

# Putting DNA to Work for Patients

## UVMHN Genomic Population Health Report

Robert S. Wildin, M.D.

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Associate Professor

Departments of Pathology & Laboratory Medicine and Pediatrics

Robert Larner, M.D. College of Medicine at UVM

Associate Medical Director of Genomic Medicine

University of Vermont Health Network



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# UVM Medical Center



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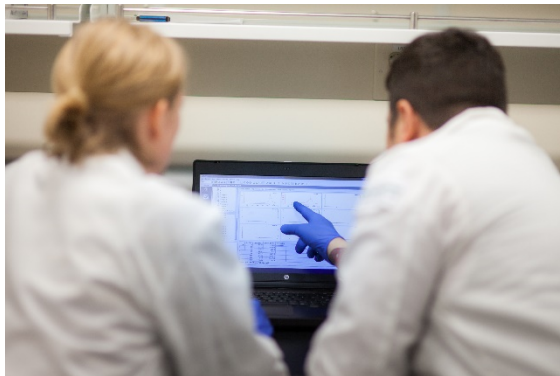
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# Genomic Medicine Program/CLIA Laboratory

## Pathology & Laboratory Medicine

- Somatic Cancer Genomics
  - NGS Testing of tumor DNA and RNA for prognosis and therapy of solid tumors (and hematologic malignancies by 12/2020)
  - Precision medicine applied to therapy of manifest disease



- Genomic Population Health
  - NGS “screening” of unselected adults for hidden genetic disease and treatment risks
  - Genotyping for variants affecting response to common medications (Spring 2021)
  - Detection of genetically-defined health risks, enabling management before symptomatic presentation



# Responsive vs. Preventative care



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Genetics  
inMedicine

ACMG POLICY STATEMENT

ACMG policy statement: updated recommendations regarding analysis and reporting of secondary findings in clinical genome-scale sequencing

ACMG Board of Directors<sup>1</sup>

Disclaimer: These recommendations are designed primarily as an educational resource for medical geneticists and other health-care providers to help them provide quality medical genetics services. Adherence to these recommendations does not necessarily ensure a successful medical outcome. These recommendations should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, geneticists and other clinicians should apply their own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from these recommendations.

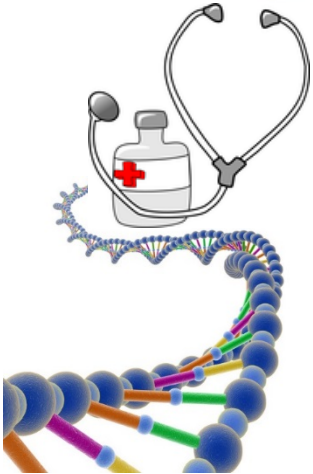
As genome-scale sequencing is increasingly applied in clinical medicine, complex issues arise regarding the extent to which primary data should be analyzed and reported. At the present time, the most common clinical application of massively parallel sequencing lies in its use as a powerful new diagnostic tool in selected patients. When such sequencing is performed, primary data files consisting of a vast number of genomic variants are generated for each individual, with that information varying greatly with regard to relevance to the specific diagnostic question. What to do with these large numbers of "secondary" or "incidental" variants (nomenclature has varied with regard to such findings; the American College of Medical Genetics and Genomics (ACMG) has now adopted "secondary findings" as standard nomenclature, as recommended by the Presidential Commission on Bioethical Issues<sup>2</sup>) has been a matter of considerable debate and discussion. Challenges include how extensively the primary data should be routinely analyzed and which of the many variants discovered should be reported to patients. These questions are especially challenging because although many of these variants are not clinically relevant or are uninterpretable, a minority may have important medical implications for the individual patient.

of severe disease that is preventable if identified before symptoms occur.

The release of this set of recommendations resulted in considerable discussion, much of it focused on whether the analysis of these 56 genes should be "mandatory" when whole-exome/whole-genome sequencing is pursued clinically, or whether patients should be able to "opt out" of such secondary analysis and reporting. This discussion was informed by a report by the Presidential Commission on Bioethical Issues regarding secondary findings and a survey administered by the ACMG to its membership in January 2014. In March 2014, the ACMG updated its recommendations, prompted in part by what appeared to be a general consensus among ACMG members and other relevant stakeholders that patients should be able to opt out of the analysis of genes unrelated to the indication for testing, and that the decision should be made during the process of informed consent before testing.

In this issue of *Genetics in Medicine*, we publish the results of this survey<sup>3</sup> and articulate the current recommendations of the ACMG with regard to the analysis and return of secondary findings when clinical genome-scale analysis

# Vision: Genomes for All

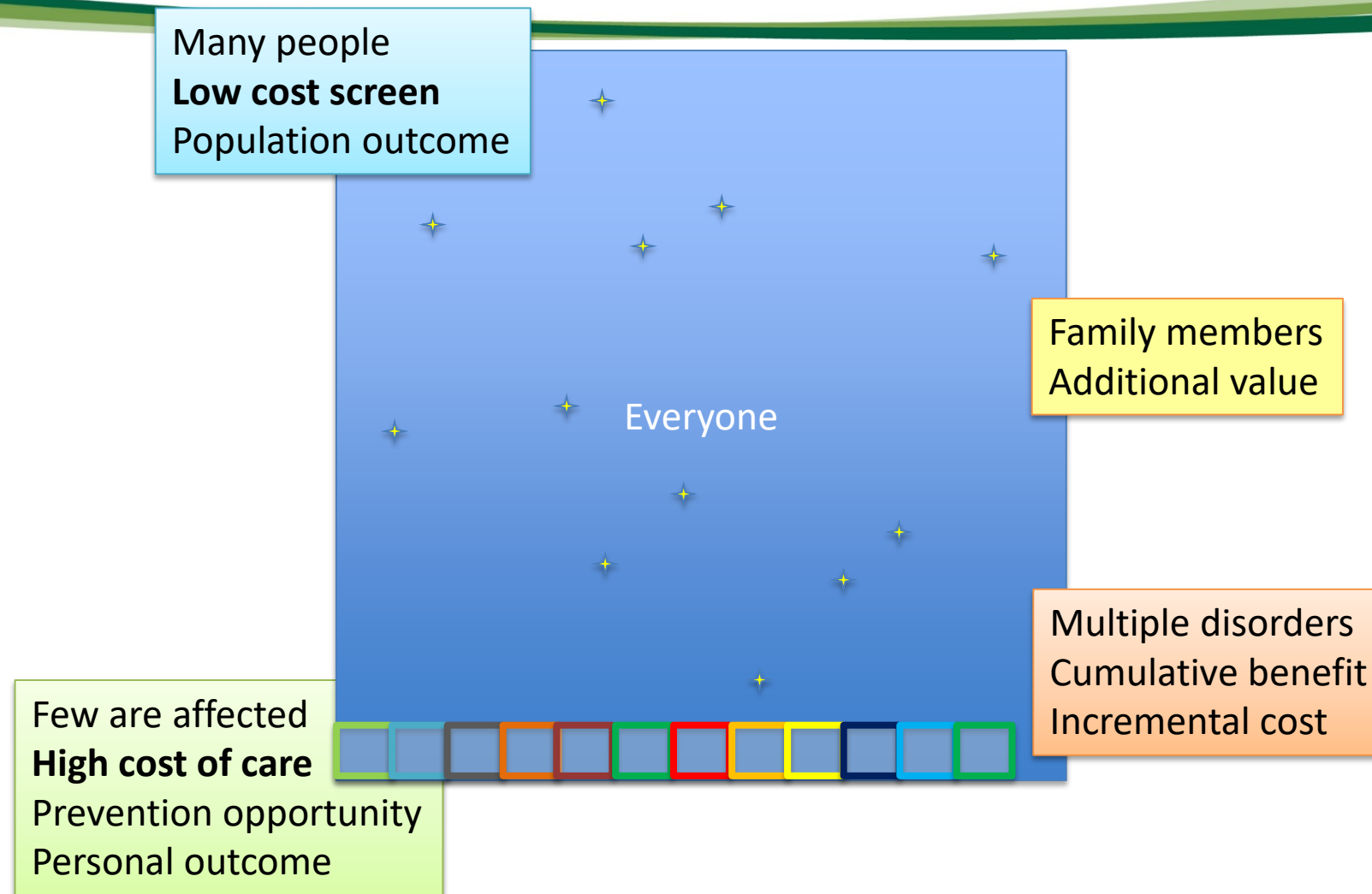


- We will integrate the use of genetic information into usual medical practice in diagnosis, prevention, and treatment
- We will demonstrate the value of genetics for health outcomes and cost for our 1 million patients

