

# McCance: Pathophysiology, 6th Edition

## Chapter 05: Genes, Environment, and Common Diseases

### Key Points – Print

#### SUMMARY REVIEW

##### Factors Influencing Incidence of Disease in Populations

1. The incidence rate is the number of new cases of a disease reported during a specific period (typically 1 year) divided by the number of individuals in the population.
2. The prevalence rate is the proportion of the population affected by a disease at a specific point in time. This rate, and the incidence rate, can be used to compare population variation in disease frequency.
3. Relative risk is a common measure of the effect of a specific risk factor. It is expressed as a ratio of the incidence rate of the disease among individuals exposed to a risk factor divided by the incidence of the disease among individuals *not* exposed to a risk factor.
4. Many factors can influence the risk of acquiring a common disease, such as cancer, diabetes, or hypertension. The factors can include age, gender, diet, exercise, and family history of the disease.

##### Principles of Multifactorial Inheritance

1. Traits in which variation is thought to be caused by the combined effects of multiple genes are polygenic.
2. The term *multifactorial* is used when environmental-lifestyle factors also are believed to cause variation in the trait.
3. Many quantitative traits (e.g., blood pressure) are multifactorial.
4. Because traits are caused by the additive effects of many genetic and environmental-lifestyle factors, they tend to follow a normal or bell-shaped distribution in populations.
5. Those diseases, however, that do not follow a bell-shaped distribution appear to be either present or absent in individuals. They do not follow the inheritance patterns of single-gene disease. Instead, such diseases may follow an underlying liability distribution. It is thought that a threshold of liability must be crossed before the disease is expressed.
6. Examples of diseases that correspond to the liability model include pyloric stenosis, neural tube defects, CL/P, and some forms of congenital heart disease.
7. Many of the common adult diseases, such as hypertension, coronary heart disease, stroke, diabetes mellitus (types 1 and 2), and some cancers, are caused by complex genetic and environmental-lifestyle factors and are thus multifactorial diseases.
8. For most multifactorial diseases, empirical risks, risks based on direct observation of data, have been derived.

9. In contrast to most single-gene diseases, recurrence risks for multifactorial diseases can change significantly from one population to another because gene frequencies, as well as environmental-lifestyle factors, can differ among populations.
10. Several criteria are used to define multifactorial inheritance: (a) the recurrence risk becomes higher if more than one family member is affected; (b) if the expression of the disease in a proband is more severe, the recurrence risk is higher; (c) the recurrence risk is higher if the proband is of the less commonly affected sex; (d) the recurrence risk for the disease usually decreases rapidly in more remotely related relatives; and (e) if the prevalence of the disease in a population is  $f$ , the risk for offspring and siblings of probands is approximately  $\sqrt{f}$ .

#### Nature and Nurture: Disentangling the Effects of Genes and Environment or Lifestyle

1. Family members share genes and a common environment; therefore, resemblance in traits, such as high blood pressure, reflects both genetic and environmental-lifestyle commonality (nature and nurture, respectively).
2. Few traits are influenced *only* by genes or *only* by environment-lifestyle. Most are influenced by both.
3. When a disease has a relatively larger genetic component, as in breast cancer, examination of family history should be emphasized in addition to lifestyle modification.
4. Two research strategies often are used to estimate the relative influence of genes and environment-lifestyle: twin studies and adoption studies.
5. Monozygotic twins originate when the developing embryo divides to form two separate but identical embryos.
6. Dizygotic twins are the result of a double ovulation followed by the fertilization of each egg by a different sperm.
7. If both members of a twin pair share a trait, they are said to be *concordant*. If they do not share the same trait, they are *discordant*.
8. Studies of adopted children also are used to estimate the genetic contribution to a multifactorial trait.
9. A genetic predisposition may interact with an environmental-lifestyle factor to increase the risk of disease; this is called a *gene-environment interaction*.

#### Genetics of Common Diseases

1. Congenital diseases are those present at birth. Most of these diseases are multifactorial in etiology.
2. Multifactorial diseases in adults include coronary heart disease, hypertension, breast cancer, colon cancer, diabetes mellitus, obesity, AD, alcoholism, schizophrenia, and bipolar affective disorder.

3. It is incorrect to assume that the presence of a genetic component means that the course of a disease cannot be altered—most diseases have *both* genetic and environmental-lifestyle aspects.