Developing HIT Tools & Infrastructure To Implement Genomic Medicine in CT

Thomas Agresta, MD, MBI
Professor of Family Medicine — UConn Health
CT Forum on DNA Testing and Genomic Medicine

January 23, 2020

Agresta@uchc.edu

UCONN
HEALTH

# Genomic Medicine In The News Case Study

- Kathy Mathes got a genetic test indicating an 84% risk for ovarian cancer by age 70
- Kathy underwent preventative surgery as a result of the high risk
- Her risk changed to a variant of uncertain significance 4 years later
- Surgery was not a necessity

A Genetic Test Led Seven Women in One Family to Have Major Surgery. Then the Odds Changed.

Two sisters, their mother and aunts showed a mutation on a BRCA gene and an elevated risk of breast and ovarian cancer

Marcus, A. (2020). A Genetic Test Led Seven Women in One Family to Have Major Surgery. Then the Odds Changed. *The Wall Street Journal*. [online] Available at: https://www.wsj.com/articles/seven-women-in-a-family-chose-surgery-after-a-genetic-test-then-the-results-changed-11576860210 [Accessed 15 Jan. 2020].

UCONN HEALTH

Lessons Learned

- Lack of integration at point of care
  - Genomic results are not effectively sent and stored in EHRs
  - Ordering Genomic tests not standardized
- Various labs report data differently
  - Commercial, Hospital system, research
  - Different levels – whole genome, variants
  - Formats – PDF, XML, metadata
  - Standards – HL7, FHIR, JSON
- Low level of understanding and interpretation at point of care
  - Clinical meaning of the results changes
  - Limited Clinical Decision Support
  - No standardized way to present information in EHRs
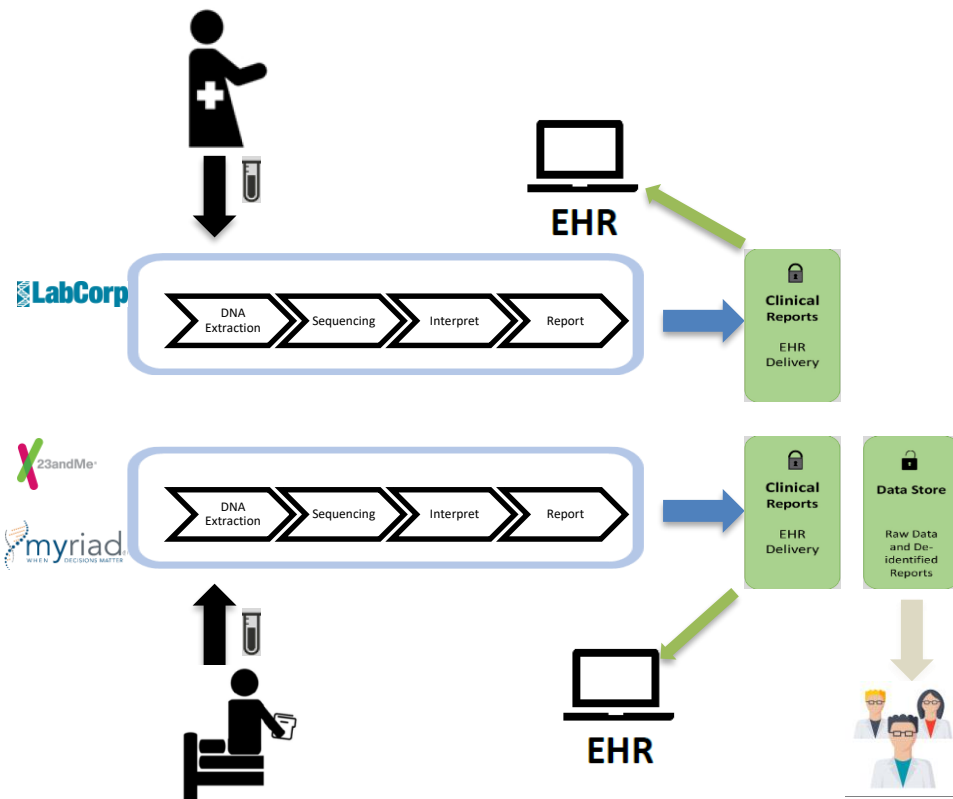
Workgroup for Electronic Data Interchange (WEDI) (2015). *Issues and Trends in Electronic Genomic Data Exchange*. Reston, VA.

Potential HIE Use Genomic Medicine in CT

Leveraging Connecticut's Health Information to Support
Genomic Medicine Workshop on March 8,2019

- Key Issues/Use Cases Addressed:
  - Clinical Decision Support Data Standardization
  - Genomics Knowledge Base
  - Clinical Decision Support Engine
  - Prior Authorization
  - Post-testing Resources
  - Pre-Testing Resources

UCONN
HEALTH

Prior Authorization Use Case Example

Prior Authorization Tool

- Information from the EHR, the tool determines risk for Genomic Condition based on guidelines and screening tests

- Queries the HIE for additional information

- Sends prior authorization request to the patient's insurance company

- Orders Genomic Test
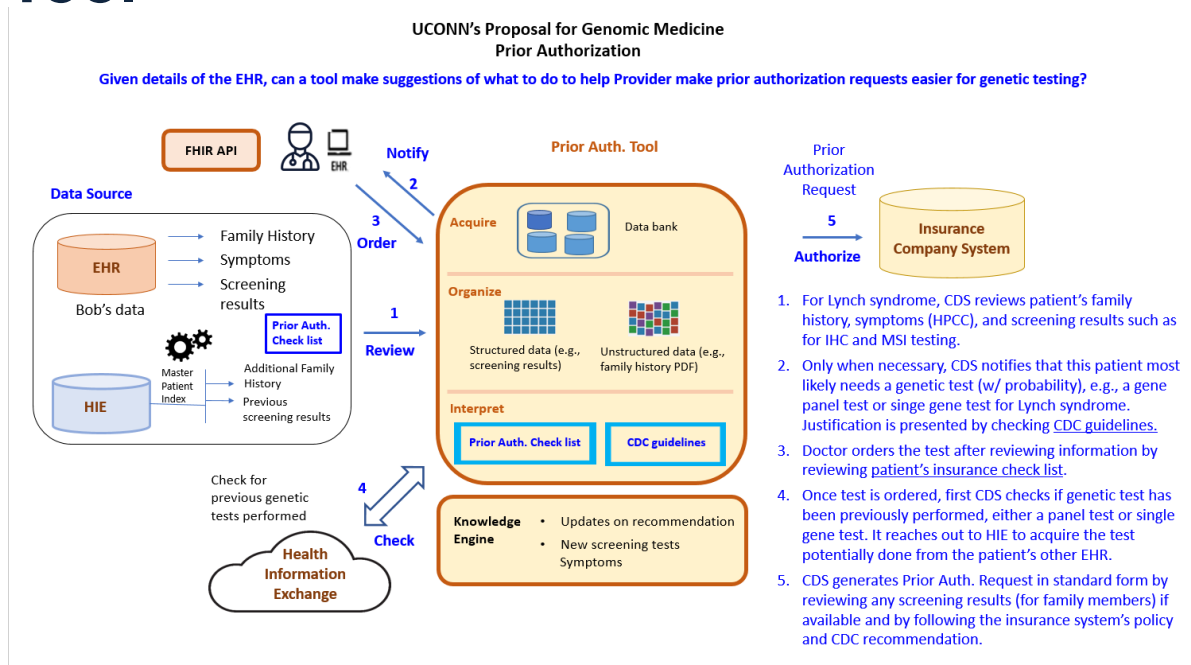
Genomic Medicine and HIT
Lessons from Medication Reconciliation

Medication Reconciliation

- Goals:

  - Improve methods of communication between providers and pharmacists

  - Improve physician, pharmacists and patients' knowledge regarding their medications

  - Improve data sharing, standardization and presentation for patient medication data

Genomic medicine

- Goals:

  - Improve methods of communication between patient's and specialists

  - Improve patient, physician and genetic counselor's knowledge regarding genetic results

  - Improve data sharing, standardization and presentation for genetic data

UCONN
HEALTH

Med Rec Polypharmacy Workgroup

- Formed legislatively in 2018
  - Under HITO / HIT Advisory Council
  - Multi-Stakeholder group with over 50 participants
- Met Sept 2018 to June 2019
  - With Facilitation / Support from the HITECH HIE funding
  - UConn Health help a "Med Wreck Hackathon" Spring 2019
- Produced a report to Legislature June 2019
  - 11 Core Recommendations
- Now Med Rec Polypharmacy Committee (MRPC) (Sept 2019- 2021)
  - Under HIT Advisory Council – with formal charter and support from HITECH funds
  - Working on Policy, Funding, Education & Outreach, Pilot projects, Prototype development

UCONN
HEALTH

Consider Genomic Medicine HIT Workgroup

- **Model on the Med Rec Process**
  - Multi-stakeholder
  - Assigned to HIT Advisory Council for support / reporting
  - Ask for reports / recommendations back to legislature
  - Explore in more depth how HIE can support Genomic Medicine
  - Consider Sponsoring Grants, Competitions, Hackathons to illicit best ideas and foster collaborations amongst stakeholders
  - Explore how to build / fund underlying HIT / HIE infrastructure required for sustained success

UCONN
HEALTH