

# Connecticut Genomics Forum:

## Genetic Counseling

**Miriam DiMaio, MSW, CGC**

Frank H. Netter MD School of Medicine at Quinnipiac University

**Samantha Wesoly, MS, CGC**

Genetics and Genome Sciences, UConn School of Medicine

**Danielle Bonadies, MS, CGC**

My Gene Counsel



# **Miriam S. DiMaio, MSW**

Frank H. Netter MD School of Medicine  
Quinnipiac University

Genetic Counselor Licensed by the State of Connecticut  
Board Certified in Genetic Counseling  
by the American Board of Medical Genetics

I have no conflicts of interest.



# Who Are Genetic Counselors?

- Genetic counselors are health professionals with specialized training, graduate degrees, and experience in medical genetics and counseling.
- They work as part of a health care team that includes physicians, nurses, social workers, laboratory personnel, and others.
- They enter the field from a variety of disciplines, including biology, genetics, nursing, psychology, public health, and social work.
- They are certified or eligible for certification by the American Board of Medical Genetics or the American Board of Genetic Counseling and can be licensed by the State of Connecticut based on established criteria.



# What Is Genetic Counseling?

Genetic counseling is the process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease.



# What Is the Role of a Genetic Counselor?

- **Interpretation** of family and medical histories and laboratory data to determine appropriate DNA tests for making a diagnosis
- **Organizing, coordinating, and facilitating** interpretation of DNA test results
- **Educating** patients and families about DNA test results, inheritance patterns, risk assessment, disease management, treatment, prevention, reproductive options, resources, and research
- **Counseling** about risks to other relatives
- **Promoting** informed choices and adaptation to the risk or condition
- **Educating** referring physicians and other health professionals about benefits, limitations, and possible outcomes of genetic testing



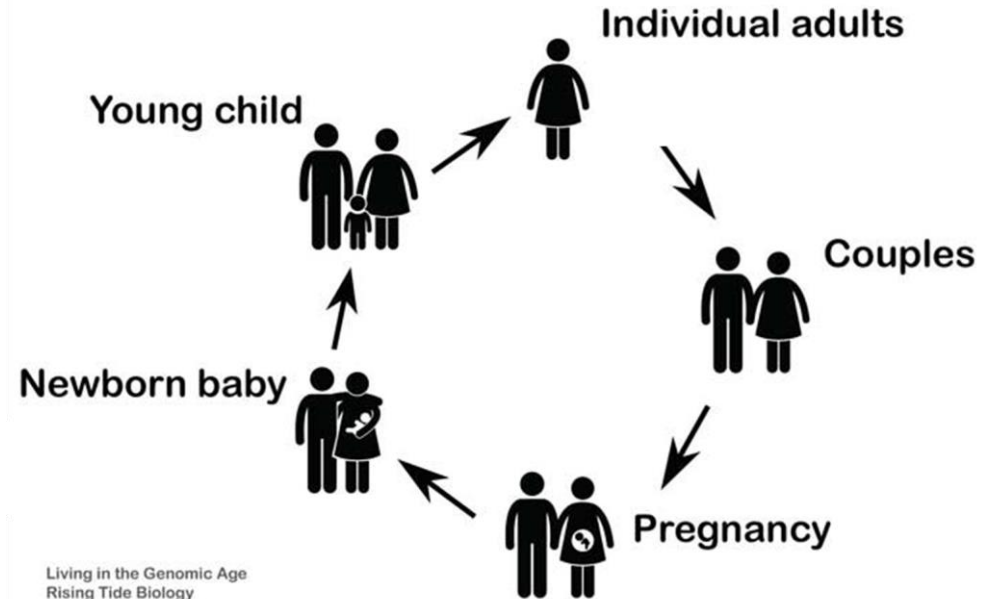
# Where Do Genetic Counselors Work?

- Hospitals
- Doctor's offices
- University and commercial genetic testing laboratories
- Research projects
- Public health initiatives
- Insurance companies
- Consumer advocacy organizations
- and more...



