http://www.aacn.nche.edu/education/pdf/BaccEssentials08.pdf) identified genetics and genomics as strong forces influencing the role of nurses in patient care. This brief overview identifies key terms and concepts and highlights essential genetics and genomics competencies for all nurses.

Genes, Genetics, and Genomics

Genes are segments of deoxyribonucleic acid (DNA) that contain genetic information necessary to control a certain physiologic function or characteristic. These segments are often referred to as *sites* or *loci,* indicating a physical or "geographic" location on a chromosome. Variant forms of a gene commonly occur within a population. When referring to a particular form of a gene, the term *allele* is used. Variant forms of a gene (variant alleles) may lead to no measureable or observable differences, may cause the person to be susceptible to clinically recognizable pathology within specific environmental contexts, may cause a clinically recognized disease or disorder, or may prove advantageous within a particular environmental context.

In earlier times, human diseases were thought to be either clearly genetic or typically environmental. However, the observation that some genetic disorders are congenital (present at birth) but others are expressed later in life has led scientists to conclude that many, if not most, diseases are caused by a genetic predisposition that can be activated by an environmental trigger. Examples of such interactions are found in single-gene disorders, such as phenylketonuria (PKU) and sickle cell disease, and **multifactorial conditions**, such as cancer and neural tube defects (NTDs). PKU is a disorder resulting from the (genetically determined) absence of an enzyme that metabolizes the amino acid phenylalanine. However, the deleterious effects in the infant are expressed only after sufficient ingestion of phenylalanine-containing substances, such as milk (environmental trigger). Even in the case of a "classic" genetic condition, such as sickle cell disease, its acute symptoms are precipitated by certain conditions, such as lowered oxygen tension, infection, or dehydration.

Congenital Anomalies