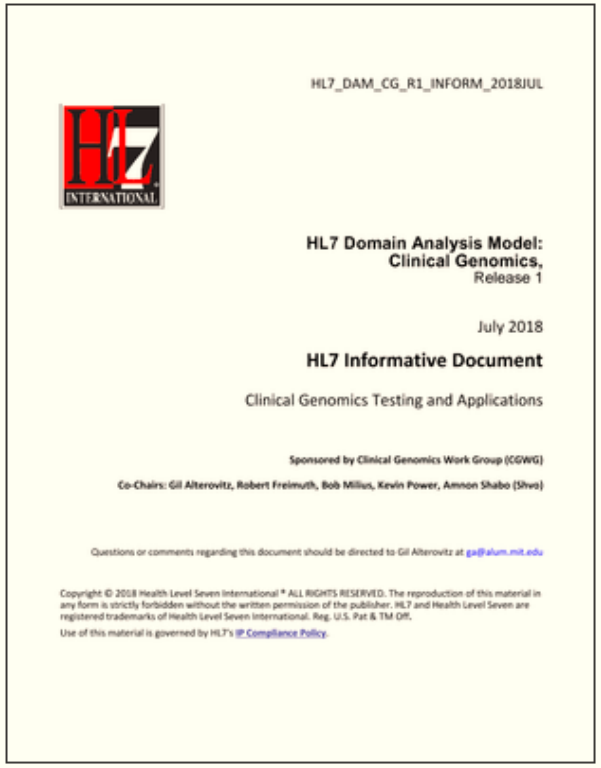
Intelligence Augmentation at its finest:
How to use genomic-based clinical informatics to change the clinical culture that supports genomics

Gil Alterovitz, PhD, FACMI, FAMIA
Director, Biomedical Cybernetics Laboratory
Assistant Professor, Harvard Medical School

HARVARD MEDICAL SCHOOL

BWH

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



HL7_DAM_CG_R1_INFORM_2018JUL

HL7 Domain Analysis Model:
Clinical Genomics,
Release 1

July 2018

HL7 Informative Document

Clinical Genomics Testing and Applications

Sponsored by Clinical Genomics Work Group (CGWG)

Co-Chairs: Gil Alterovitz, Robert Freimuth, Bob Milius, Kevin Power, Amnon Shabo (Shvo)

Questions or comments regarding this document should be directed to Gil Alterovitz at ga@alum.mit.edu

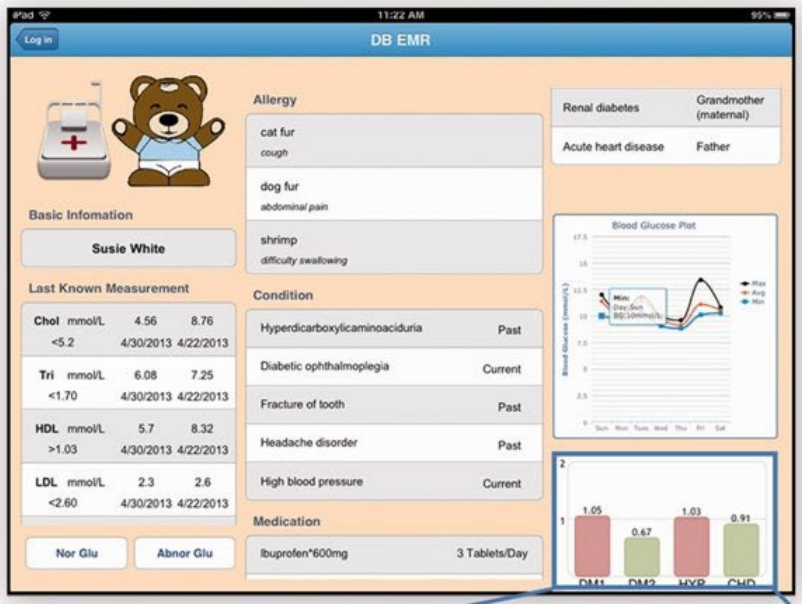
Copyright © 2018 Health Level Seven International * ALL RIGHTS RESERVED. The reproduction of this material in any form is strictly forbidden without the written permission of the publisher. HL7 and Health Level Seven are registered trademarks of Health Level Seven International. Reg. U.S. Pat & TM Off.
Use of this material is governed by HL7's IP Compliance Policy.

