



FHIR-BASED INTEROPERABILITY TO ACCELERATE GENOMIC MEDICINE RESEARCH AND PRACTICE: OPPORTUNITIES AND CHALLENGES

**DEVELOPING A CLINICAL GENOMICS INFORMATICS RESEARCH AGENDA
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DISCLOSURES

- I report honoraria, consulting, sponsored research, licensing, or co-development in the past year with Hitachi, Pfizer, RTI International, the University of California at San Francisco, MD Aware, and the U.S. Office of the National Coordinator for Health IT (via Security Risk Solutions)
- I was also an unpaid board member of the non-profit Health Level Seven International (HL7) health IT standard development organization that developed the FHIR standard; I am an unpaid member of the U.S. Health Information Technology Advisory Committee; and I have helped develop a number of health IT tools which may be commercialized to enable wider impact

WHY INTEROPERABILITY FOR GENOMIC MEDICINE?

- Discovery
 - Normalized data sets for analysis and discovery
- Clinical care
 - Patient-level care optimization
 - Population-level care optimization

HL7 FAST HEALTHCARE INTEROPERABILITY RESOURCES (FHIR)¹

- Standard for exchanging healthcare information
- Gaining rapid industry adoption, endorsed by NIH²
- Several versions (e.g., DSTU2, STU3, R4)
- Several implementation guides (IGs)
- Support for US Core IG³ now required by regulation (US Core Data for Interoperability, USCDI)

1. <https://www.hl7.org/fhir>

2. <https://grants.nih.gov/grants/guide/notice-files/NOT-OD-19-122.html>

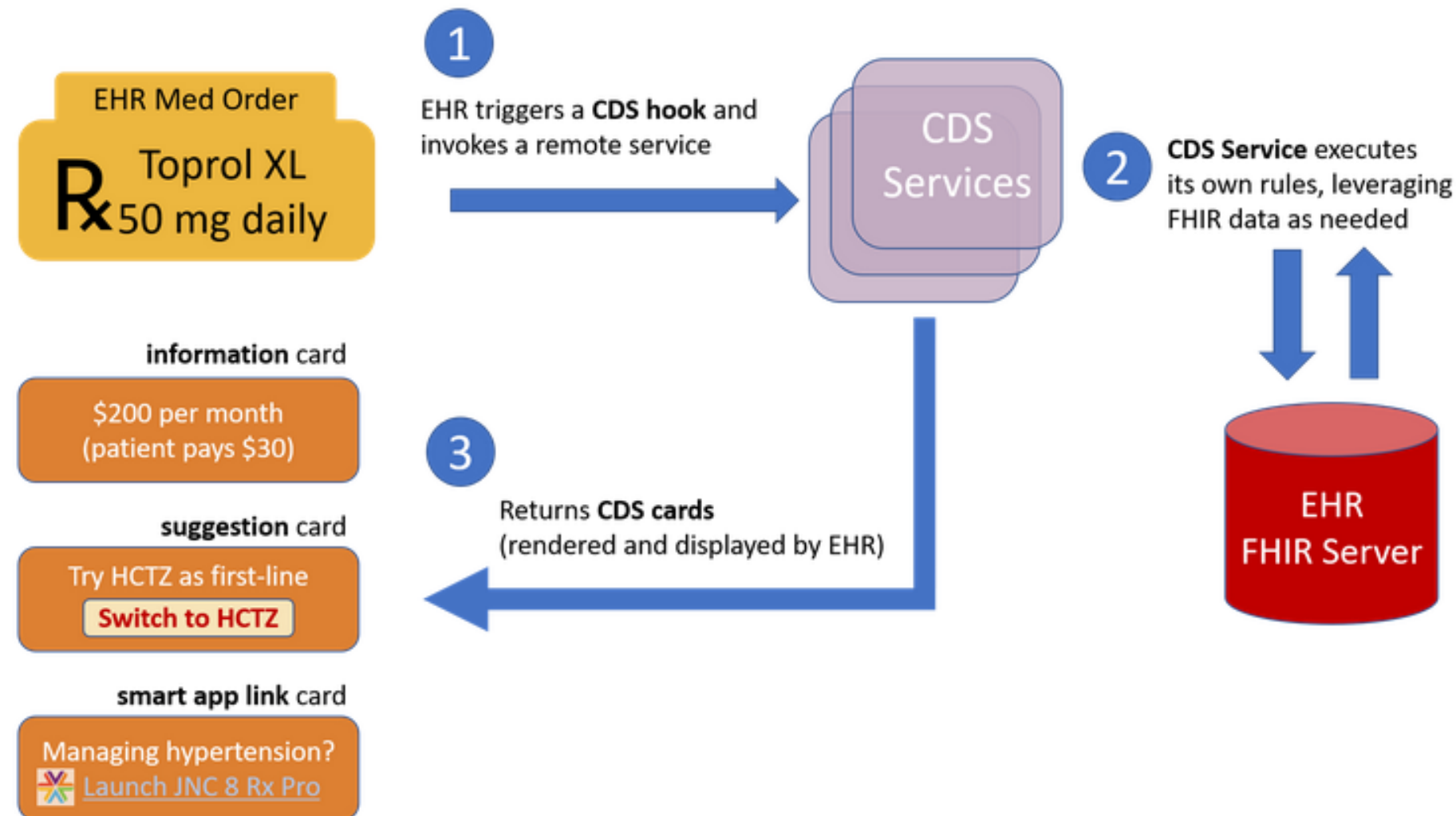
3. <https://www.hl7.org/fhir/us/core/>

SMART ON FHIR

- Allows embedding third-party apps in the EHR
- Widespread EHR vendor support
- Supports patient-facing apps (e.g., Apple Health)
- Uses OAuth 2 to delegate users' data rights to app
- Uses FHIR to read, and sometimes write, data
- May use Clinical Quality Language (CQL) and/or CDS Hooks for logic evaluation
- In some cases, may augment EHR FHIR server to add needed data or to filter out unnecessary data

CDS HOOKS

- HL7 standard since 2019; increasing use
- Companion standard to SMART on FHIR



<https://cds-hooks.hl7.org/>

CLINICAL QUALITY LANGUAGE (CQL)

- HL7 standard since 2015; widespread use
- Enables expression of computable phenotypes, decision support logic, and clinical quality measures

```
36  define "Inclusion Criteria":
37      AgeInYears() >= 18
38      and (
39          exists (Common."Active Ambulatory Benzodiazepine Rx")
40          and exists (Common."Active Ambulatory Opioid Rx")
41      )
42
43  define "Get Indicator":
44      if "Inclusion Criteria"
45          then 'warning'
46      else null
47
48  define "Get Summary":
49      if "Inclusion Criteria"
50          then 'Patient has active prescriptions for opioid pain medication and benzodiazepines'
51      else null
52
53  define "Get Detail":
54      if "Inclusion Criteria"
55          then 'Avoid prescribing opioid pain medication and benzodiazepines concurrently whenever possible'
56      else null
```

