
Web watch

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GENOMICS AND THE HUMAN GENOME

ScienceWatch newsletter reports on the “Hottest Research of 2011” (http://www.sciencewatch.com/newsletter/2012/201203/hottest_research_2012). Many articles on genomes and genomics are listed, including a number by top-ranked researcher Eric S Lander. One of the 14 “Hot Papers” to which he contributed was “A Map of Human Genome Variation from Population-Scale Sequencing”.¹ It is available under a Creative Commons licence at <http://www.nature.com/nature/journal/v467/n7319/full/nature09534.html>.

The genome is the complete set of genetic material of an organism including genes and non-coding portions of DNA (or RNA). Genomics is the study of the genome, and it is fundamental to the understanding of the relationship between genetics and disease. Topics noted in the list included genomic analysis of multiple myeloma, hypertension, and cancers of the lung, breast and pancreas.

The Human Genome Project is an international project that aims to determine the sequence of human DNA. It started in 1990, and a “rough draft” of the genome was finished in 2000 (http://www.en.wikipedia.org/wiki/Human_Genome_Project). Many other projects have expanded on the initial work.

INTRODUCTORY INFORMATION

There is a lot of general information on the web about genomics.

You can search or browse the Talking Glossary of Genetic Terms at <http://www.genome.gov/glossary/index.cfm?id>. Selecting a term brings up spoken, detailed definitions by working experts. Some terms include illustrations or 3D animations. You can also read profiles of the scientist speakers and test your gene knowledge.

There is a historical introduction with a colourful image of the human genome sequence at <http://www.australiangeographic.com.au/journal/one-this-day-in-history-human-genome-completion.htm>.

There are many videos on the web that provide a good introduction to genetics. For example, at <http://www.bioinformaticsonline.com> topics include “An introduction to bioinformatics” and “Human Genome Project”.²

There is content on medical conditions, mutations, health and therapy in the Genetics Home Reference from the US National Library of Medicine at <http://www.ghr.nlm.nih.gov/handbook/hgp/description>.

There is also an excellent range of content at the Wellcome Trust and US Department of Energy websites.

WELLCOME TRUST: THE HUMAN GENOME

The Wellcome Trust (<http://www.genome.wellcome.ac.uk>) hosts a website on the genome with general and specific information including:

- a news feed of recent discoveries;
- articles, eg comments from a number of scientists on the significance of the Human Genome Project (http://www.genome.wellcome.ac.uk/doc_WTX060064.html);

* All websites cited were viewed June 2012.

¹ See (2010) 467 *Nature* 1061.

² This website is heavily advertising based, but the speakers seem authoritative, eg Daniel Reda from Singularity University (<http://www.curetogether.com/blog/about/team>).

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- sections on “In the genome”, “Genes and the body”, “Tackling disease” and “Genetics and society”.

The “In depth” tab links to specific topics/collections, including “Single gene disorders” and the Crick papers (early drafts of the seminal 1953 James Watson and Francis Crick papers on the structure of DNA and copies of Crick’s most influential unpublished papers).

The Wellcome Trust Sanger Institute website (<http://www.sanger.ac.uk/about/history/hgp>) tells the story of the genome project and includes some entertaining cartoons. The Institute also describes its current research into genetics and disease (<http://www.sanger.ac.uk/about/what/difference.html>).

US DEPARTMENT OF ENERGY/HUMAN GENOME PROJECT INFORMATION

The US Department of Energy has a gateway site leading to its four genomic websites (<http://www.genomics.energy.gov>):

- DOE Genomic Science Program;
- Human Genome Project Information;
- Microbial Genome Program Archive;
- Image Gallery Gateway.

The Human Genome Project Information: Genome Glossary (http://www.ornl.gov/sci/techresources/Human_Genome/glossary) is browsable alphabetically, and contains cross-references from terms to related terms. The site also has content on:

- ethical, legal and social issues (societal concerns arising from the new genetics);
- medicine (diagnosing and predicting disease);
- education (teaching resources, including lesson plans, videos, webcasts and PDF documents, many aimed at the US curriculum. Includes links to websites in Spanish);
- gene gateway (guides to help novice users get started with the data from the Human Genome Project that is available through the web);
- research archive (archive of the Human Genome Project by topic).

