



THE HUMAN GENOME

Biomedical Engineering Seminar III

Pilar Serrano Sanz

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pilarserrano@hotmail.com

The Human Genome: the book of life

The hereditary substance found in all living cells is deoxyribonucleic acid, or DNA. It is composed of four chemical subunits, called bases: A, T, C and G. The order of the bases provides coded instructions for everything a cell does.

The physical unit of heredity is a gene, which is a region of DNA that controls a hereditary characteristic and it is the instructions manual for making the proteins that carry out the functions, and malfunctions of the organism. DNA molecule is part of a chromosome, which is in the nucleus of the cell (23 pairs in each cell).

The complete set of genetic instructions carried within a single cell is known as genome. Determining the order of the bases along the DNA strand is called sequencing, and this is what the *Human Genome Project* is about: to provide the genome sequence for understanding the function of genes and their role in human health and disease.

The idea of determining the human genome sequence started in 1953 when Watson & Francis discovered the double helical structure of DNA, but it is in 1995 when HGP public consortium started working and in 1998 Celera was founded. Both of them got the complete sequence in February 2001, earlier than expected because of the emergence of high throughput techniques of DNA sequence determination.

All sequence produced by these projects is in a freely accessible database.

HPG tidbits

The major impact is to reveal how similar humans are to each other and to other species. Scientists found out that there are fewer genes than what they expected: the human genome has around 30,000 genes, only twice as many in a worm or fly. This makes the unity of life more obvious, and it blows the idea of human uniqueness. The similarity with chimpanzees is about 99%, and two humans only differ 0.1%.

And this project confirms, too, that there is no genetic basis for what people describe as race is not a scientific concept. Another point is that

most of the mutation that underlie evolution and bring gradual change is on the Y chromosome, which only men have.

Medical Applications

Having the complete sequence is the beginning of efforts to identify genes and to determine their function. This project opens the way to a *new medicine*. Parents will have the option to be told their carrier status for many recessive diseases. Diseases will be identified and treated to prevent problems before symptoms, with dietary or pharmaceutical interventions. In the future, the monitoring of individual drug response profiles will be a practice.

Although the initial purpose was to give the world new tools to treat and prevent diseases, ethical questions have appeared: Is genetic manipulation right? Which cases?

Iceland: Making the map of life

DeCodeGenetics is an Iceland company that is creating a *nationwide* health database (IHD). They are going to cross it with the company's genealogical database and genotypic data. The linkage of these resources will create a powerful analytical tool to search the human genome for gene mutations that are linked to the occurrence of a particular disease. Because nearly every disease has some genetic component, the identification of disease susceptibility genes can provide a wealth of new information vital to a more thorough understanding of human common illnesses. The idea is that, by comparing a gene that is mutated in patients is likely to be the disease gene.

It will allow faster diagnostics, allowing for earlier treatment or changes in lifestyle. It will provide customized treatments, specific to disease subtype and with a lower risk of side effects, better informed and more cost effective disease management strategies and more accurate drug targeting, leading to more effective, tailor made treatment regimes

Sources

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