

Personalized Medicine, Genomics, and Pharmacogenomics: A Primer for Nurses

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Personalized medicine is the study of patients' unique environmental influences as well as the totality of their genetic code—their genome—to tailor personalized risk assessments, diagnoses, prognoses, and treatments. The study of how patients' genomes affect responses to medications, or pharmacogenomics, is a related field. Personalized medicine and genomics are particularly relevant in oncology because of the genetic basis of cancer. Nurses need to understand related issues such as the role of genetic and genomic counseling, the ethical and legal questions surrounding genomics, and the growing direct-to-consumer genomics industry. As genomics research is incorporated into health care, nurses need to understand the technology to provide advocacy and education for patients and their families.

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Genetics and genomics are rapidly changing health care by making it more personalized than ever before. Medical genetics is defined as the study of how individual genes can be identified and used for medical applications, including as markers for targeted drug therapies, to identify disease or the predilection for disease, and for tailoring treatments (Offit, 2011). In 2003, the first human genome was completely mapped, cataloging all of a person's genes (National Institute of General Medical Sciences, 2013). In the years since the advent of medical genetics, researchers have found that many factors other than the presence or absence of single genes predict disease; medical genomics is a study of all genes and other factors (e.g., epigenetic influences, environmental influences) that contribute to inheritance patterns and how medical care can be tailored to individual genomes. Pharmacogenomics is the study of how a person's genome affects his or her reaction to medicines (National Institute of General Medical Sciences, 2013). Personalized medicine employs applications from genetics, genomics, and pharmacogenomics, as well as the analysis of environmental factors, to individualize health care to an individual's specific needs. Personalized medicine represents a major change in the way health care will be delivered, and

nurses need to stay informed about its science, clinical use, ethics, economics, and social impact. Because cancer is fundamentally a genetic disease, advances in genetics and genomics have profound implications for oncology, and patients will be looking to their healthcare providers—including nurses—for guidance (Riley et al., 2012).

Background

To understand the nursing implications of genomics, one must first understand the underlying science. Although the idea of inheritance has been studied since ancient times, the era of modern genetics began in 1865 when Mendel explained the concept of discrete dominant and recessive genes and described how traits can be passed down from generation to generation, sometimes skipping a generation (Lorentz, Wieben, Tefferi, Whiteman, & Dewald, 2002). Not long after, genetic material (DNA) and chromosomes were discovered. In 1953, Watson and Crick described the double-helix structure of DNA and later showed that DNA codes for ribonucleic acid (RNA), which then codes for the production of proteins in the body (Lorentz et al., 2002). With the exception of sperm and egg cells, each human cell contains 23