Published in final edited form as:

Biol Res Nurs. 1999 October ; 1(2): 133–146.

State of the Science: Social, Psychological, and Ethical Nursing Research in Genetics

Gwen Anderson, RN, PhD

Keywords

Genetic nursing research; integrative review; ethics; genetic counseling

Integrative reviews of the literature are generally understood as one way scholars and students develop new knowledge within the discipline of nursing (Ganong 1987). This literature review integrates published nursing research pertaining to the social, psychological, and ethical implications of genetics for patients and for nursing practice. A synthesis of this literature enables the entire nursing profession to understand where we have arrived and where we are going (Feldman 1971) in terms of incorporating genetic science into nursing practice. My goal is to provide concrete suggestions for journal editors and leaders in continuing education who disseminate this knowledge to nurses. An understanding of where the cutting edge of nursing research lies will help nursing leaders envision how they can promote future growth and development of genetic knowledge for and about nursing. They will better understand how to present this knowledge to nurses who are already interested in genetic information and how to appeal to nurses who think that genetics does not relate to practice.

The first goal of this review is to identify and analyze genetic nursing research published in English that directly addresses the social, psychological, and ethical impact of genetics on nursing, patients, families, and communities. The second goal is to conceptually map the state of the science and the art of genetic nursing research based on a systematic mode of inquiry. The third goal is to interpret and infer generalizations about how this body of research relates to and expands extant nursing knowledge.

Method

Identifying and Collecting the Literature

Genetic nursing research is defined here as a systematic mode of inquiry into a clinical nursing problem or a phenomenon of concern to nurses or to patients regarding genetic research or genetic health care service delivery. Articles included in this integrative review meet the following inclusion criteria: (1) the article is authored or coauthored by a nurse and published in either a nursing or a nonnursing journal; (2) the word *genetic(s)* is used in the title of the article, abstract, or body of the paper; and (3) the topic of the research focuses on social, psychological, or ethical issues that emerge from genetic science, genetic technologies, or application of genetic tests in clinical or research settings. Excluded from this review are research articles coauthored by nurses that are not published in a nursing journal and do not include nursing implications. Also not included are genetic nursing research articles that feature

the biological, cellular, or molecular aspects of human genetics because that content area lies outside the scope of this article.

This compendium of genetic nursing research literature was obtained from online searches in 13 databases, namely, *CINAL*, *MedLINE*, *ToxLine*, *AIDSLINE*, *PsychInfo*, *CRISP*, *Life Sciences*, *HealthStar*, *PsycLIT*, *ERIC*, *Sociofile*, *GreatfulMED*, and *Cancerlit*. Only 43 genetic nursing research articles were retrieved from a search of the literature from 1965 to 1998, even though the first article was published in *Nursing Research* in 1974. These findings are significant to the discipline of nursing because they indicate that nursing practices recommended in the nearly 600 non-research articles published by nurses who are working in the context of genetic health care are not founded on nursing research-based evidence. Nurse authors are relying on scientific research and clinical protocols in genetics developed outside the discipline of nursing, as well as experiential knowledge from their clinical practice. To advance genetic nursing knowledge in the future, many more nurse scientists are needed to conduct programmatic research in genetics from the perspective of nursing (Abdellah 1991).

Mapping the Terrain of the State of the Science of Genetic Nursing Research

The 43 articles described here represent the state of the science and the art of the social, psychological, and ethical aspects of genetic nursing research. To be inclusive of quantitative, qualitative, or triangulated research studies, the author decided against using Cooper's classic description of meta-analysis. Instead, a qualitative method of content analysis described by Borgman (1990) in the field of bibliometrics was developed for this study. Borgman suggests that it is important to examine and analyze the concepts and artifacts that exist within the literature. By *concepts*, he means the terminology or the arrangement of language into meaningful typologies that trace the flow of ideas, values, practices, and concerns that capture the interest of a discipline. By *artifacts*, he means the essential or critical messages being communicated or the embodiment of what the researcher is reporting and communicating to the discipline. Borgman's analytic approach is incorporated into the following method of "modified bibliometric analysis."

Each study was analyzed individually according to all of the following characteristics: purpose, conceptual framework, research question(s), subject population, sample size and sampling method, research design, methods of data collection and analysis, major research findings, and implications for nursing. This analysis was guided by five research questions:

1. What are the themes or topics that nurse researchers are exploring?
2. What conceptual or theoretical models underpin this body of research?
3. What have nurse scientists discovered about the social, psychological, and ethical issues involved in genetics?
4. Is it possible to conceptually map the state of the science of genetic nursing research using themes discovered during this investigation?
5. How does this body of knowledge fit into the entire body of nursing knowledge?

Once the articles were analyzed, they were sorted into one of four themes: (1) family stress and coping, (2) reproductive planning, (3) case finding in communities, and (4) ethical inquiry. The articles within each theme were grouped into subthemes that describe different aspects of the overall topic. After each article was analyzed, a conceptual map or model of nursing practice was drawn to visualize how all studies in a particular theme represent the state of the science for that theme. Common concepts were identified and laid out in a pattern that represents common concepts and theoretical frameworks discussed in the studies. Each model traces the

evolution of research over time and incorporates common concepts or phenomenon of concern and implications for nursing practice, education, and research.

Family Stress and Coping

The greatest number of research studies (23 or 53%) focus on family stress and coping within families in which there is a child who has a chronic illness due to a genetic condition. This theme consists of three subthemes: (1) parental coping, (2) family coping, and (3) discovering characteristics of the child who has a genetic condition. The conceptual model most often used to guide these studies is the Folkman and Lazarus's (1988) theory of stress reduction (Lazarus and Launier 1978), which hypothesizes that effective cognitive appraisal and effective coping strategies facilitate adaptation to illness.

Parental coping was of concern to nurses who worked with families in which there was a child who had a developmental disability. Of the 11 studies, 4 compared mothers' versus fathers' ability to cope within the family context (Damrosch and Perry 1989; Gibson 1986; Heaman 1995; Van Cleve 1989, 1993a; Van Os and others 1985). Most recently, Baroni and others (1997) compared parents of healthy children with parents of a child with cystic fibrosis, in terms of their ability to cope with perceived stress resulting from newborn screening for cystic fibrosis.

The very first study by Rodgers and others (1974) assessed the psychological adjustment of children with cystic fibrosis. Most future researchers used Folkman and Lazarus's (1988) theory of stress reduction as a conceptual framework. They hypothesized that accurate cognitive appraisal and effective coping reduce stress, which facilitates adaptation to illness. Williams (1995), Samuelson and others (1992), and Heaman (1995) used Hymovich's (1988) Parent Perception Inventory to assess family coping. McCubbin (1984) and Gibson (1986) used the Coping Health Inventory for Parents (a scale developed by McCubbin and others 1979). Damrosch and Perry (1989) used a parental coping scale they developed to assess parents' psychological adaptation. Van Cleve (1989) and Samuelson (1992) used the Chronicity Impact and Coping Instrument to assess factors that affect coping. Van Os and others (1985) used the Schedule of Recent Experience to assess parental adjustment to life events. Loebig (1990) used the Impact on Family Scale to assess associations between demographic variables and mothers' self-reports of their ability to cope with the stress of having a child with spina bifida. Baroni and others (1997) used the Parenting Stress Index to measure stress on the parent-child system.

According to Van Os and others (1985), social resources, strength of the marital relationship, and low perceived pressures related to time, money, and direct demands for physical care moderated the effects of stress on health status. The question of how parents support each other emerged for Heaman (1995) as an important variable that required further investigation. Baroni and others (1997) concluded that "the degree of stress related to parenting may be exacerbated by any number of life circumstances, including medical diagnoses as well as a myriad of issues that may not come to the attention of health care providers" (p 150).

Loebig (1990) reported that mothers were dissatisfied with some aspects of the health care services. Mothers recommended that services be family focused, tailored to meet the needs of individuals and families, and better coordinated. Damrosch and Perry (1989) reported that mothers and fathers describe very different patterns of adjustment. Mothers experienced "peaks and valleys," whereas fathers described their adjustment as a steady and gradual return to a prediagnosis level of adjustment. Gibson (1986) found that parents who cope well still experienced stress-related illness. Mothers and fathers in her study reported differences in the way in which they perceived and dealt with stress. Mothers emphasized maintaining and promoting family integrity; they indicated that fathers' ability to support them was an important

coping strategy for them. Van Cleve (1989) found that parents who attended a support group reported higher coping scores, and parents who scored high on coping relied heavily on a few rather than a great variety of effective coping strategies.

Family coping was the major theme in eight studies that explored the relationship between healthy siblings and children affected with chronic illness such as cystic fibrosis and spina bifida. Seven researchers used qualitative research methods such as semi- or open-ended questions for face-to-face interviews, field notes, and in some cases a demographic questionnaire (Brissette and others 1988; Faux 1991; Feldstein and others 1996; Kruger and others 1980; Russell and others 1996; Spitzer 1992; Whyte 1992), or a Pictorial Stimuli Questionnaire to assess coping strategies (Spitzer 1992). Questions and discussions about sibling relationships centered around issues related to sibling care responsibilities, patterns of communication between sibling, concerns about the impaired sibling, situations that engender anger and rivalry (Faux 1991), family support, and the burden of care (Whyte 1992). One researcher used Lazarus's model of stress and coping to conceptualize the study (Spitzer 1992). Three authors used two nursing models: Roy's model of adaptation to illness (Russell and others 1996; Whyte 1992) and Orem's self-care framework (Kruger and others 1980); another researcher used a family-cycle model (Feldstein and others 1996) to conceptualize family processes and family functioning.

Lee and others (1997) designed a case-control study based on family systems theory to test the hypothesis that children with sickle cell disease score higher on depression and hopelessness and lower on perceived competence compared to their well siblings. Scores on hopelessness and perceived self-worth competence were no different for children who lived with sickle cell disease compared to well siblings. However, there were significant differences on the depression scores between these two groups (Lee and others 1997). Stress has its greatest impact on healthy siblings rather than affected children.

Both Lee and others (1997) and Whyte (1992) interpreted that illness creates a sense of chronic crisis in the family. A child's view of illness is based on an ability to appraise the situation accurately (Spitzer 1992). This appraisal process relies on knowledge of facts, imagination, fears, emotions, and cognitive evaluations. Spitzer (1992) discovered that imagination, fears, and emotions can undermine children's ability to accurately assess their sense of control over stressful events. Consequently, nurses must recognize that children's cognitive appraisal of a situation might be amplified by irrational fears that the child is unaware of or could be overly minimized if the child compared the current situation with a much worse case scenario that is unrealistic. Accurate cognitive appraisal contributes to a sibling's ability to communicate with the ill child about the illness and treatment regime.

Whyte (1992) found that the mother-father relationship has a great effect on how siblings are involved in the burden of providing care and how the whole family is able to adapt to a state of more or less chronic crises. Within a supportive environment, it is possible for the family's grief reaction toward the affected child to be a source of transformation for all members in a family by reframing the situation from a life of pain and suffering to one that has purpose, meaning (Kruger and others 1980), and an uplifting sense of heroism (Feldstein and others 1996). Feldstein and others 1996 found that "multigenerational patterns, myths, and belief systems contribute to the way a family" reaches out for support from professionals and nonprofessionals.

Discovering characteristics of the child who is affected with a chronic condition is a more recent phenomenon in nursing research. These researchers conducted studies between 1992 and 1996 that used a matched case control design. Monsen (1992) tested the hypothesis that a sense of autonomy and the ability to use problem-focused coping strategies facilitates

achievement of independence with self-care abilities. Folkman and Lazarus's (1988) theory of stress and coping and Orem's theory of self-care were used as the guiding framework. When comparing the scores of children who have spina bifida with healthy children, the researcher found no difference in these children's ability to use problem-focused coping or in their perception of self-care ability. This finding suggests that the presence of a handicap such as spina bifida may not necessarily interfere with a child's natural pattern of developing coping skills and self-care agency. The only variable that was different for the two groups was autonomy. This raises the possibility that autonomy may not be necessary for developing appropriate coping and self-care agency skills.

Mobley and others (1996) compared two groups of preschool children in terms of their self-perception and self-esteem. These researchers hypothesized that the families' ability to cope with and adapt to a child with spina bifida would be reflected in the child's self-perception. By comparing figure drawings produced by 21 children with spina bifida and 21 children who were physically normal, these researchers found that children with chronic illness drew fewer feet, legs, and trunks. One explanation given for this difference is that by ignoring dysfunctional body parts, children with spina bifida could maintain a positive body image. One important finding is that there was no difference in the children's perception of competence and acceptance by parents, professionals, and teachers; however, the affected children felt less happy compared to healthy children when talking to their friends. It is suggested that a negative perception of one's body image and a feeling of lack of acceptance by other children may be counterbalanced by feelings of maternal acceptance.

In a study by Williams (1992; Williams and others 1991), the researcher explored the possibility that cognitive ability might effect the capacity of children with Turner syndrome to achieve independence and appropriate problem-solving coping strategies. The findings indicate that there are no overall differences between children with Turner syndrome and children with nonverbal learning disabilities, in terms of their short-term memory and their ability to sustain attention and inhibit impulsively. The researcher suggested that children with Turner syndrome would benefit from the same kind of educational services offered to children with learning disabilities. Furthermore, school nurses can anticipate that children with Turner syndrome are at risk for alterations in nonverbal, verbal, and attention skills similar to children with nonverbal learning disabilities.

A Conceptual Model of a Holistic Approach to Family Adaptation

As depicted in this conceptual model of genetic nursing practice (see Figure 1), the phenomena of most concern to nurse researchers include cognitive appraisal, stress, uncertainty, parental coping, coping behaviors, coping patterns, social support, adaptation to chronic illness, social and psychological maladjustment, sibling rivalry, marital discord, chronic stress, chronic sorrow, negative affect, family functioning, transitions through stages of growth and development, and family involvement in the community.

Nursing research in this area began when Folkman and Lazarus's (1988) theory of stress and coping in illness was used to study families in which there is a child with a genetic condition such as cystic fibrosis, spina bifida, Turner syndrome, or Hemophilia. Nurse researchers discovered that parents of a child with a chronic genetic condition were faced with the same stresses as normal families. However, they had to cope with additional stresses such as the stress of the disease, the stress of uncertainty in reaction to the child's illness and potential death, stress on the couple relationship, and stress placed on the family as a whole. By combining qualitative research methods and two nursing models, the Roy Adaptation Model, and Orem's Self-care Model with family systems theory, a holistic approach to family coping and adaptation evolved. Expanding the focus of research to include sibling relationships between the normal and the affected child enabled researchers to better understand how healthy

siblings might respond to the affected child and how the healthy child might be adversely effected by burdens of care imposed on the family unit by the affected child and the parents. Most recently, nurses expanded this model again to investigate who the child affected by a genetic condition, really is, how the child transitions through stages of growth and development, and how the community responds to them and their needs.

Implications and Utility of This Knowledge for Nursing

This research contributes to practice-based theory by identifying and describing the main areas of concern. The task ahead is to question whether the psychological theories of stress, coping, and adaptation adequately explain the challenges that families face. Perhaps, it is time to give up borrowed theories and innovate new theories based on substantive evidence from nursing research. To extend their gaze even farther, nurse researchers must examine a much wider variety of genetic conditions that create chronic illness for children.

This research contributes a better understanding of the role of the nurse. Nurses in a wide variety of community practice settings can use this knowledge to provide education and support to families. They can use it to mobilize and coordinate community and professional services that are required for a child who makes ongoing transitions between hospital and home. These nurse researchers reiterate the significance and meaning of the nurse-patient relationship by discovering that long-term relationships are valuable. Families need consistent caring over time before they feel secure enough to disclose the inner workings of the family. Nurses need to listen and learn from each person how one person's experience affects the whole family. Future nurse researchers must emphasize the whole family in order to promote family-centered care within the framework of collaborative community services.

Reproductive Planning with New Technologies

Between 1982 and 1997, 11 (or 26%) of the genetic nursing research studies focused on this area. Some researchers investigated psychological and social factors that affect women's decisions to accept or decline prenatal genetic testing, whereas other researchers assessed the psychological and social consequences of genetic testing on pregnant women. Four subthemes emerged in this analysis: (1) factors that effect testing decisions, (2) lived experience of prenatal genetic services, (3) prenatal loss and its psychological and social effects after genetic testing, and (4) effects of reproductive technologies on maternal-fetal attachment.

Factors that effect testing decisions were of concern to nurse researchers who became involved in prenatal genetics research by asking the same question other disciplines were asking in the 1980s. At a time when researchers in other disciplines claimed that comprehension and recall of genetic knowledge was the most influential factor for making that decision, Davies and Doran (1981) found that 50% of the 66 advanced maternal age women (AMA) (older than 35 years of age) who accepted testing reported that moral reasons and a desire to have a healthy child resulted in conflicted and difficult reproductive decision making. Although 49% of respondents indicated a religious affiliation, only 15% indicated that this factor influenced their reproductive decisions. Using the Health Belief Model as a conceptual framework, these two researchers hypothesized that personal moral beliefs are different from and perhaps more influential than a women's religious affiliation in terms of prenatal genetic decision making.

Of a convenience sample of 100 parents, 25% guardians, stepparents and foster parents who cared for a child affected with cystic fibrosis, in a survey administered by Van Cleve (1993a), indicated that they would have chosen prenatal genetic screening and abortion if genetic testing had been available. Moral concerns and religious beliefs were cited as an important factor affecting this decision, in spite of the fact that only 25% indicated they attended religious services frequently. Twenty percent of respondents indicated they would use the

genetic information as an opportunity to plan future coping strategies in the event that they had to parent a child with cystic fibrosis. Only 10% of these parents said they would consult with a nurse regarding prenatal decision making.

Secondary analysis of census data, birth certificates, and retrospective surveys conducted in a single “health systems area” (Cox and others 1984a; 1984b) with 203 AMA women was conducted to assess factors that affect a woman’s decision to accept or to decline testing. Thirty variables were significantly associated with the decision to have or not have an amniocentesis. The following variables were most highly correlated to decision making: insurance coverage, prior experience with birth defects, perceived social supports, encouragement to seek a physician who is supportive of prenatal genetic testing, and a desire for decisional control over reproduction. The researchers suggested that willingness to insist on control over one’s own decisions may be the direct result of a higher socioeconomic status.

Williams and Schutte (1997) used a qualitative approach to assess psychosocial sequelae for women with a known ethnic or family history of genetic conditions after they accepted preconceptual genetic carrier testing. The women interpreted the genetic test as a way to settle uncertainty about their future reproductive options. Noncarriers of a genetic condition felt a great sense of relief knowing that they no longer had to delay childbearing. Carriers experienced intense feelings of finality and sadness because their worst fears had been confirmed.

Likewise, Winslow (1987) interviewed 12 well-educated, articulate AMA women who described what it was like to be pregnant for the first time at 35 years of age or older. Pregnancy was perceived as one more project that these women needed to manage and coordinate. They used critical and analytical thinking that “reflected a substantial feeling of control over their pregnancy project” (p 94). They sought genetic testing as part of the task of managing the project of pregnancy to reassure themselves that the fetus was normal. To manage safe passage through pregnancy, they sought the best possible prenatal care that in their minds included prenatal genetic testing.

Psychological and social effects after genetic testing following identification of a fetal malformation, and pregnancy loss were the focus of a qualitative study by VanPutte (1988). She interviewed 14 couples and 4 additional women who experienced prenatal loss following genetic diagnosis. VanPutte conceptualized the research findings as a psychological process involving five stages: (1) coping with fear of the unknown, (2) mobilization of coping strategies for decision making, (3) the need for continuity of support, (4) the need for respectful treatment, and (5) the need for follow-up counseling. VanPutte spelled out specific nursing skills that were effective for these women who endured a genetic termination and loss. “In this situation cure is impossible, and sensitive caring is essential” (p 20). Necessary nursing skills in this situation included support, anticipatory guidance, building rapport and trust, pain management and, mobilization of community resources and support systems.

To document the “lived experience” of deciding to undergo “planned birthing” of a malformed fetus, Matthews (1990) used a phenomenological method to interview 20 women who found themselves in this same situation. Of the 21 infants born, 19 of them died in the first few hours or days of life. Matthews described the findings as a psychosocial process she called the Expectancy of Loss Model, which consisted of six phases: uncertainty, verification of the malformation, preparation, reconfirmation, reparation, and resiliency. Despite feelings of loss, grief, sadness, and guilt, these women perceived that information provided during genetic counseling had a “positive impact on the experience” and they preferred “knowing” in advance because “it was the lesser of two evils” (P 173).

For 25 infertile and 13 “normal” couples all older than age 35 who had prenatal testing, qualitative research findings indicate that women experience “uncertainty and

hope” (Sandelowski and others 1991, p 167) and “iatrogenic anxiety” (Sandelowski and others 1991, p 176) as a result of genetic testing. The infertile couples compared the risk of miscarriage to “playing the odds” (p 175), as they had done while trying to become pregnant using medical technologies. Infertile women who decided not to have prenatal testing regretted their decisions later in the pregnancy because they had this haunting fear that they could have made the wrong decision (Sandelowski and others 1991).

Effects of reproductive technologies on maternal-fetal attachment was of interest to three researchers. Heidrich and Cranley (1989) conducted a time-series study to assess the effects of fetal movement, ultrasound scans, and amniocentesis on maternal-fetal attachment and perception of fetal development. They found that ultrasound scans had no effect on either fetal attachment or perception of fetal development (POFD). However, women who reported fetal movement early in pregnancy had higher attachment scores and higher POFD. This might explain why some women elect not to have genetic counseling or fetal testing: They may shy away from testing after they experience fetal movement and formulate an image in their minds of the developing fetus.

Using a case control design, Grace (1984) compared the responses of 67 AMA women with healthy pregnant women. She asked whether knowing the baby’s gender in advance or seeing the baby on the ultrasound screen influences mother-infant interaction postnatally. Grace cautioned nurses not to assume that a decision against testing was predictive of diminished mother-infant attachment in the postpartum period. Women who chose to know the baby’s gender prenatally did not score higher on the mother-infant interaction scale.

With a similar interest, Walker (1992) interviewed 88 pregnant women to document maternal reaction to knowledge of fetal gender in relation to Rubin’s (1984) maternal tasks of pregnancy. She pointed out that mothers may experience rejection of their fetus initially when it is not the desired sex. She claimed that nurses are responsible for promoting acceptance of the child despite initial disappointment experienced by mothers and perhaps, other family members.

A Model of Reproductive Planning with New Technologies

The phenomena of concern depicted in this model (see Figure 2) include maternal anxiety, maternal-fetal attachment, uncertainty, sex preferences, grief, guilt, family stress, support, crisis counseling, emotional care, stress and coping, personal control, decisional control, family relationships, moral concerns, beliefs, values, and hope.

Nurse researchers began prenatal genetic research by exploring the social and psychological factors that effect a woman’s decision to accept or decline prenatal genetic services that include alpha-fetoprotein screening in early pregnancy, ultrasound imaging, and genetic diagnosis for single gene or chromosomal disorders. With both quantitative and qualitative research studies, nurses better understand the characteristics of women who accept or decline genetic tests. Nurse researchers who became interested in the lived experience of prenatal loss following genetic testing further elucidated social and psychological sequelae of participating in prenatal technologies. These researchers again used Folkman and Lazarus’s (1988) theory of stress and coping, and family stress theory to investigate the effects of genetic testing in pregnancy. Janis and Mann’s (1977) theory of cognitive appraisal and rational choice, and the health belief model were used to explore effects of genetic testing and to explain the need for supportive counseling with emotional as well as physical caring.

Implications and Utility of This Knowledge for Nursing

The qualitative research studies produced conceptual models that describe lived experiences and psychosocial processes that emerged as a result of genetic testing and, in some cases,

prenatal loss. These models provide concrete suggestions that can be used to improve women's experiences in clinical practice. For example, one unique contribution made by nurses that is meaningful to women is their continuity and consistency of caregiving. Nurses can be assured that the intervention of fostering and maximizing a women's sense of personal control is therapeutic for women who must adjust to physical and psychological changes with pregnancy as well as cope with new reproductive technologies. Likewise, the often underrated nursing intervention of listening to patients tell their stories and validating their personal ways of knowing is critically important for women who are trying to make sense and find meaning in the experience of pregnancy and prenatal genetic testing (Anderson 1998b). With new knowledge about factors that inhibit women from undergoing testing, nurses can help reduce their fears, ease their moral uncertainty, and help them explore their personal values and beliefs. What seems to be missing from this research is recognition of cultural diversity. How this factor might alter women's experience of new reproductive services has not been pursued by nurse researchers.

Case Finding Studies

Five (or 12%) of the studies focused on locating all patients in a given region who are affected with or at risk to developing a genetic condition. The theoretical framework behind these studies is a preventative health model and a holistic model of nursing. The studies involved nurse researchers in the United States, Canada, the United Kingdom, and Australia.

The first of these kinds of studies was reported by an oncology clinical nurse specialist and a collaborative team of researchers involved in the Family High-Risk Program initiated at the University of Utah Health Science Center (Beck and others 1988). Nurses were recognized as a key link for identification of families at risk to cancer, for early detection of cancer, and for initiating behavioral changes that may very well substantially decrease cancer incidence. Within a "multidisciplinary team" (Beck and others, p 303), nurses along with administrators in regional high schools, local health departments, and primary care physicians were recruited to participate in a family cancer risk assessment, education, and evaluation program. High-risk families identified at the University Medical Center using a Health Family Tree Questionnaire were visited by community health nurses who provided "nursing interventions focused on primary and secondary prevention" (Beck and others, p 303). Nurses accepted as their responsibility the need to develop and participate in an innovative cancer prevention program that enabled them to work from a holistic framework in which health concerns are known to involve individuals, families, and communities.

Joachim and Hassall (1992) used a matched case control design to investigate 222 children diagnosed with inflammatory bowel disease in order to describe the prevalence of inflammatory bowel disease in their relatives and to try to determine whether there was a consistent pattern of familial transmission. The researchers conducted face-to-face interviews with affected children, written questionnaires were completed by the child's mother, and data were obtained from a review of the child's medical record. Three important findings were discovered. The relatives of these affected children were diagnosed with Crohn's disease and ulcerative colitis. In both affected children and in the case controls, inflammatory bowel disease showed up significantly more frequently on the mother's side of the family. A limitation of this finding is that mothers who completed the questionnaires may have underreported the occurrence of this disease in the paternal side of the family. The most frequent familial pattern of association between affected family members was sibling to sibling, followed by first cousins. This represented a horizontal transmission rather than a vertical relationship. This research generated a question that requires follow-up. Is it possible that there are two types of inflammatory bowel disease, one caused by genetic factors and the other by environmental factors?

The Folate and Neural Tube Defects Prevention Project (Bower and others 1996) included nurses as coinvestigators and as a target audience for education. The aims of the project were to inform health professionals and women of childbearing age about the need to increase folate intake to prevent neural tube defects. Pre- and postsurveys were conducted in these two target populations following an education campaign that included media promotion, supermarket, workplace, and community demonstrations to disseminate information. This research is described as the first population-based project in the world to promote increased folate intake to prevent neural tube defects.

Price and others (1996) conducted a similar study. They contacted 141 subjects identified in the Acadian Tumor Registry in the state of Louisiana to ascertain what percentage of them reported one or more first-degree relatives with pancreatic cancer. The findings from this purposive nonprobability sample were intended to serve as a pilot for a matched case control study. Data were collected using a survey questionnaire. The questions on the survey covered three broad topics: (1) demographic characteristics of persons affected with pancreatic disease, (2) the geographic location and frequencies of pancreatic cancer reported in first degree relatives, and (3) reported use of tobacco and alcohol, and dietary intake of vegetables, fruit, and pork. The researchers found that the median age for pancreatic cancer was 70 years. Tobacco was reported in 64.5% of people affected by pancreatic cancer; 55% of affected people drank alcohol; and the average age of death for affected persons who smoked was 66 years, and 78 years for those who did not smoke. These findings were useful to nurses who could help people receive an appropriate level of preventative health care services and to teach the community about health promotion and disease prevention.

The purpose of a similar study in the United Kingdom by Skirton and Glendinning (1997) was to locate all patients affected with Huntington's disease in Somerset county to assess the degree of support they received from health services and to identify deficiencies in the support that was available. They identified 31 people with Huntington's disease. "A questionnaire was designed with the help of experienced health authority and social service personnel" (p 83). The questionnaire dealt with 12 major categories of probable patient needs in a total of 300 questions. Information about patient and family demographics, the type of home setting, and the degree of disability was collected in face-to-face interviews or recorded in detailed field notes. Eighteen out of 31 people were living at home with Huntington's disease; of those, 61 % were being cared for by a spouse, 11% were cared for by the unaffected parent, 5% were cared for by a daughter, and 19% of caregivers were suffering from stress-related illness. Of affected persons, 92% thought they were receiving sufficient support and care from their family, friends, or other nonprofessional support system. However, 40% of the patients surveyed were not receiving regular contact from a member of the social service team, which is required by the National Health Services and Community Care Act of 1990. None of the patients in the study were receiving services from a physical therapist, and some families had received inaccurate information about transmission of the disease from genetic service providers. As a result of this study, the Huntington's Disease Advisory Group for Somerset devised an action plan with a view to establishing protocols that improve quality of care in this and other communities.

A Model of Genetic Nurses as Case Finders in Communities

The phenomena of concern to these nurse researchers include family health assessment, familial patterns of transmitting disease, early diagnosis to prevent disease, early intervention, and health promotion. These researchers documented a role for nurses as case finders who advocate for a model of health promotion in community settings (see Figure 3). The nurse is the frontline health care professional who seeks other disciplines when consultations are necessary.

All of these studies emphasize the importance of building and strengthening relationships among nurses, patients, families, teams of health care professionals, and community resources. As nurses become more knowledgeable about familial patterns of disease within a given community, the needs of patients and families become clearer. This enables nurses who know genetic epidemiology to create advanced practice roles related to genetic nursing. Other health care professionals are consulted or asked to serve as collaborators; the goal is to improve individual, family, and community health. As genetic epidemiology case finders, nurses are researchers, teachers, and activists in their communities. They try to improve health by shaping health care policy decisions and delivery of services.

Implications and Utility of This Knowledge for Nursing

This is an exciting new area of research that can strengthen nurses' position as change agents in communities and in the realm of health care policy. These kind of studies can be used to create new education programs and supportive services for patients and families in communities at risk to particular diseases because of their ethnic heritage, the environment, or genetic susceptibility. This research helps to clarify the role and scope of practice for nurses who have expertise in population genetics, epidemiology, and preventative health care. More research in this area is needed to describe and expand the role of the nurse in occupational and community health settings in which genetic conditions are ubiquitous in the population. Future studies should be undertaken that document family pedigrees and genetic and environmental risk factors. This data would generate many new research questions and new opportunities for collaborative inquiry. This kind of study necessitates a collaborative effort from research scientists in other disciplines. To keep track, over a long time, of enough data to evidence cause and effect associations between genetic or environmental risk factors and the outcomes of behavioral modification on human disease (or health) requires the coordinated effort of many collaborators. Nurses are essential to creating effective models for patient education, health surveillance, disease prevention, and health promotion related to genetics.

Ethical Inquiry

Nurse researchers directly focus on ethical issues related to genetic testing in only four studies (or 9% of the genetic nursing research literature). In these studies, the researchers describe ethical and moral factors that affect the decision to have or not to have genetic testing as well as ethical dilemmas or moral concerns that arise because of the availability of genetic testing. In describing how they live with the consequences of making a decision for or against testing, research subjects describe their personal values, beliefs, and moral standards.

In a qualitative study, Anderson (1998a, 1998b) interviewed 12 AMA women and their husbands separately to explore personal values and beliefs that affect their decision to accept or decline prenatal genetic testing. Seven couples who accepted testing and 5 couples who declined were interviewed. The single most important ethical finding was that couples who declined testing and couples who accepted testing did so for exactly the same reason: they believed that it was important to preserve the essence of humanness and carry this forward into the next generation of offspring.

Those who accepted testing perceived this essence of humanness as optimal health, wholeness, and well-being. These were necessary attributes because their children would need them to be able to contribute their best effort toward improving conditions for human beings all over the world. Those who declined testing perceived the essence of humanness as the dignity that inheres in every human spirit that enters into the hearts of other humans through love, compassion, suffering, enlightenment, and life itself. To this group of participants, the challenge was to see that gift even in children who have genetic conditions. Another important ethical finding in this study is that not all research subjects perceived that genetic services were

provided in a nondirective manner, even though they were quick to point out that they experienced no sense of coercion or arm twisting. However, they did describe five moral imperatives that challenge the claim that nondirectiveness is even possible (Anderson 1999). These moral imperatives are driven by a professional desire to know new information, to inform patients, to prevent morbidity and mortality in mothers and infants, to prevent pain and suffering, and to conserve public health care resources. These imperatives inhibit patients from really exploring and communicating their personal values and beliefs within the context of the therapeutic relationship. This finding questions whether nondirective counseling actually protects and fosters autonomy.

Using a qualitative approach, Van Cleve (1993b) investigated the experiences, attitudes, and concerns of seven neonatal intensive care nurses who provided direct patient care to anencephalic infants kept on life support systems as potential organ donors. Beachamp and Childress's analytic model of decision making was used to analyze interview data. Of the 110 nurses who worked on the unit at that time, 75 nurses gave care to these neonates. Surprisingly, not a single nurse refused to give care to one of the infants. The findings show some of their reasoning: 56% said they gave care without reservations, 44% said they had some reservations, 52% felt responsible to the infants, 52% thought that these donor babies provided a great good to society, 49% said they were motivated to contribute to a new frontier in medical care, 47% thought that the donorship issue did not influence their decision to participate in the program, 51% thought that nurses and doctors have different perspectives regarding ethical issues, and finally 65% of nurses questioned whether this program altered the dignity of the baby's life. The author concluded that despite the fact that some nurses struggled with their own values and beliefs, they sometimes settled for compromising positions imposed by physicians. Nurses must speak their views on these ethical situations because doing so will help shape ethical discourse concerning neonates that are born with a genetic condition incompatible with life.

In a case study in which a couple sought prenatal genetic testing due to a family history of hemophilia, Fibison and Davies (1983) inferred from this couple and from previous clinical experience that couples who undergo genetic testing expect to receive a definitive or certain medical diagnosis about the health of their fetus. The woman in this case study felt that she would rather face a dilemma than not have any information. The laboratory results following an amniocentesis procedure indicated that the fetus was a male. A fetoscopy procedure was performed to determine whether the male fetus was affected with hemophilia. However, the results were inconclusive because the sample was contaminated with maternal blood. At 19 weeks gestation, the couple faced a difficult decision, Should they elect to have an abortion when there was a 50% chance that the baby could be normal? She was asked how she would cope with the uncertainty that this baby could be a normal fetus. The researcher recognized that this ethical dilemma would create stress, and the couple would need help with their psychological adaptation to be able to cope with this and future pregnancies. The woman thought she was making the best decision she could at the time, and in a later discussion with the nurse, she said she did not regret having made that choice. She said, "I choose to believe he had hemophilia."

Tessaro and others (1997) conducted eight focus groups with women who had experienced familial breast cancer and healthy women who were eligible for testing because they were relatives. The goal was to determine women's responses to genetic services and BRAC-I testing for familial breast cancer. The women expressed a great deal of ambivalence about testing because of anticipated social, psychological, and ethical implications. On one hand, "Many women were already stressed because of having cancer due to their family history" (p 322), and they perceived that genetic information would decrease their sense of uncertainty, thus their stress. However, they were concerned about alack of proven treatment options if they did test positive. They were fearful that insurance companies would rescind their health or life

insurance if the information was not kept confidential. Those who said they would have testing done claimed that they would use that knowledge to make better health-related decisions. Another moral perspective remains unexplained but nonetheless interesting. Women described a sense of obligation to be tested in order to “fight breast cancer so that others [women in their families or in society] would not have to suffer as they had” (p 320). Finally, these women wanted their doctors to be more involved in the decision-making process; however, this was problematic to the researchers who suggested that physicians are not prepared to discuss complex genetic and ethical issues with patients.

A Model of Ethical Inquiry

In this genetic nursing practice model (see Figure 4), the availability of genetic technology creates options for testing. Simultaneously, ethical dilemmas and moral concerns happen as a result of contemplating genetic testing options. Ethical dilemmas and moral concerns affect the way people are moved to make sense of the situation and how they journey through the process of decision making. Once faced with the decision, even if they decide not to undergo genetic testing, they are transformed. They become sensitized to the multiplicity of ethical dilemmas and moral concerns that result from the advent of this technology and its use in society. Ultimately, everyone has to live with the consequences of their decision or indecision. These researchers illustrate the need for nurses to rethink familiar concepts: advocacy, authenticity, caring, believing, suffering, consent, autonomy, confidentiality, privacy, discrimination, decision making, and what it means to be human.

Implications and Utility of This Knowledge for Nursing

Genetic science and genetic technology have resulted in an ever burgeoning number of genetic screening tests, diagnostics, genetic therapies, and the new field of pharmacogenetics. These new genetic options pose moral concerns and ethical dilemmas never before witnessed by society. Nurses all over the world are beginning to face these ethical conundrums, as patients and families address genetic information and look for professional guidance while making sense and finding meaning in the decisions they must at least try to comprehend. This is an area of research that deserves much greater attention from nurse researchers. What is needed is moral philosophy for genetic nursing such as articulating a way of being with patients that reflects the ethos of nursing’s social mission in the context of genetics and cultural diversity.

Concluding Comments

Three themes recur in these studies; foremost is the notion that the family is the recipient of nursing care, and the welfare of the family is a nursing responsibility. The second is that nursing roles must be practiced in a holistic manner that fosters coordination of services provided by collaborative teams of health care professionals. Third, the holistic nature of the nurse patient relationship and how it is used as a practice modality is critical to patients who are continually required to interpret population statistics, genetic science, and new medical interventions. These three practice modalities—family practice, collaborative practice, and holistic practice—are common to genetic nursing in spite of differences in clinical settings, clinical populations, and cultures.

It is important to recognize that the four models of genetic nursing practice derived from this integrative review of the literature contain essential components for building genetic nursing knowledge. These concepts (or phenomena of concern) are recognized by all nurses in practice, research, education, and administration. The three practice modalities within these four models of genetic nursing practice link this research to the larger body of nursing theory, research, education, and practice.

References

- Abdellah F. The human genome initiative: Implications for nurse researchers. *J Profess Nurs* 1991;7(6):332.
- Anderson G. Creating moral space in prenatal genetics. *Qual Health Res: An Int and an Interdis J* 1998a;8(2):168–87.
- Anderson G. Storytelling: A holistic foundation for genetic nursing. *Holis Nurs Prac* 1998b;12(3):64–76.
- Anderson G. Nondirectiveness in prenatal genetic services: Patients read between the lines. *Nurs Eth: Int J Health Care Profess* 1999;6(2):126–36.
- Baroni M, Anderson Y, Mischler E. Cystic Fibrosis newborn screening: Impact of early intervention screening results on parenting stress. *Ped Nurs* 1997;23(2):143–51.
- Beck S, Breckenridge-Potterf S, Wallace S, Ware J, Asay E, Giles R. The family high-risk program: Targeted cancer prevention. *Onc Nurs Forum* 1988;15(3):301–6.
- Borgman, C., editor. *Scholarly communications and bibliometrics*. Thousand Oaks, CA: Sage; 1990.
- Bower C, Blum L, Watson C, Stanley F. Folate and the prevention of neural tube defects: Evaluation of a health promotion project in Western Australia. *Health Promo Int* 1996;11(3):177–87.
- Brissette S, Zinman R, Reidy M. Disclosure of psychological concerns of young adults with advanced cystic fibrosis (CF) by a nurse home visiting program. *Int J Nurs Studies* 1988;25(1):67–72.
- Cox C, Roughman K. Empirical test of the interaction model of client health behavior. *Research in Nurs and Health* 1984a;7:275–85.
- Cox C, Sullivan J, Roghmann K. A conceptual explanation of risk-reduction behavior and intervention development. *Nurs Res* 1984b;33(3):168–73. [PubMed: 6563535]
- Damrosch S, Perry L. Self-reported adjustment, chronic sorrow, and coping of parents of children with Down Syndrome. *Nurs Res* 1989;38(1):25–30. [PubMed: 2521384]
- Davies B, Doran T. Factors in a woman's decision to undergo genetic amniocentesis. *Nurs Res* 1981;31(1):56–9. [PubMed: 6214766]
- Faux S. Sibling relationships in families with congenitally impaired children. *J Ped Nurs* 1991;6(3):175–84.
- Feldman K. Using the work of others: Some observations on reviewing and integrating. *Soc Ed* 1971;44:86–102.
- Feldstein M, Baker J, Gemma P. Family stories of cystic fibrosis. *Nurs Leader Forum* 1996;2(1):14–23.
- Fibison W, Davies B. Dilemmas in prenatal diagnosis: A case study. *Iss in Health Care of Women* 1983;4:57–67.
- Folkman S, Lazarus R. The relationship between coping and emotion: Implications for theory and research. *Soc Sci in Med* 1988;26(3):309–17. [PubMed: 3279520]
- Ganong L. Integrative reviews in nursing research. *Res Nurs and Health* 1987;10(1):1–11. [PubMed: 3644366]
- Gibson C. How parents cope with a child with cystic fibrosis. *Nurs Papers* 1986;18(3):31–45.
- Grace J. Does a mother's knowledge of fetal gender affect attachment? *Mater Child Nurs* 1984;9:42–5.
- Heaman D. Perceived stressors and coping strategies of parents who have children with developmental disabilities: A comparison of mothers with fathers. *J Ped Nurs* 1995;10(5):311–9.
- Heidrich S, Cranley M. Effects of fetal movement, ultrasound scans and amniocentesis on maternal-fetal attachment. *Nurs Res* 1989;38(2):81–84. [PubMed: 2648336]
- Hymovich. *Parent perception inventory*. King of Prussia, PA: Author; 1988.
- Janis, I.; Mann, L. *Decision making: A psychological analysis of conflict, choice, and commitment*. New York: Free Press; 1977.
- Joachim G, Hassall E. Familial inflammatory bowel disease in a pediatric population. *J Adv Nurs* 1992;17(11):1310–5. [PubMed: 1430637]
- Kruger S, Shawver M, Jones L. Reactions of families to the child with cystic fibrosis. *Image: J Nurs Sch* 1980;12(3):67–72.

- Lazarus, R.; Launier, R. Stress-related transactions between persons and environment. In: Pervin, L.; Lewis, M., editors. *Perspectives in international psychology*. New York: Plenum; 1978. p. 287-325.
- Lee J, Phoenix D, Brown W, Jackson B. A comparison study of children with sickle cell disease and their non-diseased siblings on hopelessness, depression, and perceived competence. *J Adv Nurs* 1997;25(1):79–86. [PubMed: 9004014]
- Loebig M. Mother's assessments of the impact of children with spina bifida on the family. *Mater Child Nurs J* 1990;19(3):251–64.
- Matthews A. Known fetal malformations during pregnancy: A human experience of loss. *Birth Defects: Ori Art Ser* 1990;26(3):168–75.
- McCubbin, H.; McCubbin, M.; Cauble, E. CHIP: Coping Health Inventory for Parents (Form A). St. Paul, Minnesota: Family Social Science, University of Minnesota; 1979.
- McCubbin M. Nursing assessment of parental coping with cystic fibrosis. *West J Nurs Res* 1984;6(4):407–17. [PubMed: 6570387]
- Mobley C, Harless L, Miller K. Self-perception of preschool children with spina bifida. *J Ped Nurs* 1996;11(4):217–24.
- Monsen R. Autonomy, coping, and self-care agency in healthy adolescents and in adolescents with spina bifida. *J Ped Nurs* 1992;7(1):9–13.
- Price T, Payne R, Oberleitner M. Familial pancreatic cancer in South Louisiana. *Cancer Nurs* 1996;19(4):275–82. [PubMed: 8768685]
- Rodgers B, Ferholt J, Cooper C. A screening tool to detect psychological adjustment of children with cystic fibrosis. *Nurs Res* 1974;23(5):420–6. [PubMed: 4496752]
- Rubin, R. *Maternal identity and the maternal experience*. New York: Springer; 1984.
- Russell M, Reinbold J, Maltby H. Transferring to adult health care: Experiences of adolescents with Cystic Fibrosis. *J Ped Nurs* 1996;11(4):262–8.
- Samuelson J, Foltz J, Foxall M. Stress and coping in families of children with a myelomeningocele. *Arch Psychiat Nurs* 1992;VI(5):287–95.
- Sandelowski M, Harris B, Holditch-Davis D. Amniocentesis in the context of infertility. *Health Care for Women Int* 1991;12(2):167–78.
- Skirton H, Glendinning N. Using research to develop care for patients with Huntington's disease. *Brit J Nurs* 1997;6(2):83–90. [PubMed: 9116444]
- Spitzer A. Illness and treatment appraisal processes of healthy and hemophilic boys. *Res Nurs and Health* 1992;15(1):3–9. [PubMed: 1579649]
- Tessaro I, Borstelmann N, Regan K, Rimer B, Winer E. Genetic testing susceptibility to breast cancer: Findings from women's focus groups. *J Women's Health* 1997;6(3):317–27.
- Van Cleve L. Parental coping in response to their child's spina bifida. *J Ped Nurs* 1989;4(3):172–6.
- Van Cleve L. Alpha-fetoprotein testing: Opinions of parents of children with spina bifida. *Mater Child Nurs J* 1993a;21(1):20–5.
- Van Cleve L. Nurses' experience caring for anencephalic infants who are potential organ donors. *J Ped Nurs* 1993b;8(2):79–84.
- Van Os D, Clark C, Turner C, Herbst J. Life stress and cystic fibrosis. *West J Nurs Res* 1985;7(3):301–15. [PubMed: 3850699]
- VanPutte A. Perinatal bereavement crisis: Coping with negative outcomes from prenatal diagnosis. *J Perinatal Neonatal Nurs* 1988;2(2):12–22.
- Walker M. Maternal reactions to fetal sex. *Health Care for Women Int* 1992;13(3):293–302.
- Whyte D. A family nursing approach to the care of a child with a chronic illness. *J Adv Nurs* 1992;17(3):317–27. [PubMed: 1573100]
- Williams J. School-aged children with Turner's Syndrome. *J Ped Nurs* 1992;7(1):14–9.
- Williams J. Parenting a daughter with precocious puberty or Turner Syndrome. *J Ped Health Care* 1995;9(3):109–14.
- Williams J, Richman L, Yarbrough D. A comparison of memory and attention in Turner syndrome and learning disabilities. *J Ped Psychol* 1991;16(5):585–93.

- Williams J, Schutte D. Benefits and burdens of genetic carrier identification. *West J Nurs Res* 1997;19(1):71–81. [PubMed: 9030039]
- Winslow W. First pregnancy after 35: What is the experience? *Maternal and Child Nurs* 1987;12(2):92–6.

Figure 1.
Holistic approach to family adaptation to a child with a genetic condition and chronic illness.

Figure 2.
Reproductive planning with new technologies.

Figure 3.
Case finding in communities.

Figure 4.
Ethical inquiry.