

# Direct-to-Consumer Genetic Testing: Providing Personalized Medicine

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## Disclosures

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- I received gratis genetic testing for personal use from Color Genomics prior to being asked to give this talk.

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## A Note

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- Specific products and laboratories will be discussed in this talk. This does not imply endorsement by myself, my employer or the providers of the activity.

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## Presentation Outline

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- Introduction on DTC genetic tests
- Interpreting DTC genetic tests
- Available resources and next steps for patients
- The future of DTC genetic test
- Q&A

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# Presentation Outline

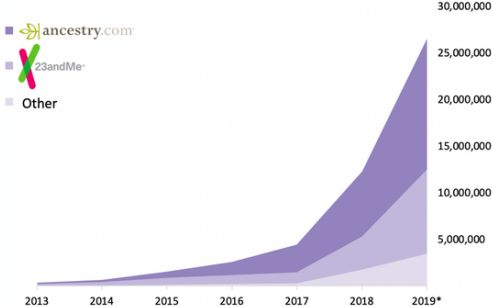
- Introduction on DTC genetic tests
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# Direct to Consumer Testing

About 30 million people are estimated to have had some type of DTC testing

Within a 4-day timespan (“Black Friday” through “Cyber Monday,” November 2017), Ancestry.com sold an estimated 1.5 million DNA kits

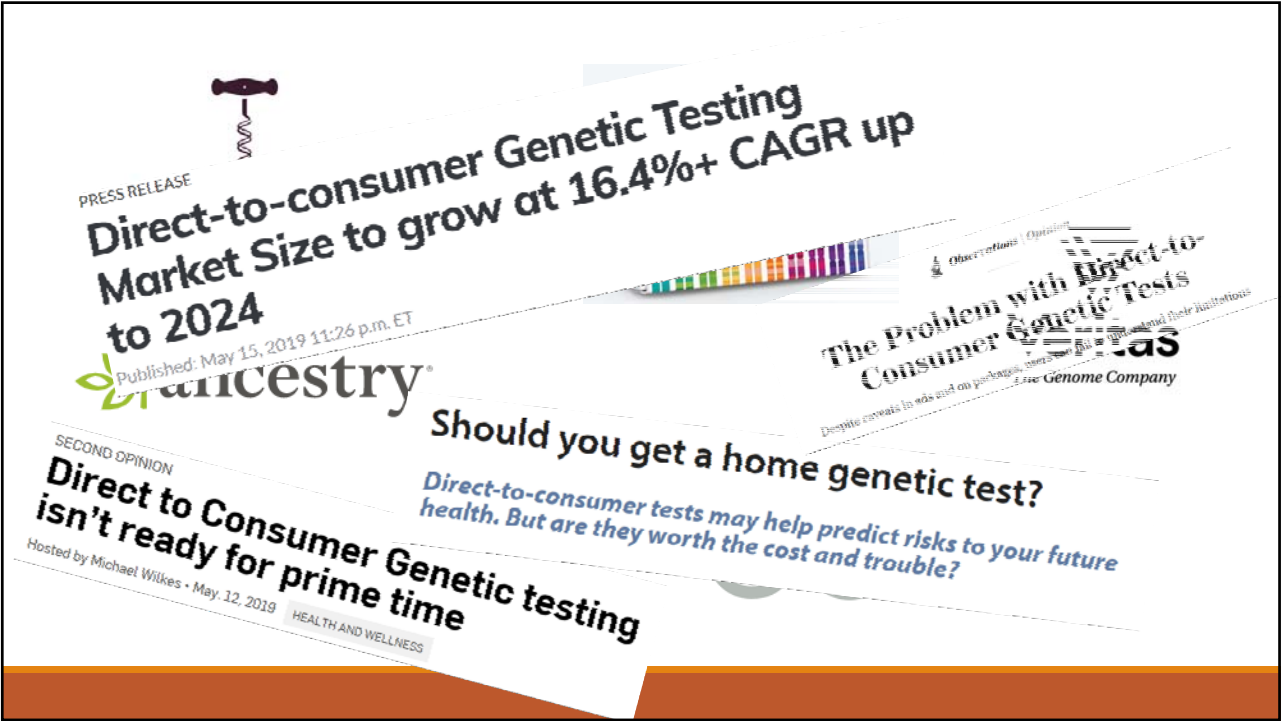
**Global Consumer Appetite For Genetic Testing Has Ballooned Over The Past 6 Years**  
Total number of consumers tested by genetic testing companies globally



\*As of January 1, 2019  
Source: MIT Technology Review, February 11, 2019  
Methodology: MIT Technology Review's estimates are based on its own reporting, data aggregated by the International Society of Genetic Genealogy, and public statements by the four largest genetic testing companies. Because the genetic testing companies release their information intermittently, MIT Technology Review used the disclosures closest to January 1 for 2012-2018. To create a figure for 2019, MIT Technology Review used data reported by Ancestry on November 29, 2018.



