

Genetic and Molecular Privacy, Discrimination, and Designer Genomes

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Faculty Affiliate, the Information Society Project, Yale Law School
May 28th, 2024

Sometimes you “win” the
genetic lottery

Large muscles

Myostatin (MSTN) homozygous nulls (-/-) give lean and large muscles

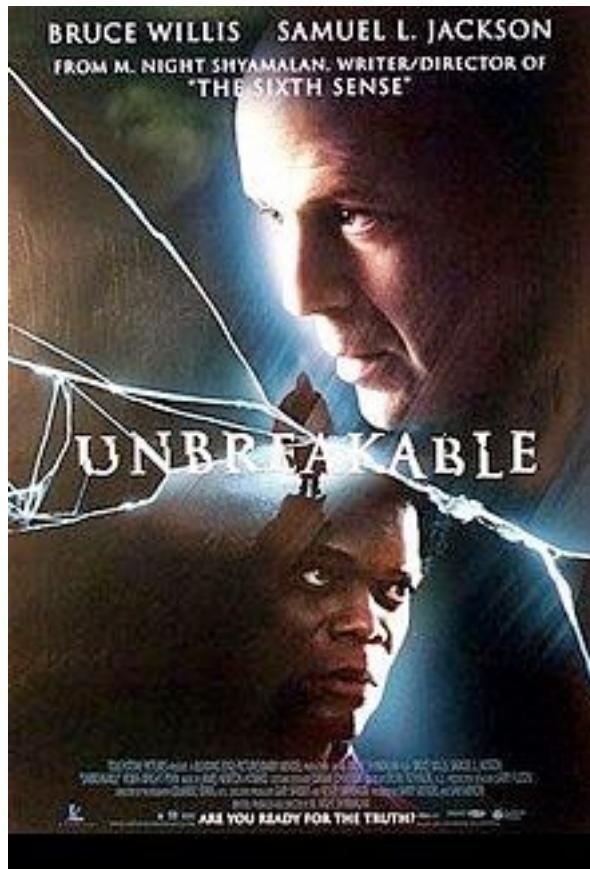


<http://thevoiceofnetizen.blogspot.com>



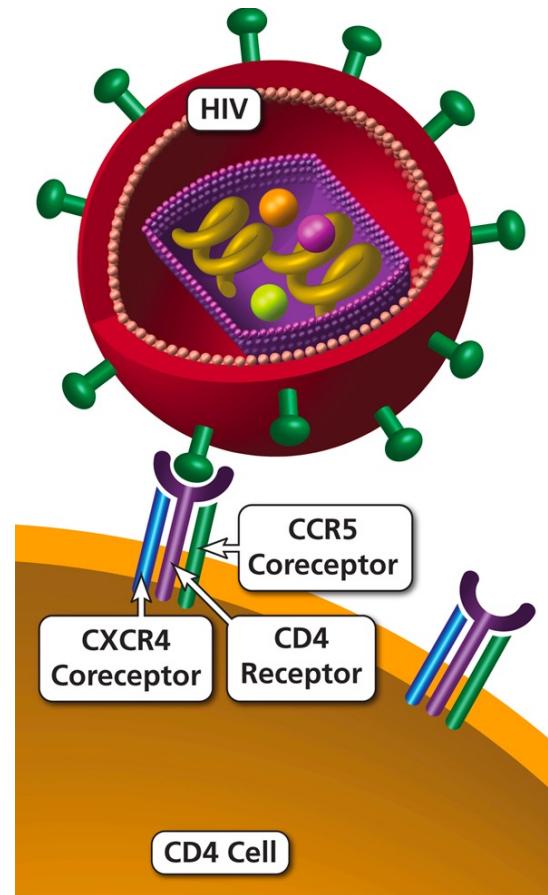
Strong bones

Low density lipoprotein receptor 5 (LRP5) heterozygotes (+/-) can have strong bones



Immunity to HIV Infection

C-C chemokine receptor type 5 (CCR5)
homozygous nulls (-/-) have HIV protection





The NEW ENGLAND JOURNAL of MEDICINE

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Long-Term Control of HIV by CCR5 Delta32/Delta32 Stem-Cell Transplantation

Gero Hütter, M.D., Daniel Nowak, M.D., Maximilian Mossner, B.S., Susanne Ganepola, M.D., Arne Müßig, M.D., Kristina Allers, Ph.D., Thomas Schneider, M.D., Ph.D., Jörg Hofmann, Ph.D., Claudia Kücherer, M.D., Olga Blau, M.D., Igor W. Blau, M.D., Wolf K. Hofmann, M.D., and Eckhard Thiel, M.D.

N Engl J Med 2009; 360:692-698 | February 12, 2009 | DOI: 10.1056/NEJMoa0802905

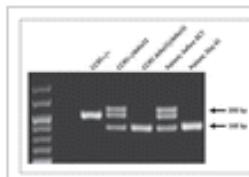
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[Abstract](#)[Article](#)[References](#)[Citing Articles \(312\)](#)

Infection with the human immunodeficiency virus type 1 (HIV-1) requires the presence of a CD4 receptor and a chemokine receptor, principally chemokine receptor 5 (CCR5). Homozygosity for a 32-bp deletion in the CCR5 allele provides resistance against HIV-1 acquisition. We transplanted stem cells from a donor who was homozygous for CCR5 delta32 in a patient with acute myeloid leukemia and HIV-1 infection. The patient remained without viral rebound 20 months after transplantation and discontinuation of antiretroviral therapy. This outcome demonstrates the critical role CCR5 plays in maintaining HIV-1 infection.

MEDIA IN THIS ARTICLE

FIGURE 1



Genotyping of CCR5 Alleles.

FIGURE 2



Sometimes you “lose” the
genetic lottery

Hypertrophic Cardiomyopathy

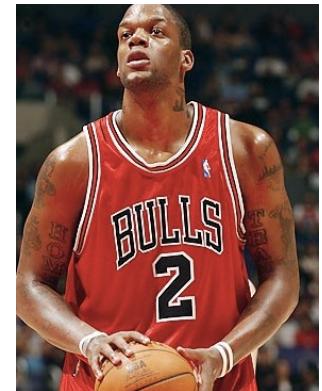
- 1 in 500
- Most common cause of sudden death in young athletes
- Left ventricular hypertrophy, outflow obstruction
- 27 different genes, 100s of mutations¹



¹ Bos, JM et al, J Am col Cardiol, 2009; 54:201-211

Reggie Lewis

Eddy Curry



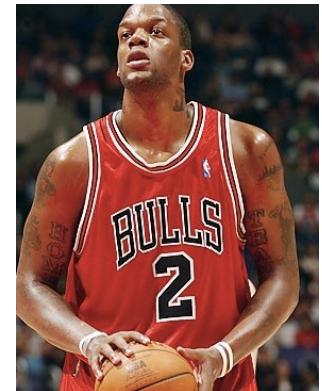
In the 2004–05 season the Bulls improved by 28 wins and made the playoffs as the 22-year-old Curry led the team in scoring.

Then, he was hospitalized with an irregular heartbeat.

This caused him to miss the last 13 games of the regular season and the entire playoffs.

On June 24, 2005, heart specialists cleared Curry to resume practice.

Eddy Curry

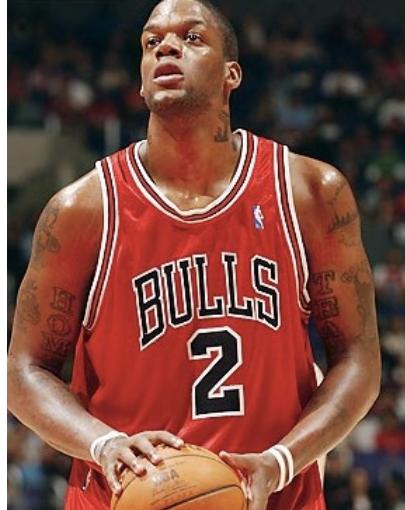


“Bulls General Manager John Paxson said he understood the privacy issues involved but insisted the Bulls’ concern was a situation similar to those of former Boston Celtics guard Reggie Lewis or Loyola Marymount star Hank Gathers—players with hypertrophic cardiomyopathy who collapsed and died.”

The Bulls had offered Curry an annuity of \$400,000 per year for 50 years if he took, and failed, the genetic test.

He refused.

Eddy Curry



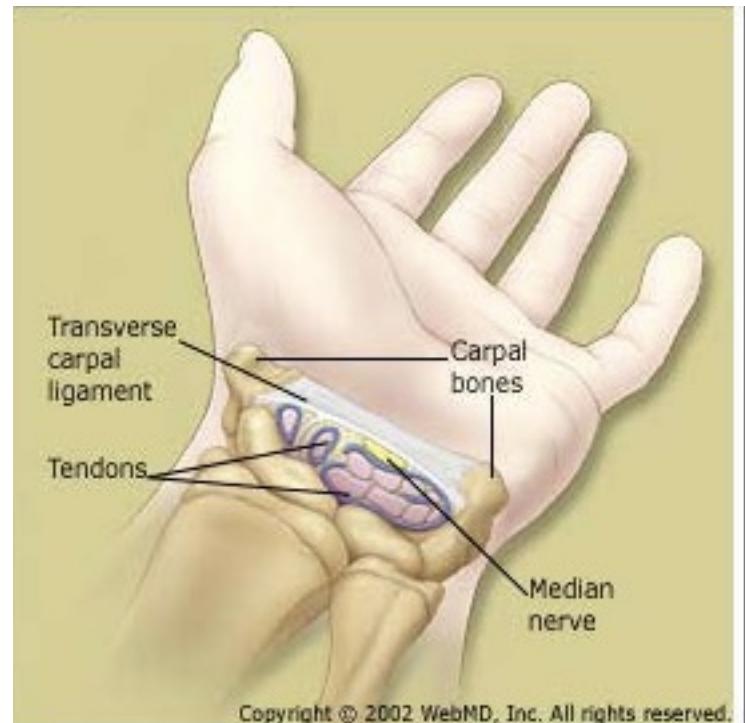
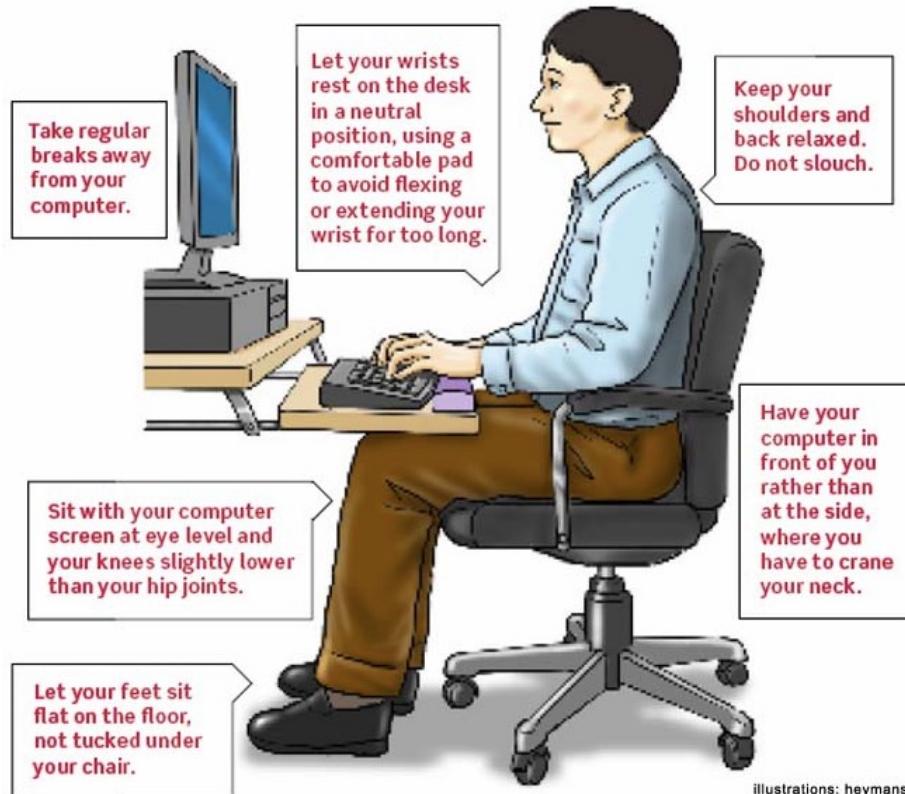
Curry was then traded to the Knicks (2005), then traded to the Timberwolves (2011), the Heat (2011), the Mavericks (2012), and then the Zhejiang Golden Bulls (2012). then, the Zhuhai Wolf Warriors (2018).

Burlington Northern and Santa Fe (BNSF) Railway



In 2001, Increasing Claims of Carpal Tunnel Syndrome at BNSF

Prevent carpal tunnel syndrome with better posture



#162500

NEUROPATHY, HEREDITARY, WITH LIABILITY TO PRESSURE PALSIES; HNPP*Alternative titles; symbols*

POLYNEUROPATHY, FAMILIAL RECURRENT
TOMACULOUS NEUROPATHY

Phenotype-Gene Relationships

Location	Phenotype	Phenotype MIM number	Pheno map key	Gene/Locus	Gene/Locus MIM number
17p12	Neuropathy, recurrent, with pressure palsies	162500	3	PMP22	601097

Clinical Synopsis**TEXT**

A number sign (#) is used with this entry because hereditary neuropathy with liability to pressure palsies (HNPP) can be caused by deletion of the gene encoding peripheral myelin protein-22 (PMP22; 601097); duplication of PMP22 causes Charcot-Marie-Tooth disease type 1A (CMT1A; 118220). Point mutation in PMP22 may result in HNPP or CMT1A.

Clinical Features

This disorder may have been described first by [De Jong \(1947\)](#) who reported a family in which 1 man and 4 women in 3 generations had recurrent peroneal neuropathy after digging potatoes in a kneeling position. Families were reported by [Davies \(1954\)](#) and by [Earl et al. \(1964\)](#). The latter group found that motor nerve conduction velocity (NCV) was reduced in some clinically normal family members. [Staal et al. \(1965\)](#) studied a family in which members in 4 generations showed transient unilateral peroneal palsies. The neuropathy manifested itself especially after prolonged work in a kneeling



Online Mendelian Inheritance in Man®

An Online Catalog of Human Genes and Genetic Disorders
Updated 8 September 2014

<http://omim.org/entry/162500>

Genetic Testing Ordered to Rule out Cause for Carpal Tunnel

The New York Times

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February 10, 2001

Commission Sues Railroad To End Genetic Testing In Work Injury Cases

By TAMAR LEWIN

The Equal Employment Opportunity Commission yesterday filed its first court action challenging genetic testing by an employer, the Burlington Northern Santa Fe Railway Company.

The commission asked a court to order the railroad to end what it called the company's nationwide policy of requiring all union members who claim work-related carpal tunnel syndrome to provide blood samples for a DNA test for a condition that may predict some forms of carpal tunnel syndrome.

According to the court papers filed in Federal District Court in Sioux City, Iowa, the railroad's employees were asked for blood samples but not asked to consent to their use for genetic testing, and at least one individual who refused to provide a sample because he suspected it would be used for genetic testing was threatened with discharge if he did not submit it.

The railroad said that it had not sought blood tests on every worker who filed a claim for carpal tunnel syndrome, and that it had not tested any employees without their consent.

"According to my understanding, there were 125 claims filed for workers compensation for carpal tunnel syndrome since March 2000, and 18 instances in which we sought genetic testing," said Richard Russack, a railroad spokesman. "I'm under the impression that all 18 said yes, and were tested. And my understanding is that we have not threatened anybody with disciplinary action, or taken any disciplinary action."

Mr. Russack said that neither the employment commission nor the rail workers union, the Brotherhood of Maintenance of Way Employees, had ever contacted the railroad about the issue or sought mediation, and that the railroad first heard of the genetic-testing issue yesterday, from a reporter. The railroad is a unit of the Burlington Northern Santa Fe Corporation, which is based in Fort Worth.

One worker named in the court papers, George Avary, contended that a month after he filed a claim for work-related carpal tunnel syndrome last October, the railroad told him that he had to provide a blood sample. The papers said that he refused to do so, and that in January, the railroad told him he would be subject to discipline for his refusal.

Two other workers named in the court papers said that they had allowed blood samples to be taken, but that they did not know the samples would be used for genetic testing. The union has also brought charges on behalf of all its members.

Carpal tunnel syndrome, which can cause pain and numbness in the hands and arms, is usually caused by repetitive motion, but in some cases may have a genetic component.

"The affidavits we have tell of workers threatened with losing their jobs because of genetic testing," said Ida L. Castro, the chairwoman of the employment commission.

EEOC vs. BNSF



In 2002, the U.S. Equal Employment Opportunity Commission (EEOC) [eeoc.gov] settled the first lawsuit alleging this type of discrimination.

BNSF was ordered to pay \$2.2 million for violating the ADA (Americans with Disabilities Act) by genetically testing or seeking to test 36 of its employees without their knowledge or consent

Maybe a change is needed?

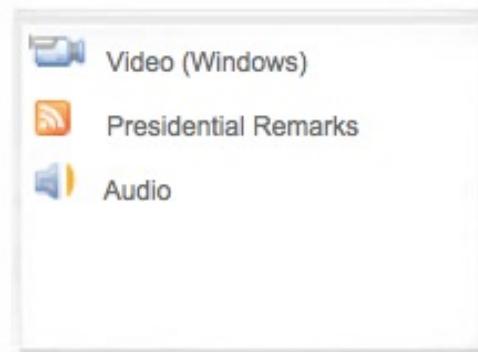


President Bush Signs H.R. 493, the Genetic Information Nondiscrimination Act of 2008

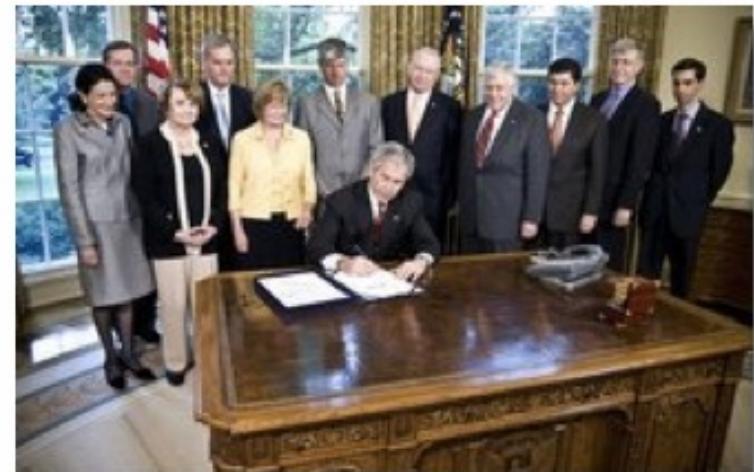
Oval Office

2:05 P.M. EDT

THE PRESIDENT: I want to thank the members of Congress who've joined us as I sign the Genetic Information Nondiscrimination Act, a piece of legislation which prohibits health insurers and employers from discriminating on the basis of genetic information. In other words, it protects our citizens from having genetic information misused, and this bill does so without undermining the basic premise of the insurance industry.



I also want to pay homage today to -- and not only to members of the Congress who are behind me, but also to Senator Ted Kennedy, who has worked for over a decade to get this piece of legislation to a President's desk. All of us are so pleased that Senator Kennedy has gone home, and our thoughts and prayers are with him and his family.



Now it's my honor to sign the Genetic Information Nondiscrimination Act.

(The Act was signed.)

Thank you. (Applause.)

END 2:06 P.M. EDT

GINA – what does it do for health insurance?

- Prohibits issuers of health insurance from discrimination on the basis of the genetic information of enrollees, for:
 - (1) eligibility,
 - (2) coverage,
 - (3) underwriting,
 - (4) premium-setting decisions.



GINA – for employment?

- Prevents employers from using genetic information in employment decisions, including: hiring, firing, promotions, pay, and job assignments.
- Prohibits employers or other covered entities (e.g. unions) from requiring or requesting genetic information and/or genetic tests as a condition of employment



What GINA does not cover

- GINA does not apply to employers with fewer than 15 employees.
- The US military, the Indian Health Service, the Veterans Health Administration, or the Federal Employees Health Program.
- The law does not cover long term care insurance, life insurance, or disability insurance.

Florida expanded GINA

Florida Enacts Sweeping Genetic Protection Law



July 1, 2020 - Florida just became first state to [enact a law](#) that protects genetic information from life, long-term care and disability insurers, which are exempt from the national protections provided by the [Genetic Information Nondiscrimination Act](#) (GINA).



FLORIDA HOUSE OF REPRESENTATIVES

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[Home](#) > [Bills](#) > [HB 1189](#)

HB 1189 -Genetic Information for Insurance Purposes

General Bill by Sprowls and Williamson (CO-SPONSORS) Aloupis; Buchanan; Burton; DiCeglie; Duggar

Genetic Information for Insurance Purposes: Prohibits life insurers & long-term care insurers from calculating premium rates based on genetic information; prohibits such insurers from taking certain actions relating to

Effective Date: July 1, 2020

Last Event: Filed on Thursday, January 9, 2020 10:40 AM

Florida becomes first state to protect DNA from life, disability insurers

HB 1189 would expand protections against using genetic data for insurance purposes.



By **Renzo Downey** on July 1, 2020

Applications

GINA and “The Mystery of the Devious Defecator”

IN THE UNITED STATES DISTRICT COURT
FOR THE NORTHERN DISTRICT OF GEORGIA
ATLANTA DIVISION

By [Fiona W. Ong](#) on June 24, 2015

POSTED IN [EMPLOYMENT DISCRIMINATION, LITIGATION](#)

JACK LOWE and DENNIS REYNOLDS,	:	
Plaintiffs,	:	
v.	:	
ATLAS LOGISTICS GROUP RETAIL SERVICES (ATLANTA), LLC,	:	CIVIL ACTION NO. 1:13-CV-2425-AT
Defendant.	:	

Atlas Logistics Group Retail Services (Atlanta), LLC (“Atlas”) operates warehouses for the storage of products sold at a variety of grocery stores. So one could imagine Atlas’s frustration when a mystery employee began habitually defecating in one of its warehouses.¹ To solve the mystery of the devious

¹ Apparently, this problem is not as rare as one might imagine. *See Ashtari, EPA Employees Asked To Stop Pooping In The Hallway*, Huffington Post (June 26, 2014 10:59 AM).

'Devious Defecator' Case Tests Genetics Law



Dennis Reynolds, left, and Jack Lowe in Atlanta in May. The men sued their employer after they were asked to take a DNA test. Bryan Meltz for The New York Times



Why the 'devious defecator' case is a landmark for US genetic-privacy law

Nature explores the impact of the first US court decision over how employers use genetic information.

Natasha Gilbert

25 June 2015



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A US company is the first to face penalties under the Genetic Information Nondiscrimination Act (GINA), a law that protects the privacy of genetic information. On 22 June, a federal court jury in Georgia awarded US\$2.25 million to two men whose employer tested their DNA, seeking to identify who had repeatedly left faeces in one of its warehouses.

The firm, Atlas Logistics Group Retail Services, a grocery distributor in Atlanta, Georgia, asked employees Jack Lowe and Dennis Reynolds to give cheek swabs in 2012. Atlas sent their DNA to a lab for genetic comparison with the offending faecal matter. The tests showed that Lowe and Reynolds' DNA was not a match.

In 2013, the workers sued Atlas. The case, nicknamed the 'mystery of the devious defecator' by US district court judge Amy Totenberg, is the first brought under GINA to go to trial. Here, *Nature* explains why the ruling matters.

EUROPE

A Forensic Approach to a Sidewalk Nuisance

By JIM YARDLEY FEB. 22, 2014

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NAPLES, Italy — Problems? Yes, conceded Tommaso Sodano, the vice mayor here, Naples has problems. Unpaid debts have reportedly topped \$2 billion. Many streets are pocked with potholes. The police department is underfunded, organized crime operates like a shadow state, and illegal dumps are scattered around what is



THE BALTIMORE SUN



OPINION MARKETPLACE SERVICES

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coders genetic

Recommend 150

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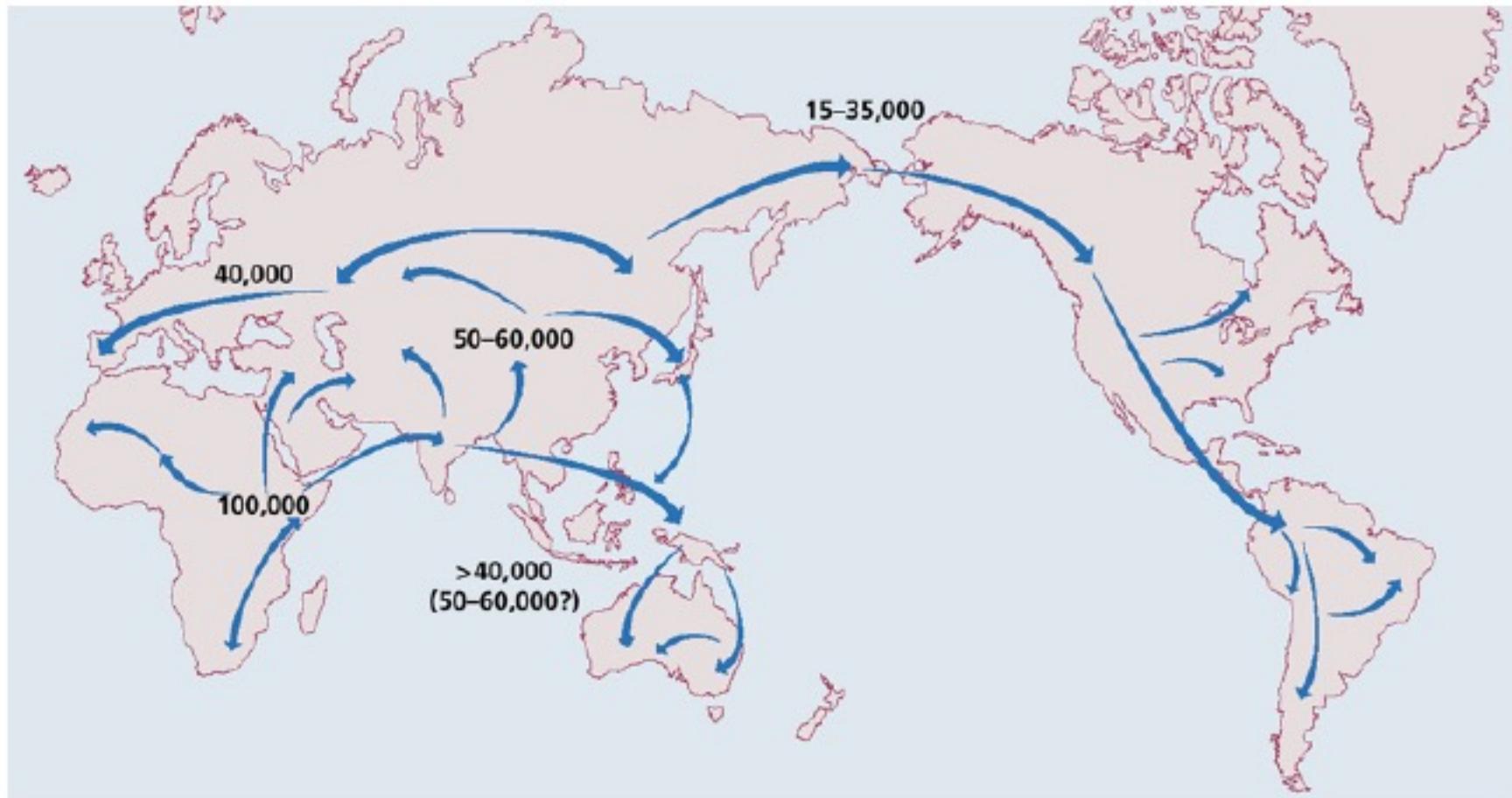
Tweet

all over the ritzy Scarlett Place condominium near the culprit: mandated DNA tests for every dog in the

poop. We bring guests over and this is what they're one who lives in this building," says Steve Frans, the of the dozens of dogs in the luxury building is behind

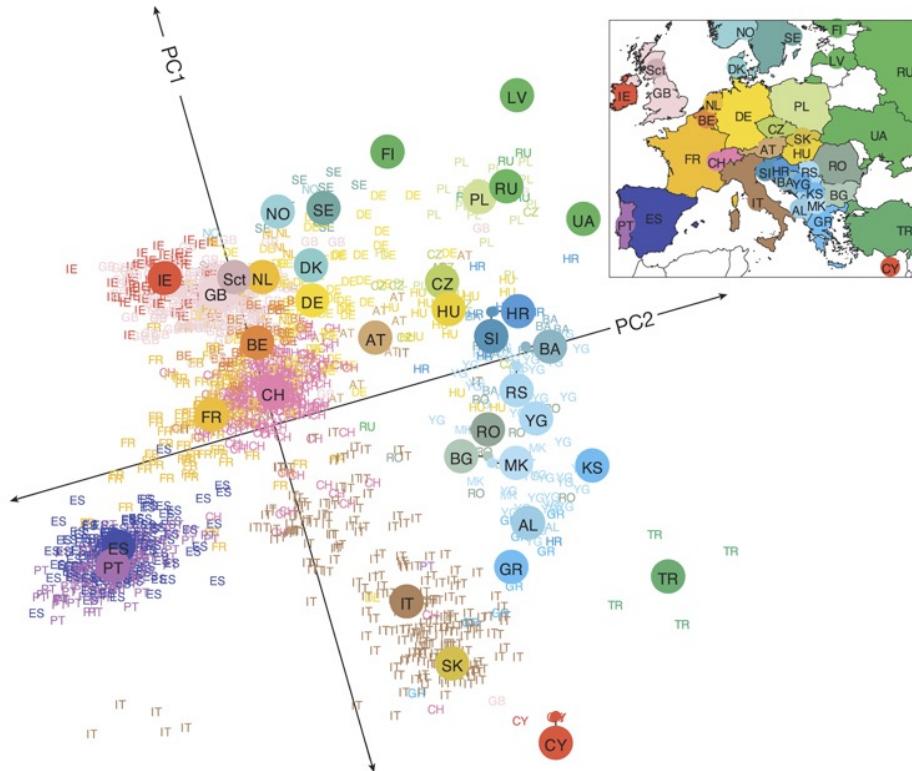
All the DNA you leave behind
tells a story

You are an echo of your ancestors

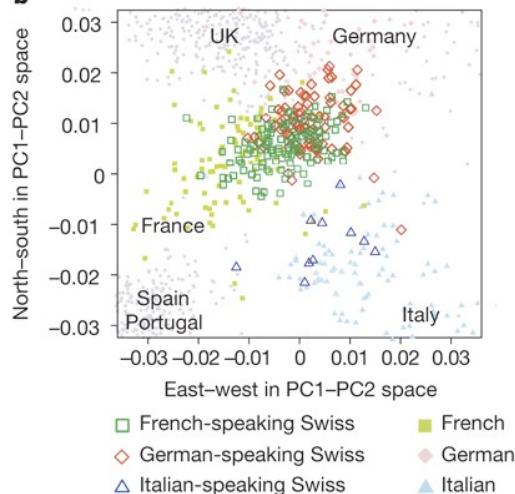


Genotype data can predict your birthplace

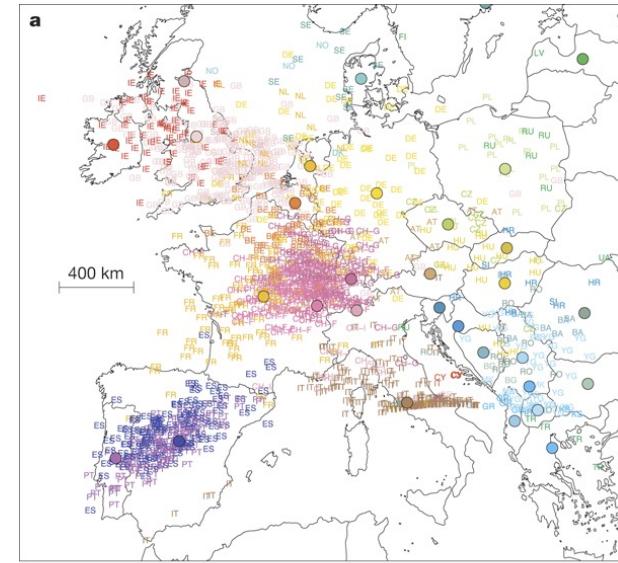
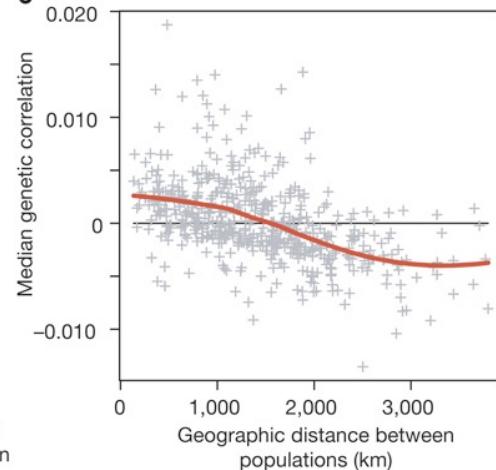
a



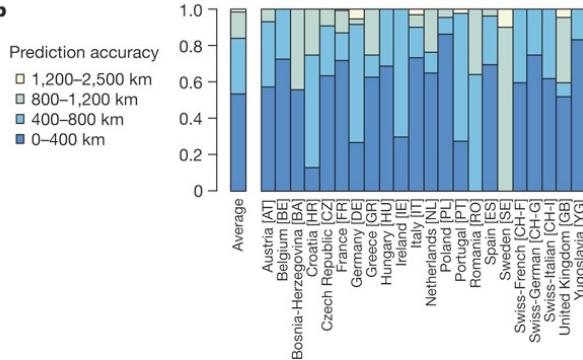
b



c



b

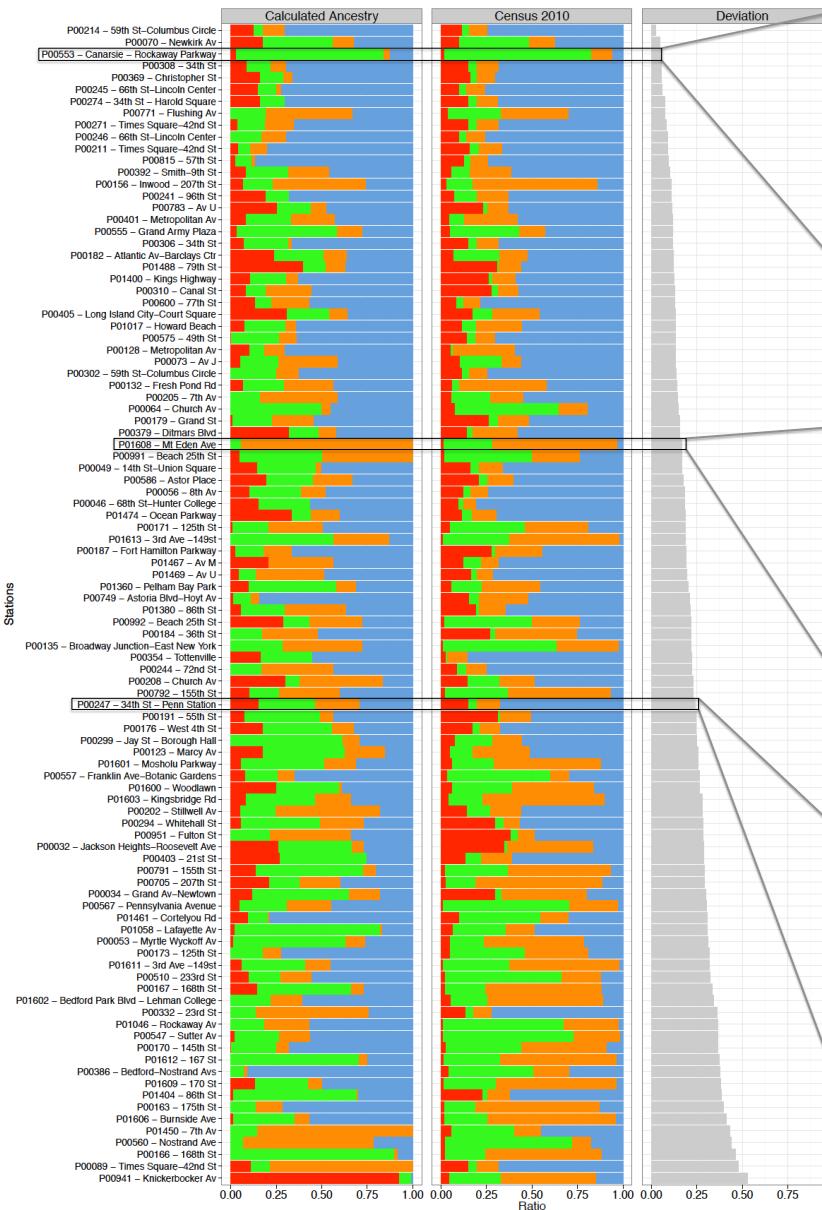


Genes mirror geography within Europe
Novembre et al., 2008

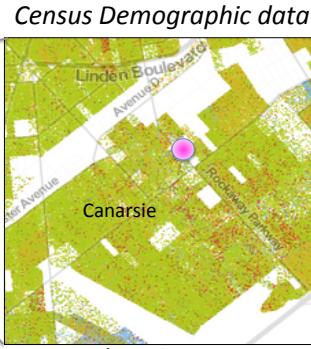
Subway DNA shows ancestry “molecular echo”

A

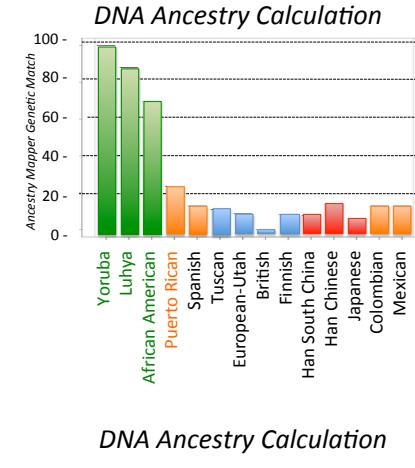
Admixture Proportion: Asian (red) Black (green) Hispanic (orange) White (blue)



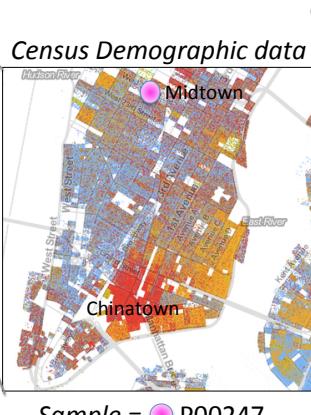
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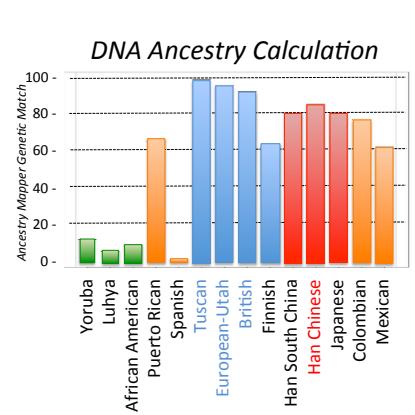
C



F



G



Artist creates faces from DNA left in public

By Natalie Angley, CNN

updated 3:10 PM EDT, Wed September 4, 2013 | Filed under: **Innovations**



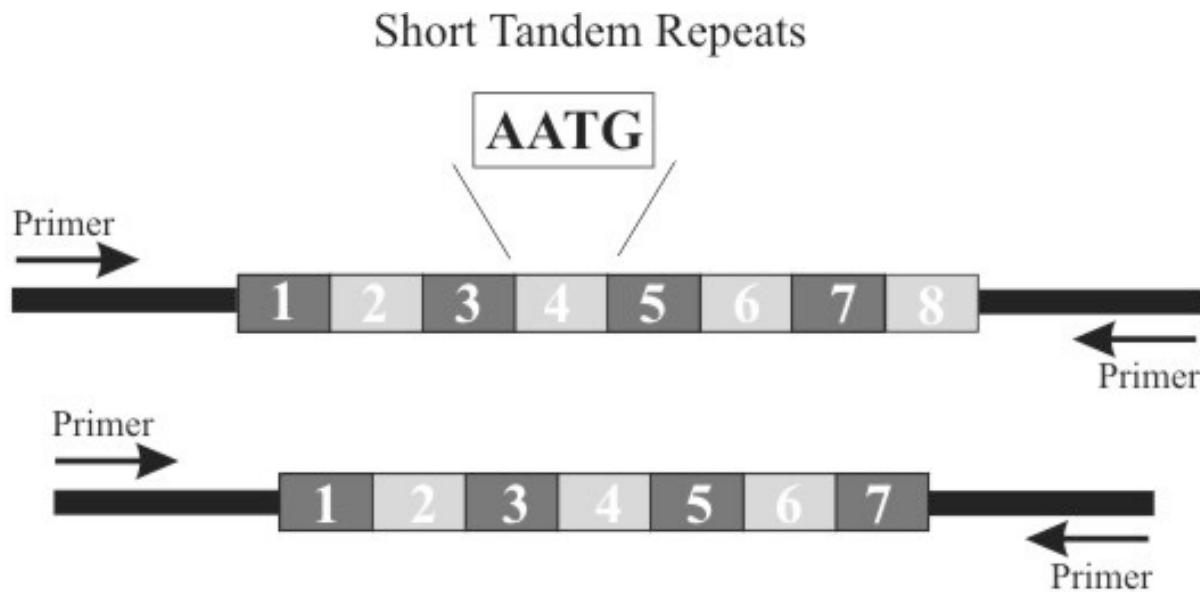
from Heather Dewey-Hagborg's "Stranger Visions"

<http://www.cnn.com/2013/09/04/tech/innovation/dna-face-sculptures/>

You are also an echo of your family

Relationship	% common DNA (average)
Identical Twin	100%
Parent/child/sibling	50%
Grandparent/Aunt/Uncle/Niece/ Nephew/Half-sibling	25%
1 st Cousin	12.5%
2 nd Cousin	3.1%
3 rd Cousin	0.8%
4 th Cousin	0.2%

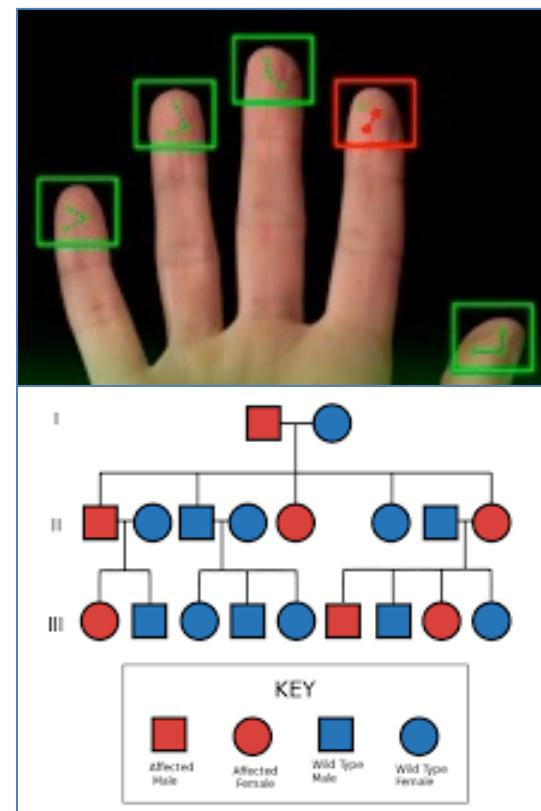
FBI's Combined DNA Index System (CODIS) – Uses 13 Core STR Loci and 7 addition loci



The flanking regions where PCR primers bind are constant

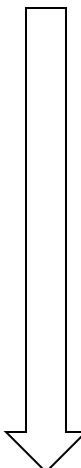
Homozygote = both alleles are the same length

Heterozygote = alleles differ and can be resolved from one another



Genetic Data and Familial Data can solve crimes

>Craig Harman's thrown brick was linked to his brother's STR markers; found & convicted



Case Solved – The Bloody Brick: Craig Harman

This was the first familial search in Great Britain in which the suspect was apprehended and convicted of the crime. In the early morning hours on March 21, 2003, Mr. Michael Little, a 53-year-old truck driver, was driving his truck on a highway in Surrey, when he drove beneath an overpass. A brick was thrown from the overpass and crashed through his windshield. It hit Mr. Little in his chest and caused fatal damage to the heart. Before Mr. Little died, he was able to bring his truck to a stop on the side of the road.

Law enforcement analyzed the blood on the brick and found two DNA profiles, one of Mr. Little and one of another unknown individual. That evening, before the brick was thrown from the overpass, a car had been burglarized in the same town. The burglar could not get the car started and he left his blood at the scene.

The police were able to extract a full DNA profile and it matched the DNA profile on the brick which killed Mr. Little. The profile was run through the DNA Database, but no match was found.



Craig Harman
Source: BBC News
Wednesday, 4 October, 2006

However, the DNA analysis established that the offender was caucasian. A police profiler looked at the details of the crime, and suggested that he was under the age of 35. Also, Surrey police believed the killer lived locally and so authorities performed a DNA dragnet screen involving 350 people from the surrounding area who volunteered to give samples. But still no match was found.

Law enforcement then decided to perform a familial search of white males under the age of 35 living in Surrey or Hampshire. [Twenty five people](#) with similar DNA were located including a relative of the suspect whose DNA matched 16 of 20 DNA markers. They interviewed the relative and discovered that he had a 19-year-old brother, [Craig Harman](#), who lived where the crime had occurred. Harman gave his DNA voluntarily and confessed. In April, 2004, Craig Harman pleaded guilty to manslaughter and was sentenced to 6 years.



Michael Little
Source: BBC News
Monday, 15 March, 2004



<http://www.dnaforensics.com/familialsearches.aspx>
http://news.bbc.co.uk/2/hi/uk_news/england/3513604.stm

CODIS may be agnostic to the subject, but society is not

PLOS MEDICINE

OPEN ACCESS

ESSAY

Do Health and Forensic DNA Databases Increase Racial Disparities?

Peter A. Chow-White , Troy Duster

Published: October 4, 2011 • <https://doi.org/10.1371/journal.pmed.1001100>

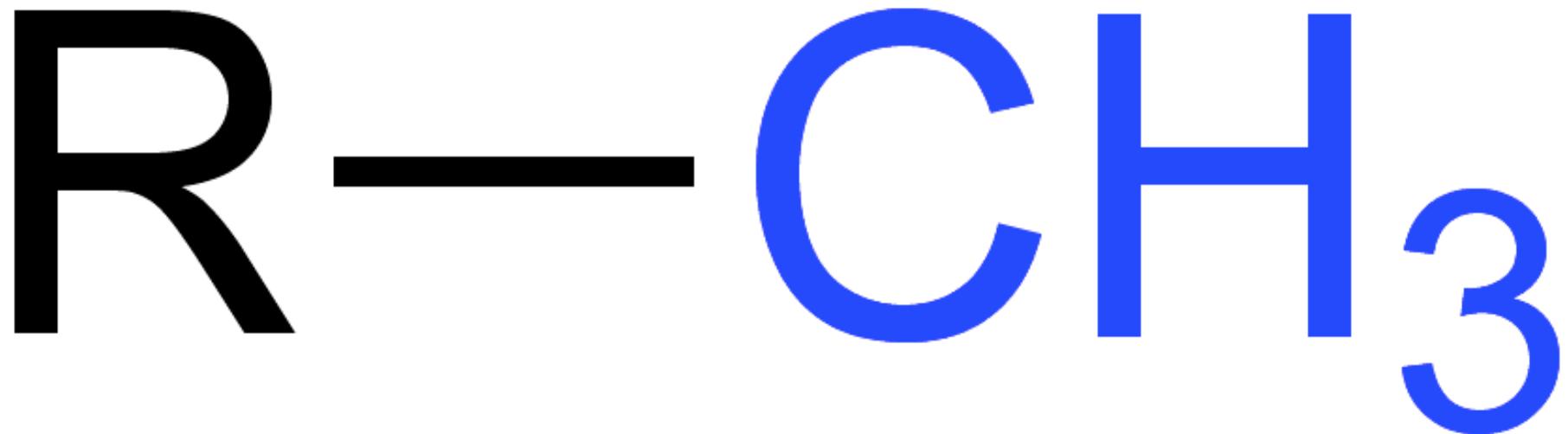
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3186804/>

https://scholarship.law.upenn.edu/cgi/viewcontent.cgi?article=1436&context=faculty_scholarship

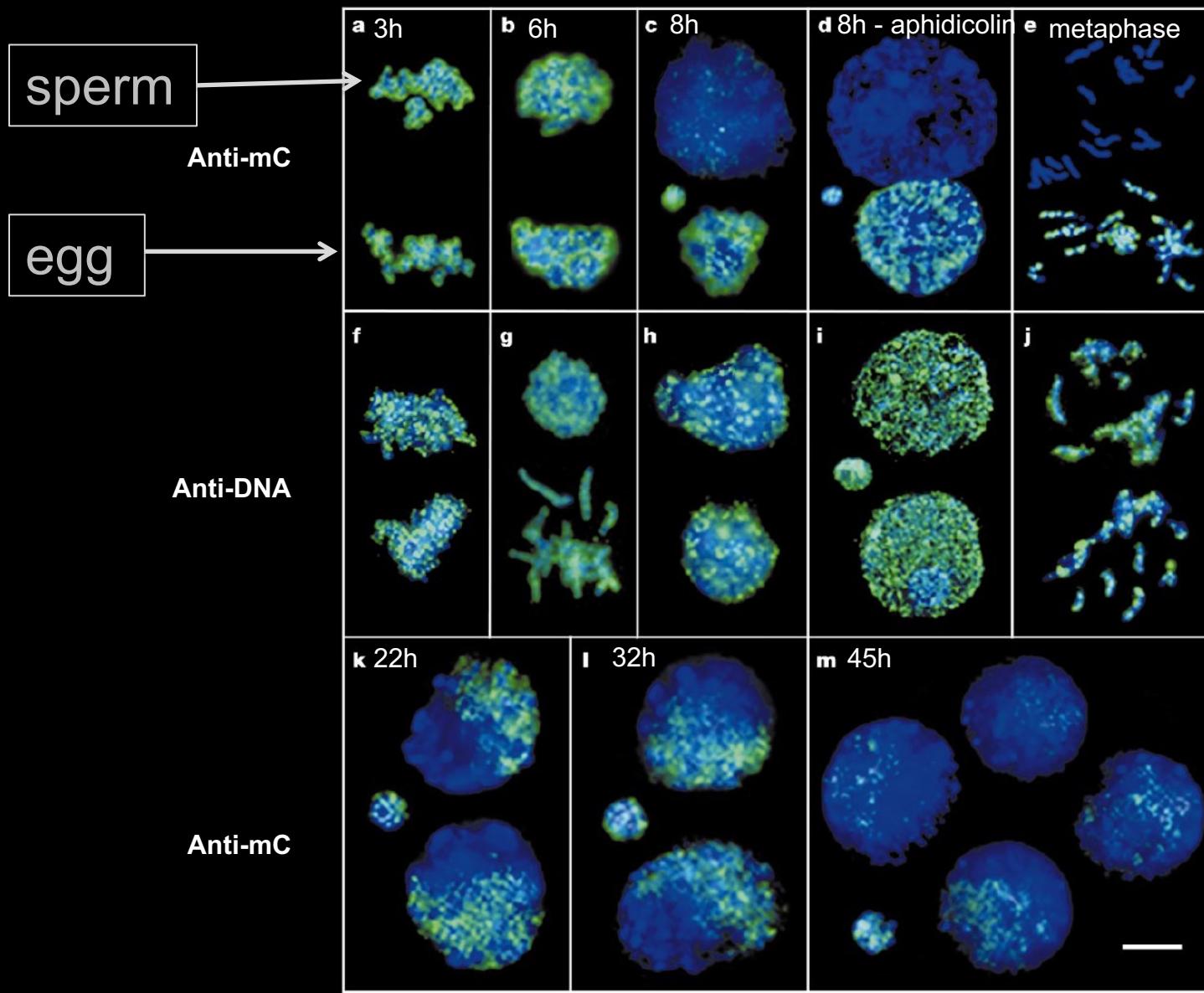
<https://www.nytimes.com/2020/01/23/opinion/dna-collection-border-privacy.html>

What other molecules
can
reveal information about you?

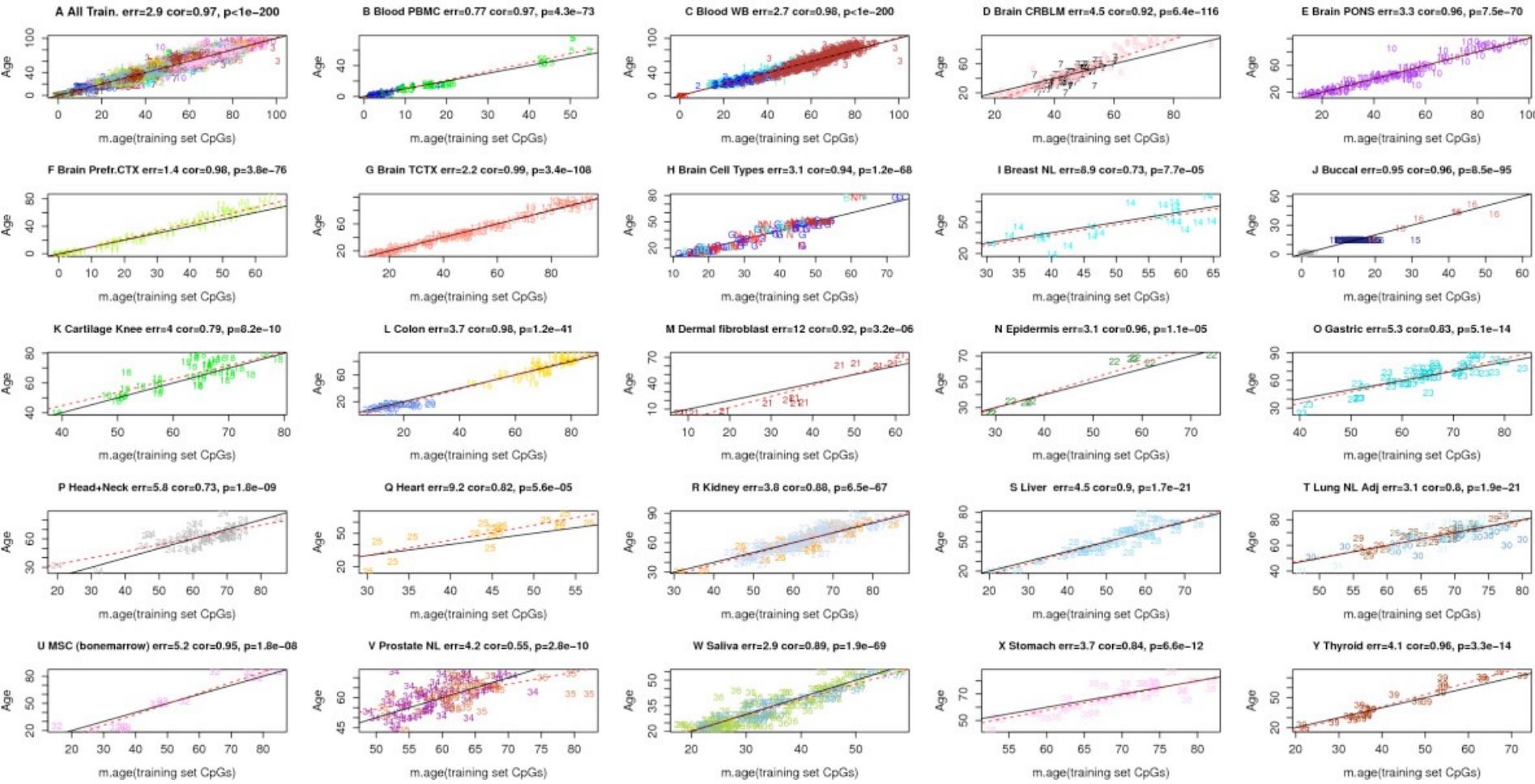
DNA Methylation (methyl-C) is
an echo of your life as well.



