Editorial: The Human Genome Project

Dear readers,

The last few decades have seen a number of exciting developments in genetics. First, Watson and Crick broke the genetic code; since then, technologic and methodologic breakthroughs have permitted the study and direct manipulation of our DNA. Now there is an international ground swell to map and sequence the human genome. The Bush administration had originally requested $128 million in last year's budget for the Human Genome Project. However, a total cost of some $3 billion is envisaged over a 15-year period for the successful completion of the project. Few people doubt the value of this research, particularly in its application to human disease and gene therapy. The knowledge of the entire sequence of the 3 billion base pairs that comprise our 100,000 or so genes will biologically define a human being. However, little has been said about normal human variation and how it reflects our evolutionary past. With the rapid acculturation of small genetic isolates and the absorption of these groups into larger aggregates, much information on our evolutionary past is being lost. To date, the Human Genome Project has failed to address this problem.

A number of laboratories are attempting to collect DNA specimens from various human genetic isolates, but these research units are financially limited and collect few samples based on families or unrelated individuals. Because of the difficulties in obtaining these specimens of DNA, access to these genetic data is limited. One solution to this problem is the creation of living cell lines from blood specimens. These cell lines could be maintained indefinitely and the DNA extracted from them would be more readily available. Some laboratories are implementing such programs. In addition, recent breakthroughs in the forensic sciences permit the extraction of DNA from hair follicles, thus facilitating comparisons between populations that are highly isolated geographically.

I recently received a letter written by several concerned scientists about the establishment of an international center for the collection, processing, and storage of DNA samples from genetic isolates. This proposal, if implemented, would result in the preservation of the range of normal human DNA variation, a consequence of our evolutionary history. I would like to share this letter with you. It is reprinted below.

It is my opinion that the Human Genome Project should include support for the establishment of an international center for the preser-

vation of DNA specimens from human populations around the world and support for field research through mandates to the National Science Foundation and the National Institutes of Health. For example, the USSR has signed an agreement through the Siberian Branch of the Academy of Sciences to support the study of DNA variation in the indigenous populations of Siberia under the Human Genome Project. It is hoped that the US government will follow suit and allocate a portion of the multiple billion dollars earmarked for the Human Genome Project for research on DNA RFLP variation in genetic isolates, in particular, those located in the New World.

M.H. CRAWFORD
Editor-in-Chief

An open letter to the ASHG, the Human Genome Project, HUGO, and others:

Around the world many cultural-linguistic-genetic isolates, or semi-isolates, are fast being absorbed into larger groups. Many of these isolates are "tribal," rural, small, and politically weak. Many have longstanding genetic or cultural adaptations to their physical, biotic, and social microenvironments. For both historical and selective reasons, the isolates have gene and haplotype pools that differ from those of major social groups.

While linguistic and ethnographic analyses of many isolates are available, their genetic composition is largely unexplored. In the interest of documenting and evaluating human biodiversity, efforts must be made during the next decade to establish a cell/DNA bank that is as representative as possible of small human populations. Such a bank should include basic information on each cell donor, such as age, sex, health status, birthplace(s) of biological parents, and possible consanguinity of parents (through second degree).

The population-evolutionary-genetic study of the human species has fallen out of favor in the United States and other research-oriented countries. However, the establishment of a human biodiversity cell/DNA bank need not be excluded from other types of study. Eanes and coworkers have already made a start for establishing such a bank for the US population (*Am. J. Hum. Genet.* 45, No. 4, Oct. 1989 supplement, p. A237). Investigators of malarial infections, of human mitochondrial DNA polymorphisms, and of other biological phenomena, let alone blood bank authorities, could easily contribute to the collection phase of this project,

ancillary to their main work.

What is needed on a national, or better international, scale is a center for collecting, processing, and storing samples. Banked materials could then be made available for a variety of genetic or other biological studies.

Time is short. Human biocultural isolates are vanishing at a fast rate. If action is to be taken, it must be done soon.

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