



**FRIENDS OF THE**  
NATIONAL LIBRARY  
OF MEDICINE

# PRECISION HEALTH: WHAT DOES IT MEAN FOR NURSING PRACTICE?

Accelerating the Integration of Precision  
Health into Nursing Practice

A VIRTUAL WEBINAR

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**Tuesday, February 6, 2024**  
**9:30am – 5:00pm Eastern Time**

# Welcome

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**Glen Campbell**

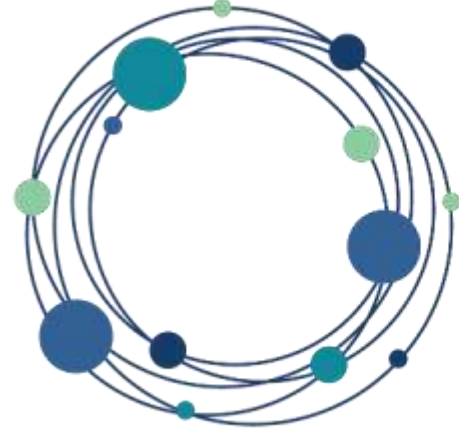
Chair, Friends of the National Library of Medicine

# Welcome

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**Glen Campbell**

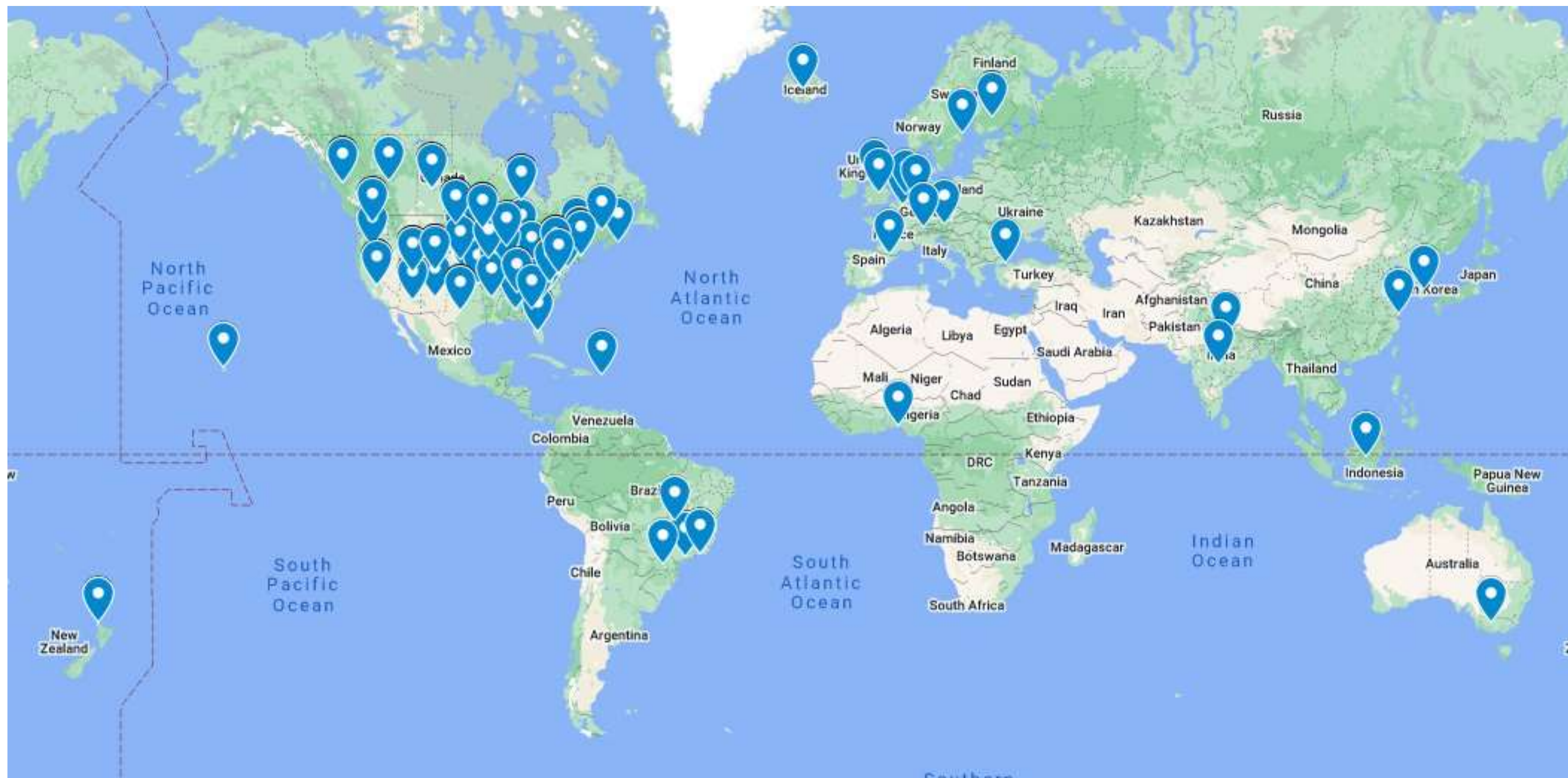
Chair, Friends of the National Library of Medicine



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**Proud to support and promote the**  
***National Library of Medicine***  
**the world's largest medical library, a center for data**  
**science research, and a vital component of NIH.**







The Friends of the National Library of Medicine supports and promotes the world's largest medical library, the National Library of Medicine (NLM), a vital part of the National Institutes of Health (NIH), the nation's premier medical research agency.

The Friends organization was formed in 1986 as a nonprofit 501(c)(3) organization to promote, publicize, and support the Library. FNLM is a coalition of individuals, medical associations and societies, hospitals, health science libraries, corporations, and foundations, dedicated to increasing public awareness and use of the NLM, as well as to support its many programs in research, education, and public service.

The Library, founded in 1836 and now located on the NIH campus in Bethesda, Maryland, is the world's largest repository of biomedical and health sciences information. It has over eight million items including books, journals, videos, historical artifacts, and receives over one billion hits per year over the World Wide Web.

The Library's constituency includes those who participate in its programs and utilize its services and databases. These are physicians, nurses, dentists, librarians and other members of the health care community, research scientists in medical centers, institutes and pharmaceutical, chemistry and communication corporations, and the public - including patients and family caregivers. Members of the FNLM Board represent all these constituencies in various parts of the country and serve without compensation, giving freely of their time and expertise.



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**Roy Simpson**  
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# Accelerating Discovery and Data-Powered Health



**Accelerate discovery and  
advance health through  
data-driven research**



**Reach more people in more  
ways through enhanced  
dissemination and  
engagement**



**Build a workforce  
for data-driven  
research and health**

# Mentoring in Medicine



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The National Library of Medicine (NLM) and Friends of NLM continue to be among the sponsors of Mentoring in Medicine (MIM). This nonprofit outreach program works with disadvantaged minority students to spark their interest in healthcare careers and, ultimately, to help diversify the healthcare workforce.

# Friends of the NLM Webinars 2024

Supporting Biomedical Discovery and Advancing Public Health



**E. Andrew Balas,  
MD, PhD**  
Vice President

**Nursing's Role in  
Precision Healthcare**  
**February 6, 2024**

**Protecting the Health of  
the Public in the Age of  
Misinformation**  
**September 2024**



**Sandra Franklin, MLS,  
AHIP, FMLA**

**Principles and Practice of  
Quality Improvement in  
Biomedical Research**  
**April 2024**

**The Emerging Role of  
Artificial Intelligence and  
the Medical Librarian**  
**Spring 2024**





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Nursing Informatics Award

**Barbara B. Frink, PhD, RN, FAAN**

Vice President and Chief Nursing  
Information Officer  
MedStar Health



MICHAEL E. DEBAKEY LIBRARIAN  
OUTREACH AND COMMUNITY  
COLLABORATION AWARD

**Steve Wilson, MLS**

Web Architect & Outreach Librarian  
Coordinator, Center for Disability  
Resources Library



MICHAEL E. DEBAKEY LIBRARIAN  
OUTREACH AND COMMUNITY  
COLLABORATION AWARD

**Heather K Moberly, MLS**

Coordinator of Veterinary Information  
and Research Services at the Texas  
A&M University's Medical Sciences  
Library



## 2023 Friends of the NLM Honorees



PAUL G. ROGERS AWARD FOR  
PUBLIC SERVICE

**Carla D. Hayden, MA, PhD**

14th Librarian of Congress



DISTINGUISHED MEDICAL SCIENCE  
AWARD

**Jennifer Doudna, PhD**

Professor of Molecular and Cell  
Biology and Chemistry  
University of California  
Berkeley



DONALD A.B. LINDBERG DISTINGUISHED  
HEALTH COMMUNICATIONS AWARD

**Anthony Fauci, MD**

Director of the National Institute of  
Allergy and Infectious Diseases  
Chief Medical Advisor to the President



## 2024 NURSING INFORMATICS AWARD



**Murielle Beene**  
**DNP, MBA, MPH, MS, RN-BC, PMP, FAAN**  
Senior Vice President and Chief Health Informatics  
Officer at Trinity Health

## 2024 MICHAEL E. DEBAKEY LIBRARY SERVICES OUTREACH AWARD



**Katherine V. Chew**  
Associate Librarian for Research/Outreach Services  
& Mortuary Sciences, Center for Allied Health  
Programs and Center for Bioethics at the Health  
Sciences Libraries, University of Minnesota

## DONALD A.B. LINDBERG DISTINGUISHED HEALTH COMMUNICATIONS AWARD



**Eric Topol, MD**  
Founding Director of Scripps Research Translational Clinic.  
Author of three bestsellers on the future of medicine: *The  
Creative Destruction of Medicine* (2010), *The Patient Will  
See You Now* (2015), and *Deep Medicine: How Artificial  
Intelligence Can Make Healthcare Human Again* (2019).



## Friends of the National Library of Medicine 2024 Gala

Tuesday, September 10, 2024

6:00 PM – 9:00 PM ET

Willard Hotel

1401 Pennsylvania Ave NW

Washington, DC, 20004



# Appreciation and Gratitude

**Jessica Nassif**

**Friends of the NLM Operations Manager**



**Chris Reddall**

**Cypher Media**



**Jake Carless**

# **PRECISION HEALTH: WHAT DOES IT MEAN FOR NURSING PRACTICE?**

## **Accelerating the Integration of Precision Health into Nursing Practice**



**Chair**  
**Kathleen McCormick**  
**PhD, RN, FAAN, FACMI, FHIMSS**



**Barbara B. Frink**  
**PhD, RN, FAAN**



**Barbara Redman**  
**PhD, RN, MBE, FAAN**



**Mary Anne Schultz**  
**PhD, MBA, MSN, RN, FAAN**



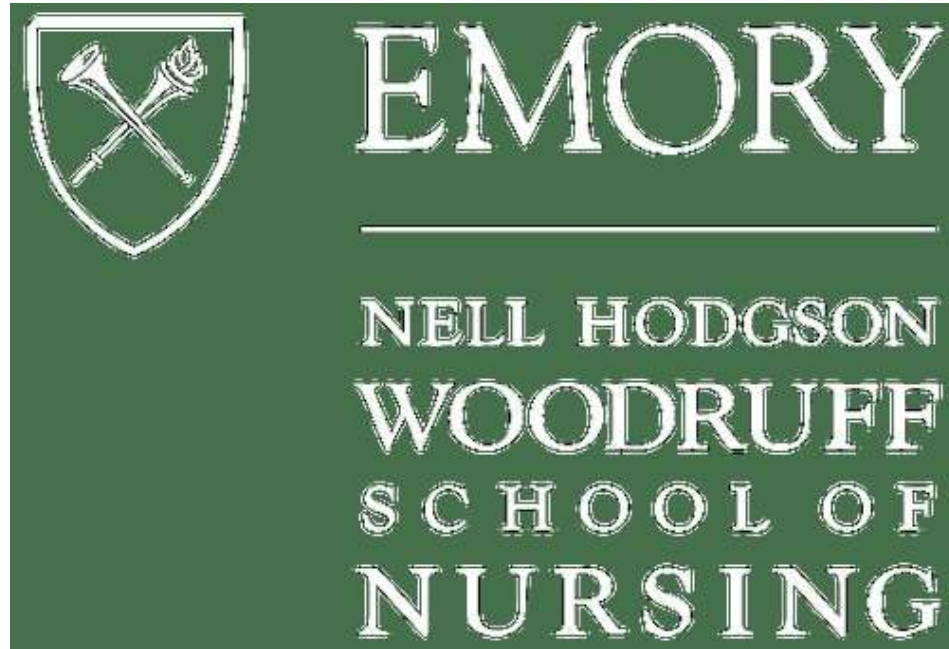
**Marisa Wilson**  
**DNSc, MHSc, RN-BC, CPHIMS, FAAN**



**Evangeline Fangonil-Gagalang**  
**PhD, RN**



## Appreciation and Gratitude



**National League  
*for* Nursing**

Emory Nursing Professional Development Center (ENPDC) is accredited as a provider of nursing continuing professional development by the American Nurses Credentialing Center's (ANCC) Commission on Accreditation



# CNE Credit Disclosures

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- ENPDC has evaluated everyone who has the ability to control content of this activity (planning committee members, subject matter experts, presenters) and found no relevant financial relationships

## **Disclosure to Learners: Awarding Contact Hours**

- To obtain contact hours participants must
  - Participate in the entire activity
  - Complete the evaluation at the end
- Certificates will be distributed at the end of the evaluation

Thanks, Diana Harmon, MSN, MHA, RN & Roy Simpson, DNP, RN, DPNAP, FAAN!



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  - Attendance at meetings of the membership.
- **\$425 – Basic + Education Program Member**
  - Basic membership
  - Four education webinars of choice during one calendar year – can be transferred.
  - Opportunity to serve on one committee.
- **\$750 – All-Inclusive Member**
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  - Access to all educational webinars during one calendar year – can be transferred.
  - One ticket to the annual Friends of the NLM Awards event – can be transferred.
  - Opportunity to serve on one committee.
  - May be asked to participate in a Friends educational event.

By joining the Friends of the National Library of Medicine, your membership dues provide increased funding for a variety of activities and programs that help move the National Library of Medicine (NLM), and society, forward. The NLM's role is critical in accelerating research and improving clinical outcomes, one that is invaluable now more than ever.

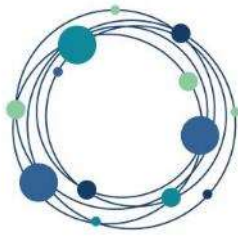


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- **Ability to purchase organizational Friends of the NLM educational program viewing rights:**
  - Academic organizations – inquire about fees.
  - Non-Academic and for-profit organizations – \$250 for up to five viewers per event or \$500 for six to ten viewers per event.
- For Further Information contact [jessican@fnlm.org](mailto:jessican@fnlm.org)





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# In Remembrance



**Marisa Wilson**  
**DNSc, MHSc, RN-BC, CPHIMS, FAAN**

**Dr. Virginia Saba**  
**Friends of the NLM**  
**Nursing Informatics**  
**Award**  
**2014**



# Introduction

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**Suzanne Bakken**  
PhD, RN, FAAN, FACMI, FIAHSI

# Marisa Wilson

DNSc, MHSc, RN-BC, CPHIMS, FAMIA, FIAHSI, FAAN

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- **Public health epidemiologist**
- **Nurse informaticist**
- **Nurse educator**
- **Mentor**
- **National and global leader**
- **Bold vision for nursing and for life**
- **Vision and planning for today's symposium**

**September 1, 1957 - January 5, 2024**

# Keynote

## Roles and Functions for Nursing Practice

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**Mary Anne Schultz**

PhD, MBA, MSN, RN, FAAN

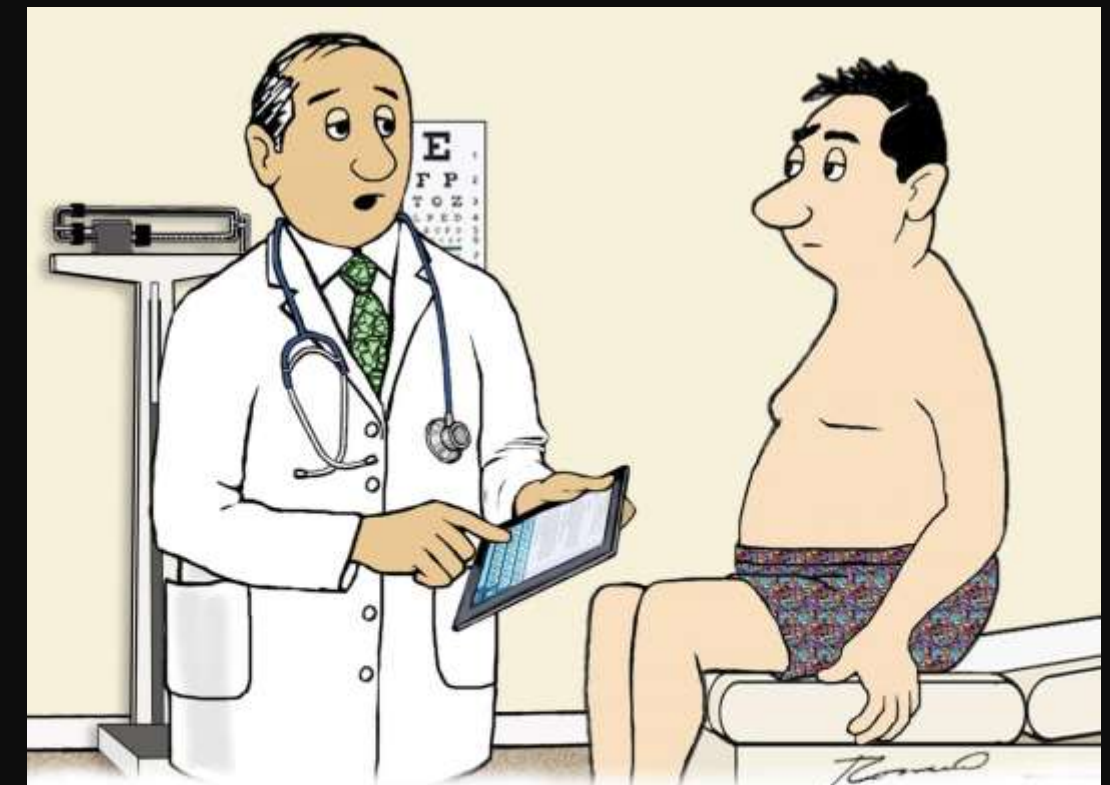


# Precision Health: Roles & Functions for Nursing Practice

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Mary Anne Schultz PhD MBA MSN RN FAAN  
Professor Emerita & Informaticist  
California State University, San Bernardino

# Doximity GPT: HIPAA-compliant A. I. Writing Assistant



"According to your HIPAA release form  
I can't share anything with you."

<http://tinyurl.com/mrkc86f7>

# Definition of Precision Health

**Precision Health** as defined by the National Institute of Health (2018) is most often described in relationship to Precision Medicine with authors identifying both commonalities and distinctions. Precision Medicine is defined as “the emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person. PH can be defined as an emerging approach to wellness which is respectful of individual lifestyle, genetics, behaviors and environmental contexts of our uniqueness.



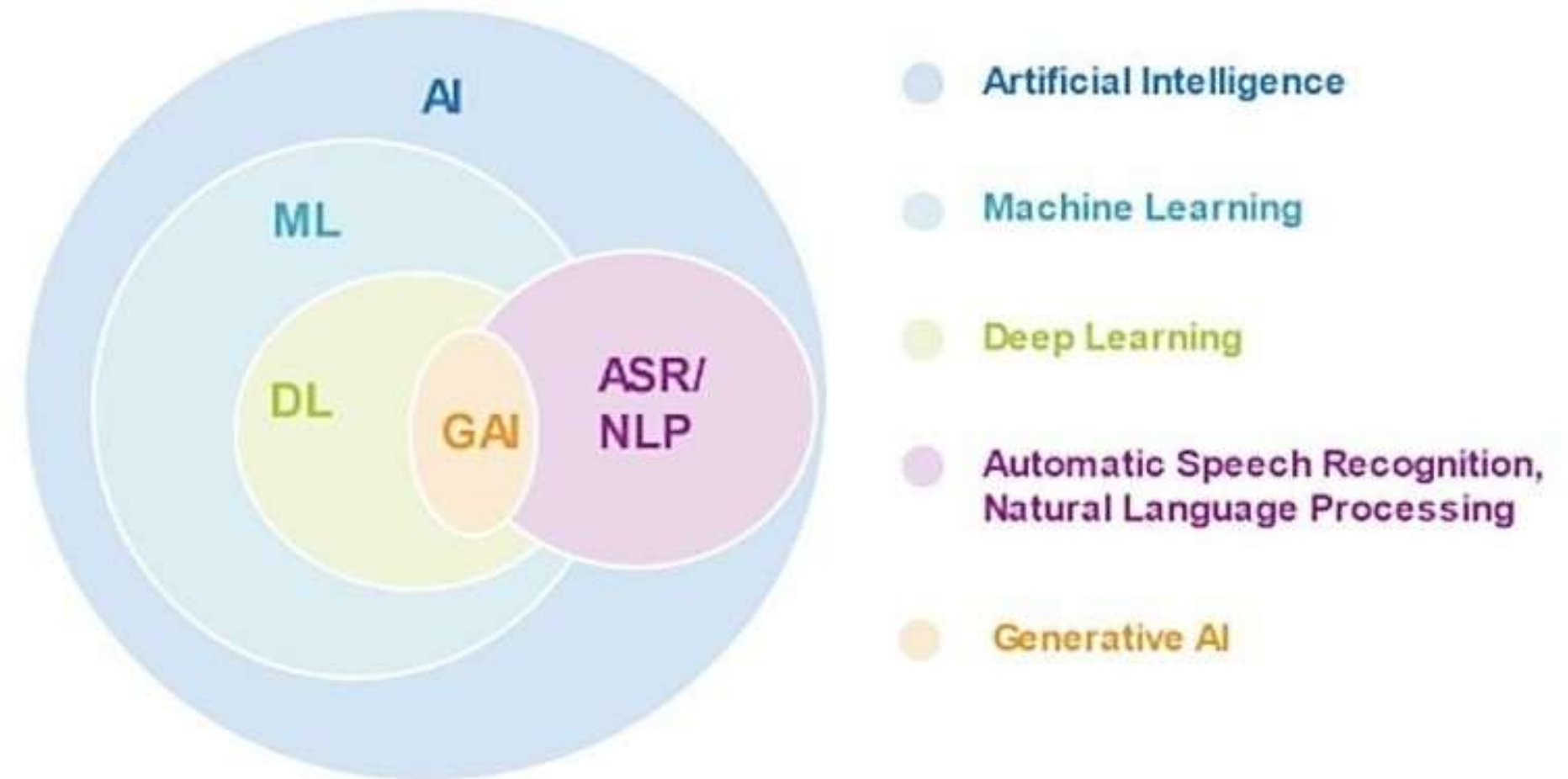
# 7 Things to Expect in Healthcare: *First*, Generative AI

<https://medicalfuturist.com/7-things-to-expect-from-ai-in-healthcare-this-year>



## What is Generative AI?

Generative AI refers to a category of artificial intelligence (AI) algorithms that generate new outputs based on the data they have been trained on. Unlike traditional AI systems that are designed to recognize patterns and make predictions, generative AI creates new content in the form of images, text, audio, and more.

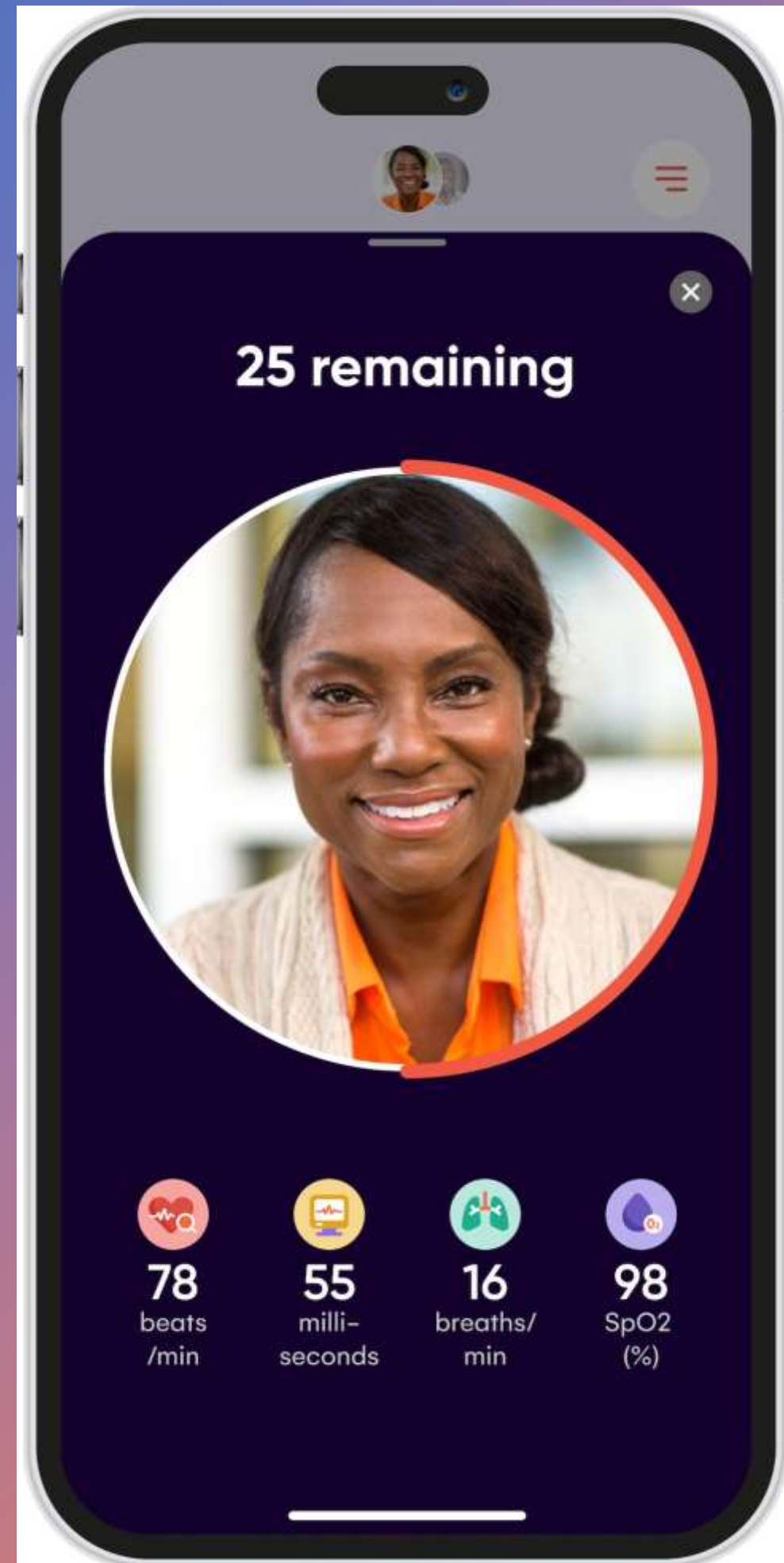




# ChatGPT Unleashed: The Future of Generative AI for K-12 and Higher Education

ChatGPT is a Generative Pre-trained Transformer (GPT) trained on OpenAI's Large Language Model (LLM)

- **Large Language Models (LLM)** are machine learning models that are designed to understand and process natural language by learning the structure and patterns of language from large datasets, enabling them to generate or process natural language text with a high degree of accuracy and fluency.
- **Generative Pre-trained Transformers (GPT)** are specific types of LLMs that have been pre-trained on large amounts of text data (i.e., ChatGPT, etc.).



# Together by Renee

First app to use **Generative AI** to make health care tasks “magically easy”

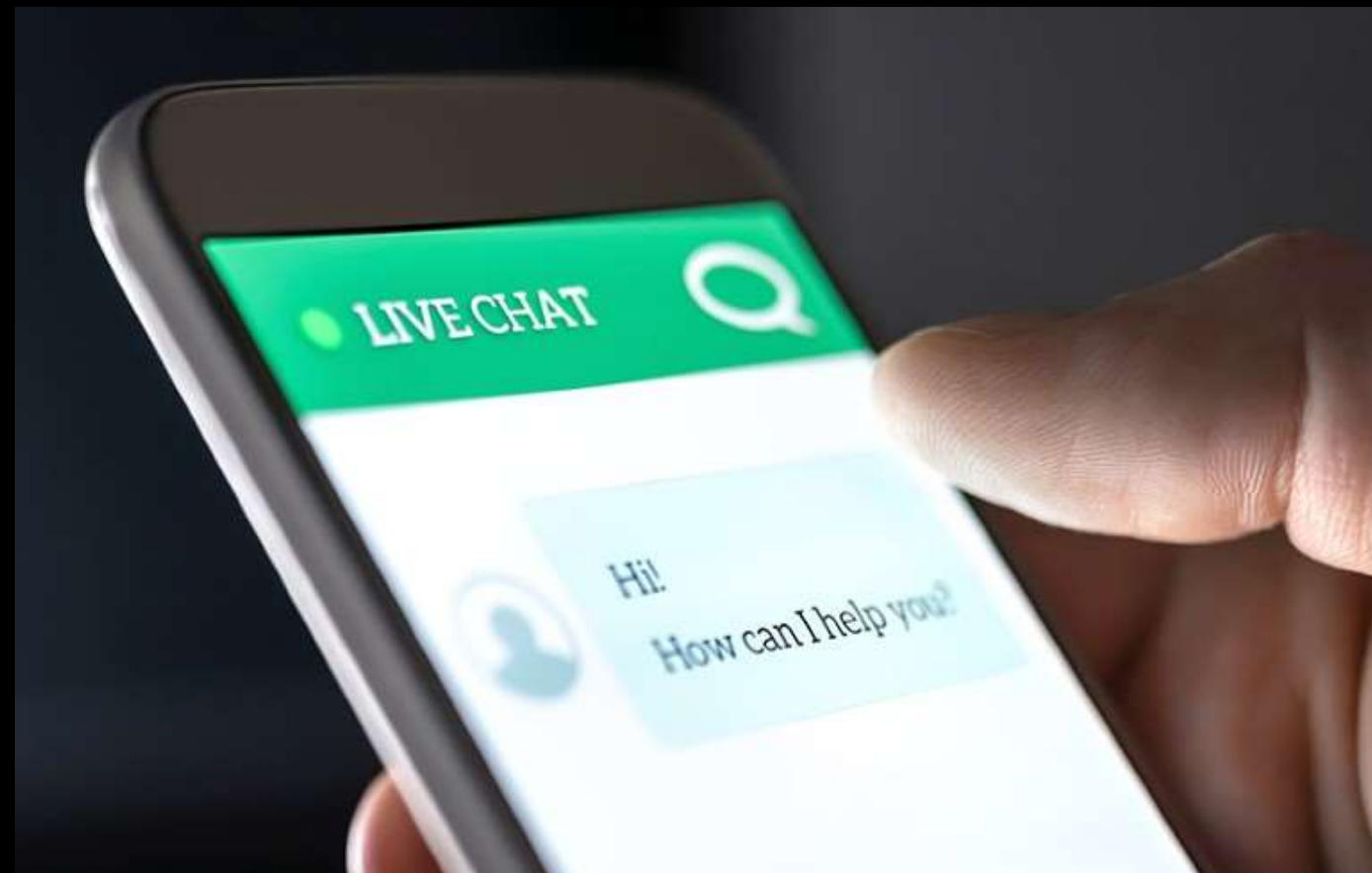
<http://tinyurl.com/yaefv9fw>

# NewsByte: The AI Arms Race: Microsoft's \$16 Billion Acquisition of Nuance

**Nuance Communications** integrates ***GPT-4*** into its ***Dragon Ambient Intelligence Platform*** to 'listen' to clinical conversation to generate the work of 'scribes'.







# 7 Things to Expect in Healthcare: *Second*, Medical LLMs Replace ChatGPT

<http://tinyurl.com/4r3yf8y8>

<https://medicalfuturist.com/7-things-to-expect-from-ai-in-healthcare-this-year>

Can ChatGPT & ChatGPT-4 be trained to diagnose medical conditions accurately?

*...if you want your patients to die...*

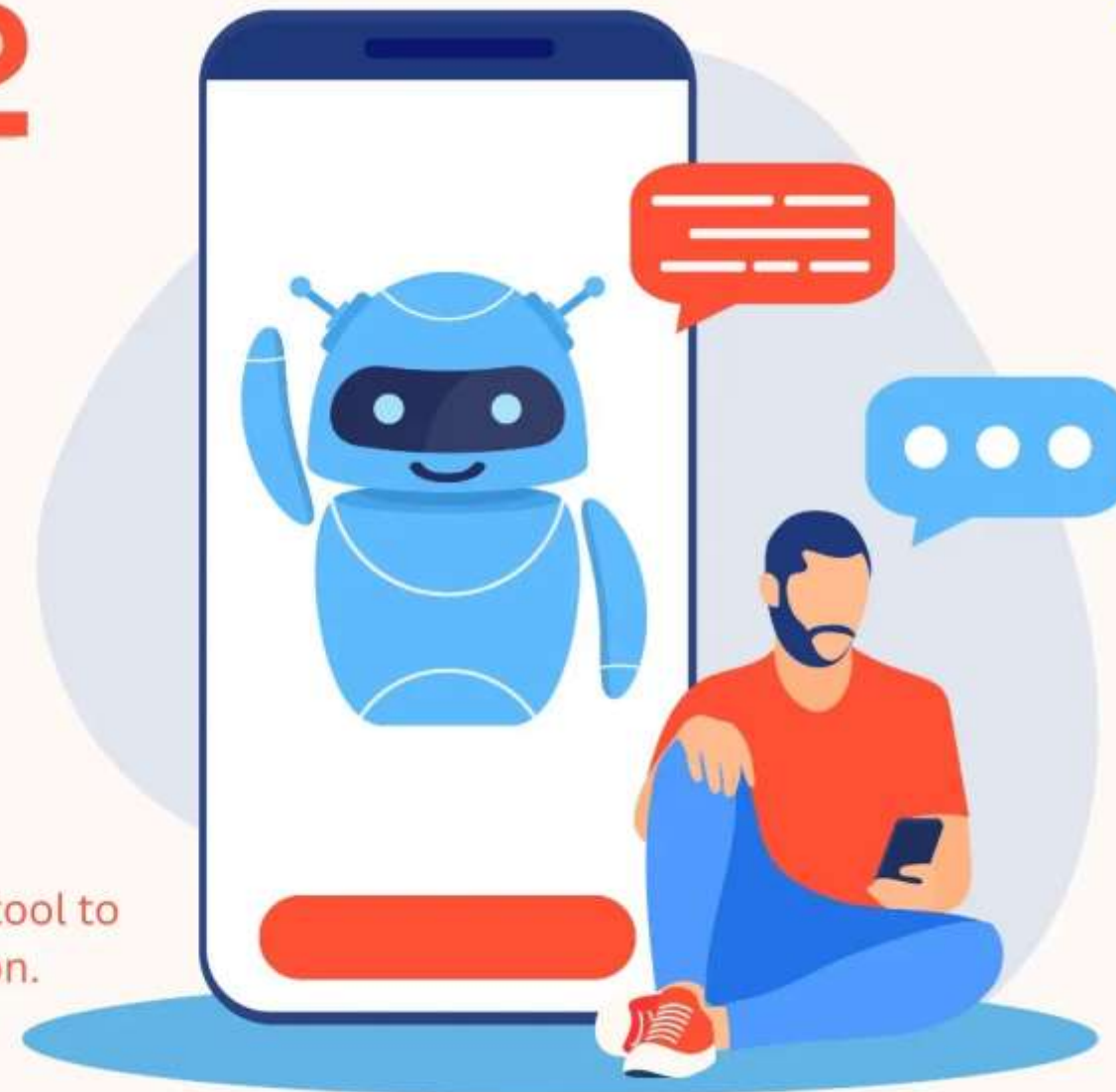


# Google & Med-Palm 2

## MED-PALM 2

**Google upgrades  
Med-PaLM for  
reliable health  
information**

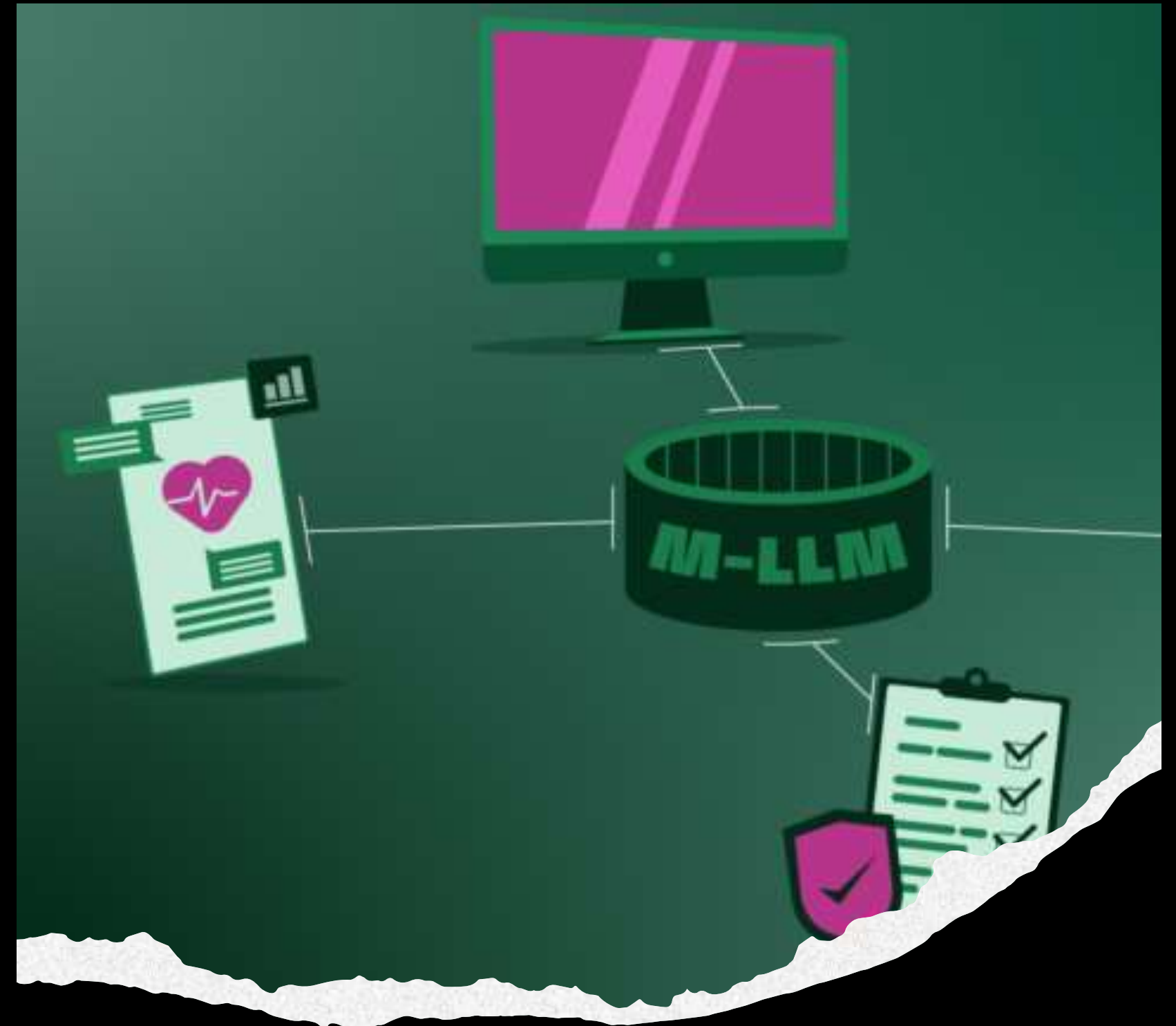
Google's revolutionary medical question-answering tool to provide accurate and reliable medical information.





# 7 Things to Expect in Healthcare: *Third*, Multimodal LLMs for Hospitals

<https://medicalfuturist.com/7-things-to-expect-from-ai-in-healthcare-this-year>



<https://medicalfuturist.com/7-things-to-expect-from-ai-in-healthcare-this-year>

# 7 Things to Expect in Healthcare: *Fourth*, AI Digital Twins

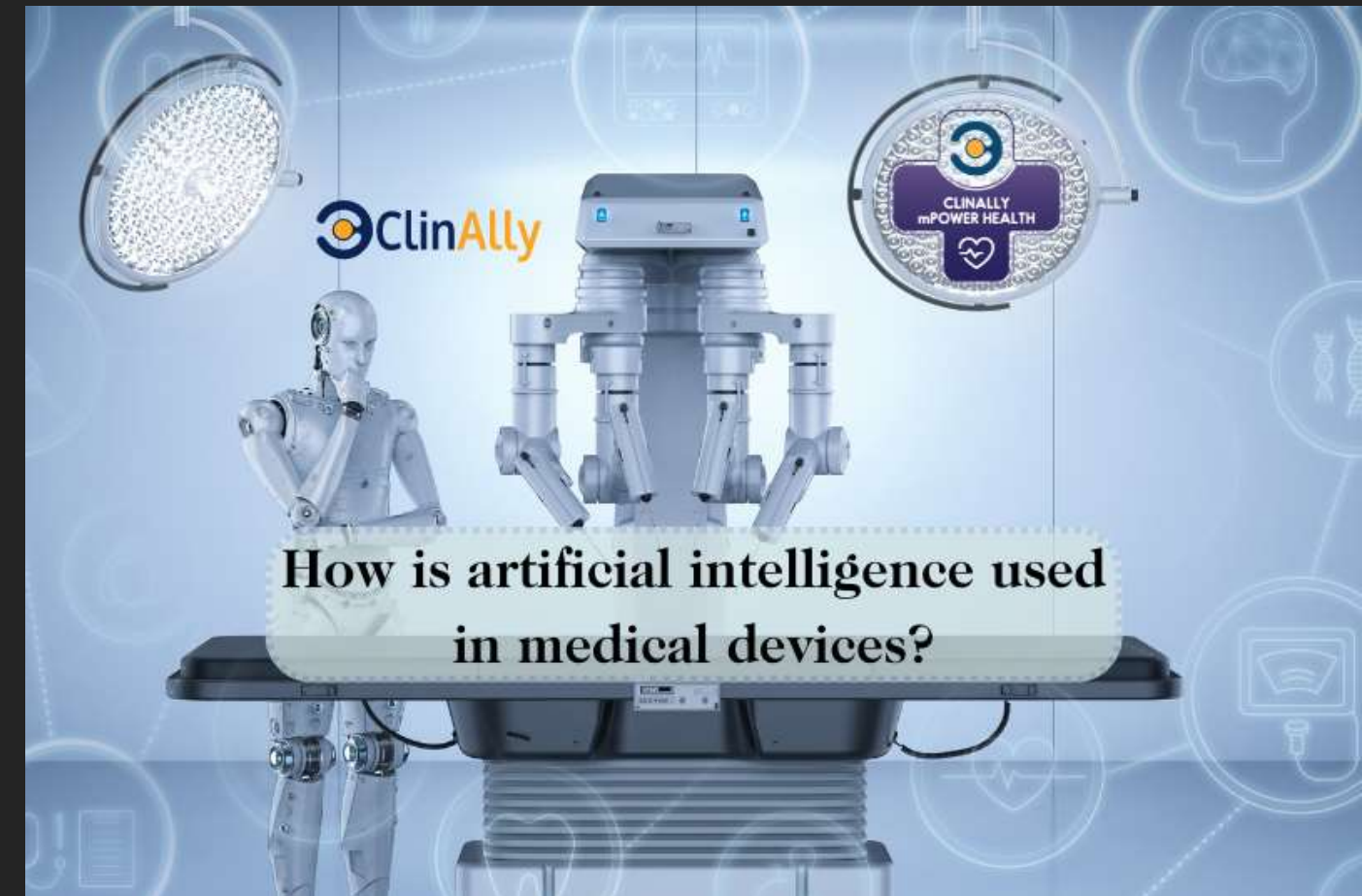
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<https://medicalfuturist.com/7-things-to-expect-from-ai-in-healthcare-this-year>


# 7 Things to Expect in Healthcare: *Fifth*, Over 1,000 FDA-approved Devices

<https://medicalfuturist.com/7-things-to-expect-from-ai-in-healthcare-this-year>



<https://app.clinally.com/the-use-of-artificial-intelligence-in-medical-devices>





**7** Things to Expect in  
Healthcare:  
*Sixth*, Influx of AI Tools for  
Patients

[www.glzlab.com](http://www.glzlab.com)

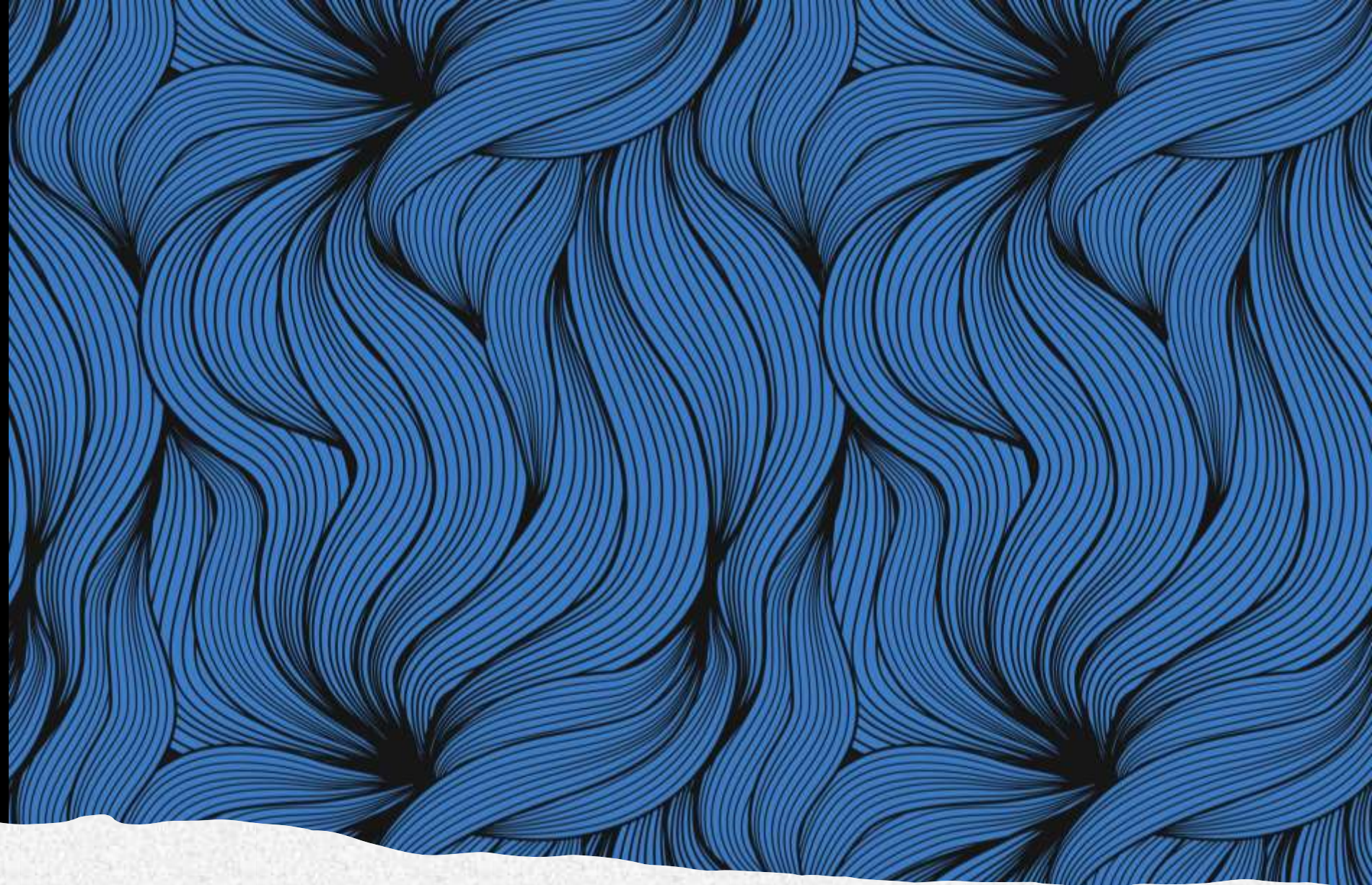
<https://medicalfuturist.com/7-things-to-expect-from-ai-in-healthcare-this-year>



# Med-PaLM

Large language model from Google Research,  
designed for the medical domain.

Google



## 7 Things to Expect in Healthcare: *Seventh*, Prompt Engineering

The Art and Science of Crafting Effective  
Inputs to Guide an AI Model Towards  
Generating the Desired Output [TMF]

<https://medicalfuturist.com/prompt-engineering-11-tips-to-craft-great-chatgpt-prompts>



# 11 Tips to Craft Great ChatGPT Prompts

Be Specific

Describe the Goal

Describe Setting &  
Context

Experiment with  
Different Prompt  
Styles

Ask it to Play Roles

Iterate and Refine  
to Delve Deeper  
and/or Get Better  
Answers

Use Your Previous  
Threads

Ask Open-Ended  
Questions

Request Examples

Use Time Wisely

Set Realistic  
Expectations (up to  
2021)

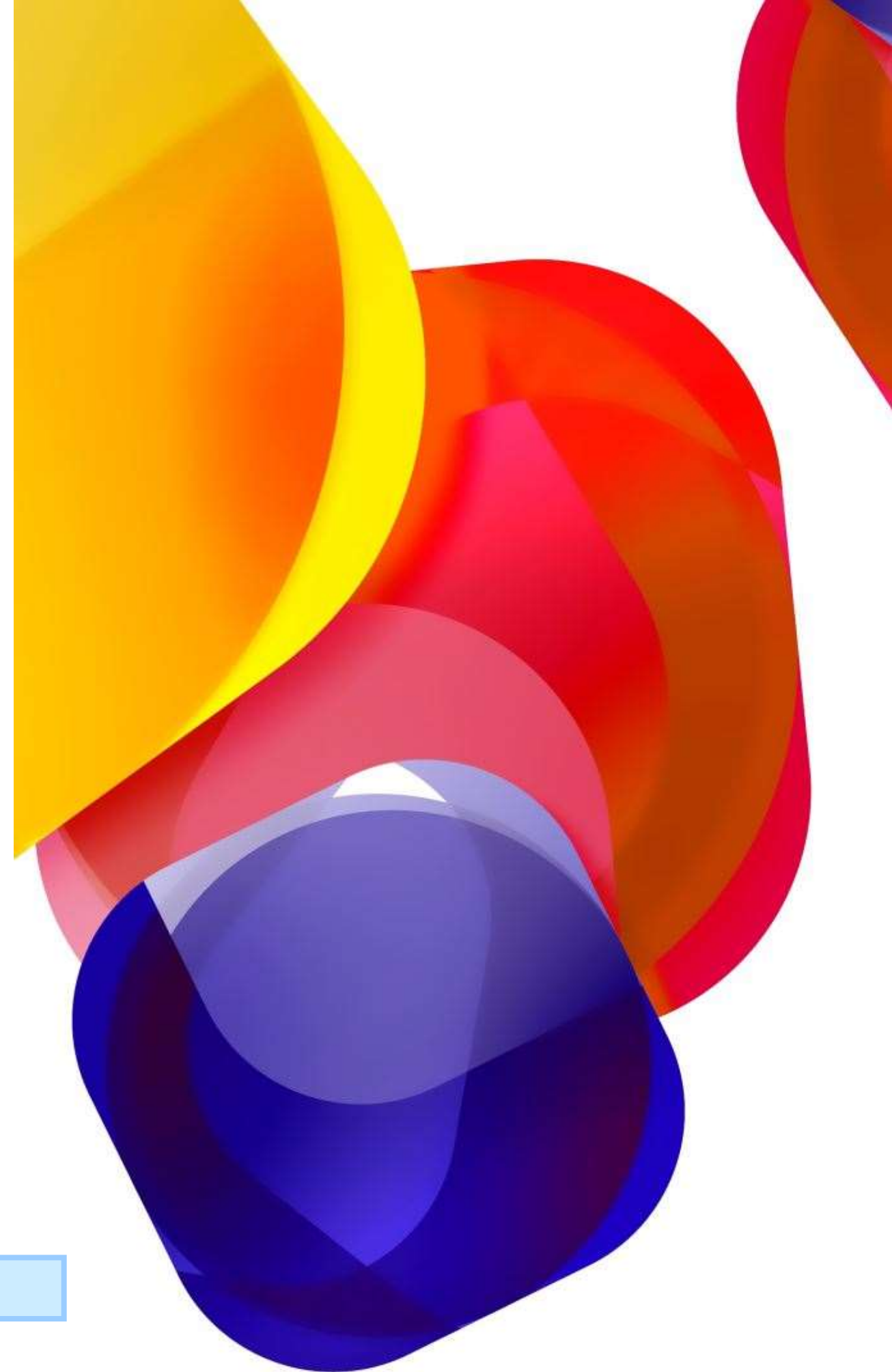


# Beyond ChatGPT: What Does ChatGPT-4 Add to HealthCare?

- Virtual Health Assistance
- Triage & Symptom Analysis
- Medication Adherence
- Data Analysis – Wearables
- Precision Patient Education
- Care Coordination
- Scheduling

<https://shorturl.at/afhy4>

Figure on next slide





**Prompt: I would like you to explain to me as a patient in a pictorial and understandable way what it means for me to have mitral regurgitation**

GPT-4: Think of the heart as a house with four rooms. The two rooms at the top (the atria) are where the blood begins its journey, and the two rooms at the bottom (the ventricles) are where the blood pumps out to the rest of the body.

Your mitral valve is like a door between one of the rooms at the top — the left atrium — and one of the rooms at the bottom — the left ventricle. When your heart beats, the mitral valve opens to allow blood to pass from the left atrium to the left ventricle, and then closes to prevent blood from returning back to the atrium.

Now, mitral valve regurgitation means that this door does not close as well as it should — it's slightly ajar. So, when the blood is pumped out of the ventricle, a little bit goes back into the atrium instead of flowing forward into the rest of the body.

It's a bit like when you have an incompletely closed door and the wind causes some of the air to enter back into the room, instead of flowing outside. But instead of wind and air, we are talking about the heart and the pumping of blood. So, the heart has to work harder to pump that 'escaping' blood, just as a fan would have to work harder to pump air out through a door which is ajar. But for the time being, as you have no symptoms, it looks like your heart is coping well with this extra effort.

**Figure 3: How ChatGPT-4 can easily and pictorially explain medical jargon to a patient.**

Source: <https://shorturl.at/afhy4>



# Companies Integrating ChatGPT



Nuance Communications	Medical note-taking	
Nabla	Transcribes video conversations	
Doximity	Prepares referrals & authorization requests	
Be My Eyes	Connects blind people with sighted volunteers	
ChatBeacon	Support through emotional assistance	
Bionic Health	Preventative health management	
Ferma.ai	Provides answers to life science questions	
MedMatch Network	Connects physicians and patients	
livewello	Genetic data analysis	
Epic Systems	Electronic medical records	
Litely	Fasting app for weight loss	
Dot Compliance	Quality management solution	
Kahun	A clinical reasoning tool	
Wondercise	Fitness content services	
Amazfit	Health management we	
EmoyaMed Private Hospital	Private hos	

“We Are Living in a Revolution Right Now”

The Medical Futurist: May 9, 2023

<https://medicalfuturist.com/17-healthcare-companies-that-already-integrated-chatgpt-infographic/>

Competency: Search & Summarization within an EHR?

--can you understand the intent behind an inquiry?

--can you communicate that in a query?

<http://tinyurl.com/5fdfhf2n>

# What Does NLP & a LLM feel like in EPIC?

Helen Waters, Meditech EVP & COO





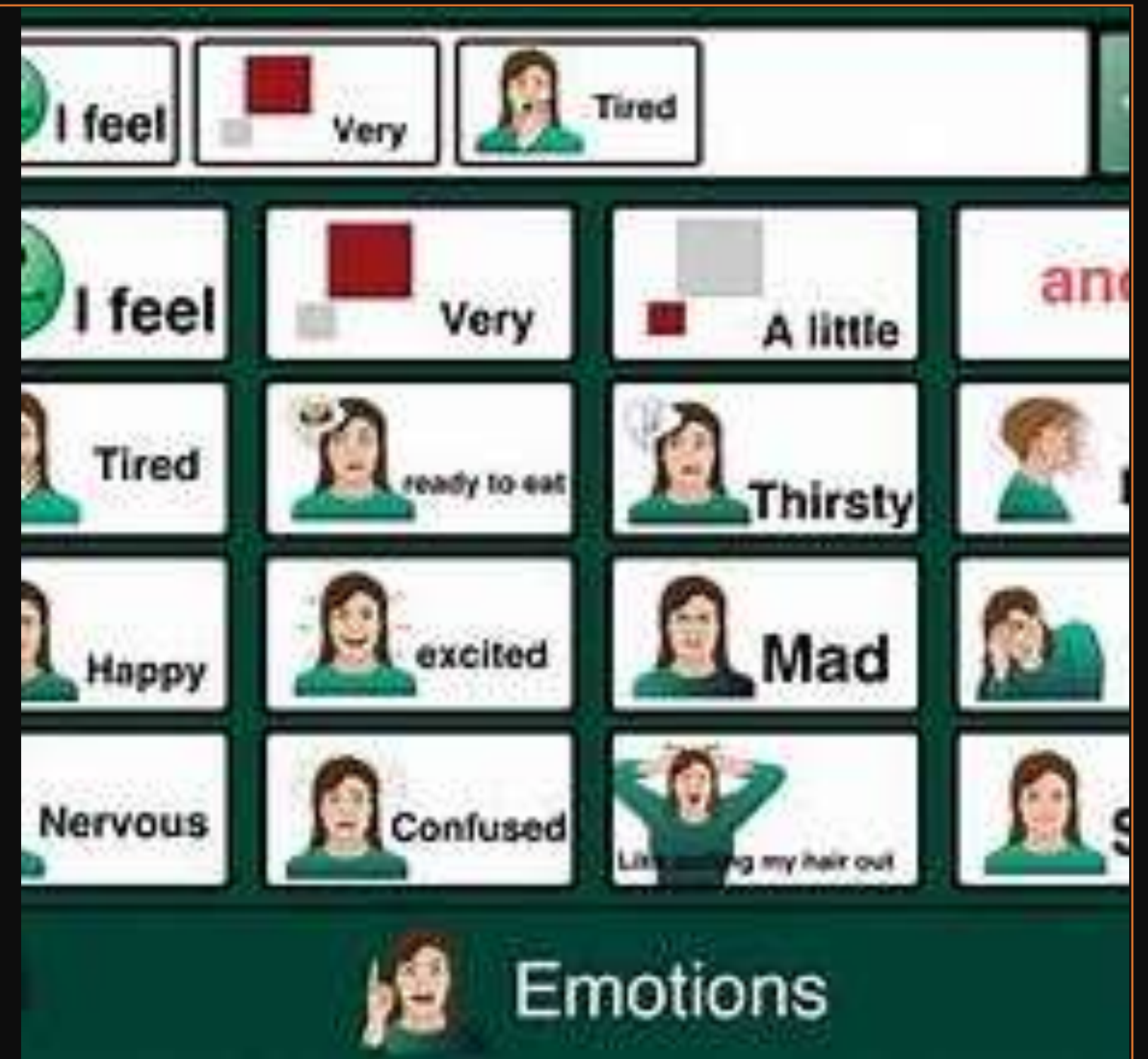
# What's Up with Cognitive Health?

<https://medicalfuturist.com/the-future-of-cognitive-health-this-is-how-digital-health-can-help>



# What More is Up with Cognitive Health?

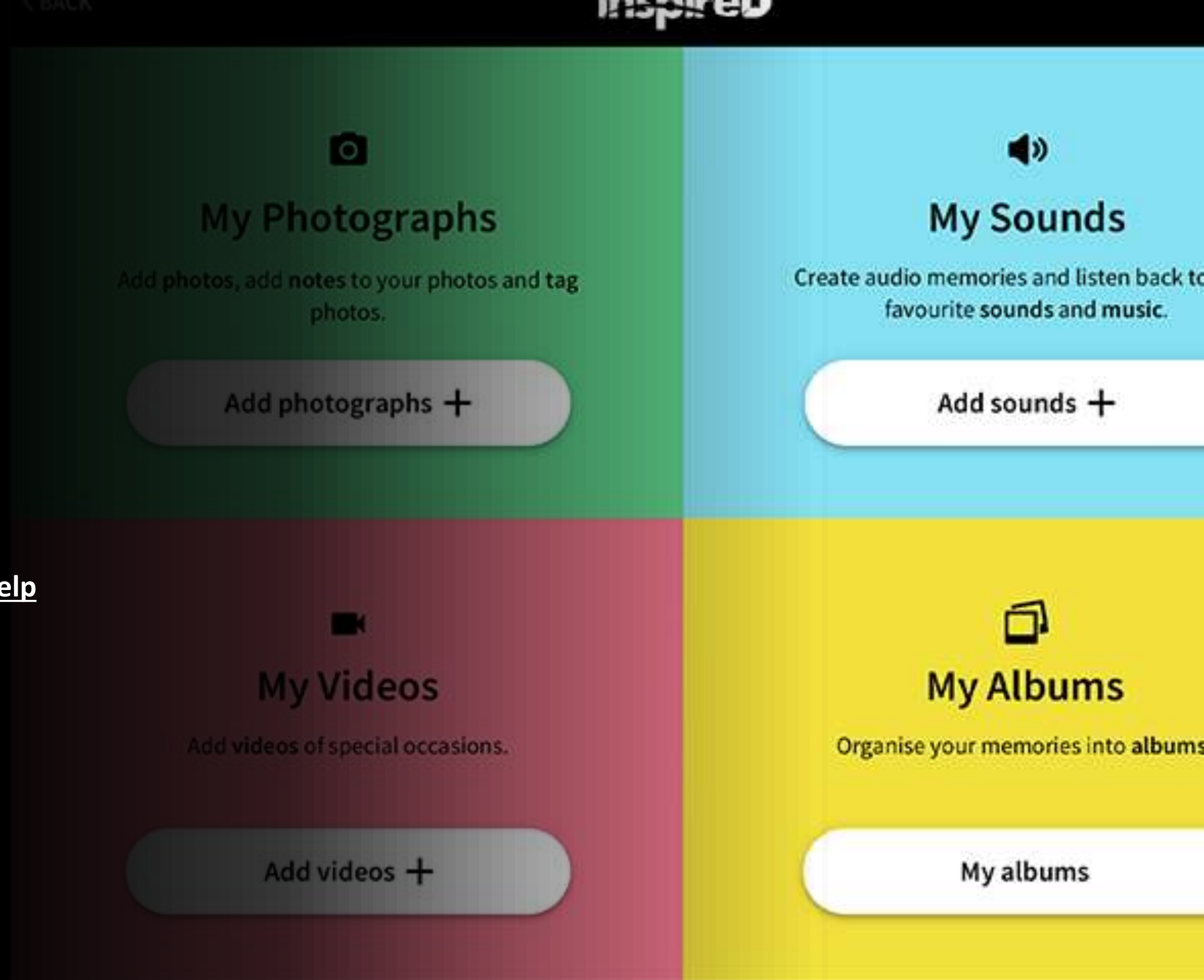
<https://medicalfuturist.com/the-future-of-cognitive-health-this-is-how-digital-health-can-help>





# What Else is Up with Cognitive Health?

<https://medicalfuturist.com/the-future-of-cognitive-health-this-is-how-digital-health-can-help>

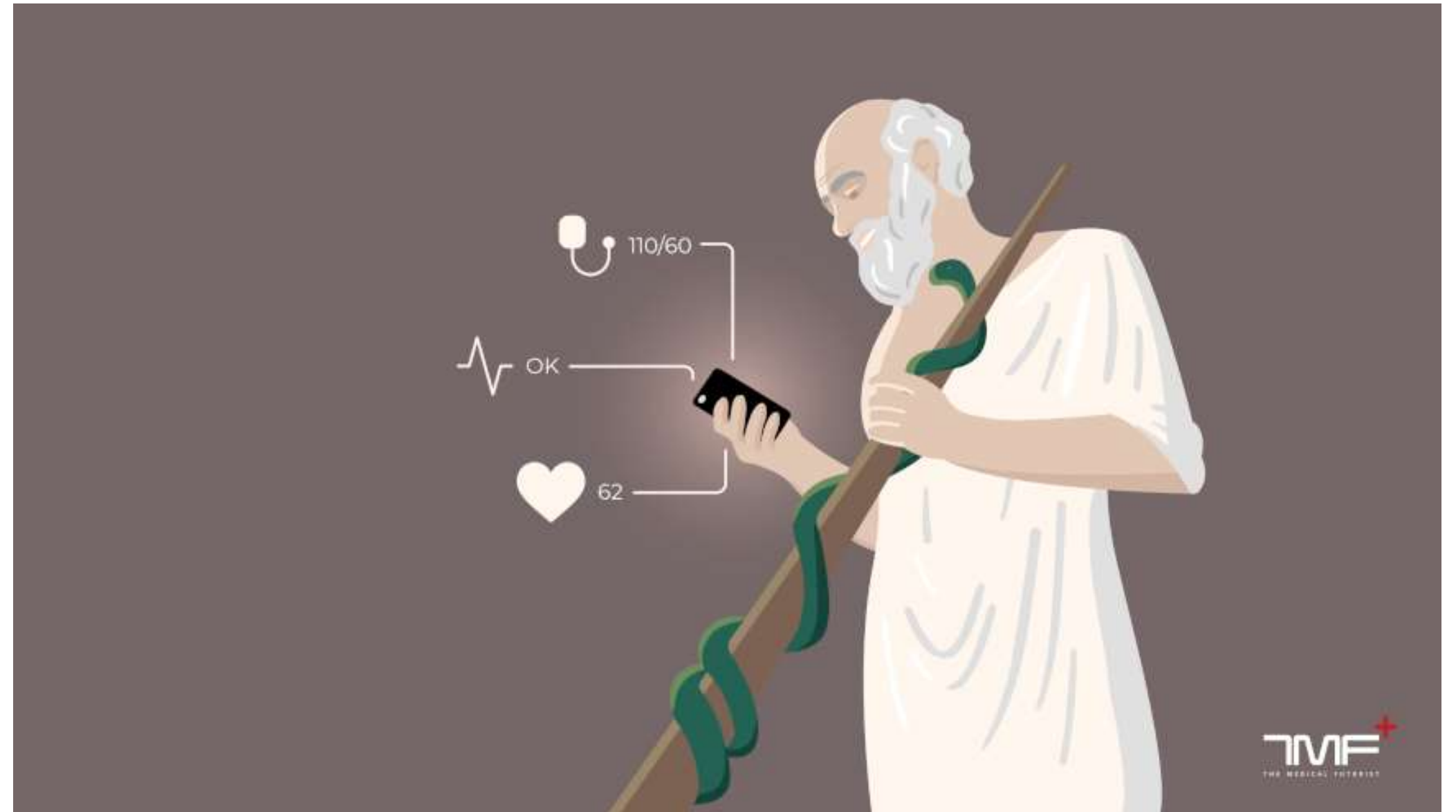







## AI steps up in healthcare: GPT-3.5 and 4 excel in clinical reasoning

# The Upgraded Hippocratic Oath



<https://medicalfuturist.com/why-an-upgraded-hippocratic-oath-is-needed-in-the-digital-era>





—

The ANA Story  
The first generalist  
provider organization  
to adopt PH policy!

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<https://tinyurl.com/2c7vusmx>



A GUIDE  
TO ARTIFICIAL  
INTELLIGENCE  
IN HEALTHCARE



2021  
edition

DR. BERTALAN MESKÓ  
THE MEDICAL FUTURIST



*From the desk of Bertalan Mesko MD  
The Medical Futurist*

<https://medicalfuturist.com>



Free Virtual Event

MARCH 24, 2022 / 12:00 – 2:00 PM EST

# AI AND MACHINE LEARNING FOR HEALTH CARE DELIVERY



REGISTER & GET FREE CASE STUDY →

## New England Journal of Medicine Catalyst

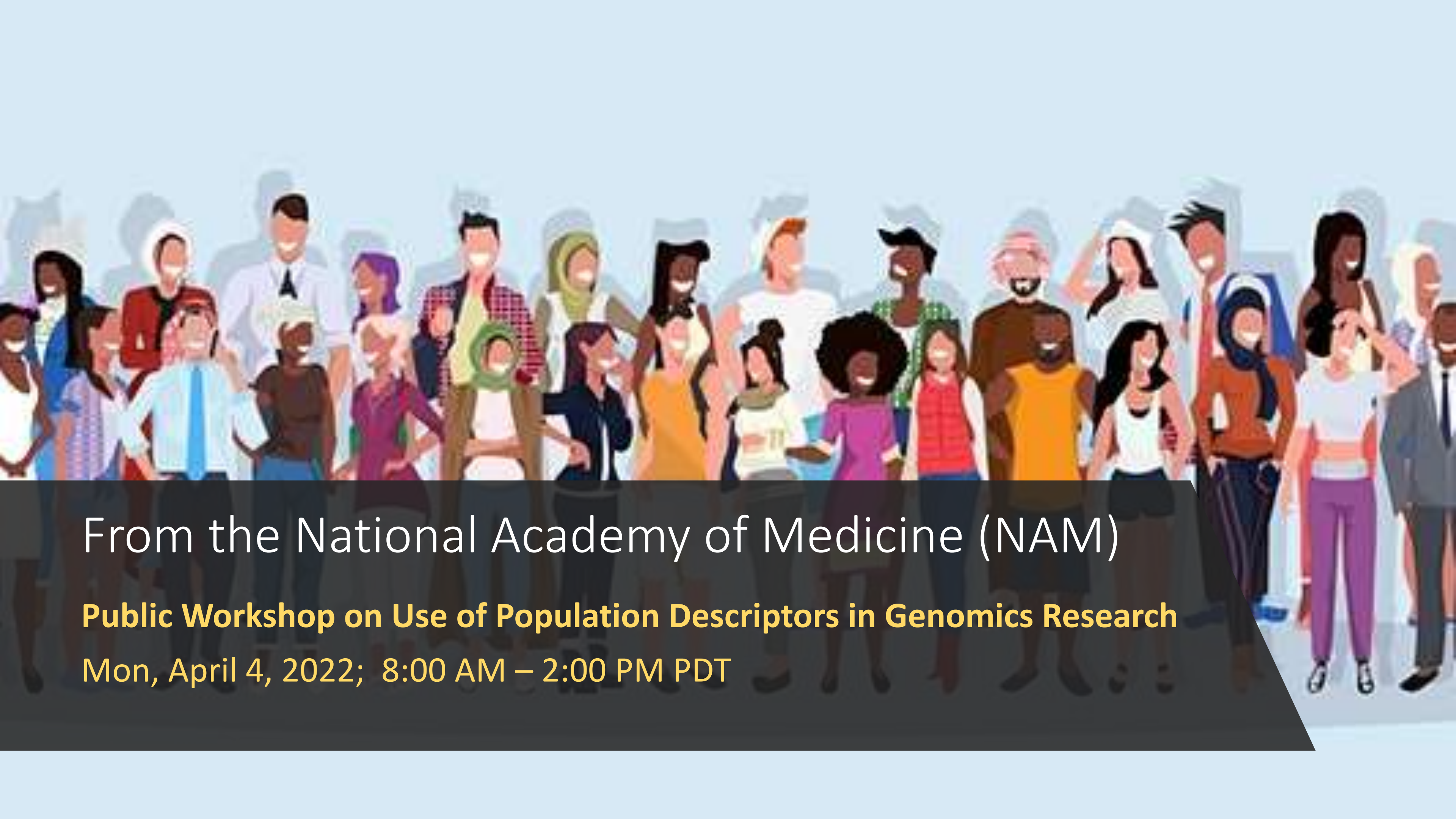
[Preventing Delayed and Missed Care by Applying Artificial Intelligence to Trigger Radiology Imaging Follow-up](#)

[How Health Systems Decide to Use Artificial Intelligence for Clinical Decision Support](#)

[Growing Use and Confidence in Artificial Intelligence for Care Delivery](#)

[Design, Implementation, and Clinical Impact of a Machine Learning-Assisted Intervention Bundle to Improve Opioid Prescribing](#)



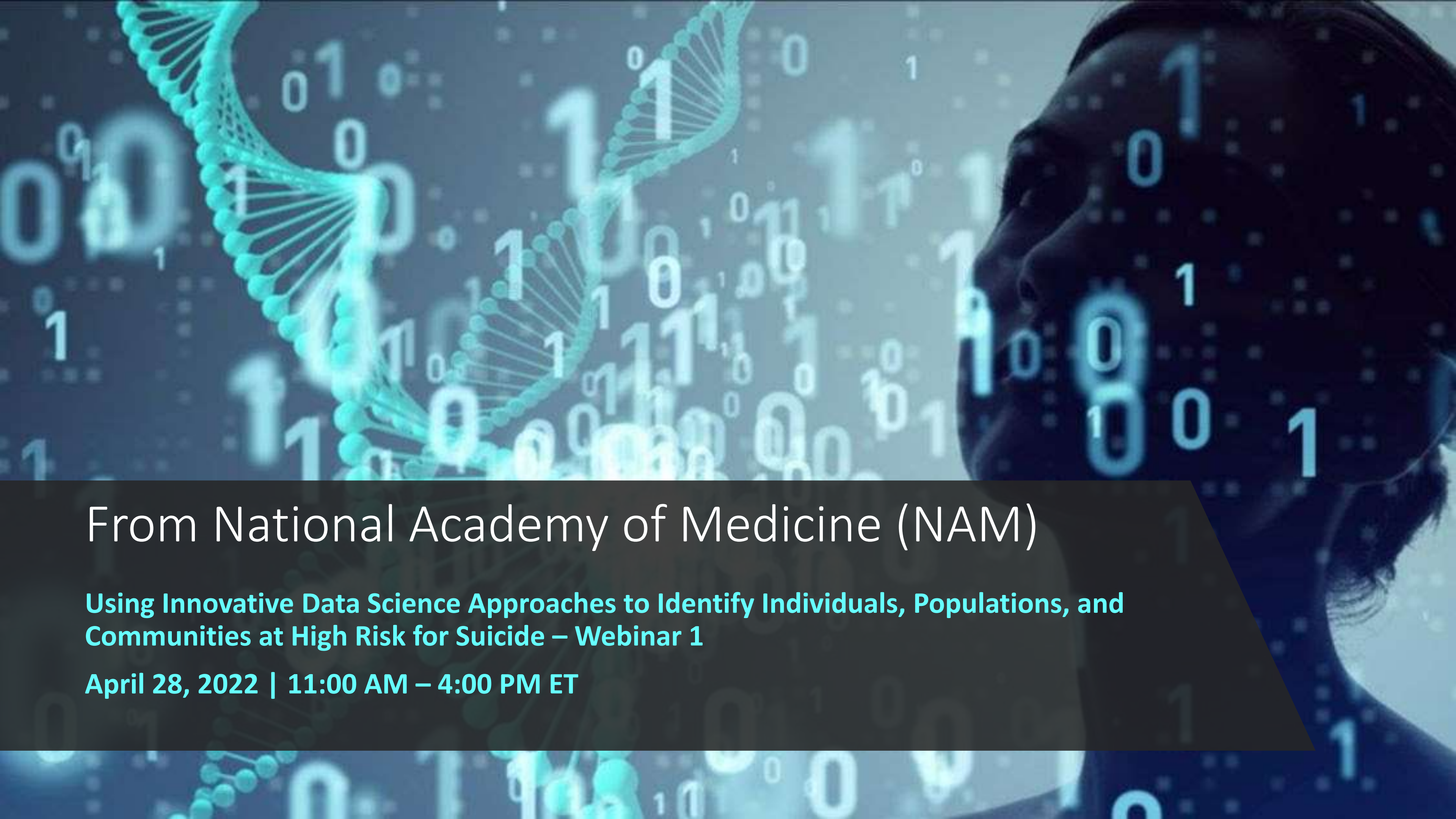


From the National Academy of Medicine (NAM)

**Public Workshop on Use of Population Descriptors in Genomics Research**

Mon, April 4, 2022; 8:00 AM – 2:00 PM PDT





# From National Academy of Medicine (NAM)

**Using Innovative Data Science Approaches to Identify Individuals, Populations, and Communities at High Risk for Suicide – Webinar 1**

**April 28, 2022 | 11:00 AM – 4:00 PM ET**

„A.I. will not replace medical professionals. However, those medical professionals that use A.I. will replace those that don't.”

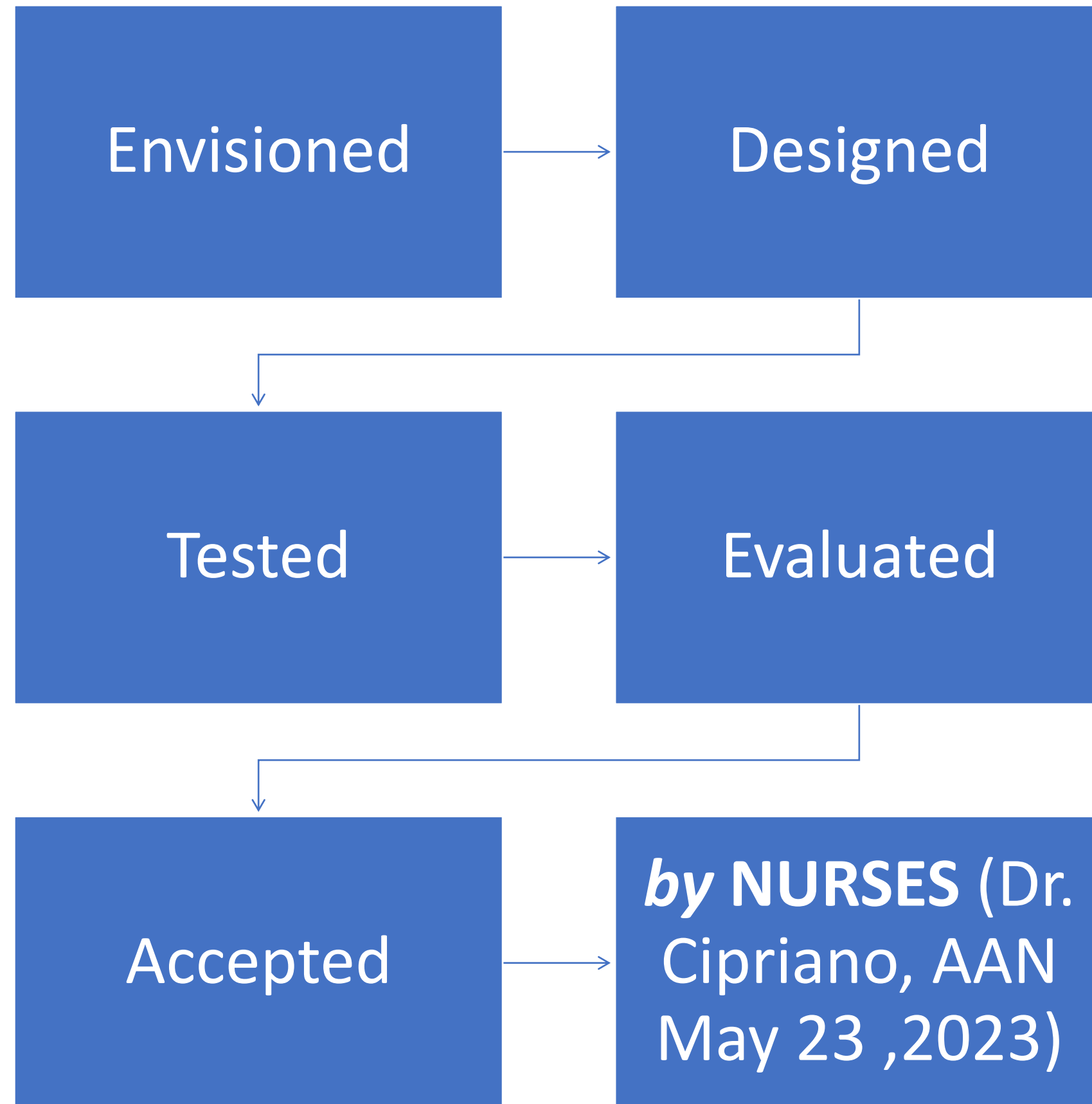
Dr. Bertalan Meskó



The Medical Futurist: <https://medicalfuturist.com>



Lastly--  
Technology  
in Health  
Care Must  
be





# Key Source Materials, Webinar

ABC News. (2023, March 17). *OpenAI CEO, CTO on risks and how AI will reshape society* [Video]. YouTube. <https://rb.gy/y2zxr>

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## Q&A

# Keynote Roles and Functions for Nursing Practice

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**Moderator: Suzanne Bakken, PhD, RN, FAAN, FACMI, FIAHSI**

- **Mary Anne Schultz, PhD, MBA, MSN, RN, FAAN**

**We'll be right back with our next session**

# **PANEL 1**

## **Precision Health in the Real-World**

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**Moderator: Kim Glassman, PhD, RN, NEA-BC, FAONL, FAAN**

- **Danielle Fournier, DNP, APRN, AGPCNP-BC, AOCNP**
- **Marilyn Hammer, PhD, DC, RN, FAAN**
- **Kevin Whitney, DNP, RN, EMT-P**

# **PANEL 1**

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- **Kevin Whitney, DNP, RN, EMT-P**



# Presentation

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**Danielle Fournier**

DNP, APRN, AGPCNP-BC, AOCNP

# Research to Practice: Precision Health Elevating Patient-Centered Nursing Care

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**Moderator: Kimberly Glassman, PhD, RN, NEA-BC, FAONL, FAAN**  
Dean, Mount Sinai, Phillips School of Nursing  
New York, NY, USA

**Danielle Fournier, DNP, APRN, AGPCNP-BC, AOCNP**  
Department of Thoracic Surgery  
MD Anderson Cancer Center, Houston, TX, USA

**Marilyn J. Hammer, PhD, DC, RN, FAAN**  
Phyllis F. Cantor Center for Research,  
Dana-Farber Cancer Institute, Boston, MA, USA

**Kevin Whitney, DNP, RN, EMT-P**  
Mass General Brigham Healthcare at Home  
Boston, MA, USA

# Disclosures

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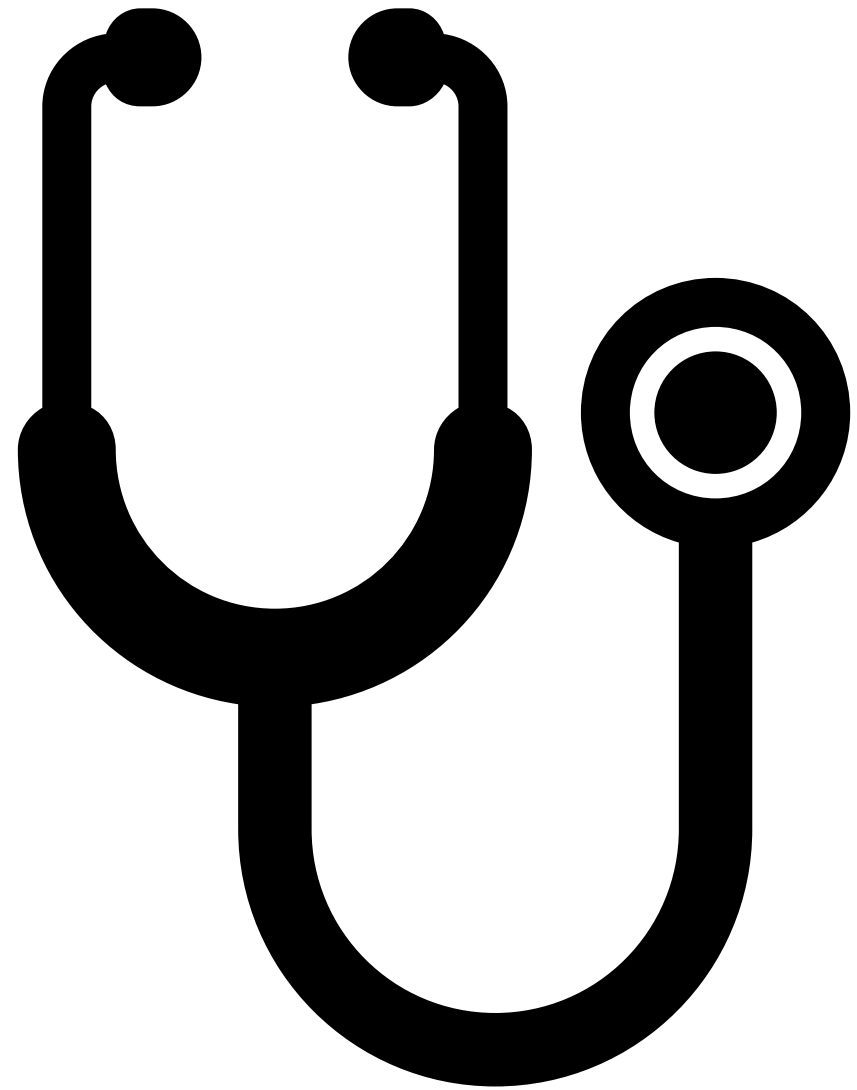
- Dr. Fournier has no financial disclosures.
- Dr. Hammer has research funding support from GSK through Pack Health, A Quest Diagnostics Company. Research is also supported by the Mittelman Foundation.
- Dr. Whitney has no disclosures.



# Presentation Objectives

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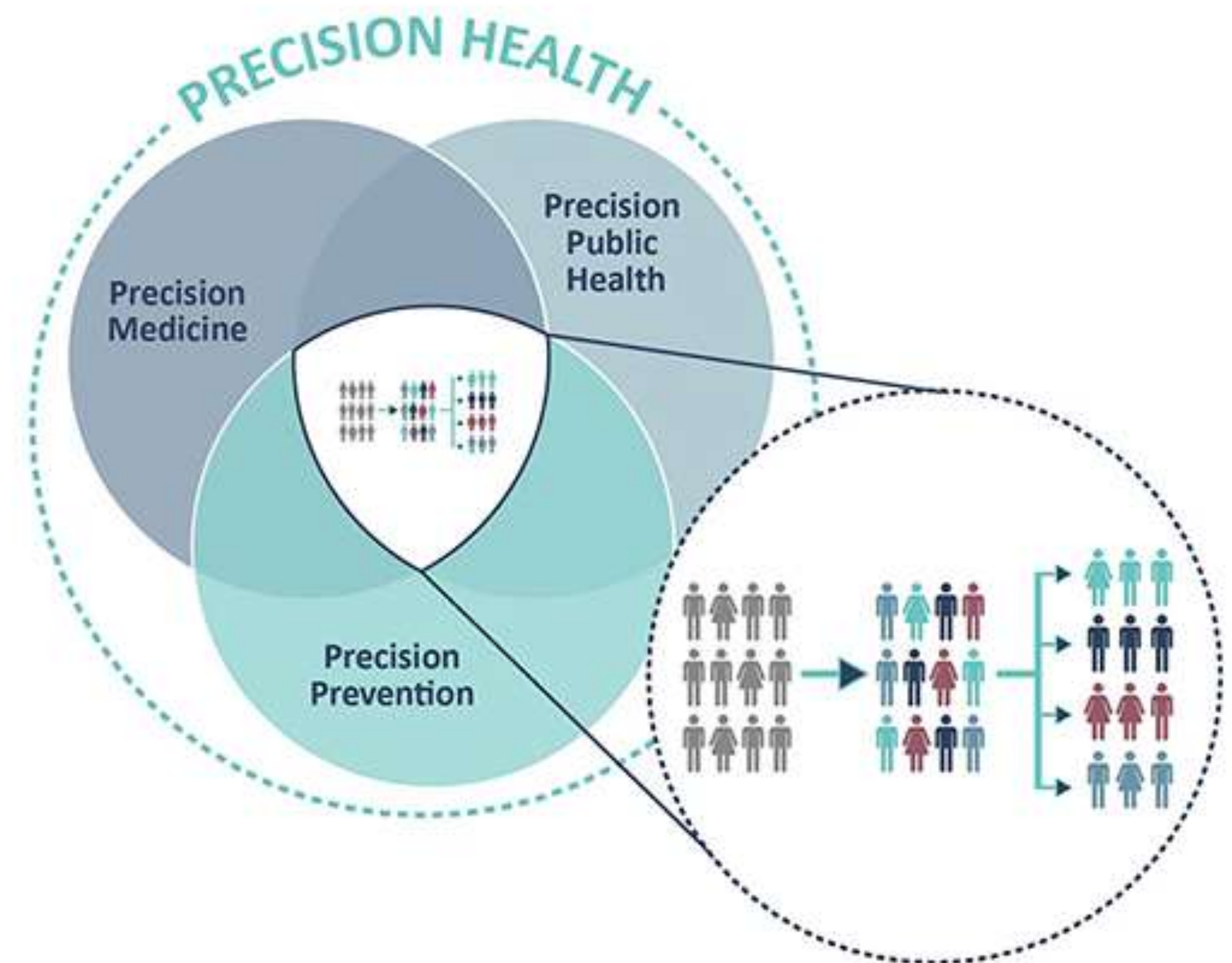
- Discuss clinical implications of precision health on the role of nursing and practice
- Evaluate nurse scientist led studies in precision health and the translation of findings into clinical nursing practice
- Describe key components of home hospital and home care models including the use of digital health technology and remote patient monitoring



# **Precision Health and Nursing Clinical Practice**

# Precision Health

- Innovative approach to tailor healthcare to the need of the individual
- Best intervention to the right patient at the right time
- Based on:
  - Genetics, genomics, -omics
  - Lifestyle
  - Social
  - Economic
  - Cultural and environmental influences

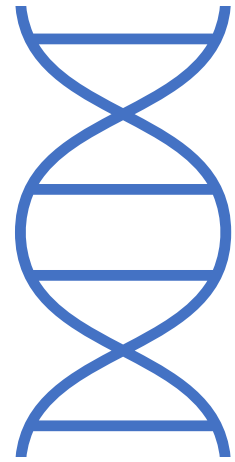




# The Five Ps of Precision Health

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Prediction



Prevention



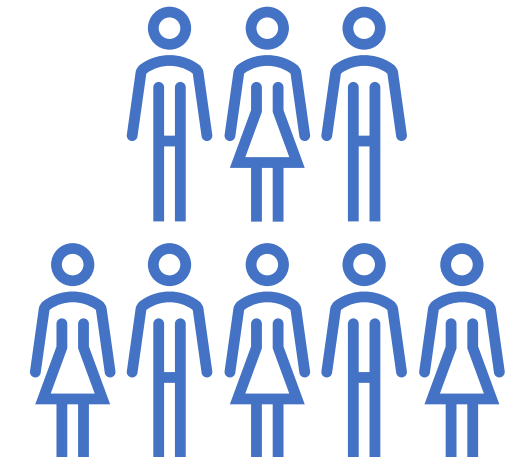
Participation



Personalization



Population









# Nursing's Role in Precision Healthcare

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- Nurses have a significant role in promoting, protecting, and optimizing health as part of multidisciplinary healthcare team
- Nursing clinical practice focuses on the personalization of care to enhance safety and clinical outcomes
- Integration of genomic information is paramount to nursing clinical practice in all practice settings
- Direct impact on risk assessment, health promotion, disease prevention, and patient education



# Implications for Nursing Clinical Practice



Obtaining a comprehensive family history



Interpreting and discussing genomic test results



Pharmacogenomic-based drug administration



Patient education



Care coordination

# Comprehensive Family History

- Risk assessment starts with obtaining a complete family history
- Accurate family health history can:
  - Help to identify personal and family risk for genomic-influenced conditions
  - Recognize the need for genomic testing
- Referral to genetics professional for further discussion and testing



# Interpretation of Genomic Results

---

- Nurses are a key resource to help patients understand their genomic testing results
  - Discuss implications for disease management
  - Encourage testing of at-risk family members
  - Facilitate referral to genetics professional or specialist as appropriate
- Connect patients with appropriate resources based on results
- Ensure communication of test results aligns with patient and family preferences
- Provide psychosocial support



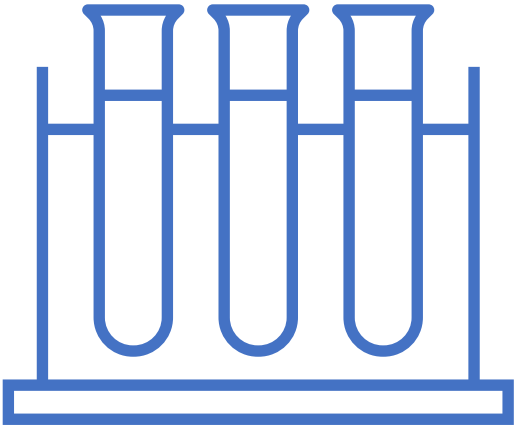
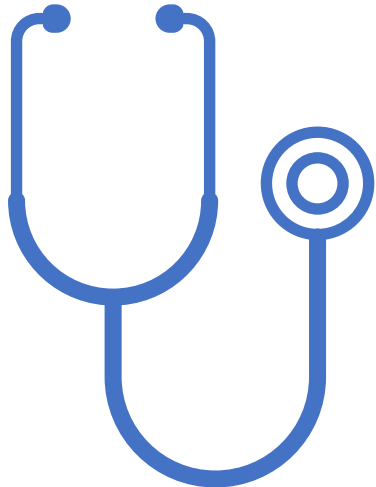
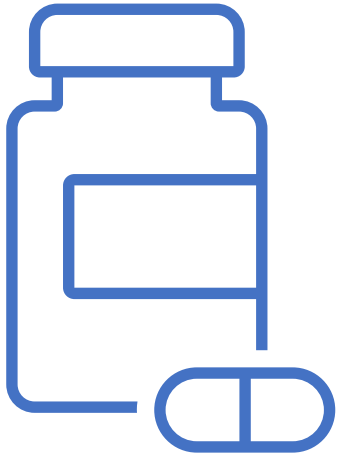
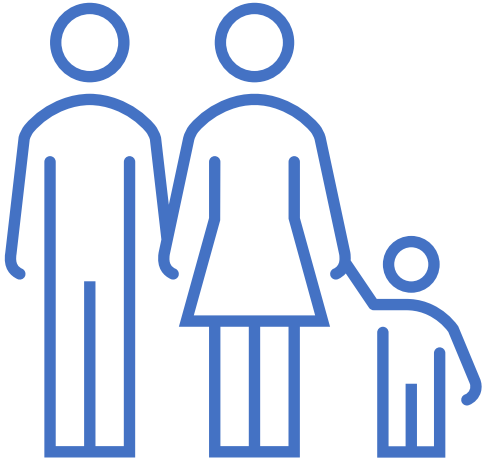






# Pharmacogenomic-Based Drug Administration

- Aimed at identifying the genomic factors that influence the efficacy or toxicity of drug therapy
- As of 2023: 570 drugs approved by the FDA with pharmacogenomic information included in drug labelling
- Potential to improve clinical outcomes
- Nurses must understand the role for pharmacogenomic testing and implications for patient care

# Patient Education

					
Risks, benefits and limitations of genomic testing	Role of genomic testing in disease management	Interventions needed based on test results	Implications for family members	Lifestyle modifications based on results	Ethical, legal and social implications

# Coordination of Care

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- Patient-centered approach to assess and meet the needs of patients while helping them navigate the healthcare system
- Bridging gaps along the care pathway through information transfer
  - Among members of one healthcare team
  - Between patient care teams
  - Between patients and caregivers
  - Across healthcare settings





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# Presentation

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**Marilyn Hammer**  
PhD, DC, RN, FAAN



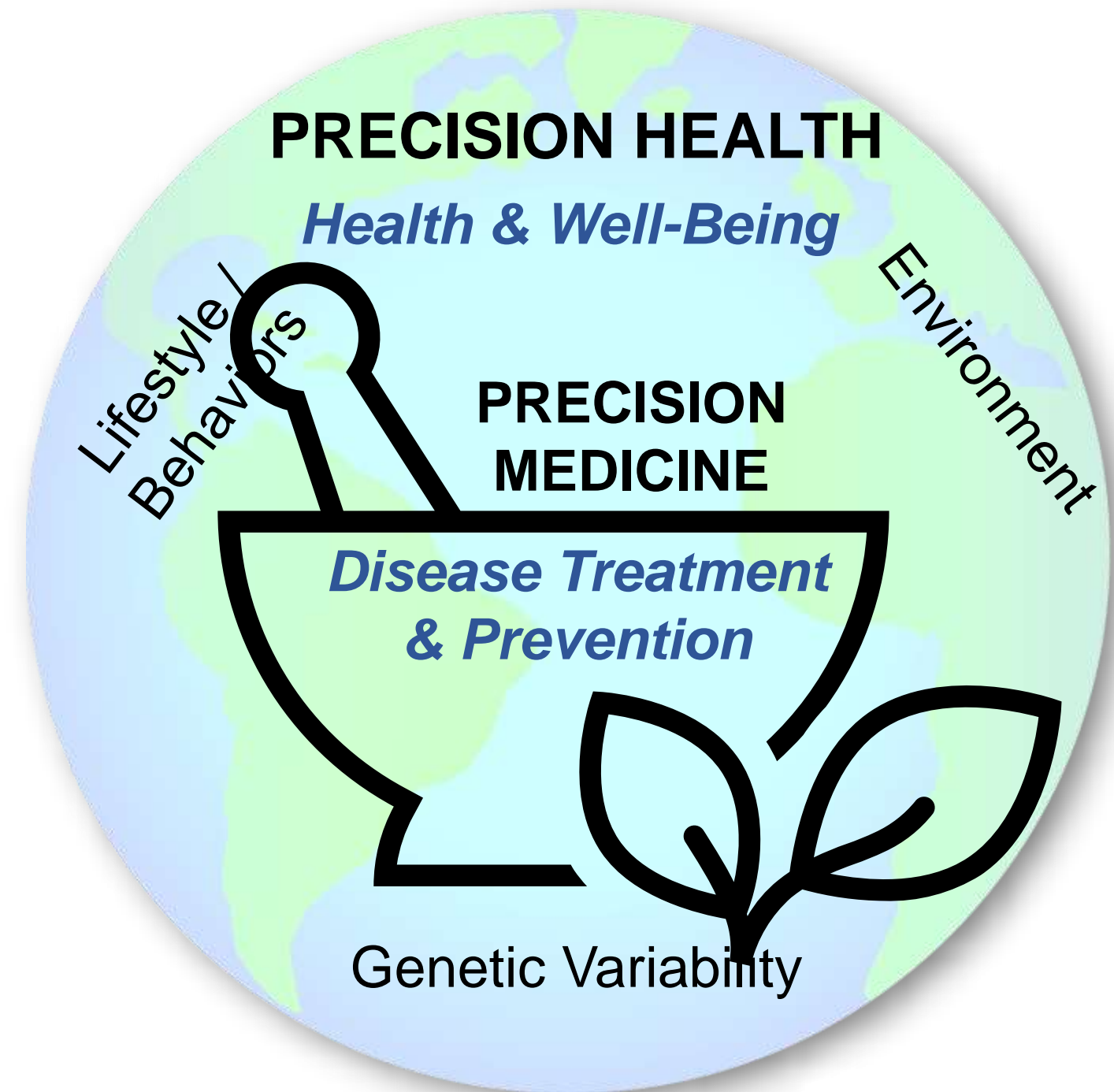
# **Precision Health & Nursing Science**



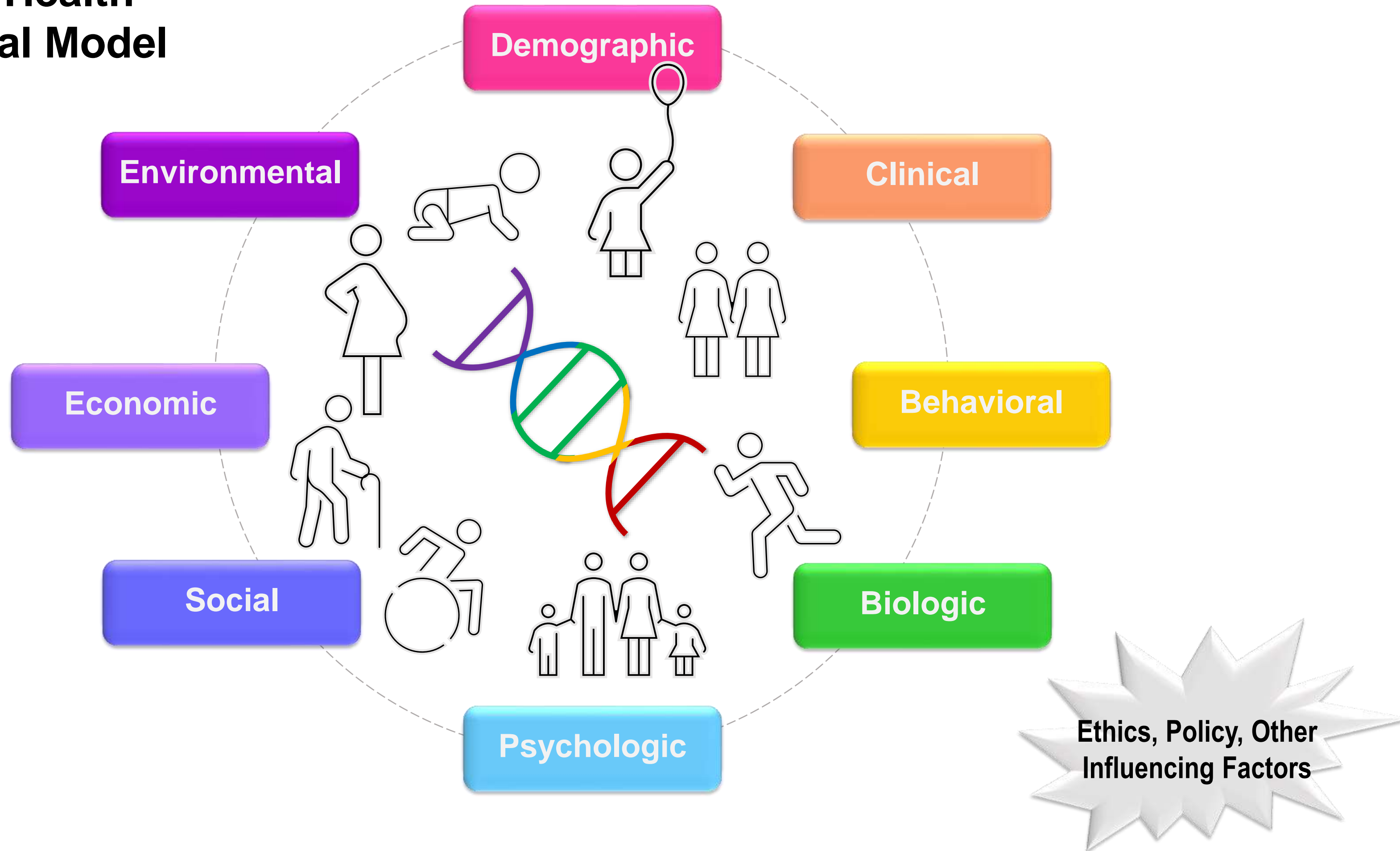
# Precision Medicine / Precision Health

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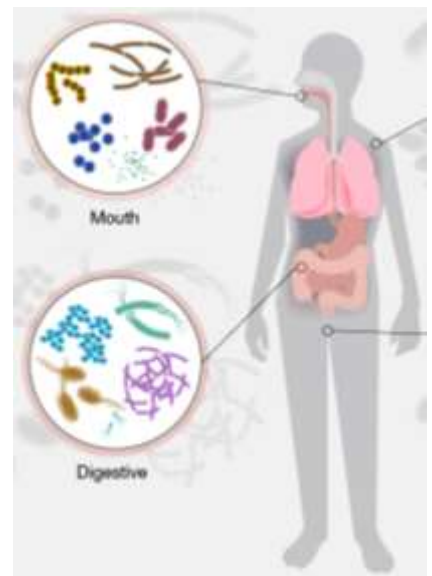
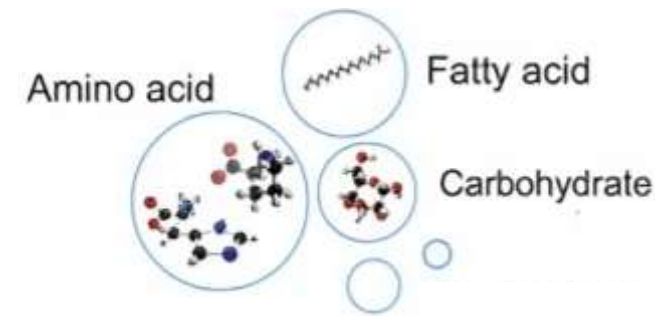
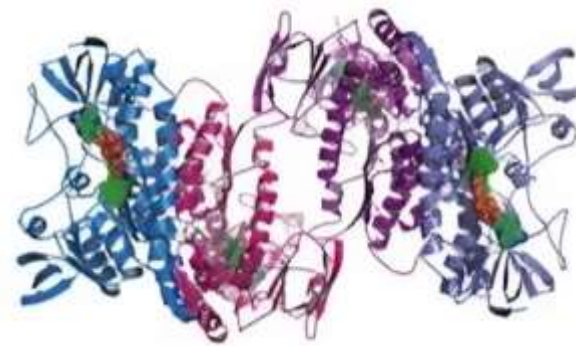
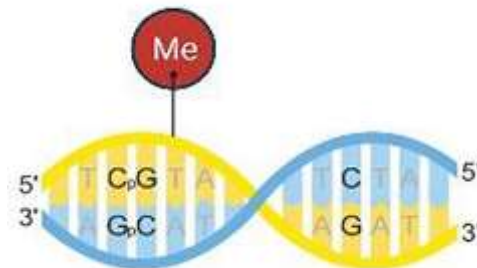
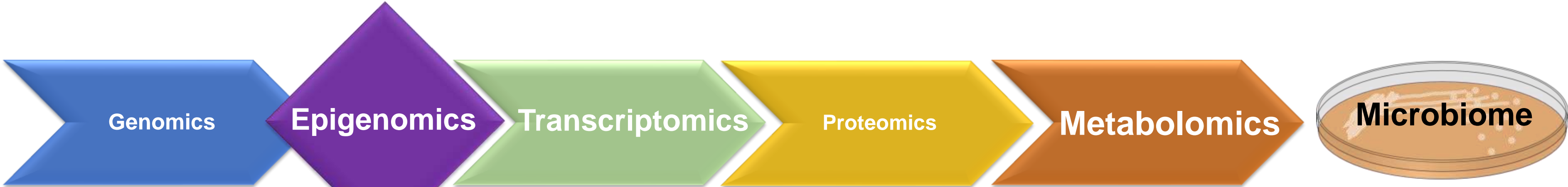
- Precision Medicine
  - Disease treatment and prevention
    - Individual genetic variability
    - Lifestyle / behaviors
    - Environment
  - “Predictive, preventive, personalized, participatory”
- Precision Health
  - Optimizing health and well-being
    - Individual genetic variability
    - Environment
    - Lifestyle / behaviors



# Precision Health Conceptual Model



# Omics-Based Approaches to Studying Symptoms and Other Patient Outcomes





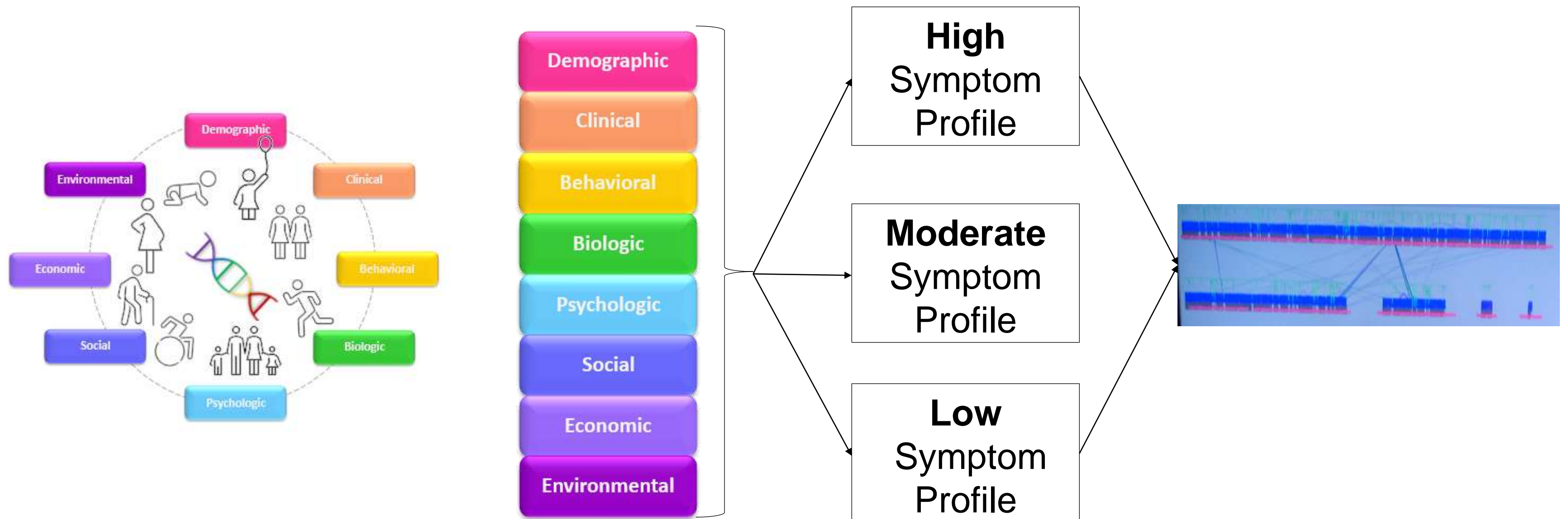
# Background

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- Individuals with cancer receiving chemotherapy experience 10 to 12 co-occurring symptoms
- The most common co-occurring symptoms
  - Pain – 39.4% - 66.4%
  - Fatigue – 25% - 99%
  - Sleep disturbance – 17% - 70%
  - Depression – 8% - 58%
- Large amounts of inter-individual variability in occurrence rates of these four symptoms
- Inflammation may be a common underlying pathway

# Study Schematic

- Latent profile analysis → identify subgroups of patients with distinct symptom profiles
- Evaluate for perturbations in inflammatory pathways between patients who had low and high levels of all four symptoms (pain, fatigue, sleep disturbance, depression)



# Methods

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- Descriptive longitudinal study
- Recruitment from seven outpatient oncology clinics
- 1,343 patients with breast, lung, GI, or GYN cancer
  - $\geq 18$  years old
  - Scheduled for 2 or more cycles of chemotherapy
- Worst pain rated on a 0 to 10 numeric rating scale
- Lee Fatigue Scale – evening fatigue ( $\geq 5.6$ )
- General Sleep Disturbance Scale ( $\geq 43.0$ )
- Center for Epidemiologic Studies Scale ( $\geq 16.0$ )



# Estimated Means for Symptom Severity Scores (n=1,140)

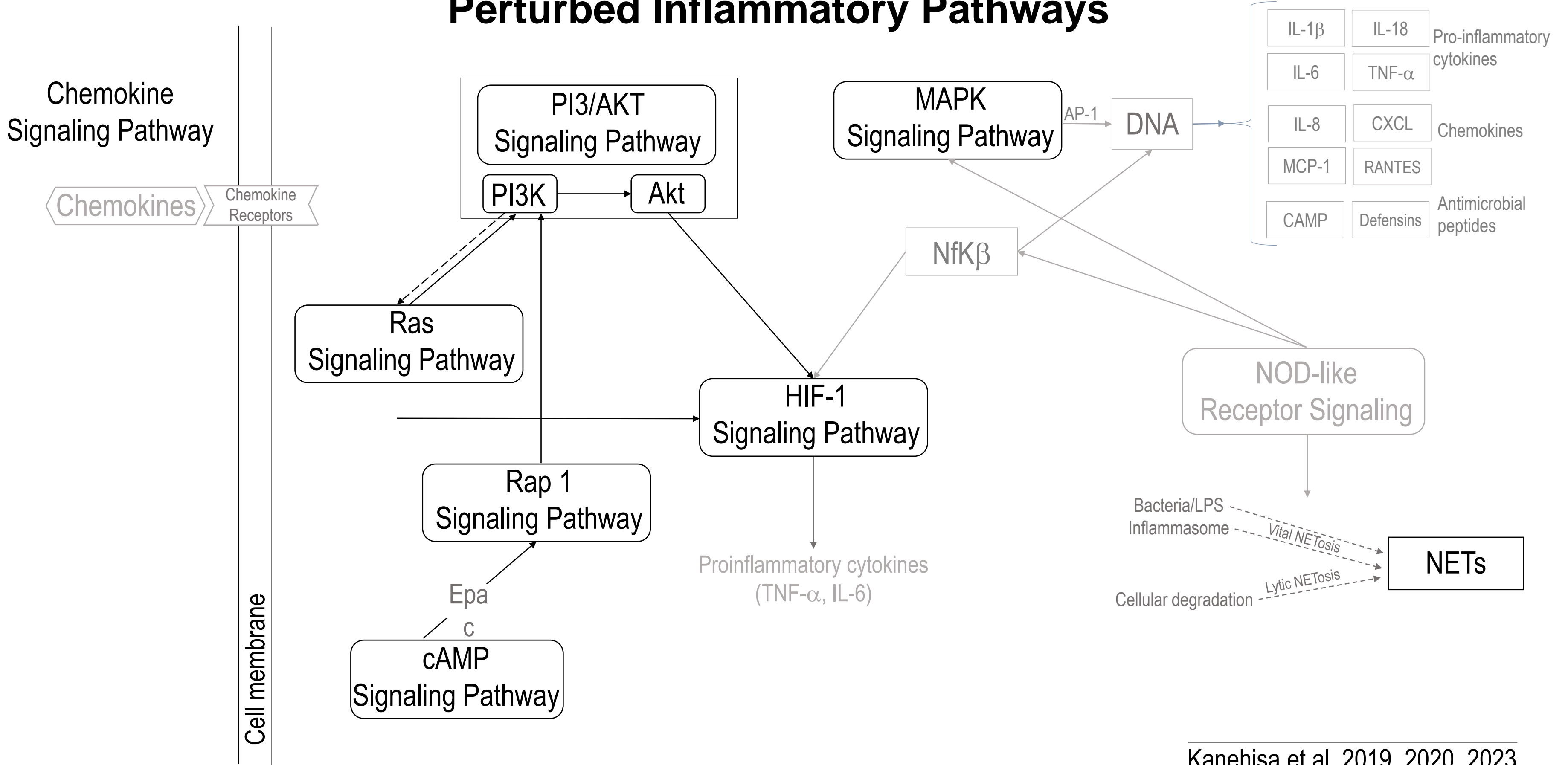
<b>Symptom</b>	<b>Low Class 44.0%</b>	<b>Moderate Class 45.1%</b>	<b>High Class 10.8%</b>
Any pain	59.8%	80.4%	94.8%
For patients with pain – worst pain intensity Moderate = 4 to 6 Severe = $\geq 7$	5.098	6.377	7.243
Evening fatigue ( $\geq 5.6$ )	4.188	6.061	7.048
Sleep disturbance ( $\geq 43.0$ )	37.170	61.423	77.609
Depression ( $\geq 16.0$ )	6.471	14.256	32.277

# Omic Analysis: Perturbed Pathways

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- Inflammatory (select pathways)
  - Chemokine signaling (hsa04062)
  - Mitogen-activated protein kinase (MAPK) (hsa04010)
  - Hypoxia inducible factor-1 (hsa04066)
  - Neutrophil extracellular trap (NET) formation (hsa04613)
  - Ras (hsa04014) and Rap1 (hsa04015) signaling pathways
  - PI3K/Akt (hsa04151)
- Endocytosis (hsa04144)
- Phagosome (hsa04145)

# Perturbed Inflammatory Pathways





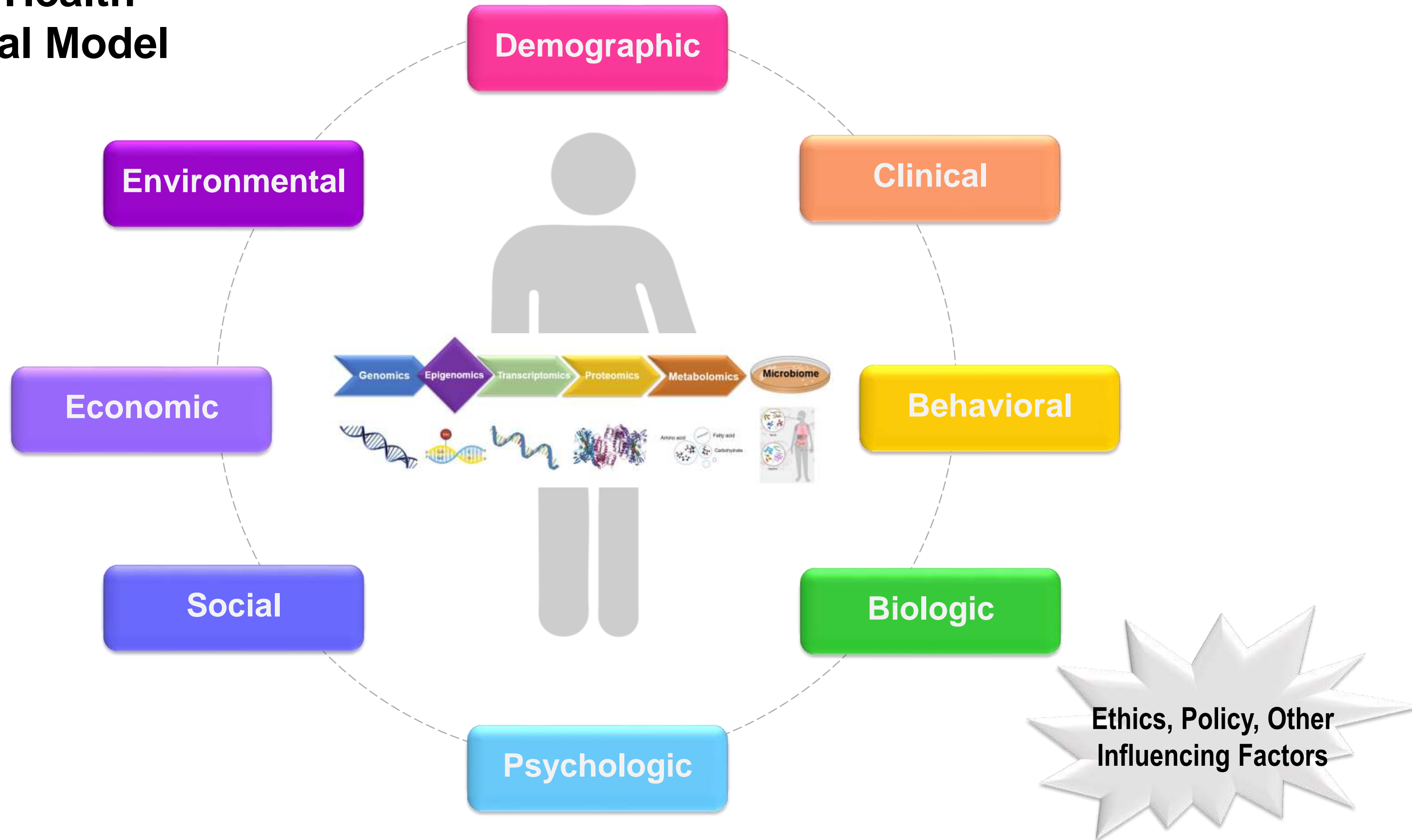
# Translation to Clinical Practice

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- Understanding associations between symptom cluster and underlying inflammatory mechanisms
- Monitoring and managing multiple co-occurring symptoms may mitigate continuous symptom burden in people undergoing treatment for cancer



# Precision Health Conceptual Model



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# Presentation

**Note: These slides are omitted as we do not have permission to distribute**

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**Kevin Whitney**  
DNP, RN, EMT-P

# Q&A

## PANEL 1

# Precision Health in the Real-World

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**Moderator: Kim Glassman, PhD, RN, NEA-BC, FAONL, FAAN**

- **Danielle Fournier, DNP, APRN, AGPCNP-BC, AOCNP**
- **Marilyn Hammer, PhD, DC, RN, FAAN**
- **Kevin Whitney, DNP, RN, EMT-P**

# LUNCH BREAK

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The webinar will continue at 12:30pm Eastern Time



# PANEL 2

## Precision Health in the Real-World

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**Moderator: Rebecca Freeman, PhD, RN, PMP**

- **Cathy Turner, BSN, MBA, RN-BC**
- **Anna Schoenbaum, DNP, MS, RN-BC, FHIMSS**
- **Johnston Thayer, MBA, RN**

# Presentation

---

**Cathy Turner**  
BSN, MBA, RN-BC



# The Future of Medicine Means Getting Personal

**Cathy Turner, MBA, RN, NI-BC**  
Chief Marketing and Nurse Executive  
Medical Information Technology, Inc.

**CONSUMER TRENDS**

**GENOMICS AT THE  
POINT OF CARE**

**COMMUNITY  
COMMITMENT**



# Genomics

## Consumer Trends

**Direct-to-consumer genetic**  
testing is spurring patient interest

---

**The role genes play in health**  
is becoming more widely  
understood

---

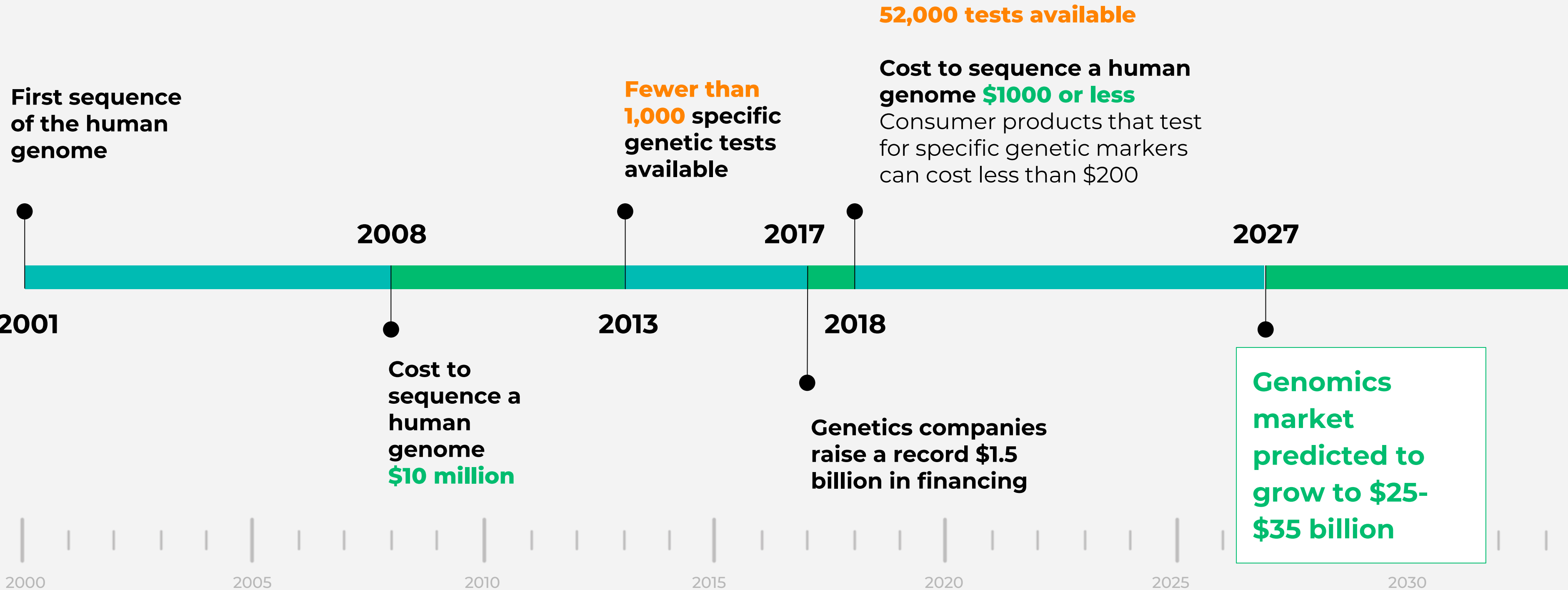
**Patients expect their providers**  
to understand their unique  
genetic profile and factor it into  
decision making





# Genomics

# What's now and what's next





**The fastest growing field**  
in healthcare today

**90% of Health Systems**  
are already ordering genetic testing today.



# Genomics

*at the Point of Care*



## IMPORT & CONSUME

genetic test results from outside labs and other facilities



## STORE & DISPLAY

key genetic data directly within patient records



## INFORM & GUIDE

at the point of care with clinical decision support

EMPOWERS  
**CLINICIANS**

# Genomics

*at the Point of Care*



## PERSONALIZED MEDICINE

Improve efficacy and reduce risk based on unique genetic profiles



## CUTTING-EDGE CARE

In the convenience of your own *community* with your own *physicians*



## IMPROVED OUTCOMES

The right therapies, at the right time, for the right patient

EMPOWER YOUR  
**PATIENTS**



# GENOMICS AND YOUR BOTTOM LINE

## EMPOWER YOUR ORGANIZATION

### **Boost Clinician Satisfaction**

Provide a more intuitive, efficient solution that saves physicians time and reduces cognitive load

### **Increase Patient Loyalty**

Keep patients from going elsewhere for genetic testing and follow-up care

### **Build Trust and Confidence**

Throughout your organization with contemporary, state-of-the-art technology





Order placed for genetic test in Expanse



Specimen is collected and sent to reference lab



Reference Lab conducts test and returns results



Expanse imports and parses data directly into patient record



Results are displayed intuitively within patient chart



Interpretation & guidance is provided  
with embedded Clinical Decision Support



# Expanse Genomics

## Ordering of genetic tests

the same way they place other orders



### ORDERING

Genetic tests ordered out of Expanse



## Reference labs return results

which are parsed and stored in a new LIS Genetics application



Reference  
Lab A

Reference  
Lab A

Reference  
Lab A

Reference  
Lab A



## Genetics application

stores selective results and makes selective genetic markers available to the Expanse EHR



### EXPANSE GENETICS APPLICATION

A new LIS application within Expanse Genomics



## Patient chart

includes enhanced chart displays for key genetic markers

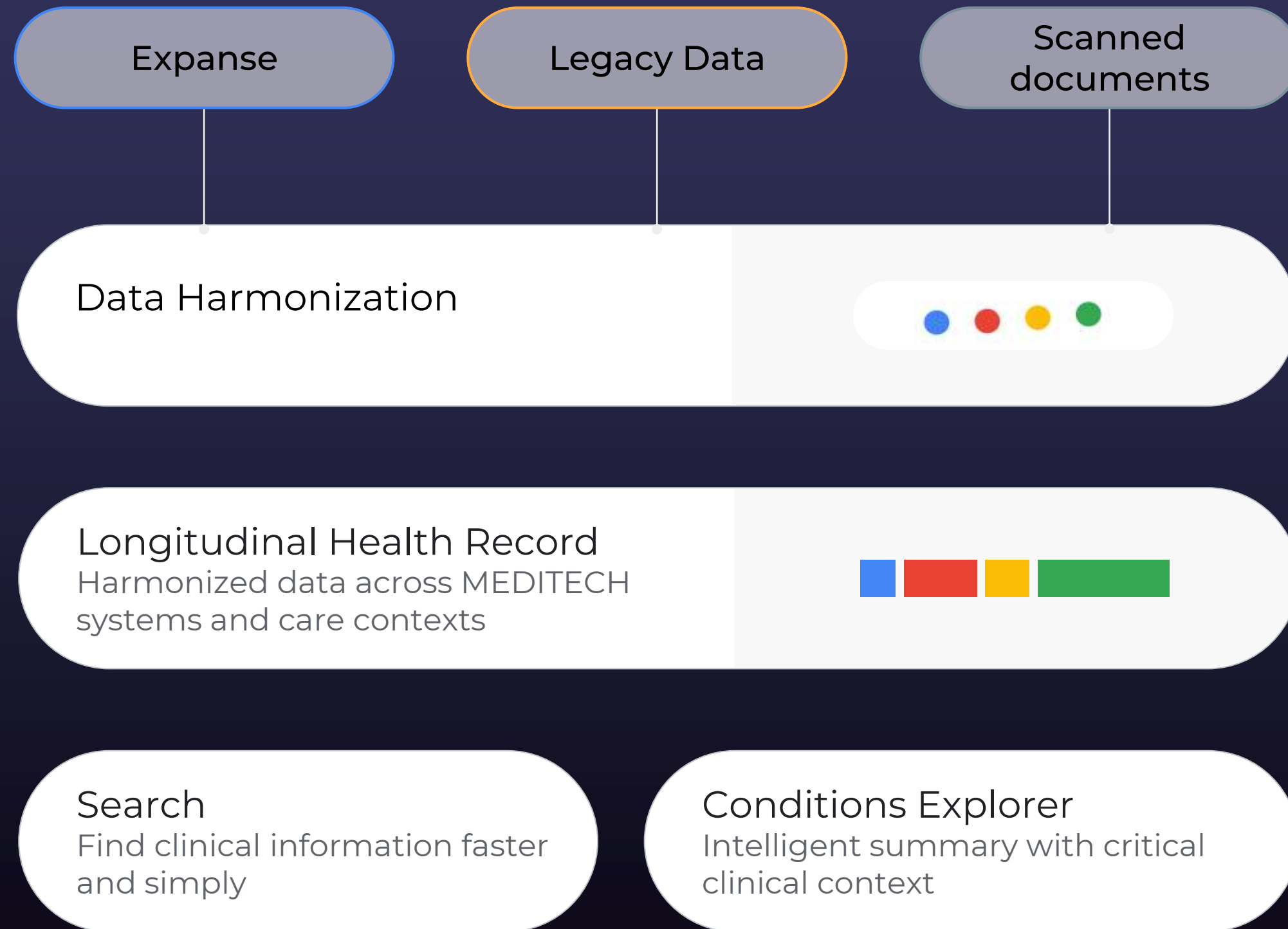


Patient  
Chart

Patient  
Chart

Patient  
Chart

# Expanded Longitudinal Health Record for a More Complete View of the Patient



# MEDITECH SOLUTIONS *powered by AI*

- The most **relevant information** is **identified**
- **Intelligently organized and presented**, highlighting meaningful data for immediate review and comparison
- All within an easy-to-view format **directly within Expanse**
- Providers and patients get back the time they need to **keep moving the care plan forward.**



## Google search and summarization

- LLM and Natural Language Processing enables concept-based searches
- Finds and presents unstructured data, including scanned and handwritten, from legacy platforms
- Synthesizes data to support clinical decision-making



HHS Announces  
**MEDITECH** as Early  
Stakeholder in  
**CANCER MOONSHOT  
INITIATIVE**



“Given the complexity of cancer care and the vast disparities between how each patient responds to treatment, the **ability to share vital patient information and cancer research between disparate healthcare systems** and EHRs can be **critical to a patient’s survival**,” says Vogt.

“**We commend the HHS and ONC** for bringing key stakeholders together to **improve patient outcomes** and look forward to our participation in this workgroup.”

USCDI+ Cancer data elements required for submission via HL7 FHIR API

The team is **already working closely** with the **ONC** to champion the **adoption of HL7 FHIR** in national standards to **support discrete data exchange**.



# Precision Medicine

at Frederick Health



“

Our precision medicine and genetics program includes hereditary genetics in oncology, neurology, and cardiology, and we're ramping up pharmacogenomics across numerous specialties....By flagging prescriptions for drug-to-gene interactions and helping to determine the most appropriate medications based on patients' genetic profiles, we look to reduce drug failures, prevent hospital readmissions, and help find patients the right medication the first time.

”



**Jackie Rice**

Vice President and  
Chief Information Officer  
Frederick Health  
*Frederick, MD*



# Expanse Genomics

Precision Medicine | Tailored and Intuitive



## Therapies and Clinical Trials

Recommendations for therapies and clinical trials specifically designed to target the mutational pathway, patient demographics, and problems

## Pharmacogenomics

In workflow alerts for over 400 medications with active guidance and recommendations

## Intuitive Interpretations

Key genetic details always up to date including interpretations from ClinVar and hot spot information

## Collaboration

Widgets designed to push the most important genetic results to the forefront and collaborate with the rest of the clinical team





# Thank You

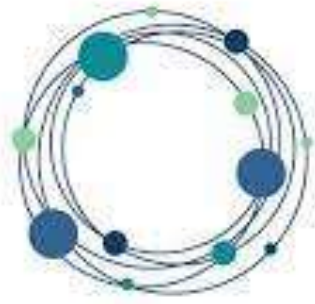
[cturner@meditech.com](mailto:cturner@meditech.com)

# Presentation

---

**Anna Schoenbaum**  
DNP, MS, RN-BC, FHIMSS





FRIENDS OF THE  
NATIONAL LIBRARY  
OF MEDICINE



# PRECISION HEALTH & THE EHR

February 6, 2024

Anna Schoenbaum, DNP, MS, RN, NI-BC, FHIMSS  
Penn Medicine, University of Pennsylvania Health System  
[Anna.schoenbaum@pennmedicine.upenn.edu](mailto:Anna.schoenbaum@pennmedicine.upenn.edu)



# Penn Medicine

University of Pennsylvania Health System

Rayland and Ruth Perelman School of Medicine

- Academic Health System- located in Pennsylvania and New Jersey
- Nation’s first hospital, founded in 1751
- First medical school in the United States
- Early adopters of EHR – 26 years ago
- 1st Cosmos and 1st of 5 health systems of Genomics Module implementations
- FutureEdge Award and AAMC Group on Information Resources (GIR) 2020 Excellence Award on Genomics Implementation

**3,600**



Licensed  
Beds

**11,581** Nurses

**2,336** APPS

**6,243** Physicians



**6.9 Million**



Outpatient  
Visits

**\$11.1 Billion**



Operating  
Revenue

**49,101**



Total  
Employees

**135,122**



Total  
Admissions

**\$1 Billion**



Total Sponsored  
Research

**696,101**



Home  
Health Visits

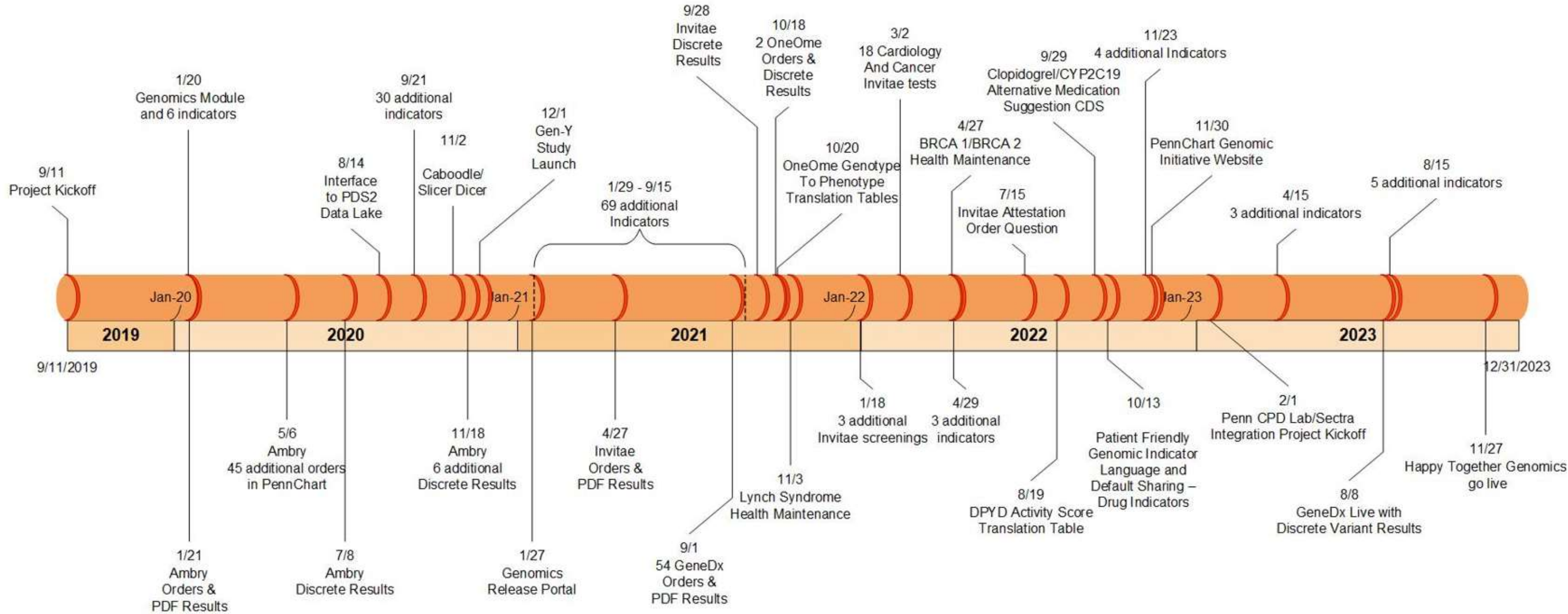


# *Why optimize the EHR for Precision Health?*

- Genetic testing results remain constant over time, providing a stable reference for patient care
- More patients are getting genetic testing (happening outside the medical model)
- Treatment plans extend beyond genetic information, encompassing various data for a comprehensive health record
- In the future, the influence genetics and genomics is expected to penetrate every facet of healthcare



# Precision Health Implementation Timeline



# *How do we optimize the EHR for Precision Health?*

01

**EHR Design**

02

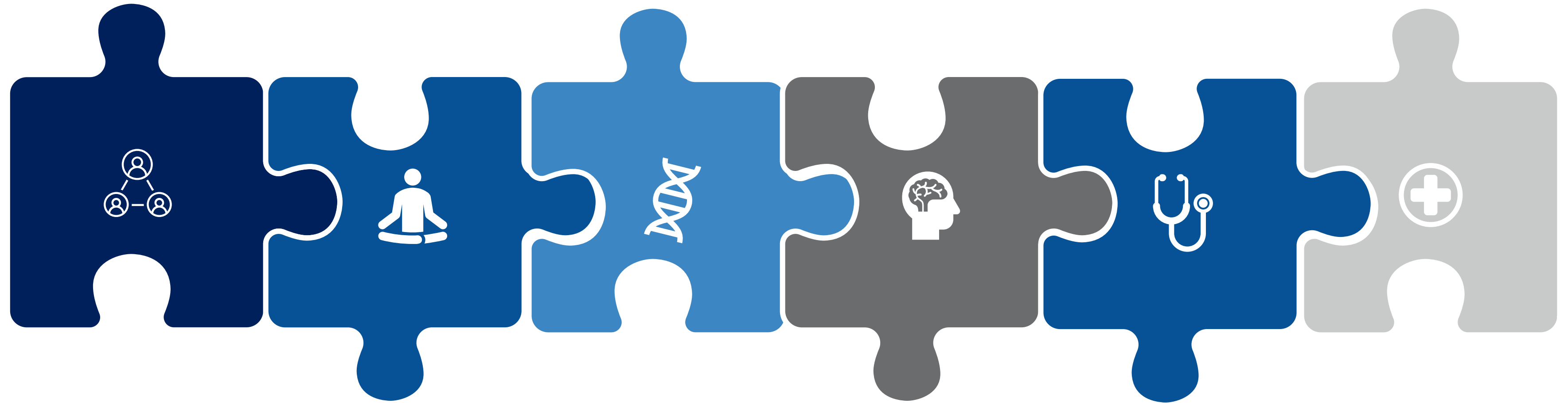
**Workflow Integration**

03

**Considerations**



# *EHR Design for Precision Health*



*Social Determinants of Health (SDOH)*

*Religion & Spirituality*

*Genetics & Genomics*

*Mental Status*

*Clinical Data*

*Other factors*

- Simplify and standardize recording of data
- Utilize widely accepted standards (HL7, FHIR, CDA) for exchanging health data



# EHR Design for Precision Health

01

## Precision Health Tab

- Create a central location within EHR that allows the storing of genomic data, including orders, results and reports
- Eliminates having to “find” genomic data for a particular patient



## Genomics Module

- Streamlining ordering
- Integration of test results from outside laboratories in to EHR
- Simplify and standardize the recording of genetic testing results
- Enabled clinical decision support tools

## Patient Portal

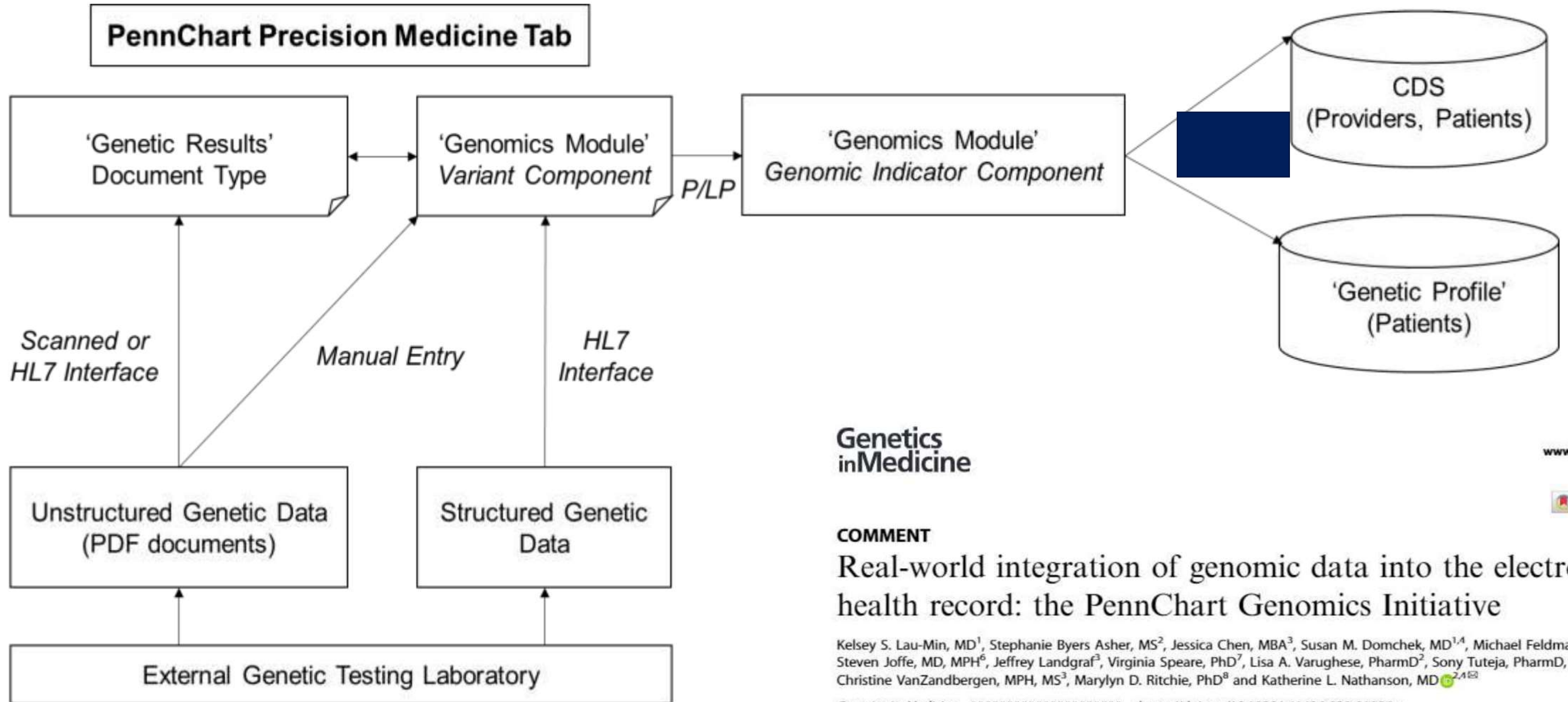
- Provide educational materials and tools for patients to make informed decisions about their care
- Design features that enable patients to engage (i.e., patient reminders)



Precision Health Tab

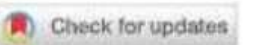


Genomics Module



Genetics inMedicine

www.nature.com/gim



COMMENT

Real-world integration of genomic data into the electronic health record: the PennChart Genomics Initiative

