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The impact of genomics on health outcomes, quality, and safety

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The Human Genome Project reached its 25th anniversary on October 1, 2015. Since the project's launch, the implications of genomics science for healthcare and nursing practice have progressed steadily. In 2016, the new spending increase to the National Institutes of Health (NIH) includes $200 million targeted toward the Precision Medicine Initiative. This initiative is intended to accelerate the use of genetic variation in healthcare with specific emphasis on cancer therapeutics, including resistance, as well as establishing a 1 million person or more American research cohort that incorporates biospecimens, diet, lifestyle, and other health information, including links to the electronic health record (EHR) for those who consent.[1]

Why should nurse managers care about advances in genomics in healthcare? The impact of genomic information and technology has the potential to improve healthcare outcomes, quality, and safety, and result in cost savings. These outcomes are directly dependent on optimizing the use of information technology in the healthcare system, including the EHR.[2] Individual genetic makeup and variation inform the risk of disease, including in the prenatal, newborn, childhood, and adult contexts; can be used as a screening tool; more precisely characterize health conditions; improve medication selection, including therapies that may be designed to target the underlying disease genomics; and inform management of symptoms.

So important are these new advances that the American Nurses Association added the concept of genetics/genomics to the second edition of its Nursing Informatics: Scope and Standards of Practice. These standards state that informatics nurses must be able to "incorporate genetic and genomic technologies and informatics into practice" and "demonstrate in practice the importance of tailoring genetic and genomic information and services to clients based on their culture, religion, knowledge level, literacy, and preferred language."[3]

Integration into informatics

In 2013, a team of genetic specialty nurses and physicians identified the influences of genetics and genomics across the healthcare continuum: preconception/prenatal care,

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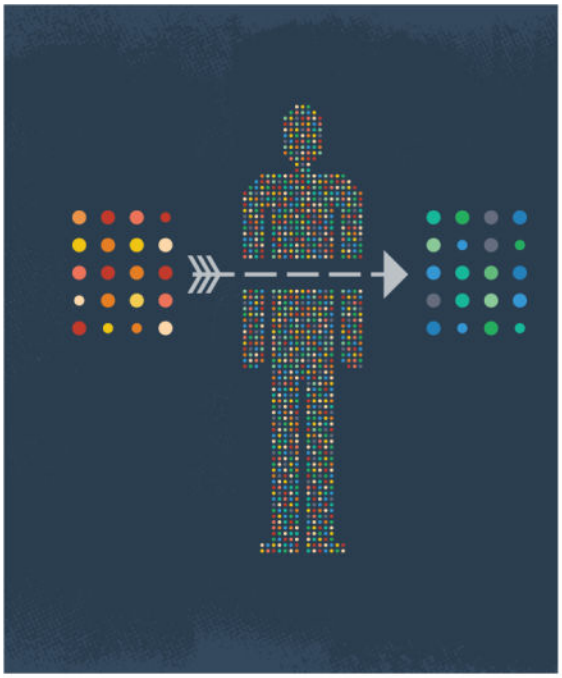
newborn screening, disease susceptibility, screening/diagnosis, prognosis and therapeutic decisions, and monitoring disease burden and recurrence.[4] (See Table 1.) A genetic analysis of a single patient can produce about 1 terabyte of data in a single encounter.[5] Therefore, when considering that genomics may be analyzed before or at the time of diagnosis and multiple times during treatment, as well as being integrated with lab data, clinical observations, tissue biopsy and other morphologic data, and imaging data, the volume of new data is so large that nurses will need to develop roadmaps for incorporation into their current practice and EHRs.