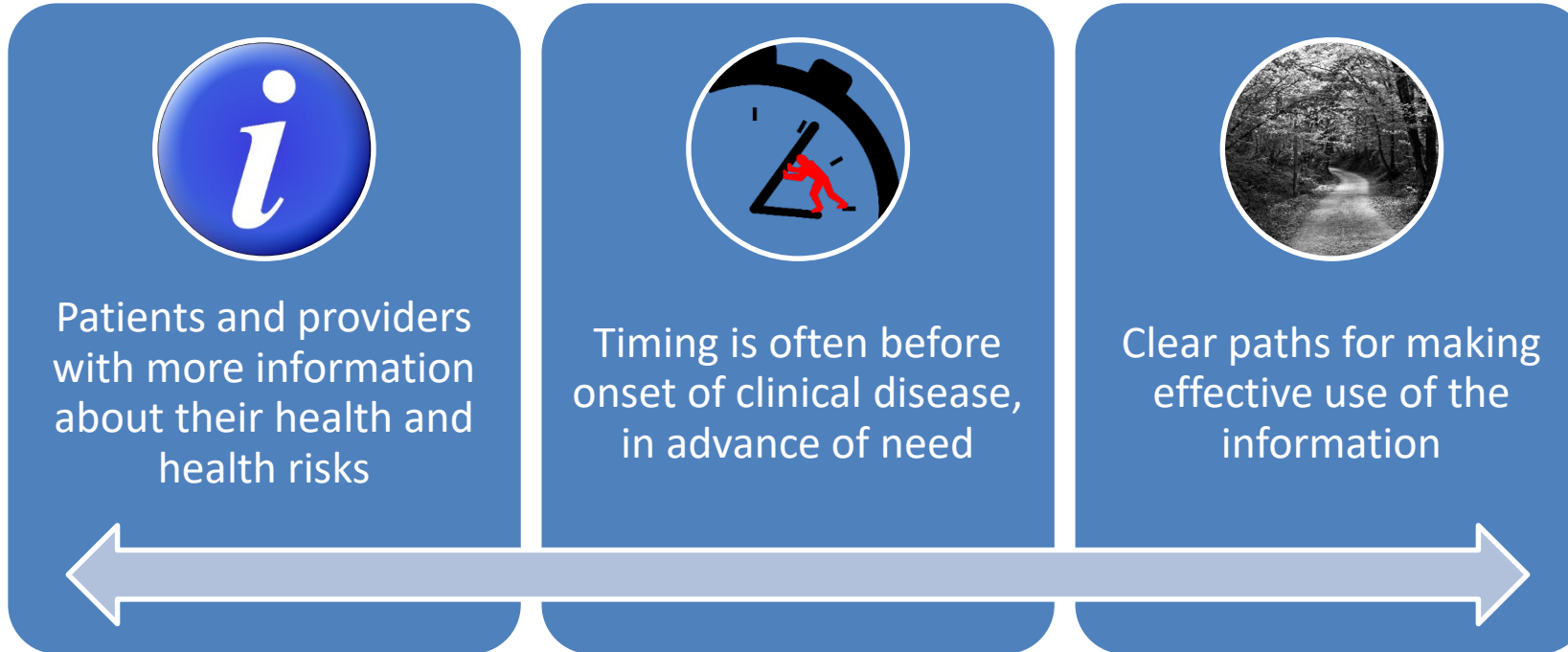




# Multiplex Genomic Screening Test – the Value Proposition



# What does it look like at the end?





# Newborn Screening is an Example of Population Health for Genetic Diseases

- VT Newborn screening is mandated by state law and detects:
  - 37 rare disorders for which an established intervention exists that can change outcomes
  - Disorders for which precision therapy (targeting the biological defect) exists
  - Disorders that require lifelong management
  - Disorders where the cost of care is low to extraordinarily high

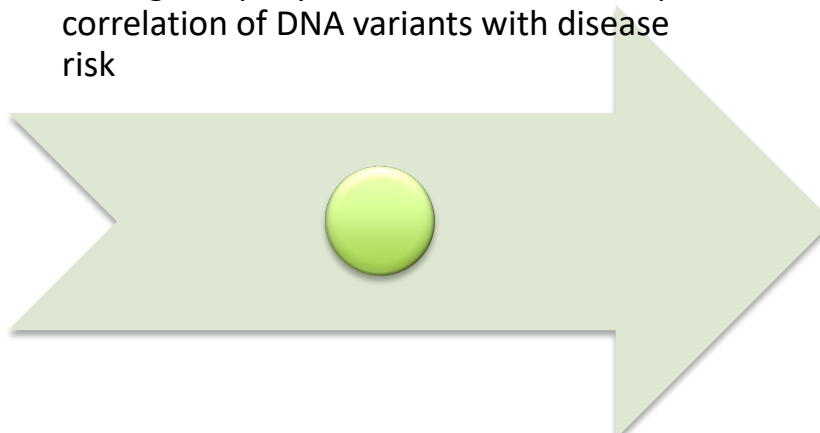
**\$203.00  
per baby**



# Clinical Test Used for Clinical Care

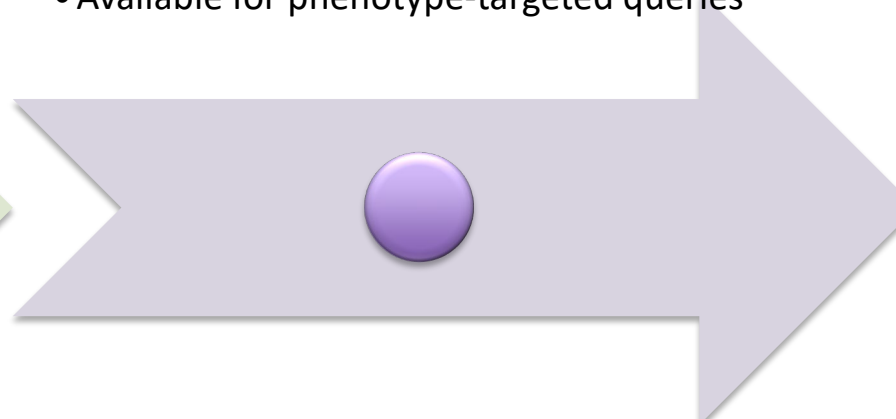
## The Genomic DNA Test

- Gene panel test set
- Performed in a clinical laboratory so results can be directly used for patient care
- Sequencing through an expert genomic testing company allows lower cost & expert correlation of DNA variants with disease risk



## The Clinical Genome Test

- (Future)
- Genome sequence
- Interpretation of clinically actionable genes, *including rare ones*
- Available for phenotype-targeted queries





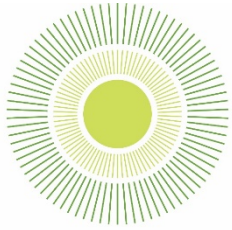
# Core Implementation Requirements

- System Leadership
- Integrate with Clinical Care
- No economic barrier
- Planning and Oversight
- Patient and Provider
  - Receptiveness
  - Competency
- Scalability mindset
- Learning enabled
  - Responds to changing knowledge
  - Enables new knowledge

- Engagement path
- Clinical sequencing and interpretation
- Clinical informatics
- Clinical care pathways
- Clinical decision support (traditional)
- Communication
- Quality mindset
- Funding
- Persistence



# Powered by Partnerships



OneCareVermont



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# Understand Your Genome (UYG)

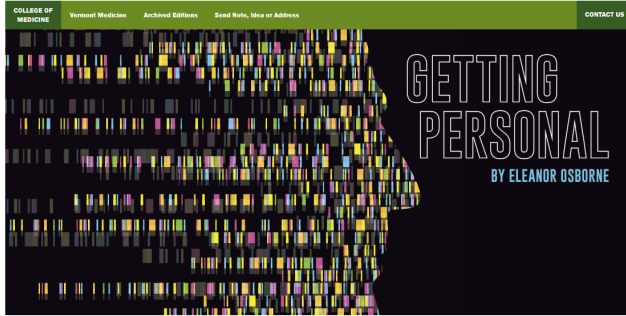


- 2012-2017 Illumina Symposium Program\*
- 70+ UVMHN system leaders and staff
- ~1200 genes sequenced
- Results returned
- Outcomes
  - At least one life-changing result
  - Several clinically important results

