Leveraging Connecticut's Health Information Exchange (HIE) to Support Genomic Medicine

May 1, 2019
Version 11
Connecticut Office of Health Strategy (OHS)

Executive Summary

According to the Office of the National Coordinator for Health Information Technology (ONC), Health Information Exchange (HIE) "allows health care professionals and patients to appropriately access and securely share a patient's medical information electronically."[1] Connecticut's statewide HIE will enable stakeholders across the health care system to securely share electronic health information to improve coordination of care, health outcomes, and reduce provider burden among myriad other benefits.

The Connecticut Office of Health Strategy (OHS) hosted this workshop to convene key stakeholders in the state with the overarching goal of discovering paths for leveraging Connecticut's Health Information Exchange (HIE) to support genomic medicine.

To understand how the statewide HIE can be used to support genomic medicine, it is important to understand the definitions of, and differences between, genomics and genetics. Based on the National Human Genome Research Institute's definitions of the two terms, *genomics* studies a person's entire genome—i.e. their complete set of genes— whereas *genetics* studies just one of those genes in particular. [2] *Genomic testing*, therefore, means mapping and studying a person's entire genome to look for variations that might show up anywhere in the code, while *genetic testing* looks for specific genetic variations in single genes or small groups of genes that are linked to certain diseases. [3]

Connecticut is uniquely positioned to become the nation's leader in leveraging the statewide HIE to support genomic medicine. To start, Connecticut is home to a singular concentration of genomic expertise in the form of Yale Center for Genomic Health and its launch of the Generations Project in 2019 that will generate cutting-edge data, Yale University's numerous biobanks, Jackson Laboratories, and Sema4. What's more, Connecticut has taken an approach toward HIE architecture *from inception* that anticipates leveraging the HIE to support genomics. Finally, as noted by presenter Dr. Michael Murray, while Connecticut has only 1% of the nation's population, the demographic layout of Connecticut's population aligns very closely with that of the entire United States, making Connecticut a reasonable model for how successful use of genomic medicine could impact the entire population.[4]

Genomic data that is successfully integrated with information support tools within the statewide HIE, and subsequently into Electronic Health Records (EHRs) in a seamless and actionable form, could enable already overloaded clinical providers to deliver better precision care. It could also facilitate the integration of care and research efforts and, importantly, further improve patient outcomes and reduce overall costs of care.

[1] HealthIT.gov. "What is HIE?" https://www.healthit.gov/topic/health-it-and-health-information-exchange-basics/what-hie

[2] National Human Genome Research Institute. "Genomics." https://www.genome.gov/genetics-glossary/genomics

[3] National Human Genome Research Institute. "Genetic Testing." https://www.genome.gov/genetics-glossary/Genetic-Testing

[4] Michael Murray, "Changing Landscape of Genomic Medicine and Opportunities to Incorporate Genomics in Connecticut" (presentation, Incorporating Genomic Medicine into Connecticut's Health Information Network, Hartford, CT, March 8, 2019).

It's important to note that HIE solutions posited by this workshop focused on leveraging communication *about* genomic medicine rather than focusing on moving or disclosing patients' actual genomic information. For example, one of the workshop's ideas was for the HIE to indicate *when* a specific genetic test would be recommended for patients. The overall goal is to ensure that consumers and providers have access to clinically-actionable genomic information that empowers them with knowledge and resources to make the best health decisions possible.

Workshop participants from diverse stakeholder groups collaboratively conceived of use cases that address ways of leveraging the statewide HIE to support actionable use of genomic medicine (See Key Findings and Ideas from Workshop for all use case ideas). Participants voted the following three use cases as actionable priorities for the state of Connecticut:

1. **Prior Authorization**: Use the HIE to push clinical information in support of prior authorization requests for medically indicated genetic tests. Payers would collaboratively write the prior authorization checklist. It's important to note that there was interest along the entire continuum of care to do this.

2. **Clinical Decision Support (CDS) Engine**: Use a genomic medicine "rules engine," curated by a collective group to ensure up-to-date information, to recommend best practices to providers. For example, the CDS engine could inform the provider when a patient is a candidate for a certain genetic test.

3. **CDS Data Standardization:** Standardize genomic data, much of which is currently in PDF form, so that it's actionable for CDS. Start with CDC Tier One genomic applications. This use case supports the work that would be done by the CDS Engine and would likely need to be developed first. The combined support for these two companion use cases indicates that using the HIE for CDS for genomic medicine is a clear priority.

To advance these ideas through to production in the HIE, OHS will conduct the following next steps. First, OHS has reviewed this white paper with the White Paper Editorial Team. Next, OHS will share the white paper with the HIT Advisory Council. Then, OHS will convene the Genomic Medicine Working Group that will further define practical use case ideas and provide input throughout the development process.

OHS would like to thank Sema4 for their generous sponsorship of the workshop. We also extend our thanks to The Lyceum for providing a truly pleasant meeting experience for workshop participants. Finally, we would like to acknowledge Velatura's help in facilitating this workshop and drafting associated documents.

Table of Contents

Genomic Medicine Workshop

The Connecticut Office of Health Strategy (OHS) hosted the Leveraging Connecticut's Health Information Exchange to Support Genomic Medicine workshop at The Lyceum in Hartford, Connecticut on March 8, 2019. In his introduction, Allan Hackney, the Health Information Technology Officer for Connecticut, aptly called the day "an opportunity to explore something innovative."

