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”

• 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.

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.
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...
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-canceravigator.github.io

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 ...
Contextualized, dynamic, actionable, and re-computable results in ...
Structured data, enabling analytics...

```json
{
    "$schema": "http://json-schema.org/draft-04/schema#",
    "definitions": {
        "variant": {
            "type": "object",
            "properties": {
                "seq_id": {
                    "$ref": "#/definitions/sha512_20"},
                "span": {
                    "$ref": "#/definitions/span"},
                "alt": {
                    "$ref": "#/definitions/sequence"}
            },
            "required": ["seq_id", "span", "alt"]
        },
        "sequence": {
            "type": "string",
            "pattern": "^[A-Z]"}
    },
    "sha512_20": {
        "type": "string",
        "pattern": "^[0-9abcdef]$",
        "minLength": 20,
        "maxLength": 128,
    }
}
```
For a specific gene mutation in a specific lung cancer patient, show information for most common KRAS in lung cancer populations.

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

- Traditional Labs
- NGS Labs

Genetics Lab Test Results
- + Additional genetic information
- + Context
- + Location/quality information
- + Whole sequence/reads

Observation
- + LOINC codes
- Observation Genetics Profiles
- + Family History/Other resource profiles
- Sequence Resource
- + Repository Reads
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 DIGITizeE 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, IU4, 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

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 on FHIR

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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**

Pre-production to production

- SMART/FHIR Genomics Pilots
- FHIR Connectathons

**Utilize standard**

**Feedback to standard**

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

**Genomics**

Standard, Use Cases, Tools, and Processes
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

Pathway toward Clinical and Genomic Integration
**Single** National Standard for Clinical Care & Research

Clinical Care

- SMART on FHIR Clinical Data
- EHR (+ GACS) System

Research

- FHIR Clinical Data
- All of Us Cohort Program, etc.

SMART Apps Clinician & Patient

Sequencing Lab

- SMART on FHIR Genomic Data
- FHIR Genomic Data
- FHIR Genomic Data

Research Data Warehouse

Pharmacogenomics Cancer Genomics ...
Where do genomics results go?
PACS Image Repository

Picture Archive Computer/Communication System

Smart Phone  Tablet  Laptop

Clinic PC

Hospital EMR EHR

Metadata
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
Gil Alterovitz

ga@alum.mit.edu

LinkedIn: