



The Office of the National Coordinator for
Health Information Technology

The Intersection Between Research and Care Delivery: Leveraging Health IT to Advance Precision Medicine

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ONC Annual Meeting
November 29, 2018



Agenda

- ONC Chief Scientist Division (CSD) Overview
- Patient-Centered Outcomes Research (PCOR) Overview
- ONC and the Precision Medicine Initiative (PMI)

ONC Chief Scientist Division Overview

- Develop and evaluate ONC's scientific efforts and activities, and recommend scientific policy to the National Coordinator
- Direct a wide variety of innovative health IT programs and projects to advance an interoperable health care system that also supports biomedical and health services research
- Promoting activities that **spur innovation, support patient-centered outcomes research, and advance precision medicine**, through the sharing of electronic health data (clinical, genomic, and personal) between patients, providers, and researchers

At the Intersection Between Research and Care Delivery

- To realize the benefits of a 21st Century health system, more needs to be done to support the sharing of electronic health data
- Current health IT infrastructure is distinct for clinical care and for research, although the two may intersect in organizations that conduct both research and deliver care

ONC Scientific Initiatives and Activities

ONC Patient-Centered Outcomes Research (PCOR) Portfolio	Launched 2013
Structured Data Capture (SDC)	2013 - 2016
Data Access Framework (DAF)	2013 - 2017
Patient-Generated Health Data (PGHD)	2015 - Present
Patient Matching, Aggregating, and Linking (PMAL)	2015 - Present
Privacy and Security Framework for PCOR (PSP)	2015 - Present
Coordinated Registry Network (CRN) for Women's Health Technologies	2017 - Present
Common Data Model Harmonization (CDM)	2017 - Present
Patient-Reported Outcomes through Health IT (PRO)	2017 - Present
ONC Precision Medicine Initiative (PMI) Projects	Launched 2015
Sync for Science	2016 - Present
Sync for Genes	2016 - Present
Advancing Standards for Precision Medicine	2018 - Present

ONC Patient-Centered Outcomes Research (PCOR) Portfolio

- **LAUNCHED:** 2013
- **MISSION:** To develop the policy, standards, and services necessary to expand the data infrastructure for patient-centered outcomes research

ONC Patient-Centered Outcomes Research (PCOR) Portfolio

- **CURRENT PROJECTS LAUNCHED IN 2017:**
 - » Coordinated Registry Network (**CRN**) for Women's Health Technologies
 - To establish a standards-based CRN and develop the tools to facilitate collection of data in three clinical registries focused on women's health
 - » Common Data Model Harmonization (**CDM**)
 - To harmonize the common data models (CDMs) in Sentinel, PCORnet, OHDSI, and i2b2 to further advance the utility and interoperability of the data within these networks for use in PCOR
 - » Patient-Reported Outcomes through Health IT (**PRO**)
 - To standardize the integration of structured PRO data into EHRs and other health IT solutions supporting the interoperable exchange of this information

The Precision Medicine Initiative (PMI)

- **LAUNCHED:** 2015
- **MISSION:** To enable a new era of medicine through research, technology, and policies that empower patients, researchers, and providers to work together toward development of individualized care



ONC and the Precision Medicine Initiative

- Accelerate innovative collaboration around pilots and testing of standards that support health IT interoperability for research
- Adopt policies and standards to support privacy and security of cohort participant data
- Advance standards that support a participant-driven approach to patient data contribution

<https://healthit.gov/topic/precision-medicine>

The *All of Us* Research Program



- **LAUNCHED:** 2018
- **MISSION:** To accelerate health research and medical breakthroughs, enabling individualized prevention, treatment, and care for all of us
 - » One million or more volunteers, reflecting the broad diversity of the U.S.
 - » Opportunities for volunteers to contribute their health data and biospecimens on an ongoing basis
 - » Data will inform a variety of research studies

<https://allofus.nih.gov/>

What is Sync for Science (S4S)?

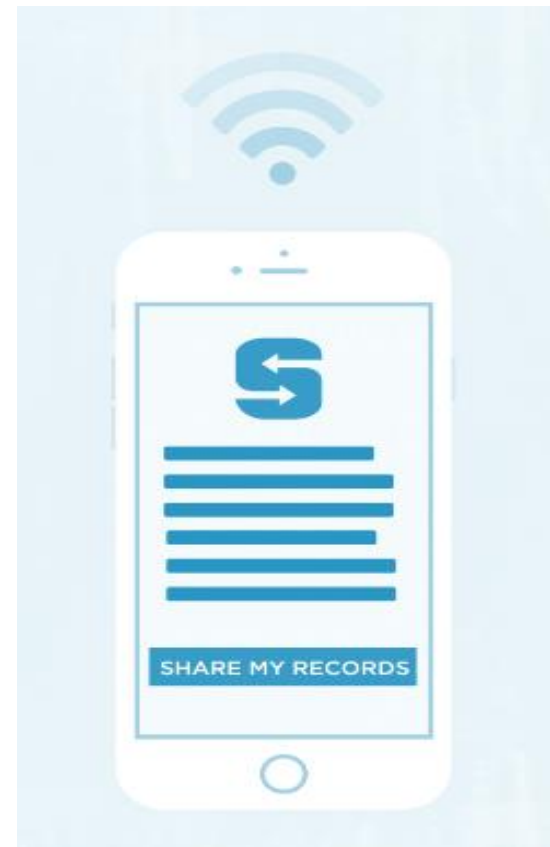


With a click of a button, you'll be able to voluntarily share your health data to help scientists perform groundbreaking research.

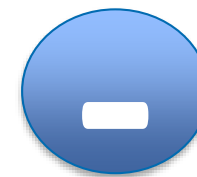
Patients Sharing EHR Data

- **LAUNCHED:** 2016
- **MISSION:** Promote patient-mediated access to data; establish structures that facilitate data sharing to the PMI cohort
- National collaboration among EHR developers, NIH, ONC, and Harvard Medical School's Department of Biomedical Informatics

<http://syncfor.science/>



Patients Sharing EHR Data



Recruitment at point of care

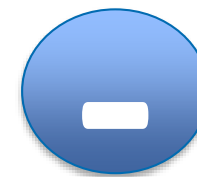
Patient trust

Well-defined data, specimens

Network creation expensive/slow

Limited cohort diversity

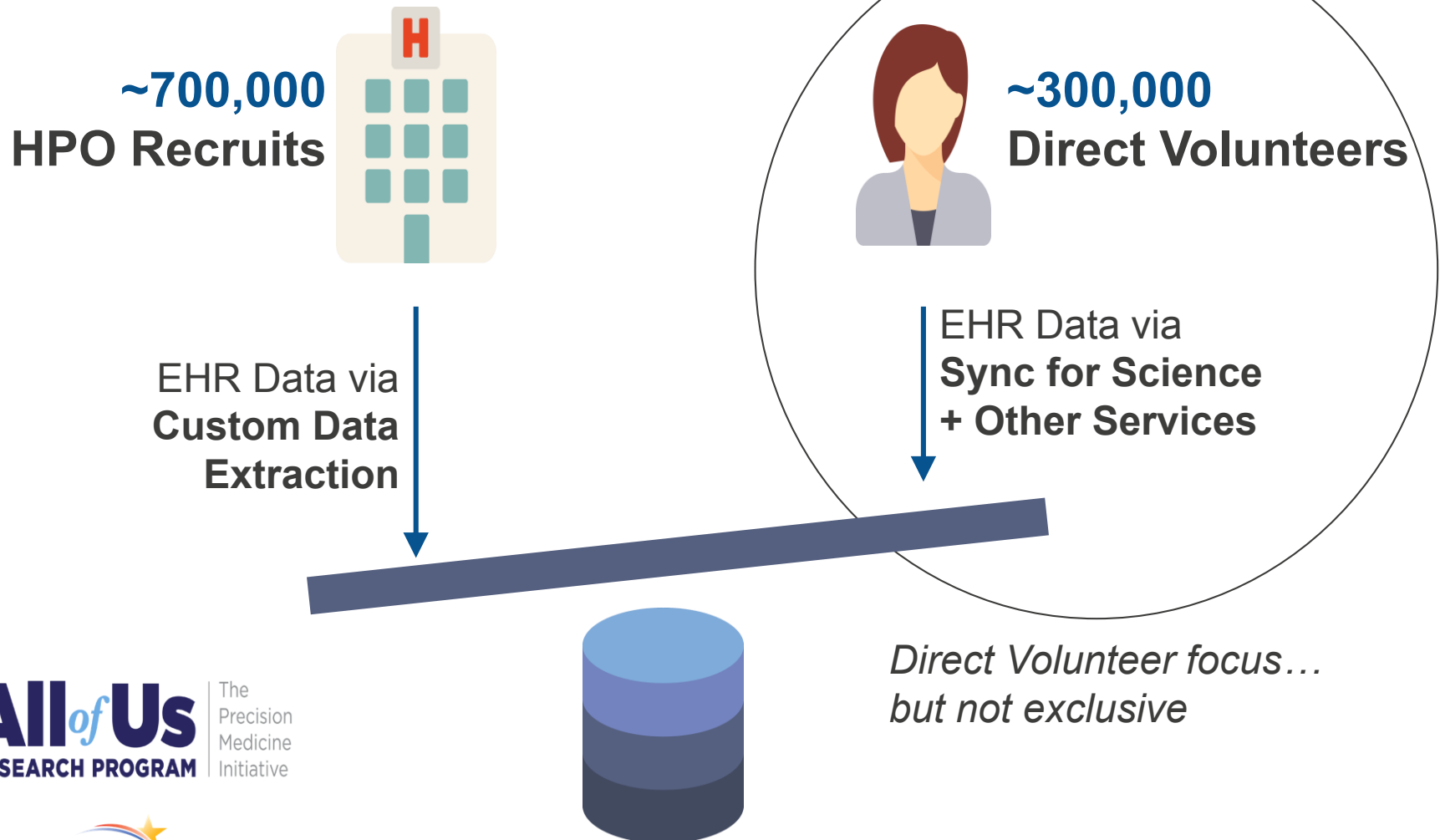
Poor coverage for rare conditions



Reach communities of patients
Recruit non-patients
Clear route to participation (PROs)

Faxing records expensive/slow
Painful EHR sharing workflows
Diversity of data formats

S4S as a Component of the *All of Us* Direct Volunteers (DV) Program



Traditional Out-of-Network Barriers



Patient ID: _____ Center ID: _____ Date: _____

Name of the Patient: _____ Age (years): _____ Sex: M F

Contact Number: _____ Address: _____

History of	Yes	Duration
Smoking		
Alcohol		
Burning in feet		
Amputation		
Heart attack/ stroke		
Diabetes		
Hypertension		
Dyslipidemia		
On ACE-i		
On ARBs		
On statins		

Family history of	Yes	No
Diabetes mellitus		
Kidney disease		
High blood pressure		
Heart disease		

Personal Details	Details
Height (cm)	
Weight (kg)	
Waist circumference (cm)	
BMI	
Education	

	Visit 1	Visit 2	Visit 3
Date of Visit			
Blood			
...			
...			
...			



