Genomic Data & Privacy

Risks & opportunities

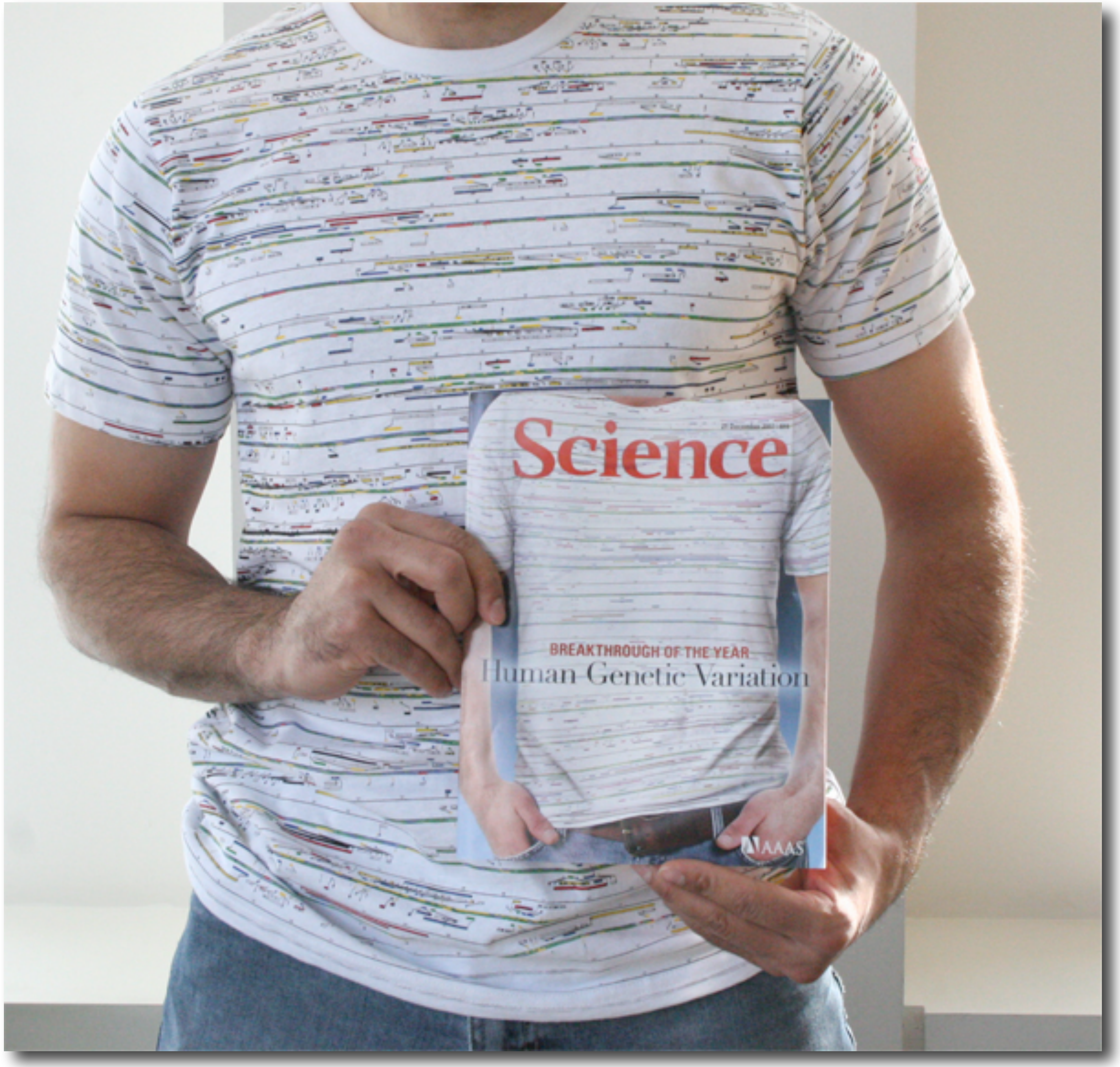
Michael Baudis | UZH BIO392 HS23

Genomic Data & Privacy

Risks & opportunities

- Why do we need a lot of data for understanding genomic variation in health and disease?
- Data sharing protocols ...
  - GA4GH Beacon
- Breaking data privacy
  - Different types of (genomic) privacy attacks
  - Beacon attacks and mitigation
  - DTC and Longe-range familial attacks
- Regulation of genome data production & access in Switzerland
- Some strategies for enabling genomic data sharing & re-use

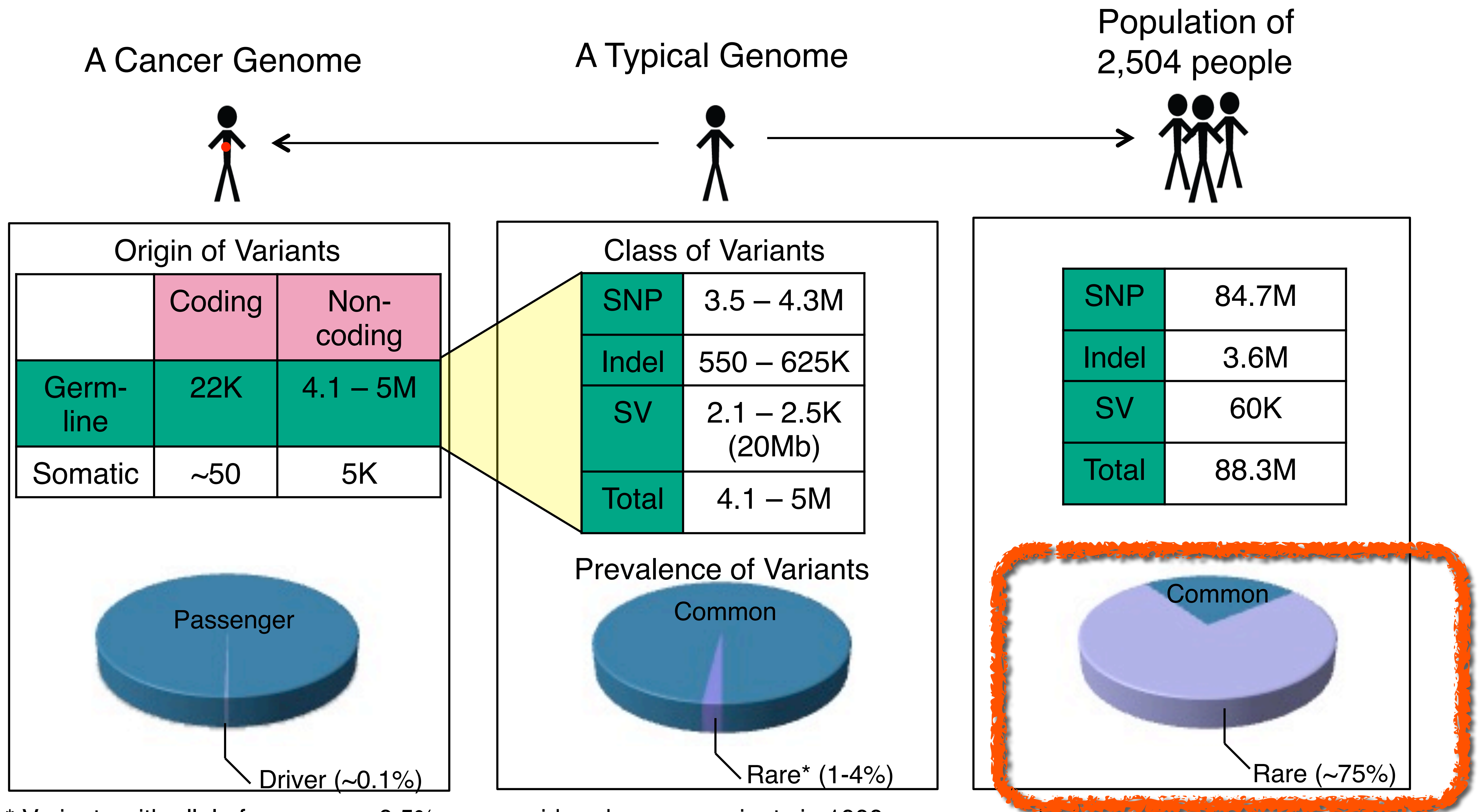
The **trouble with** human genome **variation**



Finding Somatic Mutations In Cancer

Many Needles in a Large Haystack

- a typical human genome (~3 billion base pairs) has ~5 million variants
- most of them are "**rare**"; i.e. can only be identified as recurring when sequencing thousands of people
- cancer cells accumulate additional variants, only **few** of which ("**drivers**") are relevant for the disease



Origin of Variants

	Coding	Non-coding
Germ-line	22K	4.1 – 5M
Somatic	~50	5K

Class of Variants

SNP	3.5 – 4.3M
Indel	550 – 625K
SV	2.1 – 2.5K (20Mb)
Total	4.1 – 5M

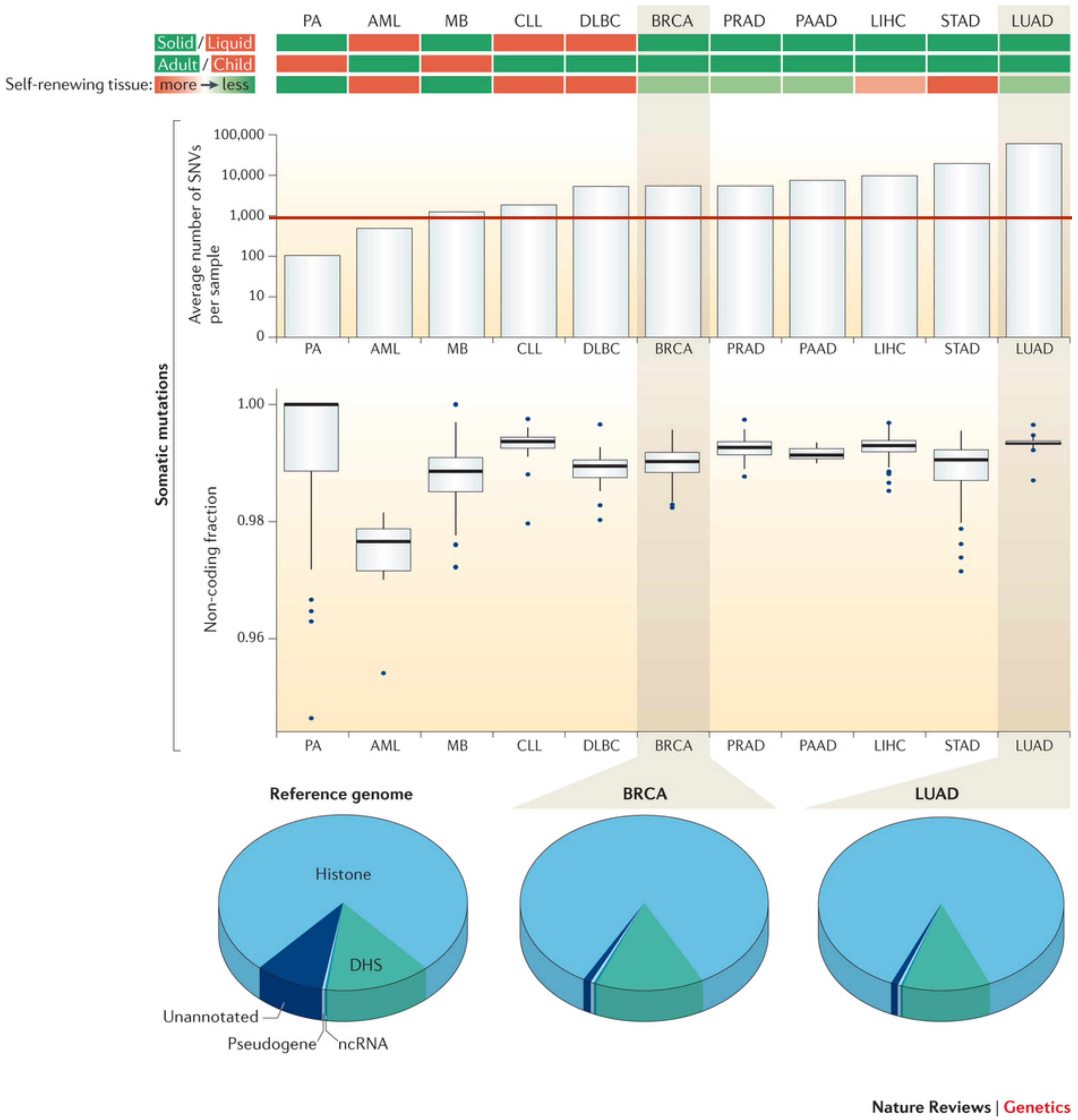
SNP	84.7M
Indel	3.6M
SV	60K
Total	88.3M

* Variants with allele frequency < 0.5% are considered as rare variants in 1000 genomes project.

The 1000 Genomes Project Consortium, Nature. 2015. 526:68-74
Khurana E. et al. Nat. Rev. Genet. 2016. 17:93-108

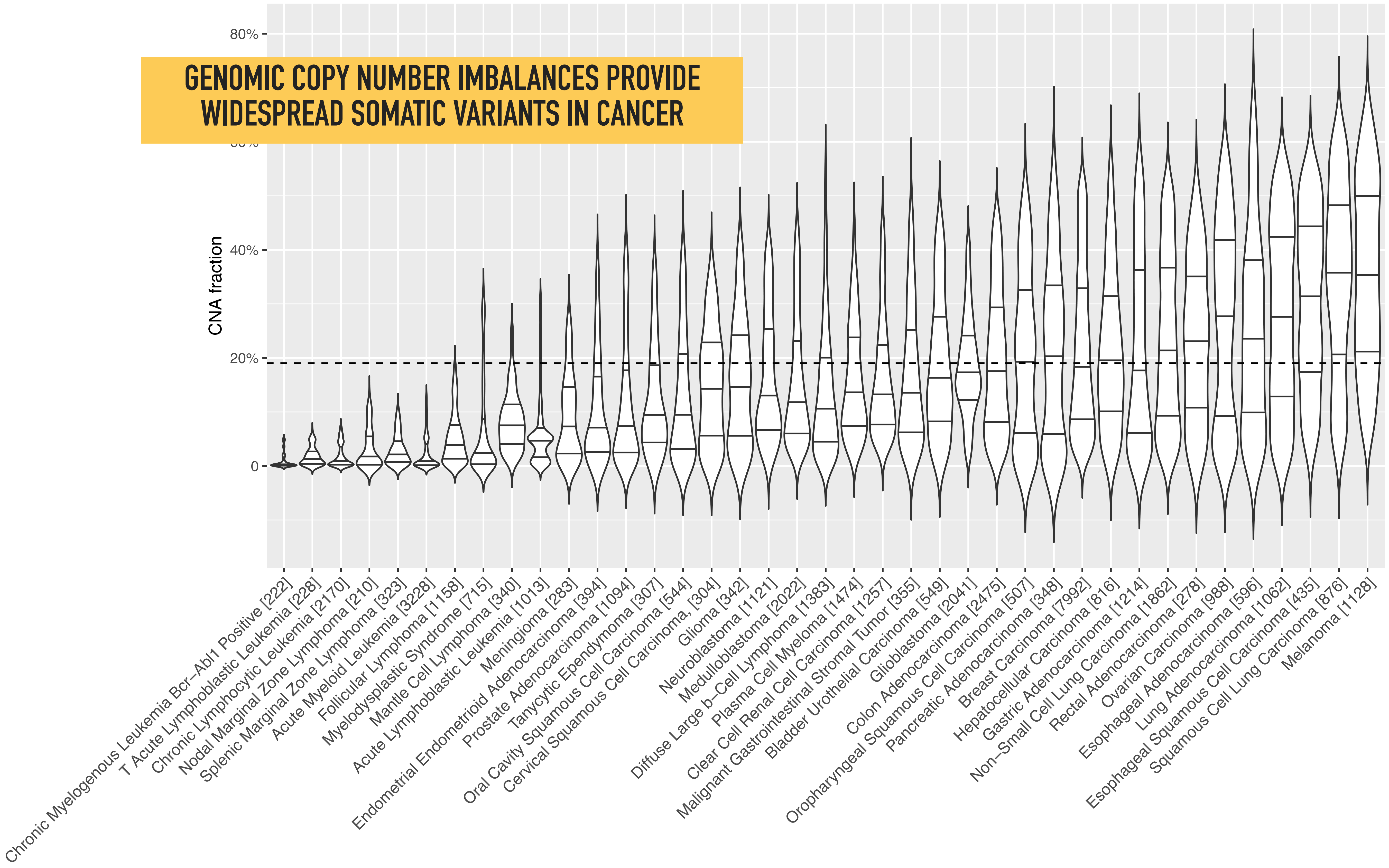
Graphic adapted from Mark Gerstein (GersteinLab.org; @markgerstein)

Quantifying Somatic Mutations In Cancer



CANCERS SHOW THOUSANDS OF SINGLE NUCLEOTIDE VARIANTS PER SAMPLE, MOSTLY IN NON-CODING REGIONS

Pan-Cancer Analysis of Whole Genomes (PCAWG) data show widespread mutations in non-coding regions of cancer genomes (Khurana et al., Nat. Rev. Genet. (2016)

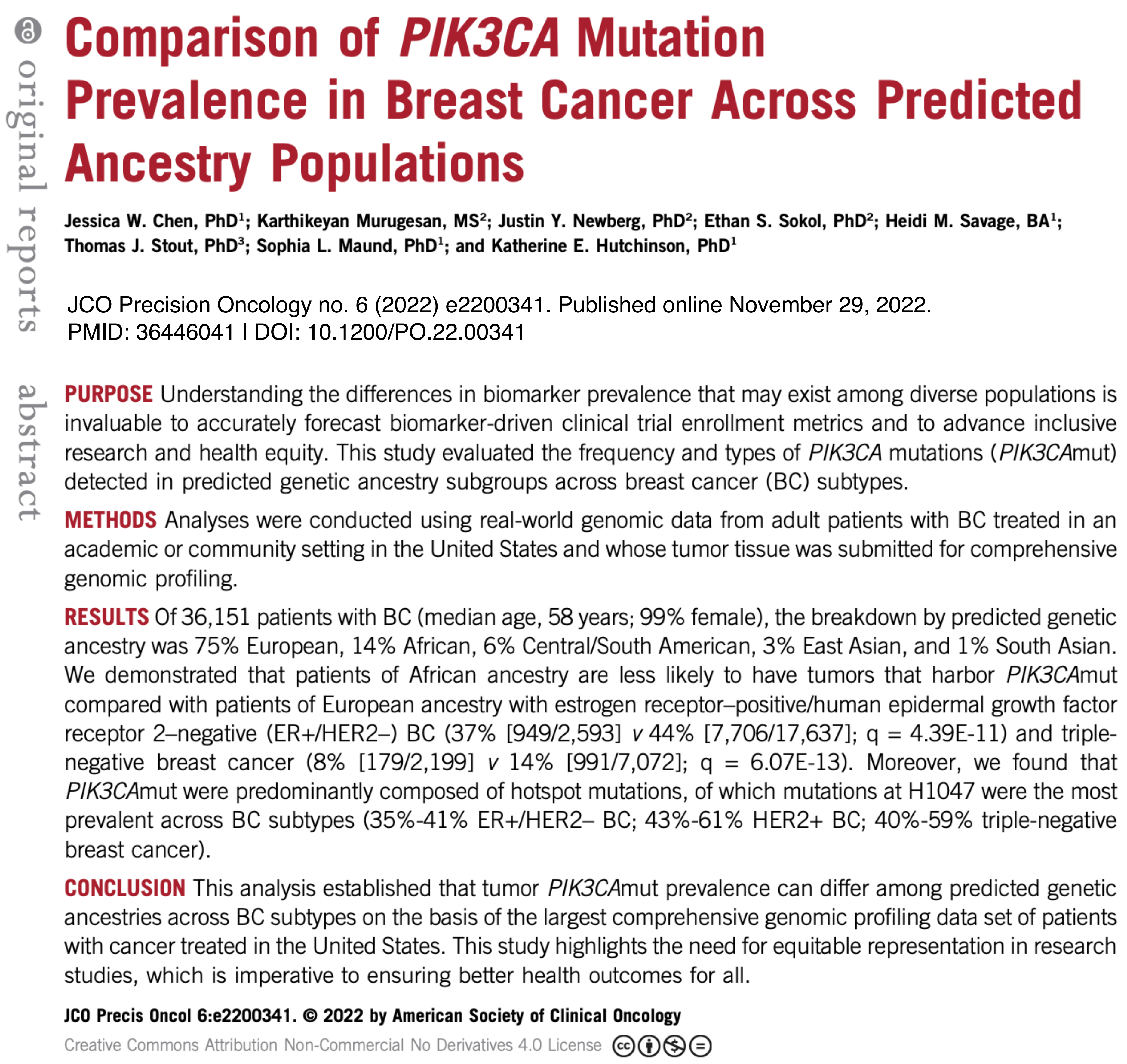


GENOMIC COPY NUMBER IMBALANCES PROVIDE WIDESPREAD SOMATIC VARIANTS IN CANCER

On average ~19% of a cancer genome are in an imbalanced state (more/less than 2 alleles); Original data based on 43654 cancer genomes from progenetix.org

Why is population representation needed?

- types and prevalences of somatic variations may vary on different ancestral backgrounds
- relevant e.g. for design of variant panels, drug selection and clinical trial statistics



original reports abstract

Comparison of *PIK3CA* Mutation Prevalence in Breast Cancer Across Predicted Ancestry Populations

Jessica W. Chen, PhD[1]; Karthikeyan Murugesan, MS[2]; Justin Y. Newberg, PhD[2]; Ethan S. Sokol, PhD[2]; Heidi M. Savage, BA[1]; Thomas J. Stout, PhD[3]; Sophia L. Maund, PhD[1]; and Katherine E. Hutchinson, PhD[1]

JCO Precision Oncology no. 6 (2022) e2200341. Published online November 29, 2022.
PMID: 36446041 I DOI: 10.1200/PO.22.00341

PURPOSE Understanding the differences in biomarker prevalence that may exist among diverse populations is invaluable to accurately forecast biomarker-driven clinical trial enrollment metrics and to advance inclusive research and health equity. This study evaluated the frequency and types of *PIK3CA* mutations (*PIK3CA*mut) detected in predicted genetic ancestry subgroups across breast cancer (BC) subtypes.

METHODS Analyses were conducted using real-world genomic data from adult patients with BC treated in an academic or community setting in the United States and whose tumor tissue was submitted for comprehensive genomic profiling.

RESULTS Of 36,151 patients with BC (median age, 58 years; 99% female), the breakdown by predicted genetic ancestry was 75% European, 14% African, 6% Central/South American, 3% East Asian, and 1% South Asian. We demonstrated that patients of African ancestry are less likely to have tumors that harbor *PIK3CA*mut compared with patients of European ancestry with estrogen receptor–positive/human epidermal growth factor receptor 2–negative (ER+/HER2–) BC (37% [949/2,593] *v* 44% [7,706/17,637]; q = 4.39E-11) and triple-negative breast cancer (8% [179/2,199] *v* 14% [991/7,072]; q = 6.07E-13). Moreover, we found that *PIK3CA*mut were predominantly composed of hotspot mutations, of which mutations at H1047 were the most prevalent across BC subtypes (35%-41% ER+/HER2– BC; 43%-61% HER2+ BC; 40%-59% triple-negative breast cancer).

CONCLUSION This analysis established that tumor *PIK3CA*mut prevalence can differ among predicted genetic ancestries across BC subtypes on the basis of the largest comprehensive genomic profiling data set of patients with cancer treated in the United States. This study highlights the need for equitable representation in research studies, which is imperative to ensuring better health outcomes for all.

JCO Precis Oncol 6:e2200341. © 2022 by American Society of Clinical Oncology

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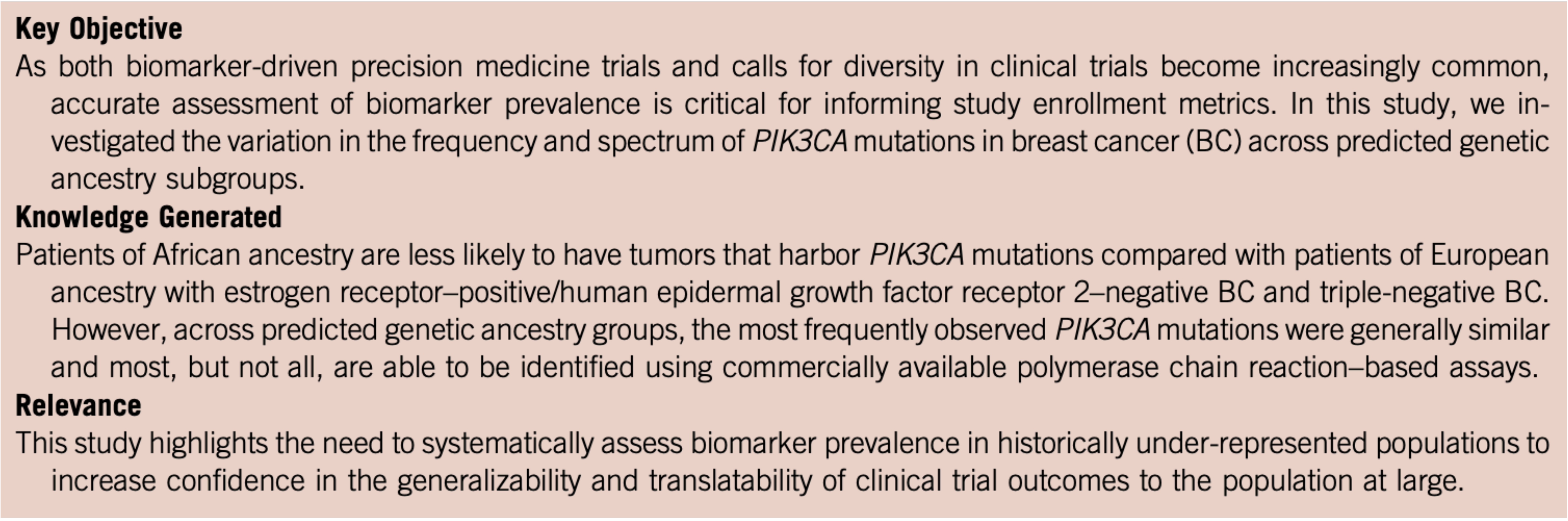


Key Objective

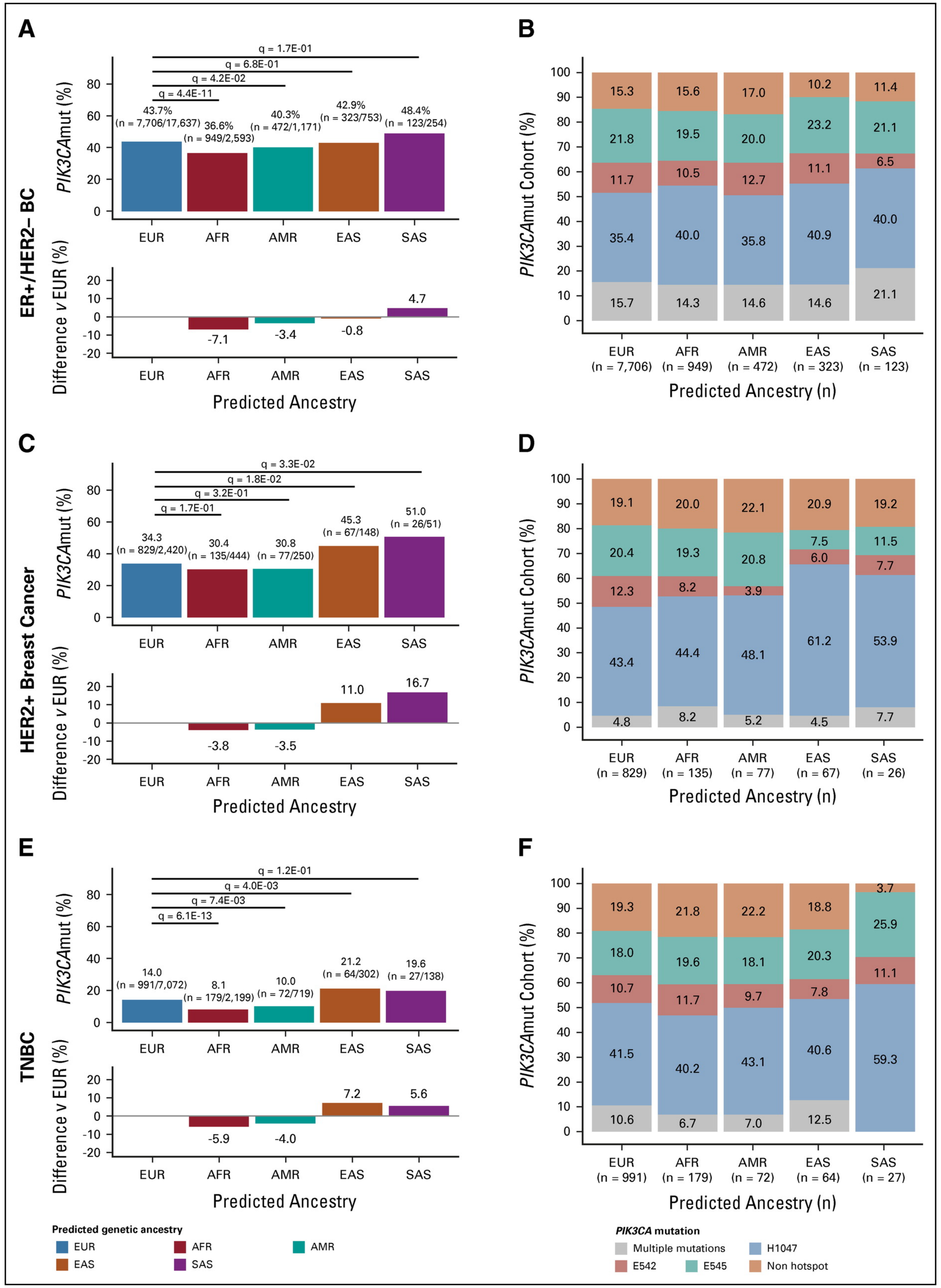
As both biomarker-driven precision medicine trials and calls for diversity in clinical trials become increasingly common, accurate assessment of biomarker prevalence is critical for informing study enrollment metrics. In this study, we investigated the variation in the frequency and spectrum of *PIK3CA* mutations in breast cancer (BC) across predicted genetic ancestry subgroups.

Knowledge Generated

Patients of African ancestry are less likely to have tumors that harbor *PIK3CA* mutations compared with patients of European ancestry with estrogen receptor–positive/human epidermal growth factor receptor 2–negative BC and triple-negative BC. However, across predicted genetic ancestry groups, the most frequently observed *PIK3CA* mutations were generally similar and most, but not all, are able to be identified using commercially available polymerase chain reaction–based assays.

Relevance

This study highlights the need to systematically assess biomarker prevalence in historically under-represented populations to increase confidence in the generalizability and translatability of clinical trial outcomes to the population at large.



Limited Population Diversity in Cancer Studies

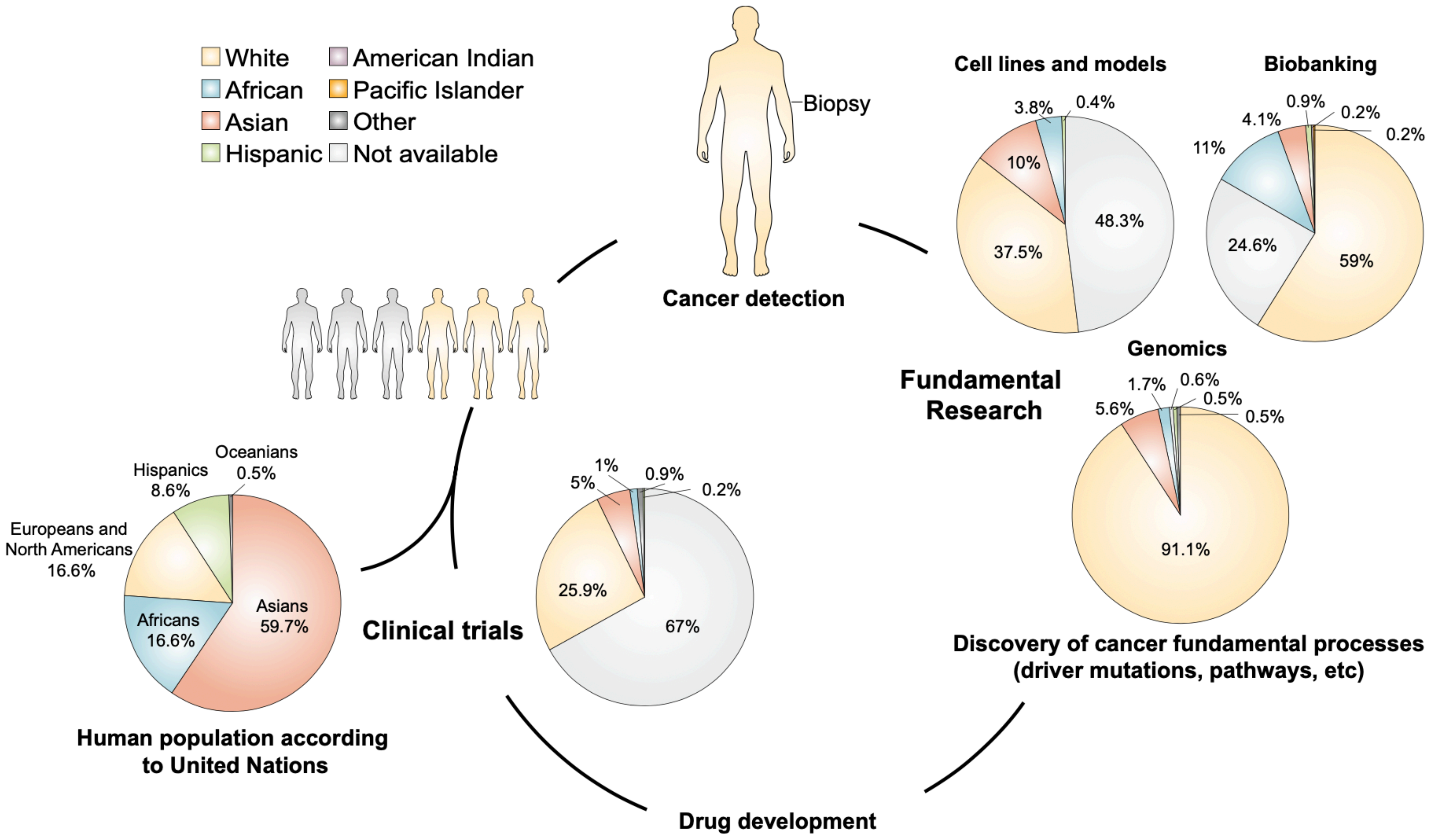


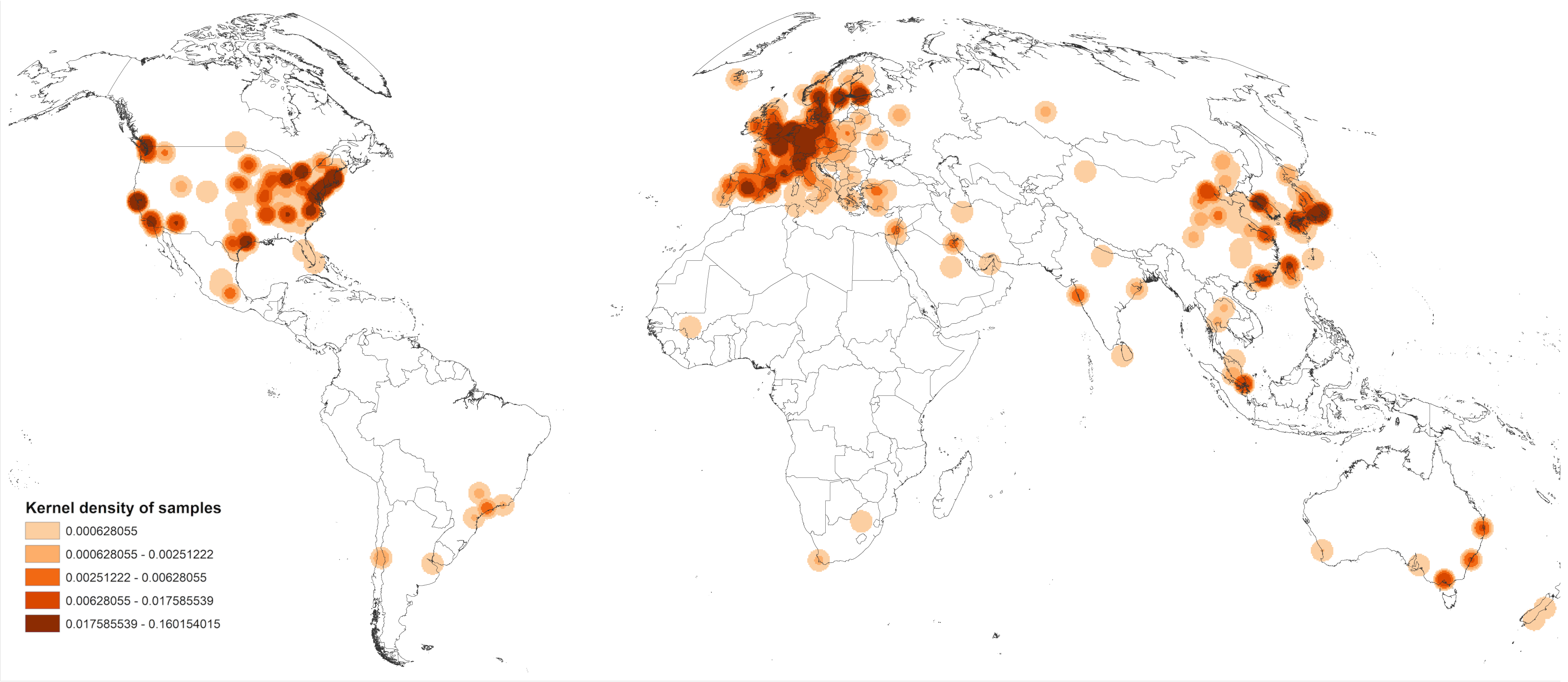
Figure 1. Racial/Ethnic disparities in cancer research. Racial/ethnic inclusion was studied in several aspects of oncological research, from cell lines and patient-derived xenografts to biobanking, genomics and clinical trials.

Guerrero S, López-Cortés A, Indacochea A, et al. Analysis of Racial/Ethnic Representation in Select Basic and Applied Cancer Research Studies. *Sci Rep*. 2018;8(1):13978.

Publication Landscape of Cancer CNV Profiling

Publication statistics for cancer genome screening studies. The graphic shows our as- sessment of publications reporting whole-genome screening of cancer samples, using molecular detection methods (chromosomal CGH, genomic array technologies, whole exome and genome sequencing).

For the years 1993-2018, we found 3'229 publications reporting 174'530 individual samples in single series from 1 to more than 1000 samples. Y-axis and size of the dots correspond to the sample number; the color codes indicate the technology used.



Global Genomic Data Sharing Can...

Global Alliance
for Genomics & Health

Demonstrate patterns in health & disease

Increase statistical significance of analyses

Lead to “stronger” variant interpretations

Increase accurate diagnosis

Advance precision medicine

200+ Genomic Data Initiatives Globally

Clinical/Genomic Medicine

Research

National

Cohorts

biobank uk

Enabling scientific discoveries that improve human health

WHOLE GENOME SEQUENCING DATA ON 200,000 UK BIOBANK PARTICIPANTS ARE NOW AVAILABLE FOR RESEARCH USE

This dataset represents the world's largest single release of Whole Genome Sequencing data

5 PETABYTES OF WGS DATA

When combined with the extensive amount of lifestyle, biochemical and health outcome data already held for the participants in UK Biobank, it will enable researchers to better understand the role of genetics for health outcomes and to advance drug discovery and development

wellcome · UKRI Medical Research Council · AMGEN · AstraZeneca · gsk · Johnson&Johnson INNOVATION · deCODE genetics · wellcome sanger institute

How Many Genomes?

RESEARCH

HEALTHCARE

60M individuals
132.5M sequences

CLINICAL TRIALS

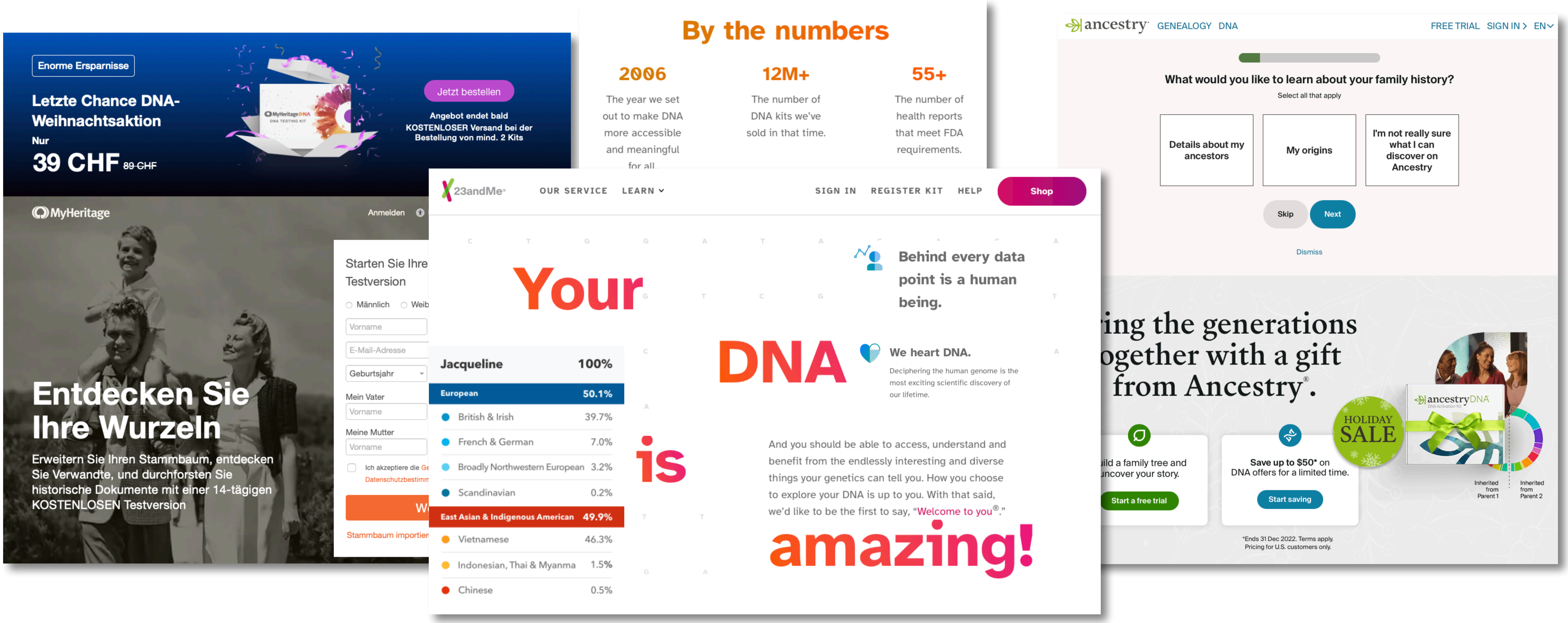
2.7-3M individuals

COHORTS

140M individuals

Direct to Consumer DNA Analyses

Population Background, Family Trees, Traits & Disease Risks...



Enorme Ersparnisse

Letzte Chance DNA-Weihnachtsaktion

Nur

39 CHF ~~89 CHF~~

Jetzt bestellen

Angebot endet bald
KOSTENLOSER Versand bei der Bestellung von mind. 2 Kits

MyHeritage — Anmelden

Entdecken Sie Ihre Wurzeln

Erweitern Sie Ihren Stammbaum, entdecken Sie Verwandte, und durchforsten Sie historische Dokumente mit einer 14-tägigen KOSTENLOSEN Testversion

Starten Sie Ihre
Testversion

○ Männlich ○ Weib

Vorname

E-Mail-Adresse

Geburtsjahr

Mein Vater

Vorname

Meine Mutter

Vorname

□ Ich akzeptiere die Ge
Datenschutzbestimm

W

Stammbaum importie

By the numbers

2006 The year we set out to make DNA more accessible and meaningful for all

12M+ The number of DNA kits we've sold in that time.

55+ The number of health reports that meet FDA requirements.

23andMe — OUR SERVICE — LEARN — SIGN IN — REGISTER KIT — HELP — Shop

Your DNA is amazing!

Behind every data point is a human being.

We heart DNA.

Deciphering the human genome is the most exciting scientific discovery of our lifetime.

And you should be able to access, understand and benefit from the endlessly interesting and diverse things your genetics can tell you. How you choose to explore your DNA is up to you. With that said, we'd like to be the first to say, "Welcome to you®."

Jacqueline	100%
European	50.1%
● British & Irish	39.7%
● French & German	7.0%
● Broadly Northwestern European	3.2%
● Scandinavian	0.2%
East Asian & Indigenous American	49.9%
● Vietnamese	46.3%
● Indonesian, Thai & Myanma	1.5%
● Chinese	0.5%

ancestry — GENEALOGY — DNA — FREE TRIAL — SIGN IN > — EN

What would you like to learn about your family history?

Select all that apply

Details about my ancestors

My origins

I'm not really sure what I can discover on Ancestry

Skip — Next

Dismiss

ing the generations together with a gift from Ancestry®.

HOLIDAY SALE

ild a family tree and uncover your story.

Start a free trial

Save up to $50* on DNA offers for a limited time.

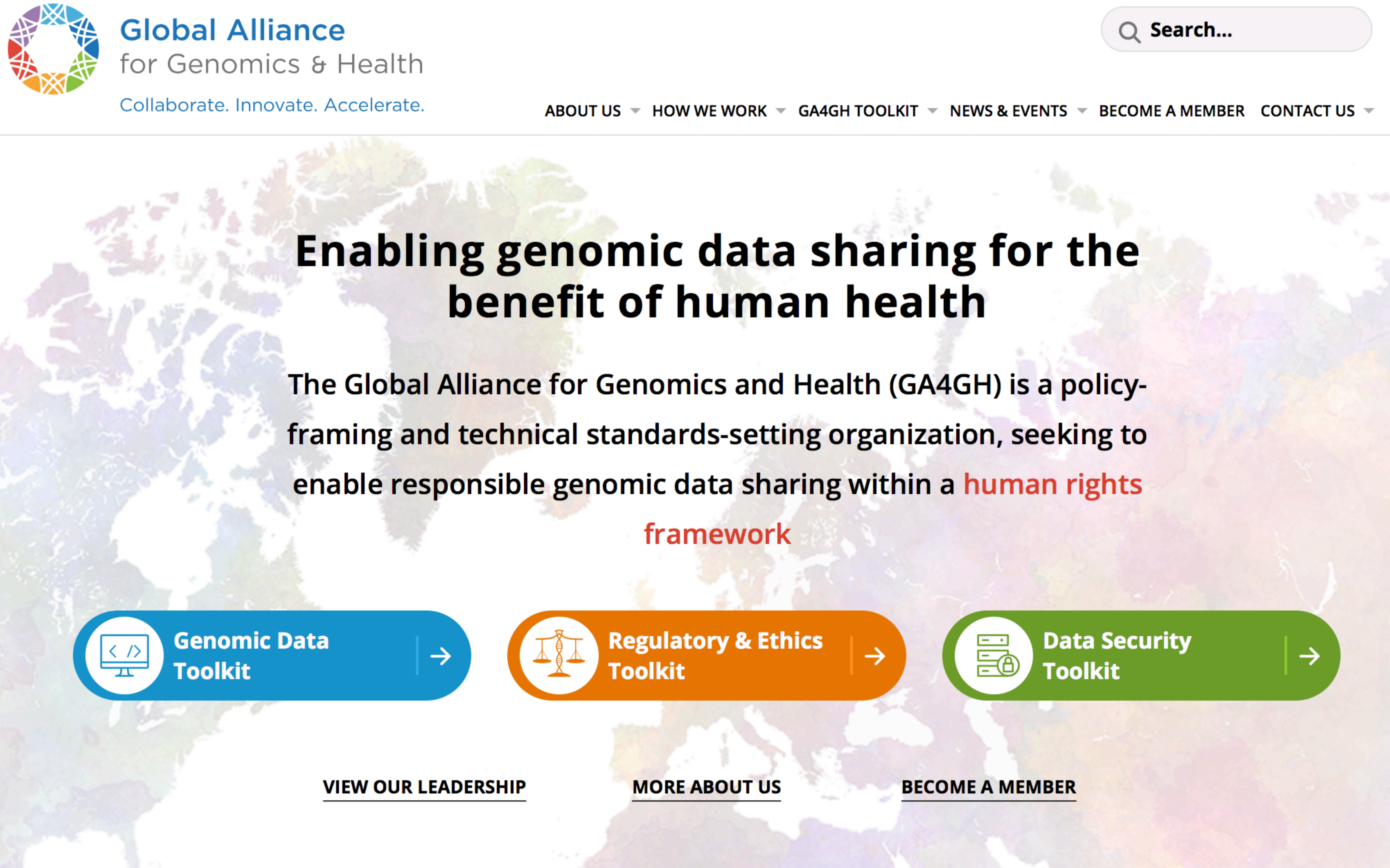
Start saving

*Ends 31 Dec 2022. Terms apply. Pricing for U.S. customers only.

The vision for genomic research: **Federation** of data

Global Alliance
for Genomics & Health

Collaborate. Innovate. Accelerate.



Enabling genomic data sharing for the benefit of human health

The Global Alliance for Genomics and Health (GA4GH) is a policy-framing and technical standards-setting organization, seeking to enable responsible genomic data sharing within a human rights framework

GENOMICS

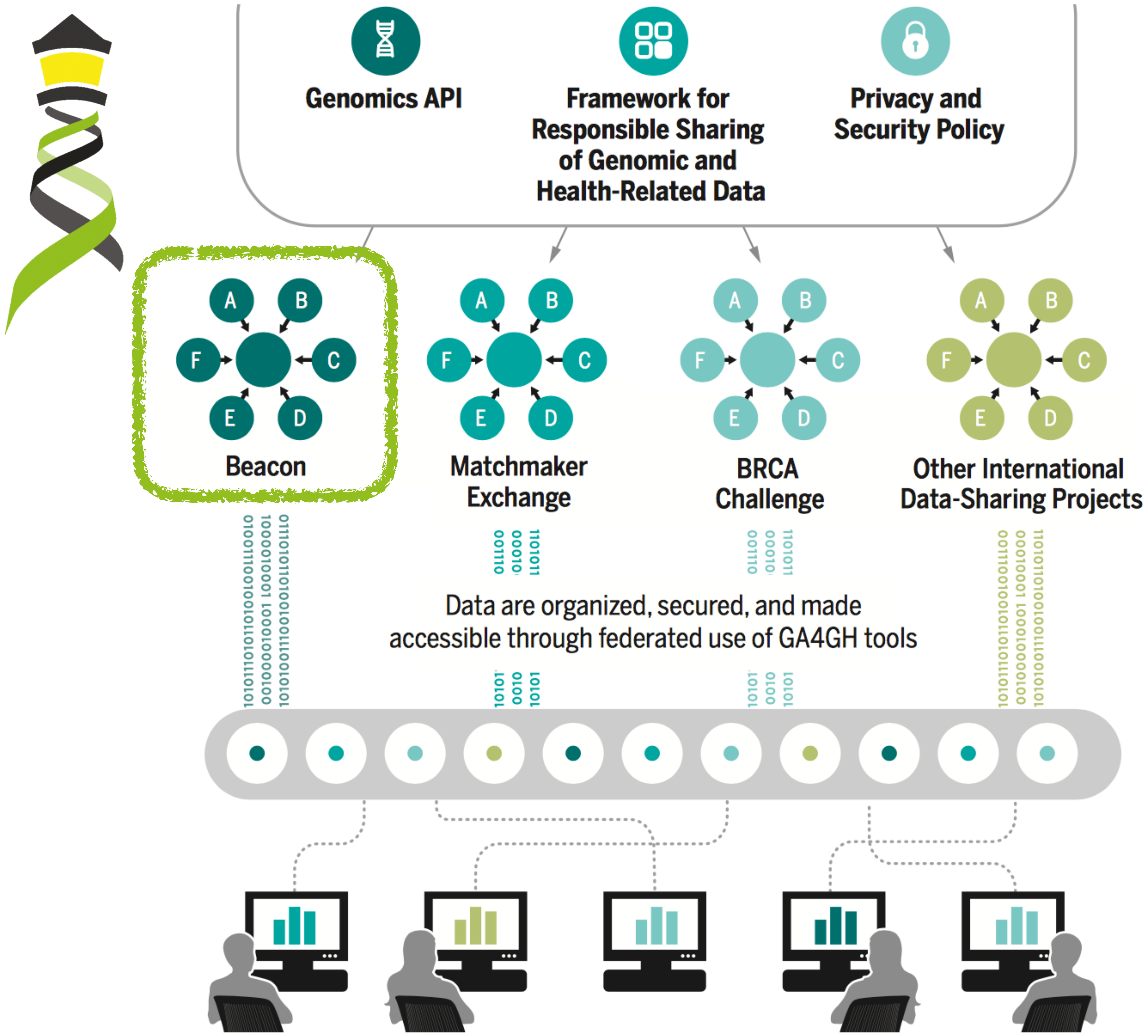
A federated ecosystem for sharing genomic, clinical data

Silos of genome data collection are being transformed into seamlessly connected, independent systems

The Global Alliance for Genomics and Health*

SCIENCE 10 JUNE 2016 • VOL 352 ISSUE 6291

A federated data ecosystem. To share genomic data globally, this approach furthers medical research without requiring compatible data sets or compromising patient identity.


Genomics API

Framework for Responsible Sharing of Genomic and Health-Related Data

Privacy and Security Policy

Beacon

Matchmaker Exchange

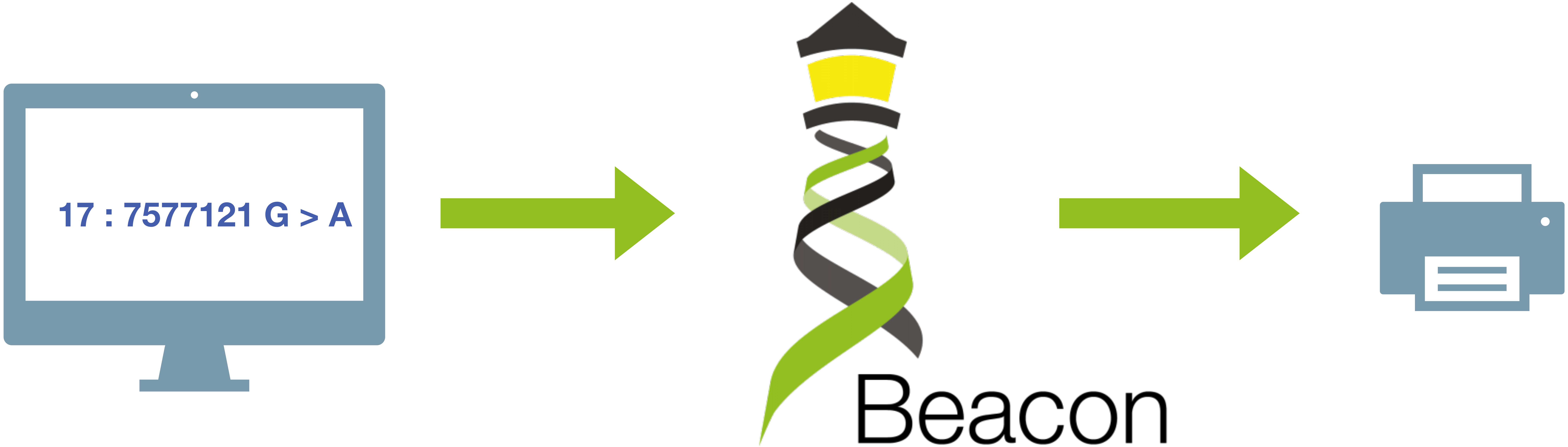
BRCA Challenge

Other International Data-Sharing Projects

Data are organized, secured, and made accessible through federated use of GA4GH tools

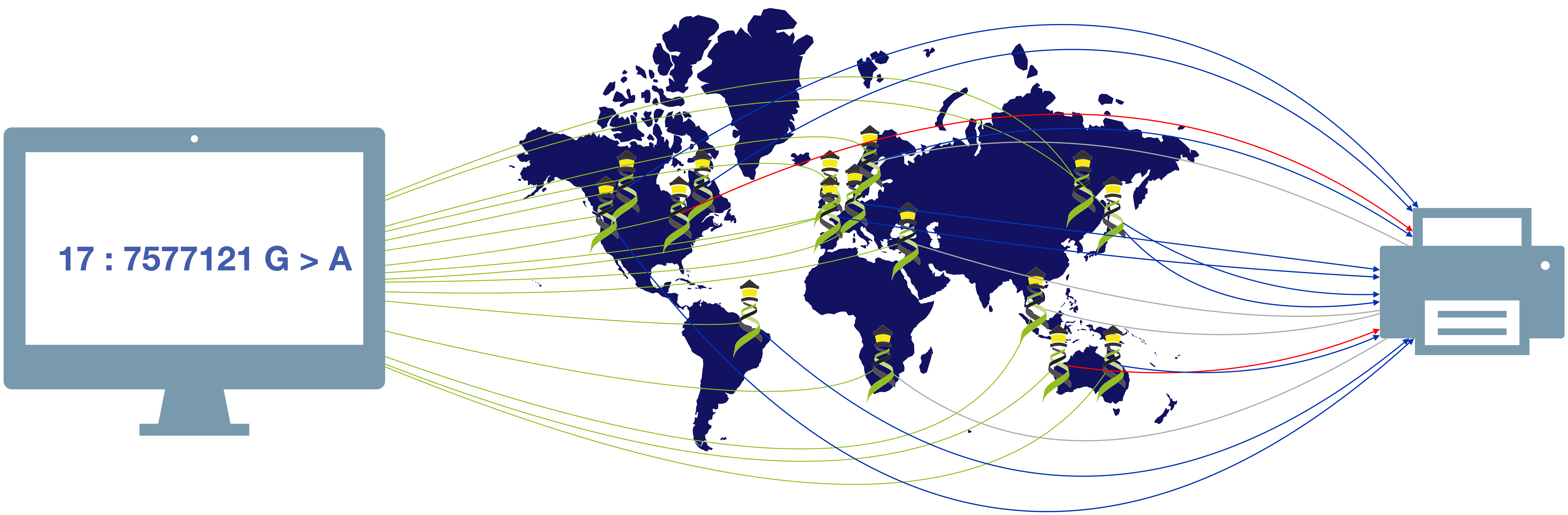
DNASTACK

Global Alliance for Genomics & Health



A ***Beacon*** answers a query for a specific genome variant against individual or aggregate genome collections

YES | **NO** | **\0**



Have you seen this variant? It came up in my patient and we don't know if this is a common SNP or worth following up.

A Beacon network federates *genome variant queries* across databases that support the ***Beacon API***

Here: The variant has been found in **few** resources, and those are from **disease** specific **collections**.

Global Alliance “Beacon” - Jim Ostell, NCBI, March 7, 2014

Global Alliance
for Genomics & Health

Introduction

… I proposed a challenge application for all those wishing to seriously engage in ***international*** data sharing for human genomics. …

1. Provide a public web service
2. Which accepts a query of the form “Do you have any genomes with an “A” at position 100,735 on chromosome 3?”
3. And responds with one of “Yes” or “No” …

“Beacon” because … people have been scanning the universe of human research for ***signs of willing participants in far reaching data sharing***, but … it has remained a dark and quiet place. The hope of this challenge is to 1) ***trigger the issues*** blocking groups … in way that isn’t masked by the … complexities of the science, fully functional interfaces, and real issues of privacy, and to 2) in ***short order*** … see ***real beacons of measurable signal*** … from ***at least some sites*** … Once your “GABeacon” is shining, you can start to take the ***next steps to add functionality*** to it, and ***finding the other groups*** … following their GABeacons.

Utility

Some have argued that this simple example is not “useful” so nobody would build it. Of course it is not the first priority for this application to be scientifically useful. …intended to provide a ***low bar for the first step of real*** … ***engagement***. … there is some utility in …locating a rare allele in your data, … not zero.

A number of more useful first versions have been suggested.

1. Provide ***frequencies of all alleles*** at that point
2. Ask for all alleles seen in a gene ***region*** (and more elaborate versions of this)
3. Other more complicated queries

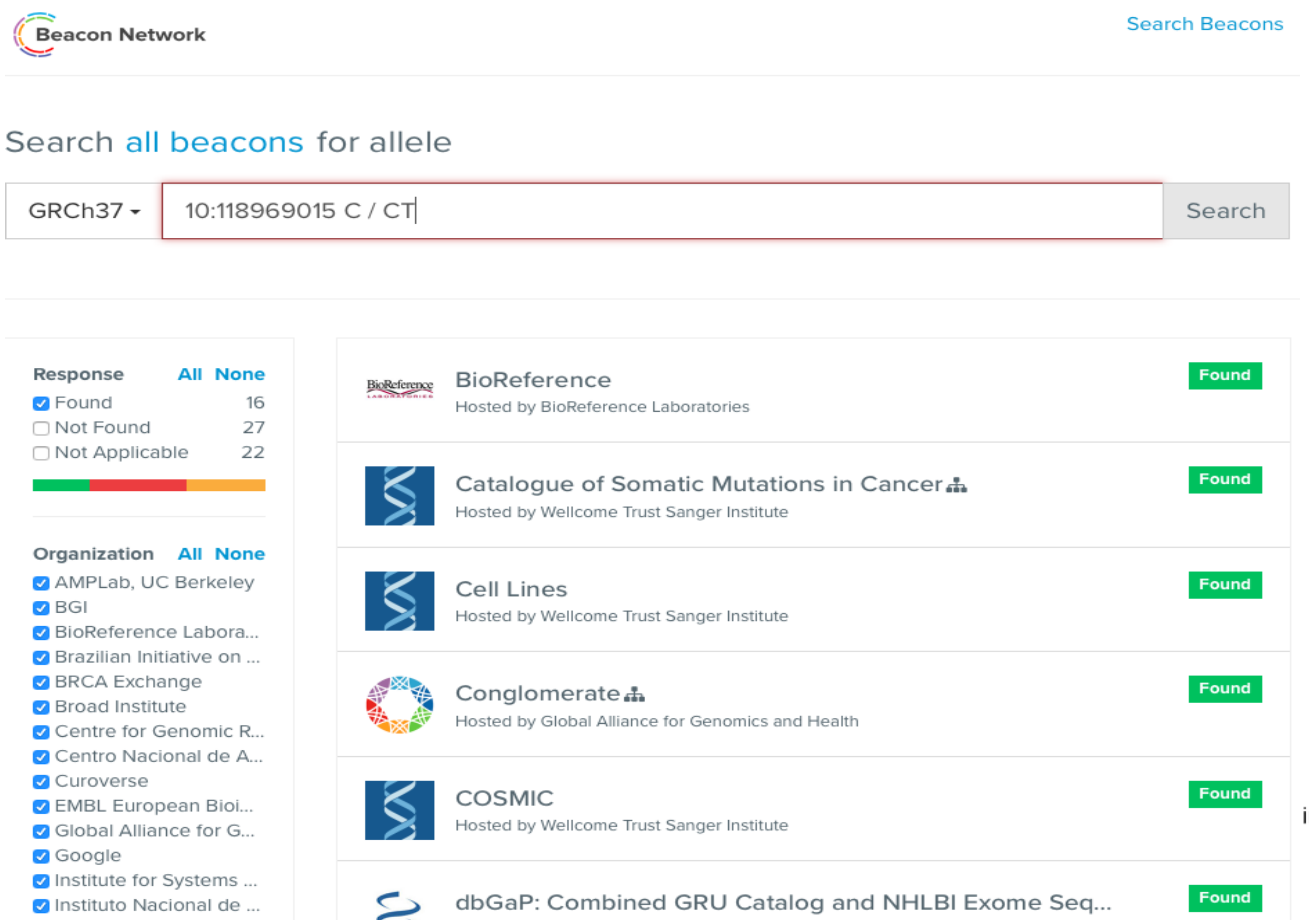
"I would personally recommend all those be held for version 2, when the beacon becomes a service."
Jim Ostell, 2014

Implementation

1. Specifying the chromosome … The interface needs to specify the ***accession.version*** of a chromosome, or ***build number***…
2. Return values … right to ***refuse*** to answer without it being an error … DOS ***attack*** … or because …especially ***sensitive***…
3. Real time response … Some sites suggest that it would be necessary to have a ***“phone home” response*** …

Beacon Project in 2016

An open web service that tests the willingness of international sites to share genetic data.



Beacon Network

Search Beacons

Search all beacons for allele

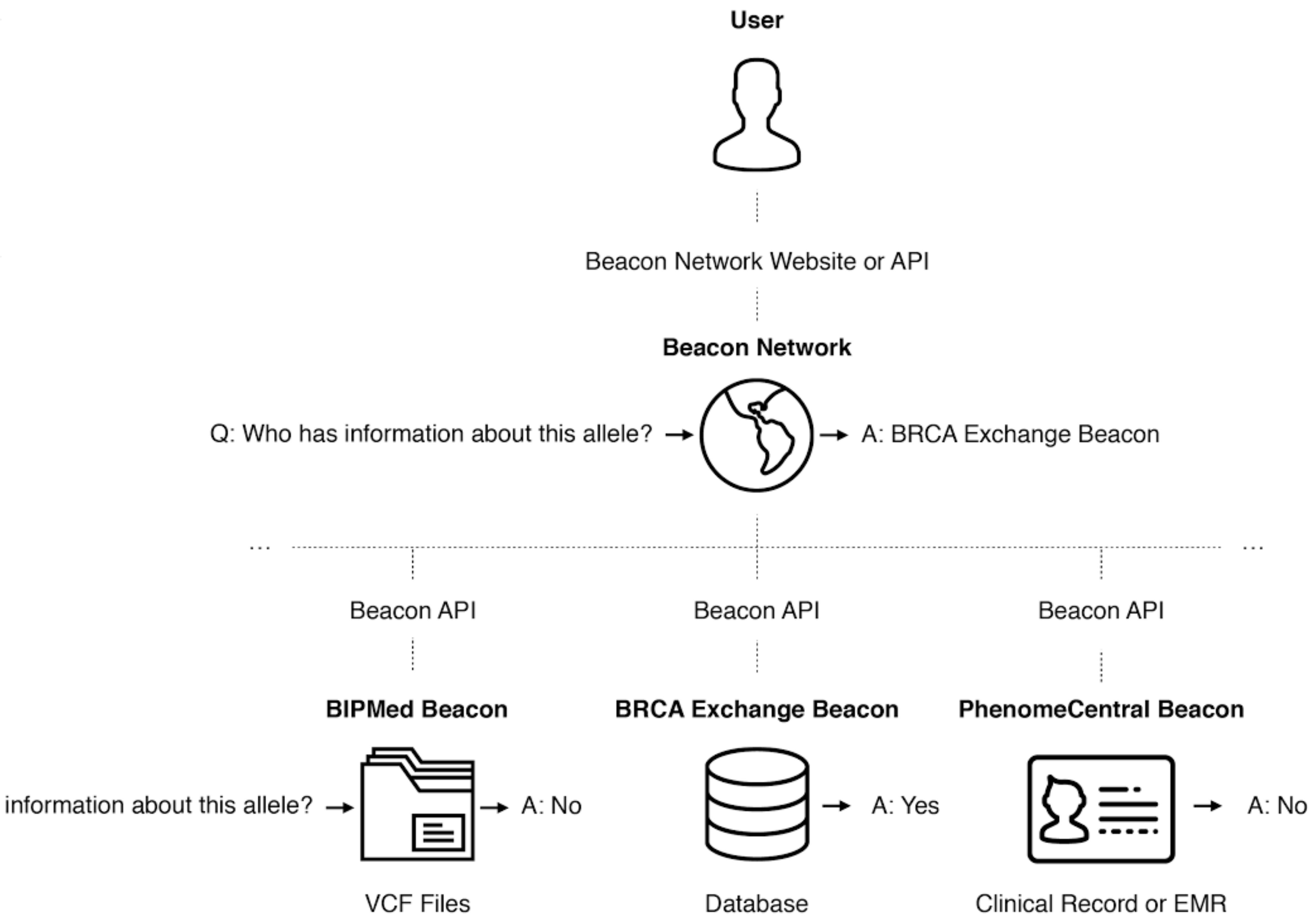
GRCh37 ▾ | 10:118969015 C / CT | Search

Response	All None
☑ Found	16
☐ Not Found	27
☐ Not Applicable	22

Organization All None

- ☑ AMPLab, UC Berkeley
- ☑ BGI
- ☑ BioReference Labora...
- ☑ Brazilian Initiative on ...
- ☑ BRCA Exchange
- ☑ Broad Institute
- ☑ Centre for Genomic R...
- ☑ Centro Nacional de A...
- ☑ Curoverse
- ☑ EMBL European Bioi...
- ☑ Global Alliance for G...
- ☑ Google
- ☑ Institute for Systems ...
- ☑ Instituto Nacional de ...

Beacon	Hosted by	Response
BioReference	Hosted by BioReference Laboratories	Found
Catalogue of Somatic Mutations in Cancer	Hosted by Wellcome Trust Sanger Institute	Found
Cell Lines	Hosted by Wellcome Trust Sanger Institute	Found
Conglomerate	Hosted by Global Alliance for Genomics and Health	Found
COSMIC	Hosted by Wellcome Trust Sanger Institute	Found
dbGaP: Combined GRU Catalog and NHLBI Exome Seq...		Found

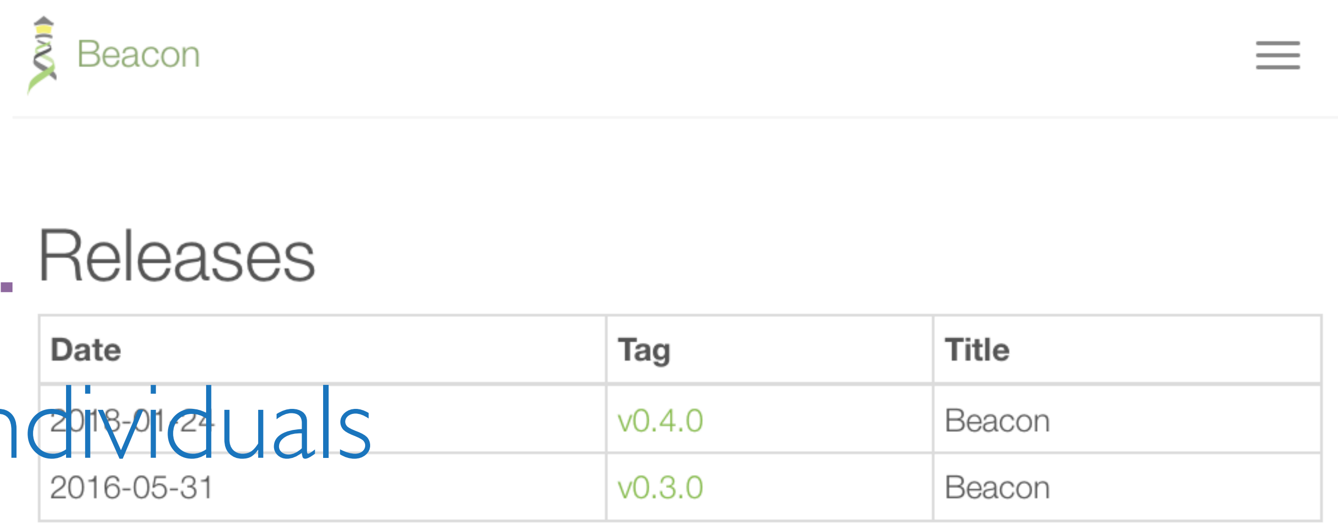


35+ Organizations

90+ Beacons

200+ Datasets

100K Individuals

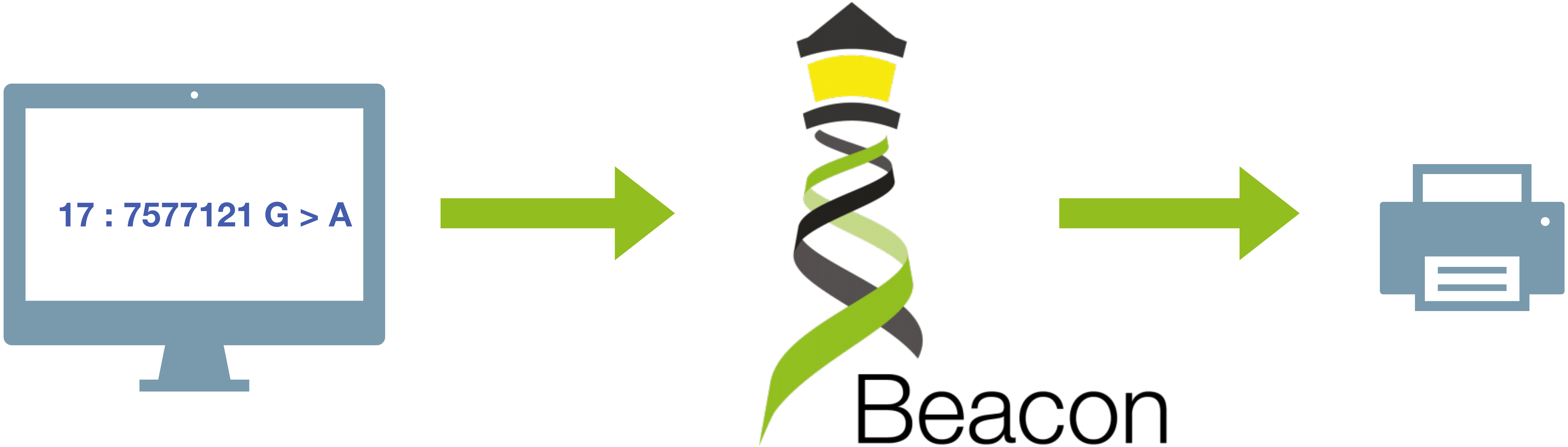


Beacon

Releases

Date	Tag	Title
2018-01-24	v0.4.0	Beacon
2016-05-31	v0.3.0	Beacon

Global Alliance for Genomics & Health



17 : 7577121 G > A

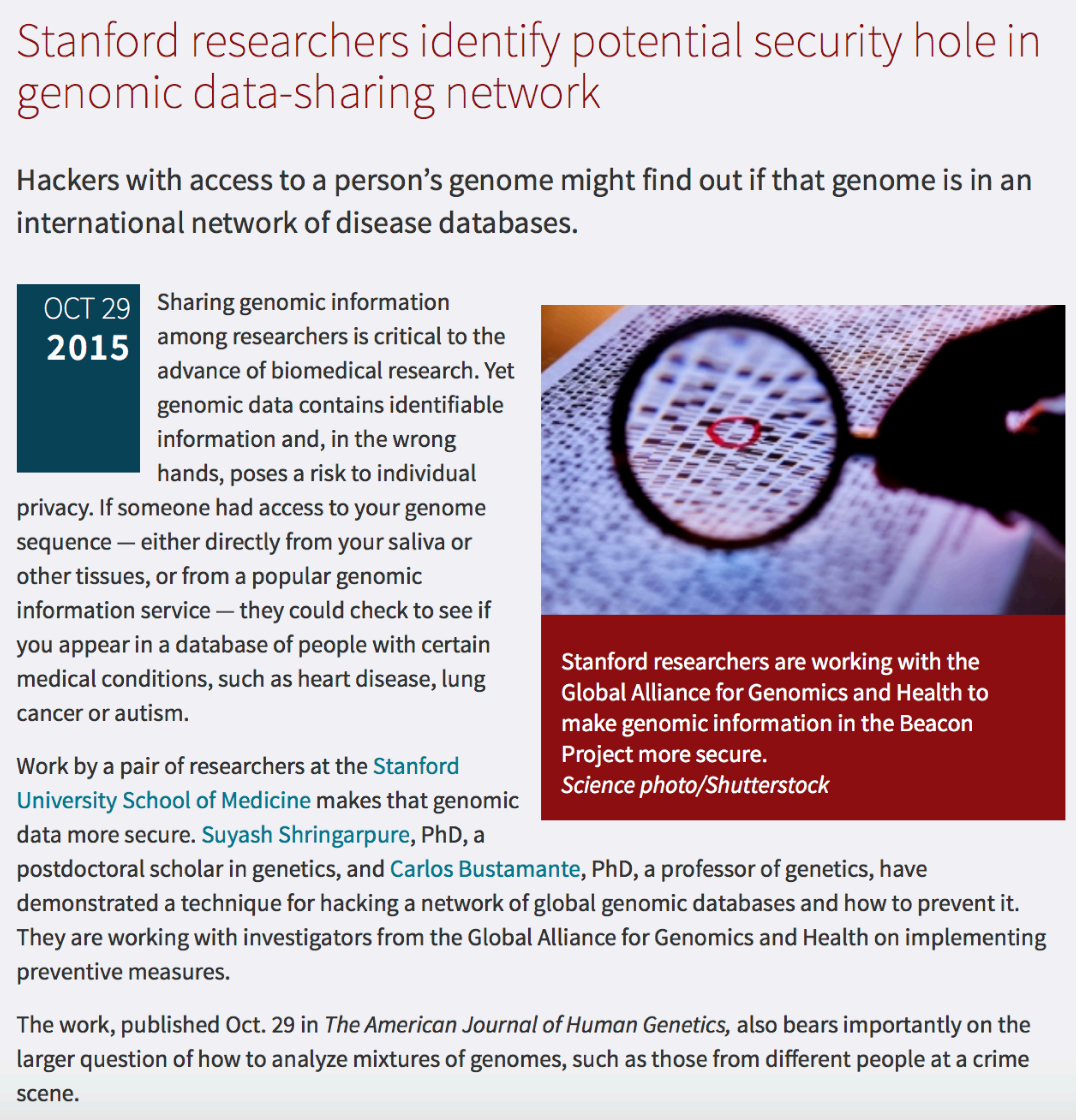
Beacon

A ***Beacon*** answers a query for a specific genome variant against individual or aggregate genome collections

YES | **NO** | **\0**

Genome *Beacons* Compromise Security?

Querying for thousands of specific SNV occurrences in a genomic data pool can identify individuals in an anonymized genomic data collection



Stanford researchers identify potential security hole in genomic data-sharing network

Hackers with access to a person's genome might find out if that genome is in an international network of disease databases.

OCT 29
2015

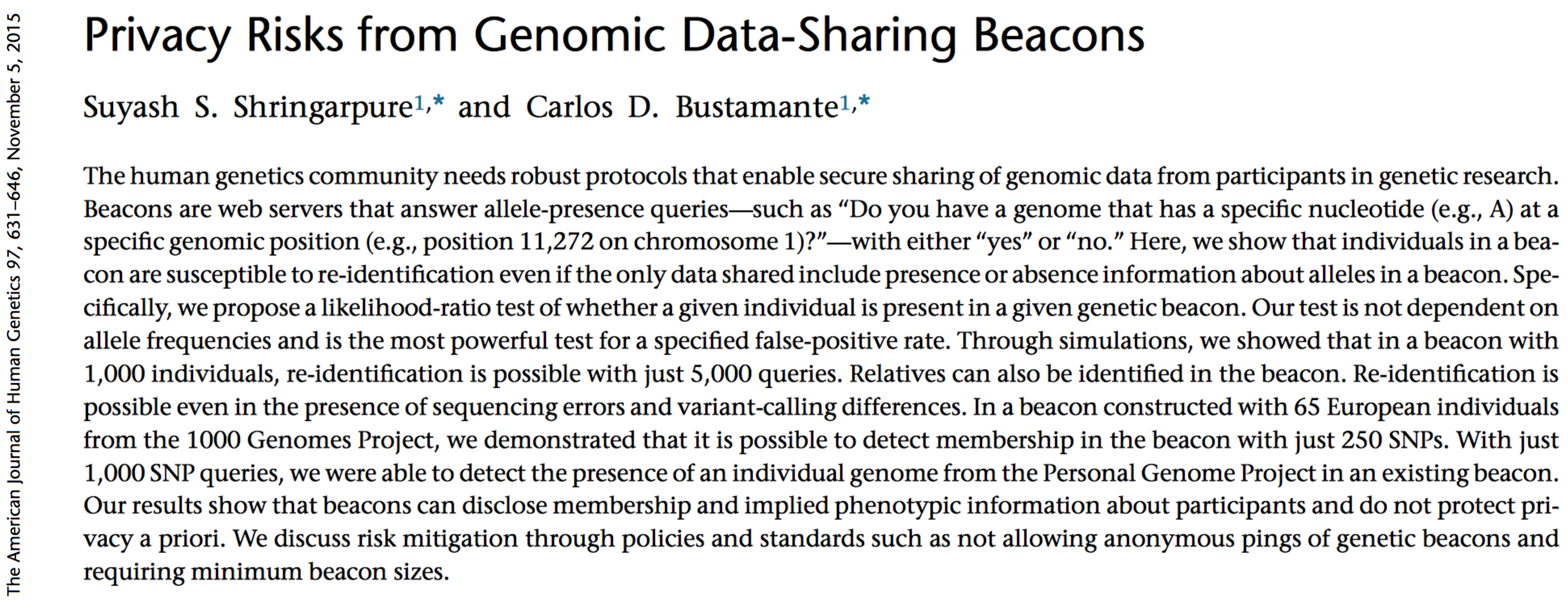
Sharing genomic information among researchers is critical to the advance of biomedical research. Yet genomic data contains identifiable information and, in the wrong hands, poses a risk to individual privacy. If someone had access to your genome sequence — either directly from your saliva or other tissues, or from a popular genomic information service — they could check to see if you appear in a database of people with certain medical conditions, such as heart disease, lung cancer or autism.

Stanford researchers are working with the Global Alliance for Genomics and Health to make genomic information in the Beacon Project more secure.
Science photo/Shutterstock

Work by a pair of researchers at the Stanford University School of Medicine makes that genomic data more secure. Suyash Shringarpure, PhD, a postdoctoral scholar in genetics, and Carlos Bustamante, PhD, a professor of genetics, have demonstrated a technique for hacking a network of global genomic databases and how to prevent it. They are working with investigators from the Global Alliance for Genomics and Health on implementing preventive measures.

The work, published Oct. 29 in *The American Journal of Human Genetics,* also bears importantly on the larger question of how to analyze mixtures of genomes, such as those from different people at a crime scene.

IDENTIFICATION OF INDIVIDUALS FROM MIXED COLLECTIONS USING RARE ALLELES



The American Journal of Human Genetics 97, 631–646, November 5, 2015

Privacy Risks from Genomic Data-Sharing Beacons

Suyash S. Shringarpure[1,*] and Carlos D. Bustamante[1,*]

The human genetics community needs robust protocols that enable secure sharing of genomic data from participants in genetic research. Beacons are web servers that answer allele-presence queries—such as "Do you have a genome that has a specific nucleotide (e.g., A) at a specific genomic position (e.g., position 11,272 on chromosome 1)?"—with either "yes" or "no." Here, we show that individuals in a beacon are susceptible to re-identification even if the only data shared include presence or absence information about alleles in a beacon. Specifically, we propose a likelihood-ratio test of whether a given individual is present in a given genetic beacon. Our test is not dependent on allele frequencies and is the most powerful test for a specified false-positive rate. Through simulations, we showed that in a beacon with 1,000 individuals, re-identification is possible with just 5,000 queries. Relatives can also be identified in the beacon. Re-identification is possible even in the presence of sequencing errors and variant-calling differences. In a beacon constructed with 65 European individuals from the 1000 Genomes Project, we demonstrated that it is possible to detect membership in the beacon with just 250 SNPs. With just 1,000 SNP queries, we were able to detect the presence of an individual genome from the Personal Genome Project in an existing beacon. Our results show that beacons can disclose membership and implied phenotypic information about participants and do not protect privacy a priori. We discuss risk mitigation through policies and standards such as not allowing anonymous pings of genetic beacons and requiring minimum beacon sizes.

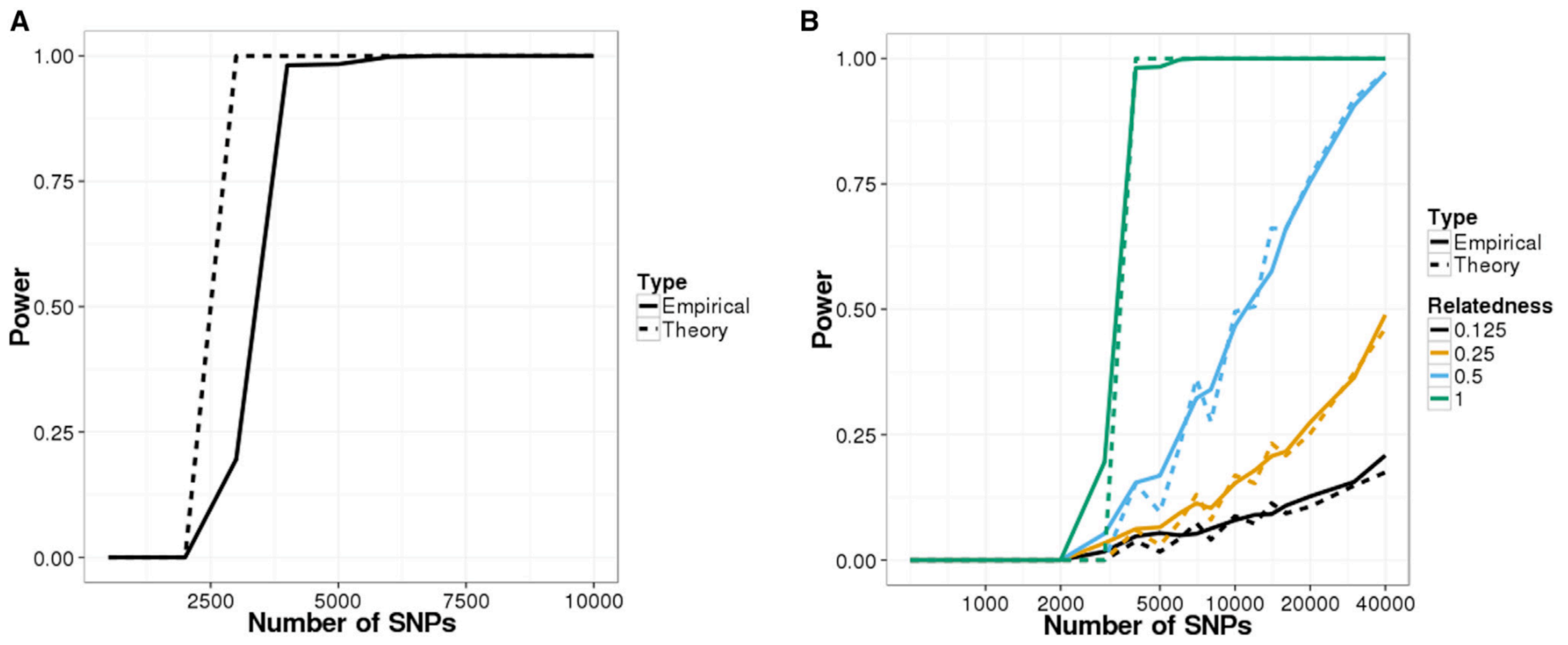
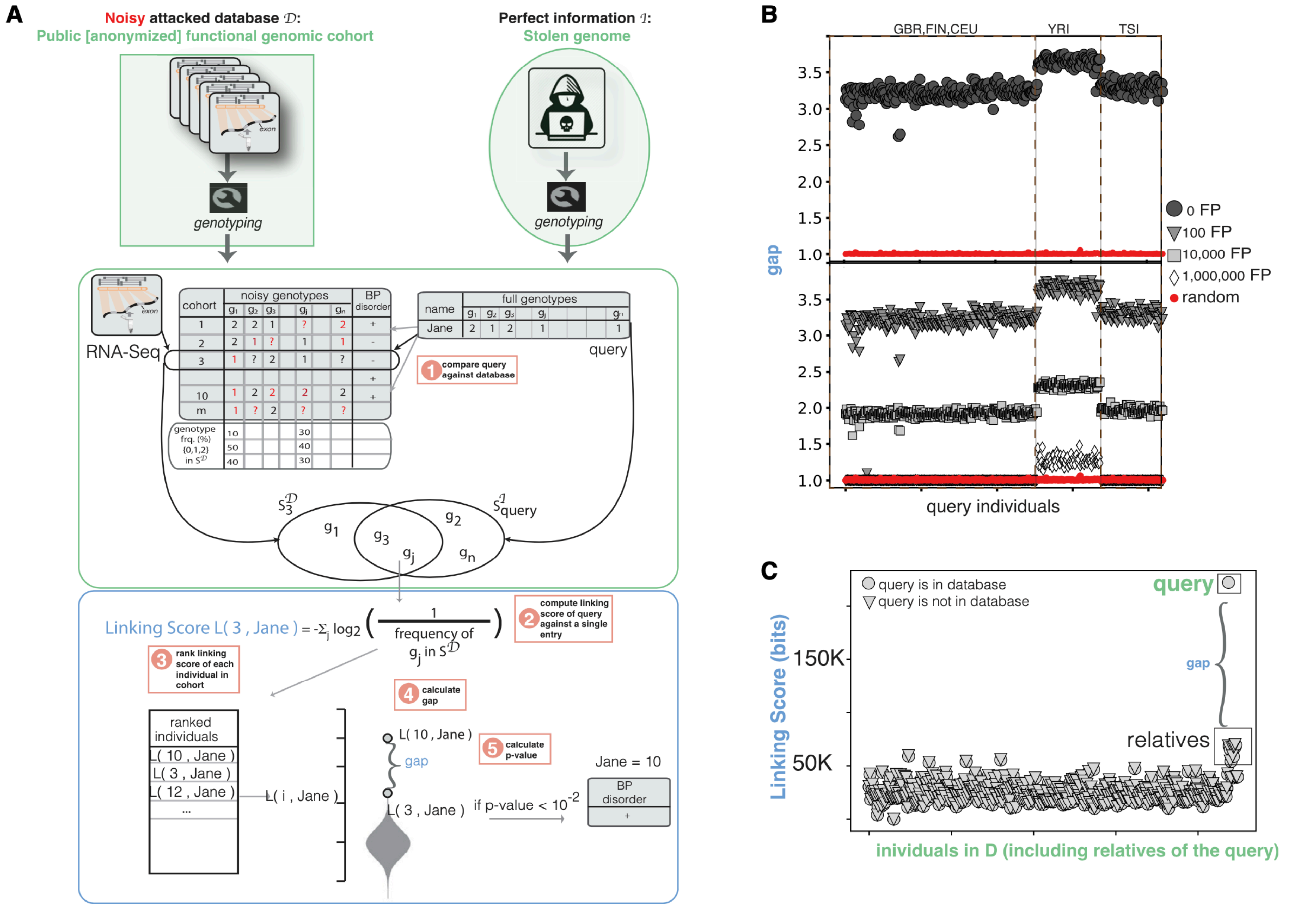


Figure 1. Power of Re-identification Attacks on Beacons Constructed with Simulated Data
Power curves for the likelihood-ratio test (LRT) on (A) a simulated beacon with 1,000 individuals and (B) detecting relatives in the simulated beacon. The false-positive rate was set to 0.05 for all scenarios.

- rare allelic variants can be used to identify an individual (or her relatives) in a genome collection without having access to individual datasets
- however, such an approach requires previous knowledge about the individual's SNPs

Information Leakage from Functional Genomics Data

- many research studies contain "functional" genomics data, e.g. from expression analyses
- such (anonymized) data may have lower protection levels than data from dedicated genotyping studies
- with a non-noisy genome of interest, attackers can generate linkage scores to identify the best match to the genomic profile



cohort	noisy genotypes g_1	g_2	g_3		g_j		g_n	BP disorder
1	2	2	1		?		2	+
2	2	1	?		1		1	-
3	1	?	2		1		?	-
								+
10	1	2	2		2		2	+
m	1	?	2		?		?	
genotype frq. (%) {0,1,2} in $S^{\mathcal{D}}$	10 50 40				30 40 30			

name	full genotypes g_1	g_2	g_3		g_j			g_n
Jane	2	1	2		1			1

Linking Score L(3 , Jane) = $-\Sigma_j \log2 \left(\frac{1}{\text{frequency of } g_j \text{ in } S^{\mathcal{D}}} \right)$

ranked individuals
L(10 , Jane)
L(3 , Jane)
L(12 , Jane)
...

Figure 1. Functional Genomics Data De-anonymization Scheme with Perfect Genomes

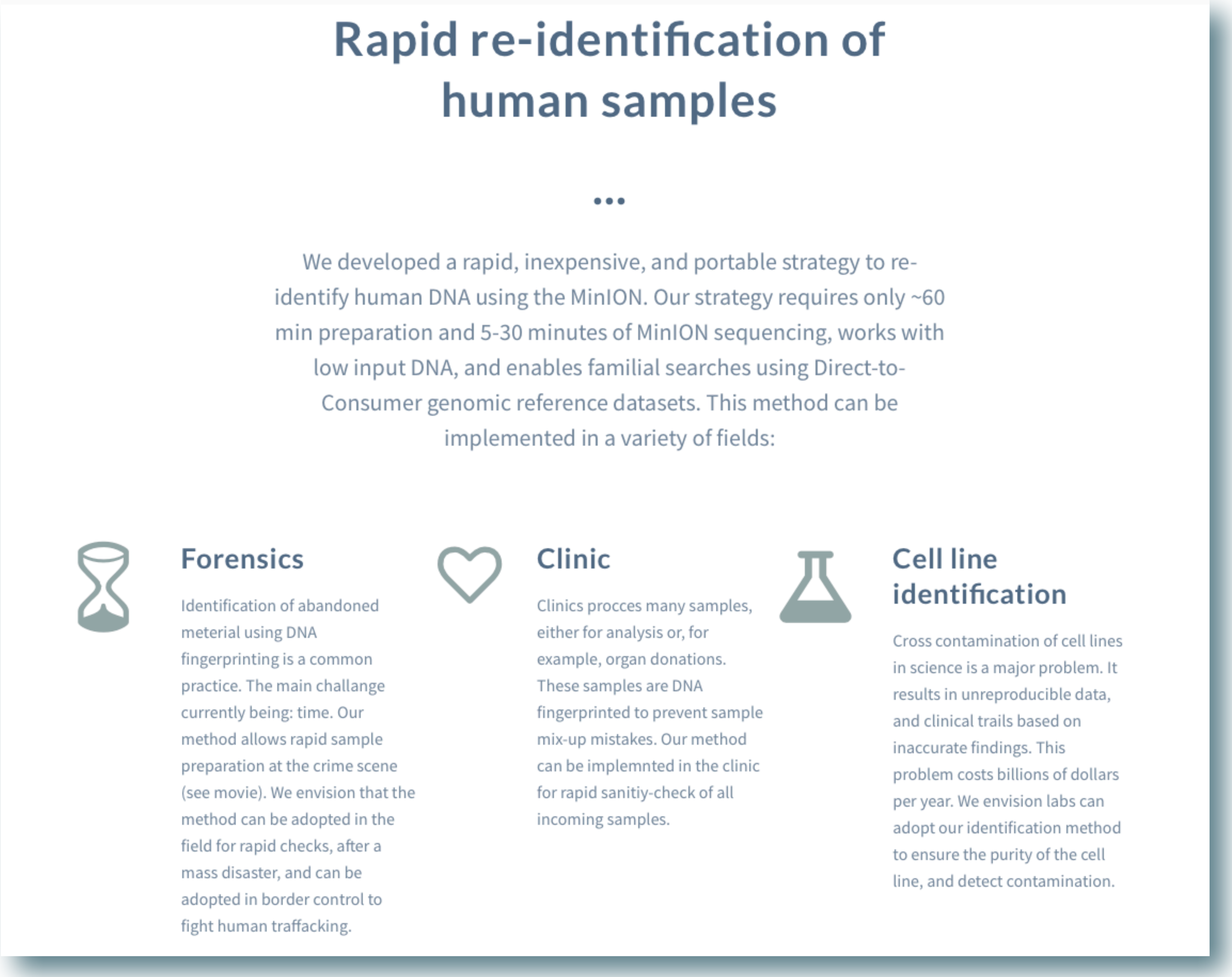
(A) Anonymized functional genomics data from a cohort of individuals can be seen as a database *D* to be attacked, which contains functional genomics reads and phenotypes for every individual in the cohort. The perfect information *I* about an individual can be the genome of an individual. After obtaining genotypes from the functional genomics reads, the attacker scores each individual in the cohort based on the overlapping genotypes between the known individual's genome and the noisy genotypes called from functional genomics. These scores are then ranked and the top-ranked individual in the cohort is selected as the known individual. See also Figure S1.

(B) *gap* values for the 1000 Genomes Project individuals in the gEUVADIS RNA-seq cohort. Red circles are the *gap* values obtained by linking a random set of genotypes to the RNA-seq panel. *gap* values are also shown after adding false-positive genotypes to the genotype set of each individual in the database.

(C) The linking scores for each individual in the functional genomics cohort after the addition of genetically related individuals to the query, with and without the query individual present in the database.

Gürsoy et al., 2020, Cell *183*, 905–917
November 12, 2020 © 2020 Elsevier Inc.
https://doi.org/10.1016/j.cell.2020.09.036

But genotyping itself is for professional labs, right?



Rapid re-identification of human samples

...

We developed a rapid, inexpensive, and portable strategy to re-identify human DNA using the MinION. Our strategy requires only ~60 min preparation and 5-30 minutes of MinION sequencing, works with low input DNA, and enables familial searches using Direct-to-Consumer genomic reference datasets. This method can be implemented in a variety of fields:

Forensics

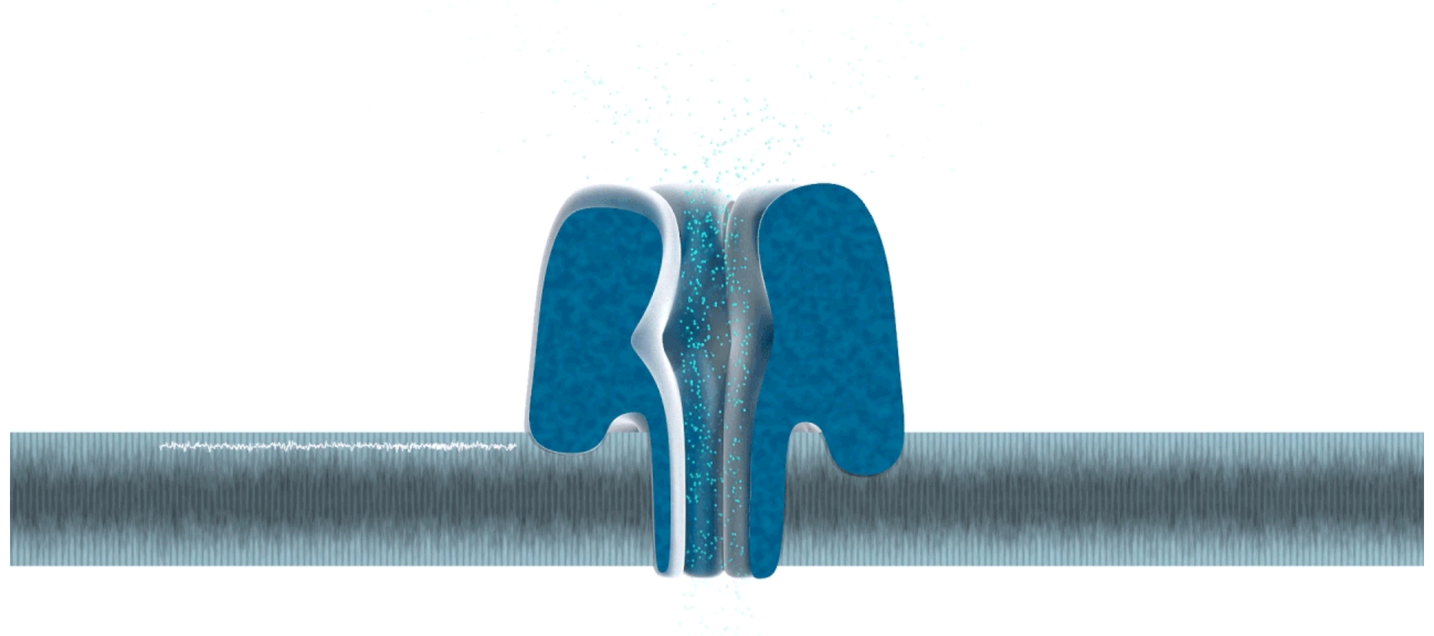
Identification of abandoned meterial using DNA fingerprinting is a common practice. The main challange currently being: time. Our method allows rapid sample preparation at the crime scene (see movie). We envision that the method can be adopted in the field for rapid checks, after a mass disaster, and can be adopted in border control to fight human traffacking.

Clinic

Clinics procces many samples, either for analysis or, for example, organ donations. These samples are DNA fingerprinted to prevent sample mix-up mistakes. Our method can be implemnted in the clinic for rapid sanitiy-check of all incoming samples.

Cell line identification

Cross contamination of cell lines in science is a major problem. It results in unreproducible data, and clinical trails based on inaccurate findings. This problem costs billions of dollars per year. We envision labs can adopt our identification method to ensure the purity of the cell line, and detect contamination.

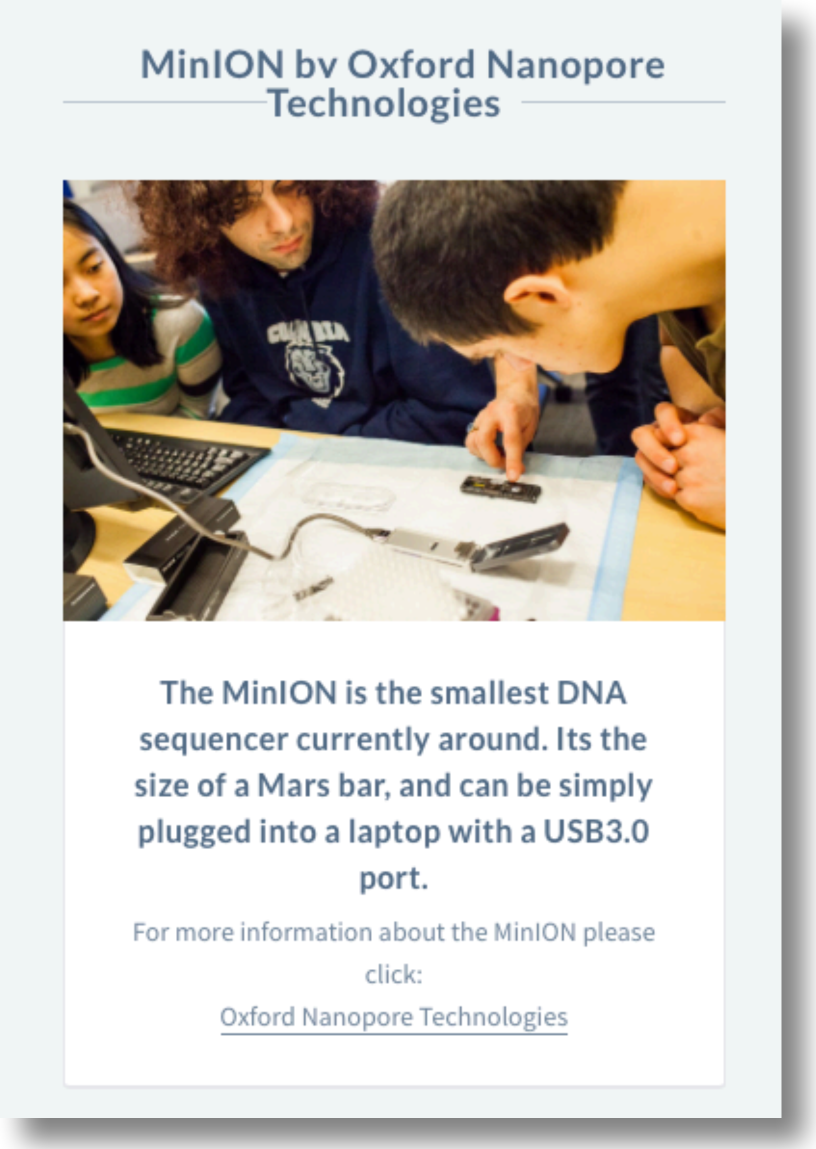


The MinION (Oxford Nanopore)
Source: Sophie Zaaijer
https://medium.com/neodotlife/nanopore-6443c81d76d3

DEMOCRATIZING DNA FINGERPRINTING

Sophie Zaaijer, Assaf Gordon, Robert Piccone, Daniel Speyer, Yaniv Erlich, 2016

ddf.teamerlich.org

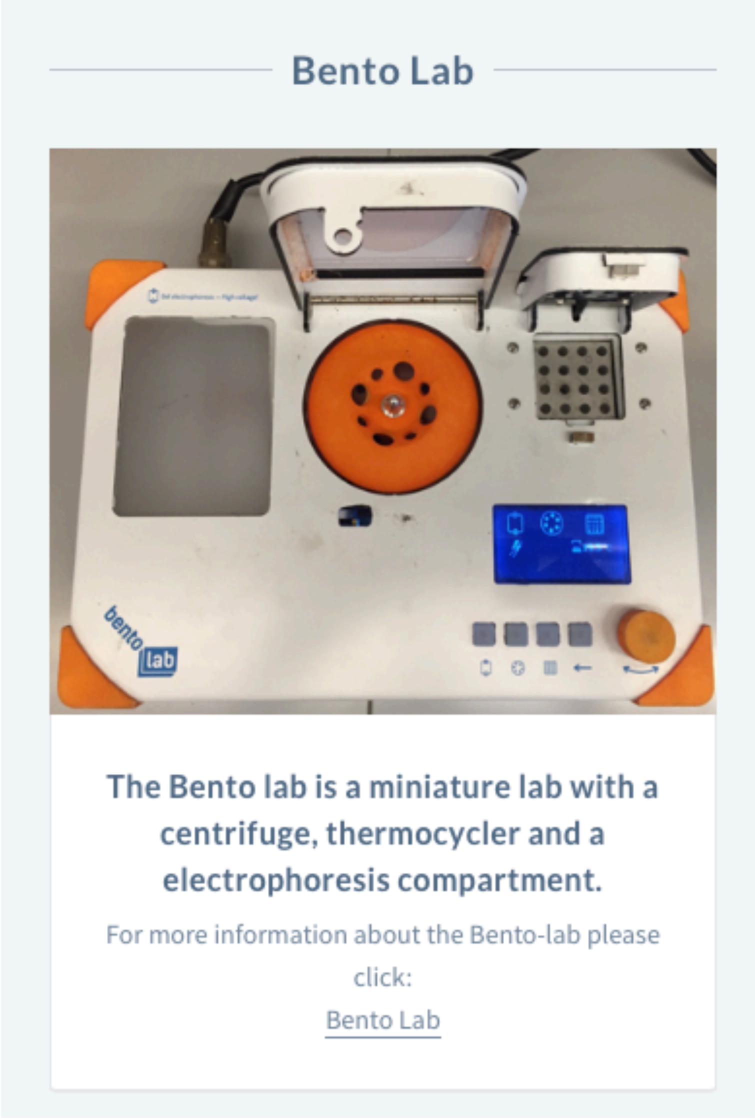


MinION by Oxford Nanopore Technologies

The MinION is the smallest DNA sequencer currently around. Its the size of a Mars bar, and can be simply plugged into a laptop with a USB3.0 port.

For more information about the MinION please click:

Oxford Nanopore Technologies

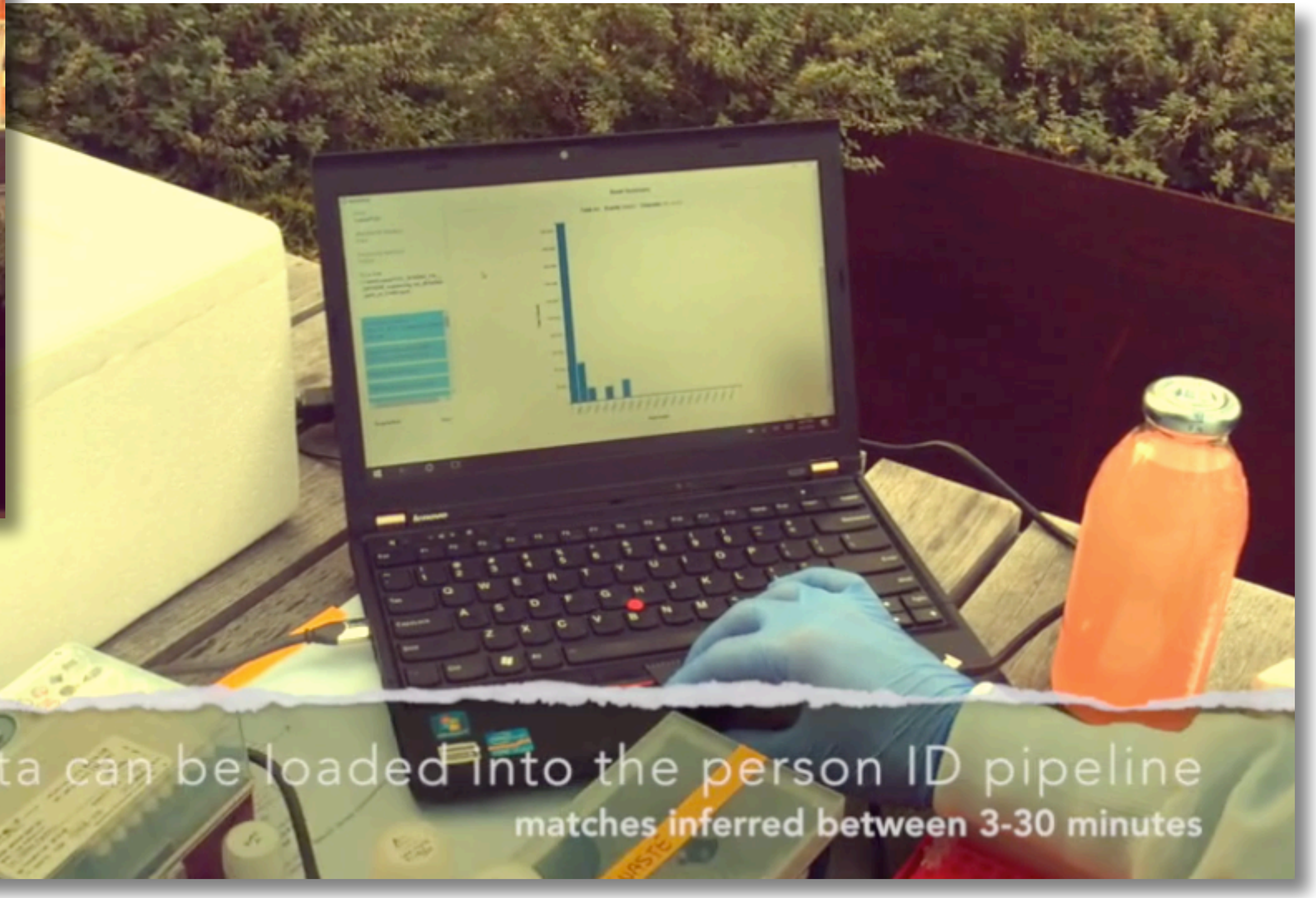


Bento Lab

The Bento lab is a miniature lab with a centrifuge, thermocycler and a electrophoresis compartment.

For more information about the Bento-lab please click:

Bento Lab



Data can be loaded into the person ID pipeline

matches inferred between 3-30 minutes

DNA sequencing for identification/fingerprinting soon "commodity" technology (in contrast with technological/data challenges in "precision medicine")

Typical Data Scopes in Genomics (Research) Collections

Biomedical and procedural "Meta"data types

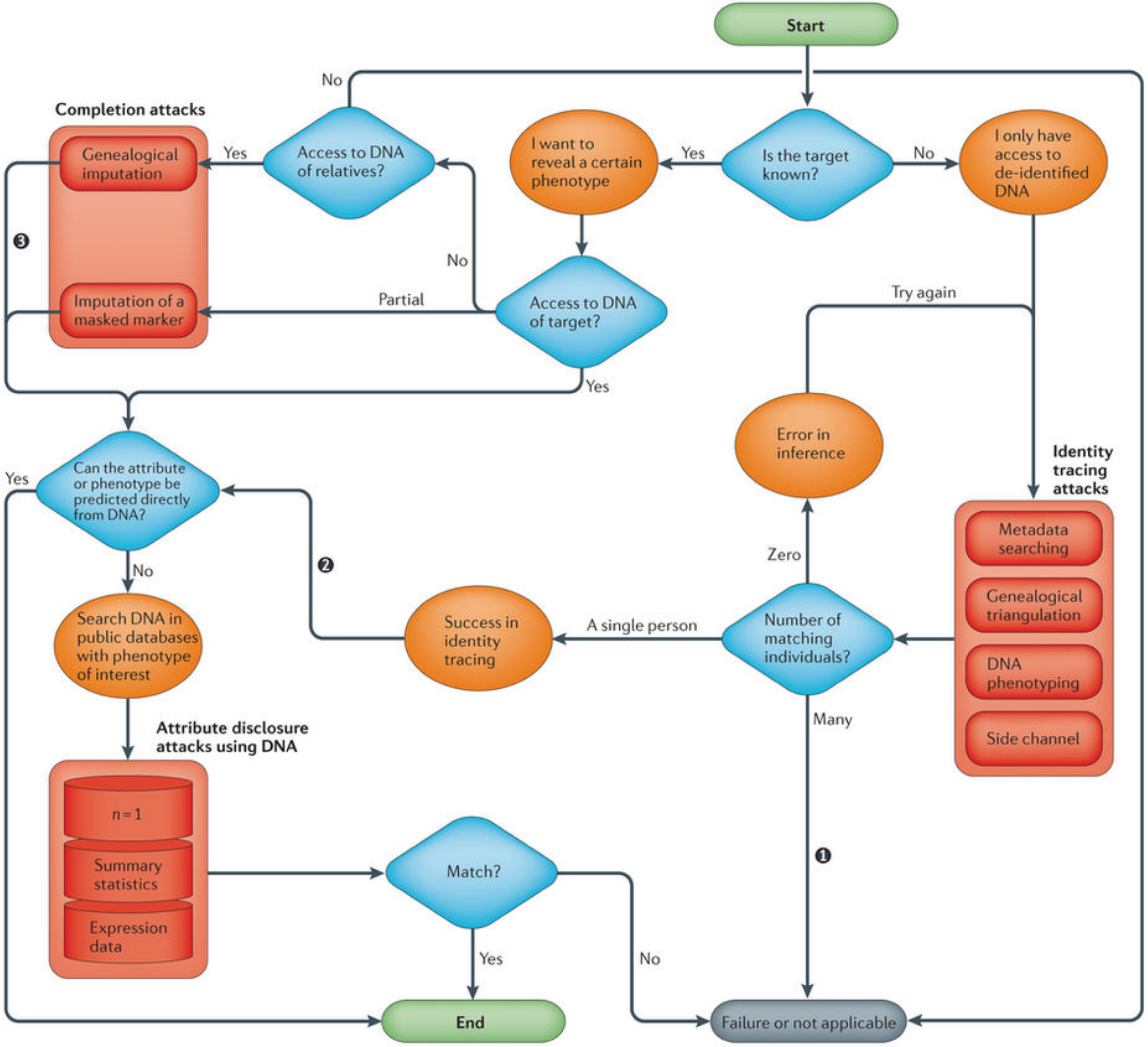
- Diagnostic classification
  - mapping text-based cancer diagnoses to standard classification systems
- Provenance data
  - store identifier-based pointers
  - geographic attribution (individual, biosample, experiment)
- Clinical information
  - **core set** of typical cancer study values:
    - ➡ stage, grade, followup time, survival status, genomic sex, age at diagnosis
  - balance between annotation effort and expected usability

Routes for breaching and protecting genetic privacy

The map contrasts different scenarios, such as identifying de-identified genetic data sets, revealing an attribute from genetic data and unmasking of data. It also shows the interdependencies between the techniques and suggests potential routes to exploit further information after the completion of one attack. There are several simplifying assumptions (black circles).

In certain scenarios (such as insurance decisions), uncertainty about the target's identity within a small group of people could still be considered a success (assumption 1). For certain privacy harms (such as surveillance), identity tracing can be considered a success and the end point of the process (assumption 2). The complete DNA sequence is not always necessary (assumption 3).

Yaniv Erlich & Arvind Narayanan. Nature Reviews Genetics 15, 409–421 (2014)



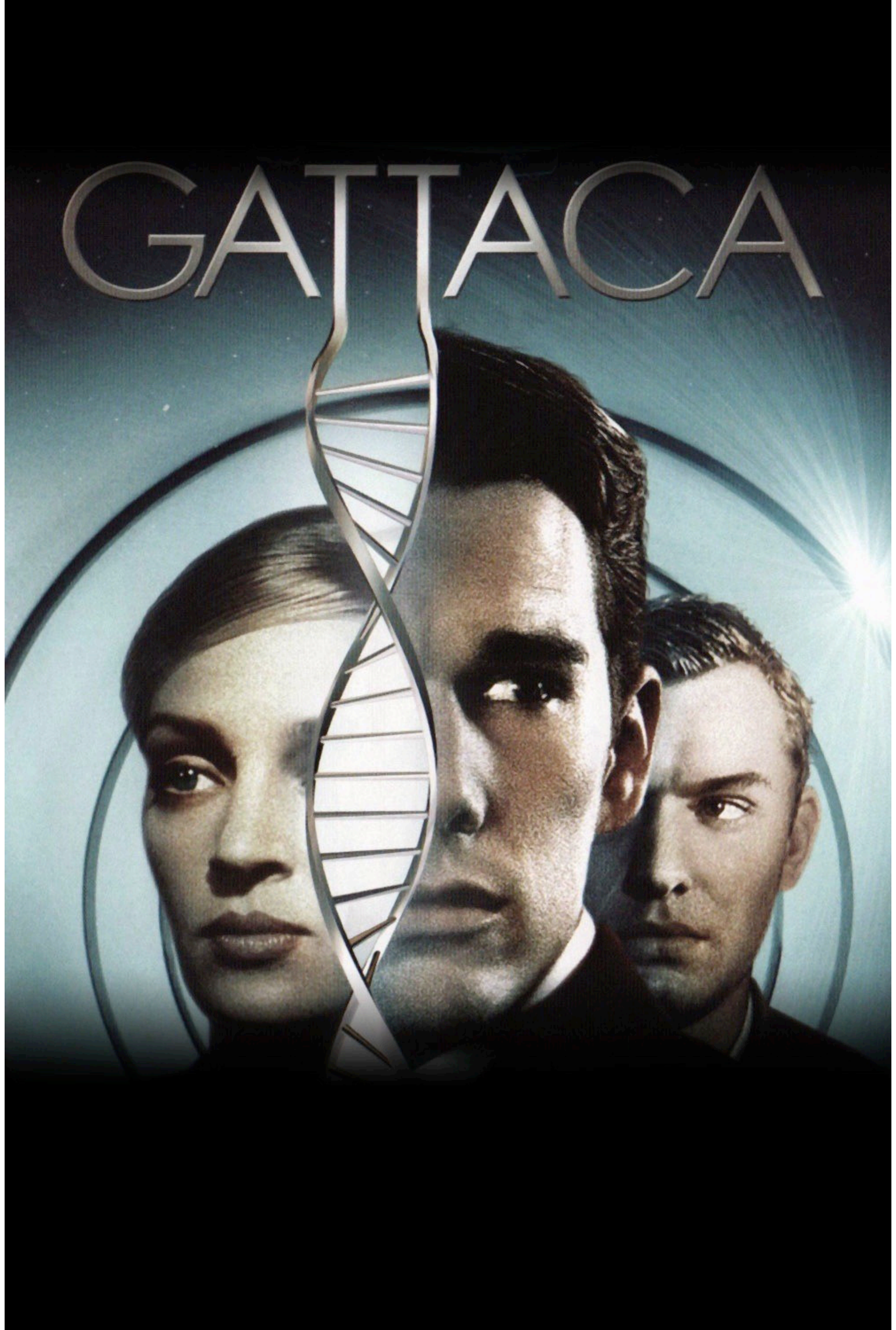
Start
Is the target known?
Yes
No
I want to reveal a certain phenotype
I only have access to de-identified DNA
Access to DNA of relatives?
No
Yes
Completion attacks
Genealogical imputation
Imputation of a masked marker
3
Access to DNA of target?
No
Partial
Yes
Try again
Error in inference
Identity tracing attacks
Metadata searching
Genealogical triangulation
DNA phenotyping
Side channel
Zero
Number of matching individuals?
A single person
Many
Success in identity tracing
2
Can the attribute or phenotype be predicted directly from DNA?
Yes
No
Search DNA in public databases with phenotype of interest
Attribute disclosure attacks using DNA
n = 1
Summary statistics
Expression data
Match?
Yes
No
1
End
Failure or not applicable

Nature Reviews | Genetics

“We’re an information economy. They teach you that in school. What they don’t tell you is that it’s impossible to move, to live, to operate at any level without leaving traces, bits, seemingly meaningless fragments of personal information. Fragments that can be retrieved, amplified . . .”

–William Gibson in "Johnny Mnemonic" (1986)

Genomes
Privacy
Society

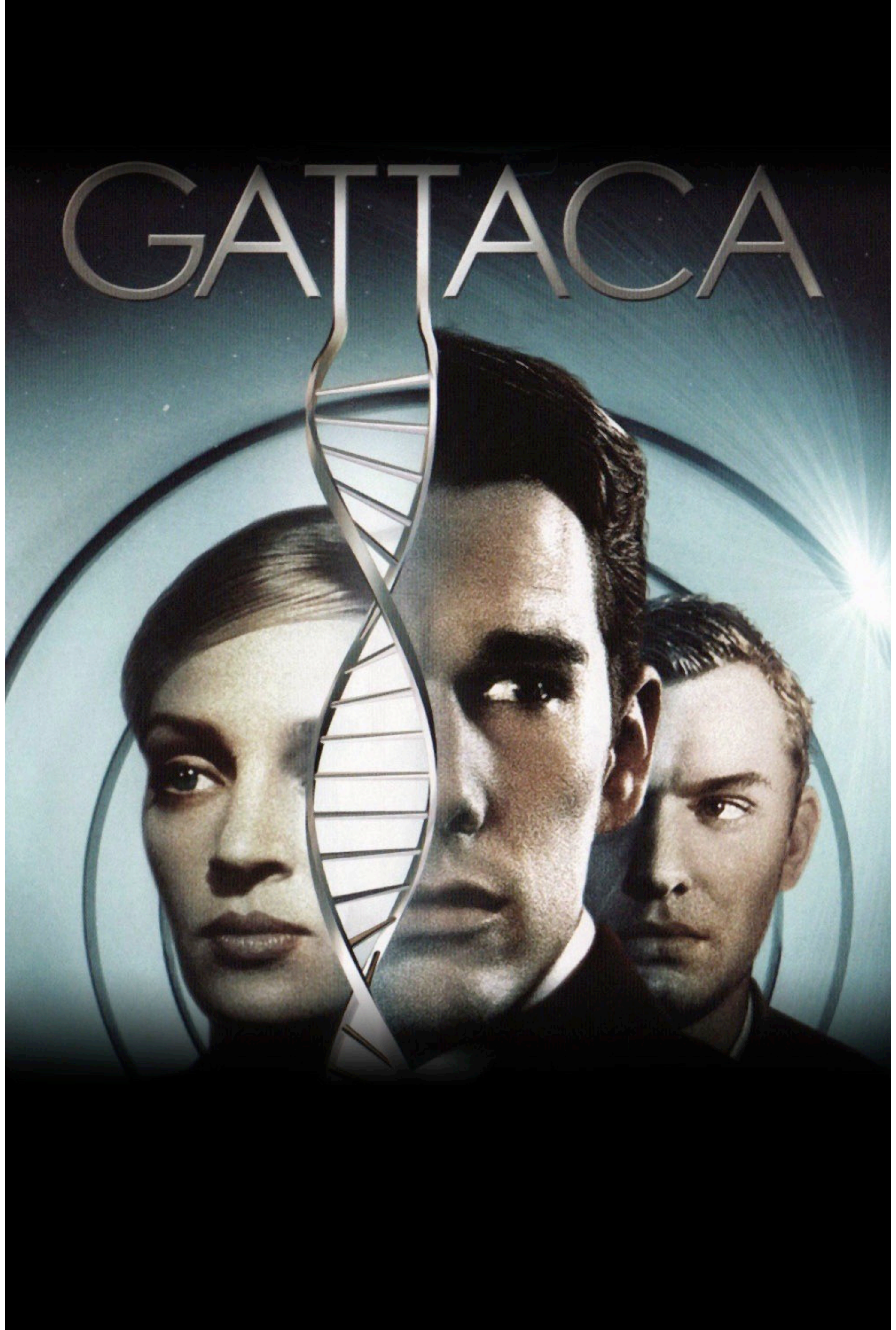


Gattaca (1997)

A genetically inferior man assumes the identity of a superior one in order to pursue his lifelong dream of space travel.

- genetic determinism
  - main character has been determined to be unsuitable for complex jobs based on genetic analysis
- genetic identification
  - the use of genetic sampling for personal identification is daily routine

With information from https://www.imdb.com/title/tt0119177/



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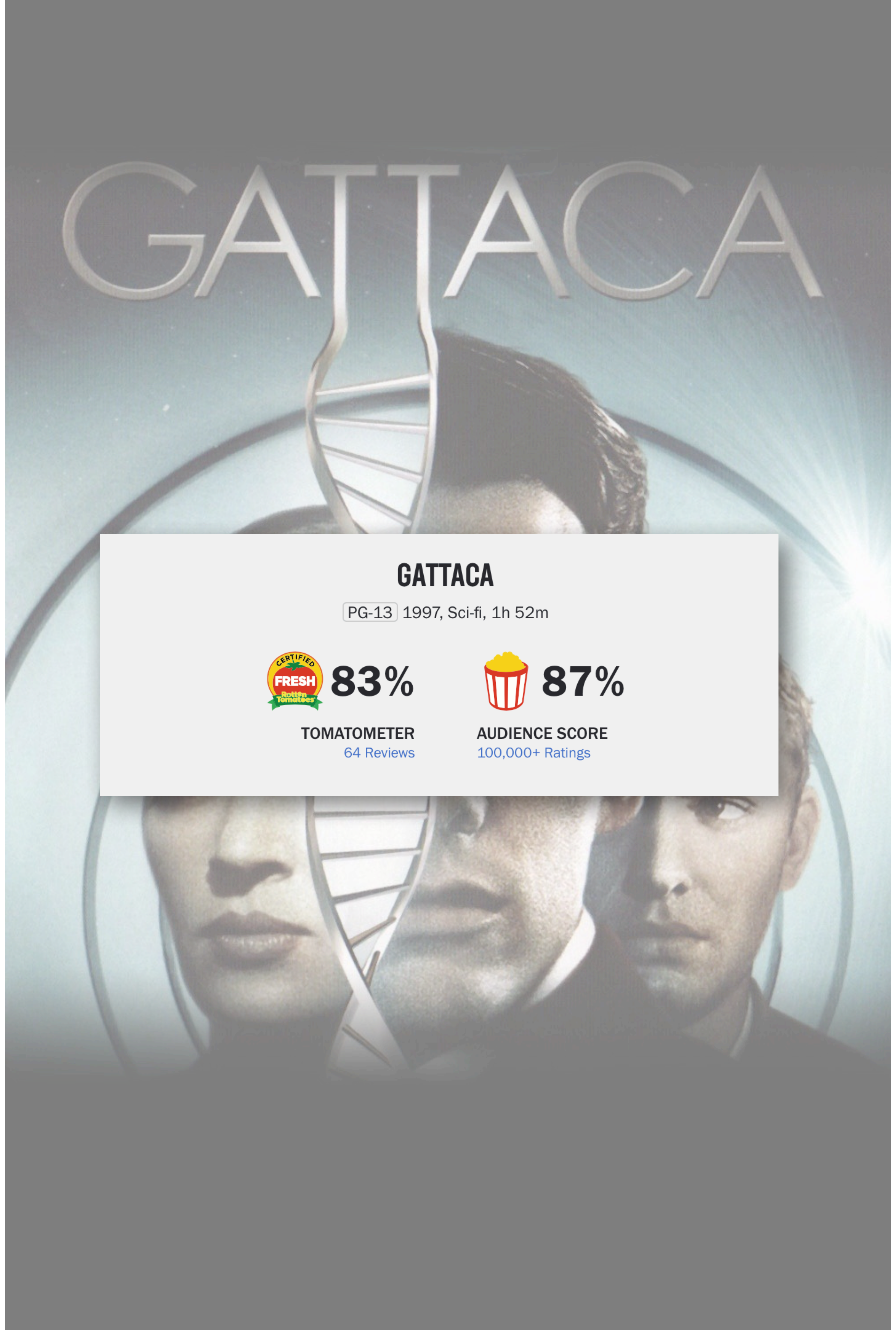


GATTACA

PG-13 1997, Sci-fi, 1h 52m

83% TOMATOMETER 64 Reviews

87% AUDIENCE SCORE 100,000+ Ratings

DTC Genomics

Direct-to-Consumer Genomic Testing

- family ancestry or genealogy
  - >7Mill customers in 2018 at ancestry.com
- DNA-based health & traits information
  - disease risk
  - carrier status
  - lifestyle information
- participation in large cohort studies

ancestry

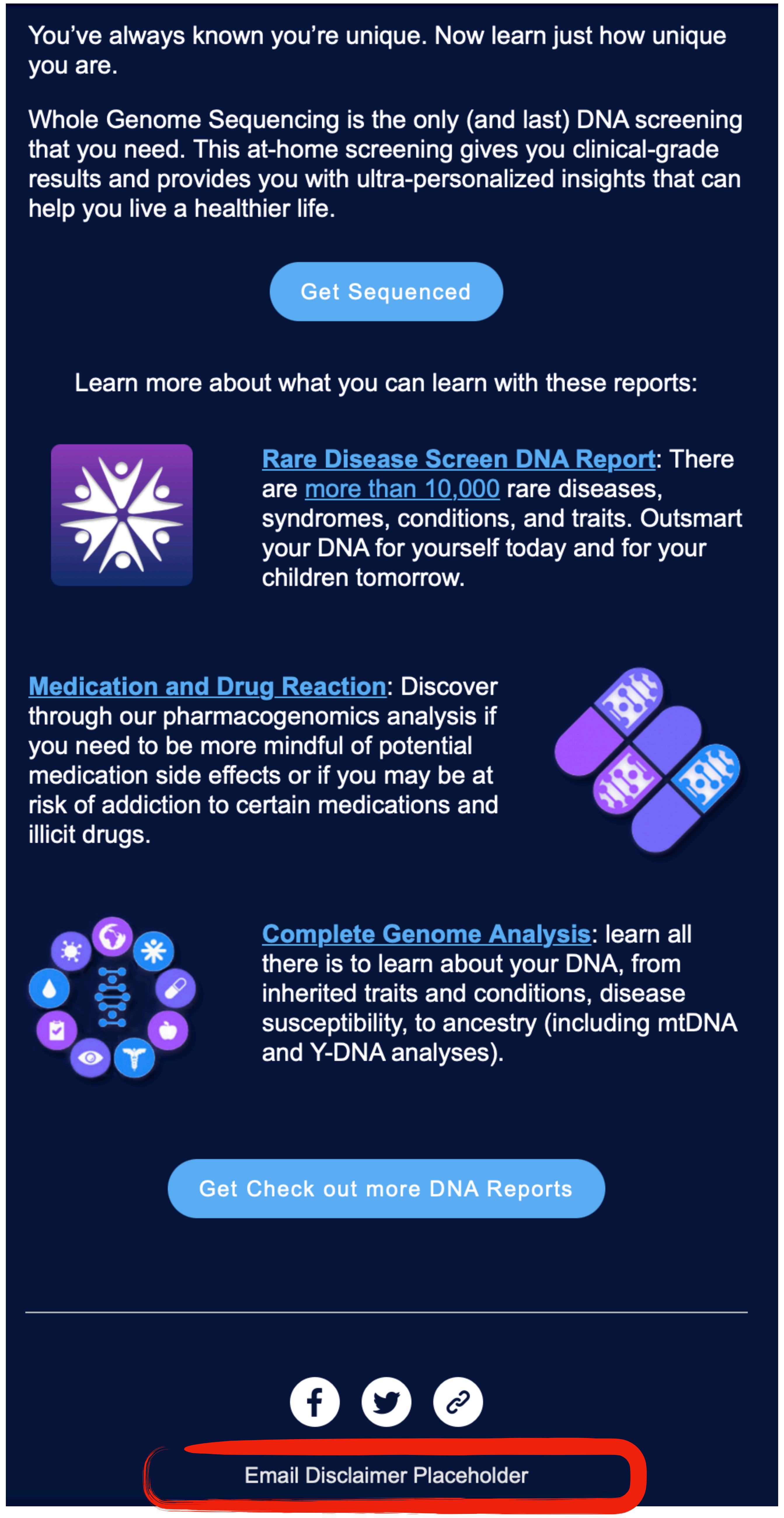
23andMe®

MyHeritage

Think Before You Spit

https://blogs.cdc.gov/genomics/2017/04/18/direct-to-consumer-2/

DTC Genomics



You've always known you're unique. Now learn just how unique you are.

Whole Genome Sequencing is the only (and last) DNA screening that you need. This at-home screening gives you clinical-grade results and provides you with ultra-personalized insights that can help you live a healthier life.

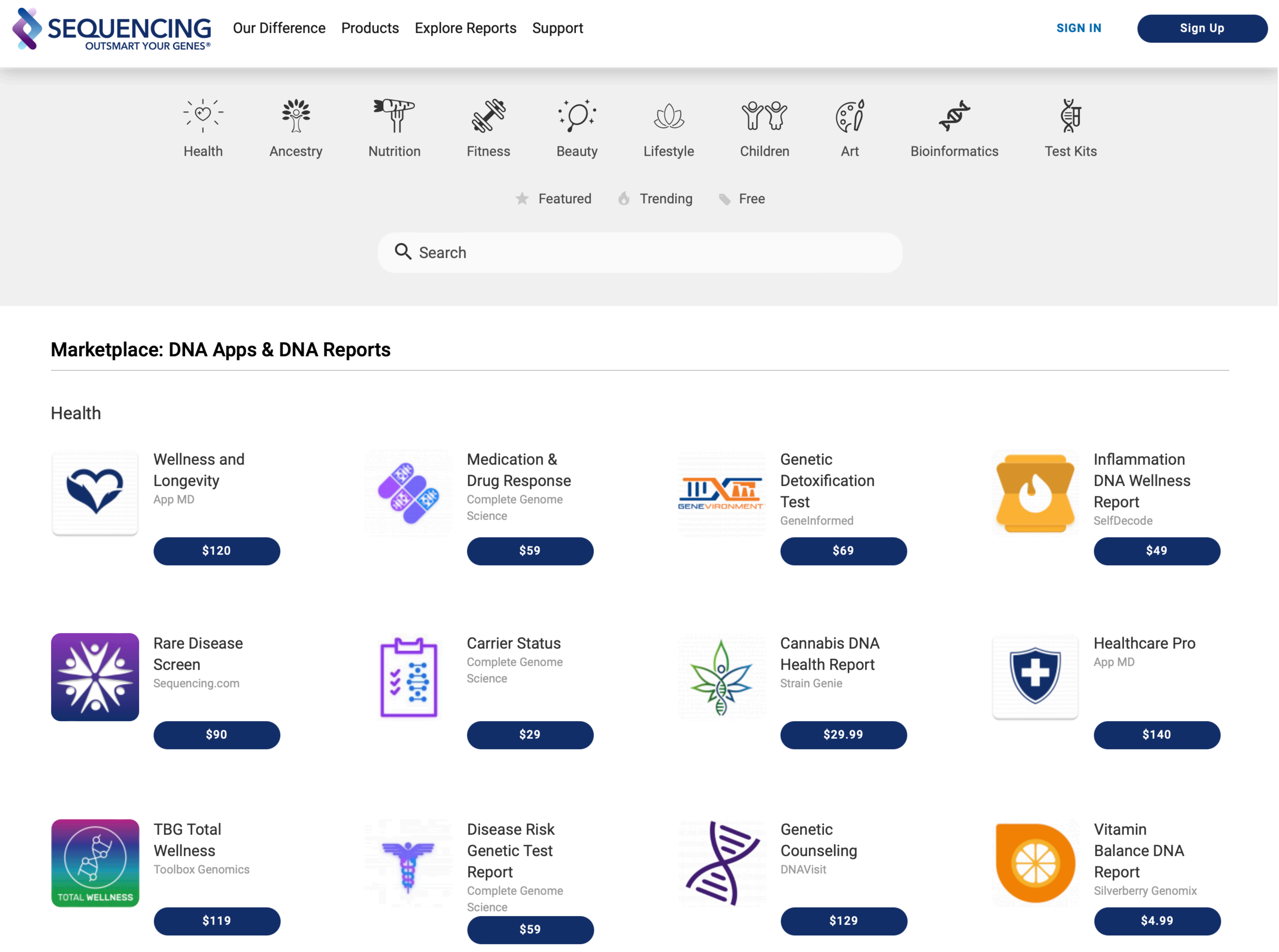
Learn more about what you can learn with these reports:

Rare Disease Screen DNA Report: There are more than 10,000 rare diseases, syndromes, conditions, and traits. Outsmart your DNA for yourself today and for your children tomorrow.

Medication and Drug Reaction: Discover through our pharmacogenomics analysis if you need to be more mindful of potential medication side effects or if you may be at risk of addiction to certain medications and illicit drugs.

Complete Genome Analysis: learn all there is to learn about your DNA, from inherited traits and conditions, disease susceptibility, to ancestry (including mtDNA and Y-DNA analyses).

Email Disclaimer Placeholder



Marketplace: DNA Apps & DNA Reports

Health

Wellness and Longevity
App MD
$120

Medication & Drug Response
Complete Genome Science
$59

Genetic Detoxification Test
GeneInformed
$69

Inflammation DNA Wellness Report
SelfDecode
$49

Rare Disease Screen
Sequencing.com
$90

Carrier Status
Complete Genome Science
$29

Cannabis DNA Health Report
Strain Genie
$29.99

Healthcare Pro
App MD
$140

TBG Total Wellness
Toolbox Genomics
$119

Disease Risk Genetic Test Report
Complete Genome Science
$59

Genetic Counseling
DNAVisit
$129

Vitamin Balance DNA Report
Silverberry Genomix
$4.99

Right to Know?

Dealing with "non-actionable" genomic predictions

- diagnostic and direct to consumer genetic tests may provide risk predictions for disease susceptibility
- most will be non-deterministic, non-actionable, and usually be associated with a very low ***absolute*** risk - but heritable
- understanding such "prognostications" is challenging & potentially fraught with errors - and opens the door to services

VANITY FAIR

EXCLUSIVE

Chris Hemsworth Changed His Life After an Ominous Health Warning

In an exclusive sit-down with *Vanity Fair,* the actor discusses movies, the future of Thor, his businesses, fatherhood, and how a genetic predisposition for Alzheimer's alters everything.

BY ANTHONY BREZNICAN

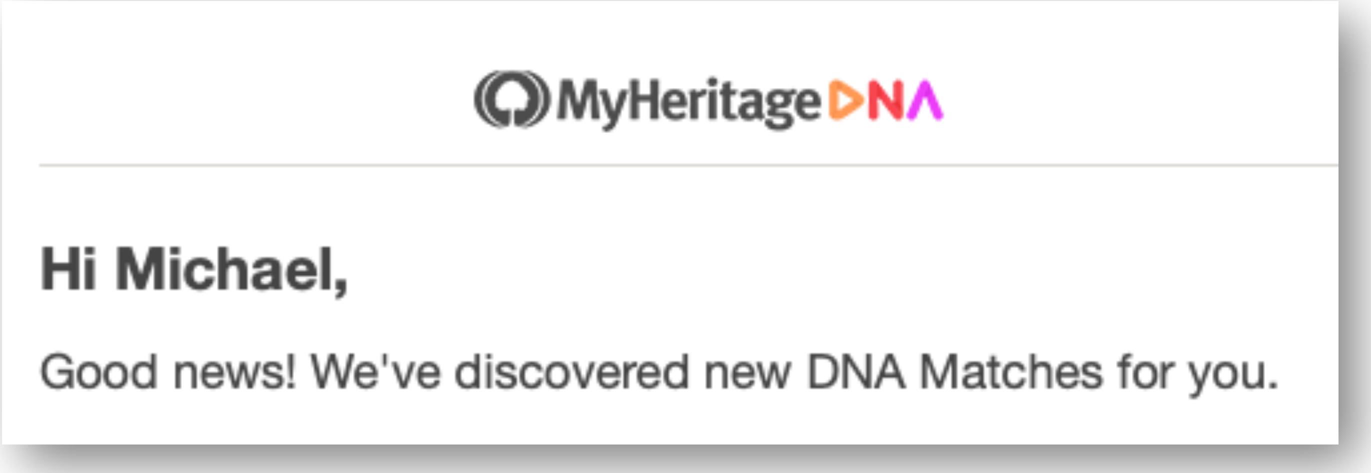
NOVEMBER 17, 2022

...His makeup includes two copies of the gene APOE4, one from his mother, the other from his father, which studies have linked to an increased risk of Alzheimer's disease. One in four people carry a single copy of the gene, but only 2 to 3% of the population have both, according to a 2021 study by the National Institutes of Health.

"For me, the positive of it was like, "Right, if I didn't know this [Alzheimer's] information, I wouldn't have made the changes I made." I just wasn't aware of any of it, so now I feel thankful that I have in my arsenal the sort of tools to best prepare myself and prevent things happening in that way."

vanityfair.com/hollywood/2022/11/chris-hemsworth-exclusive-interview-alzheimers-limitless

Long-Range Familial Searches

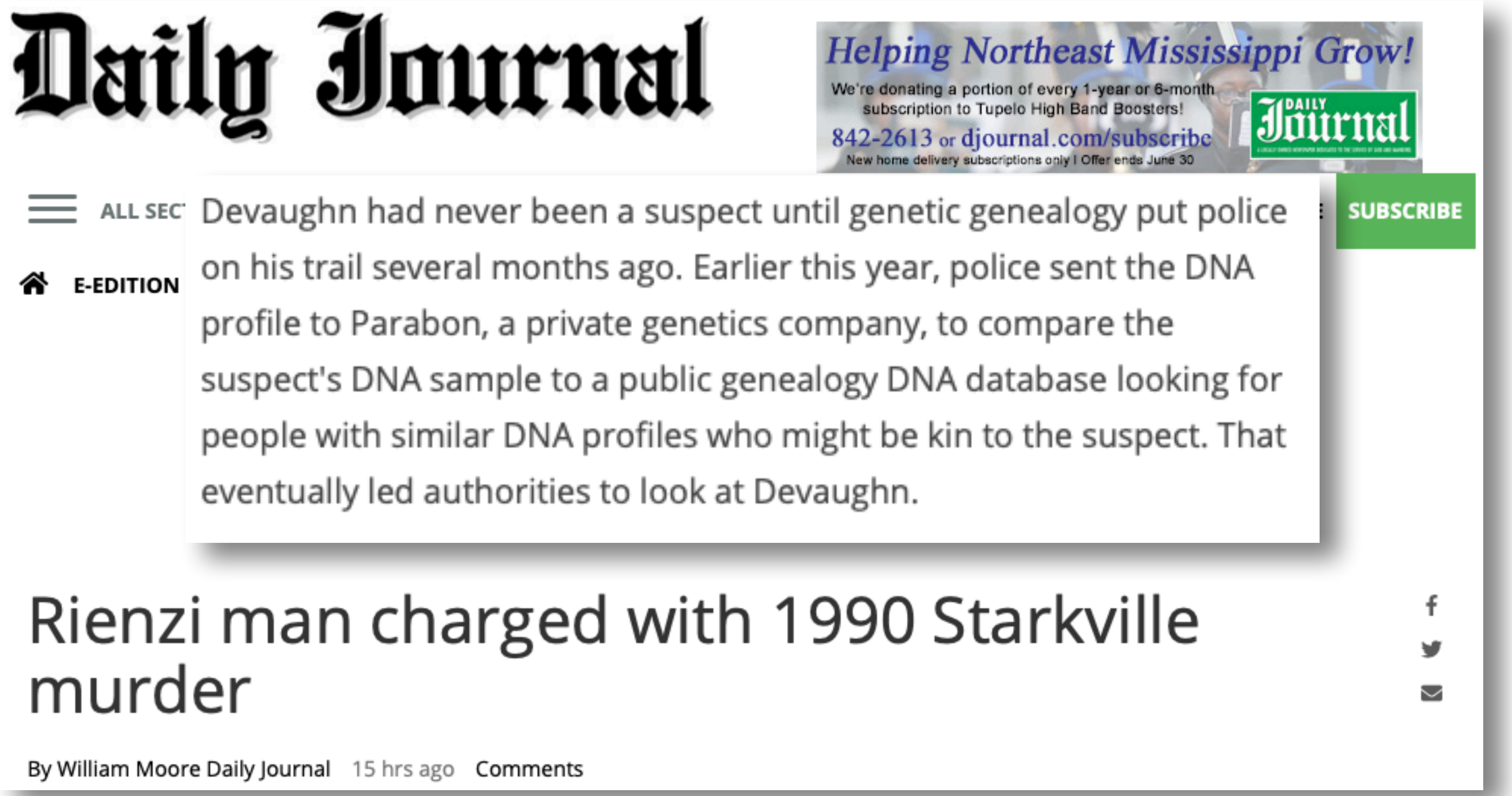


MyHeritage DNA

Hi Michael,

Good news! We've discovered new DNA Matches for you.

- Commercial, "Direct to Customer" DNA analyses are provided through independent sites and such affiliated to genealogy services (MyHeritage, Ancestry.com, 23andMe...)
- Genealogy sites identify individuals with matching haplotype blocks & provide a prediction about degree of genetic relation
- Law enforcement agencies (and who else?!) can send individual SNP profiles (e.g. recovered from evidence many years after a crime) using a *Jane Doe* identity, to identify relatives of the suspect - **long range familial search**



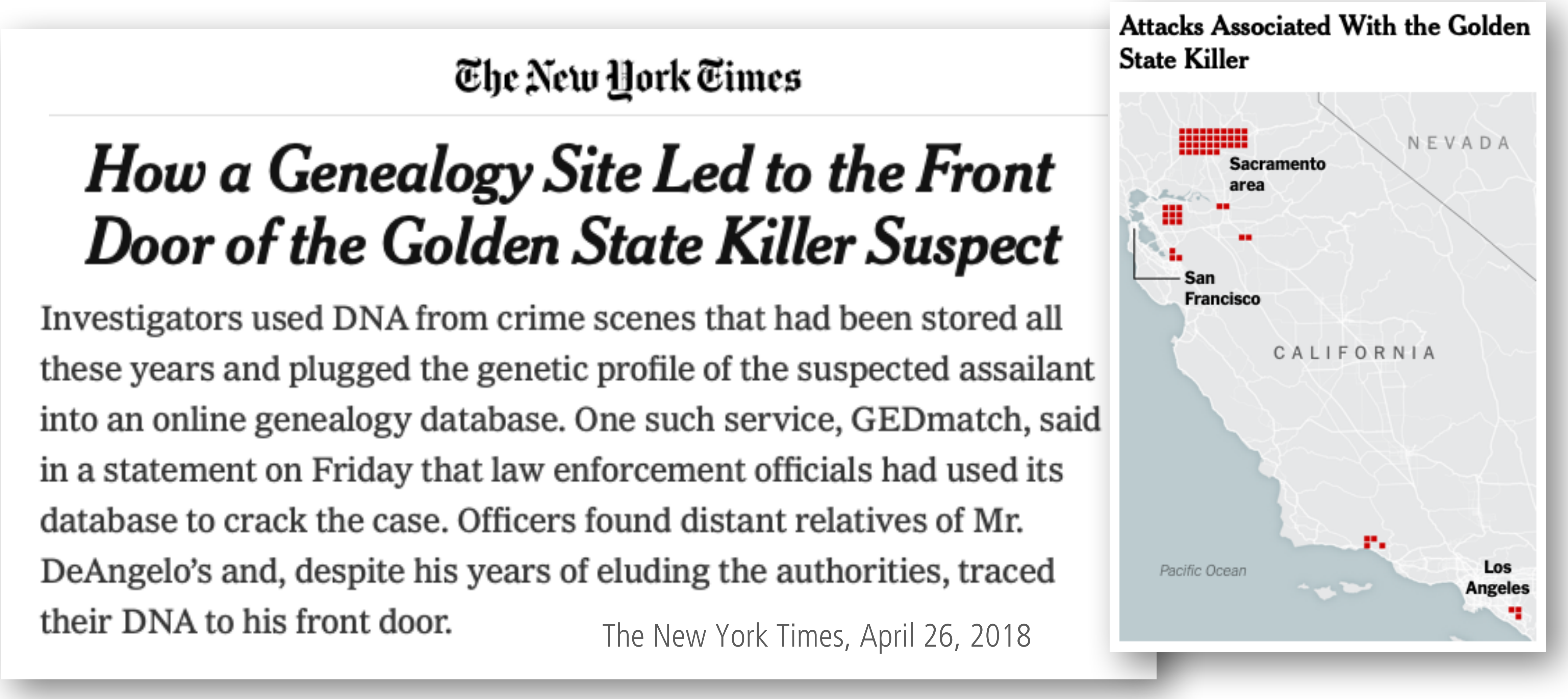
Daily Journal

Devaughn had never been a suspect until genetic genealogy put police on his trail several months ago. Earlier this year, police sent the DNA profile to Parabon, a private genetics company, to compare the suspect's DNA sample to a public genealogy DNA database looking for people with similar DNA profiles who might be kin to the suspect. That eventually led authorities to look at Devaughn.

Rienzi man charged with 1990 Starkville murder

By William Moore Daily Journal 15 hrs ago Comments

© Copyright 2018 Daily Journal, 1242 S Green St Tupelo, MS



The New York Times

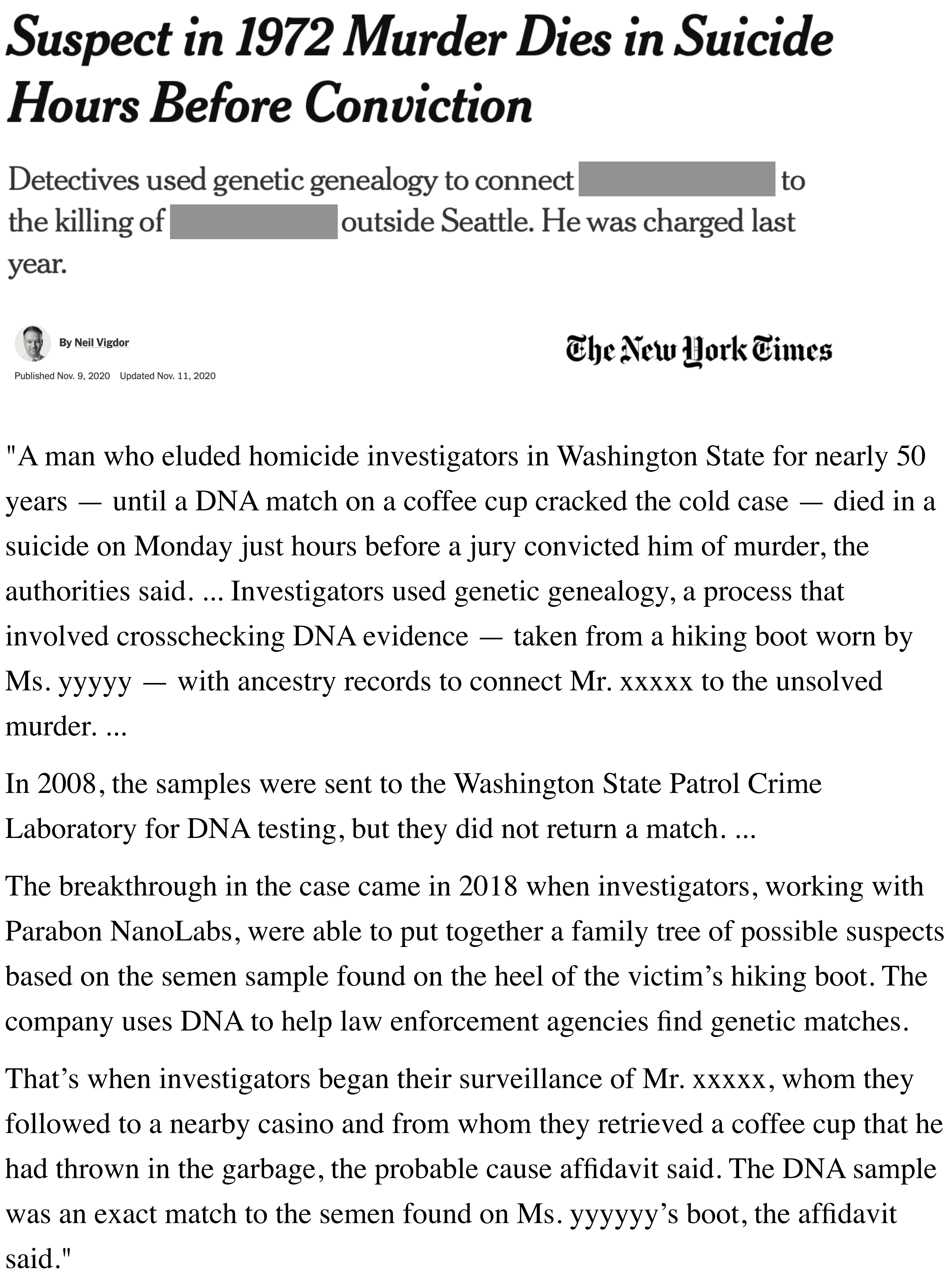
How a Genealogy Site Led to the Front Door of the Golden State Killer Suspect

Investigators used DNA from crime scenes that had been stored all these years and plugged the genetic profile of the suspected assailant into an online genealogy database. One such service, GEDmatch, said in a statement on Friday that law enforcement officials had used its database to crack the case. Officers found distant relatives of Mr. DeAngelo's and, despite his years of eluding the authorities, traced their DNA to his front door.

Attacks Associated With the Golden State Killer

The New York Times, April 26, 2018

Long-Range Familial Searches



Suspect in 1972 Murder Dies in Suicide Hours Before Conviction

Detectives used genetic genealogy to connect [redacted] to the killing of [redacted] outside Seattle. He was charged last year.

By Neil Vigdor

Published Nov. 9, 2020 Updated Nov. 11, 2020

The New York Times

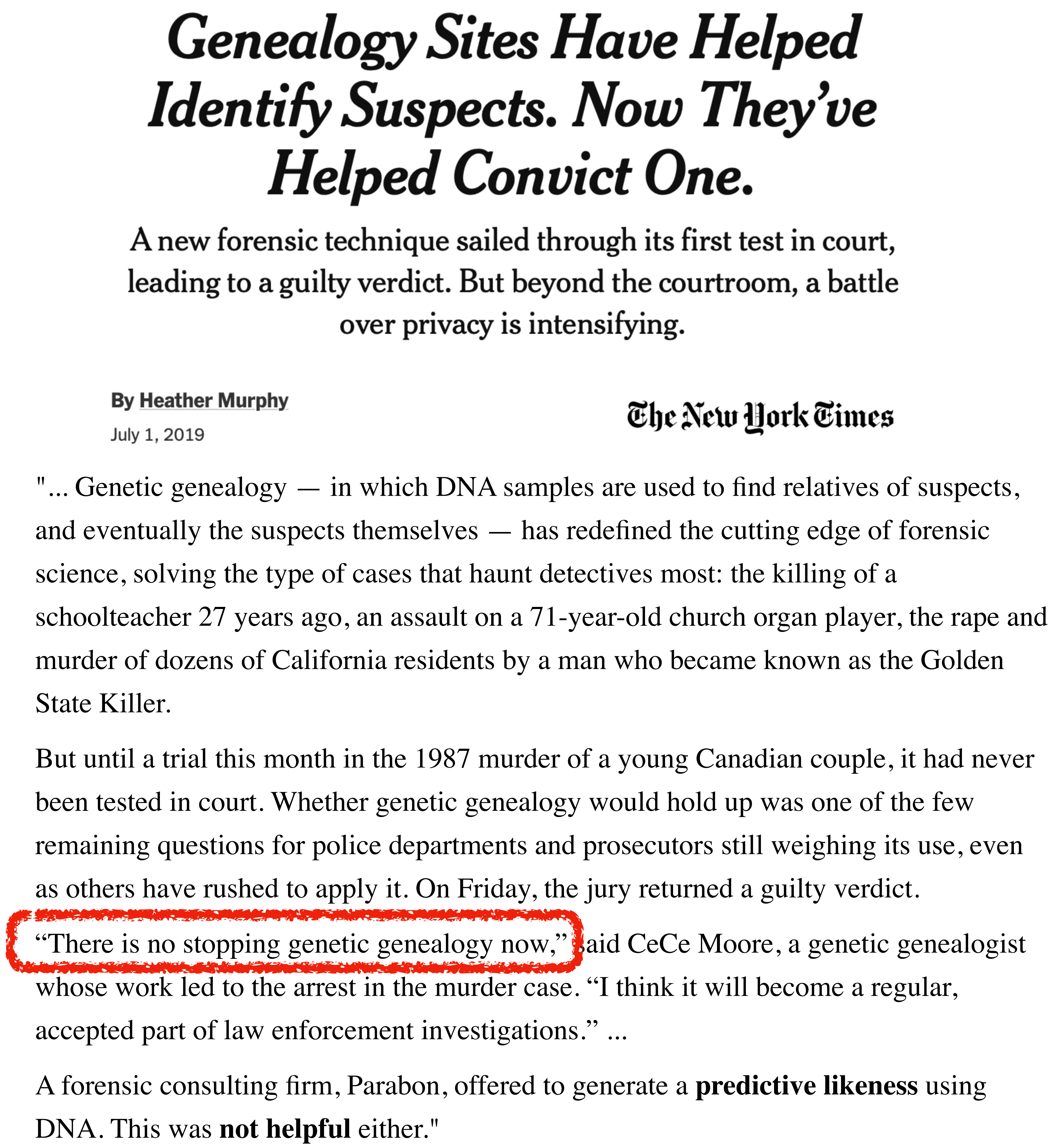
"A man who eluded homicide investigators in Washington State for nearly 50 years — until a DNA match on a coffee cup cracked the cold case — died in a suicide on Monday just hours before a jury convicted him of murder, the authorities said. ... Investigators used genetic genealogy, a process that involved crosschecking DNA evidence — taken from a hiking boot worn by Ms. yyyyy — with ancestry records to connect Mr. xxxxx to the unsolved murder. ...

In 2008, the samples were sent to the Washington State Patrol Crime Laboratory for DNA testing, but they did not return a match. ...

The breakthrough in the case came in 2018 when investigators, working with Parabon NanoLabs, were able to put together a family tree of possible suspects based on the semen sample found on the heel of the victim's hiking boot. The company uses DNA to help law enforcement agencies find genetic matches.

That's when investigators began their surveillance of Mr. xxxxx, whom they followed to a nearby casino and from whom they retrieved a coffee cup that he had thrown in the garbage, the probable cause affidavit said. The DNA sample was an exact match to the semen found on Ms. yyyyyy's boot, the affidavit said."

https://www.nytimes.com/2020/11/09/us/terrence-miller-suicide.html?referringSource=articleShare



Genealogy Sites Have Helped Identify Suspects. Now They've Helped Convict One.

A new forensic technique sailed through its first test in court, leading to a guilty verdict. But beyond the courtroom, a battle over privacy is intensifying.

By Heather Murphy

July 1, 2019

The New York Times

"... Genetic genealogy — in which DNA samples are used to find relatives of suspects, and eventually the suspects themselves — has redefined the cutting edge of forensic science, solving the type of cases that haunt detectives most: the killing of a schoolteacher 27 years ago, an assault on a 71-year-old church organ player, the rape and murder of dozens of California residents by a man who became known as the Golden State Killer.

But until a trial this month in the 1987 murder of a young Canadian couple, it had never been tested in court. Whether genetic genealogy would hold up was one of the few remaining questions for police departments and prosecutors still weighing its use, even as others have rushed to apply it. On Friday, the jury returned a guilty verdict.

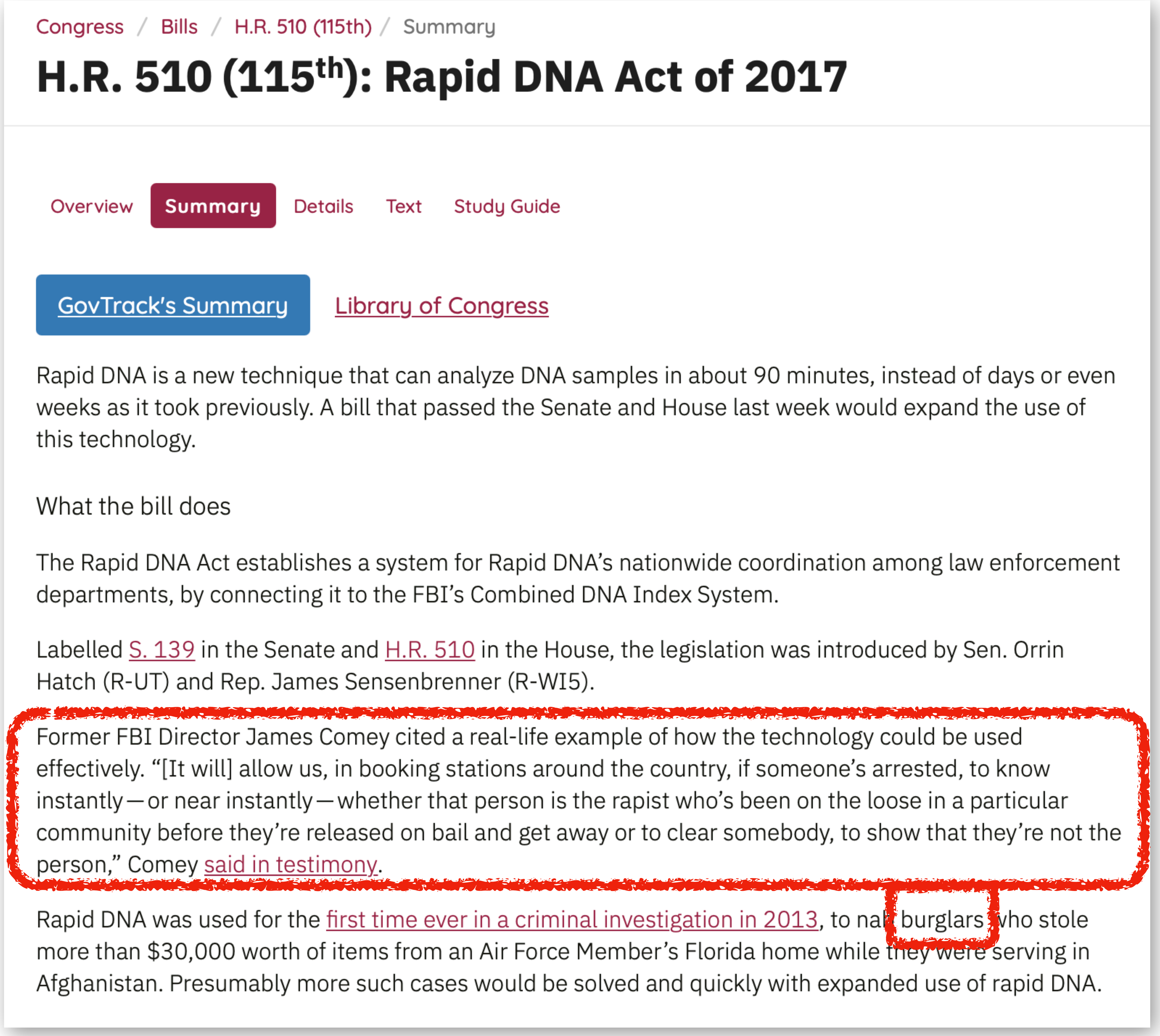
"There is no stopping genetic genealogy now," said CeCe Moore, a genetic genealogist whose work led to the arrest in the murder case. "I think it will become a regular, accepted part of law enforcement investigations." ...

A forensic consulting firm, Parabon, offered to generate a **predictive likeness** using DNA. This was **not helpful** either."

https://www.nytimes.com/2019/07/01/us/dna-genetic-genealogy-trial.html?referringSource=articleShare

Rapid DNA

Legalizing DNA Tests for DNA Indexing



H.R. 510 (115th): Rapid DNA Act of 2017

Rapid DNA is a new technique that can analyze DNA samples in about 90 minutes, instead of days or even weeks as it took previously. A bill that passed the Senate and House last week would expand the use of this technology.

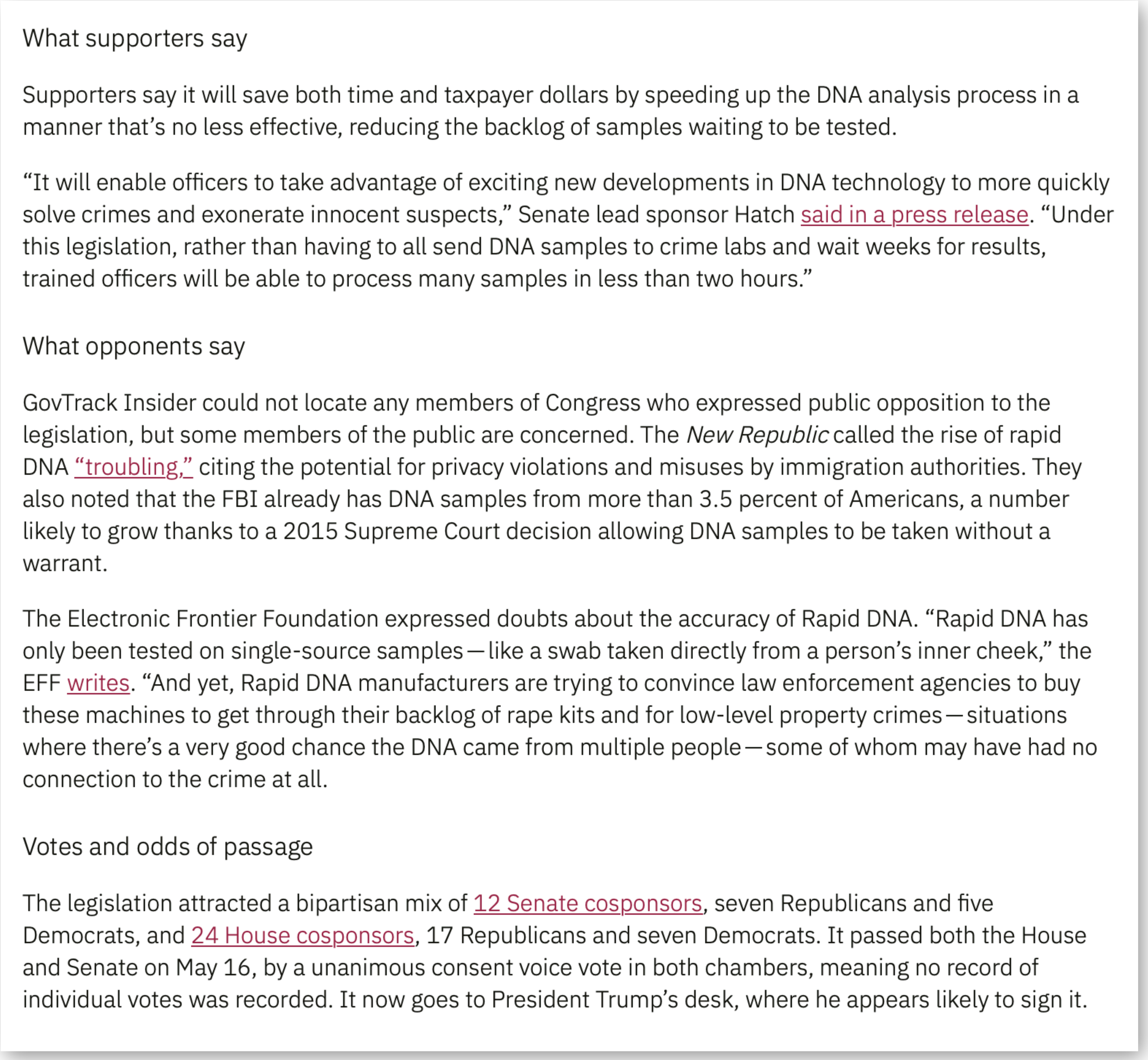
What the bill does

The Rapid DNA Act establishes a system for Rapid DNA's nationwide coordination among law enforcement departments, by connecting it to the FBI's Combined DNA Index System.

Labelled S. 139 in the Senate and H.R. 510 in the House, the legislation was introduced by Sen. Orrin Hatch (R-UT) and Rep. James Sensenbrenner (R-WI5).

Former FBI Director James Comey cited a real-life example of how the technology could be used effectively. "[It will] allow us, in booking stations around the country, if someone's arrested, to know instantly—or near instantly—whether that person is the rapist who's been on the loose in a particular community before they're released on bail and get away or to clear somebody, to show that they're not the person," Comey said in testimony.

Rapid DNA was used for the first time ever in a criminal investigation in 2013, to nab burglars who stole more than $30,000 worth of items from an Air Force Member's Florida home while they were serving in Afghanistan. Presumably more such cases would be solved and quickly with expanded use of rapid DNA.



What supporters say

Supporters say it will save both time and taxpayer dollars by speeding up the DNA analysis process in a manner that's no less effective, reducing the backlog of samples waiting to be tested.

"It will enable officers to take advantage of exciting new developments in DNA technology to more quickly solve crimes and exonerate innocent suspects," Senate lead sponsor Hatch said in a press release. "Under this legislation, rather than having to all send DNA samples to crime labs and wait weeks for results, trained officers will be able to process many samples in less than two hours."

What opponents say

GovTrack Insider could not locate any members of Congress who expressed public opposition to the legislation, but some members of the public are concerned. The *New Republic* called the rise of rapid DNA "troubling," citing the potential for privacy violations and misuses by immigration authorities. They also noted that the FBI already has DNA samples from more than 3.5 percent of Americans, a number likely to grow thanks to a 2015 Supreme Court decision allowing DNA samples to be taken without a warrant.

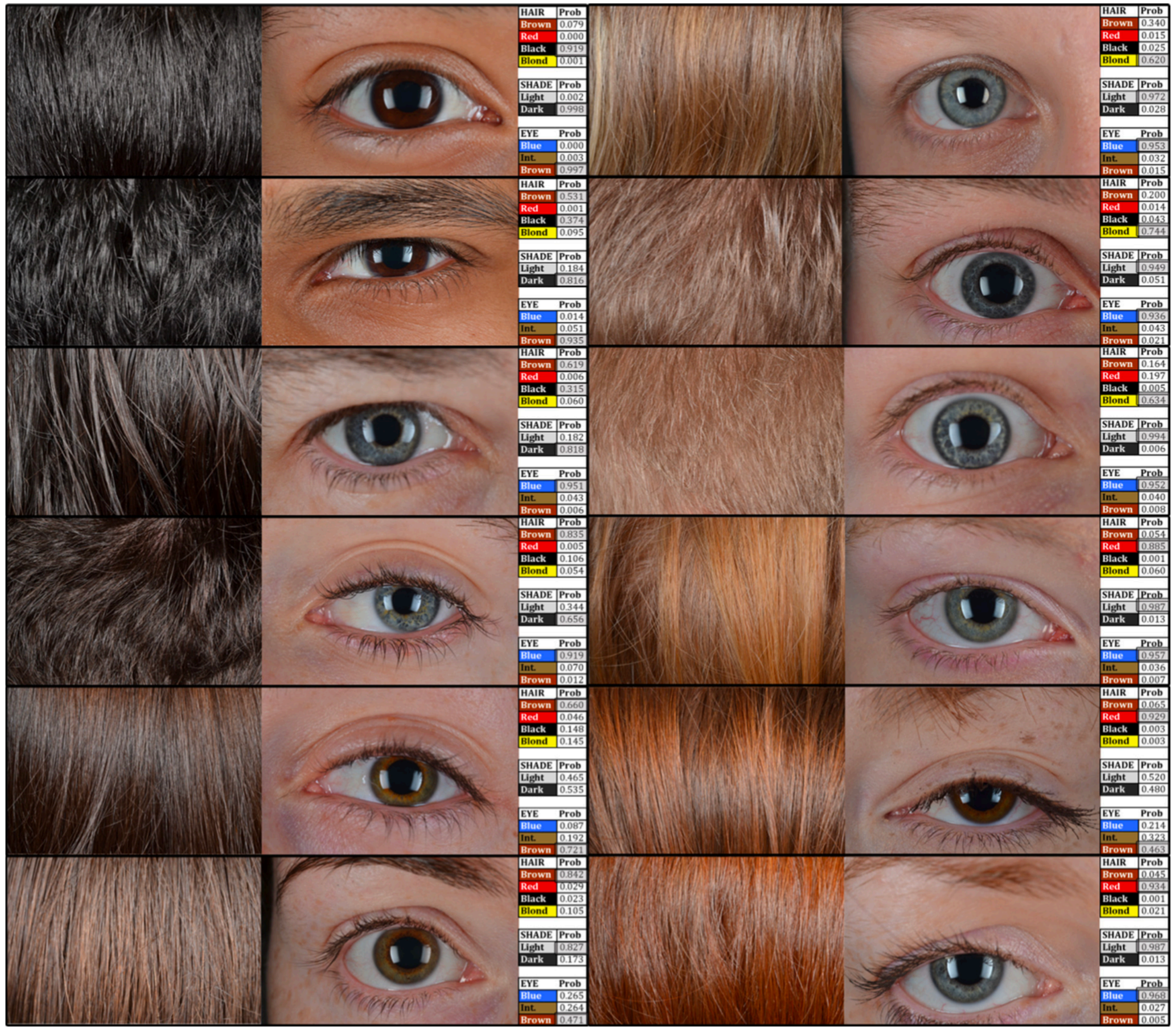
The Electronic Frontier Foundation expressed doubts about the accuracy of Rapid DNA. "Rapid DNA has only been tested on single-source samples—like a swab taken directly from a person's inner cheek," the EFF writes. "And yet, Rapid DNA manufacturers are trying to convince law enforcement agencies to buy these machines to get through their backlog of rape kits and for low-level property crimes—situations where there's a very good chance the DNA came from multiple people—some of whom may have had no connection to the crime at all.

Votes and odds of passage

The legislation attracted a bipartisan mix of 12 Senate cosponsors, seven Republicans and five Democrats, and 24 House cosponsors, 17 Republicans and seven Democrats. It passed both the House and Senate on May 16, by a unanimous consent voice vote in both chambers, meaning no record of individual votes was recorded. It now goes to President Trump's desk, where he appears likely to sign it.

https://www.govtrack.us/congress/bills/115/hr510/summary

Forensic G2P



HAIR	Prob
Brown	0.079
Red	0.000
Black	0.919
Blond	0.001

SHADE	Prob
Light	0.002
Dark	0.998

EYE	Prob
Blue	0.000
Int.	0.003
Brown	0.997

HAIR	Prob
Brown	0.531
Red	0.001
Black	0.374
Blond	0.095

SHADE	Prob
Light	0.184
Dark	0.816

EYE	Prob
Blue	0.014
Int.	0.051
Brown	0.935

HAIR	Prob
Brown	0.619
Red	0.006
Black	0.315
Blond	0.060

SHADE	Prob
Light	0.182
Dark	0.818

EYE	Prob
Blue	0.951
Int.	0.043
Brown	0.006

HAIR	Prob
Brown	0.835
Red	0.005
Black	0.106
Blond	0.054

SHADE	Prob
Light	0.344
Dark	0.656

EYE	Prob
Blue	0.919
Int.	0.070
Brown	0.012

HAIR	Prob
Brown	0.660
Red	0.046
Black	0.148
Blond	0.145

SHADE	Prob
Light	0.465
Dark	0.535

EYE	Prob
Blue	0.087
Int.	0.192
Brown	0.721

HAIR	Prob
Brown	0.842
Red	0.029
Black	0.023
Blond	0.105

SHADE	Prob
Light	0.827
Dark	0.173

EYE	Prob
Blue	0.265
Int.	0.264
Brown	0.471

HAIR	Prob
Brown	0.340
Red	0.015
Black	0.025
Blond	0.620

SHADE	Prob
Light	0.972
Dark	0.028

EYE	Prob
Blue	0.953
Int.	0.032
Brown	0.015

HAIR	Prob
Brown	0.200
Red	0.014
Black	0.043
Blond	0.744

SHADE	Prob
Light	0.949
Dark	0.051

EYE	Prob
Blue	0.936
Int.	0.043
Brown	0.021

HAIR	Prob
Brown	0.164
Red	0.197
Black	0.005
Blond	0.634

SHADE	Prob
Light	0.994
Dark	0.006

EYE	Prob
Blue	0.952
Int.	0.040
Brown	0.008

HAIR	Prob
Brown	0.054
Red	0.885
Black	0.001
Blond	0.060

SHADE	Prob
Light	0.987
Dark	0.013

EYE	Prob
Blue	0.957
Int.	0.036
Brown	0.007

HAIR	Prob
Brown	0.065
Red	0.929
Black	0.003
Blond	0.003

SHADE	Prob
Light	0.520
Dark	0.480

EYE	Prob
Blue	0.214
Int.	0.323
Brown	0.463

HAIR	Prob
Brown	0.045
Red	0.934
Black	0.001
Blond	0.021

SHADE	Prob
Light	0.987
Dark	0.013

EYE	Prob
Blue	0.968
Int.	0.027
Brown	0.005

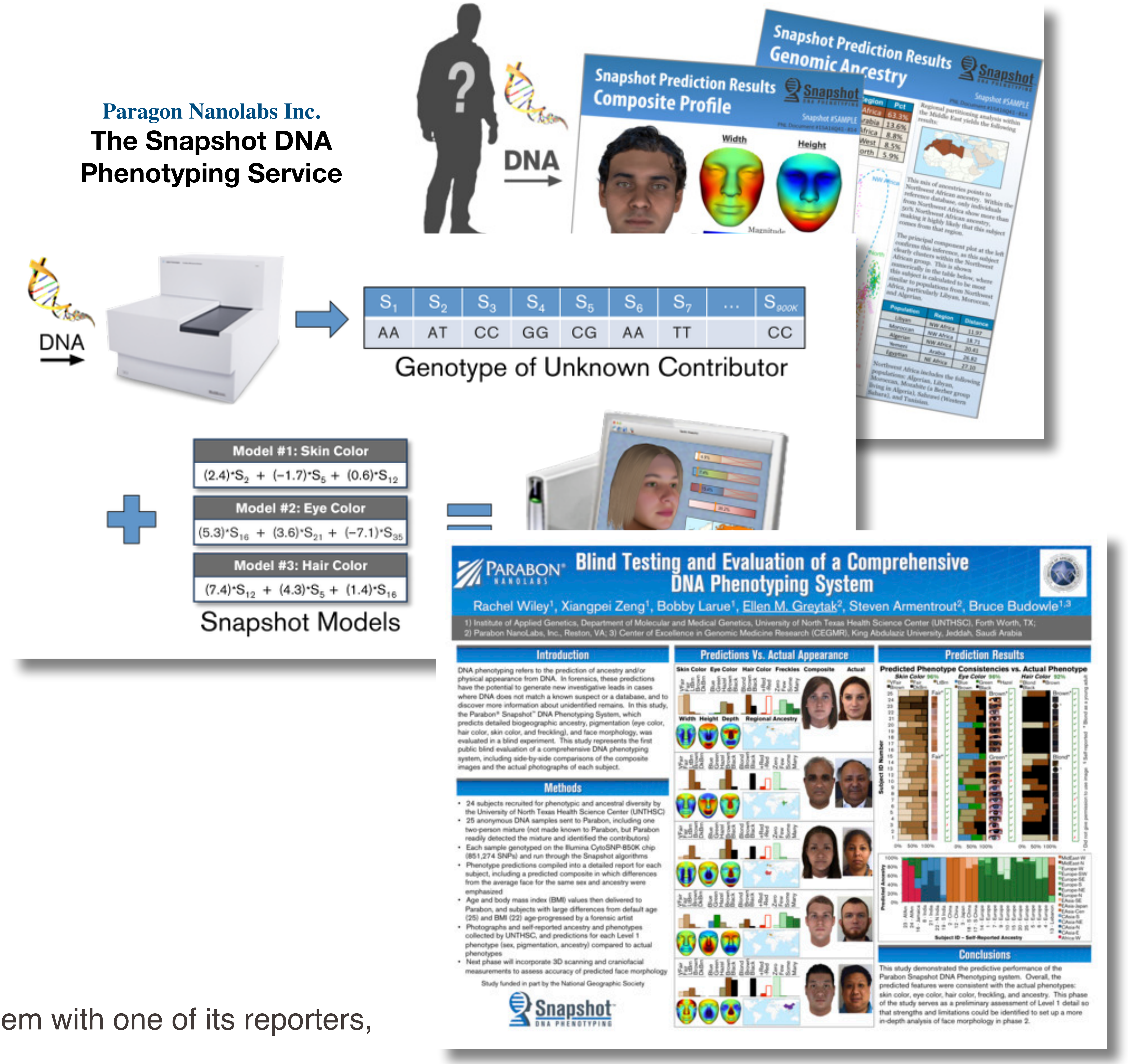
Fig. 1. Individual examples of HIrisPlex-based eye and hair color DNA prediction. Probability outcomes are provided for eye and hair color categories as obtained from complete HIrisPlex SNP profiles [50] using the enhanced IrisPlex eye color and the enhance HIrisPlex hair color prediction models [25] (http://www.erasmusmc.nl/fmb/resources/Irisplex_HIrisPlex/) for 12 individuals chosen with varying eye and hair colors. Eye and hair photographs are provided to allow visual phenotype inspection and comparison with DNA predicted conclusions. Those probabilities that led to the eye and hair color conclusions are highlighted in grey based on the highest probability rule for eye color and by using the HIrisiPlex hair color prediction guide described elsewhere [25,50]. Individual numbering is 1–6 on the left side and 7–12 on the right side. DNA-based prediction conclusions are as follows 1: black hair and brown eyes, 2: dark brown/black hair and brown eyes, 3: dark brown/black hair and blue eyes, 4: brown/dark brown hair and blue eyes, 5: brown/medium brown hair and brown eyes, 6: brown hair and brown eyes (likely with non-brown parts), 7: blond/dark blond hair and blue eyes, 8: blond hair and blue eyes, 9: blond/dark blond hair and blue eyes, 10: red hair and blue eyes, 11: red hair and brown eyes (likely with non-brown parts), and 12: red hair and blue eyes.

Manfred Kayser. Forensic DNA Phenotyping: Predicting human appearance from crime scene material for investigative purposes. Forensic Science International: Genetics 18 (2015) 33–48

Phenotyping from DNA

From DNA to "Wanted" Posters?

- association of genomic variants with phenotypic data collection
- while hair, eye color are easy targets not useful for relevant phenotypic features especially if large environmental component
- huge biases based on input/collection data
- Belgium and Germany do not allow forensic DNA phenotyping
- Switzerland: Bundesrat decision on 2020-12-04 to allow phenotyping for law enforcement purposes


Paragon Nanolabs Inc.
The Snapshot DNA Phenotyping Service

S_1	S_2	S_3	S_4	S_5	S_6	S_7	…	S_{900K}
AA	AT	CC	GG	CG	AA	TT		CC

Genotype of Unknown Contributor

Model #1: Skin Color
$(2.4)\cdot S_2 + (-1.7)\cdot S_5 + (0.6)\cdot S_{12}$

Model #2: Eye Color
$(5.3)\cdot S_{16} + (3.6)\cdot S_{21} + (-7.1)\cdot S_{35}$

Model #3: Hair Color
$(7.4)\cdot S_{12} + (4.3)\cdot S_5 + (1.4)\cdot S_{16}$

Snapshot Models

PARABON NANOLABS

Blind Testing and Evaluation of a Comprehensive DNA Phenotyping System

Rachel Wiley[1], Xiangpei Zeng[1], Bobby Larue[1], Ellen M. Greytak[2], Steven Armentrout[2], Bruce Budowle[1,3]

1) Institute of Applied Genetics, Department of Molecular and Medical Genetics, University of North Texas Health Science Center (UNTHSC), Forth Worth, TX; 2) Parabon NanoLabs, Inc., Reston, VA; 3) Center of Excellence in Genomic Medicine Research (CEGMR), King Abdulaziz University, Jeddah, Saudi Arabia

Introduction

DNA phenotyping refers to the prediction of ancestry and/or physical appearance from DNA. In forensics, these predictions have the potential to generate new investigative leads in cases where DNA does not match a known suspect or a database, and to discover more information about unidentified remains. In this study, the Parabon® Snapshot™ DNA Phenotyping System, which predicts detailed biogeographic ancestry, pigmentation (eye color, hair color, skin color, and freckling), and face morphology, was evaluated in a blind experiment. This study represents the first public blind evaluation of a comprehensive DNA phenotyping system, including side-by-side comparisons of the composite images and the actual photographs of each subject.

Methods

- 24 subjects recruited for phenotypic and ancestral diversity by the University of North Texas Health Science Center (UNTHSC)
- 25 anonymous DNA samples sent to Parabon, including one two-person mixture (not made known to Parabon, but Parabon readily detected the mixture and identified the contributors)
- Each sample genotyped on the Illumina CytoSNP-850K chip (851,274 SNPs) and run through the Snapshot algorithms
- Phenotype predictions compiled into a detailed report for each subject, including a predicted composite in which differences from the average face for the same sex and ancestry were emphasized
- Age and body mass index (BMI) values then delivered to Parabon, and subjects with large differences from default age (25) and BMI (22) age-progressed by a forensic artist
- Photographs and self-reported ancestry and phenotypes collected by UNTHSC, and predictions for each Level 1 phenotype (sex, pigmentation, ancestry) compared to actual phenotypes
- Next phase will incorporate 3D scanning and craniofacial measurements to assess accuracy of predicted face morphology

Study funded in part by the National Geographic Society

Predictions Vs. Actual Appearance

Prediction Results

Predicted Phenotype Consistencies vs. Actual Phenotype

Conclusions

This study demonstrated the predictive performance of the Parabon Snapshot DNA Phenotyping system. Overall, the predicted features were consistent with the actual phenotypes: skin color, eye color, hair color, freckling, and ancestry. This phase of the study serves as a preliminary assessment of Level 1 detail so that strengths and limitations could be identified to set up a more in-depth analysis of face morphology in phase 2.

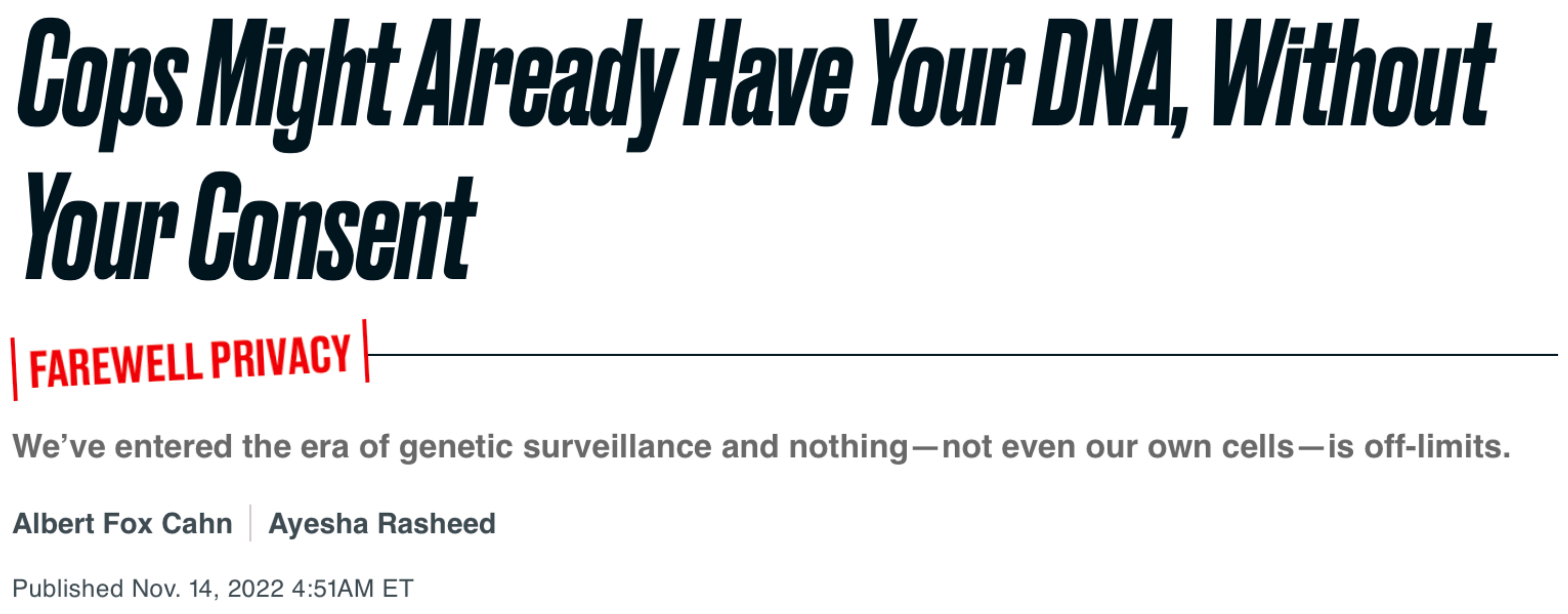
"When the New York Times ran an informal test of the Parabon system with one of its reporters, it failed badly." (ACLU.org)

https://snapshot.parabon-nanolabs.com/phenotyping

DNA & Law Enforcement

Legal minefields, hard to avoid?

- "...when police in Edmonton, Canada, released a suspect’s image, the **crude graphic** ... came **from the suspect’s DNA**."
- "...every time a **family member** sends in their swab, they’re sending in your data too..."
- "...**many players** in this growing movement offer to translate our genetic code into phenotypes (that is, observable features like eye color), often with **scant commitment to scientific accuracy**..."
- "...Veering into **pseudoscience**, they are a modern **sales pitch for** the long-discredited **phrenology** of the past. They wrongly treat race as a biological fact, rather than the social construct that it is. And in the process, they open all the **flaws of facial recognition** to new realms..."
- "...we first have to change our focus from preventing DNA collection to **preventing misuse** and managing access..."
- "The answer is simple: **Ban DNA searches** ... beyond the types of one-to-one DNA tests that are subject to judicial oversight..."



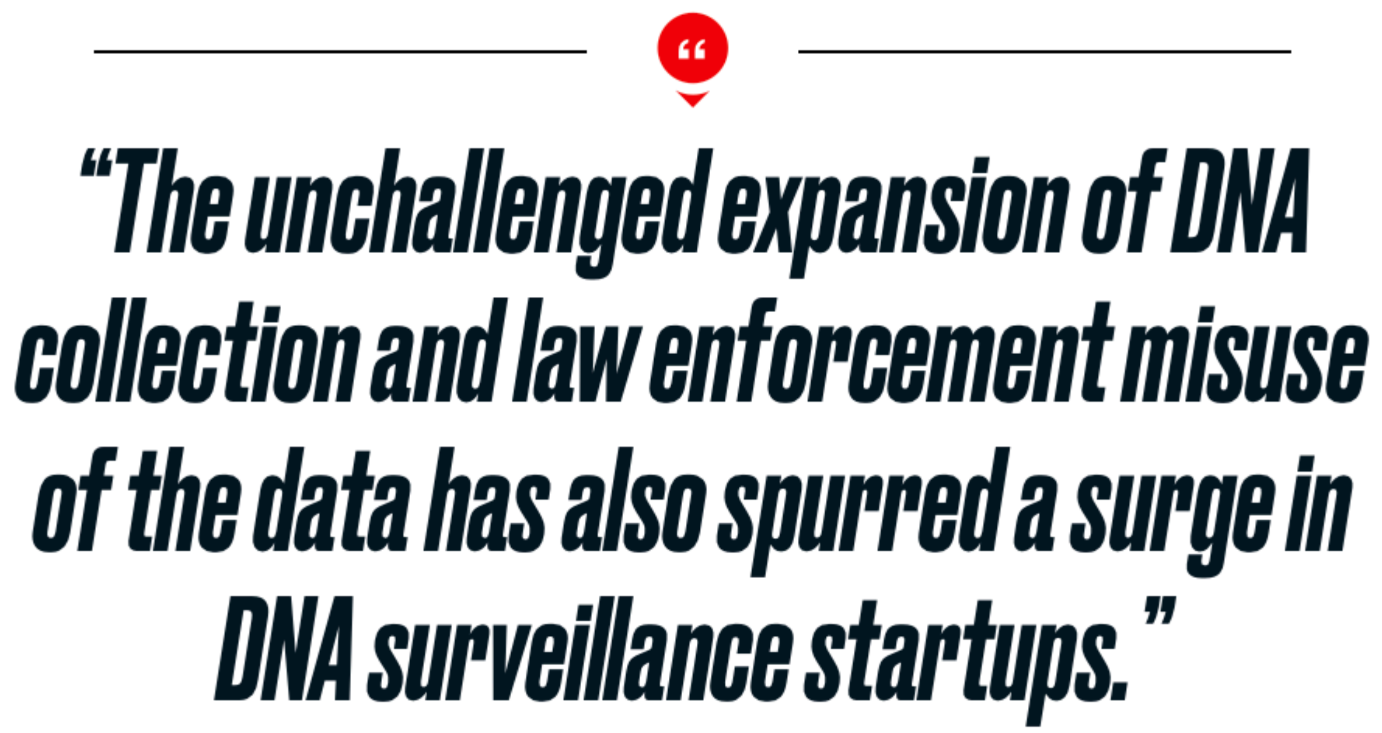
Cops Might Already Have Your DNA, Without Your Consent

FAREWELL PRIVACY

We’ve entered the era of genetic surveillance and nothing—not even our own cells—is off-limits.

Albert Fox Cahn | Ayesha Rasheed

Published Nov. 14, 2022 4:51AM ET



“The unchallenged expansion of DNA collection and law enforcement misuse of the data has also spurred a surge in DNA surveillance startups.”

thedailybeast.com/cops-might-already-have-your-dna-without-your-consent

The Swiss Way

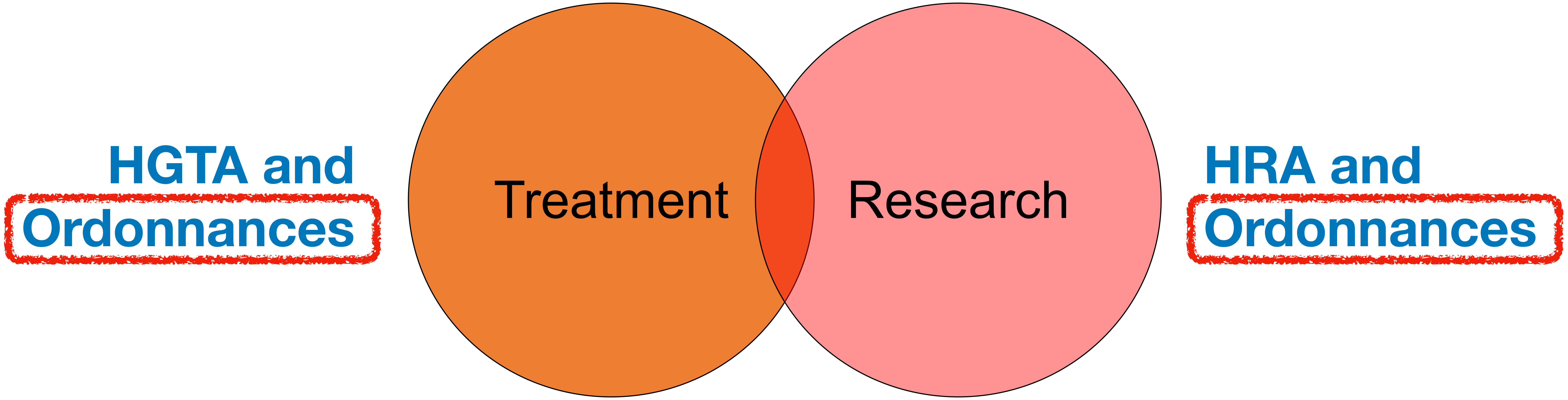
Genomic Data & Privacy Protection - The Swiss View

Relevant areas

- Medical treatment (Federal Act on Human Genetic Testing, HGTA)
- Human Research (Human Research Act, HRA)
- Tests other than for medical purposes (*new* in the HGTA from 2021 on)
- Law enforcement (Federal Act on the Use of DNA Profiles in Criminal Proceedings and for Identifying Unidentified or Missing Person, DNA Profiles Act)
- Data protection (Data Protection Act, DPA)
- ...

Content provided by Julian Mausbach | Oberassistent für Strafrecht und Strafprozessrecht

Law's View on Modern Medicine



- How do we handle the growing overlap area?

➡ unclear; current legislative movement:
HRA will relate more to HGTA in the future

HGTA : Federal
Act on Human
Genetic Testing

HRA: Human
Research Act

Content provided by Julian Mausbach | Oberassistent für Strafrecht und Strafprozessrecht

2021 Data Protection Act

Art. 5 Definitions

The following definitions apply in this Act:

a. **personal data**: all information relating to an identified or identifiable natural person;

b. **data subject**: natural person whose personal data is processed;

c. **sensitive personal data**:

1. data on religious, ideological, political or trade union-related views or activities,

2. data on health, the intimate sphere or the racial or ethnic origin,

3. genetic data,

4. biometric data which unequivocally identifies a natural person,

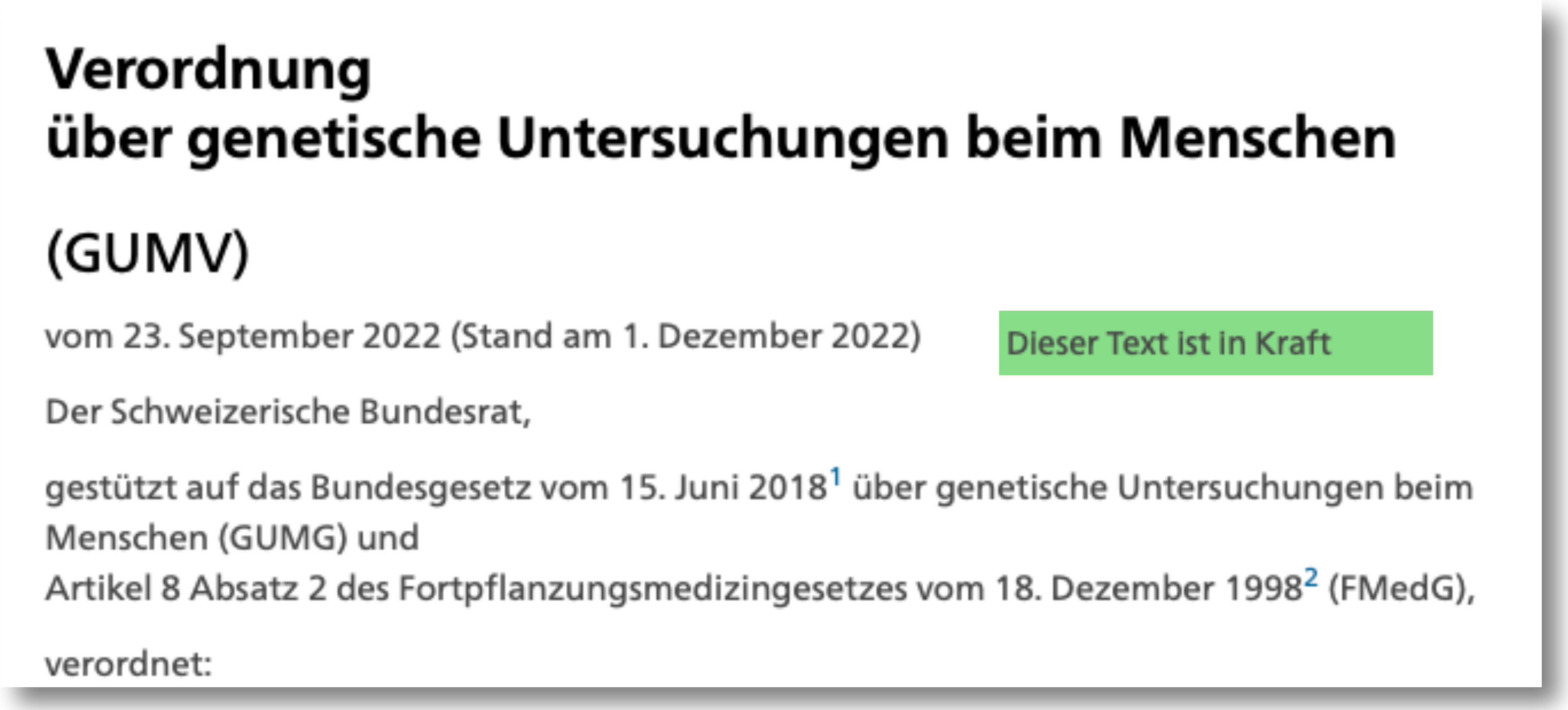
….

Therewith **Genetic Data is always sensitive data**, and especially Art. 6 Principles of data processing and **High-risk profiling**: profiling which involves a high risk to the personality or fundamental rights of the data subject, as it creates a pairing between data that enables an assessment of essential aspects of the personality of a natural person, needs to be considered deeper.

Content provided by Julian Mausbach | Oberassistent für Strafrecht und Strafprozessrecht

HGTA : Federal Act on Human Genetic Testing

HGTA new	medical field	outside the medical field	
Investigated characteristics	medical relevant	especially protective values characteristics	other characteristics
General Requirements	Non-discrimination, information and consent, right to information, right not to know, avoidance of surplus information, protection of samples and genetic data, Circulation concerning public advertising, state of science and technology, penal provisions		
Initiation	Physician	Health professional (controlled taking of samples)	Consumer **(DTC)**
Persons concerned	Persons with **and** without capacity of judgement, pregnant woman (PND)	ONLY persons with Capacity of judgement	ONLY persons with Capacity of judgement
Communication of surplus information	as a rule according to decision of the person concerned	Not allowed	Not allowed
Laboratory	subject to authorization (cyto and molecular genetic studies)	subject to authorization (cyto and molecular genetic studies)	not subject to authorisation
Employers and Insurance institutions	Studies and Recovery of Results / Data only in regulated exceptional cases	Prohibition to carry out investigations and the Recovery of Results / Data	Prohibition to carry out investigations and the Recovery of Results / Data



Verordnung über genetische Untersuchungen beim Menschen

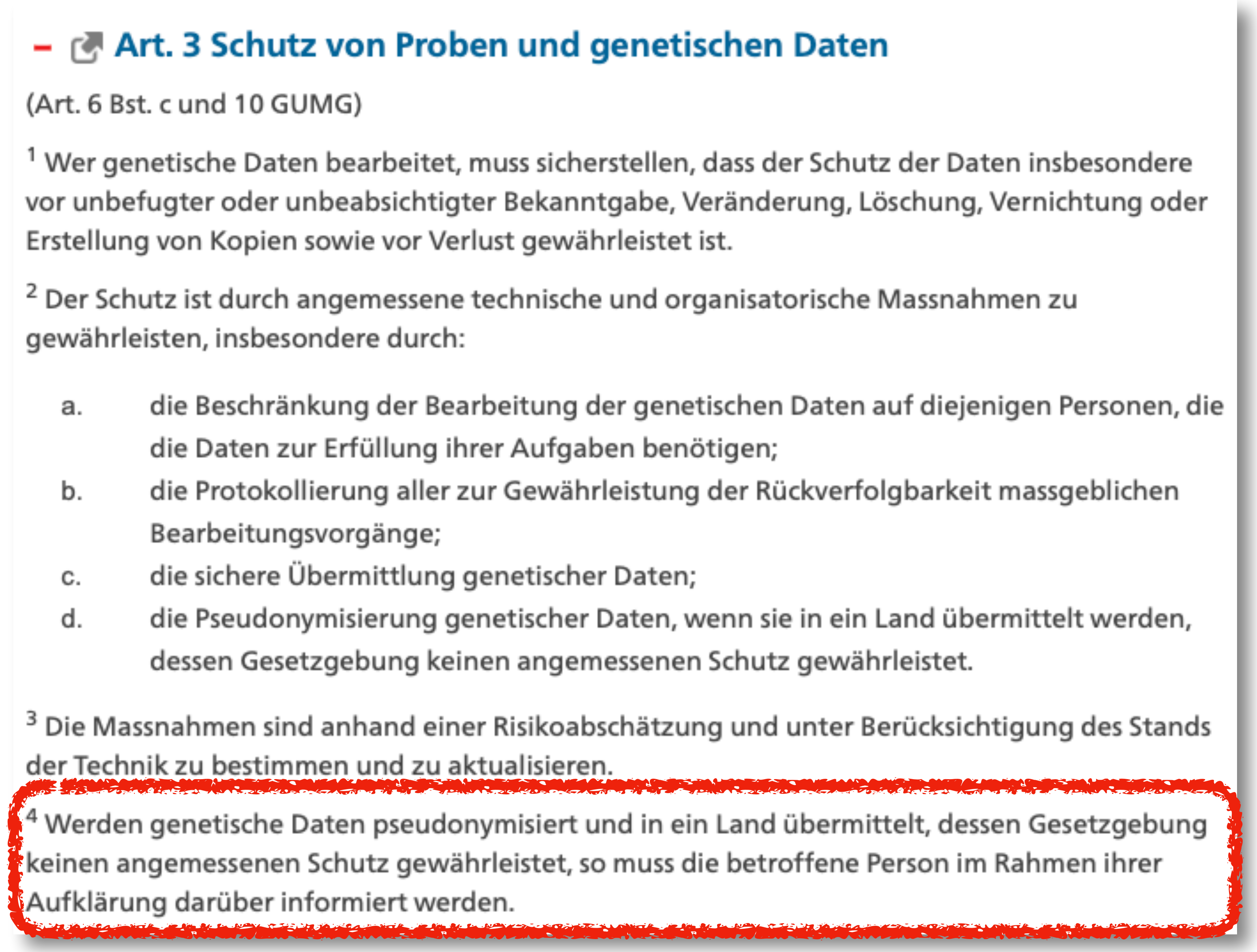
(GUMV)

vom 23. September 2022 (Stand am 1. Dezember 2022) Dieser Text ist in Kraft

Der Schweizerische Bundesrat,

gestützt auf das Bundesgesetz vom 15. Juni 2018[1] über genetische Untersuchungen beim Menschen (GUMG) und
Artikel 8 Absatz 2 des Fortpflanzungsmedizingesetzes vom 18. Dezember 1998[2] (FMedG),

verordnet:



Art. 3 Schutz von Proben und genetischen Daten

(Art. 6 Bst. c und 10 GUMG)

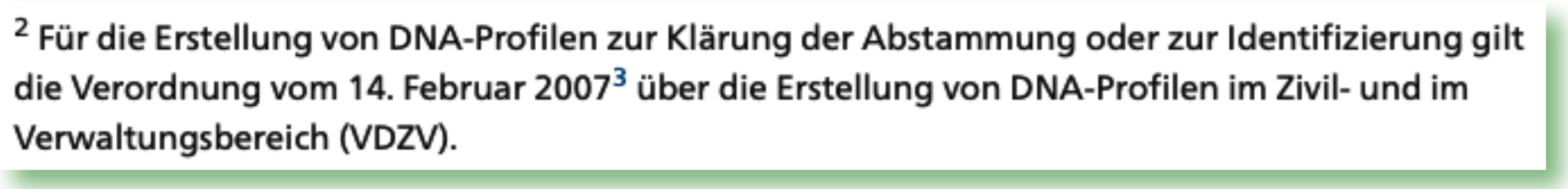
[1] Wer genetische Daten bearbeitet, muss sicherstellen, dass der Schutz der Daten insbesondere vor unbefugter oder unbeabsichtigter Bekanntgabe, Veränderung, Löschung, Vernichtung oder Erstellung von Kopien sowie vor Verlust gewährleistet ist.

[2] Der Schutz ist durch angemessene technische und organisatorische Massnahmen zu gewährleisten, insbesondere durch:

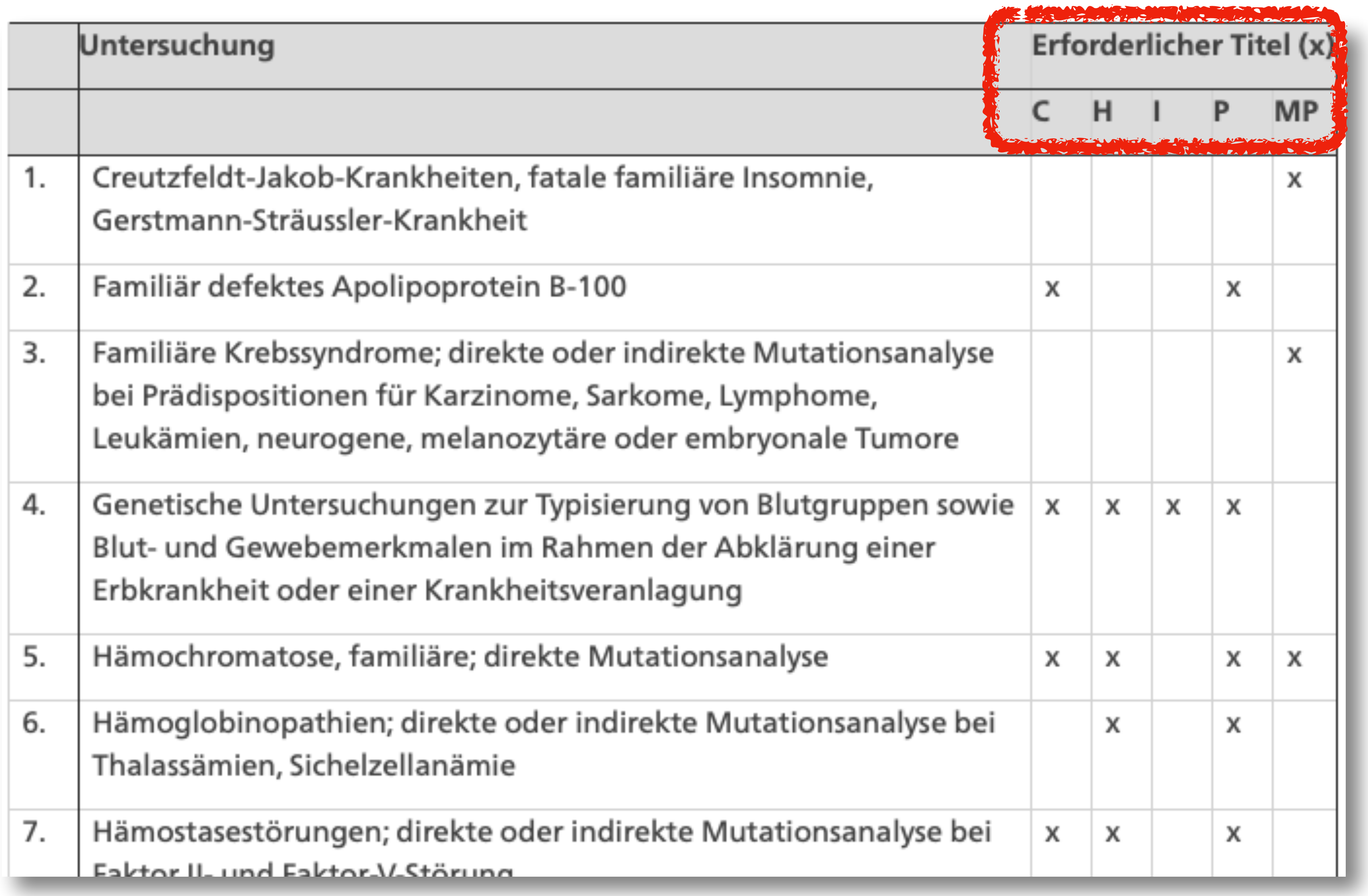
a. die Beschränkung der Bearbeitung der genetischen Daten auf diejenigen Personen, die die Daten zur Erfüllung ihrer Aufgaben benötigen;
b. die Protokollierung aller zur Gewährleistung der Rückverfolgbarkeit massgeblichen Bearbeitungsvorgänge;
c. die sichere Übermittlung genetischer Daten;
d. die Pseudonymisierung genetischer Daten, wenn sie in ein Land übermittelt werden, dessen Gesetzgebung keinen angemessenen Schutz gewährleistet.

[3] Die Massnahmen sind anhand einer Risikoabschätzung und unter Berücksichtigung des Stands der Technik zu bestimmen und zu aktualisieren.

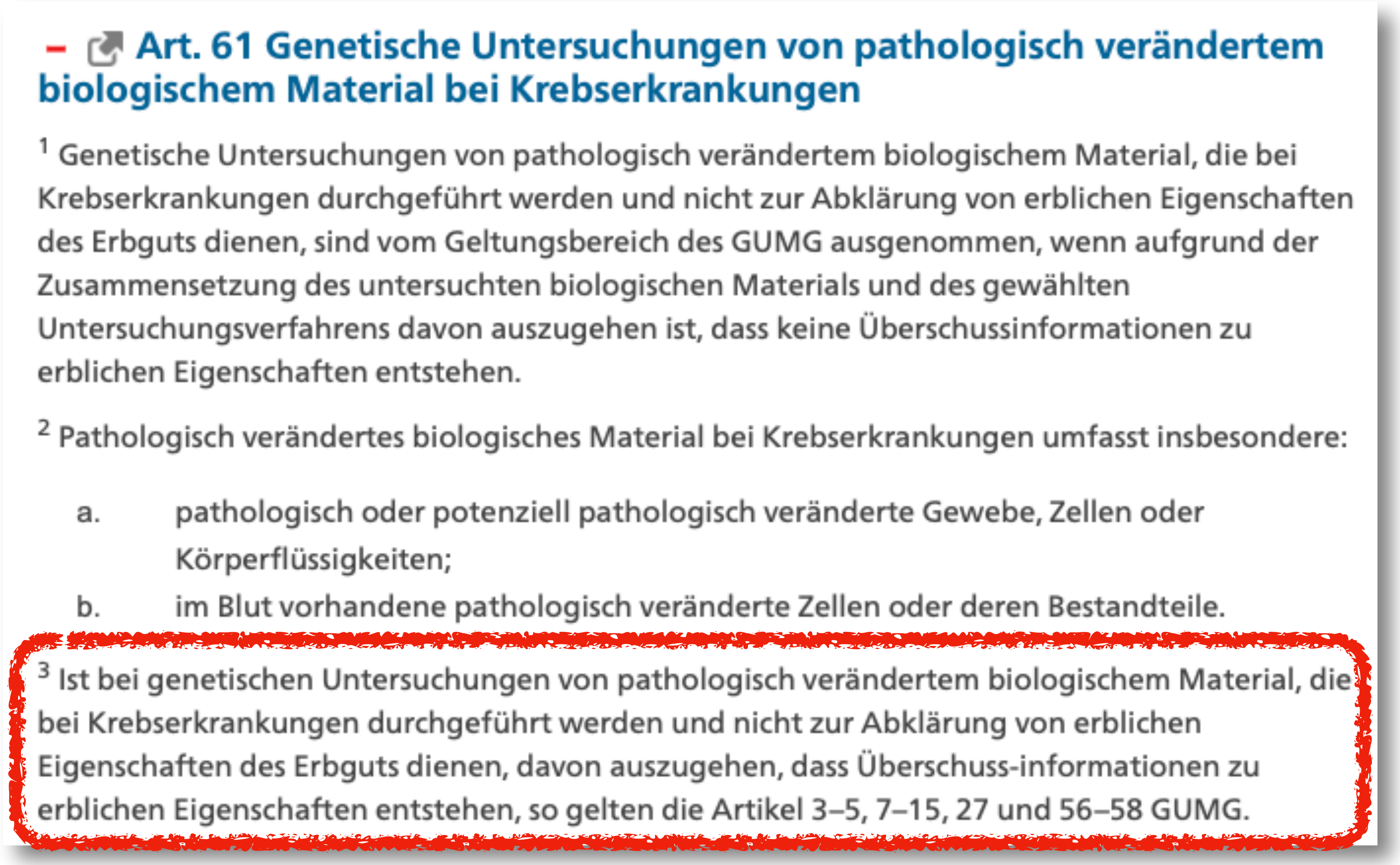
[4] Werden genetische Daten pseudonymisiert und in ein Land übermittelt, dessen Gesetzgebung keinen angemessenen Schutz gewährleistet, so muss die betroffene Person im Rahmen ihrer Aufklärung darüber informiert werden.



[2] Für die Erstellung von DNA-Profilen zur Klärung der Abstammung oder zur Identifizierung gilt die Verordnung vom 14. Februar 2007[3] über die Erstellung von DNA-Profilen im Zivil- und im Verwaltungsbereich (VDZV).



	Untersuchung	Erforderlicher Titel (x)				
		C	H	I	P	MP
1.	Creutzfeldt-Jakob-Krankheiten, fatale familiäre Insomnie, Gerstmann-Sträussler-Krankheit					x
2.	Familiär defektes Apolipoprotein B-100	x			x	
3.	Familiäre Krebssyndrome; direkte oder indirekte Mutationsanalyse bei Prädispositionen für Karzinome, Sarkome, Lymphome, Leukämien, neurogene, melanozytäre oder embryonale Tumore					x
4.	Genetische Untersuchungen zur Typisierung von Blutgruppen sowie Blut- und Gewebemerkmalen im Rahmen der Abklärung einer Erbkrankheit oder einer Krankheitsveranlagung	x	x	x	x	
5.	Hämochromatose, familiäre; direkte Mutationsanalyse	x	x		x	x
6.	Hämoglobinopathien; direkte oder indirekte Mutationsanalyse bei Thalassämien, Sichelzellanämie		x		x	
7.	Hämostasestörungen; direkte oder indirekte Mutationsanalyse bei Faktor II- und Faktor-V-Störung	x	x		x	



Art. 61 Genetische Untersuchungen von pathologisch verändertem biologischem Material bei Krebserkrankungen

[1] Genetische Untersuchungen von pathologisch verändertem biologischem Material, die bei Krebserkrankungen durchgeführt werden und nicht zur Abklärung von erblichen Eigenschaften des Erbguts dienen, sind vom Geltungsbereich des GUMG ausgenommen, wenn aufgrund der Zusammensetzung des untersuchten biologischen Materials und des gewählten Untersuchungsverfahrens davon auszugehen ist, dass keine Überschussinformationen zu erblichen Eigenschaften entstehen.

[2] Pathologisch verändertes biologisches Material bei Krebserkrankungen umfasst insbesondere:

a. pathologisch oder potenziell pathologisch veränderte Gewebe, Zellen oder Körperflüssigkeiten;
b. im Blut vorhandene pathologisch veränderte Zellen oder deren Bestandteile.

[3] Ist bei genetischen Untersuchungen von pathologisch verändertem biologischem Material, die bei Krebserkrankungen durchgeführt werden und nicht zur Abklärung von erblichen Eigenschaften des Erbguts dienen, davon auszugehen, dass Überschuss-informationen zu erblichen Eigenschaften entstehen, so gelten die Artikel 3–5, 7–15, 27 und 56–58 GUMG.

Data Ownership

- Within Switzerland, there is no coherent approach on ownership of data as such (but academic discussion is ongoing, if that is needed).
- Restrictions of usage and disclosure of data other than personal data mainly stem from contractual relationships.
- In the field of research this leads mostly to a data ownership by the research institution.

Of course the restrictions of the different acts that are in the field need to be respected (procuring data lawfully, consent for further use, etc.)

Content provided by Julian Mausbach | Oberassistent für Strafrecht und Strafprozessrecht

Way forward...

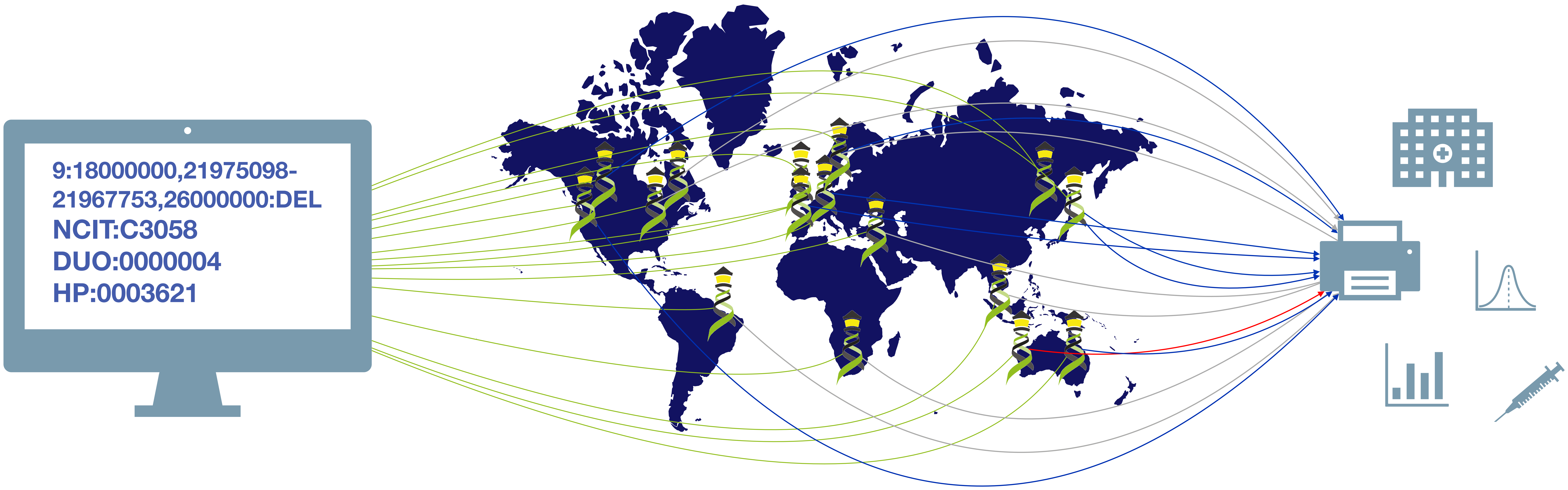
The vision for genomic research: **Federation** of data

Making Beacons Biomedical - Beacon v2

- Scoping queries through "biodata" parameters
- Extending the queries towards clinically ubiquitous variant formats
  - cytogenetic annotations, named variants, variant effects
- Beacon queries as entry for **data delivery**
  - Beacon v2 permissive to respond with variety of data types
    - Phenopackets, biosample data, cohort information ...
  - handover to stream and download using htsget, VCF, EHRs
- Interacting with EHR standards
  - FHIR translations for queries and handover ...
- Beacons as part of local, secure environments
- Authentication to enable non-aggregate, patient derived datasets
  - ELIXIR AAI with compatibility to other providers (OAuth...)

Global Alliance
for Genomics & Health

elixir



9:18000000,21975098-21967753,26000000:DEL
NCIT:C3058
DUO:0000004
HP:0003621

Have you seen deletions in this region on chromosome 9 in Glioblastomas from a juvenile patient, in a dataset with unrestricted access?

Beacon *v2* API

The Beacon API v2 proposal opens the way for the design of a simple but powerful "**genomics API**".

Making Beacons Biomedical - Beacon v2

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Definitely breaks the "Relative Security by Design" Concept!

- Interacting with EHR standards
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Generalkonsent

BENEFIT

BLOCKCHAIN

HEALTH

PRIVACY

SECURITY

CONSENT

ACCESS

Right to Research

HACKERS

LAWS

Genetic
Information
Nondiscrimination
Act

Health
Insurance
Portability and
Accountability
Act

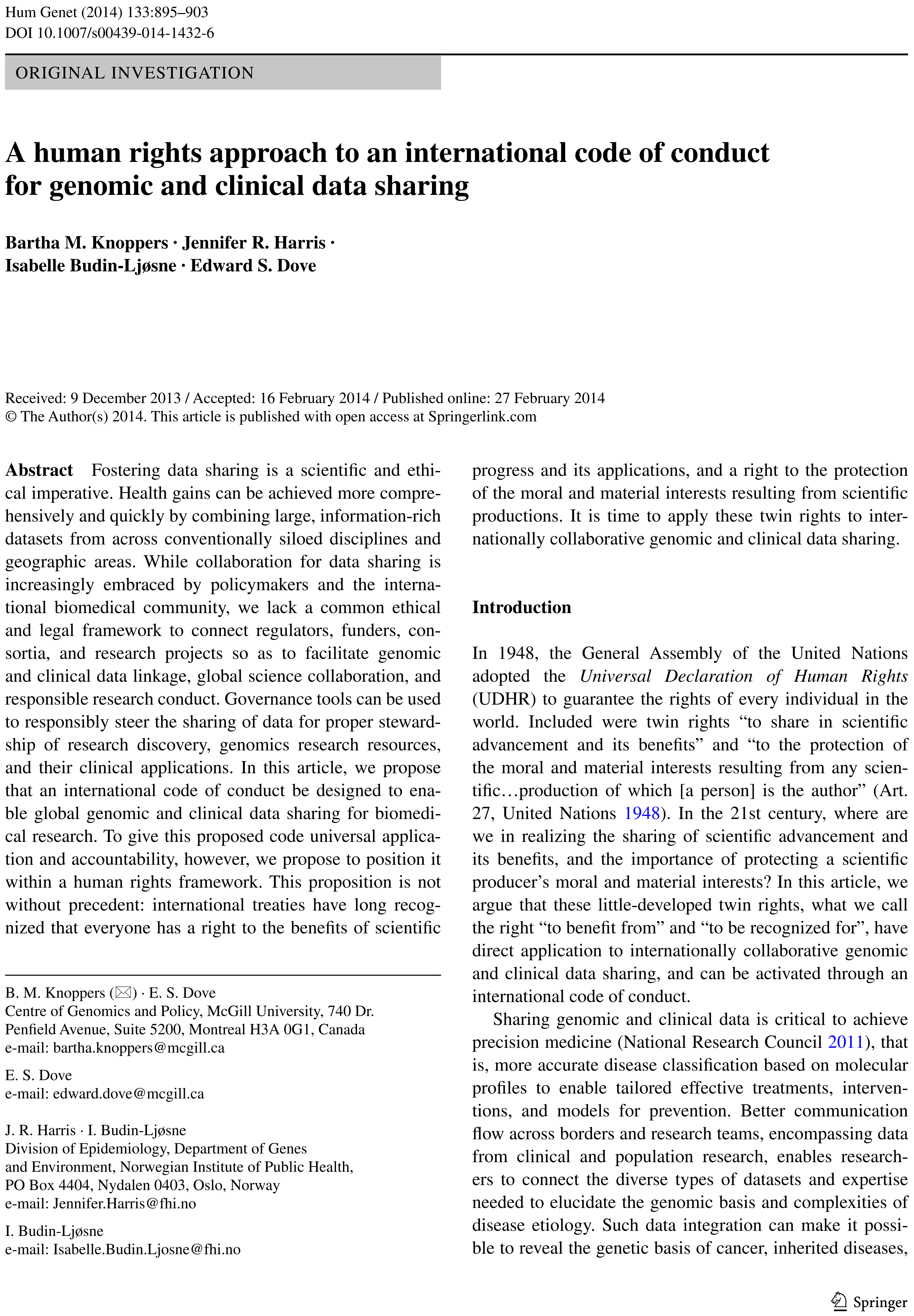
SAFETY

CRYPTOGRAPHY

The Right to Scientific Knowledge

In 1948, the General assembly of the United nations adopted the Universal Declaration of Human Rights (UDHr) to guarantee the rights of every individual in the world. Included were twin rights “to share in scientific advancement and its benefits” and “to the protection of the moral and material interests resulting from any scientific...production of which [a person] is the author” (art. 27, United nations 1948).

from *Knoppers et al, 2014*



Hum Genet (2014) 133:895–903
DOI 10.1007/s00439-014-1432-6

ORIGINAL INVESTIGATION

A human rights approach to an international code of conduct for genomic and clinical data sharing

**Bartha M. Knoppers · Jennifer R. Harris ·
Isabelle Budin-Ljøsne · Edward S. Dove**

Received: 9 December 2013 / Accepted: 16 February 2014 / Published online: 27 February 2014

Abstract Fostering data sharing is a scientific and ethical imperative. Health gains can be achieved more comprehensively and quickly by combining large, information-rich datasets from across conventionally siloed disciplines and geographic areas. While collaboration for data sharing is increasingly embraced by policymakers and the international biomedical community, we lack a common ethical and legal framework to connect regulators, funders, consortia, and research projects so as to facilitate genomic and clinical data linkage, global science collaboration, and responsible research conduct. Governance tools can be used to responsibly steer the sharing of data for proper stewardship of research discovery, genomics research resources, and their clinical applications. In this article, we propose that an international code of conduct be designed to enable global genomic and clinical data sharing for biomedical research. To give this proposed code universal application and accountability, however, we propose to position it within a human rights framework. This proposition is not without precedent: international treaties have long recognized that everyone has a right to the benefits of scientific progress and its applications, and a right to the protection of the moral and material interests resulting from scientific productions. It is time to apply these twin rights to internationally collaborative genomic and clinical data sharing.

Introduction

In 1948, the General Assembly of the United Nations adopted the *Universal Declaration of Human Rights* (UDHR) to guarantee the rights of every individual in the world. Included were twin rights “to share in scientific advancement and its benefits” and “to the protection of the moral and material interests resulting from any scientific…production of which [a person] is the author” (Art. 27, United Nations 1948). In the 21st century, where are we in realizing the sharing of scientific advancement and its benefits, and the importance of protecting a scientific producer’s moral and material interests? In this article, we argue that these little-developed twin rights, what we call the right “to benefit from” and “to be recognized for”, have direct application to internationally collaborative genomic and clinical data sharing, and can be activated through an international code of conduct.

Sharing genomic and clinical data is critical to achieve precision medicine (National Research Council 2011), that is, more accurate disease classification based on molecular profiles to enable tailored effective treatments, interventions, and models for prevention. Better communication flow across borders and research teams, encompassing data from clinical and population research, enables researchers to connect the diverse types of datasets and expertise needed to elucidate the genomic basis and complexities of disease etiology. Such data integration can make it possible to reveal the genetic basis of cancer, inherited diseases,

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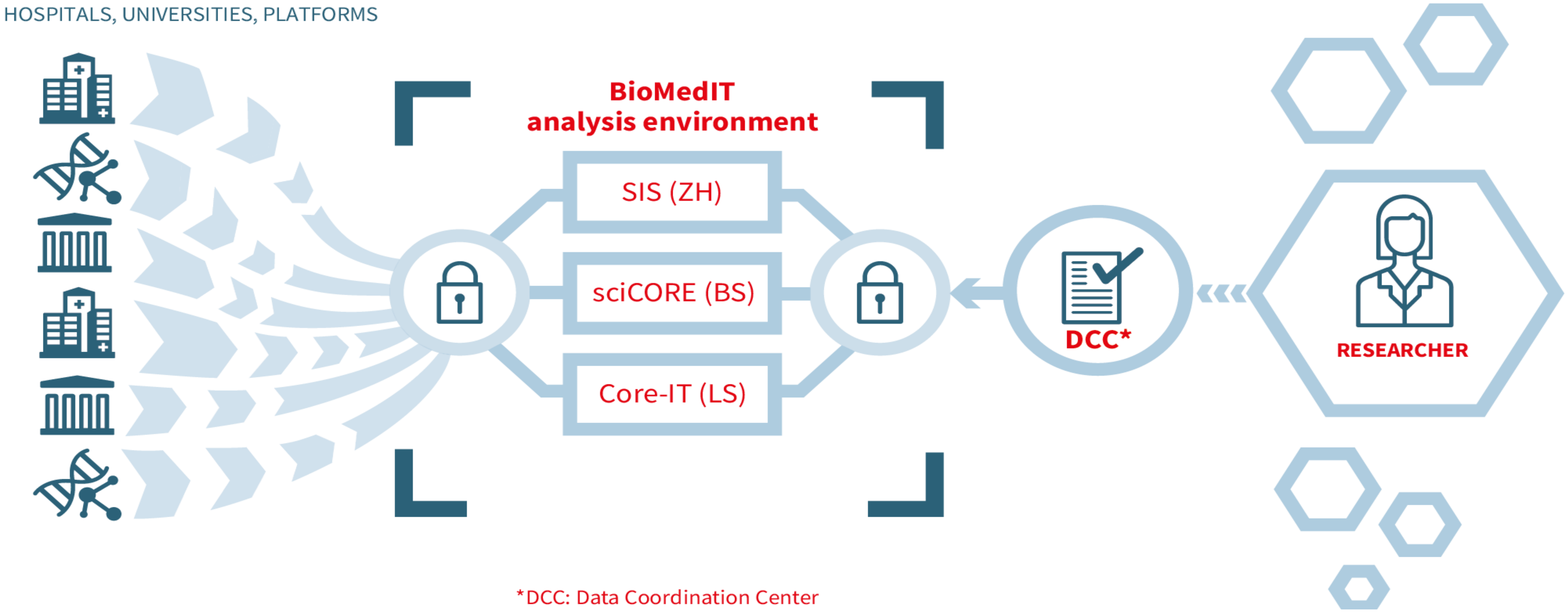
Improving Data Privacy but Empowering Beneficial Use

Intersecting Areas of Development

- Make genomic (and functional) data "obfuscated" for malicious use
  - e.g. spiking / randomization of variants in "not-disease" loci
- access protection with defined user access using standardized protocols for users' roles and permissions, in contrast to individual per user, per dataset access requests over data access committees (DACs)
  - digital "differential" consent using e.g. data use ontologies
- intentional and unintentional (!) data providers have to be protected from abuse by legal regulations - though thin line regarding "overzealous" use by law enforcement
- alternative solution for active consent
  - encrypted wide-area networking solutions with managed access control (e.g. SPHN's BiomedIT) and limited access to anonymized data (e.g. using the Beacon protocol with "handover" scenarios)
  - (genomic) data ownership by the individual "data donors, together with strong privacy protection by law

The BioMedIT network

BioMedIT provides researchers with access to a secure and protected computing environment for analysis of sensitive data without compromising data privacy



*DCC: Data Coordination Center

A project of SAMW ASSM Schweizerische Akademie der Medizinischen Wissenschaften Académie Suisse des Sciences Médicales Accademia Svizzera delle Scienze Mediche Swiss Academy of Medical Sciences

SIB Swiss Institute of Bioinformatics

Making Beacons Biomedical - Beacon v2

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  - ELIXIR AAI with compatibility to other providers (OAuth...)

Definitely breaks the "Relative Security by Design" Concept!

Mitigation by tailored implementation and security practices

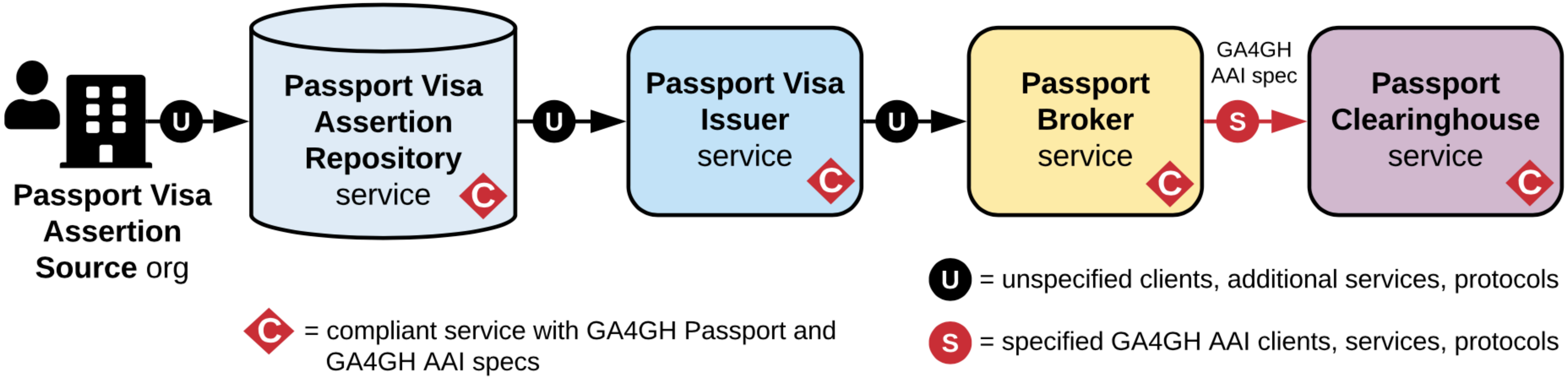
Global Alliance for Genomics & Health

elixir

GA4GH Passports

Global Alliance
for Genomics & Health

Communicating a user's data access authorizations



Passport Visa Assertion Source org

Passport Visa Assertion Repository service

Passport Visa Issuer service

Passport Broker service

GA4GH AAI spec

Passport Clearinghouse service

C = compliant service with GA4GH Passport and GA4GH AAI specs

U = unspecified clients, additional services, protocols

S = specified GA4GH AAI clients, services, protocols

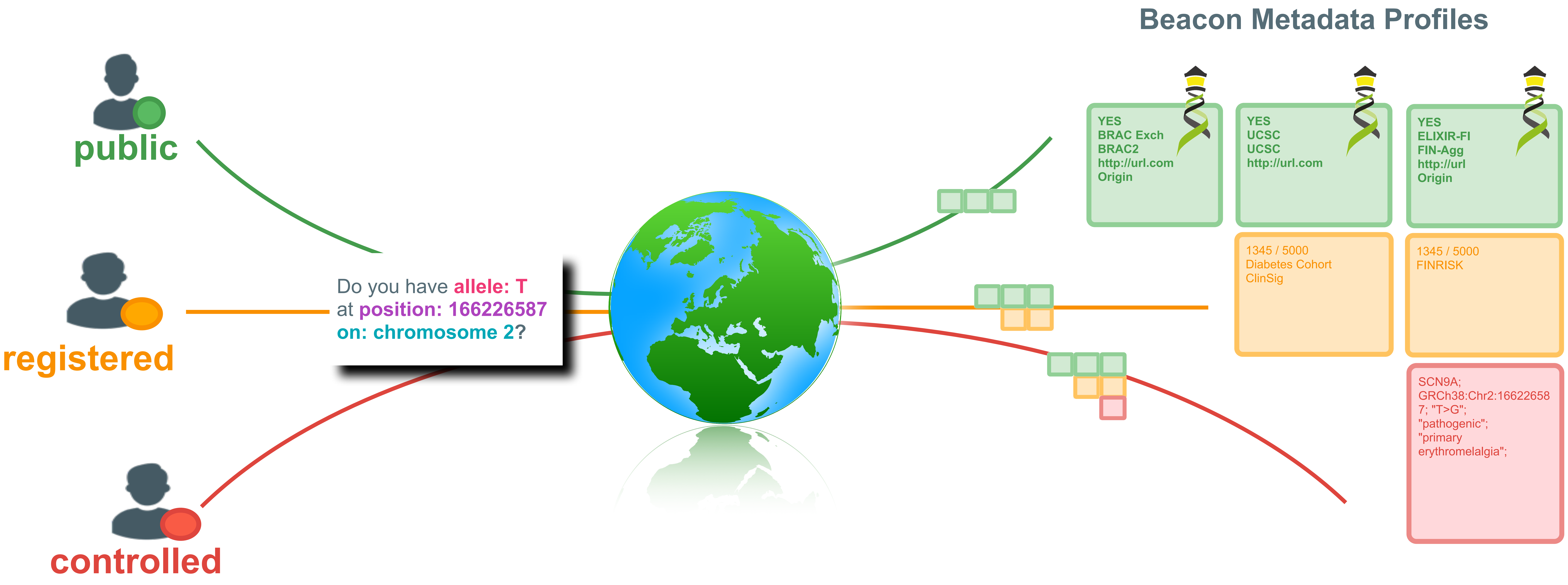
www.ga4gh.org/ga4gh-passports/

- format to communicate a user's data access authorizations based on either their role (e.g. researcher), affiliation, or access status
- works together with the GA4GH Authentication and Authorization Infrastructure (AAI) OpenID Connect Profile to streamline researchers' data access over federated data access protocols
- both standards approved in Dec 2019 with early implementation by Google Cloud services and ELIXIR

Google Cloud elixir

Empowering Beacon use through Access Levels

Integrating permissions and discovery



Beacon Metadata Profiles

public

registered

controlled

Do you have **allele: T** at **position: 166226587** on: **chromosome 2**?

YES
BRAC Exch
BRAC2
http://url.com
Origin

YES
UCSC
UCSC
http://url.com

YES
ELIXIR-FI
FIN-Agg
http://url
Origin

1345 / 5000
Diabetes Cohort
ClinSig

1345 / 5000
FINRISK

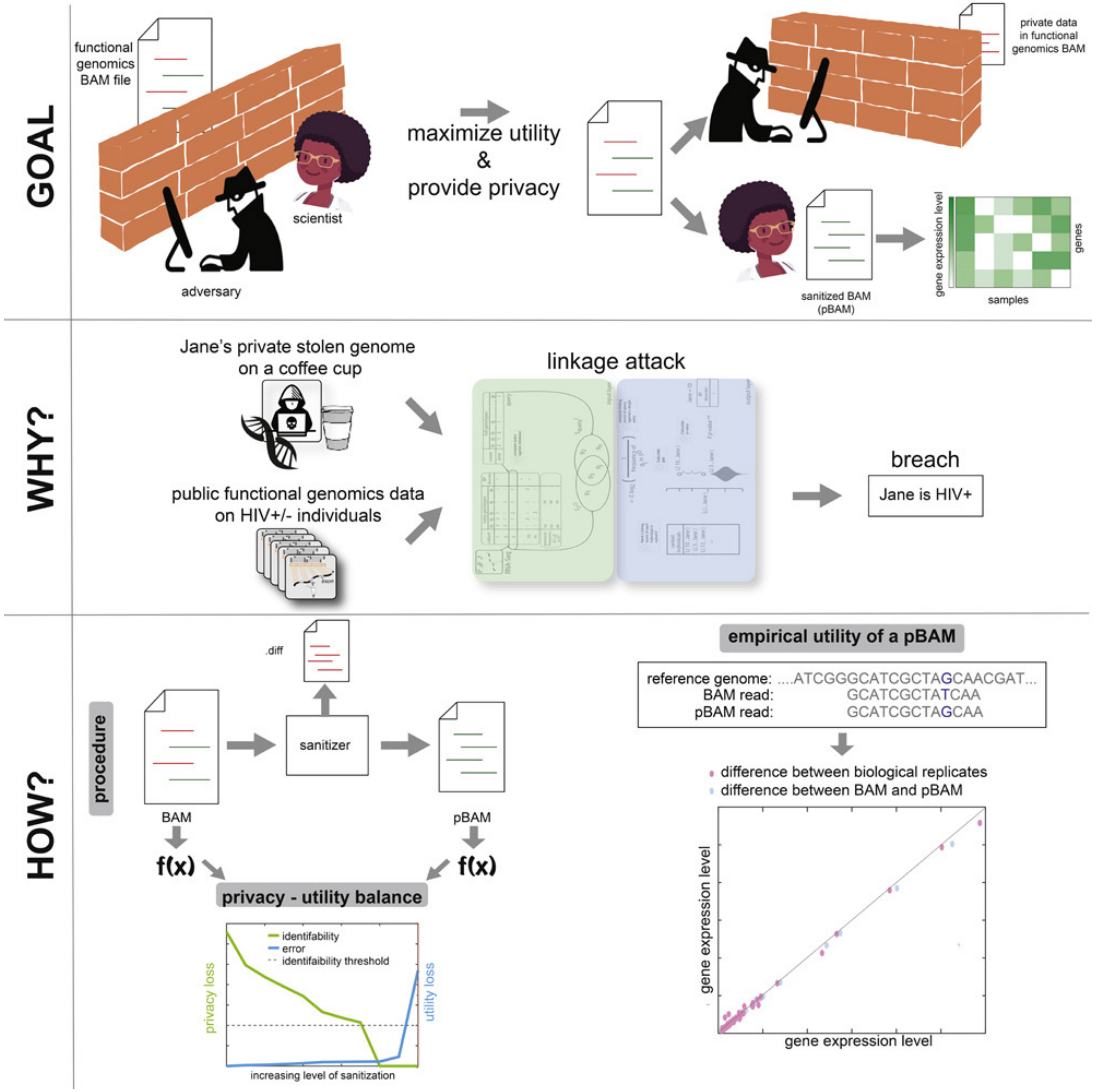
SCN9A;
GRCh38:Chr2:166226587; "T>G";
"pathogenic";
"primary erythromelalgia";

https://www.youtube.com/watch?v=LyfmvAs7LtQ&feature=youtu.be

Information Leakage from Functional Genomics Data

"Sanitize"...

- "functional" genomics data can be sanitized by removing features which are not relevant for the specific use cases
- an example could be the randomization of variant alleles in datasets where variant call specificity is of minor concern

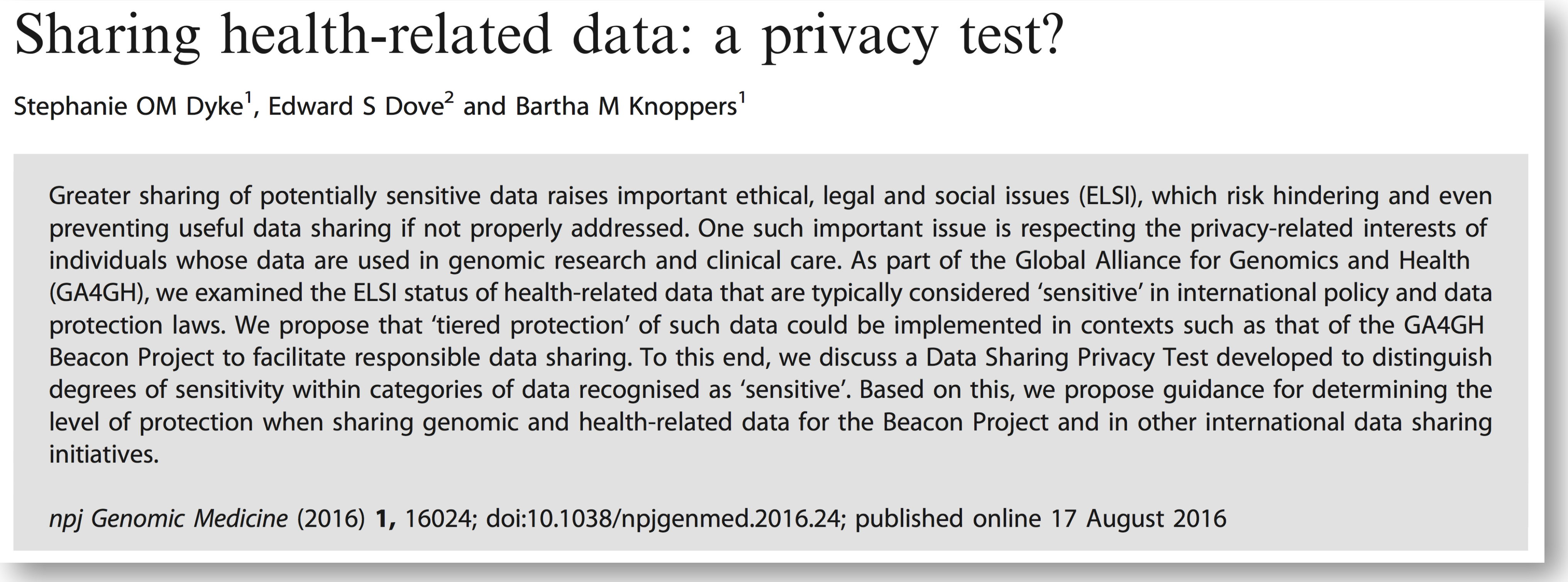

GOAL
functional genomics BAM file
scientist
adversary
maximize utility & provide privacy
private data in functional genomics BAM
sanitized BAM (pBAM)
gene expression level
samples
genes
WHY?
Jane's private stolen genome on a coffee cup
public functional genomics data on HIV+/- individuals
linkage attack
breach
Jane is HIV+
HOW?
procedure
.diff
sanitizer
BAM
pBAM
f(x)
f(x)
privacy - utility balance
identifiability
error
identifaibility threshold
privacy loss
utility loss
increasing level of sanitization
empirical utility of a pBAM
reference genome: ....ATCGGGCATCGCTAGCAACGAT...
BAM read: GCATCGCTATCAA
pBAM read: GCATCGCTAGCAA
difference between biological replicates
difference between BAM and pBAM
gene expression level
gene expression level

Gürsoy et al., 2020, Cell *183*, 905–917
November 12, 2020 © 2020 Elsevier Inc.
https://doi.org/10.1016/j.cell.2020.09.036

Health Related Data & Privacy

Considerations when evaluating risks of data sharing

- Is the genetic condition outwardly visible?
- How severe is it? (serious disease, penetrance, age of onset)
- Is it associated with what could be considered to be stigmatizing health information (e.g., associated with mental health, reproductive care, disability)?
- Is it familial (i.e., potential carrier status/reproductive implications for family/relatives)?
- Does it provide information about the likely geographical location of individuals?
- Does it provide information about ethnicity that may be considered potentially stigmatizing information?

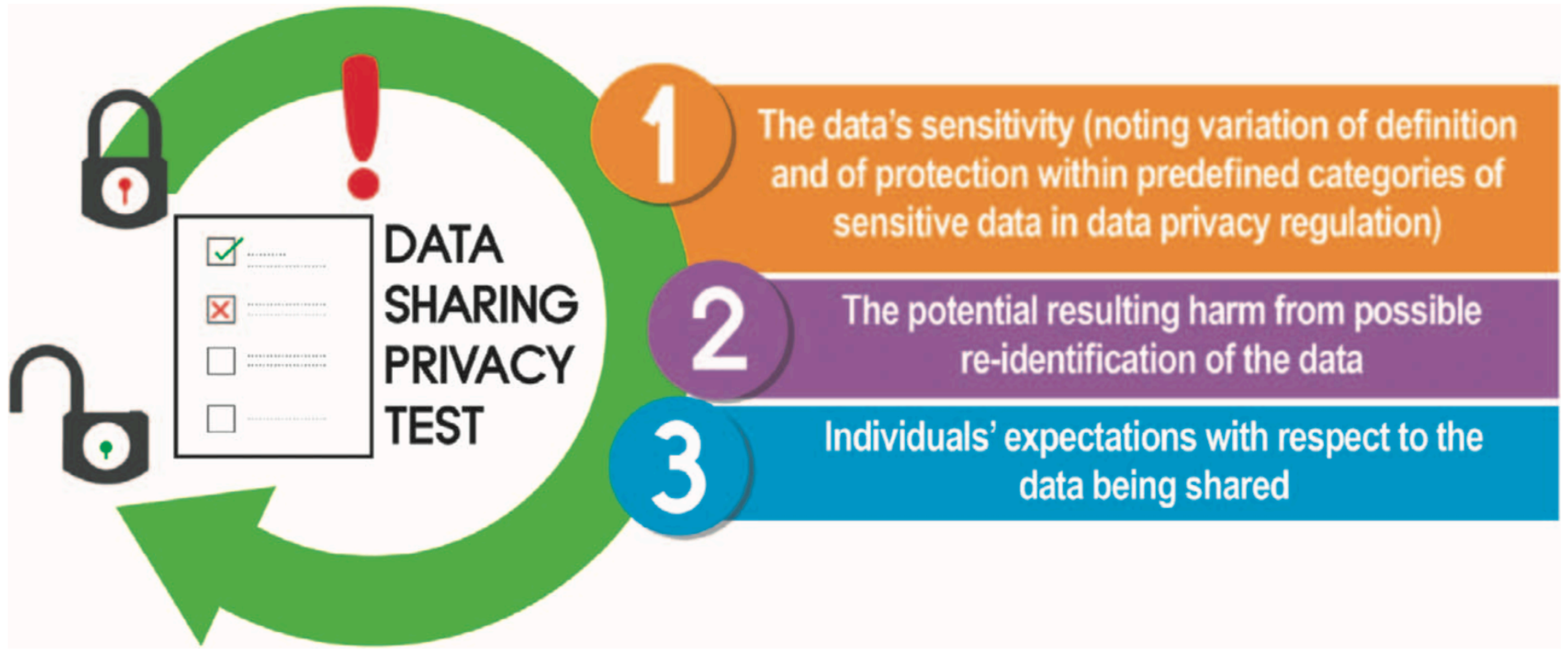


Sharing health-related data: a privacy test?

Stephanie OM Dyke[1], Edward S Dove[2] and Bartha M Knoppers[1]

Greater sharing of potentially sensitive data raises important ethical, legal and social issues (ELSI), which risk hindering and even preventing useful data sharing if not properly addressed. One such important issue is respecting the privacy-related interests of individuals whose data are used in genomic research and clinical care. As part of the Global Alliance for Genomics and Health (GA4GH), we examined the ELSI status of health-related data that are typically considered 'sensitive' in international policy and data protection laws. We propose that 'tiered protection' of such data could be implemented in contexts such as that of the GA4GH Beacon Project to facilitate responsible data sharing. To this end, we discuss a Data Sharing Privacy Test developed to distinguish degrees of sensitivity within categories of data recognised as 'sensitive'. Based on this, we propose guidance for determining the level of protection when sharing genomic and health-related data for the Beacon Project and in other international data sharing initiatives.

npj Genomic Medicine (2016) **1**, 16024; doi:10.1038/npjgenmed.2016.24; published online 17 August 2016



The data's sensitivity (noting variation of definition and of protection within predefined categories of sensitive data in data privacy regulation)

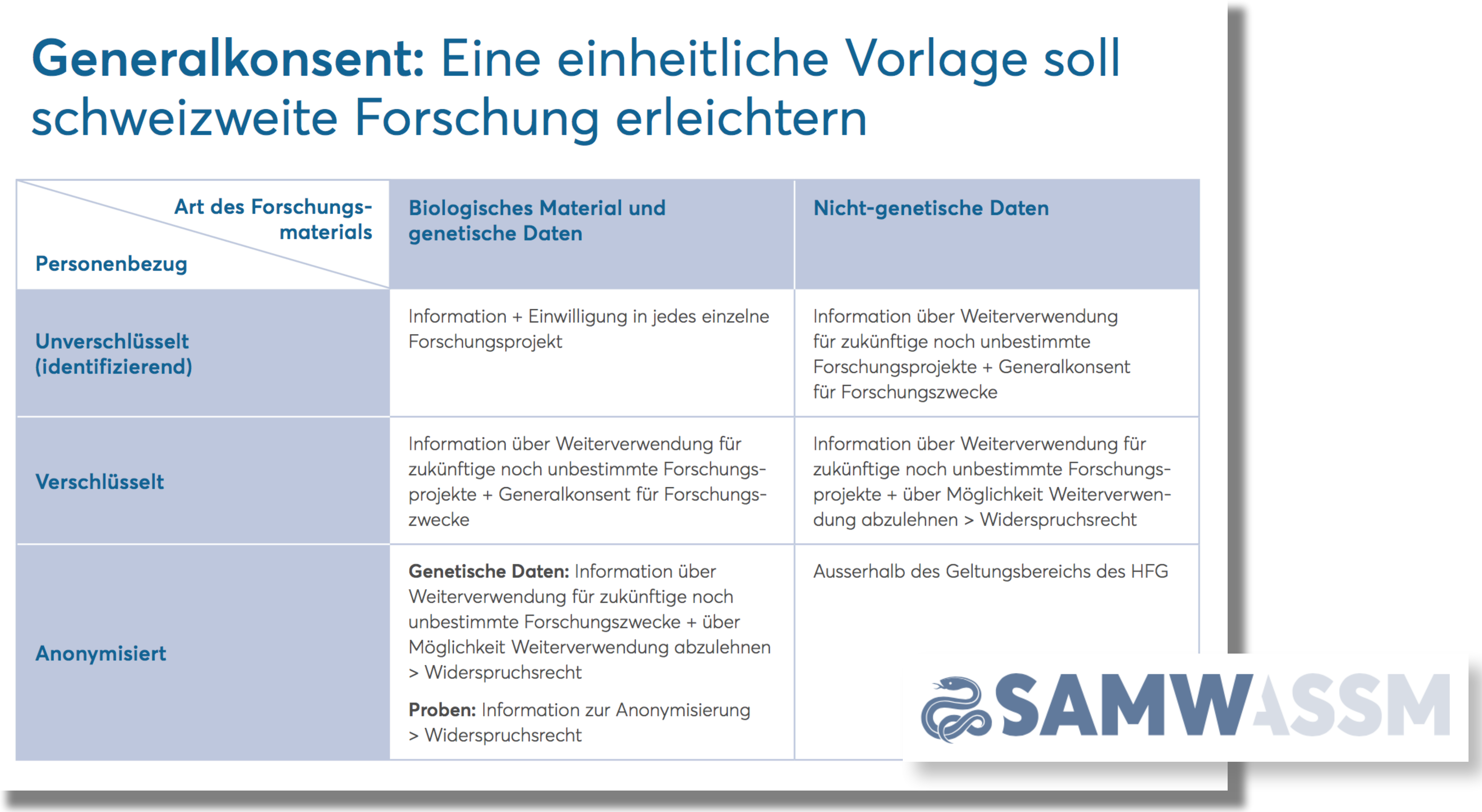
The potential resulting harm from possible re-identification of the data

Individuals' expectations with respect to the data being shared

Figure 1. The three steps of a Data Sharing Privacy Test to distinguish degrees of data sensitivity within categories of data recognised as 'sensitive'.

Modernizing Patient Consent

forward looking, transparent and technically feasible regulations for enabling access to research material and data while empowering ***patients***

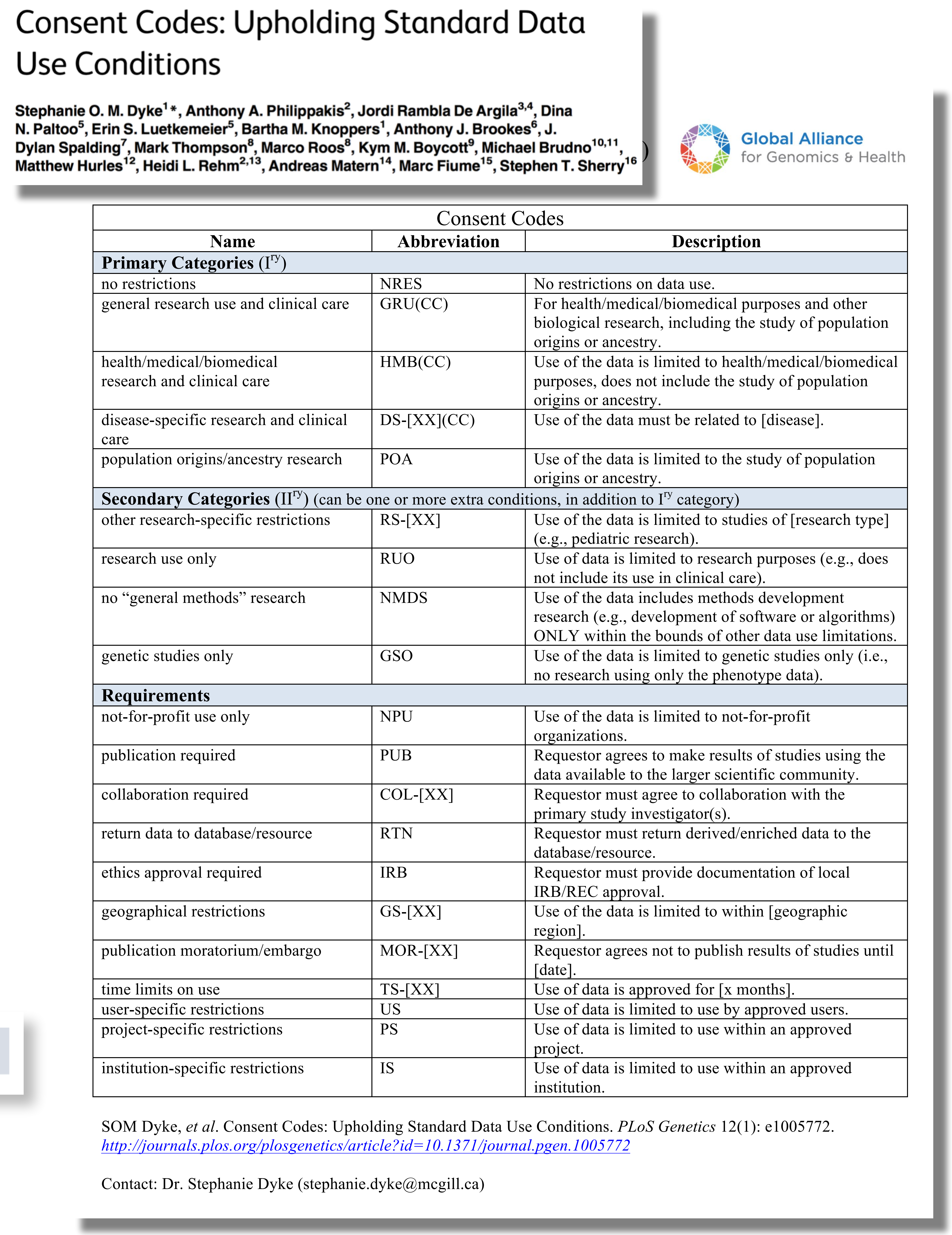


Generalkonsent: Eine einheitliche Vorlage soll schweizweite Forschung erleichtern

Art des Forschungsmaterials Personenbezug	Biologisches Material und genetische Daten	Nicht-genetische Daten
Unverschlüsselt (identifizierend)	Information + Einwilligung in jedes einzelne Forschungsprojekt	Information über Weiterverwendung für zukünftige noch unbestimmte Forschungsprojekte + Generalkonsent für Forschungszwecke
Verschlüsselt	Information über Weiterverwendung für zukünftige noch unbestimmte Forschungsprojekte + Generalkonsent für Forschungszwecke	Information über Weiterverwendung für zukünftige noch unbestimmte Forschungsprojekte + über Möglichkeit Weiterverwendung abzulehnen > Widerspruchsrecht
Anonymisiert	**Genetische Daten:** Information über Weiterverwendung für zukünftige noch unbestimmte Forschungszwecke + über Möglichkeit Weiterverwendung abzulehnen > Widerspruchsrecht **Proben:** Information zur Anonymisierung > Widerspruchsrecht	Ausserhalb des Geltungsbereichs des HFG

SAMW ASSM

Switzerland: Definition of a unified "Generalkonsent", to provide a single framework to manage permissions for access to patient derived material and related data



Consent Codes: Upholding Standard Data Use Conditions

Stephanie O. M. Dyke[1]*, Anthony A. Philippakis[2], Jordi Rambla De Argila[3,4], Dina N. Paltoo[5], Erin S. Luetkemeier[5], Bartha M. Knoppers[1], Anthony J. Brookes[6], J. Dylan Spalding[7], Mark Thompson[8], Marco Roos[8], Kym M. Boycott[9], Michael Brudno[10,11], Matthew Hurles[12], Heidi L. Rehm[2,13], Andreas Matern[14], Marc Fiume[15], Stephen T. Sherry[16]

Global Alliance for Genomics & Health

Consent Codes		
Name	**Abbreviation**	**Description**
Primary Categories (I^{ry})		
no restrictions	NRES	No restrictions on data use.
general research use and clinical care	GRU(CC)	For health/medical/biomedical purposes and other biological research, including the study of population origins or ancestry.
health/medical/biomedical research and clinical care	HMB(CC)	Use of the data is limited to health/medical/biomedical purposes, does not include the study of population origins or ancestry.
disease-specific research and clinical care	DS-[XX](CC)	Use of the data must be related to [disease].
population origins/ancestry research	POA	Use of the data is limited to the study of population origins or ancestry.
Secondary Categories (II^{ry}) (can be one or more extra conditions, in addition to I^{ry} category)		
other research-specific restrictions	RS-[XX]	Use of the data is limited to studies of [research type] (e.g., pediatric research).
research use only	RUO	Use of data is limited to research purposes (e.g., does not include its use in clinical care).
no "general methods" research	NMDS	Use of the data includes methods development research (e.g., development of software or algorithms) ONLY within the bounds of other data use limitations.
genetic studies only	GSO	Use of the data is limited to genetic studies only (i.e., no research using only the phenotype data).
Requirements		
not-for-profit use only	NPU	Use of the data is limited to not-for-profit organizations.
publication required	PUB	Requestor agrees to make results of studies using the data available to the larger scientific community.
collaboration required	COL-[XX]	Requestor must agree to collaboration with the primary study investigator(s).
return data to database/resource	RTN	Requestor must return derived/enriched data to the database/resource.
ethics approval required	IRB	Requestor must provide documentation of local IRB/REC approval.
geographical restrictions	GS-[XX]	Use of the data is limited to within [geographic region].
publication moratorium/embargo	MOR-[XX]	Requestor agrees not to publish results of studies until [date].
time limits on use	TS-[XX]	Use of data is approved for [x months].
user-specific restrictions	US	Use of data is limited to use by approved users.
project-specific restrictions	PS	Use of data is limited to use within an approved project.
institution-specific restrictions	IS	Use of data is limited to use within an approved institution.

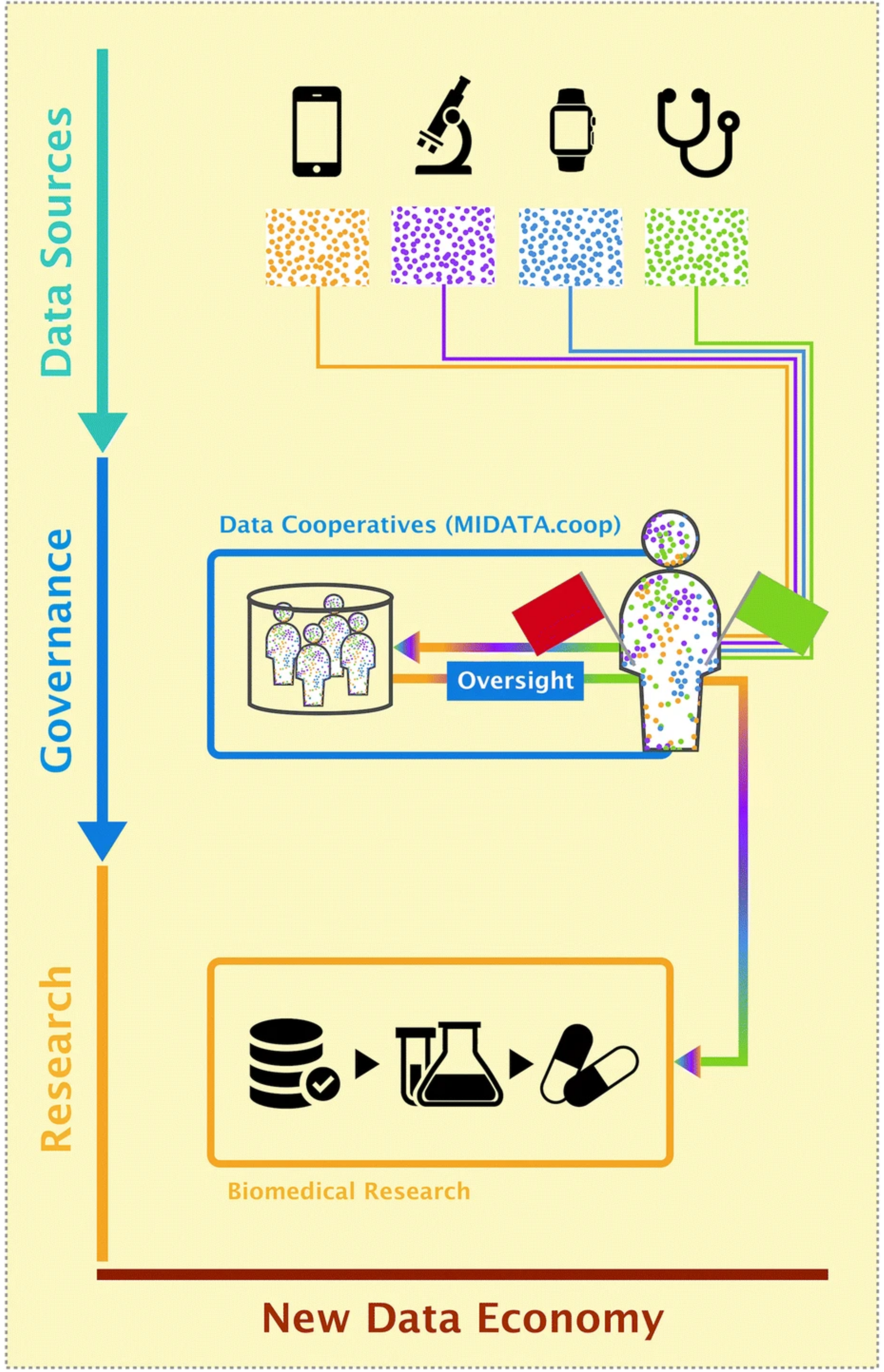
SOM Dyke, *et al*. Consent Codes: Upholding Standard Data Use Conditions. *PLoS Genetics* 12(1): e1005772. http://journals.plos.org/plosgenetics/article?id=10.1371/journal.pgen.1005772

Contact: Dr. Stephanie Dyke (stephanie.dyke@mcgill.ca)

Power to the People?!

Individuals as Owners & Managers of their Data

- (genomic) data ownership by the individual "data donors"
- supported by technological frameworks for data management and arbitration
- one vision here are "data cooperatives"
- need strong support from policy makers and financial sustainability support



graphic credit Manuel Schneider

Citizens aggregate data from different sources and make them available for research through data cooperatives. Cooperatives offer oversight mechanisms to filter data access requests and tools for the democratic governance of the data.

Blasimme, A., Vayena, E. & Hafen, E. **Democratizing Health Research Through Data Cooperatives**. *Philos. Technol.* 31, 473–479 (2018). https://doi.org/10.1007/s13347-018-0320-8

Genomic Data & Privacy - Key Areas

- **Re-identification**
  - identification of an individual based on sets of genomic variants they (or close relatives) carry - so one needs some genome data first
  - information to be gained is circumstantial (e.g. their genome is in a particular disease related dataset)
  - currently only risk with some practical use (e.g. **long-range familial attacks**)
- **Genotype-to-Phenotype (G2P) attacks**
  - determination of some disease risk or phenotypic features from a genome itself
  - needs access to genome data which is illegal in many jurisdictions (but technically more & more feasible)
  - real-world use cases are limited but abuse through wrong perception of utility
- **Genomic Determinism**
  - assignment of individual abilities and personal development trajectories from genomic profiling
  - topic of (some good, most bad) SciFi
  - but: **Wehret den Anfängen**!

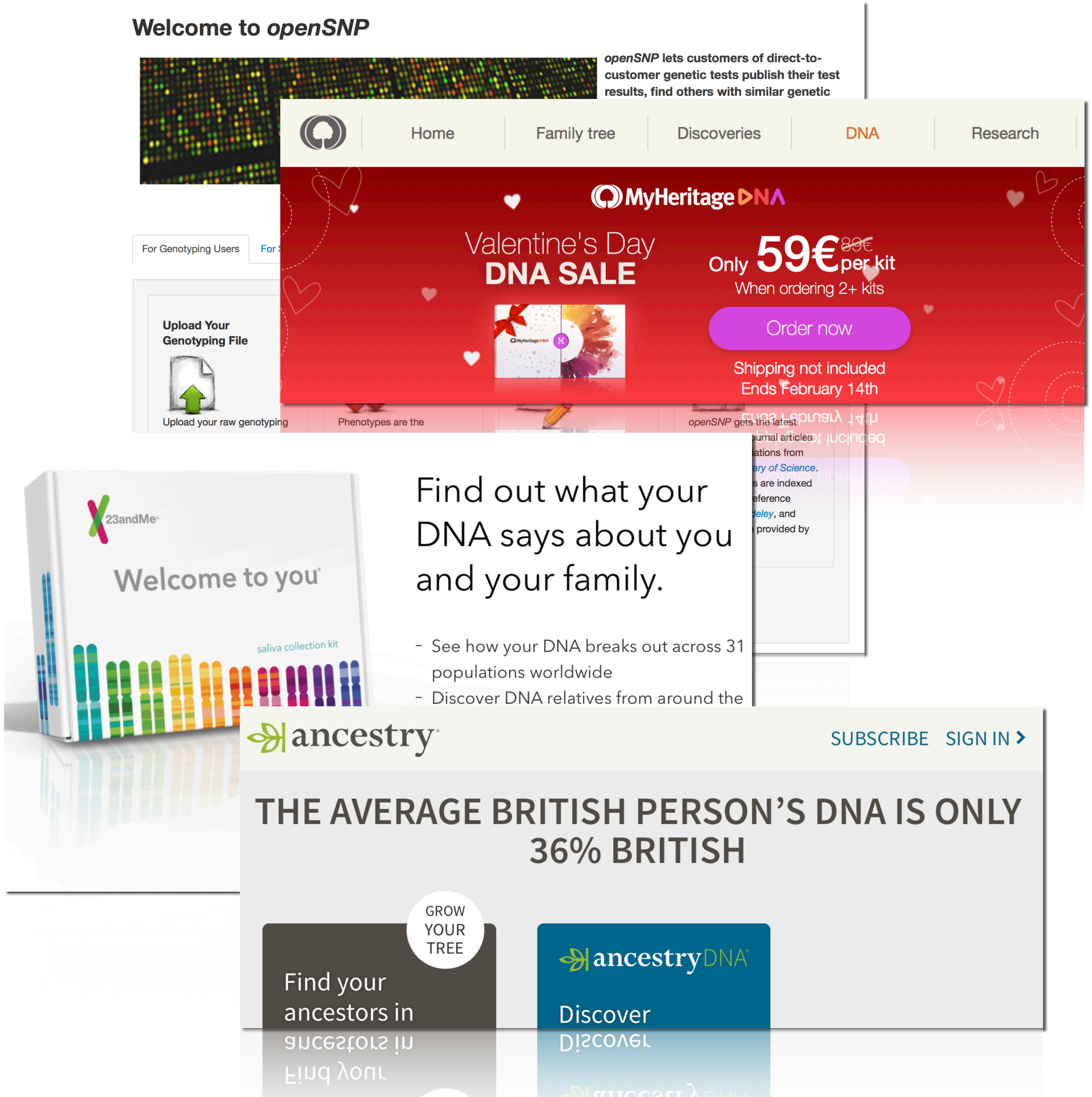
Genomic Data & Privacy - Some Take-Home Messages

- Many clinical and research applications in genomics **need vast numbers of genomes** to evaluate e.g. genotype-phenotype relationships
- Such data cannot simply be provided by a few reference data curation resources - and those again rely on multitudes of original data resources > **federated data access** + **data curation**
- Genomic data is considered to potentially expose unwilling individuals through **re-identification**/de-anonymization but also through direct information (genotype -> phenotype/disease)
- Legislative bodies and law enforcement have varying and *curious* approaches to "genomic privacy", with a mix of de-legalizing genomic data generation (e.g. in Switzerland) or strictly limiting its use while also using "eminent domain" to co-opt such data for criminal persecution in a possibly extending set of use cases

Share *YOUR* Genome data?

- The Beacon concept - balanced approach for accessing genome variant data from internationally distributed resources
- However: Genome data has the inherent “risk” of being identified and linked to a person

Solutions from Technology or Society? Discourse!



Welcome to *openSNP*

openSNP lets customers of direct-to-customer genetic tests publish their test results, find others with similar genetic

For Genotyping Users

Upload Your Genotyping File

Upload your raw genotyping

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DNA SALE

Only 59€ ~~89€~~ per kit
When ordering 2+ kits

Shipping not included
Ends February 14th

Find out what your DNA says about you and your family.

- See how your DNA breaks out across 31 populations worldwide
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THE AVERAGE BRITISH PERSON’S DNA IS ONLY 36% BRITISH

Find your ancestors in

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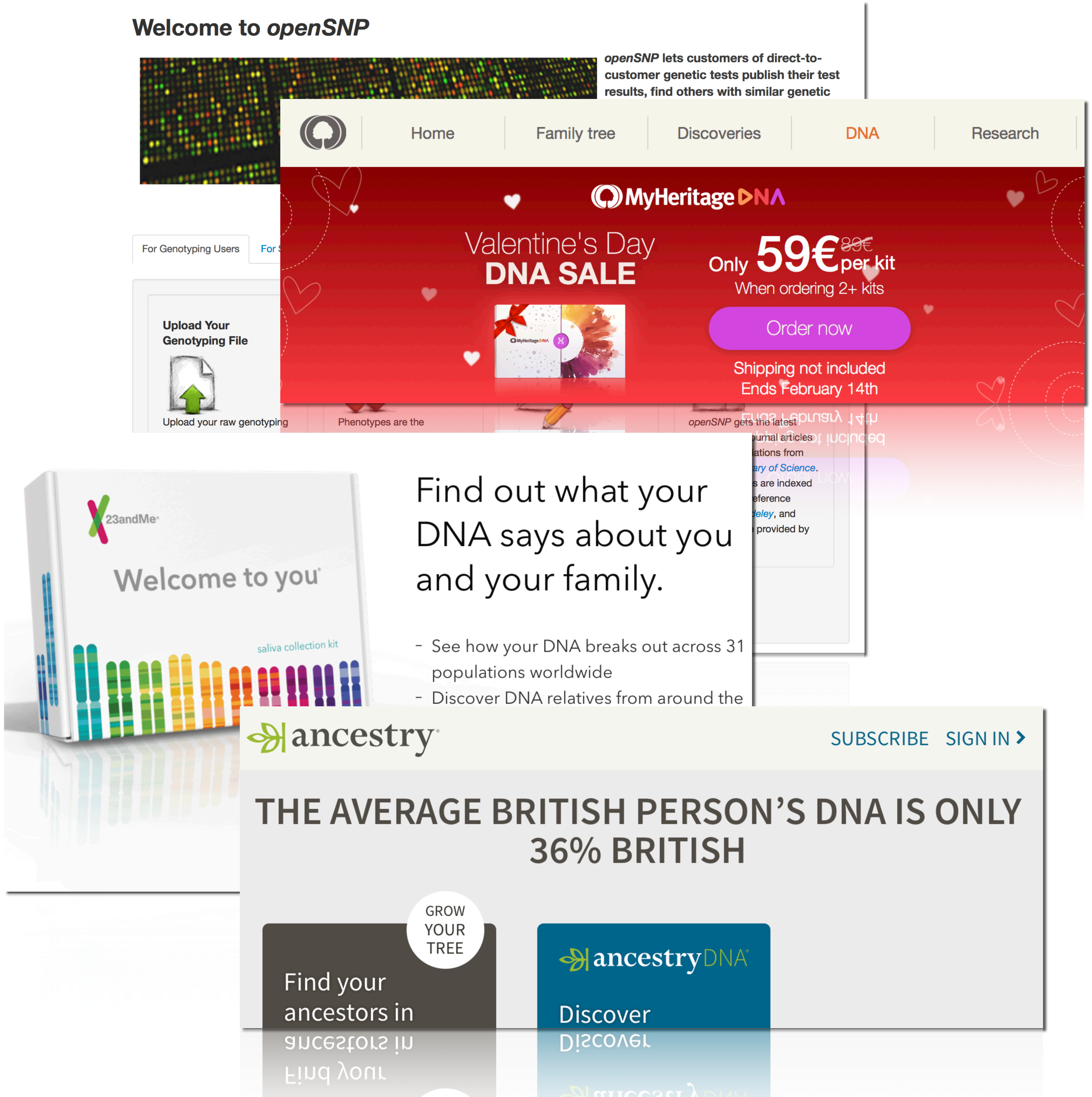
John Yuyi, NYT 2018-02-09



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BIO392 HS23

Exam

- 2023-10-11
- time: 09:30-10:30
- multiple (single + multiple) choice w/ one or two open questions

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