# Genetic Counselors Are Involved Throughout the Lifespan

- Examples of newborn screening
  - Sickle cell disease
  - Phenylketonuria (PKU)
  - Cystic fibrosis
  - Hearing loss
  - Many others



Living in the Genomic Age  
Rising Tide Biology  
[www.risingtidebio.com](http://www.risingtidebio.com)



# Case Example: Impact of Genetic Testing on Families

- An 8-month-old previously healthy boy died suddenly in his sleep of unknown reasons. An autopsy did not reveal the cause of death.
- DNA testing on a postmortem tissue sample discovered the cause of death.
- The boy's genes contained a mutation that affected his heart's electrical circuits, which can lead to fatal arrhythmias at any stage of life.
- Because the underlying genetic cause was identified, other family members could be tested to determine if they are at risk for dying suddenly due to the same heart problem.
- Testing of the deceased boy's parents and three siblings showed that his mother and two of his siblings have the same genetic defect and thus are at risk for sudden death.
- They are now receiving medical treatment to prevent sudden death.



# Impact of Genetic Testing on Families: Not an Isolated Example

- At least several percent of children who die of sudden infant death syndrome (SIDS) have a genetic mutation predisposing to fatal cardiac arrhythmias
- 30% of swimming-related drownings have a similar genetic arrhythmia syndrome
- ~30% of people under 35 who die suddenly of unexplained causes with a normal autopsy have similar genetic mutations that affect the heart's electrical system

*Behere & Sweindling Ann Pediatr Cardiol 2015 (8(3): 210-220. Kenny & Martin Arch Dis Child 2011: 96:5-8; Tester Mayo Clin Proc 2011 Oct;86(10):941; Klaver et al Int J Cardiol 2011 Oct 0;152(2):162-7*



# What are the Standards and Protocols that Genetic Counselors Follow for Counseling and DNA testing?

Guidelines regarding indications for DNA testing based on professional society recommendations:

- American College of Medical Genetics and Genomics
- National Society of Genetic Counselors
- American Society of Clinical Oncology
- American College of Obstetrics and Gynecology
- Society for Maternal Fetal Medicine
- Society for Vascular Surgery
- American College of Cardiology Foundation/American Heart Association
- American Medical Association
- Others



# Genetic Testing and Counseling: Continuing Challenges

## Complexity of Interpretation of Genetic Testing Results

- Tidal wave of genetic information now available
- Consequences of specific DNA changes is sometimes complex and not always clear cut
- DNA testing may reveal an unexpected finding that may be medically important

## Genetics Education of Physicians:

- Lack of genetics knowledge of primary care physicians and other specialists
- Rapid advances in the field
- Shortage of genetic counselors (and medical geneticists) to meet growing demands for services
- Medical school genetics curricula:
  - Insufficient emphasis on genetic basis of health and disease
  - Curriculum lags behind current genetics knowledge



# Summary

- Genetic counselors work in concert with physicians and other health professionals to ensure that DNA testing, and interpretation and dissemination of test results to patients and referring physicians are performed with high standards
- The explosion of knowledge in genetics and the rapidly increasing use of DNA-based tests in health management requires more health professionals with training in genetics.
- Clear guidelines and safeguards need to be adopted uniformly to protect DNA information



# Connecticut Genomics Forum:

## Genetic Counseling

**Samantha Wesoly, MS, CGC**

Genetics and Genome Sciences, UConn School of Medicine

Connecticut Children's Division of Genetics

[swesoly@connecticutchildrens.org](mailto:swesoly@connecticutchildrens.org)



# Safety and Security of DNA Data

**The laws governing genetic data in the United States vary by state**

- Different federal and state laws, regulations, official guidance, and professional standards that currently regulate the field of genomics
- Genetic anonymity is almost never guaranteed
- Need consensus among policymakers about the future where almost everyone will have DNA testing
- NIH funded LawSeq project: assessing field's biggest challenges and seeking a consensus about standards that regulate the privacy of DNA

# Protections in Federally Funded Research

## Specific protections exist for individuals who participate in research studies

- A federal law protecting human subjects, called the US Common Rule, mandates that you be informed of how your data might be shared prior to signing a consent document
- In 2016, Congress passed the [21st Century Cures Act](#), which also provides any federal research subjects with a certificate of confidentiality.
- This restricts the release of your genetic data to law enforcement or other government agencies
- If that information were to somehow be illegally obtained, through a hack or some other breach, it would be inadmissible in court

# Protections in the Medical Record

**Genetic information is incorporated into the medical record regularly**

- Information becomes a piece of personal health data governed by HIPAA
- Data can't be given to a school or employer
- Law enforcement agencies are entitled to access it without a warrant if you are the victim of a crime or a suspect in a criminal investigation