*\*Now supported by Genome Medical*

# Leadership Engagement

Vermont Medicine Magazine



"I was doubting myself, then I was sure it was the right thing to do, then I was doubting myself again—it was an endless loop of uncertainty. Finding out put an end to the cycle. It was powerful." - Michael Towle

[http://contentmanager.med.uvm.edu/vtmedicine/getting\\_personal](http://contentmanager.med.uvm.edu/vtmedicine/getting_personal)



Debra Leonard, M.D., Ph.D., and Michael Towle

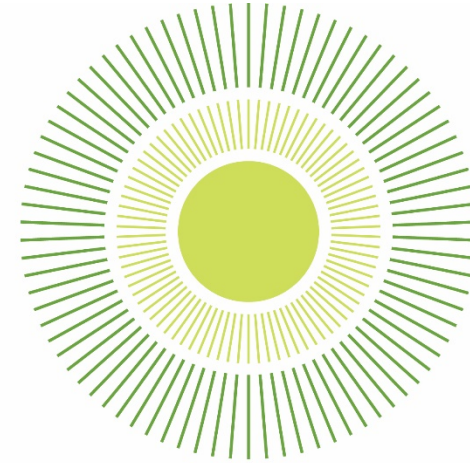


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# Partner with Accountable Care Organization

- \$0 cost
  - Care Innovation Waiver
- Long-term impact
  - cost and outcomes analysis



OneCareVermont

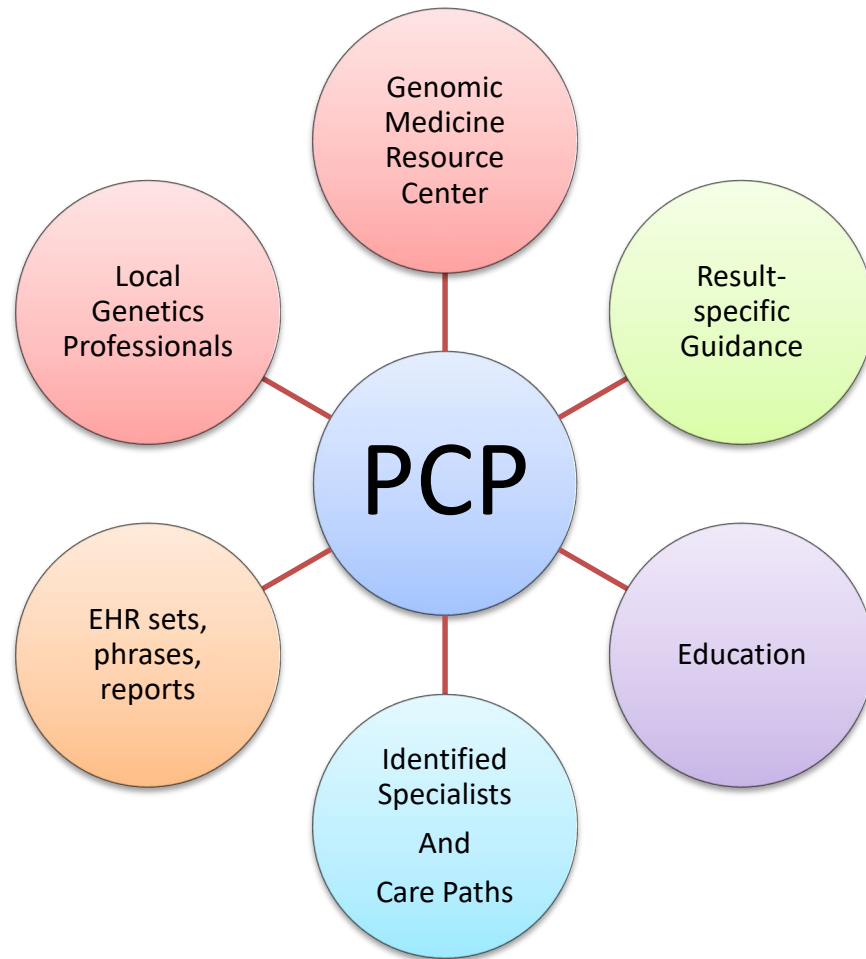


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# Primary Care Toolbox



# Clinical pathways to integrate genomic testing into patient care

Identify patients, educate about the test

Obtain and ship specimen

Integrate genomic results into EHR

Guide PCPs and work with specialists to coordinate care

Multi-disciplinary conferences to learn and optimize care

Obtain clinical informed consent

Perform genomic test & report interpretation

Counsel patient (& family), as appropriate

Test family members ("Cascade testing")



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# Care Pathway Partners in UVMHN

Cardiology

Oncology

Pulmonary and Anesthesiology

Pharmacy

Medical Ethics

Healthcare Value Experts

*Patient and Family Advisors*

Primary Care



Aaron Reiter, MD

Oncology

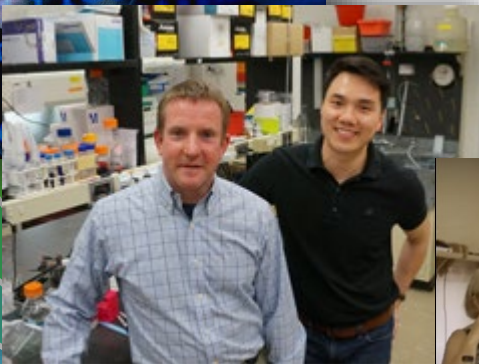


Marie Wood, MD

Cardiology



Daniel Correa de Sa, MD



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# What is “the Genomic DNA Test”?

Test for genetic differences that can cause diseases in the tested person or their family members

Invitae  
Pro-Active  
**Health Screen** –  
147 genes for  
health risks

- If positive, blood relatives get free testing

Invitae  
Comprehensive  
**Carrier Screen** –  
301 genes for  
recessive disorders

- Low cost partner testing

Total of 431 genes

- Some genes are reported on both panels

In the future

- Pharmacogenomic variation (drug-gene interactions)
- Genome sequencing (in place of panels)
- Polygenic Risk Scores ??????

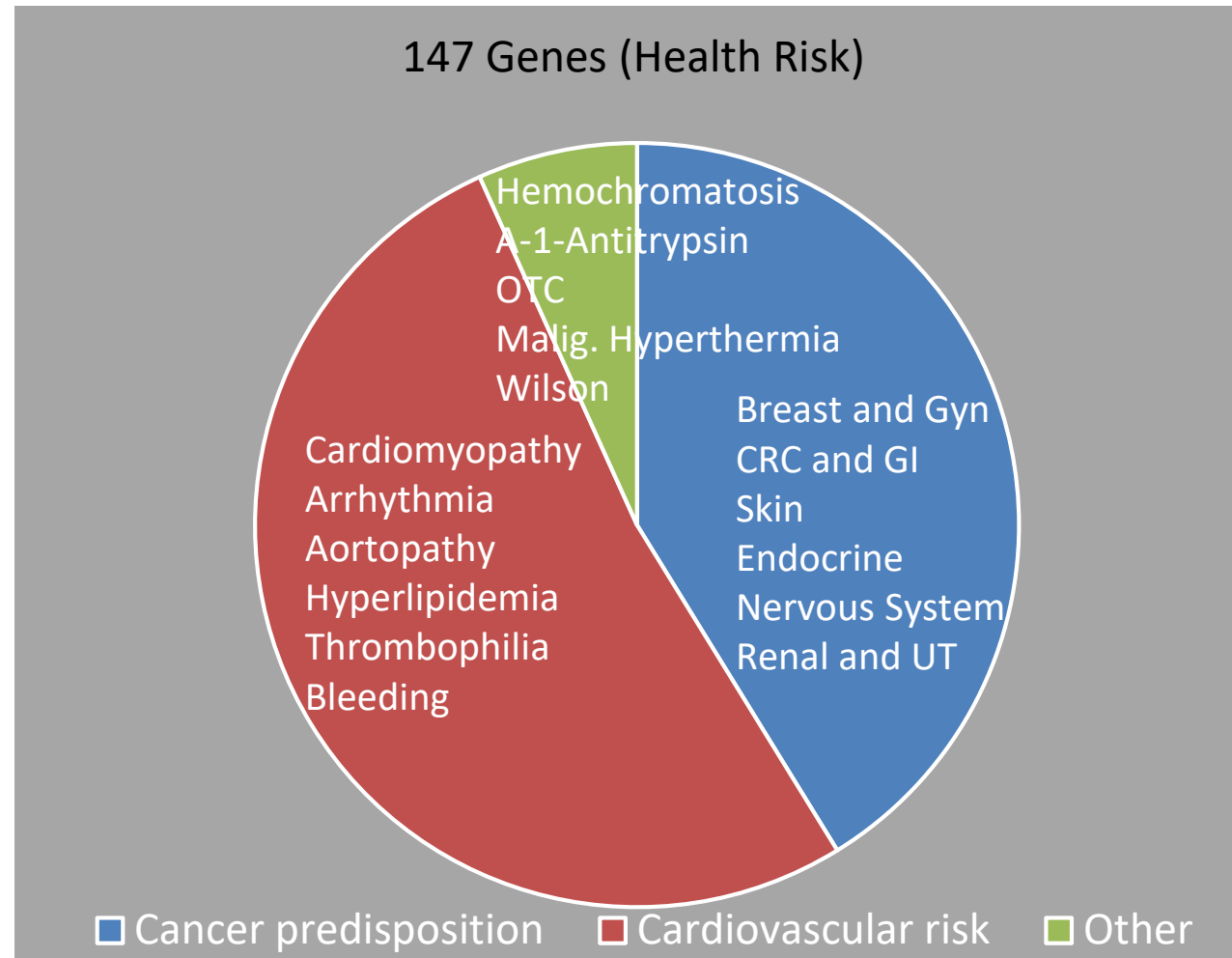
Variants of Uncertain Significance (VUSs) are NOT REPORTED



