



## The Human Genome Project - What's in it for us?



DNA Double Helix

Researchers worldwide have spent years mapping the entire genetic sequence of a human being. This human blueprint gives them the most fundamental and far-reaching insight into human biology and human medicine imaginable. The next step is to put it to good use.

### What is a genome?

A genome is the entire double helix shaped DNA in an organism. DNA

is made up of four chemicals called bases. These are abbreviated A (adenine), T (thymine), C (cytosine), and G (guanine). The human genome has around 3.2 billion pairs of bases. The sequence of these pairs is unique to each organism - humans, mice, fruit flies, or spiders!

### How will this affect our health care?

Dr. Francis Collins, Director of the National Human Genome Research Institute in Bethesda, describes the human genome as a giant instruction book with 3 billion letters in its set.<sup>1</sup> Each cell in our body carries all this information, but uses it differently depending on its type - blood cell, muscle cell, neuron, etc. The human genome is 99.9% the same for every person on earth. The 0.1% variation determines physical characteristics including the risk for different diseases. Certain conditions like heart disease, diabetes, asthma, and schizophrenia tend to run in families, but they are not inherited in a simple way. The next phase in research will discover the variations in individuals' genomes that put them at risk for illness. While most of us know the tenets of preventive healthcare, we tend to ignore these generalities. Would we not respond better to recommendations based on our own "user manual?" This will be possible in 10 to 15 years. Scientists are working to provide a complete personal genetic sequence for \$1000; currently it would cost \$10 million! This blueprint would become part of our medical record, and could guide decisions for screening and therapy.

### What about ethical issues?

Naturally there are concerns about genetic discrimination by employers and insurance companies. New legislation and policies on privacy and discrimination will be necessary to protect individuals. The National Human Genome Resource Institute (NHGRI) is collecting such information in a free database containing Federal and State laws and other legislative, administrative, and executive materials on many issues that will be affected.<sup>2</sup>

### New advances are speeding up genomic medicine

The genetic variation between individuals, though tiny, still translates into 10 million points where codes can vary - called polymorphisms. The HapMap, based on the analysis of DNA samples from around the world, identifies the "neighborhoods" of the human genome where polymorphisms are found for specific diseases. Gene sequencing speed has been boosted by a process that shatters DNA strands, allowing simultaneous sequencing of the fragments which are then knitted together again by computer.<sup>3</sup> As these new techniques evolve,

they will become more accurate, less expensive, and faster and will transforming science and medicine as we know it today.

### Genetic testing?

It's already here! Newborn screening (NBS) in the U.S. is the first and largest example of systematic, population-wide genetic testing. Newborn infants are screened for up to 50 specific metabolic, hormonal, and blood disorders as mandated by individual state health departments. Early detection and treatment for diseases such as cystic fibrosis has improved thousands of children's lives. Nevertheless, NBS is facing significant ethical and clinical challenges, posed by rapidly evolving technologies along with new economic and social forces.<sup>4</sup>

We already know enough to dodge some of our genetic bullets. For example, 5 - 10% of all breast cancers are hereditary. Genetic testing can determine if an individual has the BRCA1 cancer-predisposing mutation. Carrying a certain gene mutation does not automatically confer disease, but such awareness can inform decisions for life-style modification and treatment.

### Why not learn more?

The National Library of Medicine has launched a Genetics Home Reference Site which offers consumer-friendly information on genetics, genetic disorders and conditions, genetic testing, and gene therapy.<sup>5</sup> You can browse by specific gene or by condition. Aetna IntelliHealth and the Harvard Medical School have partnered to provide a Genetic Testing Guide,<sup>6</sup> which features articles, illustrations, videos, and an interactive decision tool.

### Resources

1. "Medical mapper." Potempa, A. *Anchorage Daily News*, Mar. 28, 2006: E1.
2. NHGRI Policy and Legislation Database. <http://www.genome.gov/LegislativeDatabase>
3. "The science speeds up." Komaroff, A.L. & J. Labaar. *Newsweek*, Dec.12, 2005: 57. Full text in Academic Search Premier database. <http://sled.alaska.edu/databases/home.html>
4. "Newborn screening: Complexities in universal genetic testing." Green, N.S. et al. *American Journal of Public Health*, Nov. 2006: 1955. Full text in Academic Search Premier database. <http://sled.alaska.edu/databases/home.html>
5. *Genetics Home Reference*. National Library of Medicine. <http://ghr.nlm.nih.gov/>
6. *Genetic Testing Guide: Information, Options, Decisions*. Aetna IntelliHealth & Harvard medical School. <http://www.intelihealth.com/genetics>
7. *The science behind the Genome Project*. <http://tinyurl.com/2g9k6r>
8. *Human Genome Resources* [including downloads]. <http://www.ncbi.nlm.nih.gov/genome/guide/human/>