Share your genome?

Dealing with very personal data.

Professor Adam Hedgecoe, Associate Director CESAGEN
ESRC Centre for Social and Economic Aspects of Genomics,
Cardiff University
Dr Liz Lyon, Associate Director, Digital Curation Centre,
University of Bath, UK

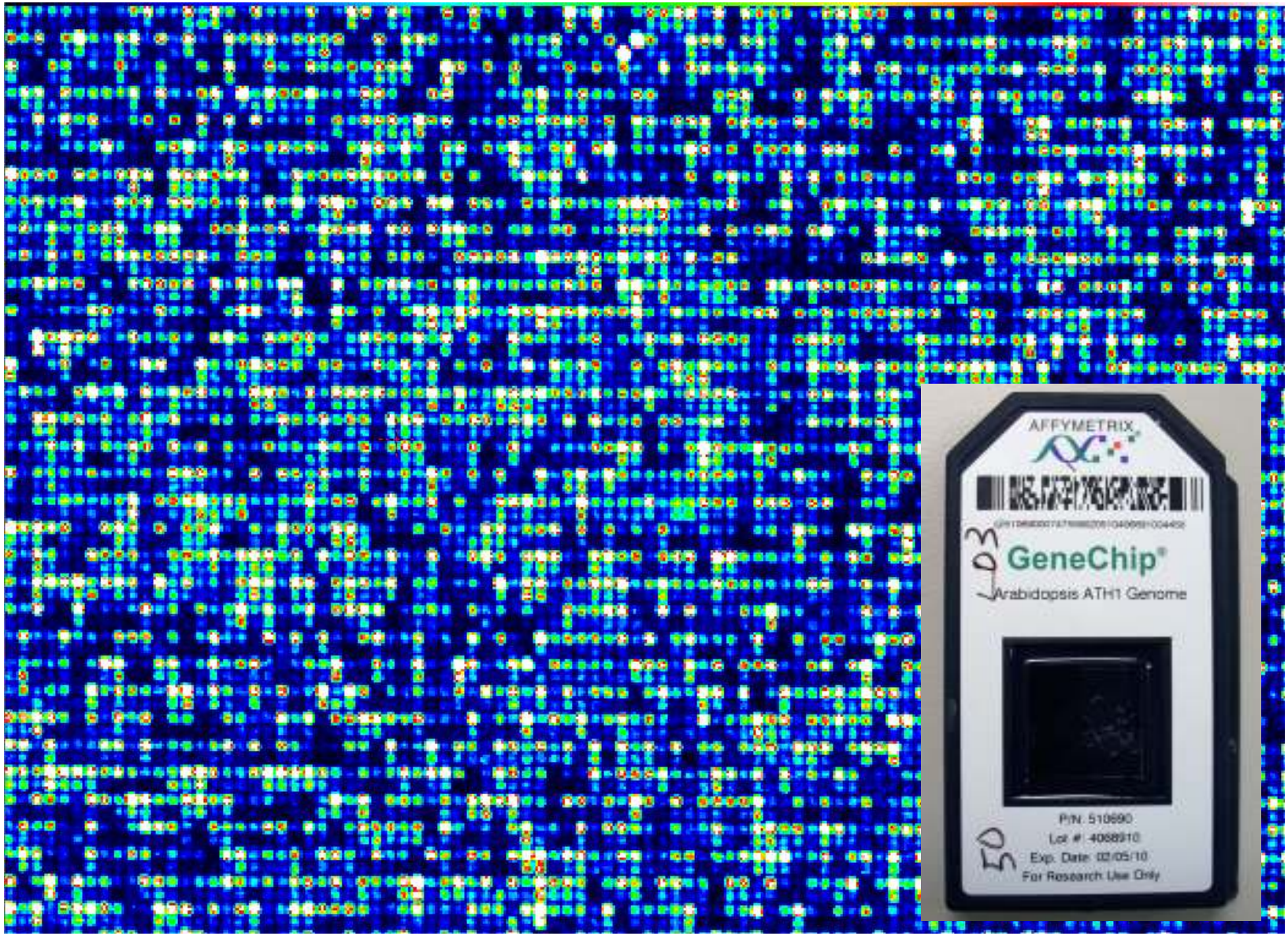
7th IDCC Symposium, Bristol, December 2011