A roadmap for determining if genetic and genomic findings are clinically relevant is a project called ClinGen, an NIH-funded resource dedicated to building an authoritative central resource that defines the clinical relevance of genomic variants for use in precision medicine implementation. ClinGen is aimed at improving patient care through accelerating the understanding of genomic variation in healthcare through data sharing, knowledge curation, and technology development. Three questions are raised in considering whether a clinical variation is known: Is the gene associated with the disease? Is this variant causative? Is this information actionable? Working groups are establishing data models and standards for integrating these finding into EHRs.[6]

Work is also ongoing related to workflow and algorithm pathways for inclusion of genetic, genomic, and pharmacogenomic information into user-friendly clinical decision support (CDS) formats in the EHR. For example, St. Jude's Hospital has developed a model workflow with supportive CDS for pharmacogenomic tests into its EHR.[7] In addition, several national initiatives have been established to facilitate strategies to integrate genomics into practice, including Implementing Genomics in Practice, or IGNITE, and the Electronic Medical Records and Genomics, or eMERGE, Network.

Another relevant implementation project is Displaying and Integrating Genetic Information Through the EHR, or DIGITizE. At the Institute of Medicine (IOM) Roundtable on Translating Genomic-Based Research for Health, several vendors discussed a vision for implementation into the EHR. The IOM is launching pilot studies with vendors that concentrate on pharmacogenomic examples. The DIGITizE working groups are also developing an implementation guide, the Logical Observation Identifiers Names and Codes database, and an Allele Registry with ClinGen.[8]

Relevant to nursing informatics is the need to ensure that the family history section in the EHR elicits a minimum of three generations and the physical assessment section includes genetic and environmental information and risk factors.[9] Nursing informaticists need to identify current genetic and genomic information resources, such as the Pharmacogenomics Knowledge Base, or PharmGKB, website and the Clinical Pharmacogenetics Implementation Consortium guidelines, which should be included in EHRs. We also need to work on policies regarding access to genomic information stored within the EHR. Lastly, we need to understand the unique issues of privacy and security related to the use and potential misuse of genomic information.

Transitioning into nursing

Because genetics and genomics are becoming more relevant to the outcomes, quality, and safety of patient care, many nurse leaders are seeking to include genomic competencies in practice. Genetic/genomic competencies have been established for all RNs regardless of academic degree, clinical role, or specialty.[10] Genetic/genomic competencies are also available for other disciplines, including medicine, pharmacy, and physician assistants.[11–13]

Nurse leaders have studied how to diffuse genetic and genomic information into nursing practice to improve the quality of care and safety. The study enrolled Magnet® hospital champion dyads (administrator/educator pairs) and trained them to develop, implement, and evaluate a 1-year education intervention program. Twenty-one Magnet hospitals and two control environments participated in the study. The hospitals utilized several online learning resources to improve their knowledge gap.

In order to take the next step to integrate knowledge of genomics into their healthcare environment, the champions identified policies that needed to be changed, developed, or expanded. Several of the environments engaged in staff development activities, including booklets, pocket cards, identifying consultation resources, and encouraging staff to participate in workshops. Other settings engaged in genetic grand rounds. Communication included several media approaches, but a very popular strategy was the 1-page monthly series called GeneSplash with up-to-date information on genetic and genomic facts for particular diseases.[14,15] Currently, this group of study participants and researchers has partnered to build an online toolkit of all effective strategies used by both educators and administrators with an anticipated launch later in 2016.

Most of the Magnet hospital environments identified obstacles and challenges to moving forward. The study demonstrated awareness of innovation, but no integration of information

into the EHR. One of the most influential champions was the CNO, who was identified as the most important leader requiring genetic and genomic competency sufficient enough to plan for future integration into practice and the financial costs identified as a barrier.

The resources in Table 2 supplement the multiple resources provided by the Genetics/Genomics Competency Center. They aren't inclusive, but are meant to facilitate continuing learning. Integration into practice does require a basic understanding of genetics, genomics, and pharmacogenomics. (See Table 3.) The message isn't to acquire a PhD in genetics/genomics or bioinformatics, but rather develop a sufficient underpinning in genetics/genomics based on your role to establish a roadmap within your environment to incorporate this technology and knowledge into clinical practice. The impact of these discoveries on healthcare decision making potentially affects a large proportion of your patients.