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## Two Major Categories of Testing

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- Direct to Consumer Genetic Testing (DTC-GT)
  - Genetic tests ordered by an individual without the involvement of a health care professional
- Consumer-Initiated Genetic Testing (CI-GT)
  - Genetic tests ordered by an individual but require a health care professional to sign off

Weissman, Scott. "DTC Genetic Testing 201."  
<https://www.nsgc.org/p/bl/et/blogid=59&blogaid=1057>

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## Direct to Consumer Genetic Testing

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- Type of tests: Ancestry, genetic traits, some disease risk, entertainment
- Methodology: Generally uses genotyping of predefined single nucleotide variants
- Many give back raw data which can be interpreted through 3<sup>rd</sup> party websites
- Lab Examples:
  - 23andMe, Ancestry.com, FamilyTreeDNA, Genos, MyHeritage, Helix (some tests)

Weissman, Scott. "DTC Genetic Testing 201."  
<https://www.nsgc.org/p/bl/et/blogid=59&blogaid=1057>

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## Consumer-Initiated Genetic Testing

- Type of tests: genes known to cause a hereditary risk of disease, which can include cancer, cardiac and carrier testing; pharmacogenetic testing
  - Methodology: Uses “full” gene sequencing
  - Raw data is not generally available
  - Lab Examples:
    - Color, Invitae, OneOme, Veritas (closed), Helix (some tests), JScreen
- Weissman, Scott. “DTC Genetic Testing 201.”  
<https://www.nsgc.org/p/bl/et/blogid=59&blogaid=1057>

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## Benefits and Limitations of DTC-GT

### BENEFITS

- Allows consumers access to their genetic information
- Possibly encourage consumers to change their behaviors
- Research opportunities through partnerships between the lab and other companies

### LIMITATIONS

- Testing type/methodology is not the same as clinical testing
- Concerns about consumer misunderstanding of the utility/limitations of the testing
- Concerns about privacy protections
- Potential to find unexpected information
- Results require confirmation in a clinical lab prior to using for clinical management

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# Benefits and Limitations of CI-GT

## BENEFITS

- Medical-grade test
  - Performed in a CLIA-certified lab
- Improved access compared to traditional testing
- Involvement of HCP who is familiar with test, often includes post-test genetic counseling

## LIMITATIONS

- May only report pathogenic or likely pathogenic variants
  - Variants of uncertain significance may not be reported
- May have limited methodology
  - Gene sequencing only
- May not be the “complete” test
  - Only selected genes/variants may be included

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# Step One (The Most Important Step)

- Determine what testing has been performed:
  - Wellness/trait association
  - FDA-approved testing from a DTC company
  - 3<sup>rd</sup> party analysis of raw data from DTC testing
  - Consumer-initiated testing

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# Wellness/Trait Markers

“Info-tainment” : Limited Clinical Utility

Helix Wellness

**Does my DNA really impact my weight and wellbeing?**

You might be surprised how much you can learn about your day-to-day from your DNA. Helix Wellness can get you started with eight fascinating and personal genetic traits.

Traits included:

- **Body mass index (BMI) introduction**  
It's no secret that genetics plays a role in weight, since two people who eat the same foods can have very different responses. You'll learn about just one of the many genes that influences BMI, the FTO gene, and how it influences your weight.

