small to reliably visualize under a light microscope but are still clinically relevant. Fragile, or weak, sites associated with expanded triplet repeats have been identified on both the autosomes and the X chromosome. A classic example is fragile X syndrome.
Contiguous gene syndromes are disorders characterized by a microdeletion or microduplication of smaller chromosome segments, which may require special analysis techniques or molecular testing to detect (Bar-Shira, Rosner, Rosner, et al, 2006).

Chromosome anomalies typically affect large numbers of genes; however, a **single-gene disorder** is caused by an abnormality within a gene or in a gene's regulatory region. Single-gene disorders can affect all body systems and may have mild to severe expressions. Single-gene disorders display a Mendelian pattern of dominant or recessive inheritance that was first delineated in the mid-nineteenth century by Gregor Mendel's experiments with plants.

Mendelian inheritance laws allow for risk prediction in single-gene disorders; however, phenotypic expression may be altered by incomplete penetrance or variable expressivity of the responsible allele. An allele is said to have **reduced** or **incomplete penetrance** in a population when a proportion of persons who possess that allele do not express the phenotype. An allele is said to have **variable expressivity** when individuals possessing that allele display the features of the syndrome in various degrees, from mild to severe. If a person expresses even the mildest possible phenotype, the allele is penetrant in that individual.

Role of Nurses in Genetics

All nurses need to be prepared to use genetic and genomic information and technology when providing care. The professional practice domains of the essential genetic and genomic competencies include applying and integrating genetic knowledge into nursing assessment; identifying and referring clients who may benefit from genetic information or services; identifying genetics resources and services to meet clients' needs; and providing care and support before, during, and after providing genetic information and services (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009). Often a nurse is the first one to recognize the need for genetic evaluation by identifying an inherited disorder in a family history