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Nursing Informatics and Epigenetics

An Interdisciplinary Approach to Patient-Focused Research

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PURPOSE

Nursing informatics (NI) is in a unique position to improve the future of patient outcomes through an interdisciplinary approach to research. With readily available epigenetic data and patient-centered clinical data, NI can be at the forefront of performing epigenetic research to enhance patient care and precision medicine and to provide a framework for incorporating epigenetic-related disease information into clinical decision support tools. This will not only enhance the clinical aspect of the EHR but also help move epigenetic research toward a patient-focused research model. Performing epigenetic research will allow NI to develop the necessary skills and knowledge to lead the effort to incorporate genetic information into patient records for future health outcome improvements.

KEY POINTS

- Nursing informatics as a specialty of nursing is in a unique position to conduct epigenetic research that can enhance precision medicine.
- Epigenetic research involves data-driven analysis of genetic changes that occur due to patient lifestyle and environmental factors.
- Nursing informatics should be at the forefront of epigenetics research to provide the patient data and framework for incorporating epigenetic research into patient care.

Nursing Informatics

The American Nurses Association (ANA) recognized NI as a sub-specialty in nursing in 1992 and updated the scope and standards of NI practice in 2014 to include more functional areas such as genomics and research.[1] These new functional areas are in addition to the roles traditionally held by NI specialists, which include supporting the EHR, barcode medication administration, education of nursing staff on computer use, communication, and collaboration.[2,3] Since its beginnings, NI has evolved and grown with the nursing profession to allow nurse clinicians the ability to collect and understand data generated during patient care.[4] Incorporated within the framework of NI is the understanding of transforming clinical data into information, which is then translated into knowledge and used to develop wisdom.[5] This continuum starts with the understanding that data drive the processes that evolve into understanding, whether in using technology or providing direct patient care. Historically, ANA defined NI as working within the domains of nursing science, information science, and computer science.[1] Building on this definition, a new conceptual model of NI is being introduced that includes the three domains from the ANA with the addition of a fourth domain, cognitive science (see Figure 1).[6,7]

A major component of NI is an understanding of nursing as a science, art, and profession, which is represented by the nursing science domain. This knowledge must include at least an understanding, if not expert knowledge, of nursing workflow, patient advocacy, care delivery, critical thinking, inter-professional collaboration, and understanding of overall nursing processes.[8,9] Through the collection and dissemination of nursing data, NI has the capacity to impact patient scheduling, resource planning, management of care, discharge planning, and communicating patient information; NI expertise also plays an instrumental role in patient safety, change management, and use of data systems.[10]

The information science domain involves the knowledge of information structure, processing, and technology. Nursing informatics is concerned with the legitimate access to and use of data, information, and knowledge, with a focus on the capture, management, and processing of health information.[11,12] In 1998, information science was introduced into the NI curriculum at a BSN program for teaching undergraduate nursing students NI components of information and technology,[13] demonstrating the importance of information science in relation to NI.

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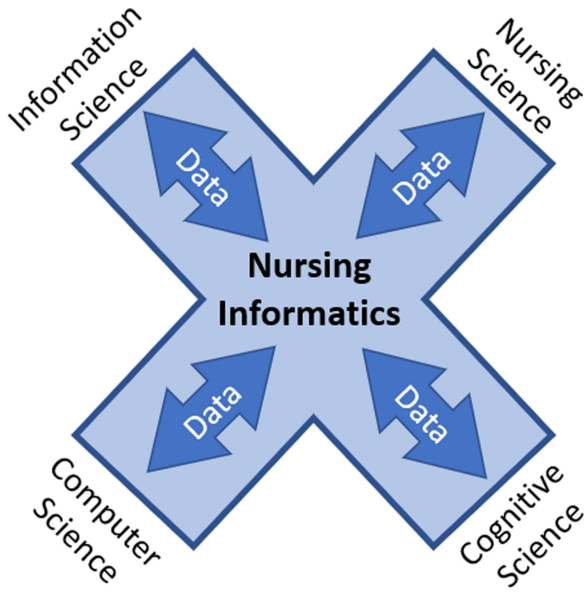

FIGURE 1. Conceptual model of NI.

The third domain in the model is computer science, where NI brings a nursing perspective to the implementation and integration of clinical applications and serves as a bridge between technology, data analysis, and patient care.[14] Clinical-technical fluency, both using and teaching computer fluency, is a key characteristic of NI. In fact, an early definition stated that NI is the application of computer technology to all fields of nursing.[15]

The fourth and final domain is cognitive science, where NI is expected to have knowledge of learning, teaching, and communication of data that reach across the healthcare continuum, even if technology is not available.[9,15,16] Cognitive science was initially introduced as a required domain for NI knowledge by Turley[6] in 1996 but not adopted in the ANA conceptual model. Stillings et al[17(p2)] note that cognitive science works to understand the mind through perception, thinking, and understanding, with many cognitive scientists using computers.

This model adds to the understanding that NI is a nursing specialty that requires continual application of data collection methods and analysis, making data the core component of an NI conceptual model. It represents the interaction between NI and non-nursing science domains using clinical patient data as the bridge and making collaboration an essential role of NI. This model provides the foundation for NI to participate in precision medicine research and predictive health models using big data and genetic information.

Epigenetics

Epigenetics is the study of changes that occur in DNA expression due to lifestyle, environment, and disease processes.[18] Though the genome (DNA sequence) of each cell is fairly consistent in each cell of the body (see Figure 2), the epigenome can change over time with exposure to environmental factors through a process called methylation (DNAm).[19,20]

Cytosine is one of four nucleotides that make up the building blocks of DNA, which looks like a twisted ladder or double helix (see Figure 2). The nucleotides (cytosine, guanine, adenine, and thymine) are nitrogen-based and bind together across the helix as the "rungs" of the ladder. Cytosine always pairs with guanine (CG base pair), and adenine always pairs with thymine (AT base pair). Of the four nucleotides, cytosine is the only one that can have an extra chemical attached through a covalent bond, which is called a methyl group. It is this methyl group attachment to cytosine that is called DNAm and is what regulates genes to turn them off and on.[21] When going up the strand of DNA, the nucleotides are joined to sugar molecules, which are joined by phosphate groups. It is this that makes each strand, and the two strands are held together by the base pairs.[22]

DNAm generally occurs when cytosine is followed immediately by a guanine nucleotide, referred to as a cytosine-phospo-guanine dinucleotide (CpG site), not to be confused with a CG base pair across the DNA strand.[23] Approximately 4%–6% of cytosines appear in CpG dinucleotides,

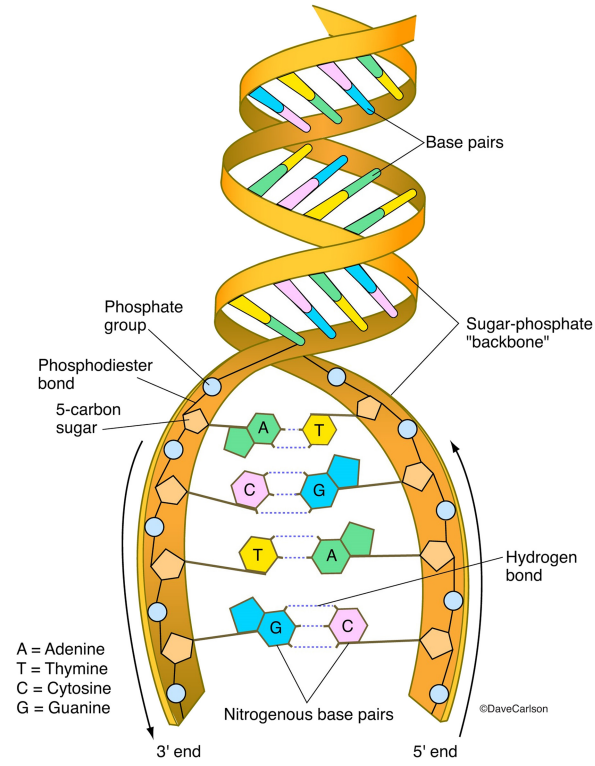

FIGURE 2. DNA structure including dinucleotide base pairs of adenine to thymine and cytosine to guanine. Copyright: Dave Carlson (www.CarlsonStockArt.com).

