FHIR Genomics Profile Proposal

This project aims to create Fast Healthcare Interoperability Resources (FHIR) profiles for access to specific genetic information as FHIR resource instances.

The Artifacts to be produced as part of this project include:

- Define profiles on selected FHIR resources that define additional search criteria for retrieving those resources.
- Create profiles on selected FHIR resources identifying minimum data and corresponding constraints and value sets for query response.
- Define a FHIR genomics implementation guide that organizes and provides context for the above artifacts.

Profile balloting plans

STU3

Profile Details

The following resource is proposed:

- Sequence

The following profiles are proposed:

- Observation for Genetics
- Diagnostic Report for Genetics
- Diagnostic Order for Genetics
- Family member history for genetics analysis
- Profile for Consensus Sequence Block
- Profile for HLA Genotyping Results

Profiled Resource(s)

The following FHIR resources are profiled:

- Observation
- Sequence
- DiagnosticReport
- DiagnosticOrder
- FamilyMemberHistory
Constraints to be Applied

The following constraints are applied to the FHIR resources as necessary:

- Constrain the cardinality of the data elements within the resource.
- Constrain the mustSupport attribute of the data elements within the resource.
- Define searchParameters as necessary for the resource.
- Constrain vocabularies for the resources as applicable.

Extensions to be Applied

The following extensions are applied to resources as appropriate:

- Clinical genomics specific extensions for the various FHIR resources.

Example Scenarios

- Query Examples for each resource.
- Query Result Examples for each resource.

Scope of coverage

- Subject: patient
- Disciplines: Patient Care, Clinical Research, Public Health
- Delivery Environment: Inpatient, Ambulatory, Community, Emergency, etc.

Realm

Clinical genomics.

Ownership

Owning committee name

Clinical Genomics

Contributing or Reviewing Work Groups

- None

Expected implementations

gForge Users

- Gil Alterovitz
- Gaston Fiore

FHIR Profile Development Project Insight ID

1110

Plans

Timelines

- July 2016

Balloting Requirements

- Ballot with next FHIR DSTU or Normative Edition

Desired Ballot Date

- End 2016