

# Direct-to-Consumer Genetic Testing

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# Conflicts of Interest

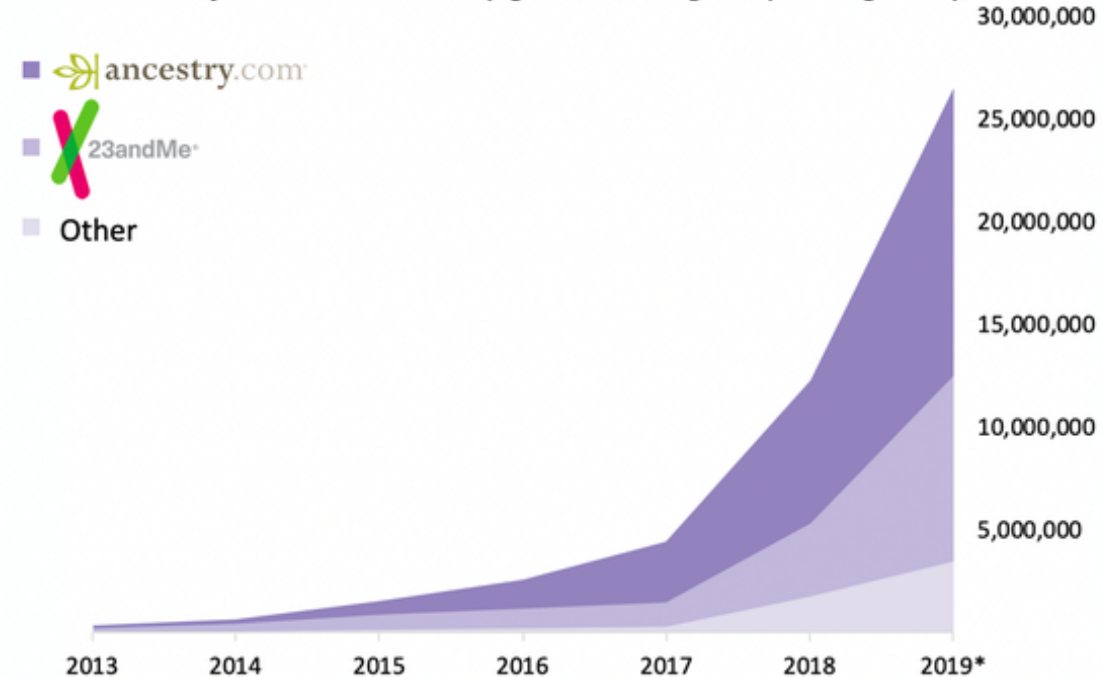
- I have no conflicts of interest to declare.
- The views expressed in this presentation are those of the author and do not reflect the official policy of the Department of Army/Navy/Air Force, Department of Defense, the U.S. Government, or the Uniformed Services University.