Traditional Out-of-Network Barriers



via E 79th St 17 min
Best route
[DETAILS](#)

via F 70th St 16 min
2.6 miles

12:19 AM–12:46 AM 27 min

Patient ID: _____ Center ID: _____ Date: _____

Name of the Patient: _____ Age (years): _____ Sex: M F

Contact Number: _____ Address: _____

History of	Yes	Duration
Smoking		
Alcohol		
Burning in foot		
Amputation		
Heart attack/ stroke		
Diabetes		
Hypertension		
Dyslipidemia		
On ACE-I		
On ARBs		
On statins		

Family history of	Yes	No
Diabetes mellitus		
Kidney disease		
High blood pressure		
Heart disease		

Personal Details	Details
Height (cm)	
Weight (kg)	
Waist circumference (cm)	
BMI	
Education	

	Visit 1	Visit 2	Visit 3
Date of Visit			
Blood			
Urine			
ECG			
ECG			
ECG			
ECG			





March 2016

The Latest on Health Information Technology from ONC

EH/EMR	Health Innovation	From the ONC Desk	ONC Programs
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[Health IT Buzz](#) > [Health Innovation](#) > [NIH and ONC Launch the Sync for Science \(S4S\) Pilot: Enabling Individual Health Data Access and Donation](#)

NIH and ONC Launch the Sync for Science (S4S) Pilot: Enabling Individual Health Data Access and Donation

March 21, 2016, 11:46 am / [Jon White, M.D.](#) / Acting National Coordinator for Health IT, Office of the National Coordinator for Health IT,
[Josephine Briggs, M.D.](#) / Interim Director, Precision Medicine Initiative Cohort Program, and
[Josh Mandel, M.D.](#) / Research Scientist, Harvard Medical School Department of Biomedical Informatics

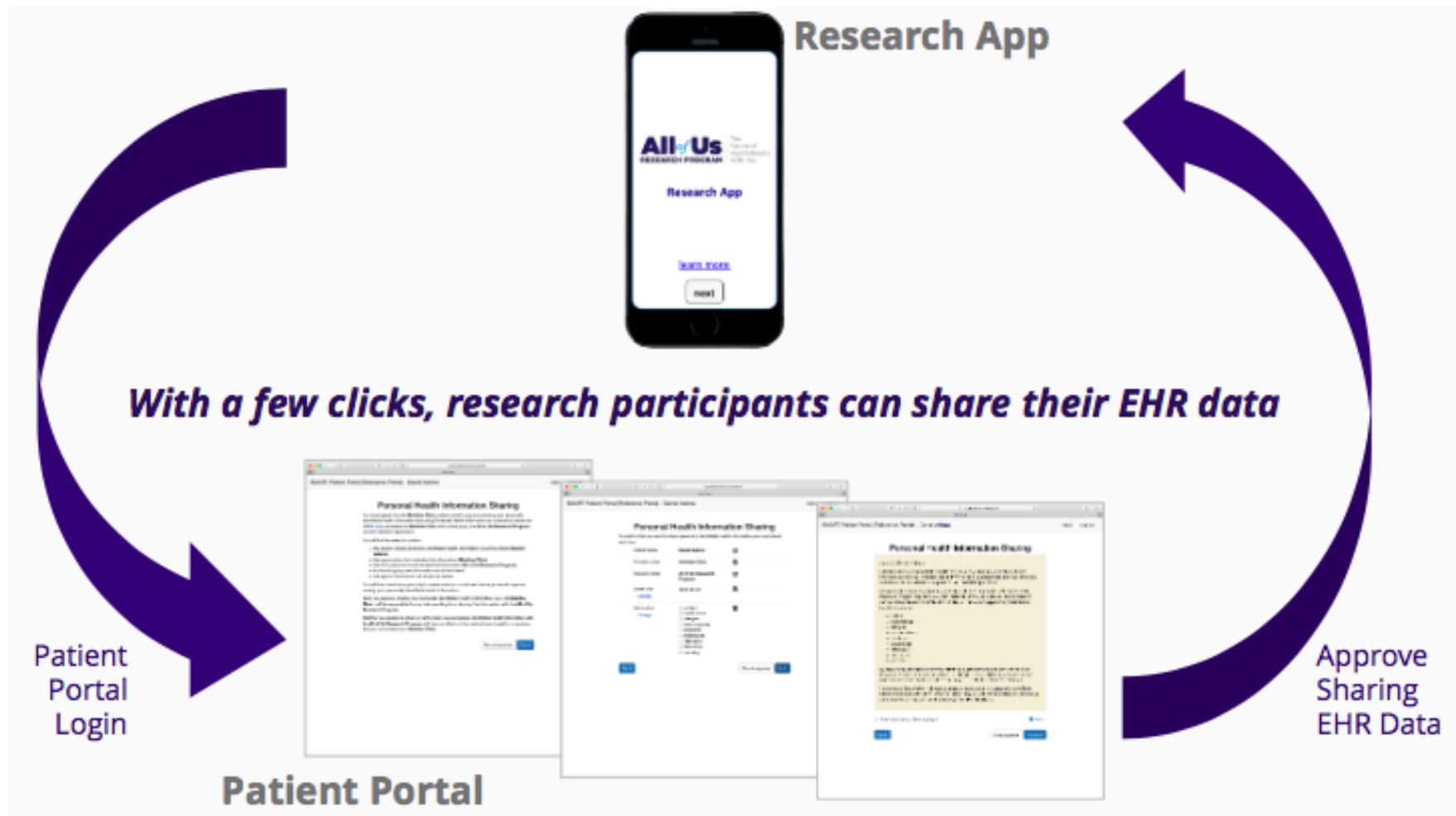
- NIH + Harvard Medical School + EHR Vendors
- Leveraged **HIPAA** and **Meaningful Use Stage 3** + 2015 **EHR Certification**
 - » Patient has right to access her/his EHR data (HIPAA)
 - » Patient application access *so patients can share their data via apps* (MU3)
 - » Common Clinical Data Set *covering a core set of summary elements* (MU3)
- Built on **open standards** for healthcare data + security
- Emphasizes **automated testing** of EHR vendor systems

