

All of Us Genetic Return of Results: Infrastructure, support, and communication of DNA results

December 7, 2022

Anastasia Wise, on behalf of the *All of Us* GHR3 team

All of Us Research Program

Agenda for Today

All of Us Research Program Genetic Return of Results

Infrastructure

- Participant choices
- DNA results

Support

- Support Center and genetic counseling
- Learning Center

Communication

- Comprehension and user testing
- Cultural awareness

1 M
or more
across

Inviting

1 Million

or more people
across the
United States



Data available from **372,000+** participants

80%

are from
underrepresented
communities



45%

are from racial
and ethnic
minority groups

Infrastructure

Participant Choice in Consent and Data Shared



1. Create an Account



3. Agree to Share Electronic Health Records (EHR)



5. Answer Health Surveys



2. Review the Consent



4. Complete the Consent to Get DNA Results

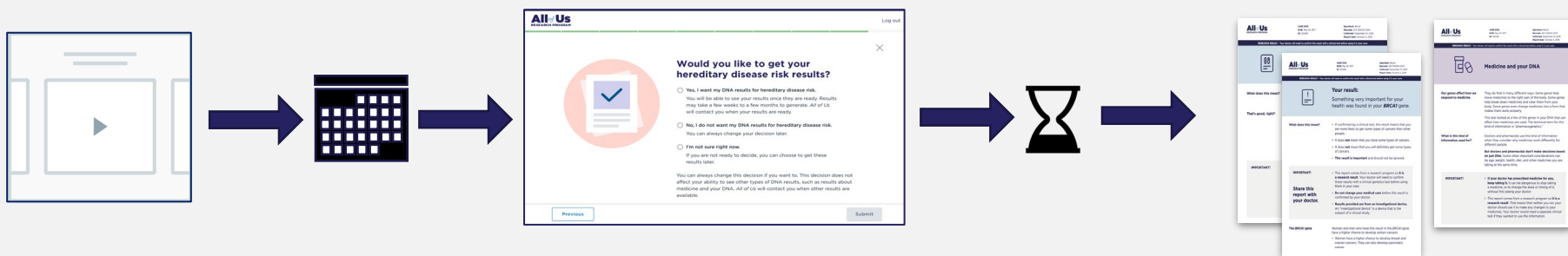


Participant Choice for DNA Results

Consent to Get DNA Results

Informing Loops

Genetic Return of Results



Informing loops **provide participants choice** on what types of DNA results they want after consent and the ability to **change** their minds

DNA results are generated when a participant says “yes” and can take from seconds for genetic traits to months for Hereditary Disease Risk results to produce

Genotyping and Whole Genome Sequencing



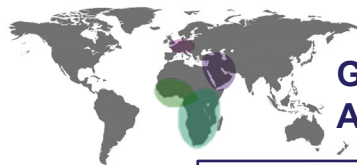
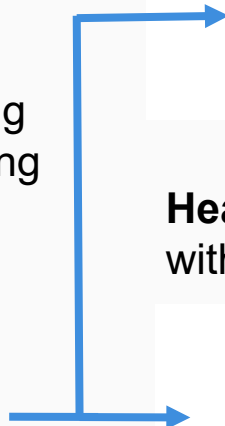
Biobank: biospecimen processing and DNA extraction



Genome Centers: genotyping and whole genome sequencing

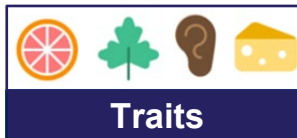


DRC: data repository and sharing with researchers



Genetic Ancestry

Genetic Ancestry and Traits starts with data from genotyping



Traits

Health-Related Research Reports starts with data from whole genome sequencing



Hereditary disease risk results
(HDR)



Medicine and your DNA results
(PGx)

Two Choices for Genetic Ancestry and Traits

DNA Results

You'll see all of your DNA results here when they're ready. See [options for your DNA results](#).

Genetic ancestry and trait results

5 results



Genetic ancestry

Genetic ancestry can be very interesting, but you may also learn information you didn't expect. [Learn more](#)

View Results



Bitter taste perception

Learn what your genes can tell you about how you taste bitter things.

View Results



Cilantro preference

Your genes play a role in whether you love cilantro or think it tastes like soap.

View Results



Earwax type

Flaky or sticky? Earwax type is in your genes.

