counseling is only conducted at the urgency stage to build awareness of the importance of genetic counseling. In fact, in 2014, the WHO stated that every 1 million population in each country must have one genetic counselor(1). Therefore, if the Indonesian communities are above 200 million, there should be at least 200 professional genetic counselors. To reach this number is still far away, although the number of health workers is enormous, especially nurses. In almost every health service places, nurses are the most considerable number of health workers(2).

A lot of research can be used as reference material in understanding the urgency, goals, management, process, and evaluation in carrying out genetic counseling. One of the exciting writings that we can read includes an article written by Rujito and Gazhali (2010) entitled *Initiating Development of Genetic Counseling Services in the Health Service Unit: A Preliminary Study*, published by the Indonesian Medical Magazine. This article can learn about the opportunities and challenges in providing genetic counseling services in health facilities in Indonesia(3).

Genetic counseling is a communication process undertaken to address genetic diseases affected in the family(4). The history of gene counseling begins in Sweden in the 1940s, although there was no term genetic counseling at that time. However, the practice of health services provided is a Genetic counseling activity that we know today. This process aims to help individuals or families know and understand disease management, prognosis, genetic basis, possible recurrence, treatment options, or genetic testing and help them make choices according to their personal and family situation(5).

However, genetic counseling interventions carried out in hospitals and other health services are still considered insufficient. Several studies have demonstrated patient dissatisfaction with congenital disease treatments performed in healthcare facilities(6). Also, healthcare providers' roles and responsibilities to care for patients with genetic diseases have not been well identified. This shows that the standard genetic counseling services carried out by health professionals such as doctors, nurses, and genetic counselors have not adequately addressed the needs of patients(6,7,8).

In practice, genetic counseling is often faced with a dilemma, especially when it comes to explaining disease risk factors and the results of genetic testing. Patients often experience increased levels of depression, anxiety, and distress due to genetic counselors' information regarding their illness. This shows a misconception between the purpose of gene counseling intervention with facts and realities in the field. Therefore, a study on the concept of genetic counseling should be carried out. The purpose of the intervention does not conflict with the reality in practice(9).

This study's objective is to sustain the theory development of Genetic Counseling as a gold standard to improve self-care awareness, increase the quality of life, and reduce stress levels among patients and families with genetic diseases. *Is it relevant to the self-care theory by Orem?*

SELFCARE THEORY

There are several concept models. One of them is the "self-care" model introduced by Dorothea E. Orem. Orem developed this nursing concept model in early 1971, where he published it under the title "Nursing Concepts of Practice Self Care." This model initially focused on individuals, then the second edition in 1980 was developed on multiperson units (families, groups, and communities). In the third edition, constructed theories include self-care theory, self-care deficit theory, and nursing system theory(10).

CENTRAL PHILOSOPHY

The basic concept of Selfcare is active awareness, while genetic counseling is passive awareness. The nursing theory is based upon the philosophy that all "patients wish to care for themselves." They can recover more quickly and holistically if they are allowed to perform their self-care to the best of their ability(11). Orem's self-care deficit nursing theory emphasized establishing nursing perspectives regarding human and practice.

Genetic counseling is a part of Nursing Intervention to optimize the role of Nurses as a counselor to improve patients and families who suffer congenital abnormalities that can be inherited to the next generation. Nurses are not the only educator about the physical condition but also provide counseling to resolve psychosocial problems (QoL, Stress, and Anxiety Level)(12).

Meta-paradigm

A meta-paradigm is a set of theories or ideas that provide a structure for how a discipline should function. In Genetic Counseling, Patients and families decide (Active Awareness) of some alternatives or treatment management options, screening tests, and function of relation role (such as family). Nurses as a genetic counselor who investigate, inform, counsel, negotiate, confirm, overall as a facilitator to guide patients and families to make a decision(13).

In its implementation, genetic counseling interventions can be carried out for patients and families who experience numerical and structural chromosomal aberrations such as Down Syndrome, Fragile X Syndrome, Autism Spectrum Disorder, Diabetes Mellitus, and hypertension. Many diseases can be taken as examples of clients or counseling genetic objects because many conditions involve genetic roles and functions.

Regarding self-care theory, Genetic counseling correlates with the nursing meta-paradigm(14-15). The implementation of the meta-paradigm can be explained as follows.

1. Person

Individuals or groups who could not continuously maintain self-care for life and health, recovery from illness/trauma or copying, and their effects. Justifies the decision-making process by presenting with health problems or situations requiring decision making. As an individual, the patient has the right to comprehend the disease's function comprehensively. Also, genetic counseling services that are carried out need to consider the patient and family's physical and psychological condition.