Kelsey S. Lau-Min, MD<sup>1</sup>, Stephanie Byers Asher, MS<sup>2</sup>, Jessica Chen, MBA<sup>3</sup>, Susan M. Domchek, MD<sup>1,4</sup>, Michael Feldman, MD, PhD<sup>5</sup>, Steven Joffe, MD, MPH<sup>6</sup>, Jeffrey Landgraf<sup>3</sup>, Virginia Speare, PhD<sup>7</sup>, Lisa A. Varughese, PharmD<sup>2</sup>, Sony Tuteja, PharmD, MS<sup>2</sup>, Christine VanZandbergen, MPH, MS<sup>3</sup>, Marylyn D. Ritchie, PhD<sup>8</sup> and Katherine L. Nathanson, MD<sup>2,4,8</sup>

Genetics in Medicine \_#####\_; https://doi.org/10.1038/s41436-020-01056-y



## Genomic Testing Considerations

- Develop multi-disciplinary team (genetics providers, molecular pathologists, researchers, pharmacists, IS & informatics)
- Close collaboration with privacy officers and legal team
- Partnership with commercial testing laboratories
- Utilize new interface capability (3-12 mo. point to point interface to 4 - 5 wks. with Epic's Aura platform)



## Clinical Decision Support

- Integrate decision support into workflows at the point of care
- Alerts for potential drug-gene interactions and personalized treatment recommendations



## Care Coordination

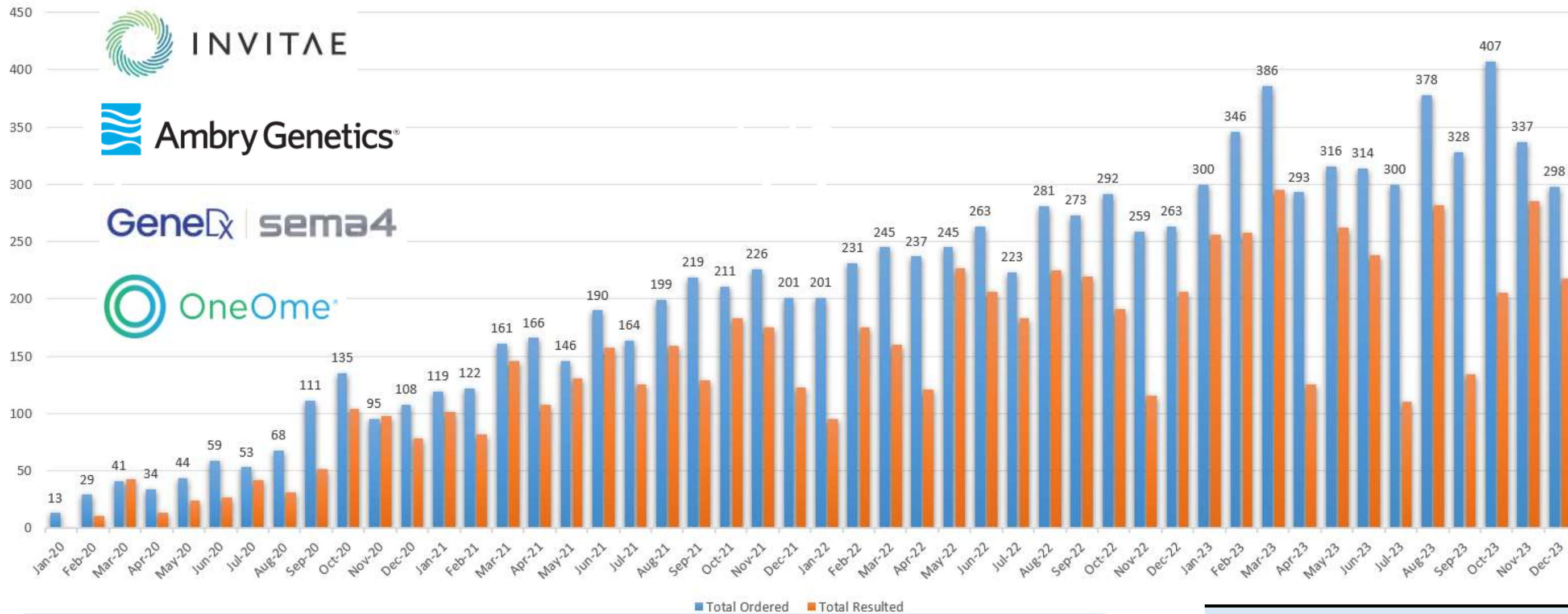
- Integrate data around population health, social care, medication therapy
- Identify high risk patients for screening and surveillance
- Design workflows in coordinating precision health interventions (customized health maintenance reports)
- Ensure communication between care providers, case managers, specialists, and genetic counselors





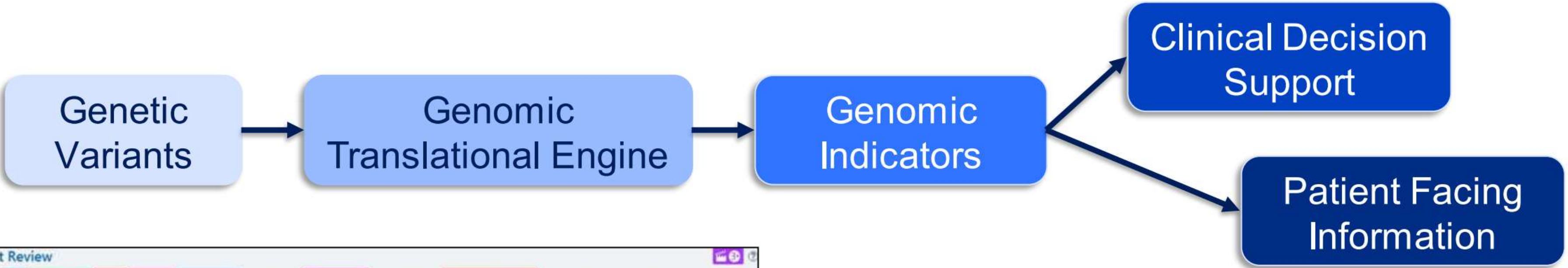
# Genomic Testing Workflow

### Total Genetic Tests Ordered and Resulted by Month



- Over **422** unique genetic testing orders (genes + panels)
- Over **300** different ordering providers
- **10,175** genetic testing orders have been placed/resulted as part of clinical care since 1/21/2020

New Lab/Vendor Requests	Type(s)	Note
Myriad	Germline	AURA
Guardant	Somatic	AURA
Foundation Medicine	Somatic	AURA
Caris	Somatic	AURA
Natera	Germline	AURA
Penn CPD	Somatic	Internal



**Chart Review**

Encounters Labs Imaging Procedures Cardiology Medications Other Orders Precision Medicine Episodes Letters Notes

Snapshot Health Screening RECENT VISITS/RESULTS Snapshot

Medicare Advantage

Reminders and Results  
None

Implants  
No implants to display

**Genomic Indicators**

SDHB Related Hereditary Pheochromocytoma/Paraganglioma Syndrome  
Updated by User, System Default

Indicator Description  
Mutations in the SDHB gene are associated with increased risk of Pheochromocytomas and Paragangliomas.

ibuprofen (ADVIL PO)  
lidocaine 5 % patch  
MELOXICAM PO  
naproxen 500 mg tablet  
prednisONE 10 mg tablet

**Immunizations/Injections**  
New immunizations from outside sources are available for reconciliation

**Significant History/Details**  
Smoking: Light Tobacco Smoker, 1 ppd, 8 pack-years  
Smokeless Tobacco: Current User  
Alcohol: Yes

**Specialty Comments**  
No comments regarding your specialty

**Family Comments**  
None

**Social Determinants of Health**

**myPENN MEDICINE**

Ccwttest Health Visits Messaging Billing Resources Profile

**My Genetic Profile**

This page shows what we know about your genes, including how they might affect the way you respond to certain medications and whether you are predisposed to certain conditions.

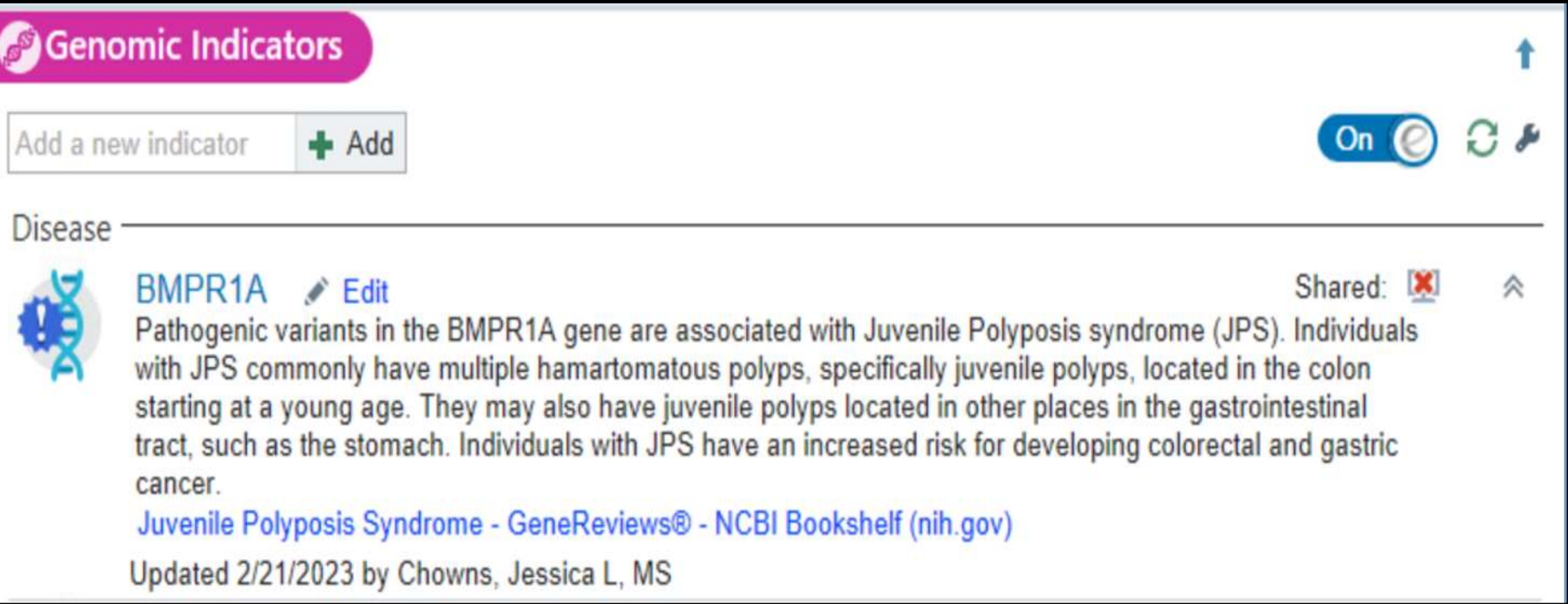
**Genetic Findings of Interest**  
This section contains genetic testing findings related to your health that might require special attention.

**Hereditary Breast and Ovarian Cancer**  
BRCA1 Related  
Based on your recent genetic test result, you may be at risk of developing certain cancers. [More information...](#)

**LMNA-Related Cardiac Disease**  
Based on your recent genetic test, you either have or are at risk to develop familial cardiomyopathy and arrhythmias. If you have a heart condition continue to follow-up with your care team as they recommend. If you do not have a heart condition, you need to have your heart checked with certain tests periodically. If you would like more information, please speak with a cardiologist.



# Genomic Indicators



**Genomic Indicators**

Add a new indicator

**Disease**

**BMPR1A**  Shared:

Pathogenic variants in the BMPR1A gene are associated with Juvenile Polyposis syndrome (JPS). Individuals with JPS commonly have multiple hamartomatous polyps, specifically juvenile polyps, located in the colon starting at a young age. They may also have juvenile polyps located in other places in the gastrointestinal tract, such as the stomach. Individuals with JPS have an increased risk for developing colorectal and gastric cancer.

[Juvenile Polyposis Syndrome - GeneReviews® - NCBI Bookshelf \(nih.gov\)](#)

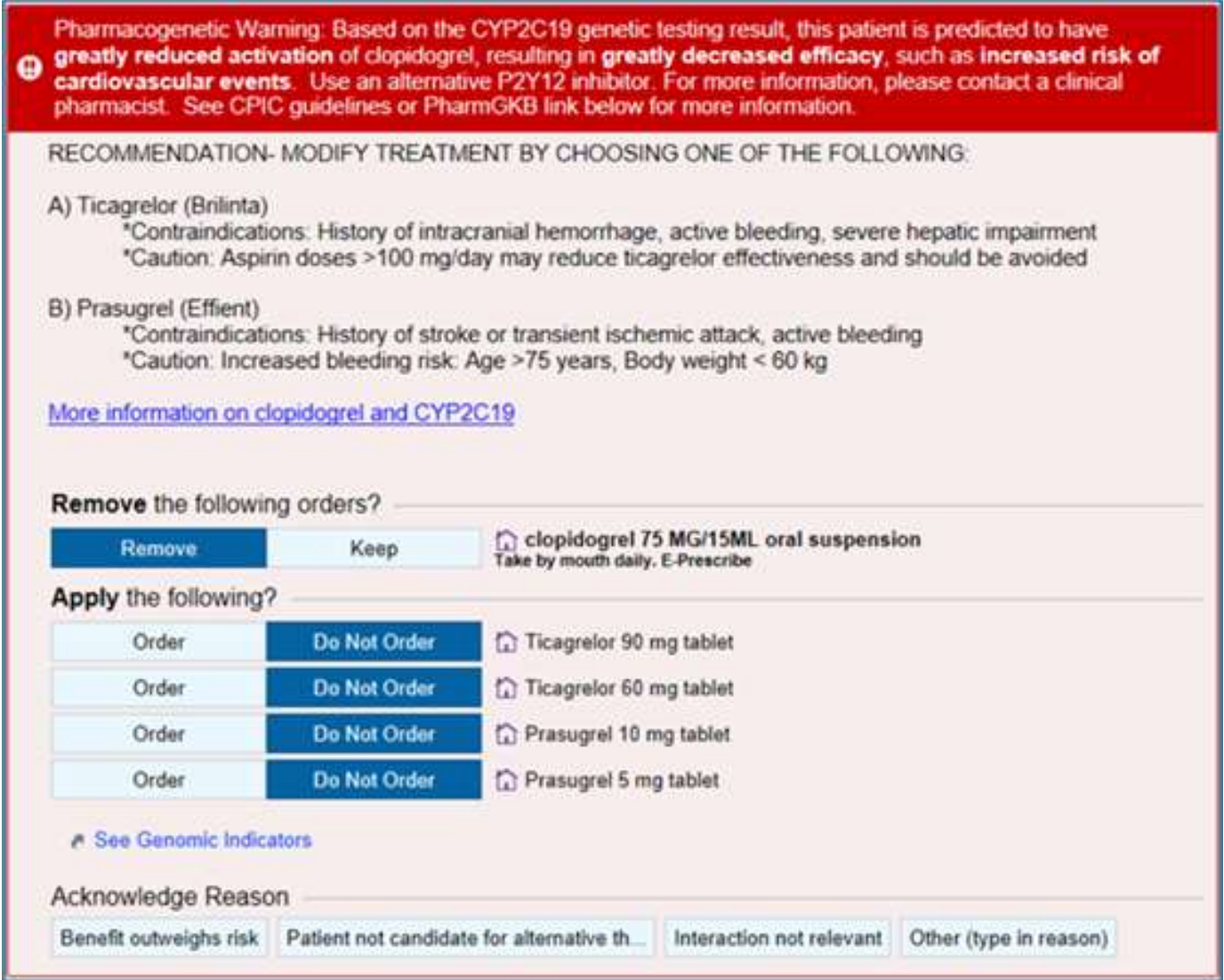
Updated 2/21/2023 by Chowns, Jessica L, MS

© 2024 Epic Systems Corporation

- Examples: *BMPR1A; GREM1; SMAD4; BROP1; RAD51C*
- Function like a ‘flag’ on charts, displaying in the Genomic Indicators activity and Summary reports
- Automatically added to patient charts based on results
  - *Can also be manually added*

# Best Practice Alerts

Pharmacogenomics: Suggests alternative medications for CYP2C19/ Clopidogrel interaction



**Pharmacogenetic Warning:** Based on the CYP2C19 genetic testing result, this patient is predicted to have **greatly reduced activation** of clopidogrel, resulting in **greatly decreased efficacy**, such as **increased risk of cardiovascular events**. Use an alternative P2Y12 inhibitor. For more information, please contact a clinical pharmacist. See CPIC guidelines or PharmGKB link below for more information.

**RECOMMENDATION- MODIFY TREATMENT BY CHOOSING ONE OF THE FOLLOWING:**

A) Ticagrelor (Brilinta)  
 \*Contraindications: History of intracranial hemorrhage, active bleeding, severe hepatic impairment  
 \*Caution: Aspirin doses >100 mg/day may reduce ticagrelor effectiveness and should be avoided

B) Prasugrel (Effient)  
 \*Contraindications: History of stroke or transient ischemic attack, active bleeding  
 \*Caution: Increased bleeding risk: Age >75 years, Body weight < 60 kg

[More information on clopidogrel and CYP2C19](#)

**Remove the following orders?**

clopidogrel 75 MG/15ML oral suspension  
 Take by mouth daily. E-Prescribe

**Apply the following?**

<input type="button" value="Order"/>	<input checked="" type="button" value="Do Not Order"/>	<input type="button" value="Home"/> Ticagrelor 90 mg tablet
<input type="button" value="Order"/>	<input checked="" type="button" value="Do Not Order"/>	<input type="button" value="Home"/> Ticagrelor 60 mg tablet
<input type="button" value="Order"/>	<input checked="" type="button" value="Do Not Order"/>	<input type="button" value="Home"/> Prasugrel 10 mg tablet
<input type="button" value="Order"/>	<input checked="" type="button" value="Do Not Order"/>	<input type="button" value="Home"/> Prasugrel 5 mg tablet

[See Genomic Indicators](#)

**Acknowledge Reason**






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# Health Maintenance

## Lynch Syndrome, BRCA1/2 Carriers

*Annual exams, imaging, testing*

Health Maintenance			
<a href="#">Address Topic</a>   <a href="#">Remove Override</a>   <a href="#">Edit Modifiers</a>   <a href="#">Report</a>   <a href="#">Refresh</a>   <a href="#">Guidelines</a>			
Topic	Due Date	Frequency	Date Completed
<b>Current Care Gaps</b>			
BRCA1 Gyn Onc Appointment Needed	 Overdue - never done	1 year(s)	
BRCA1/2 CREP Appointment Needed	 Overdue - never done	1 year(s)	
BRCA1/2 Breast MRI Needed	 Overdue since 10/5/2016	1 year(s)	10/5/2015 - MAM...
BRCA1/2 Mammogram Needed	 Overdue since 7/11/2017	1 year(s)	 7/11/2016 - M...

# Other Considerations for Precision Health



## Training & Education

- Provide training for healthcare professionals on interpreting and using precision health data
- Develop educational programs to keep clinicians updated on advancements in genomic medicine



## Data Governance & Security

- Establish data governance policies to ensure the security and privacy of proper distribution and handing of genomic information
- Understand liability for genomic data storage (variants not reported or reclassified)

# Wisdom from Wilson

*“my lived experiences as a stage 4 cancer patient who also works and interacts with a large international group of patients and supporters, we need to be sure not to overshadow the important considerations:*

- *SDOH*
- *Religion/spirituality*
- *Access to health care*
- *Mental status*
- *Genomics, biomarker information*

*In my support group of patients with the same cancer that I have, what we choose to do varies based on so many more factors than just our cancer type and biomarkers....*

*....always look at the decisions of others through their personalized health care plan.”*







# Presentation

---

**Johnston Thayer**  
MBA, RN

# PRECISION HEALTH

*Delivered at the point of care*





**“The era of EMIR  
genomics has begun.”**

-Ingo Helbig  
(August 19, 2020)



# GENOMICS DATA LANDSCAPE

*Clinical genomics in the context of the electronic health record*

## Genomic Data Management

Incorporate discrete genomic results into patients' records



## Decision Support & Education

Provide genomically-informed point-of-care guidance, education, and content



## Analytics and Research

Combine phenotypic and genomic data to perform research and make new discoveries



## Specialties Integration

Leverage genomic data in specialty workflows, such as Care Paths and Protocol Selection





# Genomics



Technology enables...

- ❖ Storing massive volumes of information
- ❖ Complex interpretation of data
- ❖ Tracking and surfacing context relevant insights at the point of care (e.g., pharmacogenomics)





# PRECISION MEDICINE FOR EVERYONE, ANYWHERE

We all know the promise...



Ordering the right medication, the first time



Precision cancer treatments



Unlocking the genetic origins of disease

...and the historical barriers



Expensive, time-consuming connections to labs



Text-based PDF result reports



Complexity and difficulty of genomic data



# ORDERS AND RESULTS ANYWHERE



**e**Care  
Everywhere








# TURNKEY ACCESS TO GENETICS

*with native connections to specialty diagnostic labs through Aura*



**200K**



**tests ordered per month**

2.8x increase in the last year

**\$100,000**

*savings per lab connection*

**2 WEEK**

*technical implementation time*

**ZERO**

*extra steps for your clinicians*



**KNOWLEDGEBASES**

# GENOMICS CLOUD

**SCALABLE COMPUTE**

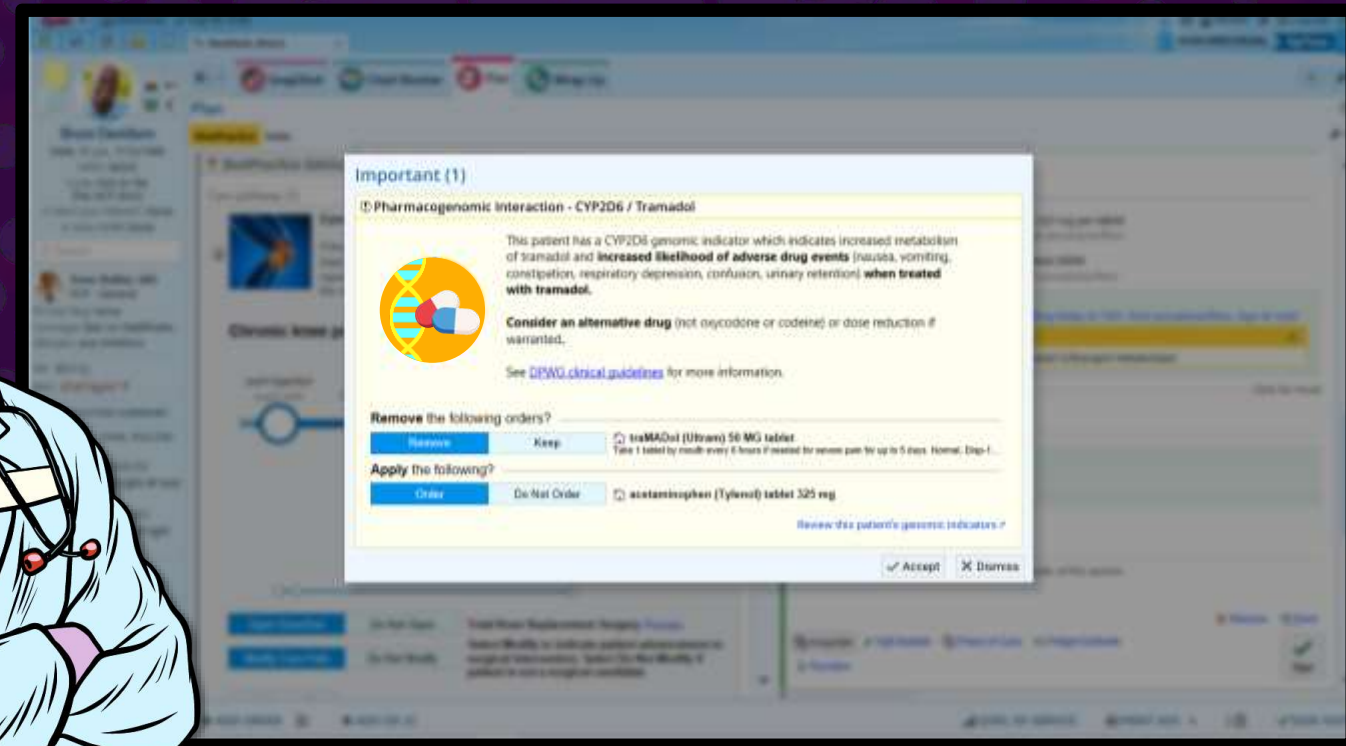
**MASSIVE DATASETS**

Powered by  
**Nebula**

**SEQUENCE  
DATA**



**LABS**








# ACTIONABLE GENOMIC INSIGHTS TO IMPROVE CLINICAL CARE



Based on your genetic testing, it seems like we should change your treatment plan.

**Important (1)**

**Pharmacogenomic Interaction - CYP2D6 / Tramadol**

 This patient has a CYP2D6 genomic indicator which indicates increased metabolism of tramadol and **increased likelihood of adverse drug events** (nausea, vomiting, constipation, respiratory depression, confusion, urinary retention) **when treated with tramadol.**

**Consider an alternative drug** (not oxycodone or codeine) or dose reduction if warranted.

See [DPWG clinical guidelines](#) for more information.

Remove the following orders?

Remove  Keep  traMADol (Ultram) 50 MG tablet  
Take 1 tablet by mouth every 6 hours if needed for severe pain for up to 5 days. Normal. Disp-1...

Apply the following?

Order  Do Not Order  acetaminophen (Tylenol) tablet 325 mg

[Review this patient's genomic indicators](#)

Accept  Dismiss

**GENETIC VARIANT DECISION SUPPORT**



# ACTIONABLE GENOMIC INSIGHTS TO IMPROVE CLINICAL CARE



Based on your genetic testing, it seems like we should change your treatment plan.

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Remove the following orders?

<input type="checkbox"/>	<input type="checkbox"/>	traMADol (Ultram) 50 MG tablet
<input type="checkbox"/>	<input type="checkbox"/>	Take 1 tablet by mouth every 6 hours if needed for severe pain for up to 5 days. Normal, Disp-1...

Apply the following?

<input type="checkbox"/>	<input type="checkbox"/>	acetaminophen (Tylenol) tablet 325 mg
--------------------------	--------------------------	---------------------------------------

Accept  Dismiss

## Genomic Variant Results

Expand All Collapse All

**Clinical Genome Sequencing** Order: 1957345  
Collected 8/15/2022 9:12 AM Status: Edited Result - FINAL

**Germline — Detected**

- BMP2 (Pathogenic)** Risk for pulmonary hypertension  
Heterozygous
- PTGIS (Likely pathogenic)** Risk for essential hypertension  
Homozygous

HGVS Name: NM\_000961.4(PTGIS):T>C (Substitution) Homozygous  
Type: Simple Allelic State: Homozygous  
ClinVar-V (v 1): VCV000007864

**Interpretation**  
Research is still being established, but this variant in PTGIS is indicative of possible familial essential hypertension.

**Sequence Change**

DNA Change:	Transcript:	Molecular
T>C	NM_000961.4 (RefSeq-T)	Consequence: Splice Donor Variant

**Region**

Cytogenetic Location:	Genome Assembly:	Chromosome:	DNA Region:
20q13.13	GRCh38	20	49511026

**Pharmacogenomic Results**

- Likely Significant**
- CYP2C19 \*17/\*17 CYP2C19 Ultrarapid Metabolizer
- CYP2D6 \*1/\*1x3 CYP2D6 Ultrarapid Metabolizer

Genotypes: CYP2D6 \*1/\*1x3 Type: Pharmacogenomic Genotype

**Associated Phenotype Information**

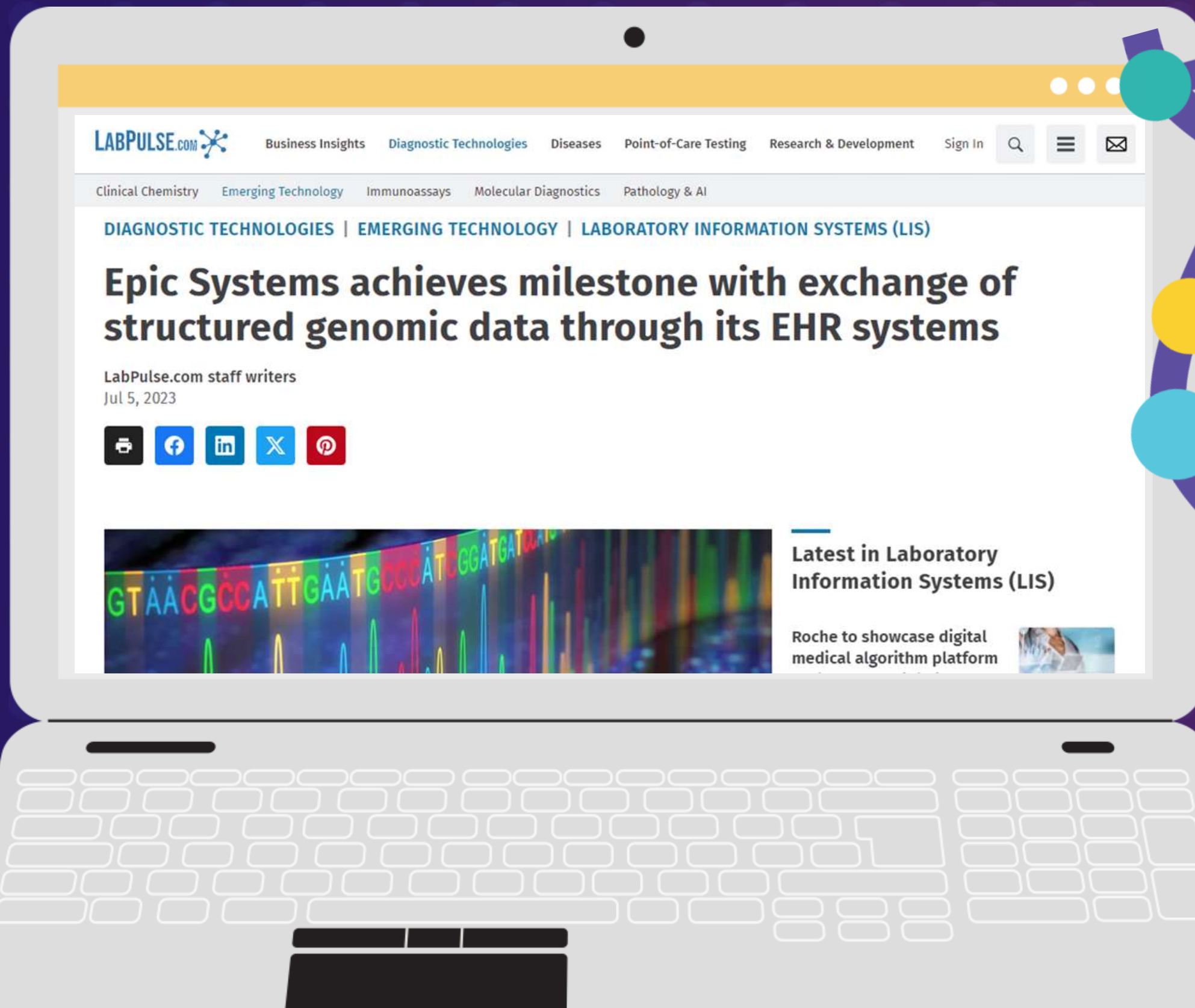
Effect on Drug Metabolism:	Effect on High-Risk:
Ultrarapid metabolizer	Risk: High risk

- Possibly Significant**
- CYP2C19 CYP2C19 Ultrarapid Metabolizer - Variant Details

**GENETIC VARIANT DECISION SUPPORT**



# PRECISION MEDICINE, MADE COLLABORATIVE



# ***ACTIONABLE GENOMIC INSIGHTS FOR NURSING***





# ACTIONABLE GENOMIC INSIGHTS FOR NURSING

**Jessica Hubbard**  
Female, 39 y.o., 12/26/1984  
Pronouns: she/her/hers

**CURRENT PROGRAMS**  
Whole Person Care  
Enrolled

**SOCIAL DETERMINANTS**  
[Icons for food, housing, car, weather]

**Mammography BRCA Positive**  
Due: Yearly

Has Genomic Indicator  
BRCA: Breast Cancer Genetic Risk

The screenshot shows a patient's EHR profile. On the left, a dark blue sidebar contains the patient's name, photo, and demographic information. Below this, there are sections for 'CURRENT PROGRAMS' (Whole Person Care Enrolled) and 'SOCIAL DETERMINANTS' (represented by four icons: a fork and knife, a house, a car, and a weather cloud). The main content area is a light blue grid. A white pop-up window is overlaid on the grid, displaying a 'Mammography BRCA Positive' alert with a 'Due: Yearly' reminder. To the right of the alert, a legend explains the 'Has Genomic Indicator' and 'BRCA: Breast Cancer Genetic Risk'.



# ACTIONABLE GENOMIC INSIGHTS FOR NURSING

**Matthews, Bruce** ✕

**Assessments**

1343	📅	Reassess Pain Level after administering tramadol (ULTRAM) tablet 50 mg	Doc
1345	📅	Assess for Adverse Drug Event	Doc

[Genomic Indicators](#) [Order Details](#) [Edit](#) [Skip](#)

[+ Task](#) [📅 Appt](#) [📅 Patient Event](#)

***THANK YOU***

# Q&A

## PANEL 2

# Precision Health in the Real-World

---

**Moderator: Rebecca Freeman, PhD, RN, PMP**

- **Cathy Turner, BSN, MBA, RN-BC**
- **Anna Schoenbaum, DNP, MS, RN-BC, FHIMSS**
- **Johnston Thayer, MBA, RN**



**We'll be right back with our next session**

## **PANEL 3**

# **Precision Health: Implications for Curriculum**

---

**Moderator: Connie Delaney, PhD, RN, FAAN, FACMI**

- **Erin Dickman, DNP, RN, OCN**
- **Kathleen Calzone, PhD, RN, AGN-BC, FAAN**
- **Patricia Sharpnack, DNP, RN, CNE, NEA-BC, ANEF, FAAN**

# PANEL 3

## Precision Health: Implications for Curriculum

---

**Moderator: Connie Delaney, PhD, RN, FAAN, FACMI**

- **Erin Dickman, DNP, RN, OCN**
- **Kathleen Calzone, PhD, RN, AGN-BC, FAAN**
- **Patricia Sharpnack, DNP, RN, CNE, NEA-BC, ANEF, FAAN**

# Presentation

---

**Erin Dickman**  
DNP, RN, OCN

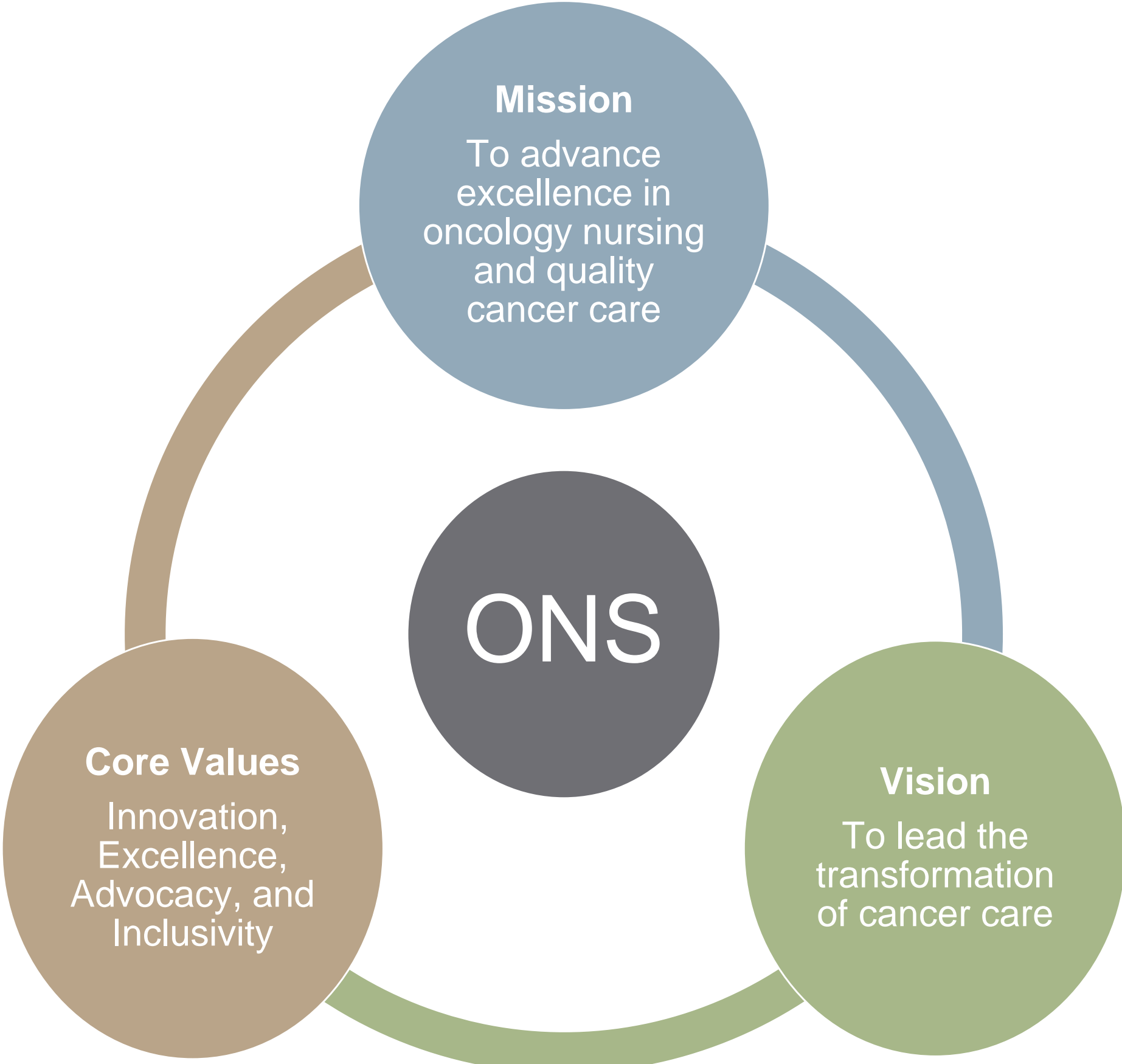




# Designing Multimodal Genomics and Precision Oncology Education for Nurses

**Erin Dickman, DNP, RN, OCN**  
Oncology Clinical Specialist  
Oncology Nursing Society

# Oncology Nursing Society (ONS)



# The Role of the Oncology Nurse

Adopt	Adopt and encourage the use of standardized, consistent terminology
Educate	Educate patients and family members about the biomarker testing process
Participate	Participate in the consent process and/or collect the biospecimen
Understand	Understand possible outcomes of testing and implications for cancer care
Identify	Identify when to refer to a genetics professional for possible germline testing
Provide	Provide emotional support to patients and families
Advocate	Advocate for precision treatment options



# ONS Member Genomics' Survey

50% would describe their knowledge of genomics as good; 45% would describe it as poor

44% report low confidence in discussing genomics with patients

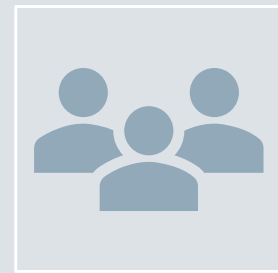
49% report low confidence discussing genomics with healthcare providers

99% responded they would see value in learning more about genomics

93% responded they would attend a course on genomics on their own time

99% responded that ONS should offer programs on genomics

# A Human-Centered Design Approach



## Internal Data

Landscape analysis  
Expert advisory board  
Member survey



## External Data

Literature review  
Analyzed efforts of other organizations

# Our Strategic Approach

## Awareness

- Infographic
- Chapter media toolkit
- Social media
- Learning library
- Call to action
- Press release
- Campaign with hashtag

## Knowledge

- Taxonomy
- Publications
- Podcasts
- Online course
- Video series
- Synchronous workshops

## Point of Care Tools

- Quick guides
- Algorithms
- Checklists
- Discussion tools
- Patient education
- Genetic disorder reference sheets
- Biomarker database





Visual



Auditory



Kinesthetic



Reading



Writing

# Multi-Modal Teaching

# Guiding Principles

01

Start with foundational content

02

Provide a variety of lengths and formats

03

Make education accessible

# Genomics and Precision Oncology Learning Library



The ONS Genomics Advisory Board members have compiled a comprehensive list of learning resources for your quick reference!

Patient Education- New!

Clinical Practice Resources- New!

Learning Activities

Genomic "Glad You Asked" Video Series

ONS Articles

ONS Genomics Taxonomy

Books

Podcasts

Genomics External Resources

Learn More About the ONS Genomics Advisory Board



+

+

+

+

+

No paid membership needed, just an ONS registered account.





# Genomics Taxonomy



The ONS Genomics Advisory Board members have created a comprehensive glossary of genomics related terms for your quick reference.

*As ONS works to update and expand available genomic education, resources and publications, use of current terminology consistently is a priority to help avoid confusion and reflect the state of the evidence. We are in the midst of a terminology transition as we move from genetics into genomics. The goal is for all oncology nurses to be fluent in not just these terms, but the underlying concepts and their applications in practice. This taxonomy serves as foundation for terminology that will begin to be represented in ONS materials. As the evidence base continues to evolve and expand, so will this taxonomy. You can refer to the [outline of the taxonomy](#) to find the categorization of terms.*

[Genomics Taxonomy Terms Table of Contents](#)

[Genome Foundations](#)

[Mode of Inheritance](#)

[Biomarkers](#)

[Biomarker Testing](#)

[Variant](#)

[Variant Subcategories](#)

[Incidental Finding](#)

[Genomics Taxonomy References](#)

## Learn Cancer Care

Articles reviewing venous access devices, cancer genetics, immunotherapy, survivorship care, and more.

[Resources »](#)



40% PROGRESS

Brain

Breast Cancer

Lung Cancer

Other lung

Prostate Cancer

Liver

Skin Cancer

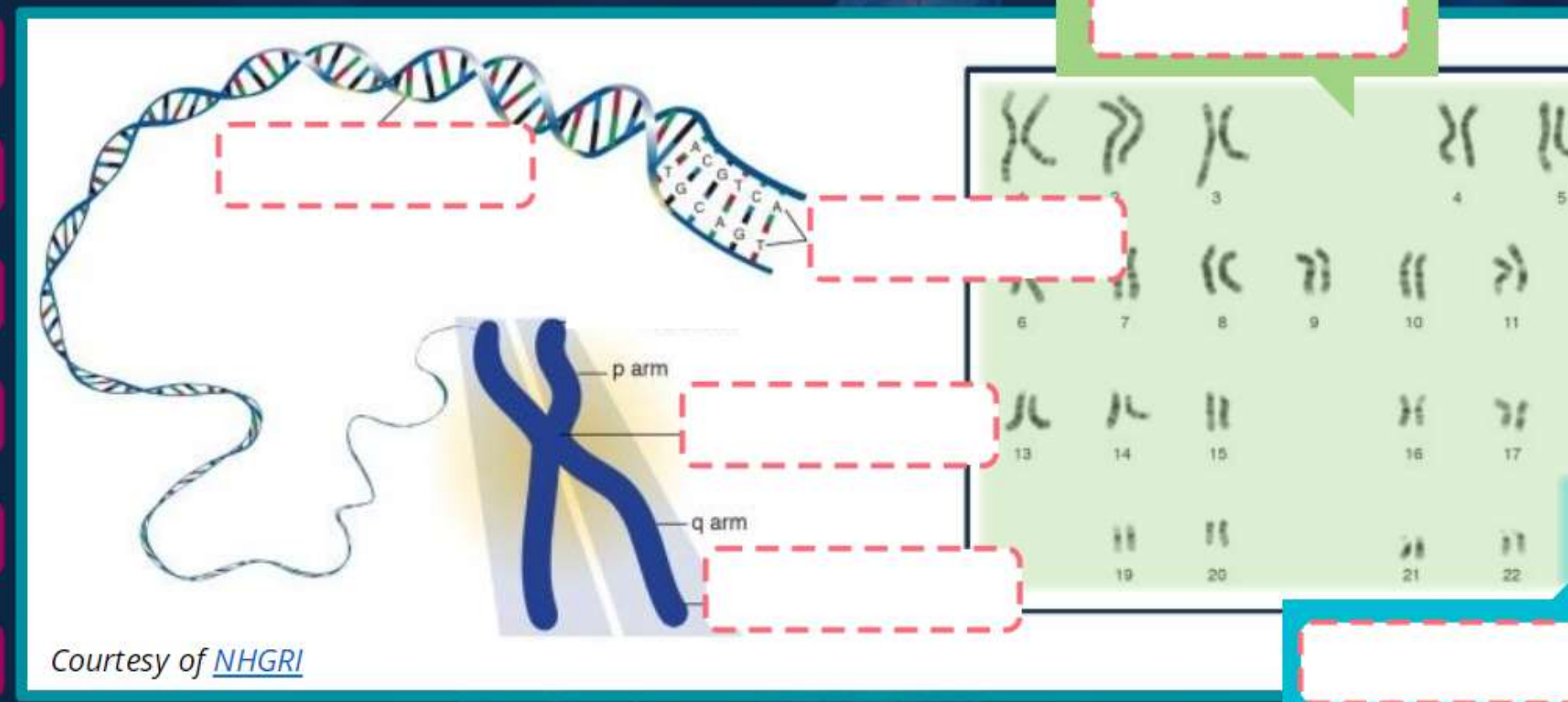
Adrenal gland

Bone

Select the buttons to see main sites of metastasis.

# Knowledge Check

- Centromere
- Base pairs
- Telomere
- Autosomes
- DNA
- Sex chromosomes



Drag and drop each term to its location.

SUBMIT

## Objectives

- Articulate the importance of genomics to oncology nursing in the context of precision oncology.
- Discuss foundational genomic concepts.
- Describe the genomic basis for cancer development and progression.
- Define biomarker and discuss methods of biomarker testing.

# Genomic Foundations for Precision Oncology Course









# Glad You Asked Video Series



Answers questions posed by ONS members



14 videos



3–7-minute videos



Topics range from being broad to specific

**Glad you Asked: Questions for Family History**  
Oncology Nursing Society

What relatives should be included in assessment of family history?

**Degrees of Relationship**

- First-degree relatives : parents, siblings, children
- Second-degree relatives : half-siblings, aunts, uncles, grandparents, nieces & nephews
- Third-degree relatives : first cousins

## Overview

Listen to ONS Genomics Advisory Board member, Mary L. Schmitt, MS, APRN, FNP-BC, AOCNP as she speaks to what is meant by a wild-type gene. Learn how this information could help plan cancer treatment.

**Glad You Asked: Wild-Type**  
Oncology Nursing Society

Nurses must embrace the science and effectively teach their patients about unique genomic characteristics which drive their cancer and how this information provides guidance in the selection of optimal therapy.

# Clinical Practice Resources

Infographics

Discussion Tools

Quick Guides

Toolkits

### Biomarker Testing in Non-Small Cell Lung Cancer Discussion Tool

**WHAT IS BIOMARKER TESTING?**  
Biomarker testing is the use of a laboratory test to measure genes, proteins, or other substances (biomarkers) found in blood or tissue. It can provide insights into what drives cancer to grow.

**WHY DO WE USE BIOMARKER TESTING?**  
We use it to gather individualized information about the cancer to choose treatments specifically designed to target what makes the cancer grow. Some therapies only work for people whose cancer shows a certain biomarker.

**WHEN TO TEST FOR BIOMARKERS?**  
At diagnosis of stage IV or metastatic disease with a nonsquamous type\*  
Early-stage EB-IIIa nonsquamous type that has been removed surgically  
In some situations among patients with squamous cell carcinoma  
\*Adenocarcinoma is the most common subtype to have biomarkers present.

**HOW IS BIOMARKER TESTING DONE?**  
A tissue sample from a tumor biopsy is sent to a lab for analysis. | One or two vials of blood are collected from a blood draw and sent to a lab for analysis of the DNA shed by the cancer into the bloodstream.

**WHEN WILL YOU GET THE RESULTS?**  
Your doctor or nurse will get the results in approximately two to three weeks. It is important to have these results before beginning treatment so your care plan is customized for your specific type of cancer.

**OTHER BENEFITS:**  
MORE EFFECTIVE | OFTEN FEWER SIDE EFFECTS

**YOUR HEALTHCARE PROVIDER MAY DECIDE TO RETEST AT ANY TIME DURING YOUR TREATMENT IF THEY FEEL IT IS NECESSARY.**

### WHEN TO REFER TO A GENETICS PROFESSIONAL

**WHY IS IT IMPORTANT TO KNOW IF YOUR PATIENT'S CANCER IS HEREDITARY?**

- Helps the patient to choose the right cancer therapy
- Informs the patient and the patient's family of their risk and allows them to get tested

**HOW TO IDENTIFY A PATIENT WITH HEREDITARY RISK:**

- Update medical history.
- Update family history.
- Evaluate for risk of hereditary cancer syndrome and refer if appropriate.

**HEREDITARY RISK RED FLAGS:**

- Younger than 50 years old with a cancer diagnosis
- Personal history of more than one cancer diagnosis
- Multiple family members with cancer
- Diagnosed with a rare cancer

**OTHER HEREDITARY RISK FACTORS:**

Personal or family history of:

- Male breast cancer
- Ovarian cancer
- Retinoblastoma
- Pheochromocytoma
- Medullary thyroid cancer
- Triple negative breast cancer (younger than 60 years old)
- Breast, colon or endometrial cancer (younger than 50 years old)
- Known genetic pathogenic variant
- Pancreatic cancer
- >20 adenomatous polyps; >5 serrated polyps with 2 polyps 10 cm or >20 serrated polyps
- Paranglioma
- MSI high colon or endometrial cancer
- Multiple cancers in an individual or a family

**If the patient has at least 1 of these risk factors:**

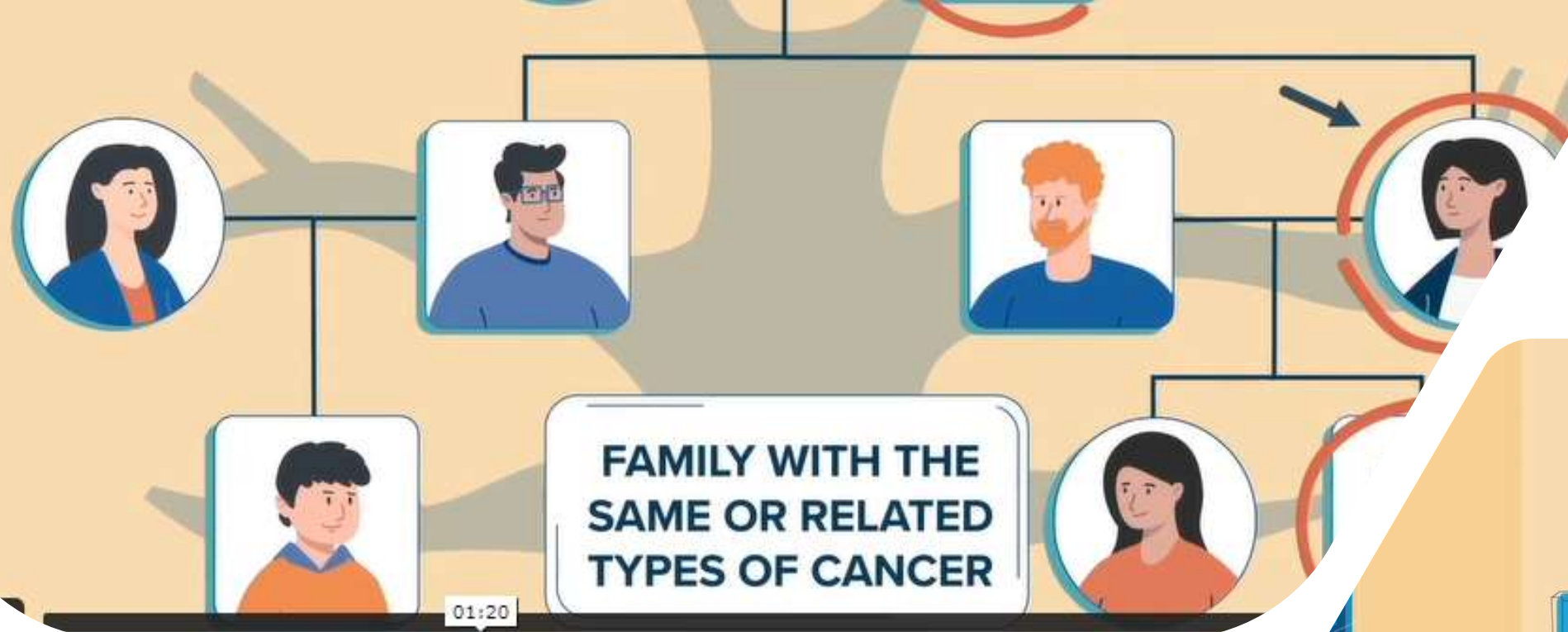
- Refer to a genetics professional
- Continue to update and reevaluate the patient's medical history and family history every year

**ONS Oncology Nursing Society**

National Comprehensive Cancer Network. (2020). Genetic/Familial High Risk Assessment: Breast, Ovarian and Pancreatic. Version 1.2020. Retrieved from www.nccn.org  
National Comprehensive Cancer Network. (2018). Genetic/Familial High Risk Assessment: Colorectal version 1.2018. Retrieved from www.nccn.org  
Sierczynska, M., Krasnowska, K., Jaskolska, A. L., & Kozłowski, P. (2019). Large-scale meta-analysis of mutations identified in panels of hereditary cancer-related genes - Providing evidence of cancer predisposition genes. *Open Access Oncology*, 13(12), 412-462. DOI: 10.1080/19370191.2019.161627  
This resource was developed by ONS through a sponsorship from AstraZeneca.







01:20



# Genetic Counseling and You

- Aunt Cathy, colon cancer, age 56
- Grandpa Jerry, stomach cancer, age 74
- Cousin Anna, endometrial cancer, age 42
- Brother Tom, prostate cancer, age 66

03:24



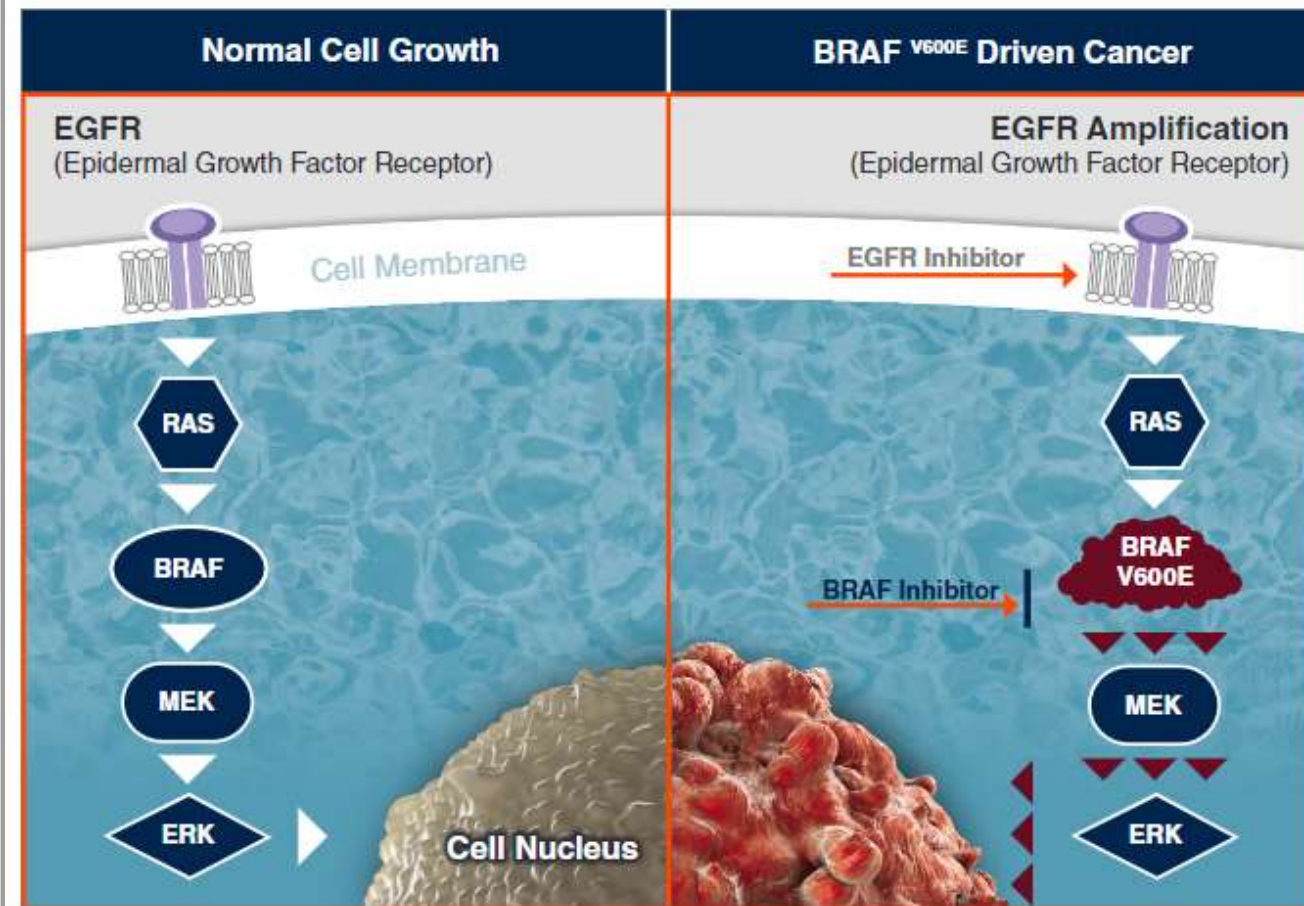


# Biomarker Quick Guide: BRAF

## What is BRAF?

*BRAF*\* is a protein-coding gene which is part of the normal cellular signaling pathway, often described as the EGFR/RAS/MEK/ERK pathway also known as the MAPK (mitogen activated protein kinase) pathway. The *BRAF* proto-oncogene controls cell growth, differentiation, migration, and apoptosis.

\**BRAF*—B-raf proto-oncogene serine/threonine-protein kinase



Adapted from: National Cancer Institute/Kelly Crotty, <https://www.cancer.gov/news-events/cancer-currents-blog/2019/colorectal-cancer-braf-triplet-targeted-therapy>

## What does it mean when there is a driver variant in the BRAF gene?

When there is a driver variant in the BRAF gene, the gene is continually in the “on” position with uncontrolled cellular growth and proliferation. One of the most common BRAF driver variants is located at codon number 600 and is known as the V600E variant.



# NEXT-GENERATION SEQUENCING SAMPLE REPORT

Many companies offer biomarker testing that use next-generation sequencing (NGS) technology. While the reports may vary in format, they typically include similar content. Highlighted below are the sections most relevant to nurses.

Additional Resource: ONS Biomarker Database, <https://n.ons.org/biomarkers>

Patient		Physician		Specimen	
Name:	Ordering Physician:	Facility:	Specimen Type: FFPE	Specimen Site: Left lung	Tumor Type: Lung adenocarcinoma
Disease:	Medical Faculty ID:	Pathologist:	Collection Date: 3/30/2022	Date of Analysis: 4/10/2022	
Date of Birth:	Additional Recipients:				
Sex:					
Health Record #:					

CLINICALLY SIGNIFICANT VARIANTS					
A	B	C	D	E	F
Actionable Variant	Classification	FDA-Approved Therapies With Clinical Benefit	Therapies Not Likely to Be Effective	FDA-Approved Therapies in Different Cancer Type	Potential Clinical Trials
EGFR amplification; T790M	Tier I	Osimertinib (Tagrisso®)	Gefitinib (Iressa®), erlotinib (Tarceva®), afatinib (Gilotrif®), dacomitinib (Vizimpro®)	None	60 current trials

G Immunotherapy Biomarkers		H Prognostic or Diagnostic Variants			
Biomarker	Actionability	Biomarker	Classification	Associated Cancer Types	Associated Clinical Outcomes
Microsatellite status: MS-Stable	No therapies or clinical trials	TP53	Prognostic	Ovarian, esophageal, colorectal, head and neck, lung, leukemia, sarcoma, testicular, melanoma, cervical	More aggressive disease, increased rates of resistance to chemotherapy, shorter survival
Tumor Mutational Burden: 8 Muts/Mb	No therapies or clinical trials				

### A Actionable Variant

Variants associated with clinical utility meaning that it may be predictive of response or nonresponse to certain cancer therapies

### B Classification

Variants are classified into four tiers based on evidence from the literature supporting their clinical significance. Variants in Tier I are pathogenic and are likely associated with FDA-approved therapies, whereas variants in Tier IV are benign or likely benign.

### C FDA-Approved Therapies With Clinical Benefit

Therapies indicated for the biomarker and primary cancer diagnosis, approved by the FDA (Food and Drug Administration)

### D Therapies Not Likely to Be Effective

Therapies that are not clinically effective at treating tumors with the identified actionable variant

### E FDA-Approved Therapies in Different Cancer Type

Therapies approved to treat tumors found to have the actionable variant in other cancer types

### F Potential Clinical Trials

Number of open clinical trials based on the actionable variant. Usually, a report will also contain a more detailed list of potential trials (see page 6 of this resource for an example).

### G Immunotherapy Biomarkers

Biomarkers such as tumor mutational burden (TMB) and microsatellite instability (MSI) may help predict how well the tumor may respond to immune checkpoint inhibitors. There may be options with certain tests to also see PD-L1 expression scores.

### H Prognostic or Diagnostic Variants

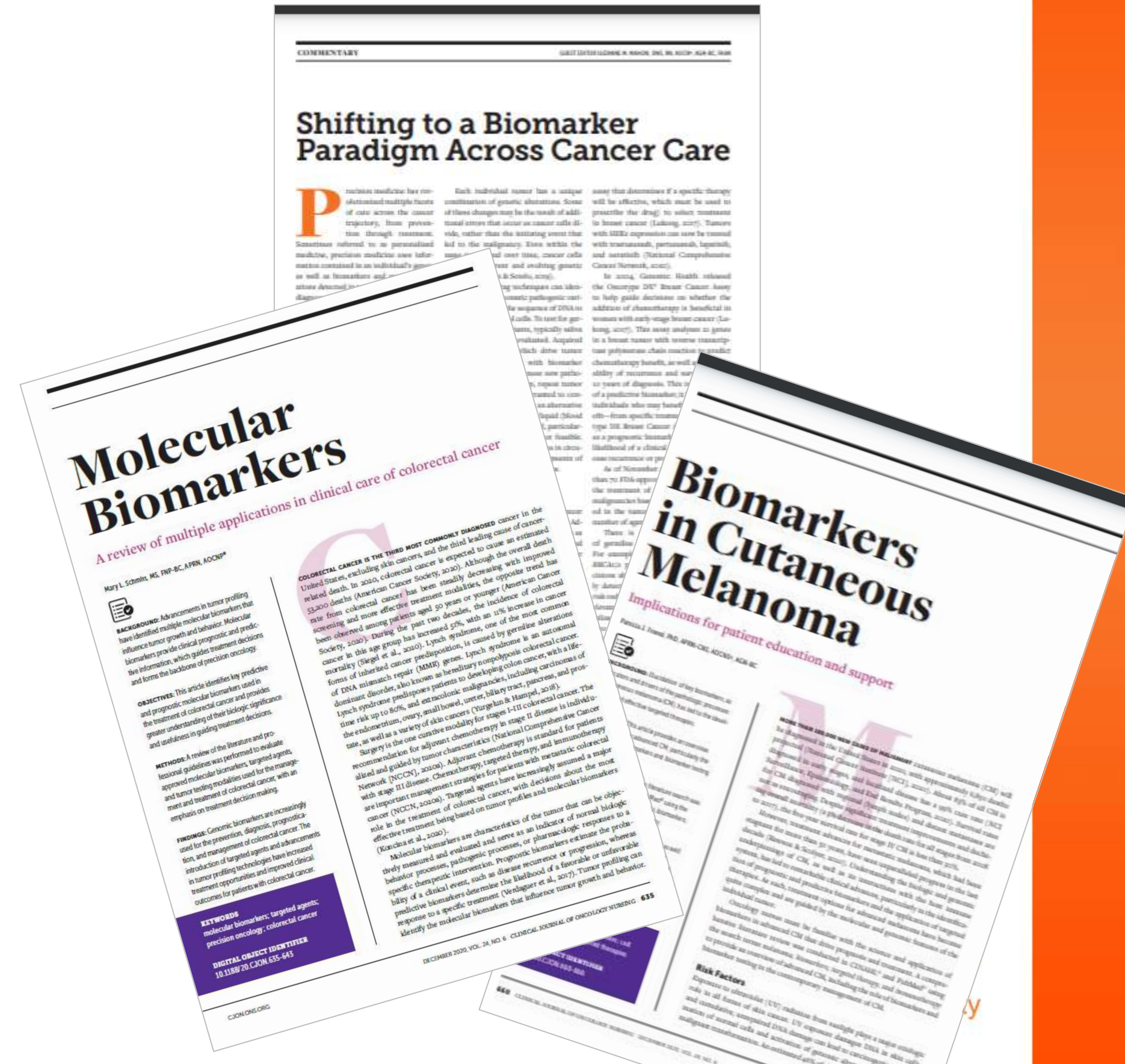
Variants associated with disease prognosis or diagnosis



# ONS Podcasts and Publications

## Interviews with Key ONS experts that include additional resources to supplement the episode

- What you Need to Know About At-Home Genetic Testing
- Understanding Genomics in Oncology Nursing
- Hereditary Cancer Genetics-ONS Congress
- Navigating Genomics and its Effect on Cancer Care
- The History of Genomics in Cancer Care
- Nurse Innovators Increase Access to Biomarker Testing with ONS Hackathon
- Biomarker Testing Improves Outcomes for Patient with Non-Small Cell Lung Cancer
- Genomics Must be a Priority in Oncology Nursing Education





# Welcome, Learn more about Biomarkers

Curated for oncology nurses by oncology nurses to bring precision oncology and the most recent biomarker advances to the point of care

Learn More

Search by biomarker, cancer type, or targeted therapy.

Search

Refine by:

BRCA1 X

Cancer Types

Targeted Therapies

Showing 1-1 of 1 Results

## BRCA1

Show Details >>

Prostate Cancer

### Implications for Patient Care

BRCA1 is a susceptibility biomarker, and pathogenic variants in germline testing indicate increased risk of breast and other cancers. BRCA1 is a prognostic biomarker, and germline variants are...

### Quick Links

[Testing](#)

[Additional Considerations](#)



Updated 12/12/2023

# BRCA1

## Prostate Cancer

*BRCA1* (Breast Cancer gene 1) is a DNA repair gene that identifies and corrects double strand errors as part of the HRR DNA pathway. Every human has two copies (alleles) of *BRCA1*, one inherited from each parent. Variants in *BRCA1* may be germline or develop somatically during carcinogenesis. [1,2].

If you want to see all biomarkers associated with Prostate Cancer, [click here](#).

### Implications for Patient Care

*BRCA1* is a [susceptibility biomarker](#), and pathogenic variants in germline testing indicate an increased risk of breast and other cancers.

*BRCA1* is a [prognostic biomarker](#), and germline variants are associated with aggressive prostate cancer [1].

*BRCA1* is a [predictive biomarker](#), and germline and somatic variants in genes that repair DNA damage are associated with the HRR pathway and may indicate response to PARP inhibitor therapy [1,3].

### Targeted Therapies

#### [Olaparib \(Lynparza®\)](#)

##### PARP Inhibitor

Indicated for the treatment of adult patients with pathogenic or likely pathogenic germline or somatic HRR gene-altered mCRPC who have progressed following prior treatment with enzalutamide or abiraterone [4,5,6].

#### [Patient Education](#)

## Related Links

[Genomics and Precision Oncology Learning Library](#)

[Genomics Taxonomy](#)

[Biomarker Testing Quick Guide](#)

[When to Refer to a Genetics Professional Quick Guide](#)

[Genetic Disorder Reference Sheet: BRCA1 and BRCA2 Hereditary Cancers](#)

[Next-Generation Sequencing Toolkit](#)

# Current Genomics Advisory Board



Kathleen Calzone,  
PhD, RN, AGN-BC,  
FAAN



Patricia Friend,  
PhD, APRN-CNS,  
AOCNS, AGN-BC



Patricia A. Kelly,  
DNP, APRN, CNS,  
AGN-BC, AOCNS



Danielle Fournier,  
DNP, APRN,  
AGPCNP-BC,  
AOCNP



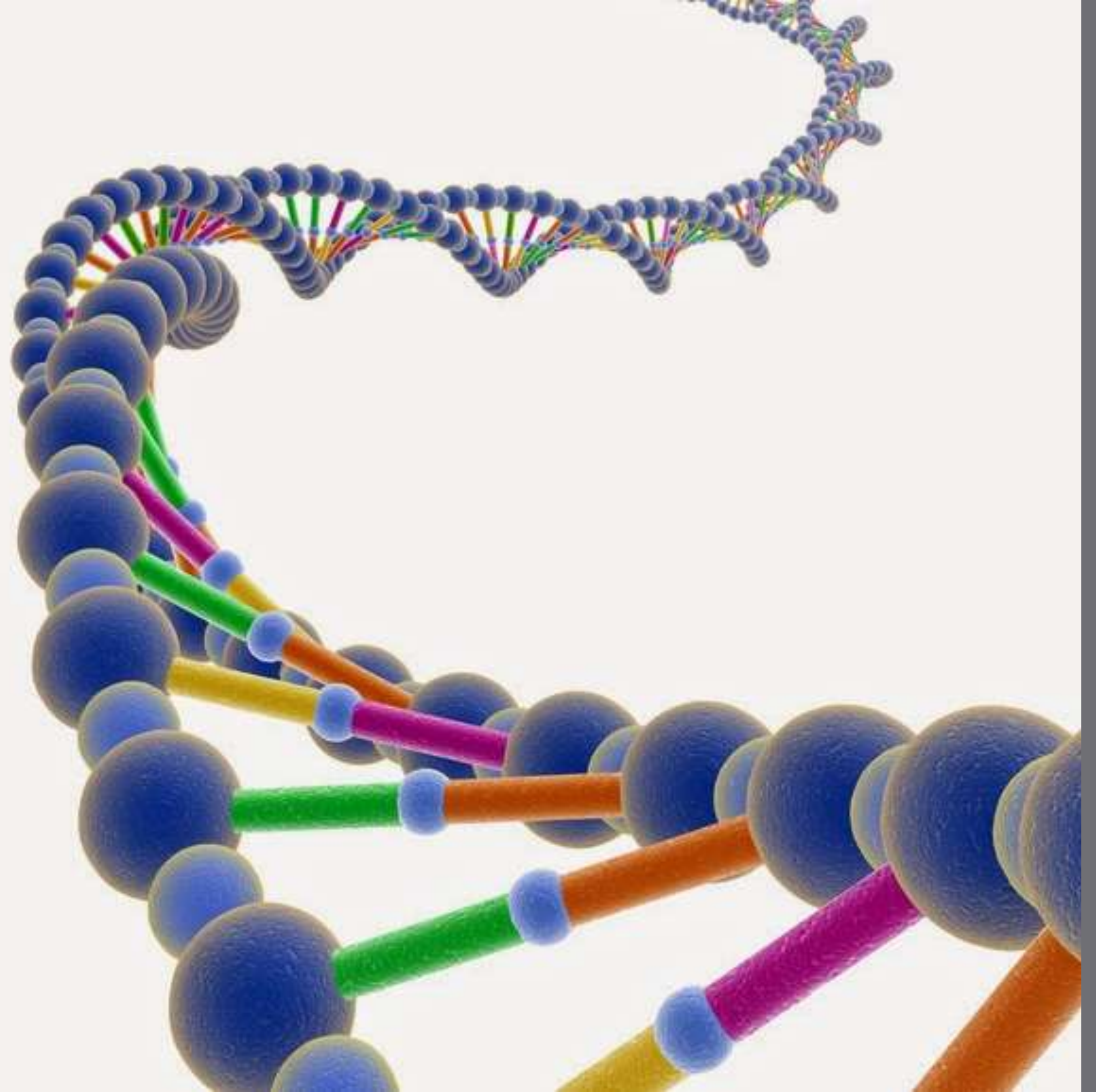
Suzanne M Mahon  
DNS RN AOCN®  
AGN-BC FAAN



Mary L. Schmitt,  
MS, APRN, FNP-  
BC, AOCNP



What's  
Next?





# Thank you

**For additional questions, please contact:**

Erin Dickman: [edickman@ons.org](mailto:edickman@ons.org)



# Presentation

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**Kathleen Calzone**  
PhD, RN, AGN-BC, FAAN

# Genomics: What Does Every Nurse Need to Know

*Kathleen Calzone, PhD, RN, AGN-BC, FAAN*

*Research Geneticist; Head, Genomic Healthcare Section*

*Genetics Branch, Center for Cancer Research*

*National Cancer Institute, National Institutes of Health, USA*



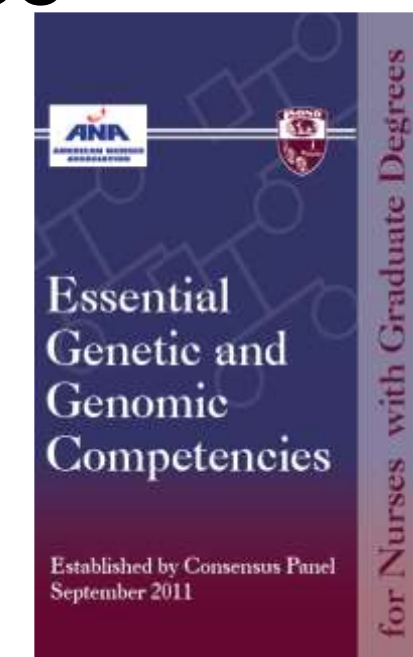
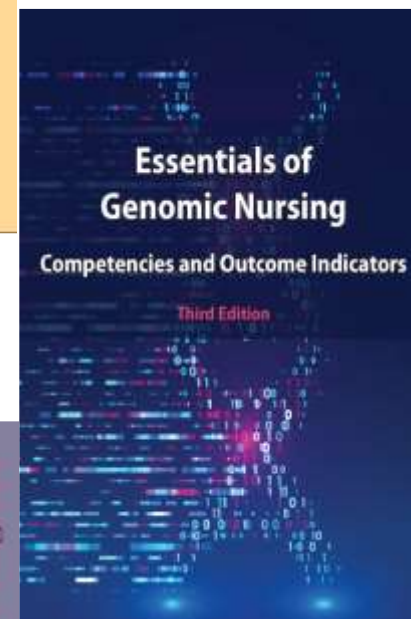
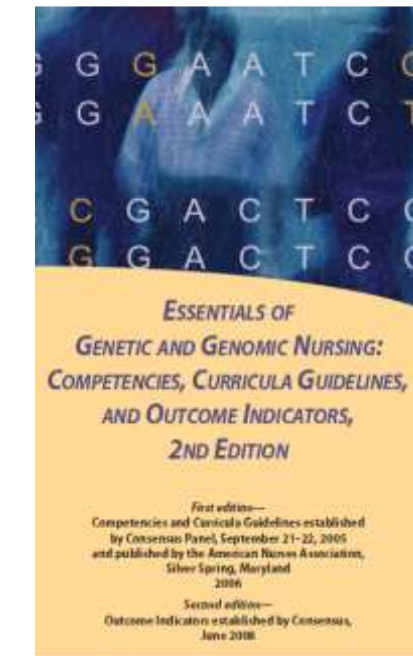
**NATIONAL CANCER INSTITUTE**  
Center for Cancer Research

2/6/2024



# Genomic Competencies: Collaboration with American Nurses Association

- Establish the minimum basis for nurses to deliver competent genomic nursing care
  - Competencies developed by a panel of nurse leaders from clinical, education, research and organizational settings
    - Endorsement
    - National Nursing Workforce Study
  - Second edition established outcome indicators consisting of learning outcomes and suggested clinical practice indicators
  - Third edition required because nursing competency and practice implementation in the US is highly uneven
    - Delphi study, revision, public comment, in production
    - National Nursing Workforce Study
  - Graduate competencies developed using similar panel
    - Under revision



Jenkins, J., et al. (2007). Establishing the essential nursing competencies for genetics and genomics. JNS. PMID: 17393960

Calzone, K., et al. (2011). Establishing the outcome indicators for the essential nursing competencies and curricula guidelines for genetics and genomics. JPN. PMID: 21596359

Greco, K., et al. (2011). Development of the essential genetic and genomic competencies for nurses with graduate degrees. ARNR. PMID: 22891504

Calzone, K. et al. (2024). Update to the Essential Genomic Nursing Competencies and Outcome Indicators. Submitted

# Four Practice Competency Domains

- **Nursing Assessment: Applying/Integrating Genomic Knowledge**
  - Demonstrates an understanding of the relationship of genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness.
  - Collects, reviews, and updates personal and family health history to include any genomic testing and environmental and other risk factors.
  - Conducts health and physical assessments that incorporate knowledge about known or potential environmental, genomic, and other risk factors (e.g., behavioral, lifestyle).
  - Assesses clients' knowledge, perceptions, and responses to genomic information about themselves and their family members.

# Four Practice Competency Domains, continued

## ■ Identification

- Evaluates assessment data to identify clients who may benefit from specific genomic information and services.
- Identifies credible, accurate, appropriate, and current genomic information, resources, services, and technologies specific to given clients.
- Identifies ethical, ethnic or ancestral, cultural, religious, legal, fiscal, and societal issues related to genomic information and technologies.
- Recognizes issues that undermine the rights of all clients for autonomous, informed genomic-related decision-making and voluntary action.



# Four Practice Competency Domains, continued

## ■ Referral Activities

- Facilitates referrals for specialized genomic services for clients as needed.

## ■ Education Care and Support

- Develops a plan of care in collaboration with the interdisciplinary team that incorporates genomic assessment information.
- Facilitates clients' access to credible, accurate, appropriate, and current genomic information, resources, services, and technologies.
- Advocates for autonomous, informed genomic-related decision-making.
- Demonstrates in practice the importance of tailoring genomic information and services that are responsive to the unique attributes of every person.

# Four Practice Competency Domains, continued

## ■ Education Care and Support, continued

- Uses health promotion and disease prevention practices that consider genomic influences as well as personal and environmental risk factors.
- Provides genomic health care in collaboration with interdisciplinary professionals and when possible clients and their families.
- Performs interventions appropriate to clients' genomic health care needs.
- Evaluates the impact and effectiveness of genomic interventions on clients' outcomes.

# Competency Outcome Indicators

- Specific Areas of Knowledge
- Clinical Performance Indicators

Domain: Professional Practice Nursing Assessment: Applying/Integrating Genomic Knowledge	
Demonstrates an understanding of the relationship of genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness.	
Specific Areas of Knowledge	Clinical Performance Indicators
<p>Relationship of genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness</p> <p>Relationship of genomics to normal physiology and pathophysiology, including:</p> <ul style="list-style-type: none"> <li>■ basics of gene function and genomic variants in individuals and populations</li> <li>■ germline variants, somatic variants, and polymorphisms</li> <li>■ selected variants associated with single-gene disorders, and chronic disease</li> <li>■ concept of genotype and phenotype</li> <li>■ selected genotype predictors for disease prognosis and treatment</li> </ul> <p>Basic principles of pharmacogenomics:</p> <ul style="list-style-type: none"> <li>■ polymorphisms and drug metabolism</li> <li>■ selected examples (e.g., Warfarin and CYP polymorphisms)</li> </ul> <p>Patterns of disease associated with single-gene and multifactorial inheritance</p>	<p>Collect a client's personal and three-generation family health history to assess for genomic factors that impact the client's health.</p> <p>Identify potentially significant information from the family health history.</p> <p>Identify a client who might benefit from referral to genetic specialists or information resources.</p> <p>Facilitate appropriate referral to genetic specialists, accurately documenting and communicating relevant history and clinical data.</p> <p>Describe a typical client journey that might be experienced in the process of genetic counseling.</p> <p>Describe the genomic factors that contribute to the variability of responses to pharmacologic agents.</p> <p>Incorporate genomic health assessment data into routinely collected biopsychosocial and environmental assessments of health and illness parameters in the client, using culturally sensitive approaches.</p> <p>Identify resources available to assist the client seeking genomic information or services, including the types of services available.</p>



# Genomics is a Quality and Safety Healthcare Issue

- Adverse Drug Events are adverse events that occur in **correctly prescribed and administered** medications
- 2017-2019 there were approximately 6 Emergency Department (ED) visits per 1000 persons per year that were associated with medications ADEs of which 38.6% required hospitalization
- 14.9% were anticoagulants and 21% were associated with therapeutic use
- Common anticoagulants such as warfarin and clopidogrel both have long standing pharmacogenomic Clinical Pharmacogenetics Implementation Consortium (CPIC<sup>®</sup>) guidelines
- There are 26 CPIC<sup>®</sup> evidence-based guidelines covering >140 common medications, i.e. anti-depressants, anti-coagulants, pain control, antibiotics, and anti-emetics with associated FDA drug labeling

Budnitz, D.S., (2021). US emergency department visits attributed to medication harms, 2017-2019. JAMA, PMID: 34609453.  
Clinical Pharmacogenetics Implementation Consortium. (2024). Clinical Pharmacogenetics Implementation Consortium.  
<https://cpicpgx.org/>

# Nurses with Prescriptive Privileges

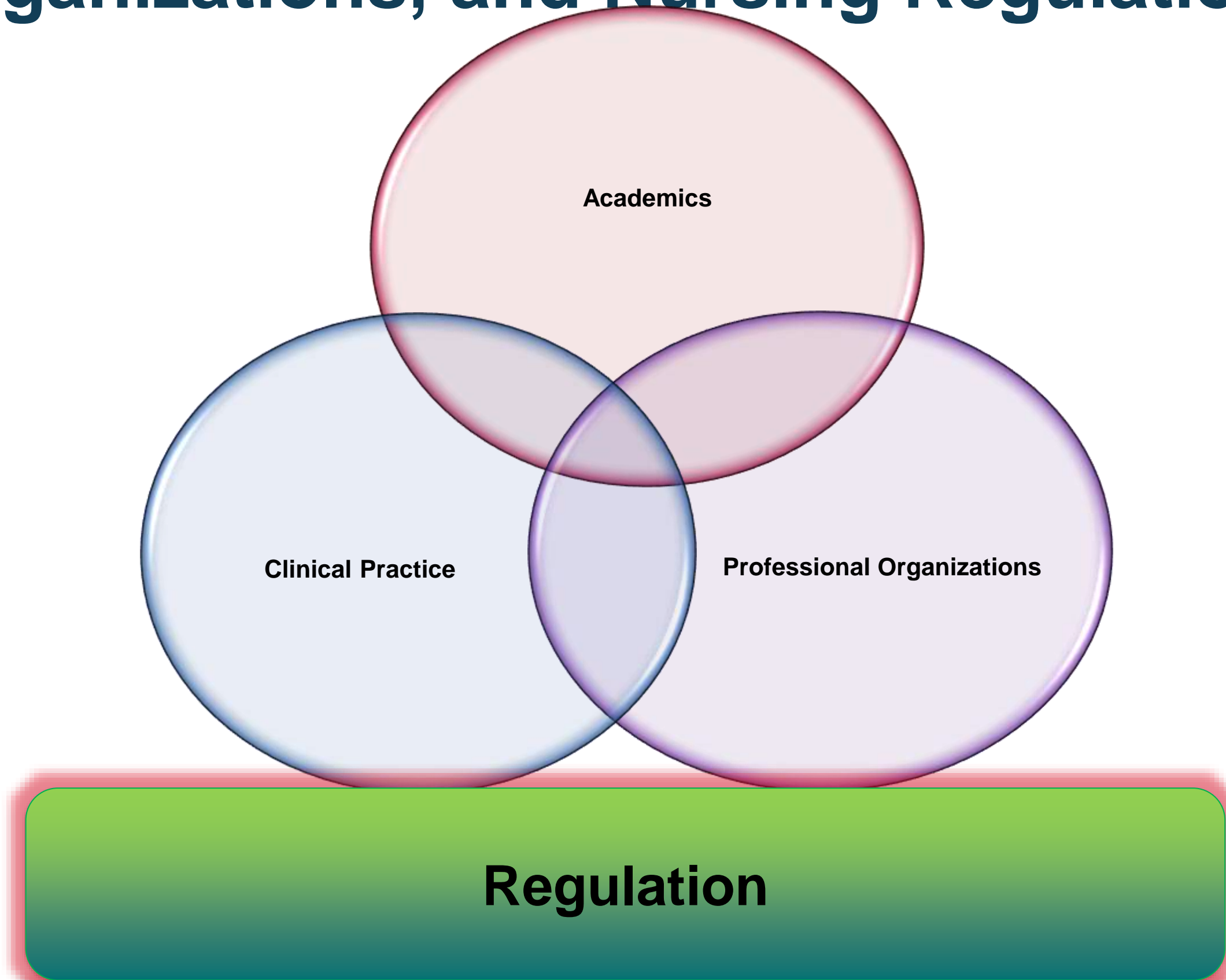
- Most (84%) reported they had never utilized CPIC<sup>®</sup> Guidelines.
- Utilization of pharmacogenomic testing defined as ordering a test in the past year was low (29%)
- When asked why they have not ordered a pharmacogenomic test, 48% indicated they did not know what test to order
- 52% reported they had no pharmacogenomic curricular content
- Nurses who graduated more recently were more likely to have reported some pharmacogenomics in their curriculum ( $p=0.031$ )
- Pharmacogenomic curricular content was associated with self-reported familiarity with pharmacogenomics ( $p=0.045$ ) but NOT confidence in their pharmacogenomic knowledge ( $p=0.615$ )

# Genomics: A Biospecimen Is Not Always Required





# Genomic Implementation Involves the Interface Between Academics, Clinical Practice, Nursing Organizations, and Nursing Regulation



# Genomics is Complex Competency

- Nursing has little to no foundational underpinning in genomics
  - Differs from other change initiatives
- Outcomes of appropriately used genomics applications may not be observable
  - Lack of observability slows adoption rates
- The language of genomics is not understood by general healthcare providers
  - Limits capacity to read and understand the literature and talk with colleagues
- Genomic applications benefit from infrastructure
  - Point of care decision support, documentation capacity, i.e. pedigree
  - Requires leadership to have sufficient competency to recognize value of supporting these efforts

# Genomics is Complex Competency, continued

- Waiting for the future workforce to solve this problem is unrealistic
  - Some studies have shown that faculty genomic knowledge is equivalent to the students
  - Existing US licensure and certification exams have limited to no genomic content
- The existing training model that includes clinical experiences is not feasible in the absence of clinical integration
  - Novel strategies such as the use of standardized patients for training need to be considered
- This is an interprofessional competency issue
  - You may not have a colleague to go to for help



# Thank you

Calzonek@mail.nih.gov  
240-760-6178



# Presentation

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**Patricia Sharpnack**

DNP, RN, CNE, NEA-BC, ANEF, FAAN



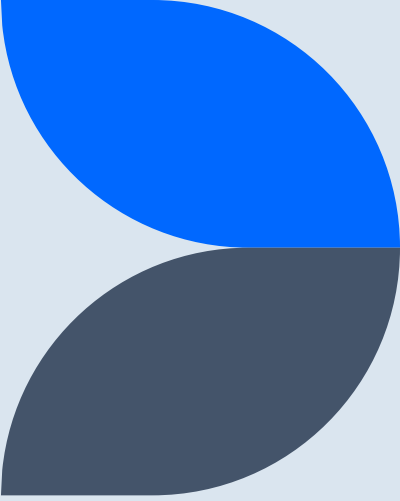
# Designing a Framework for Competency-Based Education

Patricia A. Sharpnack DNP, RN, CNE, NEA-BC, ANEF, FAAN





# NLN Vision Statement on CBE



Nursing education is at a crossroads. Challenges include growing workforce demand, nurses' disillusionment with practice realities, long-standing disparities in care delivery, acknowledged history of racism and bias within the profession and its educational processes, digital transformation, and employer dissatisfaction with new graduate capabilities (Kavanagh & Sharpnack, 2021). All these challenges prompted nursing education leaders across the nation, including the National League for Nursing, to reevaluate nursing education methods and long-standing practices and turn toward CBE as a model for the future.

# ANA Membership Assembly

## June 2021



We are suggesting outreach efforts to ANA members for discussions on establishing a strategic initiative to integrate Precision Health and Genomics (PH&G) into basic and advanced nursing practice. This would include but not be limited to:



1. Establish entry-level and advanced nursing competencies for Precision Health that will inform policy and practice recommendations.



2. Update the Genomic Nursing Competencies for Nurses with Graduate Degrees

# ANA Continued...



3. Integrate the PH&G competencies into all nursing scopes and standards of practice inclusive of practice specialties.



4. Assess the state of PH&G Nursing capacity in the existing nursing workforce to inform an education initiative and provide the basis by which to measure outcomes.



5. Address deficits in nursing knowledge, skills, and attitudes (KSAs) uncovered in the PH&G nursing capacity assessment. This should include demonstration projects leading to evidence-based best practices







# Development of the Toolkit:

## Primary goals

Provide resources to facilitate the identification and mapping of key competencies for practice-ready nurses.

Develop strategies for nursing programs to integrate competency-based education into the curriculum with a particular emphasis on assessments as key for measuring learning and mastery of competencies.

Identify tools and resources to assist nursing programs in developing a competency-based education curriculum including the collection and use of data for facilitating learning and mastery and systematic evaluation of learning outcomes.



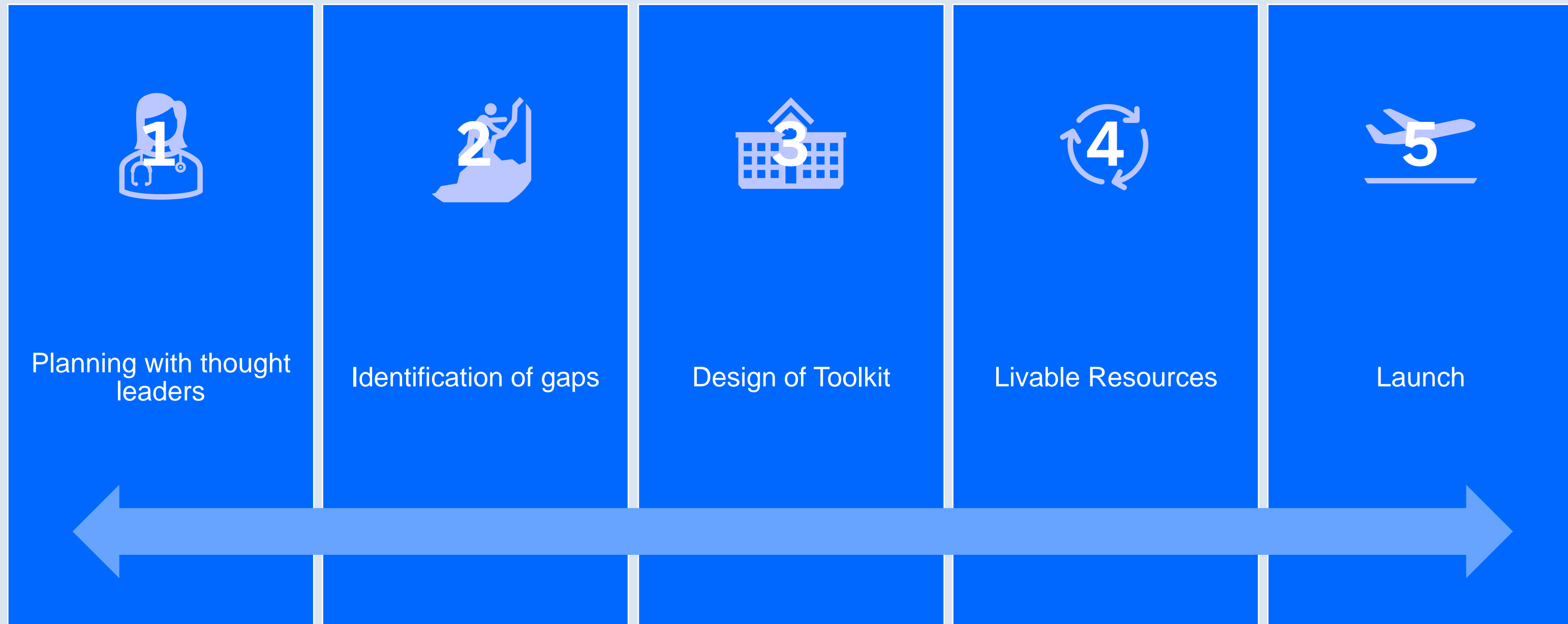
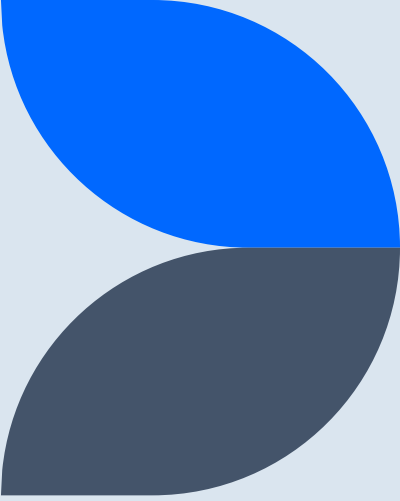
# Framework



Assessment focus	Proposed methods of assessment
Readiness to cope with challenges in practice	Same as does + Entrustment-based assessments (e.g., discussion)
Performance integrated into workplace	360 <sup>o</sup> assessment, Case-based discussions, Clinical competency assessments, DOPS, Multi-source feedback, Portfolios, Work place-based assessment
Performance in controlled settings	OSCEs, Practicals, Simulations, Standardized clients / patients
Application and Manipulation of knowledge, Relationships between concepts and principles	Case presentations, Essays, Gaming, Extended matching MCQs, Problem-solving approaches
Fact gathering, Processes, Scientific principles	Essays, Oral exams, Reports, Traditional MCQs, Various tests

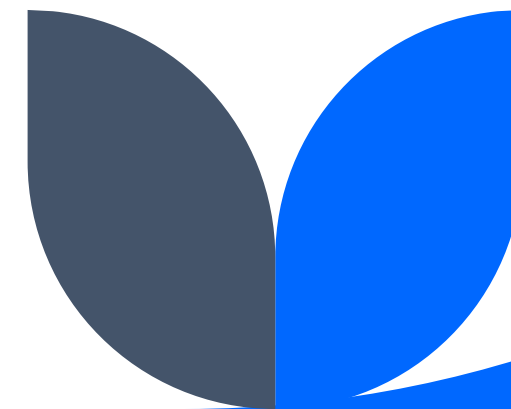


# Plan for toolkit launch



# Toolkit Components

- I. **Introduction** – (NLN mission, core values, The WHY, relevance)
- II. **Background** (Research)
  - a. Framework
  - b. What CBE is and what it isn't?
  - c. How can it be tailored to *schools*?
  - d. Information about what has been done in CBE—implementation, assessment, measurement/evaluation
  - e. How schools can modify CBE to meet their needs (bite-size pieces)
- III. **Implementation of CBE** – (livable and usable resources – how to...) NOT based on AACN Essentials -
  - a. Phases of implementing CBE (the how-to)
  - b. How to get buy-in (buy-in may be differentiated for different groups, e.g., deans, faculty)
  - c. Exemplars – examples of schools that have implemented CBE fully and those that have implemented CBE partially
  - d. Steps in an orientation plan for faculty
- IV. **Clinical and didactic** – include simulation
  - a. Differentiation of clinical and didactic
  - b. How to align clinical and didactic
  - c. EPAs included in this area
- V. **Curriculum redesign**
- VI. **Systematic Evaluation** (assessment and measurement) –
- VII. **Recommendations**
- VIII. **Conclusions**



# Future Plans

- 2024 Summit
- Workshops
- Others





The future is not inevitable; it is inventible.



