nature genetics

Human Variome Microattribution Reviews

The journal has now begun to commission reviews on variation at selected loci as a way to demonstrate that microattribution can produce the required incentive for high-quality community annotation of the human genome.

Our aim is to produce a publication workflow that is open to all journals and that draws on the expertise of all those with a stake in understanding variation at a particular region of the human genome. Two international meetings this year could provide timely opportunities to discuss the implementation, priorities and software still required to make this work.

In April 2007, we highlighted an initiative called the Human Variome Project (HVP; http://www.nature.com/ng/journal/v39/n4/full/ng0407-423.html), a global effort to describe and annotate human genetic variation. Since then, many of the project's recommendations have been adopted or proposed for grant funding. The practical implementation of an incentive system and publication workflow has received considerable thought, and we will endeavor to keep this discussion prominent on the journal's blog (http://blogs.nature.com/ng/freeassociation/2007/11/towards_a_hermeneutics_of_quan.html).

A Human Variome Microattribution Review will consist of two components. First, a peer-reviewed locus published on the Variome Microattribution Browser will display the actual numbers of articles and database entries that refer to each variant, indexed to the genome at the variant nucleotide itself. Each variant nucleotide will also link via an annotation table to the data in the original study. Annotations that have been subject to peer review will be indicated in black. Unreviewed annotations and microattributed wiki comments indexed to each nucleotide will be in gray. The browser will have simple features to aid peer review: the genome coordinates of a locus under review will be marked, along with the deadlines for publication and the contact details of the coordinating author of the locus review, to prevent duplication of effort.

Concurrently with the locus review, a high-profile article will be commissioned by a participating journal to summarize the phenotypes and pathogenicity of all variants at a particular locus. The article and its associated browser publication will be published and cited together, much as the text, figures and supplementary information of a paper currently are. The annotation of the locus and writing of the article will be coordinated by locus-specific databases and experts on individual genes and loci, and will incorporate as authors all who have entered valid variant annotations into participating public databases by the commissioning deadline.

It is not the intention of the HVP to duplicate the work of existing journals, databases or browsers, but rather to provide an incentive process whereby all can receive credit for genome annotation via microattribution and high-level journal articles. All the underlying data should be deposited in stable public databases, such as NCBI and EBI. A Variome server would provide only that microattribution not available in source databases or journals. Functions can be distributed to journals, databases and browsers rather than centralized.

The Human Genome Organization (http://www.hugo-international.org/) might be the ideal organization to guarantee the credentials of all who are interested in annotating the human genome. All researchers change institutional affiliation during their career, and many even change their surname. And to complicate matters further, an estimated 330 million people share the surnames Wang, Li, Zhang and Liu. So, before collaborative annotation of the genome can be attempted, it will be necessary to uniquely identify all the participants. Each participant's presence would take the form of an individual home page in a social network. Such a page might provide a photo, secure contact email, institutional affiliations, CV, and lists of current publications and papers of interest, grants and collaborative networks. As journals develop author IDs and databases assign unique handles for data depositors, the HUGO Net page might become the definitive place where a genome researcher might authoritatively disambiguate his or her disparate identities.

We believe that it will be necessary to focus immediately on the software, workflows and priorities needed to produce Human Variome Microattribution Reviews. As we are all, in a sense, stakeholders in the human genome and there is such a diversity of expertise and opinion available worldwide, we have great hopes for two international meetings this year, the International Congress of Genetics in Berlin, July 12–17 (http://www.geneticsberlin2008.com/), and the HUGO Human Genome Meeting in Hyderabad, September 27–30 (http://hgm2008.hugo-international.org/). We propose to coordinate input at satellite sessions to those meetings and would welcome suggestions from interested parties. ■