UKOLN is supported by: JISC, Microsoft Research

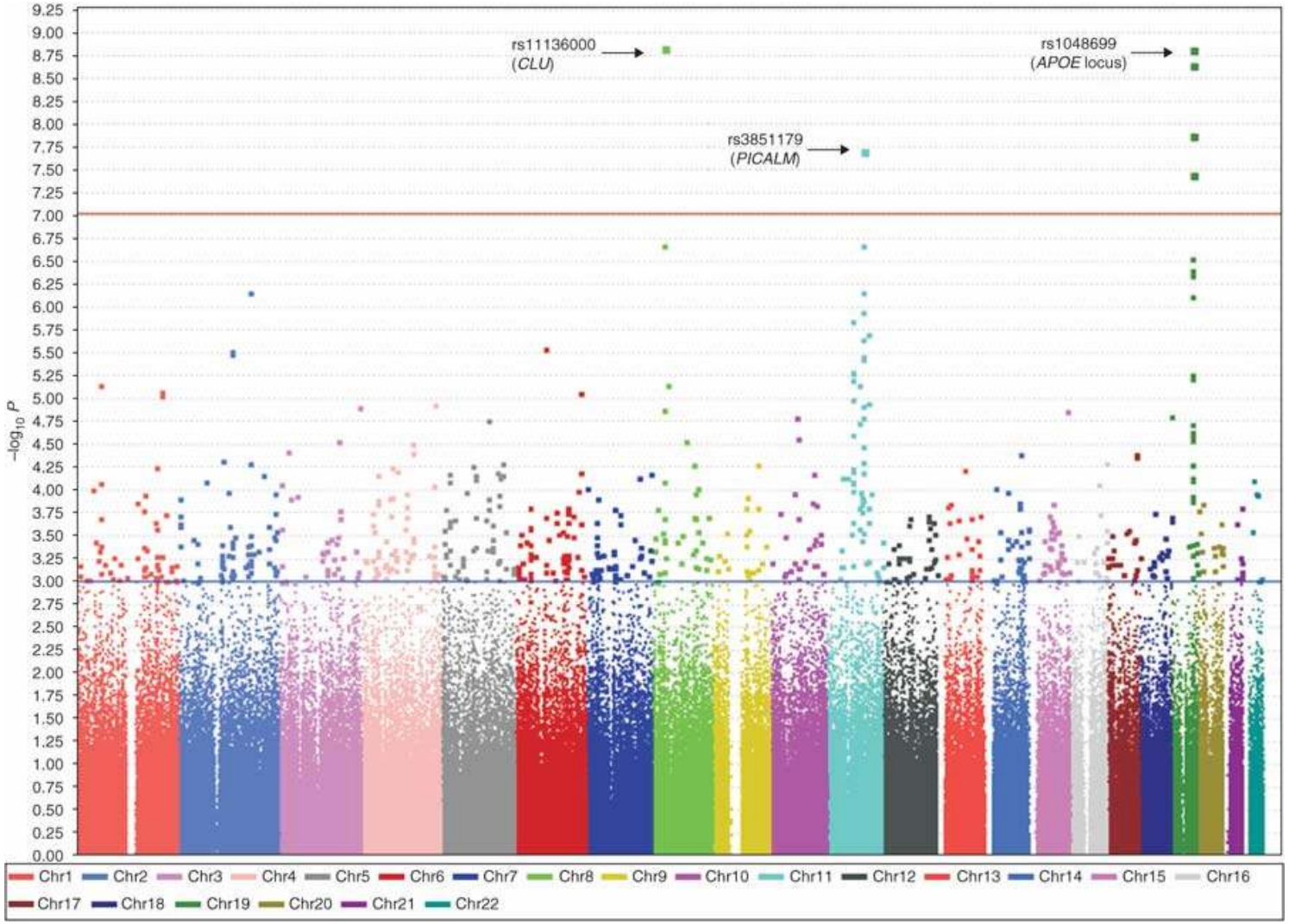
genomicsnetwork
cesagen
CARDIFF UNIVERSITY
PRIFYSGOL CAERDYDD

UKOLN
DCC
UNIVERSITY OF BATH
THE SUNDAY TIMES UNIVERSITY OF THE YEAR 2011-12

One of the major results of the Human Genome Project was the development of new technologies…



These technologies have led to 'big data' approaches to research into genetic disease.



Harold et al (2009) Genome-wide association study identifies variants at *CLU* and *PICALM* associated with Alzheimer's disease *Nature Genetics* 41: 1088 - 1093

Which in turn has led to the marketing of this genomic data directly to consumers.

Start filling in the gaps with your DNA

"Because I had given my doctor information from 23andme, he got to a diagnosis much faster. 23andme saved my life." Kirk C.

$99*

Our new low price for all!
Was $199

Order Now »

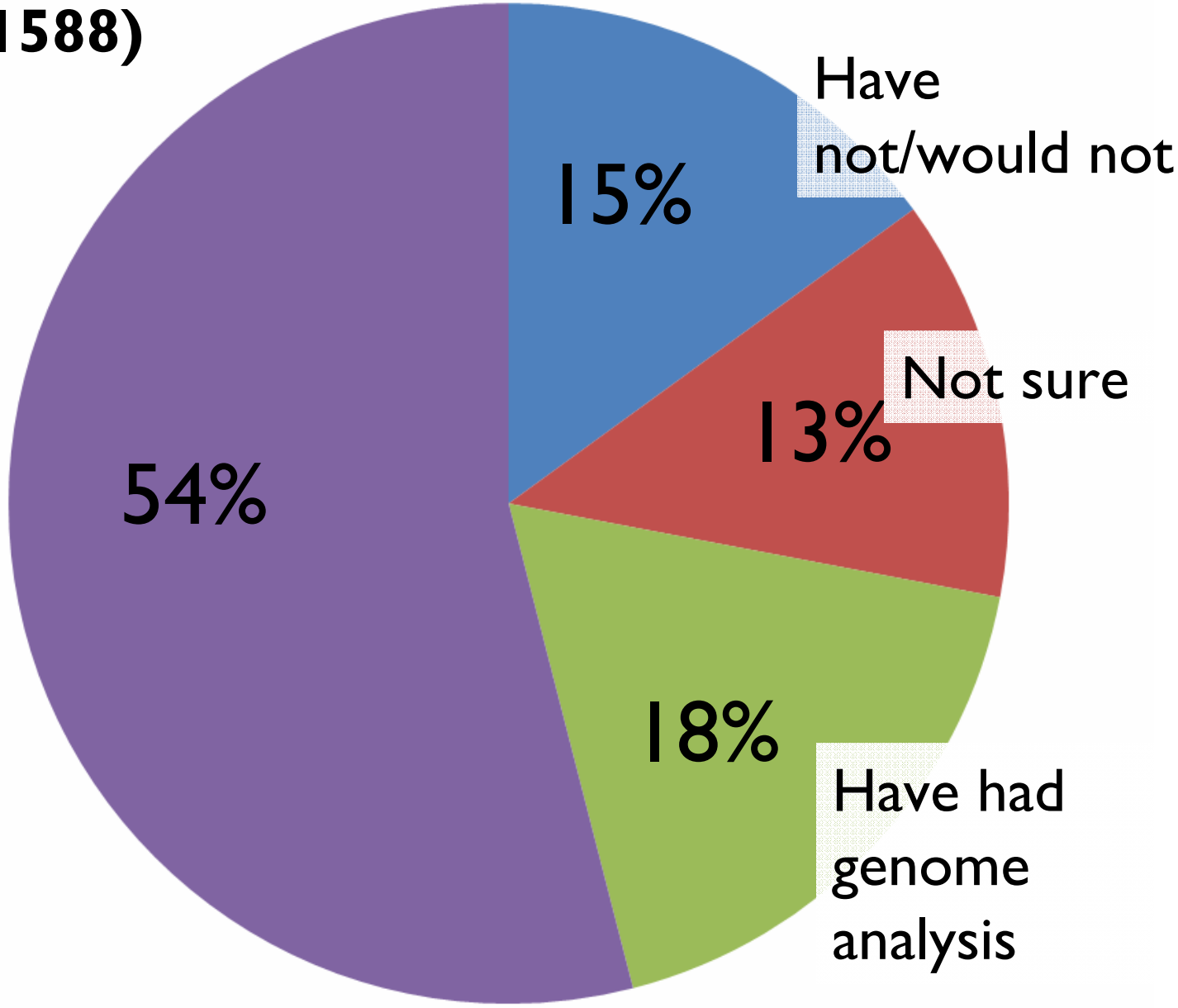
*Requires a 1-year commitment to the Personal Genome Service® at $9/mo. Order for $399 without commitment.

❶ Get Your Kit ❷ Provide Saliva

Gain insight into your traits, from ba
factors for 97 diseases. Know your
thinners to coffee. And uncover you

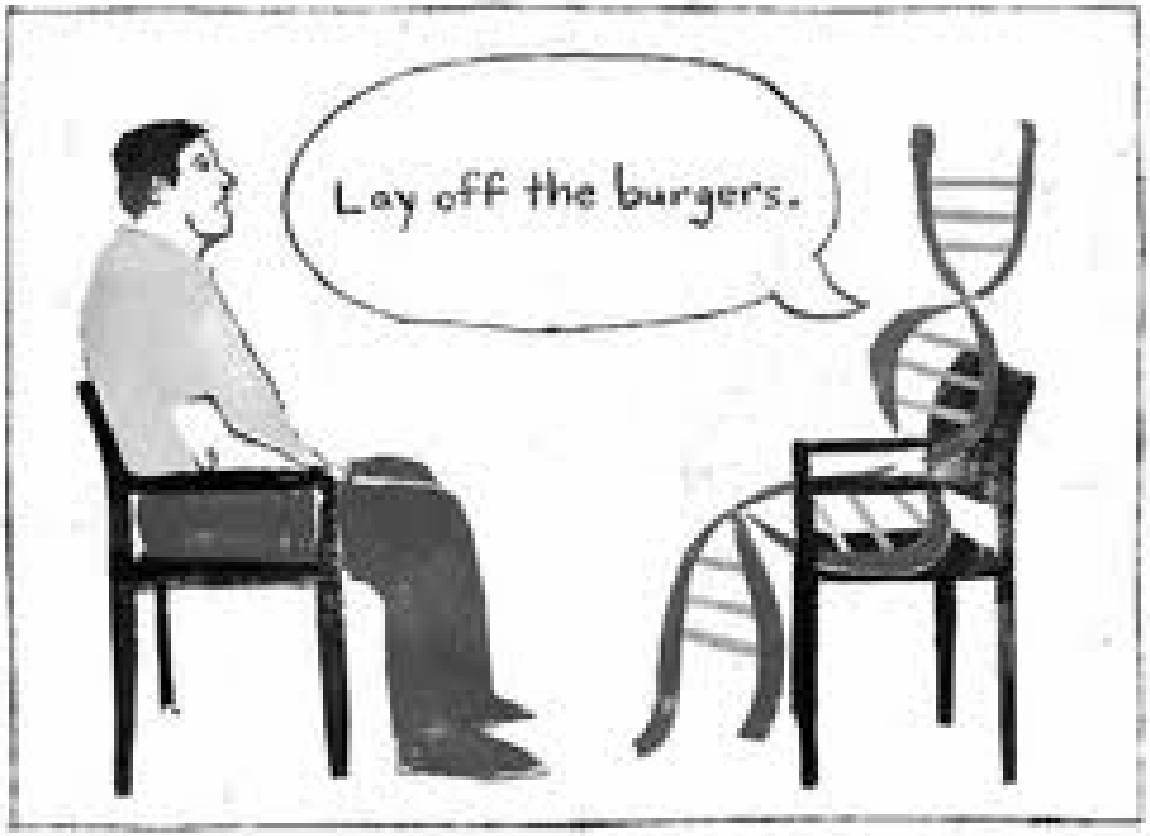
Overview | Discover Health & Ancestry | Keep Your Doctor Informed | Participate In Research

In a recent 2011 survey, *Nature* asked its readers whether they had, or would consider, a genome analysis (n=1588)