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

http://blog.hl7.org/hl7-publishes-valuable-clinical-genomics-domain-analysis-model

Apps

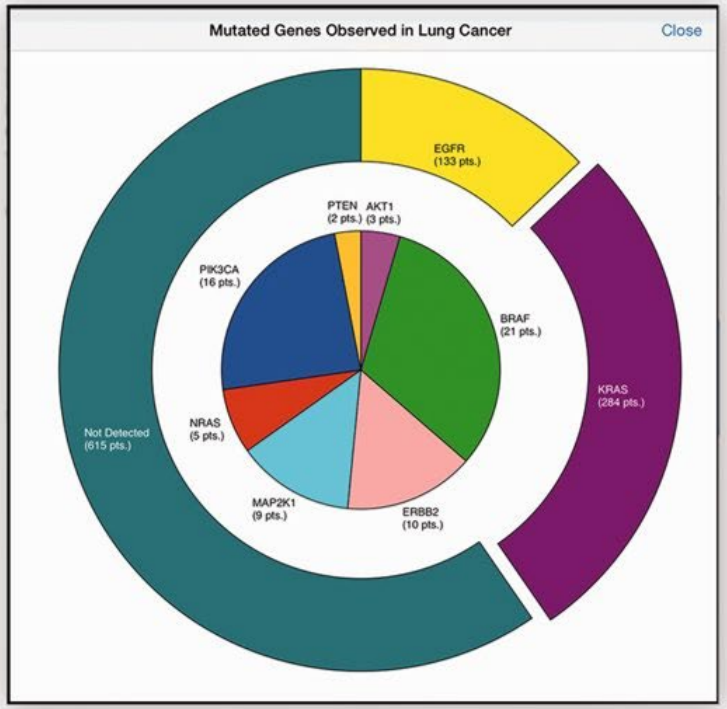
A



B

SMART Genomics Advisor

C



SMART Precision Cancer Medicine

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

iPad 11:03 AM 83%

Patient Search

Patient name

[redacted] (MRN: [redacted])

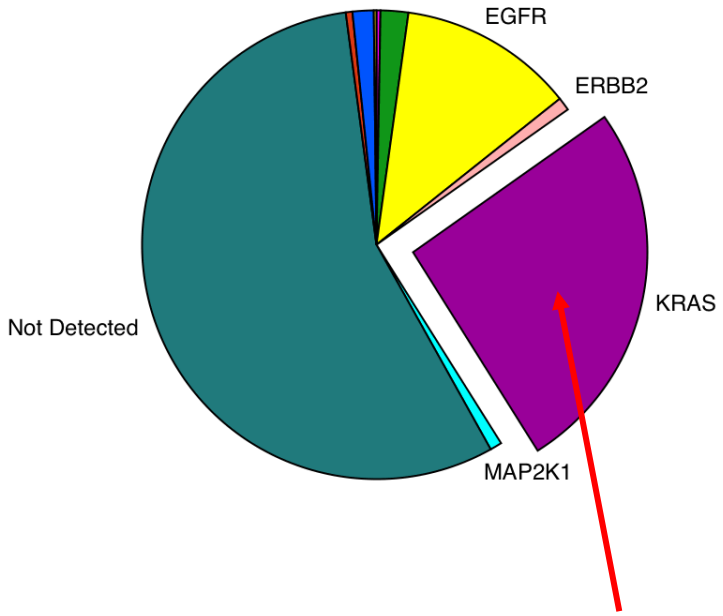
Male, [redacted]
Diagnosis: Lung Cancer
Mutation: KRAS G12C

