Patient portals and EHR portability: Moving data with the patient

Gil Alterovitz, PhD, FACMI

Patient Portals

- Online website or mobile application
- Secure storage and access to personal health/medical information

Portal Limitations

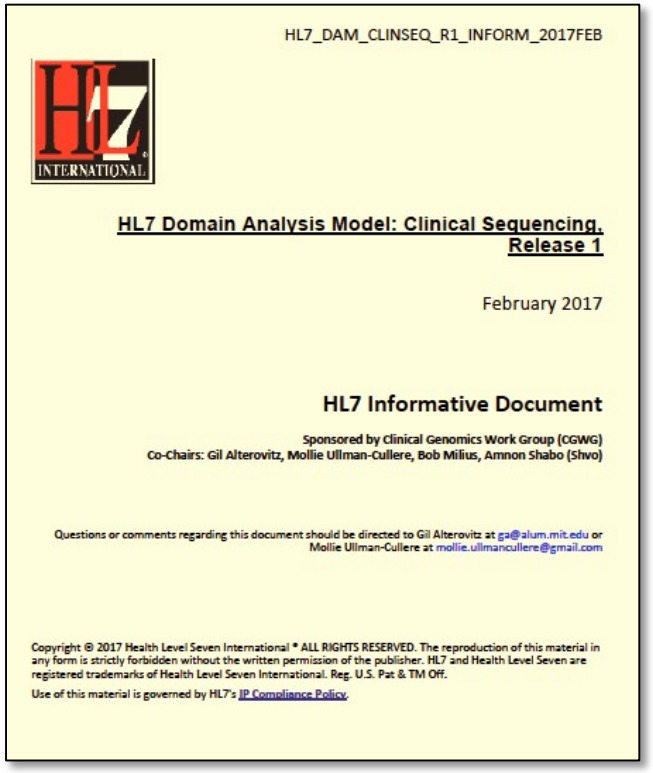
- **Few use cases**
  - Many portals originally designed as one way communication of lab results to patients.
  - Typically designed for patients to use on their own, before/after health care visits
  - Lack of integration of complex/genomic information

- **Closed environments**
  - Hard to add new use cases (often depends on EHR vendor)
  - Closed environment can prevent third-party innovation
  - Limited ability for analytics
  - Interoperability issues
  - Access may be limited (e.g. based on coverage, provider, etc.)


SORRY, SON...THERE'S NO APP FOR THAT

There are many use cases in genomics…

Use Cases:
"HL7 Domain Analysis Model: Clinical Sequencing"



HL7_DAM_CLINSEQ_R1_INFORM_2017FEB

HL7 Domain Analysis Model: Clinical Sequencing, Release 1

February 2017

HL7 Informative Document

Sponsored by Clinical Genomics Work Group (CGWG)
Co-Chairs: Gil Alterovitz, Mollie Ullman-Cullere, Bob Milius, Amnon Shabo (Shvo)

Questions or comments regarding this document should be directed to Gil Alterovitz at ga@alum.mit.edu or Mollie Ullman-Cullere at mollie.ullmancullere@gmail.com

- Publication contains list of use cases and workflows for clinical genomics- with focus on clinical sequencing
- Published Feb 2017
- Work has formed basis for several national/international programs
- Used as guide for analyzing use cases by a number of provider institutions.

https://www.healthcare-informatics.com/news-item/interoperability/hl7-publishes-domain-analysis-model-clinical-sequencing

Some Key Use Cases

Family History

Estimating disease risk using germline tests, family pedigree, and clinical data

Precision Cancer

Somatic (tumor) sequencing for diagnosis/prognosis

HLA Typing

Immunogenomic sequencing for organ matching, etc.

NGS Assay Evaluation/Regulatory

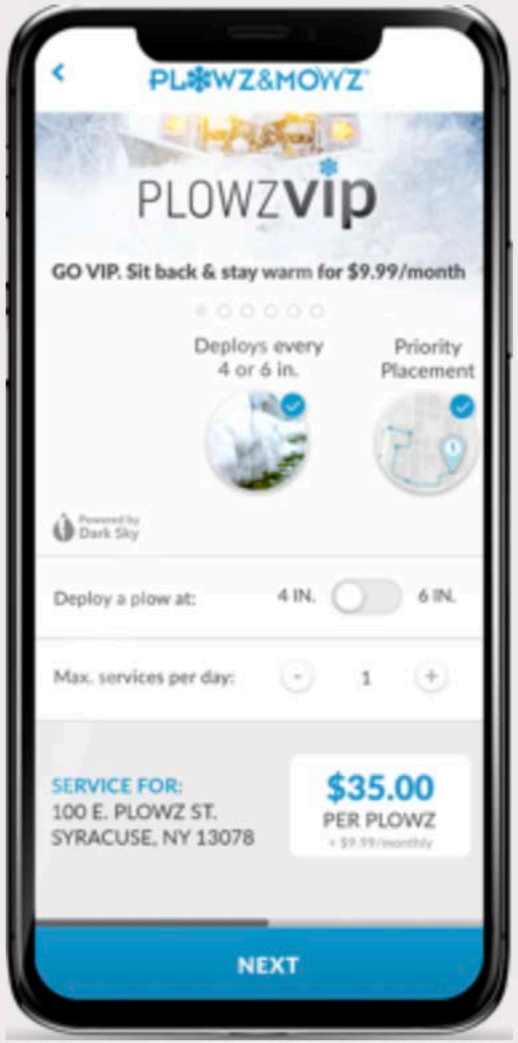
Benchmarking NGS quality of vendor to “gold standard”

Pharmacogenomics

Germline and somatic sequencing for drug selection, rejection, dosing

And, patients want to be engaged in their own care and data use/sharing…

ACTUALLY, DAD... THERE'S A NUMBER OF APPS FOR THAT.


PLOWZ&MOWZ
PLOWZvip
GO VIP. Sit back & stay warm for $9.99/month
Deploys every 4 or 6 in.
Priority Placement
Deploy a plow at:
4 IN.
6 IN.
Max. services per day:
1
SERVICE FOR:
100 E. PLOWZ ST.
SYRACUSE, NY 13078
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PER PLOWZ
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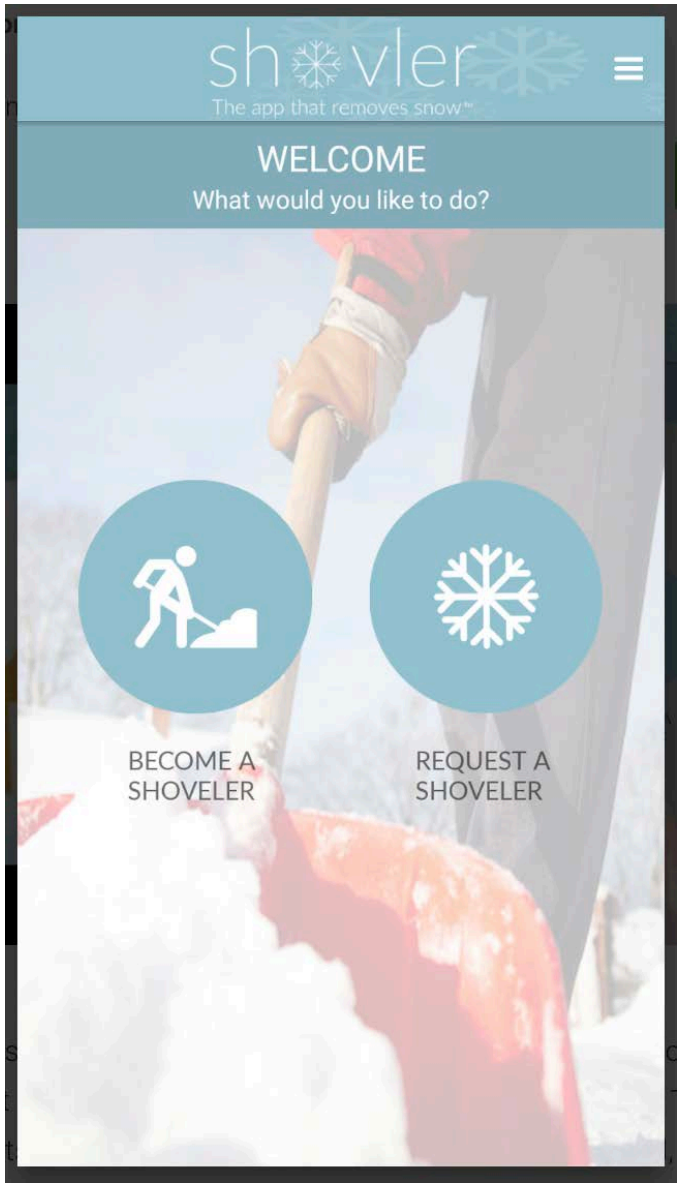
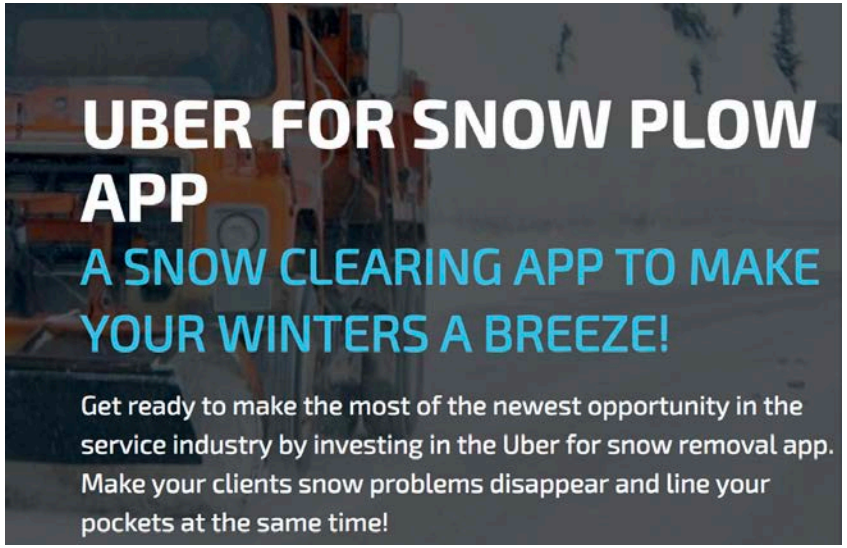

shovler
The app that removes snow™
WELCOME
What would you like to do?
BECOME A SHOVELER
REQUEST A SHOVELER


UBER FOR SNOW PLOW APP
A SNOW CLEARING APP TO MAKE YOUR WINTERS A BREEZE!
Get ready to make the most of the newest opportunity in the service industry by investing in the Uber for snow removal app. Make your clients snow problems disappear and line your pockets at the same time!

And, the same may become true for genomics by creating an EHR ecosystem…

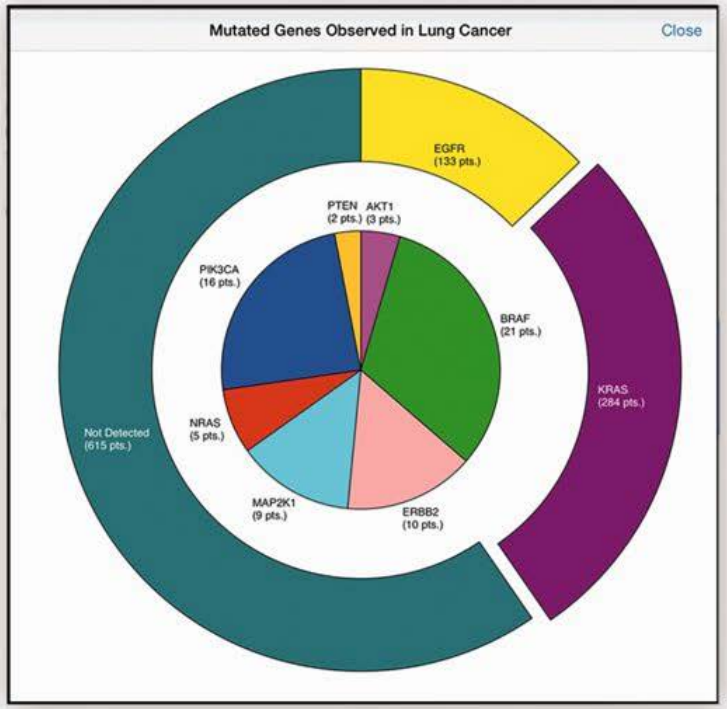
Apps

A

B

SMART Genomics Advisor

C



SMART Precision Cancer Medicine

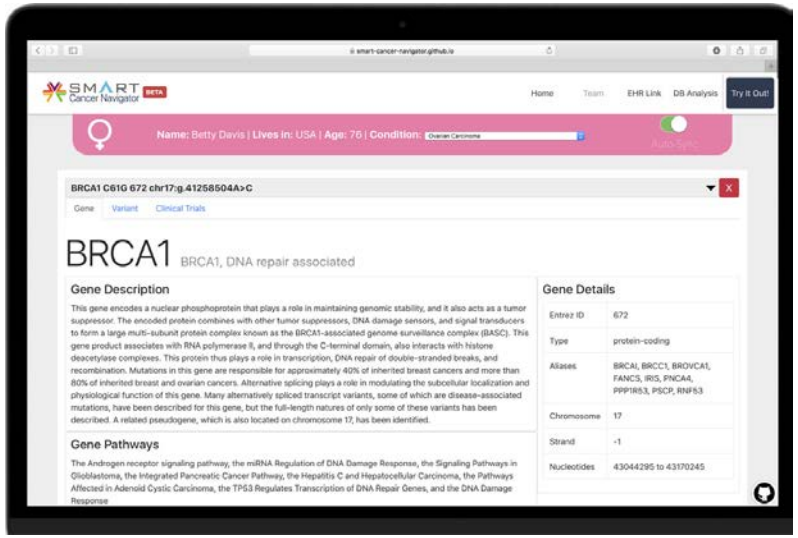
Alterovitz, et al, SMART on FHIR Genomics: Facilitating standardized clinico-genomic apps, JAMIA, 2015.

Also, providers want to leverage extra-EHR resources/services

SMART Cancer Navigator

- A Framework for Implementing ASCO Workshop Recommendations to Enable Precision Cancer Medicine

- Securely links patient-specific data from EHRs via FHIR and multiple laboratory/reference knowledge bases for information and treatment options.



https://smart-cancer-navigator.github.io

J Warner, I Prasad, M Bennett, M Arniella, A Beeghly-Fadiel, K Mandl, and G Alterovitz, SMART Cancer Navigator: A Framework for Implementing ASCO Workshop Recommendations to Enable Precision Cancer Medicine, JCO Precision Oncology 2018:2, 1-14.

SMART on FHIR®©

How do we enable this?

Moving Forward

- **Few use cases -> Many use cases**
  - Enable an apps, app stores ecosystem

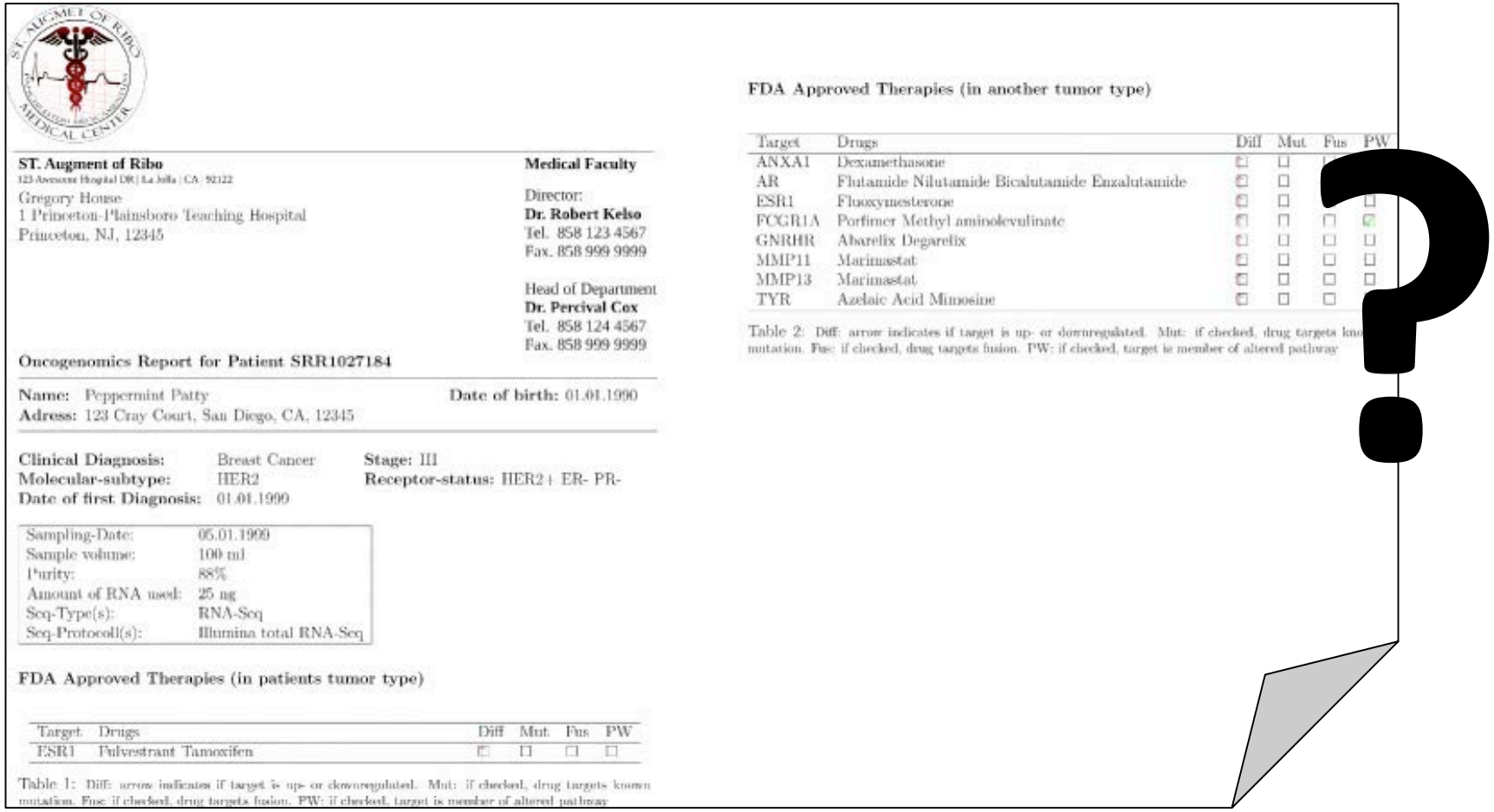
- **Closed environments -> Open environments to foster innovation**
  - Enable standards-based environments and tools

How best to engage with

genomics lab results

to improve patient care?

Data buried in PDFs, **or** …



ST. Augment of Ribo
123 Awesome Hospital DR | La Jolla | CA 92122
Gregory House
1 Princeton-Plainsboro Teaching Hospital
Princeton, NJ, 12345

Medical Faculty
Director:
Dr. Robert Kelso
Tel. 858 123 4567
Fax. 858 999 9999

Head of Department
Dr. Percival Cox
Tel. 858 124 4567
Fax. 858 999 9999

Oncogenomics Report for Patient SRR1027184

Name: Peppermint Patty **Date of birth:** 01.01.1990
Adress: 123 Cray Court, San Diego, CA, 12345

Clinical Diagnosis: Breast Cancer **Stage:** III
Molecular-subtype: HER2 **Receptor-status:** HER2 + ER- PR-
Date of first Diagnosis: 01.01.1999

Sampling-Date:	05.01.1999
Sample volume:	100 ml
Purity:	88%
Amount of RNA used:	25 ng
Seq-Type(s):	RNA-Seq
Seq-Protocoll(s):	Illumina total RNA-Seq

FDA Approved Therapies (in patients tumor type)

Target	Drugs	Diff	Mut	Fus	PW
ESR1	Fulvestrant Tamoxifen	☐	☐	☐	☐

Table 1: Diff: arrow indicates if target is up- or downregulated. Mut: if checked, drug targets known mutation. Fus: if checked, drug targets fusion. PW: if checked, target is member of altered pathway

FDA Approved Therapies (in another tumor type)

Target	Drugs	Diff	Mut	Fus	PW
ANXA1	Dexamethasone	☐	☐	☐	☐
AR	Flutamide Nilutamide Bicalutamide Enzalutamide	☐	☐	☐	☐
ESR1	Fluoxymesterone	☐	☐	☐	☐
FCGR1A	Porfimer Methyl aminolevulinate	☐	☐	☐	☑
GNRHR	Abarelix Degarelix	☐	☐	☐	☐
MMP11	Marimastat	☐	☐	☐	☐
MMP13	Marimastat	☐	☐	☐	☐
TYR	Azelaic Acid Minosine	☐	☐	☐	☐

Table 2: Diff: arrow indicates if target is up- or downregulated. Mut: if checked, drug targets kno
mutation. Fus: if checked, drug targets fusion. PW: if checked, target is member of altered pathway

Contextualized, dynamic, actionable, and re-computable results in …

Structured data, enabling analytics…

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04/schema#",

   "definitions": {
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       "properties": {
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        "span": {"$ref": "#/definitions/span"},
        "alt": {"$ref": "#/definitions/sequence"}
       },
       "required": ["seq_id", "span", "alt"]
    },

    "sequence": {
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       "pattern": "^[A-Z]$"
    },

    "sha512_20": {
       "type": "string",
       "pattern": "^[0-9abcdef]$",
       "minLength": 20,
       "maxLength": 128,
    }
   }
}
```

iPad 11:03 AM 83%

Patient Search

Patient name

[redacted] (MRN: [redacted])

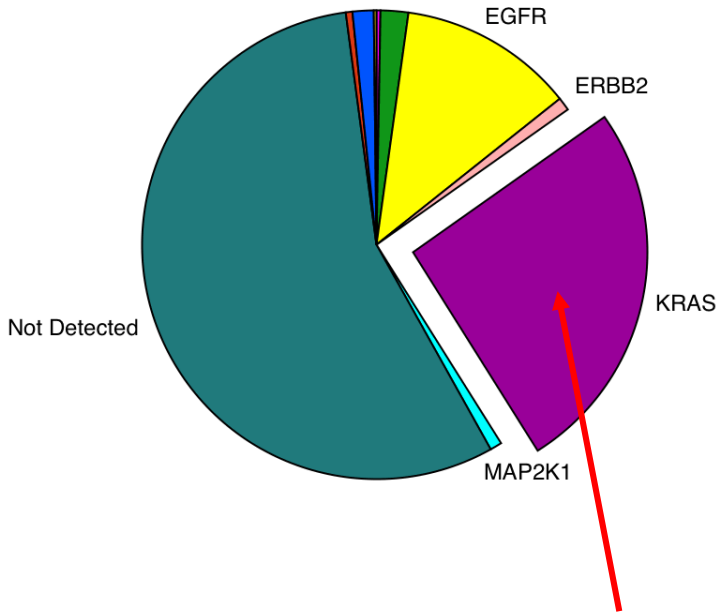
Male, [redacted]
Diagnosis: Lung Cancer
Mutation: KRAS G12C

Date of birth

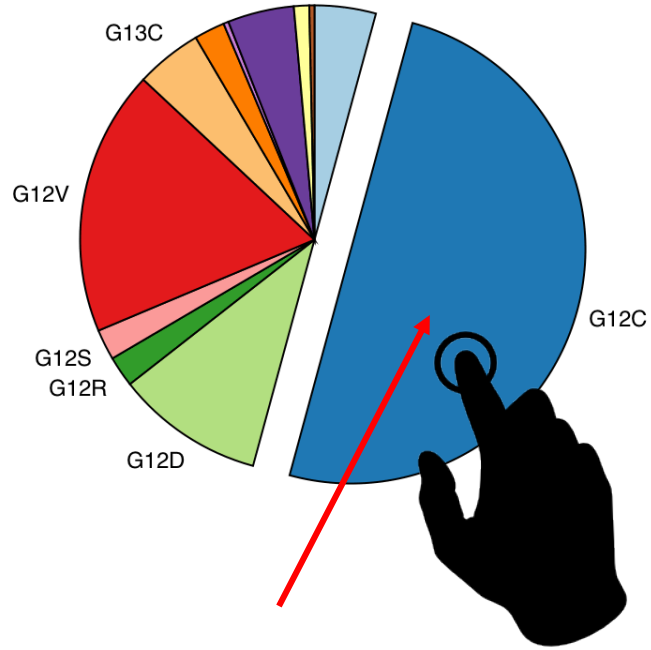
Medical record number

Warner & Alterovitz. SMART Precision Cancer Medicine, JAMIA, 2016.

Mutated Genes Observed in Lung Cancer



Observed Variants in Lung Cancer Patients with KRAS Mutation



For a specific gene mutation in a specific lung cancer patient, show information for most common KRAS in ***lung cancer*** populations

MRN Search Patient Search About

Vision

Unified Clinical and Genomic Data Standard

clinical data ◆ genomics (omics) ◆ precision medicine

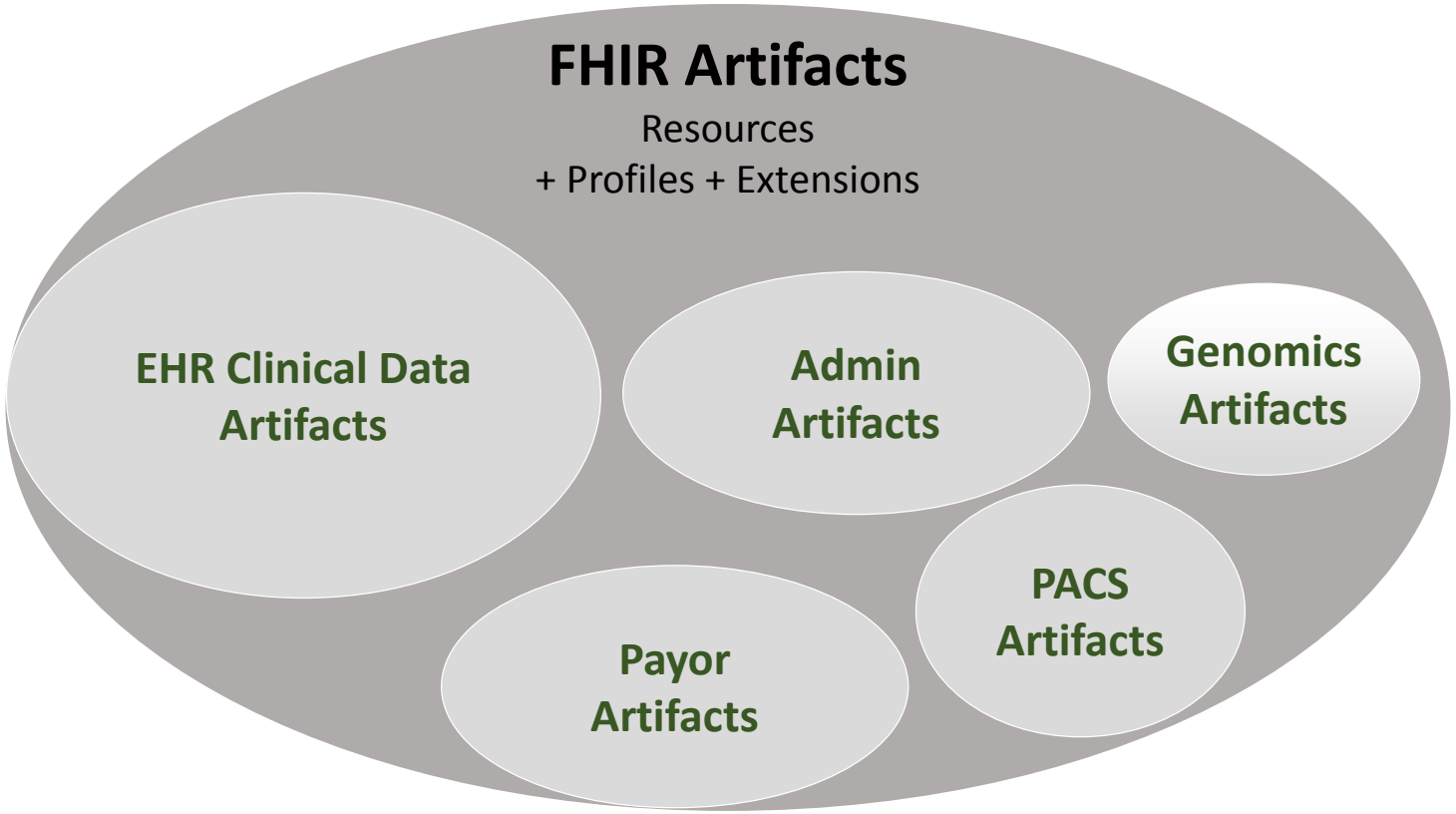
Modern Technology

web-friendly ◆ RESTful ◆ resource-based

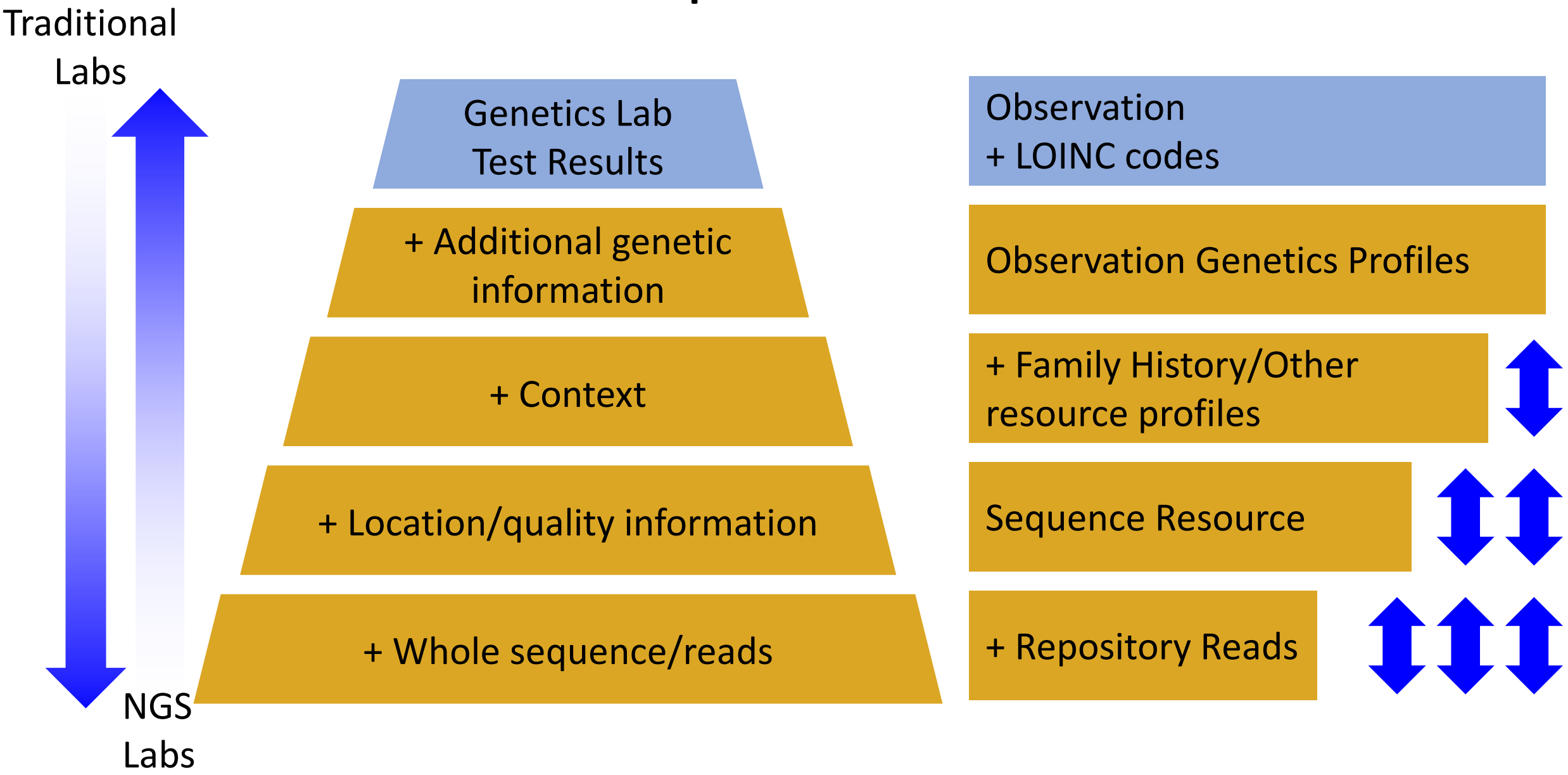
Workflow Ready

queryable ◆ granular ◆ on-demand

Fast Healthcare Interoperability Resources (FHIR)



Incremental Adoption of FHIR Genomics



Precision Medicine Initiative (All of Us) Cohort Program RFA

Describe potential utilization of current and emerging standards to facilitate data exchange and analysis, such as:

- Standards for capture and representation of family health history such as SNOMED CT and HL7 Version 3 Implementation Guide: Family History/Pedigree for familial relationships.
- HL7 DIGITizE Actions Collaborative draft LOINC specification for pharmacogenomics.
- HL7 Clinical Genomics WG standards including CDA R2 Clinical Genetics Reporting, Clinical Genomics Pedigree Model, HL7 Genetic Testing Results Message (V2), and Clinical Sequencing Domain Analysis Model (DAM).
- SMART on FHIR Genomics standards to support development of clinico-genomic apps to communicate clinical genomics data between EHR systems.
- Open ID Connect, OAuth and UMA for individual authorization and authentication
- More complete authorization standards (e.g., IHE XUA, IUA, etc.) to ensure authorization standards are compatible across disparate networks.
- Global Alliance for Genomics and Health (GA4GH) standards to address computable consent for

SMART on FHIR Genomics standards to support development of clinico-genomic apps to communicate clinical genomics data between EHR systems.

SMART on FHIR Genomics: Facilitating Standardized Clinico-Genomic Apps

ABSTRACT

BRIEF COMMUNICATION

Alterovitz, et al, SMART on FHIR Genomics: Facilitating standardized clinico-genomic apps, JAMIA, 2015.

SMART on FHIR

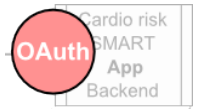
API

Resource oriented, everything a **URL**

Data Model

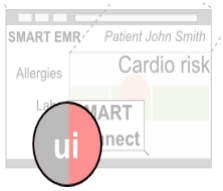
Context (container, user, patient)

Medical (problems, allergies, …)



Authentication

Consistent delegation, web standards (OAuth)



UI

Standards-based integration (HTML5)

SMART®

SMART on FHIR

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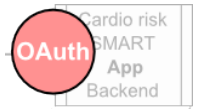
FHIR



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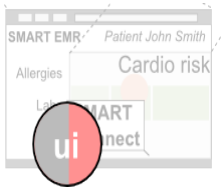

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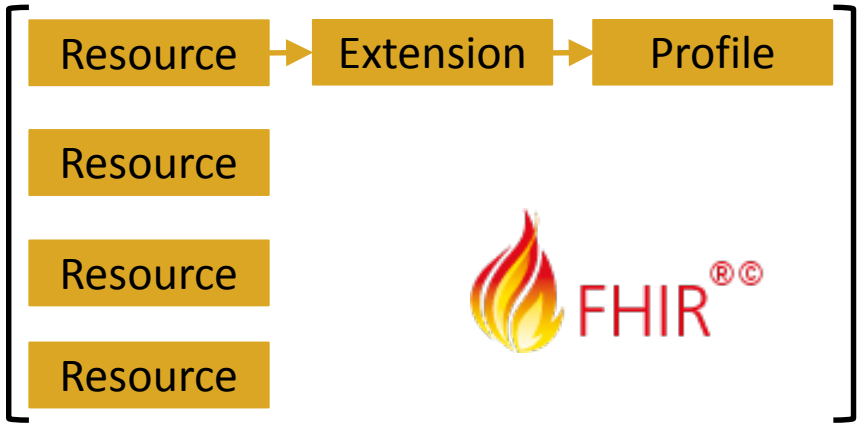
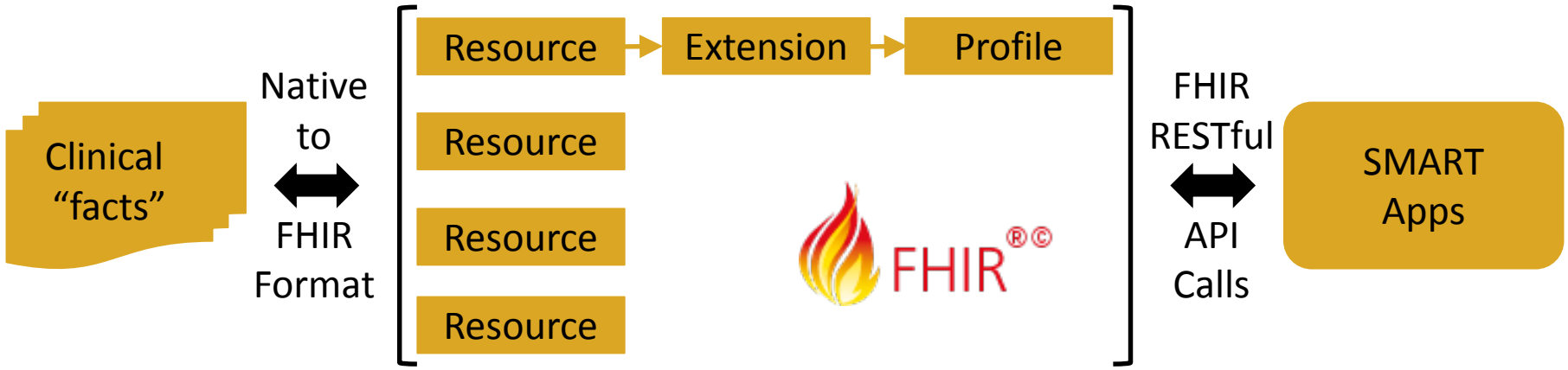
SMART®

SMART on FHIR on Genomics

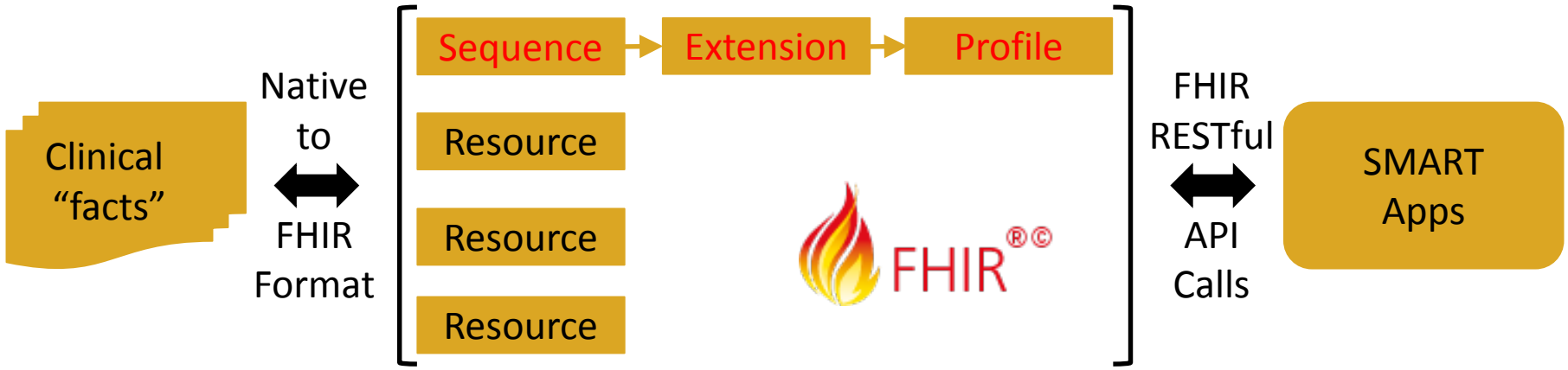


SMART on FHIR on Genomics



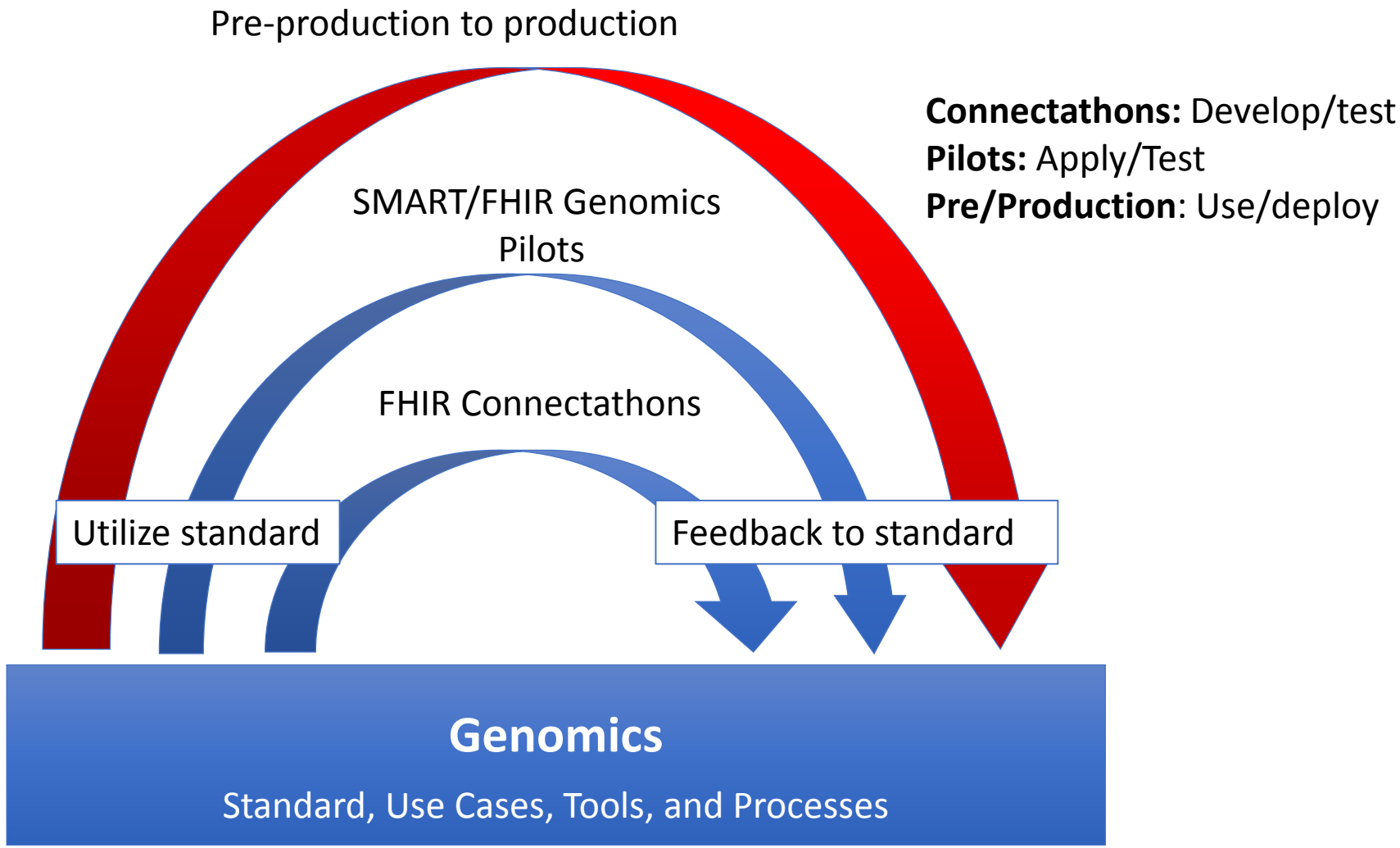
Moving data in and out of native systems to third-party apps

SMART on FHIR with Genomics



Landscape of Adoption Programs

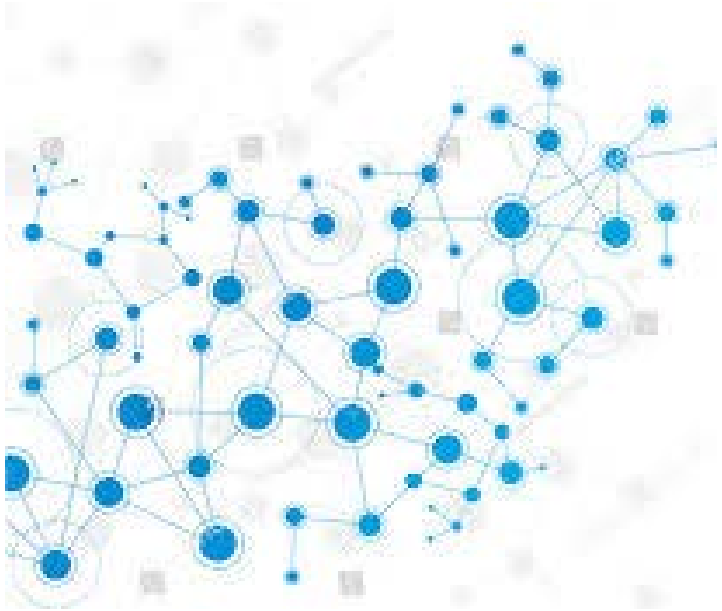
and Iterative Genomics Standard Feedback



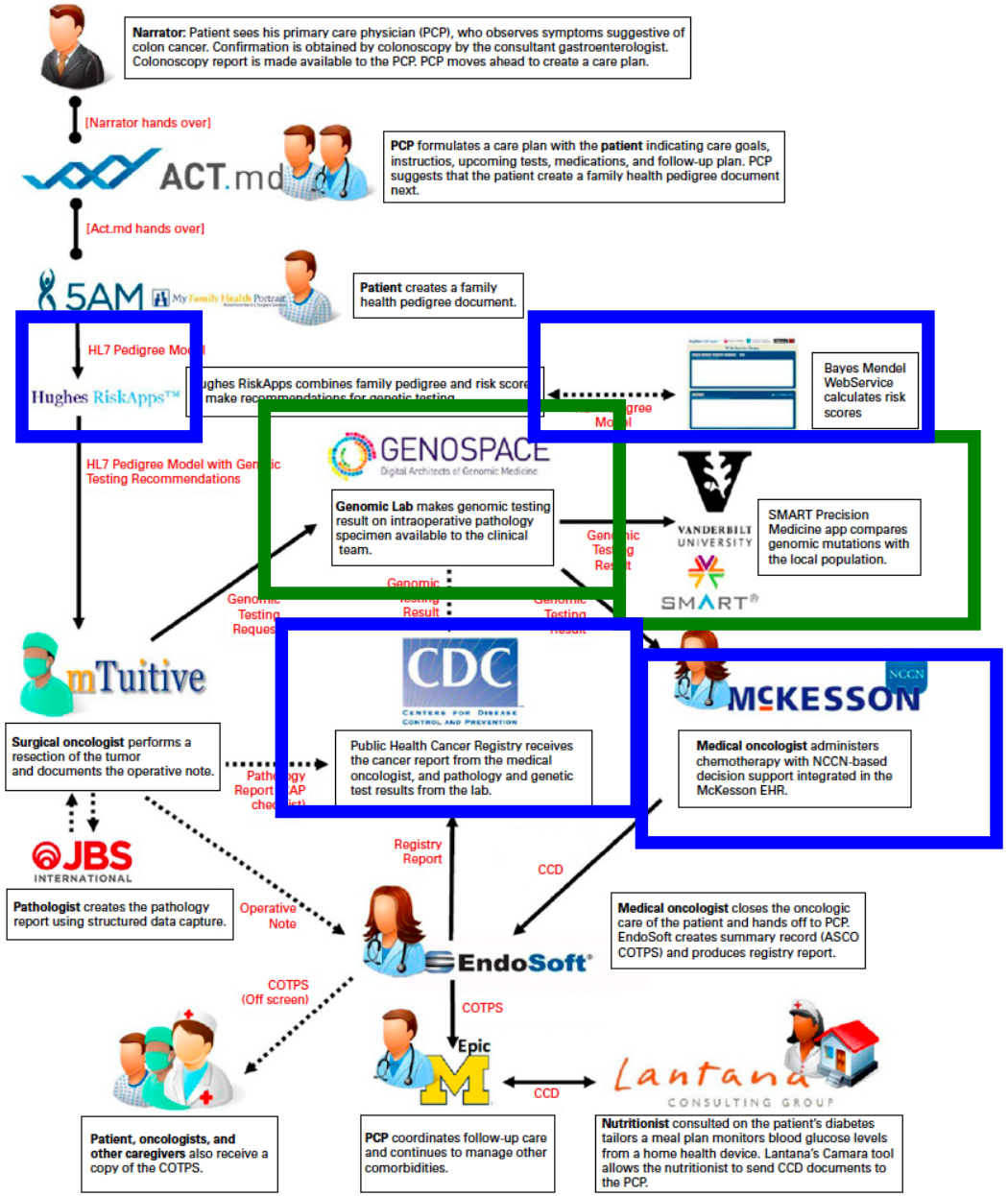
Connectathons: Develop/test
Pilots: Apply/Test
Pre/Production: Use/deploy

Toward a Genomics-Empowered Ecosystem

- Move away from point-to-point thinking toward networked ecosystem
- Need ability to communicate in heterogeneous ecosystem with multiple parties
- Create ability to communicate different levels of clinical genomic information
- Create metrics for measuring different speed in adoption



SMART on FHIR Pilots Ecosystem Example



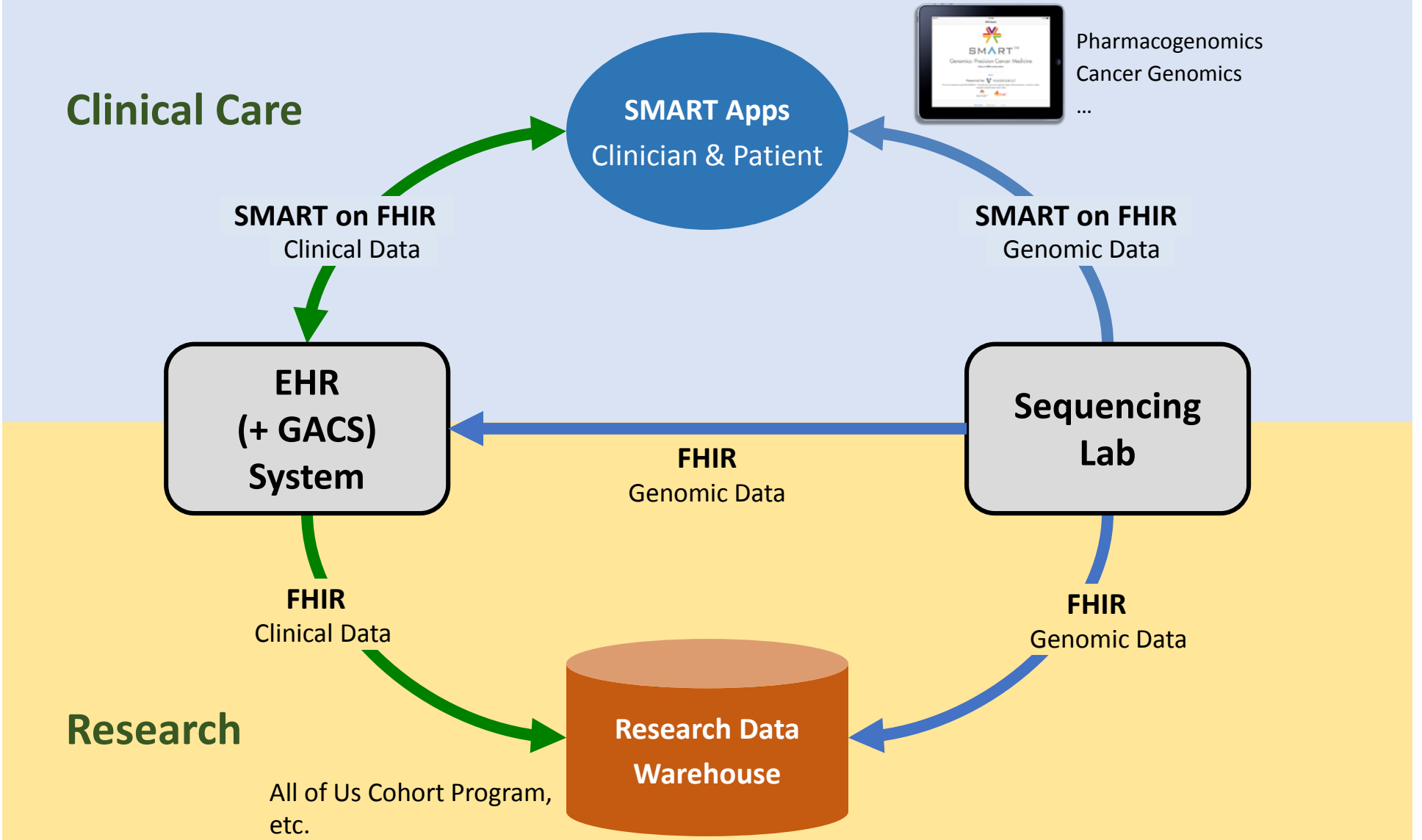
Org. support SMART/FHIR

Org. pilots of SMART/FHIR

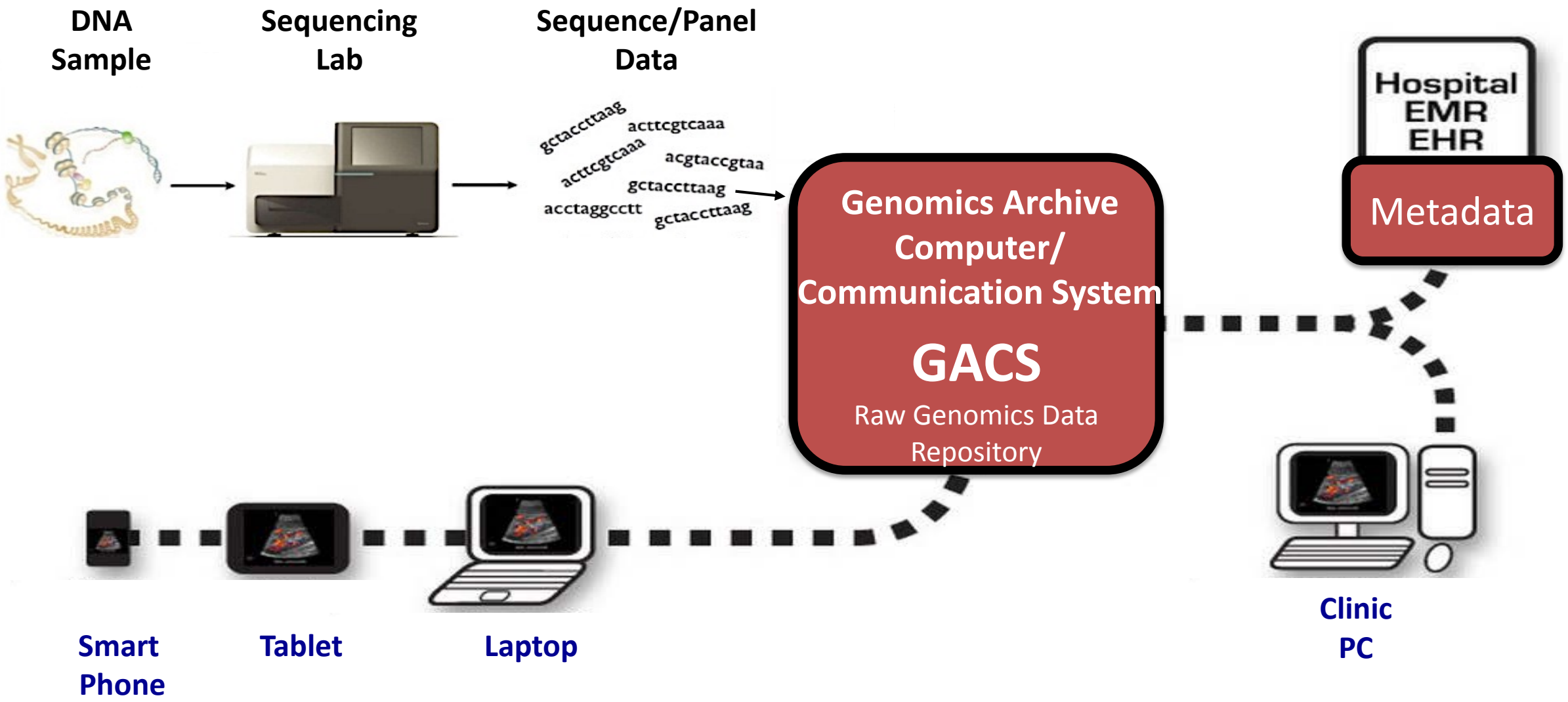
Data Sharing to Support the Cancer Journey in the Digital Era. Journal of oncology practice, 2016.

Pathway toward Clinical and Genomic Integration

Single National Standard for Clinical Care & Research



Where do genomics results go?


DNA Sample
Sequencing Lab
Sequence/Panel Data
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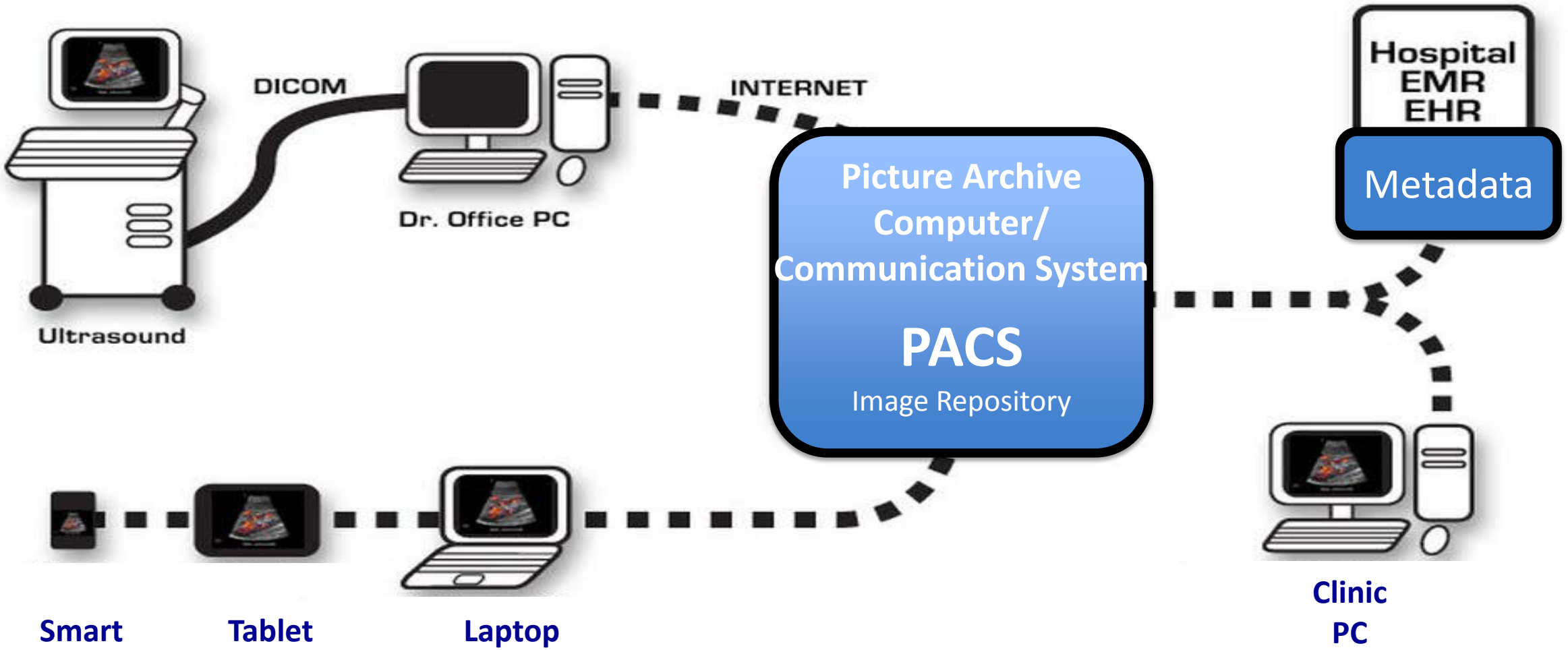
acttcgtcaaa
acttcgtcaaa
acgtaccgtaa
gctaccttaag
acctaggcctt
gctaccttaag
Genomics Archive Computer/ Communication System
GACS
Raw Genomics Data Repository
Hospital EMR EHR
Metadata
Smart Phone
Tablet
Laptop
Clinic PC

Precedent


DICOM
INTERNET
Dr. Office PC
Ultrasound
Picture Archive
Computer/
Communication System
PACS
Image Repository
Hospital
EMR
EHR
Metadata
Smart
Phone
Tablet
Laptop
Clinic
PC

Looking Forward…

- SMART on FHIR-powered cloud-based servers with patient apps
  - App stores enable patients to customize experience based on needs
    - Patient ability to control information sharing in real-time for clinical/genomic information.
- Apps that enable patients and providers to “collaborate” on care
  - Screens built for provider-patient engagement
- Apps designed for genomic care coordination
  - Patient control of information/sharing

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