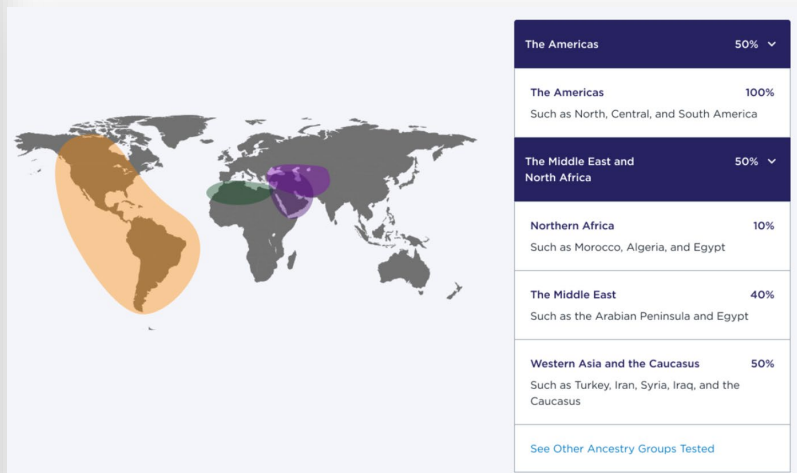
View Results



Lactose intolerance

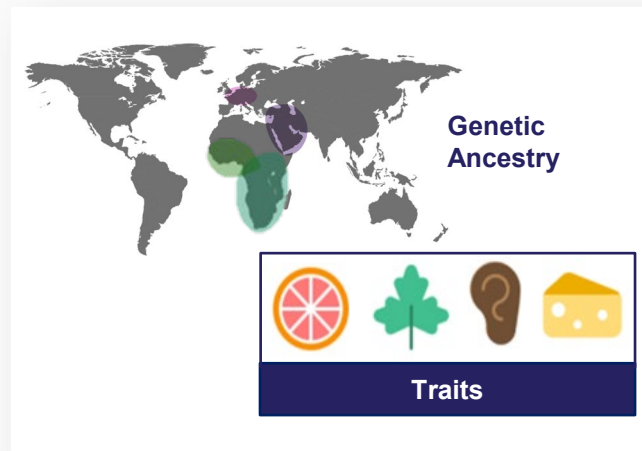
Your genes have a say in how well you digest lactose products like milk.

View Results



Genetic Ancestry and Traits (as of November 2022)

- **Over 180,000 participants sent notifications so far** (email, push, and SMS based on participant preference) to choose if they want to receive results
- **71% complete** genetic ancestry and traits **informing loop choice** (128k)
 - **88%** of those that view any result **view any of the 4 trait results** (bitter taste perception, cilantro preference, earwax type, and lactose intolerance) (113k)
 - **97%** of those that view any result **viewed their genetic ancestry results** (125k)



Public Announcement: December 10th, 2020 [press release](#) and [NIH Director's Blog](#)

Two Health-Related Research Report Choices at Launch



Hereditary Disease Risk
(HDR)



Medicine and Your DNA
(PGx)

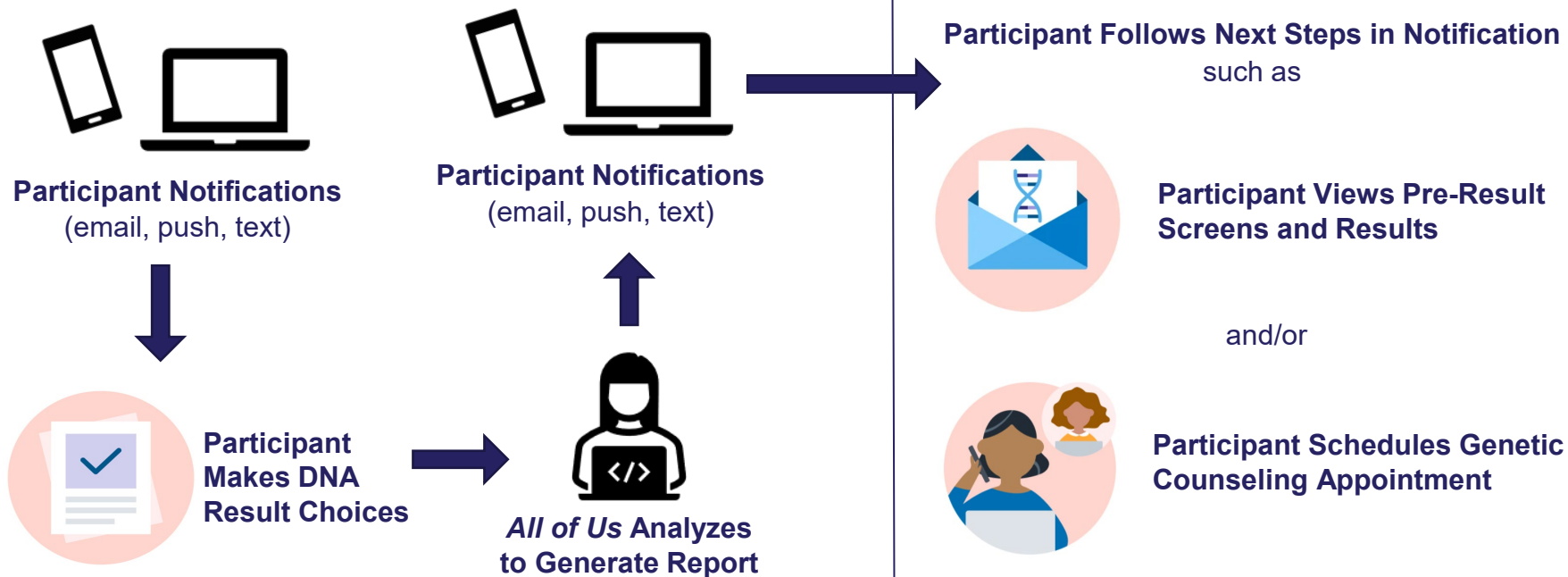
59 genes

Serious health conditions such as certain kinds of cancer or heart disease

7 genes

Genes that can affect how medicines are used

Participant Experience: Choice to Results



Participant notifications do not reveal results. Results can only be accessed by participants through the Genomics Platform portal online and genetic counseling.

DNA Results Available Online and as PDF Download

 **RESEARCH RESULT** - Your doctor will need to confirm this result with a clinical test before using it in your care.

[← Back](#)

Hereditary disease risk results

Your results

Risk with the *BRCA1* gene

Next steps



YOUR RESULT:

Something very important for your health was found in your *BRCA1* gene.

What does this mean?

This result means that you are more likely to get some types of cancers than other people.

It does **not** mean that you have some types of cancers. It does **not** mean that you will definitely get some types of cancers. This result is **important** and should not be ignored.

Important!

- [Share this report with your doctor.](#)
- This report comes from a research program, so **it is a research result**. Your doctor will need to confirm these results with a clinical DNA test before using them in your care.
- **Do not change your medical care** before this result is confirmed by your doctor.
- **Results provided are from an investigational device.** An "investigational device" is a device that is the subject of a clinical study.



All of Us
RESEARCH PROGRAM

JANE DOE
DOB: May 25, 1977
ID: 123456

Specimen: Blood
Barcode: 223 234234 2343
Collected: September 15, 2018
Report date: July 14, 2022

RESEARCH RESULT - Your doctor will need to confirm this result with a clinical test before using it in your care.



Your result:

Something very important for your health was found in your ***BRCA1*** gene.

What does this mean?

- If confirmed by a clinical test, this result means that you are more likely to get some types of cancers than other people.
- It does **not** mean that you have some types of cancers.
- It does **not** mean that you will definitely get some types of cancers.
- **This result is important** and should not be ignored.

IMPORTANT!

Share this report with your doctor.

- This report comes from a research program so **it is a research result**. Your doctor will need to confirm these results with a clinical genetics test before using them in your care.
- **Do not change your medical care** before this result is confirmed by your doctor.
- **Results provided are from an investigational device.** An "investigational device" is a device that is the subject of a clinical study.

Support

Support for Genetic Ancestry and Traits

- Participants contact Support Center 1st
- Escalate genetics questions not answered by FAQs
- Staffed by support agents and **genetic counselors**
- **English and Spanish**
- Interpretation available in >200 languages

Participant has a genetics question



Call



Email



Chat

Support Center replies with FAQ or escalates

Questions?

Our advisors are ready to answer your questions.
We're open from 7 a.m. to 10 p.m. Eastern, excluding public holidays.



