Genomic Testing and Privacy Issues

Emiliano De Cristofaro
University College London
https://emilianodc.com

Who Am I?

- Reader in Security & Privacy Technology, UCL Computer Science (2013–)
- Head of Information Security Research Group (2018–)
- Faculty Fellow at the Alan Turing institute (2018–)
- Research Scientist, Xerox PARC (2011–2013)
- PhD in Computer Science, University of California (2011)

- Cybersecurity
- Privacy Enhancing Technologies
- Privacy and Ethical Issues in AI
- Data-Driven measurements of “interesting” issues on the Web

Privacy in Computer Science

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Mostly defined as a set of **information security** properties…

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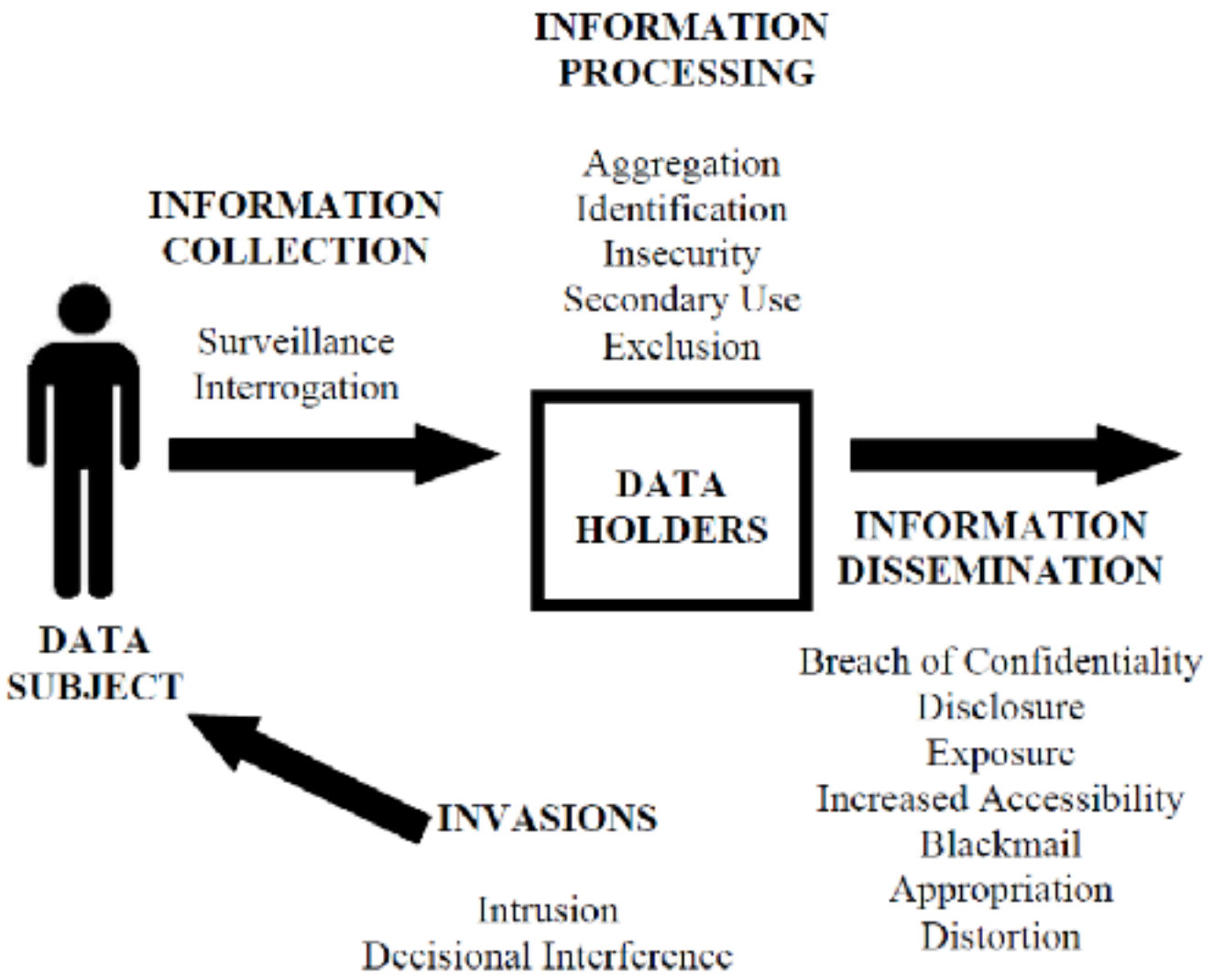
Control: giving control to the individual about the use of their personal information

Self-actualization: allowing the individual to use their information environment to further their own aims

Taxonomy of Privacy Harms



Solove, Daniel J. "A taxonomy of privacy." University of Pennsylvania Law Review (2006): 477-564.

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Despite promising not to sell its members' personal information to others, a company does so anyway

Ex

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Reporters dece
record the pers

New X-ray devi
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The governmer
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A company ma

Despite promis
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GCHQ data collection violated human rights, Strasbourg court rules

Spies breached right to privacy in programme revealed by Edward Snowden, judges say

The GCHQ building in Cheltenham. Photograph: GCHQ/PA

Two families of privacy technologies

George Danezis. Introduction to Privacy Technologies. 2018

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E.g.: Tunnel encryption (SSL/TLS)

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George Danezis. Introduction to Privacy Technologies. 2018

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k-out-of-n honest third parties

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“Keeping honest services safe from insiders / employees”

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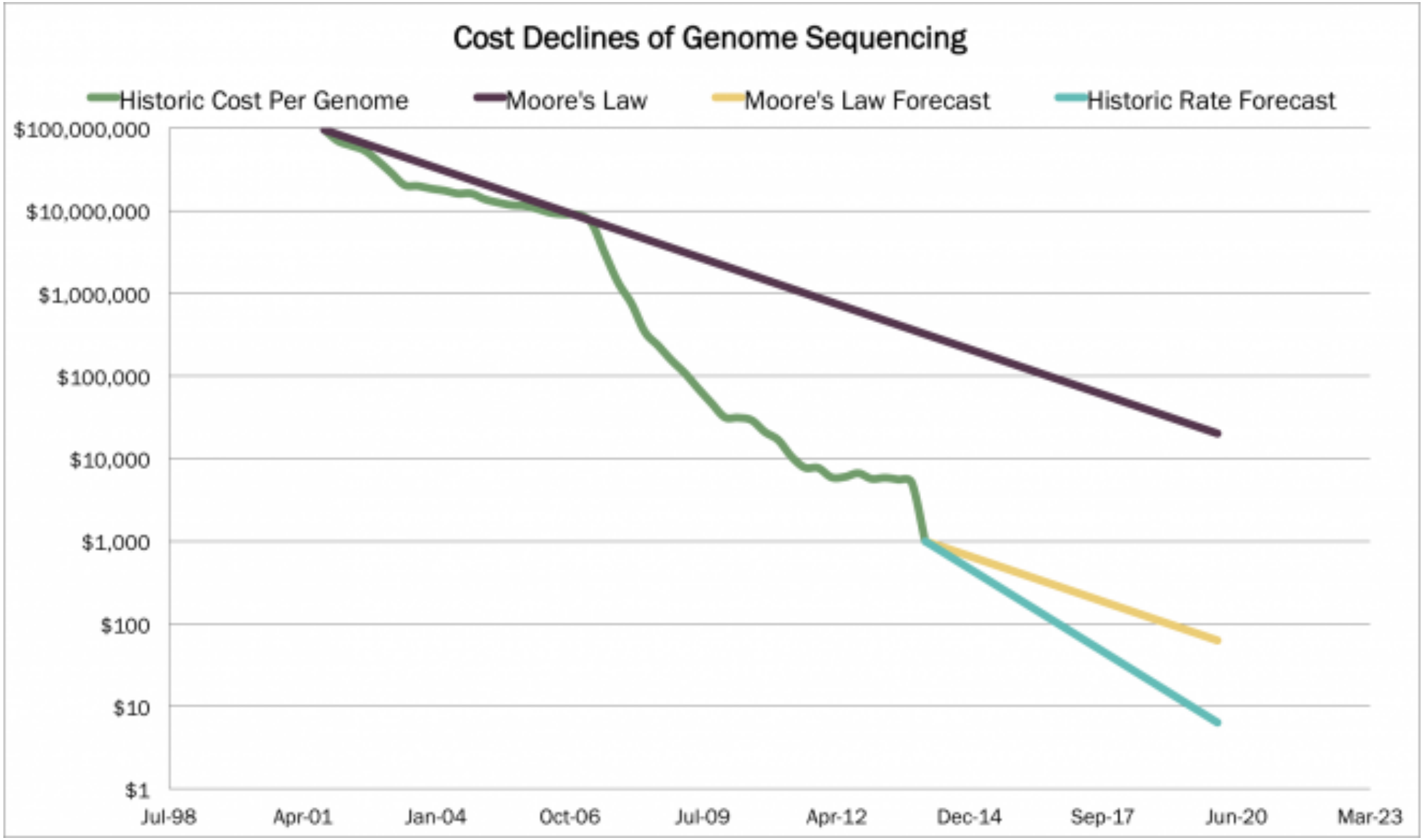
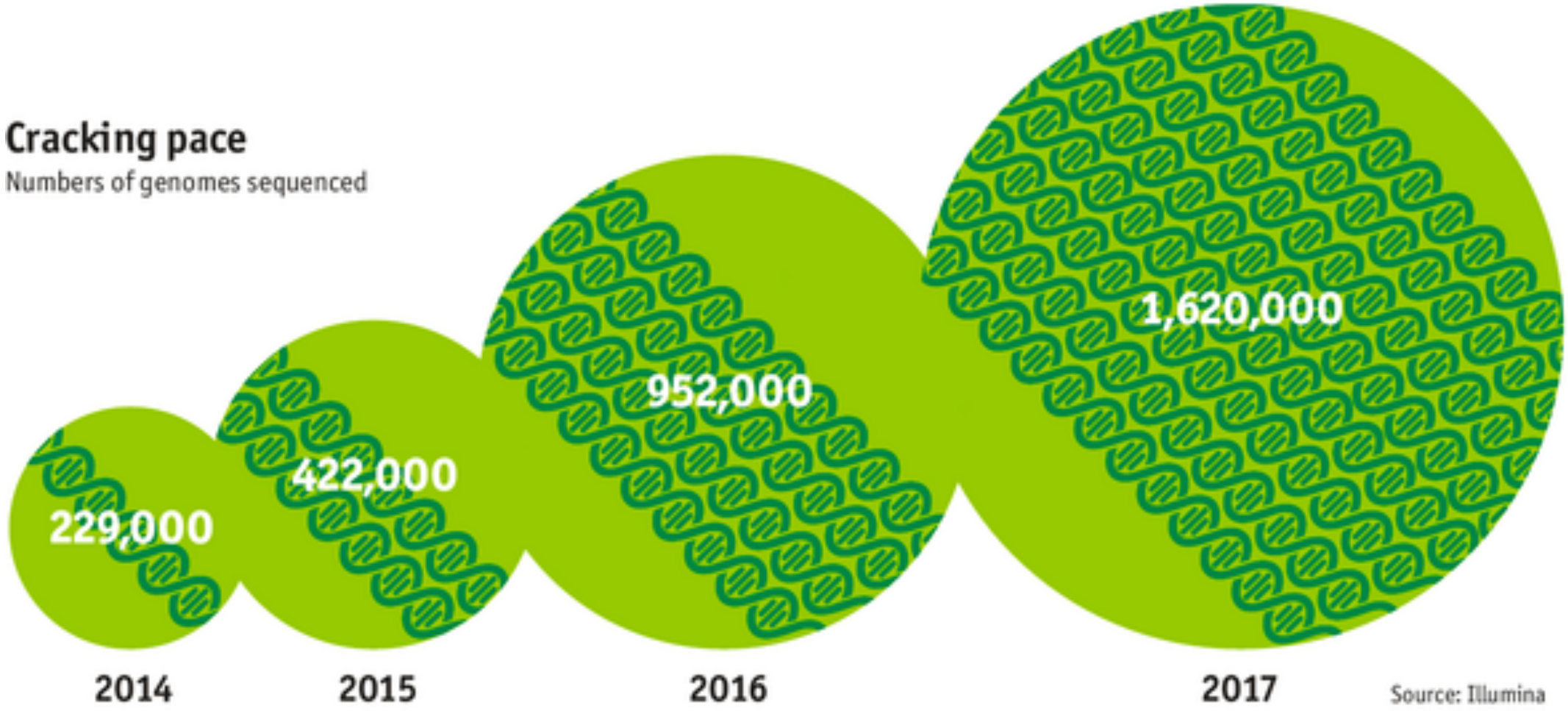
E.g., Tor

May relay on service integrity if auditing is possible

A Case Study: Genomic Testing



From: James Bannon, ARK



From: The Economist

9

How to read the genome?

Genotyping

Testing for genetic differences using a set of markers

Sequencing

Determining the full nucleotide order of an organism's genome

1/05/2011 @ 4:57PM | 30,075 Views

The First Child Saved By DNA Sequencing

+ Comment Now + Follow Comments

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Comprehensive whole genome sequence analyses yields novel genetic and structural insights for Intellectual Disability

Farah R. Zahir ✉, Jill C. Mwenifumbo, Hye-Jung E. Chun, Emilia L. Lim, Clara D. M. Van Karnebeek, Madeline Couse, Karen L. Mungall, Leora Lee, Nancy Makela, Linlea Armstrong, Cornelius F. Boerkoel, Sylvie L. Langlois, Barbara M. McGillivray, Steven J. M. Jones, Jan M. Friedman † and Marco A. Marra †

BMC Genomics 2017 18:403

https://doi.org/10.1186/s12864-017-3671-0

Received: 4 November 2016 | Accepted: 29 March 2017 | Published: 24 May 2017

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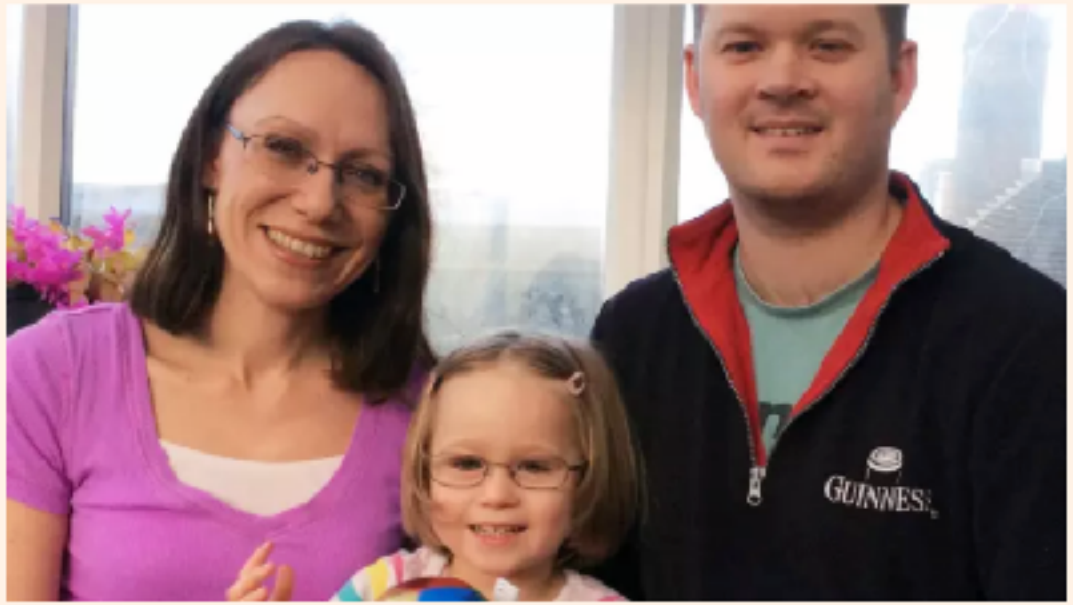
Faster and cheaper DNA sequencing brings new hope to patients



