Genetics Policy Hub
An Initiative of the National Coordinating Center
for the Regional Genetics Networks

Webinar Series

Direct-to-Consumer Testing – August 2023

This project is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) under Cooperative Agreement #UH9MC30770 from 6/2020-5/2024 for $800,000 per award year.

Genetics Policy Hub

Webinar Speakers

Nguyen Park, DMSc, PA-C, DFAAPA

ISCC-PEG DTC-GT Workgroup Member

Molly Caisse, MPH

Project Manager, NCC

GPH
Genetics Policy Hub

Learning Objectives

- Describe the importance of direct-to-consumer (DTC) genetic testing
- Identify current policies related to direct-to-consumer genetic testing

GPH
Genetics Policy Hub

Webinar Outline

- Basics of DTC Genetic Testing
- DTC Genetic Testing Information in GPH (NCC)
  - Overview of NCC and GPH
  - DTC Genetic Testing Specific Information
- Speaker on DTC Genetic Testing
- Q&A

GPH
Genetics Policy Hub

Definition of Direct-to-Consumer Genetic Testing

Direct-to-Consumer (DTC) genetic testing are tests that are ordered directly from the laboratory by the individual being tested, often without the input of a medical provider.

GPH
Genetics Policy Hub
An Initiative of the National Coordinating Center
for the Regional Genetics Networks

Background on Direct-to-Consumer Genetic Testing

Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG)

Composed of educators and healthcare practitioners, including physicians, physician assistants, pharmacists, and genetic counselors

Direct-to-Consumer Genetic Testing FAQ for Healthcare Professionals

The Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG) provides a venue for individuals to collaborate and develop educational resources. The opinions expressed in this resource do not reflect the view of all ISCC-PEG members, the National Institutes of Health, the Department of Health and Human Services, or the United States government.

genome.gov/iscc

The 3 W’s related to Direct-To-Consumer Genetic Testing (DTC-GT)