# Direct-to Consumer Genetic Testing

GENETIC TESTS THAT ARE MARKETED DIRECTLY TO CONSUMERS WITHOUT THE INVOLVEMENT OF HEALTH CARE PROVIDERS

## 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.

BUSINESS  
INSIDER  
INTELLIGENCE

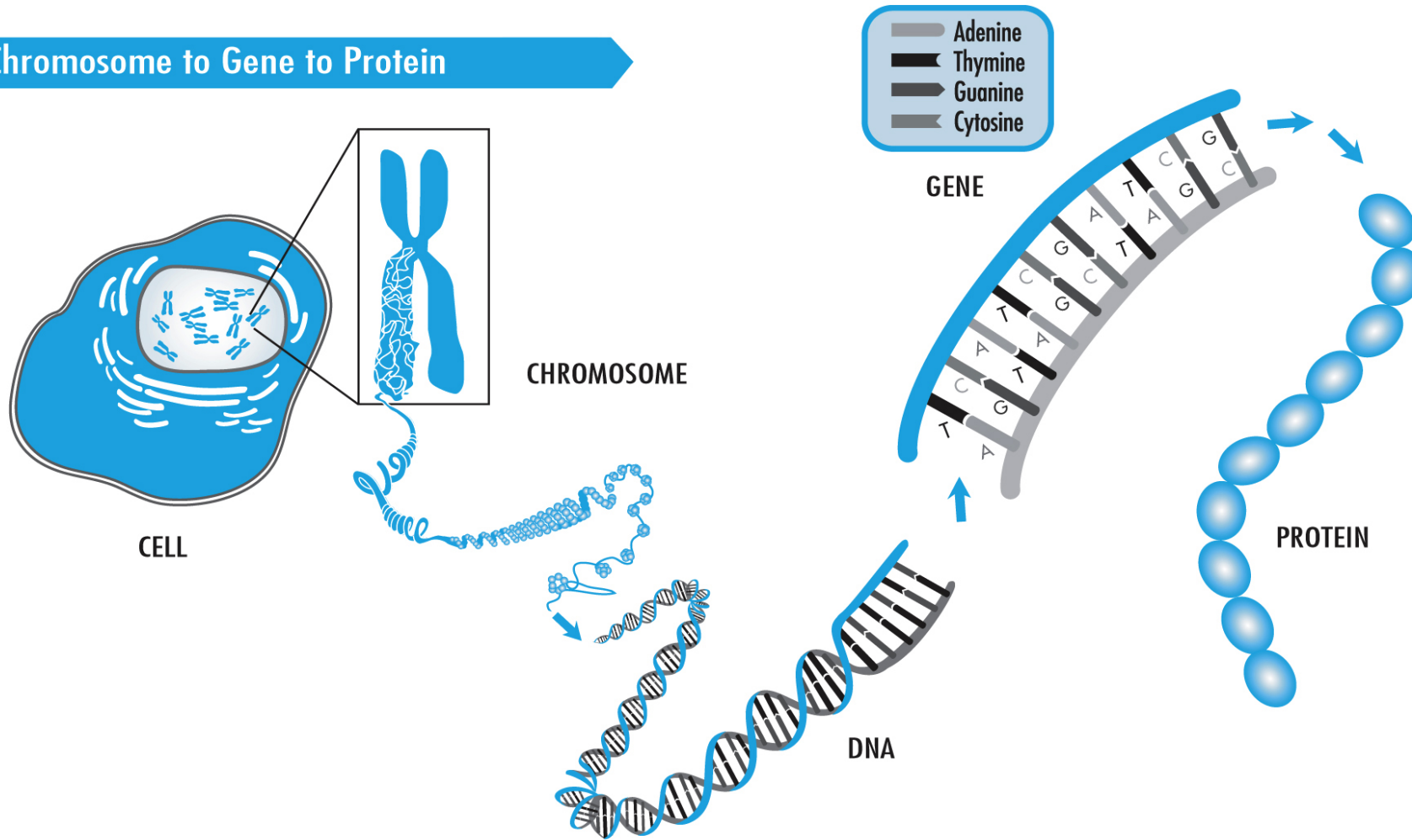
# Types of Results from DTC Testing

- Ancestry or Genealogy
- Kinship
- Lifestyle/Recreational
  - General wellness, athletic ability
- Disease Risk and Health
  - Cancer Predisposition, Carrier Status, Pharmacogenomics

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# Chromosome to Gene to Protein



# Genome Analogy



Genome

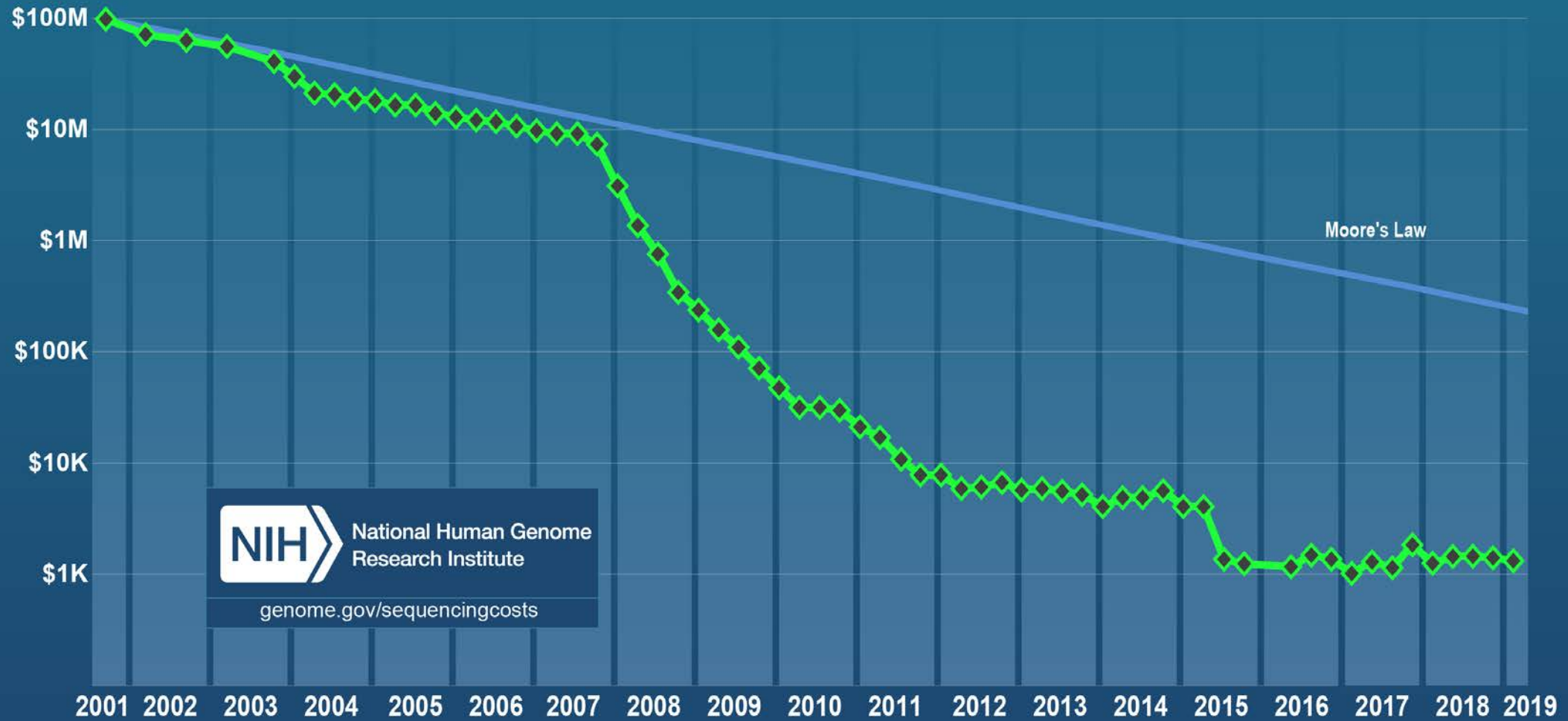


Chromosome



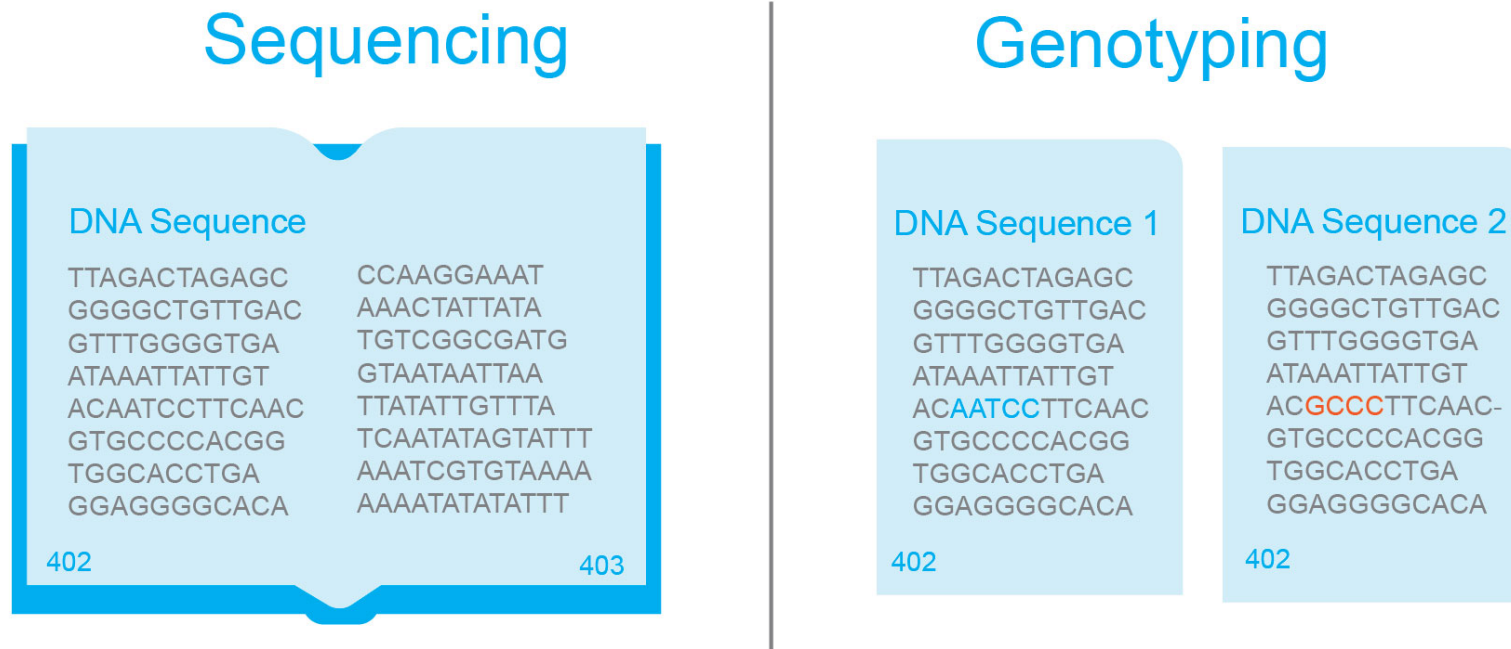
Gene

# Cost per Genome





# Sequencing vs. Genotyping



**Figure 1.** Illustrates the analogy of a book being used to compare sequencing to genotyping. Sequencing (left) is like reading out all of the letters in a book. Genotyping (right) is like looking at a specific section of a specific page, and comparing to others of the same page.

# Genotyping Example



**Figure 5.** Conceptual illustration of SNP rs1815739, related to muscle performance. The T allele, (top) codes a stop codon (amino acid code X), while the C allele (bottom) codes an arginine (amino acid code R). One publication found that within a small number of female Olympic athletes, sprinters were associated with the R allele (rs1815739(C)), while none of the female sprinters were XX homozygote.

# Benefits of DTC Testing

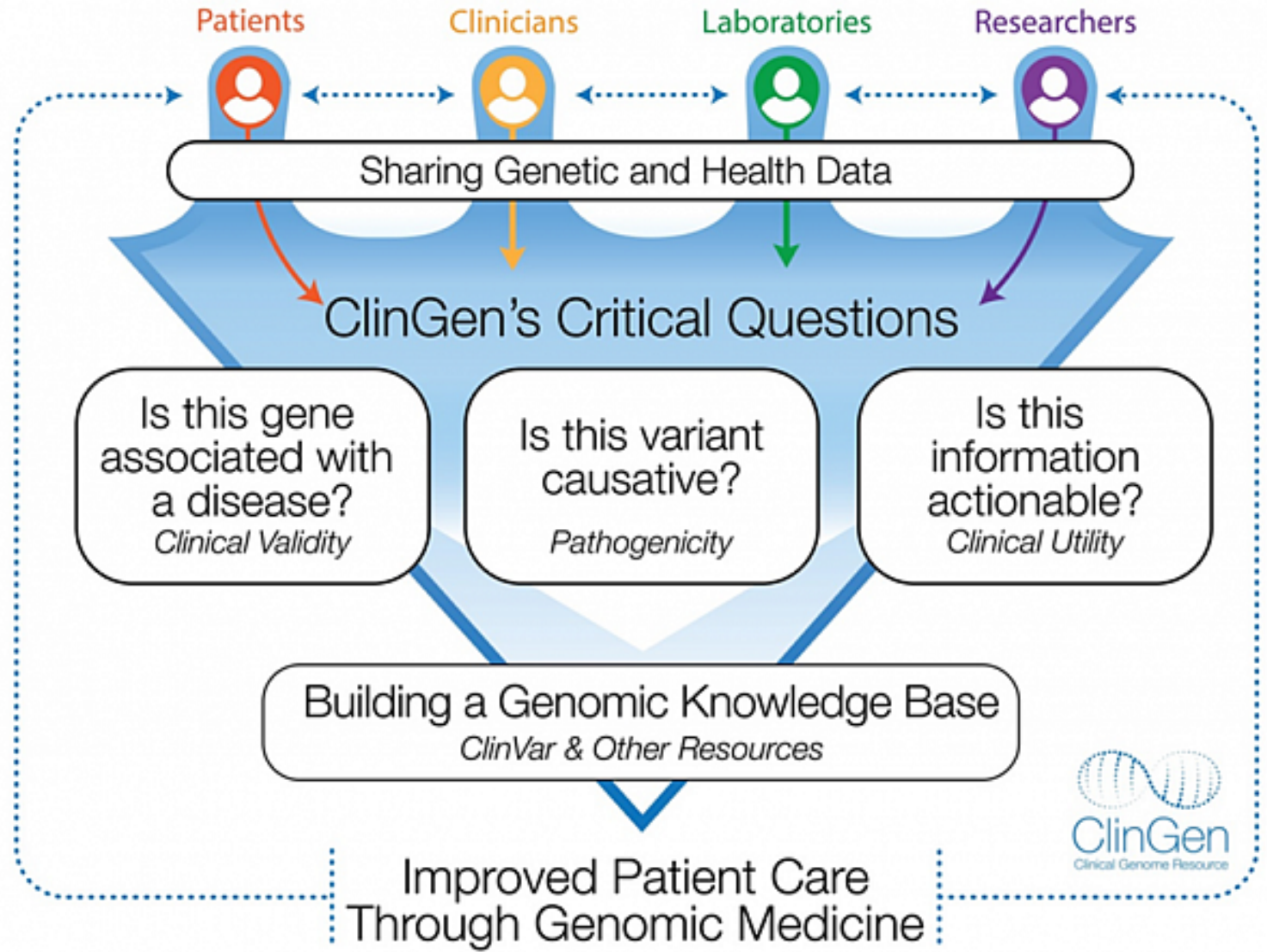
- Awareness of genetic diseases.
- Personalized information.
- Often less expensive than clinical genetic testing.
- Collection is usually simple and noninvasive, and results are available quickly.
- Your data may aid further medical research.

# Cons

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- Scientific evidence may be limited.
- May not be applicable to health conditions of interest.
- Does not definitively diagnose or rule-out disease.
- Results may be inaccurate, incomplete, or misunderstood.
- May reveal unexpected information
  - Health, family relationships, or ancestry.
- Little oversight or regulation.
- Genetic privacy may be compromised.
- May impact ability to obtain life, disability, or long-term care insurance.

# Scientific Evidence



# Applicability

Four tests have FDA approved marketing authorization for associated health risk

- All through one company, 23andMe
  - Carrier testing for Bloom Syndrome
  - Carrier testing for genetic health risk
    - Multiple variants for ~10 conditions
  - Pharmacogenetics report
  - Selected variants in *BRCA1/BRCA2*
    - 3 out of ~3700 pathogenic variants

# Accuracy- Case Example

Patient brought in DTC results that indicated 3 disease-causing changes

1. Increased risk of breast and ovarian cancer
  2. Increased risk of colorectal cancer
  3. Increased risk of early-onset Alzheimer's disease
- Sample sent to clinical laboratory to confirm results
  - 0/3 of these findings were clinically confirmed- all false positives

# Unexpected Information

## With genetic testing, I gave my parents the gift of divorce

By George Doe | Sep 9, 2014, 7:50am EDT

f   SHARE





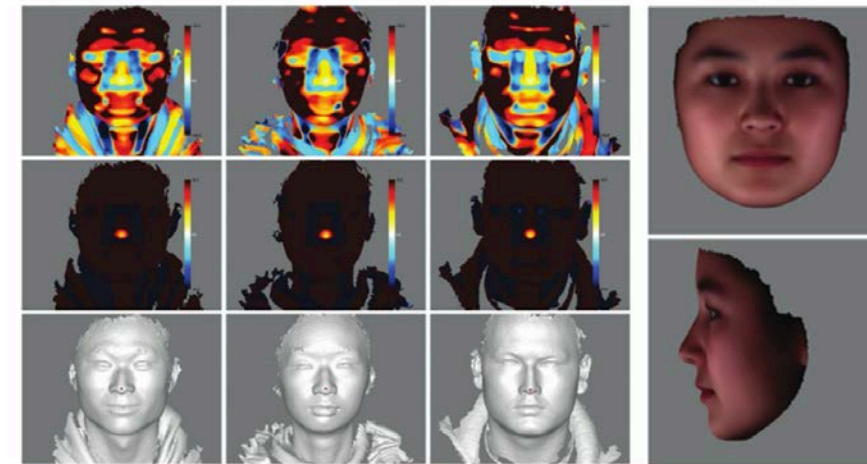
# Military Specific Concerns

- Individual
  - Impacts on Career progression
  - Accession/Retention
- Military Health System
  - Accession/Retention
  - Healthcare utilization
- National Security
  - Use of information by hostile actors
  - Release of unexpected information

The New York Times

## *China Uses DNA to Map Faces, With Help From the West*

Beijing's pursuit of control over a Muslim ethnic group pushes the rules of science and raises questions about consent.



Images from a study in 2013 on 3-D human facial images. BMC Bioinformatics

# National Security Concerns

## Additional concerns for the service member



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DEC 20 2019

MEMORANDUM FOR: SEE DISTRIBUTION

SUBJECT: Direct-to-Consumer Genetic Testing Advisory for Military Members

It has come to the attention of the DoD that some direct-to-consumer (DTC) genetic testing companies are encouraging DoD personnel to purchase genetic ancestry and health information through the offering of military discounts or other incentives. These DTC genetic tests are largely unregulated and could expose personal and genetic information, and potentially create unintended security consequences and increased risk to the joint force and mission.

Exposing sensitive genetic information to outside parties poses personal and operational risks to Service members. DTC genetic tests that provide health information have varying levels of validity, and many are not reviewed by the Food and Drug Administration before they are offered, meaning they may be sold without independent analysis to verify the claims of the seller. Possible inaccuracies pose more risk to DoD military personnel than the public due to Service member requirements to disclose medical information that affects readiness (see DoD Instruction 6025.19, "Individual Medical Readiness"). Testing outside the Military Health System is unlikely to include a clear description of this risk.

Moreover, there is increased concern in the scientific community that outside parties are exploiting the use of genetic data for questionable purposes, including mass surveillance and the ability to track individuals without their authorization or awareness.

Until notified otherwise, DoD military personnel are advised to refrain from the purchase and/or use of DTC genetic services.

Joseph D. Kernan  
Under Secretary of Defense for Intelligence

James N. Stewart  
Assistant Secretary of Defense for Manpower  
and Reserve Affairs, Performing the Duties  
of the Under Secretary of Defense for  
Personnel and Readiness

“These DTC genetic tests are largely unregulated and could expose personal and genetic information, and potentially create unintended security consequences and increased risk to the joint force and mission.”

“Testing outside the Military Health System is unlikely to include a clear description of this risk.”

“Until notified otherwise, DoD military personnel are advised to refrain from the purchase and/or use of DTC genetic services.”

# QUESTIONS?

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