

# 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

7<sup>th</sup> IDCC Symposium, Bristol, December 2011

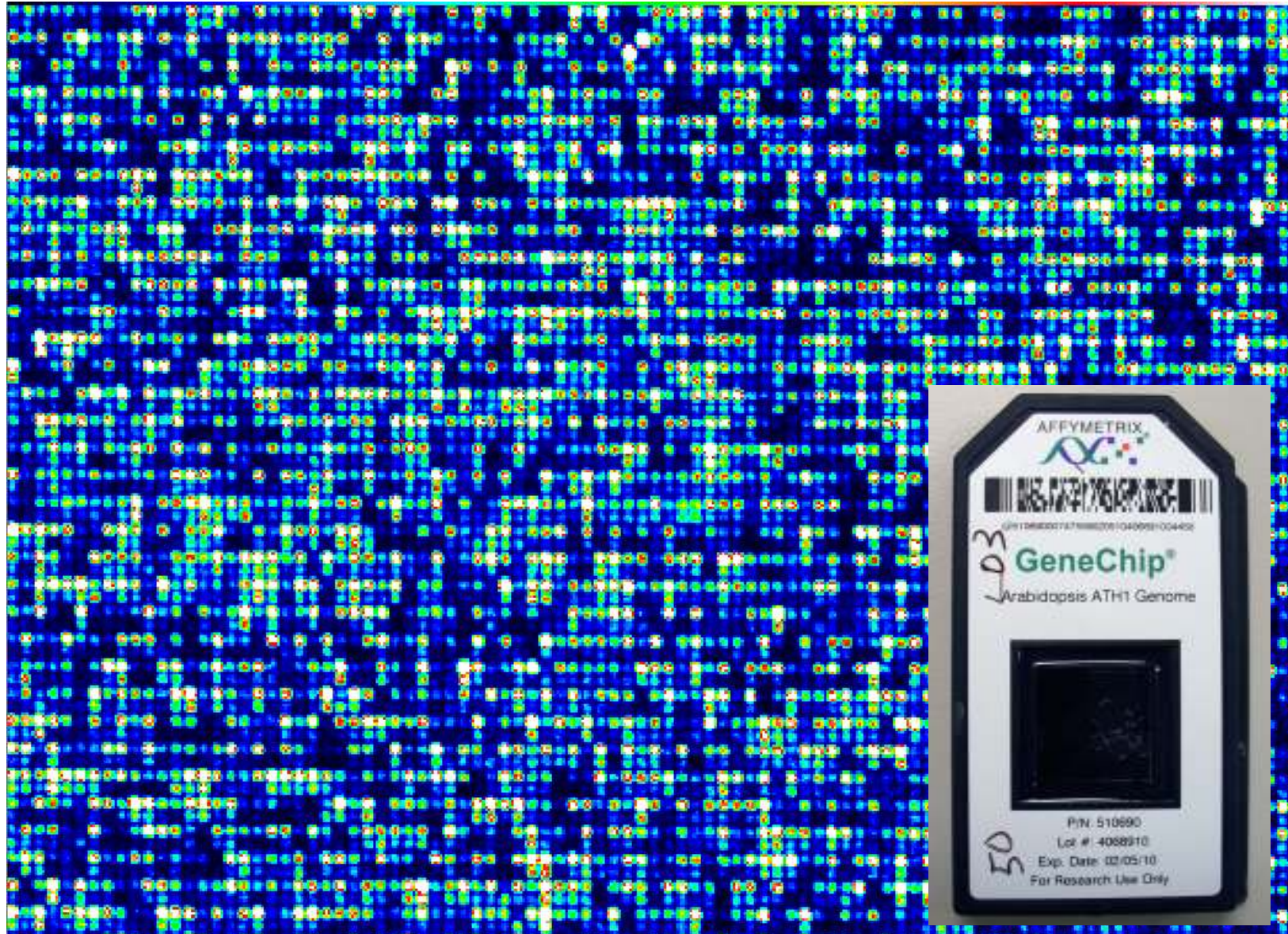


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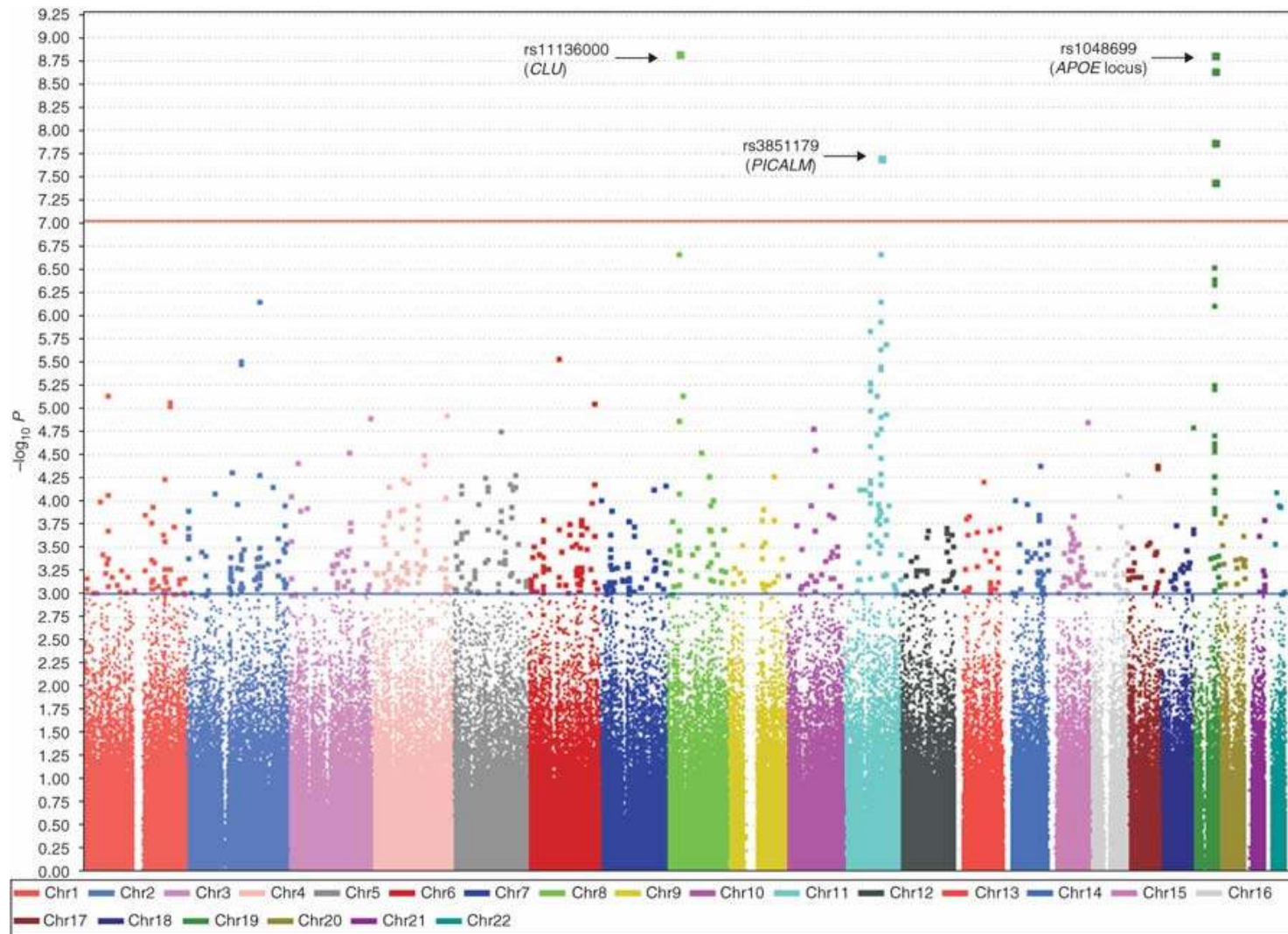
UKOLN is supported by: **JISC**  
Microsoft  
**Research**



**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.

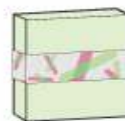
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\*Requires a 1-year commitment to the [Personal Genome Service](#) at \$9/mo. [Order for \\$399](#) without commitment.

