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Original Article

A theoretical framework for interaction of nursing discipline with genetics and genomics

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ABSTRACT

Background: Since the completion of the Human Genome Project, health science has been strongly influenced by the advances in genetics and genomics. However, the progress of embracing genetics and genomics into nursing discipline is limited. One of the main barriers is lack of understanding on the relevancy of genetics and genomics to nursing discipline.
Objectives: This paper aims to synthesize and develop a theoretical framework for the interaction of nursing discipline with genetics and genomics.
Methods: Through content analysis and constant comparative method, a theoretical framework was developed from synthesis of the studies regarding nursing and genetics/genomics indexed in multiple English and Chinese databases.
Results: Four main theoretical statements were constructed in the framework: 1) There are three ways to show how genetics and genomics can influence nursing discipline: a new specialty, new technologies and a new lens; 2) The significant contribution of nursing discipline to genetics and genomics lies in how nurses could focus on the association between human responses and genes and how nurses could advocate for their clients in the genetic and genomic era; 3) A paradigm shift occurs after a constant interaction of nursing discipline with genetics and genomics; 4) Implementation strategies could be used to facilitate the integration of genetics and genomics to nursing discipline and advance the paradigm shift.
Conclusions: The framework will help to understand the relationship between nursing discipline and genetics and genomics and implicate the future studies integrating genetics and genomic science into nursing discipline.

1. Introduction

The Human Genome Project was started in 1997 and achieved the goals of mapping the entire three billion pairs of the human genome in 2003 [1]. This was not considered an end but a beginning to the era of advancing genetics and genomics technology and integrating them in health care [2]. It laid a foundation of developing and refining many new genomic technologies [3]. The quick progress of genetic and genomic science has significantly influenced the ways of screening, diagnosing, treating and preventing disease, and monitoring therapy effectiveness in health care [4].

The science of genetics is focused on exploring and explaining the impact of individual or single gene or chromosome changes, most of which are individually quite rare, on health [5,6]. The broader term, genomics, considers the interactions between and within genes, regulatory sequences, the environment, and other psychosocial and cultural factors [5,6]. As the emphasis is moving towards the understanding that an individual's genes themselves are not acting in isolation but many factors impact overall health of an individual, the basic science of genetics has involved into genomic healthcare [5].

Genetics and genomics are reaching people everywhere and have emerged as important science and knowledge for all

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healthcare professionals. Nurses, who represent the largest contingent of health care providers in the world and play a pivotal role in delivering quality health care services, are also expected to acquire the knowledge and skills to apply genetic and genomic technologies [7]. In fact, the implications of genetics and genomics on nursing practices have been recognized for a long time. A few initiatives and attempts have been conducted by the International Society of Nurses in Genetics (ISONG) and other national institutions, including integrating genetics and genomics in the baccalaureate and graduate curricula, certification or licensure examinations, and continuing education in nursing discipline [8,9]. Also, the ISONG and other institutions have provided nursing education as long as 25 years via international or national nursing conferences [8]. In addition to that, they have developed and maintained networking and exchange programs for nurses around the world to learn about the latest information and cutting edge knowledge in other areas or countries [8].

In spite of these efforts, limited progress in the integration of genetic and genomic science into nursing practice still existed [10]. Recent studies showed that nurses from different countries can barely show any competency on genetics and genomics and few of them were well-prepared to deliver genetics and genomics based health care to clients [11–13]. Although barriers, which were identified to fully integrate genetics and genomics into nursing practice, were multifactorial, one of the most fundamental problems is understanding on the relevancy between nursing discipline and genetic and genomic science [14–16].

The purpose of this paper was to synthesize and develop a theoretical framework for the interaction of nursing discipline with genetics and genomics, which can also act as implications of facilitating the integration of genetics and genomics in nursing practice.

2. Methods

2.1. Design

Theory development provides a way of identifying and expressing key ideas about the essence of practices [17]. There are four different approaches for middle-range theory generation: 1) induction/deduction through the literature on research and practice, 2) deduction from the application of grand theories, 3) combination of existing nursing and non-nursing middle-range theories, and 4) derivation from theories of other disciplines that relate to nursing [18]. This study used the first approach and the framework was developed through the synthesis of the studies regarding nursing discipline and genetics/genomics.