# Thank you

[psharpnack@ursuline.edu](mailto:psharpnack@ursuline.edu)



## Q&A

### PANEL 3

# Precision Health: Implications for Curriculum

---

**Moderator: Connie Delaney, PhD, RN, FAAN, FACMI**

- **Erin Dickman, DNP, RN, OCN**
- **Kathleen Calzone, PhD, RN, AGN-BC, FAAN**
- **Patricia Sharpnack, DNP, RN, CNE, NEA-BC, ANEF, FAAN**



**We'll be right back with our next session**

## **PANEL 4**

# **Equity, Ethical and Legal Issues in Precision Health**

---

**Moderator: Dorcas Kunkel, DNP, RN/PHN, CNE, CPHIMS**

- **Edwin Aroke, PhD, CRNA, FAANA, FAAN**
- **Elaine Petty, RN, MALS**
- **Laurie Badzek, LLM, JD, MS, RN, FNAP, FAAN**

# PANEL 4

## Equity, Ethical and Legal Issues in Precision Health

---

**Moderator: Dorcas Kunkel, DNP, RN/PHN, CNE, CPHIMS**

- **Edwin Aroke, PhD, CRNA, FAANA, FAAN**
- **Elaine Petty, RN, MALS**
- **Laurie Badzek, LLM, JD, MS, RN, FNAP, FAAN**

# Presentation

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**Edwin Aroke**

PhD, CRNA, FAANA, FAAN



# Precision Health Equity: Can precision health reduce disparities in health care?

Edwin N. Aroke, PhD, CRNA, FAANA, FAAN  
Associate Professor

# Disclosure



National Institute of  
Arthritis and Musculoskeletal  
and Skin Diseases



**AANA  
FOUNDATION**



**UAB**

**Center for Addiction and Pain  
Prevention and Intervention**

**UAB**

**MINORITY HEALTH & HEALTH DISPARITIES  
RESEARCH CENTER**

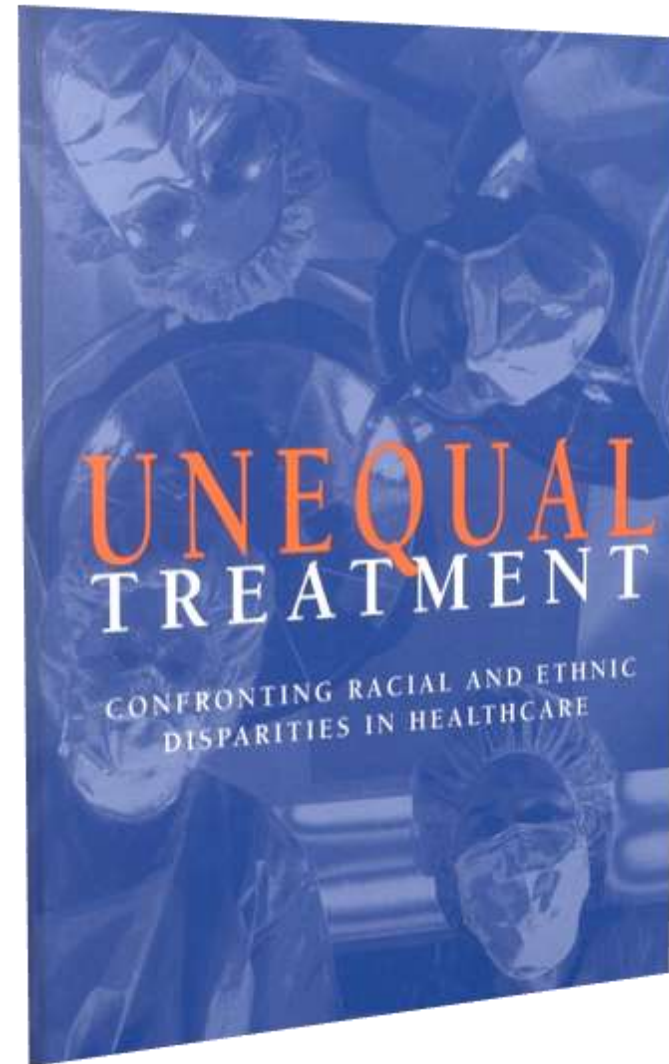
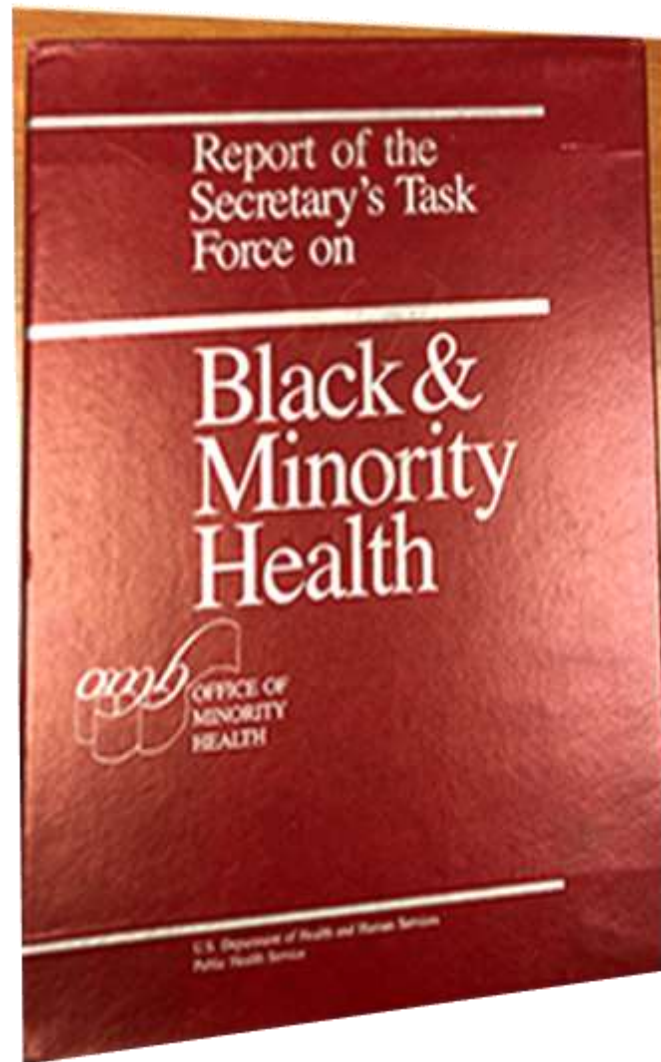
The University of Alabama at Birmingham



National Institute  
on Minority Health  
and Health Disparities

**Thank you to funders, participants, lab members, and collaborators**

# Healthcare disparities are well-documented



African American baby is more than twice as likely to die in the 1<sup>st</sup> year of life than a white baby

Racialized minorities experience more greater pain severity and pain-related disability

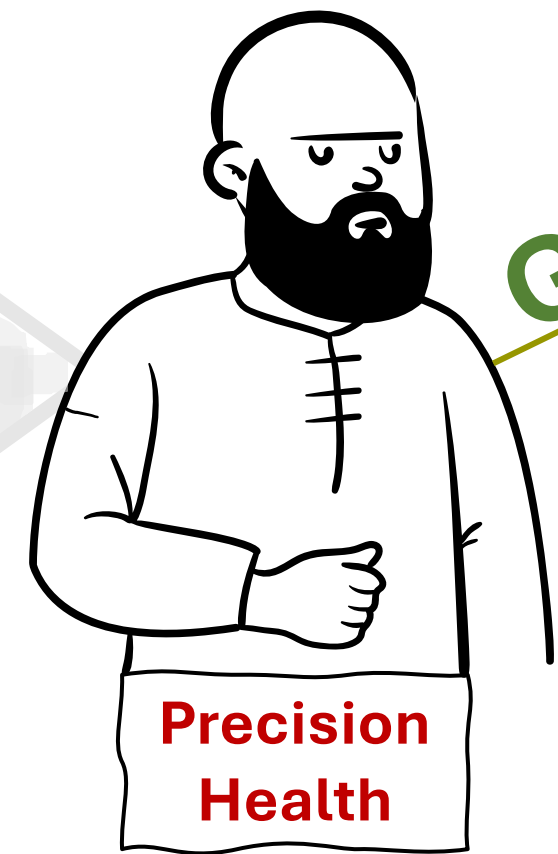
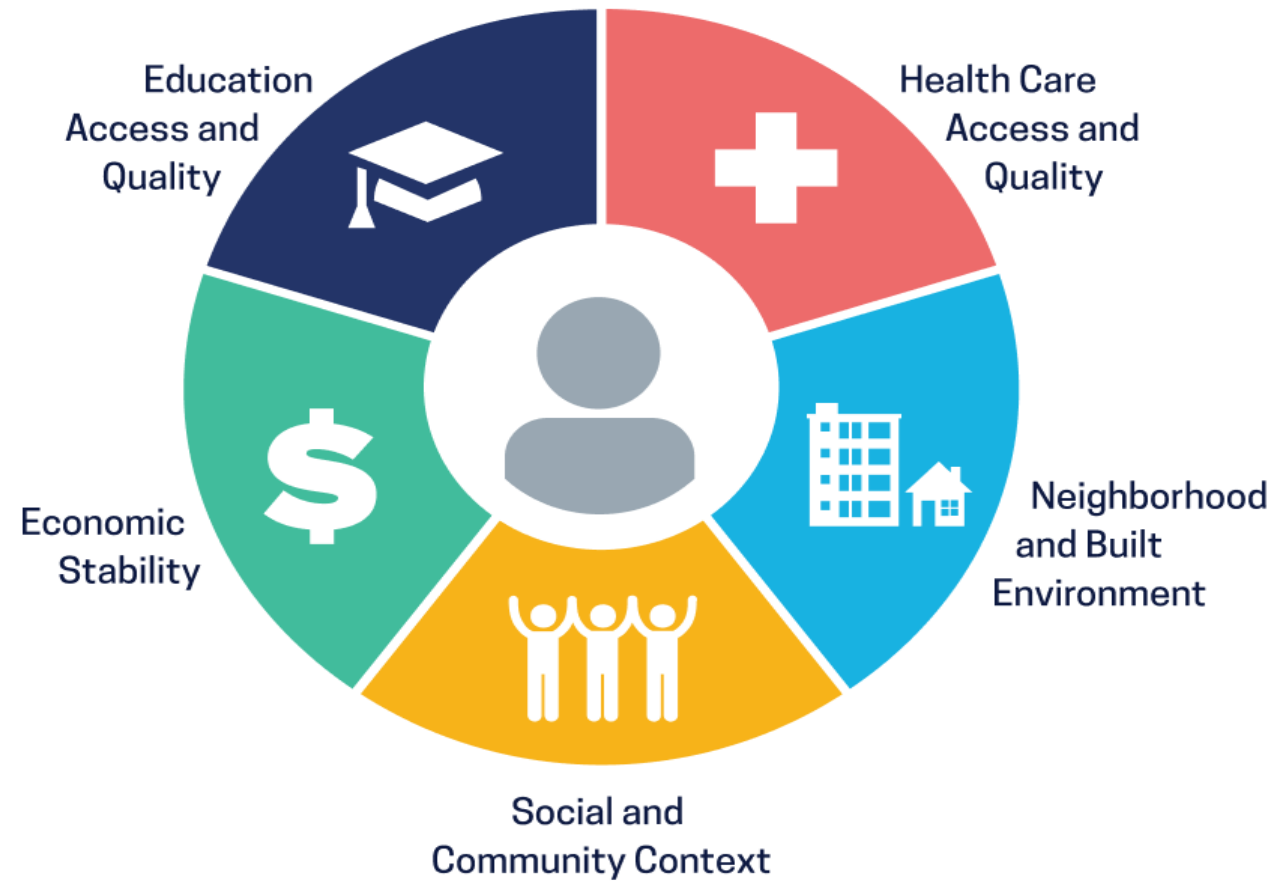
Healthy People 2030



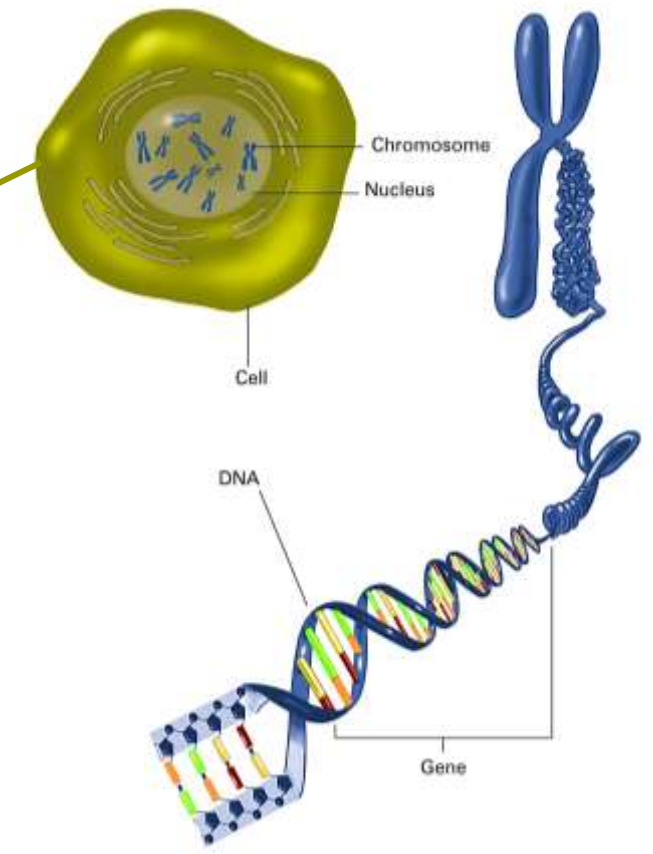
What is the “something” that is so damaging to the health of underrepresented individuals?

# Root causes of health disparities

## Social Determinants of Health



**Genes**



**SDOH accounts for about 80% of variance in health outcomes**

- Humans are over 99% identical**
- Genetics/genomics
  - Transcriptomics
  - Metabolomics
  - Proteomics
  - Pharmacogenetics

**Race is a social construct, NOT a biological variable**





# How do social experiences get under the skin and cause health disparities?



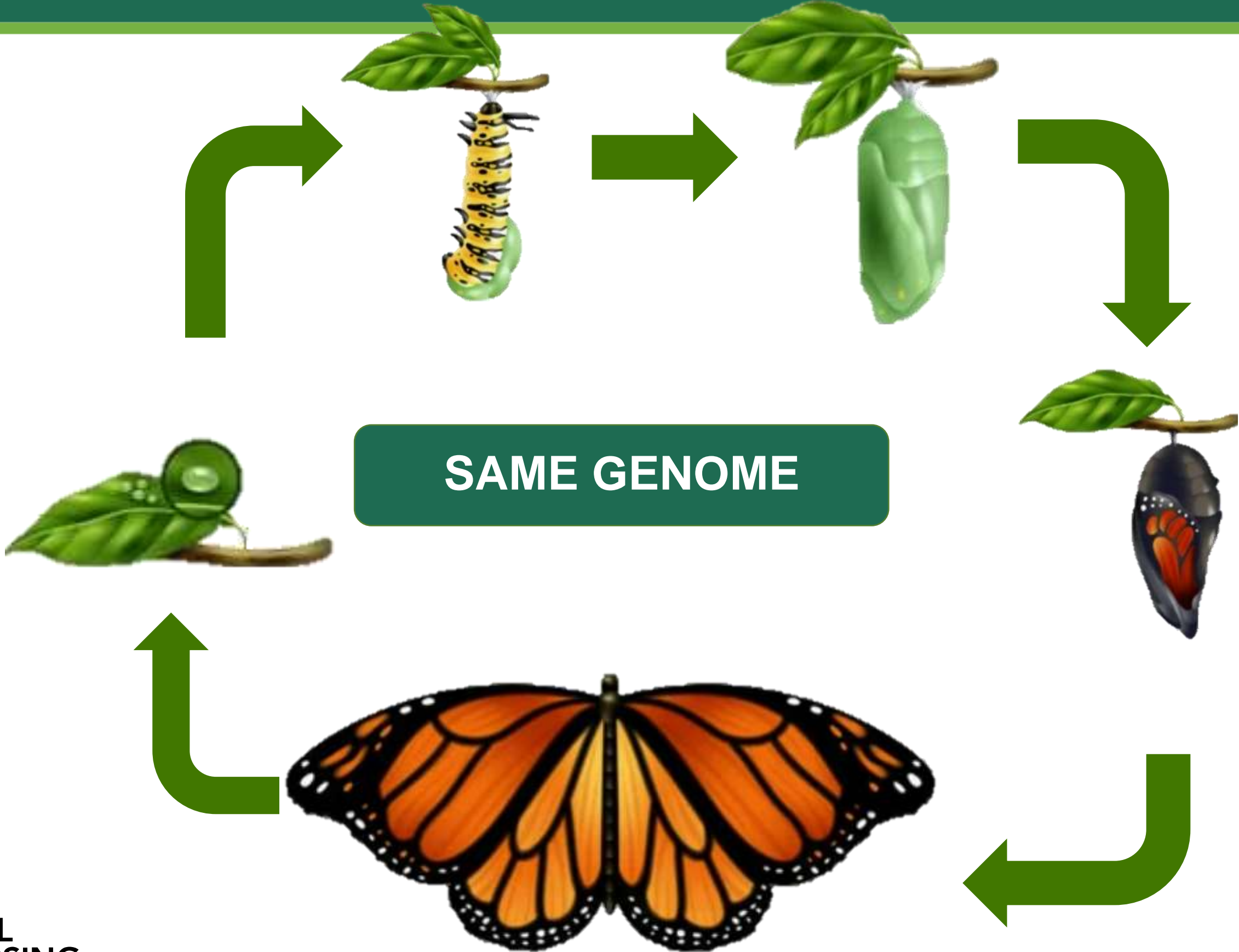
The University of Alabama at Birmingham

## Epigenetics

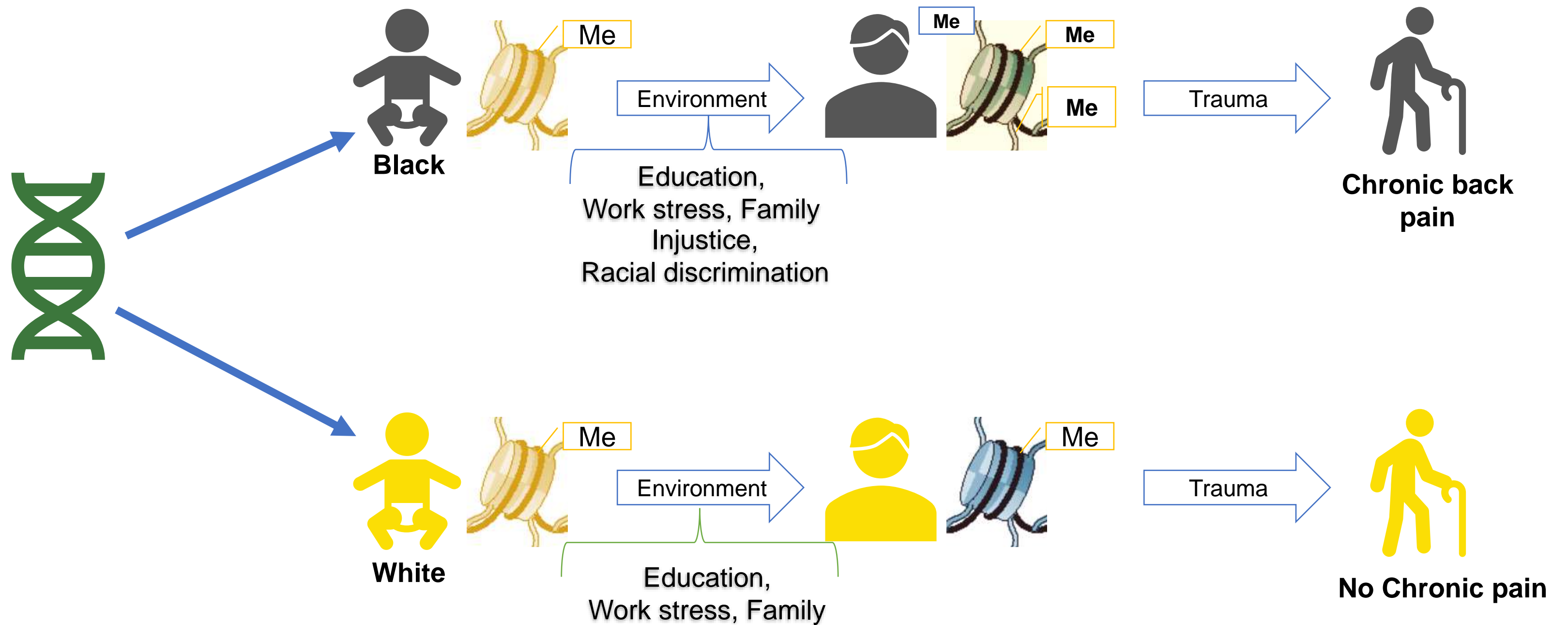


Image courtesy of NIEHS

# Why the epigenome?

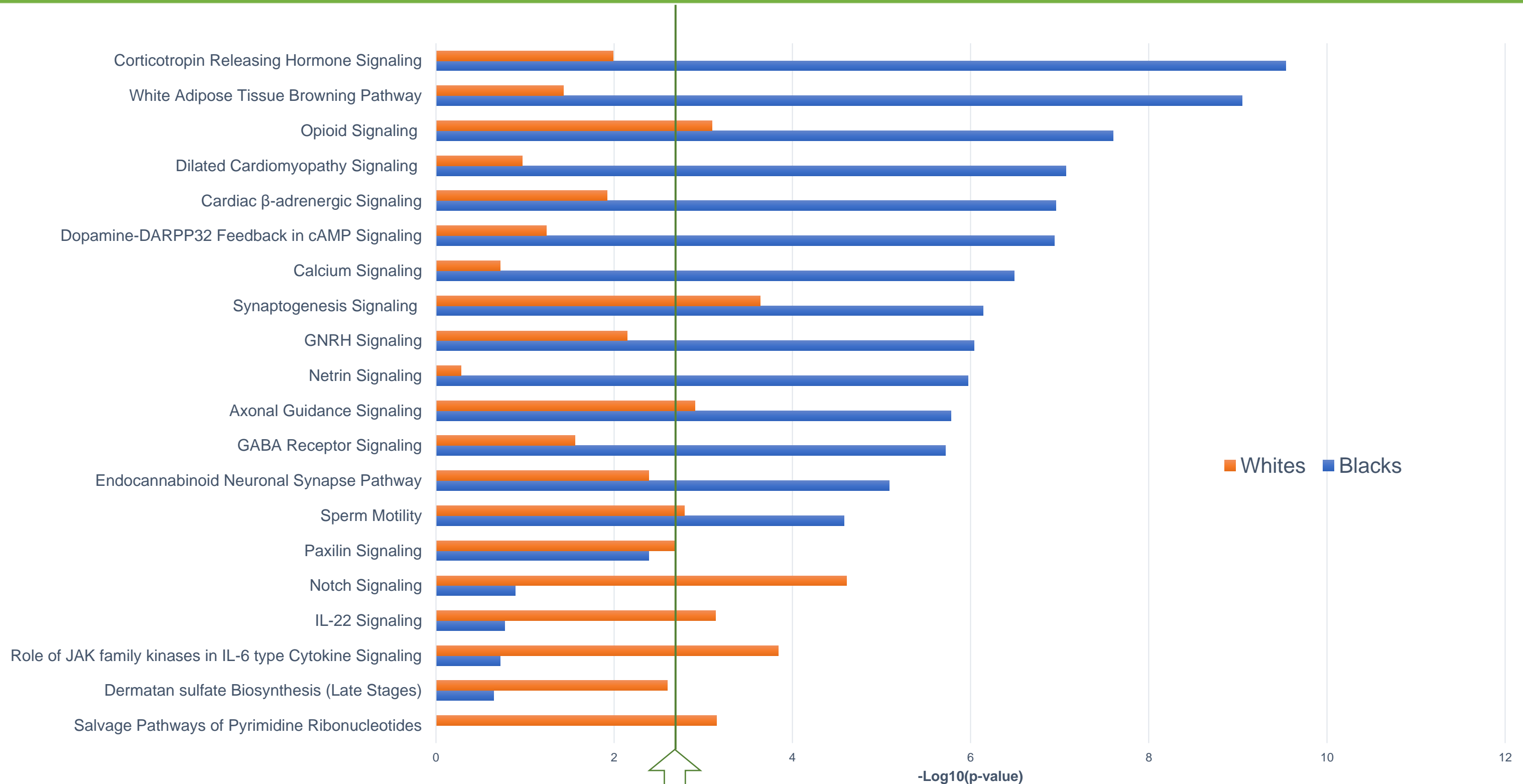


# Could epigenetics help explain racial disparities in chronic pain?



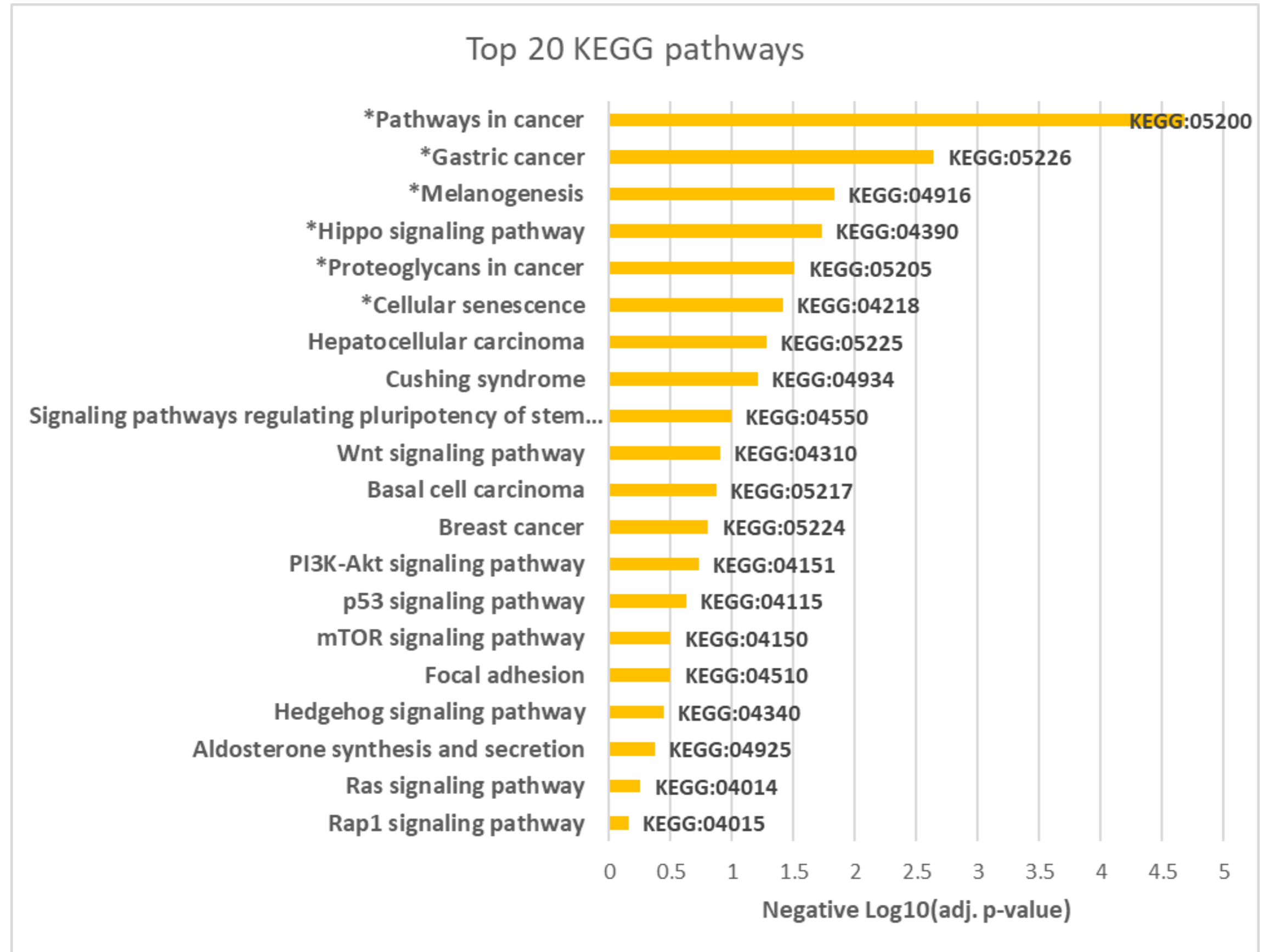


# Pain and stress related genomic pathways are more significant in Blacks than Whites with chronic low back pain



# Internalized stigma is linked to worse pain outcomes

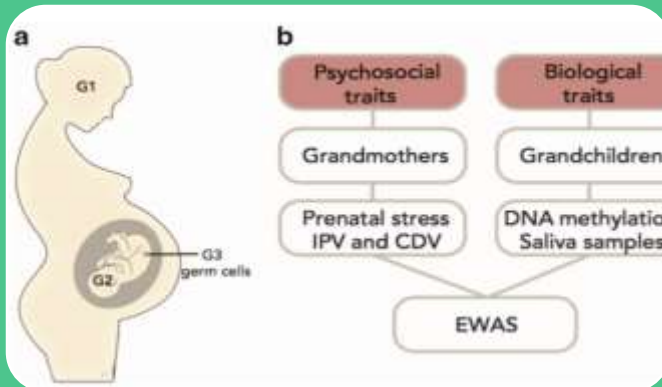
Pathways previously associated with stress and stress-related disorders are enriched by differentially methylated genes between adults with low versus high internalized stigma



# Transgenerational transmission of stress and trauma



Maternal psychosocial stress epigenetically alter stress genes in offsprings



Grandmother stress from intimate partner abuse alter DNA methylation in 3<sup>rd</sup> generation offsprings



Perceived discrimination linked differential DNA methylation in African American women with hypertension



# Advancing health equity with precision health

Move beyond race as a proxy and directly measure concepts like racism, injustice, discrimination

Lack of diversity and representation in genomics studies, including the reference genome

Ethical, legal, social, and access concerns

Epigenetic Changes are Dynamic and Reversible

The potential for precision health to revolutionize treatments is in its infancy

Holistic approach to care management that is tailored to the person's needs

Integrating lifestyle, environmental, and genetic information can foster health equity

# References

- Aroke, E. N., Joseph, P. V., Roy, A., Overstreet, D. S., Tollefsbol, T. O., Vance, D. E., & Goodin, B. R. (2019). Could epigenetics help explain racial disparities in chronic pain?. *Journal of pain research*, 701-710. <https://doi.org/10.2147/JPR.S191848>
- Aroke, E. N., Jackson, P., Meng, L., Huo, Z., Overstreet, D. S., Penn, T. M., ... & Goodin, B. R. (2022). Differential DNA methylation in Black and White individuals with chronic low back pain enrich different genomic pathways. *Neurobiology of Pain*, 11, 100086. <https://doi.org/10.1016/j.ynpai.2022.100086>
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- Serpeloni, F., Radtke, K., de Assis, S. G., Henning, F., Nätt, D., & Elbert, T. (2017). Grandmaternal stress during pregnancy and DNA methylation of the third generation: an epigenome-wide association study. *Translational Psychiatry*, 7(8), e1202. <https://doi.org/10.1038/tp.2017.153>

# Presentation

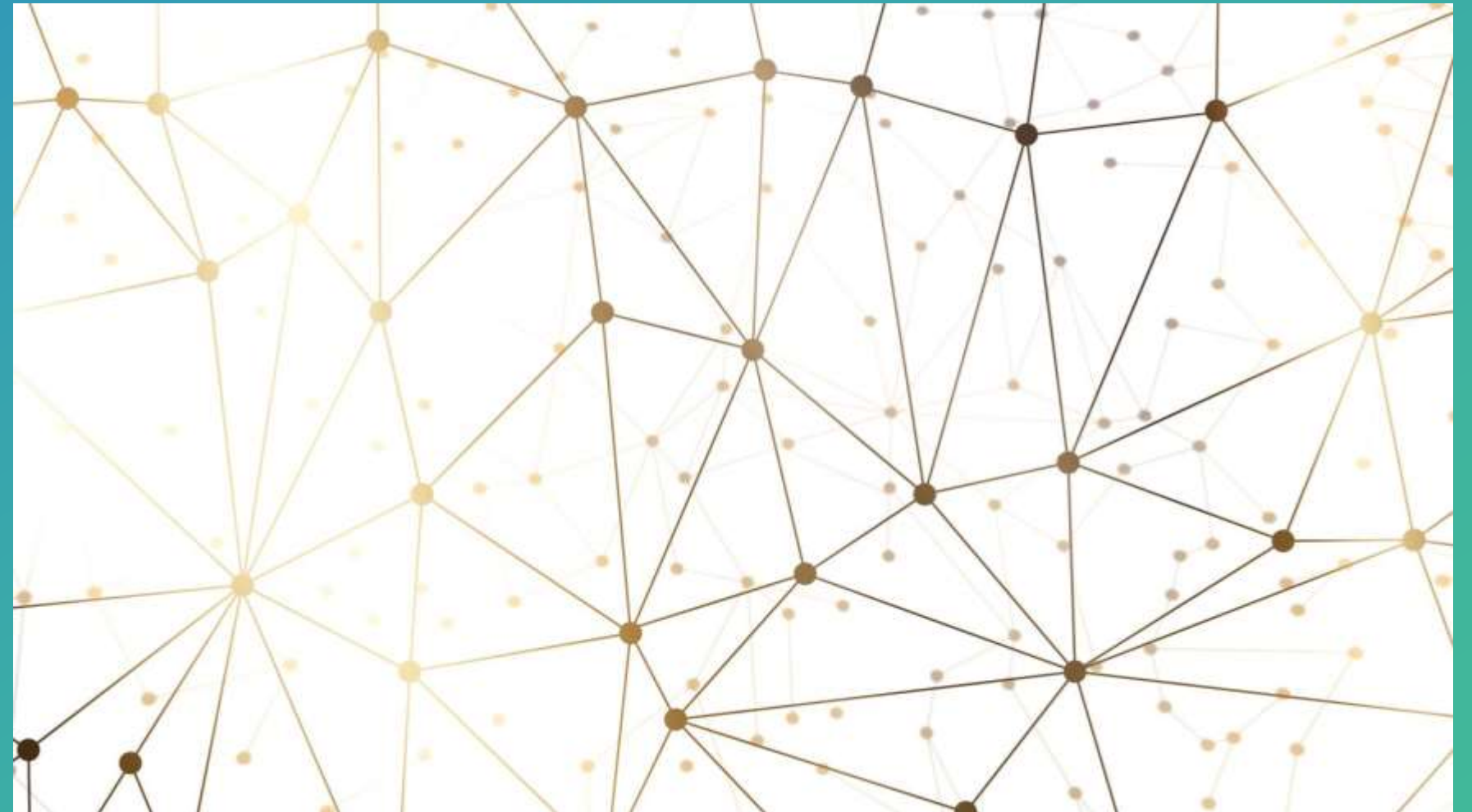
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**Elaine Petty**  
RN, MALS



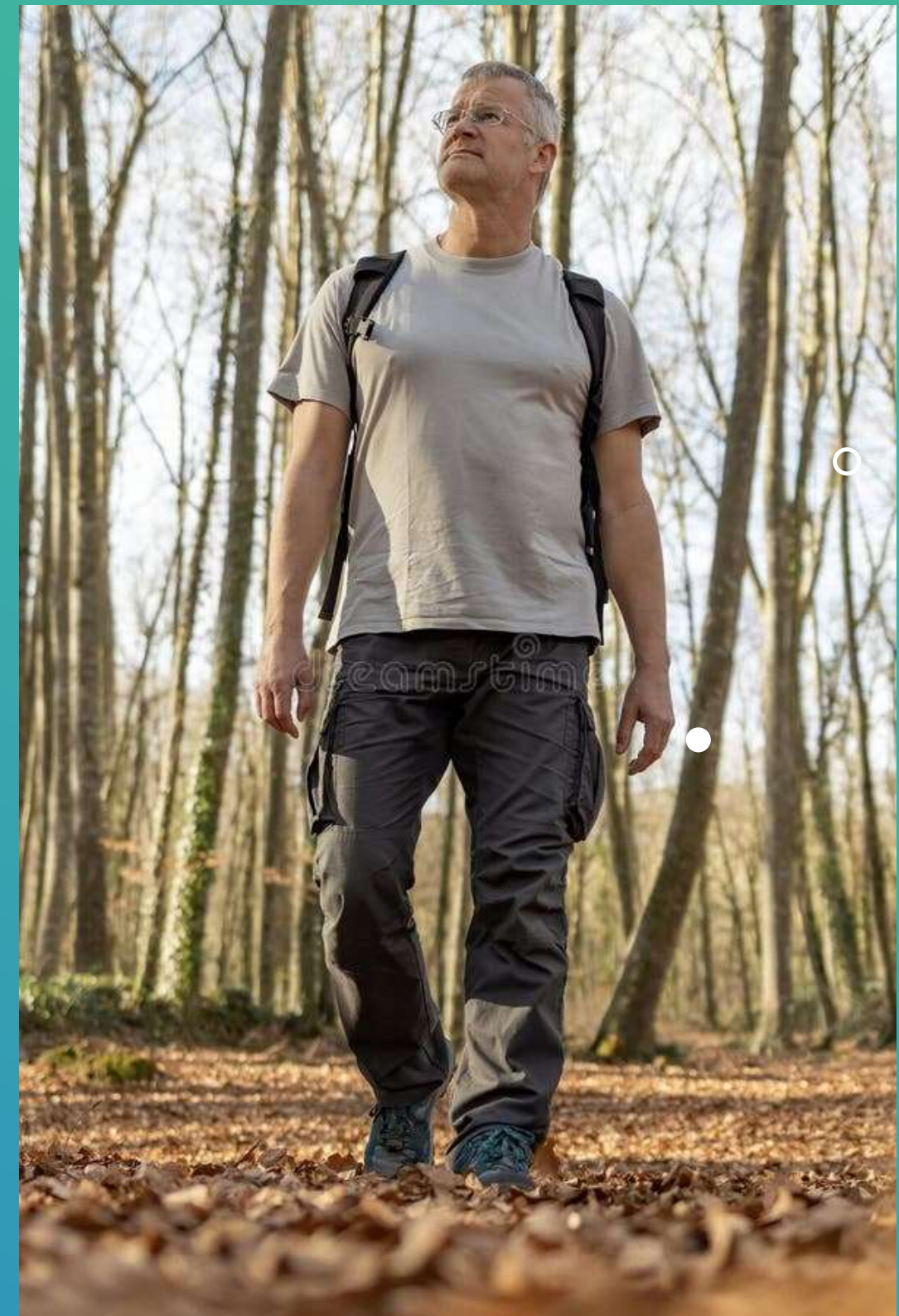
# ETHICAL RISKS & POTENTIAL HARMS IN PRECISION HEALTH

Elaine Petty, RN, MALS  
Pellegrino Center for  
Clinical Bioethics  
Georgetown University





# GENE THERAPY





# 23 and Me

# DNA Testing for Dogs





---

## Leon Kass

physician, scientist, philosopher

*The benefits of biomedical progress are obvious, clear, and powerful. The hazards are much less well appreciated.*



# Asking the right questions

Research & science asks:

“What can we do?”

Ethics asks:

“What should we do?”

“How & when should we do it?”

---

# What are some of the right questions to ask regarding ethics of pertinent issues, including possible dilemmas and risks?

Vast amounts of data is a key aspect of facilitating precision medicine

- Q: Who has access to data?
- Q: Who interprets the data? When should we use it? What scenarios should be prohibited?
- Q: What kind of education is necessary to interpret data?
  - Physicians, nurses, genetic counselors
- Q: What kinds of systems are required to manage & analyze vast amounts of patient data?

Unique and expanded role of nurses

- Q: What kind of engagement will be needed for patients to be part of the process?
  - Challenges concerning increased complexity of informed consent, family history, dynamic consent that changes over time, use of personal devices, additional findings.
  - How to ensure patients are fully informed about the risks & benefits?
  - How to build patient trust & engagement?
- Q: How to consider the therapeutic relationship - the patient as a person - as central?
- Q: How to design systems to ensure that omic testing aligns with patient preferences?





# Concerns from patient point of view

- Privacy & security of data
- Economic impact
- Possible harms, including psychosocial harms
- Risk for discrimination by certain groups
  - Insurance companies & employers
- Risks in the process of acquiring informed consent
- Mistrust in the provider and in medical research
- Issues with the diagnostic accuracy of precision medicine
- Changes in doctor-patient relationship

# Four Principles of Bioethics

## Autonomy

- Free & informed decision-making
  - Informed consent (pt education, risks & benefits, support, time)
  - Considering individual health literacy, community-based societies where therapeutic decisions are not made on basis of pts will alone

## Justice

- Based on fairness for all without discrimination
- Access for all people – can not be dependent socioeconomic status, ethnicity or limited by geography

## Beneficence

- Whether & under which conditions PM really has the potential to improve patients' quality of life – the life they themselves would judge as “good”

## Non-Maleficence

- Data security, privacy – risk of misuse of data
- Changes in the doctor-patient relationship due to focus on multi-omics data and whether the patient's subjective experience is thereby eclipsed.



# Edmund Pellegrino

physician, bioethicist, philosopher

The practice of medicine is not just a science, but also an art that requires a deep understanding of human nature and values. Bioethics should be grounded in the virtues of compassion, empathy, and respect for human dignity. To make ethical decisions, you must be an ethical person.



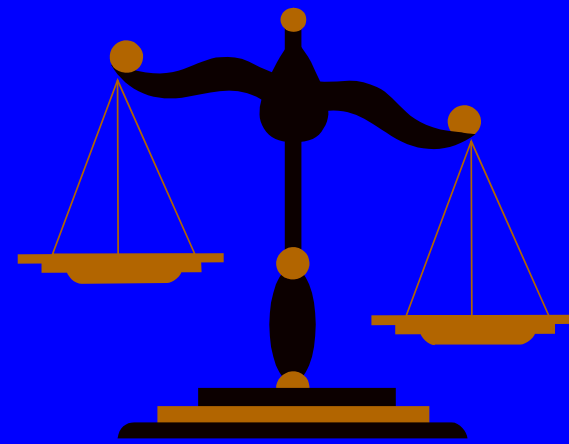
# Presentation

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**Laurie Badzek**

LLM, JD, MS, RN, FNAP, FAAN

# Legal Issues in Precision Health



Laurie Badzek, LLM, JD, MS, RN, FNAP, FAAN  
Ross and Carol Nese College of Nursing  
Penn State University

# Presentation Roadmap

1. Refresher on “the law”
2. Identify common areas of liability related to precision health
3. Some thoughts how to avoid pitfalls



# Introduction

Caring for patients potentially presents ethical and legal challenges for all nurses.

# Sources of Law

Constitutional

Legislation and Regulation

Judicial and Common Law

U. S.  
Constitution  
Bill of Rights



# Types of Law

- Substantive vs. Procedural
- Public vs. Private
  - Criminal
  - Civil
    - » contract law
    - » tort law
      - intentional
      - unintentional





# TORTS

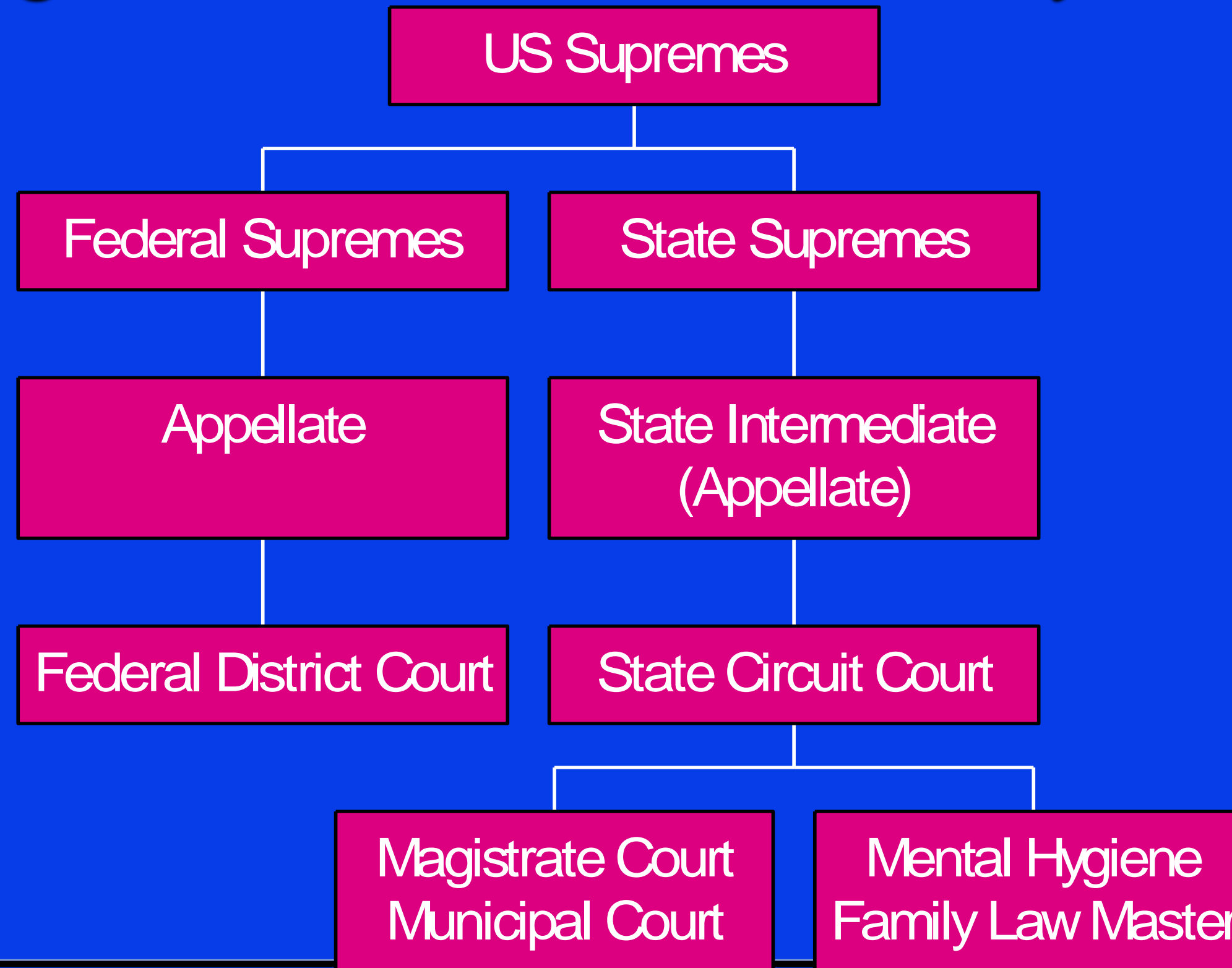
## Intentional

- defamation\*
- invasion of privacy\*
- assault & battery
- trespass
- int'l infliction of emotional distress\*

## Unintentional

- negligence
- malpractice

# Organization of the US Court System



# Precision Health and the Law

Covers topical areas related to

Health

Medicine

Genes

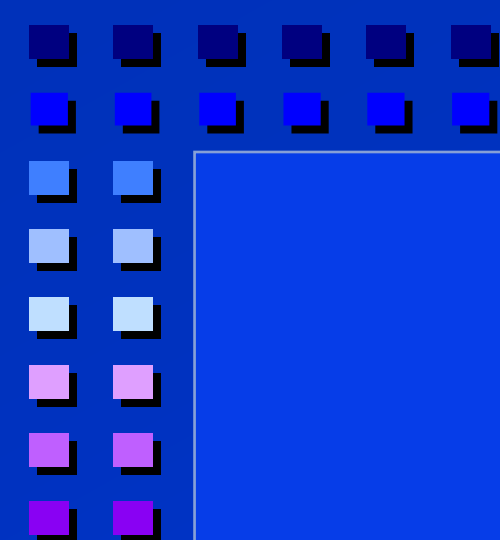
The Environment

Lifestyles

Research

And More.....





