Genomics Study

HL7 WGM
May 11, 2022

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Design Patterns

• "Definition" resources are resources that describe clinical, administrative or other events that can potentially occur. They do so in a manner that is independent of a particular patient or subject and independent of any particular timeframe.

• Any sort of description of an activity that is "desired" where the description is specific as to the subject of the activity and the approximate timing of the activity would be considered a "Request".

• An "event" is any description of an activity that has already taken place or that is currently ongoing. It includes resources that primarily describe the "result" of an activity or what was found (e.g., a condition or observation).
Activity Definition (definition)

• Activity to be performed
• Part of a workflow, protocol, or catalog
• Does not indicate action has been performed or intent to act
• This is an abstract Task, which is a specific instance of an activity
• VERY generic
Plan Definition (definition)

- Defined group of actions to be taken, often with conditional elements or decision points
- Represents workflows, CDS, protocols
- Can contain hierarchical Activity Definitions
- Each action specifies what, when, and whether it should occur
- Too detailed?
Procedure (event)

• Action that is or was performed on or for a patient, or for locations or devices
• Provides summary info about the occurrence of the procedure
• Procedures generate Observations and Diagnostic Reports
• basedOn ServiceRequest | CarePlan
• part of Procedure | Observation
• Subject 1..1 Patient | Device |... (NOT specimen!)
• Report 0..* DxRpt | DocRef – Any report resulting from the procedure
Task

• Activity that can be performed and has been initiated
• Focused on tracking state, not operational details
• partOf Task
• Input, output
Genomic Study
Identifier : identifier
Type : CC
Subject : Patient
startDate : datetime
basedOn : [ServiceRequest | Task]
Endpoint : Endpoint
Note : Annotation
Description : string
ReasonCode : CC
ReasonReference : [Cond|Obs|…]

Observed Variation (profile)
Quality
Confidence
ACMG classification
AMP tier
See Mol Var models

Molecular Variation
See Mol Var models
Mol Var includes names (HGVS, ISCN, HLA, PGx)

Molecular Sequence
See Mol Seq model
“Called” sequence, via Observed Sequence

Genomic Analysis
Identifier : identifier
Subject : Patient
Specimen : identifier
Date : datetime
MethodType : [CC?]
ProtocolDefinition : ActivityDefinition
ProtocolPerformed : Procedure?
GenomicRegionStudied : TBD

Genomic File
Endpoint : endpoint
GeneratedBy : GStudy | GAssay
Date : datetime
Filetype: [CC??] (VCF, BAM, etc)
RegionCalled : TBD

Discussion

Not shown: implication, interpretation, etc
Genomic Analysis Definition
url : uri 0..1
Identifier : Identifier 0..1
Version : string 0..1
Title : string 0..1
ProtocolDefinition : PlanDefinition 0..1
MethodType : CC 0..*
VariationTypeDetected : CC 0..*
GenomicRegionStudied : DocRef 0..*

Bold = reference resource
Normal = from ref resource
Italics = extensions for genomics
Event (Base) «Pattern»

- **identifier**: Identifier [0..*]
- **instantiatesCanonical**: canonical [0..*] «Definition»
- **instantiatesUri**: uri [0..*]
- **basedOn**: Reference [0..*] «Request»
- **partOf**: Reference [0..*] «Event»
- **researchStudy**: Reference [0..*] «ResearchStudy»
- **status**: code [1..1] «EventStatus!»
- **statusReason**: CodeableConcept [0..1]
- **code**: CodeableConcept [0..1]
- **subject**: Reference [1..1] «Patient|Group»
- **encounter**: Reference [0..1] «Encounter»
- **occurrence[x]**: DataType [0..1] «dateTime|Period|Timing»
- **recorded**: dateTime [0..1]
- **reported[x]**: boolean [0..1] «Reference(Patient|RelatedPerson|Practitioner|PractitionerRole|Organization)»
- **location**: Reference [0..1] «Location»
- **reason**: DataType [0..*] «Condition|Observation|DiagnosticReport|DocumentReference»
- **note**: Annotation [0..*]

Performer

- **function**: CodeableConcept [0..1]
- **actor**: Reference [1..1] «Practitioner|PractitionerRole|Organization|CareTeam|Patient|Device|RelatedPerson»
Genomic Analysis Definition
- **url**: uri 0..1
- **Identifier**: Identifier 0..1
- **Version**: string 0..1
- **Title**: string 0..1

**ProtocolDefinition**: PlanDefinition 0..1
- **MethodType**: CC 0..*
- **VariationTypeDetected**: CC 0..*
- **GenomicRegionStudied**: DocRef 0..*

**Genomic Analysis Event**
- **Identifier**: identifier 0..*
- **InstantiatesCanonical**: canonical Def 0..*
- **InstantiatesUri**: uri 0..*
- **Subject**: Patient 0..1
- **Date**: datetime 0..1
- **Note**: Annotation 0..*
- **ProtocolPerformed**: Procedure 0..1
- **Specimen**: identifier | Ref(Specimen) 0..1
- **Title**: string 0..1

**Genomic Study**
- **Identifier**: identifier 0..*
- **Type**: CC 0..1
- **Subject**: Patient 1..1
- **startDate**: datetime 0..1
- **basedOn**: ServiceRequest | Task 0..1
- **Endpoint**: Endpoint 0..1
- **Note**: Annotation 0..1
- **Description**: string 0..1
- **Reason**: CodeableReference 0..*

**Input**
- **File**: Ref(DocRef) 0..1
- **Type**: CC 0..1
- **GeneratedBy**: GStudy | GAnalysisActivity 0..1
- **Subject**: Patient 0..1
- **Role**: CC 0..1

**Output**
- **File**: Ref(DocRef) 0..1
- **Type**: CC 0..1

Explicitly capture called region
Record genome build/ref used for analysis
Input.subjectused to specify roles for comparison analyses