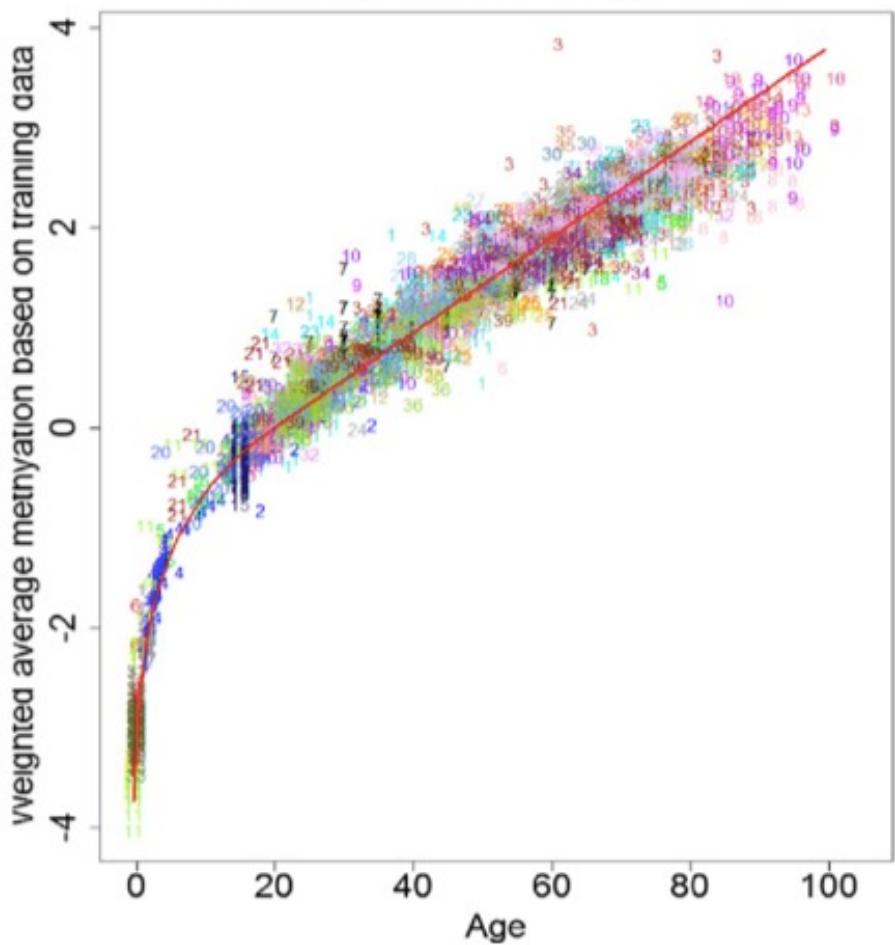
mC is important on day one



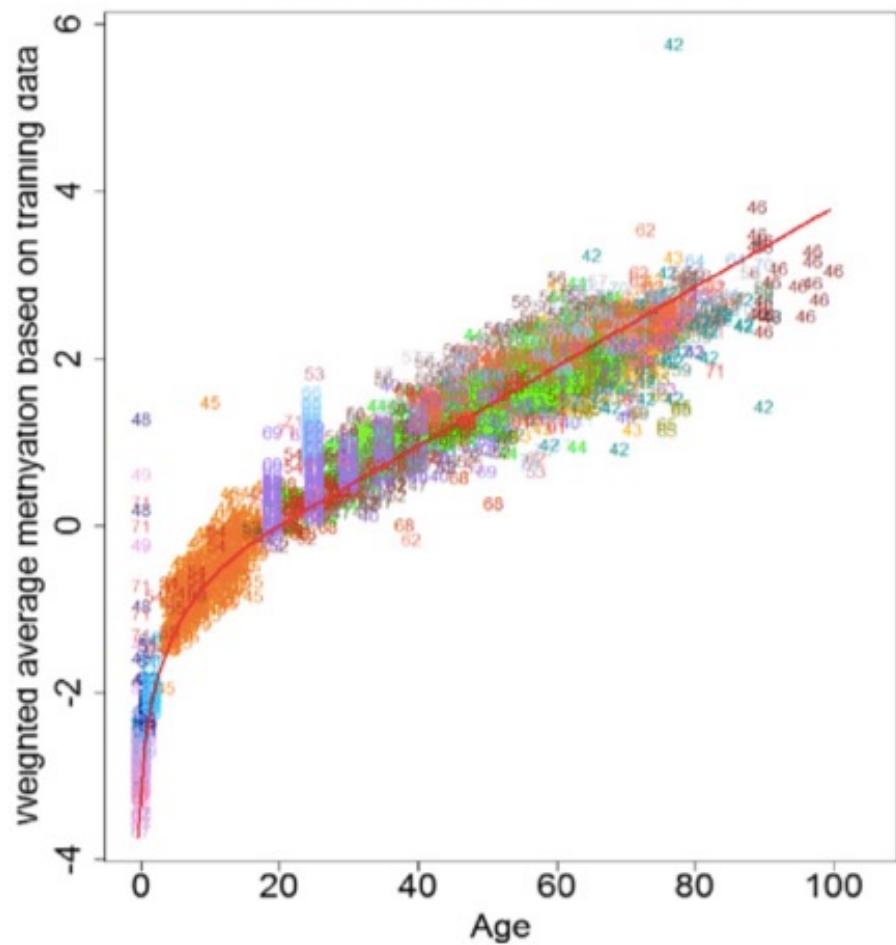
“Epigenetic age” increases in all tissues as time passes



B Training data $\text{cor}=0.92, p<1e-200$



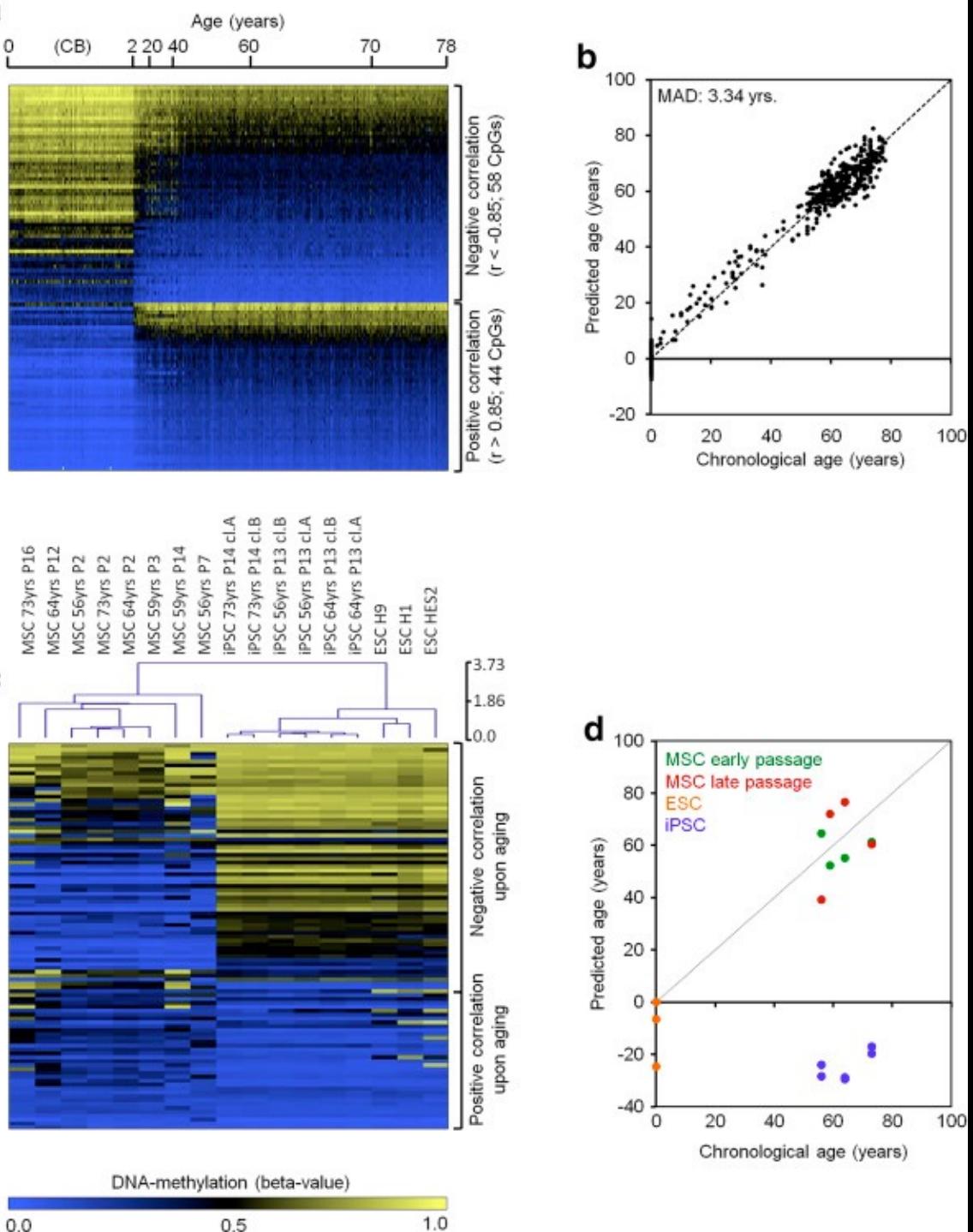
C Test data $\text{cor}=0.92, p<1e-200$



Horvath S. "DNA methylation age of human tissues and cell types." *Genome Biology*. 2013;14(10):R115.

The Epigenetic marks can reveal your age.

Weidner CI *et al.*,
“Aging of blood can be
tracked by DNA
methylation changes at
just three CpG sites.”
Genome Biology 2014
Feb 3;15(2):R24.



They can also give the age of blood at a crime scene

Forensic Genomics, Ahead of Print |

Epigenetic Forensics for Suspect Identification and Age Prediction

Jonathan Foox, Daniela Bezman, Priyanka Vijay, Kylie Getz, Kamolwat Ratanachai, Justin W. Davis, Keith Booher, Xiaojing Yang,

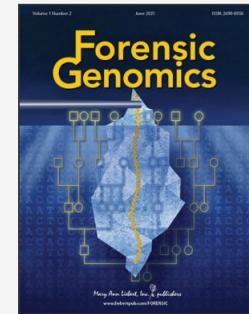
Cem Meydan  and Christopher E. Mason  

Published Online: 29 Jul 2021 | <https://doi.org/10.1089/forensic.2021.0005>

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<https://www.liebertpub.com/doi/abs/10.1089/forensic.2021.0005>

Epigenetic services

Cygenia provides service for epigenetic methods in diagnostics of disease, biological analysis based on pyrosequencing of the DNA-methylation level at specific genomic regions. Our information about epigenetic analysis is provided [here](#).

1) Epigenetic diagnostics of disease.

Particularly malignant diseases are associated with specific changes in the DNA-methylation pattern. Therefore a viable alternative to further strengthen diagnostics of diseases – such as le

2) Prediction of biological age.

There is a growing perception that aging is associated with highly reproducible DNA-nucleotide changes in the genome. Cygenia provides methods to estimate "biological age" based on three genomic parameters that impact on aging ([read more](#)).

3) Quality control of replicative senescence

It is important for scientists to carefully characterize their cell preparations – particularly in regenerative medicine but also in basic research. On the other hand, the cells undergo changes that need to be taken into consideration ([read more](#)).

4) Quality control of pluripotent cells

Cygenia provides Epi-Pluri-Score analysis for validation of successful reprogramming ([read more](#)).

5) Study design and bioinformatics



Epigenetic Biomarker to Support Classification into Pluripotent and Non-Pluripotent Cells

Michael Lenz, Roman Goetzke, Arne Schenk, Claudia Schubert, Jürgen Veeck, Hatim Hemeda, Steffen Koschmieder, Martin Zenke, Andreas Schuppert & Wolfgang Wagner*

Scientific Reports 5, Article number: 8973 (2015)

doi:10.1038/srep08973

[Download Citation](#)

Received: 08 October 2014

Accepted: 11 February 2015

Published online: 10 March 2015



- [Antibodies](#) >
- [Cell Culture Kits](#) >
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- [Growth Factors](#) >
- [iPSC Kits](#) >
- [Media/Reagents](#) >
- [Transfection Reagent](#) >
- [Virus Production](#) >

Human iPS Cell Line (Episomal, HFF)

Footprint-free human iPS (induced pluripotent stem) cells (HFFs) by ectopic expression of OCT4, SOX2, KLF4, and c-MYC genes.

Human iPS Cell Line (Episomal, MSC)

Footprint-free human iPS (induced pluripotent stem) cells (MSCs) by ectopic expression of OCT4, SOX2, KLF4, and c-MYC genes.

Human iPS Cell Line (Episomal, PBMC)

Footprint-free human iPS (induced pluripotent stem) cells (PBMCs) by ectopic expression of OCT4, SOX2, KLF4, and c-MYC genes.

Human iPS Cell Line (Retroviral)

Human iPS (induced pluripotent stem) cell line was derived using retroviral vector-mediated gene transfer for expression of OCT4, SOX2, KLF4, and c-MYC genes.

Mouse iPS Cell Line

Mouse induced pluripotent stem cell line derived using retroviral vector or drug selection.

Human Neural Stem Cell Line

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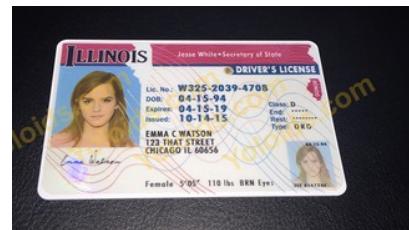
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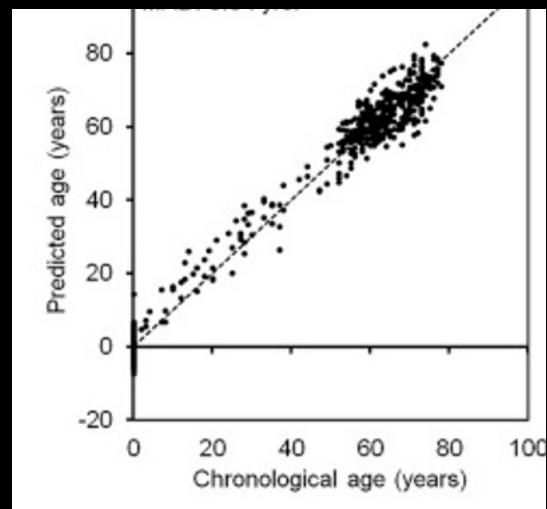
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How will we check age in the future?



IT'S TIME FOR CONGRESS TO UPDATE OUR GENETIC NONDISCRIMINATION LAW

Congress should act now to prevent epigenetic discrimination before it becomes widespread.

MAY 24, 2023 - 10:45AM



Crystal Grant, Former Technology Fellow, ACLU Speech, Privacy, and Technology Project

The Genetic Information Nondiscrimination Act (GINA) was enacted in 2008 to protect Americans from being widely discriminated against by a then new technology: genetic testing. The law was written after genetic discrimination emerged in the 1970s. At the time, programs to screen and identify genetic carriers of sickle cell anemia, a disease which afflicts many Black Americans, were being mandated by states. These mandated screening programs targeted Black people, perpetuating racial bias and stigmatization. Congress acted to make this genetic discrimination illegal. But now, GINA is 15 years old and needs to be updated to reflect a new threat of abuse of biological information – epigenetic discrimination. Because the law was written before many modern advances in the field of genetics, it is unclear whether its protections will extend to the novel types of information that will soon be generated from millions of Americans.

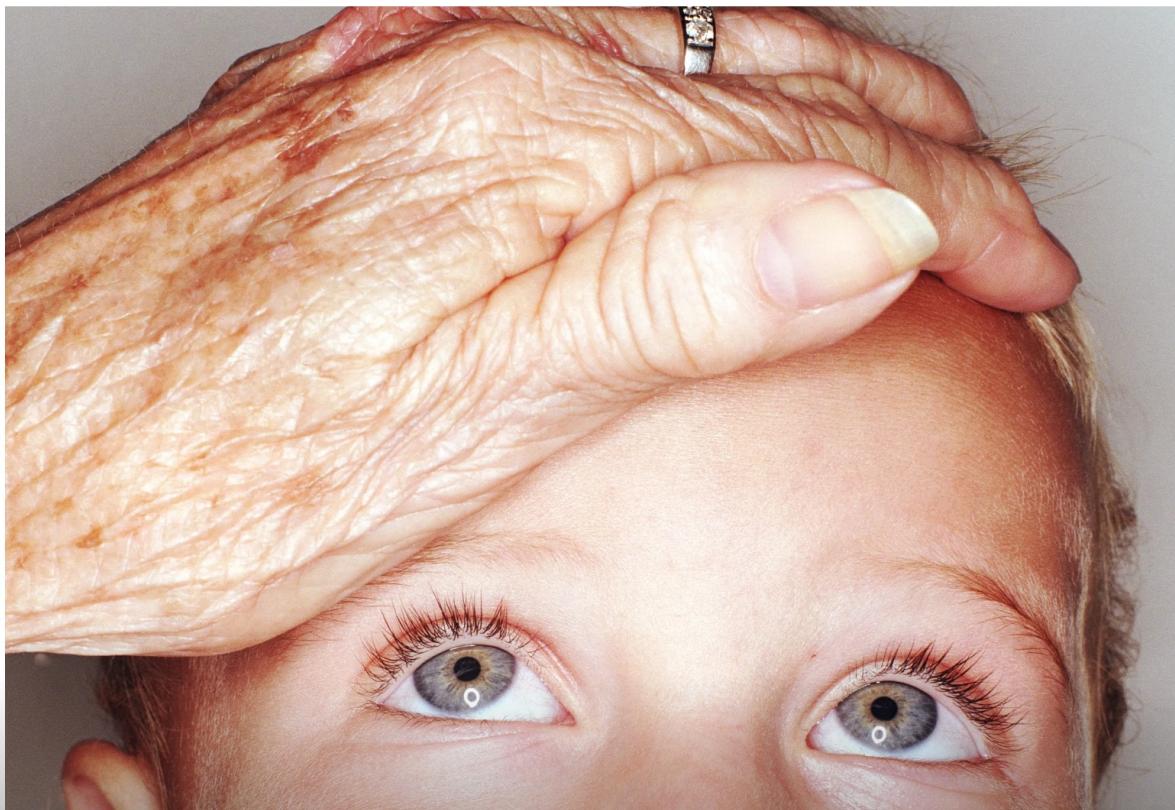
Epigenetic age

WIRED

BACKCHANNEL BUSINESS CULTURE GEAR IDEAS SCIENCE SECURITY

How Old Are You, Really? New Tests Want to Tell You

About a dozen such consumer tests are now on the market, but the science of reading DNA for insights about longevity is still young.



<https://www.wired.com/story/how-old-are-you-really-new-tests-want-to-tell-you/>

Each person has
a unique microbiome



Article | OPEN | Published: 04 September 2018

GePMI: A statistical model for personal intestinal microbiome identification

Zicheng Wang, Huazhe Lou, Ying Wang, Ron Shamir, Rui Jiang & Ting Chen

npj Biofilms and Microbiomes **4**, Article number: 20 (2018) | Download Citation ↓

Even after antibiotic treatment or fecal microbiota transplantation, the individual k-mer signature still maintains a certain specificity.

<https://www.nature.com/articles/s41522-018-0065-2>

Microbiomes raise privacy concerns

DNA from microbes living on the human body can be used to identify individuals.

Ewen Callaway

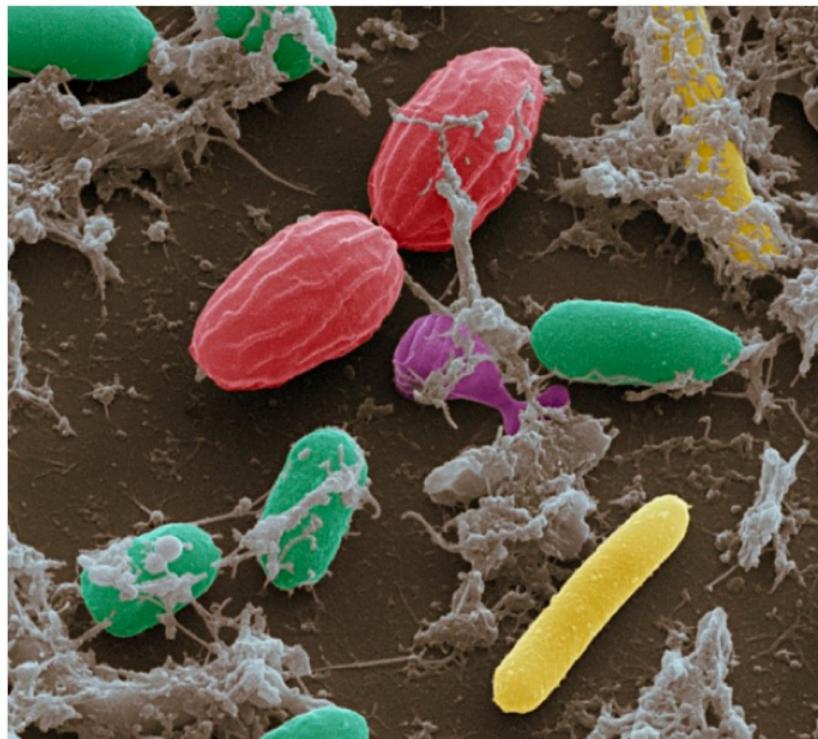
11 May 2015



PDF



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Eye of Science/Science Photo Library

DNA from bacteria in human faeces could be used as a 'gut print' to identify individuals.

Call it a 'gut print'. The collective DNA of the microbes that colonize a human body can uniquely

Each shoe has a unique
microbiome



Global City Sampling Day (gCSD)
<http://metasub.org/>

ARTICLE | ONLINE NOW

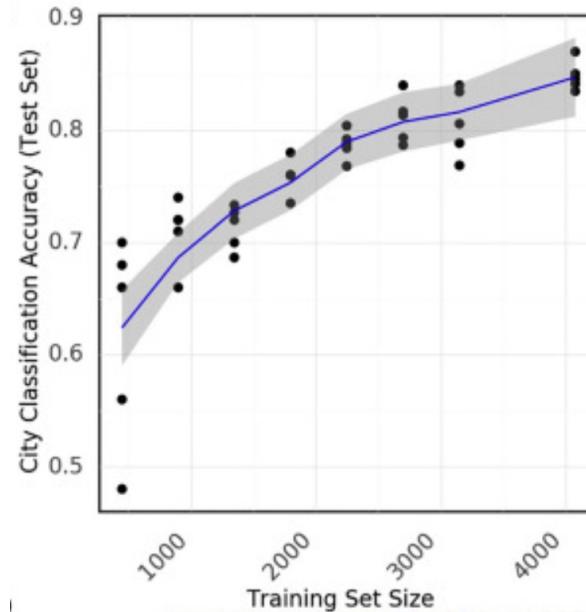
A global metagenomic map of urban microbiomes and antimicrobial resistance

David Danko⁶⁸ • Daniela Bezdán⁶⁸ • Evan E. Afshin • ... Sibo Zhu • Christopher E. Mason   •

The International MetaSUB Consortium • Show all authors • Show footnotes

Open Access • Published: May 26, 2021 • DOI: <https://doi.org/10.1016/j.cell.2021.05.002>

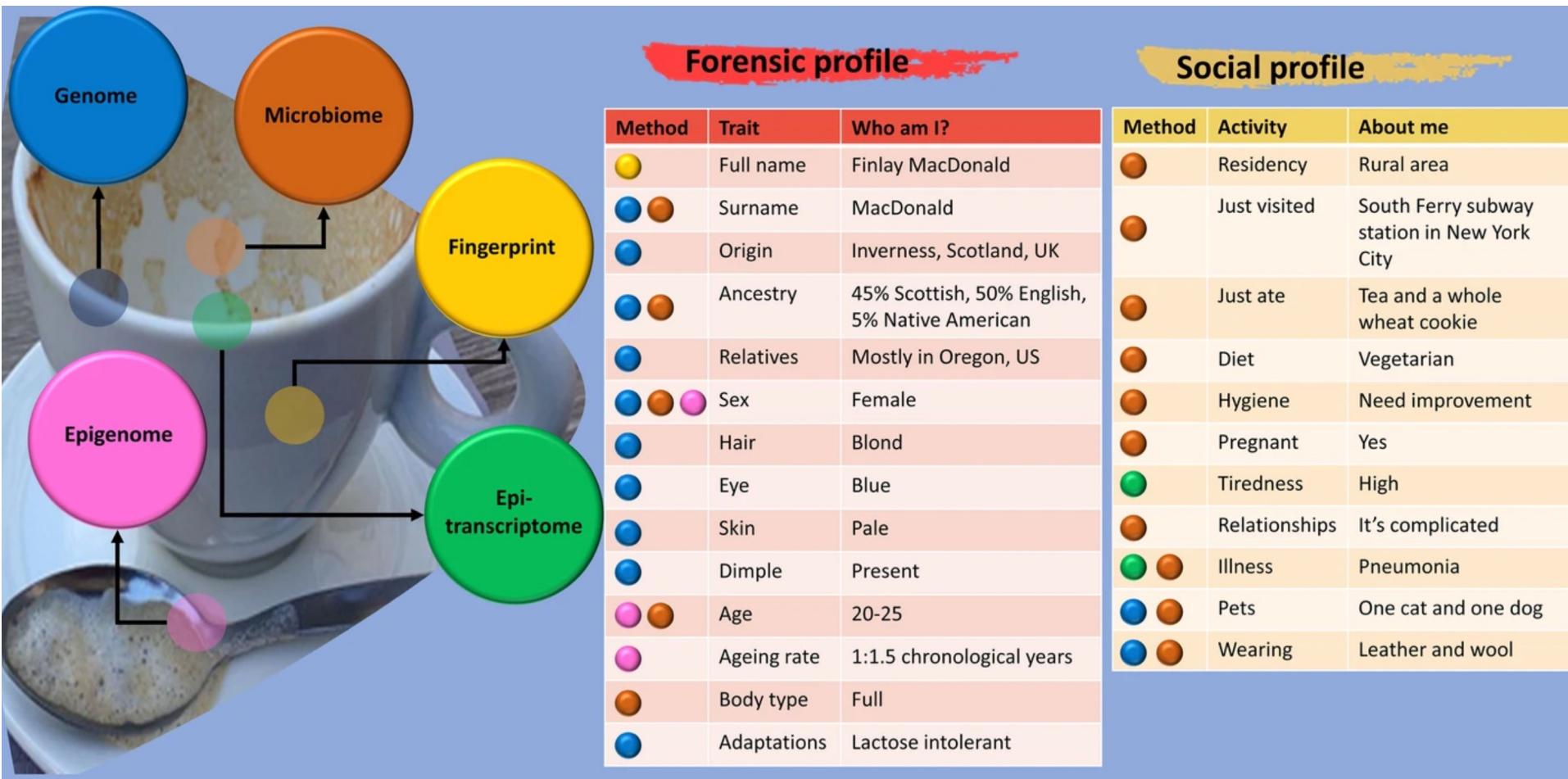
Each city
has a unique
microbiome profile



Nearly 90% accuracy for city-of-origin prediction

[https://www.cell.com/cell/fulltext/S0092-8674\(21\)00585-7](https://www.cell.com/cell/fulltext/S0092-8674(21)00585-7)

Cross-kingdom molecular forensics and informatics can reduce genetic privacy



Can all these methods be
legal? or patented?

Depends on:
“Expectation of Privacy”

Kyllo v. United States



Supreme Court of the United States

Argued February 20, 2001

Decided June 11, 2001

Full case name Danny Lee Kyllo v. United States

Citations 533 U.S. 27 [\(more\)](#)

121 S. Ct. 2038; 150 L. Ed. 2d 94;
69 U.S.L.W. 4431; 2001 U.S.
LEXIS 4487; 2001 Cal. Daily Op.
Service 4749; 2001 Daily Journal
DAR 5879; 2001 Colo. J. C.A.R.
2926; 14 Fla. L. Weekly Fed. S 329

Prior history On writ of certiorari to the United States Court of Appeals for the Ninth Circuit

Holding

Thermal imaging of a home constitutes a Fourth Amendment "search" and may be done only with a warrant.



Could you patent a gene?

A bit of history



New Haven, CT

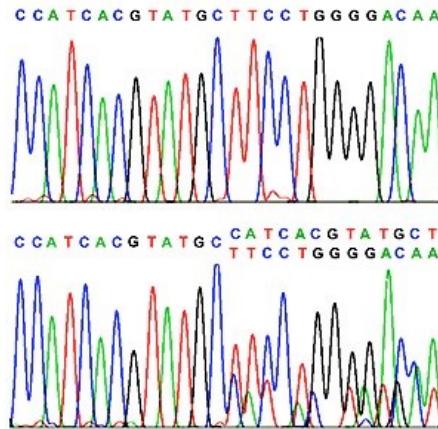
A bit of history -2005

Law & Order Genomics Unit

- I. First Division: Stem Cell Research:
- II. Second Division: Creating Organisms and Biomes
- III. Third Division: Interventions in the Human Genome
- IV. Fourth Division: Selected issues in the genetic frontier

Information Society Project
Yale Law School

I was a post-doc looking for disease genes,
but there may be hundreds of them



POLICY FORUM

INTELLECTUAL PROPERTY

Intellectual Property Landscape of the Human Genome

Kyle Jensen and Fiona Murray*

California, Isis Pharmaceuticals, the former SmithKline Beecham, and Human Genome Sciences. The top patent assignee is Incyte Pharmaceuticals/Incyte Genomics, whose IP rights cover 2000 human genes, mainly for use as probes on DNA microarrays.

Although large expanses of the genome are unpatented, some genes have up to 20 patents asserting rights to various gene uses

defined through amino acid sequences. (See table S1 for a sensitivity analysis.)

Our results reveal that nearly 20% of human genes are explicitly claimed as U.S. IP. This represents 4382 of the 23,688 of genes in the NCBI's gene database at the time of writing (see figure, right). These genes are claimed in 4270 patents within 3050 patent families (28).



Physical mapping of genes into 300-kb segments

patent claiming a gene represents the number of genes that show the loci of highly



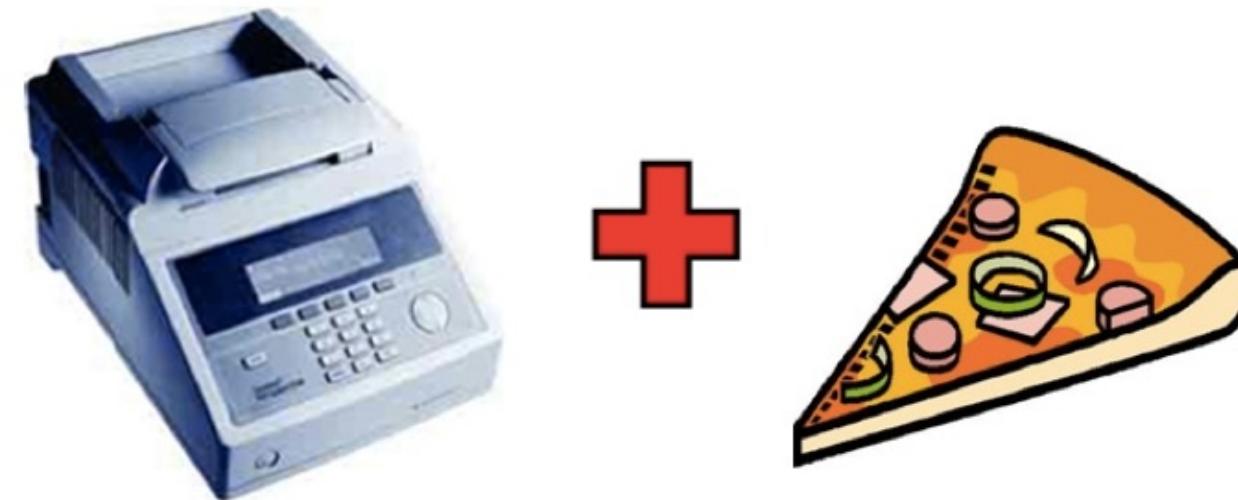
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PCR and Pizza Night

**Open House October 20th.
Join us for PCR and Pizza
night!**

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<http://www.genspace.org/>



New York City's C

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CCR5 Δ32
HIV immunity

So, since all of your birthdays are so close this year (again! what are the odds!!?!)?, Joan and I wanted to give the gift of genomics! So, us four gents will find a day to go to the Personal Genotyping workshop at Genspace NYC, and we can go and sequence our own DNA! The courses are offered most Saturdays, so we'll just find a time and go anytime in the next month. More details are at genspace.org, and also pasted below.



New York City's Community Biolab

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Genspace NYC

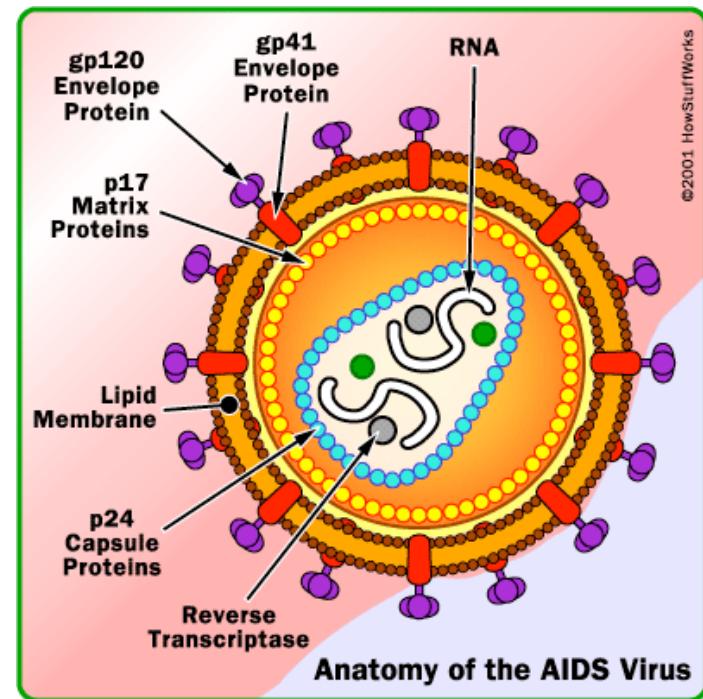
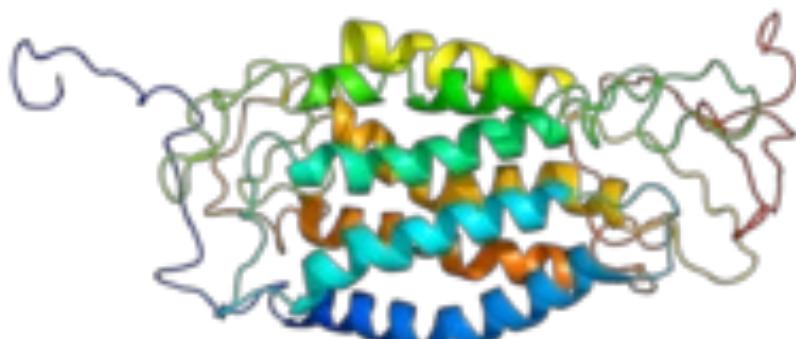


Gift certificate for a
one-day personal
genotyping workshop
at Genspace



Yes, that gene

C-C chemokine receptor type 5 (CCR5) homozygous nulls (-/-) have HIV protection



This was an infringing act

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37
COMMENTS

PHARMA & HEALTHCARE

7/31/2011 @ 5:06PM | 18,940 views

Private companies own your DNA - again

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Cartoon (c) by Cathy Wilcox

Many scientists cheered last year when a federal judge ruled that human genes couldn't be patented. The case involved Myriad Genetics, which holds the patent rights on two genes, BRCA1 and BRCA2, that are associated with increased risks for breast and ovarian cancer. Thanks to these patents, you can't look these genes in your own body without paying a fee to Myriad. Sounds ridiculous, right? Well, that was the state of gene patents until last May, when judge Robert Sweet ruled that the Myriad's patents were invalid.

But now the courts have reversed themselves again. In [a 105-page decision](#), two federal judges decided that the whole matter comes down to the meaning of the word "isolated." I kid you not.

!!!!

Infringement is unavoidable
for a modern-day geneticist.

A patent must be:

1. New (1790)
2. Useful (1790)
3. Non-Obvious (1952)

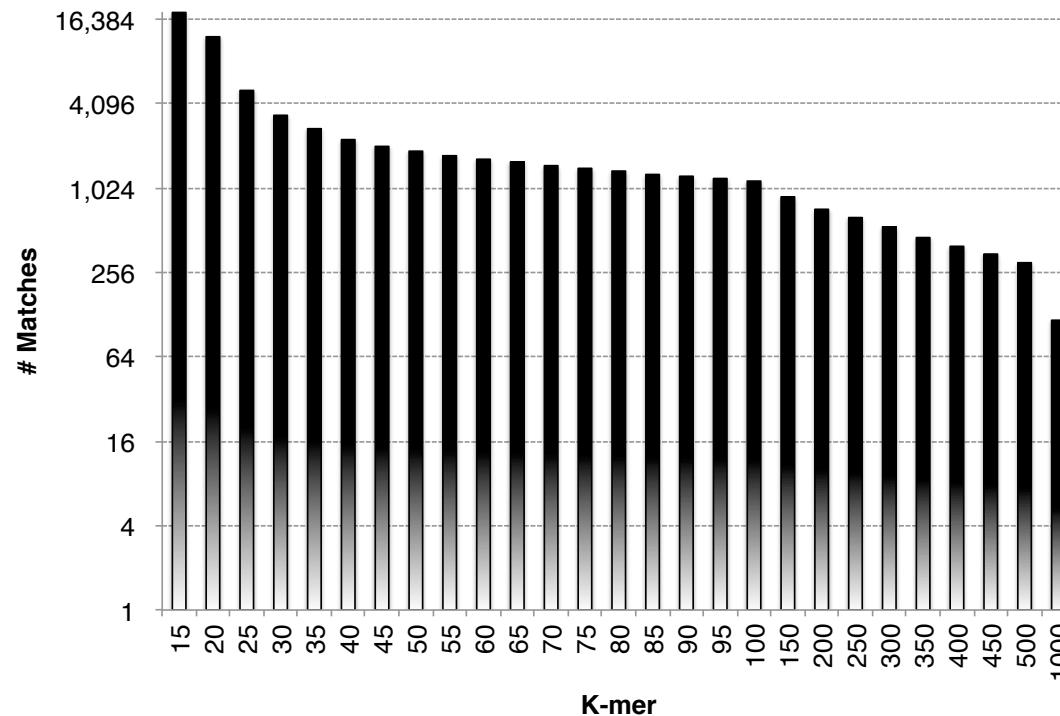
“The Congress shall have Power To ... promote the Progress of Science and useful Arts, by securing for limited Times to Authors and Inventors the exclusive Right to their respective Writings and Discoveries.” **Article I, Section 8, of the United States Constitution**

Claim #1. An isolated DNA coding for a BRCA1 polypeptide, said polypeptide having the amino acid sequence set forth in SEQ ID NO:2 (the BRCA1 cDNA).

Claim #2. The isolated DNA of claim 1, wherein said DNA has the nucleotide sequence set forth in SEQ ID NO:1 (the BRCA1 gene).

Pervasive sequence patents cover the entire human genome

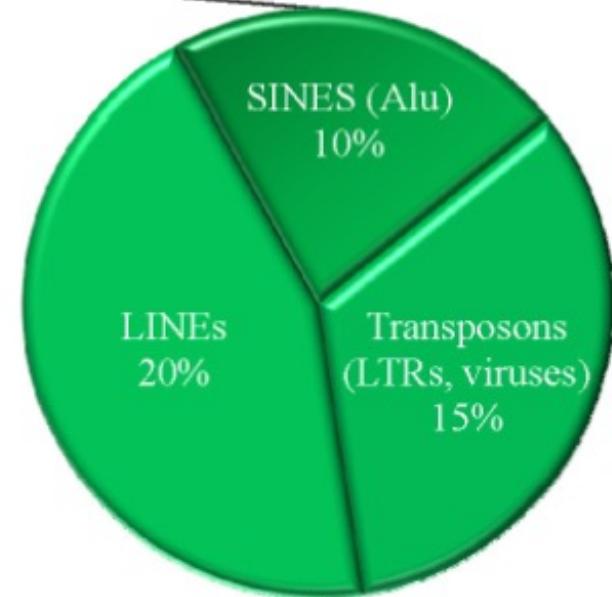
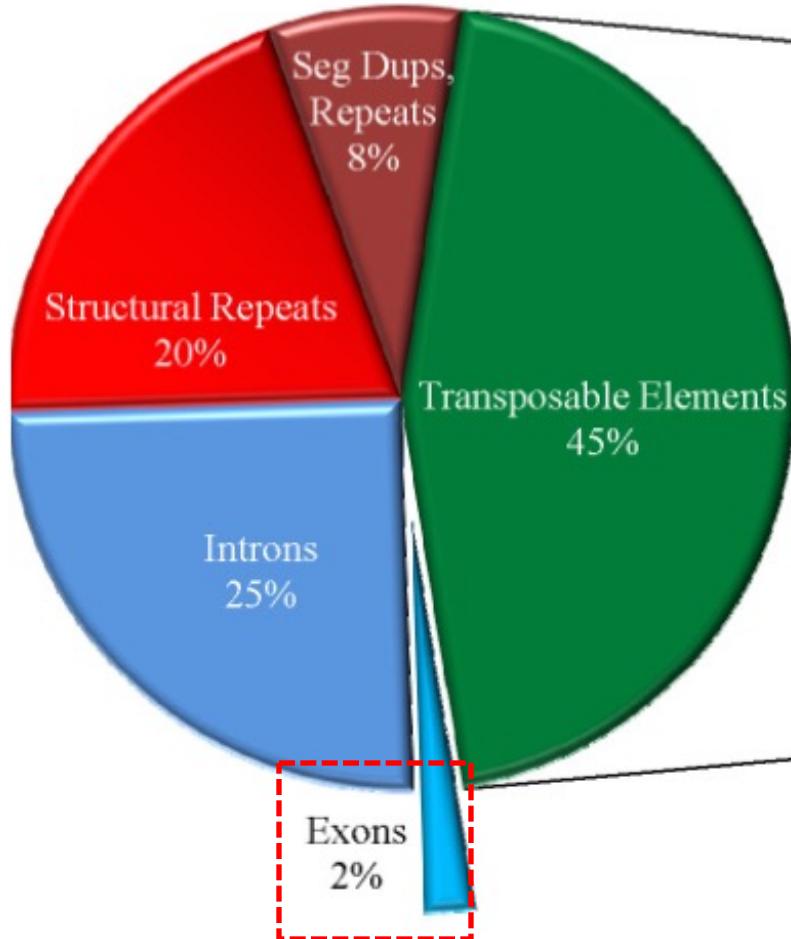
Jeffrey Rosenfeld^{1,2} and Christopher E Mason^{*3,4,5}



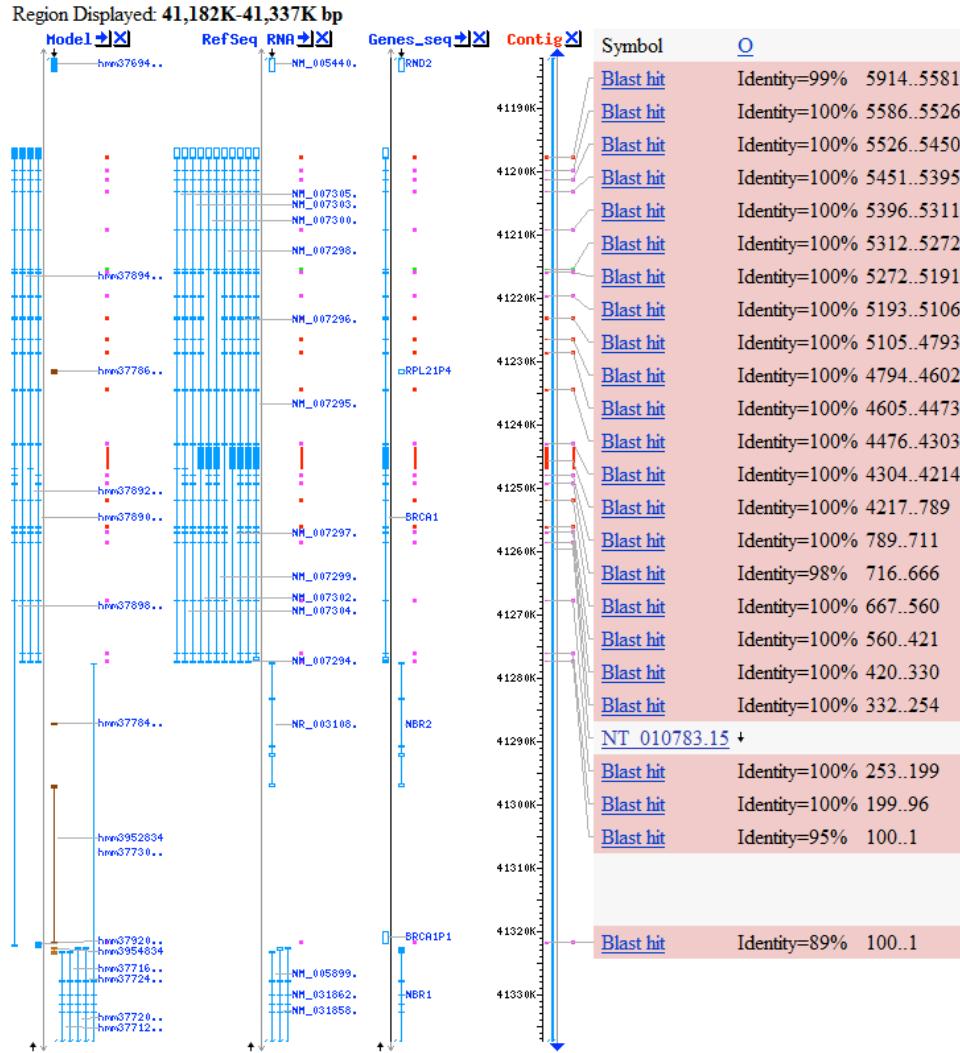
How many genes?

100% of genes have one 15-mer that matches another gene

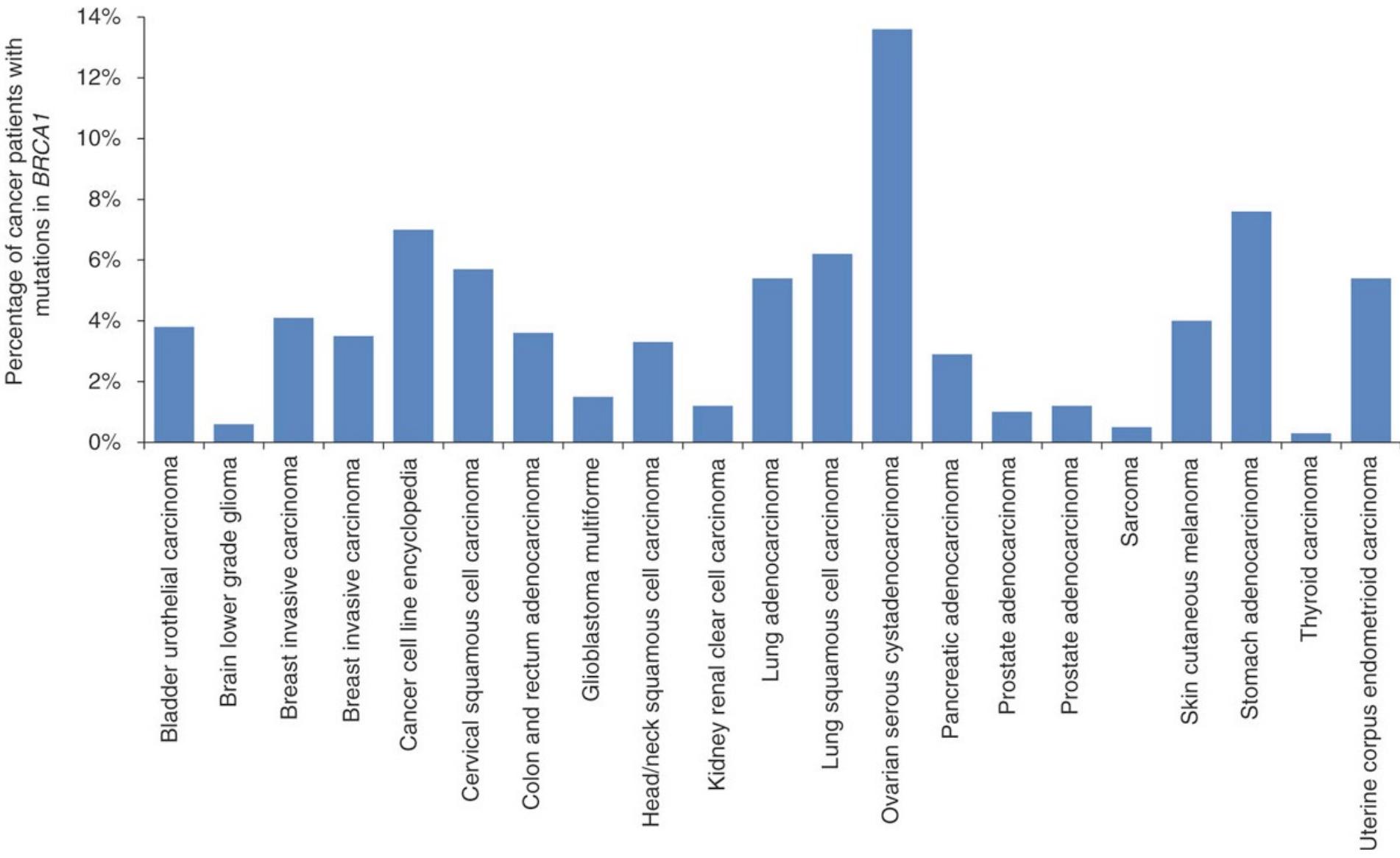
Matches occur across the entire human genome,
including pseudogenes



Even BRCA1 has a psuedogene in the human genome (BRCA1P1)



BRCA1 mutations are important in most cancers, not just breast/ovarian



**BRCA1 has
Pleiotropy**

UNITED STATES DISTRICT COURT
SOUTHERN DISTRICT OF NEW YORK

ASSOCIATION FOR MOLECULAR PATHOLOGY;
AMERICAN COLLEGE OF MEDICAL GENETICS;
AMERICAN SOCIETY FOR CLINICAL PATHOLOGY;
COLLEGE OF AMERICAN PATHOLOGISTS;
HAIG KAZAZIAN, MD; ARUPA GANGULY, PhD;
WENDY CHUNG, MD, PhD; HARRY OSTRER, MD;
DAVID LEDBETTER, PhD; STEPHEN WARREN, PhD;
ELLEN MATLOFF, M.S.; ELSA REICH, M.S.;
BREAST CANCER ACTION; BOSTON WOMEN'S
HEALTH BOOK COLLECTIVE; LISBETH CERIANI;
RUNI LIMARY; GENAE GIRARD; PATRICE FORTUNE;
VICKY THOMASON; KATHLEEN RAKER,

09 Civ. 4515 (RWS)

Briefs filed in 2009, 2010...

Plaintiffs,

ECF Case

v.

UNITED STATES PATENT AND TRADEMARK
OFFICE; MYRIAD GENETICS; LORRIS BETZ,
ROGER BOYER, JACK BRITTAINE, ARNOLD B.
COMBE, RAYMOND GESTELAND, JAMES U.
JENSEN, JOHN KENDALL MORRIS, THOMAS PARKS,
DAVID W. PERSHING, and MICHAEL K. YOUNG,
in their official capacity as Directors of the University
of Utah Research Foundation,

SUPPLEMENTAL
DECLARATION OF
CHRISTOPHER E.
MASON

Defendants.

I, Christopher E. Mason, declare under penalty of perjury:

1. I previously submitted a declaration in this case on August 20, 2009. I am currently

Assistant Professor of Computational Genomics at Weill Cornell Medical College in New York

City. I also hold an appointment at the tri-institutional training program on computational
biology and medicine between Rockefeller University, Memorial Sloan-Kettering Cancer Center,
and Cornell University. I perform research, publish, and teach courses on whole genome

Opinions

The Supreme Court should invalidate the patent on human DNA

By Jeffrey A. Rosenfeld and Christopher E. Mason April 5, 2013

Jeffrey Rosenfeld is an assistant professor of medicine at the New Jersey Medical School and a member of the High Performance and Research Computing Group. Christopher E. Mason is an assistant professor of computational genomics at Weill Cornell Medical College and affiliate fellow of the information society project of Yale Law School.

Asked in 1955 whether his polio vaccine was patented, Jonas Salk replied, “There is no patent. Could you patent the sun?” With that, Salk debunked the misguided notion of patenting objects found in nature. His polio vaccine was not a new invention but an inactive form of the natural polio virus.

[Letters to the Editor](#)

Patents for DNA molecules help promote innovation

April 12, 2013

Regarding Jeffrey A. Rosenfeld and Christopher E. Mason's April 7 Sunday Opinions commentary, "[Who owns your DNA? Not who you think.](#)":

The patents for which Mr. Rosenfeld and Mr. Mason criticized my company were essential to developing diagnostic tools that have been used by more than 1 million women to understand their hereditary risks of breast cancer and ovarian cancer. We did not patent human genes from anyone's body. Rather, our patents protect synthetic molecules created in the lab. This is no different than thousands of other patents issued in the past 30 years.

Were these molecules derived in part from natural material? Sure. But that is true of many patents. Labs routinely turn naturally found molecules into innovative medicines and get patent protection.

Simply put, the U.S. patent system made Myriad's tests possible. Our research cost more than \$500 million, and it took 17 years for us to get to the break-even point. Notably, our tests are



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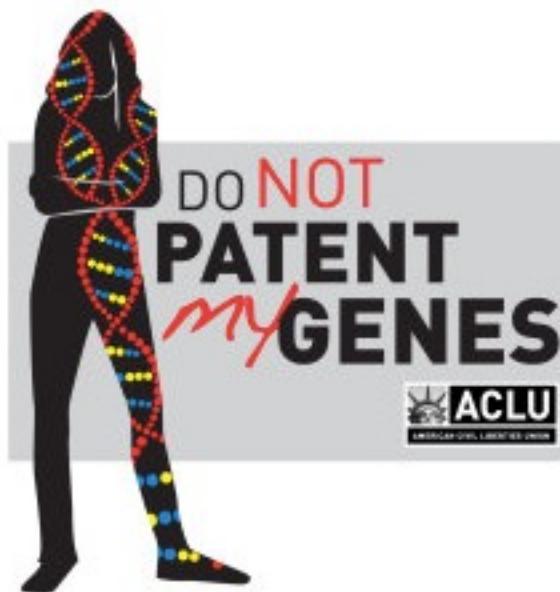
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There is something fundamentally wrong with allowing companies to patent our genome.

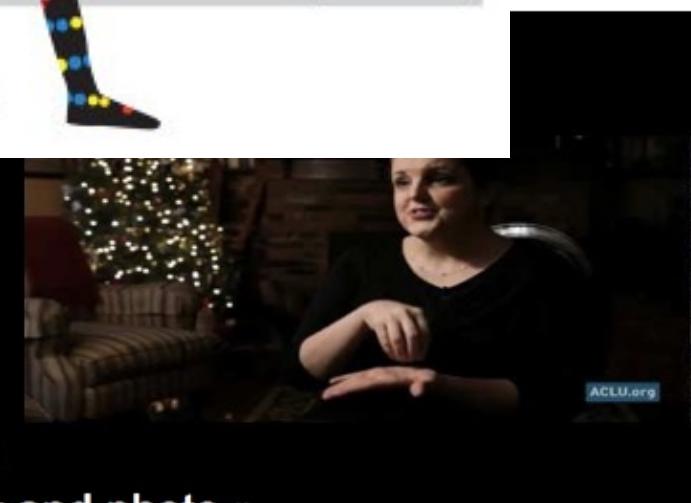
Take Back Our Genes

The government should not be granting private entities patents on our genes. Moreover, granting patents on genetic information violates the First Amendment.

The ACLU petitioned the Supreme Court to hear arguments in a case challenging a patent on breast and ovarian cancer. Stand with us and share your story.



Slideshow



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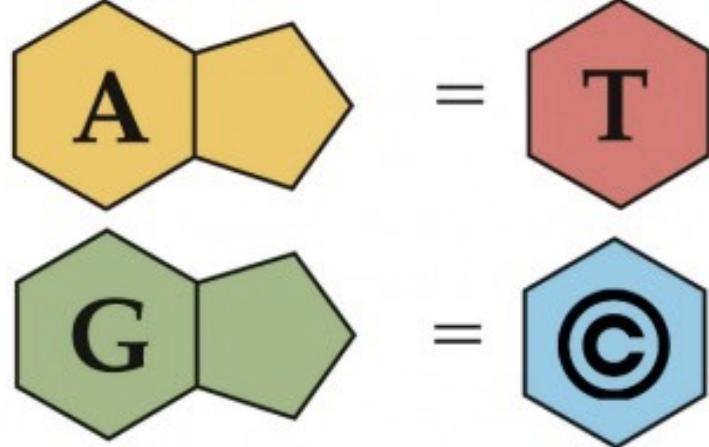
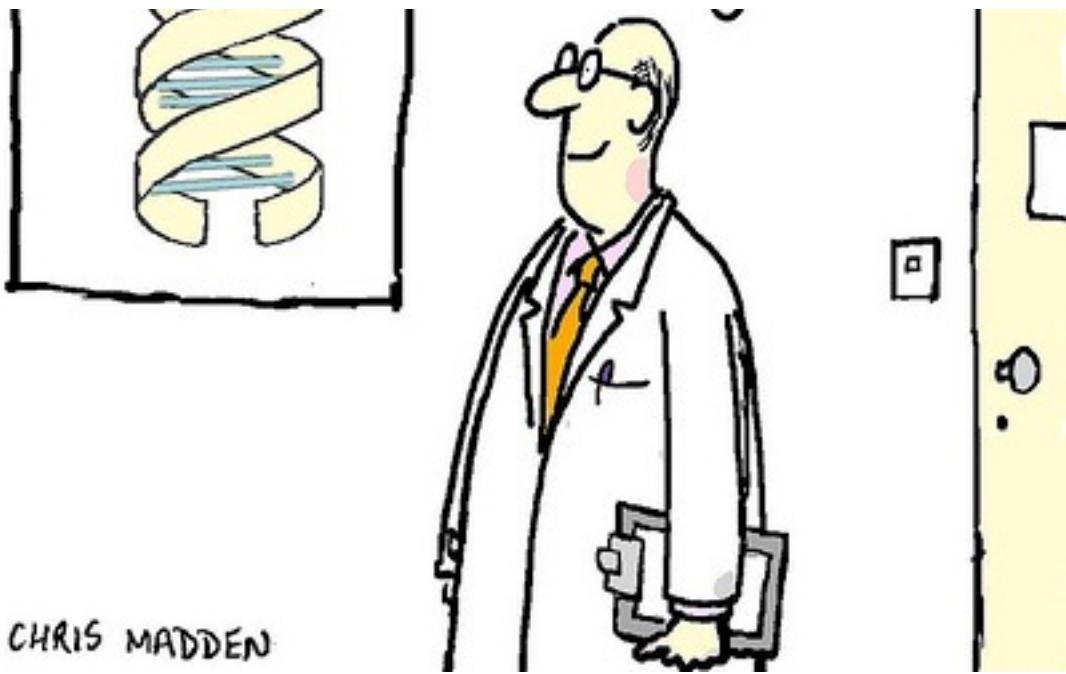
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NEWSFEED

**Stand with the
ACLU as we
defend
reproductive
freedom
across the
country.**

Genomic Liberty For All?

Published: MARCH 29, 2013





April 15th, 2013 – Supreme Court heard
case challenging the BRCA1/2 patents

June 13th, 2013 – Claims **rejected**

\$4400

to

\$250

in one day.

The Battle continues

New Bill Would Bring Back Terrible Software and Genetic Patents

BY JOE MULLIN | AUGUST 18, 2022



A [recently introduced patent bill](#) would authorize patents on abstract ideas just for including computer jargon, and would even legalize the patenting of human genes. The “Patent Eligibility Restoration Act,” sponsored by Sen. Thom Tillis (R-NC), explicitly overrides some of the most important Supreme Court decisions of the past 15 years, and would tear down some of the public’s only

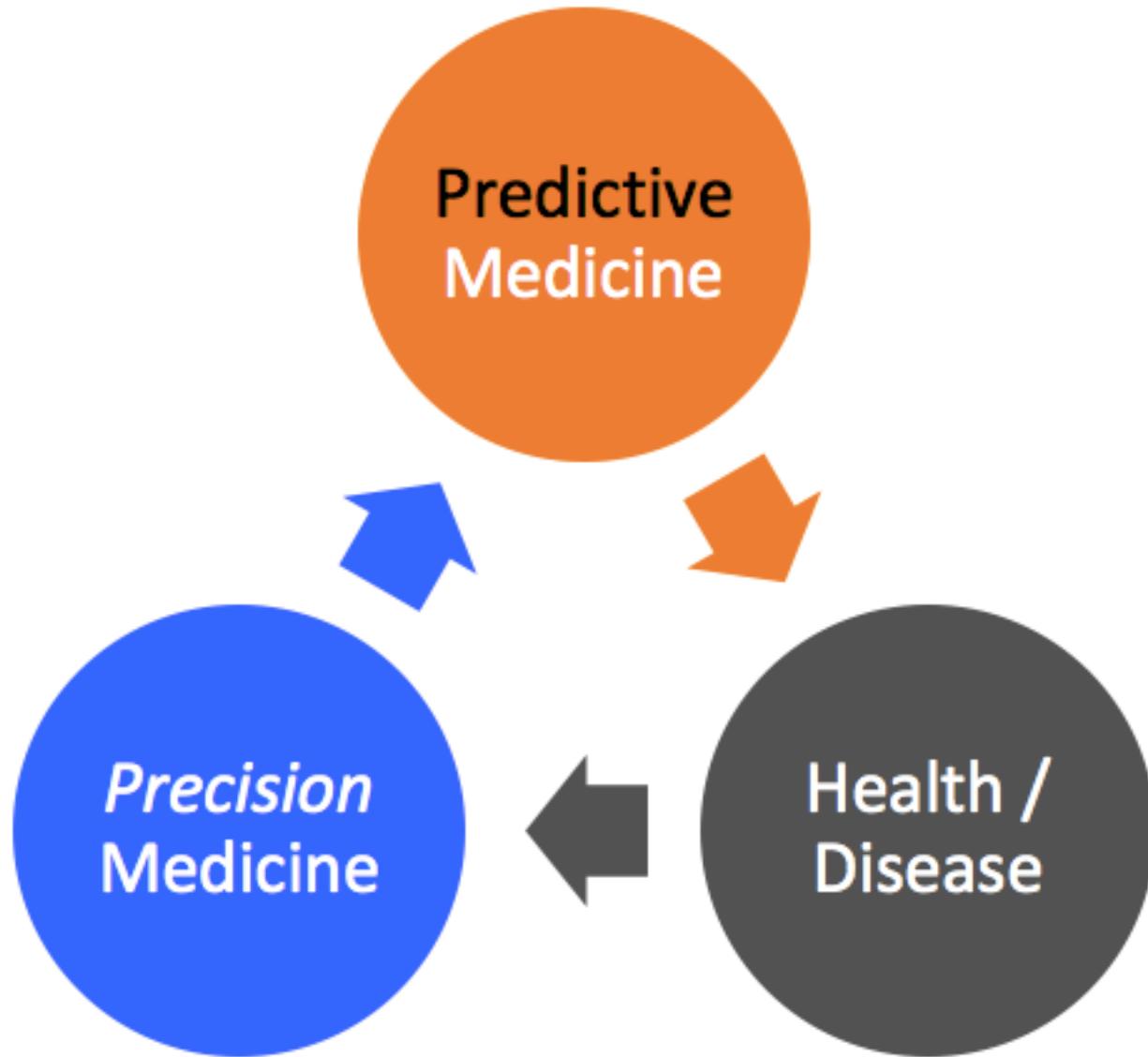
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Could we just correct
the “bad genes?”

Designer Genomes

The model



Human Genome Editing

RESEARCH ARTICLE

CRISPR/Cas9-mediated gene editing in human tripronuclear zygotes

Puping Liang, Yanwen Xu, Xiya Zhang, Chenhui Ding, Rui Huang, Zhen Zhang, Jie Lv, Xiaowei Xie, Yuxi Chen, Yujing Li, Ying Sun, Yaofu Bai, Zhou Songyang, Wenbin Ma, Canquan Zhou[✉], Junjiu Huang[✉]

Guangdong Province Key Laboratory of Reproductive Medicine, the First Affiliated Hospital, and Key Laboratory of Gene Engineering of the Ministry of Education, School of Life Sciences, Sun Yat-sen University, Guangzhou 510275, China

[✉] Correspondence: hjunjiu@mail.sysu.edu.cn (J. Huang), zhoucanquan@hotmail.com (C. Zhou)

Received March 30, 2015 Accepted April 1, 2015

ABSTRACT

Genome editing tools such as the clustered regularly interspaced short palindromic repeat (CRISPR)-associated system (Cas) have been widely used to modify genes in model systems including animal zygotes and human cells, and hold tremendous promise for both basic research and clinical applications. To date, a serious knowledge gap remains in our understanding of DNA repair mechanisms in human early embryos, and in

pressing need to further improve the fidelity and specificity of the CRISPR/Cas9 platform, a prerequisite for any clinical applications of CRISPR/Cas9-mediated editing.

KEYWORDS CRISPR/Cas9, β-thalassemia, human tripronuclear zygotes, gene editing, homologous recombination, whole-exome sequencing

NATURE | NEWS



Chinese scientists genetically modify human embryos

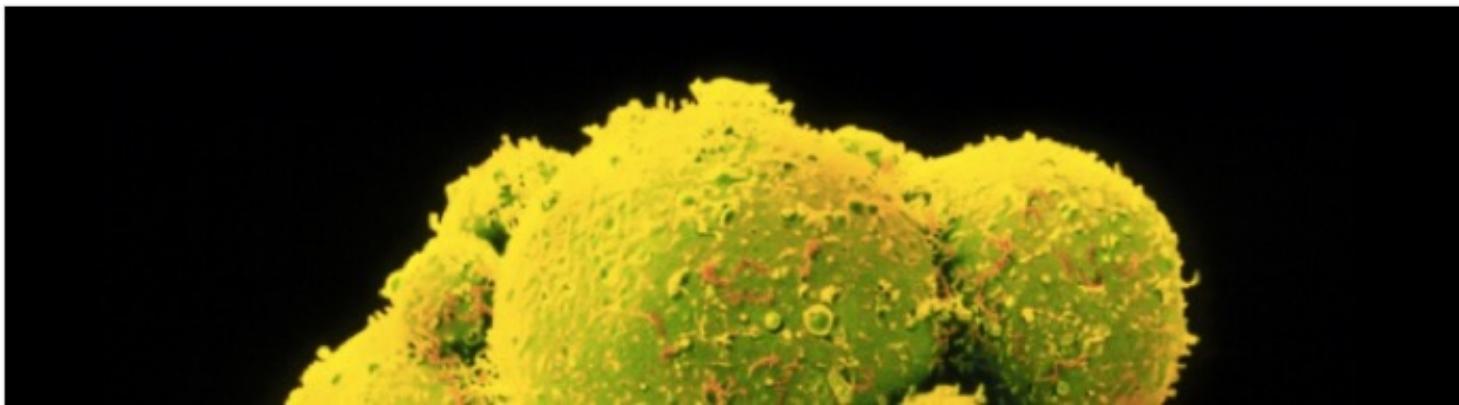
Rumours of germline modification prove true — and look set to reignite an ethical debate.

David Cyranoski & Sara Reardon

22 April 2015



[Rights & Permissions](#)



NATURE | NEWS



Gene-editing record smashed in pigs

Researchers modify more than 60 genes in effort to enable organ transplants into humans.

Sara Reardon

06 October 2015

<http://www.nature.com/news/gene-editing-record-smashed-in-pigs-1.18525>



Search

NEWS · 28 NOVEMBER 2018

CRISPR-baby scientist fails to satisfy critics

He Jiankui gives talk about controversial claim of genome editing babies, but ethical questions remain.

David Cyranoski



<https://www.nature.com/articles/d41586-018-07573-w>

CCR5 can help limit West Nile Virus risk

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In This Issue

CCR5 saves lives

[Heather L. Van Epps](#)DOI: [10.1084/jem2028iti4](https://doi.org/10.1084/jem2028iti4) | Published October 17, 2015**Douglas M. Durrant, Brian P. Daniels, TracyJo Pasieka, Denise Dorsey and Robyn S. Klein** *Journal of Neuroinflammation* 2015 12:233<https://doi.org/10.1186/s12974-015-0447-9> | © Durrant et al. 2015

Received: 18 June 2015 | Accepted: 25 November 2015 | Published: 15 December 2015

<https://jneuroinflammation.biomedcentral.com/articles/10.1186/s12974-015-0447-9><http://jem.rupress.org/content/202/8/1015.2>

**CCR5 has
Pleiotropy**



CENTER of EXCELLENCE
for ENGINEERING BIOLOGY



GP-write Consortium

<http://engineeringbiologycenter.org/>

NIH's \$190M Somatic Cell Genome Editing Program

 National Institutes of Health
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Somatic Cell Genome Editing

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Somatic Cell Genome Editing

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[Program Meetings](#)

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NIH to Launch Genome Editing Research Program

[Read Press Release](#)

The NIH Common Fund is launching the Somatic Cell Genome Editing program to help develop safe and effective genome editing

Visit commonfund.nih.gov/editing for more information

Program Snapshot

Many common and rare diseases are caused by changes to the genetic code. Genome editing technologies present an exciting prospect for treatments and possibly even cures for these diseases. The NIH Common Fund's **Somatic Cell Genome Editing** program aims to develop quality tools to perform

 CGE
Somatic Cell Genome Editing

Program Background

- September 1, 2017 - NIH Council of Councils presentation  (at time 2:53)
- September 1, 2017 - NIH Council of Councils slides 

What is Genome Editing?

Visit NHGRI's [Genome Editing website](#) to learn more about this technology.

Join the Common Fund Mailing List for Updates



<https://commonfund.nih.gov/editing>

Repair of *MYBPC3* (hypertrophic cardiomyopathy)

nature International weekly journal of science

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Archive > Volume 548 > Issue 7685 > News > Article

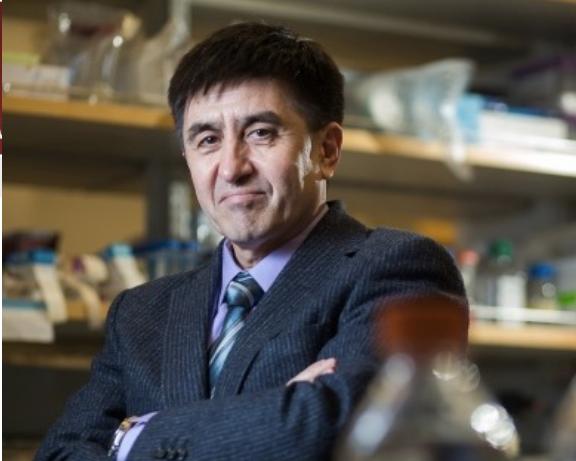
NATURE | NEWS

CRISPR fixes disease gene in viable human embryos

Gene-editing experiment pushes scientific and ethical boundaries.

Heidi Ledford

02 August 2017



Leah Nash/NYT/Redux/eyevine

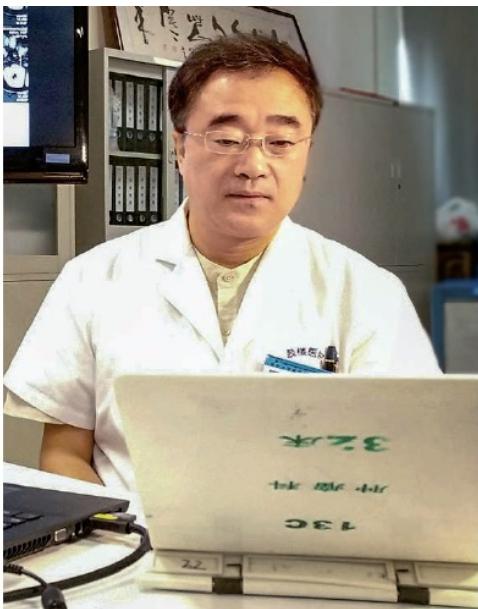
Reproductive biologist Shoukhrat Mitalipov and his team used

<http://www.nature.com/news/crispr-fixes-disease-gene-in-viable-human-embryos-1.22382>

GENOME EDITING

China sprints ahead in CRISPR therapy race

Human trials are using the genome-editing technique to treat cancers and other conditions



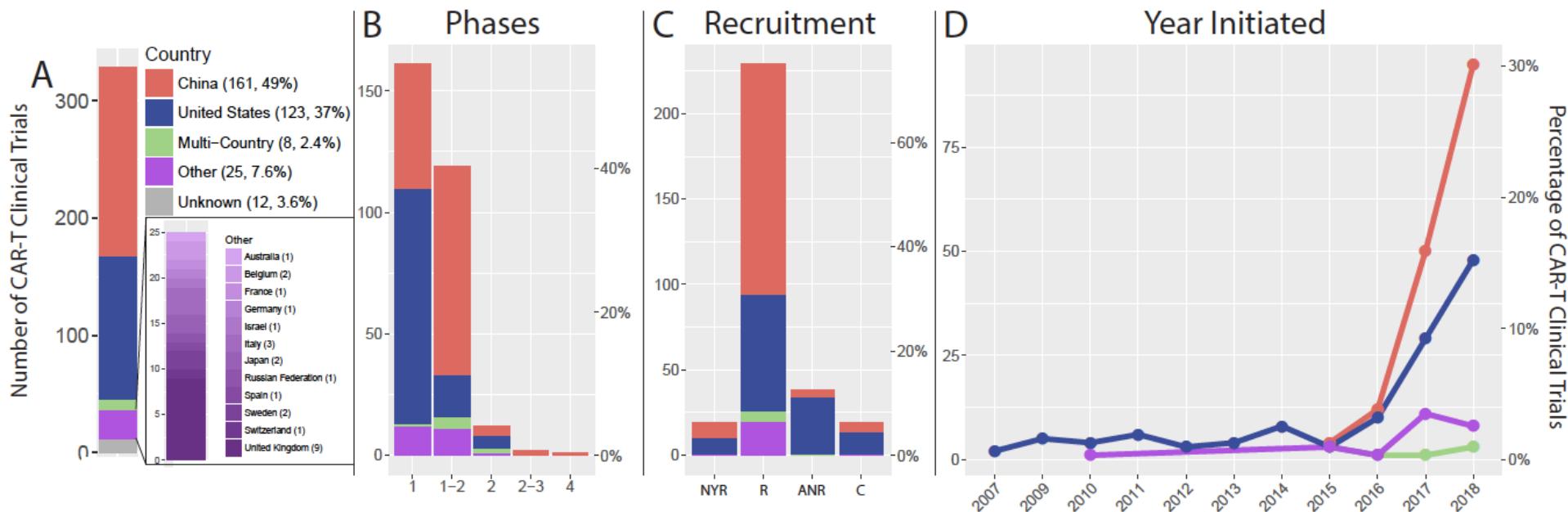
Liu Baorui is using CRISPR on two scourges in China: gastric and nasopharyngeal cancers.

Bringing CRISPR into the clinic in China

Chinese researchers are pioneering the genome-editing tool CRISPR in human therapy. Their rapid advances are driven in part by a rising cancer burden and a paucity of experimental drugs when conventional treatments fail.