Phone
(844) 842-2855



Email
help@joinallofus.org



Chat
[Start Online Chat >](#)

GCR Call Center replies to question



Call

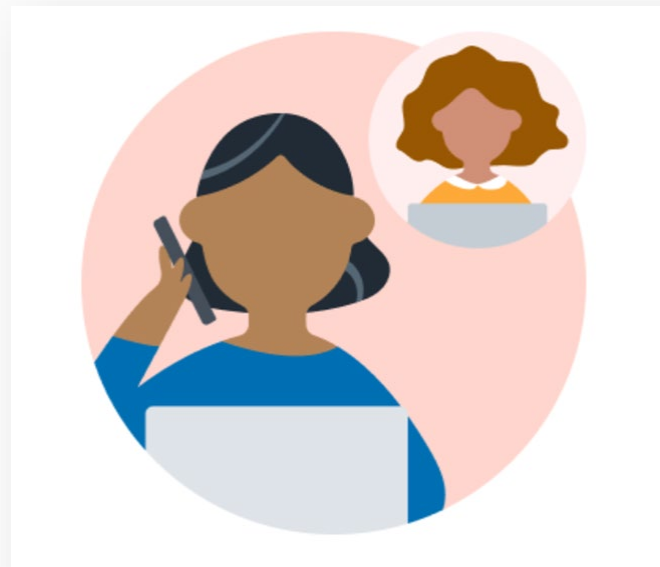


Email

>3 times
make FAQ

Genetic Counseling Appointment Scheduling

- **Participant next step to schedule a genetic counseling appointment** - result may not be pathogenic or likely pathogenic finding
- **All participants** with Hereditary Disease Risk or Medicine and Your DNA results can schedule
- Available for participants and family members (when participant present), health care providers also can call
- **Genetic counseling notes available** to participants in Genomics Platform portal online
- **No cost** to participants, can have multiple appointments
- **English and Spanish** with interpretation available in >200 languages



Appointments available within ~48 hours of notification

Learning Center

- **Additional educational content** for participants that want to learn more

LEARNING CENTER > DNA & GENOMICS



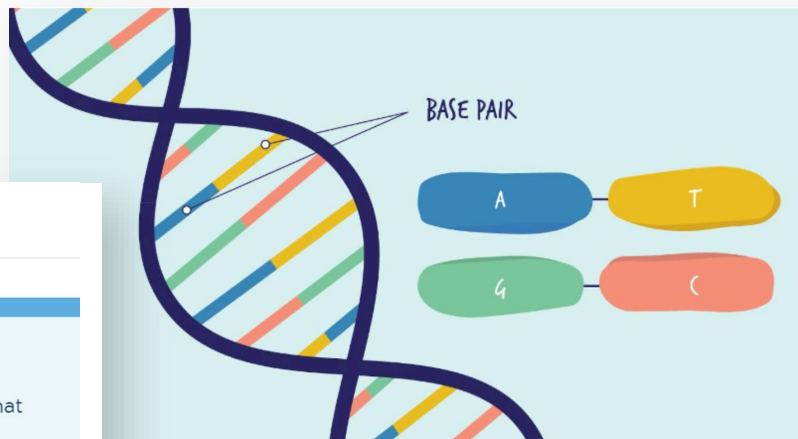
DNA & Genomics

DNA is an important source of information about health. Learn about the role of genes, what DNA does, and how genetic testing works.

Genes and Health

How do genes affect health and disease? ^

Our bodies are very complex. They require teamwork from many kinds of cells, which are made up of molecules. Proteins are specific molecules that carry out nearly everything happening inside our bodies. Genes are the instructions that tell your cells how to make different proteins. Each protein has its own job to do to keep our bodies running. A change in a gene's DNA can affect how well these proteins do their job. This can affect our health or potentially cause disease.



Includes:

- Topics to explore
- Images
- Videos

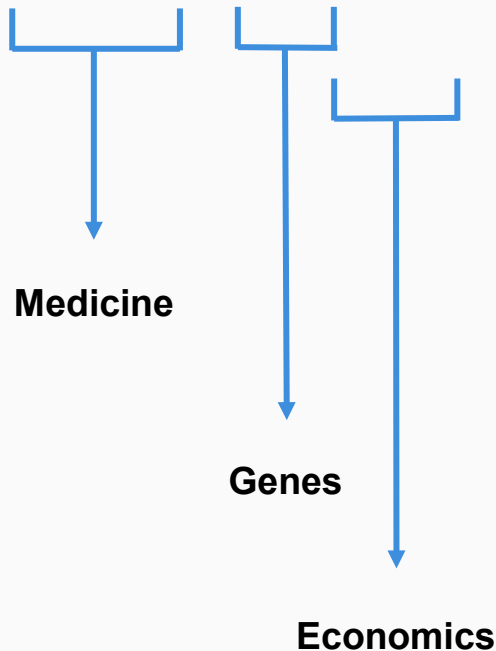
Communication

DNA Results: Comprehension and User Testing

Pharmacogenomics



Medicine and Your DNA



Learn where your ancestors may have lived.