Quick search of legal literature in  
Nexus-Uni using terms  
“Precision Health”  
“Precision Medicine” :

Big Data Breaches Breaches

Patent Infringement



# Areas to look out for

- Data Security and Privacy, Data Sharing issues
- Discrimination
- Testing/Profiling
- Regulatory Compliance
- Patent disputes
- Product liability claims
- Consumer and Patient Rights



# More areas to look out for

- Informed consent
- Scope of Practice: competency & malpractice
- Patient/client communications
  - Documenting/HIPAA





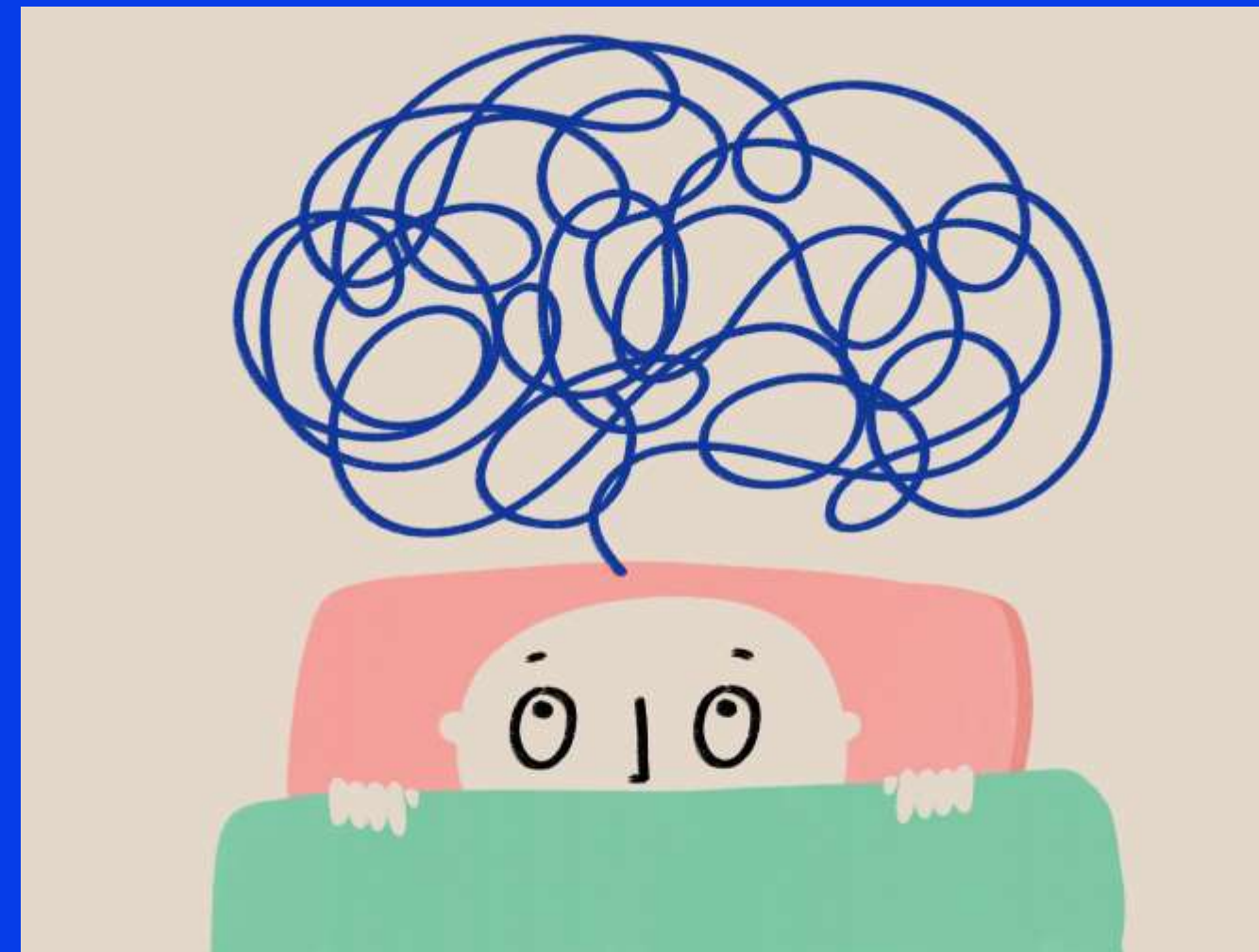
# Uncertainty

About the risks

What harm?

What enforcement?

What penalty?



# Communication

- Communication is often the key
- The patient/client record provides continuity of care
- Complete and accurate information = quality care, standard of care
- Participate in CEUs, Review competencies and be aware of standards

Thank You!!



## Q&A

### PANEL 4

# Equity, Ethical and Legal Issues in Precision Health

---

**Moderator: Dorcas Kunkel, DNP, RN/PHN, CNE, CPHIMS**

- **Edwin Aroke, PhD, CRNA, FAANA, FAAN**
- **Elaine Petty, RN, MALS**
- **Laurie Badzek, LLM, JD, MS, RN, FNAP, FAAN**

**We'll be right back with our next session**

## **PANEL 5**

# **Research and Funding in Precision Health**

---

**Moderator: Rebecca Schnall, PhD, MPH, BSN, FAAN, FACMI**

- **Elizabeth Eisenhauer, PhD, RN**
- **Theresa Koleck, PHD, RN**
- **Nonniekaye Shelburne, CRNP, MS, AOCN**

# **PANEL 5**

## **Research and Funding in Precision Health**

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**Moderator: Rebecca Schnall, PhD, MPH, BSN, FAAN, FACMI**

- **Elizabeth Eisenhauer, PhD, RN**
- **Theresa Koleck, PHD, RN**
- **Nonniekaye Shelburne, CRNP, MS, AOCN**



# Presentation

---

**Elizabeth Eisenhauer**  
PhD, RN



# **Staying Informed about Precision Health: A Curated Set of Tools for Nurses**

---

**Friends of the  
National Library of Medicine  
February 6th, 2024  
Elizabeth R. Eisenhauer,  
PhD, MLS, RN**

# Information Use

---

- How patients, nurses, others seek, access, understand, and use information (“information behavior”)
- **Low knowledge levels** (Lipkus et al., 2011, Skirton et al., 2012; Wright et al., 2019)
- **Information needs** (Cogdill, 2003; Clarke et al., 2013; Tan et al., 2023)
- **Information sources** (Fossum, et al., 2022; McKnight, 2006)



# Information Tools-Facilitators

---

- End-user/stakeholder involvement
- Desirable, feasible, and viable
- Avoids jargon
- Functionality: Easy to learn/use
- Effective  
(completes task/result is accurate)
- Efficient  
(not too time-consuming)
- Integrates into workflow
- Practical – fulfills a real need
- Reliable (works consistently)
- Satisfaction (subjective reactions)
- Permanency  
(no broken links please!)
- Impact (on DM, individual, & org.)
- Organizational support

# Barriers

---

- Lack of time
- Lack of knowledge
- Lack of information literacy skills
- Difficulties with test ordering and reimbursement
- Lack of interest/perception of evidence or relevancy
- Need for information/IT security
- Need for decision support

# If you build it....

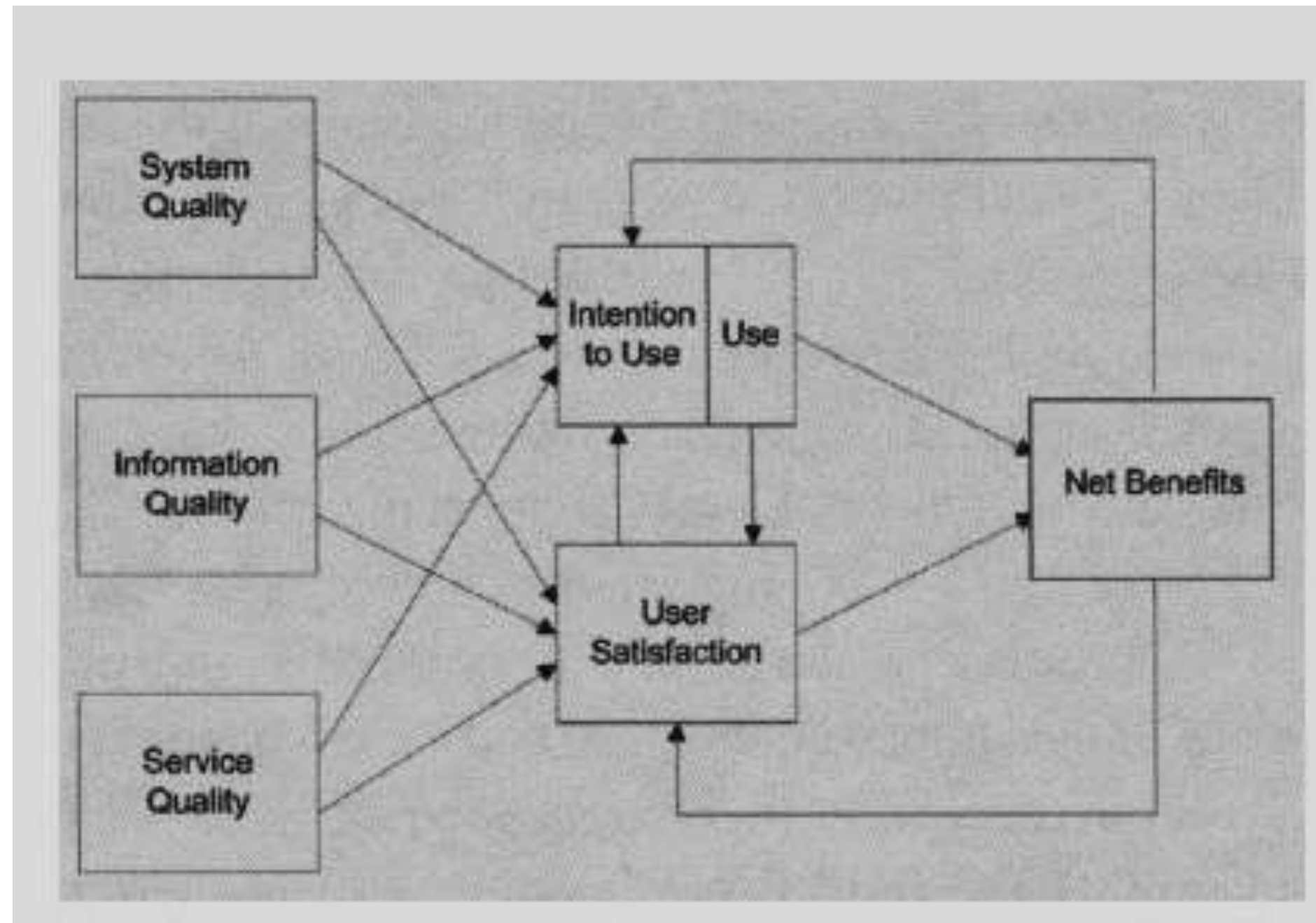
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- ....they might **NOT** come!
- Implementation strategy needed!
- Evaluation imperative!



# Information Systems Success

---



# Tools for Precision Health

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- **Genetics and Genomics**
- Newborns' Health
- Pharmacogenetics (PGx)
- Oncology

- **Social Determinants of Health**

- Screening

- Great example:

Cordova-Ramos et al., 2023

"We Don't Want to Screen for  
the Sake of Screening"

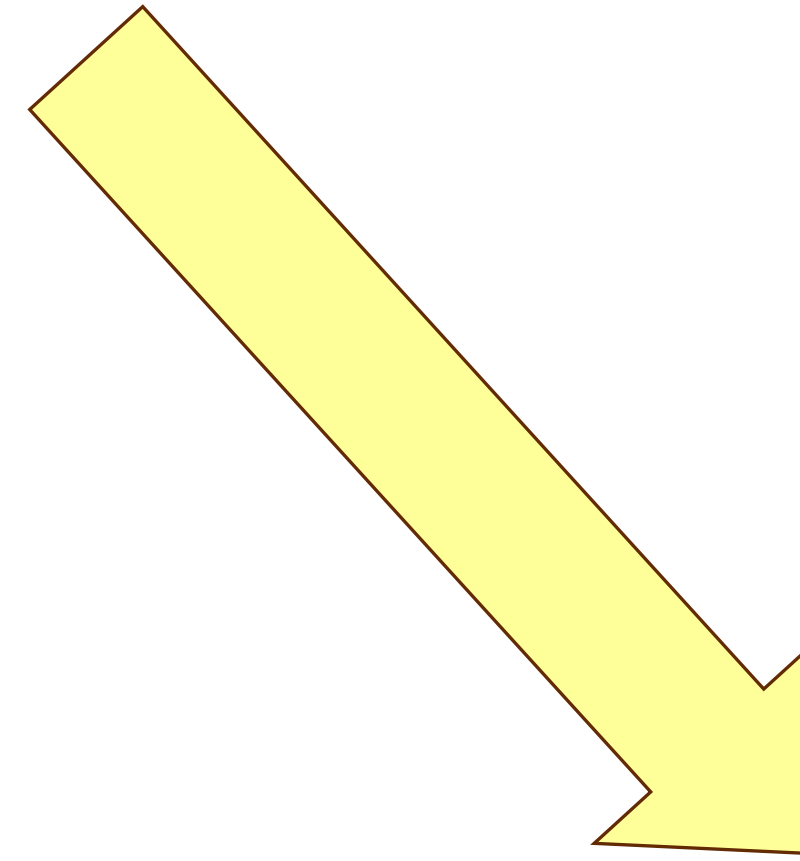
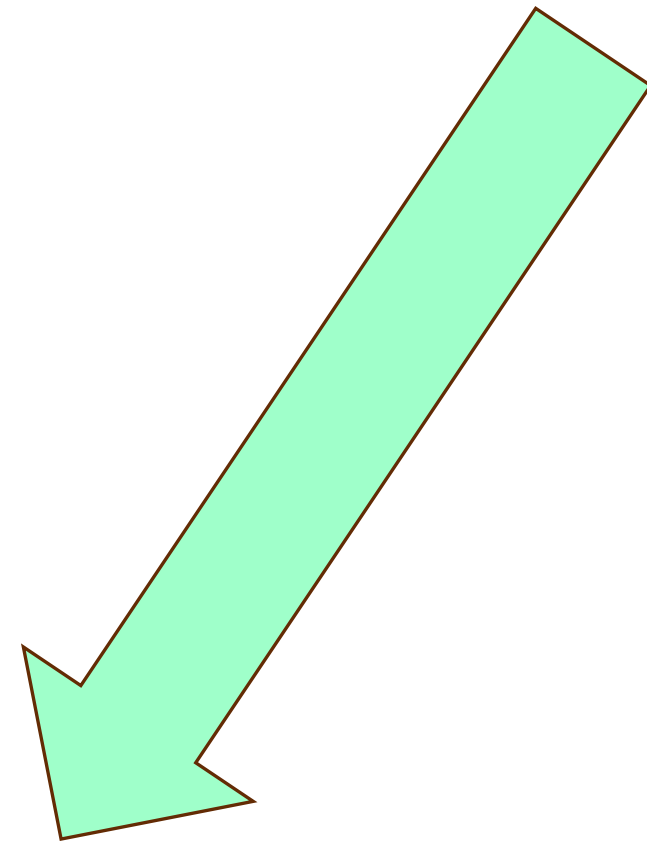
# Questions

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- What tools are available to help practicing nurses learn about of genomic applications in the clinical setting?
  - How can they stay up-to-date?



# Newborns' Health



---

Traditional Newborn  
Screening

Research Projects  
WGS of Newborns

# Traditional Newborn Screening (NBS)

---

- ~4 million births annually in U.S.
- Nearly all newborns
- Important Public Health Program
- Identifies > 12,900 of newborns / year with treatable conditions
- Not without controversy.....
- NBS specimens (“blood spots”) are being stored and used for research
- Consent for screening is assumed; Consent for research varies by state

# Research Projects about WGS of Newborns

Genomics England Generation Study

BabySeq2, Harvard, Cambridge, MA, USA

NewbornsInSA, South Australia

Perigenomed, France

GUARDIAN Study, Boston Children's Hospital, Columbia University, New York-Presbyterian, and the New York State Department of Health in collaboration with GeneDx and Illumina

ScreenPlus, New York State

North Carolina's Early Check Program, a partnership with Illumina and GeneDx

Australia's BabyScreen+ study

Screen4Care, a multinational project in Europe

FirstSteps study, Greece

BeginNGS: Newborn Screening by WGS, Rady Children's Institute for Genomic Medicine, San Diego, CA, USA



# Resources for Nurses

---

- Baby's First Test (2024). What is newborn screening?  
<https://www.babysfirsttest.org/>
- March of Dimes. (August 31, 2020). Newborn screening: Your baby's first tests. <https://www.marchofdimes.org/find-support/blog/newborn-screening-your-babys-first-tests>
- National Human Genome Research Institute. (2022). Informed Consent. <https://www.genome.gov/about-genomics/policy-issues/Informed-Consent>

# NewSTEPs

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- NewSTEPs is a program funded through a cooperative agreement between the Association of Public Health Laboratories (APHL) and the Genetic Services Branch of the US Health Resources and Services Administration (HRSA)
- Repository that captures comprehensive NBS data (access required)
- Aggregate counts of confirmed cases of core RUSP diseases
- <https://www.newsteps.org/nbs-disorders>


# Pharmacogenomics (PGx)

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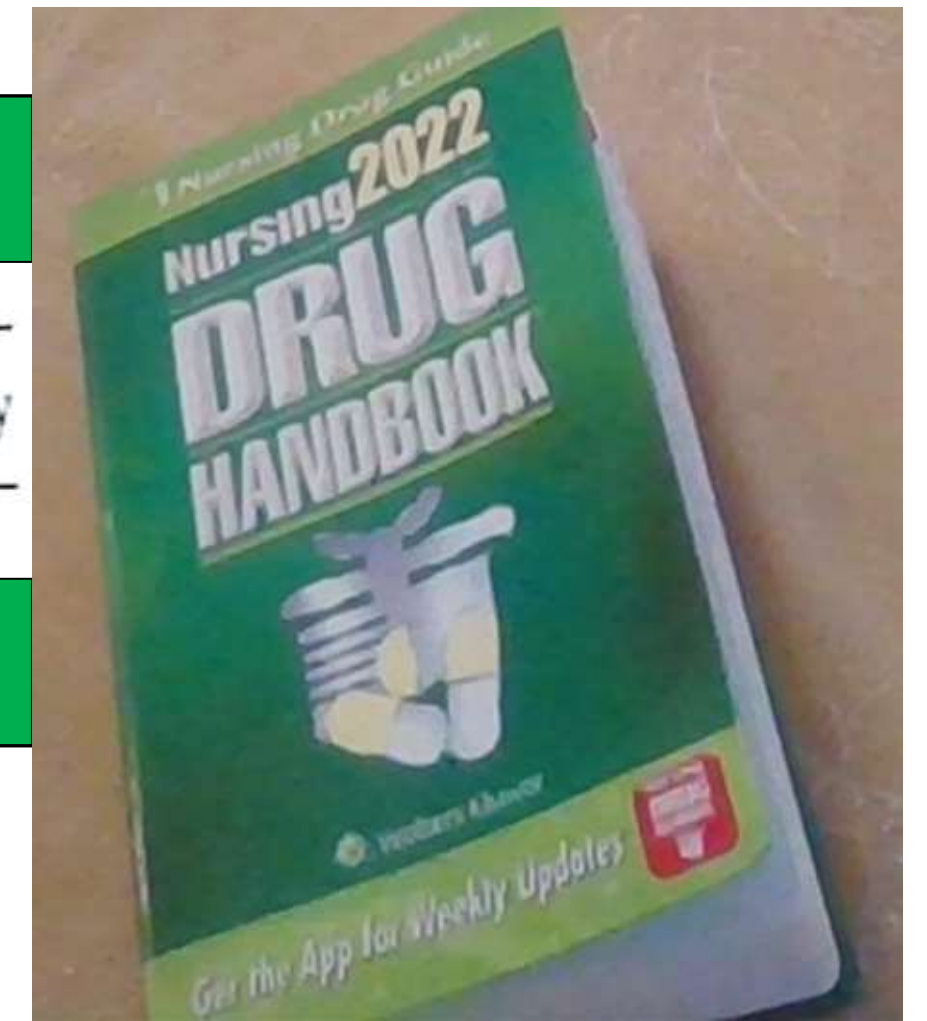
- Frequent information need of clinicians
- Drug information
- Frequent information source
- Drug reference book



# Pharmacogenomics (PGx)



Adjust-a-dose (for all indications).	Dosage adjustment needed for all indications
Genetic considerations used to select and guide drug therapy	
Clinical alert	



# Key Nursing Actions

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- Observation
- Information
- Support

# Observe, Support, Inform

---

“Nurses must be prepared to not only catch their own errors, but also the errors of healthcare providers, pharmacists, and others in the chain of medication administration” (Durham, 2015, p.1).



# Pharmacogenomics Knowledge Base (PharmGKB):

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- PharmGKB is a comprehensive resource that curates knowledge about the impact of genetic variation on drug response for clinicians and researchers
- <http://www.pharmgkb.org/>

# Clinical Pharmacogenetics Implementation Consortium (CPIC®)

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- CPIC's goal is **to address barriers to clinical implementation of pharmacogenetic tests** by creating, curating, and posting freely available, peer-reviewed, evidence-based, updatable, and detailed gene/drug **clinical practice guidelines**.
- Facilitate use of pharmacogenetic tests for patient care
- <https://cpicpgx.org/>

# Video Case Study

The screenshot shows a video player interface. At the top, there is a search bar with 'MedGen' and a 'Search' button. Below the search bar are links for 'Limits' and 'Advanced'. The video content is a case study titled 'Case Study' with the following text:

- Patient presents with Acute Coronary Syndrome, including evidence of coronary artery atherosclerosis.
- Angioplasty is scheduled
- Prescribe clopidogrel for 3-6 months
- Patient expresses concern about effectiveness given her father's experience with clopidogrel
- Practitioner researches the pharmacogenetics of clopidogrel response to consider a change in prescription.
- Patient is a poor metabolizer of clopidogrel (\*3/\*3 for CYP2C19)
- Practitioner instead prescribes prasugrel

At the bottom of the video frame, there is a caption: 'believed to be metabolized by CYP2C19, as this Medical Genetics Summaries article describes.' The video player also shows a 'Subscribe' button for the channel.

## NCBI Pharmacogenetics Resources for Clinical Care



National Library of Medicine  
108K subscribers

Subscribe

22



Share

Save



[HTTPS://WWW.YOUTUBE.COM/WATCH?V=4MHZV8LQTA0](https://www.youtube.com/watch?v=4MHZV8LQTA0)



# PGx Resources

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National Library of Medicine. (Jan 23, 2019). NCBI Pharmacogenetics Resources for Clinical Care.

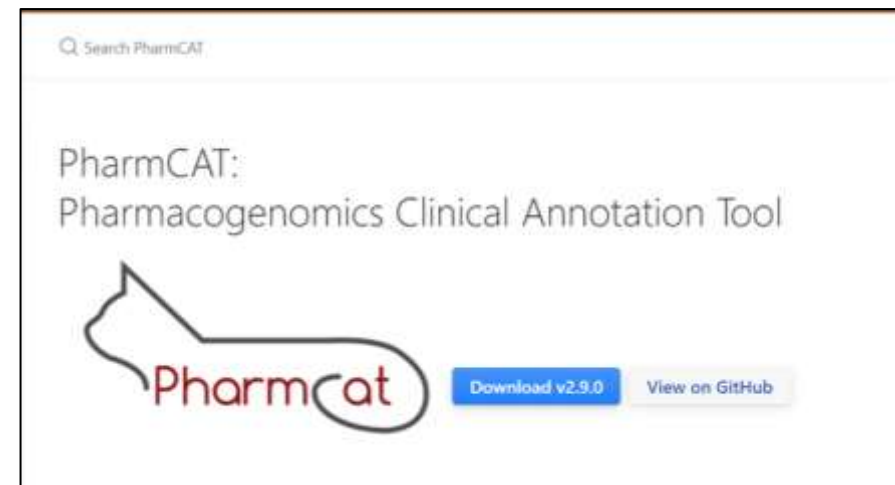
<https://www.youtube.com/watch?v=4Mhzv8LQTa0>

Explore and navigate NCBI MedGen, ClinVar, GTR, & DailyMed

- MedGen: <https://www.ncbi.nlm.nih.gov/medgen>
- Genetic Testing Registry: <https://www.ncbi.nlm.nih.gov/gtr/>
- ClinVar: <https://www.ncbi.nlm.nih.gov/clinvar/>
- DailyMed: <https://dailymed.nlm.nih.gov/>

# PharmCAT: A Pharmacogenomics Clinical Annotation Tool

---



- Extract relevant variants from genomic dataset
- Match to guidelines
- Generate a report with prescribing recommendations

# \*Oncotype DX<sup>®</sup> Breast Recurrence Score

---

- Gene expression profiling (MeSH)
- Score calculate risk of recurrence and likelihood of benefit from chemotherapy in ER-positive, HER2-negative, early-stage breast cancer
- May spare patients the effects of chemotherapy



# Oncotype DX<sup>®</sup> Breast Recurrence Score

---

- Schaafsma et al., 2021; Seer data 2004-2015
- 34% of all ER+ breast cancers tested in 2015
- Users = Good outcomes
- Lower use in non-whites -Why?
- Addition of a nurse navigator increased Oncotype DX tests for eligible patients from 26% to 88% (McAllister & Schmitt, 2015, p.11).

# Exact Sciences (Genomic Health)

---

- Tools for Patients and Providers
- Discussion Guides
- Videos
- Research
- Exact Sciences Corporation. (n.d.). Precision Oncology.  
<https://precisiononcology.exactsciences.com/>

# Fallowfield et al.

---

- Health literacy
- Numeracy expertise
- Communication skills
- Tolerance of uncertainty
- In both the patient and the clinician



# Fallowfield et al.

---

- 8-hr workshop Talking About Risk in the context of GEnomic Tests (TARGET)
- Adapted to 6-hr workshop for nurses
- Empowering RNs
- Implications for workforce retention
- Need increased professional development opportunities

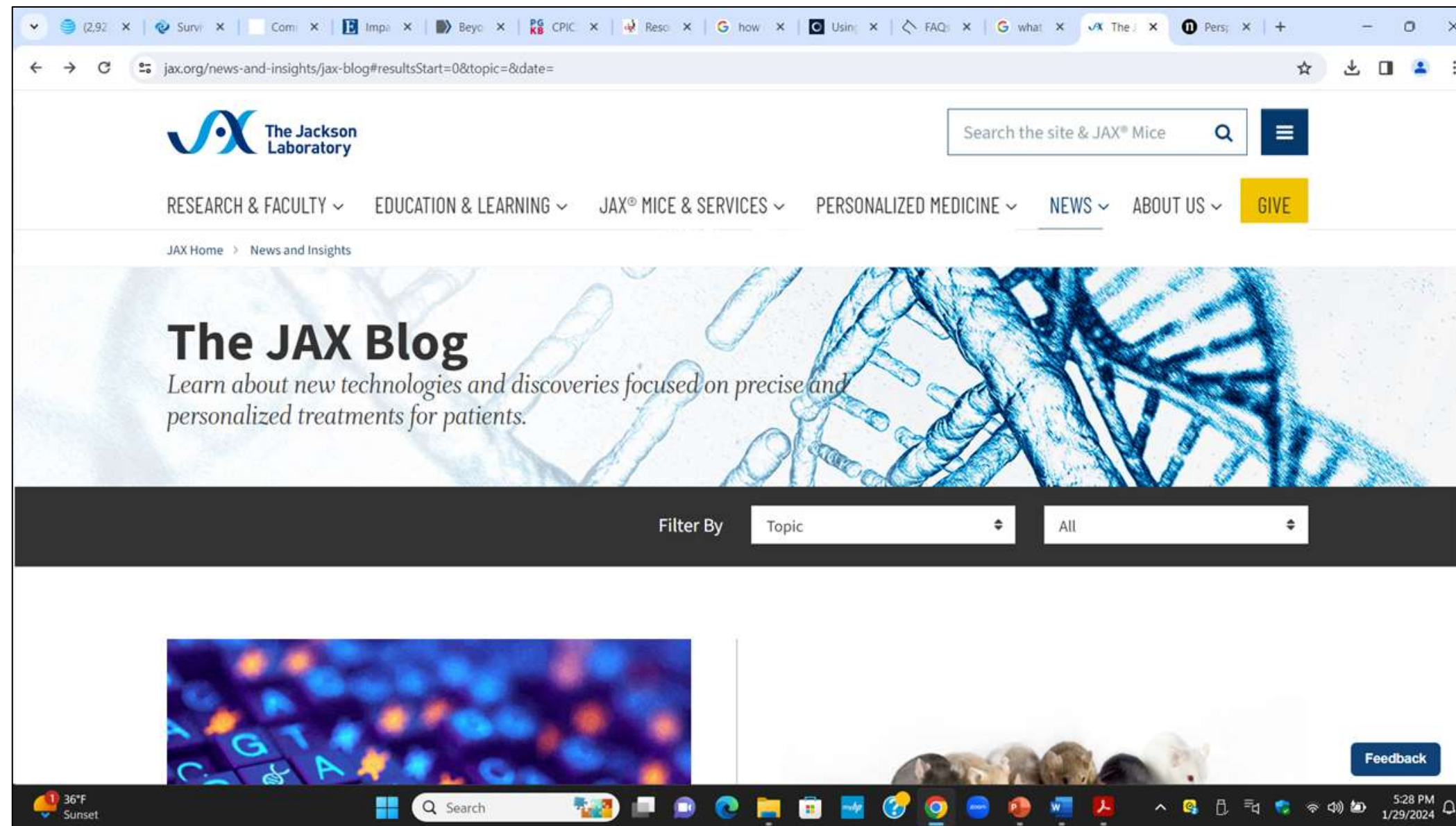
# Staying Up-to-Date

The screenshot shows an email newsletter from GenomeWeb. At the top, there is a black bar with the text "Email not displaying correctly? [View it in your browser.](#)". Below this is the GenomeWeb logo on the left and the title "Daily News" in large bold letters on the right. Underneath the title is the subtitle "The Latest Headlines From The GenomeWeb Newsroom".

On the left side, there is a "Breaking News" section with a blue header. Below it is a news item with a bookmark icon, the title "Thermo Fisher Scientific, Pfizer Ca Colorectal Cancer Care", and the text "Published on Nov 28, 2023". Below this is a "Featured in Depth" section with a blue header.

On the right side, there is a dark grey sidebar containing navigation links: [Customer Service](#), [About us](#), [Contact](#), [Advertise](#), [Privacy Policy](#), [Manage Newsletter Preferences](#), [Opt out of the Daily News Bulletin](#), and [Unsubscribe from all GenomeWeb mailings](#). Below these links is the GenomeWeb logo and the copyright notice: "© 2024 GenomeWeb, a business unit of Crain Communications. 685 Third Avenue, 10th Floor, New York, NY 10017 212-210-0100 All Rights Reserved. [View it in your browser.](#)"

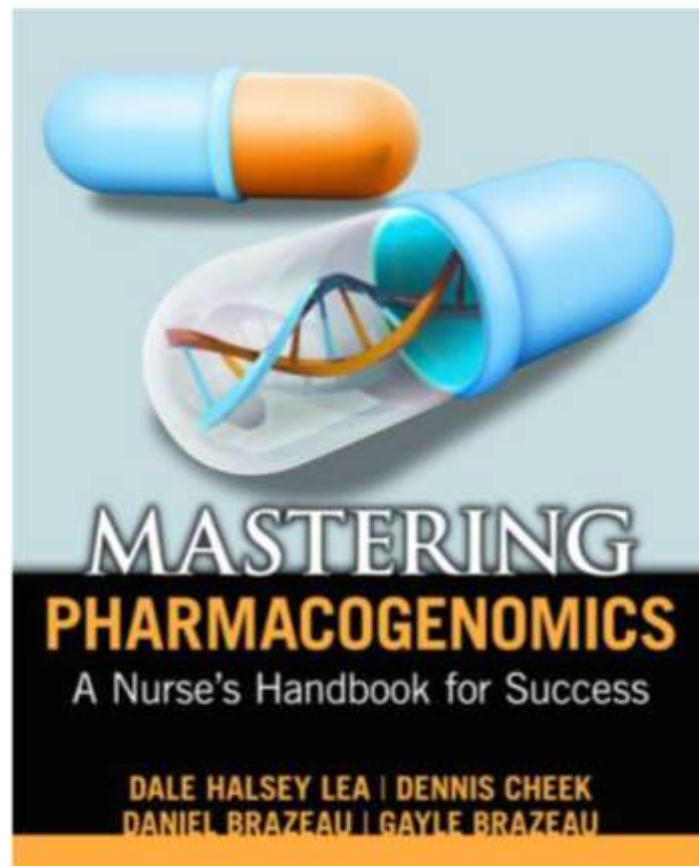
# The JAX Blog





# General Resources

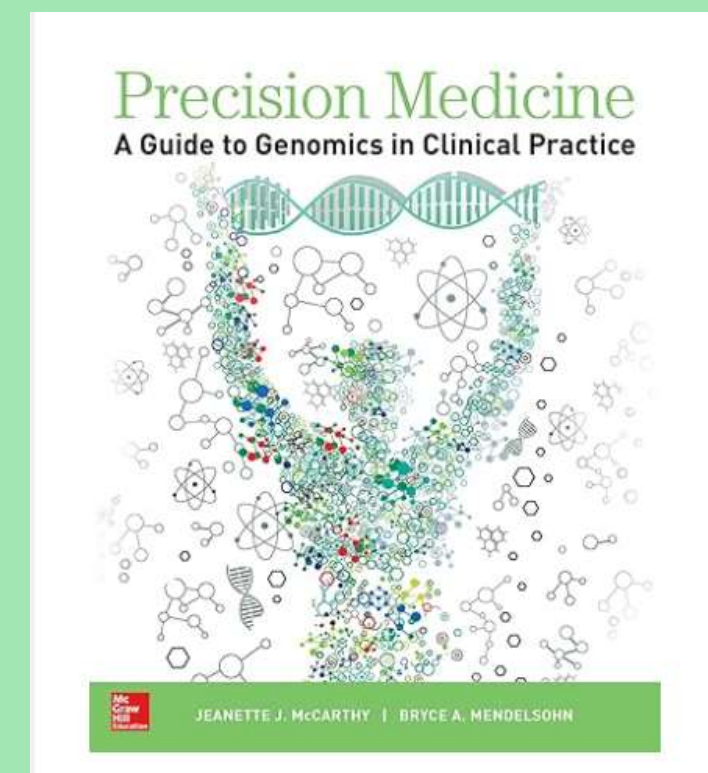
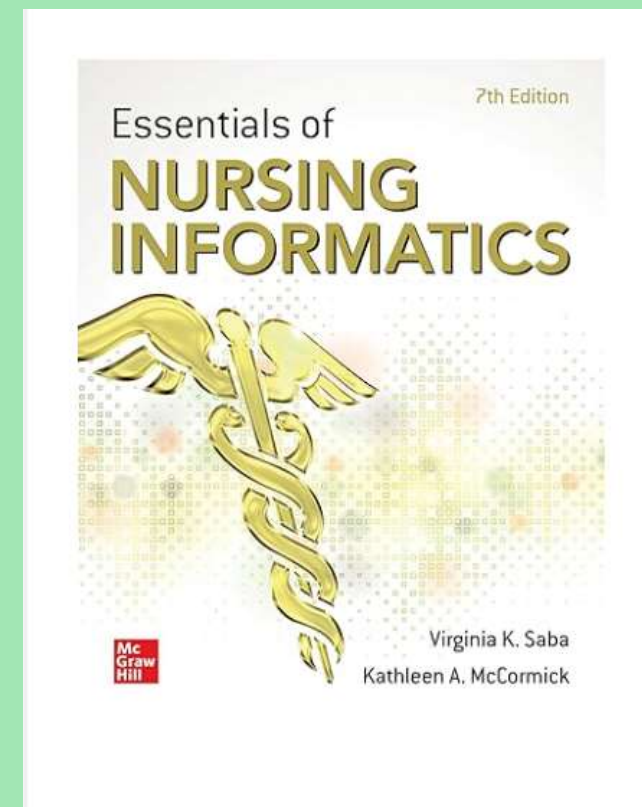
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Lea, D. H., Cheek, D., Brazeau, D. Brazeau, G. (2015). Mastering pharmacogenomics : a nurse's handbook for success. Sigma Theta Tau International. **(Doody's 4 star review)**

# General Resources

Saba, V. K. & McCormick, K. A. (2021). Essentials of nursing Informatics (7th ed.). McGraw-Hill Education. **Doody's Core Titles 2023!** McCarthy, J. J., & Mendelsohn, B. (2017). Precision medicine: A guide to genomics in clinical practice. **Doody's Core Titles 2022!**



# ISONG

---

**International Society of Nurses in Genetics. (2023).**

Genomics Education Resources Repository

Comprehensive genomics education resources compiled by  
the ISONG global membership and education committees

<https://www.isong.org/ed-resources-repository>



# General Resources



## Genomic Competencies for Nurses from Theory to Application: An Online Course

**Increase Your Genomic Literacy**

Are you a DNP- or PhD-prepared nurse, faculty or student and ever wish you had a better understanding of genome science, clinical genetics, and their social and ethical implications?

Now is your chance to enroll in a FREE, Flexible, NIH-funded self-paced online course, beginning September 2022 and January 2023!

*Genomic Competencies for Nurses from Theory to Application: An Online Course* will increase genomic literacy among a diverse group of doctorally prepared nurses and students. Participants will gain expertise in genetic and genomic competencies, resulting in significant benefits for nursing science, the nursing workforce, and ultimately, for patients.

Learn from local and national scholars in genomics, ethics, nursing and transcultural care in this self-paced course. We recommend completing within 6-12 weeks.

**Who is Eligible to Participate?**  
Doctoral-level (PhD or DNP) nurses including faculty, clinicians, researchers and students interested in applying genomic knowledge to their research, evidenced-based practice, education and advocacy efforts. No genetic experience required.

**Course Outcomes**  
To improve nurses' genetic literacy and genomic science literacy to benefit patient/population health care outcomes, particularly those from underrepresented groups.

**Course Content**

Module 0: Genetic Primer (optional)	Module 3: Clinical Management
Module 1: Risk Assessment and Interpretation	Module 4: Ethical, Legal, and Social Implications
Module 2: Genetic Education, Counseling, Testing, and Results Interpretation	Module 5: Research – Translation to Practice
	Module 6: Professional Role and Leadership

**Professional Development**  
This professional development opportunity is offered through Duquesne University School of Nursing and funded by National Human Genome Research Institute of the National Institutes of Health. This material is based upon work supported by the National Institutes of Health under Grant No. (R25HG012228). Any opinions, findings, and conclusions or recommendations expressed in this material are those of the author(s) and do not necessarily reflect the views of the National Institutes of Health.

Contact hours will be offered upon completion.

Duquesne University School of Nursing is accredited as a provider of nursing continuing professional development by the American Nurses Credentialing Center's Commission on Accreditation.


**Questions?**  
Please contact Elizabeth Davidson, MA  
Coordinator of Professional Development and Continuing Education Events  
davidson@duq.edu | 412.396.2072

**Learn More**

# General Resources

An official website of the United States government [Here's how you know](#) ▾

NIH National Library of Medicine

 **MedlinePlus** **25** YEARS of health information  
Trusted Health Information for You

Search MedlinePlus





[About MedlinePlus](#) [What's New](#) [Site Map](#) [Customer Support](#)


[Health Topics](#) [Drugs & Supplements](#) [Genetics](#) [Medical Tests](#) [Medical Encyclopedia](#) [Español](#)

Home → Genetics

## Genetics


Find consumer-friendly information about the effects of genetic variation on human health.




### Genetic Conditions

Learn about the signs and symptoms, causes, and inheritance of more than 1,300 health conditions with a genetic basis.




### Genes

Find information about the function of more than 1,400 genes and see how changes in these genes are related to health conditions.



### Chromosomes and mtDNA

Read about each of the 23 pairs of human chromosomes and mitochondrial



### Related Health Topics

- [Genes and Gene Therapy](#)
- [Genetic Disorders](#)
- [Genetic Testing](#)



# BioDecide<sup>©</sup>





# Acknowledgement

---

Dr. Marcelline Harris, PhD, RN, FACMI

Associate Professor Emerita

University of Michigan

School of Nursing

# Questions?

---

## Thank you!

Elizabeth Eisenhauer, PhD, MLS, RN

[e.eisenhauer@att.net](mailto:e.eisenhauer@att.net)

# Presentation

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**Theresa Koleck**  
PHD, RN



# Precision health and nursing science: Exemplars from nursing research

**Theresa A. Koleck, PhD, RN**

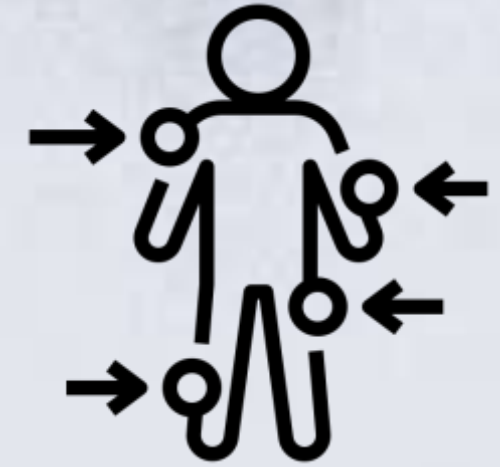
University of Pittsburgh

[tak150@pitt.edu](mailto:tak150@pitt.edu)



**Theresa A. Koleck, PhD, RN**

Assistant Professor  
School of Nursing  
University of Pittsburgh



*My program of research is dedicated to mitigating symptom burden in patients diagnosed with chronic conditions using omics-based approaches and informatics/data science techniques.*

*“Precision health, a growing trend in the health sciences, is defined as healthcare tailored to the need of the individual based on their genetic, genomic, or omic composition within the context of their lifestyle, social, economic, cultural and environmental influences to help them achieve optimal health status ... Precision nursing should be evidence-based as well as person-based in terms of values and expectations.”*

*Shiow-Ching Shun, Journal of Nursing Research, April 2023*





*“Nursing science has made monumental contributions to symptom science through research on biological and clinical features, as well as the influence of environmental, behavioral, social, and economic factors in the manifestation of symptoms and their resolution. Nursing in the omic era represents precision health that focuses on each individual’s personal risk for disease conditions or effectiveness of treatments that are estimated directly from individual’s unique combination of genetic/genomic and environmental risk factors.”*

*Mei Fu and colleagues, International Journal of Nursing Sciences, January 2020*



**University of Pittsburgh**  
**School of Nursing**  
**Genomics of Patient Outcomes HUB**

*Enhancing interdisciplinary, biobehavioral research that uses genomics and other state-of-the-science omics approaches to advance the science of patient outcomes*



**Yvette P. Conley, PhD, FAAN**

Distinguished Service Professor of Nursing,  
Professor of Human Genetics, Associate  
Dean for Research and Scholarship  
School of Nursing, University of Pittsburgh

Using omics approaches to  
understand patient  
outcomes after  
neurological insult



**Sheila A. Alexander, PhD, RN, FCCM**

Associate Professor  
School of Nursing  
University of Pittsburgh

Genetic, genomic, and  
protein biomarkers of  
acute brain recovery



**Christopher C. Imes, PhD, RN**

Assistant Professor  
School of Nursing  
University of Pittsburgh

Tailored interventions in  
adults with suboptimal  
sleep health to improve  
sleep, wellness, fatigue, and  
cardiometabolic health







**Monica A. Wagner, PhD, RN**

Assistant Professor  
Frances Payne Bolton  
School of Nursing  
Case Western Reserve University

Molecular changes  
and the development  
of chronic pain in  
cancer survivors



**Maura McCall, PhD, MSN, RN, OCN**

Postdoctoral Fellow  
Frances Payne Bolton  
School of Nursing  
Case Western Reserve University

Omics of symptoms  
and adherence  
in women with  
breast cancer



**Komal Singh, PhD, RN**

Nurse Scientist  
Mayo Clinic

Chemotherapy-induced  
symptoms and  
disruption in the gut  
microbiome factors



**Susan Grayson, RN, PhD(c)**

Doctoral Student  
School of Nursing  
University of Pittsburgh

Psychoneurological symptoms  
and cancer genomic instability in  
metastatic breast cancer



**Myeong-ga Cho, MSN, RN**

Doctoral Student  
School of Nursing  
University of Pittsburgh

Epigenetic mechanisms of  
cognitive decline in  
women with breast cancer



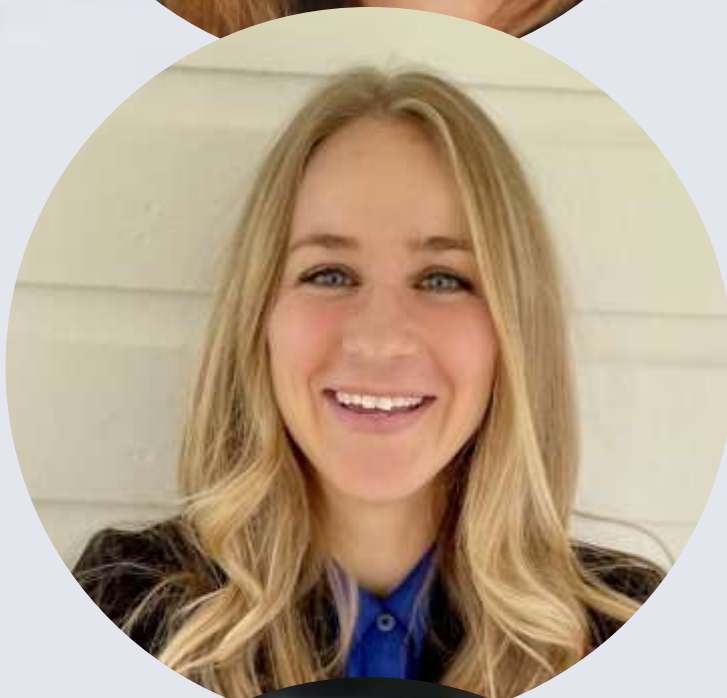


**Jessica A. Davis, PhD, RN,  
CCRN, IBCLC**

Postdoctoral Fellow

Department of Pediatrics, School of Medicine  
University of Pittsburgh

Contribution of nutrition and feeding practices and microbiome composition to growth in infants with congenital heart diseases



**Lacey W. Heinsberg, PhD, RN**

Postdoctoral Associate

Department of Human Genetics  
University of Pittsburgh

Intergenerational transmission of chronic disease, with a focus on pregnancy outcomes and early life growth



**Carolyn S. Harris, PhD, RN**

Postdoctoral Scholar

School of Nursing  
University of Pittsburgh

Risk factors that contribute to the development of symptom clusters and inter-individual variability



Pitts



# Predictors of Unrelieved Symptoms in *All of Us* Research Program Participants with Chronic Conditions



## Disclosures:

- Dr. Koleck is an *All of Us* Researcher Ambassador through Pyxis Partners [Pyxis Partners: OD028404].
- The *All of Us* Research Program has not reviewed, endorsed, or approved this presentation or research.



# One of the largest, richest, most diverse biomedical datasets of its kind

Inviting at least

**1 Million** people from across the United States



Data available from **413,450+** participants

**75%**  
are from communities underrepresented in biomedical research



**45%**  
are from racial and ethnic minority groups

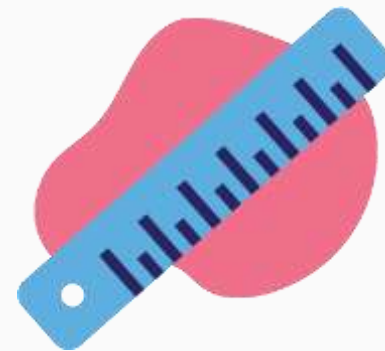
Data as of April 2023

# By securely capturing a range of different data types

The *All of Us* Research Program's Data and Research Center (DRC) curates a range of different data types as part of the data collection process.