2.2. Data sources and searches

English and Chinese databases were searched, including MEDLINE (Ovid), EMBASE(Ovid), CINAHL (EBSCO), PsycINFO (Ovid), Nursing and Allied Health (ProQuest), and the Cochrane Library (CENTRAL, Cochrane Database of Systematic Reviews, and DARE), WANFANG, CNKI and VIP. Search terms included a combination of subject headings, terms, and keywords such as “genes”, “genomics”, “nurse” and “nursing”. The date limits were set until December 2017. The initial database search retrieved 1042 records with 890 articles remaining after the duplicates of the initial records were removed.

2.3. Study screening

After removal of duplicates, a two-stage process was used for screening. First, two researchers (JH, LY) independently screened all the titles and abstracts of the studies in relation to the eligibility criteria through Covidence [19]. Inclusion criteria were those nursing studies which were in genetic and genomic science. Three-hundred-seventy-four articles were remained after title/abstract screening. Next, the full texts of the included articles were read and identified. Any discrepancies were resolved by discussion with the third reviewer (YY), when necessary. A final count of 32 studies was included in the review. The details of the screening process were displayed in a PRISMA flow chart (See Fig. 1).

2.4. Data analysis

Content analysis [20] and Miles, Huberman and Saldana's constant comparative method [21] were used to analyze the data. Through constant analysis comparison, meaningful units of the transcribed text, such as paragraphs, sentences and words, were extracted to codes, condensed into concepts or statements and grouped into the framework [22,23]. Recommended by Walker and Avant, concepts are the basic building blocks of theory [17]. A concept is a mental image of a phenomenon, an idea, or a construct in the mind about a thing or an action [17]. A statement declares a relationship of some kind between two or more concepts or asserts the existence of the concept [17]. Finally, a theory is an internally consistent group of relational statements that presents a systematic view about a phenomenon and that is useful for description, explanation, prediction, and prescription or control of the phenomenon [17].

3. Overview of the framework

A Theoretical Framework interpreting the Interaction of Nursing Discipline with Genetics and Genomics through a bidirectional approach is presented in Fig. 2. In the framework, genetic and genomic science are applied to nursing discipline through providing a new specialty, a new technology and a new lens. At the same time, nursing discipline could advance the development of genetic and genomic by bringing worldviews and different perspectives through dealing with human responses and advocating for the rights of patients in genetic and genomic era. Nursing discipline is focused on the association between genes and human responses, including physiological, psychological and behavioral responses. Also, nursing discipline plays a significant role in advocacy for individuals and families to consider and address psychological, social, ethical, and legal issues related to genetics and genomics. In the constant bidirectional interaction of Nursing Discipline with Genetics and Genomics, a paradigm shift may happen in nursing science. However, the process of paradigm shift may be discomforting, and scientific revolution is a far-reaching and long-term goal. The process of translating genetics and genomics into nursing discipline is challenging and needs more efforts. Thus, the activities of knowledge translation are necessary to guide the process of knowledge translation. Also, implementation strategies need to be tailored based on three significant factors, including evidence-based innovation, potential adopters and practice environment.

There are four levels of theories in nursing: meta-theory, grand theory, middle-range theory and micro-range theory [18]. Middle-range theories are described as “those between the minor hypotheses of day to day research and unified theory (p39).” [24] Thus, the middle range level is below the more philosophical or grand theories and above empirical generalizations framed as hypotheses The theoretical framework presented in this paper is a middle-range theory, which explains the empirical world of nursing discipline in the genetic and genomic era [25]. Although it's specific in context (nursing in genetics and genomics era), it can be general to cross multiple clinical populations and to encompass similar phenomena [25]. In addition, as a major role of middle-

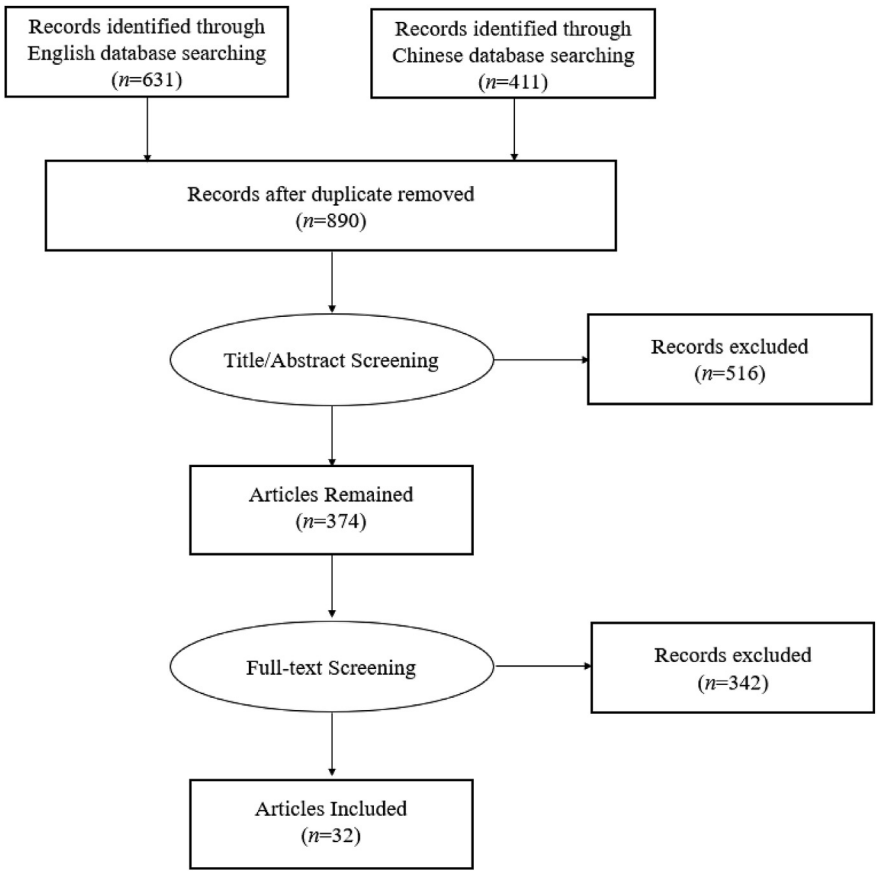

Fig. 1. Selection process of the included studies.

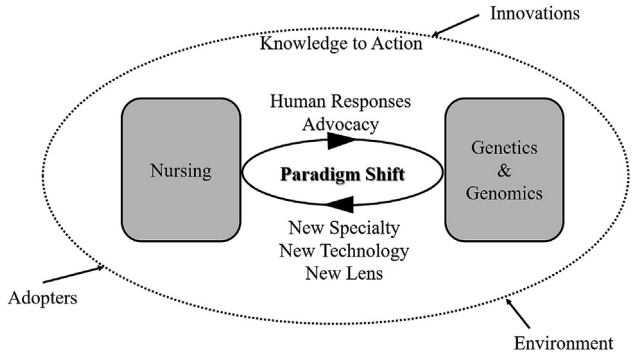

Fig. 2. Interaction of nursing with genetics and genomics.

range theories is to direct the prescriptions of micro-range theory aimed at concrete practice and prediction [17], this proposed framework could direct further studies on measurement and interventions of genetic nursing care and also refine the substantive content of nursing science in genetic and genomic field.

4. How genetics and genomics could be applied in nursing discipline?

4.1. Genetics and genomics as a new specialty

Since 1990s, a small number of nurses have obtained training and education to become genetic nurses and specialize in genetic and genomic health care [26]. In 2002, at the 15th Annual ISONG Conference, five genetic nurses shared their visions and experiences for how nurses could become specialized in genetic and genomic health care [27]. The ISONG developed the scope and standards of clinical practice and professional performances for nurses in genetics and genomics in 2007 and updated the guideline with and American Nurses Association in 2016 [28]. These specialty nurses get involved in clinical genetics providing genetic testing, diagnosis, counseling and management to individuals and families with or at risk of genetic conditions, such as chromosomal abnormalities, single gene disorders, birth defects or hereditary cancers [28]. However, as genetics and genomics influences all domains of health care and patient outcomes across the whole life span [29], it should not only be considered a specialty in discipline of nursing, but also a new technology for all nurses to use in their practices in the future.

4.2. Genetics and genomics as a new technology

Advances in genetics and genomics led to rapid changes in health care and added new scientific technologies to the health care practice. These genetic and genomic technologies, similar to informatics, mobile devices and artificial intelligence, are enhancements and extensions of the clinical practices nurses implement every day since nursing became a profession [30]. If nurses integrate genetic and genomic science into their assessment, these assessment practices may include: obtaining family history, developing and updating family pedigree, asking about and evaluating environmental factors (e.g. diet, exercise, sleep patterns, etc.) influencing gene expression, evaluating signs, lab results and symptoms related to genomic conditions, asking individuals and their family members about what they know or want to know about the genomic aspects

of their condition [31,32]. Based on a more comprehensive assessment, nurses could make a more personalized care plan catering to each patient's or family's specific needs. The nursing care plan can also be initiated according to patients' or families' genetic and genomic conditions. The nursing care could include but not limited to: educating the individual or family about genomic factors related to the individual's or family's conditions and other pertinent information, teaching the individual or family about risk factors for health conditions based on the family history and pedigree information, discussing preventive measures, available genetic testing and treatment options, and assisting with arranging a genetics referral if indicated and needed [31,32].

4.3. Genetics and genomics as a new lens

Genetic and genomic science brought new lens to nursing discipline and changed the way in which nurses think about health and disease and also the ways in which nurses deliver health care, including nursing assessment, diagnoses, interventions and management, illness prevention and health promotion [33]. After the Human Genome Project was completed, multiple genetic variants or changes have been illustrated as being associated with the development of all common diseases [4]. Detecting the presence of genetic and genomic variants could enable nurses to stratify patients into different genetic levels of disease categories, from which prevention or treatment methods could be provided accordingly [4]. For example, some forms of diabetes are directly related to a change in a single gene [34]. Some patients who are diagnosed with type 1 diabetes can now be tested for one of monogenic diabetes. The appropriate treatment for these patients is not injecting insulin, but giving oral sulfonylureas [34]. Moreover, it is now well understood that a large proportion of variation in response to medications is also genetically determined [35]. Thus, pharmacogenomics has been established and the goal is personalized medicine, which is to identify genetic differences comprehensively among persons and to correlate specific genetic features or combinations of genetic features with the differential risk of human diseases or the efficacy of certain therapeutic interventions [36]. For example, tricyclic antidepressants are metabolized by the CYP2D6 enzyme. A person who has a variation in the gene coding for the enzyme that makes him or her a poor metabolizer could have toxic plasma concentrations and side effects such as dry mouth, hypotension, sedation, tremor, or event cardiotoxicity. The idea of using individual genetic and genomic profiles could not only guide nurses' assessment, monitoring and evaluation before, during and after using medications, but also provide a direction to nurses in using this information to personalizing nursing interventions.

5. How nursing discipline could contribute to genetic and genomic science?

A Genomic Nursing State of the Science Advisory Panel, in 2012, developed a nursing research blueprint for genomic nursing science, including full range of genetic and genomic research in all care settings and diverse populations [16,37,38]. Nursing discipline could bring particular worldviews and different perspectives to genetic and genomic science not only through research, but also through clinical practice, education, and leadership, as nursing discipline is a distinct health care science and a stable epistemic community [39]. Nursing is defined as "the protection, promotion, and optimization of health and abilities, prevention of illness and injury, facilitation of healing, alleviation of suffering through the diagnosis and treatment of human response, and advocacy in the care of individuals, families, groups, communities, and populations [40]." While the medical profession has typically focused on the interaction of genes and disease [41], nursing is focused more on the how genes interact with human response and how nurses advocate for their clients in genetic and genomic era.

