ResearchConnection
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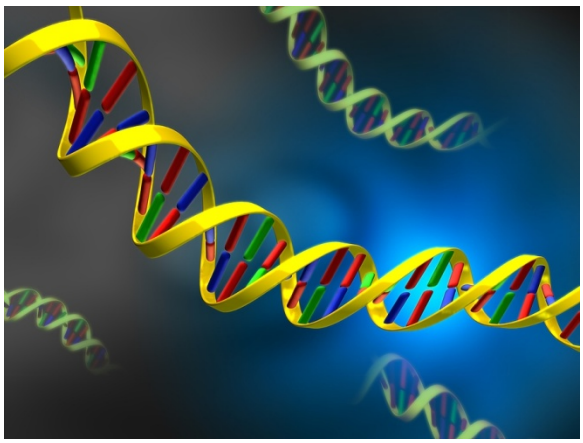
A Genomics Primer

What is a genome?

Each human being is a collection of about 100 trillion cells, each with its own function. The DNA contained within each of these cells carries the instructions needed to build and maintain the many different types of cells. This complete set of DNA instructions is a genome.



The genome or "genetic blueprint" of any given individual (except for identical twins and cloned organisms) is unique. However, what's surprising is that the genomes of any two people are very similar (at least 99.8% the same). But that tiny portion of the genome that *does* vary among humans makes every person genetically unique. DNA variations affect everything from the colour of your eyes to your risk of disease and your response to drugs.

Also surprising is what is often referred to as "junk" DNA: about 99 per cent of the genome has no *known* function, although many scientists now think that the term "junk" is outdated as much of the so-called "junk" may actually play an important role in the regulation of gene activity.

What is genetic or DNA sequencing? And what is next generation sequencing?

The term "DNA sequencing" refers to methods for determining the order of the nucleotide bases – adenine, guanine, cytosine and thymine – in a molecule of DNA. DNA can be extracted from humans through blood and other samples.

The first DNA sequences were obtained in the early 1970s by academic researchers using laborious methods based on two-dimensional chromatography. There have been

many advances since then, but today this field is very much driven by technology involving highly sophisticated machines.

The newest type of technology, called “next-generation sequencing“, has the potential to dramatically accelerate biological and biomedical research by enabling the comprehensive analysis of genomes, transcriptomes and interactomes to become inexpensive, routine and widespread, rather than requiring very costly production-scale efforts. The high demand for low-cost sequencing has driven the development of high-throughput technologies that can produce millions of sequences at once.

What was the Human Genome Project?

Started in 1990 and completed in 2003, the overall goal of the Human Genome Project (HGP) was to understand the genetic makeup of the human species.

The $3 billion international effort was coordinated by the US Department of Energy and the National Institutes of Health. Over 2,000 scientists across six countries were involved in the process of sequencing the DNA of each human chromosome using sophisticated computer programs.

Some other goals of the HGP were to:

- identify all of the approximately 20,000–40,000 genes in human DNA for further biological study
- determine the complete sequences of the 3 billion chemical base pairs that make up human DNA
- address the ethical, legal and social issues that could arise from the project

The HGP also focused on several non-human organisms, such as *E. coli,* the fruit fly and the laboratory mouse. It was one of the largest investigational projects in modern science.

What is a genetic mutation?

Each of us contains many slight variations in our genomes that make us unique. Most of these variations have little or no impact on our health. But sometimes, if a DNA letter is missing or wrong in a gene's instructions, it may produce a damaged protein, extra protein or no protein at all. These are genetic mutations, and they can cause serious health problems. For example, cystic fibrosis is a hereditary disease caused by a mutation in just a single gene. This gene, called CFTR, was discovered in Canada. In people with cystic fibrosis, the mutated CFTR gene produces a protein that doesn't work properly, resulting in numerous chronic health problems.

The transmission of genetic mutations from one generation to the next helps to explain why many diseases, including certain cancers, run in families.

What is comparative genomics?

Comparative genomics examines and compares the genome sequences of different species – human, rat and a wide variety of other creatures from roundworms to roosters. Since the mid 1990s, over 180 organisms have had their genomes sequenced. By comparing the human genome to genomes of other organisms, researchers can identify areas of similarities and differences and better understand a gene's structure and function. For example, researchers have found that two-thirds of human genes known to be involved in cancer have counterparts in the fruit fly. Research using organisms such as flies, worms and mice offers a valuable way to understand the function and regulation of genes and how they are disrupted in diseases like cancer.

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