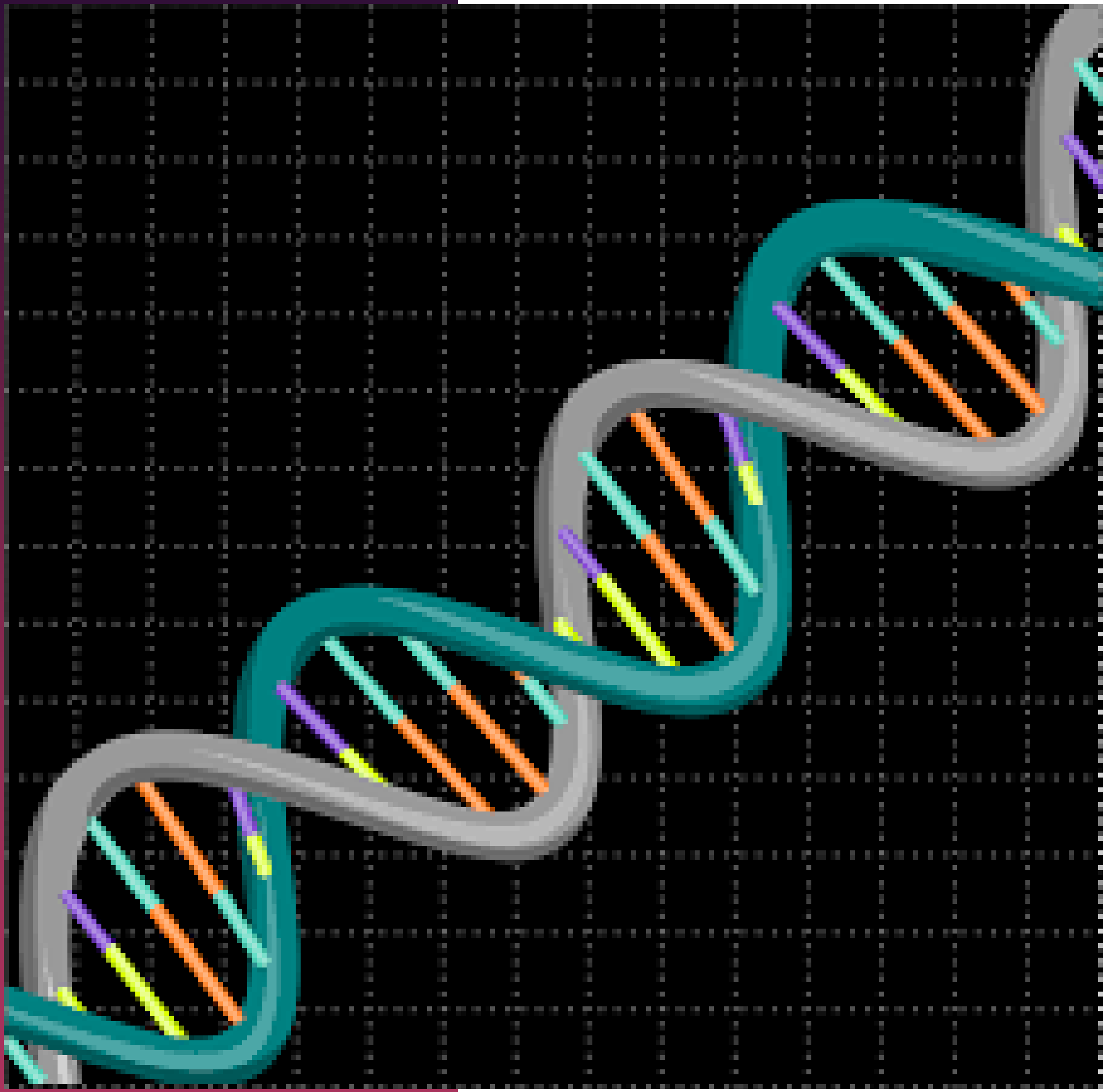
1. What
2. Who
3. Why

What: Concepts related to DTC Genetic Testing

- Information provided
- Limitations of testing

Genetic Testing Models

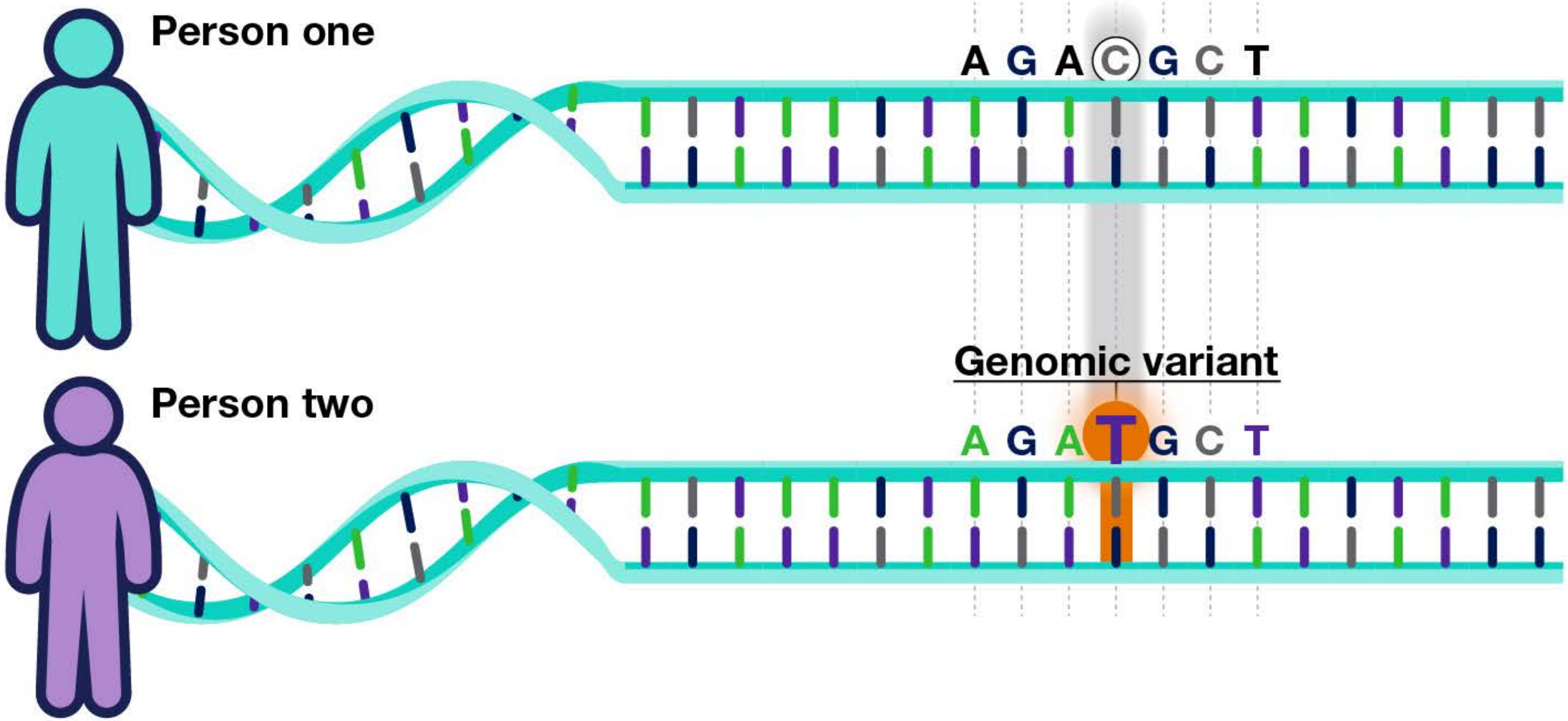
	Description	**Who orders the test?**	**How is consent obtained before the test?**	**Who discloses the results?**
Direct-to-consumer genetic testing (DTC-GT)	Healthcare professional not involved	Consumer	Direct to consumer via written information	Report issued directly to consumer
Provider-mediated genetic testing (PM-GT)	Consumer-initiated with healthcare professional involvement	Consumer or healthcare professional	Direct to consumer with/without healthcare professional	Report issued directly to consumer with optional access to healthcare professional
Clinic-based genetic testing	Facilitated by healthcare professional	Healthcare professional	By healthcare professional	Facilitated by healthcare professional; integrated into clinical care



What types of disease risk and health results does DTC-GT provide?

- Ashkenazi Breast Cancer (*BRCA*) variants
- Parkinson’s disease risk variants
- Dementia (APOe4) risk allele
- Macular Degeneration risk
- Factor V Leiden and Prothrombin (most common variant)
- Numerous carrier risk variants
- Familial hypercholesterolemia (some of the variants)
- Polygenic risk factors for DM2

Genomic Variant



https://www.genome.gov/Health/Genomics-and-Medicine/Polygenic-risk-scores

No Mr. (or Ms.) Perfect

"We are all flawed mutants."

- -Francis S. Collins, MD, PhD

We <u>all</u> have these glitches in our DNA.

0.4 percent of variation distinguishes you from other members of our species.

- Some glitches are significant, and others are not.

Types of Variants

- **Pathogenic**: responsible for causing disease
- **Likely pathogenic**: probably responsible for causing disease
- **Variant of uncertain significance (VUS)**: not currently confirmed to play a role in the development of dz
- **Likely benign**: Probably not responsible for causing disease
- **Benign**: Not responsible for causing disease.

Report types

Non-health related

Health/Wellness related

Ancestry
(composition, haplogroups, relatives)

Traits
(hair color, taste)

Paternity

Recreational
(wine preference)

Nutrigenomics
(diet supplementation)

Sports/fitness
(performance, injury)

Skin
(sun damage, skin type)

Longevity
(telomeres)

Carrier screening

Pharmacogenomics

Hereditary cancer

Adult onset disease
(Mendelian and multifactorial)

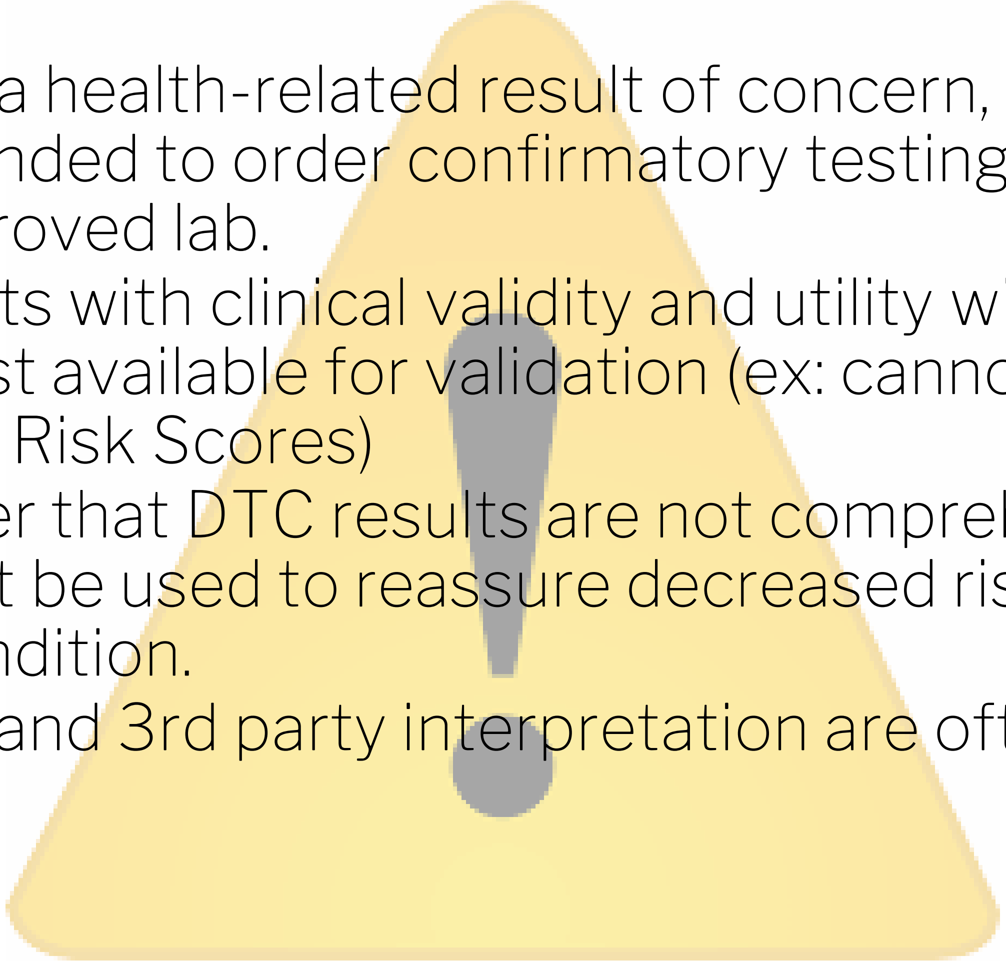
DTC-GT Example: Health Predispositions

REPORT	GENE(S)	VARIANTS	RELEVANT ETHNICITIES
BRCA1/BRCA2 (Selected Variants)	BRCA1 and BRCA2	3	Ashkenazi Jewish
Age-Related Macular Degeneration	ARMS2 and CFH	2	European
Alpha-1 Antitrypsin Deficiency	SERPINA1	2	European
Celiac Disease	HLA-DQB1 and HLA-DQA1	2	European
Familial Hypercholesterolemia	LDLR and APOB	24	All populations
G6PD Deficiency	G6PD	1	African
Hereditary Hemochromatosis (HFE-Related)	HFE	2	European
Hereditary Thrombophilia	F2 and F5	2	European
Late-Onset Alzheimer's Disease	APOE	1	All populations
Parkinson's Disease	LRRK2 and GBA	2	European, Ashkenazi Jewish, North African Berber

For patients who pursued DTC-GT:

- A positive result/ variant detected does not mean that they will invariably go on to develop the health condition.
- Additional testing may be needed to confirm or refute the results.
- Negative, absent, undetected or reassuring results may be a false negative.
- Additional testing may or may not be covered by health insurance.

Can DTC results be used in the management of patient care?



- If there is a health-related result of concern, it is recommended to order confirmatory testing from a CLIA-approved lab.
- Only results with clinical validity and utility will have a clinical test available for validation (ex: cannot validate Polygenic Risk Scores)
- Remember that DTC results are not comprehensive and should not be used to reassure decreased risk for a health condition.
- Raw data and 3rd party interpretation are often incorrect.

Who is affected by DTC-GT?

Privacy concerns- Who owns the data?

- GINA
- Current legislation

Who is getting tested?

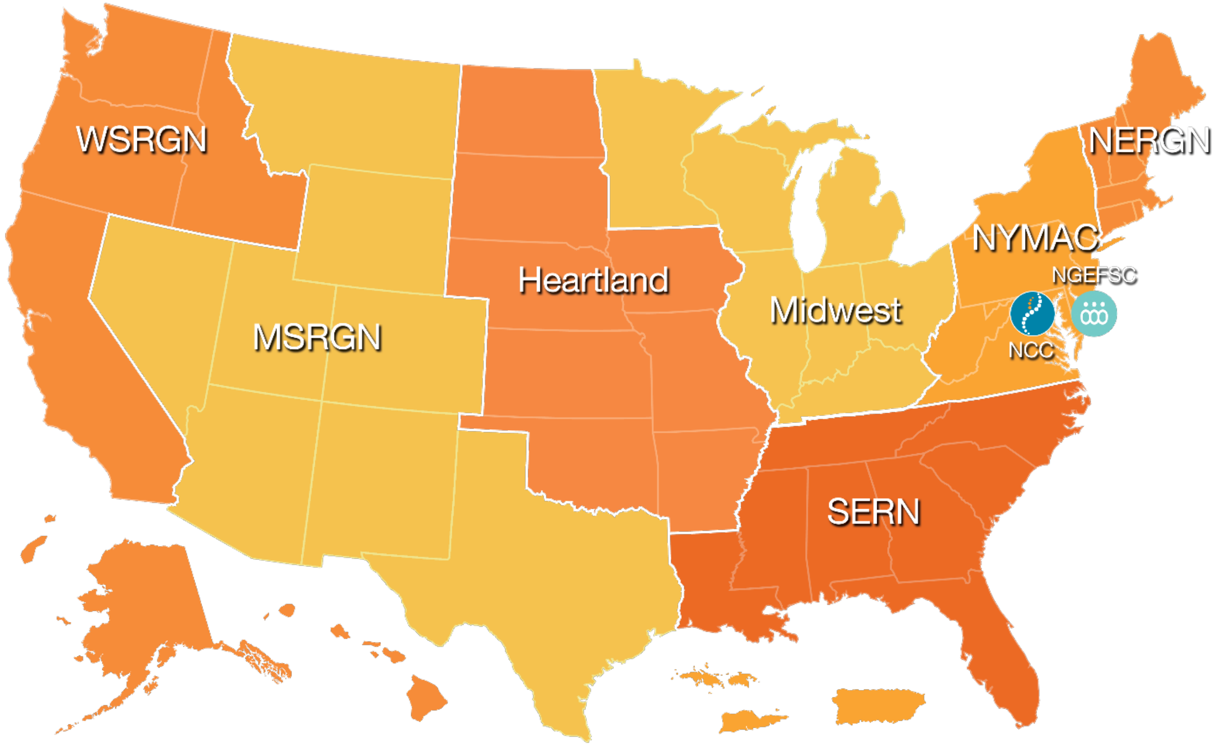
Who is affected?

GPH
Genetics Policy Hub
An Initiative of the National Coordinating Center
for the Regional Genetics Networks

Genetics Policy Information in GPH

Mission

Funded by the Health Resources and Services Administration (HRSA), the Regional Genetics Networks (RGNs), the National Coordinating Center for the Regional Genetics Networks (NCC), and the National Genetics Education and Family Support Center (NGEFSC or Family Center) mission is to improve access to genetic services for underserved populations.



Core Focus Areas

Genetics and Genomics Education

Telegenetics

Family Engagement and Partnership

Genetics Policy Hub

GPH
Genetics Policy Hub

Genetics Policy Hub Background

With the passage of the Affordable Care Act, the NCC and regional system began to develop specific resources to help support the genetic service delivery system through the lens of policy.

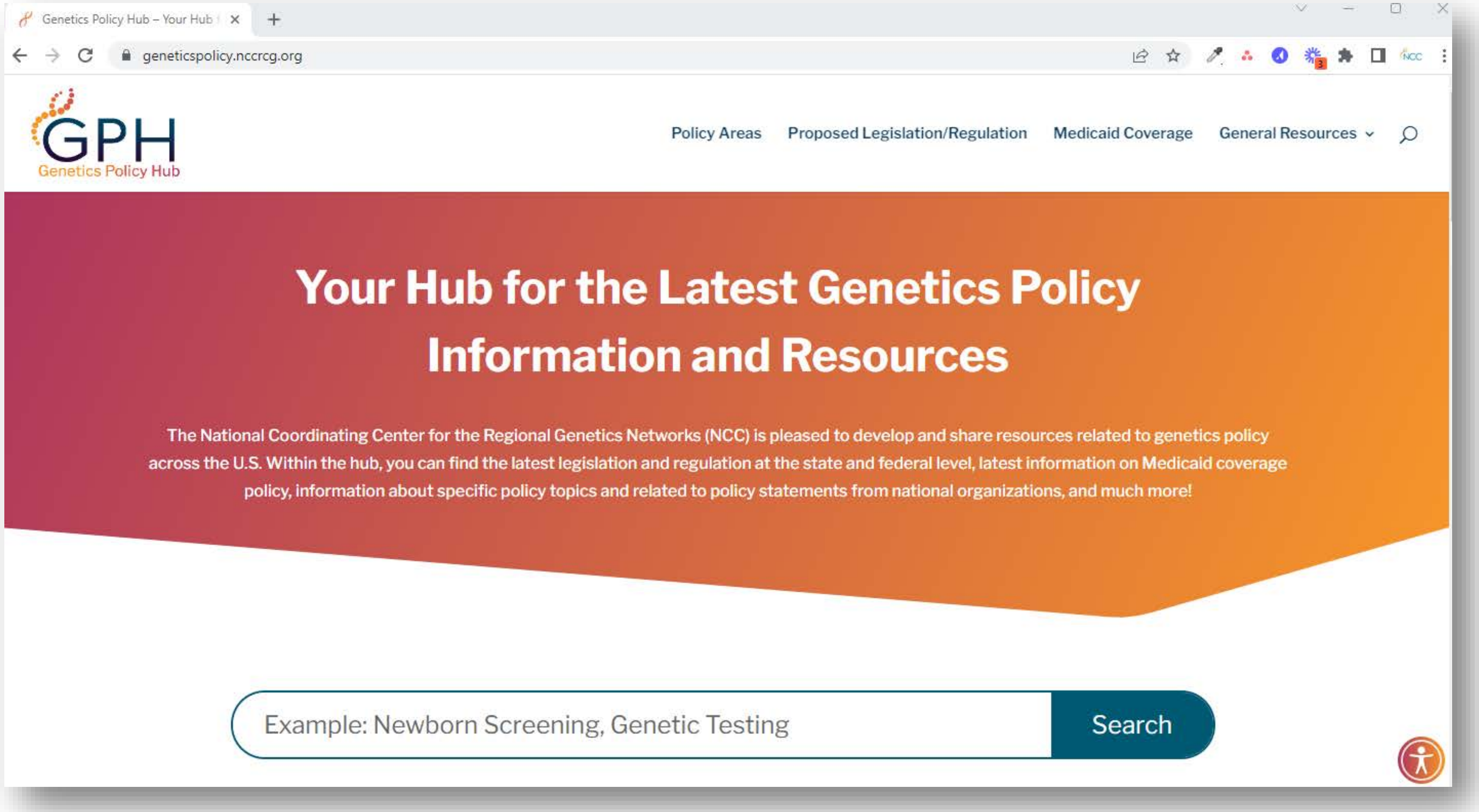
Since 2017, NCC has been tasked by HRSA to accomplish the following related to genetics policy:

- Monitor, analyze, track, and report on national policy issues related to the genetics care service delivery system with a focus on finance and insurance coverage; genetics workforce; and emerging issues in the use of genetic testing and technologies.
- Disseminate information on practices, policies, and resources related to the genetics care delivery system to states and stakeholders.

Genetics Policy Hub Background

geneticspolicy.nccrcg.org

NCC has developed multiple resources over the years to meet this goal and recently compiled all resources in a new policy website, called the Genetics Policy Hub.



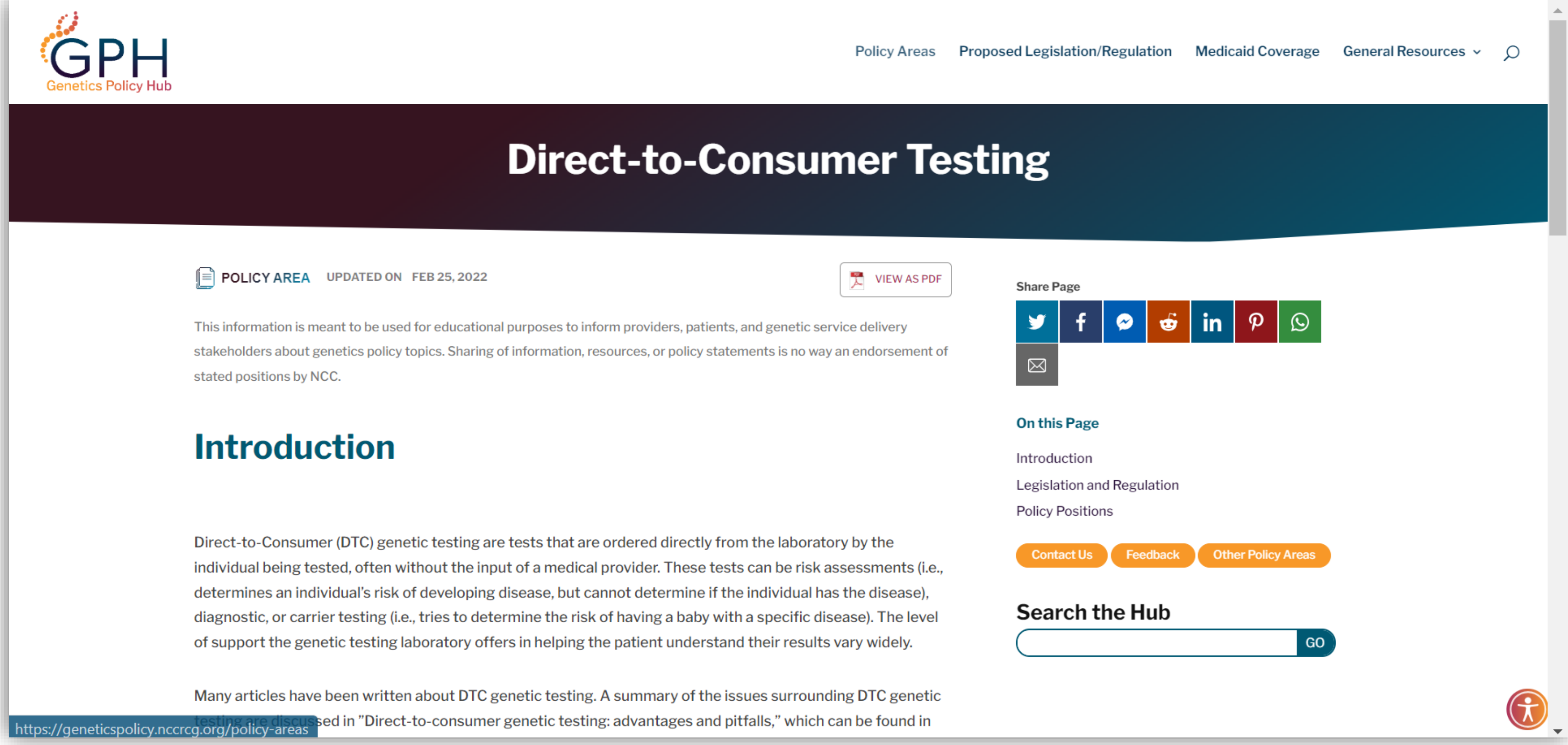
Your Hub for the Latest Genetics Policy Information and Resources

The National Coordinating Center for the Regional Genetics Networks (NCC) is pleased to develop and share resources related to genetics policy across the U.S. Within the hub, you can find the latest legislation and regulation at the state and federal level, latest information on Medicaid coverage policy, information about specific policy topics and related to policy statements from national organizations, and much more!

GPH
Genetics Policy Hub

DTC Genetic Testing Policy Area

geneticspolicy.nccrcg.org/policy-area/direct-to-consumer-testing/



Direct-to-Consumer Testing

This information is meant to be used for educational purposes to inform providers, patients, and genetic service delivery stakeholders about genetics policy topics. Sharing of information, resources, or policy statements is no way an endorsement of stated positions by NCC.

Introduction

Direct-to-Consumer (DTC) genetic testing are tests that are ordered directly from the laboratory by the individual being tested, often without the input of a medical provider. These tests can be risk assessments (i.e., determines an individual's risk of developing disease, but cannot determine if the individual has the disease), diagnostic, or carrier testing (i.e., tries to determine the risk of having a baby with a specific disease). The level of support the genetic testing laboratory offers in helping the patient understand their results vary widely.

Many articles have been written about DTC genetic testing. A summary of the issues surrounding DTC genetic testing are discussed in "Direct-to-consumer genetic testing: advantages and pitfalls," which can be found in

