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

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Assistant Professor, Harvard Medical School
Use Cases:
“HL7 Domain Analysis Model: Clinical Sequencing”

- 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
For a specific gene mutation in a specific lung cancer patient, show information for most common KRAS in **lung cancer** populations.
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
### VCF Upload

**Uploaded file:** `H0038681-CYP2C19.txt`

### VCF Information

The SMART Cancer Navigator can accept Variant Call Format (VCF) files and display them in an organized fashion, allowing patients to easily inspect any gene variants that they may have.

To import your VCF file, click the "Upload" button. Then, click the "Try it Out!" tab to see detailed information regarding your variants.

To view your variants from the app, simply click the "Login" button.

Please click here to see how to properly format the VCF file name.

<table>
<thead>
<tr>
<th>Gene</th>
<th>Description</th>
<th>Variation</th>
</tr>
</thead>
<tbody>
<tr>
<td>CYP2C19</td>
<td>Activity</td>
<td>1587 C/T</td>
</tr>
<tr>
<td>CYP2C19</td>
<td>Activity</td>
<td>1587 C/C</td>
</tr>
<tr>
<td>CYP2C19</td>
<td>Activity</td>
<td>1587 T/T</td>
</tr>
<tr>
<td>CYP2C19</td>
<td>Activity</td>
<td>1587 T/C</td>
</tr>
<tr>
<td>CYP2C19</td>
<td>Activity</td>
<td>1587 G/A</td>
</tr>
</tbody>
</table>

*Add new variant*
COVID19 Navigator

EHR Logins

Logins
- Login to VA
- Download VA Data
- Login to CMS
- Download CMS Data

Server Information
The SMART Cancer Navigator connects to two medical records servers: Veterans Affairs (VA) and Centers for Medicare and Medicaid Services (CMS).

The app can connect to these servers to display data about a patient’s conditions.

Name: Gilberto712 | Zip Code: 39004 | Age: 94 | Condition:
- VA: J96.11 Chronic respiratory failure with hypoxia
- VA: 5374008 Coronary Heart Disease
- VA: G30.9 Alzheimer's disease, unspecified
- VA: 22298006 Myocardial infarction
- VA: 2296.74 Personal history of sudden cardiac arrest
- CMS: 70219 OTHER SIBORHEIC KERATOSIS
- CMS: 70999 SKIN DISORDER NOS
- CMS: 7082 SEBACEOUS CYST
- CMS: 7038 DISEASES OF NAIL NEC
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.
## Conditions

<table>
<thead>
<tr>
<th>Category</th>
<th>Condition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age</td>
<td>Asthma</td>
</tr>
<tr>
<td>Autoimmune disorders</td>
<td>Chronic liver disease</td>
</tr>
<tr>
<td>Chronic respiratory disease</td>
<td>Heart Failure</td>
</tr>
<tr>
<td>Hypertension</td>
<td>Immune system</td>
</tr>
<tr>
<td>Race_Other vs. White</td>
<td>Respiratory system disease</td>
</tr>
</tbody>
</table>

## Legend

These are conditions that have been shown to possibly increase the risk of severe outcomes in COVID-19 patients. Click each condition to see information on recent studies. If you are logged into the VA server, some conditions may be restricted.

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### Age

- **Number of papers:** 33
- **Proportion of significant findings for severity:** 67%
- **Proportion of significant findings for fatality:** 84%

<table>
<thead>
<tr>
<th>Study Name</th>
<th>Date</th>
<th>Severity Significance</th>
<th>Fatality Significance</th>
</tr>
</thead>
<tbody>
<tr>
<td>Extent of prior lung irradiation and mortality in COVID-19 patients with a cancer history</td>
<td>2020-06-20</td>
<td>Not Significant</td>
<td>Not Significant</td>
</tr>
<tr>
<td>Clinical Characteristics and Outcomes of Patients With Diabetes and COVID-19 in Association With Glucose-Lowering Medication</td>
<td>2020-06-14</td>
<td>Significant</td>
<td>Significant</td>
</tr>
<tr>
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<td>Significant</td>
<td>Significant</td>
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<tr>
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<td>Role of Drugs Affecting the Renin-Angiotensin-Aldosterone System on Susceptibility and Severity of COVID-19. A Large Case-Control Study from Zhejiang Province China</td>
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<td>Significant</td>
<td></td>
</tr>
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</table>
Standard for Clinical Care & Research

Clinical Care

SMART on FHIR
Clinical Data

EHR
(+ GACS)
System

FHIR
Clinical Data

Research

Research Data Warehouse

All of Us Cohort Program, etc.

SMART Apps
Clinician & Patient

FHIR
Genomic Data

Sequencing Lab

FHIR
Genomic Data

Pharmacogenomics
Cancer Genomics
...
Individual

Data estate

Smart Contract

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