

## 6.2 Decoding the Human Genome



Scientists are close to completing the genetic blueprint for a human being, thanks in large part to Office of Science funding. DOE was the first federal agency to propose that the human genome could be sequenced, and it launched the Human Genome Project in 1986. Today, DOE's Joint Genome Institute (a consortium of three laboratories) is one of the 16 institutions that constitute the Human Genome Sequencing Consortium, which recently announced completion of a working draft of the human genome. About one-fourth of the chemical sequence was finished and another half was in near-finished form or better. Contributing to that achievement was the JGI's completion of high-quality draft sequences of chromosomes 5, 16, and 19, which together contain some 12,000 genes,

including those implicated in forms of kidney disease, prostate and colorectal cancer, leukemia, hypertension, diabetes, and atherosclerosis. The JGI's DNA Production Genomics Facility is one of the most productive and cost-effective public-sector DNA sequencing laboratories in the world.

**Scientific Impact:** The large-scale sequencing work will provide a framework for efficiently answering many questions in biology, such as the number of human genes, recently estimated to be much lower (30,000) than previously thought (100,000). The JGI aims to develop and use new sequencing and computational technologies with the goal of discovering and characterizing the basic principles and relationships underlying living systems.

**Social Impact:** Armed with the DNA sequence of the human genome, scientists will be able to identify more genes responsible for countless diseases and develop diagnostic and treatment approaches. Genomic studies also should answer profound questions, such as why some people are able to defend themselves against the AIDS virus and others are not.

**Reference:** "Initial sequencing and analysis of the human genome. The Genome International Sequencing Consortium." *Nature* 409, 860-921 (15 February 2001).

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