**413,350+**  
Survey Responses



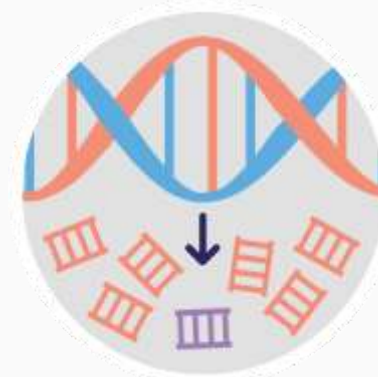
**337,500+**  
Physical Measurements



**312,900+**  
Genotyping Arrays



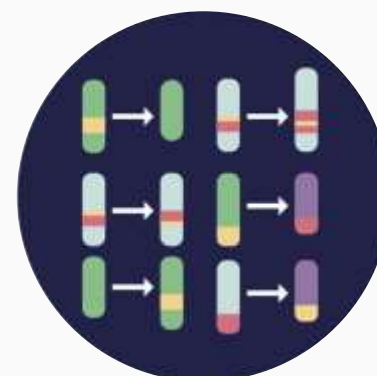
**287,000+**  
Electronic Health Records



**245,350+**  
Whole Genome Sequences (WGS)



**15,600+**  
Fitbit Records  
*NEW! Sleep Data*



**11,350+**  
Structural Variants  
*NEW! In 2023*



**1,000+**  
Long-Read Sequences  
*NEW! In 2023*



# Predictors of Unrelieved Symptoms in *All of Us* Research Program Participants With Chronic Conditions

- Over half of American adults are diagnosed with a chronic condition
- Increased risk for unrelieved, co-occurring symptoms



Journal of Pain and Symptom Management

Available online 9 September 2022

In Press, Corrected Proof ?



Original Article

## Predictors of Unrelieved Symptoms in *All of Us* Research Program Participants With Chronic Conditions

Caitlin Dreisbach PhD, RN <sup>1, 2</sup>, Susan Grayson BSN, RN <sup>3</sup>, Katelyn Leggio MSN, RN <sup>4</sup>, Alex Conway MS <sup>3</sup>,  
Theresa Koleck PhD, RN <sup>3</sup> ✉

<https://pubmed.ncbi.nlm.nih.gov/36096320/>

*All of Us Research Program offers unique opportunity to study symptoms and predictors of symptoms in individuals diagnosed with 1 or more chronic conditions!*

# Purpose (Part 1)

- To identify symptom phenotypes of patients diagnosed with one or more common chronic conditions

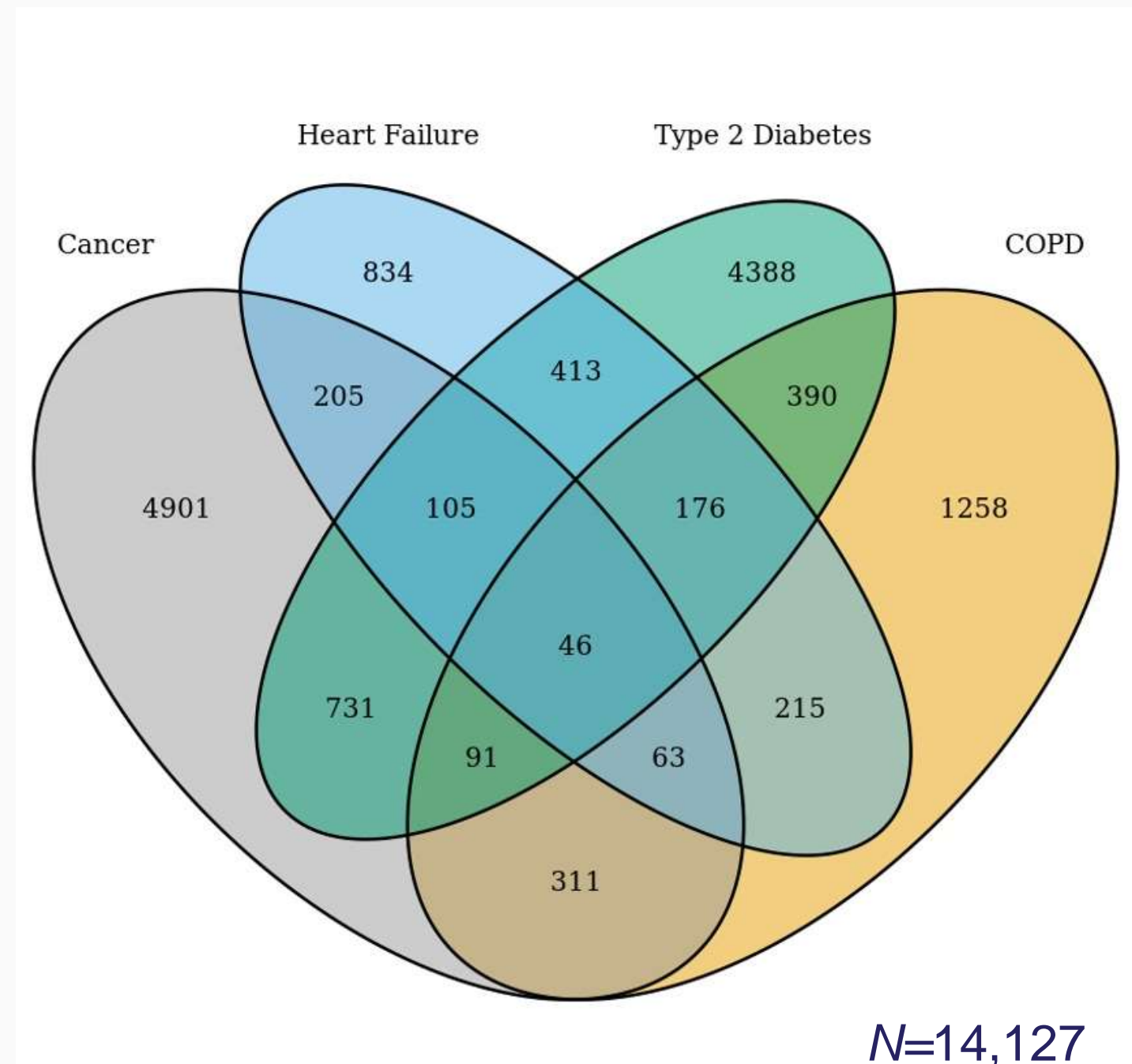
*In the past 7 days...*



Surveys

- How would you rate your **pain** on average?
- How would you rate your **fatigue**?
- How often have you been bothered by **emotional problems** such as feeling anxious, depressed or irritable?

(from 372,000+ participants)  
Including lifestyle, access to care, medical history, and data from nearly 100,000 participants on their experiences during the COVID-19 Pandemic



## Purpose (Part 2)

- To understand factors that predict membership to highly symptomatic phenotypes



### Health-related

- General health
- Physical health
- Mental health

### Demographic

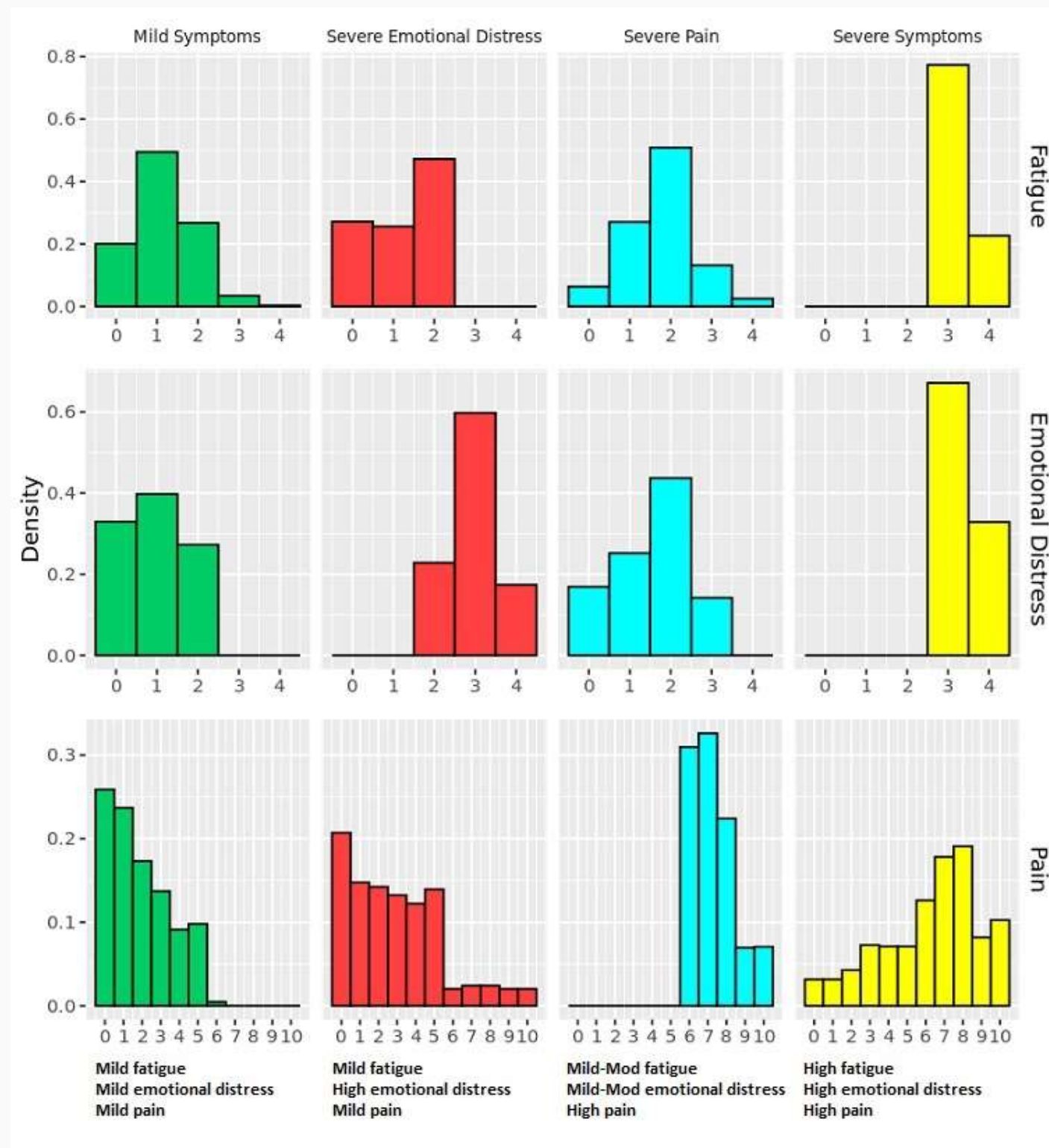
- Age
- Gender identity
- Race/ethnicity
- Marital status
- Level of education
- Insurance status
- Employment status
- Annual household income

### Healthcare access and utilization

- Foregoing and/or delaying medical care
- Having a usual place for medical care
- Time since last seen a health care professional
- Ease of understanding health information
- Being treated with respect by the provider
- Being asked about opinions or beliefs by the provider



# Results



Symptom Phenotypes

*Most notably, foregoing or delaying medical care and rating mental health as poor or fair increased the odds of belonging to a highly symptomatic phenotype.*

# Making Health Discoveries Possible

The *All of Us* Program wouldn't be possible without the generosity of our participants and the dedication of our researchers to enable health discoveries.



@AllofUsResearch  
@AllofUsCEO  
#JoinAllofUs



# Create an *All of Us* account

## Get started today

STEP  
**1**

CONFIRM YOUR INSTITUTION'S  
AGREEMENT

STEP  
**2**

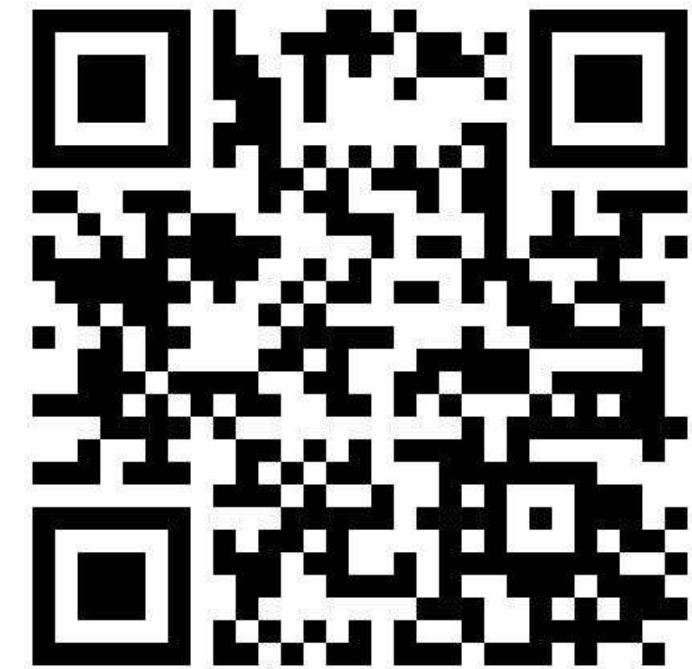
CREATE AN ACCOUNT AND  
VERIFY YOUR IDENTITY

STEP  
**3**

COMPLETE THE MANDATORY  
TRAINING

STEP  
**4**

SIGN THE DATA USER CODE OF  
CONDUCT (DUCC)



[allof-us.org/Register](https://allof-us.org/Register)



*“The spectrum of precision health spans one’s genetic code to their zip code and offers the opportunity for nurse scientists to lead the way in advancing symptom and self-management science. Precision health can stimulate discovery in many areas across the lifespan but the impact of precision health is only beginning to be realized. In order to reach the goal of precision health, approaches must be applied throughout the stages of research translation from basic science to clinical research and ultimately at the population level to improve health and prevent disease.”*

*Kathleen Hickey and colleagues, Nursing Outlook, July-August 2020*







Thank you!



# Presentation

---

**Nonniekaye Shelburne**  
CRNP, MS, AOCN



# NIH...Turning Discovery into Health<sup>®</sup>

## *Precision Medicine Research Initiatives*

*Nonniekaye Shelburne CRNP, MS, AOCN<sup>®</sup>*

*Program Director*

*Epidemiology and Genomics Research Program*

*Division of Cancer Control and Population Sciences*

*National Cancer Institute*

## Outline:

- NIH Precision Medicine Initiative
- Precision Risk Identification
- Precision Treatment Decision Making
- Implementation Science in Genomics

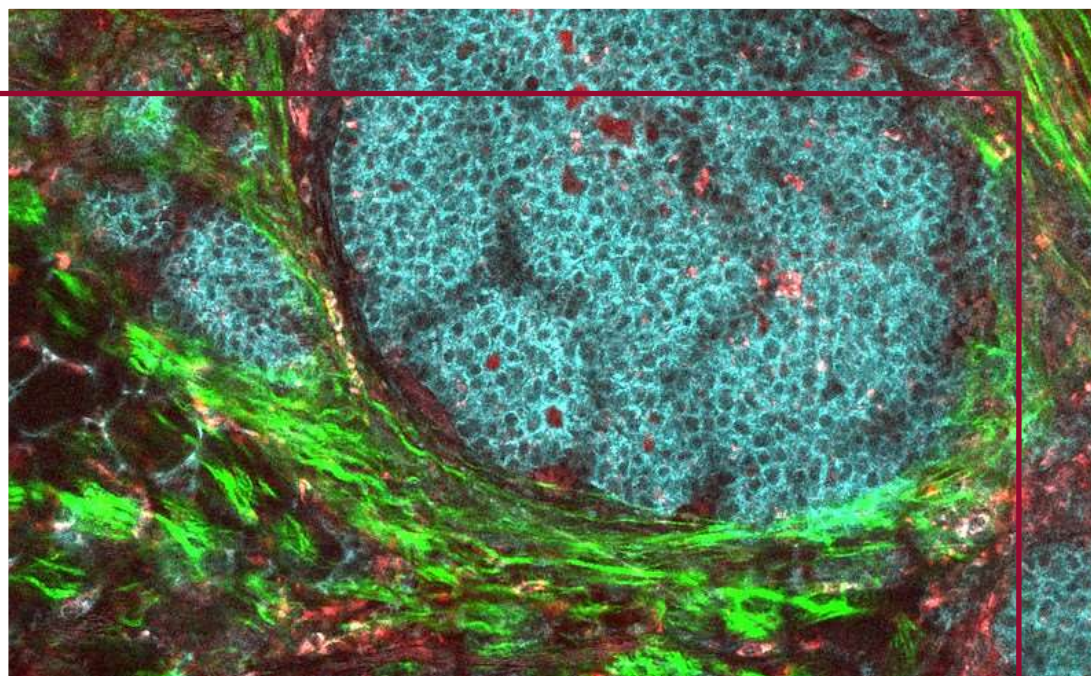
# NIH...Turning Discovery into Health®

- **Mission:** to seek fundamental knowledge about the nature and behavior of living systems and to apply that knowledge to enhance health, lengthen life, and reduce illness and disability.
- NIH supports:
  - **Basic research...**to fuel progress
  - **Translational research...**to move basic discoveries forward
  - **Clinical research...**to turn discoveries into prevention, treatments, and cures
  - **A creative and diverse workforce...**since people are our most important resource
  - **A balanced research portfolio...**to ensure high return on investment for U.S. taxpayers

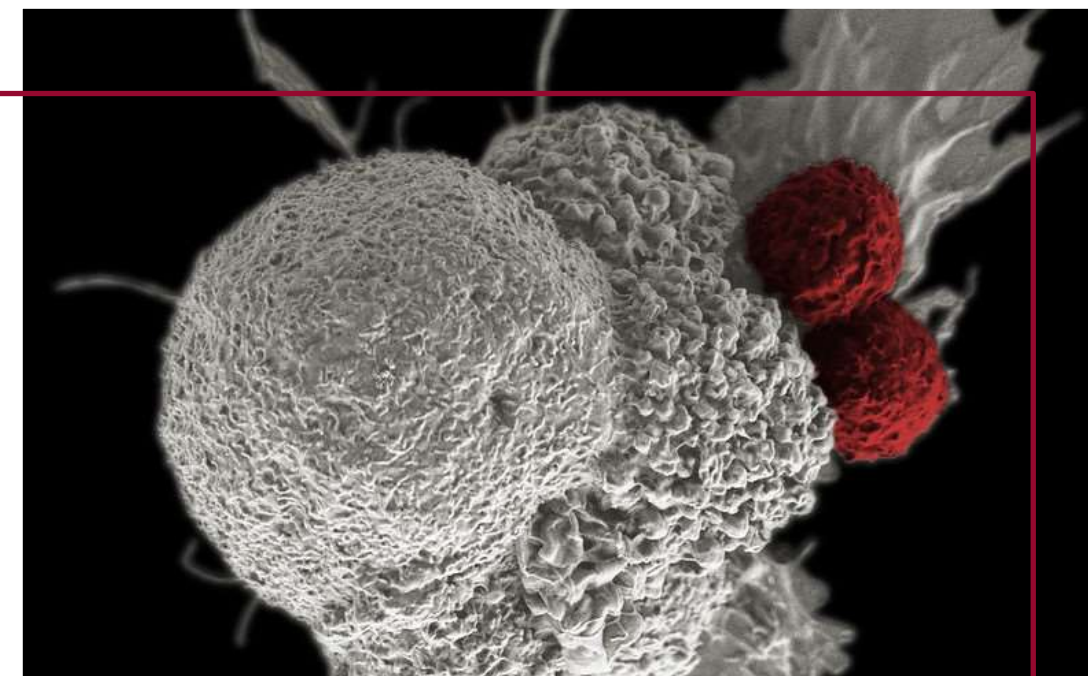
## The Promise of Precision Medicine



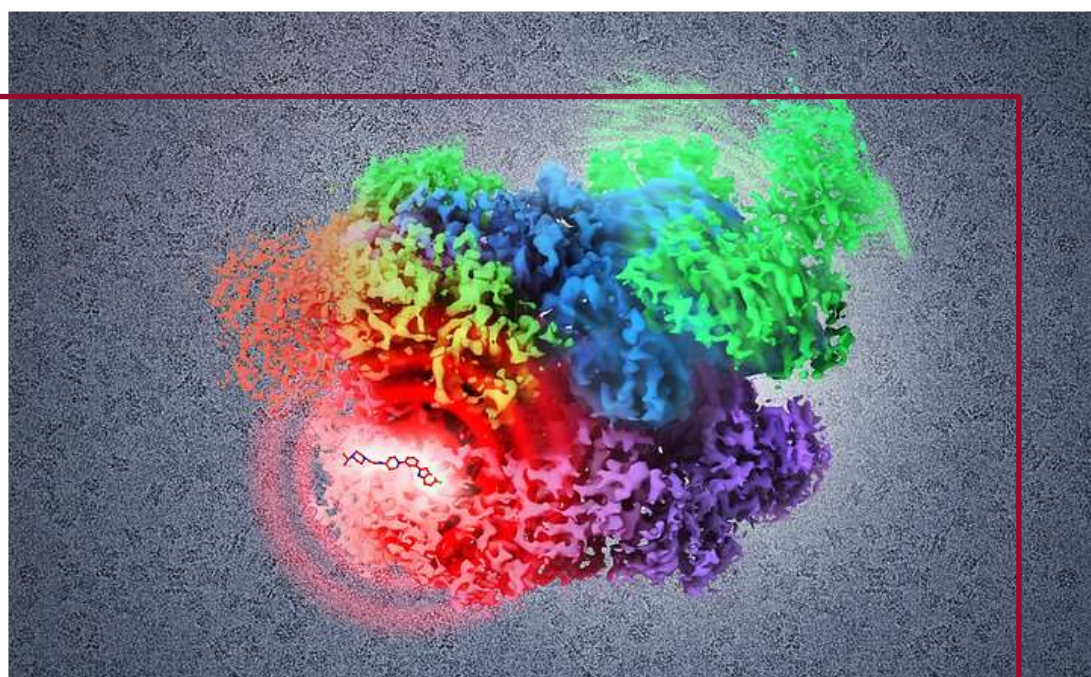
# The Promise of Precision Medicine



Precision Oncology



Cancer Immunotherapy



Pharmacogenomics

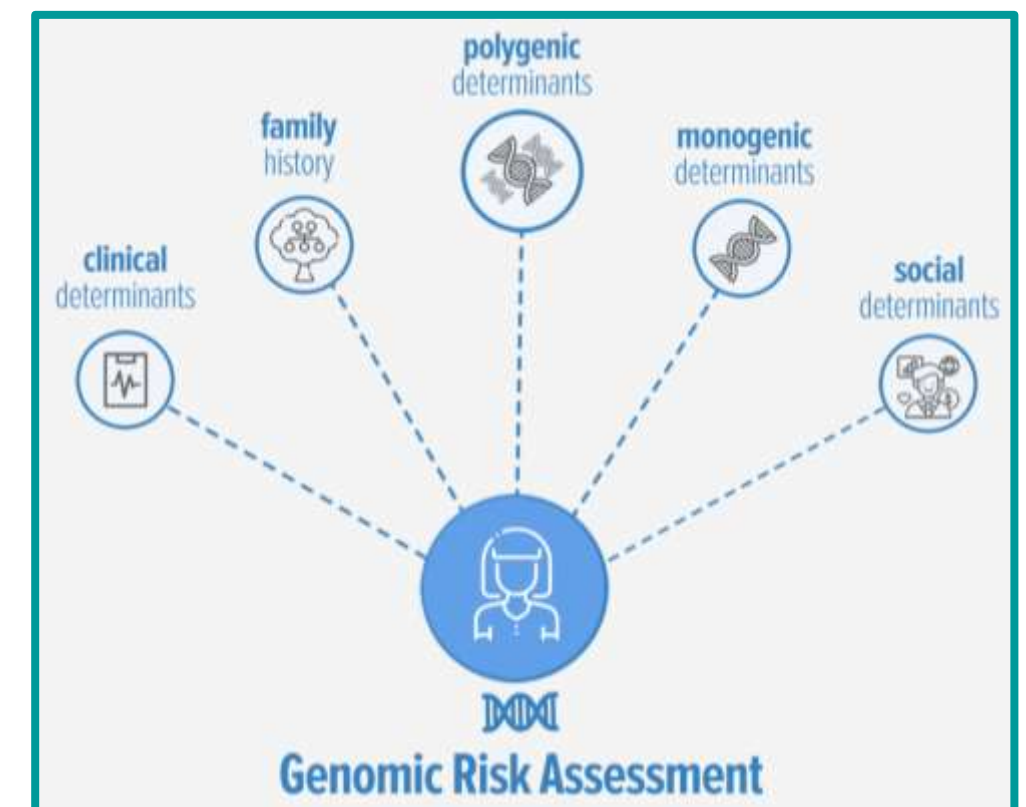


Rare Diseases



# Precision Risk Identification

- Polygenetic Risk Scores (PRS)
  - Establish clinical utility of using genetic data to estimate risk
  - Understand contribution clinical PRS to race-based health disparities
- Genomic Risk Assessment and Management Network (eMERGE )
  - How to best validate and implement genome informed risk assessments
  - Asthma, aFib, coronary heart disease, hypercholesterolemia, chronic kidney disease, cancer, obesity, type 1 and 2 diabetes





# Precision Risk Identification

- Right time
- Benefits/Harms
- Return of Results
- Actionable results

## The Promise and Perils of Next-Generation DNA Sequencing at Birth

### What pathways exist for newborn genetic sequencing?

- **Clinical** (e.g., in the NICU) (Goldenberg).
- **Direct through health care providers** (e.g., providers ordering a test from an outside company or partner) (Goldenberg).
- **Direct to consumers** (Goldenberg).
- **Supplemental** to current public health newborn screening, but not a replacement (Bonhomme).

Each of these pathways has implications for **consent, returning results, follow-up** (Goldenberg), and **cost** (Veenstra).

### What are some of the benefits and harms to consider?

- **Making sequencing more available** can increase early identification, provide families valuable information, and limit diagnostic odyssey (Hu).
- **Accessible follow-up care** is important for reducing burdens to families, **increasing equity**, and using improved health outcomes as measures of success (Hu, Woolford).
- Sequencing could increase preexisting health system inequities; therefore, it is important to **consider intersecting health disparities** (Goldenberg).

The National Academies of Sciences, Engineering, and Medicine convened a workshop to examine the use of DNA sequencing to supplement traditional newborn screening for conditions that are treatable but not clinically evident in the newborn phase. This infographic provides high-level takeaways from individual speakers on considerations for newborn genome sequencing.

### What are some of the policy and regulatory implications to consider?

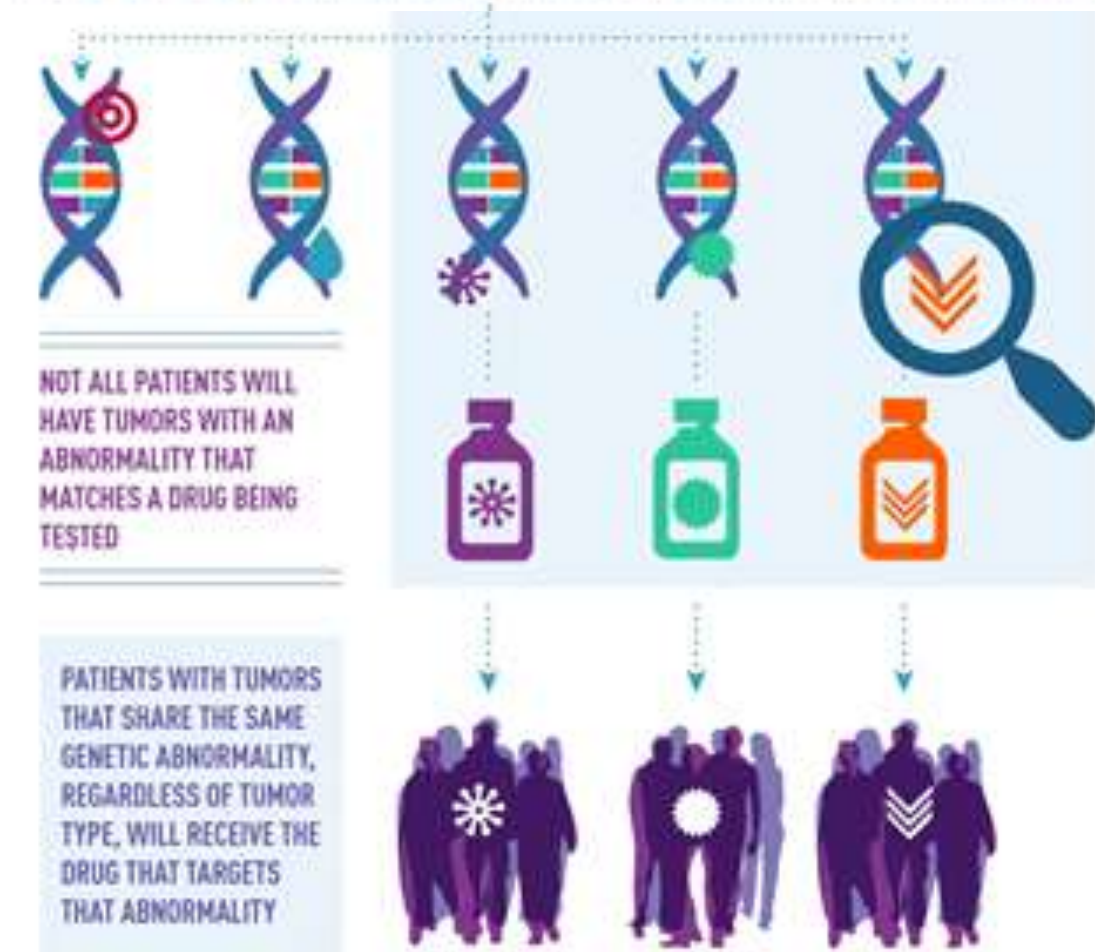
- Regulatory considerations include **protecting privacy**, addressing concerns about **data security**, and ensuring **ethical data stewardship** (Grant, Peay, Powell).
- Widespread sequencing raises concerns about **improper use of DNA data**, including possible misuse by law enforcement (Grant).

### How does sequencing affect communities, and how can community members be involved?

- **More voices are needed at the table** for successful implementation of newborn DNA sequencing (Fletcher, Hu, Klein).
- Perspectives are needed from **people living with genetic disorders** (Woolford), their **families** (Hu), and **groups who have been underrepresented in genomics research and minoritized in health care** (Bonhomme, Fletcher, Goldenberg).



# Precision Treatment Decision Making



\*NCI Molecular Analysis for Therapy Choice

[www.cancer.gov/nci-match](http://www.cancer.gov/nci-match)  
To learn more, call 1-800-4-CANCER



## ComboMATCH Trials Recruiting Patients

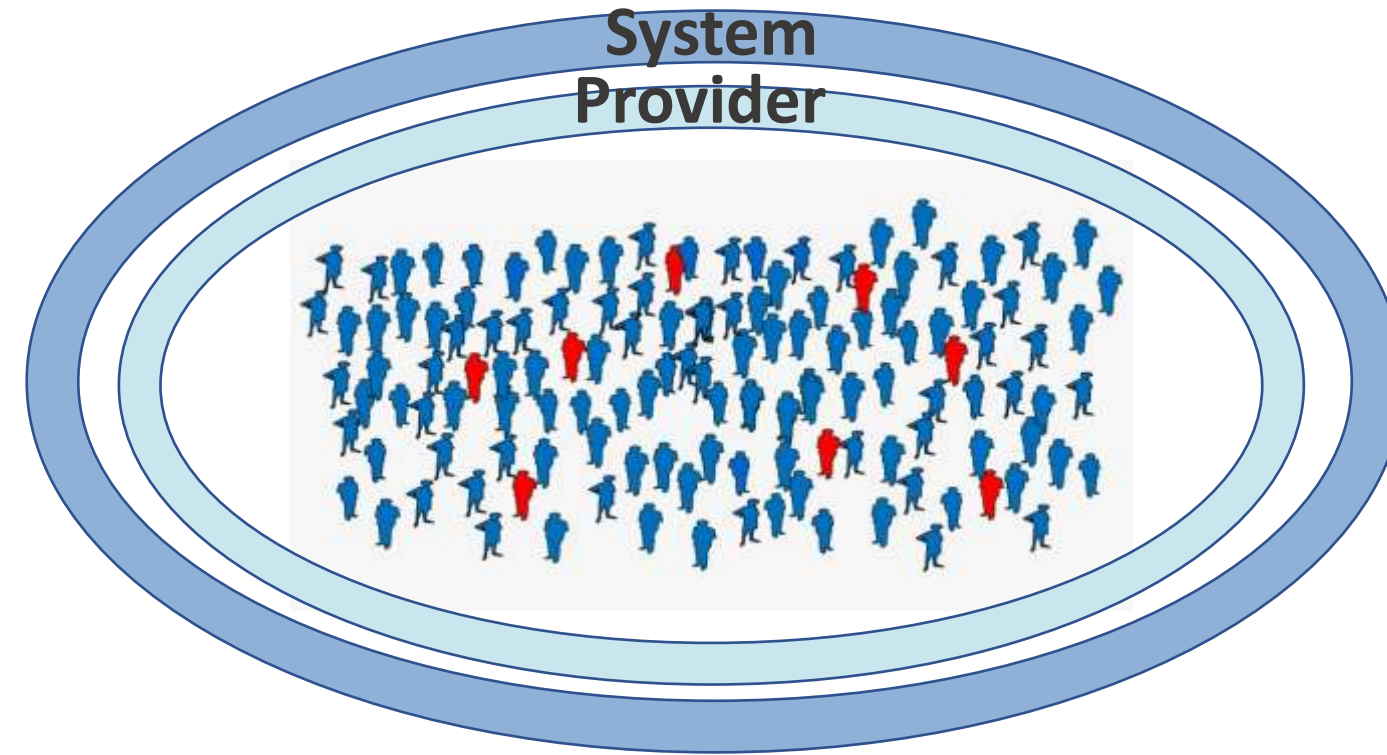
Arm	Targeted genetic change	Drug(s)	Cancer type
EAY191-E4	Taxane resistance	Nilotinib and paclitaxel	Solid tumors previously treated with a taxane
EAY191-S3	Activating <i>AKT</i> mutation	Ipatasertib and paclitaxel	Solid tumors
EAY191-N2	<i>NF1</i> nonsense or frameshift mutations or <i>NF1</i> gene deletion	Binimetinib and fulvestrant	Hormone receptor-positive, HER2-negative metastatic breast cancer
EAY191-N4	<i>RAS</i> pathway mutation	Selumetinib and olaparib	Ovarian or endometrial cancer
EAY191-A6	<i>MAPK</i> pathway mutations	Binimetinib and chemotherapy	Previously treated advanced biliary cancer
EAY191-A3	<i>RAS</i> pathway mutation	Palbociclib and binimetinib	Ovarian, pancreatic, and other solid tumors

<https://www.cancer.gov/about-cancer/treatment/nci-supported/nci-match>

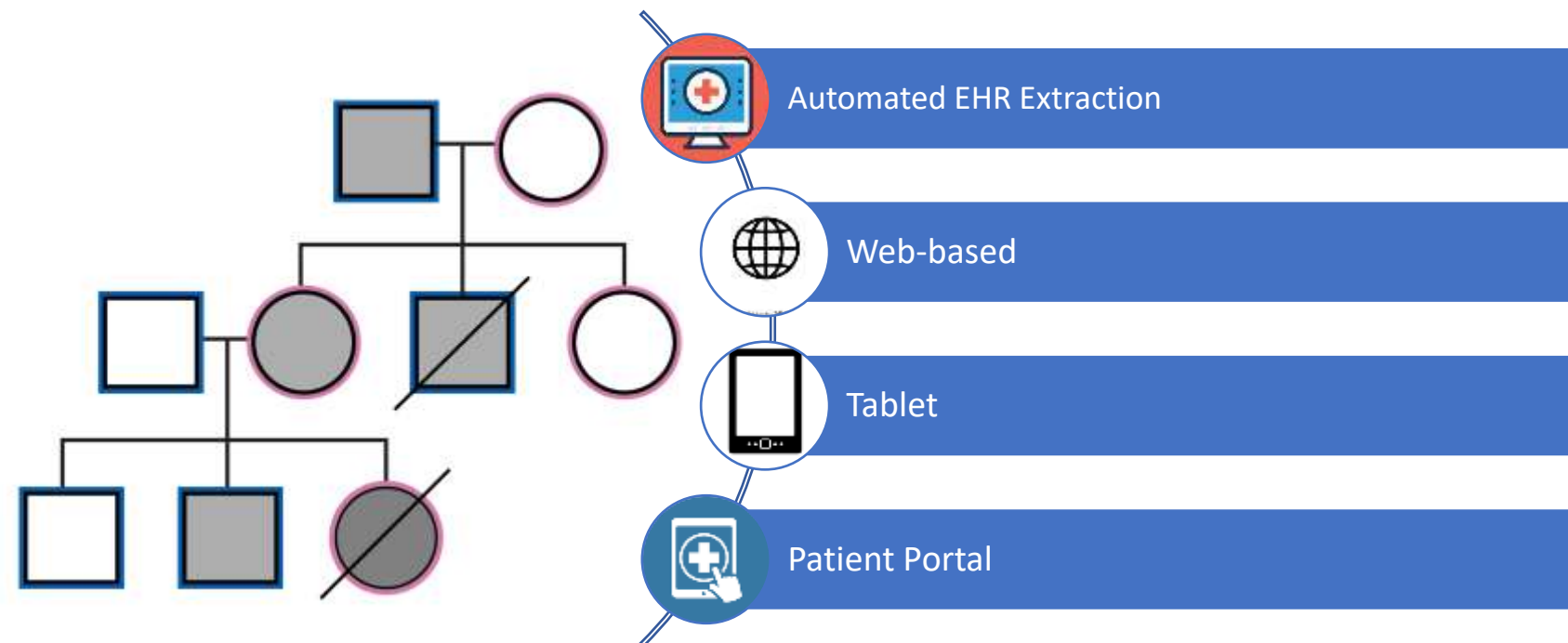
<https://www.cancer.gov/about-cancer/treatment/nci-supported/combomatch#treatment-arms-that-are-open-and-enrolling-patients>

O'Dwyer, PJ, et al. *Nature Medicine*. June 2023 .

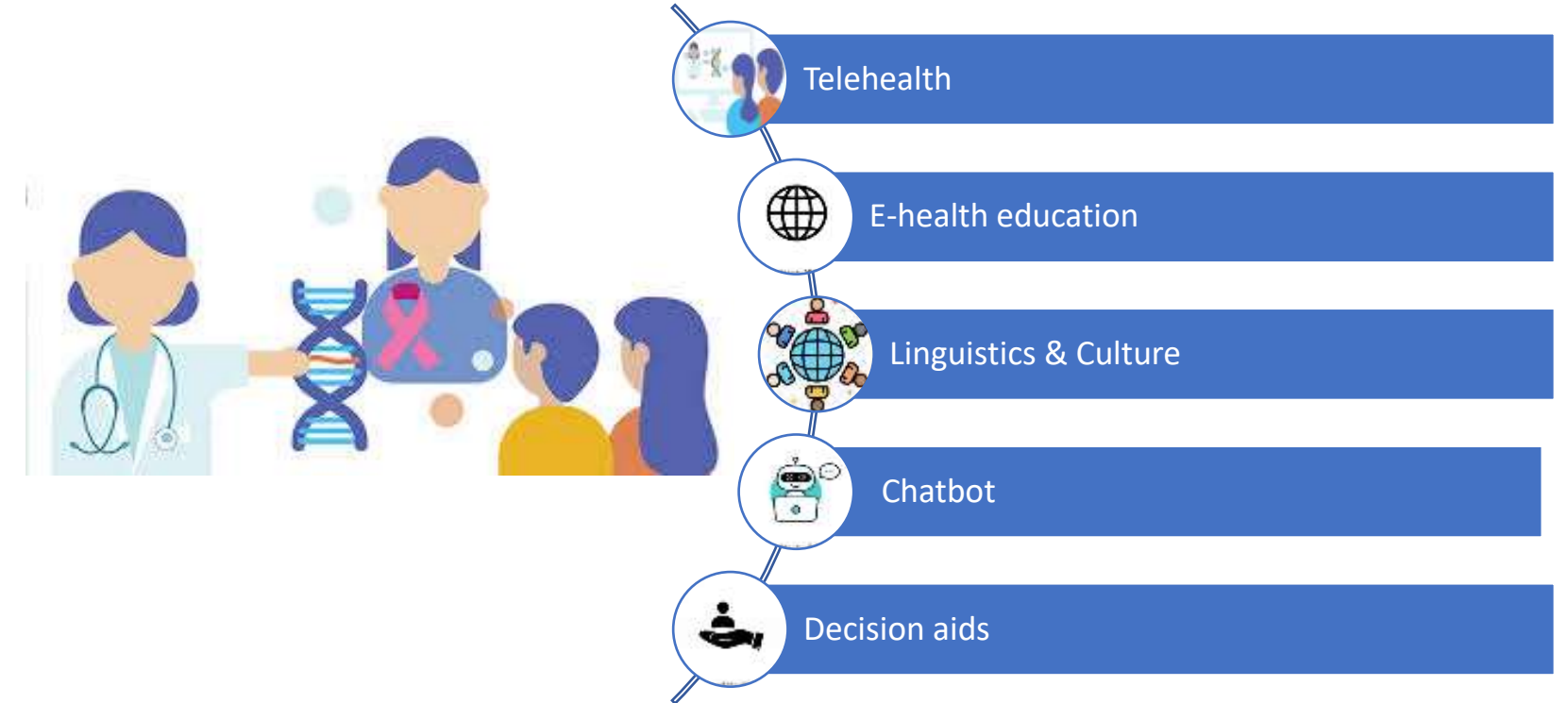
# Implementation Science



## Family Hx Collection/Risk Alert



## Augment Genetic Counseling





# Evidence Based Cancer Control Programs (EBCCP)

<https://ebccp.cancercontrol.cancer.gov>

## Transforming Research into Community and Clinical Practice

The **EBCCP (formerly RTIPs)** website is a searchable database of evidence-based cancer control programs and is designed to provide program planners and public health practitioners easy and immediate access to program materials.

 Search Now

## Breast Cancer Risk & Genetic Testing

### PROGRAM SYNOPSIS

Designed to enhance knowledge in the decision-making process for genetic testing for breast cancer susceptibility, this intervention is a computer-based decision aid that educates women about breast cancer, heredity, and the benefits and limitations of testing so they can make informed decisions. The study showed increased knowledge of breast cancer and genetic testing.



# Thank you

*Nonniekaye Shelburne CRNP, MS, AOCN®*

*Program Director  
National Cancer Institute  
[nshelburne@nih.gov](mailto:nshelburne@nih.gov)*



**NATIONAL  
CANCER  
INSTITUTE**

[www.cancer.gov](http://www.cancer.gov)

[www.cancer.gov/espanol](http://www.cancer.gov/espanol)

## Q&A

### PANEL 5

# Research and Funding in Precision Health

---

**Moderator: Rebecca Schnall, PhD, MPH, BSN, FAAN, FACMI**

- **Elizabeth Eisenhauer, PhD, RN**
- **Theresa Koleck, PHD, RN**
- **Nonniekaye Shelburne, CRNP, MS, AOCN**

**We'll be right back with our final session**

# **Closing Comments**

## **Precision Health in Nursing: Inception to Praxis**

---

**Evangeline Fangonil-Gagalang**  
PhD, MSN, RN



# **Closing Comments**

## **Precision Health in Nursing: Inception to Praxis**

---

**Evangeline Fangonil-Gagalang**  
PhD, MSN, RN

# Precision Health

From Concept to Praxis

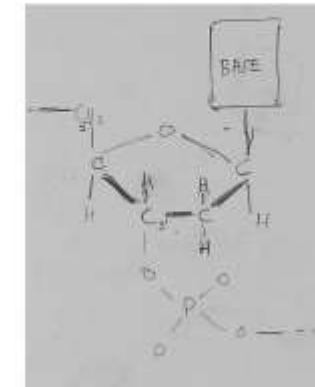
Dr. Evangeline Fangonil-Gagalang, Ph.D., MSN, RN  
Undergraduate Nursing Program Director/Assistant Professor

# Paving the Way for Precision Medicine

## The Discovery of the Double Helix, 1951-1953



## Defining the Genetic Coding Problem, 1954-1957



<https://www.genome.gov/>

<https://www.genome.gov/human-genome-project>

<https://profiles.nlm.nih.gov/spotlight/sc/feature/biographical-information>

<https://www.nature.com/scitable/topicpage/discovery-of-dna-structure-and-function-watson-397/>



# Precision Medicine

“Change will not come if we wait for some other person or some other time. We are the ones we've been waiting for. We are the change that we seek.”

Barack Obama



**THE PRECISION MEDICINE INITIATIVE**



# The PMI Promise

- biomedical discoveries
- patient-powered research
- advanced tools

# Precision Medicine?

- What is it?
- What is the definition of Precision Medicine?
- What does it mean for Nursing?
- How does it affect Nursing?
- What is the role of the nurse?
- What do nurses need to do differently?





Now, Precision Health

## Nine Years Later

- Roles and Functions of the Nurse
  - Dr. Mary Anne Schultz

## Nine Years Later

- Clinical Practice
  - Dr. Danielle Fournier
  - Dr. Marilyn Hammer
- Home Care
  - Dr. Kevin Whitney



## Nine Years Later

- Technology
  - Ms. Cathy Turner
  - Dr. Anna Schoenbaum & Mr. Johnston Thayer  
(Electronic Health Record)

## Nine Years Later

- Education & Curricula
  - Dr. Erin Dickman
  - Dr. Kathleen Calzone
  - Dr. Patricia Sharpnack

## Nine Years Later

- Research
  - Dr. Elizabeth Eisenhauer
  - Dr. Theresa Koleck



# Nine Years Later

- Research

U.S. Department of Health & Human Services National Institutes of Health

NIH National Institutes of Health  
All of Us Research Program

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**The future of health begins with you.**

The *All of Us* Research Program is inviting one million people across the U.S. to help build one of the most diverse health databases in history. We welcome participants from all backgrounds. Researchers will use the data to learn how our biology, lifestyle, and environment affect health. This may one day help them find ways to treat and prevent disease.

JOIN NOW!

<https://allofus.nih.gov/>

# Nine Years Later

- Research

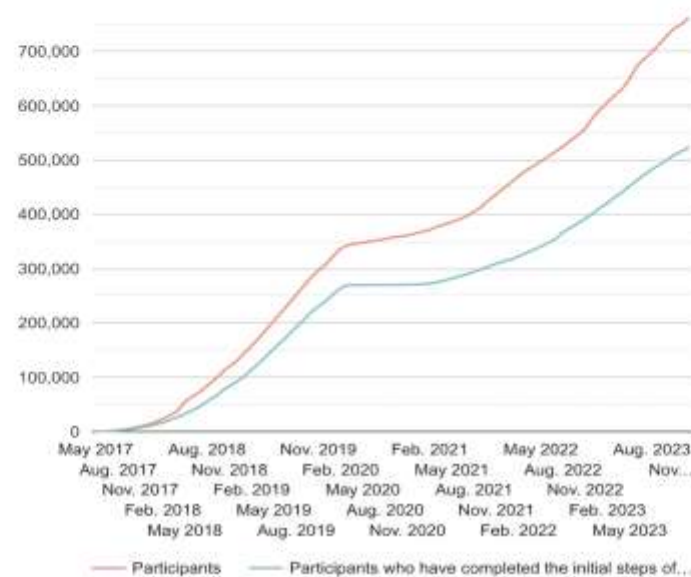
## Participants at a Glance



## Enrollment Numbers

This graph represents participants who have consented to join the program and those who have completed all initial steps of the program. The initial steps are consenting, agreeing to share electronic health records, completing the first three surveys, providing physical measurements, and donating at least one biospecimen to be stored at the biobank.

*The following numbers are approximated to protect participants' privacy. Numbers are updated as of February 5, 2024.*



# Nine Years Later

- Research

The screenshot shows the NIH All of Us Research Program website. At the top, it identifies the U.S. Department of Health & Human Services and the National Institutes of Health. The main header includes the NIH logo, the text "National Institutes of Health All of Us Research Program", and navigation links for "Log In" and "JOIN NOW". A search bar is also present. Below the header, a navigation menu lists "About", "Get Involved", "Funding and Program Partners", "Protecting Data and Privacy", and "News and Events". The central focus is a yellow banner titled "Data Now Available in the Researcher Workbench". Below this banner, seven data points are displayed with corresponding icons: Survey Responses (413,350+), Physical Measurements (337,500+), Genotyping Arrays (312,900+), Electronic Health Records (287,000+), Whole Genome Sequences (245,350+), Fitbit Records (15,600+), and Long-Read Sequences (1,000+). At the bottom of the data section, the text "All of Us Resources Beyond Data" is visible.

Category	Count
Survey Responses	413,350+
Physical Measurements	337,500+
Genotyping Arrays	312,900+
Electronic Health Records	287,000+
Whole Genome Sequences	245,350+
Fitbit Records	15,600+
Long-Read Sequences	1,000+



# Thanks!

## Any questions?

You can find me at

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- [LinkedIn](#)



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