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# What is “the Genomic DNA Test”?



# Patient Information

- Brief animated video
- Tri-fold brochure
- FAQ document
- Genes tested list
- Informed consent form
- Next steps  
(after consented and ordered)
- Forms for providers



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The University of Vermont Medical Center > Departments & Programs > Genomic Lab > Genomic Population Health and The Genomic DNA Test

## GENOMIC POPULATION HEALTH AND THE GENOMIC DNA TEST

UVM Medical Center offers the Genomic DNA Test to help you and your health care providers use your genes to inform your health care.

Genomic Population Health and The Genomic DNA Test

**COVID-19 ALERT:** Get the care you need, safely and when you need it. Video visits are available and safety measures are in place for required in-person visits to practices, Urgent Care and the Emergency Department. Call your provider with questions about your health or upcoming visits. [Learn more about the ways you can safely receive care here.](#)

### The Genomic DNA Test

Your health is important to us. Many things determine your health. One important factor is your genetics. Understanding any differences in your DNA can provide valuable health information to guide your health care. We are offering the Genomic DNA Test so that you and your health care providers may know and use the information in your genes to inform your health care.

Watch the short overview video and read the information documents to learn more.

UVMHVN Genome Test Overview Animation

Watch later Share

### Test Information for Patients

Please read these downloadable PDF documents for more information about the Genomic DNA Test. Cost and eligibility are addressed in both the brochure and the FAQ document.

- [Patient Brochure](#)
- [Frequently Asked Questions \(FAQ\)](#)
- [Genes Tested](#)
- [Informed Consent Form](#)
- [Next Steps \(after provider orders test\)](#)

**Saliva Sample Option:** If your provider orders your test, you may ask for a saliva (spit) collection kit to be mailed to you instead of a blood sample drawn. You will collect the sample at home and ship it back. Note that certain test parts (pharmacogenomics) may not be available when using saliva instead of blood. The following links describe collecting saliva at home and then shipping it using a pre-paid

### In The News

- [VPR: For One Vermont Man, Sequencing His Whole Genome Solved A Life of Pain](#)
- [WSJ: Why Knowing Your Genetic Data Can Be a Tricky Proposition - PDF](#)
- [BFP: Genomes Unlock Mystery of Diseases - PDF](#)

### LEARN

- [Apolipoprotein E-4 Genetic \(DNA\) Test](#)
- [Genetics](#)

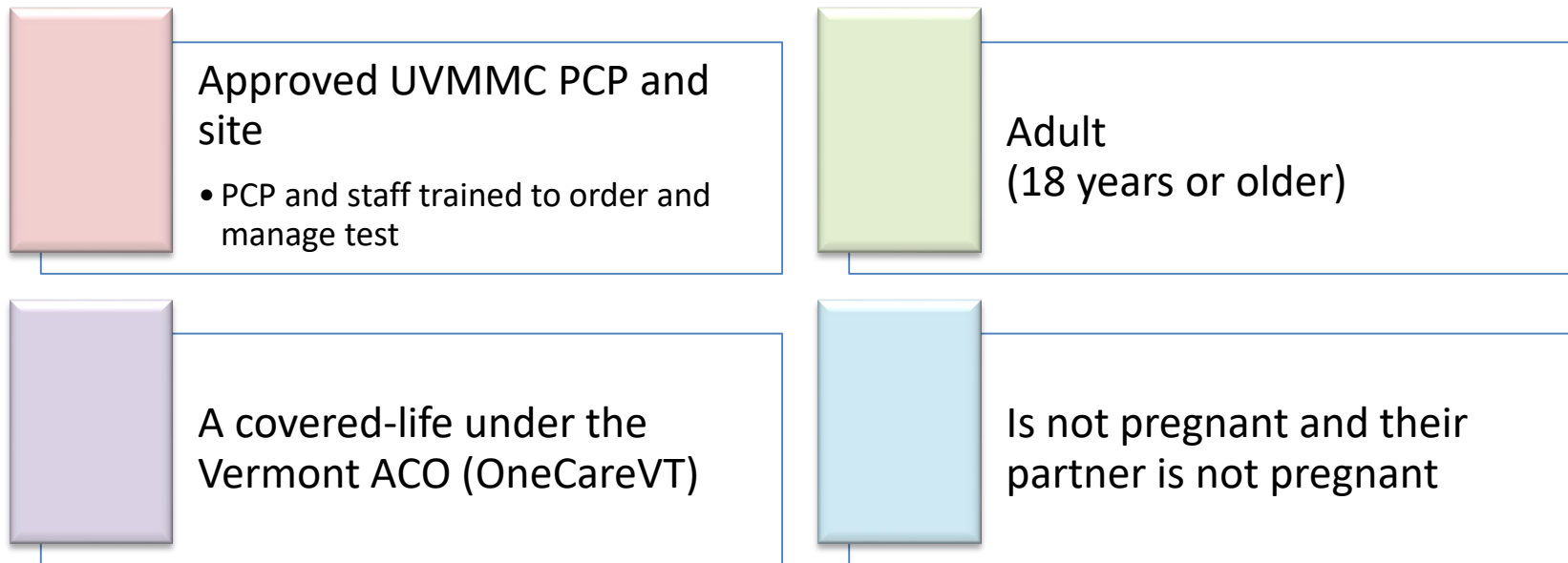
### BLOGS

- [Inside the Double Helix: What is Pharmacogenomics & How Will It Change Medicine?](#)
- [Bioinformatics: What is Personalized Genomic Medicine?](#)
- [Bioinformatics: How to Read Your "Genome Book"](#)

Subscribe by email

<https://uvmhealth.org/genomicDNAtest>

# Who is eligible to get the test?



Not to be used to diagnose suspected genetic disease







# How does it work?

## For the PCP and staff

- Identify eligible patients
- Introduce the test at visit using patient information folder and video
- Refer genetics questions to GMRC
- Complete paperwork and order
- Review results, print and mail
- Appropriate discussion of results
- Order follow up actions, document

## For the patient

- Review information
- Ask questions
- Sign Informed Consent
- Blood draw (4cc x 2, lavender)
- Get call from PCP office re results
- Receive printed results in the mail
- Appropriate genetic counseling

# Obtaining a DNA Sample



- Blood – 4ml EDTA x2
- Saliva – Orogene™ kit supplied by Invitae





# How long does the test take to result?

Typically 3  
weeks



Tell patient  
~1 month



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# Where do I find the results?



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# What do the results look like?

- 3-part PDF in EHR
  - Genomic Medicine Action Plan (GMAP) – 1-2 pages
  - Pro-active Health Screen Test Report (Invitae) – several pages
  - Comprehensive Carrier Screen Test Report (Invitae) – many pages

**INVITAE COMPREHENSIVE CARRIER SCREEN RESULTS**

**About this test**  
This carrier test evaluated 301 genes for genetic changes (variants) that increase the risk of having a child with a genetic disorder. Knowledge of these variants can provide information that can be used to assist with family planning.

**Summary**  
This test shows the presence of clinically significant genetic changes. Significant changes were identified in the remaining genes evaluated.

Results	Gene	Variant(s)
CARRIER	Hemochromatosis type 1	HFE c.845G>A

**Next steps**

- See the table above for recommendations regarding testing.
- Even for genes that have a negative test result, there is always a "residual risk." See the table below for residual risks.
- Genetic counseling is recommended to further explain the results, which may point to health information that merits additional testing.
- All patients, regardless of result, may wish to consider additional testing, such as hemoglobin electrophoresis, if this has not already been done.
- Individuals can register their tests at <https://www.invitae.com> for next steps.

**INVITAE PROACTIVE TESTING RESULTS**

**Test performed**  
Sequence analysis and deletion/duplication testing of the 147 genes listed in the Genes Analyzed section.

**RESULT: CARRIER**

**A single clinically significant genetic change was found in the HFE gene, which is a carrier for a hereditary condition.**

Gene	Variant	Zygosity	Carrier Status
HFE	c.845G>A (p.Cys282Tyr)	heterozygous	Carrier

**Next steps**

- A carrier result is medically important and you should consider discussing this with your provider. This result indicates you do not have any personal genetic risk factors for the condition. It is important to note that at least a baseline, population-level risk remains for developing a disease. Appropriate screenings are still recommended. A complete family history may provide additional information. Genetic counseling is recommended to discuss the implications of this result and to help you understand your options.
- Consider sharing this result with relatives as they may also be at risk. Details on how to share your results are available at [www.invitae.com/family](https://www.invitae.com/family).
- Register your test at [www.invitae.com/patients](https://www.invitae.com/patients) to download a digital copy of your report and learn more about how your results can help inform your health.

**Genomic Medicine Action Plan (GMAP) – NEGATIVE HEALTH SCREEN AND CARRIER SCREEN**

The Genomic DNA Test is a screening test for certain genetic changes. It is created to help patients and their provider use the results to make decisions about their health. The GMAP is a set of messages, not requirements. The patient's provider will discuss the messages with the patient and their family.

**Invitae Test Report**  
Type of report: Proactive Health Screen  
Accession ID: [Invitae Accession Number - Genetic Health Screen](#)  
and  
[Invitae Accession Number - Comprehensive Carrier Screen](#)  
Date of Report: 3/5/2020

**Result Summary** – see the associated report(s)  
**No reportable genetic health risk or carrier variants were found in the genes tested (Negative).**

Messages	The patient should...	The provider should...
<b>Act (do now)</b>	Tell your provider if you have a genetic disease, or if you have a family history of a genetic disease. They may want to do other testing.  Share these results with your family members. Share the "Be Aware" information below with your family members.	Update and review the family history. A strong family history may suggest health risks not found with this test. Additional evaluation may be indicated.  Encourage the patient to share this information with family members.
<b>Be aware</b>	A negative test lowers your risk for certain conditions. It does not mean that you don't have any genetic risks or diseases, and it does not mean that your children have no genetic risks. This result means that no DNA difference was found in the genes tested that experts consider important for your health. To learn more, read the test's FAQ document, or speak with a genetics expert at the Genomic Medicine Resource Center (no cost).  Genetic counseling: If you want to speak with a genetics expert about these results and what they mean for you and your family, contact the UVMHC Genomic Medicine Resource Center. Send an email to <a href="mailto:DNAtest@uvmhealth.org">DNAtest@uvmhealth.org</a> or call (802) 847-8135. There is no cost for this service.	If the patient or a family member is diagnosed with or is suspected to have genetic disease, whether or not related to the genes on this test, review this result and contact the testing laboratory to discuss. A genetic counseling visit may be appropriate.  <i>If the patient wishes to share their DNA information with researchers, they can complete an online consent on LunaDNA. If they wish to share their health record data on LunaDNA, they must have a MyChart account. Please help them register for MyChart in that case.</i>
<b>Come back to it</b>	Review this result with your primary care provider every few years. New knowledge may reveal new health value in your DNA differences.	Review the clinical relevance of these results every few years. The interpretation of the test data can be updated in the future using new knowledge.

# Using the results The Action Plan (GMAP)

- Title
- Disclaimer
- Demographics and report info
- Result summary
- Patient Messages and Provider Messages
- Message groups in A-B-C framework
  - Act (do now)
  - Be Aware
  - Come back to it

## Genomic Medicine Action Plan (GMAP) – NEGATIVE HEALTH SCREEN AND CARRIER SCREEN

The Genomic DNA Test is a screening test for certain genetic health risks and carrier status. This GMAP is created to help patients and their provider use the results in the test report for better health. It does not replace the Laboratory Reports. It is intended to be used along with the official Laboratory Reports. The GMAP is a set of messages, not requirements. The patient and the provider should together decide if the messages make sense for the patient and their family, and contact the Resource Center with questions.



Patient	Invitae Test Report(s) - associated reports	Action Plan prepared by:
Name: «Name»	Type of report: Pro-active Testing and Comprehensive Carrier Screen	Christine Giummo, CGC
DOB: «dob»	Accession ID:	Robert Wildin, MD
MRN: «MRN»	«Invitae_accession_number_Genetic_Health»	Date prepared: 3/5/2020
PCP: «PCP»	and «Invitae_accession_number_comprehensiveness»	
	Date of Report: ____ and ____	

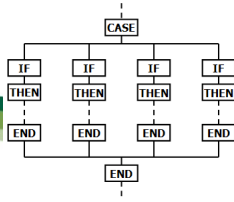
### Result Summary - see the associated report(s)

**No reportable genetic health risk or carrier variants were found in the genes tested (Negative).**

Messages	The patient should...	The provider should...
<b>Act (do now)</b>	<p>Tell your provider if you have a genetic disease, or if you have a family history of a genetic disease. They may want to do other testing.</p> <p>Share these results with your family members. Share the "Be Aware" information below with your family members.</p>	<p>Update and review the family history. A strong family history may suggest health risks not found with this test. Additional evaluation may be indicated.</p> <p>Encourage the patient to share this information with family members.</p>
<b>Be aware</b>	<p>A negative test lowers your risk for certain conditions. It does not mean that you don't have any genetic risks or diseases, and it does not mean that your children have no genetic risks. This result means that no DNA difference was found in the genes tested that experts consider important for your health. To learn more, read the test's FAQ document, or speak with a genetics expert at the Genomic Medicine Resource Center (no cost).</p> <p><u>Genetic counseling</u> If you want to speak with a genetics expert about these results and what they mean for you and your family, contact the UVMGC Genomic Medicine Resource Center. Send an email to DNAtest@uvmhealth.org or call (802) 847-8135. There is no cost for this service.</p>	<p>If the patient or a family member is diagnosed with or is suspected to have genetic disease, whether or not related to the genes on this test, review this result and contact the testing laboratory to discuss. A genetic counseling visit may be appropriate.</p> <p><i>If the patient wishes to share their DNA information with researchers, they can complete an online consent on LunaDNA. If they wish to share their health record data on LunaDNA, they must have a MyChart account. Please help them register for MyChart in that case.</i></p>
<b>Come back to it</b>	<p>Review this result with your primary care provider every few years. New knowledge may reveal new health value in your DNA differences.</p>	<p>Review the clinical relevance of these results every few years. The interpretation of the test data can be updated in the future using new knowledge.</p>