The genomic medicine workshop welcomed renowned guest speakers from Yale Center for Genomic Health and Geisinger Health System and brought together key stakeholders in the state with the overarching goal of discovering paths for leveraging the statewide HIE to support genomic medicine.

This workshop began the new process by which Connecticut's statewide HIE will convene stakeholders to conceive of use cases and initiate working groups moving forward. Future workshops will continue to explore topics of critical interest to stakeholders and the statewide HIE.

Workshop Goals

To ensure that the workshop produced actionable ideas in a relatively short period of time, stakeholders united around the following goals:

- Discuss opportunities for Connecticut to advance as a national leader in genomic medicine and information exchange by leveraging innovative architecture and local subject matter expertise

- Outline practical and achievable proposals to leverage the statewide health information exchange to support genomic medicine

- Employ Use Case Factory™ model to identify practical use case opportunities for prototyping genomics in the HIE

- Acknowledge, not solve, industry challenges (e.g. Consent, Security, etc.)

Next Steps

- White paper and white paper review with White Paper Editorial Team

- Share white paper and findings with HIT Advisory Council

- Convene Genomic Medicine Working Group

- Begin work on priority actionable use cases identified by workshop

- Continue to develop use cases to further leverage the statewide HIE to support genomic medicine

- ■ Establish Connecticut as national leader and model for leveraging the statewide HIE to support genomic medicine

Presenters

- ● **David Carey, PhD**, Associate Chief Research Officer at Geisinger Health System and Professor and Chair, Department of Molecular and Functional Genomics, Geisinger
- ● **Michael Murray, MD, FACMG, FACP**, Director for Clinical Operations for Yale School of Medicine and Yale New Haven Hospital's Center for Genomic Health
- ● **Sabina Sitaru, PMP**, Interim Chief Operating Officer for Connecticut's statewide Health Information Exchange

Panelists

- ● **Allan Hackney, CISM, CRISC**, Health Information Technology Officer for Connecticut
- ● **Robert Zavoski, MD, MPH**, Medical Director of Connecticut Department of Social Services
- ● **Thomas Agresta, MD, MBI**, Professor and Director of Medical Informatics Family Medicine, Director of Clinical Informatics Center for Quantitative Medicine, Informatics Leader at Connecticut Institute for Primary Care Innovation (CIPCI)
- ● **Victoria Veltri, JD, LLM**, Executive Director for Connecticut Office of Health Strategy

About the Author

The mission of Connecticut's Office of Health Strategy (OHS) is to implement comprehensive, data driven strategies that promote equal access to high quality health care, control costs and ensure better health for the people of Connecticut. Learn more about OHS at portal.ct.gov/OHS.

Editors

OHS would like to thank the following people for their feedback on the white paper:
- ● **Thomas Agresta, MD, MBI**, Family Physician, Professor and Director of Medical Informatics - UConn Health
- ● **Danielle Bonadies, MS, CGC**, Director of Genetics – My Gene Counsel
- ● **David Carey, PhD**, Associate Chief Research Officer at Geisinger Health System and Professor and Chair, Department of Molecular and Functional Genomics, Geisinger
- ● **Mehul Dalal, MD, MSc, MHS**, Chronic Disease Director – CT Department of Public Health
- ● **Allan Hackney, CISM, CRISC**, Health Information Technology Officer – CT Office of Health Strategy
- ● **Allen Hsiao, MD, FAAP**, Associate Professor of Pediatrics and of Emergency Medicine, Chief Medical Information Officer – Yale School of Medicine & Yale New Haven Health

- **Michael Murray, MD, FACMG, FACP**, Director for Clinical Operations – Yale Center for Genomic Medicine
- **Polly Painter**, Director, Strategic Relations for Precision Medicine and Population Health – Yale School of Medicine
- **Robert Tafuri, MD**, Medical Operations Director – Anthem
- **Amy Taylor,** Vice President, Western Region- Community Health Center
- **Beverly Tenenholz**, Genetic Counselor/Coordinator of Genetic Counseling Program – Hartford Healthcare

Genomic Medicine and Healthcare IT in Connecticut Today

Genomic medicine uses a patient's genomic information to determine their risk for certain diseases and identify possible courses of care. Currently, the CDC recommends genomic applications for three Tier 1 conditions: Hereditary Breast and Ovarian Cancer Syndrome (*BRCA1/2*), Lynch Syndrome (LS, colorectal and endometrial cancer), and Familial Hypercholesterolemia (FH, heart attacks and heart disease).[5] Identification can lead to early detection or prevention, cascade testing for family members who may be at risk, and pharmacogenomic treatment options—identifying which drugs a person may respond best to based on their specific genomic makeup.

The more genomic data that becomes available to researchers, the more they may be able to associate diseases with specific genetic conditions, like the CDC's Tier 1 conditions above. Likewise, more genomic data also advances the field of pharmacogenomics, which can provide information about whether a certain drug will be effective or produce unwanted side effects for a specific patient: this will increase opportunities for precision medicine for patients. And, if genomic information is made accessible and actionable through the statewide health information exchange network, it could reduce the burden on already overloaded providers.

This workshop focused on practical genomics with the goal of coming up with actionable ideas—i.e. projects that are within reach in the next year—for leveraging Connecticut's statewide Health Information Exchange to support genomic medicine research that's being done across the state.