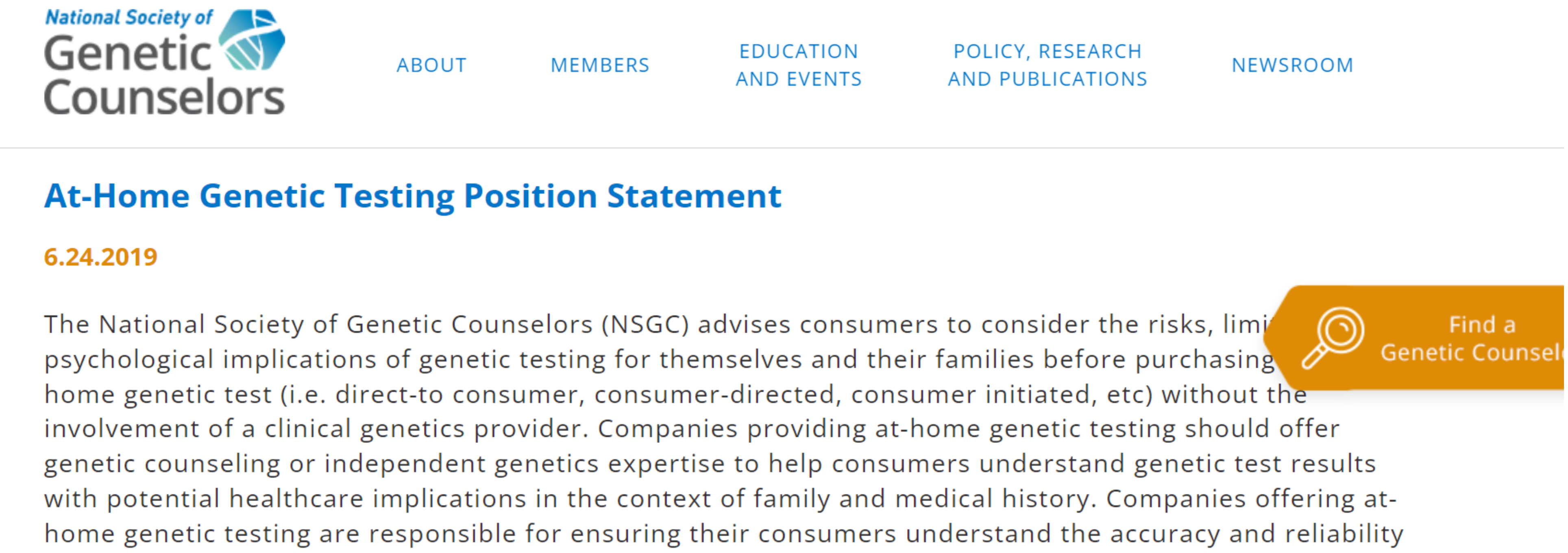
GPH
Genetics Policy Hub

DTC Genetic Testing Basics in the GPH Policy Area

AAFP

Direct to Consumer Genetic Testing

The AAFP recognizes that genetic testing is a complex process that should address risk for disease state, prevention, diagnosis, disease management, and implications for family members and reproductive health. Consumers should be informed regarding the utility of genetic testing in determining risks for future health state, the possibility of inconclusive results, and if or how this information could be acted upon by that individual and/or family members. The AAFP encourages the FDA to assess and oversee analytical validity, clinical validity, and accuracy of claims made by companies performing direct to consumer genetic testing.



At-Home Genetic Testing Position Statement

6.24.2019

The National Society of Genetic Counselors (NSGC) advises consumers to consider the risks, limi psychological implications of genetic testing for themselves and their families before purchasing home genetic test (i.e. direct-to consumer, consumer-directed, consumer initiated, etc) without the involvement of a clinical genetics provider. Companies providing at-home genetic testing should offer genetic counseling or independent genetics expertise to help consumers understand genetic test results with potential healthcare implications in the context of family and medical history. Companies offering at-home genetic testing are responsible for ensuring their consumers understand the accuracy and reliability

Genetics Policy Hub

Proposed And Enacted Legislation Related to DTC Genetic Testing

geneticspolicy.nccrcg.org/legislative-policies/

- Currently in 2023, there are 2 enacted and 1 introduced state bill related to Direct-to-Consumer Genetic Testing:
  - Enacted:
    - Tennessee HB 1310
    - Texas HB 2545
  - Introduced: Minnesota (SF 1138/HF 1520)

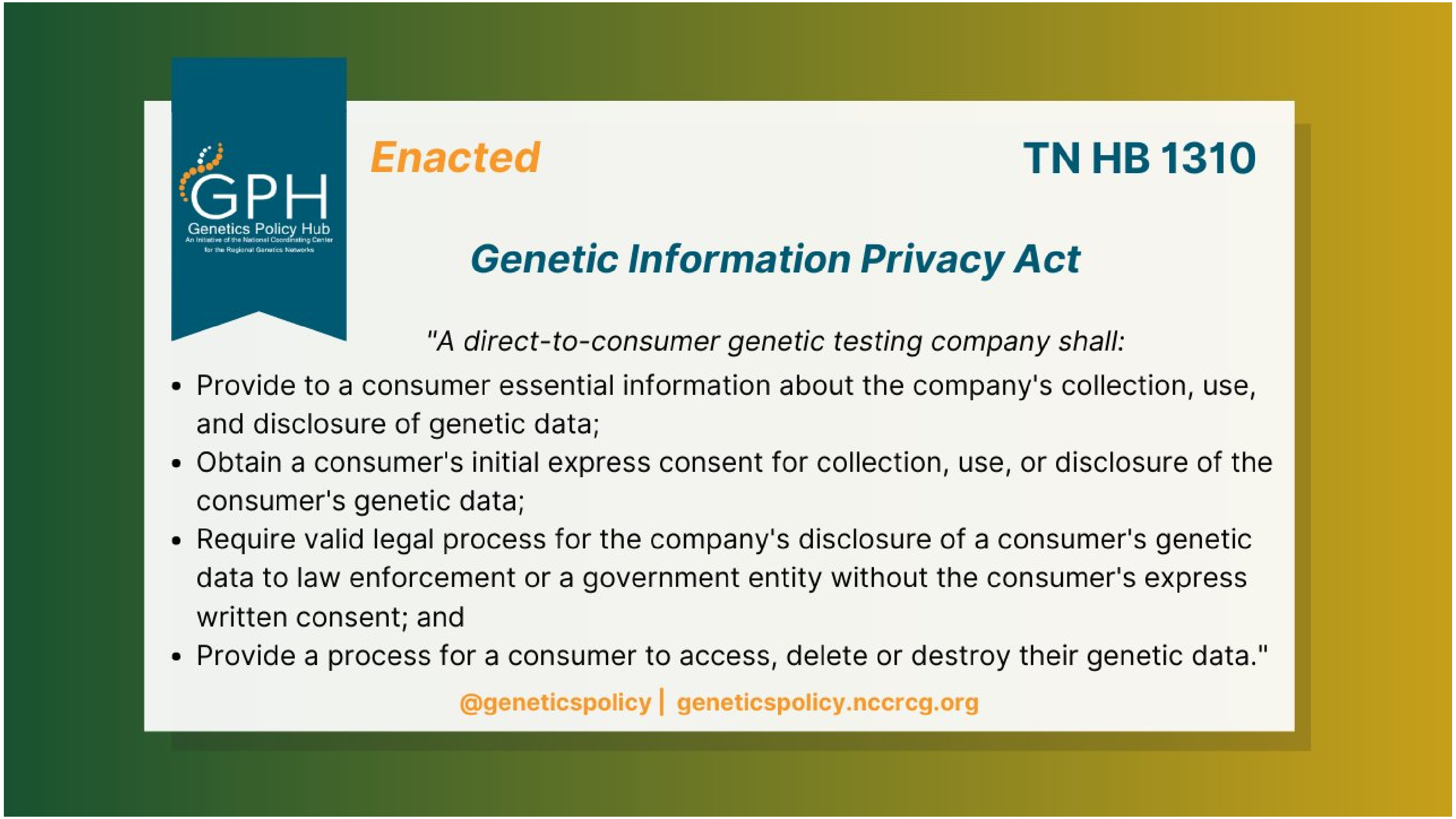
- In 2022, 2 state bills were enacted related to Direct-to-Consumer Genetic Testing:
  - Maryland (HB 866)
  - Kentucky (HB 502)

Genetics Policy Hub

2023 Sample Enacted Legislation Related to DTC Genetic Testing

https://bit.ly/TennDTC

— Enacted in Tennessee on April 28, 2023:



Genetics Policy Hub
An Initiative of the National Coordinating Center for the Regional Genetics Networks

Enacted

TN HB 1310

Genetic Information Privacy Act

"A direct-to-consumer genetic testing company shall:

- Provide to a consumer essential information about the company's collection, use, and disclosure of genetic data;
- Obtain a consumer's initial express consent for collection, use, or disclosure of the consumer's genetic data;
- Require valid legal process for the company's disclosure of a consumer's genetic data to law enforcement or a government entity without the consumer's express written consent; and
- Provide a process for a consumer to access, delete or destroy their genetic data."

