FHIR & CDS Hooks support of Pharmacogenomics workflows

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http://www.elimu.io
Our roots

Est. 2005

+  

Est. 2012

=  

Est. 2017
Clinical scenario

Conrad Smart
*Rheumatoid arthritis*

Dr. Good
*Wants to prescribe azathioprine*

Azathioprine (Imuran)
*Will it work? Will it harm?*
Clinical decision support outcomes

- **Full dose ok**: TPMT gene normal on both sides
- **Suggest half dose**: TPMT gene abnormal variant on one side
- **Do not prescribe**: TPMT gene abnormal variant on both sides
- **Need more info**: Genetic test results not found/required
Workflow enablers

FHIR

CDS Hooks®
Clinical Intelligence Platform

1. EHR Rx workflow
2. Terminology Service
3. Data Retrieval Service
4. PGx Rules Engine
5. Information Card
   Suggestion Card
   Launch App (Sapphire)

- FHIR
- CCD
- Genomics
- CDS Hooks Server
- Terminology Service
- PGx Rules Engine
Genetic variant on paternal side

Reference Sequence

Patient’s Sequence

Maternal

Paternal

A₁ B₁ C₁

A₁ B₁ C₁

A₁ B₂ C₁

A₁ B₁ C₁
FHIR representation of genetic variant

`<Observation xmlns="http://hl7.org/fhir">
  <extension url="http://hl7.org/fhir/StructureDefinition/observation-geneticsDNASequenceVariantName">
    <valueCodeableConcept>
      <text value="NM_000367.4(TPMT):c.238G>C (p.Ala80Pro)"/>
    </valueCodeableConcept>
  </extension>
  ...

“HGVS” syntax – a formalism for representing variants

NM_000367.4(TPMT):c.238G>C (p.Ala80Pro)

- Reference sequence
- Gene (optional)
- mRNA variation
- Protein variation (optional)
SMART on FHIR App Builder

Build apps in minutes
Embed your app into workflows
No coding required
Demo