It is important to note that many of the other industry challenges surrounding genomic medicine—consent, security, education needs, ethics—were acknowledged but tabled for future workshops and working groups. While addressing these challenges is critical to furthering genomic medicine, it would not be possible to solve all of them in a one-day workshop.

---

[5] "Tier 1 Genomics Applications and their Importance to Public Health," CDC online, Last modified March 6, 2014, https://www.cdc.gov/genomics/implementation/toolkit/tier1.htm.

> *"Help us decide where we can invest so we can come forward with something we can
> actually implement." – Allan Hackney, HITO for Connecticut*

A Vision for Genomic Medicine and Health Information Exchange in Connecticut

The vision for Connecticut's statewide HIE is to create a "Network of Networks," which would provide a secure mechanism for providers and caregivers to connect, support the bi-directional exchange of health data, leverage existing data-sharing initiatives and services, and establish the ability to participate with national interoperability initiatives. The "Network of Networks" would be able to function effectively, in part, by establishing a trust framework with legal agreements with all Trusted Data Sharing Organizations.

Furthermore, Connecticut is taking a Use Case Factory™ approach to identify high value data-sharing activities and will continue to convene stakeholders to collaboratively conceive of priority use cases as happened at the Genomic Medicine and HIE workshop. The Use Case Factory™ process guides ideas through development and into production in the HIE.

Leveraging the statewide HIE to support genomic medicine could help address multiple stakeholder needs that must be met before genomic information can be actionable for patient care. To start, providers need support to know when genetic testing is recommended and access to readable, actionable results from genetic testing that is streamlined into their workflow. They also need to know when and how to connect patients with genetic counselors.

Patients benefit most when they can go over pre-test possibilities and actionable results with genetic counselors or providers that have apt knowledge and skills paired with seamless access to genetic counseling tools and information. Finally, to streamline prior authorizations, providers need an easy-to-use, EHR-integrated prior authorization process to facilitate bidirectional communication of appropriate risk factor information and approval from payers about whether genetic testing will be covered through insurance. As experience and research accumulate, payers, providers, and patients would also benefit from knowing the long-term benefits and risks associated with genetic testing for early detection and/or prevention as this could both dramatically improve patient outcomes and reduce costs to payers in the long run.

Informative presentations by leading industry researchers prepared workshop participants to identify high-value use cases to address these varied stakeholder needs. Dr. David Carey emphasized that Geisinger Health System's MyCode genomic medicine project, much like Connecticut's statewide HIE, developed a process and infrastructure *from inception* that would enable patients and providers to receive important results in an actionable way and would provide paths for researchers to discover new target drugs. Geisinger has conducted numerous patient focus groups and Dr. Carey emphasized the importance of patient and provider education. From focus groups, Geisinger has found that

patients want to know about results that provide a positive actionable benefit. To learn more about the Geisinger Health MyCode project, please refer to Dr. David Carey's presentation in Appendix C.

Dr. Michael Murray from Yale Center for Genomic Health highlighted several specific opportunities for leveraging the statewide HIE to support genomic medicine. Dr. Murray presented the CDC's three Tier 1 genomic applications: Hereditary Breast and Ovarian Cancer Syndrome (*BRCA1/2* associated cancer), Lynch Syndrome (LS, colorectal and endometrial cancer), and Familial Hypercholesterolemia (FH, heart attacks and heart disease).[6] Because these conditions are both relatively common and well understood, the identification of these genetic risks opens the door to evidence based next steps for the patient and for family members. Currently, there are no statewide initiatives pursuing identification of these risks, so Connecticut has the opportunity to lead the nation in integrating clinical decision support information into the statewide HIE to ensure that all patients who should be having these CDC-recommended genetic tests are actually getting them.

Take ovarian cancer for example: ovarian cancer is the 5th leading cause of cancer death in Connecticut.[7] Since 2007, the evidence-based recommendation by the National Comprehensive Cancer Network is that all women with ovarian cancer be offered testing for *BRCA1/2*; however, one study found that only 10-20% of the women who qualified to have the test actually had it.[8] Targeted quality improvement efforts at two healthcare provider organizations have demonstrated improved adherence to the recommendation to greater than 85%. Connecticut has an opportunity to demonstrate an adherence rate at a state level through collaborations between the statewide HIE, the Connecticut tumor registry, and healthcare provider organizations.

The possibilities for statewide clinical decision support extend beyond genetic testing for certain diseases: Dr. Murray also gave an overview of pharmacogenomics, pointing out that genomic information can clue providers in to which drugs may or may not be effective for a particular patient. Dr. Murray's full presentation is available for reference in Appendix D.

Key Findings and Ideas from Workshop

To generate actionable use case ideas for leveraging the statewide HIE to support genomic medicine, small groups of workshop participants mapped care plans for two personas—synthetic patients with realistic medical histories. One persona had poorly controlled

---

[6] "Tier 1 Genomics Applications and their Importance to Public Health," CDC online, Last modified March 6, 2014, https://www.cdc.gov/genomics/implementation/toolkit/tier1.htm.

[7] "Ovarian Cancer in Connecticut Fact Sheet" Connecticut State Department of Public Health, Last modified August 2016, https://authoring.ct.gov//-/media/Departments-and-Agencies/DPH/Tumor-Registry/pdf/OvarianCancerpdf.pdf.