# What does PCP office do with the results?

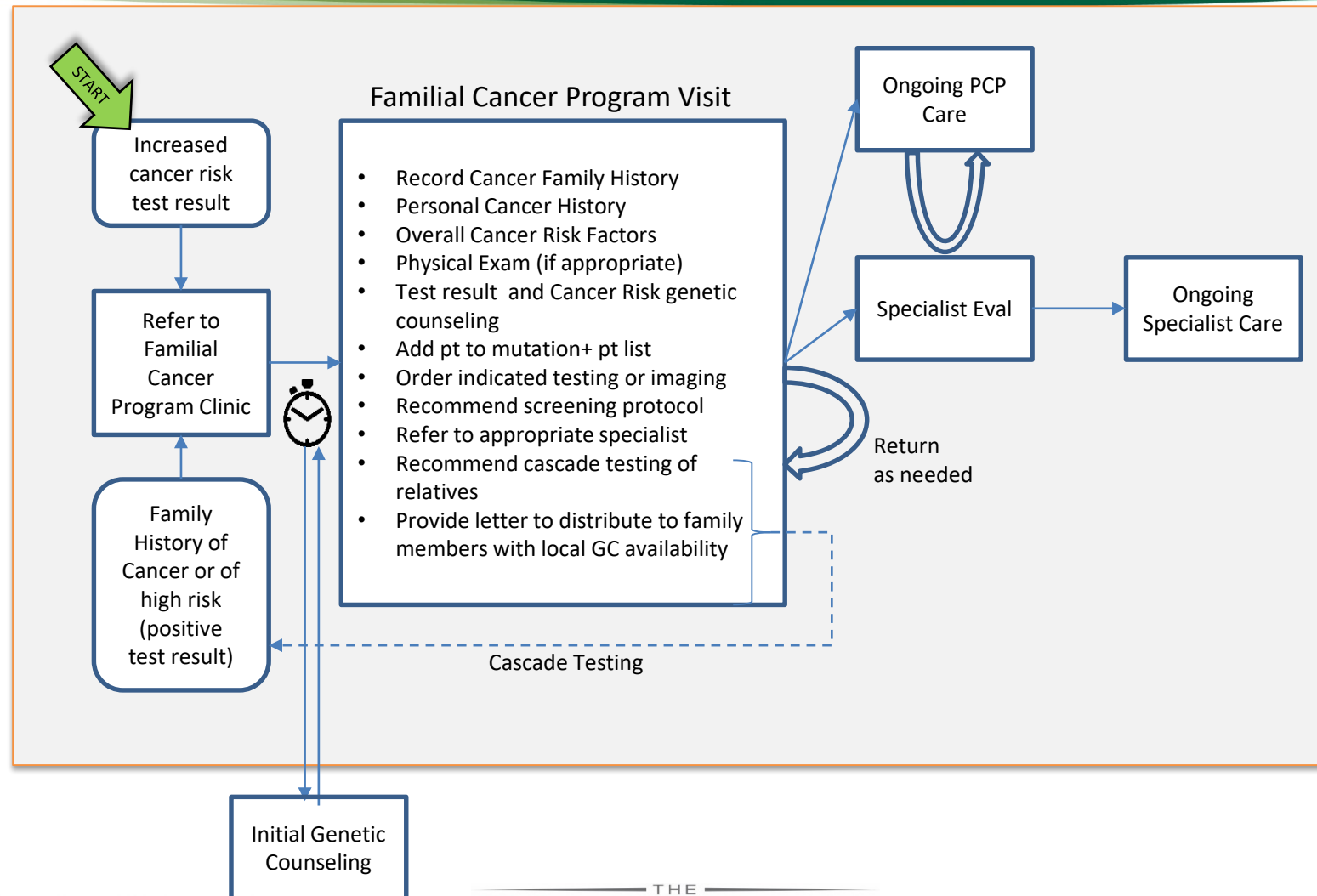
## (example triage scheme)



Result Class	Actions
Negative, Negative	<b>Support staff</b> prints and mails result packet to patient
Carrier results only	Also: <ol style="list-style-type: none"> <li>1. Consider partner testing</li> <li>2. <b>Nurse</b> calls patient, encourages reading of mailed items, discusses partner testing</li> <li>3. Recommends contacting Resource Center <b>if questions or genetic counseling is desired</b> and appropriate to reproductive or family member status</li> </ol>
Positive Health Risk result	Also: <ol style="list-style-type: none"> <li>4. <b>Provider</b> calls patient, or telemed./face-to-face visit</li> <li>5. Review Genomic Medicine Action Plan; implement actions when appropriate to overall patient context;</li> <li>6. <b>Recommend</b> contacting Resource Center for genetic counseling (or FCP referral for cancer-risk result).</li> </ol>
Combined, Complex, or Ambiguous result	Also: <ol style="list-style-type: none"> <li>7. <b>Strongly recommend</b> contacting Resource Center for genetic counseling</li> </ol>