5.1. The interaction between genes and human responses

Most nursing interventions are targeted towards management of symptoms or symptom clusters, which is one type of the patient responses. A strong research interest in identifying the impact of individual genetic and genomic variation on symptoms and symptom management has emerged and this research direction has also been listed in the blueprint for genomic nursing science. Recent studies demonstrated that the intensity of irritable bowel syndrome symptoms, such as diarrhea, bloating, colonic transit delay or even perception of the illness, are linked to different genes [42–45].

Nursing is also more focusing on psychological or behavioral responses, such as posttraumatic stress disorder, cognitive impairment, smoking, obesity and sleep [41]. If more research showed that these psychological or behavioral responses are truly gene related, nursing will modify the traditional perceptions of these issues and develop more appropriate interventions to optimize patient outcomes [30,33].

5.2. Advocacy for their clients in genetic and genomic era

This new knowledge, genetics and genomics, and its applications within the delivery of health care carry with psychological, social, ethical, and legal issues related to testing, recording, sharing and storage of genetic/genomic information [46]. Nurses must be anticipated and prepared to advocate for the rights of all individuals by addressing issues and problems associated with the vast scientific progress in human genetics [46,47].

5.2.1. Psychological and social issues

The genetic/genomic information could lead to potential emotional, psychological and social stress on individuals, family members, and communities. Nurses could use holistic nursing principles to guide the process of providing genetic and genomic information in a nondirective and supportive way that allow patient and their families to make informed decisions that are best suited to their needs and values [48,49]. Nurses could assess individuals' culture, knowledge, language skills and developmental stage, tailor the information to meet the needs of individuals with divers educational levels and learning needs, and then use appropriate communication to increase their understanding of genetic and genomic information [50]. Also, nurses should be aware of personal values and beliefs of individuals or families which would influence the nursing care and the provided support. Respecting patient autonomy is an important guiding principle [50]. Finally, nurses could work in a partnership with patients, family members and other professions in the management of psychological and social issues, recognize the potential expertise of individuals, family members with genetic/genomic healthcare needs and also ensure the appropriate referral to other genetic specialties if necessary [50].

5.2.2. Ethical issues

There are many ethical issues that have surfaced along with advances in the science of genetics and genomics. Nurses must ensure that use of technology and scientific advances are compatible with the safety, dignity and rights of people. Principlism is the most commonly used approach in the ethics of genetics and genomics in clinical care, including autonomy, confidentiality, beneficence and non-maleficence [5]. An ethical problem arises when these important ethical principles contradict with each other. Genetic information is different from other kinds of health information, as it is

more sensitive and provides information about the person being tested and about his or her family members [6]. For example, one of patient's paternal grandfather has Huntington disease. The patient chooses to have predictive genetic testing to see if he carries the allele for Huntington disease and decides whether he/she needs any preventive treatments, but his/her father doesn't. When the nurse gets the results showing positive, the nurse knows something not only about the patient, but also about his/her father. In one hand, nurses have a duty to abide by the ethical principle of beneficence and need to make decisions based on what will benefit the patients and families. In the other hand nurses need to adhere to the ethical principle of patient autonomy, confidentiality and non-maleficence. In this case, the duty to keep the patient's genetic information confidential may be stronger than the right of his or her family members to be warned that they are at risk. Most recommendations of professional and governmental organizations are trying to balance the principles of maximizing benefit and minimizing harm, even if it is only psychological harm [6].

5.2.3. Legal issues

Advances in genetic testing and genetic health care promise to make things better for most people seeking treatment for many diseases. However, many Americans report that they are afraid of having genetic testing done because they do not want to be victims of genetic discrimination [6]. They are afraid that they might pay much higher premiums for health insurance or lose jobs if their increased genetic risk for a major health problem is documented on their health records. Although the Genetic Information and Nondiscrimination Act was signed into law in the USA in 2008, more legal issues need to be considered and addressed [6]. Nurses are obliged to participate in international or national interdisciplinary organizations and conferences to actively represent nurses' voices with issues in genetics and genomics [27]. Also, they could engage into the oversight and regulations of genetic testing and other genetic technologies and ensure equal access among all populations [27]. More importantly, nurses could support and get involved in national legislation that advocates genetic privacy and prevent genetic discrimination [27].

