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Learning about the Human Genome. Part 1: Challenge to Science Educators. ERIC Digest.

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We have reached a new milestone in our quest to understand ourselves. On February 15, 2001, the International Human Genome Sequencing Consortium published a landmark report: "Initial sequencing and analysis of the human genome" ("Nature," Vol. 409). As I thumb through the maps of human chromosomes presented in the report, I am both amazed and humbled by the accomplishment. The international collaboration of specialists in computing, mathematics, molecular genetics, technology, and other sciences to accomplish this task and usher us into the modern era of biotechnology has been extraordinary. At the same time, the potential consequences of using the resulting information and skills are humbling. "We've now got to the point in human history where for the first time we are going to hold in our hands the set of instructions to make a human being." John Sulston, UK Sanger Centre (For more reactions, see: http://news.bbc.co.uk/hi/english/sci/tech/newsid_807000/807126.stm).

Decoding the human genome brings new meaning to "the information age." In a few short years we have progressed beyond creating digital books and encircling the globe with the World Wide Web to documenting the full text of the genetic code that describes how to assemble and operate a human being. We may not yet know how to interpret all the text, let alone follow the recipe, but it is being recorded in full detail on the Web for anyone in the world to see. From the perspective of digital information, this is the book of human life, and decoding it is a phenomenal accomplishment of scientific thinking and our creative use of technology. The only thing more astounding is that this genetic recipe has been stored, read, and translated by every nucleated cell in every human that has ever lived.

But what are we to make of it? Presented here are some initial ideas about what high school students and informed parents can learn about the human genome. First, a few words about the source of recent findings. On October 1, 1990, a project-the Human Genome Project (HGP)-aimed at mapping and sequencing the entire genetic code of humans began. Funding of the project in the United States has been provided by the National Institutes of Health and the U.S. Department of Energy, with the goal of sequencing the entire human genome within 15 years. Due to advances in technology and vigorous competition, the project is currently well ahead of schedule and costing less than anticipated. A guide to the project and its accomplishments to date has been edited by Dennis and Gallagher (2001), and the rich context of events and issues leading up the project have been provided by Bishop and Waldholz (1999). A response to the ramifications-from interpreting the past to considering the future-of the HGP has been contributed by Ridley (1999). These and other authors characterize the accomplishments of the HGP as more than a momentous achievement; rather, the sequencing of the human genome is viewed as the beginning of a revolution in knowledge.

THE EMERGING LEGACY OF THE HGP

As spectacular as the accomplishments of the HGP have been, its ongoing importance will be revealed through what it enables us to do over time. The information gathered from the Project will fuel biological and medical research for years to come, transforming both science and how we use genetic information.

TRANSFORMING HOW SCIENCE IS DONE

The HGP has introduced fundamental changes in the way biological research is done (Butler, 2001). In addition to the advent of large-scale, international, multidisciplinary collaboration in pursuing an ambitious goal, we've seen the further transformation of biology into a computational science with huge data sets to mine. The online publishing of results through the World Wide Web is also notable and will likely further the trend toward nearly real time dissemination of research findings. Science educators will need to revise their descriptions of scientific enterprise to reflect these new ways of gathering, interpreting, and disseminating information.

DEVELOPING NEW TOOLS TO COMBAT DISEASE

Information and techniques associated with the HGP are leading to new approaches to examining and understanding the causes and mechanisms of diseases. A review issue of Pathology [Vol.195 (1)] was devoted to "Genomic Pathology-A New Frontier" Also, a publication by the Department of Energy, "Genomics and Its Impact on Medicine and Society: A 2001 Primer" (Available online at http://www.ornl.gov/hgmis/publicat/primer2001/index.html), outlines potential applications of genome research to medicine and disease control, including the following:

* Improving diagnoses of diseases

* Detecting genetic predispositions to disease

* Creating drugs based on molecular information

* Using gene therapy and control systems as drugs

* Designing "custom drugs" based on individual genetic profiles

EXAMINING THE HUMAN CONDITION

The ability to perform detailed analyses of DNA sequences enables both genetically describing individuals and discerning the genetic heritage of individuals and groups over time. Some of the applications of these procedures are outlined by "Genomics and Its Impact on Medicine and Society," including he following:

* Risk Assessment

Evaluating the health risks faced by individuals who may be exposed to radiation (including low levels in industrial areas) and to cancer-causing chemicals and toxins.

* Bioarchaeology, Anthropology, Evolution, and Human Migration

Studying evolution through germline mutations in lineages

Studying migrations of different population groups based on maternal genetic inheritance

Studying mutations on the Y chromosome to trace lineage and migration of males

Comparing breakpoints in the evolution of mutations with ages of populations and historical events

* DNA Identification

Identifying potential suspects whose DNA may match evidence left at crime scenes

Exonerating persons wrongly accused of crimes

Identifying crime, catastrophe, and other victims

Establishing paternity and other family relationships

Matching organ donors with recipients in transplant programs

THE HGP CHALLENGE FOR SCIENCE

EDUCATORS

Despite the rapid progress and success of the HGP, little attention has been given to the project and its findings within the standard school curriculum. Some have focused on the ethical dimensions (Morris, 1994; Rifkin, 1998), and others have focused on the challenge of teaching about DNA sequencing (Morvillo, 1997). As McInerney (1996) has pointed out, there has been no educational revolution in response to the biological revolution regarding our understanding of human genetics and the human genome. Given the inevitable role that genetics will increasingly have as a central feature of health care and public policy, it is crucially important that non-specialists come to understand genetics and its many applications. McInerney identified four challenges in translating the complexities of modern genetics to non-specialists:

* Teaching for Conceptual Understanding.

Conventional instruction in genetics typically is preoccupied with isolated facts, extensive vocabularies, simplistic single-gene traits, and typologies rather than variation.

* The Nature of Science.

As Mcinerney said, "Poor public understanding of how the scientific community generates and validates new knowledge likely is a more critical deficiency than is the public's lack of familiarity with any given piece of that knowledge." The best way to learn about the nature of science is to actually do science in the classroom, and there are many genome-related investigations appropriate for the high school classroom.

* The Personal and Social Impact of Science and Technology.

The pace at which advances in our understanding of genetics is raising once-hypothetical issues calls for greater attention to ethics and public policy matters in the science classroom. We must move beyond simplistic debates to engage students in critical analysis of arguments, reasoning, and views.

* The Principles of Technology.

Technology-dependent endeavors such as the HGP and genetic medicine highlight the

need to promote greater attention to the principles of technology in science classes.

The resources identified in Part 2 of this Digest are provided to assist educators in meeting these challenges. An indication of the potential linkages between the HGP and the National Science Education Standards is included, along with Web resources and instructional materials that can serve as starting places in developing school instruction and public outreach programs.

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