Genetic ancestry results are estimates based on the information we have right now. This means your results will not tell you the origin of your ancestors for sure and they may change as we learn more.

People whose ancestors have less precise results. ?

Also, genetic ancestry results can affect your health care.

To create your genetic ancestry results, we compare your DNA to publicly available DNA data sets. These data sets have more DNA information from people with European genetic ancestry. This means genetic ancestry results may be more precise for people with European genetic ancestry. We hope to improve the estimates as we collect more data from people with other genetic ancestries.

likely to have less

affect your health

DNA Results: Cultural Awareness

Risk if your sex assigned at birth was female

[Why does this matter?](#)

If this result is confirmed by a clinical DNA test, it means:

- You have a higher chance of developing breast cancer, ovarian cancer, and pancreatic cancer.

This chart shows the risk for people assigned female at birth with this result in the *BRCA1* gene compared to the general population. *

Breast cancer

Risk by age 80

↑ Risk with a confirmed *BRCA1* result ¹ : 59-78%



👤 Average risk in the U.S. ² : 13%

Why does this matter?

Some diseases have different risks, treatments, or prevention plans based on a person's sex assigned at birth. Sex assigned at birth is based on the reproductive organs you are born with, your DNA, or both.

- This may or may not match your own gender identity. Gender identity is how you view yourself and what you call yourself. Your gender may be female, male, man, non-binary, transgender, woman, multiple genders, no gender, or something else. It can also change over time.
- Intersex people have variations in reproductive organs, genitalia, hormones, and/or anatomy that may be different from their sex assigned at birth or those often associated with female and male assignments. These differences may be present at birth or later.
- A doctor or health care provider who understands your unique situation can best address how to care for you.

Return of Genetic Results is an Iterative Process

All of Us Research Program Genetic Return of Results

Infrastructure

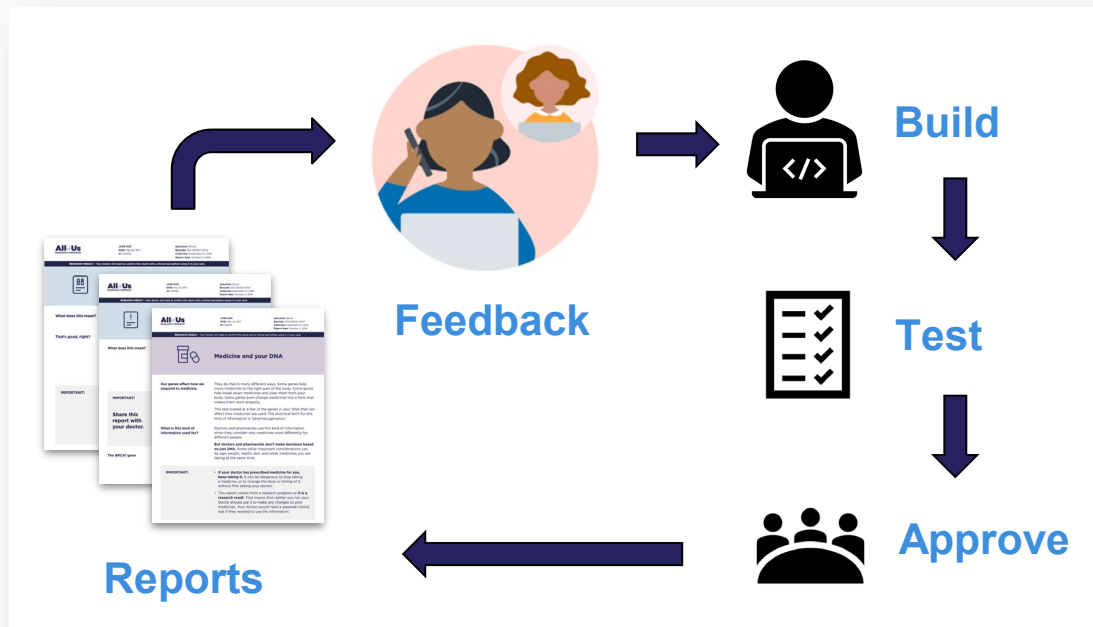
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Thank You!



<https://www.joinallofus.org>
<https://www.joinallofus.org/go>



National Institutes
of Health

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