COMMERCIAL EXPLOITATION AND PATENTING

As well as international scientific research into the human genome, there are also many commercial sequencing projects, and attempts by companies to patent human genes. In 1999, BBC News noted that Celera Genomics had sought 6,000 human gene patents, a move that outraged scientists (<http://www.news.bbc.co.uk/2/hi/science/nature/485951.stm>). Celera now holds numerous patents based on its discoveries (https://www.celera.com/celera/intellectual_property).

Robert Cook-Deegan and Christopher Heaney have reviewed the use of patents in genomics and human genetics (<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2935940>). The final version of the review can be purchased at <http://www.annualreviews.org/doi/pdf/10.1146/annurev-genom-082509-141811>.³

Recently the Australian Commonwealth government introduced the *Intellectual Property Laws Amendment (Raising the Bar) Bill 2012*⁴ (<http://www.comlaw.gov.au/Details/C2012A00035>). This Bill was designed to protect cancer researchers who wanted access to genetic material patented by corporations. Melissa Parke MP thinks this Bill did not go far enough and is lobbying the governing Labor party to accept a new private members’ Bill banning patents on all human genes (<http://www.abc.net.au/lateline/content/2012/s3502732.htm>).

³ Cook-Deegan R and Heaney C, “Patents in Genomics and Human Genetics” (2010) 11 *Annual Review of Genomics and Human Genetics* 383. First published online as a Review in Advance on 29 June 2010, DOI: 10.1146/annurev-genom-082509-141811.

⁴ Since enacted as the *Intellectual Property Laws Amendment (Raising the Bar) Act 2012* (Cth), No 35.

INFORMATION MANAGEMENT AND PRIVACY

The amount of data generated by the human genome project offers challenges in information management and sharing.

In 1991, Pearson and Soll discussed the Human Genome Project as “a model for the rapid dissemination of genome and related information in biology and medicine”.⁵ In developments they probably could not have imagined, data from the 1000 Genomes Project is now publicly available on the Amazon Web Services (AWS) cloud (<http://www.nih.gov/news/health/mar2012/nhgri-29.htm>) and also through the 1000 Genomes website (<http://www.1000genomes.org/data>).

The ethical issues to do with the easy availability of human genome sequence data are currently being discussed in the US (<http://www.healthdatamanagement.com/news/privacy-genetic-genomic-data-hhs-obama-44219-1.html>; <https://www.federalregister.gov/articles/2012/03/27/2012-7329/request-for-comments-on-issues-of-privacy-and-access-with-regard-to-human-genome-sequence-data#p-3>) and Australia. Timothy Smith, (<https://www.theconversation.edu.au/sharing-is-caring-we-need-open-access-to-genetic-information-6695>) argued “for the open sharing of clinical trial data among the medical research community” while Wendy Bonython and Bruce Arnold (<http://www.theconversation.edu.au/sharing-isnt-always-caring-genetic-privacy-must-come-first-6843>) argued that “while the potential benefits of sharing genetic information are enormous, they cannot be allowed to come at the expense of individuals’ rights to privacy of their medical information”.

Consent to Research (<http://www.weconsent.us/about>; <http://www.afr.com/p/technology/consent-open-source-for-gene-research-xDyWRCTT4HQjHTfRJ8jnfN>) is a not-for-profit company that provides legal tools, user engagement systems, and technology to enable people to choose to share their data, with the aim of creating “the world’s largest pool of openly available, user-contributed data about health and genomics”.

AND NOW ON TO THE CONNECTOME ...

While research into the genome has provided extraordinary insight into human biology, there is still much that we do not know. Work is proceeding on mapping the human connectome – the complete description of the physical wiring of the human brain and nervous system. Get started at:

- <http://www.humanconnectomeproject.org>;
- http://www.ted.com/talks/sebastian_seung.html;
- <http://www.scholarpedia.org/article/Connectome>;
- <http://www.guardian.co.uk/science/2012/may/07/quest-connectome-mapping-brain>.

VIDEOS ABOUT WRITERS

This YouTube video explains how James Frazer indexed books in preparation for writing his anthropological work *The Golden Bough* (<http://www.youtube.com/watch?v=X5OeMCSBOlo>). He wrote 12 volumes over a period of 20 years, using hundreds of footnotes. To keep track of his research, he recorded notes from his books, and then indexed these notes in a receipt book with carbon paper. One example of an index entry was “Princess, coming of age of a Zulu”.

Writer and speaker Christopher Hitchens died in December 2011. This memorial video by Alex Gibney shows excerpts from interviews and speeches that highlight Hitchens’ ideas (<http://www.vanityfair.com/video/2012/04/1573674109001?playlistid=1569514829001>).

⁵ Pearson ML and Soll D, “The Human Genome Project: A Paradigm for Information Management in the Life Sciences” (1991) 5 *The FASEB Journal* 35. Available at <http://www.ncbi.nlm.nih.gov/pubmed/1991581> and <http://www.fasebj.org/content/5/1/35.full.pdf>.

INTERNATIONAL TIME ZONES

I have a regular teleconference with people from Europe, Japan and the United States. The meeting agenda is sent with a link to the [timeanddate.com](http://www.timeanddate.com) website, which displays the time in different cities. This is handy, especially when daylight saving stops or meeting times change (<http://www.timeanddate.com/worldclock/fixedtime.html?month=3&day=2&year=2012&hour=21&min=0&sec=0&p1=0>).

CHASING THE SUN

Speaking of international time zones, “Chasing the sun” is a service by health libraries that “provides an after business hours online professional reference service, taking advantage of global time differences between countries to offer out-of-hours librarian support for clinicians” (<http://www.health.sa.gov.au/libguides.com/content.php?pid=213850&sid=1779306>).

Cheryl Hamill wrote to the aliaHEALTH mailing list on 18 April 2012 reporting the introduction of a similar service for radiologists, saying “Got to give it to those South Australian health librarians – they’re ahead of their time – only been doing that since – what – 2004?”⁶

FREE ONLINE COURSES

MIT and Harvard are planning to offer free online courses through their EdX technological platform. Courses will include video lesson segments, quizzes and student-paced learning (<http://www.newsbreaks.infotoday.com/Digest/MIT-and-Harvard-Launch-a-Revolution-in-Education-82444.asp>).

MIT has offered free courses through MIT OpenCourseware for some time (<http://www.ocw.mit.edu/courses>, <http://www.ocw.mit.edu/courses/most-visited-courses>) and Stanford offered an online course in artificial intelligence, which had 160,000 enrolments (<http://www.smh.com.au/technology/technology-news/rise-of-online-teaching-sets-a-clicking-pace-for-universities-20120427-1xqae.html>)

The non-profit Khan Academy (<http://www.khanacademy.org>) offers free online training with a selection of over 3,200 educational videos. Most of the content is science and mathematics videos, though other subjects including art history and microeconomics are offered.

Australian companies are involved in the development of software for online course provision (<http://www.smh.com.au/it-pro/business-it/teaching-startups-the-new-cool-kid-20120430-1xtuc.html>).

FOR FUN

If you ever have to choose a course to study, first go to the cartoon on this page (<http://www.xkcd.com/1052>) and sing along to the tune of “Modern Major-General”. (A lovely 1929 version of Gilbert and Sullivan’s operetta *The Pirates of Penzance* is available in the Internet Archive at <http://www.archive.org/details/GilbertSullivanThePiratesOfPenzance>; the song is in Act 1 at 31 min 41 sec.)

While in the mood for humour, you could also consider doing a laughter yoga course (<http://www.laughteryoga-australia.org>) or one on the biopsychosociology of humour (<http://www.handbook.unsw.edu.au/undergraduate/courses/2007/GENT1401.html>).

⁶ House C and Williams R, “The World Class in Radiology Services” (2012) 122 *Health Service Journal* 25.