

ETHICS WATCH

The \$1,000 genome: ethical and legal hurdles

There is much buzz these days about 24-hour, whole-genome genotyping of individuals for under US \$1,000. Craig Venter has announced that he will tackle this '\$1,000 genome', and some futurists are thinking of how it might be used in medicine¹. Although the technology for rapid whole-genome genotyping is not yet developed, the tools for doing so might soon be available².

Solving the technical problems fortunately gives us time to resolve the ethical, legal and social issues (ELSI) that this endeavour presents before it is ready for 'prime time'. Take the question of ownership of one's DNA, still an unresolved issue in many jurisdictions. Individuals have the right to decide whether tissue that contains their DNA is removed from their body, but few personal legal rights have been established over the genotyping or testing of DNA that is legally acquired or sloughed off a person.

To gain public acceptance of sequencing an individual's whole genome, and the testing that this will make possible, legal protections for consent and privacy are essential. However, at present, only 15 US states explicitly protect the rights of consent to testing, and Canada, Australia and most European countries do not explicitly protect an individual's rights to refuse testing and to have privacy in their test results³.

We also need to make clear rules about when genetic information might, or might not, be used in job and insurance decisions. To support a ban on genetic discrimination is easy, but there are some legitimate uses of genetic testing in both settings. A more balanced solution is needed, but we have yet to identify what it would be.

Finally, holders of patents on genes and DNA sequences could block rapid sequencing of the whole genome of an individual by making excessive royalty demands or refusing to license those uses⁴. Changes in the patent system, to allow freer use of gene patents while protecting incentives for innovation, are also needed.

All is not lost, however. Attention to the ethical, legal and social issues raised by the human genome project has identified most of the relevant issues, and this has helped to shape a consensus about how ethically to tackle many issues, including research with stored samples, biobanking, testing minors, setting standards for genetics counselling and the need to rethink patent law.

The prospect of a \$1,000 genome is a welcome prod for legal authorities to finish the work that the genomics, medical and bioethics communities have done on ELSI questions. It is time for clear, legal recognition of people's rights to control the acquisition, testing, use and privacy of their DNA. The \$1,000 genome has the potential to increase greatly access to the medical pay-offs of genomics. To get there, we will need to get our ethics and laws straight as well.

John Robertson



Diptych: 'Yin/Yang sienna', by Jacques Deshaies (2002) (detail).

REFERENCES ¹Pollock, A. Genome pioneer will start center of his own. *New York Times* A18, 15 August 2002 | ²Nature Science Update. *24-hour Genome Dawns* [online] <<http://www.nature.com/nsu/o20923/020923-2.html>> (24 September 2002) | ³Roche, P. A. & Annas, G. J. Protecting genetic privacy. *Nature Rev. Genet.* **2**, 392–396 (2001) | ⁴Merz, J. F. Patents limit medical potential of sequencing. *Nature* **419**, 878 (2002)

GENOMICS

Sea squirt genome released

The sea squirt *Ciona intestinalis* is an unusual organism with an unusual history. For more than a century, biologists have pored over this marine invertebrate due to its peculiar anatomical features and controversial taxonomy. It is now widely accepted that this species is a primitive chordate — that is, it has a cartilaginous column resembling a spine — and so, it is suitably placed to tell us about the evolutionary steps leading from invertebrates to vertebrates such as ourselves. The draft sequence of the *C. intestinalis* genome — which has been 18 months in the making — has now been published by Dehal and colleagues and represents the first genome of an invertebrate chordate to be published. In their paper, the authors present a preliminary analysis of the gene content of *Ciona* and how it compares with those of other sequenced animals.

The 160 million bp *Ciona* genome was sequenced using a whole-genome shotgun approach and is freely available on the web. It was found to contain ~16,000 protein-coding genes. This is similar to the number of genes in *Drosophila melanogaster* (~14,000) and *Caenorhabditis elegans* (~19,000), but around half the number estimated to be present in puffer fish (~31,000) and human (~30,000). It has been argued that extensive gene duplication was important in the evolution of the vertebrates. Comparative studies of gene families in *Ciona* and the vertebrates support this idea, as most of the genes that are present as multiple copies in vertebrates only have a single representative in *Ciona*.

The two physiological innovations that characterize vertebrates are their complex central nervous system and their adaptive immune system. A major evolutionary question is how and when these features arose. Genes required for these functions are absent from the *Ciona* genome, suggesting that they originated specifically in the vertebrate lineage. By contrast, there are *Ciona* homologues of genes required for other aspects of vertebrate



Adult sea squirt, image courtesy of Nori Satoh.

development that are absent in the fly and worm genomes, such as thyroid hormone production and detection, and heart development.

As well as shedding light on the origin of the vertebrates, interesting evolutionary insights can be gained from unique features of the *Ciona* genome. This is the first animal that has been found to have genes involved in the synthesis of cellulose. A putative cellulose synthase has been identified that is likely to be involved in the production of the outer tunic of the adult sea squirt. This gene shares homology with those found in nitrogen-fixing bacteria and so might have been acquired by an ancestor of *Ciona* through horizontal gene transfer from a bacterial genome. Also of interest is a new family of receptors that contain two domains — a caspase domain and a region with homology to proteins involved in *Notch* signalling — that have never been observed before in a single protein, thereby providing an example of domain shuffling.

This paper illustrates the power of genome projects to give insight into evolutionary processes. Sea squirts might be only distantly related to humans, but their genome sequence adds another perspective to that given by other sequenced vertebrate and invertebrate genomes. Undoubtedly, the release of the sea urchin genome, expected in the next ten months, will complement this work and will provide more information about meta-zoan evolution.

Catherine Baxter

References and links

ORIGINAL RESEARCH PAPER Dehal, P. et al. The draft genome of *Ciona intestinalis*: insight into chorate and vertebrate origins. *Science* **298**, 2157–2167 (2002)

WEB SITE

***Ciona* genome project:** <http://genome.jgi-psf.org/ciona4/ciona4.home.html>