6. Paradigm shift

The process of nursing profession actively applying and contributing to genetic and genomic science could lead to a paradigm shift in nursing science. Paradigm shift is a concept developed by Thomas S. Kuhn in the most cited book of twenties century: The Structure of Scientific Revolutions [51]. In his theory, paradigm shift is the critical point of scientific revolutions happened in normal science [51].

Normal science means that research firmly based upon one or more past scientific achievements, which some particular scientific community acknowledges for a time as supplying the foundation for its further scientific activity and practice [51]. Paradigm is the matrix of the community rules and commitments shared by practitioners of a particular scientific field, including specific vocabulary and discourses, accepted theoretical claims and established scientific techniques for obtaining the previous achievements [51]. Same to other scientific communities, paradigms help nursing profession to bound their discipline through creating avenue of inquiry, formulating questions, selecting methods, defining area of relevance, and establishing or creating meaning. However, a striking feature of normal science is that doing research is to discover what is known in advance and results from the research add to the scope and precision of the paradigm which is being applied [51]. Thus, normal science is considered conservative, with scientists building on rather than questioning existing science [51].

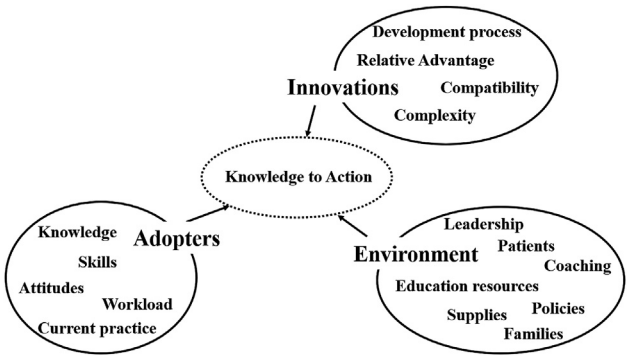

Fig. 3. Three factors influencing knowledge translation.

However, the stage of normal science is not stable and will be changed when new or abnormal phenomena are repeatedly uncovered by scientific research in its own or other field [51]. In this case, advances in genetics and genomics are the new phenomena. The incremental findings in genetics and genomics are loosening theoretical stereotypes in nursing discipline and are necessary for a fundamental paradigm shift, but it is just a beginning. The paradigm shift will be complete when the paradigm has been adjusted so that the new specialty, new lens and technologies of genetics and genomics become expected in the shifted paradigm. The result of the paradigm shift is a scientific revolution making nursing professionals able to see nature in a different way [51].

7. Implementation strategies to facilitate the shift

Scientific revolutions are non-cumulative developmental episodes and lead to significant changes in ways of thinking and doing [51]. As the implications of the changes are discomforting and far-reaching, it is difficult for individuals involved to embrace and integrate the discoveries into their daily practice. Previous studies showed that education initiatives alone are not enough to bring genetic and genomic science into nursing discipline. Knowledge translation activities are necessary to be applied to facilitate the process [52–56]. Actually, more theories and findings showed that assessing barriers and facilitators and tailoring implementation strategies are one of the most important steps in the process of knowledge to action [57]. In this framework, three key factors categorized by Ottawa Model of Research Use, which can influence knowledge translation by tailoring implementation strategies, were highlighted [58]. These three factors are evidence-based innovation, potential adopters and practice environment (see Fig. 3) [58]. More recently, leadership has been suggested as a critical and independent factor in facilitating and sustaining knowledge translation among different professions [59,60].

8. Conclusion

A theoretical framework was synthesized and developed based on the published articles regarding nursing discipline and genetic and genomic science. We believe that the proposed framework can promote the application of genetics and genomics into nursing discipline by helping to understand how genetic and genomic science could be integrated into nursing discipline.

Conflicts of interest

The authors disclose no conflict of interest.

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Appendix A. Supplementary data

Supplementary data related to this article can be found at https://doi.org/10.1016/j.ijnss.2018.08.004.

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