<https://cql.hl7.org/>

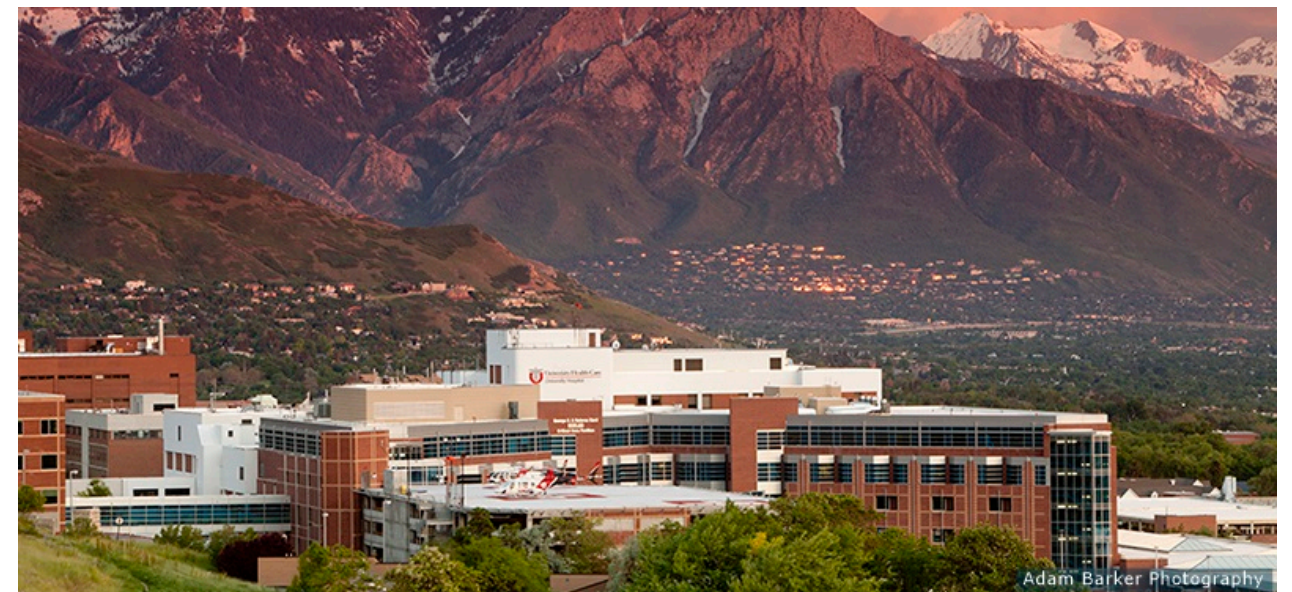
FHIR BULK DATA ACCESS (FLAT FHIR)¹

- Per-patient FHIR access approaches are too slow for population-level data retrieval
- FHIR Bulk Data Access could address this issue
- Researchers could leverage normalize large data sets retrieved through Flat FHIR
- This approach is still in development