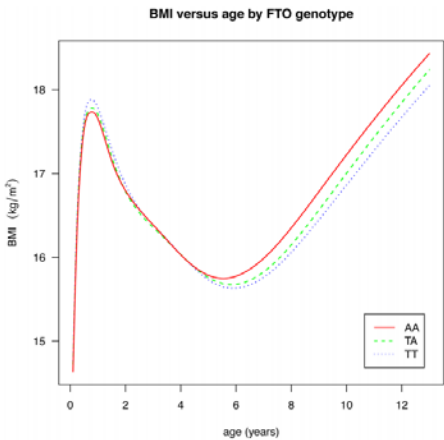


Figure 2. Curves of median BMI by age and genotype at rs9939609, estimated by the LMS method and adjusted for study and sex. doi:10.1371/journal.pgen.1001207.g002

PMID 21379325

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## Wellness/Trait Markers

- Main benefit: Changing consumer behavior
  - 23% had a positive lifestyle change (specifically improved dietary and exercise practices, quit smoking)
  - 7% had subsequent preventive checks

PMID 28664264

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## Clinical testing from DTC Companies

### 23ANDME

(FDA-APPROVED, CLIA-CERTIFIED)

#### Health Predisposition Reports :

- Age-related macular degeneration (2 variants); Alpha-1 Antitrypsin deficiency (2 variants); BRCA1/2 (3 variants); Celiac disease (2 variants); Familial hypercholesterolemia (24 variants); G6PD Deficiency (1 variant); Hereditary amyloidosis (3 variants); Hereditary hemochromatosis (2 variants); Hereditary thrombophilia (2 variants); Late-onset Alzheimer’s disease (1 variant); MUTYH-Associated polyposis (2 variants); Parkinson’s disease (2 variants)

Carrier Status Reports\*: 40+ disorders

<https://www.23andme.com/dna-reports-list/>

### ANCESTRY.COM

(CLIA-CERTIFIED)

#### Cancer Risk:

- BRCA1/2: 27 variants
- Lynch syndrome: 12 variants in 4 genes

#### Carrier status:

- Selected variants for cystic fibrosis, sickle cell anemia, Tay-Sachs disease

#### Heart and Blood Health

- MYBPC3 and MYH7- associated cardiomyopathy (9 variants)
- Familial hypercholesterolemia (9 variants)
- Hereditary Hemochromatosis (2 variants)
- Hereditary Thrombophilia (2 variants)

<https://www.ancestry.com/health/variants>

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# DTC Raw Data Analysis

- Many DTC companies, such as Ancestry.com and 23andMe give users the option to download their raw data
- Raw data can then be analyzed through a 3<sup>rd</sup> party site, such as:
  - Promethease
  - GenomeGenie
  - LiveWello

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# Promethease

- “Literature retrieval system that builds a personal DNA report based on connecting a file of DNA genotypes to the scientific findings cited in SNPedia”



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# Promethease

- “Literature retrieval system that builds a personal DNA report based on connecting a file of DNA genotypes to the scientific findings cited in SNPedia”

rs3738579(T,T)

1.5x - 2x increased risk for cervical cancer, HNSCC, and breast cancer

rs3738579 represents a SNP in the 5' UTR region upstream of the RNASEL gene. A study of patients diagnosed with carcinoma of the uterine cervix, head and neck squamous cell carcinomas (HNSCC), and breast cancer found 1.5x-2x increased risk for all three cancer types for the rs3738579(T,T) genotype, while finding decreased risk (0.5x) for rs3738579(C,T) heterozygotes. rs3738579(C,C) homozygotes had 0.6x less risk for cervical cancer but increased risk for HNSCC (1.4x) and breast cancer (1.8x). Although statistics were not reported per genotype, a combination of data from all three cancer forms over all genotypes provided strong statistical evidence for rs3738579 as a cancer marker, with a p-value of 4.43x10<sup>-5</sup>.  
[View more info](#)

Bad

Report

36.1%

2013-11-07

0.2392

1

182566801

3

2018-12-05

### rs3738579

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A study of patients diagnosed with carcinoma of the uterine cervix, head and neck squamous cell carcinomas (HNSCC), and breast cancer found 1.5x-2x increased risk for all three cancer types for the rs3738579(T,T) genotype, while finding decreased risk (0.5x) for rs3738579(C,T) heterozygotes. rs3738579(C,C) homozygotes had 0.6x less risk for cervical cancer but increased risk for HNSCC (1.4x) and breast cancer (1.8x). [PMID 18575592]

Although statistics were not reported per genotype, a combination of data from all three cancer forms over all genotypes provided strong statistical evidence for rs3738579 as a cancer marker, with a p-value of 4.43x10<sup>-5</sup>. [PMID 18575592]

Note that the research cited above was published over a decade ago, and there has been no follow-up or replication to our knowledge. It would be best to consider the research preliminary and perhaps of little to no clinical significance (until it is confirmed or replicated in a larger sample).