# Protections by Commercial Laboratories

## University-based and commercial laboratories can use remaining DNA sample for various purposes

- Samples are de-identified and can be used for:
  - Quality improvement of equipment
  - Internal research projects
  - Test development
- Offer “opt out” forms to destroy the patient’s DNA sample after the clinical test is complete
- Several states, including New York, have implemented strict privacy protections for genetic information
  - Limits additional tests that can be performed on samples without the individual’s prior consent
  - Requires destruction of sample within 60 days of completion of the test
- Every lab is different and has different requirements, but all labs need to be CLIA-certified

# Accessing Genetic Data

**Patients can access their genetic data by sending a request to the lab that performed their genetic testing**

- Patients obtain the report that comes from the lab, but they have to specifically request raw, unfiltered DNA data
- Patient empowerment by obtaining their own data
- Patients can then run their raw data through third party websites
  - Promethease
  - Genetic Genie

# Case Vignette

## 40 year old healthy woman who ordered 23&Me and ran her data through a third party vendor

- Report listed several variants in the *COL3A1* gene
  - Associated with vascular Ehlers-Danlos syndrome, a potentially fatal connective tissue disorder
- Presented in clinic with significant anxiety that she had this genetic condition
- Genetic counseling session included the utility of third party vendors and the difference between disease causing genetic changes and benign genetic changes
- Patient left the session better understanding the complexities of genetic data

# Connecticut Genomics Forum:

## Genetic Counseling

**Danielle Bonadies, MS, CGC**

Co-Founder and Director of Genetics

My Gene Counsel

[Danielle@MyGeneCounsel.com](mailto:Danielle@MyGeneCounsel.com)



# Avenues of Testing

## Provider Initiated

- Traditional medical test
- Most comprehensive
- Provider recommends and coordinates testing
- Diagnostic tests
- Often covered by health insurance when certain criteria are met

## Patient Initiated

- Patient initiated, but...
- Doctor “approved”
- Typically full genetic analysis/sequencing
- Generally self-pay

## Direct-to-Consumer

- Initiated by consumer
- No MD/clinician involved
- Often ordered online
- Genotyping for particular variants/SNPs
- Patient self-pay



# BRCA1/2: Breast and Ovarian Cancer Syndrome

CANCER	AVG RISK	BRCA1+ RISK	BRCA2+ RISK
Female Breast	12-13%	50-85%	40-70%
Ovarian	1-2%	25-60%	10-20%
Prostate	11%	increased	20-30%
Male Breast	<1%	1-2%	7%
Pancreatic	<1%	2-3%	2-5%
Melanoma	1-2%	increased	3-5%





## AGE 18

- Become familiar with your breasts as breast self-exams may be helpful.



## AGE 25-29

- Become familiar with your breasts.
- Breast self-exams may be helpful.
- Clinical breast exams every 6-12 months.
- Yearly breast MRI with contrast.



## AGE 30-75

- Become familiar with your breasts.
- Breast self-exams may be helpful.
- Clinical breast exams every 6-12 months.
- Yearly breast MRI with contrast.
- Yearly mammogram.



## AFTER AGE 75

- Breast imaging schedule should be discussed with your health care provider.



## RISK REDUCTION

- Discuss the option of risk-reducing mastectomy (RRM) with your health care providers. Cover these topics: degree of breast cancer risk reduction, surgical risks, reconstruction options, and the emotional, financial, and social aspects of RRM.
- Consider chemoprevention medications (Tamoxifen and Evista) to decrease the risk of breast cancer.

# Hereditary Cancer Testing: A Comparison

- Full gene analysis BRCA
- Most accurate and comprehensive
- Must be ordered by health care provider
- Must meet insurance criteria for coverage
- Pre and post test counseling recommended

Provider Initiated



- Full gene analysis with some testing limitations
- Patient initiated, but must have MD sign off
- Does not know patient
- Skirts FDA
- Self-pay
- Post-test counseling offered if positive

