



# Frumkin, 2e

## Part 1: Methods and Paradigms

**Chapter 6: Genetics and Environmental Health**

# Genetics

- Genetics, the study of individual genes, has expanded to include **genomics**, which is the study of all the genes that make up an organism.
- As regards environmental health, genetics is important for the gene-environment interaction.
- People respond differently to exposure to toxins as a result of genetic variability.

# Susceptibility

- Certain genes cause individuals to be susceptible to certain diseases; these are called **susceptibility genes**.
- Genes also determine our **environmental response machinery**, the biologically conditioned responses to exposure.



# Human Genome Project

- In 2001, scientists in the **Human Genome Program (HGP)** completed the sequencing of the human genome, providing a complete roadmap to the locations of the approximately 30,000 human genes.
- The genome sequence information will be used as a reference in the process of learning about individual human **genetic variation**.

# Genetic Variation

- Variations in genes or groups of related genes result in **phenotypes**. Phenotypes can describe physical traits such as hair color, behavioral features such as anxiety, and specific physiological susceptibilities or responses to gene-environment interactions.
- Genetic variation among individuals is typically due to insertions and deletions of DNA known as **indels**, and **single nucleotide polymorphisms (SNPs)**, which are normal variations of one letter of the genetic code.
  - *SNPs tend to occur in patterns, or blocks, of associated, inherited alleles called **haplotypes**.*

# Environmental Genome Project

- **Environmental Genome Project (EGP) ( 2005 ).**  
The EGP uses the candidate gene approach to identify and characterize human genetic variability in selected genes thought to be involved in susceptibility to toxicant-induced disease.
- The environmentally responsive genes selected for study by the EGP tend to fall into eight categories: cell cycle, DNA repair, cell division, cell signaling, cell structure, gene expression, apoptosis, and metabolism.



# Advances in Exposure Assessment

- Methodologies are emerging to characterize dose-response curves at the molecular level (such as gene expression, protein expression , metabolism, and so forth).
- High-throughput analytical tools along with the computational tools necessary to interpret massive amounts of data have allowed the development of the scientific paradigm known as **systems biology**.
- In systems biology the goal is to understand the functioning and responses of the entire organism by integrating information about its elements (such as genes and proteins) with knowledge about these elements ' interrelationships.

# The Genes, Environment and Health Initiative

- **The Genes, Environment and Health Initiative** seeks to analyze genetic variation in groups of patients with specific illnesses, and to produce and validate new methods for monitoring environmental exposures that may interact with a genetic variation to result in human disease.



# A Challenge to Investigators

- Those investigating gene-environment interactions often assume that their knowledge about exposure is correct, and seek out genes that determine responses to a particular compound of exposure.
- However, it could be that their knowledge about exposure is incorrect, a fact that is overlooked in the current gene-focused research.

# Theomics Technologies

- Many of the traditional fields in biology have embraced an –omics component, a capability of studying biological phenomena on the genome scale.
- **Pharmacogenetics** examines the response of individual genes to medicine; it includes **pharmacogenomics**, which looks at drug response over the entire genome, identifies drug targets, screens compounds for medicinal activity, and characterizes response phenotypes.
- **Toxicogenomics** takes a systems biology approach to an organism's response to an exposure.
  - ***Proteomics**, the study of the proteome, the global expression of proteins in a cell, and **metabolomics**, which involves the identification of metabolites, or suites of metabolites, in body fluids as they relate to particular responses have also become important fields of study in environmental health.*

# Epigenetics

- Changes in the mechanisms that carry out the genetic code, such as messenger RNA, are another important route by which exposure can lead to disease.
- These changes can happen as a result of individuals' normal development, exposure to environmental chemicals, ingestion of pharmaceuticals, or diet.
- The study of these types of modifications to single genes or sets of genes is known as **epigenetics**.
- Analysis of these changes across the entire genome is referred to as **epigenomics**.