

REVIEWS

Genetic nursing: Reflections on the 20th century

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Abstract

Background: Nursing has a long tradition in caring for individuals, families, and communities who have or are at risk to diseases that are genetically mediated. A comprehensive review of genetic nursing literature is described in order to better understand priorities, gaps, and future directions. A comprehensive review of the genetic nursing literature in the 20th century is important because it reflects how nursing used borrowed knowledge from medicine and applied it to nursing practice. We propose that nursing developed practice based knowledge in the 20th century and this provides a valuable example of how our discipline incorporates new science and technology.

Objectives: This paper describes the results of a bibliometric study designed to analyze 1,067 publications collected between 1906 and 1999 that represent the vast majority of literature covering the field of genetic nursing. Our purpose is to provide an overall summary of where genetic nursing has come over 100 years and to make overall suggestions about future endeavors to advance genomic nursing competencies. Our intention is to describe the historical integration of genetic and genome science into nursing knowledge and application to nursing practice in the 20th century.

Methods: This study collates a wide variety of publications including qualitative and quantitative research, editorial or opinion papers, case reports, clinical updates, science application and utilization papers, concept analysis papers, and previous evidence-based reviews. Data abstracted from 1,067 publications reported in frequencies and percentages provides descriptive analysis of the evolution and translation of borrowed genetic and genomics science into practical knowledge for nursing.

Results: This study illustrates that nursing knowledge is generated not only by nursing research but also from application and synthesis of borrowed knowledge from molecular, cellular, biological, and medical sciences and technologies taught in genome science, clinical genetic medicine, and the discipline of genetic counseling. Nurse and non-nurse authors collaborate to contribute to the translation and transfer of genetic and genome science into nursing education, research and practice.

Conclusions: This study clearly illustrates that nursing developed practice-based knowledge adopted from the specialty of clinical genetic services and applied it to nursing practice. This historical review describes 100 years of genetic science in healthcare used by nurses to better care for patients and families. Nurses in the future will be challenged to develop educational curricula, strategies and tools to measure competency in genomic nursing. Future collaboration across disciplines and in the context of academic practice partnerships would greatly aid nurses' efforts to translate genetic and genomic science into practice, education, research, and healthcare policy.

Key words

Genomics, Genetics, Genetic nursing, Bibliometrics, Competencies, Evidence-based practice, Nursing research

Background and significance

The human genome was mapped in 2003^[1] and more than 3,000 gene tests are now available for genetically mediated diseases and conditions^[2]. Professional standards of care are incorporating genetic testing for screening, diagnosing, and treating the genetic component of the ten leading causes of death in the USA^[3] including a variety of common complex diseases such as cancer, heart disease, neurologic disorders, mental health, diabetes, hypertension, and obesity.

Information about genome science is on the Internet, in libraries, popular magazines, newspapers, and textbooks this puts genetic and genomic information at their fingertips of nurse executives, educators, managers, clinicians and students. However, they must know how to access, analyze, interpret, and make sense out of it, before they can use it in nursing curricula, for quality improvement projects at the bedside, and for designing research aimed at modifying patient behavior and health outcomes in a genomics era.

Even with publication of the scope and standards of genomic nursing practice^[4], and the genomic nursing competencies for graduate^[5] and undergraduate students^[6] the uptake of genetics and genomics knowledge into academic and clinical curricula, and RN performance competencies lags well behind this rapidly advancing science. The nursing professional has not yet integrated genetic science into education or practice in a systematic or standardized manner.

We undertook a synthesis of a comprehensive historical review of the genetic nursing literature. The ultimate aim of this evidence-based practice study was to describe the history of integrating genetics knowledge into nursing's body of knowledge and to identify recommendations for incorporating genetics knowledge into nursing practice, education, and research based on lessons learned in the 20th century. Retracing the historical evolution of the genetic nursing literature can help future generations of nurses understand how nursing as a practice profession translates evolving new sciences into nursing practice.

Study design

This evidence-based practice study uses a descriptive bibliometric design as described by Cooper^[7], Ganong^[8], Borgman^[9], Broome^[10] and Pritchard^[11]. We quickly realized that a traditional evidence-based practice design would not fit our purpose of a historical review. One objective of this bibliometric design was to develop and test a systematic method of capturing, coding, and analyzing diverse types of genetic nursing literature in periodicals, including research and non-research, clinical papers, opinion papers, and newsletters, chapters in textbooks, and monographs. By selecting bibliometrics, the intent is to explain how genetics and genomics science and technology are translated into bedside nursing practice, education, and research.