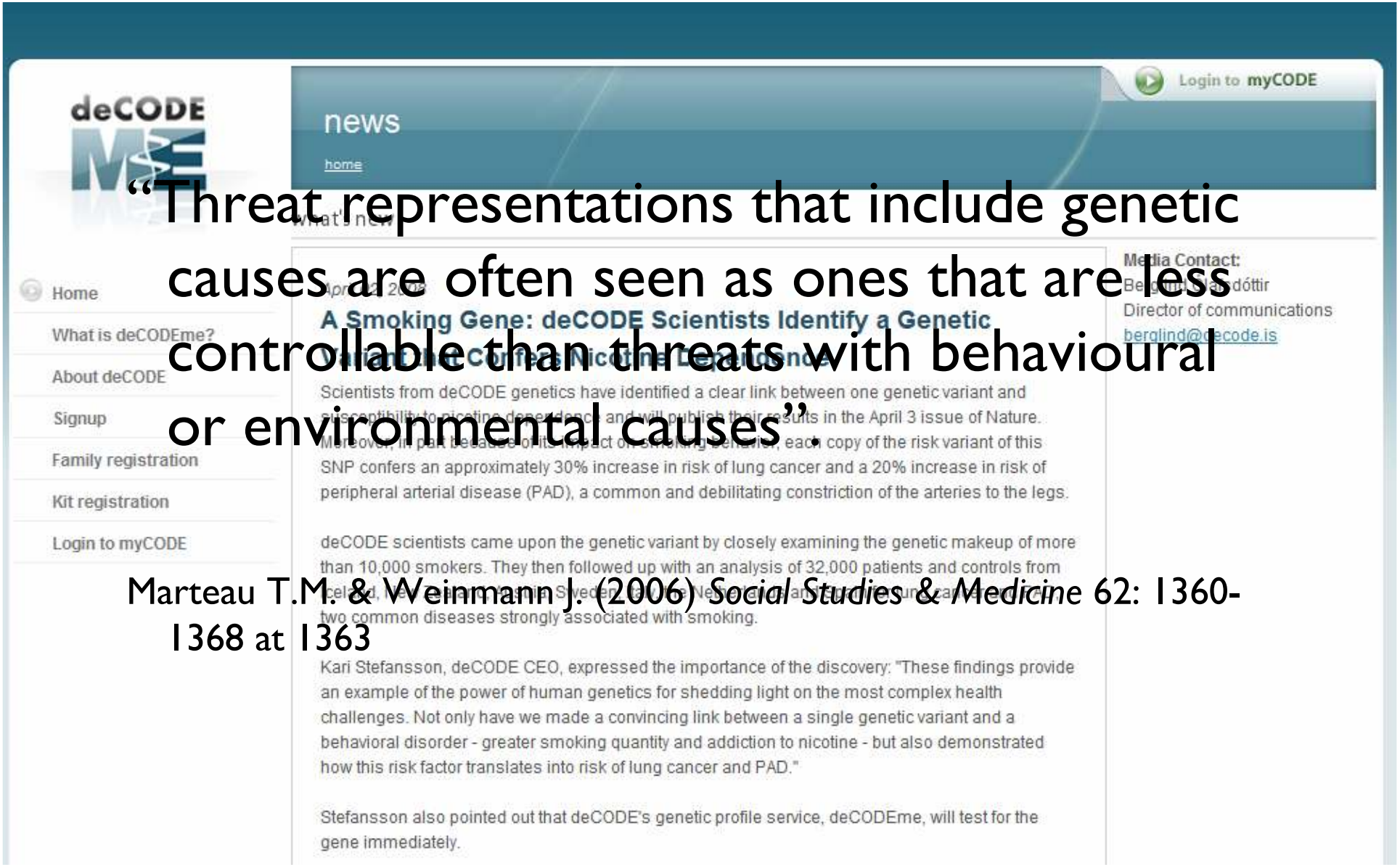
nature

This raises three core issues around the direct-to-consumer (DTC) marketing of these tests.



As yet these tests have limited clinical value, and give little useful information to consumers.

DTC genomics assumes a rational basis for people's decision making around health behaviours.



"Threat representations that include genetic causes are often seen as ones that are less controllable than threats with behavioural or environmental causes".

Marteau T.M. & Weinmann J. (2006) *Social Studies & Medicine* 62: 1360-1368 at 1363

The DTC model circumvents the traditional doctor-patient context for delivery of sensitive information

UC Berkeley adjusts freshman orientation's gene-testing program

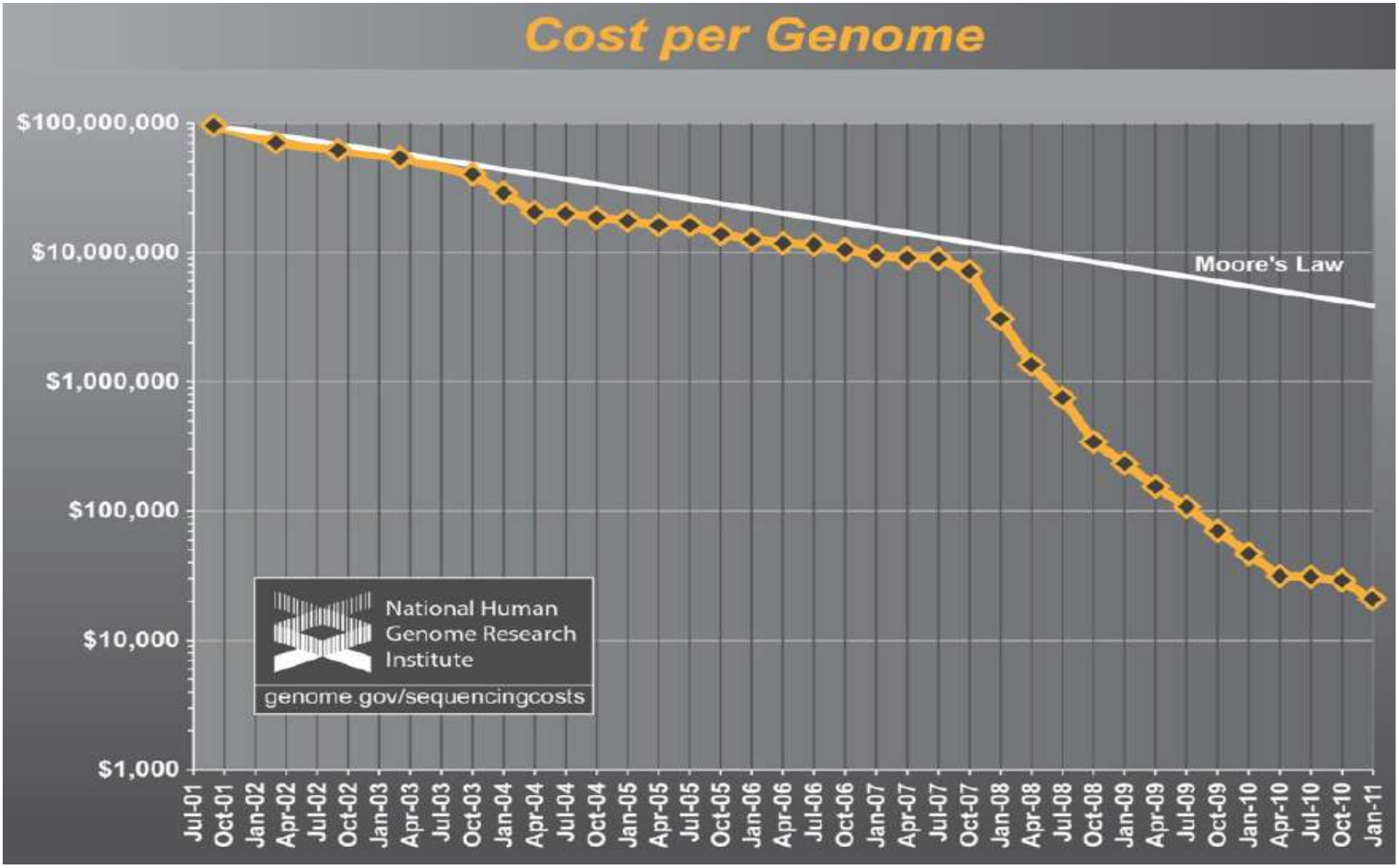
To comply with regulations on how DNA samples should be handled, the university is restricting the project by presenting only the collective results of all the participants.

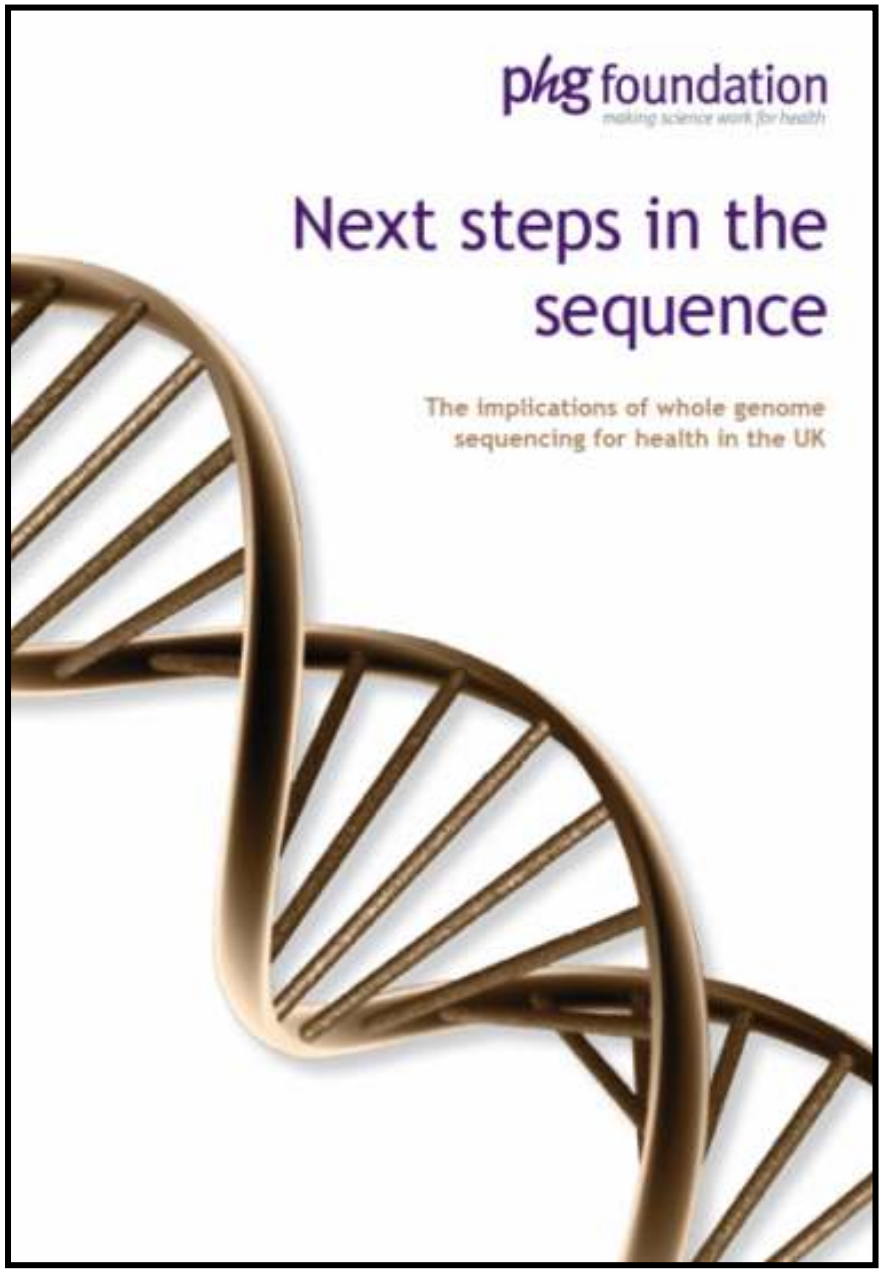
August 13, 2010 | By Larry Gordon, Los Angeles Times

UC Berkeley announced on Thursday a rollback in its controversial plan for voluntary genetics testing of incoming students, part of an orientation program called "Bring Your Genes to Cal."

In response to a state Public Health Department ruling on how DNA samples should be handled, UC Berkeley scientists reluctantly abandoned the idea to have freshmen and transfer students individually and confidentially learn about three of their own genetic traits. Instead, only collective results for all the 1,000 or so participants will be available and discussed at the orientation seminars next month.

Beyond genetic <u>testing</u>, full-genome sequencing is dropping in price and raising its own issues.





http://www.phgfoundation.org/reports/10364/

Debate is promoted and structured by various organisations, research projects & reports. …

Research in this area is underpinned by key 'ideological' belief in the value of open data

A critical new component of the Project is the selection of 2,500 DNA samples from 27 populations around the world. Each participant has provided explicit consent for full and public release of DNA samples and full sequence data….

- 1000 Genomes from 27 populations around the world
- Each participant provided explicit consent for full release

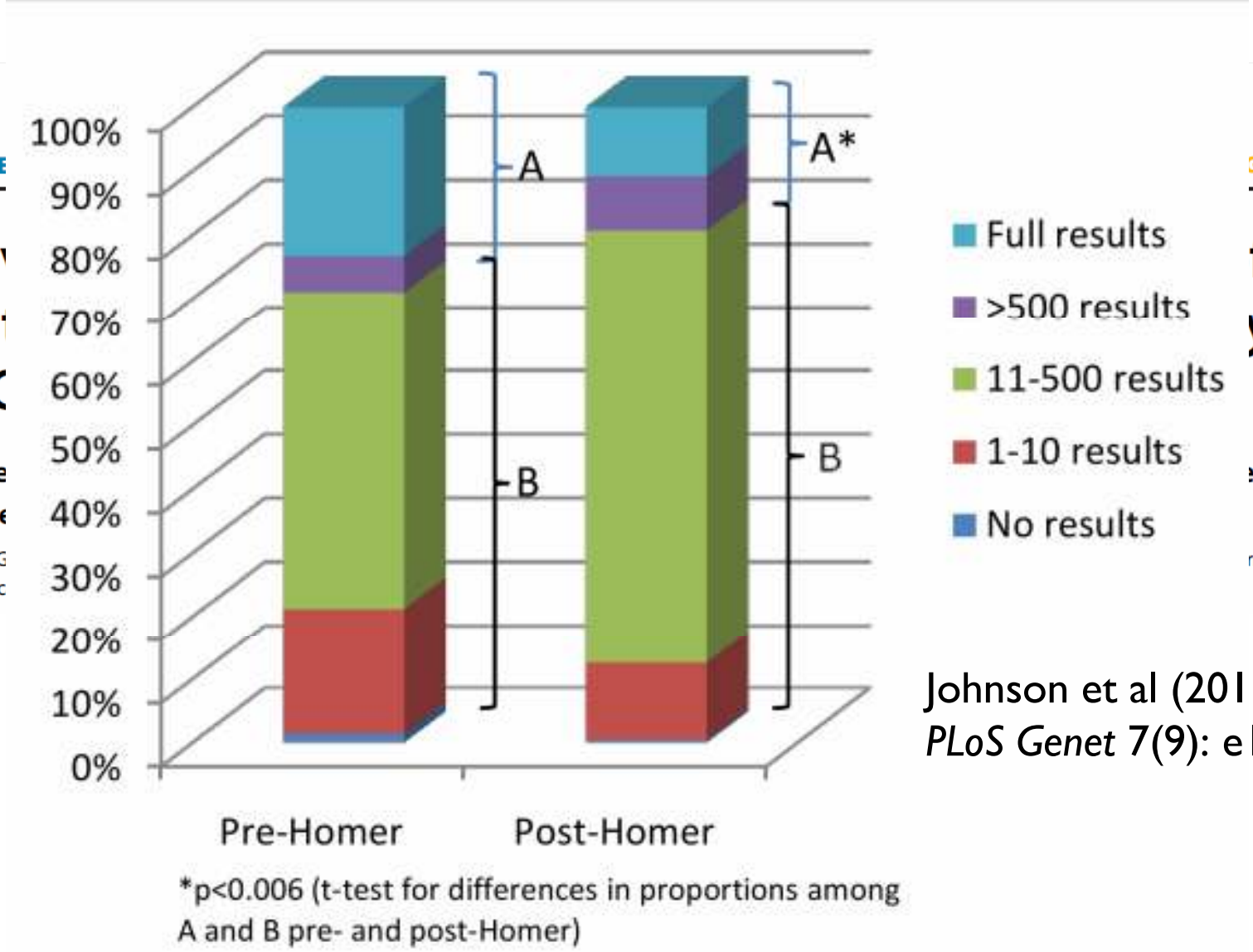
"We are committed to make these data public to make certain that any institution or researcher around the world can access and work with our datasets to better understand common disease"

Jun Wang, Ph.D., associate director of the Beijing Genomics Institute in Shenzhen, China, 1000 Genomes Project steering committee.

"Free and open access to genome data has had a profoundly positive effect on progress."

Francis Collins, *Nature, April 2010*

Yet other approaches to genomics have run into the tension between openness and privacy.



Johnson et al (2011)
PLoS Genet 7(9): e1002269.

In sequencing, these tensions can be found in the case of Hendrikje van Andel-Schipper

BBC Mobile | News | Sport | Weather | iPlayer | TV

NEWS HEALTH

DNA sequenced of woman who lived to 115

By Helen Briggs
Health editor, BBC News website

The entire DNA sequence of a woman who lived to 115 has been pieced together by

The woman, whose identity is being kept secret, and is known only as W115, is the oldest person to have her genes mapped

The woman had some rare genetic changes

The study, reported at a scientific conference in Canada, suggests she had genes that protected against dementia.

Further work could give clues to why some people are born with genes for a long life, says a UK scientist.

It is more than 10 years since the first draft of the human genetic code was revealed.

Since then, perhaps a few hundred individuals have had their genes mapped in full, as the technology to "read" DNA gets better and cheaper.

The woman, whose identity is being kept secret, and is known only as W115, is the oldest person to have her genes mapped.

She donated her body to medical science, allowing doctors to study her brain and other organs, as well as her entire genetic code.

Related Stories

Read/write your own genetic code

"Sequencing the genome of the world's oldest woman is an important starting point"

The secret identity of W115 is easy to discover given she has her own Wikipedia page

Hendrikje van Andel-Schipper

From Wikipedia, the free encyclopedia

Hendrikje "Henny" van Andel-Schipper (née **Schipper**) (Dutch pronunciation: [hɛndrikjə van andəl ʃxipər][stress?]) (29 June 1890 – 30 August 2005) was the oldest person ever in the history of the Netherlands (breaking the previous record of Catharina van Dam on 26 September 2003), and from 29 May 2004 was thought to be the oldest recognized person in the world until her death (although her case was later superseded by the one of María Capovilla of Ecuador). She became the oldest living person in the Netherlands on 16 February 2001, at the age of 110 years 232 days. If not counting disputed cases, she remains one of the 20 oldest people ever.

Contents [hide]

Hendrikje van Andel-Schipper

Henny van Andel on her 113th birthday

Born	29 June 1890 Smilde, Drenthe Netherlands
Died	30 August 2005 (aged 115 years 62 days) Hoogeveen, Drenthe Netherlands
Known for	Supercentenarian
Spouse	Dick van Andel (lived: 1889–1959, married: 1939–1959)

Biography [edit]

Van Andel-Schipper was born as Hendrikje Schipper in Smilde, a small village in Drenthe. She was born prematurely and there were doubts that she would survive. However, thanks to the continuous care of her grandmother during her first four weeks, she recovered. At the age of five on her first day of school, she was sick again and was removed from the school on advice of a local doctor. Her father, who was head of the local school, taught her to read and write.