Putting it into practice

With the $1,000 genome in reach—the capacity to sequence a genome for $1,000, a cost comparable to other medical tests—the clinical utility of genetic/genomic information will continue to expand.[4,16] This information means the right diagnosis and the right treatment at the right dosage in contrast to the historic approach that a given treatment works for most people with minimal toxicity. The greatest barrier to practice translation remains healthcare providers and the infrastructure within which they practice.[17] This is a call to nurse leaders that health outcomes, quality, safety, and, ultimately, cost containment depend on maximizing genomics knowledge and accelerating its translation into practice.

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Table 1

Genomics and the healthcare continuum[4]

Healthcare continuum	Genomic application example	Clinical application example
Preconception/prenatal	Germline genetic testing for recessive conditions	Preconception testing for carrier status in prospective parents for genetic variants associated with recessive conditions, such as sickle cell disease, cystic fibrosis, and Tay Sachs disease[18]
	Cell-free fetal DNA from maternal plasma	Less invasive strategy compared with amniocentesis for assessing fetal genomic variations that have health implications such as fetal aneuploidy[19]
Newborn screening	State-mandated newborn screening; not all recommended screening tests are genetic tests, but they screen for indications of the need for further genetic evaluation	Approximately 4 million newborns screened annually using dried blood spot cards for conditions such as immunodeficiency disorder and congenital heart disease[20]
Disease susceptibility	Germline genetic testing	Inherited cancer syndromes, such as hereditary breast ovarian syndrome, associated with mutations in BRCA1 and BRCA2[21]
		Familial hypercholesterolemia associated with mutations in LDLR, APOB, and PCSK9[22]
Screening and diagnosis	Stool DNA testing	FDA-approved test that can be used for screening purposes instead of colonoscopy[23]
Prognosis and therapeutic decisions	Targeted therapies	Therapy based on tumor genomic variation, such as epidermal growth factor receptor somatic mutation in non–small cell lung cancer and tyrosine kinase inhibitors[24]
	Tumor profiling	Basket trials, such as the Molecular Analysis for Therapy Choice, or NCI-MATCH, designed to identify somatic mutations/amplifications/translocations in patient tumor samples and assign patients to agents/regimens based on tumor genomics and not histology[25]
	Pharmacogenomics	Individuals who carry HLA-B*57:01 have an increased risk of hypersensitivity to the antiretroviral drug abacavir[26]
Monitoring disease burden and recurrence	Pharmacogenomics	Symptom management such as pain control; CYP2D6 to determine whether an individual can convert codeine into the active metabolite morphine[26]

Table 2

Selected online resources

American Nurses Credentialing Center	http://nursecredentialing.org/AdvancedGenetics or http://nursecredentialing.org/InformaticsNursing
ANA Essential Genetic and Genomic Competencies for Nurses with Graduate Degrees	www.nursingworld.org/MainMenuCategories/EthicsStandards/Genetics-1/Essential-Genetic-and-Genomic-Competencies-for-Nurses-With-Graduate-Degrees.pdf
ANA Essentials of Genetic and Genomic Nursing: Competencies, Curricula Guidelines, and Outcome Indicators, 2nd edition	www.aacn.nche.edu/education-resources/Genetics__Genomics_Nursing_Competencies_09-22-06.pdf
CDC	www.cdc.gov/genomics
Clinical Pharmacogenomics Implementation Consortium	https://www.pharmgkb.org/page/cpic
Electronic Medical Records and Genomics (eMERGE) Network	www.genome.gov/27540473
Genetics/Genomics Competency Center	www.g-2-c-2.org
GenomeWeb	www.genomeweb.com
Implementing Genomics in Practice (IGNITE)	www.genome.gov/27554264
PharmGKB	https://www.pharmgkb.org

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Table 3

Basic definitions

Genetics: The study of individual genes and their impact on relatively rare single-gene disorders
Genomics: The study of all of the genes in the human genome together, including their interactions with each other, the environment, and other psychosocial and cultural factors
Pharmacogenomics: The study of the influences of genetic variation on medication and adverse events