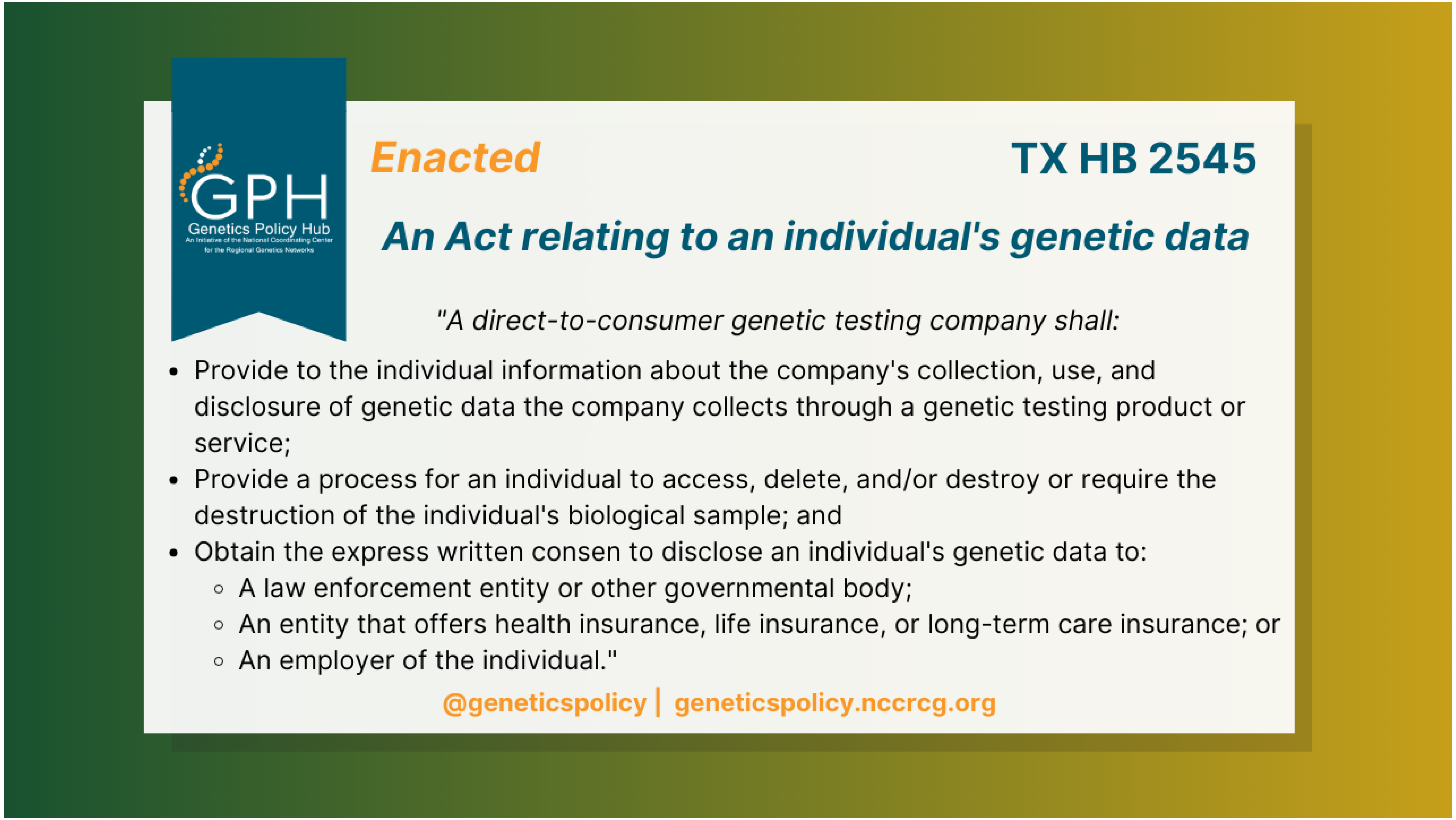
@geneticspolicy | geneticspolicy.nccrcg.org

Genetics Policy Hub

2023 Sample Enacted Legislation Related to DTC Genetic Testing

https://bit.ly/TexasDTC

Enacted in Texas on June 17, 2023:



Genetics Policy Hub
An Initiative of the National Coordinating Center for the Regional Genetics Networks

Enacted **TX HB 2545**

An Act relating to an individual's genetic data

"A direct-to-consumer genetic testing company shall:

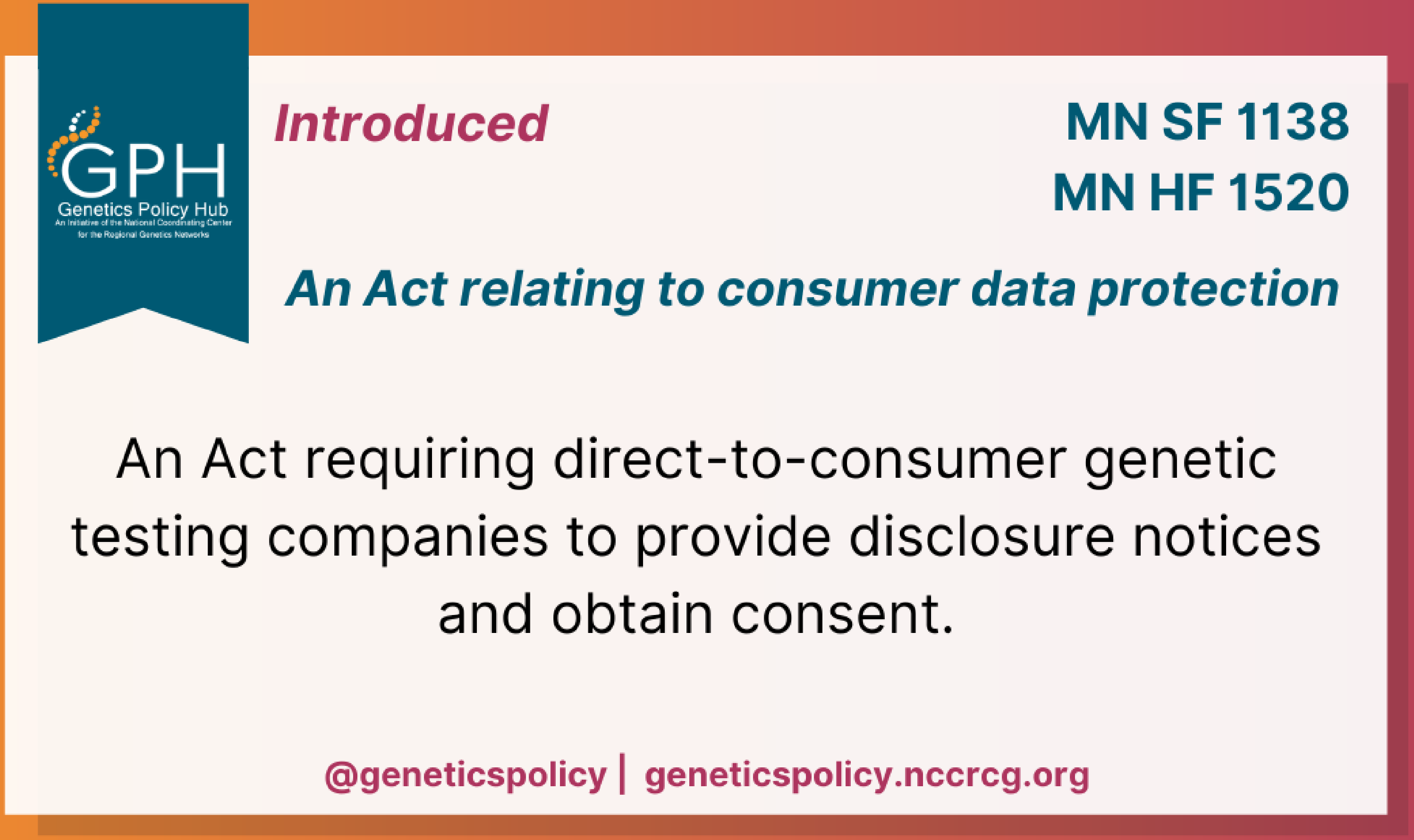
- Provide to the individual information about the company's collection, use, and disclosure of genetic data the company collects through a genetic testing product or service;
- Provide a process for an individual to access, delete, and/or destroy or require the destruction of the individual's biological sample; and
- Obtain the express written consen to disclose an individual's genetic data to:
  - A law enforcement entity or other governmental body;
  - An entity that offers health insurance, life insurance, or long-term care insurance; or
  - An employer of the individual."