Date of birth

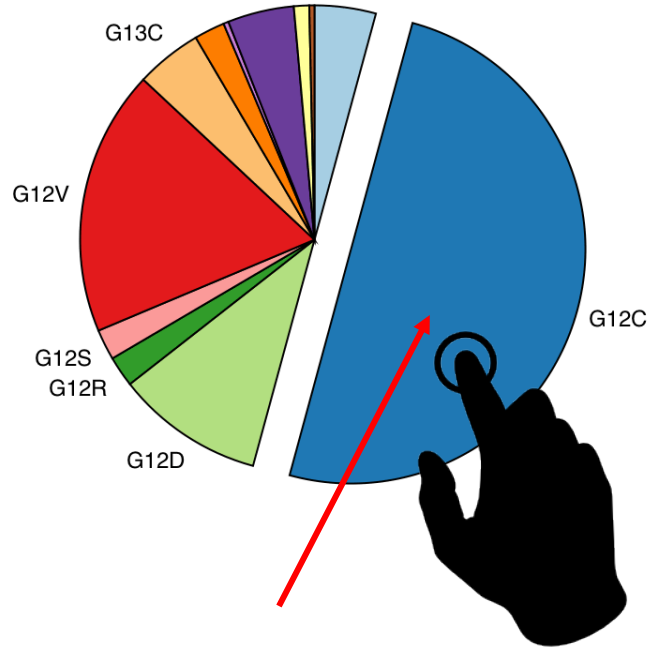
Medical record number

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

Mutated Genes Observed in Lung Cancer



Observed Variants in Lung Cancer Patients with KRAS Mutation



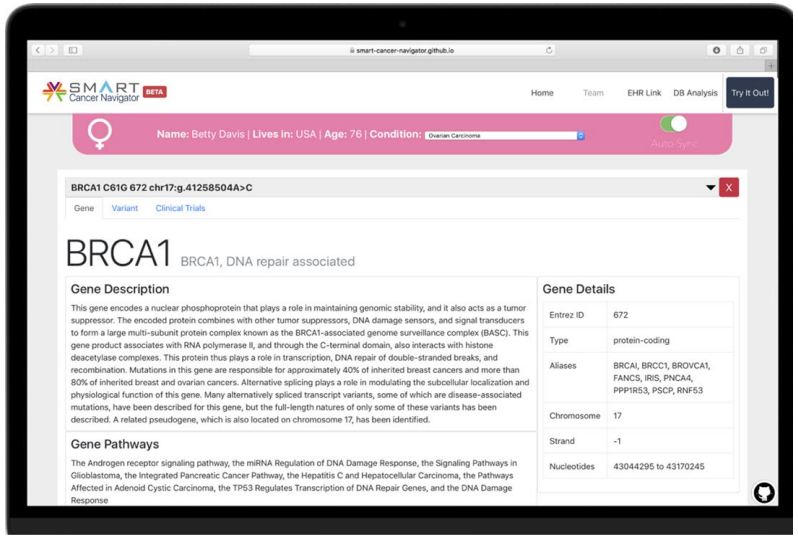
For a specific gene mutation in a specific lung cancer patient, show information for most common KRAS in ***lung cancer*** populations

MRN Search Patient Search About

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.