Categories:

Orientation minus

Stabilized minus

Geno	Mag	Summary
(C,C)	1.1	0.6x decreased risk for cervical cancer, but 1.4x increased risk for HNSCC and 1.8x increased risk for breast cancer (reported in 2008)
(C,T)	1.1	0.5x decreased risk for cervical cancer, HNSCC, and breast cancer (reported in 2008)
(T,T)	1.1	1.5x - 2x increased risk for cervical cancer, HNSCC, and breast cancer (reported in 2008)

Reference GRCh38 38.1/141

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ORIGINAL RESEARCH ARTICLE

Genetics  
inMedicine

Open

False-positive results released by direct-to-consumer genetic tests highlight the importance of clinical confirmation testing for appropriate patient care

Stephany Tandy-Connor, MS, Jenna Guiltinan, MS, Kate Krempely, MS, Holly LaDuca, MS, Patrick Reineke, BS, Stephanie Gutierrez, BS, Phillip Gray, PhD and Brigette Tippin Davis, PhD, FACMG

Confirmed alterations 60%

False positives 40%

COL3A1 (n=1)

CHEK2 (n=1)

TP53 (n=1)

BRCA1 (n=3)

BRCA2 (n=4)

MLH1 (n=2)

ATM (n=1)

Figure 1 False-positive variants in clinically actionable genes. The pie chart on the left indicates of the variants analyzed, 60% were confirmed and 40% were false positives. The pie chart on the right shows which genes were involved with the false-positive cases and how often those false calls were detected in this study.

PMID 29565420

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Table 3 Classification discrepancies

Gene	Variant	DTC/third party <sup>a</sup>	Ambry <sup>b</sup>	ClinVar <sup>c</sup>	ESP <sup>d</sup>	1000 Genomes <sup>e</sup>	dbSNP <sup>f</sup>
ATM	p.M1040V (c.3118A>G)	Increased risk	Benign	Benign	1.36%	0.95%	1.48%
BRCA1	p.Q356R (c.1067A>G)	Increased risk	Benign	Benign	4.59%	2.81%	3.97%
BRCA2	p.N372H (c.1114A>C)	Increased risk	Benign	Benign	23.32%	24.26%	24.44%
COL3A1	p.A698T (c.2092G>A)	Increased risk	Benign	Benign	21.39%	21.16%	19.16%
COL5A1	c.655-8689C>T	Increased risk	Deep intronic—benign	N/A	N/A	N/A	N/A
COL5A1	c.654+2749A>G	Increased risk	Deep intronic—benign	N/A	N/A	N/A	N/A
COL5A1	c.1827+399C>T	Increased risk	Deep intronic—VUS	N/A	N/A	N/A	N/A
COL5A1	c.1827+1142T>C	Increased risk	Deep intronic—benign	N/A	N/A	N/A	N/A

DTC, direct to consumer; N/A, not available; VUS, variant of unknown significance.  
aVariant classification provided by the DTC company or a third-party interpretation service. bVariant classification provided by Ambry. cVariant classification provided in ClinVar (clinical laboratory submissions only). dExome Sequencing Project population frequency database. e1000 Genomes population frequency database. fdbSNP population frequency database.

PMID 29565420

Consumer Initiated Genetic Testing

- Hereditary Cancer Syndromes:
  - Hereditary breast and ovarian cancer, Lynch syndrome, prostate cancer, thyroid cancer, etc.
- Cardiovascular Disease:
  - Aortopathies, arrhythmias, cardiomyopathies, familial hypercholesterolemia
- Carrier testing:
  - Testing to determine the chances of having a child with a condition due to the parent being a carrier for the disorder
- Pharmacogenomic testing
- Newborn genetic screening
- Whole exome or whole genome sequencing:
  - Analysis of the coding portions of genes (exome) or entirety of genome for sequence variants

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## Web-Based Resources for Consumers

- Genetics Home Reference:

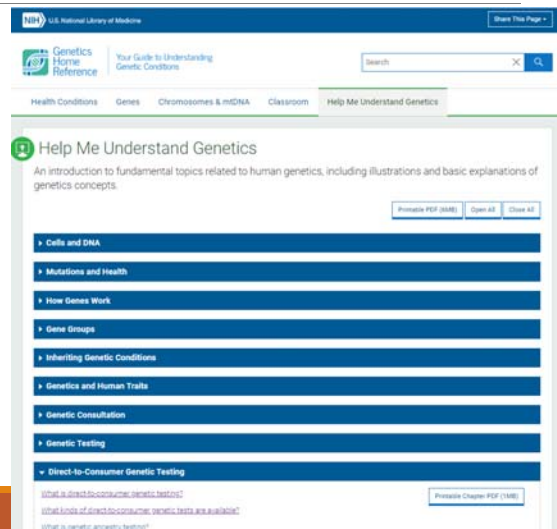
<https://ghr.nlm.nih.gov/primer>

- National Human Genome Research Institute:

<https://www.genome.gov/dna-day/15-ways/direct-to-consumer-genomic-testing>

- Questions about Genetic Discrimination:

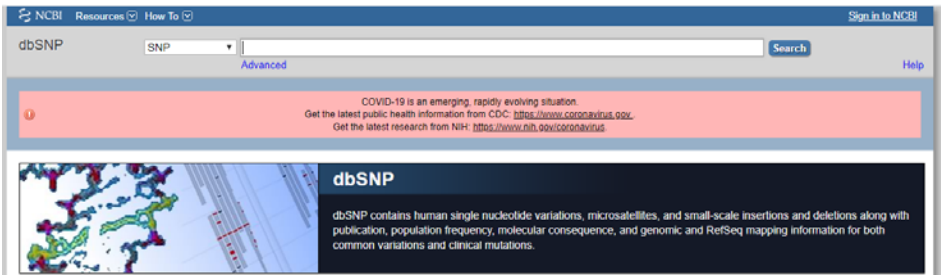
<http://ginahelp.org/>



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# Variant Interpretation

- dbSNP: <https://www.ncbi.nlm.nih.gov/snp/>
- Reference SNP “rs” numbers



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dbSNP

Short Genetic Variations

Search for terms  
Examples: rs268, BRCA1 and...

Welcome to the Reference SNP (rs) Report

All alleles are reported in the [Forward orientation](#). Click on the [Variant Details tab](#) for details on Genomic Placement, Gene, and Amino Acid. HGVS names are in the [Aliases tab](#).

Reference SNP (rs) Report

[Switch to classic site](#)

[Download](#)

rs2696245

Organism

Homo sapiens

Position

chr17:50187356 (GRCh38.p12)

Alleles

C>T

Variation Type

SNV Single Nucleotide Variation

Frequency

C=0.030350 (3811/125568, TOPMED)  
C=0.03201 (1004/31366, GnomAD)  
C=0.0242 (121/5008, 1000G) [\(-6 less\)](#)  
C=0.0192 (86/4480, Estonian)  
C=0.0158 (61/3854, ALSPAC)  
C=0.0135 (50/3708, TWINSUK)  
C=0.007 (4/600, NorthernSweden)  
C=0.000 (0/214, Vietnamese)  
C=0.02017 (482/23900, ALFA Project)

Clinical Significance

Not Reported in ClinVar

Gene : Consequence

COL1A1 : Intron Variant

Publications

0 citations

Genomic View

[See rs on genome](#)

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# Variant Interpretation

- ClinVar: <https://www.ncbi.nlm.nih.gov/clinvar/>
  - Aggregate of information about genomic variation

ClinVar

Genomic variation as it relates to human health

Search ClinVar

Advanced search

About

Access

Submit

Stats

FTP

Help

Was this helpful?

Follow

Print

Download

NM\_000179.2(MSH6):c.187T>C (p.Ser63Pro)

Cite this record

Interpretation:

Conflicting interpretations of pathogenicity  
Likely benign(1);Uncertain significance(7)

Review status:

★☆☆ criteria provided, conflicting interpretations

Submissions:

8 (Most recent: Mar 6, 2020)

Last evaluated:

Dec 30, 2019

Accession:

VCV000220796.8

Variation ID:

220796

Description:

single nucleotide variant

Variant details

Conditions

Allele ID:

221252

Gene(s)

Variant type:

single nucleotide variant

Variant length:

