



# 2010 Summer Institute in Public Health Genomics:

*Translating Genomics into Policy and Practice*

Presented by  
The University of Washington  
Center for Genomics and Public Health

**SUPPLEMENT FOR VIDEO 4:  
PHARMACOGENOMICS**

## VIDEO 4

# PHARMACOGENOMICS

### THEMES COVERED IN THIS LECTURE:

- Varied response rate to drugs
- Factors to consider when assessing pharmacogenomic tools
- Graphical cost-effectiveness evaluation



## KEY CONCEPTS

- *Genomics*: A branch of genetics that investigates the entire genome (not only genes) and intragenomic relationships, e.g. gene and protein interactions.
- *Pharmacogenomics*: A branch of pharmacology that investigates the influence of genetic variation on drug response in patients.
- *Concepts from Video 3, Decision Analysis*
- *The “Central Dogma” of Biology*: The traditional dogma of DNA → mRNA → protein.
- *Protein synthesis*: The combined processes of transcription and translation, by which a protein is synthesized from a sequence of DNA.
- *Enzyme activity*: Enzymes are specialized proteins that synthesize, decompose, or assist in chemical reactions. For example, numerous enzymes catalyze the process of protein synthesis.
- *Cell membrane transporters*: Other specialized proteins that regulate the flow of ions and molecules across the cell membrane. Some are enzymes that actively facilitate transportation across the membrane; others serve as molecular “tunnels” or “escorts.”

## KEY CONCEPTS

- *Point & chromosomal mutations:* Point mutations cause the substitution of a single nucleotide with another, or a deletion or duplication of a single nucleotide or base pair. Chromosomal mutations involve large scale changes, e.g. duplications of entire chromosomes or the translocation of an entire part of a chromosome to another.
- *Oncogene & tumor suppressor genes:* Oncogenes can potentially cause cancer. Tumor suppressor genes protect a cell from progressing on the path to cancer.
- *Alleles:* The specific variation of a gene.
- *Genetic polymorphism:* The existence together of many forms of DNA sequences at a specific locus within the population.
- *Penetrance & expressivity:* The first term refers to how many people in a population will actually display a phenotype if possessing a certain phenotype; the second term refers to the extent to which a particular phenotype is “expressed” in one individual.
- *Thiopurine methyltransferase:* An enzyme that methylates thiopurine compounds. TPMT is best known for its role in the metabolism of the thiopurine drugs such as azathioprine, 6-mercaptopurine and 6-thioguanine, used in acute lymphoblastic leukemia, autoimmune disorders (e.g., Crohn's disease, rheumatoid arthritis), and organ transplant recipients.