[8] Erica Bednar, et al. "A universal genetic testing initiative for patients with high-grade, non-mucinous epithelial ovarian cancer and the implications for cancer treatment," *Gynecologic Oncology* 146, no. 2: 399-404, accessed March 27, 2019, http://dx.doi.org/10.1016/j.ygyno.2017.05.037.

diabetes and one was recently diagnosed with ovarian cancer. For each persona, groups mapped two care plans: one in which the patient received genetic testing, and one in which they did not. Groups then used the care plans they generated to look for the points at which information about the genetic testing should be shared by asking, "When should the information be shared?" and "By or with whom?". (Persona Case Studies can be found in Appendix E.)

These points of possible information exchange along with key questions that arose during the exercise (such as: How does a provider look for results if the results aren't in their own EHR? Who has access to results? Is there a way for providers to know if a patient is at higher risk of other cancers or a candidate for clinical trial? Who pays for the test? When is it covered and when not?), lead groups to identifying actionable use cases.

It's important to note that the use case ideas emphasize leveraging *when* genomic approaches would be beneficial and sharing information *about* genetic testing rather than actually moving or disclosing a patient's genomic information. The goal of these use cases, and of leveraging Connecticut's statewide HIE to support genomic medicine in general, is to ensure that genomic information is clinically advantageous to consumers and providers.

Workshop participants came up with thirteen use case ideas that would leverage Connecticut's HIE to support genomic medicine. Participants were given stickers with varying dollar amounts on them and asked to vote for the three use cases they saw as priorities and as feasible to begin planning and implementing within the next year. The chart below shows the voting results.
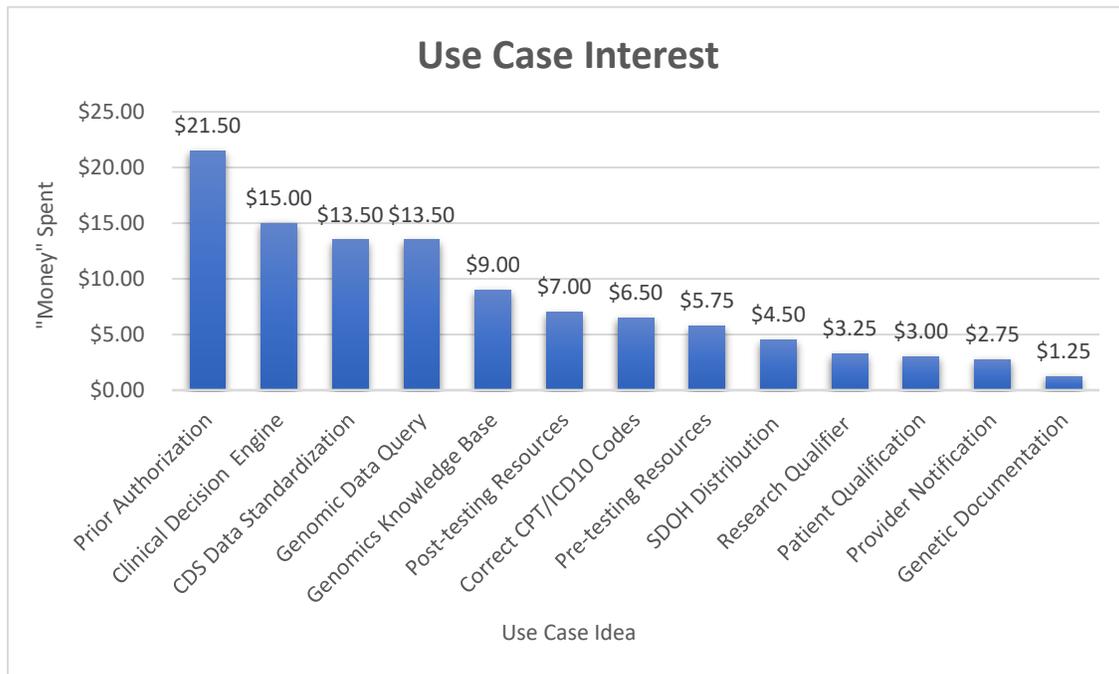
Prior Authorization

The Prior Authorization use case received the most support, coming in with 27% more "dollar" support than the use case that follows it, which was the largest margin between any of the use cases. This high level of support from an incredibly diverse group of stakeholders in the room indicates that there is interest along the entire continuum of care to use the HIE in support of prior authorization for genetic testing.

In this use case, the HIE would be used to automate the gathering of clinical information for prior authorization requests for medically indicated genetic tests. In other words, the HIE would give the payer clinical information on the patient that supports the medical recommendation for performing the genetic test. Participants voiced the supplementary idea that the HIE could encourage payers to collaboratively write the prior authorization checklist.

Support for this use case also hinged around the fact that, while the cost of genetic testing has dropped dramatically in recent years, some tests remain expensive. It would be beneficial for patients to be able to know in real time from their providers whether a test would be covered and, if not, how much it would cost.

OHS was surprised that the prior authorization use case came so prominently to the forefront at the workshop. Because use case ideas hadn't been socialized previously within the payer community, OHS didn't yet have a sense of their point of view. It was exciting to see such enthusiastic support for this idea not only from payers but also from the entire continuum of care.