INSTITUTION	CONDITION	INTERVENTION	STATUS
Affiliated Hospital to Academy of Military Medical Sciences, Beijing	HIV infection	CCR5 gene modification	Recruiting patients
First Affiliated Hospital of Sun Yat-sen University, Guangzhou	Cervical cancer	Disrupt human papillomavirus	In planning
Chinese PLA General Hospital, Beijing	Leukemia	Modified T cell receptors	Recruiting patients
Peking University, Beijing	Prostate cancer	PD-1 knockout T cells	In planning
Peking University, Beijing	Bladder cancer	PD-1 knockout T cells	In planning
Peking University, Beijing	Bladder cancer	PD-1 knockout T cells	In planning
Affiliated Nanjing Drum Tower Hospital of Nanjing University Medical School, Nanjing	Gastric and nasopharyngeal cancers	PD-1 knockout T cells	Recruiting patients
Hangzhou Cancer Hospital, Hangzhou	Esophageal cancer	PD-1 knockout T cells	Recruiting patients
Sichuan University, Chengdu	Lung cancer	PD-1 knockout T cells	Recruiting patients

An unprecedented explosion of CAR-T trials



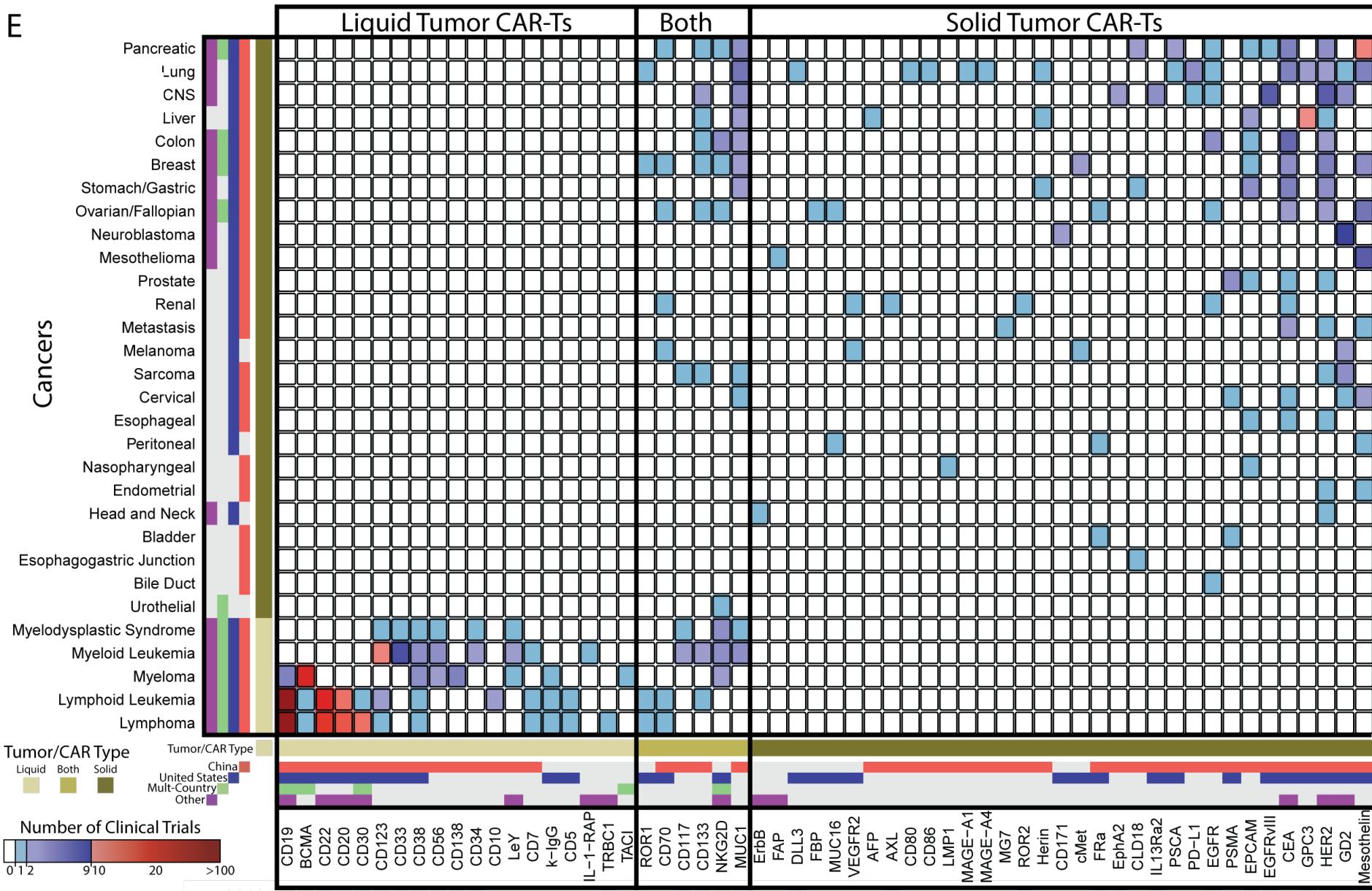
Analysis | Published: 06 January 2020

The therapeutic landscape for cells engineered with chimeric antigen receptors

<https://www.nature.com/articles/s41587-019-0329-2>

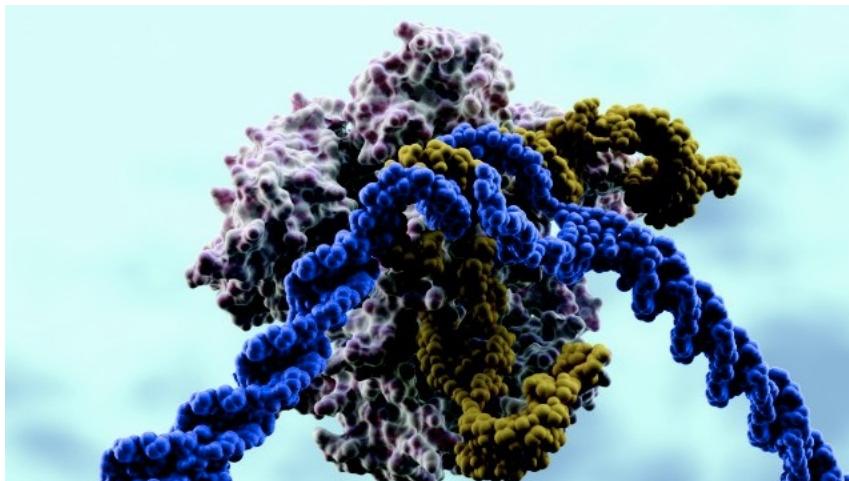
CAR targets span many cancer types now

E



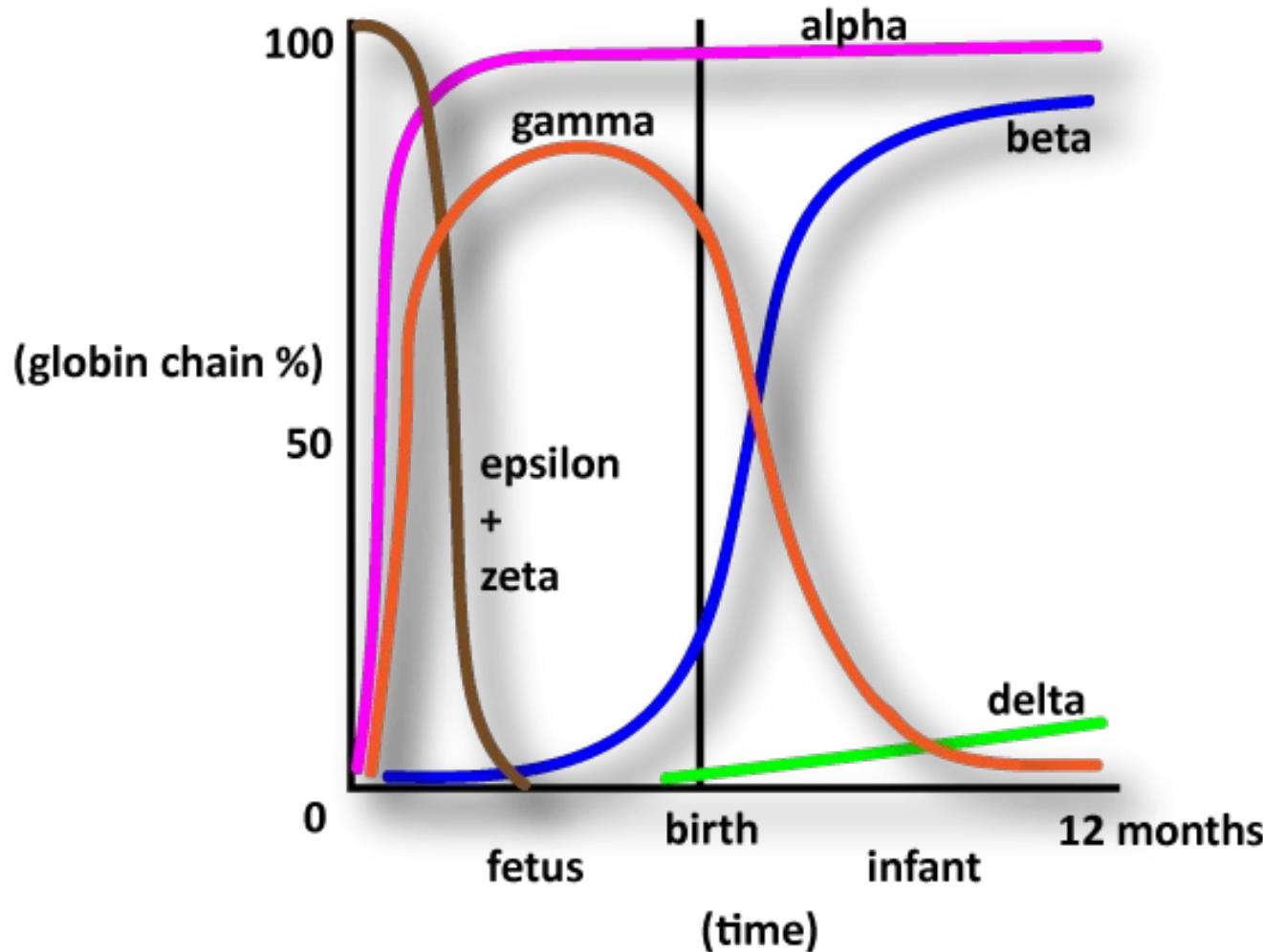
Adult trials for sickle cell disease and beta-thalassemia began in 2018

- CTX001 for editing BCL11A
- Custom Cas enzyme (Cpf1) for editing adult hemoglobin



<https://cen.acs.org/articles/95/web/2017/12/CRISPR-gene-editing-coming-clinic.html>

If alpha has a problem, why not bring back gamma?



Some successes already for Transfusion-dependent β-thalassemia (TDT) and sickle cell disease (SCD)

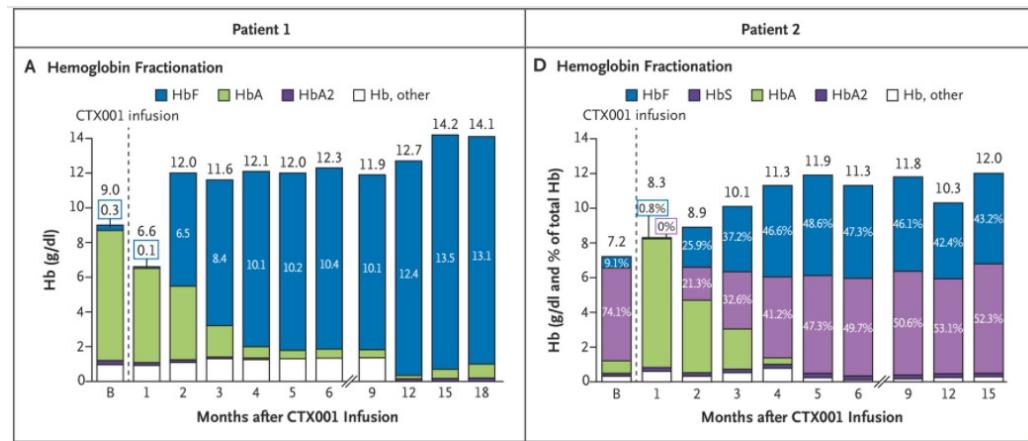
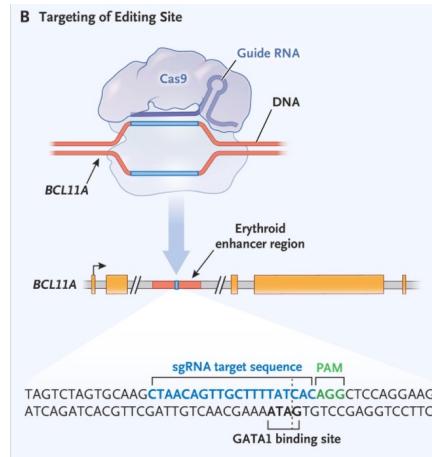
ORIGINAL ARTICLE BRIEF REPORT

January 21, 2021

N Engl J Med 2021; 384:252-260

CRISPR-Cas9 Gene Editing for Sickle Cell Disease and β-Thalassemia

Haydar Frangoul, M.D., David Altshuler, M.D., Ph.D., M. Domenica Cappellini, M.D., Yi-Shan Chen, Ph.D., Jennifer Domm, M.D., Brenda K. Eustace, Ph.D., Juergen Foell, M.D., Josu de la Fuente, M.D., Ph.D., Stephan Grupp, M.D., Ph.D., Rupert Handgretinger, M.D., Tony W. Ho, M.D., Antonis Kattamis, M.D., Andrew Kernytsky, Ph.D., Julie Lekstrom-Himes, M.D., Amanda M. Li, M.D., Franco Locatelli, M.D., Markus Y. Mapara, M.D., Ph.D., Mariane de Montalembert, M.D., Damiano Rondelli, M.D., Akshay Sharma, M.B., B.S., Sujit Sheth, M.D., Sandeep Soni, M.D., Martin H. Steinberg, M.D., Donna Wall, M.D., Angela Yen, Ph.D., and Selim Corbacioglu, M.D.



Prime editing makes things even better

Article | Published: 21 October 2019

Search-and-replace genome editing without double-strand breaks or donor DNA

Andrew V. Anzalone, Peyton B. Randolph, Jessie R. Davis, Alexander A. Sousa, Luke W. Koblan, Jonathan M. Levy, Peter J. Chen, Christopher Wilson, Gregory A. Newby, Aditya Raguram & David R. Liu



Nature **576**, 149–157 (2019) | [Cite this article](#)

the target site and encodes the desired edit. We performed more than 175 edits in human cells, including targeted insertions, deletions, and all 12 types of point mutation, without requiring double-strand breaks or donor DNA templates. We used prime editing in human

From epigenetic detection, to correction

Cell

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ARTICLE

Rescue of Fragile X Syndrome Neurons by DNA Methylation Editing of the *FMR1* Gene

X. Shawn Liu, Hao Wu, Marine Krzisch, Xuebing Wu, John Graef, Julien Muffat, Denes Hnisz, Charles H. Li, Bingbing Yuan, Chuanyun Xu⁵, Yun Li, Dan Vershkov, Angela Cacace, Richard A. Young, Rudolf Jaenisch⁶

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DOI: <https://doi.org/10.1016/j.cell.2018.01.012> | CrossMark

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Summary Full Text Methods Images/Data References Related Articles

Highlights

- Targeted demethylation of CGG repeats by dCas9-Tet1 reactivates *FMR1* in FXS cells
- Demethylation of CGG repeats induces an active chromatin status for *FMR1* promoter
- Methylation-edited FXS neurons behave similarly as wild-type neurons
- FMR1* reactivation by dCas9-Tet1 is sustainable in a human/mouse chimeric model

Summary

Fragile X syndrome (FXS), the most common genetic form of intellectual disability in males, is caused by silencing of the *FMR1* gene associated with hypermethylation of the CGG expansion mutation in the *FMR1* promoter. This study shows that targeted demethylation of the CGG repeats by dCas9-Tet1 reactivates the *FMR1* gene in FXS cells, leading to an active chromatin status for the *FMR1* promoter. The methylation-edited FXS neurons behave similarly as wild-type neurons. The reactivation is sustainable in a human/mouse chimeric model.

Highlights

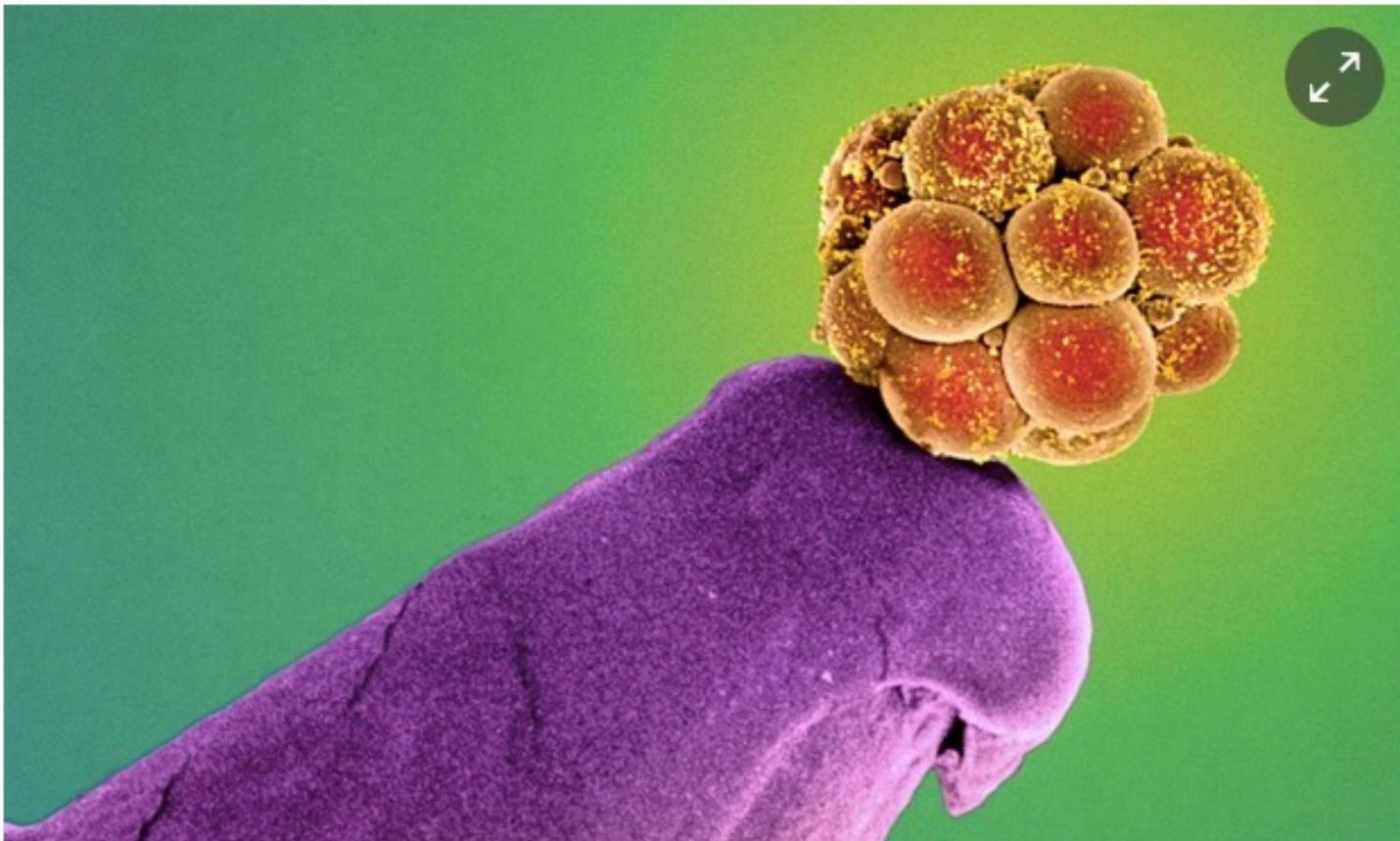
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Crispr: is it a good idea to 'upgrade' our DNA?

New genome-editing technology has the potential to eliminate genetic disease by making changes to our DNA that will pass down the generations. Such modification is currently banned in the UK but could that be about to change?



'Three-parent baby' test success could mean human trials in two years

To avoid hereditary diseases, not (just) to create superbabies.



David Lumb
06.08.16 in Medicine

19
Comments

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MPs vote in favour of 'three-person embryo' law

In historic debate Commons votes for controversial change to genetics law to allow mitochondrial transfer



MPs vote in favour of 'three-person' babies. Source: ITN

Perspective

“People get comfortable with technologies,” she says, citing how over time society has become relaxed about the use of IVF.

“I suspect this will be the same.”

– Dr. Jennifer Doudna.

Questions?