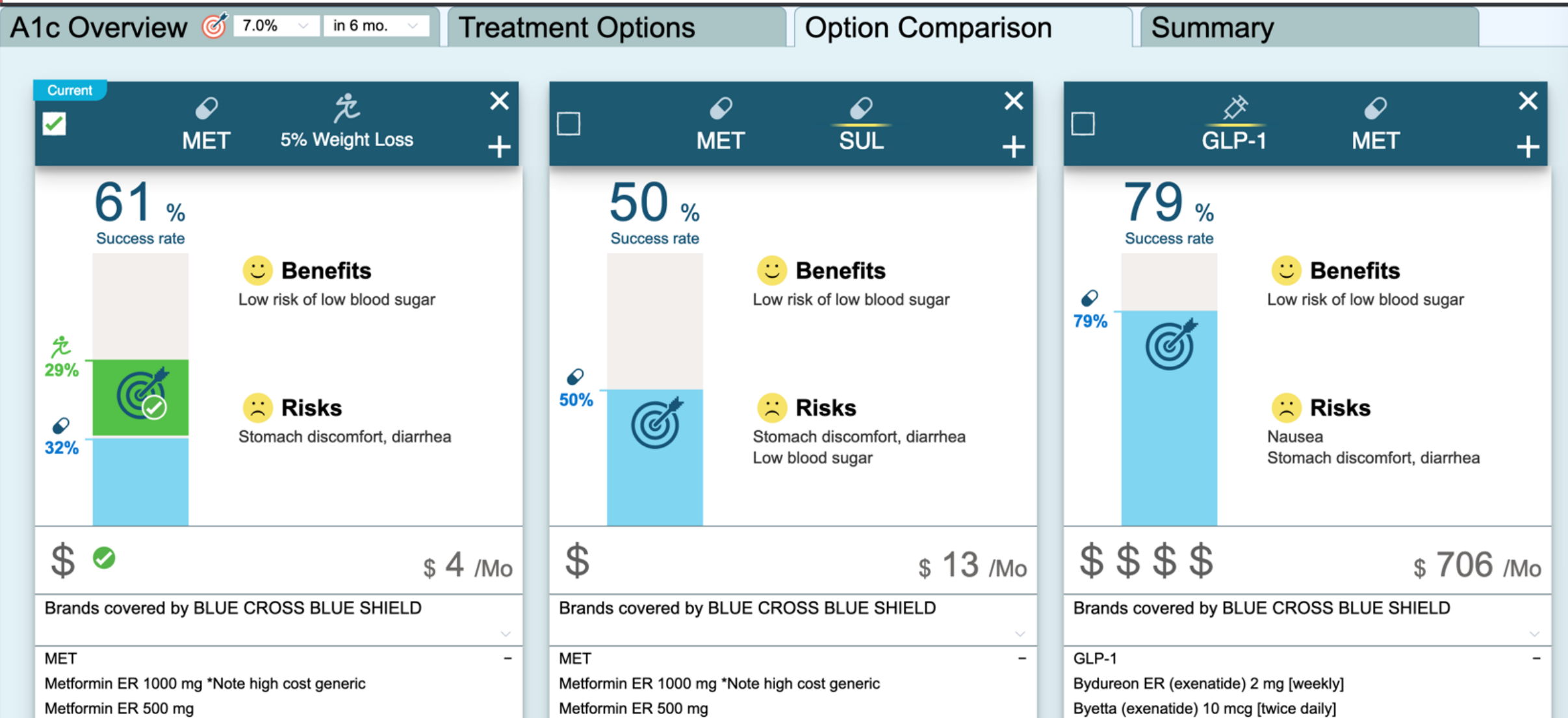
1. <https://hl7.org/fhir/uv/bulkdata/>

UNIVERSITY OF UTAH REIMAGINE EHR INITIATIVE

- Multi-stakeholder initiative started in 2016 leveraging FHIR and related standards
- Early leader spanning research and operations
- Agile, collaborative, innovative environment
- Steering committee co-chaired by CIO & CMIO
- >10 solutions
- >\$30M in grants



PERSONALIZED MEDICINE FOR DIABETES THROUGH SMART ON FHIR



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- Collaboration with Hitachi
- AI-driven Rx guidance (predictive model, AUC 0.87)
- Accounts for individuals' insurance coverage

