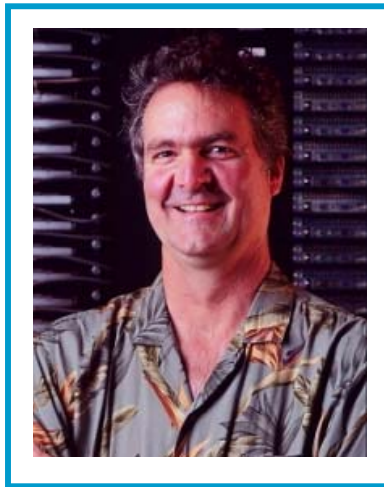**The CSU Information Science and Technology Center (ISTeC)**
and the Department of Computer Science

Present

Dr. David Haussler

Presidential Chair of Computer Science
Professor of Computer Science
Director of the Center for Biomolecular Science and Engineering
University of California, Santa Cruz

Friday November 14, 2003

The UCSC Human Genome Browser
ISTeC Distinguished Lecture
3:10 p.m.

and

Comparing the Human, Mouse and Rat Genomes
Computer Science Distinguished Lecture
4:30 p.m.

Colorado State University Campus
Lory Student Center Theater

Lectures are free and open to the public.
Please join us for receptions at 2:30 and 4:00 in the Lory Student Center Art Lounge.
Additional information is available at (970) 491-5862.

ABSTRACTS
The UCSC Human Genome Browser

Each day about 4,000 people visit the web site http://genome.ucsc.edu/ at the University of California at Santa Cruz and together request about 140,000 web pages of information about the human genome and the genomes of related species. A meld of computer science, genetics and molecular biology has developed into a new tool for exploring our shared genetic inheritance. This genome browser is a kind of interactive microscope on the recently completed reference sequence of the human genome, allowing users to zoom into specific regions in the vast landscape of 3 billion DNA bases that make up the human chromosomes. Advanced users can upload their own "tracks" of data and view it mapped to the genome sequence, while the typical user chooses from the standard tracks of information provided by UCSC and dozens of other labs worldwide. These tracks show the latest predictions on the locations of human genes, human genome variations within our population, comparisons between the human genome and the genomes of mouse, rat and puffer fish, and many more types of information. We'll give a live interactive demonstration of the UCSC genome browser to let everyone get a feel for the possibilities of this new mode of doing genetics and biomedical research, many of which are as yet unexplored.

Comparing the Human, Mouse and Rat Genomes

The reference sequence of the human genome was recently produced, along with drafts of the mouse and rat genomes. Data and bioinformatic analysis of these are available on the genome browser at http://genome.ucsc.edu, a site that is now receiving more than 140,000 page requests per day. We'll tell a bit of the history of this site, which was where the first publicly accessible working draft of the human genome was posted. Then we'll discuss comparative genomic methods to predict functional elements in these genomes, used in some of the analysis presented at that site. A statistical estimate based on a simple measure of conservation between short orthologous segments in the human and mouse genomes suggests (very roughly) that about 5% of the human genome shows signs of being under Darwinian selection (in particular, purifying selection), but does not definitively tell us which segments are under selection. We are using new models of molecular evolution to address this question. These models include context dependencies between substitutions at adjacent bases and allow for different rates of evolution in different parts of the chromosomes, as has been confirmed in recent analysis. These are being used to find regions of the human genome that are evolving like coding exons in genes, and occasionally lead to the prediction of a new human gene. Deeper analysis using genome data from 12 vertebrates in the region of the human cystic fibrosis gene has also allowed us to pinpoint a host of previously unexplored non-coding conserved elements in the human genome. Determining the function of these elements poses an intriguing problem. Finally, we'll look at the large scale rearrangements that have happened in the evolution of the human and mouse genomes, in an analysis that reveals chromosomal rearrangement "hotspots."

DR. DAVID HAUSSLER

Dr. Haussler and Jim Kent, a grad student at UCSC, developed the algorithm and software that assembled the human genome for the Human Genome Project. Their "working draft" of the genome has been instrumental to biomedical researchers in identifying genes related to specific diseases, in understanding how genetic variations affect susceptibility to diseases and responses to drugs, and in the design of new drugs.

Dr. Haussler obtained his Computer Science doctorate in 1982 at the University of Colorado. He is a Howard Hughes Medical Institute Investigator, and holds the University of California Presidential Chair in Computer Science at UC, Santa Cruz. He is the Director of the Center for Biomolecular Science and Engineering at UCSC, and scientific co-director of the multi-campus Institute for Bioengineering, Biotechnology and Quantitative Biomedical Research at the University of California, Berkeley, San Francisco and Santa Cruz.

His honors include Scientist of the Year by *Research and Development Magazine* in 2001, Fellow of the American Association for the Advancement of Science, and Boston Biomedica/Clinical Ligand Assay Society Distinguished Scientist of the Year in 2002.

Information Science and Technology Center (ISTeC)
ISTeC is a university-wide organization for promoting, facilitating, and enhancing CSU's research, education, and outreach activities pertaining to the design and innovative application of computer, communication, and information systems. For more information please see ISTeC.ColoState.edu.