Descriptive bibliometrics is defined as "the application of mathematics and statistical methods to shed light on the processes of written communication and on the nature and course of development of a discipline (in so far as this is displayed through written communication), by means of counting and analyzing the various facets of written communication"^[12, p. 294]. Another definition explicit to our historical purpose and the research design is, "the assembling and interpretation of statistics relating to books and periodicals...to demonstrate historical movements"^[13, p. 348]. The purpose of this study is to describe a historical overview of the integration of genetic science into nursing practice, education and research across the 20th century.

The following steps depict the process of this bibliometric study: a) formulating the purposes and research questions; b) defining 'genetic nursing literature'; c) developing inclusion and exclusion criteria; d) conducting the literature searches

and systematically organizing the literature; e) designing a data abstraction/ collection tool including 31 variables; f) pilot-testing of the tool on a sample of 200 articles, then revising the tool; g) reading, coding, and analyzing all the articles; h) running descriptive statistics to analyze the data; and i) interpreting the findings in an historical trajectory over the 20th century.

The aim of the study is to explore the following knowledge development and science translation questions:

- 1) In what clinical specialties is genetics introduced into nursing literature and how has that changed over time?
- 2) What nursing concepts, models, and theories are applied to caring for patients who have or are at risk for a genetic condition? What concepts particular to nursing comprise the phenomena of concern related to genetic nursing for basic and advanced practice nurses, and for researchers?
- 3) How do nurses describe their roles in genetic services? How have these roles changed over time? What are the current models of providing genetic health care and how do nurses fit into interdisciplinary models of delivering services?
- 4) What policies or administrative decisions most influenced nursing's implementation of clinical practice roles in genetics?
- 5) What guidelines do nurses follow when informing patients and families about the ethical, legal, and psychosocial implications of genetic testing?
- 6) Who in nursing is involved in conducting programmatic research in genetics? In what ways does this body of knowledge call for change in nursing practice?
- 7) What patient outcomes are identified by nurse authors as important to patients in the context of genetic healthcare?

For our study, we defined 'genetic nursing literature' as a publication that contained discussions about nurses, nursing science, education, ethics and the practice implications for nursing in the context of genetics which is the study of genes (defined as the principles and concepts of genetics such as mutations and the biological mechanism of inheritance, or human diseases arising from single gene or multifactorial disorders).

Literature searches

An examination of the Cumulative Index to the Nursing and Allied Health Literature (CINAHL) for the years 1906-1999 revealed that of the 121,961 journal articles that discuss nurses and nursing, only 726 (0.6%) discussed genes and/or genetics within nursing; and 569 (0.5%) discuss embryology congenital anomalies and "birth defects". Published articles were ascertained via search 33 databases dealing with nursing, health care, medicine, pharmacy, allied health, education, ethics, psychology, and sociology. Total citations obtained from all databases included 6,230 individual citations. The strategy used to search all possible literature databases was based upon a method developed by Cooper^[7]. Two hundred and two searches were conducted. Of these searches, twenty were conducted by hand on databases available only in print spanning the years 1900 through 1980 (i.e., CINL, CINAHL, and NSI). Thirty-three individual databases and indices were searched using 41 keywords (see Table 1).

The first task was to enter the keywords, nurses and nursing, then combine these with terms such as genes and genetics, then combine these with 41 other keywords until all possible combinations and even overlapping citations were captured. Identification of the key words was an iterative process whereby the list grew as new terms were identified in the literature itself. All databases (both electronic and print) were searched using the same key words and search strategy. Each publication retrieved was examined using the inclusion and exclusion criteria to determine whether a publication fit our definition of genetic nursing literature. The full citation of each article was entered into bibliographic software (Procite) to

create a master bibliography, every articles was coded by a ‘case’ number, for easy retrieval of both the article and the completed data abstraction form. A second copy of every publication was marked with the same code number and stored in file folders labeled by a theme from an inductive qualitative analysis. Each co-investigator stored a complete set of all the publications.

Table 1. Key Words

Nurse	Hereditary disease
Nursing	Tumor suppressor
Genetics	Medical
Genetic disease	Inherited disease
Genes	Nursing practice
Oncology	Nursing education
Oncogenes	Nursing models
Neoplasms	Patient education
Chromosomes	Philosophy
Chromosome abnormalities, aberrations	Knowledge
Congenital anomalies, abnormalities	Research
Birth defects	Utilization
Prenatal diagnosis	Review
Fetal	Observation
Cancer	Social support
Genetic	Nurse Practitioner
Counseling	Spina bifida
Fetal abnormalities	Hemoglobinopathy
Fetal disease	Sickle cell
Infant disease	Cystic fibrosis
Newborn disease	

Inclusion and exclusion criteria

Publications were included if they met the inclusion criteria using our definition of ‘genetic nursing literature’. They were published in nursing and non-nursing journals and included nurse and non-nurse authors. Another criteria for inclusion required publications to discuss patients, families, and communities with or at risk for a disease with a single gene mutations, a genetic inheritance pattern in the family or a chromosomal aberration. The following types of literature were excluded: conference proceedings, abstracts of papers, theses and dissertations, popular magazines, newspaper articles and papers published by nurse coauthors with physicians in medical journals with no mention of nursing implications. All articles had to include a discussion of nursing implications; therefore, articles by nurses and non-nurses that were published in nursing or other journals that did not contain nursing implications were excluded from this study.

Creating the data abstraction form

A qualitative content analysis described by Borgman ^[9] in the field of bibliometrics was undertaken to identify broad themes and phenomena of concern discussed by the authors in the genetic nursing literature. The entire pile of articles was sorted by content area into one of 18 themes by reading the abstract; each paper was assigned a numerical code derived from the theme. Two articles from each of the theme categories were randomly selected for a full review by both co investigators; the purpose was to identify the content areas that make up the concept of genetic nursing knowledge by reading across a spectrum of content areas and types of articles. This analytic discussion and process resulted in identification of 31 variables and their relevant sub-concepts which were identified and coded for each article (see Table 2).

The data abstraction form was pilot tested on 200 articles with double coding by three reviewers which yielded an inter-rater reliability rating of 0.78 using the Cohen Kappa; then all 1,067 articles were coded.

Table 2. List of variables in the data abstraction form used to code every publication for data collection and analysis

Year	Ethical implications discussed
Country of Author	Legal implications discussed
Name of Journal	Social implications discussed
Type of document	Religious or spiritual implications discussed
Continuing education credits (CEUs)	Political/policy implications discussed
Number of authors	Phenomenon of concern to nurses in the context of genetics
Author credentials	Concerns experienced by patients as discussed by authors
Certification	Nursing interventions discussed in the context of genetics
Non nurse author discipline	Genetic nursing interventions named in the context of caring for patients/ families
Population discussed in the paper	Patient outcomes
Body system as the area of focus	Clinical practice recommendations/ guidelines/ standards of nursing practice in genetics
Clinical practice setting	Conceptual models and theories used to frame discussion of genetic nursing practice/ education/ research
Type of topic	Policy statements and recommendations
Research based publication	
Research approach	
Type of genetic science knowledge included	
Type of genetic phenomena identified and discussed	

Data abstraction/collection

Each publication was read, then checked for accuracy in terms of the inclusion and exclusion criteria, and coded using the data abstraction instrument. One reviewer read each article, then reviewed each question on the data abstraction form to code the data by circling every subcategory of content in every variable that was identified (or discussed) in the article. The reviewer based every coding decision on a binary question: was the content included or not included. Each coded data abstraction form was identified with the same code number as the publication and was stapled to its respective publication. This set of articles was stored alphabetically (first author's name) by year. An Excel spreadsheet was created to enter the data for the 31 variables including a variable number of sub-concepts. At frequent intervals, the same single reviewer transferred the data from the data collection form to an Excel spreadsheet using the 'case number' for each article. This strategy enabled checking inter-rater reliability as well as accuracy of data entry. The SPSS file was backed up on two computers and a storage disk.

Statistical analysis of the data

The data from each data abstraction form was entered by 'case number' into an Excel spread sheet and copied into SPSS. The coded data were analyzed using the Statistical Package for the Social Sciences (SPSS), version 19 for Windows. The initial phase of data analysis entailed frequency tables and scatter plots of all variables to check for outliers and accuracy of data entry. Descriptive statistics were conducted to report frequency and percentages.

A description of the genetic nursing literature in the 20th century

In total 1,067 publications were analyzed and included in this analysis. Two-thirds of the literature was published during the decade of the 1990s. Nearly three quarters of the literature (69.6%) was published in the United States (US), and over 20.0% was published in the United Kingdom (UK), and 4.4 % in Canada. Countries of publication include Australia, New

Zealand, Singapore, and Jamaica. In all, 700 of the 743 articles published in the US appeared during the 1980s and 1990s. Eighty-seven percent of the literature appeared in nursing journals. Articles appeared in 250 journals with the majority of authors publishing only one article. The journals: *Oncology Nursing Form*, *American Journal of Nursing*, *Home Healthcare Nursing*, *Public Health Nursing*, and the *Journal of Continuing Education in the Health Professions* each published between 60 and 22 articles. Most journals published one or only a few articles.

The most frequently recorded authorship was among Master's prepared nurses (26% of first authors and 41% among all authors). While many publications did not list the author's credentials (12.8%), over two-thirds of the first authors were nurses. Just under a fourth of the literature (20%) was published by doctorate prepared nurses as first authors; 8% of nurse authors held a bachelor's degree. Approximately 8% of the first authors were physicians with nurse co-authors and 11% of these articles were published in *medical journals*. The data suggest that nurses who were engaged in pursuing academic advancement were reading and writing about genetics in collaboration with physicians and that collaboration enabled integration and dissemination of this new knowledge. Nearly a third of the authors held a nursing certification (29%) and identified one of numerous nursing specialties (26%). Only a very small percentage of the articles provided Continuing Education Credits (4%). The vast bulk of the literature (85%) was not research-based. The largest proportion of research publications (10%) was categorized as quantitative research: correlational descriptive studies.

The research questions and related findings

Analysis and results of the genetic nursing literature address eight research questions.

Question 1: In what clinical specialties is genetics introduced into nursing literature and how has that changed over time?

While most of the literature (57%) was written for nurses in any clinical setting, half of the articles (50%) focused on the principles and mechanisms of genetics bioscience knowledge such as: DNA mutations, single gene disorders, genetic testing, technology or procedural knowledge for using genetics in clinical practice. Nurse authors diffused genetics into nursing by reiterating borrowed knowledge from clinical medicine and genetic biosciences and described it in the context of diseases, management of patients, or as science based primers. Nurse authors understood that the basic science of genetics had to be included in nursing literature in order for nurses to incorporate this new knowledge into practice.

Sixteen percent of the publications discussed adult genetic diseases in acute care settings, while 20% of the publications described nurses' roles involving genetics in acute care settings. This first question was considered in two major parts—the target population (such as children, adults, families, communities) and the body system or clinical specialty area of concentration (such as cardiovascular, respiratory, cancer/oncology). Nurse authors diffused the genetic component of disease in two domains: a) descriptions of common diseases affecting large segments of the population and b) patients affected across their life span (e.g., cancer). Conditions that largely affect only adults (both men and women) were second most frequent (e.g., Alzheimers disease), followed by conditions that affect children (including adolescents e.g., cystic fibrosis) and infants (e.g., Down Syndrome). Conditions that affect pregnant women (such as prenatal diagnosis and reproductive technologies) were fourth most frequent.

The literature at the beginning of the 20th century contained reports of conditions affecting newborns and children. Discussions about conditions affecting adults, families, and communities, including overview discussions of genetics (or genetic primers) were added in the 1950s. Discussions of genetics with regard to pregnancy and reproduction began in the 1960s with identification of *inheritance patterns involved in 'birth defects'* which paralleled the use of genetic technologies to detect chromosomal aberrations and rare single gene disorders diagnosed prenatally, or in infancy, childhood, or adulthood. The emergence of nursing content pertaining to genetics paralleled developments in genetic bioscience at that time. For example, delayed physical development in newborns was a content area in the earliest published articles. Cystic fibrosis, muscular dystrophy, hemophilia, and mental retardation were topics for prescribing

acute and home care nursing practice in the 1950s. Discussions of disease prediction and prevention (newborn screening and treatment for phenylketonuria, for example) started in the 1950s and proliferated in the 1960s. “Mental subnormality” and “congenital dislocation of the hip” were topics of concern in the 1960s. Prenatal genetics and pediatric conditions appeared next along with emerging discussions about genetics and cancer. Psychiatric conditions such as depression and Alzheimer’s disease, ethics and healthcare policy topics are the most recent areas of publication in the genetic nursing literature. The clinical specialty of cancer was the largest single area of concentration with articles emerging in the 1980s and growing in volume during the 1990s as cancer genetics science evolved.

Question 2: What nursing concepts, models, and theories have been applied to caring for patients who have or are at risk for a genetic condition? What concepts particular to nursing comprise the phenomena of concern related to genetic nursing for basic and advanced practice nurses, and for researchers?

The most prominent model or theory evident in the genetics nursing literature (37%) was a medical model of disease management that promotes early diagnosis, early intervention, disease prevention, and medical management or cure of disease. The second most prominent was an educational, information-giving model which was seen in 18% of the literature. This paralleled the goals and processes of the new discipline of ‘genetic counseling’ and the goal of genetic testing, in clinical practice by teams consisting of genetic specialty nurses and physicians and later, by genetic counselors and physicians.

The models and theories guiding the genetic nursing literature, indeed, the discipline of nursing, were largely focused on illness management and information-giving (education model) throughout the entire century. Nursing theories were evident in only a small number of publications. The nursing theories of Orem (5 articles), Roy (5 articles), Neuman (2 articles), Watson (1 article), and Leininger (1 article) are surprisingly few considering the importance of nursing theory in the discipline. The nursing literature began to discuss holism, decision-making models, and nursing theories in the 1980s and 1990s. While the models and theories gradually became more diverse by the end of the 20th century, they focused on providing care, support, and education of patients, families, and communities, and remained culturally sensitive throughout the century. A model of illness acceptance, management, and adaptation is discussed in 70% of the literature; while not named as such, this finding indicated wide acceptance of Roy’s model of adaptation. The community health model appeared in the 1900s and reappeared in the late 1990s as nurses became reinvented in public health in the community and population-wide administration of genetic health care services. A community health model including epidemiology, population services, and health services administration was seen in 20% of the literature.

Phenomena of concern to nurses, particularly providing nursing care, information-giving to patients and families, and advocacy was evident in publications across the entire 20th century. Providing support to patients and families appeared as early as the 1950s, and support for family decision-making appeared in the 1970s. The concept of caring appeared in 49% of the literature including discussions of caring and its ramifications in nursing management of health conditions (implementation of the nursing process components of assessment, diagnosis, goal-setting, planning, intervening, and evaluating care). Information-giving and providing health teaching to patients and families was seen in 86% of the literature. Supporting patients and families, while not present as frequently was evident in 62% of the literature.

Most of the literature (99.4%) provided the reader with some discussion of genetic mechanisms, some of which noted only the mode of inheritance. Nearly half of the literature (38%) provided the anatomical, physiological, dysmorphic features, and/or psychological mechanisms; and nearly one half (45%) described genes, DNA chromosomes or proteins involved in the conditions discussed. Approximately 42% of the publications were devoted to discussions of single diseases such as cancer in terms of chromosomal anomalies, single gene disorders, and multifactorial conditions. It is apparent that nurses authors intended to translate genetic and genomic science knowledge into practical knowledge for nurses at the bedside. Approximately 10% of the literature reported research on patients and families in terms of genetic diseases, ethical or psychological issues related to genetic testing.

More than half (66%) of the literature discussed 'genetic risk factors' associated with disease and genetic testing. The second most common genetic concept in the literature was epigenetics (9%) (ie., the relationship between genes and environmental changes that affect protein expression at the cellular level). The use of borrowed knowledge from genetic science was essential to teach nurses about advances in science and technology and to provide a basic understanding of concepts, definitions and principles of genetics and genomics as nurses began to understand patterns of inheritance in families, DNA alterations and single gene mutation diseases, and how personal lifestyles and the environment impacted illness and disease risk in families. Nine percent of the articles contain the concept of a holistic view of health with an understanding of the interplay between risk factors and health or illness and disease.

Question 3: How do nurses describe their roles in genetic services? How have these roles changed over time? What are the current models of providing genetic health care and how do nurses fit into interdisciplinary models of delivering services?

Nursing roles that have a genetic component were coded when they were identified in the text and described here as genetic nursing activities or role expectations. Approximately 218 (20%) of the publications discussed roles for nurses in genetics. In contrast, 39% included updates about the genetic science or technologies related to single diseases and how that knowledge impacted clinical practice such as disease management that informed nurses how to provide or improve patient care. The second most frequent topic of concern to nurses was describing the process of genetic counseling services (21%). This finding is consistent with the expectation that nurses would refer patients and families to this type of service. In contrast, only 16 of the articles (1.5%) discussed family history assessment and pedigree construction as part of the nursing role. A majority of the literature emphasized providing or managing nursing care (80%), giving family support (74%), providing educational materials (74%), fostering hope (67%), reducing upset in patients and families (59%), exploring the socio-cultural aspects of family life (59%), and providing caring, empathy, sympathy, compassion or emotional/ spiritual care to patients (19%). Less than one third of the literature identified a role for nurses in following up genetic services (27%), coordinating health care services after a diagnosis of a genetic condition (26%), or referring the patient and family to a support group (27%). Incorporating new knowledge into practice was emphasized in the context of nurses' existing roles rather than assuming new skills or practice competencies.

Prior to the 1950s, the literature described the nurse's role as information-giving, coordination and follow-up of health care teams that provided specialized services to patients and families who had a genetic condition and disabilities. In the 1950s, the literature began to urge nurses to refer patients and families to support groups. This was consistent with a trend that had been gradually increasing throughout the 20th century as individuals with disabilities began to receive greater acceptance in society. It was not until the 1950s that the nursing literature began to recommend that nurses record family histories and pedigrees; assess family histories for risk of a genetic condition; provide counseling for genetic conditions; and make a referral to a specialized genetic service program where family history taking was known to be part of the genetic counseling service.

Collectively, only 14% of the literature drew attention to the nurse's personal values and beliefs, ethics, or cultural sensitivity as a component of the applications of genetic technologies in health care. Nursing roles relating to decision-making by and advocating for patients and families, was present in 5% of the literature. Nurses were expected to incorporate genetics knowledge into their practice in clinically relevant specialties that affected people across the life span. Over time, nurses progressed from concentrating on delivery of direct care services to coordinating health care teams, case management, administration of genetic health care information in specialty clinics, and leadership in health care research related to genetics services in collaboration with physicians.

The findings indicate that nurses' responsibility in care at the bedside involved understanding genetic science knowledge and how genetic phenomena create risk for genetic conditions in families. Gene identification discussions first appeared in the 1960s and all forms of genetic phenomena, including the concepts of genetic linkage, genetic testing, and gene-based therapeutics proliferated across the 1980s and 1990s as the genetic and genomic science emerged.

Question 4: What policies or administrative decisions most influenced nursing's implementation of clinical practice roles in genetics?

Only 56 of the publications or 4% of the literature offered continuing education credits (based upon submission of test answers on a score sheet). These articles were designed to provide instructional information to the reader and offered a readily accessible form of genetic nursing education. This finding is very surprising considering that continuing education is mandatory for licensure and is an effective way to disseminate new knowledge into a practicing discipline.

The genetics nursing literature shows that nurses have taken leadership roles in many areas including but not limited to research, practice, education, policy, and administration. The recommendations made for these roles have come mainly from publications in the 1990s. These include expanding nursing knowledge specialization to keep pace with the trend of sub-specialization, proliferation of managed care and case management responsibilities, respect for patient autonomy and bringing the patient's voice into health care practice and policy arenas. These published authors encouraged nurses to be involved in providing direct care and education to patients, families, and communities about genetic conditions and possible risks for genetic conditions. Actual policy recommendations were sparse in the literature. There is little evidence that nursing contributed to policy development in the broader fields of public health, health promotion, school nursing, or human rights and research ethics as related to genetics and genomic healthcare.

Because 65% of the literature was exclusively authored by one author and 31% of the literature authored by two to five authors, cooperation between nurses with other health care providers is clearly evident. The contributions of non-nurse authors to nursing's body of knowledge supports collaborative roles in delivery of services to diverse populations, and demonstrates the importance of cross-disciplinary involvement in clinical genetics research, and collaborative efforts to disseminate genetic science and technology knowledge. Policy related to collaborative practice, education or research for nursing was non-existent.

Question 5: What guidelines do nurses follow when informing patients and families about the ethical, legal, and psychosocial implications of genetic testing?

Approximately 790 publications (71%) discussed social issues surrounding a particular health condition and its genetic aspects. These issues include socioeconomic, political, cultural, interpersonal (familial and community) concerns. Specific references to social status, cultural practices, and intra-familial relationships affected by genetic status were coded as present, if they appeared in the publication. Discussions of social implications (including family, community, socioeconomic, and cultural aspects) were sparse until the 1950s, with the largest segment of literature (533 publications) appearing in the 1990s.

Legal implications of genetic science and health care with a genetic component were sparse (8 articles) prior to the 1980s. In the 1990s, 223 publications appeared (20%) with discussions about laws, public policy, and regulations associated with genetics, genetic status, screening, and related topics. The literature provides nurses with knowledge about laws, regulations, and policies with regard to genetic testing. The impact of these on patients and families was not explored in any detail.

Over half of the literature (53%) emphasized the ethical principle of justice (i.e., access to care and information) and the topic received some mention in 93% of the literature. The second most frequently discussed area of ethics was beneficence in 49% of the articles; discrimination on the basis of genetic status was discussed in 29% of the literature. Approximately 45% of the literature had some discussion of the principle of autonomy, personal choice, and informed consent with regard to genetic services, testing, and biotherapeutics. The literature reveals that nurses were focused on the traditional principles of justice, beneficence, and autonomy when providing information about the ethical implications of genetic testing. Narrative ethics was discussed in two articles and issues related to an ethic of caring or feminist ethics were not mentioned.

Discussions of ethical implications of genetics and health care were present from the beginning of the genetic nursing literature with the ethical principles of justice and beneficence appearing from the 1900s onward. However, in general, discussions of ethics were sparse until the 1960s when autonomy received attention more widely in healthcare. Again, the 1970s had relatively few contributions discussing ethics, but the diversity and depth of discussions increased in the 1980s and 1990s. As mirrored in the medical genetics literature, questions and concerns about confidentiality and discrimination received considerable attention in the last twenty years.

Approximately 103 publications (9%) discussed religion and/or spirituality surrounding a health condition with a genetic aspect. These include descriptions of prayer, support of God, and other spiritual aspects of health and illness; two thirds of this literature (70) appeared in the 1990s.

Question 6: Who in nursing is involved in conducting programmatic research in genetics? In what ways does this body of knowledge call for change in nursing education and practice?

All the research-based reports (a total of 175) used descriptive or quasi-experimental methods; in the 1960s two studies based on qualitative methodologies appeared. The largest segment (133 reports) appeared in the 1990s, of these, 100 reported using quantitative methods. Discussions of pharmacogenetics were sparse (nine articles total) in all decades.

Nurses authored the majority of nursing research literature in genetics. Only a handful of researchers are publishing articles based on a program of research. Nurse researchers, most often conducted a single study. The genetic nursing research literature supported nursing practices that emphasize giving information about genetic risks, counseling patients and families about genetic concerns, providing educational materials, fostering hope, reducing fears, listening, and exploring the socioeconomic and cultural features of family life.

The nursing research literature, particularly in the past two decades, called nurses to continue their commitment to caring for clients in culturally sensitive ways even when these may not have economic advantages for healthcare. Exploring the economic value of caring, listening, and fostering hope among families in nursing tends to be less valued for nurses in genetics. The literature over the 20th century shows that nurses systematically provided care, education, and support for persistent concerns for patients such as: lack of knowledge, fears of death or a limited future life, and grappling with the difficulties of decision-making related to genetic testing and disclosure of test results to family members.

The first recommendations for nurses to increase their knowledge in genetics appeared in the 1960s. Since then, particularly in the 1990s nurses were urged by nursing leaders to become involved in academic education, research, public policy and the legislative process as it relates to genetics. However, there is a dearth of publications on these topics before the 1980s and virtually nothing that spans across international borders or across disciplines.

Question 7: What patient outcomes are identified by nurse authors as important to patients in the context of genetic healthcare?

The most frequent patient concern that authors discussed in the nursing literature was lack of knowledge about their condition and/or genetic health status as seen in 33% of literature that discussed patient concerns. Patients' and families' experience of fear about their limited future life including concerns that a genetic condition could be disabling and/or lead to an early death was present in 19% of the literature. Decision-making appeared in 17% of the literature; and concerns about the economic burden of their genetic condition appeared in 9% of the literature.

Beginning in the 1900s when the earliest publications emerged, the phenomena of concern to patients focused on parental concern over delayed physical development of a child. Later in time, authors included lack of knowledge about a particular genetic condition, and fear of a child's or adult's limited future. Discussions about guilt first appeared in the 1950s; uncertainty, fear, and the full range of concerns appear with greater depth of discussion in the 1980s and 1990s.

Discussions of coping with a genetic condition including stress, anxiety, sadness, deteriorating health, grief, and stigmatization appeared first in the 1960s.

Early in the century and up to the 1960s some nurse authors mentioned prescriptive statements to parents about future reproduction that now would be considered eugenic. Progressively nurses became more sensitive to their own values and beliefs about reproduction and the care of children with disabilities as the science advanced from identifying the cause of 'lethal' conditions to management of chronic conditions. Care of children with cleft lip and palate also appeared in the early decades with attention to the precise feeding techniques and pre-and postoperative care of the surgical site. The genetics of congenital hip dysplasia, hemophilia and the muscular dystrophies were among the first genetic conditions explained in the earliest nursing literature. Nursing of adults and children recovering from corrective procedures to the heart received attention beginning in the 1940s with advances in successful amelioration of congenital heart defects.

The most common patient outcome was decreased morbidity and/or mortality in 410 publications (38%). Increasing knowledge and comprehension of the genetic inheritance pattern in the family for a genetic condition was a concern for patients and families as the most consistently discussed aspect of patient concerns by appearing in nearly every publication. These two patient outcomes cut across the literature in the entire 20th century, as did psychological adjustment, early diagnosis, prevention, and quality of life issues.

The literature began to discuss reduction in stress, anxiety, grief and willingness to use preventive health care measures in the 1960s. The 1970s saw a relative decrease in diversity and depth of discussions and volume of all types of articles. There was an increase in content about comfort with decisions for testing and treatments, family stability, and quality of life during the 1980s and 1990s but only in 2% of the literature.

Discussion

The majority of publications included and described the basic bioscience of genetics in terms of disease risk or the etiology of disease. While risk assessment was identified in 20% of the publications, this concept described the impact of environmental exposure risks on DNA mutations, familial inheritance patterns, and the need for referral to a genetic specialist rather than on an RN's role to conduct interviews to gather family medical histories and construct a pedigree which is necessary to determine risk.

The authors of genetic nursing knowledge in the 20th century included detailed descriptions of terms, definitions, concepts, and principles of genetics and genomics. This finding verifies the importance of nurses learning basic genetic principles and biosciences as new knowledge emerges and evolves across time. The core competencies for undergraduate nurses define genetics as the "study of individual genes and their impact on relatively rare single gene disorders" [6, p. 20], and identifies the need for nurses to use this knowledge in patient education and nursing interventions. This systematic review of the genetic nursing literature describes in detail the type of genetic bioscience knowledge and its relevance to practice more clearly than stated in the core competencies. Acknowledgment of the basic sciences needed for nurses is an essential step in developing educational curricula and tools to measure competency. This literature showed that nurse authors described RN roles as giving information, *referring to a genetic specialist* to assess genetic risks, educating patients, families and communities, providing psychological and decision making support and accessing resources, and information when facing genetic illness and/or the risk for genetic illness. Nurses were concerned with helping patients and their families attend to nursing care needs of affected family members, support coping with fear, anxiety, grief, suffering and increasing family stability, use of prevention information, and autonomous decision making. During this historical period, nurse authors documented the inclusion of family history taking or pedigree construction in nursing roles in less than 3% of the literature. This finding is echoed in recent literature [14] that reveals that nurses in practice still question whether family history taking and pedigree construction are part of their role. Nurse leaders in education and clinical practice still face the challenge of incorporating this core competency into RN performance competencies and practices. Publications by nurse educators in the 20th century were minimal but all authors reiterated the need for genetic education for nurses. A

few authors emphasized the need for genetics content in textbooks and undergraduate curricula, preparation of nursing faculty to teach genetics, and genetic education offered by national nursing organizations. Standardization of curricular content based on the findings in this review in combination with published scope and standards of practice and the core competencies would greatly facilitate nurses' opportunities for continuing education and academic programs that specialize in genetics and genomics.

Conclusions

This bibliometric study covered the entire 20th century of published genetic nursing knowledge from the very first printed articles and books beginning in 1906. This review encompasses the largest compendium of publications discussing genetics and nursing gathered to-date. The task of archiving and analyzing all these references, while time consuming and tedious, provides valuable information for describing the integration of genetics into nursing practice, education and research. An understanding of the key features of this literature draws attention to the need for concerted effort on the part of Deans, nurse educators and nursing executives to demand genetic nursing education for all students and practicing nurses and to advance education and credentialing for an advanced nursing practice specialty in genetics and genomics. Academic, clinical and continuing education should be developed to increase nurses' basic and advanced practice knowledge of genetic and genomic aspects of disease, prevention strategies and uses of genomic science in health care. The current challenge for nurses is to incorporate genomics, epigenetics, pharmacogenomics, and personalized medicine into practice and education while researching the impact on individuals, families, and societies.

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