She had a love of theatre from a young age, but after her mother objected she decided not to pursue a career in acting and became a needlework teacher instead.

The supercentenarian-to-be lived with her parents until she was 47 years old. At the age of 46, she met her future husband Dick van Andel, who worked in Amsterdam. She left her parents' home at the age of 47 and married van Andel, a tax inspector, at the age of 49 in 1939, taking the hyphenated name of van Andel-Schipper.

During the Second World War, she and her husband moved to Hoogeveen, where she had to sell jewellery to help pay for food during the German occupation. Her husband died from cancer in 1959.

Van Andel-Schipper underwent a mastectomy in 1990 after being diagnosed with breast cancer at the age of 100.[1] She continued to live on her own before moving into a retirement home at the age of 105.

She became the oldest recognized woman in Europe on the death of Maria Teresa Fumarola Ligorio in May 2003, and the oldest recognized person in Europe on the death of Joan Riudavets in March 2004. The death of Charlotte Benkner in early May 2004 left her second-oldest recognized in the world behind Ramona Trinidad Iglesias-Jordan, whose death later that month left her apparently the world's oldest at 113 years 335 days. It was the first time since the 1980s that no one had been recognized as over 114. However, during the next year, "Aunt Hennie" outlived several prior "world's oldest" titleholders, including Mitoyo Kawate, Ramona Trinidad Iglesias-Jordan, Eva Morris, Marie Brémont and Maud Farris-Luse.

One solution is to rely on public participation and take ‘open data’ to its natural conclusion.

Personal Genome Project

Home | Project Overview | Participation Overview | PGP Community | DONATE

Volunteers from the general public working together with researchers to advance personal genomics.

We believe individuals from the general public have a vital role to play in making personal genomes useful. We are recruiting volunteers who are willing to share their genome sequence and many types of personal information with the research community and the general public, so that together we will be better able to advance our understanding of genetic and environmental contributions to human traits. Learn more about how to participate in the Personal Genome Project.

Project Overview. The PGP hopes to make personal genome sequencing more affordable, accessible, and useful for humankind. Learn more about our mission.

Want to participate? We aim to enroll 100,000 informed participants from the general public. Learn more about participation in the PGP and how you can get involved.

Meet our volunteers. Participants may volunteer to publicly share their DNA sequence and other personal information for research and education. Meet the "PGP-1K".

Participant Login

Login Now

Project News

Subscribe to our newsletter.

Oct 5, 2011: PGP-HMS prepares for national blood collection campaign, adds hundreds of walk-in clinics to network. See list.

Sep 10, 2011: KPGP publishes 32 genomes of Korean participants. More.

The PGP posts medical data…

Name	Start Date	End Date	Dosage	Frequency
Cabergoline	2000-01-01		0.5 mg Tablet	Take 1, every other day

Personal Genome Project

Log in

Public Profile -- hu720B20

Public profile url: https://my.personalgenomes.org/profile/hu720B20

Google Health

Demographic Information

Date of Birth	1954-01-19 (57 years old)
Gender	Male
Weight	210lbs (95kg)
Height	
Blood Type	A+
Race	White

Conditions

Name	Start Date	End Date
Arthritis, left hip	2010-01-01	

As well as sequence data and its interpretation

About Genomes Editing guide Recent changes Contributors Download

Variant report for huE80E3D (PGP4: Misha Angrist) CGI var file, build 36

- Name: huE80E3D (PGP4: Misha Angrist) CGI var file, build 36
- This report: evidence.personalgenomes.org/genomes?fe9f72be9699820adc9af9e001500e02189adc84
- public profile: my.personalgenomes.org/profile/huE80E3D
- Download: source data (373 MB), dbSNP and nsSNP report (126 MB)
- Show debugging info

Genome report | Insufficiently evaluated variants | Coverage | Metadata

Show rare (f<10%) pathogenic variants | Show all

Show All entries Search:

Variant	Clinical Importance	Impact	Allele freq	Summary
CPT2-S113L	High	Well-established pathogenic Recessive, Carrier (Heterozygous)	0.78%	This is the most common variant associated with late-onset carnitine palmitoyltransferase deficiency, which is classically viewed as recessive. Many patients are heterozygous for this, but are presumably compound heterozygous.
TREM2-R47H	High	Uncertain pathogenic Recessive, Carrier (Heterozygous)	0.78%	Unreported, predicted to be damaging. Other recessive mutations in this gene cause polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy (a severe genetic disorder, usually lethal by age 50).
CC2D2A-G776R	High	Uncertain pathogenic Recessive, Carrier (Heterozygous)	0.78%	Unreported, predicted to be damaging. Other recessive mutations in this gene cause Joubert Syndrome and Meckel Syndrome.

Gene search

"GENE" or "GENE A123C":

search

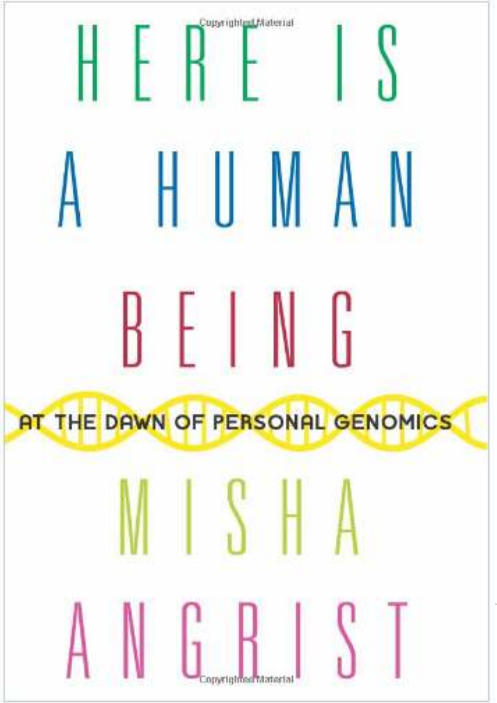
Log in

Google login

Yahoo login

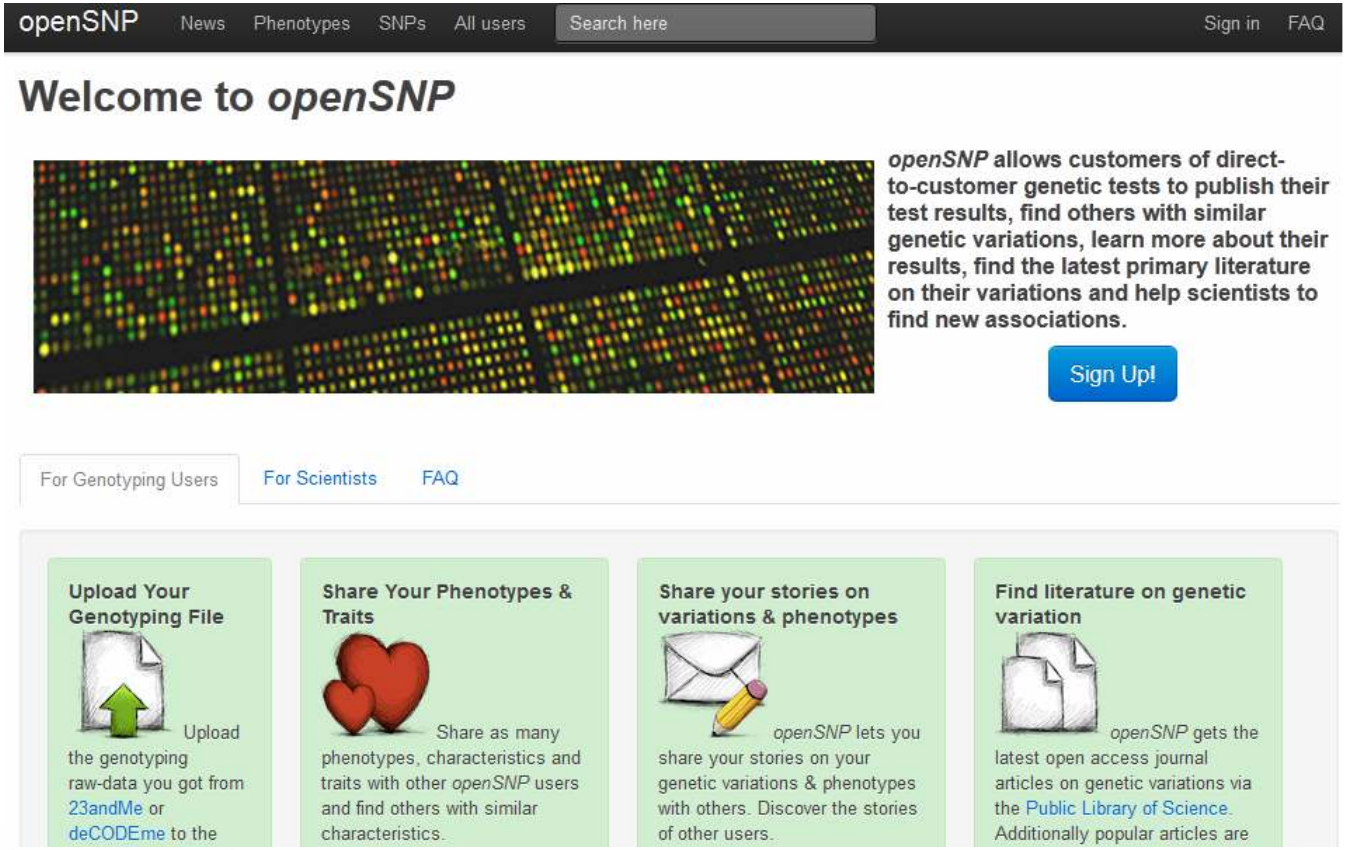
OpenID URL:

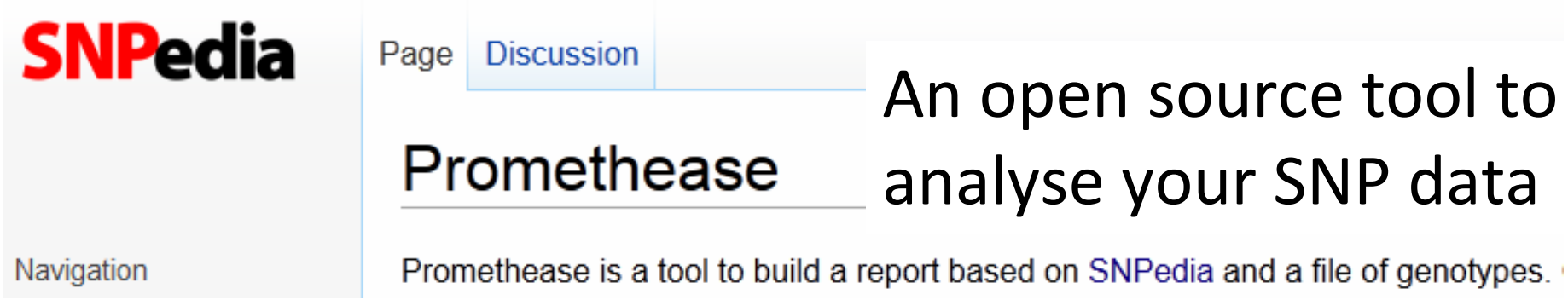
Log in



openSNP: share your phenotype too?

- Launched October 2011
- By 3 Masters students in Frankfurt





An open source tool to analyse your SNP data

Some areas for discussion?

- Wider awareness? Getting attention?
- Understanding the full potential of shared genome (and phenotype) data
- Implications for:
  - University data infrastructure providers
  - PIs
  - Students
  - You
- Advocacy & guidance? What is needed?
  Who should provide it? To whom?
  - Consent guidelines, legal concerns, ethical issues, data policy, good practice….