and approximately 60%-85% of these CpG sites are normally methylated, depending on the cell itself.[24] DNAm can occur due to environmental exposure to toxins, diet, and existing diseases.[25] Studies have linked DNAm to gene expression and disease processes, because DNAm can cause the gene to not be expressed (see Figure 3), and the removal of a methyl group can cause the gene to be expressed.[27–29]

DNAm can be measured and compared using discrete methylation differential data. If comparing two cells, one from a disease process such as cancer and the other a control, the methylation of the exact same CpG site can be measured to determine the methylation level, which is reported as being hyper-methylated or hypo-methylated in the diseased cell. Measuring the level of DNAm is a data-driven task, as there are approximately 3000000000 nucleotide pairs representing approximately 30000 to 40000 genes on 23 pairs of chromosomes in human DNA.[30] When considering the number of cytosine nucleotides that can potentially be methylated, the amount of data becomes exorbitant, with analysis and interpretation becoming daunting tasks.

Statistical analysis and programming packages that allow for analysis of methylation differential, in either CpG sites or clusters of methylated CpG sites called differentially methylated regions, are available and commonly used in epigenetics research. The work of using and developing these tools and programming packages is referred to as bioinformatics,[31] which according to the National Institutes of Health is a combination of biology and computer science that deals with the collection and analysis of biological data.[32] Whereas epigenetics can be performed strictly using bioinformatics tools and statistical analysis, when combined with nursing and clinical data, NI-driven epigenetics research can enhance patient-centered care, improve patient outcomes, and contribute to the advancement of precision medicine.[33,34]

CURRENT STATE

Although there are many potential benefits of NI-driven epigenetics research, little exists in the literature. Despite the ANA indicating that genomics (the study of genes) and research are now part of the NI scope of practice,[1] and epigenetics playing such a significant role in the overall health of the patient population, a literature search in PubMed revealed 396 articles in which "Nursing Informatics" and "bioinformatics" were combined, and an identical search in CINAHL and MEDLINE returned 146 articles. When "epigenetics" replaces "bioinformatics" in the search, the count reduces to 19 and 12, respectively. Though not a thorough and complete literature search, this rudimentary search shows that NI and epigenetics research is either not being conducted or not being published in nursing-related journals. Table 1 provides the search results and filters the count of articles by nursing-related journals. Only one article was found that combines NI and epigenetics with methodological considerations published in a nursing journal.[35] The article focuses on ethical considerations for DNAm study sample collection and analysis. Although research articles and continuing education are available from NI specialists,[36,37] they do not involve epigenetics, and the vast majority of NI publications deal with medication administration, care coordination issues, and data integration.[38]

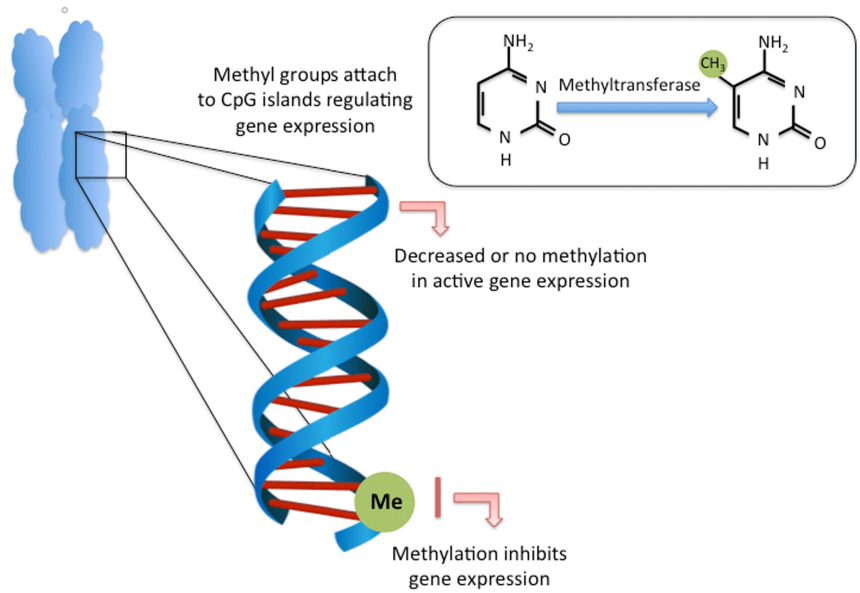

FIGURE 3. Methylation of cytosine from methyltransferase that can lead to inhibition of gene expression. From *Sperm DNA Methylation, Infertility and Transgenerational Epigenetics*.[26] Used by permission.

Table 1. Literature Search Results for Epigenetic and Bioinformatic Research Using NI and Keywords (Bioinformatics and Epigenetics) Found Anywhere in the Document

Database	NI and Bioinformatics		NI and Epigenetics	
	All Journals	Nursing Journals	All Journals	Nursing Journals
PubMed	396	55 (13.9%)	19	2 (10.5%)
CINAHL and MEDLINE	146	20 (13.7%)	12	1 (8.3%)

Percentage in parentheses represents nursing journal entries compared with all journal entries.

In recent years, there have been indications of advancements in nursing and epigenetic research. For example, a genomic knowledge matrix for nursing science was introduced in 2019 to guide genomic content education in nursing curricula.[39] The article indicates that an understanding of both epigenetics and bioinformatics is necessary for nursing to conduct genomic research but provides no indication that NI is necessary, despite mentioning that clinical patient data should be used. Additionally, older frameworks introduced for nursing to conduct genetic and epigenetic research also failed to indicate that NI should be an integral component of the research team.[40] In fact, in a 2013 priority list for NI research, integrating genomic data into the EHR was last in a list of 20 research topic areas; no other epigenetic research-related topics were included in the priority list, and a 2021 study listed genomics as 32 of a 36-item priority research list.[41,42] Among other priorities of NI research listed were clinical decision support (#2 on the 2021 list), Big Data (#3 on the list), and data science (#25 on the list), all of which are components of genomic and epigenetic research. Nursing informatics theory got a higher priority (#30 of 36) than genomics, indicating that some NI researchers are focusing on topics that do not put the patient at the center of data analysis or NI research. Finally, a 2020 study of NI literature between 1962 and 2018 concluded that NI has the ability to influence other disciplines.[38]

Bioinformatics and epigenetics naturally share a close relationship, given that bioinformatics tools are generally used to measure and quantify epigenetic changes to the genome,[18,24,31] and bioinformaticians look for patterns within DNA or proteins. These tools are generally computer programs that provide statistical analysis of patterns and trends in DNA sequences or methylation data. When combined with NI, clinical patient data can be used to enhance research for discovering physiologic-specific trends in disease processes. More work could be done to improve patient care outcomes and quality of care if epigenetics were given a higher priority in NI-related research projects.

DISCUSSION

Nursing informatics as a nursing specialty needs to be on the forefront of epigenetic research. With the ready availability of patient-centered data and genetic/epigenetic testing being conducted on a wider range of patient populations, the need for NI-based research to encourage the growth of data to knowledge is essential. This will provide the necessary framework for future integration of epigenetic markers and findings into a seamless clinical decision support tool in most, if not all, EHRs. This will allow for a more precise, patient-centered, readily available precision medicine mechanism that will benefit all healthcare specialties.

Given the nature of NI as being data focused and epigenetics research being data intensive, it seems only natural that the two should come together to provide patient-centered research opportunities. Cognitive science deals with learning, teaching, and communicating data; information science deals with information structure and processing; and bioinformatics has been defined as a blending of biology and computer science. Therefore, three of the four domains (cognitive science, computer science, and information science) in the NI conceptual model (Figure 1) could easily be applied to bioinformatics. Furthermore, NI adds the unique nursing science focus and can help all clinical specialties combine the statistical relevance of methylation studies with the clinical relevance of disease processes and patient outcomes. Nursing informatics is a natural lead for patient data-specific research, given the nature of NI knowledge of the clinical data generated during clinical care. Figure 4 is a conceptual representation showing where NI can augment the current relationship between epigenetics and bioinformatics to create an area of specialized precision medicine.

Nursing informatics is a complex, wide-ranging profession that, when present within an organization, can positively

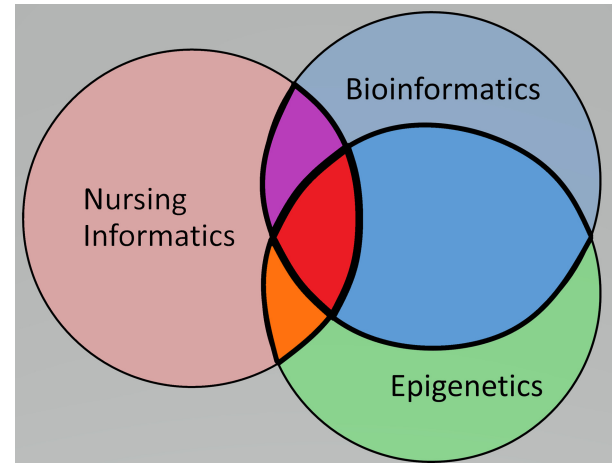

FIGURE 4. Conceptual model of overlap between NI, bioinformatics, and epigenetics.

impact the quality of patient care services. While technology and information-producing sources continue to grow and thrive within the healthcare industry and there remains a need for improving the overall quality of patient care, NI will remain a presence within the profession and practice of nursing. Epigenetics plays an important role in the overall health and well-being of the entire population and is a factor in the disease processes affecting the patients within the healthcare organization.

As informatics nurse specialists, we should consider each data source that contains patient information as a potentially important source to develop information, knowledge, and wisdom about the patients our organizations serve, not just the databases maintained by our organizations. By recognizing health or disease patterns in patients cared for across specialties and through learning and developing new methods for analyzing and presenting data, NI is uniquely positioned to research patient-centered care based on clinical findings in addition to disease states. Public data sources about our patient populations, such as cancer genome and specific disease registries, can be used for secondary data analysis. By being at the forefront of epigenetic research, NI can help improve patient outcomes, provide needed guidance for incorporating patient genomic data into EHRs, and work with other professions to enhance epigenetic research.

Online programs exist, some of which are free, that provide training on epigenetics understanding and research, and many of the tools used in epigenetic research are both free and maintained through government research grants and so are kept up to date and are trustworthy. The National Human Genome Research Institute as well as the National Institute of Nursing Research both sponsor free conferences for nursing to learn about genetic research, making epigenetic research from secondary data relatively inexpensive. Organizations will see the benefits and improved quality outcomes of the patient populations served through NI epigenetic research.

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Corrigendum to: Cocreation of Virtual Online Poverty Simulations in Google Slides: An Interprofessional Collaboration Between Students and Their Professors

Based on communication from the authors of a previously published study (Killam LA, McAuliffe JC, Hutton SA, Lefebvre ME, Campbell EE, Robinson RP, Ghartey K. Cocreation of virtual online poverty simulations in google slides: an interprofessional collaboration between students and their professors. CIN: Computers, Informatics, Nursing. 2022;40[1]: 1–6), the authors clarified that their affiliations should read as follows:

Author affiliations: School of Nursing, Queen's University (Mrs Killam), School of Health Sciences, Nursing, and Emergency Services (Mrs Killam, Mr McAuliffe, Mss Campbell, Robinson, and Ghartey); and School of Justice, Community Services and General Studies, Cambrian College (Ms Hutton and Mrs Lefebvre); Greater Sudbury, Canada.

Reference

Killam LA, McAuliffe JC, Hutton SA, Lefebvre ME, Campbell EE, Robinson RP, Ghartey K.. Cocreation of virtual online poverty simulations in google slides: an interprofessional collaboration between students and their professors. *CIN: Computers, Informatics, Nursing*. 2022;40(1): 1–6.

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