1 bp

Submitted interpretations and evidence

Interpretation (Last evaluated)	Review status (Assertion criteria)	Condition (Inheritance)	Submitter	Supporting information (See all)
Uncertain significance (Apr 18, 2018)	criteria provided, single submitter (Counsyl Autosomal Dominant Disease Classification criteria (2015)) Method: clinical testing	Hereditary nonpolyposis colorectal cancer type 5 Allele origin: unknown	Counsyl Accession: SCV000789355.2 Submitted: (Jun 20, 2018)	Evidence details
Uncertain significance (Oct 31, 2018)	criteria provided, single submitter (ACMG Guidelines, 2015) Method: clinical testing	Turcot syndrome Endometrial carcinoma Hereditary	Fulgent Genetics,Fulgent Genetics Accession: .....	Evidence details Publications Published (1) DOI: 10.1038/gim.2015.30

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# Management Resources for Providers

- American College of Medical Genetics and Genomics
- American College of Cardiology
- American College of Obstetrics and Gynecology
  - Follow-up information for carrier testing
- National Comprehensive Cancer Network: [www.NCCN.org](http://www.NCCN.org)
  - Recommendations for screening for hereditary cancer syndromes
- Pharmacogenomics:
  - Clinical Pharmacogenomics Implementation Consortium (CPIC)
  - PharmGKB

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# National Comprehensive Cancer Network

NCCN

National Comprehensive Cancer Network®

NCCN Guidelines®

NCCN Compendia

NCCN Templates®

Educational Events & Programs

Subscriptions & Products

Clinical & Business Resources

NCCN Global

NCCN Guidelines®

NCCN Guidelines® & Clinical Resources

NCCN Guidelines®

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NCCN Guidelines for Treatment of Cancer by Site

NCCN Guidelines for Detection, Prevention, & Risk Reduction

NCCN Guidelines for Supportive Care

NCCN Guidelines for Specific Populations

NCCN Guidelines for Patients

NCCN GUIDELINES FOR DETECTION, PREVENTION, & RISK REDUCTION

Breast Cancer Risk Reduction ▶

Breast Cancer Screening and Diagnosis ▶

Cervical Cancer Screening ▶

Colorectal Cancer Screening ▶

Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic ▶


Genetic/Familial High-Risk Assessment: Colorectal ▶

Lung Cancer Screening ▶

Prostate Cancer Early Detection ▶

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CPIC: Clinical Pharmacogenetics  
Implementation Consortium



Genes-Drugs

CPIC assigns CPIC levels to genes/drugs with (1) [PharmGKB Clinical Annotation Levels of Evidence](#) of 1A, 1B, 2A and 2B, or (2) a [PharmGKB PDx level](#) for FDA-approved drug labels of "actionable pgx", "genetic testing recommended", or "genetic testing required", or (3) based on nomination to CPIC for consideration.

The levels (A, B, C, and D) assigned are subject to change; only those gene/drug pairs that have been the subject of guidelines have had sufficient in-depth review of evidence to provide definitive CPIC level assignments.

Note that only CPIC level A and B gene/drug pairs have sufficient evidence for at least one prescribing action to be [recommended](#). CPIC level C and D gene/drug pairs are not considered to have adequate evidence or actionability to have prescribing recommendations.

- [View CPIC's process for assigning CPIC levels](#)
- [View CPIC's levels for gene/drugs](#)
- [View CPIC's process for prioritizing CPIC guidelines](#)

CPIC invites [feedback](#) on existing and planned gene/drug guidelines.

Search:

Download this table (CSV) - Last modified: May 7, 2020

# (in sort)	GENE (UNIQUE = 127)	DRUG (UNIQUE = 340)	GUIDELINE	CPIC LEVEL	PHARMGKB LEVEL OF EVIDENCE	PKS ON FDA LABEL	CPIC PUBLICATIONS (PMID)
1	HLA-B	abacavir	<a href="#">Guideline</a>	A	1A	Testing required	<ul style="list-style-type: none"><li>24541393</li><li>22278737</li></ul>
2	HLA-B	abacavir	<a href="#">Guideline</a>	A	1A		<ul style="list-style-type: none"><li>22272949</li><li>25526338</li></ul>
3	CYP2C19	esomeprazole	<a href="#">Guideline</a>	A	1A		<ul style="list-style-type: none"><li>23498477</li><li>22830260</li></ul>

CPIC® Guideline for Clopidogrel and CYP2C19

Most recent guideline publication:

[Clinical Pharmacogenetics Implementation Consortium Guidelines for CYP2C19 Genotype and Clopidogrel Therapy, 2013 update \(September 2013\) \[2\]](#)

Updates since publication:

March 2019: The FDA-approved label for clopidogrel (Plavix) was recently updated (September 2016) and warns that patients who are CYP2C19 poor metabolizers may have diminished effectiveness of the drug as compared to patients with normal CYP2C19 function. The drug label suggests that a different platelet P2Y12 inhibitor be used in patients identified as CYP2C19 poor metabolizers. The FDA label change does not alter the recommendation from the authors that based on available evidence, the CPIC guideline is most applicable to ACS/PCI patients.

Tables and figure in the main manuscript of the guideline:

Table 1. Assignment of likely CYP2C19 phenotypes based on genotypes

Table 2. Antiplatelet therapy recommendations based on CYP2C19 status when considering clopidogrel for ACS/PCI patients

Figure 1. Algorithm for suggested clinical actions based on CYP2C19 genotype when considering treatment with clopidogrel for ACS patients undergoing PCI

Supplement to: Clinical Pharmacogenetics Implementation Consortium Guidelines for CYP2C19 Genotype and Clopidogrel Therapy, 2013 update (September 2013) [2]

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## Available Clinical Genetics Resources

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- Laboratory Resources
  - Many labs performing testing offer genetic counseling as a service to users
  - 23andMeBlog: provides information on a variety of common topics
- National Society of Genetic Counseling ([www.NSGC.org](http://www.NSGC.org))
  - Traditional in-person genetic counseling
  - Telehealth genetic counseling
- American College of Medical Genetics and Genomics ([www.ACMG.net](http://www.ACMG.net))
- Personalized Genomic Medicine Clinics

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## Reasons for Referral to Clinical Genetics

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- Provide disease-specific evaluation, counseling and recommendations
- Coordination of cascade testing for family members or testing for reproductive partners
- Tracking variant re-classification

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## Case Example

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- 25 yo female undergoes DTC genetic testing due to multiple medical concerns
- DTC testing identifies a heterozygous pathogenic variant in the *GBA* gene associated with Gaucher disease
- Patient receives raw data and uses a 3<sup>rd</sup> party website to analyze her test results
  - Identifies C677T and A1298C variants in the *MTHFR* gene
- Presents to her PCP with these results who refers her for a clinical genetics evaluation

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## Case Example (continued)

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- Review of results with the geneticist and genetic counselor
  - *MTHFR* variants: not likely clinically relevant
    - These variants are present in up to 50% of individuals in some ethnic groups and have been linked to a variety of medical concerns. Studies are inconclusive or conflicting about the role of *MTHFR* in these disorders.
  - *GBA* variant: carrier for Gaucher disease
    - However, only 3 variants were included on the test.
- In the evaluation: history of anemia is shared, and splenomegaly is appreciated on physical exam
- Genetic testing is ordered for full analysis of *GBA*, which identifies a second variant, thus confirming a diagnosis of Gaucher disease in the patient
- Treatment with enzyme replacement therapy is initiated

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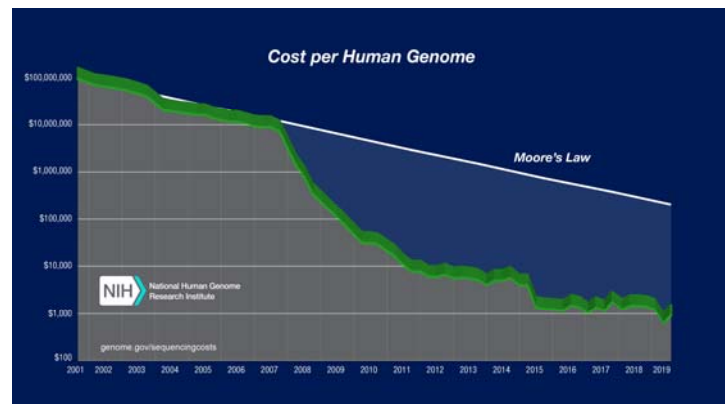
## Increased Access to Genetic Testing

### Decreasing Cost

- 2007: \$1000
- 2010: \$300-400
- Now: \$99

### Broadening test menu

### Insurance coverage for tests



<https://www.genome.gov/about-genomics/fact-sheets/DNA-Sequencing-Costs-Data>

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# New Service Models

- Telehealth Services
  - COVID-19 pandemic has increased the availability of genetics telehealth services
  - Currently limited, though, as genetic counselors are not CMS-recognized providers
- New workflows to ensure that access isn't a barrier to care

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# Primary Care

Role of PCPs in ordering genetic testing

Increased expectations that PCPs will understand and utilize test results in clinical care

**Annals of Internal Medicine®**  
LATEST   ISSUES   CHANNELS   CME/MOC   IN THE CLINIC   JOURNAL CLUB   WEB EXCLUSIVES   AUTHOR INFO

THIS ISSUE   NEXT ARTICLE  
**ORIGINAL RESEARCH** | 19 APRIL 2016  
**Consumer Perceptions of Interactions With Primary Care Providers After Direct-to-Consumer Personal Genomic Testing**  
Cathelijne H. van der Wouden, BSc; Deanna Alexis Garere, ScD, CGC; Anke H. Maitland-van der Zee, PharmD, PhD; Mack T. Ruffin IV, MD, MPH; J. Scott Roberts, PhD; Robert C. Green, MD, MPH; for the Impact of Personal Genomics Study Group† \*  
[Article, Author, and Disclosure Information](#)

1026 participants

63% planned to share results with PCP

At 6 months, only 27% reported having done so

Among participants who discussed the results, 35% were very satisfied with the encounter and 18% were not satisfied at all

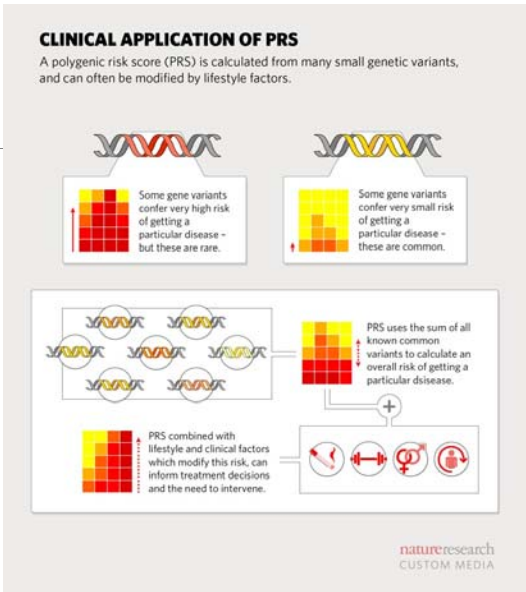
PMID 26928821

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# New Testing Options

- Polygenic Risk Scores
- Summation of many common variants to derive an overall risk of developing a particular disease
  - Currently available:
    - 23andMe (diabetes)
    - Color (CAD)
    - Helix (prostate cancer)

Big question: how to use these results in clinical care?



<https://www.nature.com/articles/d42473-019-00270-w>

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However, has the public’s interest already started to decrease?



<https://www.businessinsider.com/veritas-genetics-shutters-us-operations-2019-12>  
<https://www.cnbc.com/2019/08/25/dna-tests-from-companies-like-23andme-ancestry-see-sales-slowdown.html>

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## Take Home Points

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- Not all “direct-to-consumer” genetic testing is created equal
- Important to determine what type of testing has been performed prior to taking action
  - Consider whether the results need to be confirmed in a clinical lab
- Providers have many resources available to help patients interpret their results
- When in doubt, clinical genetics experts are available to help

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## Summary of Resources

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- For Consumers:
  - Genetics Home Reference: <https://ghr.nlm.nih.gov/primer>
  - National Human Genome Research Institute: <https://www.genome.gov/dna-day/15-ways/direct-to-consumer-genomic-testing>
  - Questions about Genetic Discrimination: <http://ginahelp.org/>
- Variant Interpretation:
  - dbSNP: <https://www.ncbi.nlm.nih.gov/snp/>
  - ClinVar: <https://www.ncbi.nlm.nih.gov/clinvar/>
- Clinical:
  - National Society of Genetic Counselors
  - American College of Medical Genetics and Genomics
  - American College of Cardiology
  - American College of Obstetrics and Gynecology
  - National Comprehensive Cancer Network: [www.NCCN.org](http://www.NCCN.org)
  - Clinical Pharmacogenomics Implementation Consortium (CPIC): <https://cpicpgx.org/>
  - PharmGKB: <https://www.pharmgkb.org/>
- Miscellaneous:
  - 23andMe Blog: <https://blog.23andme.com/>

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## Presentation Outline

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- Introduction on DTC genetic tests
- Interpreting DTC genetic tests
- Available resources and next steps for patients
- The future of DTC genetic test
- Q&A