@geneticspolicy | geneticspolicy.nccrcg.org

Genetics Policy Hub

2023 Sample Proposed Legislation Related to DTC Genetic Testing

- Introduced in MN on February 2, 2023
- Currently referred to Subcommittees



Genetics Policy Hub
An Initiative of the National Coordinating Center for the Regional Genetics Networks

Introduced

MN SF 1138
MN HF 1520

An Act relating to consumer data protection

An Act requiring direct-to-consumer genetic testing companies to provide disclosure notices and obtain consent.

@geneticspolicy | geneticspolicy.nccrcg.org

Genetics Policy Hub

2022 Sample Enacted Legislation Related to DTC Genetic Testing

- Maryland (HB 866) – An Act Concerning Genetic Information Privacy Consumer Protection and Forensic Genealogy
  - Enacted May 29, 2022
  - "Regulates the use of genetic data by direct-to-consumer genetic testing companies"
  - "Obtain certain consents from consumers before collecting, using, or disclosing the consumer's genetic data
  - "Protect genetic data and provide for certain disclosures to law enforcement and other government agencies."
- Kentucky (HB 502) - An Act relating to genetic information
  - Enacted April 8, 2022
  - "To safeguard the privacy, confidentiality, security, and integrity of a consumer's genetic data"
  - Similar language as MD HB 866 and other bills mentioned

Download Our DTC Genetic Testing Policy Brief

bit.ly/GeneticDTCPolicy

Policy Overview

August 2023

Direct-to-Consumer (DTC) Genetic Testing

The Genetics Policy Hub (GPH), a program of the National Coordinating Center for the Regional Genetics Networks, is a policy education website where you can explore state-based policies, ranging from Medicaid to policies being proposed by state governments to federal policies, ranging from proposed legislation to policy statements written by professional organizations. This policy overview highlights the data within GPH related to genetics privacy. This document is for informational purposes only, and specific questions about the information presented should be directed to the government agencies or organizations discussed. If you have questions about our methodology or feedback on what should be added to GPH, please get in touch with geneticspolicy@nccrcg.org.

Direct-to-Consumer (DTC) genetic testing are tests that are ordered directly from the laboratory by the individual being tested, often without the input of a medical provider. These tests can be risk assessments (i.e., determines an individual's risk of developing disease, but cannot determine if the individual has the disease), diagnostic, or carrier testing (i.e., tries to determine the risk of having a baby with a specific disease). The level of support the genetic testing laboratory offers in helping the patient understand their results vary widely.

Enacted and Proposed Legislation and Regulation

Updated Weekly

3 pieces of legislation are enacted or proposed related to direct-to-consumer genetic testing.

The enacted legislation:

- TN HB 1310, TX HB 2545: requires direct-to-consumer genetic testing companies to obtain consent and provide information regarding the collection, use, and disclosure of the consumer's genetic data

The proposed legislation:

- MN SF 1138/MN HF 1520: would require direct-to-consumer genetic testing companies to provide disclosure notices and obtain consent

https://geneticspolicy.nccrcg.org/legislative-policies/

Medicaid Policies

Updated Annually Each Fall

State Medicaid programs do not specifically state policies related to direct-to-consumer genetic testing. However, there may be broad policies protecting use of genetic data. Check your state's general Medicaid policies by visiting your program's website.

Updated as needed

8 Policy statements related to direct-to-consumer genetic testing from professional organizations

Professional organizations write policy statements to communicate what they believe is best for their community. Many professional organizations in the genetic system have written policy statements about direct-to-consumer genetic testing. These statements range from general policies on consumer protections to specific policies on how research should be conducted.

https://geneticspolicy.nccrcg.org/policy-area/direct-to-consumer-testing/

geneticspolicy.nccrcg.org

@geneticspolicy

This project is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) under Cooperative Agreement #UH9MC30770 from 6/2020-5/2024 for $800,000 per award year. This information or content and conclusions are those of the author and should not be construed as the official position or policy of, nor should any endorsements be inferred by HRSA, HHS or the U.S. Government.

GPH

Genetics Policy Hub

An Initiative of the National Coordinating Center for the Regional Genetics Networks

Deep Dive into Policy Perspectives

Question

Genetic Information Nondiscrimination Act (GINA) protects people from discrimination in the following situations:

a. Disability insurance
b. Healthcare insurance
c. Long-term care insurance
d. None of the above
e. All of the above

Genetic Information Nondiscrimat-ion ACT (GINA)

GINA prohibits health insurers from discrimination based on the genetic information

Genetic information cannot be used to determine if someone is eligible for insurance or to make coverage, underwriting or premium-setting decisions.

Health insurers may not request or require individuals or their family members to undergo genetic testing or to provide genetic information

Answer

Genetic Information Nondiscrimination Act (GINA) protects people from discrimination in the following situations:

a. Disability insurance
b. Healthcare insurance
c. Long-term care insurance
d. None of the above
e. All of the above

Question

GINA protects workers from having genetic information used against them in which of the following instances?

a. Companies of all sizes

b. The military

c. Hiring

d. All of the above

Exceptions to Title II of GINA

Answer

GINA protects workers from having genetic information used against them in which of the following instances?