S4S EHR Vendors – Implemented Patient Portal for S4S Pilot




eClinicalWorks





Research App – Share Your EHR Data



1

Electronic Health Record Data

Share my electronic health record (EHR) data ☒ On ☐ Off

Shared Sync for Science Medical Data

At the moment, you do not share your electronic health record (EHR) data from your health care provider.


[+ add a new health care provider](#)

2

Add Health Care Provider

Please enter the code from the brochure you received at your health care provider.

Enter signup code



Don't have a provider code?

Contact your health care provider to see if they are supported. We only support Sync for Science at the moment. You should be able to get provider code if your health provider supports Sync for Science.

[Learn more about how to get the code \(eligibility\)](#)

3

Confirm Health Care Provider

You have chosen to share your electronic health records (EHR) from **Berkshire Health Systems**.

Next, you will be directed to the (patient portal) provided by **Berkshire Health Systems**.

You can use your **Berkshire Health Systems** account information to log in.

Do you wish to proceed?

S4S at the EHR Patient Portal

Patient
Portal
Login

The image displays three overlapping screenshots of the SMART Patient Portal (Reference Portal) for Daniel Adams, illustrating the 'Personal Health Information Sharing' consent process.

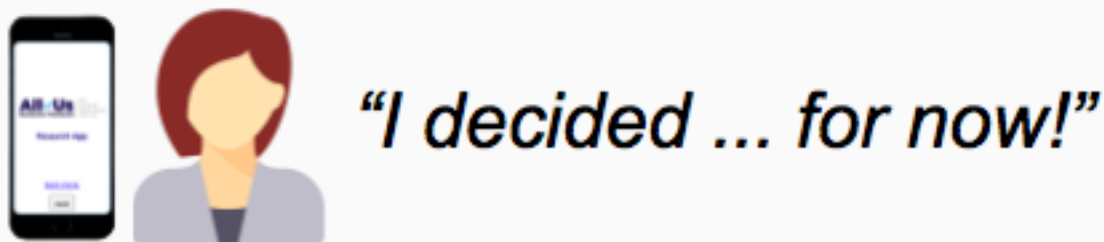
Left Screenshot: Shows the initial consent screen. It explains that the user has signed into the MyBlue Ohio patient portal to approve sharing their personally identifiable health information (PHI) and has agreed to the MyBlue Ohio Research Program. It lists the types of information that would be shared: the person whose PHI is shared, the organization that maintains the information (MyBlue Ohio), the program that will receive the information (All of Us Research Program), and the type of information that would be shared. It also states that the user will have the opportunity to review what they've approved before they would approve sharing their personally identifiable health information. A 'Do not approve' button and a 'Next' button are visible.

Middle Screenshot: Shows the 'Personal Health Information Sharing' screen with a table of information to be shared. The table has columns for 'Patient name', 'Provider name', 'Facility name', 'Date of birth', and 'Information'. The 'Patient name' is Daniel Adams, 'Provider name' is MyBlue Ohio, 'Facility name' is All of Us Research Program, 'Date of birth' is 2010-01-01, and 'Information' is a list of categories: patient, medication, allergies, immunizations, problems, procedures, vital signs, laboratory, and imaging. A 'Do not approve' button and a 'Next' button are visible.

Right Screenshot: Shows the 'Personal Health Information Sharing' screen with a list of categories of information to be shared. The categories are: patient, medication, allergies, immunizations, problems, procedures, vital signs, laboratory, and imaging. A 'Do not approve' button and a 'Next' button are visible.

Approve
Sharing
EHR Data

Research App – Return Paths



Electronic Health Record Data Sharing Successful

Thank you.

You have successfully shared your health data from Berkshire Health Systems to the *All of Us Research Program*.

We will periodically access your health data from Berkshire Health Systems.

You can stop sharing data with the *All of Us Research Program* at any time by visiting your profile.

[Return To Profile](#)

Electronic Health Record Data Sharing Not Completed

We were unable to access your health records.

If you need help, call 1 (844) 842 2855 or email help@joinallofus.org.

[Try Again](#)

[Return to Profile](#)

Research App – Cancel Sharing

Electronic Health Record Data

Share my electronic health record (EHR) data ☒ On ☐ Off

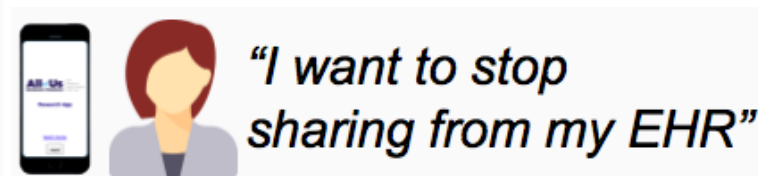
Shared Sync for Science Medical Data

We've connected to your electronic health records.
You're sharing your data from:

Fairfax Family Practice (SMART)

remove

[+ add a new health care provider](#)



Are you sure?

You will stop sharing your data from {insert hospital}, but data you shared before will still be available to researchers.

Cancel

Remove

SYNC FOR SCIENCE

Demonstration
of patient
workflow for
data sharing

TRY IT

Sync for Science Demonstration

This demo shows how S4S helps patients share clinical data with researchers. The public-facing components are:

demo portal

a mock EHR "portal" where a patient can sign in and make the decision to share data with an app

demo app

a mock research application where the patient can share EHR data

These components are available as part of an open-source reference implementation at: <https://github.com/sync-for-science/reference-stack-docker> .

[Demo](#)

S4S Standards – SMART on FHIR (OAuth 2.0 + FHIR)

- **OAuth 2.0**

- » Security authorization standard
- » Modern web technology
- » Helps users delegate permissions to apps, e.g., to share data



- **FHIR (Fast Healthcare Interoperability Resources)**

- » Clinical data standard
- » Modern web technology
- » Breaks data into "Resources" like patient, condition, immunization



Initial S4S Data Scope – Common Clinical Data Set (CCDS)

- Patient demographics (name, birthdate, race, ethnicity, ...)
- Allergies and intolerances
- Immunizations
- Lab results
- Medications (administrations, dispensations, orders, statements)
- Patient documents (*)
- Problems
- Procedures
- Smoking status
- Vital signs



S4S Test Suite – Verifying EHR Compliance with S4S

S4S Test Suite ✓ **Tests Complete!**

Feature: Allergies and intolerances ✗

Scenario: Correct resource type

Scenario: Resources are valid FHIR content

Scenario: Results exist

Scenario: Resources have ids

Scenario: All references will resolve

Scenario: All the codes are valid

Scenario: Resources fulfill the Argonaut Allergies profile

App asks for authorization

App receives authorization request

App exchanges authorization code for access token

App uses a refresh token to obtain a new access token

User revokes authorization

Tests complete!

103 scenarios passed, 0 failed, 13 skipped, 7 warnings

376 steps passed, 7 failed, 58 skipped

Coding systems

System	Count	Errors	Recognized
http://hl7.org	94	0	✓
http://hl7.org/fhir/observation-category	47	0	✓
urn:oid:1.2.840.114350.1.13.0.1.7.4.398296.6000	18	0	✗
urn:oid:1.2.840.114350.1.13.0.1.7.4.398296.6001	18	0	✗
http://named.info/sct	13	0	✓
urn:oid:1.2.840.114350.1.13.0.1.7.2.857366	10	0	✗
http://www.nlm.nih.gov/research/umls/rxnorm	9	0	✓
http://argonaut.fhir.org/extension-codes	7	0	✗
urn:oid:2.16.840.1.113883.6.90	6	0	✗
http://hl7.org/fhir/condition-category	6	0	✓
http://hl7.org/fhir/condition	2	0	✓
http://hl7.org/fhir/condition	2	0	✗
http://www.nlm.nih.gov/research/umls/rxnorm	2	0	✗
urn:oid:1.2.840.114350.1.13.0.1.7.2.895590	1	0	✗
http://www.nlm.nih.gov/research/umls/rxnorm	1	0	✓
http://hl7.org/fhir/ValueSet/allergy-status	1	0	✗
2.16.840.1.113883.6.90	1	0	✗
2.16.840.1.113883.5.104	1	0	✗
urn:oid:2.16.840.1.113883.6.90	1	0	✗
urn:oid:1.2.840.114350.1.13.0.1.7.18.766076.4030	1	0	✗
urn:oid:1.2.840.114350.1.13.0.1.7.18.766076.4040	1	0	✗
http://hl7.org/fhir/hdtt	1	0	✗

Demo

Big Picture – Long-Term Strengths of S4S

- **Quick** Setting up access is fast and easy for participants
- **Cheap** Incremental cost for sharing EHR data closer to \$0.01 than \$100
- **Ubiquitous** Widely available to participants across the country
- **Transparent** Participants understand what is happening & which data are shared
- **Deterministic** Providers/researchers do not need to guess or match patients::participants
- **Structured data** Common structures and vocabularies for core clinical, imaging, payor, lab data
- **Free-text data** Access to free-text notes and other unstructured clinical components

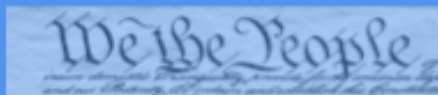
Growing the S4S Platform – Not Just Research

Medical Research



Person-Centered Apps

Consumer Health-Related



Sync for Science

Patients exercising their HIPAA rights to share their health data as they choose

**Provider
Systems**

**EHR
Aggregator**

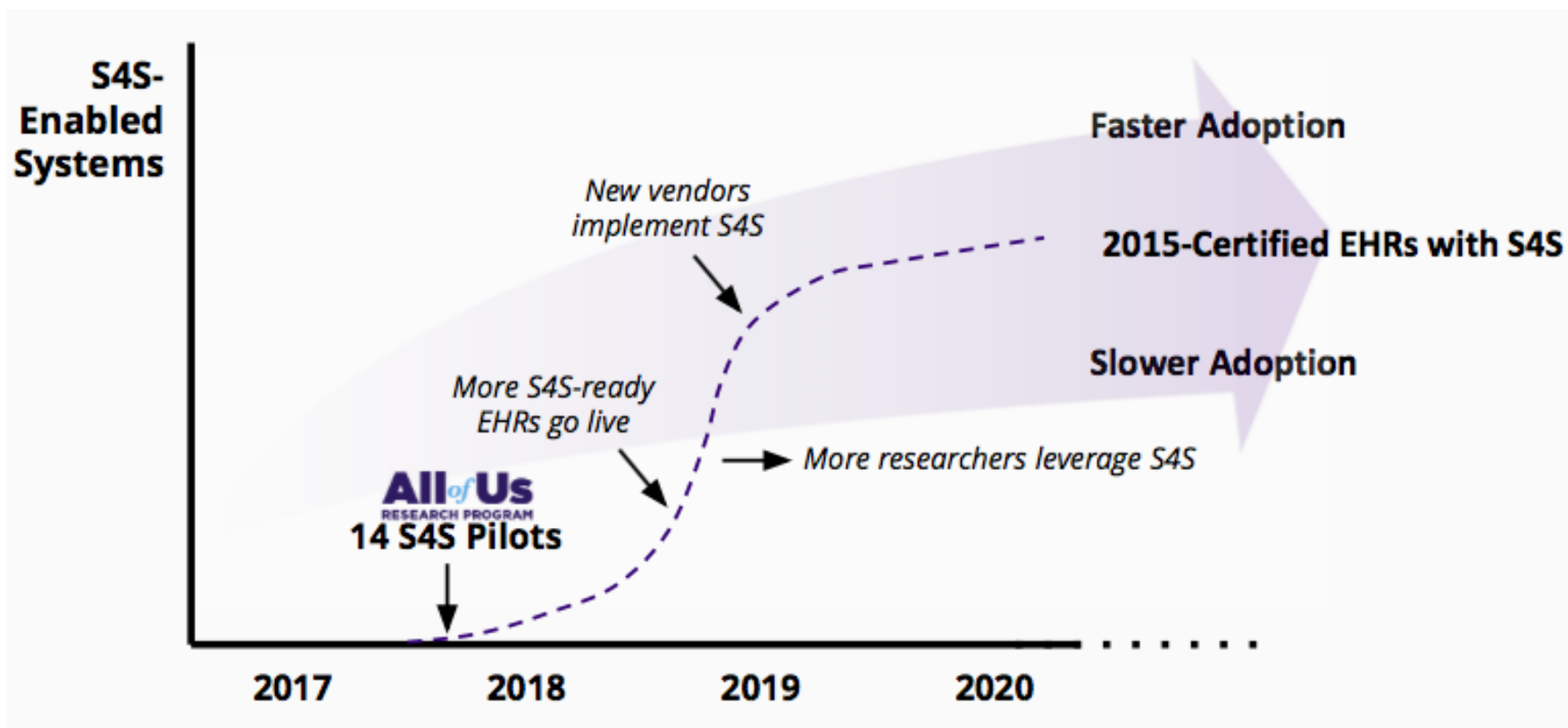
**Patient
Portals**

**Payor
Systems**

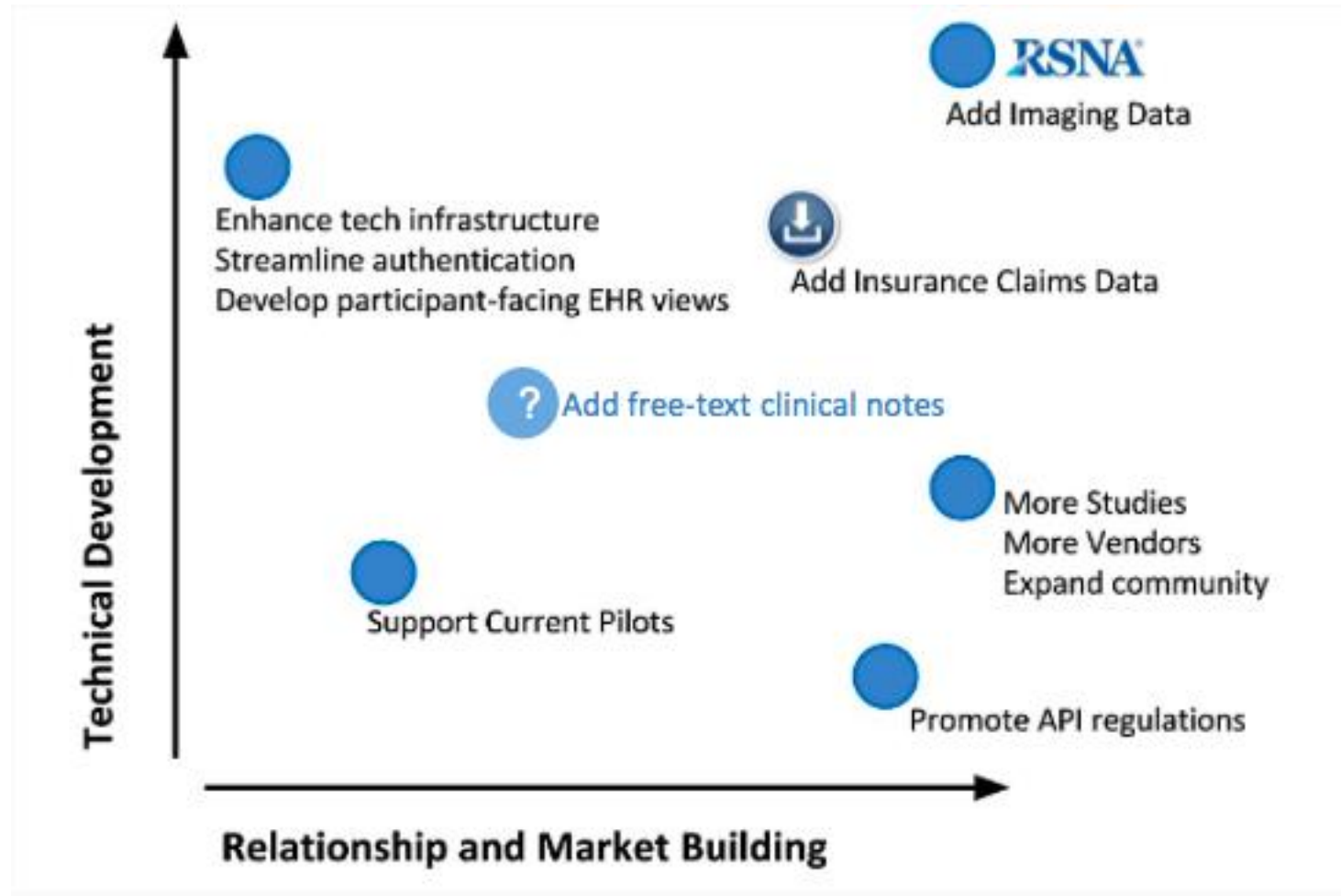
**Imaging
Systems**

**Lab
Systems**

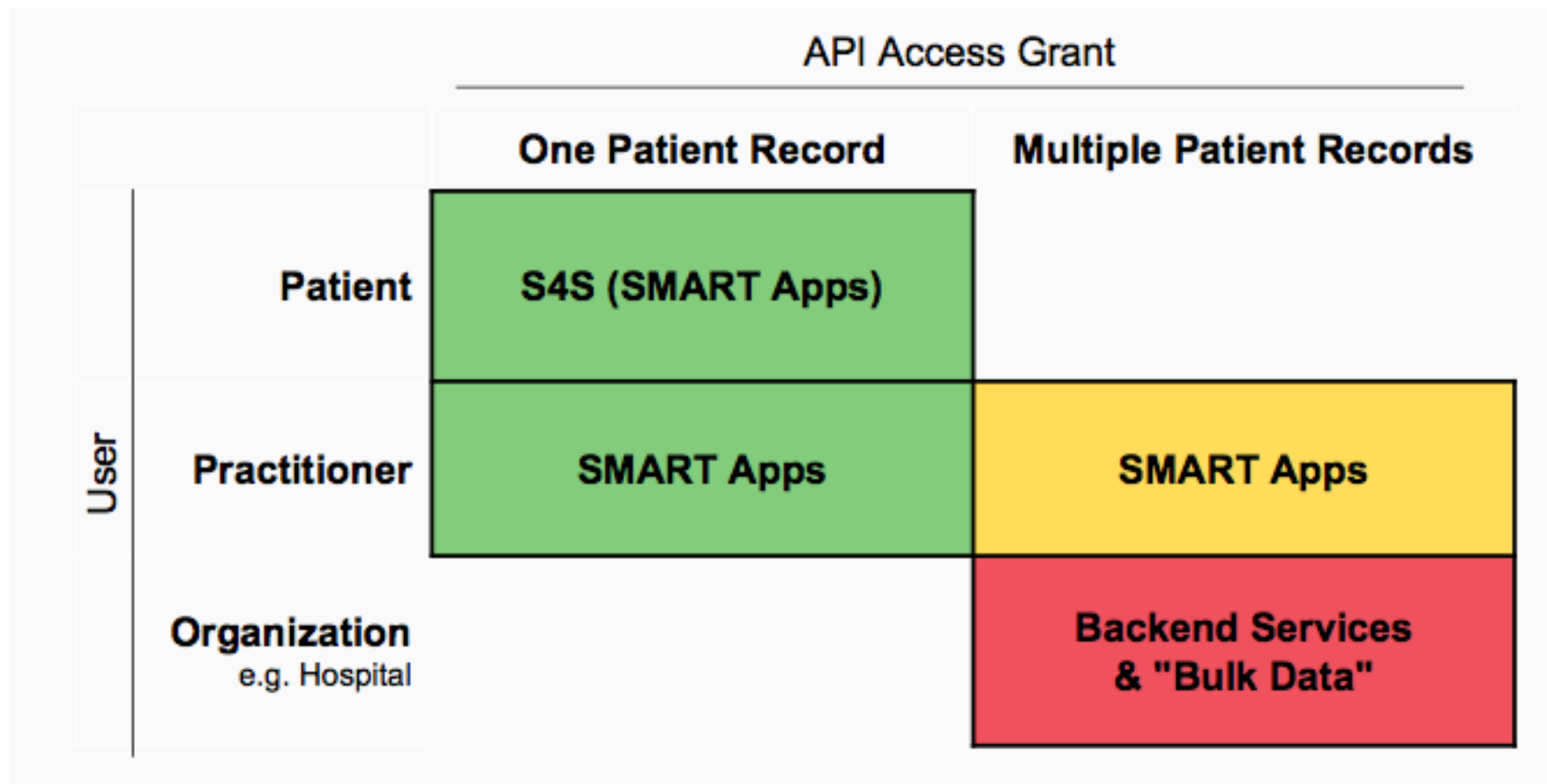
Growing the S4S Presence – Adoption Dynamics



Growing the S4S Platform in Year 2 – More Partners, More Data



S4S in the Interoperability Landscape

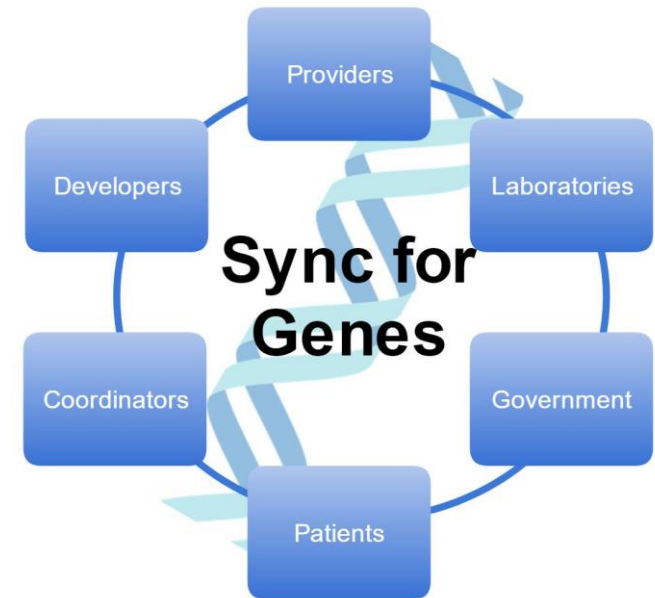


Sync for Science Application Programming Interface (API) Privacy and Security

- Technical and administrative testing, analysis, and assessment of APIs developed under the S4S Pilot Project
- Outlines Key Privacy and Security Considerations for API implementation
 - “... as long as healthcare APIs are implemented with appropriate privacy and security safeguards in place, APIs can add value to individual-directed sharing of health information.”
- Report: <https://www.healthit.gov/sites/default/files/privacy-security-api.pdf>

Sync for Genes

- **LAUNCHED:** 2016
- **MISSION:** To standardize the sharing of genomic information between laboratories, providers, patients, and researchers
- **CONCLUDED** Phase 1: Standardizing Genomic Data
 - » Updated HL7 FHIR clinical genomic profile and genomic specifications
 - » ***Final Report:***
https://www.healthit.gov/sites/default/files/sync_for_genes_report_november_2017.pdf



Sync For Genes Phase 2 Pilot Focus Areas

- **Utah Department of Health: Newborn Screening**
 - » Share data with healthcare providers in real-time for optimal clinical care and outcomes for at-risk and vulnerable newborns
- **Weill Cornell Medicine: Cancer Genomic Decision Support**
 - » Convert discrete genomic results to FHIR® and pair this data with a physician facing app that provides supplemental knowledge of variant results to pilot clinical decision support scenarios in cancer genomics
- **National Marrow Donor Program (NMDP): Patient/Donor Matching**
 - » Convert existing Next Generation Sequencing (NGS)-based human leukocyte antigen HLA genotyping information to FHIR® without losing completeness of the data
- **Lehigh Valley Health Network (LVHN): Pharmacogenomics**
 - » Leverage genomic content for diagnosis, care management, and pharmacogenomics to support the advanced cardiology program in their newborn screening program

Testing the HL7® FHIR® Specifications

- HL7 FHIR Connectathon Testing Goals
- Expected outcomes
 - » Identify what types of local modifications (customizations) pilot sites made in both the mapping and exchange activities
 - » Refine the FHIR® Clinical Genomics resource by identifying gaps in the specification

Identified Challenges and Gaps

- **Genomic Diagnostic Reports**

- » Do not always contain the same information
- » Different use cases have unique data needs
- » Exploring how to handle large raw data files such as variant call format (VCF)

- **Industry Wide**

- » Data modeling and semantics
- » Data provenance
- » Privacy and security of sensitive data
- » Ethical issues regarding the use of genomic data
- » Storage of the large data files
- » Patient access to their genomic data
- » Cost of integration

Next Steps

- Phase 2 underway
 - » Preparing for the HL7 Connectathon in January 2019
 - » Project will complete in Spring 2019
- Semantic Modeling Project underway
 - » Project will complete in Summer 2019

Advancing Standards for Precision Medicine

- **LAUNCHED:**
September 2018
- **MISSION:** To advance standards development and use for sensor and social determinants of health data
- Collaboration with [Open mHealth](#)



21st Century Cures Act

- Provides resources to expand the Precision Medicine Initiative, find cures and better tailor treatments to the individual patient needs
- ONC is working to implement Cures Act provisions by:
 - » Improving the flow and exchange of electronic health information
 - » Enhancing the usability, accessibility, and privacy and security of health IT

The Path to Personalized Medicine (2010)

“When the federal government created the national highway system, it did not tell people where to drive — it built the roads and set the standards for safety. Those investments supported a revolution in transportation, commerce, and personal mobility. **We are now building a national highway system for personalized medicine, with substantial investments in infrastructure and standards.** We look forward to doctors' and patients' navigating these roads to better outcomes and better health.”

-Margaret A. Hamburg, M.D., and Francis S. Collins, M.D., Ph.D.

<https://www.nejm.org/doi/full/10.1056/NEJMp1006304#t=article>



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