Jessica suffers from a rare condition that was diagnosed through DNA analysis

Clive Cookson FEBRUARY 28, 2017 1

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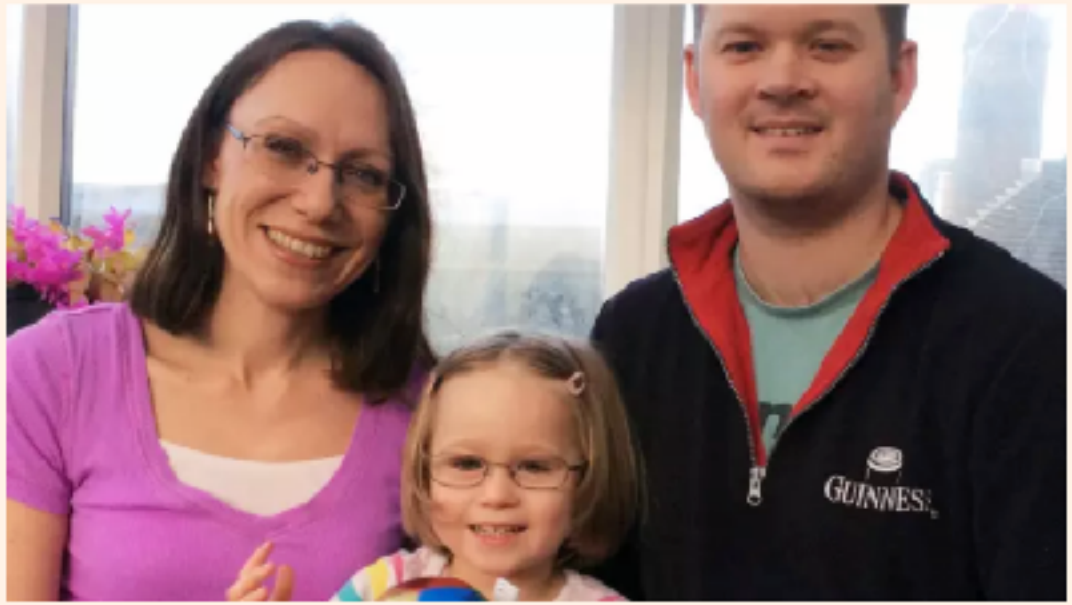

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Clive Cookson FEBRUARY 28, 2017 1

Three critically ill children helped by speedy genome sequencing

'Angelina Jolie effect' boosted genetic testing rates, study suggests

Actor's call for women to seek testing for breast and ovarian cancer mutations raised screening rates but may not have reached those most at risk

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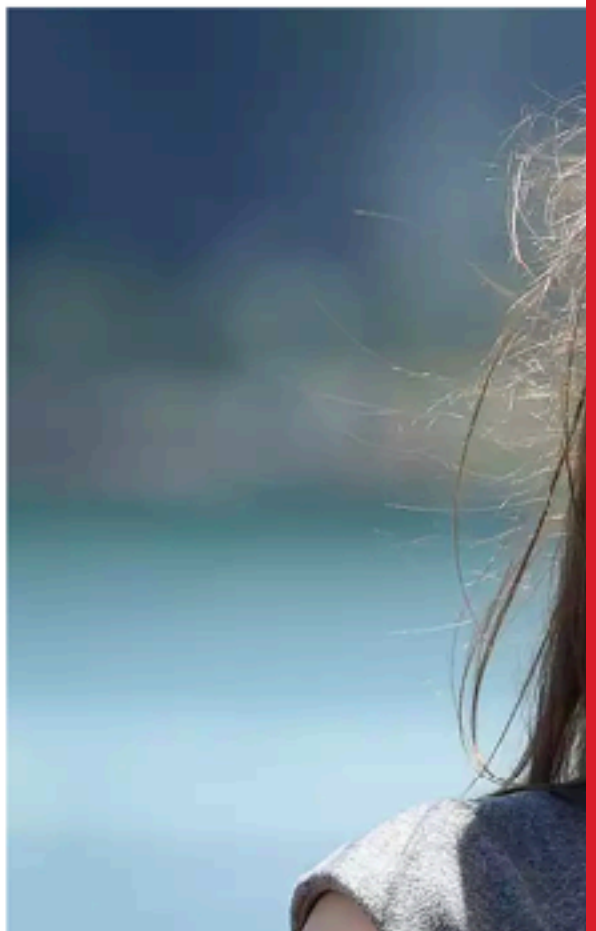