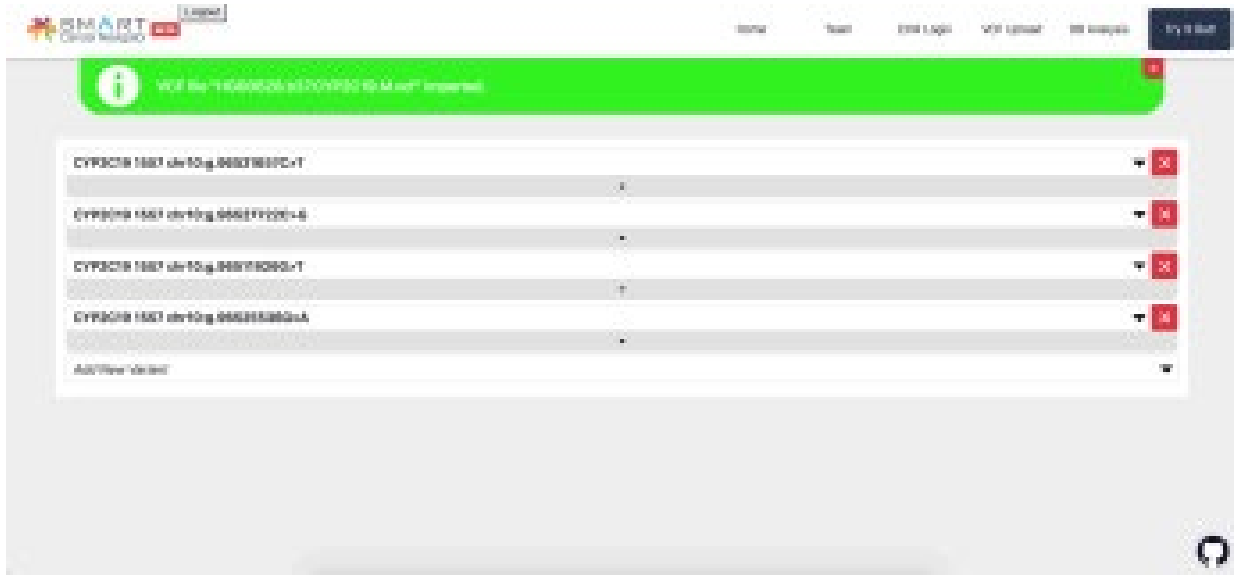


https://smart-cancer-navigator.github.io

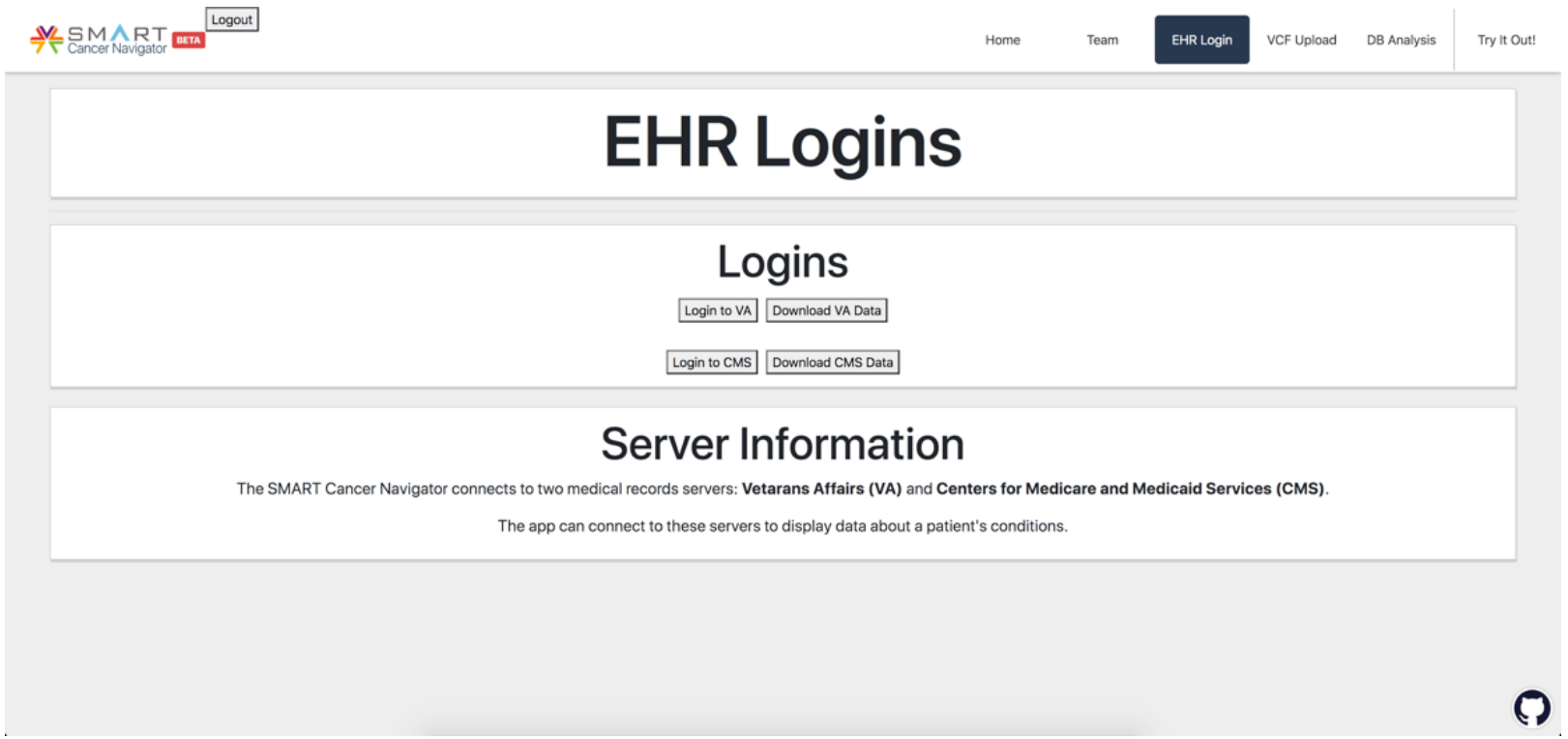
J Warner, I Prasad, M Bennett, M Arniella, A Beeghly-Fadiel, K Mandl, and G Alterovitz, SMART Cancer Navigator: A Framework for Implementing ASCO Workshop Recommendations to Enable Precision Cancer Medicine, JCO Precision Oncology 2018:2, 1-14

