Assessing Nurse Practitioner Practices Regarding Genetics and Genomics in Healthcare Services in the United States

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Abstract

Background. Nurse practitioners are well-positioned to identify patterns of genetic risk and refer individuals for genetic healthcare services. This study aimed to assess nurse practitioners’ knowledge, competency, attitudes, adoption, and confidence regarding genetics and genomics in their practice.

Methods. We employed a descriptive, cross-sectional survey design. Data was collected online using the Genetics and Genomics in Nursing Practice Survey.

Results. Of 106 participants, 84.1% (n=89) were working in a clinical setting. The knowledge score was 9.55±1.4 out of 12; 59% (n=62) of participants rated their knowledge of genetics as “good” or “excellent.” Only 8% (n=7) referred patients for genetics services, and 19.8% (n=21) were collecting full family history in practice.

Conclusion. Although the knowledge level of nurse practitioners was high, adoption of genetics was low in collecting and utilizing family history and referral to genetic services. There is a need for research investigating factors influencing the adoption of genetics activities into practice.

Keywords: nurse practitioner, genetics, genomics, practice

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Introduction

Recent developments in genetics and genomics have led to an increase in the use of advanced diagnosis and treatment strategies in clinical practice (Calzone, Kirk, et al., 2018; Manolio et al., 2019; Regan et al., 2019). Genetics involves the study of a specific and limited number of genes and their roles in inheritance; in contrast, genomics is a more recent term that describes the study of the entirety of a human’s genes (National Human Genome Research Institute [NHGRI], 2018). The advances in genetics and genomics have implications across the healthcare continuum and impact all healthcare providers regardless of their role or clinical specialty (Calzone et al., 2010; Calzone, Jenkins, et al., 2018; Calzone, Kirk, et al., 2018; Hu et al., 2018). Genetics and genomics have the potential to improve health care and outcomes by identifying individuals at increased risk of disease and, in some cases, through personalized screening, treatment, and management options (Halloran, 2015; King & Smith, 2020; Reed et al., 2019). The integration of genomic health care into practice requires a well-prepared workforce that can make genomic healthcare services more accessible for people who may benefit from the genetic and genomic discoveries (Calzone, Kirk, et al., 2018).

It is imperative to equip healthcare providers, including nurses, with the necessary knowledge and competency to integrate genomics into daily healthcare practice (Reed et al., 2019). The focus has been given to efforts, especially in the USA, to integrate genetics and genomics into undergraduate nursing curricula. In 2008, the Essentials of Genetic and Genomic Nursing: Competencies, Curricula Guidelines, and Outcome Indicators were published to define essential genetic and genomic competencies for all registered nurses (American Nurses Association [ANA], 2009). However, nursing has been facing challenges in the integration of genetics into nursing education, and there is substantial evidence showing a lack of confidence
and competence in genomics among registered nurses globally (Calzone, Kirk, et al., 2018; Donnelly et al., 2017; Read & Ward, 2016, 2018; Tonkin et al., 2020). Lack of educational guidelines, frameworks in genetic education (Calzone, Kirk, et al., 2018; Tonkin et al., 2020), and qualified faculty remain obstacles to equipping nurses at all practice levels with essential knowledge and competency in genetics and genomics (Read & Ward, 2016).

**Literature Review**

All nurses across different levels of education and scopes of practice must be equipped with genetics and genomics knowledge; however, nurse practitioners have strategic importance for integrating genetics into practice. A nurse practitioner is a registered nurse with specialized, advanced education and clinical practice competency to provide health care for diverse populations in various settings (American Association of Nurse Practitioners, 2019). Nurse practitioners function in the roles of clinician, scientist, educator, and patient advocate. The scope of nurse practitioner practice has expanded steadily over the last decades, and nurse practitioners have become vital members of healthcare services, caring for patients from preconception to death (Seibert & Darling, 2013). With increasing roles in every healthcare setting, nurse practitioners are especially well-positioned to provide person-centered and genetically informed healthcare (Hoffman et al., 2016; King & Smith, 2020). A well-prepared nurse practitioner workforce can lead the translation of genomic discoveries into daily health care practice (Seibert & Darling, 2013).

Standards of education and practice for nurse practitioners were published in the *Essential Genetic and Genomic Competencies for Nurses with Graduate Degrees* (Greco et al., 2012) and the *Genetics/Genomics Nursing Scope and Standards of Practice* (ANA, 2016). These
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guidelines aimed to identify essential genetic and genomic competencies for nurse practitioners to incorporate genetics and genomics into clinical and nonclinical nursing roles (ANA, 2016). However, there is little evidence on whether nurse practitioners are equipped to meet these standards. Studies measuring competency in genetics have primarily focused on registered nurses, undergraduate nursing students, and faculty (Calzone et al., 2013; Seven et al., 2017; Ward, 2017), as well as other non-nursing health care providers such as physicians (Ha et al., 2018; Shields et al., 2008). This is a critical gap in our understanding of the nurse practitioners’ level of knowledge and competency in genetics and genomics to integrate genomics into practice. By understanding the current knowledge and competencies of nurse practitioners regarding genetics-related healthcare services, we can inform the design of a formal curriculum and continuing education in this area. Therefore, the overall purpose of the study was to assess nurse practitioners’ knowledge, competency, attitudes, adoption, and confidence regarding genetics and genomics in their practice. The two specific aims of the study were (1) to evaluate the level of nurse practitioners’ knowledge, competency, attitudes, adoption, and confidence regarding genetics and genomics in their practice; and (2) to evaluate the factors affecting their knowledge level regarding genetics and genomics.

Methods

Design and Recruitment

This study used a descriptive, cross-sectional research design. Approval was obtained from the University of Massachusetts Amherst Institutional Review Board. Recruitment was carried out using two main strategies, which were mailing Listservs and social media. The recruitment goal was 150 nurse practitioners calculated with a 99% confidence level and 10%
margin of error with unknown population size and a 10% attrition using the Raosoft sample size calculator (http://www.raosoft.com/samplesize.html). An invitation to potential participants for the survey was sent to the University of Massachusetts Amherst postmaster’s doctor of nursing practice program and faculty email distribution lists, as well as the email list of the Massachusetts Coalition of Nurse Practitioners. Participants were encouraged to share the link with their colleagues.

**Sample**

The study population included board-certified nurse practitioners in the United States, primarily those practicing in Massachusetts. The inclusion criteria were: (1) board certification as a nurse practitioner in any of the primary specialties (family, adult-geriatric, pediatric, acute care, and women’s health) offered by either the American Nurses Credentialing Center or American Academy of Nurse Practitioners Certification Board; and (2) the ability to read English. There were 134 entries in the online survey; however, only 106 of these entries were complete. Therefore, 106 nurse practitioners were included in the study sample. Of the participants, 84.2% (n = 89) were actively engaged in patient care as part of their role (Table 1). This figure was determined by the question “Are you actively taking care of patients?”

**Instrument**

This study aimed to assess knowledge, competency, attitudes, adoption, and confidence of genetics and genomics in nursing practice among nurse practitioners; therefore, we used the revised version of the Genetics and Genomics in Nursing Practice Survey (GGNPS). For this study, the GGNPS was adapted for nursing practice from a survey targeted at family physicians.
using domains of the diffusion of innovation (DOI) theory by Calzone et al. (2013). The GGNPS is a 60-item survey that uses multiple response methods (including “select all that apply”), pick lists, multiple choice, yes/no, true/false, and Likert scales. It is divided into eight parts, which mainly correlate to the DOI theory domains of knowledge, persuasion, adoption, decision-making, implementation, and confirmation (Calzone et al., 2013; Calzone et al., 2016). In addition to sociodemographic characteristics, the domains include knowledge/competency, attitudes, decision/adoption, confidence, the social system of genetics, and genomics in nursing practice (Figure 1). The domains provide the framework for studying factors that potentially influence the integration of genetics into nursing education and practice. Items from the attitudes, receptivity, confidence, social system, and adoption domains are analyzed individually; they are not combined to calculate an overall score. A knowledge score is calculated by combining scores on responses to 12 items measuring genomic knowledge. Responses to each of the knowledge items are first marked as correct or incorrect. A total knowledge score is calculated as the number of correct answers out of 12, with a minimum possible score of 0 and a maximum possible score of 12. The calculation of the total knowledge score is restricted to individuals responding to all 12 items (Calzone et al., 2012).

Figure 1.

Measured factors affecting integration of genetic and genomics into practice.
The authors of the GGNP conducted reliability testing of the GGNP using data from registered nurses working in Magnet-designated hospitals. Weighted kappa, which calculates interrater reliability, showed a low to moderate test/retest reliability, with the poorest performing questions being knowledge-based, where participants may have been guessing (Calzone et al., 2016). Therefore, Calzone et al. (2016) revised the instrument and eliminated the weakest items to increase the reliability. Content and face validity of the GGNPS was established by Plavskin et al. (2019) via content expert feedback and assessment of a content validity index. This survey has been used in recent studies (Calzone et al., 2013, 2016) and translated into different languages such as Turkish (Yesilcinar et al., in press). In this study, we used the revised version of the GGNPS, which is publicly available through the National Human Genome Research Institute (2018).

**Data Collection**

Data collection occurred with an online survey between March and May 2020. The invitation email was sent to approximately 4036 Massachusetts Coalition of Nurse Practitioners members, 190 graduate students, and 38 faculty members of University of Massachusetts Amherst, Elaine Marieb College of Nursing. It is unknown how many potential participants read the invitation email or how many participants were recruited from each distribution list. The invitation email included the survey link using the Research Electronic Data Capture (REDCap) platform. The first page of the survey informed participants about the purpose of the study and its methods. Participants were informed that clicking “continue” at the bottom of the page was considered consent to participate. Approximately 7–14 days after the initial invitation, a
reminder email was sent to the distribution lists. This message included a thank-you for those
who had already completed the survey and a request to complete the survey for those who had
not yet participated. The survey took approximately 15–20 minutes to complete. No incentive
was provided for participation. The data was collected anonymously, stored securely with
REDCap, and accessed only by researchers to maintain confidentiality. Data was reported only
in aggregate form.

**Analysis**

Statistical analysis was performed using the Statistical Package for the Social Sciences
(SPSS), version 26.0. To address the first aim of the study—to evaluate the level of nurse
practitioners’ knowledge, competency, attitudes, adoption, and confidence regarding genetics
and genomics in their practice—descriptive statistics were used and reported as mean, standard
deviation, frequency, and percentage, as appropriate. For the second study aim—to evaluate the
factors affecting their knowledge level regarding genetics and genomics—bivariate analysis,
including the Mann-Whitney U test, t-test, Kruskal-Wallis test, and the one-way analysis of
variance (ANOVA) were used. The Mann-Whitney U test (for data that is not normally
distributed) and t-test (for normally distributed data) were used to compare differences between
two independent groups with a continuous dependent variable. Kruskal-Wallis test and one-way
ANOVA was used to compare differences between two or more independent groups. A statistical
significance was accepted as \( p < 0.05 \) for statistical analyses.
Results

Sociodemographics

Of the 106 participating nurse practitioners described in Table 1, the mean age was 49.7 ± 12.7 years (min. 26, max. 77); the mean years of practicing as a registered nurse or nurse practitioner was 23.3 ± 13.8 years (min. 2, max. 56). Of participants, 38.7% (n = 41) had a nursing curriculum that included genetics content, and 33% (n = 35) had attended any courses that included genetics since their licensure. Almost 83% (n = 87) of participants practice in Massachusetts, and 86.8% (n = 92) identified as white. Only 17.9% (n = 19) of participants had heard or read about the Essential Competencies and Curricula Guidelines for Nurses in Genetics and Genomics.

Table 1.
Demographic characteristics of nurse practitioners.

|                      | mean ± SD     |
|----------------------|---------------|
| Age                  | 49.5 ± 12.7   |
| Years in nursing     | 23.3 ± 13.8   |
| Time in patient care (%) | 74.4 ± 33.7   |

|                      | n  | %    |
|----------------------|----|------|
| Sex                  |    |      |
| Female               | 98 | 92.5 |
| Male                 | 5  | 4.7  |
| Declined/No answer   | 3  | 2.8  |
| Race/ethnicity       |    |      |
| White                | 92 | 86.8 |
| Black                | 7  | 6.6  |
| Asian                | 3  | 2.8  |
| Other                | 3  | 2.8  |
| Hispanic/Latino      | 1  | 0.9  |
| State                |    |      |
| MA                   | 87 | 82.2 |
| NH                   | 3  | 2.8  |
| NY                   | 2  | 1.9  |
| MD                   | 2  | 1.9  |
| Other                | 12 | 11.3 |
| Degree               |    |      |
| BSN                  | 2  | 1.9  |
| MSN                  | 80 | 75.5 |
Knowledge, Competency, and Attitudes

For the GGNPS knowledge questions, the mean total knowledge score was $9.55 \pm 1.4$ (min. 4, max. 12) out of 12 in this study. The question with the highest proportion of correct responses was “Do you think that genetic risk (e.g., as indicated by family history) has clinical relevance for ovarian cancer?” with 99.1% (n = 105) responding correctly. Table 2 shows the percentage of correct answers for the knowledge questions of the GGNPS.

Regarding the GGNPS competency questions, 61.3% of participants self-rated their understanding of the genetics of common diseases as “good” or “excellent”, and 59% self-rated their knowledge as “good” or “excellent.” Of nurse practitioners, 49.1% (n = 52) correctly answered that a single gene variant does not cause the most common diseases such as diabetes and heart disease. Regarding the GGNPS attitude questions, 96.2% (n = 102) of participants agreed that genetics could improve patients' treatment decisions; however, only 65.1% (n = 69) thought it would be important for nurses to be educated on the genetics of common diseases. Table 3 shows the domains of competency and attitudes towards genetics and genomics in nursing practice.
Table 2.

Correct responses to the knowledge questions of the GGNPS.

|                                                                                           | n  | %  |
|--------------------------------------------------------------------------------------------|----|----|
| A family history that includes only 1st degree relatives such as parents, siblings, and children should be taken on every new patient. | 40 | 37.7 |
| A family history that includes 2nd and 3rd-degree relatives such as grandparents, aunts, uncles, and cousins should be taken for every new patient. | 78 | 73.6 |
| Family history taking should be a key component of nursing care.                           | 102| 96.2|
| There is a role for nurses in counselling patients about genetic risks.                    | 96 | 90.6 |
| Do you think that genetic risk (e.g., as indicated by family history) has clinical relevance for breast cancer? | 105| 99.1 |
| Do you think that genetic risk (e.g., as indicated by family history) has clinical relevance for colon cancer? | 105| 99.1 |
| Do you think that genetic risk (e.g., as indicated by family history) has clinical relevance for coronary heart disease? | 105| 99.1 |
| Do you think that genetic risk (e.g., as indicated by family history) has clinical relevance for diabetes? | 103| 97.2 |
| Do you think that genetic risk (e.g., as indicated by family history) has clinical relevance for ovarian cancer? | 105| 99.1 |
| Extent to which family history supports clinical decisions (such as administering drugs prescribed). | 89 | 84 |
| The DNA of sequences of two randomly selected healthy individuals of the same sex are 90-95% identical. | 32 | 30.2 |
| Most common diseases such as diabetes and heart disease are caused by a single gene variant. | 52 | 49.1 |

**Adoption, Confidence, and Social System**

Decision/adoption is operationally described as self-reported collection and assessment of family history and self-reported facilitation of referrals to genetic service (Plavskin et al., 2019) in the GGNPS. Of the nurse practitioners who participated in the study, eight are not working in a clinical role; therefore, a total of 88 nurse practitioners who are primarily providing patient care
in clinical settings are included in the analysis of decision/adoption. While 46.6% (n = 41) of practicing nurse practitioners had been approached by a patient about genetics, only 8% (n = 7) had referred a patient to genetic services. In the past three months, the collection of complete family history was reported by 19.8% (n = 21), but 37.9% (n = 33) reported using family history to facilitate clinical decisions or recommendations. Of participants, 82.1% (n = 87) reported that they were confident discussing how family history affects recommended screening intervals, and only 41.5% (n = 44) were confident in providing information about the risks of genetic testing for common diseases.

Table 3.

The Genetics and Genomics in Nursing Practice Survey item responses: Competency and attitudes.

| Domain/question                                                                 | Answers | n   | %   |
|---------------------------------------------------------------------------------|---------|-----|-----|
| **Competency**                                                                  |         |     |     |
| When patients indicate a disorder in the family, which of the following pieces  |         |     |     |
|   of information do you collect in your standard family history assessment?     |         |     |     |
| Each family member’s:                                                          |         |     |     |
| • Age at diagnosis of a condition                                              |         | 60  | 56.6|
| • Relationship to the patient                                                  | Always  | 101 | 95.3|
| • Race or ethnic background                                                    |         | 41  | 38.7|
| • Age at death from a condition                                                |         | 67  | 63.2|
| • Both sides of the family (maternal/paternal)                                 |         | 94  | 88.7|
| Thinking about how you support clinical decisions, how important do you think  |         |     |     |
|   each of the following is to consider?                                        |         |     |     |
| • Genetic test result                                                          |         | 53  | 59.4|
| • Family history                                                               | Essential | 89 | 84  |
| Have you heard or read about the Essential Competencies and Curricula           |         | 19  | 17.9|
|   Guidelines for Nurses in Genetics and Genomics?                              | Yes     |     |     |
| Please rate your understanding of the genetics of common diseases.             | Good or | 65  | 61.3|
|                                                                                  | excellent |     |     |
In describing your genetic/genomic knowledge, would you consider it to be…

|                      | Good or excellent |
|----------------------|-------------------|
|                      | 62                |
|                      | 59                |

**Attitudes**

How important do you think it is for the nurse to become more educated about the genetics of common diseases?

|                      | Very important |
|----------------------|----------------|
|                      | 69             |
|                      | 65.1           |

Whether you think each of the following would be a potential advantage of integrating genetics of common diseases into your practice:

| Advantage                                | 102 | 96.2 |
|------------------------------------------|-----|------|
| Better treatment decisions (e.g., which drugs to prescribe) | 102 | 96.2 |
| Improved services to the patients        | 93  | 87.7 |
| Better adherence to clinical recommendations among patients | 84  | 79.2 |

Whether you think each of the following would be a potential disadvantage of integrating genetics of common diseases into your practice:

| Disadvantage                                      | 58  | 54.7 |
|---------------------------------------------------|-----|------|
| Would take too much time                          | 58  | 54.7 |
| Not reimbursable/too costly                       | 75  | 70.8 |
| Need to “retool” professionally                   | 40  | 37.7 |
| Increase patient anxiety about risk               | 52  | 49.1 |
| Would increase insurance discrimination           | 78  | 73.6 |

The GGNPS describes the social system as any clinical site where nurses are employed and that influences nursing practice. Only 35.8% (n = 38) of nurse practitioners reported that senior staff considers genetics to be part of the nurse practitioner role, and 31.1% (n = 33) agreed that senior staff considers genetics as a part of their own role. Table 4 shows the items related to the decision/adoption, confidence, and social system domain of the GGNPS.

**Table 4.**
The Genetics and Genomics in Nursing Practice Survey item responses: Adoption, confidence, and social system.

| Decision/adoption (n = 88)* | Answers | n   | %   |
|-----------------------------|---------|-----|-----|
| In the past three months, how often have you collected a complete family history from a patient that includes the | Always  | 21  | 19.8 |
following components: information on disorders from three generations, and age at diagnosis and death for each affected family member?

| Question | Yes | No | Percentage |
|----------|-----|----|------------|
| In the past three months, has any patient initiated a discussion with you about genetics? | 41 | 29 | 46.6 |
| In the past 3 months, how often have you used family history information when facilitating clinical decisions or recommendations for your patients? | Frequently | 33 | 37.9 |
| In the past 3 months, how often have you facilitated referrals to genetic services? | Frequently | 7 | 8 |

### Confidence

| Task | Yes | No | Percentage |
|------|-----|----|------------|
| Decide what family history information is needed to tell something about a patient’s genetic susceptibility to common diseases. | 84 | 20 | 79.2 |
| Discuss how family history affects recommended screening intervals. | Confident | 87 | 82.1 |
| Decide which patients would benefit from a referral for genetic counselling and possible testing for susceptibility to common diseases. | Confident | 60 | 56.6 |
| Access reliable and current information about genetics and common diseases. | Confident | 68 | 64.2 |
| Give patients information about the risks of genetic testing for common diseases. | Confident | 44 | 41.5 |
| Give patients information about the benefits of genetic testing for common diseases. | Confident | 56 | 52.8 |
| Give patients information about the limitations of genetic testing for common diseases. | Confident | 48 | 45.2 |
| Facilitate referrals for genetic services for common diseases. | Confident | 67 | 63.2 |

### Social system

| Question | Yes | No | Percentage |
|----------|-----|----|------------|
| Did your nursing curriculum include genetics content? | Yes | 41 | 38.7 |
| Since licensure, have you attended any courses that included genetics as a major component? | Yes | 35 | 33 |
| Do you intend to learn more about genetics? | Yes | 69 | 65.1 |
| Would you be able to attend a course during work hours? | Yes | 48 | 45.7 |
| Would you attend a course on your own time? | Yes | 76 | 73.1 |
| Do you think your senior staff members see genetics as an important part of your role? | Yes | 38 | 35.8 |
Do you think your senior staff members see genetics as an important part of their role?  

|   | Yes | 33 | 31.1 |

Note: *Decision/adoPTION only applies to nurse practitioners who see patients in clinical settings.

In this study, the actual knowledge score of nurse practitioners was significantly associated with the self-rated understanding and knowledge of genetics (p = 0.037, p = 0.045, respectively). Thinking that nurses have a role in genetics was also significantly associated with the total knowledge score (p < 0.05). Table 5 shows the comparisons between knowledge scores and some characteristics of nurse practitioners.
Table 5.

The comparison of knowledge score and characteristics of nurse practitioners.

| Variable                                                                 | n (%)  | Statistical value | p    |
|--------------------------------------------------------------------------|--------|-------------------|------|
| **n = 103**                                                              |        |                   |      |
| **Variable**                                                             |        |                   |      |
| Know/heard about the Essential Competencies and Curricula Guidelines for Nurses in Genetics and Genomics* |        |                   |      |
| Yes                                                                      | 18 (17.5) | 0.465             | 0.643|
| No                                                                       | 85 (82.5) |                   |      |
| **Degree****                                                             |        |                   |      |
| Baccalaureate                                                            | 2 (1.9)  | 0.079             | 0.961|
| Master                                                                   | 80 (77.7) |                   |      |
| Doctorate                                                                | 24 (23.3) |                   |      |
| **Years in Nursing***                                                    |        |                   |      |
| –                                                                        | –      | –0.053            | 0.590|
| **The nursing program included genetics***                              |        |                   |      |
| Yes                                                                      | 41 (39.8) | 1.935             | 0.057|
| No                                                                       | 65 (63.1) |                   |      |
| **Attended course***                                                     |        |                   |      |
| Yes                                                                      | 35 (34)  | 1.079             | 0.293|
| No                                                                       | 70 (68)   |                   |      |
| **Age*** (divided by the median)                                        |        |                   |      |
| < 49.7 years                                                             | 48 (46.6) | 0.895             | 0.373|
| > 49.7 years                                                             | 54 (52.4) |                   |      |
| **Self-rated understanding of the genetics of common diseases****        |        |                   |      |
| Excellent                                                                | 6 (5.8)   | 03.402            | 0.037*|
| Good                                                                     | 59 (57.3) |                   |      |
| Poor                                                                     | 41 (39.8) |                   |      |
| **Self-rated genetic/genomic knowledge ****                              |        |                   |      |
| Excellent                                                                | 7 (6.8)   | 3.204             | 0.045*|
| Good                                                                     | 55 (53.4) |                   |      |
| Poor                                                                     | 43 (41.7) |                   |      |
| **There is a role for nurses in genetics**                               |        |                   |      |
| Agree                                                                    | 98 (95.1) | 17.164            | 0.000*|
| Disagree                                                                 | 4 (3.9)    |                   |      |
| Don’t know                                                               | 6 (5.8)    |                   |      |

Notes: A statistical significance was accepted as p < 0.05.
*t-test
** Kruskal-Wallis test
*** Correlations
**** ANOV
Discussion

Knowledge, Competency, and Attitudes

Discrepancies were found between the knowledge scores of nurse practitioners in this study and the level of knowledge reported in the literature. In this study, the knowledge score of nurse practitioners in genetics and genomics was $9.55 \pm 1.4$ out of 12 questions answered correctly, or an average of 79.5%. This was considered a high knowledge score. While Hoffman et al. (2016) focused on pharmacogenetics practices, they found an average preintervention knowledge score of 3.4 out of 5 in a pilot sample of only five nurse practitioners. Furthermore, Edwards et al. (2011) showed that only 17% to 31.7% of participating nurse practitioners responded correctly to each knowledge question regarding hereditary colorectal cancer prior to their intervention.

Although the Whitt et al. (2016) study focused on nurse practitioner students, they found that less than half of the nurse practitioner students could define 53.8% of the genetic terms on the pre-course survey. These findings show lower knowledge scores, but this might be due to the small sample size, the focus on a specialized area of genetics, or the focus on nurse practitioner students rather than practicing nurse practitioners. The limited number of studies focusing on nurse practitioners and variation in results supports the need to assess genetic and genomic knowledge in broader samples of nurse practitioners in the United States. Assessing the current genetics and genomics knowledge of nurse practitioners is the first step to developing tailored educational strategies for nurses with advanced degrees, who often have more independent roles and responsibilities in health care services.

While this study focused exclusively on nurse practitioners, other studies in the literature mainly investigated the genetics and genomics knowledge among nursing students, nurses
practicing in different clinical areas, or nursing faculty (Calzone et al., 2013; Calzone, Kirk, et al., 2018; Maradiegue et al., 2005; Read & Ward, 2016, 2018; Ward & Barbosa-Leiker, 2018). Some of these studies (Read & Ward, 2016; Ward & Barbosa-Leiker, 2018) reported lower objective knowledge scores across nurses, nursing students, and faculty compared to our study; however, the authors of these studies used the Genomic Nursing Concept Inventory (GNCI), which tends to be more difficult than other knowledge measurements, including the GGNPS. Read and Ward (2016) found a mean knowledge score of 14.93 out of 31 among faculty and students, and Ward & Barbosa-Leiker (2018) found mean scores of 50% and 78% at their second and third time-points in their reliability study. A few studies have used the GGNPS with broader nursing samples, although the number of nurse practitioners in these studies was not specified (Calzone et al., 2013, 2016). The mean knowledge score among a diverse group of nurses in the Calzone et al. (2013) study, which used the GGNPS, was 8.99 out of 12; this is similar to our results, despite their sample’s more heterogeneous educational levels. More studies utilizing practice-based assessment for nurse practitioners will provide a comprehensive evaluation of the integration of genetics into practice.

In this study, the knowledge scores of the nurse practitioners were not associated with age, educational level, years in practice, having a genetics course in the curriculum, or having attended any other courses that covered genetics since the licensure. Contrary to our findings, Calzone et al. (2012) reported that knowledge scores among their respondents measured by GGNPS increased with education level. However, their population was much more heterogeneous in terms of education level, including nurses with training that ranged from diplomas to doctoral degrees. Furthermore, the limited studies on the effect of educational interventions during degree programs showed that these interventions were associated with
increased knowledge of genetics concepts among nurse practitioner students (Sloand et al., 2018; Whitt et al., 2016). Thus, our findings may be due to our participants’ more homogenous educational level and similar genetics and genomics content in master’s of science in nursing and doctorate in nursing practice curricula.

In this study, participant nurse practitioners’ self-rating of their understanding of genetics and their agreement that nurses have a role in genetics were associated with their knowledge score. This finding may show that nurse practitioners who have positive attitudes about the nursing role in genetics health care and a greater understanding of genetics are more likely to seek more information regarding genetics and genomics. Although Calzone et al. (2012) reported that nurses across various education levels (diploma, associate degree, master’s, doctorate) self-rated their knowledge and understanding as poor, their results reflect the anticipated difference between the different levels of education.

Despite a generally positive attitude towards genetics, this study found a high number of nurse practitioners were concerned about privacy and costs. Regarding attitudes toward genetics and genomics, 65.1% (n = 69) of nurse practitioners reported that it is very important for nurses to become more educated about the genetics of common diseases. A majority of them also agreed that genetics has advantages in better treatment decisions, improved services, and better adherence to clinical recommendations for patients. However, more than two-thirds of nurse practitioners reported that genetic services might create cost or reimbursement problems and increase insurance discrimination. Since only a small percentage (17.9%) of the nurse practitioners reported having read the Essential Competencies and Curricula Guidelines for Nurses in Genetics and Genomics, they may also be unaware of the US Genetic Information Nondiscrimination Act of 2008 (GINA). Although GINA does not protect the risk for
discrimination from life and disability insurances, this US law protects against health insurance
and job discrimination based on genetic testing or family history (US Department of Labor, n.d.).
The nurse practitioners in this study may also be unaware that most insurance plans in the United
States cover medically necessary genetic testing. This result shows the need for educational
initiatives, including topics such as cost, reimbursement, and protection from discrimination
related to genetic services.

**Adoption, Confidence, and Social System**

The percentage of nurse practitioners who stated, in this study, that they used family
history was higher than those who collected a full, three-generation family history. Among the
nurse practitioners working in clinical settings, 19.8% (n = 21) collected a complete family
history in the last three months, but 37.9% (n = 33) reported using family history to facilitate
clinical decisions or recommendations in their practice. This conflicting finding may indicate
that these participants may have been using family history in clinical decisions without
collecting complete family histories. Poor clinical competency in genetics and genomics can
lead to underutilization of genetic services, failure to refer for genetic counselling and testing,
mismanagement of genetic risk, and under- or overutilization of healthcare services (Calzone et
al., 2016; Ha et al., 2018; Halloran, 2015). Every nurse practitioner needs to know how to
collect complete three- generation family history (called a pedigree), recognize disease patterns
in a family, and facilitate the genetic counselling process (Reed et al., 2019). In this study, the
percent of nurse practitioners correctly indicating what information is necessary when collecting
a family history ranged from 56.6% (n = 60) for age at diagnosis of a condition to 95.3% for the
relationship to the patient. Creating a pedigree can help the clinician collect family history and
visualize the
disease patterns more clearly. Multiple tools have been developed to facilitate this process (Reed et al., 2019).

Self-rated knowledge was slightly higher in this study when compared to the literature. Only 59% (n = 62) of nurse practitioners in this study rated their knowledge as “good” or “excellent.” Similarly, Maradiegue et al. (2005) found low self-rated knowledge on multiple topics in genetics, including developing a pedigree. Only 34% of their respondents reported that they would be comfortable talking to a patient with a genetic condition. In another study, Laird and Raudonis (2020), 60.5% of nurse practitioners were confident in their knowledge of hereditary cancer syndromes, but only 40% took a three-generation family history. These findings show that nurse practitioners are not prepared to take a complete family history and recognize patterns that may indicate a hereditary predisposition to inform their clinical decision.

Poor adoption of genetics may be related to a lack of systemic support. Only 8% (n = 7) of the nurse practitioners in this study had facilitated a referral to a genetics service in the last three months, and slightly more than half of the nurse practitioners could identify which patients would benefit from such referral. This low use of referral may reflect a lack of support from administrators or collaborative physicians, as only 35.8% (n = 38) felt senior staff viewed genetics as an important part of the nurse practitioner role. Laird and Raudonis (2020) found that 37.7% of nurse practitioners would appropriately refer patients who meet criteria due to personal or family history of cancers to genetics, which was considered a low percentage. Additionally, Hoffman et al. (2016) reported that nurse practitioners rated the system support they received as 2.4 out of 5 on average, and self-reported their ability to implement pharmacogenomic testing as 3 out of 5 after participating in an educational intervention. It is possible that this lack of support may not only prevent nurse practitioners from seeking continuing education on genetics topics,
but also limit their exposure to the processes that can be used to facilitate appropriate referrals to genetic counselling. A study using dyads of administrators and nurse educators at Magnet-designated hospitals to develop genomic awareness strategies showed improvements in receptivity and adoption among nurses, in addition to an increased perception that genomics was considered important by administrative staff (Calzone, Jenkins, et al., 2018). This demonstrates how the social system, seen here with administrative support, can encourage nurse practitioners to integrate genetics into their practice and further their education on the topic.

A lack of confidence may also limit the ability of nurse practitioners to educate their patients on genetics and genomics. Less than half of nurse practitioners in this study were confident in their understanding of the risks (41.5%, n = 44) and the limitations (45.2%, n = 48) of genetic testing for common diseases. As part of the referral, nurse practitioners need to prepare patients and families for what to expect, communicate relevant information to the genetics team, and follow up with the patient after using genetic services (Reed et al., 2019). Reed et al. (2019) emphasized that facilitating the genetic counselling process is one way in which every nurse practitioner can integrate genetics into practice. As nurse practitioners are frequently the first point of contact for patients in primary care, it is essential to fill this gap in their clinical practice through educational interventions to improve knowledge and understanding of genetic testing (King & Smith, 2020; Laird & Raudonis, 2020).

Although a majority of nurse practitioners had not attended a nursing program that included genetics in the formal curriculum, there was a strong interest in pursuing further education on genetics and genomics. Only 38.7% (n = 41) of participants had a nursing curriculum that included genetics content. However, 33% (n = 35) had attended courses covering genetics and genomics post-licensure, and 65.1% (n = 69) of the nurse practitioners indicated
their intention to learn more about genetics. These findings show the need for genetic education for nurse practitioners. Several studies describe educational interventions to improve the knowledge, skills, and competency of nurse practitioners and nurse practitioner students in genetics and genomics (Blazer et al., 2011; Edwards et al., 2011; Hoffman et al., 2016; Sloand et al., 2018; Smania, 2016; Whitt et al., 2016). Research shows that these educational interventions can successfully increase knowledge and interest in furthering knowledge of genetics (Calzone, Jenkins, et al., 2018; St. Martin et al., 2017), and awareness of genetics, which would translate genetics to professional practice (Sloand et al., 2018). However, the outcomes of these studies are often measured in knowledge or confidence, with little emphasis on practice. Considering the findings of this study in terms of knowledge, competency, confidence, and social systems reported by nurse practitioners, tailored educational initiatives for nurse practitioners that, considering social system–related factors of concern, include current social and legal issues in genetics, are needed.

Conclusion

Even though the nurse practitioners in this study had a relatively high level of knowledge in genetics and genomics compared to results from other, similar studies, they still had low confidence and adoption of genetics into their practice. The areas of low confidence and adoption included low rates of taking complete family histories, using family history to facilitate clinical decisions, and referring patients to genetics services. This suggests that while nurse practitioners might have theoretical knowledge in genetics and genomics, there are factors that continue to prevent nurse practitioners from integrating genetics into clinical practice.
Our study findings suggest a need for integrating genetics and genomics into formal nurse practitioner programs and continuing nursing education, using the *Essential Competencies and Curricula Guidelines for Nurses in Genetics and Genomics* as a guideline (ANA, 2009). Moreover, continuing nursing education for nurse practitioners should be tailored to fill specific educational gaps, including key concepts in genetics and genomics; social and legal issues in genetics; family histories collection; pedigree drawing; and facilitation of referrals to genetic services. Priority in future research should be given to identifying factors beyond the knowledge of genetics and genomics that influence the adoption of genetics and genomics into the clinical practice of nurse practitioners.

**Strengths/Limitations**

The strength of our study is that it provides a baseline assessment of nurse practitioners’ practices regarding genetics- and genomics-related health care services in this study sample. This study is one of the few studies that focuses exclusively on the nurse practitioner population using a validated, comprehensive survey. However, the study does have some limitations. First, the convenience sample may cause selection bias, as nurse practitioners with a greater interest in genetics may have participated at higher rates. There may also be geographic differences in the practice patterns of nurse practitioners, as Massachusetts did not allow for independent nurse practitioner practice at the time of data collection. Further research is needed with a more geographically and clinically diverse sample of nurse practitioners in the United States. Additionally, the survey instrument used was not explicitly developed for nurse practitioners, and was thus adapted to fit this population. Therefore, there could be aspects of practice environment affecting adoption that were not examined in this study.
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