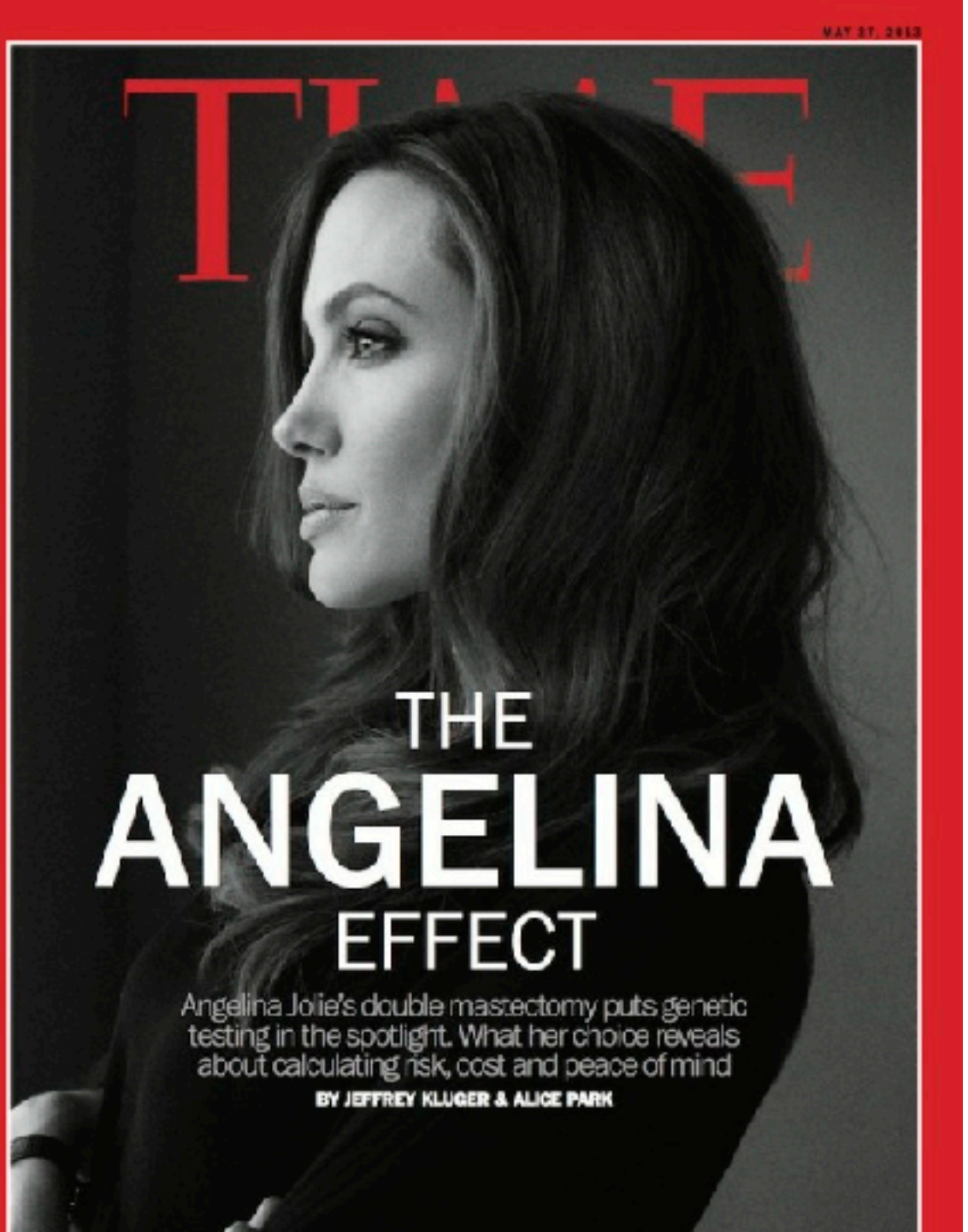

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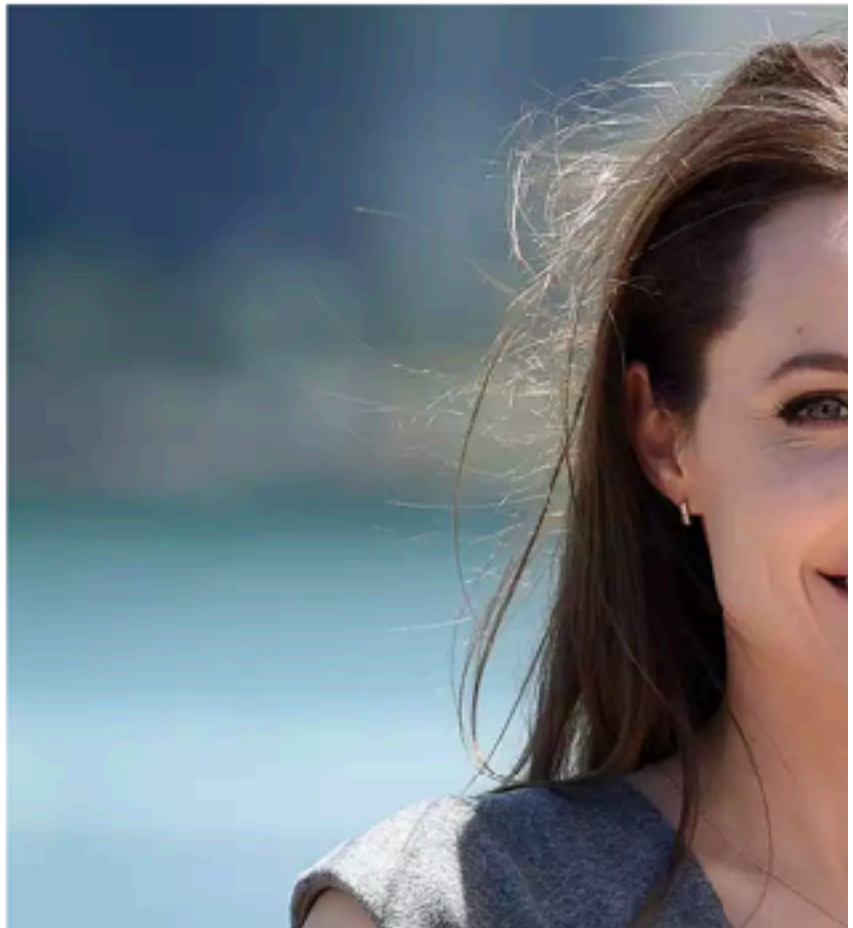

Angelina Jolie gene testing for all?

By James Gallagher
Health and science correspondent, BBC News

18 January 2018

Share

Testing all women for the "Angelina Jolie gene", even if not considered at risk, would prevent cancers, save lives and is cost effective, say doctors.

Genetic Risk Factors (11)

REPORT	RESULT
Alpha-1 Antitrypsin Deficiency	Variant Absent; Typical Risk
Alzheimer's Disease (APOE Variants)	ε4 Variant Absent
Early-Onset Primary Dystonia (DYT1-TOR1A-Related)	Variant Absent; Typical Risk
Factor XI Deficiency	Variant Absent; Typical Risk
Familial Hypercholesterolemia Type B (APOB-Related)	Variant Absent; Typical Risk

See all 11 genetic risk factors...

Inherited Conditions (43)

REPORT	RESULT
Beta Thalassemia	Variant Present
ARSACS	Variant Absent
Agenesis of the Corpus Callosum with Peripheral Neuropathy (ACCPN)	Variant Absent
Autosomal Recessive Polycystic Kidney Disease	Variant Absent
Bloom's Syndrome	Variant Absent

See all 43 carrier status...

Traits (41)

REPORT	RESULT
Alcohol Flush Reaction	Does Not Flush
Bitter Taste Perception	Can Taste
Blond Hair	28% Chance
Earwax Type	Wet
Eye Color	Likely Brown

See all 41 traits...

Drug Response (12)

REPORT	RESULT
Proton Pump Inhibitor (PPI) Metabolism (CYP2C19-related)	Rapid
Warfarin (Coumadin®) Sensitivity	Increased
Phenytoin Sensitivity (Epilepsy Drug)	Increased
Sulfonylurea Metabolism	Greatly reduced
Abacavir Hypersensitivity	Typical

See all 12 drug response...

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See

Genetic Ethnicity

Southern European	37%
West African	20%
British Isles	13%
Native South American	9%
Finnish/Volga-Ural	9%
Eastern European	6%
Uncertain	6%

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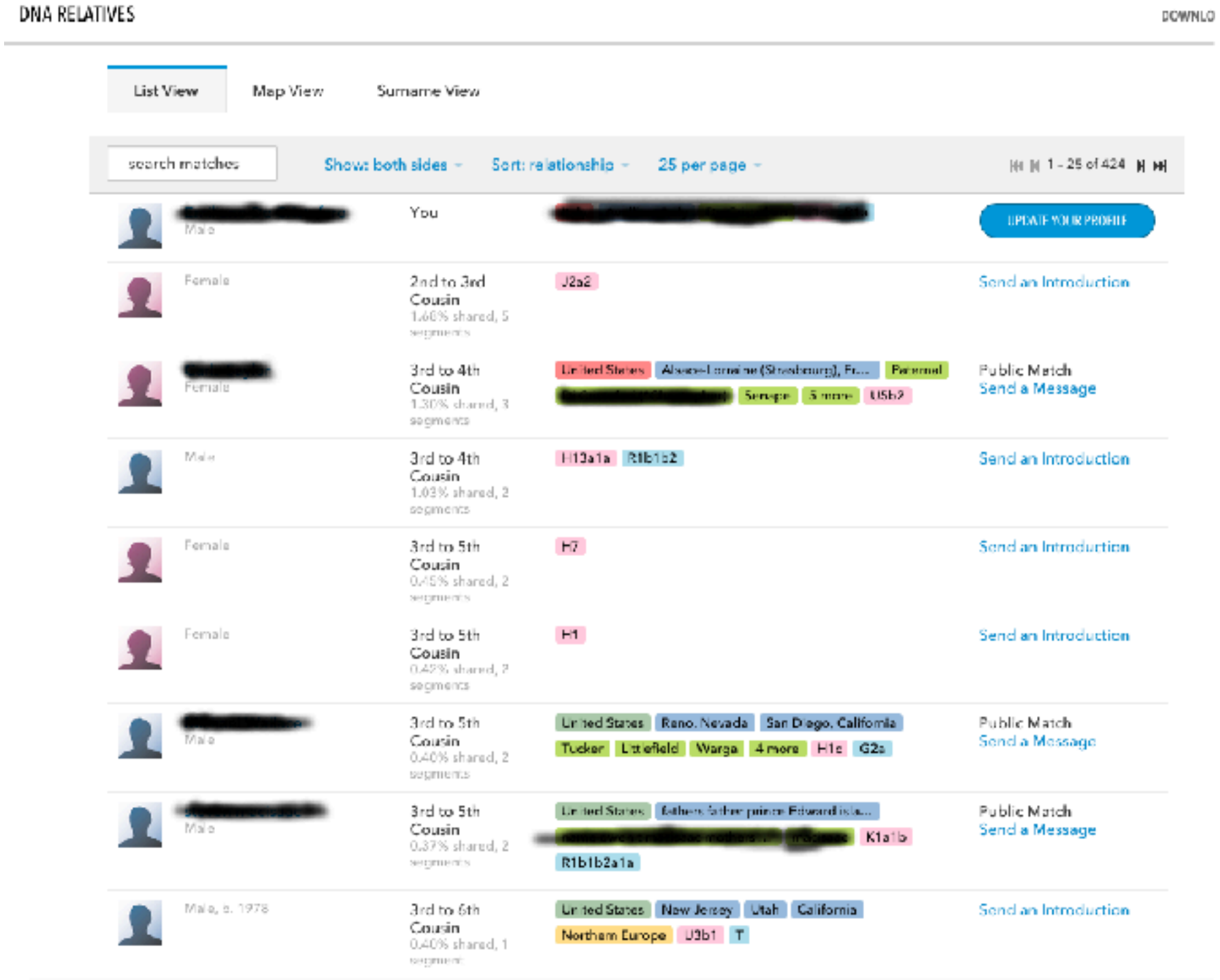
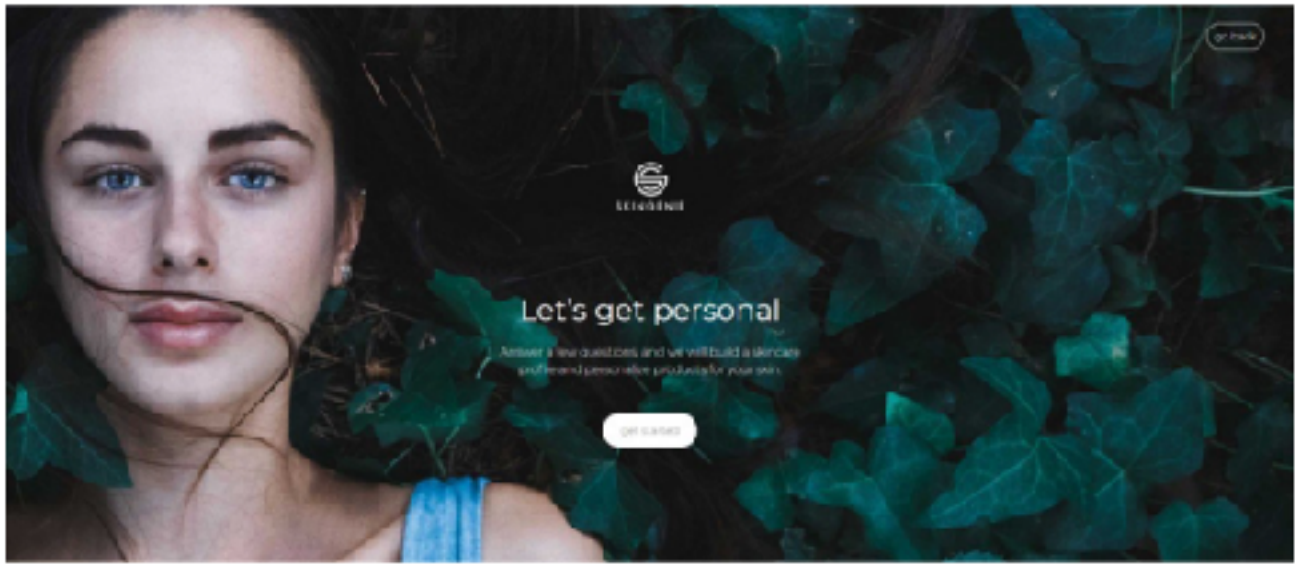
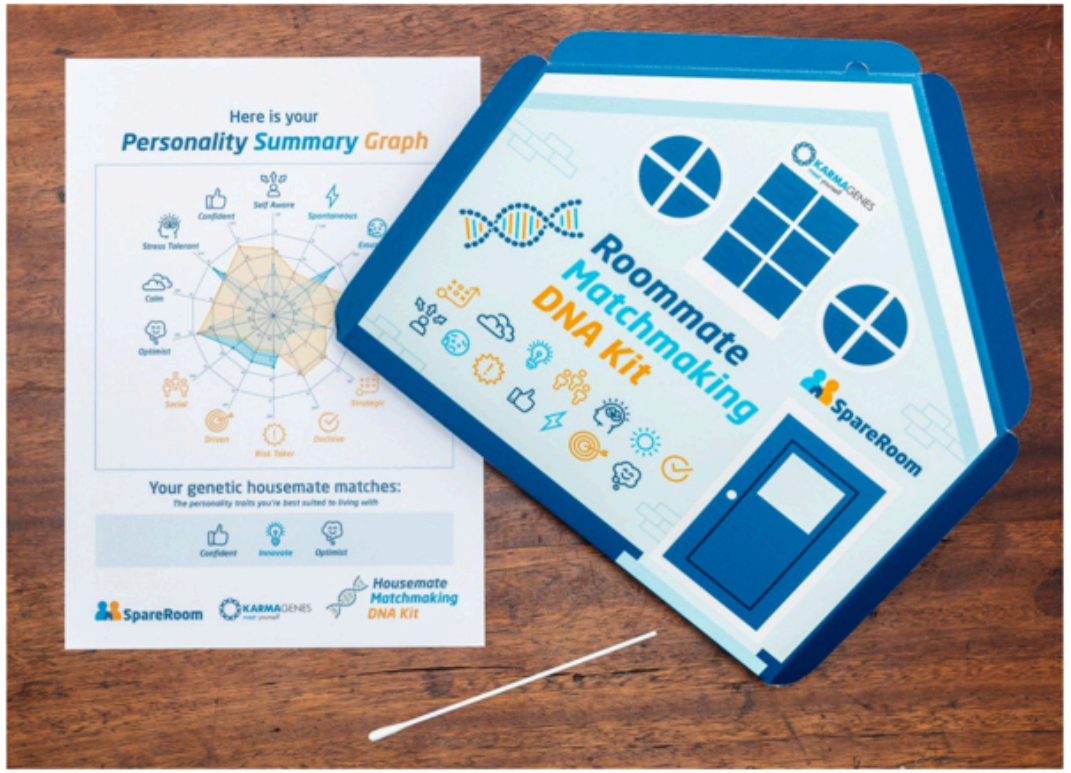

DNA RELATIVES
DOWNLO
List View
Map View
Surname View
search matches
Show: both sides
Sort: relationship
25 per page
1 - 25 of 424
Male
You
UPDATE YOUR PROFILE
Female
2nd to 3rd Cousin
1.66% shared, 5 segments
J2a2
Send an Introduction
Female
3rd to 4th Cousin
1.30% shared, 3 segments
United States
Personal
Senegal
5 more
U5b2
Public Match
Send a Message
Male
3rd to 4th Cousin
1.03% shared, 2 segments
H13a1a
R1b1b2
Send an Introduction
Female
3rd to 5th Cousin
0.45% shared, 2 segments
H7
Send an Introduction
Female
3rd to 5th Cousin
0.42% shared, 2 segments
H1
Send an Introduction
Male
3rd to 5th Cousin
0.40% shared, 2 segments
United States
Reno, Nevada
San Diego, California
Tucker
Littlefield
Warga
4 more
H1c
G2a
Public Match
Send a Message
Male
3rd to 5th Cousin
0.37% shared, 2 segments
United States
K1a1b
R1b1b2a1a
Public Match
Send a Message
Male, c. 1978
3rd to 6th Cousin
0.40% shared, 1 segment
United States
New Jersey
Utah
California
Northern Europe
U3b1
T
Send an Introduction