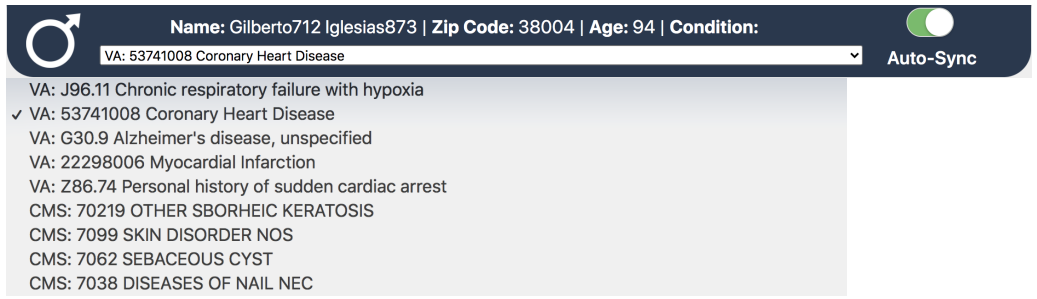
SMART on FHIR®©

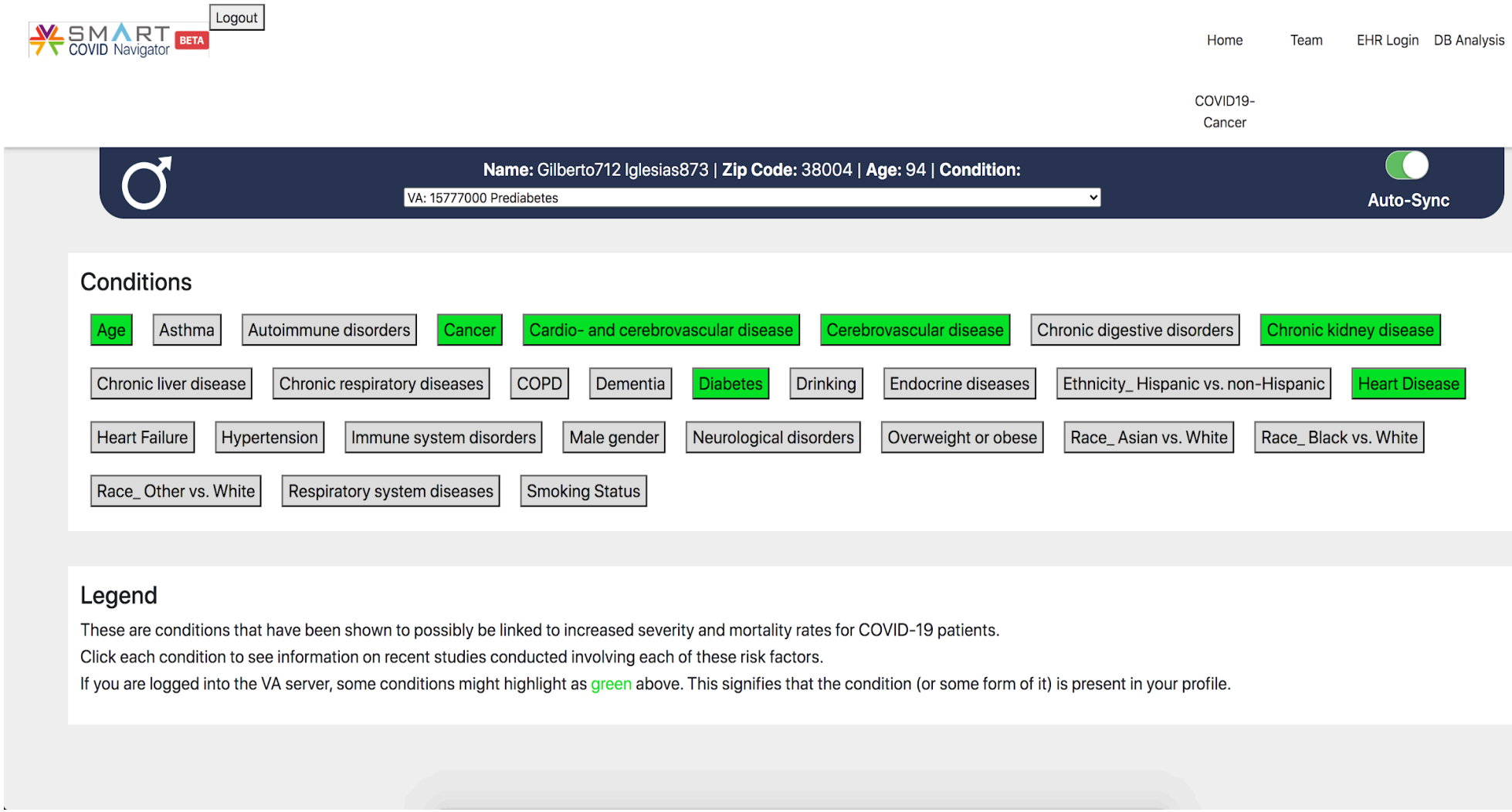
VCF Upload
VCF Information

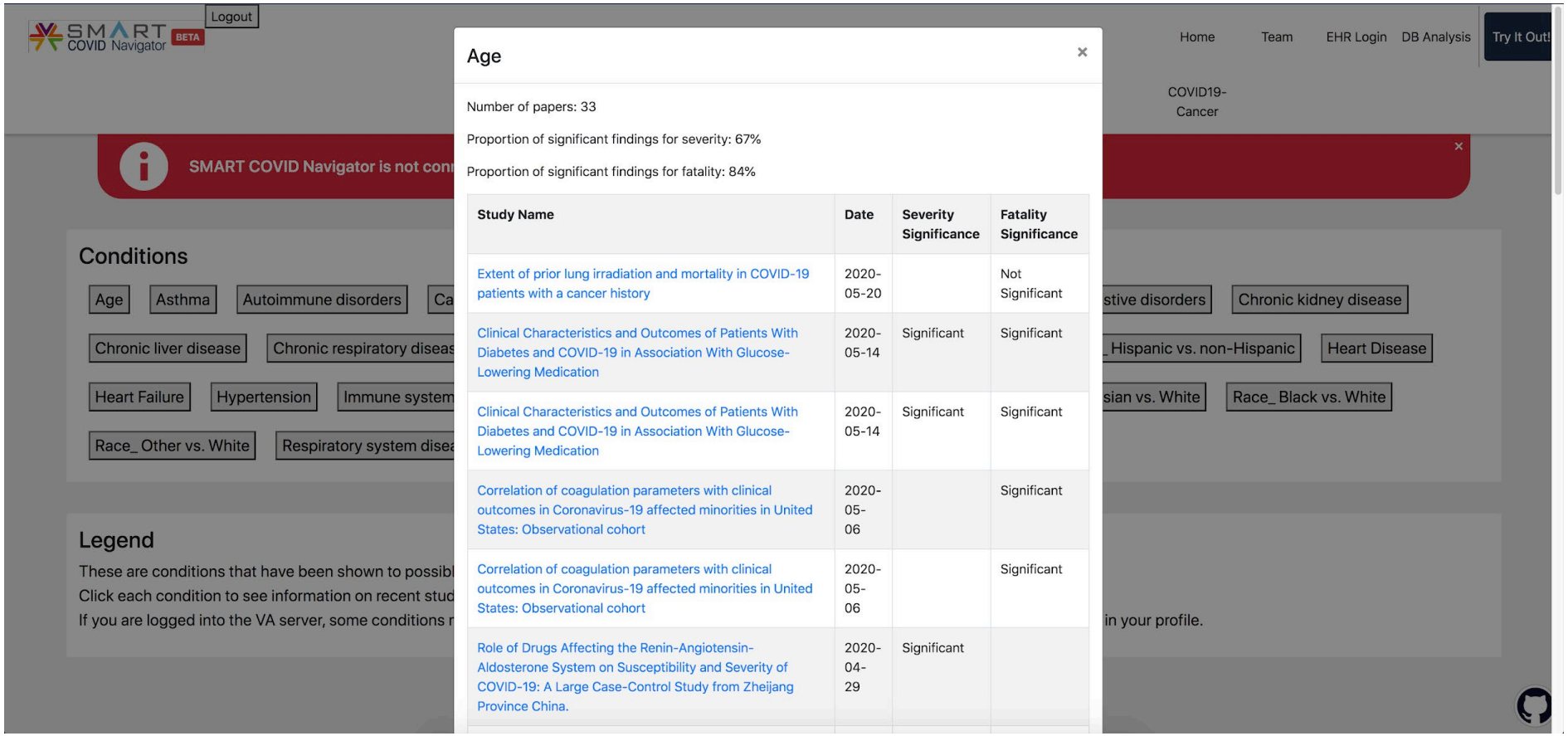


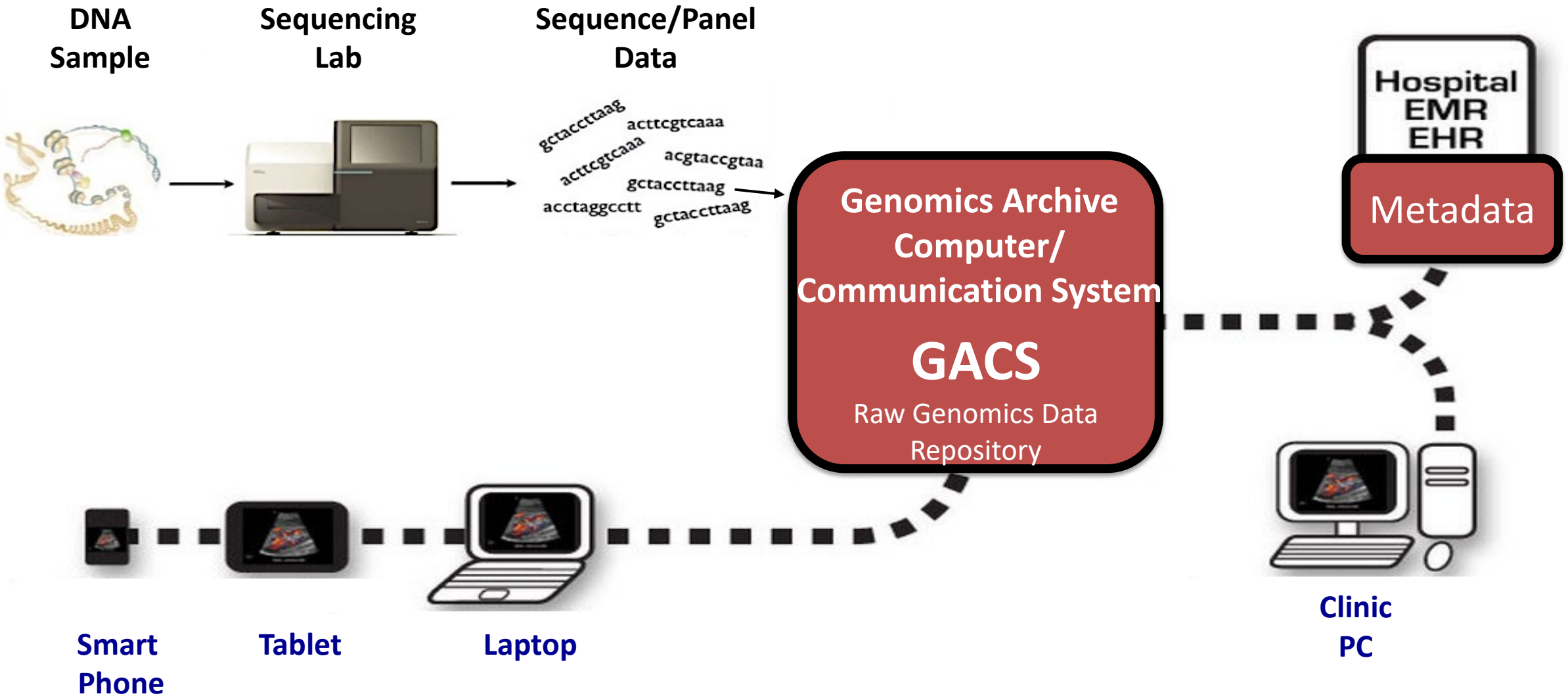
COVID19 Navigator



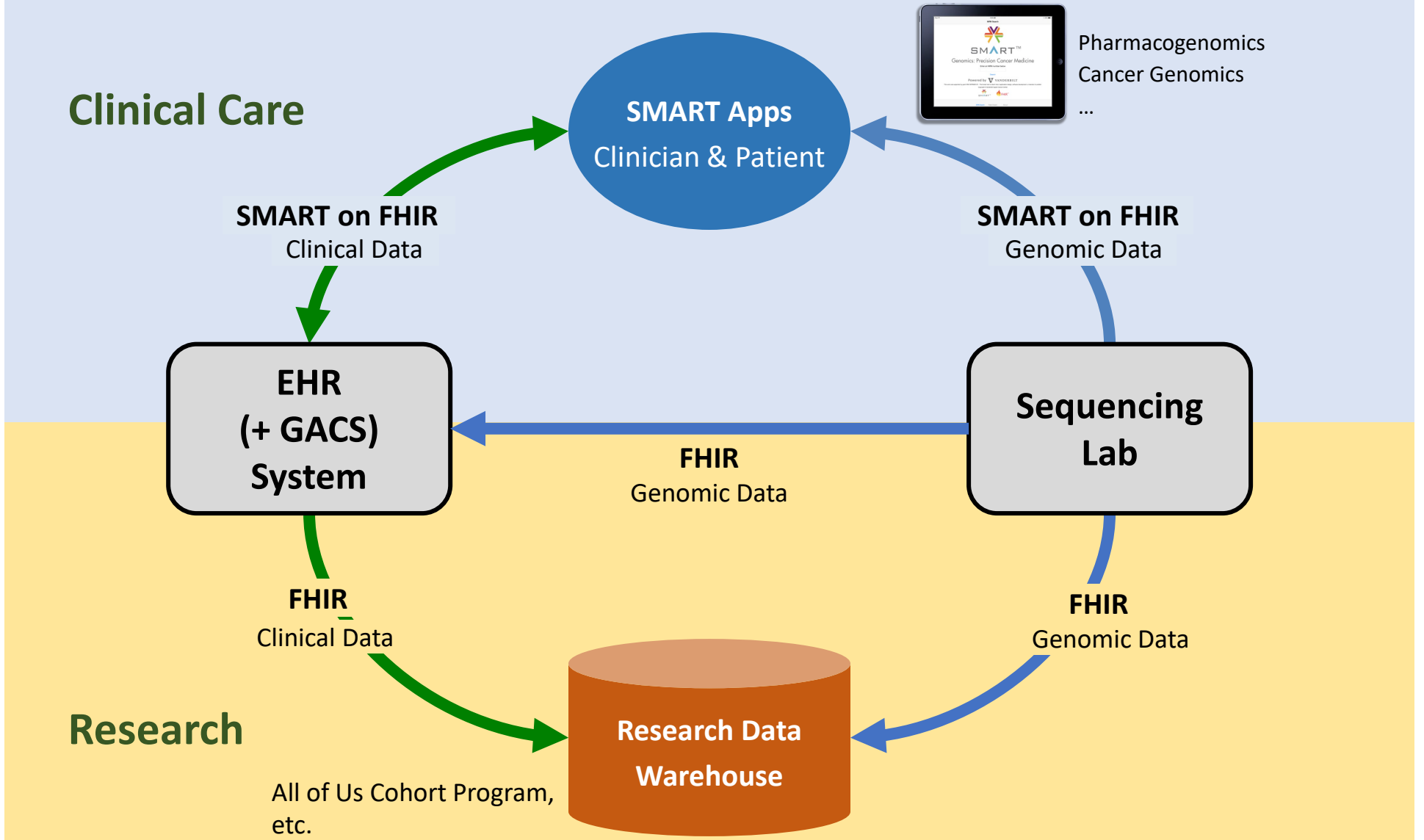



SMART COVID Navigator BETA
Logout
Home
Team
EHR Login
DB Analysis
Try It Out!
COVID19-Cancer
Name: Gilberto712 Iglesias873 | Zip Code: 38004 | Age: 94 | Condition:
VA: 15777000 Prediabetes
Auto-Sync
Conditions
Age
Asthma
Autoimmune disorders
Cancer
Cardio- and cerebrovascular disease
Cerebrovascular disease
Chronic digestive disorders
Chronic kidney disease
Chronic liver disease
Chronic respiratory diseases
COPD
Dementia
Diabetes
Drinking
Endocrine diseases
Ethnicity_ Hispanic vs. non-Hispanic
Heart Disease
Heart Failure
Hypertension
Immune system disorders
Male gender
