



## Uses of Data from the Human Genome

Information adapted from "Medicine and the New Genetics" U. S. Department of Energy,  
[http://www.ornl.gov/TechResources/Human\\_Genome/medicine/medicine.html](http://www.ornl.gov/TechResources/Human_Genome/medicine/medicine.html)

### 1.) Gene Testing – diagnosis and prediction of disease and disease susceptibility

Currently (Summer, 2003), there are more than 900 genetic tests available. As we identify more genes associated with specific disorders, that number will grow.

- Carrier screening, which involves identifying unaffected individuals who carry one copy of a gene for a disease that requires two copies for the disease to be expressed
- Preimplantation genetic diagnosis
- Prenatal diagnostic testing
- Newborn screening
- Presymptomatic testing for predicting adult-onset disorders such as Huntington's disease
- Presymptomatic testing for estimating the risk of developing adult-onset cancers and Alzheimer's disease
- Confirmational diagnosis of a symptomatic individual
- Forensic/identity testing

### 2.) Gene therapy – treating genetic disorders by correcting defective genes

- A normal gene may be inserted into a nonspecific location within the genome to replace a nonfunctional gene. This approach is most common.
- An abnormal gene could be swapped for a normal gene through homologous recombination.
- The abnormal gene could be repaired through selective reverse mutation, which returns the gene to its normal function.
- The regulation (the degree to which a gene is turned on or off) of a particular gene could be altered.

One of the challenges of gene therapy is delivering the desired functional gene into cells.

Methods of delivery:

- Viruses (retroviruses, adenoviruses, adeno-associated viruses, herpes simplex virus) – immune system complications
- Direct delivery – inefficient
- Liposomes
- Human artificial chromosome - experimental

3.) Pharmacogenomics – study of how an individual's genetic inheritance affects the body's response to drugs.

- Drug development – researchers can explore drugs to act on specific protein targets associated with disease
- Personalized medicine – prescribing the right drug for the patient based on genetic profile rather than by trial and error, could significantly reduce number of adverse reactions to medication
- More accurate dosing
- More accurate diagnoses resulting in more appropriate drug therapy

4.) Studying human genetic variation and our evolutionary history

- History of human migrations
- Genetic comparisons with our closest relatives

5.) Understanding mechanisms of disease and of normal cell function at the molecular level