One example, a mother who has a child with Down Syndrome (DS) first comes to the hospital or other health provider to ask about her child's condition. At this stage, the nurse, as a counselor, does not need to provide an in-depth and complete explanation. Provide simple information that the client can understand so that the client's psychological burden is not too heavy. However, make a plan for scheduled meetings with specific topics in stages. Therefore, genetic counseling is achieved by reducing the risk and psychological burden.

2. Health

Individuals or groups' ability to meet the demands of self-care that play a role in maintaining and increasing the structural integrity of functions and developments. Satisfaction with the decision making process and decision. We could physically monitor the signs and symptoms in Down syndrome patients, such as up slanting eye, upset ear, prominent ear, macroglossia, micrognathia, and Mongolian face. Also, intellectual disability and autism are common symptoms that often appear in DS patients. Chromosome abnormalities 21 and 14, until now, cannot be repaired. The development of research has not been able to answer the most sophisticated therapy's ability to repair abnormal chromosomes(16).

Thus, all physical abnormalities experienced by DS sufferers cannot be corrected. But intellectual disabilities or mental impairments can be improved or optimized by various therapy methods such as cognitive-behavioral therapy. The treatment model could enhance DS patients who experience intellectual disability or cognitive impairment(17).

3. The Meta-paradigm implementation was also carried out in environment

Patient space (Home, workplace, area of recreation, travel) and Health Care Space (Appointment, e-mail, Phone contact, Hospital, ED, Clinic Offices). The implementation of the meta-paradigm of genetic counseling is comprehensive in environmental aspects. The nurse's attention to the patient's environment includes a playground, school place, place of activity, workplace, relationships in interaction, interaction instruments, and other things related to the environment.

4. Nursing

Services that are deliberately chosen or activities are undertaken to assist individuals, families, and community groups in maintaining care that includes structural integrity, functions, and development. Work on strategies to implement and support the decision in the patient's own space. The partnership with the healthcare team needs to build.

In Indonesia, genetic counseling intervention is not yet widespread in nursing because there is no health service regulation on genetic counseling issued by policymakers, in this case, the Ministry of Health, a government agency. The opportunity to learn about genetic counseling, which has a standard educational curriculum for genetic counselors, only exists at Diponegoro University(18).

As a result, study information and studies on genetic counseling have not been widely accepted in the nursing profession. However, nurses as counselors have ample opportunity to learn the concepts and theories of gene counseling interventions to be applied in nursing practice. One study reported that knowledge and competence increased after receiving comprehensive training within one month in providing genetic counseling interventions(19). Therefore, nurses' opportunity to develop genetic counseling interventions is very wide open so that the role of nurses as counselors can be carried out optimally.

SELFCARE SYSTEM

Genetic Counseling as an Intervention of Family Nursing has an objective to Improve Self Care awareness(20). Nursing as a professional service, in its application, must be based on a solid scientific foundation of nursing. Thus, nurses must think logically and critically in analyzing and identifying the phenomenon of human response. Many forms of knowledge and critical thinking skills must be performed in each client's situation, including using nursing models in the nursing process. Each model can be used in nursing practice as needed(21).

There is a nursing system related to genetic counseling:

1. Interpersonal Aspect

Relationships in their family, a harmonious family, will support positive psychosocial outcomes, such as managing conflict in a family, conducting family traveling, recreation, delivering an idea, critics, etc.

2. Social Aspect

Relationships in social space. How to prevent bullying from a classmate, chose education to improve potential talent, angry management, etc.

3. Procedural Aspect

Practicing necessary family skills so that they could anticipate changes that occur.

4. Technical Aspect

Teach the family about basic techniques that can be done at home, such as doing the right compress, relaxation, deep breathing technique, etc.

CATEGORIES

Genetic counseling can improve self-care awareness related to the construction of patients' and family's knowledge. Categories of Nursing System(21) can be divided into three groups of patients and families:

1. Wholly Compensatory

Overall assistance is needed for clients who cannot control and monitor their environment and do not respond to stimuli, such as Autism Spectrum Disorder/Syndrome, Intellectual Disability, etc.

2. Partially Compensatory

Partial assistance is needed for clients who have limited mobility due to illness or accident, such as Duchenne Muscular Dystrophy, Fragile-X Syndrome of Ataxia, and gangrene of Type II Diabetes Mellitus, Dementia, hemophilia, thalassemia, etc.

3. Supportive Education

Educational support is needed by clients who require it to be studied to perform independent care, such as hypertension.

CONCLUSION

Genetic counseling is relevant to Orem's self-care theory as a gold standard to improve self-care awareness, increase life quality, and reduce stress among patients and families with genetic diseases. This theory's development can be carried out in follow-up studies that focus on implementing public policies in improving health care for patients with genetic disorders. The government needs to construct an adequate education curriculum policy, training management, competency testing, genetic counselor certification, and formal oversight institutions. All of these prerequisites will be one of the initial efforts for stable genetic counseling intervention. Also, the protocol study of genetic counseling still needs to be studied more deeply.

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