Patient Initiated



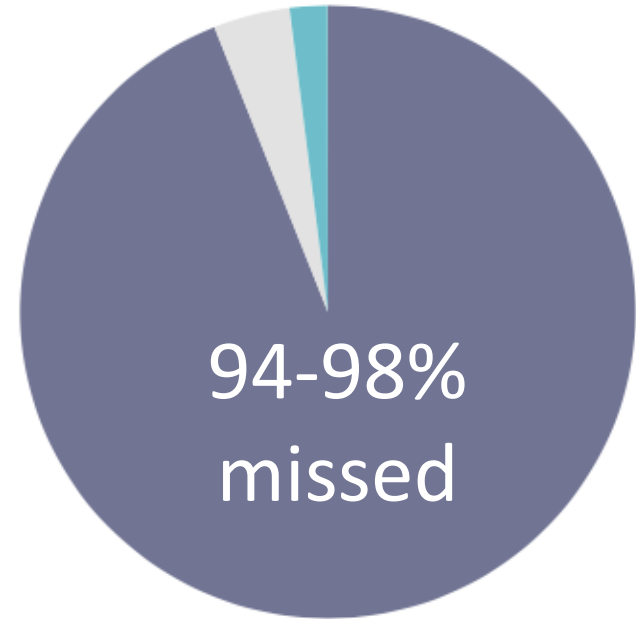
- Three of >1800 BRCA mutations
- Jewish ancestry
- FDA recommends confirmatory testing
- False reassurance with “negative” results
- 100s of tests; only 23andMe FDA approved

Direct-to Consumer



# Hereditary Cancer Testing: A Comparison

- ~400,000 patients who were BRCA+ through medical lab
- Analyzed data to see how many would have been missed if that patient had taken a DTC test



# More than 26 million people have taken an at-home ancestry test

The genetic genie is out of the bottle. And it's not going back.

by Antonio Regalado

---

Feb 11, 2019

# Impact to Patients

## *The Online Gene Test Finds a Dangerous Mutation. It May Well Be Wrong.*

Third-party analysis of raw DNA is not as rigorous as that done in a certified laboratory. But many consumers don't understand that their results are not conclusive.



By Gina Kolata

July 2, 2018

YOUR HEALTH

## Results Of At-Home Genetic Tests For Health Can Be Hard To Interpret

June 18, 2018 · 4:58 AM ET  
Heard on Morning Edition



ROB STEIN



GENETICS

## Another Reminder That Consumer DNA Tests Are Not 100% Accurate



Kristen V. Brown

3/28/18 3:55pm · Filed to: 23ANDME ▾

78.5K 35

Home / The Scientist

## Study Finds Inaccuracies in 40 Percent of DTC Genetic Testing Results

An analysis of 49 patient samples finds high proportions of false positives and misinterpretation.

BUSINESS

Mar 28, 2018  
SHAWNA WILLIAMS

## Genetic tests ordered by doctors race to market, while 'direct-to-consumer' tests hinge on FDA approval

By IKE SWETLITZ @ikeswetlitz / MARCH 16, 2018

## How Accurate Is Direct-To-Consumer Genetic Testing? From Gold(ish) To Garbage



Ellen Matloff Contributor

May 28, 2018, 02:18pm · 10,551 views · #IfIOnlyKnew

# Testing Without Guidance = Misinterpretation

ORIGINAL ARTICLE

## Errors in Genetic Testing: The Fourth Case Series

Meagan B. Farmer, MS, CGC, MBA,\*† Danielle C. Bonadies, MS, CGC,\*  
Suzanne M. Mahon, DNSc, RN, AOCN, AGN-BC,‡ Maria J. Baker, PhD, FACMG, MS, LCGC,§  
Sumedha M. Ghatge, MS, CGC,|| Christine Munro, MS, MPH, LCGC,¶ Chinmayee B. Nagaraj, MS, LCGC,#  
Andria G. Besser, MS, CGC,\*\* Kara Bui, MS, CGC,†† Christen M. Csuy, MS, CGC,‡‡  
Brianna Kirkpatrick, MS, LCGC,§§ Andrew J. McCarty, MS, LCGC,¶¶ Shelly Weiss McQuaid, MS, CGC,¶¶  
Jessica Sebastian, MS, LCGC,## Darci L. Sternes, MS, LCGC,\*\*\*  
Leslie K. Walsh, MS, MPH, LCGC,## and Ellen T. Matloff, MS, CGC\*

**Purpose:** In this ongoing national case series, we document 25 new genetic testing cases in which tests were recommended, ordered, interpreted, or used incorrectly.

**Methods:** An invitation to submit cases of adverse events in genetic testing was issued to the general National Society of Genetic Counselors List-serv, the National Society of Genetic Counselors Cancer Special Interest Group members, private genetic counselor laboratory groups, and via social media platforms (i.e., Facebook, Twitter, LinkedIn). Examples highlighted in the invitation included errors in ordering, counseling, and/or interpretation of genetic testing and did not limit submissions to cases involving genetic testing for hereditary cancer predisposition. Clinical documentation, including pedigree, was requested. Twenty-five cases were accepted, and a thematic analysis was performed. Submitters were asked to approve the representation of their cases before manuscript submission.

**Results:** All submitted cases took place in the United States and were from cancer, pediatric, preconception, and general adult settings and involved both medical-grade and direct-to-consumer genetic testing with raw data analysis. In 8 cases, providers ordered the wrong genetic test. In 2 cases, multiple errors were made when genetic testing was ordered. In 3 cases, patients received incorrect information from providers because genetic test results were misinterpreted or because of limitations in the provider's knowledge of genetics. In 3 cases, pathogenic genetic variants identified were incorrectly assumed to completely explain the exercises

genetic counseling delivery models, expanding the genetic counseling workforce, improving genetics and genomics education of nongenetics health care professionals, addressing health care policy barriers, and more. Genetic counselors have also positioned themselves in new roles to help patients and consumers as well as health care providers, systems, and payers adapt to new genetic testing technologies and models. The work to be done is significant, but so are the consequences of errors in genetic testing.

**Key Words:** Cancer genetic testing, direct-to-consumer screening and testing, genetic counseling, genetic counseling delivery models, genetic services, genetic testing, genetic testing adverse events, genetic test misinterpretation, genomics, multigene panel testing

(*Cancer J* 2019;25: 231–236)

### ERRORS IN GENETIC TESTING: THE FOURTH CASE SERIES

The availability of genetic testing is growing at an exponential rate. In a 2018 study providing an overview of the current genetic testing landscape, authors estimated that there were approximately 75,000 genetic tests on the market, with 10 new tests being introduced daily.<sup>1</sup> Fourteen percent of these tests, and 2 to 3 of the new tests introduced per day, were panel tests, a category that includes

# The Problem



- 28-year-old healthy woman
- Carried a BRCA1 mutation
- Opted for bilateral mastectomy
- Presented for genetic counseling
- Results were reviewed
- Carried a benign genetic finding
- Did not need preventive surgery

# The Problem

- 47-year-old woman, 2 teenage boys
- Diagnosed with Stage IIIC ovarian cancer
- Tested negative 2 years prior
- Seen for genetic counseling
- Sent in new DNA sample
- Lab called us, confused
- Found to carry a BRCA2 mutation 2 years earlier

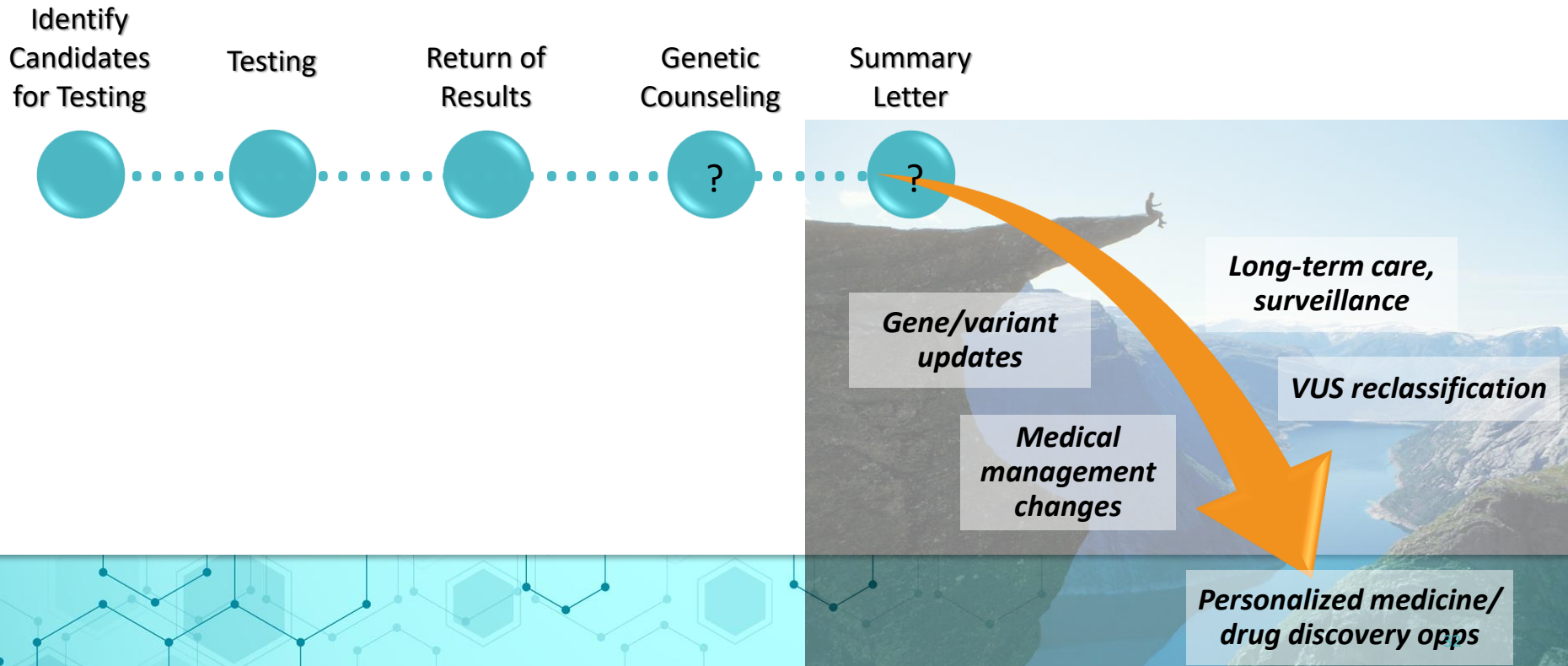


# Scope

- Rapidly changing field, thousands of genes
- Clinicians have 12 minutes/patient
- High rate of misinterpretation amongst clinicians and patients
- Errors lead to unnecessary preventative surgery, wrong treatments
- Between 100 million and 2 billion human genomes sequenced by 2025
- Virtually all clinical trials now include genetic testing
- Researchers will need to return results to patients and families

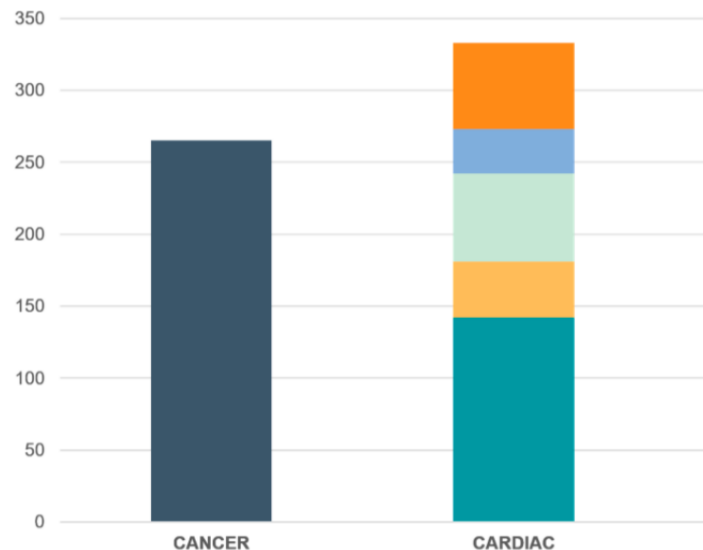


# Current State



# Continuous Medical Management Changes

**600+ Changes Within the ACMG 59 Genes**



**Within 5 Years**

# Future = Genetic Testing +

- Tools for patients and providers
- Comprehensive, searchable resources
  - Patient experience
  - Provider experience
- Accessible, scalable information
- Referrals to genetic counselors
- Push notifications and updating reports
  - Variant reclassifications
  - Medical management updates
- Engagement and retention
  - Outcome data
  - Clinical trial info and invitations



**Precision Medicine**

# State's Role to Ensure Safety, Security & Integrity of Precision Medicine

- CT workforce studies
- Several insurers now only pay for testing if ordered by a genetic counselor
- Accreditation by the commission on cancer
- Funding support of training programs to address shortage of genetic counselors
- Widen scope of licensure of genetic counselors to ensure high standards of care
- Support H.R. 3235: Access to Genetic Counselor Services Act of 2019: Centers for Medicare & Medicaid Services (CMS) recognition to bill
- Provide tools for all types of providers to incorporate genetics into their practice



# Connecticut Genomics Forum:

## Genetic Counseling

**Miriam DiMaio, MSW, CGC**

Frank H. Netter MD School of Medicine at Quinnipiac University

**Samantha Wesoly, MS, CGC**

Genetics and Genome Sciences, UConn School of Medicine

**Danielle Bonadies, MS, CGC**

My Gene Counsel