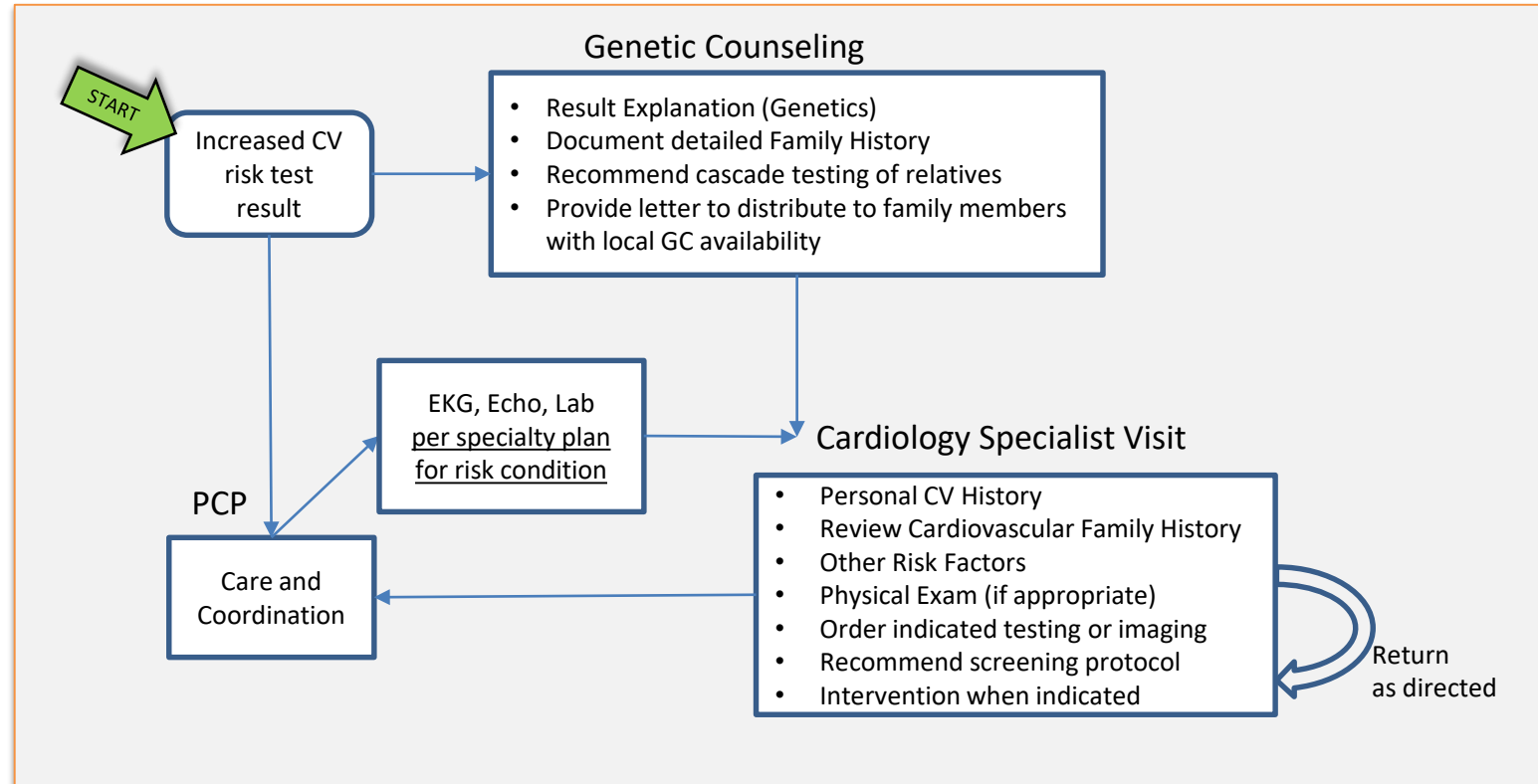


# Cancer Risk Care Pathway (v1.1)



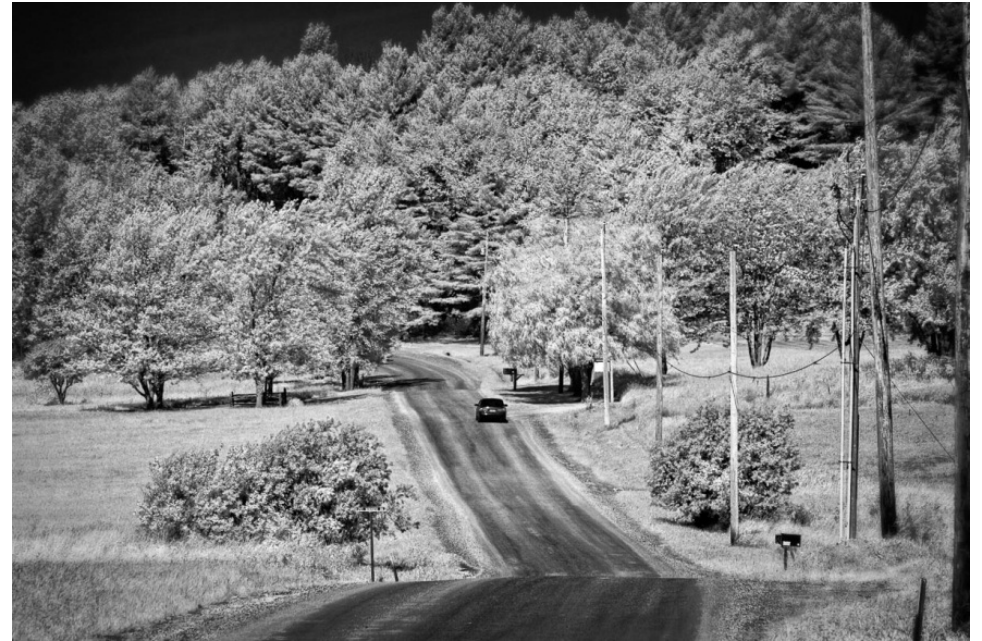


# Cardiovascular Risk Care Pathway

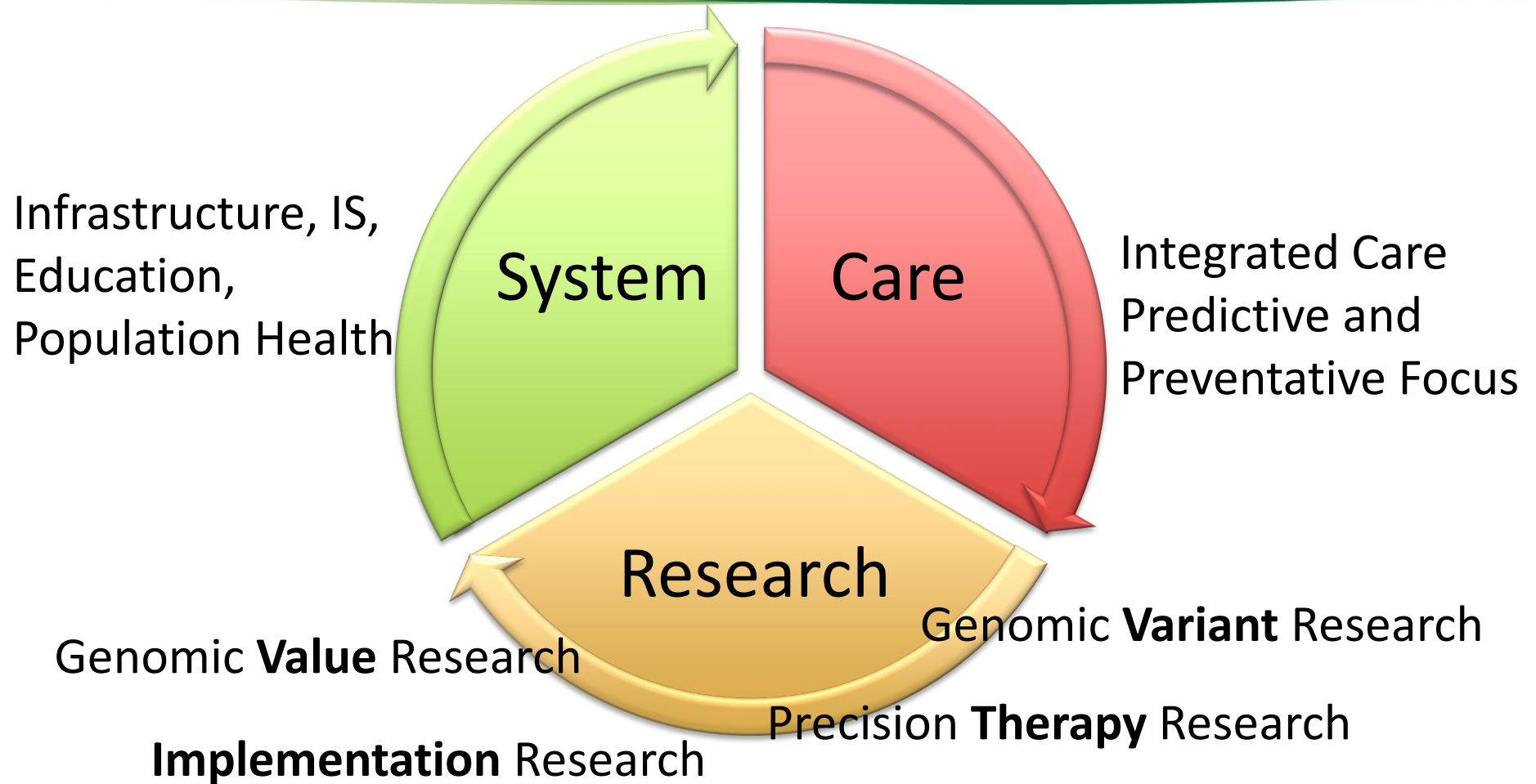


# A Genome is a Journey

- New evidence drives new value
- Updated reports are pushed when variant classification changes



# Learning Healthcare Systems



# Opportunities for Learning and Research

## Business Intelligence (OneCareVT ACO)

- Health value
- Population value
- Aggregate costs

## LunaPBC, LunaDNA platform

- “Broker” between individuals and interested researchers
- Based on fairness and confidentiality framework created by the Genetic Alliance
- Patients control their data and get shares in the company
- Genomic variant data and health record data (MyChart)

## IRB-approved research

- “I want to study this patient population” using direct EHR data, send surveys, test interventions, etc.





# Partners in Genomic Value Research

*The Center for Health Services Research*



Adam Atherly, PhD



Sarah Nowak, PhD



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## How it works

### CONTRIBUTE YOUR INFORMATION

You can easily copy your DNA information and your health history into LunaDNA's secure database.

### ACCELERATE DISCOVERY

LunaDNA lets researchers know that your information exists. If researchers want to use your data, LunaDNA will ask your permission first.

### PRIVATE + SECURE

LunaDNA makes data sharing anonymous. You control what information is shared for research, and you can stop sharing at any time. You still own your information, while giving researchers permission to study it.

### EARN SHARES

You earn ownership shares in LunaDNA for your data contribution and benefit in the value created from medical breakthroughs.

Your home for health discovery



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## Help medical researchers connect DNA variation to genetic diseases



University of Vermont Health Network (UVMHN) has partnered with LunaDNA to help research and drive medical breakthroughs.

Participating in the UVMHN Genomic DNA Test enables you to contribute your health and DNA information to LunaDNA and learn about your potential health risks.

### Why it matters

Your unique health and DNA information can help researchers study how DNA variation is related to disease. When your information is combined with that of others, more opportunities are created for medical researchers to discover treatments and cures for diseases.

The future of health care discovery can be shaped by what you share today.

**YOU HAVE THE POWER TO MAKE A DIFFERENCE  
AND CONTRIBUTE TO HEALTH CARE DISCOVERY**



To learn more, or to register, go to  
[learn.lunadna.com/UVMHN](https://learn.lunadna.com/UVMHN)

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LunaDNA lets researchers know that your information exists. If researchers want to use your data, LunaDNA will ask your permission first.

### PRIVATE + SECURE

LunaDNA makes data sharing anonymous. You control what information is shared for research, and you can stop sharing at any time. You still own your information, while giving researchers permission to study it.

