On February 15, 2001, the International Human Genome Sequencing Consortium published a draft sequence and initial analysis of the 3.2 gigabase human genome (Sachidanandam et al., 2001). Eric Lander, Director of the Whitehead Institute Center for Genomic Research stated, “We are standing at an extraordinary moment in scientific history. It is as though we have climbed to the top of the Himalayas. We can for the first time see the breathtaking vista of the human genome.” (U.S. Department of Energy, 2001).

While experts like Eric Lander readily appreciate these sights, students studying genetics from traditional textbooks and labs have little frame of reference from which to evaluate the utility and complexities of the Human Genome Project. Many students have the misconception that by sequencing the entire human genome, virtually all genes are identified and their function known. They often believe that a single gene is responsible for a single trait, and any disease is the result of one “bad gene.” Despite this simplistic viewpoint, students are usually unaware of the vast amount of personally relevant genetic information that is available in the human genome databases by a few clicks on the Internet. No password required.

The project presented here allows students to develop their own incentive for researching the human genome databases, and along the way, to resolve for themselves some genetic misconceptions. Several good computer-based projects have been described previously that introduce students to the fields of genomics and bioinformatics (Puterbaugh & Burleigh, 2001; Smith & Emmeluth, 2002; Morvillo et al., 2000; Wefer, 2003). Our goal was to develop a hands-on exploratory project that combines such computer database research with laboratory experimentation, and is individualized in order to maximize student ownership and interest. We developed a multi-week project where students research and design a PCR assay to identify a gene of personal interest in their own genome. This project is driven by the student’s interest in the topic and the need to design his/her own experiment, allowing students a firsthand experience with the challenges and rewards of deriving information from the Human Genome Project. We incorporated this project into an upper-level Molecular Genetics course, but it could be adapted readily for courses at the introductory undergraduate or advanced high school level.

The project is spread over an eight-week period (see Table 2). In the first two weeks, students explore human traits or diseases of interest to them, search the literature and molecular databases of the Human Genome Project to pinpoint genes potentially involved in the characteristic, and locate the DNA sequence corresponding to such a gene. In the third week, students design their own PCR primers to amplify a portion of the gene, and over the subsequent three weeks, they conduct a PCR assay using their own cheek cell DNA as a template to identify the gene in their own genome. Students communicate their findings with each other twice during the course of the project. At the completion, our students were fully equipped to view the human genome from the top of Mt. Everest.

The Base Camp

The National Center for Biotechnology Information (NCBI) Web site (www.ncbi.nlm.nih.gov) is the base camp for our project, as the work of thousands of scientists is collected here in the public genomic databases. We begin by introducing students to the various tools on the NCBI Web site, explaining how anyone can access human genomic DNA sequences, search an annotated version of the human genome, find relevant literature, and actually see the chromosomal position of a gene and find its corresponding DNA sequence. For a guide to using the NCBI Web site, see http://www.ncbi.nlm.nih.gov/Education/index.html.

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