Clinical Decision Support (CDS) Engine

The Clinical Decision Support Engine received the second highest dollar amount of votes. The CDS Engine would be a "rules engine" or comprehensive database of "rules" that results in various actions. Namely, the engine would recommend best practices to providers. For example, following the National Comprehensive Cancer Network's recommendation that all women with ovarian cancer get the *BRCA1/2* test, the CDS Engine could see that a patient had been recently diagnosed with ovarian cancer and automatically inform the patient's provider that she is a candidate for the *BRCA1/2* test.[9]

Additionally, a CDS rules engine could be triggered for a prescriber when there is a genetic test indicating that the medication being selected is poorly metabolized in a given patient. In the case of opioids for example, this could prevent a potential overdose situation, while in a patient with depression it might indicate the potential benefit of choosing an alternate therapy.

---

[9] Erica Bednar, et al. "A universal genetic testing initiative for patients with high-grade, non-mucinous epithelial ovarian cancer and the implications for cancer treatment," *Gynecologic Oncology* 146, no. 2: 399-404, accessed March 27, 2019, http://dx.doi.org/10.1016/j.ygyno.2017.05.037.

There was a lot of conversation throughout the day that centered around reducing provider burden. This use case would streamline best practices for genomic medicine into the workflow of providers in an easily accessible, actionable way, thereby decreasing provider overload, increasing adherence to recommendations for genetic testing, and ultimately positively impacting patient outcomes and possibly reducing costs for payers in the long term.

CDS Data Standardization

Coming in third, the Clinical Decision Support Data Standardization use case is a companion use case to the CDS Engine. In fact, while the engine had more support, this use case is likely a prerequisite to a working CDS Engine. In this use case, the HIE would be used to standardize genomic information data so that it can be actionable for Clinical Decision Support. Workshop participants pointed out that a lot of genomic data currently exists in non-standard PDF form. A starting point would be the three CDC Tier 1 genomic applications.

This use case addresses one of the main challenges with genomic information which is that there isn't a standardized way of storing or transmitting data. This can make data difficult to interpret or even to access in the first place for already overloaded providers. Non-standardized data also makes any centralized form of CDS nearly impossible.

Genomic Data Query

Coming in tied with CDS Data Standardization, the Genomic Data Query use case would use the HIE to push the location of genomic information into the provider's EHR so that the provider knows where to look for a patient's genomic information. As its name suggests, this use case would also allow providers to query for the location of patient genomic information results if the information isn't pushed in to the EHR or if the provider doesn't have an EHR.

This use case would help address the fact that genomic information is stored in myriad ways in myriad locations. If a patient thinks they've gotten a test, but there's no record of it in their chart, this use case could enable the provider to search for a result rather than go on a time-consuming hunt. This would of course require the "testing organization" to share information with the HIE infrastructure. Additionally, consent models may limit the sharing of past information on testing that happened before the implementation of the use case.

Genomics Knowledge Base

The Genomics Knowledge Base use case is one of the more large-scale use case ideas to come from the workshop. The idea is to build a knowledge base that serves as a sort of genomic data dictionary that could be used within the HIE to support many of the other use cases in this list, in particular CDS, post- and pre-testing resources, and prior authorization. Potentially, Connecticut's genomic experts could even work with genomic experts in other states to collectively maintain the database and ensure that it's up-to-date.

This use case could reduce the burden on providers by powering CDS with current research that's curated by experts in the field. It could also empower clinicians, who don't traditionally have training in genomics, to begin incorporating this data into the care of their patient populations. The Genomics Knowledge Base would also help position Connecticut as the national leader in the genomic medicine and HIE space.

Post-Testing Resources

The Post-Testing Resources use case would give providers resources and decision support after a patient's genetic test results have come in. These resources would include next steps, e.g. what to do with the results, and education resources for the provider and patient. Post-testing resources could also include genetic counselor suggestions. The Post-Testing Resource use case would ensure that genetic and genomic testing results are actionable for patient care. It is also important for health plans to know if their members have received proper genetic counseling by the ordering physicians to prepare them for their test results. It would also allow for making recommendations for further testing, particularly for family members who may be at risk.

Correct CPT and ICD10 Codes

Correct CPT and ICD10 Codes use case, as its name suggests, would use the HIE to make sure that correct codes get used for genetic tests. Accurate coding would support prior authorization, collection and transport of data, the CDS Engine, and discovery of genetic tests being done.

Pre-Testing Resources

The Pre-Testing Resources use case is a companion to the Post-Testing Resources use case. After a patient has been flagged as a candidate for a specific genetic test, the Pre-Testing Resources use case would share resources with the provider about what the next steps are for ordering the test: e.g. how to get testing done, to whom to refer the patient, who should order the test, etc.

In tandem with the Post-Test Resources use case, this use case makes sure that providers have the support they need to make genomic information clinically actionable. It would reduce provider burden by providing action steps rather than just flagging that a patient is a candidate for a certain test.

SDOH Distribution

In this use case, the HIE would be used to collect and distribute Social Determinants of Health to policy makers and the patient's care team. ICD10 codes have Z codes for SDOH, and Medicaid is encouraging all providers to use these codes. Tracking SDOH could have a tie-in to epigenetics, "an emerging field of science that studies heritable changes caused by the activation and deactivation of genes without any change in the underlying DNA sequence of the organism."[10] However, the primary focus of workshop participants seemed

---

[10] National Human Genome Research Institute. "Epigenetics." https://www.genome.gov/genetics-glossary/Epigenetics

to be on the fact that SDOH significantly impact a person's health outcomes. Even though there wasn't a direct link to genomic medicine, there was a lot of energy and urgency around using the HIE to facilitate the tracking and dissemination of SDOH. The level of support and concern could indicate this as a future topic for another OHS workshop and subsequent working group.