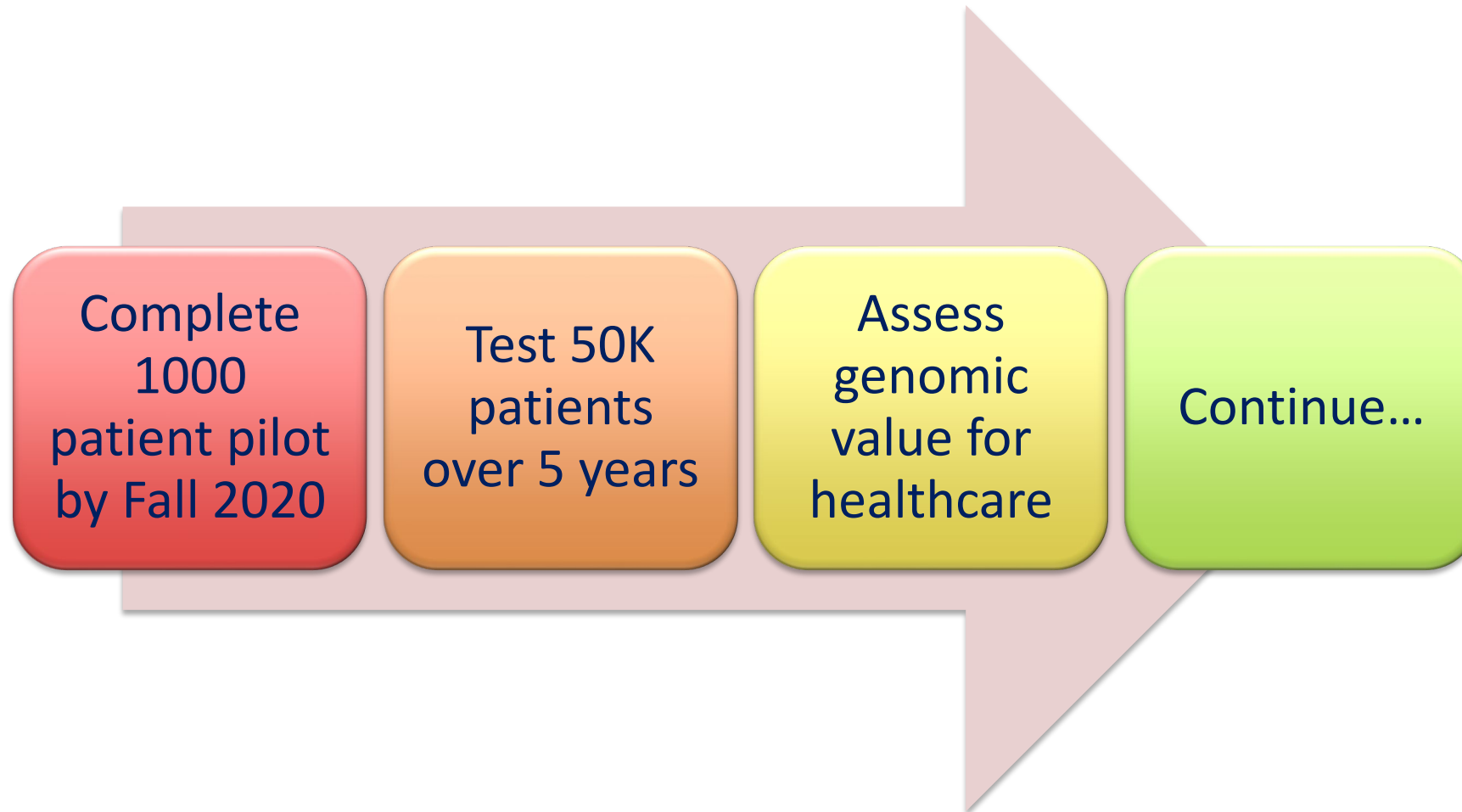
### EARN SHARES

You earn ownership shares in LunaDNA for your data contribution and benefit in the value created from medical breakthroughs.

Your home for health discovery



# Near-term Goals



# Test Results Summary as of 9/22/2020

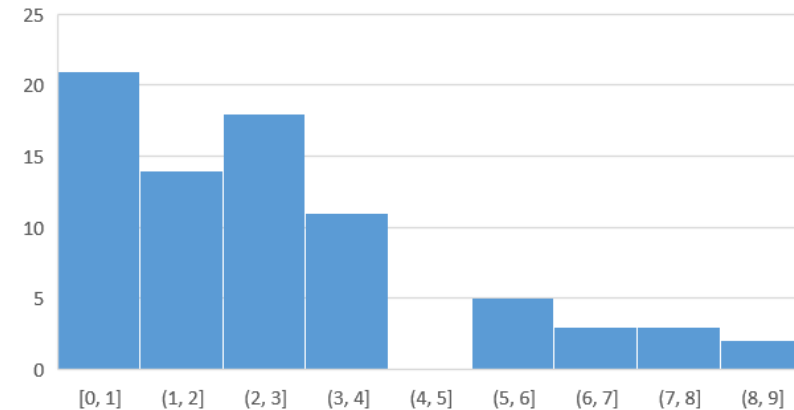
Testing started November 2019

Sites: 3  
Providers: 9

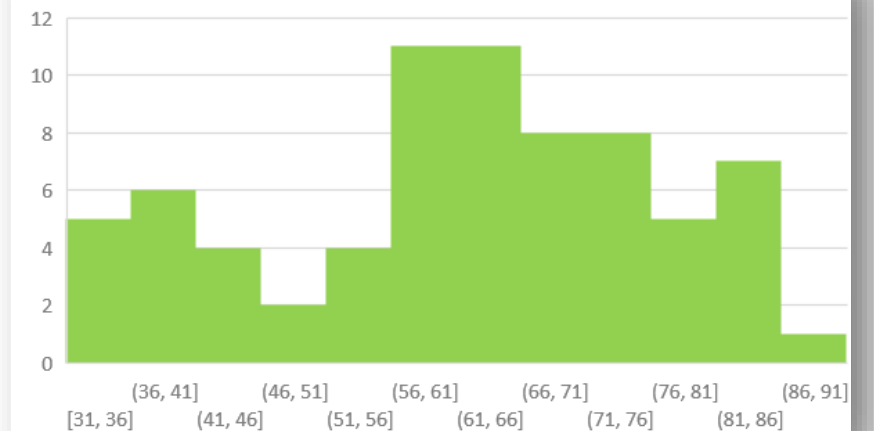
Results  
returned

76

Monthly Tests since 11/1/2019



Tested count by age group





# Testing Status (9.2020)

Measure	Count
Patient Count	76
Patients <i>without</i> any variants	16 (21%)
Patients with <i>any</i> variant	60 (79%)
Patients with at least 1 <i>carrier</i> variant	59 (76%)
Variants placing <i>tested person at increased risk</i>	23 (17%)
Total number of unique variants identified	136 (avg 1.8/individual)



# Dominant and Recessive Disorders Detected (first 76 pts)

GENE	PATIENTS	DISORDER
BRCA1	1	Hereditary Breast and Ovarian Cancer
CHEK2	2	Susceptibility to Breast and Prostate Cancer/Li-Fraumeni Syndrome 2
DMD	1	Carrier susceptibility to early onset cardiomyopathy
DSG2	1	Dilated cardiomyopathy/ARVD10
F2	1	Thrombophilia (dys- or hypo-thromboproteinemia)
F5	2	Factor V Leiden
MITF	1	Hereditary Melanoma/Renal carcinoma Risk
NBN	1	Hematologic disease susceptibility
PMS2	1	Hereditary non-polyposis colorectal cancer (HNPCC)
PRKAR1A	1	Carney Complex (neoplasias)
SDHD	1	Hereditary paraganglioma/pheochromocytoma syndrome
SERPINA1	1	Low risk Alpha-1-antitrypsin deficiency (Z allele heterozygote)
SERPINC1	1	Antithrombin III thrombophilia
TP53 ?mosaic	1	Li-Fraumeni Syndrome/Bone marrow failure/CHIP

GENE	PATIENTS	DISORDER
CFTR	1	Atypical cystic fibrosis/CAVD
DNAH5	1	Primary Ciliary Dyskinesia 3
G6PD	1	Favism (G6PD deficiency)
HFE	3	Hereditary Hemochromatosis type 1

# The Genomic Medicine Team



Debra Leonard, MD, PhD



Niki Sidiropoulos, MD



David Seward, MD, PhD



Robert Wildin, MD



Ken Hampel, PhD



Christine Giummo, MS, CGC



Roberta Francis



Denise Francis



Jordan Armstrong



Margaret Cameron



Denise Bonyun, MS, CGC



# Questions & Discussion



The University of Vermont  
LARNER COLLEGE OF MEDICINE

THE  
University of Vermont  
HEALTH NETWORK



# How to view test value in terms of Actionable Knowledge

KNOWLEDGE	Have Hidden Disease/Risk	<u>Don't</u> have Hidden Disease/Risk
PRE-TEST	0	0
POST-TEST	90-99% (minus any false positives)	50-99% (100% minus false negatives)



# Funding