SkinGenie



SKINGENIE

SpareRoom

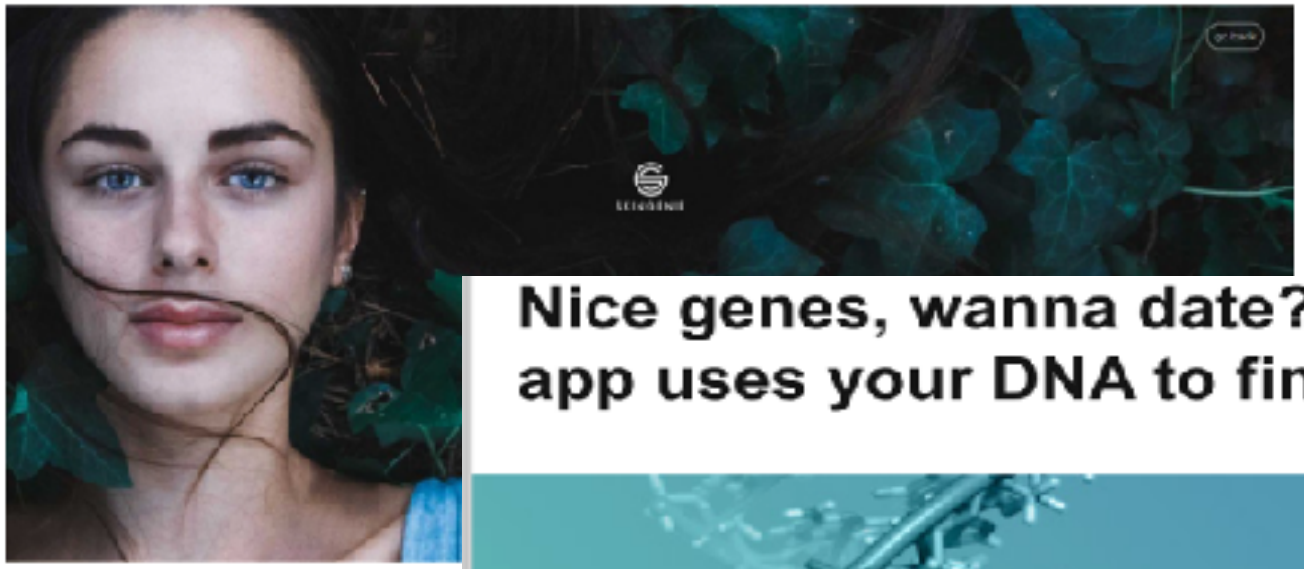


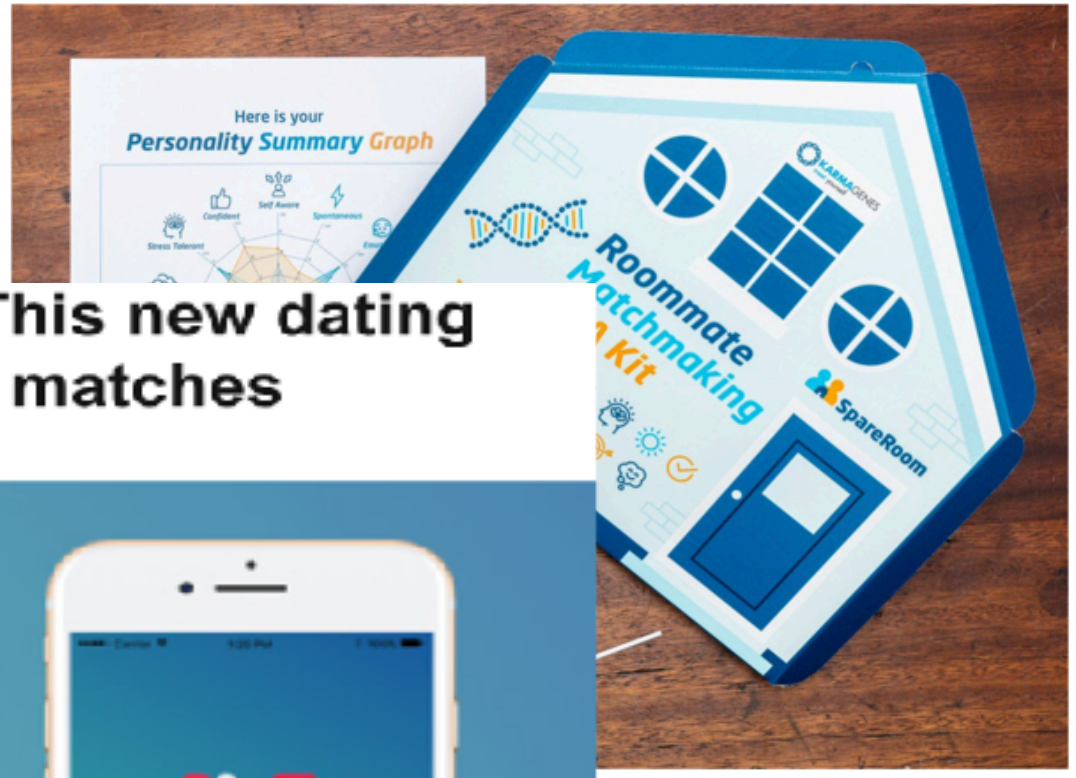
SPAREROOM

VINOME

SkinGenie

SpareRoom

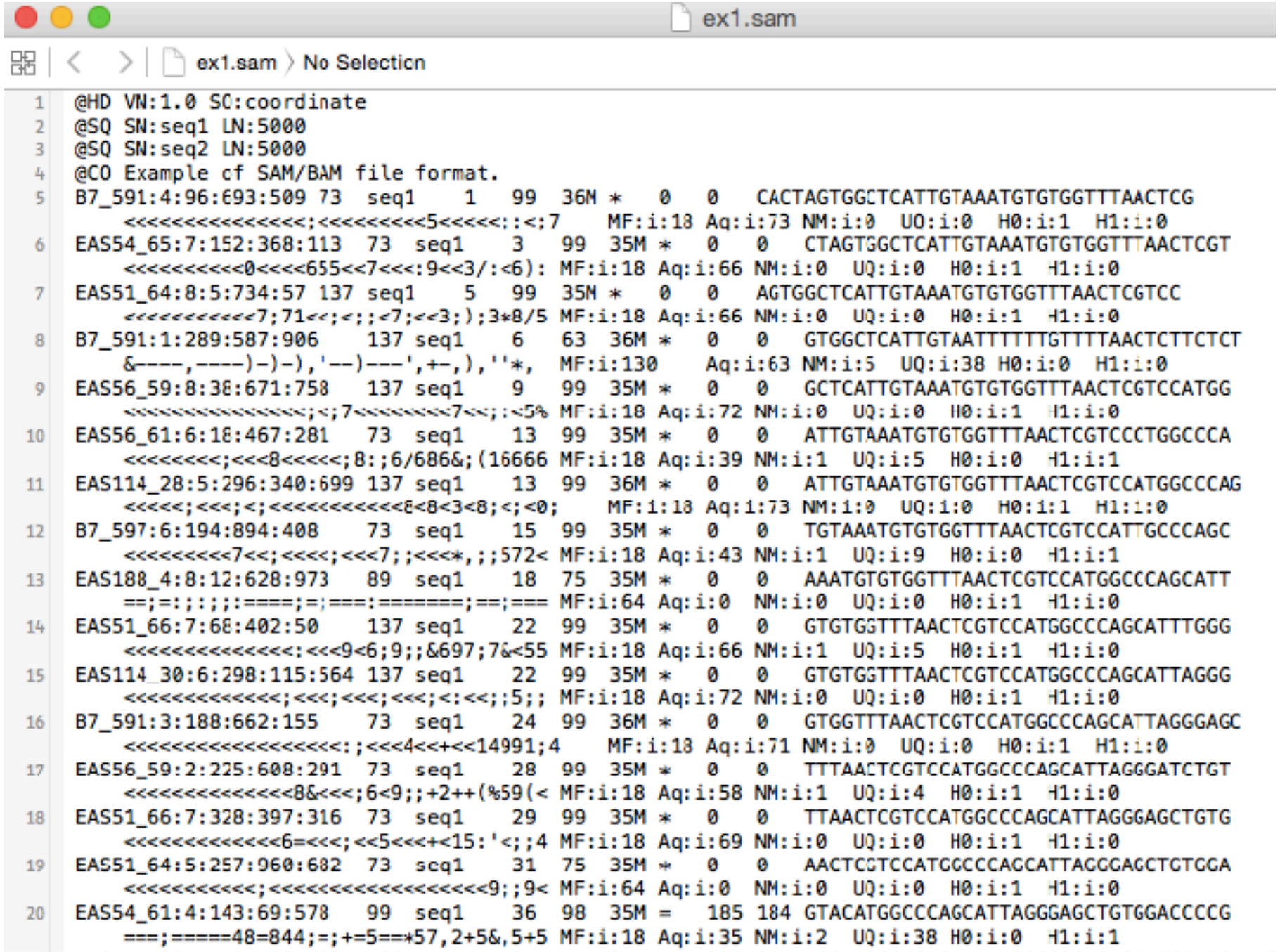




Nice genes, wanna date? This new dating app uses your DNA to find matches

SKINGENIE

VINOME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ex1.sam 〉 No Selection
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@SQ SN:seq1 LN:5000
@SQ SN:seq2 LN:5000
@CO Example of SAM/BAM file format.
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But… not all data is created equal!

Health Data Hacking

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Anthem Hacking Points to Security Vulnerability of Health Care Industry

By REED ABELSON and MATTHEW GOLDSTEIN FEB. 5, 2015

The New York Times

Health Data Hacking

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The New York Times

Anthem: one of US largest health insurers

