The Human Genome Project

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The Human Genome Project, so far is a 250 million dollar effort to write out the map of all our genes. The HGP is publicly funded by the National Institute of Health and the Department of Energy and includes four large-scale centers in the US and one near Cambridge, England, plus labs in Japan, France, Germany, and China. There are also several private companies, including Celera Genomics that are also sequencing the human genome. The goal of the HGP is to identify all of the estimated 50,000 to 150,000 genes in the nucleus of the human cell, where they are located on the chromosomes, by a process called mapping, and to determine the genetic information encoded by the order of the DNA's chemical subunits, by a process called sequencing.

The human genome is composed of 50,000 to 150,000 genes that are located on the 23 pairs of chromosomes found in the nucleus of a human cell. A single human chromosome may contain more than 250 million DNA base pairs and the entire human genome may contain more than 3 billion DNA base pairs.

DNA is a double stranded molecule shaped like a twisted ladder, known as a double helix. It is composed of linked chemical compounds known as nucleotides. The nucleotide is made up of a sugar, known as deoxyribose, a phosphate compound, and any one of four bases (adenine, thymine, guanine, and cytosine). The sugar and the phosphate make up the parallel sides of the ladder while each base on one side pairs up with another base on the other side, to make up the rungs of the ladder. Adenine always pairs with thymine and guanine always pairs with cytosine.

A gene is a section of the DNA ladder that has a unique sequence of base pairs, and the genetic code is specified by the order of these base pairs.

There are two main categories of gene mapping techniques. One technique is called linkage mapping, which identifies the order of genes along a chromosome. The other technique is called physical mapping, which can detect the actual spacing of the genes on the chromosomes. Both techniques of mapping use detectable physical or molecular characteristics that differ from one human being to another and are passed on from generation to generation. These characteristics are called genetic markers.

Human linkage maps used to be created by following inheritance patterns in families for many generations. Now, researchers create linkage maps by comparing the order of genetic markers to the position of the target gene.

Physical mapping techniques use robotics, lasers, and computers to measure the distances between genetic markers. What they do is, they breakdown DNA into many pieces and clone them to test them for the presence or absence of specific genetic landmarks. Some of the clones have the same genetic landmarks, which is from the overlapping of the chromosome. The overlapping determines the order of the landmarks on the chromosome and the sequence of the pieces of DNA taken from the chromosome.

In order to determine the actual sequence of nucleotides, a very detailed physical map is needed. What takes place is, specific pieces of DNA are cloned and modified so that each piece ends in a fluorescent form of one of the four nucleotides. The modified nucleotide is then detected by a laser, which tells the exact number of nucleotides in that piece. The exact number of nucleotides is then analyzed by a computer, which reconstructs the sequence of the base pairs in the original DNA molecule.

About a week and a half ago, Celera Genomics announced that its scientists have published an accurate assembly of the human genome and an initial interpretation of the sequence. Celera estimates that there are only 26,500 to 30,000 genes, which is a big difference from the earlier estimation of 50,000 to 150,000 genes. Their publication also revealed that humans are 99.9 percent genetically identical. The 0.1 percent of genes that vary, lets say a simple single-nucleotide polymorphism (SNP), a T in one of your gene sequences where somebody else has a C, can cause trouble.

In extreme cases, a drug that saves one person might kill another person. For example, the type II diabetes drug Rezulin has been linked to more than 60 deaths from liver toxicity. In the future, a genetic test can make it possible to determine whether a drug can treat you effectively or determine if you face the chance of being killed by that drug.

In an effort called pharmocogenomics, drug companies are collecting information to make medicines tailored to specific genes. In the future, a doctor may prescribe one version of a drug based on your genetic profile, while he prescribes a different version of the same medicine to another person.

Much like any other scientific breakthrough, the HGP has many controversial ethical, legal, and social issues. One of the biggest issues right now is, the appropriateness of patenting human-gene sequences for commercial use.