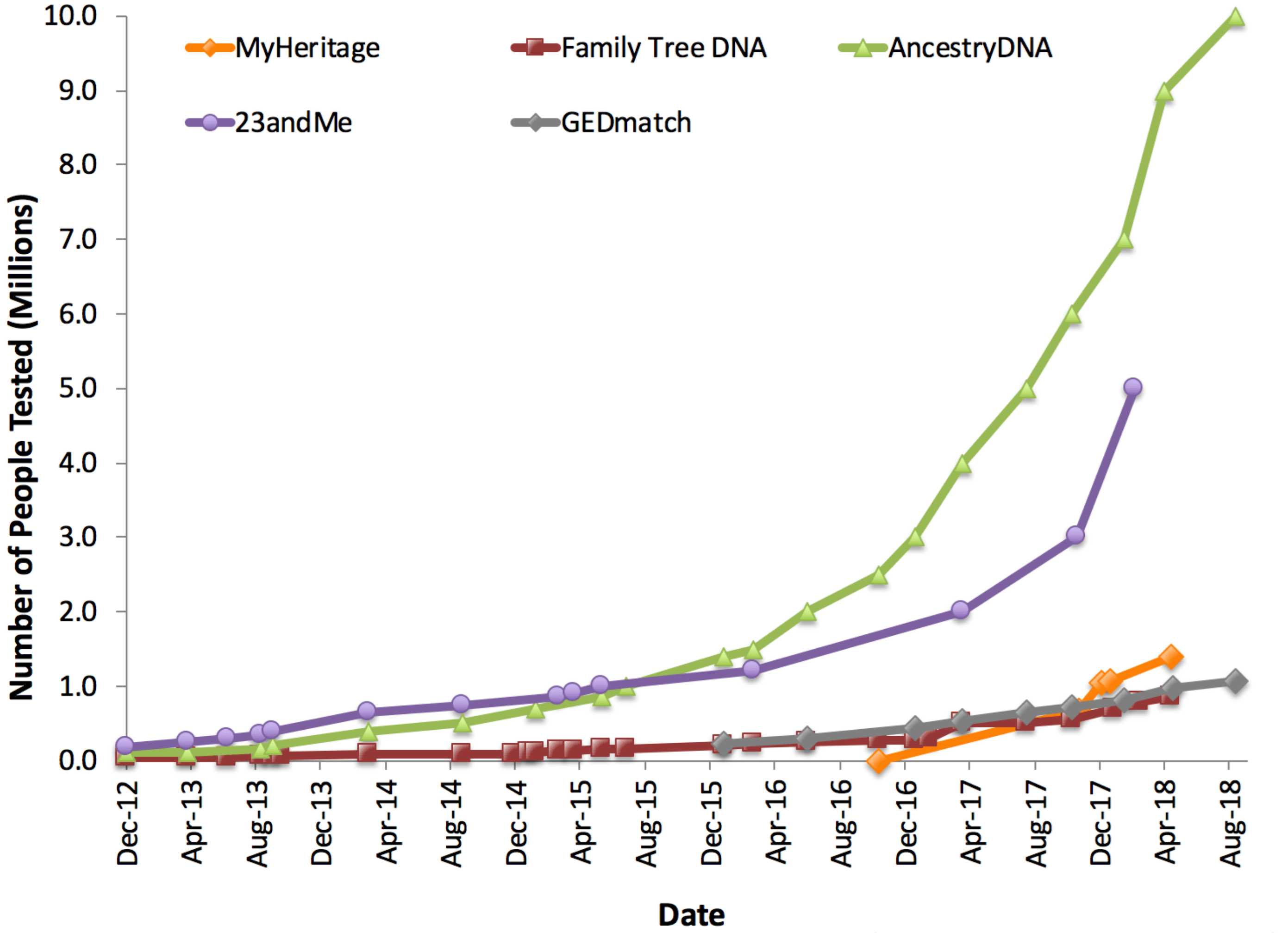
a. Companies of all sizes

b. The military

c. Hiring

d. All of the above

Consumer genomic databases rapidly growing



© 2018 by Leah Larkin, www.theDNAgeek.com. Sources: Company press releases (AncestryDNA, 23andMe, MyHeritage), ISOGG wiki "Autosomal DNA testing comparison chart" edit history (FTDNA), personal communcations (GEDmatch)

Who: Categories to Consider

Who is getting Tested?

- Everyone
- Adoptees
- People conceived using donor eggs and sperm

What if an adoptee is considering DTC-GT for health information?

There are limitations of DTC-GT for health information.

If adoptees test negative for disease risks and health results via DTC-GT, they may have a false sense of security because they may carry a pathogenic variant not included in DTC-GT.

If they test positive for a genetic variant related to disease, they should confirm these results with clinical genetic testing.

What if the person considering DTC-GT was conceived using donor eggs or sperm and wants to use DTC-GT to obtain health information?

- DTC-GT results may violate donor agreements for anonymity.
- Patients have the option to connect with their donor egg and/or sperm bank to enquire about genetic screening that was performed.
- Consider out-of-pocket clinical genetic testing

Why: Ramifications of DTC Genetic Testing

Why is DTC genetic testing being performed?

Why is it important to consider family history?

Why is it important to understand the ramifications of genetic testing?

Long-term planning

Effects on family

Benefits of DTC-GT

- Increases awareness about genetic conditions
- Can remove some of the fear and stigma that may be associated with genetic testing and with discussions around family health history
- Easy, inexpensive, no insurance or medical system involved
- Empowers, informs, entertains
  - Ancestry
  - Health
  - Paternity
  - Sports/Fitness/Nutrition
  - ‘Benign’ genetic trait

Limitations of DTC-GT

- Testing/interpretation methods are limited
- Does not test for all pathogenic variants for a particular condition, so results cannot be used to rule out genetic condition
- Even if DTC-GT shows a potentially actionable result (e.g., a test detects a BRCA1 pathogenic variant), these results need to be confirmed with a clinical genetic test; therefore, DTC-GT may add an unnecessary step to the process, as well as additional time and cost
- Results obtained from the tests should not be used for diagnosis or to inform treatment decisions.
- For ancestry results, DTC-GT may reveal a genetic relationship to biological relatives that is unexpected and possibly distressing.
- Concerns about privacy and use of data
- Limited informed consent

Why: Ramifications on Long-term Planning & Family

- Ancestry
- Gate-keeping
- Parent-child
- Siblings
- Guilt/Blame

Genomics

GENOMICS

Our genes can suggest what diseases we *might* be predisposed to, but it's an incomplete picture of human health.

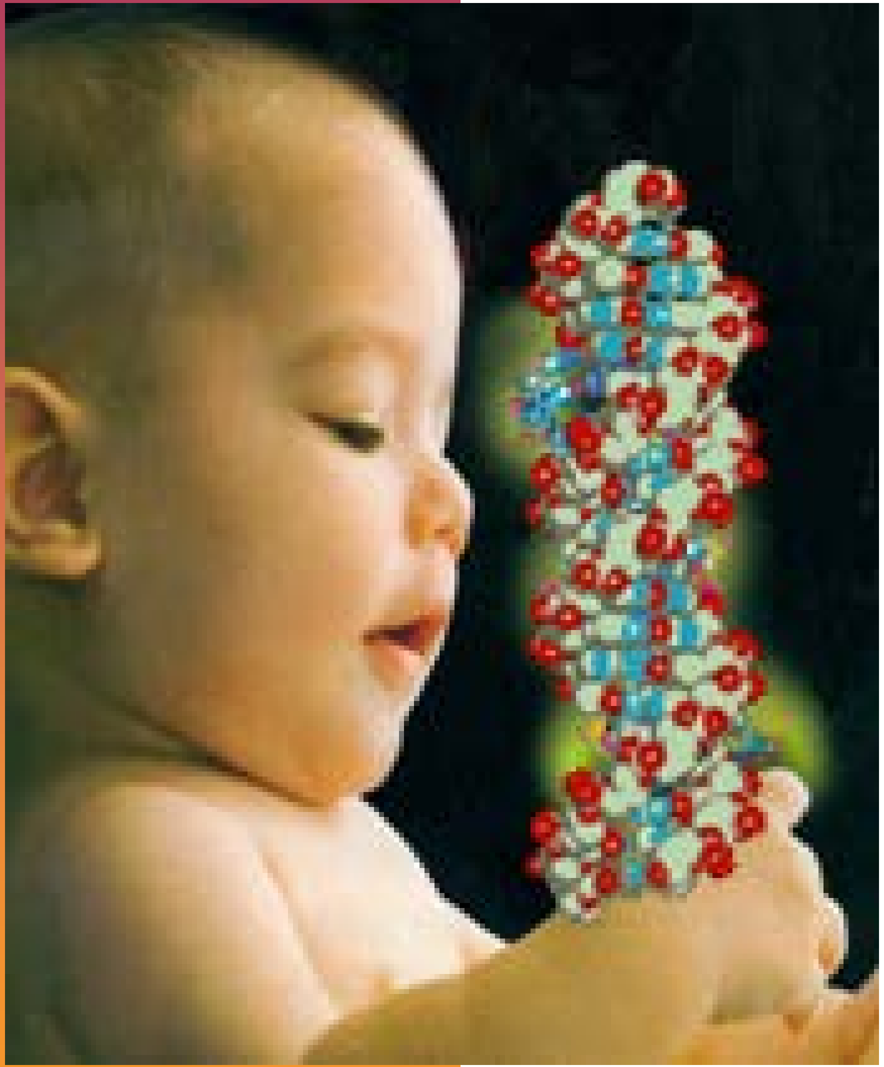
PHENOTYPE

A snapshot of the current state of health that can be used to prevent, diagnose and treat disease or improve health.

LIFESTYLE/ENVIRONMENT

External factors like diet, exercise, medications, microbiota and even where we live influence our metabolic state.

Individual differences make us who we are



Personalized Medicine…starts with a family history

Family History

CDC study:

<30 % of Americans have actively collected health information from their relatives.

96% believe this information is important to know.

Genetics Policy Hub
An Initiative of the National Coordinating Center
for the Regional Genetics Networks

Question and Answer

GPH
Genetics Policy Hub
An Initiative of the National Coordinating Center
for the Regional Genetics Networks

Connect With Us

mcaisse@nccrcg.org

www.nccrcg.org

www.geneticspolicy.nccrcg.org

www.phgw.org

@nccrcg

@nccrcg

@nccrcg

@nccrcg

@nccrcg
@geneticspolicy