Research Qualifier

The Research Qualifier use case would leverage the HIE to connect patients with clinical trials for which they qualify. Patients would have the option to opt-in to being informed about clinical research for which they qualify. Opting-in would flag their record, prompting a notification to be sent to the research organization. This could advance clinical research on genomic interventions and wouldn't necessarily have to be limited to genomic research trials. This use case could also improve patient outcomes by helping to test and develop new and effective treatment options.

Patient Qualification

The Patient Qualification use case would send a message to a patient's care team as part of the care summary message when a patient has had a positive result for certain diagnosis codes (like ovarian cancer). The message would say that the patient is a candidate for X genetic test and that it's best clinical practice to get the test. This use case, especially when combined with the Pre- and Post-Testing Resources use cases, would dramatically reduce provider burden by assuring access to current clinical recommendations at the point of care.

Provider Notification

The Provider Notification use case would simply add a flag to already-transmitted ADT notifications alerting providers that a patient's genome has been mapped or that a patient has a genetic test result and subsequently letting providers know where results can be found. This could be used for genome mapping, but it could also be used for any sort of genetic testing to reduce duplication.

Genetic Documentation

The Genetic Documentation use case would provide documentation to the payer from the medical record that the patient received genetic counseling. Currently, cases are getting denied because there is no information in the EHR that a patient has received genetic counseling, and receiving genetic counseling is often a prerequisite for the coverage of genetic testing.

Conclusion

> *"I'm ready to go. I'm looking for recommendations that are practical. It's got to be what can we do now." – Allan Hackney, HITO for Connecticut*

Connecticut is uniquely positioned to become the national leader in leveraging a statewide HIE to support genomic medicine. This cutting-edge work will not only impact patients, providers, and payers in Connecticut, it will also serve as a reasonable national model since the population distribution of the state is a close match to that of the country. By linking innovative medicine with a fully interoperable health information exchange, Connecticut has the opportunity to further genomic research, reduce provider burden, reduce overall costs of care, and most importantly improve patient outcomes.

The results of the Leveraging Connecticut's Health Information Exchange to Support Genomic Medicine workshop will be shared with Connecticut's Health IT Advisory Council. Next, the Genomic Medicine Working Group will convene to begin reviewing the actionable use case ideas and provide input throughout the development process. The goal is to have priority genomic medicine use cases in production in the HIE within a year.

OHS would like to thank the seven people from the workshop who volunteered to be part of the ongoing Genomic Medicine Working Group, and we would like to invite others who are interested in joining this pioneering working group, or future working groups that will be convened by OHS, to contact Allan Hackney at Allan.Hackney@ct.gov.

Editorial Team Recommendations for the Genomic Medicine Working Group

General Considerations

- Collaborate with some of the other genomic testing being done in Connecticut including UConn Health Center Personalized Medicine initiatives and Community Health Center Inc's participation in the NIH-funded ALL of Us project (https://allofus.nih.gov/), which is gathering patient samples tied to clinical data for whole genome-wide association research studies. Community Health Center Inc is one of the only Federally Qualified Health Centers (https://www.joinallofus.org/en/chc) involved in this work nationwide.
- Consider the health equity, access, and SDOH dimensions of prioritized use-cases by examining the following questions:
  - What strategies are in place to engage populations that typically do not access healthcare? Are there strategies to address barriers to access (e.g. trust, financial) and barriers to follow-through (i.e. lack of transport to genetic counseling appointment)?

♦ Are there design elements that need to be considered up front to minimize risk of bias? (i.e. will the clinical information required for prior authorizations be uniformly available by race and ethnicity, if not, what is the risk that prior authorization approvals may systematically bias one population over another?)

♦ What strategies are in place to ensure that healthcare providers working with traditionally vulnerable populations (low income, Medicaid etc.) have access to, are aware of, and can implement the proposed use cases?

● Consider discussing and differentiating between personalized medicine and precision medicine to make sure stakeholders understand the differences and how they apply to the statewide HIE's work to support genomic medicine.

● Consider working with Editorial Team members' student / intern groups to discuss potential participation in literature review to support Working Group.

CDC Tier One Genomic Applications

● Conduct literature review of United States Preventative Services Task Force (USPTF) recommendations for CDC Tier 1 genomic applications and associated screenings as primary care providers are much more likely to be influenced by USPTF. Sources could be addressed both within the body of the text and included in the bibliography.

● Consider conducting research on the estimated number or percentage of individuals affected by the three CDC Tier 1 genomic applications as well as race/ethnicity breakdowns.

Further Considerations for Use Cases

● Prior Authorization
 ♦ When prioritizing use cases in the working group, consider that prior authorization could benefit from the CDS Engine and CDS Data Standardization as a backbone since the standardization of the request and a CDS evaluation of when to order a test would be helpful.

● CDS Engine
 ♦ Consider using CDS Hooks, which may be a good method to call the CDS Engine via web-service.
 ♦ Consider using FHIR® to transmit data in a standardized format.
 ♦ Discuss how such rules would be integrated with various EHRs for point of care decision-making and/or incorporated into provider population health management strategies.

● CDS Data Standardization
 ♦ Explore further uses of this beyond the initial use case for CDS that would enable not-yet-envisioned CDS or additional use cases for research etc.

● Genomics Knowledge Base
 ♦ Consider reaching out to research teams conducting similar work at Vanderbilt, Northwestern, and NIH.

● Pre- and Post-Testing Use Cases

◆ Clarify when to offer genetic counseling: pre-test?, post-test?, pre- and post-test?, counseling only for people who get a positive result? Consider payment for counseling and the patient's decision based on personal preferences for counseling.

Bibliography

Bednar, Erica, Holly Oakley, Charlotte Sun, Catherine Burke, Mark Munsell, Shannon
Westin, and Karen Lu. "A universal genetic testing initiative for patients with high-
grade, non-mucinous epithelial ovarian cancer and the implications for cancer
treatment." *Gynecologic oncology* 146, no. 2 (2017): 399-404.
http://dx.doi.org/10.1016/j.ygyno.2017.05.037.

Beitsch, Peter, Pat Whitworth, Kevin Hughes, Rakesh Patel, Barry Rosen, Gia Compagnoni,
Paul Baron, *et al.* "Underdiagnosis of Hereditary Breast Cancer: Are Genetic Testing
Guidelines a Tool or an Obstacle?" *Journal of Clinical Oncology* 37, no. 6 (2018): 453-
460. https://doi.org./10.1200/JCO.18.01631.

Caraballo, PJ, SJ Bielinski, JL St. Sauver, and RM Weinshilboum. "Electronic Medical Record-
Integrated Pharmacogenomics and Related Clinical Decision Support Concepts."
Clinical Pharmacology and Therapeutics 102, no. 2 (August 2017): 254-264.
https://doi.org/10.1002/cpt.707.

Carey, David. "Leveraging the resources of an integrated health system for precision
health." Presentation at the Incorporating Genomic Medicine into Connecticut's
Health Information Network Workshop, Hartford, CT, March 2019.

Dolin, RH, A. Boxwala, and J. Shalaby. "A Pharmacogenomics Clinical Decision Support
Service Based on FHIR and CDS Hooks." *Methods of Information in Medicine* 57, no.
02 (2018): e115-e123. https://doi.org/10.1055/s-0038-1676466.

"Epigenetics." National Human Genome Research Institute.
https://www.genome.gov/genetics-glossary/Epigenetics

"Genetic Testing." National Human Genome Research Institute.
https://www.genome.gov/genetics-glossary/Genetic-Testing

"Genomics." National Human Genome Research Institute.
https://www.genome.gov/genetics-glossary/genomics

Herr, Timothy, Josh Peterson, Luke Rasmussen, Pedro Caraballo, Peggy Peissig, and Justin
Starren. "Pharmacogenomic clinical decision support design and multi-site process
outcomes analysis in the eMERGE Network." *Journal of the American Medical
Informatics Association* 26, no. 2 (February 2019): 143-148.
https://doi.org/10.1093/jamia/ocy156.

Luzum, Jasmine, and Jason Cheung. "Does cardiology hold pharmacogenetics to an

inconsistent standard? A comparison of evidence among recommendations." *Pharmacogenomics* 19, no. 15 (September 2018): 1203-1216. https://doi.org/10.2217/pgs-2018-0097.

Murray, Michael. "Changing Landscape of Genomic Medicine and Opportunities to Incorporate Genomics in Connecticut." Presentation at the Incorporating Genomic Medicine into Connecticut's Health Information Network Workshop, Hartford, CT, March 2019.

"Ovarian Cancer in Connecticut Fact Sheet." Connecticut State Department of Public Health online. Last modified August 2016. https://authoring.ct.gov//-/media/Departments-and-Agencies/DPH/Tumor-Registry/pdf/OvarianCancerpdf.pdf.

Sturm, Amy, Joshua Knowles, Samuel Gidding, Zahid Ahmad, Catherine Ahmed, Christie Ballantyne, Seth Baum, *et al*. "Clinical Genetic Testing for Hypercholesterolemia: JACC Scientific Expert Panel." *Journal of the American College of Cardiology* 72, no. 6 (August 2018): 662-680. https://doi.org/10.1016/j.jacc.2018.05.044.

"Tier 1 Genomics Applications and their Importance to Public Health." CDC online. Last modified March 6, 2014. https://www.cdc.gov/genomics/implementation/toolkit/tier1.htm.

Trivedi, Bijal. "Medicine's future?" *Science* 358, no. 6362 (October 2017): 436-440. https://doi.org/10.1126/science.358.6362.436.

"What is HIE?" HealthIT.gov. https://www.healthit.gov/topic/health-it-and-health-information-exchange-basics/what-hie

Williams, Marc, Melissa Kern, Virginia Lerch, Johnathan Billet, Janet Williams, and Gregory Moore. "Implementation of a patient-facing genomic test report in the electronic health record using a web-application interface." *BMC Informatics and Decision Making* 18, no. 32 (2018). https://doi.org/10.1186/s12911-018-0614-x.