60 to 80 million *unencrypted* records stolen in the hack (revealed in February 2015)

Social security numbers, birthdays, addresses, email and employment information and income data for customers and employees, including its own chief executive

US Healthcare “Wall of Shame”

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Around 2 declared breaches per week, each affecting 500+ people

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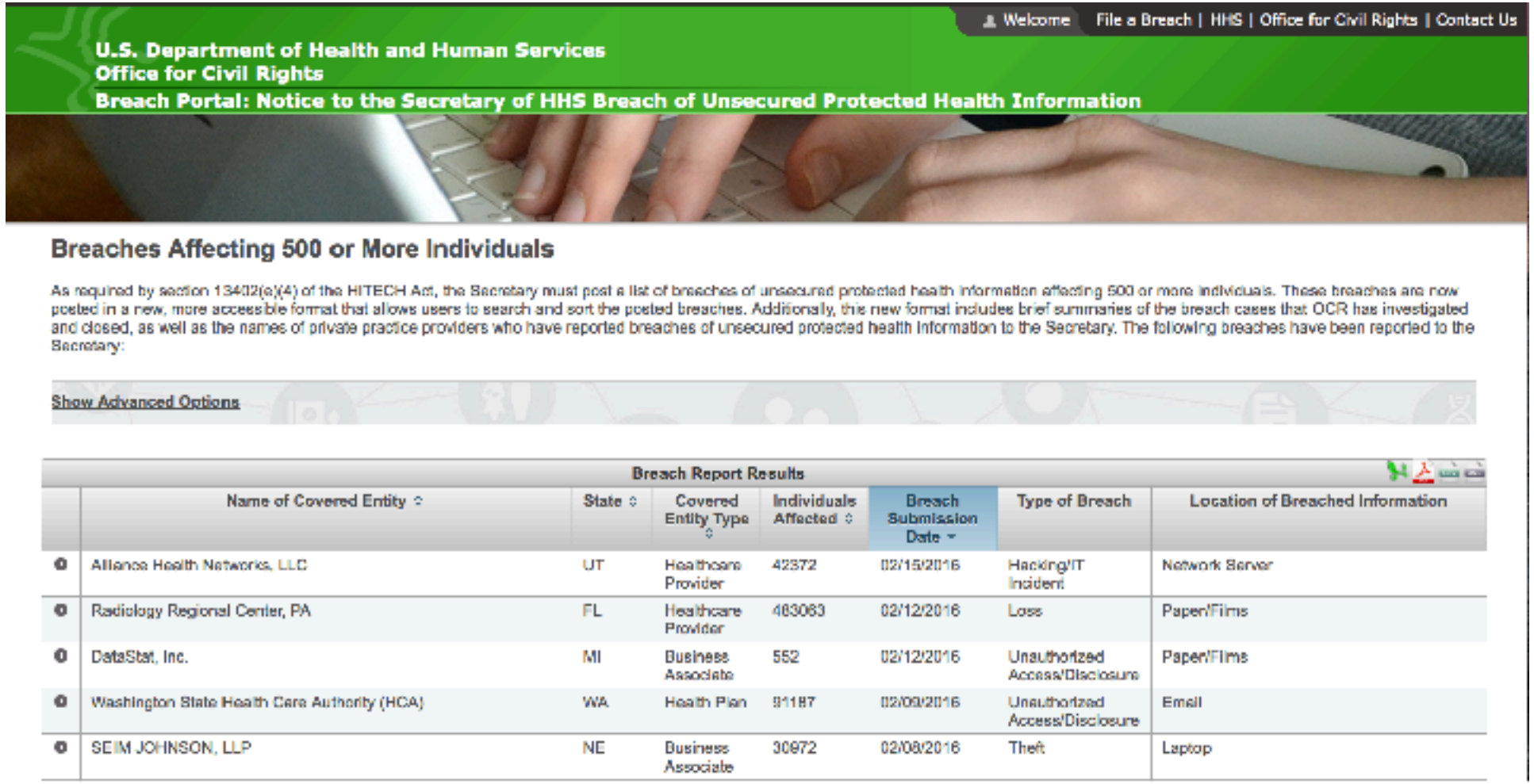
https://ocrportal.hhs.gov/ocr/breach/breach_report.jsf



De-Anonymization

TECH 4/25/2013 @ 3:47PM | 17,111 views

Harvard Professor Re-Identifies Anonymous Volunteers In DNA Study

+ Comment Now + Follow Comments

A Harvard professor has re-identified the names of more than 40% of a sample of anonymous participants in a high-profile DNA study, highlighting the dangers that ever greater amounts of personal data available in the Internet era could unravel personal secrets.