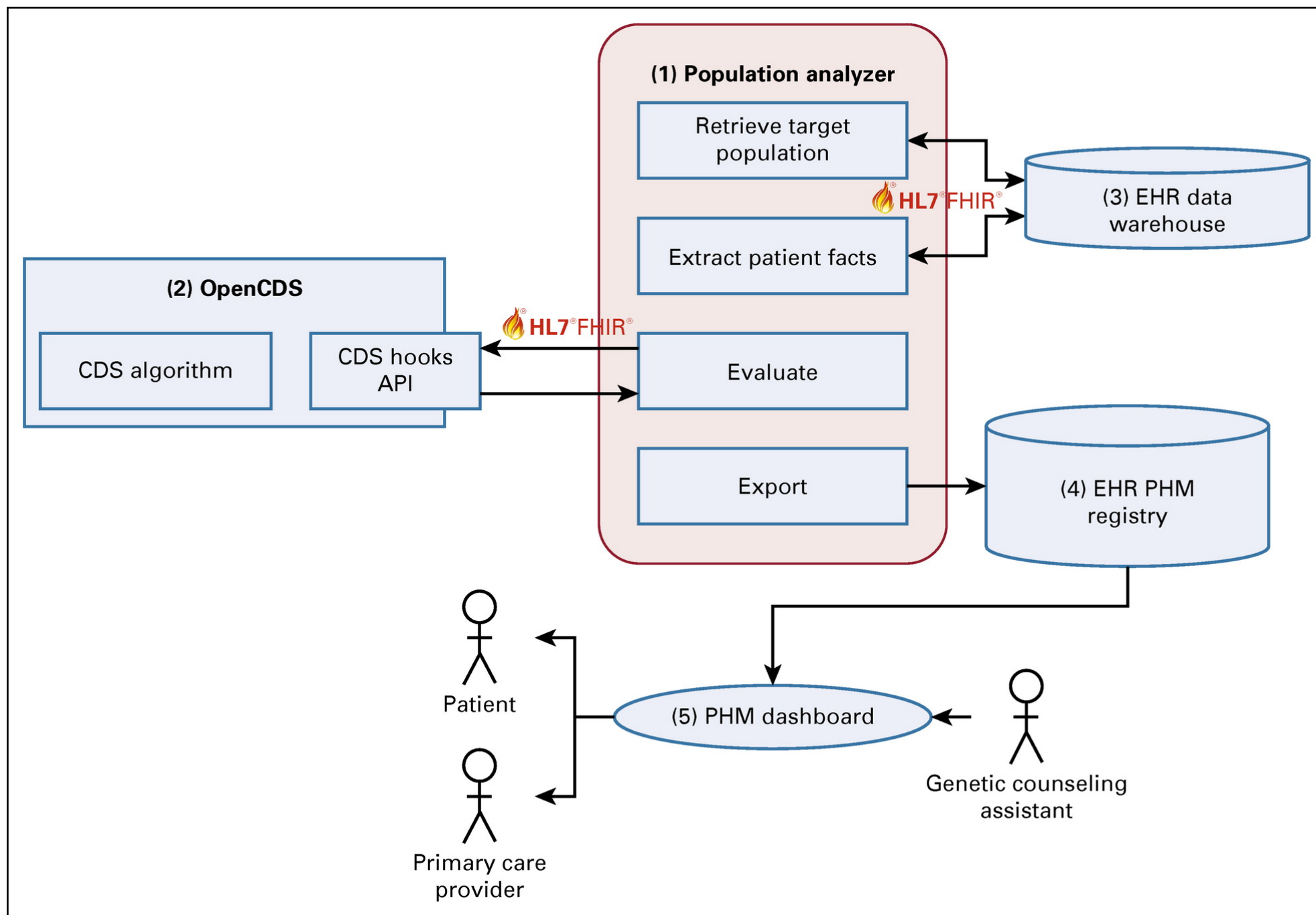
PERSONALIZED LUNG CANCER SCREENING WITH SMART ON FHIR

The screenshot shows the 'LUNG CANCER SCREENING RISK CALCULATOR' interface. On the left, under 'About the patient', it states 'This patient is **eligible** according to the USPSTF guidelines' with a 'View eligibility guidelines' button. Below this is a reminder: 'Remember: Do not recommend screening if you think the patient isn't healthy enough to withstand treatment.' and a 'Demographics' section. The main area has four tabs: 'Personalized Risk Assessment' (selected), 'Questions frequently asked by patients', 'Why personalize lung cancer screening recommendations?', and 'Evidence Basis and Development'. The 'Personalized Risk Assessment' tab displays the result: 'Screening benefits likely outweigh harms'. It lists three bullet points: '5.08% risk of dying of lung cancer in 6 years', 'Need to screen 99 patients to avoid 1 lung cancer death', and 'Lung cancer mortality benefit balanced by harms like false positive findings and possible overdiagnosis'. Below this are three buttons: 'Discourage Screening' (orange), 'Preference Sensitive' (grey), and 'Encourage Screening' (purple). A person icon stands on the 'Encourage Screening' button. At the bottom, there are 'Refresh' and 'Order LDCT' buttons. On the right side of the bottom bar, there are three status indicators: 'Shared decision making has not been previously done.', 'Shared decision making (SDM) done. Patient declines/defers screening.', and 'Shared decision making (SDM) done. Patient elects screening.'.

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- AHRQ R18 (PI: Kawamoto)
- Low-dose chest CT screening could save more lives than breast cancer screening (10,000/yr)
- Shared decision making using a decision aid required by CMS and USPSTF
- Current screening rate nationally: ~13%

FHIR-BASED POPULATION HEALTH MANAGEMENT



- Goal: identify and manage individuals at elevated risk of breast or colorectal cancer
- NCI U24 (PIs: Del Fiol, Kawamoto) & NCI U01
- ~13% of us are at elevated risk; most unaware
- AI used to extract data from free text, educate via chatbot

Del Fiol G, Kohlmann W, Bradshaw RL, Weir CR, Flynn M, Hess R, Schiffman JD, Nanjo C, Kawamoto K (2020). *JCO Clin Cancer Inform*, 4, 1-9.

KEY CHALLENGES

- Data normalization
- Execution performance
- EHR vendor support and standardization beyond the basics
 - Detailed clinical models
 - Genomics support
 - Population-based approaches
- The “last mile” of translating into practice

RECOMMENDATIONS

- Coordinate and synergize with the broader health IT community
- Recognize that hard problems largely persist (e.g., for data normalization)
- Work together to accelerate key needed developments, e.g., US Core Data for Interoperability
- Facilitate the full translation lifecycle, including the “last mile” of clinical implementation

THANK YOU!

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