1 Get Your Kit



2 Provide Saliva



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

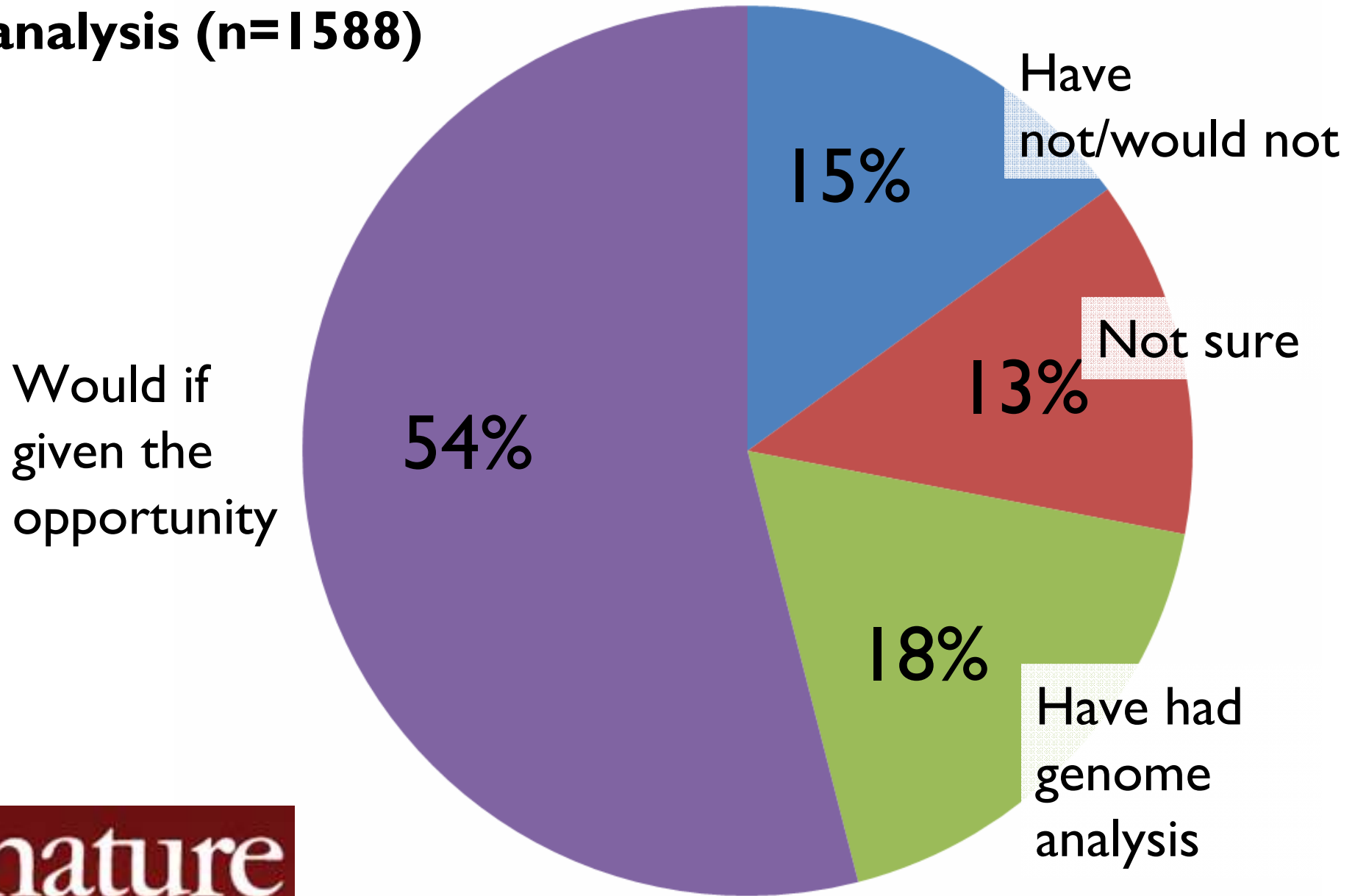
Overview

Discover Health & Ancestry

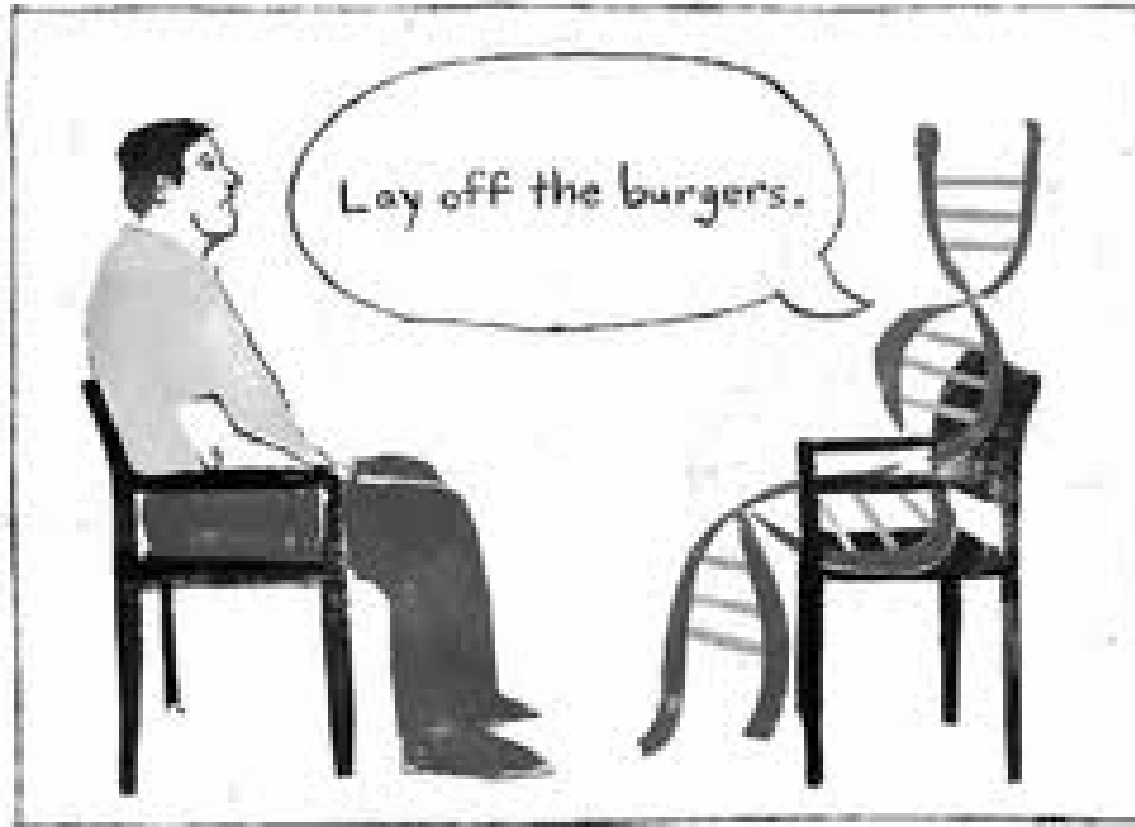
Keep Your Doctor Informed

Participate In Research

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



**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.**

The image shows a screenshot of the deCODE ME website's news section. The page has a dark blue header with the deCODE ME logo on the left and a 'Login to myCODE' button on the right. Below the header, there is a navigation menu with links for Home, What is deCODEme?, About deCODE, Signup, Family registration, Kit registration, and Login to myCODE. The main content area features a news article titled 'A Smoking Gene: deCODE Scientists Identify a Genetic Variant that Confers Nicotine Dependence'. The article text discusses the discovery of a genetic variant linked to nicotine dependence and its association with lung cancer and peripheral arterial disease (PAD). A quote from Kari Stefansson, deCODE CEO, is included, highlighting the importance of the discovery. On the right side of the article, there is a 'Media Contact' section for Berglind Þorvaldóttir, Director of communications, with the email address berglind@decode.is. The article is dated April 2, 2006.

news  
home

deCODE ME

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

Home  
What is deCODEme?  
About deCODE  
Signup  
Family registration  
Kit registration  
Login to myCODE

April 2, 2006

**A Smoking Gene: deCODE Scientists Identify a Genetic Variant that Confers Nicotine Dependence**

Scientists from deCODE genetics have identified a clear link between one genetic variant and susceptibility to nicotine dependence, and will publish their results in the April 3 issue of Nature. Moreover, in part because of its impact on smoking behavior, each copy of the risk variant of this SNP confers an approximately 30% increase in risk of lung cancer and a 20% increase in risk of peripheral arterial disease (PAD), a common and debilitating constriction of the arteries to the legs.

deCODE scientists came upon the genetic variant by closely examining the genetic makeup of more than 10,000 smokers. They then followed up with an analysis of 32,000 patients and controls from Iceland, Sweden, Norway, and the Netherlands. The gene is located on chromosome 15 and is associated with two common diseases strongly associated with smoking.

Kari Stefansson, deCODE CEO, expressed the importance of the discovery: "These findings provide an example of the power of human genetics for shedding light on the most complex health challenges. Not only have we made a convincing link between a single genetic variant and a behavioral disorder - greater smoking quantity and addiction to nicotine - but also demonstrated how this risk factor translates into risk of lung cancer and PAD."

Stefansson also pointed out that deCODE's genetic profile service, deCODEme, will test for the gene immediately.

Media Contact:  
Berglind Þorvaldóttir  
Director of communications  
[berglind@decode.is](mailto:berglind@decode.is)

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

## Los Angeles Times

### UC Berkeley adjusts freshman orientation's gene-testing program

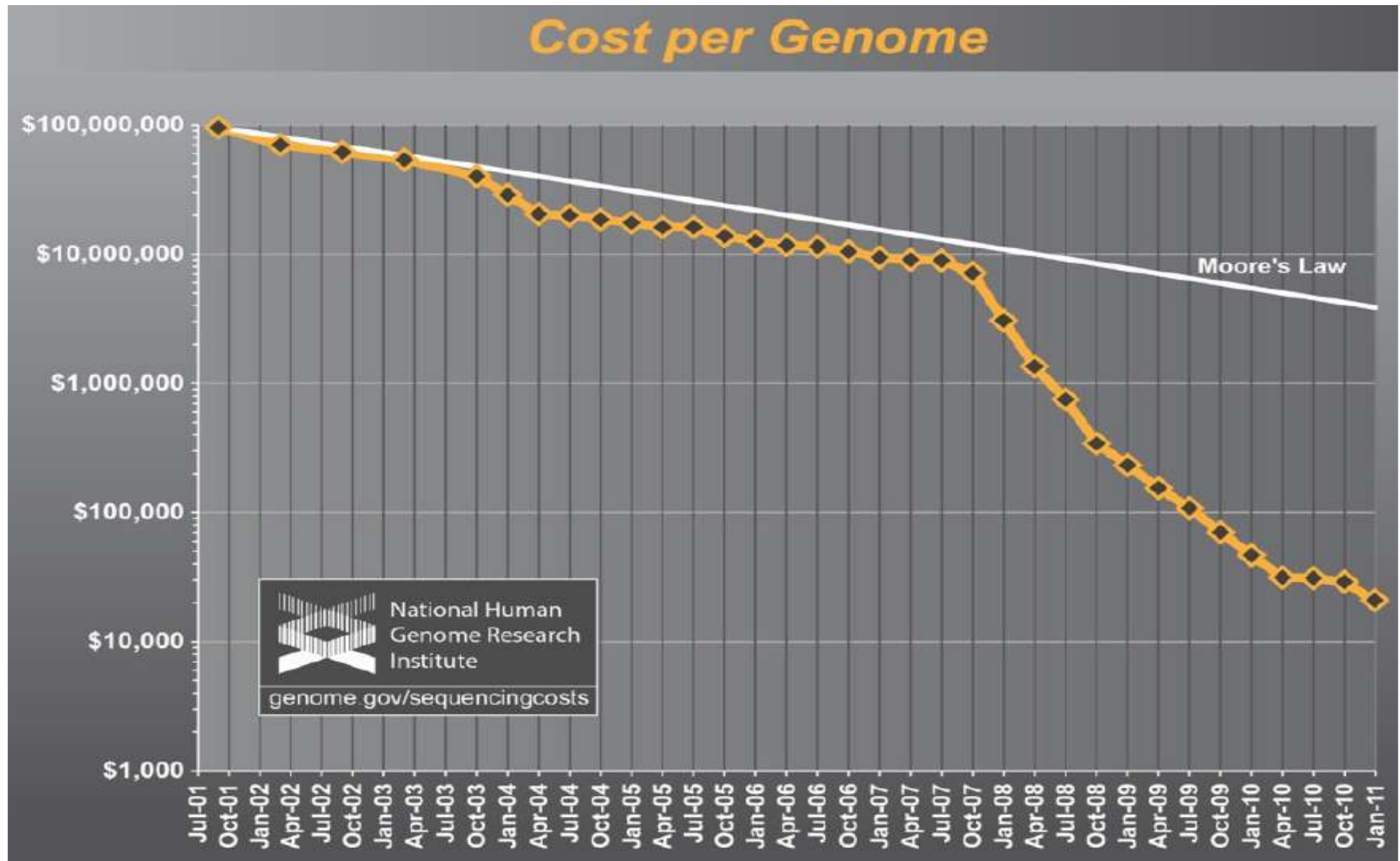
*To comply with regulations on how DNA samples should be handled, the university is restricting the project by presenting only the collective results of all the participants.*

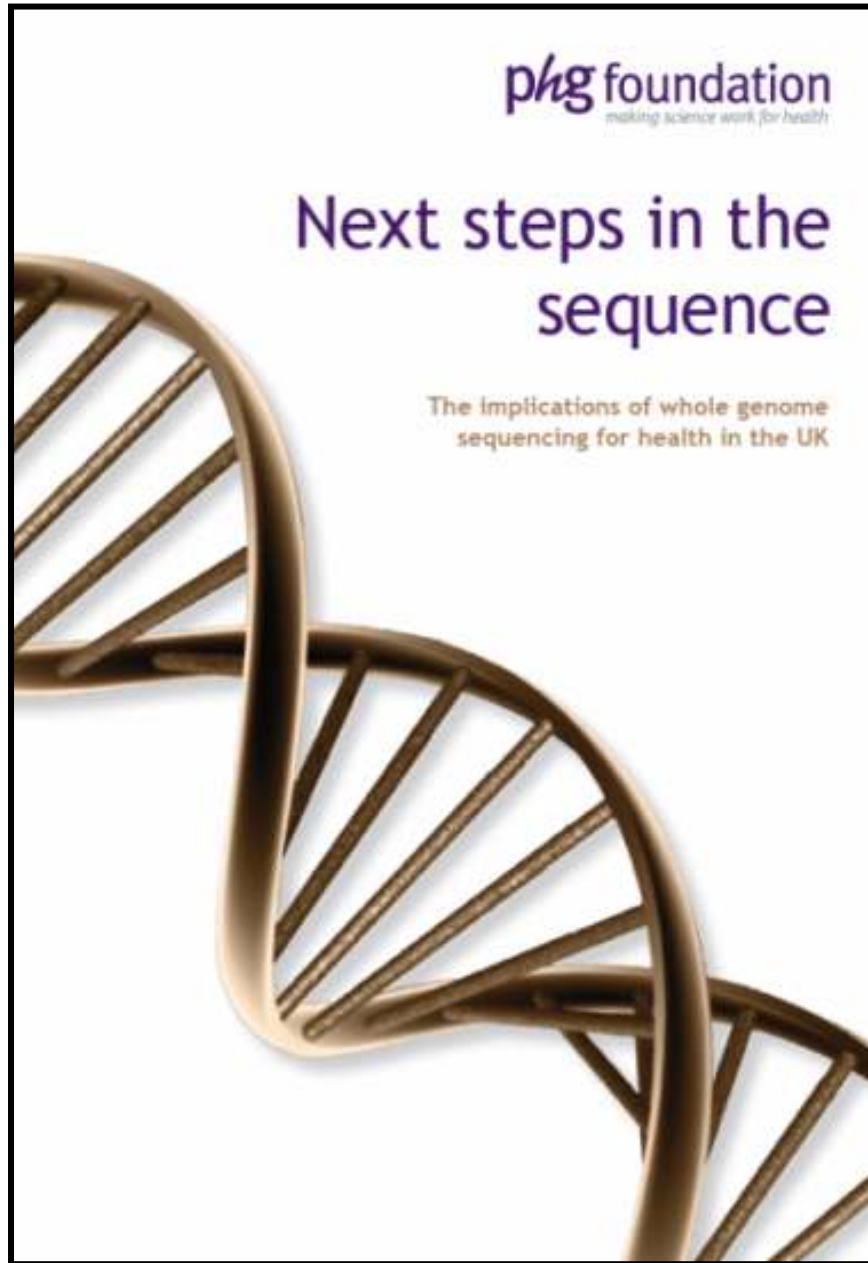
**August 13, 2010** | By Larry Gordon, Los Angeles Times

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 testing, full-genome sequencing is dropping in price and raising its own issues.





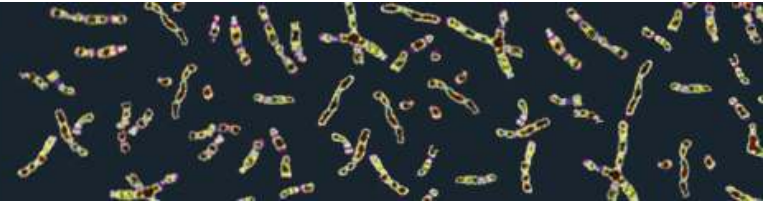
**Debate is promoted and structured by various organisations, research projects & reports. ...**

*<http://www.phgfoundation.org/reports/10364/>*

# Research in this area is underpinned by key ‘ideological’ belief in the value of open data

## 1000 Genomes

A Deep Catalog of Human Genetic Variation

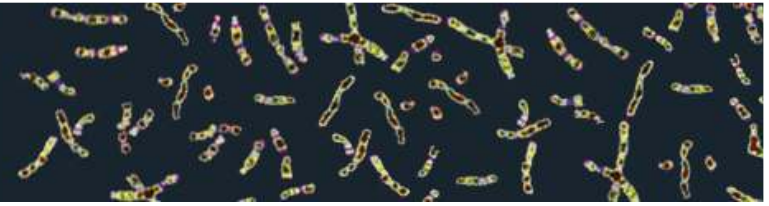


A critical new component of the Project is the selection of 2,500 DNA samples from 27 populations around the world. Each participant has provided explicit consent for full and public release of DNA samples and full sequence data....

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

# 1000 Genomes

A Deep Catalog of Human Genetic Variation

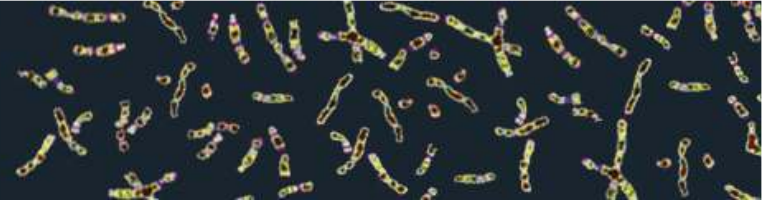


***“We are committed to make these data public to make certain that any institution or researcher around the world can access and work with our datasets to better understand common disease”***

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

## 1000 Genomes

A Deep Catalog of Human Genetic Variation



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

Francis Collins, *Nature*, April 2010

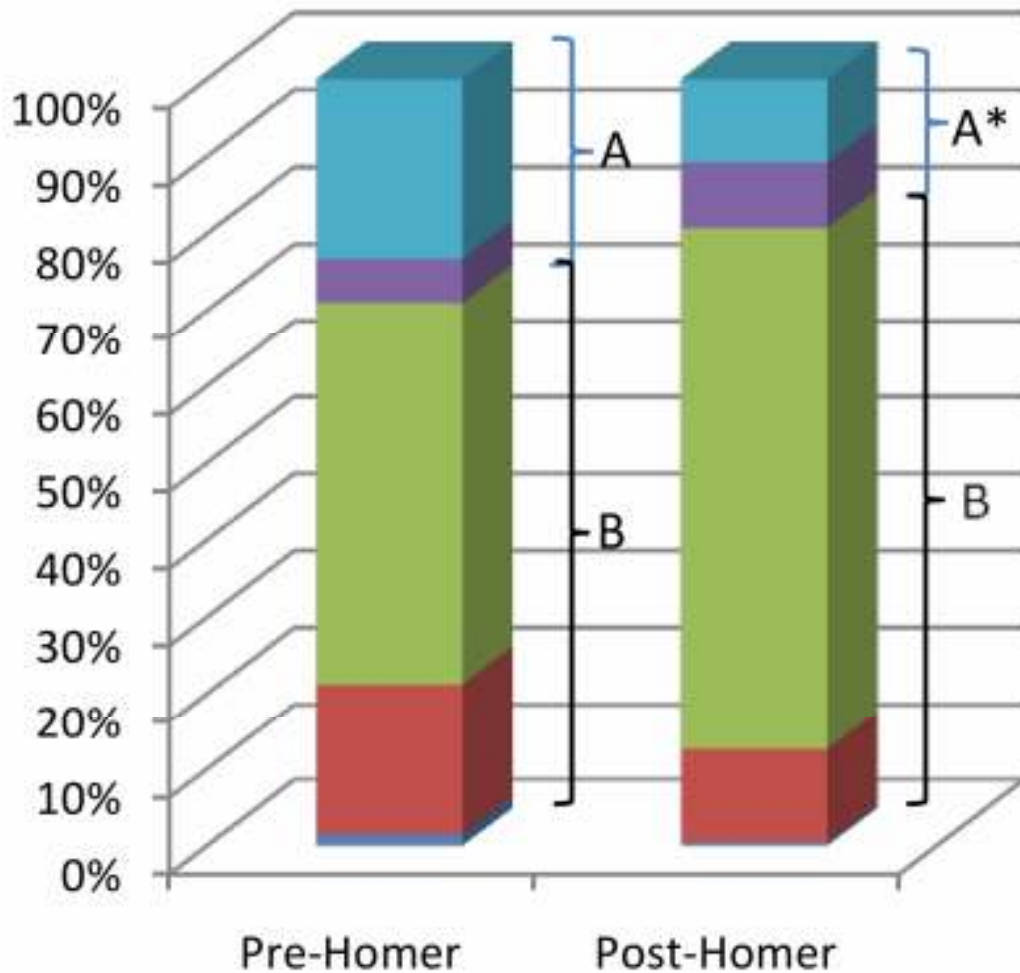
# Yet other approaches to genomics have run into the tension between openness and privacy.

OPEN ACCESS

Resolution  
DNA  
SNP C

Nils Home  
John V. Pe

1 Translational G  
States of Americ



\* $p < 0.006$  (t-test for differences in proportions among A and B pre- and post-Homer)

GENETICS

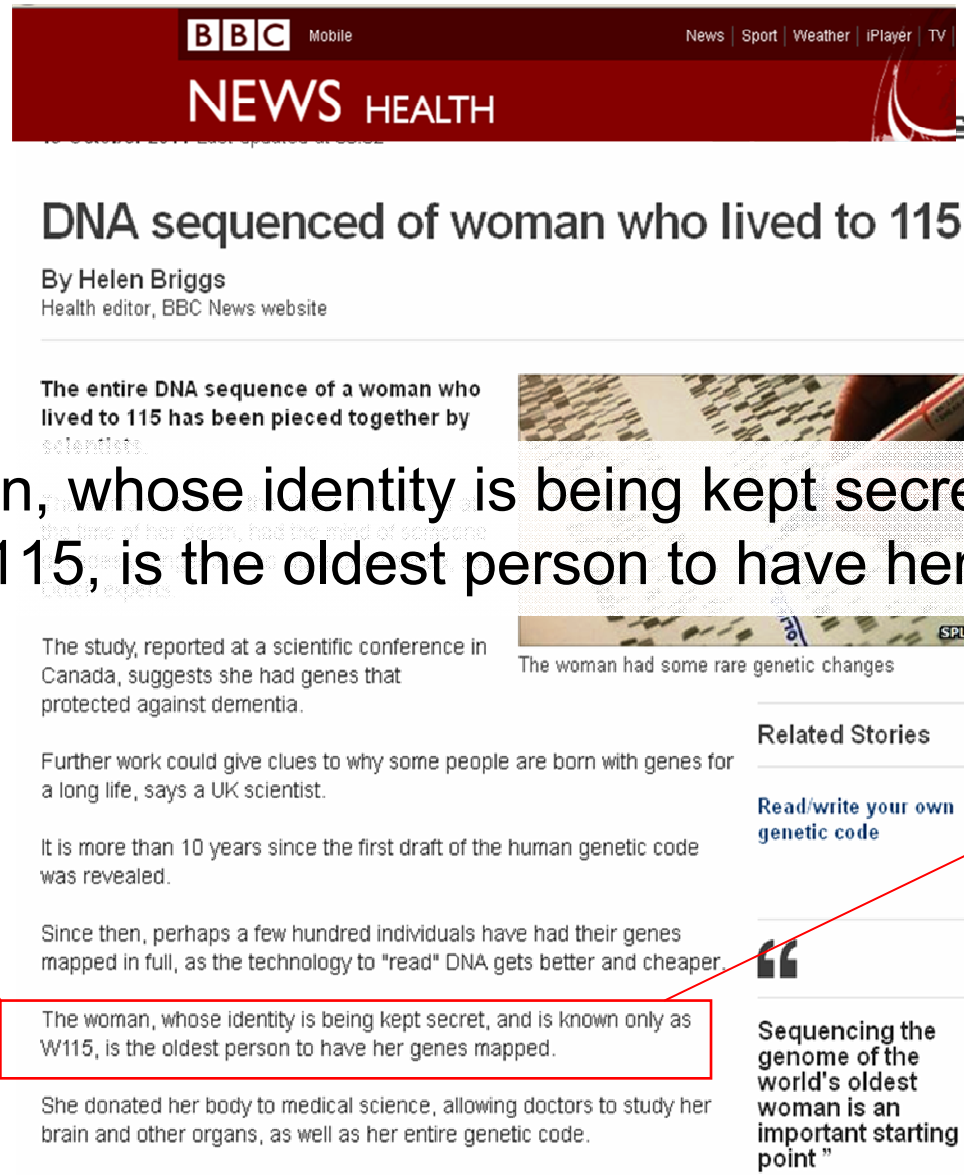
f  
y

- Full results
- >500 results
- 11-500 results
- 1-10 results
- No results

ehling<sup>1</sup>,  
nia, United

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

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



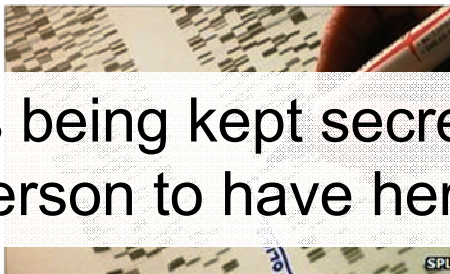
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## NEWS HEALTH

### 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 scientists.



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



# The secret identity of WII5 is easy to discover given she has her own Wikipedia page



WIKIPEDIA  
The Free Encyclopedia

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Contents  
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## Hendrikje van Andel-Schipper

From Wikipedia, the free encyclopedia

**Hendrikje "Henny" van Andel-Schipper** (née **Schipper**) (Dutch pronunciation: [hɛndrɪkʲə van andɛl ʃɪpər]<sup>[*stres-?*]</sup>) (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

- Biography
- Cause of death
- Post-mortem brain analysis
- Status as "world's oldest person"
- Wisdom
- Genome
- See also
- References
- External links

## 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 [Hoozeveen](#), 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.<sup>[*1*]</sup> 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](#).

Hendrikje van Andel-Schipper



Henny van Andel on her 113th birthday

<b>Born</b>	29 June 1890 <div><span><span></span></span> <a href="#">Smilde</a>, <a href="#">Drenthe</a>, <a href="#">Netherlands</a></div>
<b>Died</b>	30 August 2005 (aged 115 years 62 days) <div><span><span></span></span> <a href="#">Hoozeveen</a>, <a href="#">Drenthe</a>, <a href="#">Netherlands</a></div>
<b>Known for</b>	<a href="#">Supercentenarian</a>
<b>Spouse</b>	<div><span>Dick van Andel</span> (lived: 1889–1959, married: 1939–1959)</div>

# 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

<b>Date of Birth</b>	1954-01-19 (57 years old)
<b>Gender</b>	Male
<b>Weight</b>	210lbs (95kg)
<b>Height</b>	
<b>Blood Type</b>	A+
<b>Race</b>	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](http://evidence.personalgenomes.org/genomes?fe9f72be9699820adc9af9e001500e02189adc84)
- public profile: [my.personalgenomes.org/profile/huE80E3D](http://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
<a href="#">CPT2-S113L</a>	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.
<a href="#">TREM2-R47H</a>	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).
<a href="#">CC2D2A-G776R</a>	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

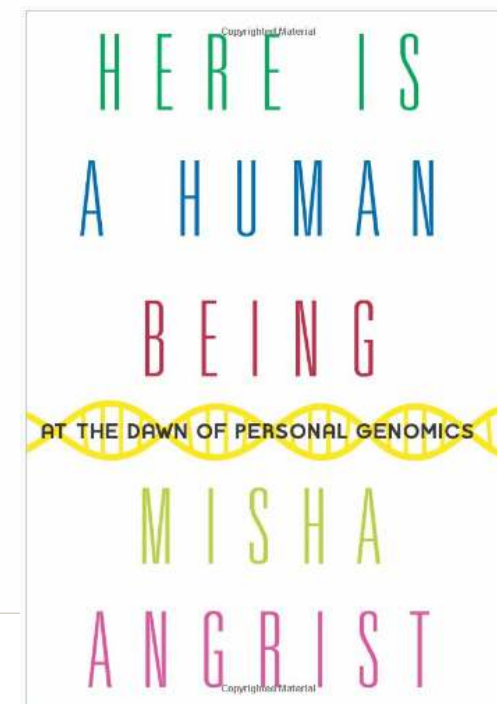
### Log in

Google login

Yahoo login

OpenID URL:

Log in



# openSNP: share your phenotype too?

- Launched October 2011
- By 3 Masters students in Frankfurt

The screenshot shows the openSNP website homepage. At the top, there is a navigation bar with links for 'openSNP', 'News', 'Phenotypes', 'SNPs', and 'All users', along with a search bar and 'Sign in' and 'FAQ' links. Below the navigation bar, the main heading reads 'Welcome to openSNP'. A large image of a DNA microarray is displayed, with a text box to its right stating: 'openSNP allows customers of direct-to-customer genetic tests to publish their test results, find others with similar genetic variations, learn more about their results, find the latest primary literature on their variations and help scientists to find new associations.' A blue 'Sign Up!' button is positioned below this text. Below the main heading, there are three tabs: 'For Genotyping Users', 'For Scientists', and 'FAQ'. The main content area features four green boxes with icons and text: 1. 'Upload Your Genotyping File' with a document icon and an upward arrow, describing the upload of raw data from 23andMe or deCODEme. 2. 'Share Your Phenotypes & Traits' with a red heart icon, encouraging users to share characteristics and find others with similar traits. 3. 'Share your stories on variations & phenotypes' with an envelope icon, allowing users to share their genetic stories. 4. 'Find literature on genetic variation' with a document icon, linking to the Public Library of Science for open access journal articles.

**SNPedia**

Page **Discussion**

**Promethease**

An open source tool to analyse your SNP data

Navigation

Promethease is a tool to build a report based on [SNPedia](#) and a file of genotypes.

# 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....