Harvard Professor Latanya Sweeney

From the onset, the Personal Genome Project,

Melissa Gymrek et al. *"Identifying Personal Genomes by Surname Inference."* Science Vol. 339, No. 6117, 2013

Aggregation

Re-identification of aggregated data

Statistics from allele frequencies can be used to identify genetic trial participants [1]

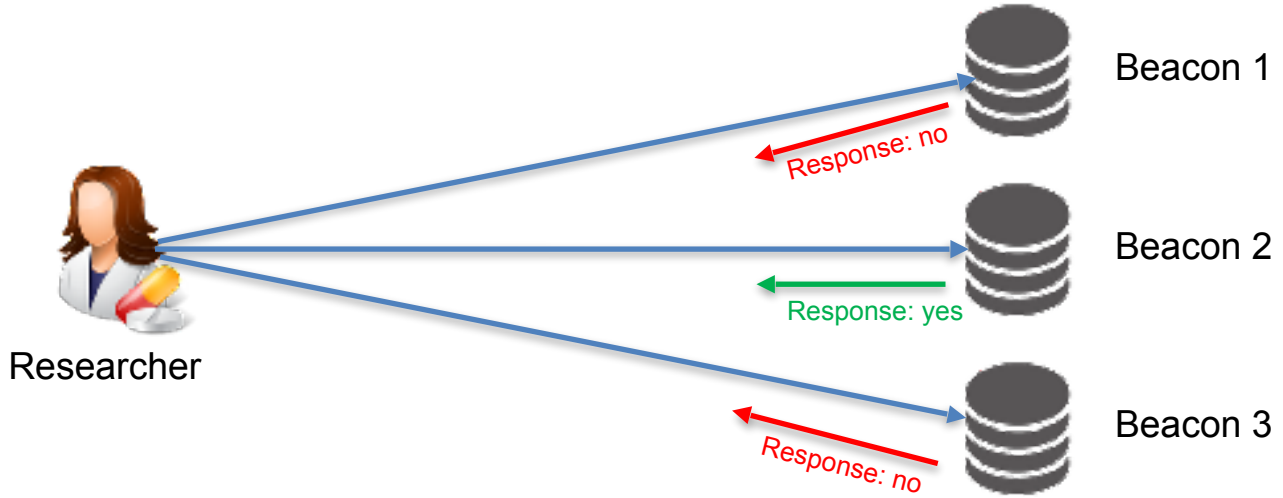
Membership Inference

Presence of an individual in a group can be determined by using allele frequencies and his DNA profile [2]

Against individuals contributing their microRNA expressions to scientific studies [3]

[1] R. Wang et al. “Learning Your Identity and Disease from Research Papers: Information Leaks in Genome Wide Association Study.” CCS, 2009
[2] N. Homer et al. Resolving individuals contributing trace amounts of DNA to highly complex mixtures using high-density SNP genotyping microarrays. PLoS Genetics, 2008
[3] M. Backes et al. Membership privacy in MicroRNA-based studies. CCS, 2016.

GA4GH Beacon Project

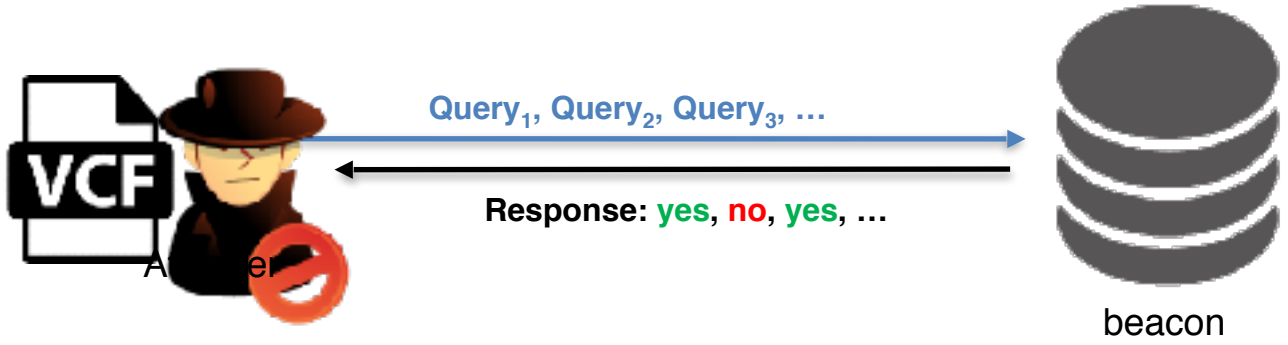


Main features:

Allows researchers to quickly query multiple database to find the sample they need; encourages cross-borders collaboration among researchers

Only minimal responses back in order to mitigate privacy concerns

Shringarpure-Bustamante's Attack



Shringarpure SS, Bustamante CD. Privacy risks from genomic data-sharing beacons. The American Journal of Human Genetics. 2015 Nov 5;97(5):631-46.

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Can be extremely dangerous if the beacon is associated with a sensitive phenotype (e.g., cancer)

Family Relationship Disclosure

Family Relationship Disclosure

Vox

THE GOODS EXPLAINERS POLITICS & POLICY WORLD CULTURE SCIENCE & HEALTH MORE

With genetic testing, I gave my parents the gift of divorce

By George Doe | Sep 9, 2014, 7:50am EDT

SHARE

Family Relationship Disclosure

reddit r/23andme Search r/23andme LOG IN

23andMe

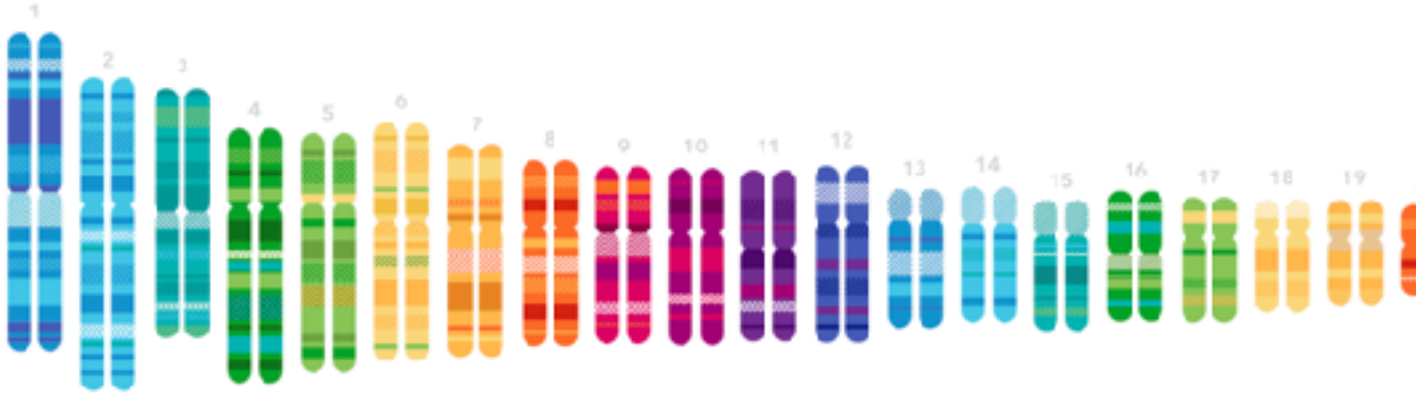
Ancestry
5 reports

Genetic Health Risks*
14 reports

Traits
214 reports

Carrier Status*
404 reports

Wellness
54 reports



Posts

Join the discussion BECOME A REDDITOR

Posted by u/23andmethrowaway22 7 months ago

132

How 23 and Me helped me find my birth father.

(copied from a Facebook post I wrote about this, after taking out identifying data)

Background: I'm 30, female, just took my 23 and Me test in November.

Donor-conceived people are tracking down their biological fathers, even if they want to hide

Thanks to DNA tests and the internet, 'anonymous' sperm donation is a thing of the past

Emily Chung, Melanie Glanz, Vik Adhopia · CBC News ·
Posted: Jan 25, 2018 5:00 AM ET | Last Updated: August 20

Deportations on the back of consumer genetic tests worry scientists

BY BRIAN OWENS | 31 AUGUST 2018

Canada's border enforcement agency appears to be using genetic tests and DNA ancestry sites to determine country of origin for would-be deportees

Some issues specific to genomes

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Genome is treasure trove of sensitive information

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Genome is the ultimate identifier

Access to one's genome ≈ **access** to **relatives'** genome

Are these “new” threats?

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We all leave cells behind after all…

Hair/saliva can be collected and sequenced?

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…scalability…

Looking Ahead…

How do we overcome the “greater good” vs “privacy” dichotomy?

How do we empower users with informed decisions about their health and genomic data?

How do reason about ethical issues around relatives?


parc
A Xerox Company


MARIE CURIE ACTIONS


Google
Google
Faculty Research Awards

Thank you!