Neurological disorders
Overweight or obese
Race_ Asian vs. White
Race_ Black vs. White
Race_ Other vs. White
Respiratory system diseases
Smoking Status
Legend
These are conditions that have been shown to possibly be linked to increased severity and mortality rates for COVID-19 patients.
Click each condition to see information on recent studies conducted involving each of these risk factors.
If you are logged into the VA server, some conditions might highlight as green above. This signifies that the condition (or some form of it) is present in your profile.


SMART COVID Navigator BETA
Logout
Home
Team
EHR Login
DB Analysis
Try It Out!
COVID19-Cancer
SMART COVID Navigator is not con
Conditions
Age
Asthma
Autoimmune disorders
Chronic kidney disease
Chronic liver disease
Chronic respiratory disea
Hispanic vs. non-Hispanic
Heart Disease
Heart Failure
Hypertension
Immune system
sian vs. White
Race_ Black vs. White
Race_ Other vs. White
Respiratory system disea
Legend
These are conditions that have been shown to possibl
Click each condition to see information on recent stud
If you are logged into the VA server, some conditions r
in your profile.
Age
Number of papers: 33
Proportion of significant findings for severity: 67%
Proportion of significant findings for fatality: 84%
Study Name
Date
Severity Significance
Fatality Significance
Extent of prior lung irradiation and mortality in COVID-19 patients with a cancer history
2020-05-20
Not Significant
Clinical Characteristics and Outcomes of Patients With Diabetes and COVID-19 in Association With Glucose-Lowering Medication
2020-05-14
Significant
Significant
Clinical Characteristics and Outcomes of Patients With Diabetes and COVID-19 in Association With Glucose-Lowering Medication
2020-05-14
Significant
Significant
Correlation of coagulation parameters with clinical outcomes in Coronavirus-19 affected minorities in United States: Observational cohort
2020-05-06
Significant
Correlation of coagulation parameters with clinical outcomes in Coronavirus-19 affected minorities in United States: Observational cohort
2020-05-06
Significant
Role of Drugs Affecting the Renin-Angiotensin-Aldosterone System on Susceptibility and Severity of COVID-19: A Large Case-Control Study from Zheijang Province China.
2020-04-29
Significant


DNA Sample
Sequencing Lab
Sequence/Panel Data
gctaccttaag
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

Standard for Clinical Care & Research



Individual

Smart Contract

Data estate

Shared (e.g. family, other individuals, research, etc)

Gil Alterovitz
ga@alum.mit.edu

LinkedIn: