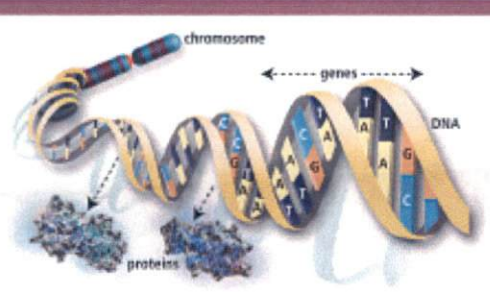


Human Genome Landmarks

Selected Genes, Traits, and Disorders

www.ornl.gov/hgmis/posters/chromosome

perovics.energy.gov



A Closer Look

Chromosomes are tightly coiled microscopic structures made of DNA, which consists of four different building blocks called bases (A, C, G, and T). The base bases are repeated millions of times to form chromosomes. The entire human genome consists of 22 pairs of chromosomes with sex chromosomes in every pair coming from each parent. With few exceptions (e.g., red blood cells), each of the billions of cells in the human body contains a complete set of chromosomes—the genome.

The color-coded figures in this poster are simplified representations (karyograms) of chromosomes, which can be distinguished by observing their features through a light microscope. Each feature includes length, position of a characteristic region (centromere), and the particular pattern of light and dark bands that result from treatment with stains.

Genes are chromosome pieces whose base sequence (A, T, C, G) encodes the instructions for making proteins, which are the body's building blocks. Each of the many thousands of different proteins required for life, humans have an estimated 25,000 genes, with an average length of about 2,000 bases. Genes make up less than 2% of human DNA. The remaining DNA has important but still unknown functions that may include regulating genes and stabilizing the chromosome structure.

Researchers hunt for disease-associated genes by looking for large changes, found only in the DNA of affected individuals. Sometimes, disorders and traits map to particular chromosomes, and such genes are called chromosomal. Some, such as cystic fibrosis (CF) and sickle cell anemia (SC), are caused by base sequence changes in a

single gene. Many common diseases such as diabetes, hypertension, deafness, and cancer have more complex causes that may be a combination of sequence variations in several genes on different chromosomes, in addition to environmental factors.

In April 2003, scientists in the International Human Genome Project (IHP) completed a highly accurate, 3 billion base pair human genome reference DNA sequence. The completion of the IHP coincided with the 50th anniversary of the publication of the structure of the DNA molecule by James Watson and Francis Crick. This achievement earned them the Nobel Prize and opened the new field of molecular biology that led to the completion of the IHP. DNA sequences are being obtained for the genomes of many other organisms, as well as are critical for comparative studies leading to a greater understanding of human biology and that of all living organisms.

A new era of Customized Medicine, knowing the DNA sequence is important because it affects responses to particular medicines, vulnerability to infections and toxins, and even intelligence. Sequence variations also can cause or contribute to such disorders as those caused by the gene.

These new data and powerful DNA analysis tools will usher in a new era of medicine that could allow doctors to detect disease at risk for stages, make more accurate diagnoses, and customize drugs and other medical treatments to fit an individual's own DNA sequence. The newfound understanding of gene functions will lead to more focused and effective treatments with fewer side effects.

For More Information

- Human Genome Project Information: Comprehensive IHP publications and a look at the "raw" data of the 2.9 billion letters. www.ornl.gov/hgmis
- Encyclopedia and its Impact on Science and Society. A Primer. www.ornl.gov/hgmis
- Genetic Education Resources. www.ornl.gov/hgmis
- NIH Consensus (CI). Expertise from scientists and patient genomes. Available for browsing and other applications. www.ornl.gov/hgmis
- Genetics in Society and the Bioethics Resources for students and teachers. www.ornl.gov/hgmis
- NIH Joint Consensus Initiative. Tools for Integrated High-Throughput Genotyping and Computational Analysis. <http://www.ornl.gov/hgmis>
- EMBLA Legal and Social Issues. Implications surrounding use of genomic data. www.ornl.gov/hgmis

Legend

- Blue: Regions affecting the structure of light and dark bands on human chromosomes (used to identify chromosomes)
- Red: Centromere or centromeric region of each chromosome
- Orange: Chromosomal regions that are in actively replicating or "open" chromatin
- Yellow: Regions of high GC content
- Green: Regions of high AT content

Source Information

Sequences from the Human Genome Project and other public domain genome sequences were used to generate this poster. The poster was designed by the Center for Genome Sciences and Policy, University of North Carolina at Chapel Hill. The poster is available for reproduction and distribution as of July 2003. Listing of genes on this chromosome poster is based on the Human Genome Project (HGP) data from the International Human Genome Project (IHP) and the Human Genome Project (HGP) data from the International Human Genome Project (IHP).

Gene Gateway

Step-by-step instructions for using the Web to learn about

Genetic Disorders

- Causes, inheritance, symptoms, diagnosis, treatments
- Associated genes
- Support groups and organizations
- Genetic health professionals
- Articles and other materials

www.ornl.gov/hgmis