Appendix A: Workshop Agenda

- 9:30 AM   Welcome & Introduction
- 10:00 AM   Benefits and Opportunities of Sharing Genomic Data
  - Michael Murray, MD
  - David Carey, PhD
- 11:00 AM   Questions for the Panel – Tabletop Exercise
- 11:30 AM   Lunch Break
- 12:00 PM   Panel Discussion
  - Allan Hackney, MD
  - David Carey, PhD
  - Michael Murray, MD
  - Robert Zavoski, MD
  - Thomas Agresta, MD
  - Victoria Veltri, JD
- 12:45 PM   Connecticut Health Information Exchange Overview
- 1:00 PM   Case Studies – Tabletop Exercise
- 1:45 PM   Short Break
- 2:00 PM   Identifying New Opportunities – Tabletop Exercise
- 3:00 PM   Summary and Next Steps
- 3:30 PM   Adjourn

Meet Our Panel

Allan Hackney, CISM, CRISC
- Health Information Technology Officer for Connecticut
- https://commonimpact.org/about/bio/hackney

David Carey, PhD
- Associate Chief Research Officer at Geisinger Health System and Director of Weis Center for Research
- https://www.geisinger.edu/research/research-and-innovation/find-an-investigator/2018/04/04/13/27/david-carey

Michael Murray, MD, FACMG, FACP
- Director for Clinical Operations for Yale School of Medicine and Yale New Haven Hospital's Center for Genomic Health
- https://medicine.yale.edu/genetics/people/michael_murray.profile

Robert Zavoski, MD, MPH
- Medical Director of Connecticut Department of Social Services
- http://www.ct.gov/oha/lib/oha/equitycommission/documents/bios/robzbio.pdf

velatura

Thomas Agresta, MD, MBI
- Professor and Director of Medical Informatics Family Medicine, Director of Clinical Informatics Center for Quantitative Medicine, Informatics Leader at Connecticut Institute for Primary Care Innovation (CIPCI)
- http://facultydirectory.uchc.edu/profile?profileId=Agresta-Thomas

Victoria Veltri, JD, LLM
- Executive Director for Office of Health Strategy
- https://portal.ct.gov/OHS/About/Director

Appendix B: Complete Slide Deck

(Double click on image to open complete slide deck as a PDF)

Appendix C: David Carey, PhD, "Leveraging the resources of an integrated health system for precision health"

(Double click on image below to open complete presentation as a PDF)

Leveraging the resources of an integrated health system for precision health

David J. Carey, PhD
Geisinger Health System

Appendix D: Michael Murray, MD, "Changing Landscape of Genomic Medicine and Opportunities to Incorporate Genomics in Connecticut"

(Double click on image below to open complete presentation as a PDF)

OHS CONNECTICUT Office of Health Strategy

Appendix E: Case Studies

Case Study #1: Alex Gonzales

Scenario:
- 67 year old male
- Smoker with poorly controlled diabetes. He notes that skips his diabetes medication half the time.
- He presented to the emergency room with a cold and blue left foot.
- An urgent balloon and stent procedure re-established circulation in his foot.
- On discharge, his clinical team provides prescriptions including Prasugrel, however, when Alex's wife asked about cost of medicine she found out that out-of-pocket cost will exceed $100/month, she says they can't afford it

Alex's wife explains that he received a test in New Haven last year to help decide what medicines will work and asked his provider if he has that result.

Questions:
1 – What happens next in the path of care with genetic testing / without genetic testing?
2 – What information goes to patient, doctor, hospital, and health plan and when?

OHS CONNECTICUT Office of Health Strategy
Yale SCHOOL OF MEDICINE

Case Study #2: Joyce Smith

Scenario:
- 57 year old female
- Recent diagnosis of ovarian cancer
- Pathology report finds that Joyce has Epithelial Ovarian Cancer
- No family history of cancer, specifically no history of breast or ovarian cancer

When reviewing diagnosis with her provider, the gynecologic surgeon recommends that she undergo a genetic test for *BRCA1/2* and other syndromic cancers.

Questions:
1 – What happens next in the path of care with genetic testing / without genetic testing?
2 – What information goes to patient, doctor, hospital, and health plan and when?

OHS CONNECTICUT Office of Health Strategy
Yale SCHOOL OF MEDICINE

velatura

Appendix F: Attendee List

Organization	Last Name	First Name
Anthem	Tafuri	Robert, MD
CHC, Inc.	Schmitt	Natalie
CHC, Inc.	Taylor	Amy
Consultant	Chatterjee	Supriyo, SB
Consumer Advisory Board	Manohar	Velandy, MD
DPH	Aye	Diane
DPH - Tumor Registry	Gonsalves	Lou
DPH - Genomic Office	Lillpopp	Jonathon
DSS	Zavoski	Robert, MD
Geisinger Health System	Carey	David
Hartford Healthcare	Tenenholz	Beverly
HIE Entity	Johnson	Lauri
HIE Entity	Mills	Brittney
HIE Entity	Schnyder	John
HIE Entity	Sitaru	Sabina
Institute of Living at Hartford Hospital	Ruano	Gualberto, MD
My Gene Counsel	Bonadies	Danielle
OSC	Czunas	Sandra
State of CT / OHS	Gerratana	Terry
OHS	Hackney	Allan
OHS	Schaefer	Mark
OHS	Veltri	Vicki
Sema4	Keeling	Jonathon
Sema4	Xue	Ming
UConn AIMS	Fontes	Alan
UConn Health	Agresta	Tom, MD
UConn Health	Hayden	Kate
Velatura	Carpenter	Brianne
Velatura	Kaye	Jill
Velatura	Kosowski	Lauren
Velatura	Ort	Summer
Velatura	Wilkening	Rick
Yale Center for Genomic Health	Murray	Michael, MD
Yale Center for Genomic Health	Painter	Polly
Yale Center for Research Computing	Peskin	Eric
Yale School of Medicine	Hsiao	Allen, MD

Appendix G: Pictures