FHIR
ClinicalProfile Resource

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Harold Solbrig
solbrig@jhu.edu

https://github.com/hsolbrig/clinicalprofileexamples/blob/master/ClinicalProfiles.pdf

Goal

Clinical Profiles summarize and demonstrate the features of a population.

14.7.1 Scope and Usage

14.7.1.1 Motivations for a clinical profile registry

Data science thrives when practitioners have open access to data resources and information. In translational science, this ideally spans basic science "omics" data to and including clinical data. However, clinical data by its nature is sensitive and confidential, and can rarely be made freely available outside clinical record systems and controlled research repositories. However, data about clinical data, such as statistical summaries, frequencies, distributions, and relative counts does not contain any personal health information, and when presented about aggregated persons cannot be re-identified. Such summaries comprise the core of clinical profiles.

The vision for a clinical profile registry is to include clinical profiles for diseases and phenotypically similar cohorts that are machine readable and consisting of aggregated statistics and facts about disease behavior derived from patient records. For example, a Fanconi anemia profile would include summary information about serum hemoglobin levels (e.g., mean, standard deviation, median value, and decile cut-points of the distribution) among other clinical variables and covariates across a group of patients with this condition. It is also attractive to consider demographic subsets of a clinical profile e.g., summary information about clinical profile measures by race, sex, and age groups. Correspondingly, for chronic and common conditions such as hypertension, profiles about cohorts of treated persons may be separate from untreated persons. For conditions such as asthma, characteristics of persons in an acute exacerbation would differ materially from those presently asymptomatic.

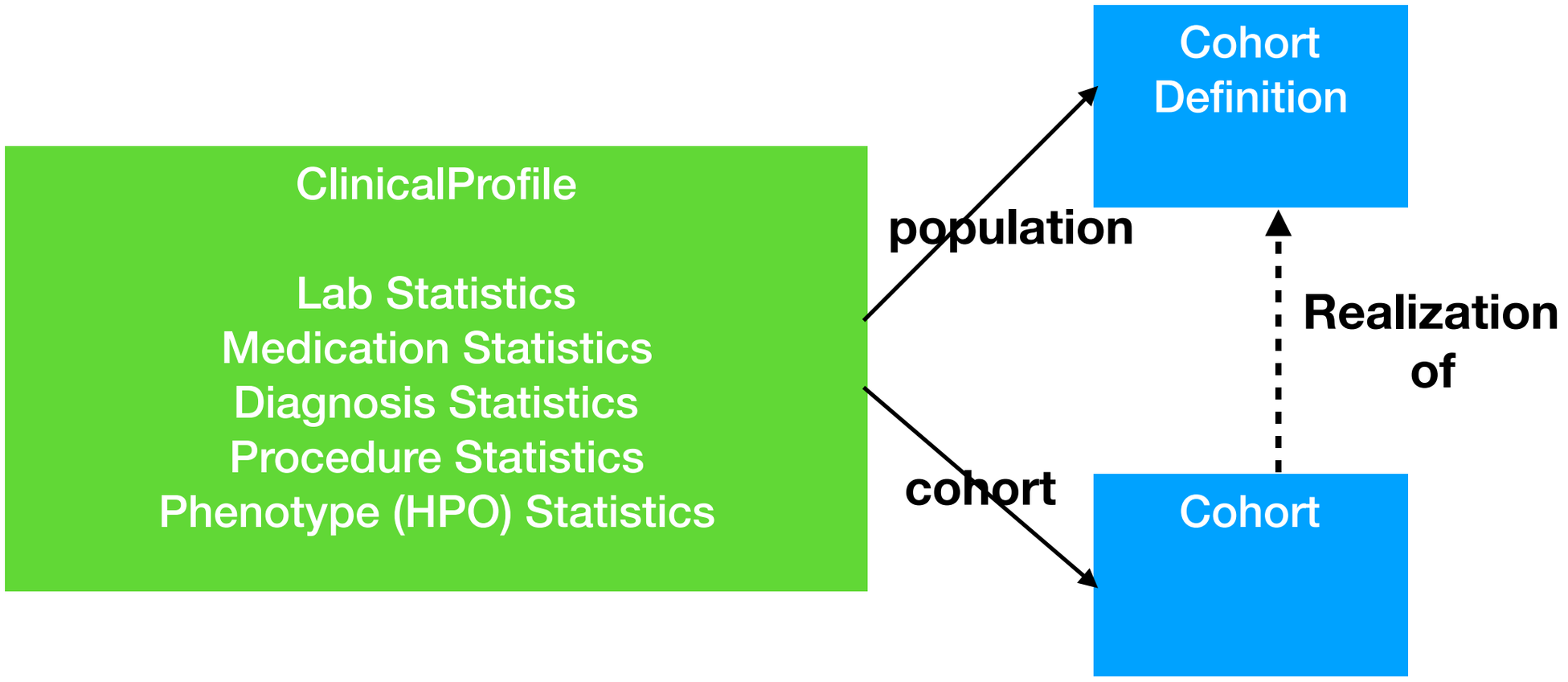
14.7.2 Background and Context

14.7.2.1 Clinical profile data model requirements

As an initial step to defining the model requirements for a clinical profile, we identify core data (demographics, medications, common lab values, and coded information such as co-morbidities and procedures), some condition-specific measures (e.g pulmonary function tests for patients for asthma) that are readily available in electronic health records (EHR). The transition from patient-level data into a clinical profile would require reducing those data into to appropriate summary measures. For example, a comorbidity may be rendered in a clinical profile as the fraction of persons in a population expressing that comorbidity. Additionally, clinical profiles might express a degree of correlation between variables when they are significant, such as hemoglobin and hematocrit.

To make such profiles useful to data scientists, they must be machine readable and conform to a published model. The present Clinical Profile FHIR Resource definition poses an initial model for containing profiles and relevant metadata. Each profile can be called with a FHIR API, and subsequently parsed using standard FHIR renditions such as JSON, XML, and RDF. The model defines the elements, boundaries, and relationships among the parsable data.

Architecture



Population Definition

- Uses FHIR Group Resource
- Provides an “abstract” definition of what belongs in a study population
- Long term goal: descriptive (today) —> formal —> computable
- A population definition is referenced by (potentially) many *Population Descriptions* (Actual language is being discussed in the EBMonFHIR initiative)

Group Definition

8.3.3 Resource Content

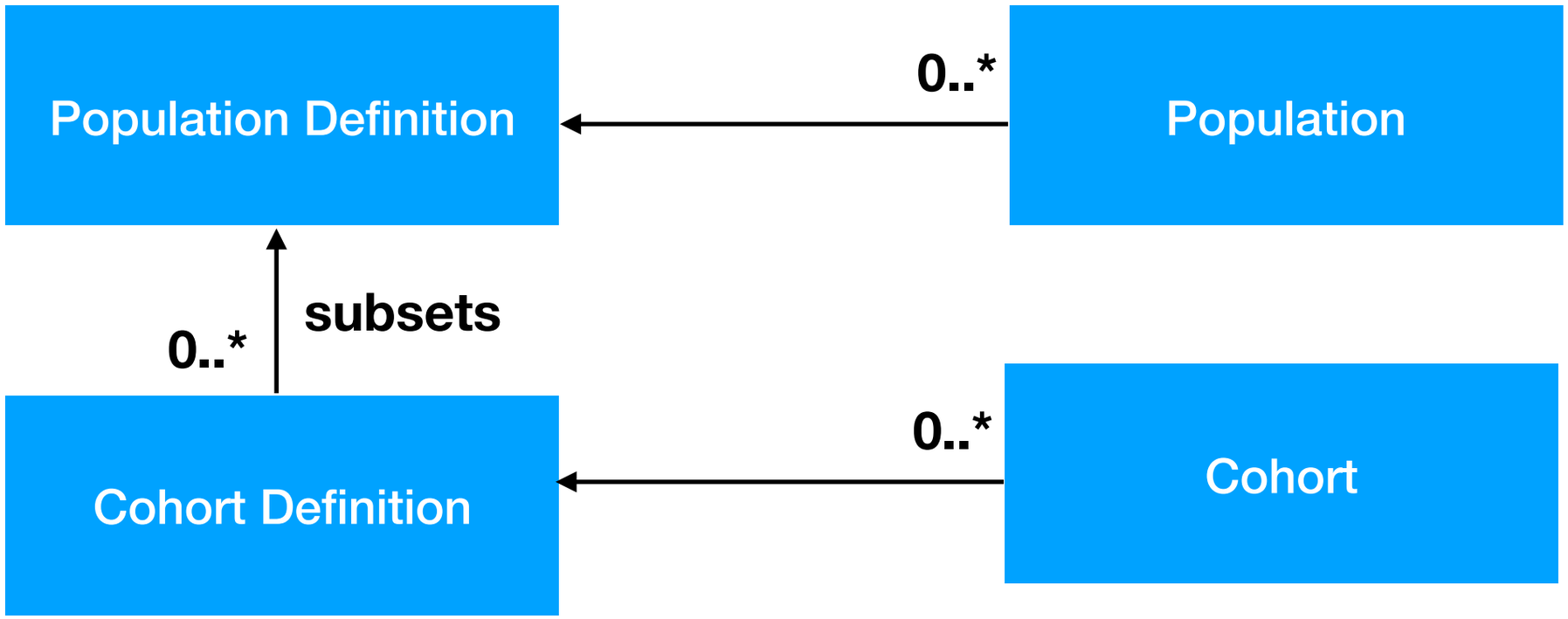
Structure | UML | XML | JSON | Turtle | R3 Diff | All

Structure

Name	Flags	Card.	Type	Description & Constraints
Group	I TU		DomainResource	Group of multiple entities *+ Rule: Can only have members if group is "actual"* Elements defined in Ancestors: id, meta, implicitRules, language, text, contained, extension, modifierExtension
identifier	Σ	0..*	Identifier	Unique id
active	Σ	0..1	boolean	Whether this group's record is in active use
type	Σ	1..1	code	person | animal | practitioner | device | medication | substance GroupType (Required)
actual	Σ I	1..1	boolean	Descriptive or actual
code	Σ	0..1	CodeableConcept	Kind of Group members
name	Σ	0..1	string	Label for Group
quantity	Σ	0..1	unsignedInt	Number of members
managingEntity	Σ	0..1	Reference(Organization | RelatedPerson | Practitioner | PractitionerRole)	Entity that is the custodian of the Group's definition
characteristic		0..*	BackboneElement	Include / Exclude group members by Trait
code		1..1	CodeableConcept	Kind of characteristic
value[x]		1..1		Value held by characteristic
valueCodeableConcept			CodeableConcept	
valueBoolean			boolean	
valueQuantity			Quantity	
valueRange			Range	
valueReference			Reference()	
exclude		1..1	boolean	Group includes or excludes
period		0..1	Period	Period over which characteristic is tested
member	I	0..*	BackboneElement	Who or what is in group
entity		1..1	Reference(Patient | Practitioner | PractitionerRole | Device | Medication | Substance | Group)	Reference to the group member
period		0..1	Period	Period member belonged to the group
inactive		0..1	boolean	If member is no longer in group

https://model.clinicalprofiles.org/group.html

Population / Cohort model



Playing with Populations and Cohorts

Binder

ClinicalProfile

Name	Flags	Card.	Type	Description & Constraints
ClinicalProfile	TU		DomainResource	Results of a measure evaluation Elements defined in Ancestors: id, meta, implicitRules, language, text, contained, extension, modifierExtension
identifier	Σ	0..*	Identifier	Additional identifier for the ClinicalProfile
status	?! Σ	1..1	code	complete | draft | error ClinicalProfileStatus (Required)
population	Σ	1..1	Reference(Group)	The base population against which this profile was generated
cohort	Σ	1..1	Reference(Group)	The cohort within the population that this profile represents
date	Σ	0..1	dateTime	When the profile was generated
source		0..*	Identifier	Identifier(s) from where the profile was acquired
reporter	Σ	1..1	Reference(Organization | Practitioner | PractitionerRole | Location)	Who is reporting the data
lab		0..*	BackboneElement	Laboratory profile entry

https://model.clinicalprofiles.org/clinicalprofile.html

ClinicalProfile Lab

lab		0..*	BackboneElement	Laboratory profile entry
code	Σ	1..*	CodeableConcept	Lab code (LOINC) LOINC Codes (Example)
count	Σ	1..1	integer	Total number of lab tests
frequencyPerYear		0..1	decimal	Frequency of this lab ordered/reported per patient per year
fractionOfSubjects		0..1	decimal	Fraction of subjects with this lab
scalarDistribution		0..1	BackboneElement	Scalar sample summary
units	Σ	1..1	SimpleQuantity	Units of scalar result
min	Σ	1..1	decimal	Minimum value
max	Σ	1..1	decimal	Maximum value
mean	Σ	1..1	decimal	Mean
median	Σ	1..1	decimal	Median
stdDev	Σ	1..1	decimal	Standard deviation
decile		0..*	BackboneElement	Decile partitions
nth		1..1	integer	Particular decile (10, 20, …)
value		1..1	decimal	Cutoff value for this decile
normalizedHigh		0..1	decimal	Normalize high normal range
normalizedLow		0..1	decimal	Normalize low normal range
fractionAboveNormal		0..1	decimal	Fraction of samples above normalized normal range
fractionBelowNormal		0..1	decimal	Fraction of samples below normalized normal range

Scalar Distribution

scalarDistribution		0..1	BackboneElement	Scalar sample summary
units	Σ	1..1	SimpleQuantity	Units of scalar result
min	Σ	1..1	decimal	Minimum value
max	Σ	1..1	decimal	Maximum value
mean	Σ	1..1	decimal	Mean
median	Σ	1..1	decimal	Median
stdDev	Σ	1..1	decimal	Standard deviation
decile		0..*	BackboneElement	Decile partitions
nth		1..1	integer	Particular decile (10, 20, ...)
value		1..1	decimal	Cutoff value for this decile
normalizedHigh		0..1	decimal	Normalize high normal range
normalizedLow		0..1	decimal	Normalize low normal range
fractionAboveNormal		0..1	decimal	Fraction of samples above normalized normal range
fractionBelowNormal		0..1	decimal	Fraction of samples below normalized normal range
correlatedLabs		0..1	BackboneElement	Correlated laboratory tests
topn		0..1	integer	Number of correlations cutoff (e.g. top 10)
abscorrelation		0..1	decimal	Minimum absolute value of correlation coefficient
entry		0..*	BackboneElement	Correlated lab
labcode	Σ	1..*	CodeableConcept	Lab code (LOINC) or Lab Group Code LOINC Codes (Example)
coefficient	Σ	1..1	decimal	Correlation coefficient
correlatedDiagnoses		0..1	BackboneElement	Correlated diagnosies

Correlations
Labs and Dx

correlatedLabs		0..1	BackboneElement	Correlated laboratory tests
topn		0..1	integer	Number of correlations cutoff (e.g. top 10)
abscorrelation		0..1	decimal	Minimum absolute value of correlation coefficient
entry		0..*	BackboneElement	Correlated lab
labcode	Σ	1..*	CodeableConcept	Lab code (LOINC) or Lab Group Code LOINC Codes (Example)
coefficient	Σ	1..1	decimal	Correlation coefficient
correlatedDiagnoses		0..1	BackboneElement	Correlated diagnosies
topn		0..1	integer	Number of diagnoses cutoff (e.g. top 10)
abscorrelation		0..1	decimal	Minimum absolute value of correlation coefficient
entry		0..1	BackboneElement	Correlation entry
code	Σ	1..1	CodeableConcept	Diagnosis code(s) SNOMED CT Clinical Findings (Example)
coefficient	Σ	1..1	decimal	Correlation coefficient

Correlations

Medications, Procedures and Phenotypes

coefficient	Σ	1..1	decimal	Correlation coefficient
correlatedMedications		0..1	BackboneElement	Correlated medications
topn		0..1	integer	Number of medications cutoff (e.g. top 10)
deviation		0..1	decimal	Deviation cutoff
entry		0..*	BackboneElement	Correlation entry
category		0..1	CodeableConcept	Indicates the type of medication dispense (for example, where the medication is expected to be consumed or administered (i.e. inpatient or outpatient)) Medication dispense category codes (Example)
lab[x]	Σ	1..1		Medication code(s) SNOMED CT Medication Codes (Example)
labCodeableConcept			CodeableConcept	
labReference			Reference(Medication)	
coefficient	Σ	1..1	decimal	Correlation coefficient
correlatedProcedures		0..1	BackboneElement	Correlated procedures
topn		0..1	integer	Number of rocedures cutoff (e.g. top 10)
abscorrelation		0..1	decimal	Minimum absolute value of correlation coefficient
entry		0..*	BackboneElement	Correlation entry
code	Σ	1..*	CodeableConcept	Procedure code(s) Procedure Codes (SNOMED CT) (Example)
coefficient	Σ	1..1	decimal	Correlation coefficient
correlatedPhenotypes		0..1	BackboneElement	Correlated phenotypes
topn		0..1	integer	Number of phenotypes cutoff (e.g. top 10)
abscorrelation		0..1	decimal	Minimum absolute value of correlation coefficient
entry		0..*	BackboneElement	Correlation entry
code	Σ	1..1	CodeableConcept	Phenotype codes
coefficient	Σ	1..1	decimal	Correlation coefficient

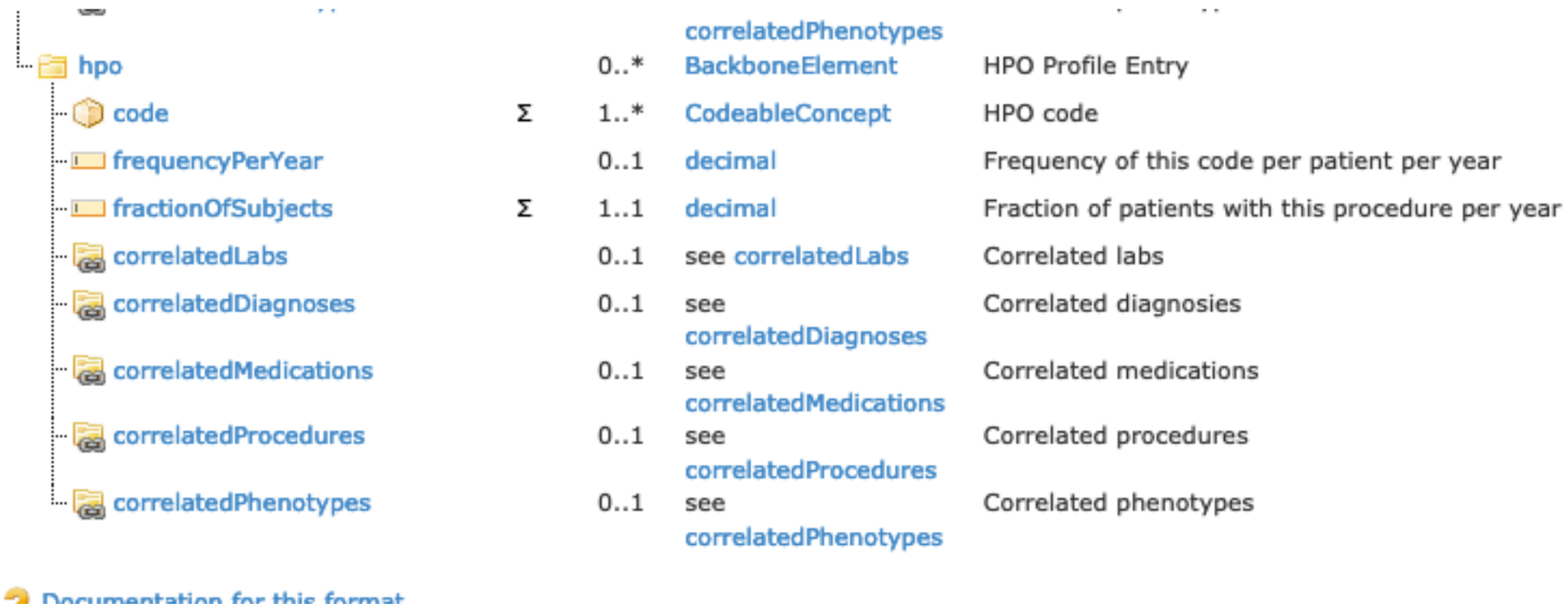
Medications

medication		0..*	BackboneElement	Medication profile entry
category		0..1	CodeableConcept	Indicates the type of medication dispense (for example, where the medication is expected to be consumed or administered (i.e. inpatient or outpatient)) Medication dispense category codes (Example)
medication[x]	Σ	1..1		Medication(s) being profiled SNOMED CT Medication Codes (Example)
medicationCodeableConcept			CodeableConcept	
medicationReference			Reference(Medication)	
dosage		0..1	BackboneElement	Details of how medication was taken
text		0..1	string	Free text dosage instructions e.g. SIG
site		0..*	CodeableConcept	Body site(s) administered to SNOMED CT Anatomical Structure for Administration Site Codes (Example)
route		0..*	CodeableConcept	Path(s) of substance into body SNOMED CT Route Codes (Example)
method		0..*	CodeableConcept	How the drug was administered SNOMED CT Administration Method Codes (Example)
dose		0..*	SimpleQuantity	Average amount of medication per dose
rate[x]		0..1		Dose quantity per unit of time
rateRatio			Ratio	
rateQuantity			SimpleQuantity	
treatementDuration		0..1	decimal	Duration of treatment (in 1 year)
frequencyPerYear		0..1	decimal	Frequency of treatments per patient per year
fractionOfSubjects	Σ	1..1	decimal	Fraction of patients in cohort treated with this drug
correlatedLabs		0..1	see correlatedLabs	Correlated labs
correlatedDiagnoses		0..1	see correlatedDiagnoses	Correlated diagnosies
correlatedMedications		0..1	see correlatedMedications	Correlated medications
correlatedProcedures		0..1	see correlatedProcedures	Correlated procedures
correlatedPhenotypes		0..1	see	Correlated phenotypes

Procedures

correlatedPhenotypes				
procedure		0..*	BackboneElement	Procedure profile entry
code	Σ	1..*	CodeableConcept	ICD-10-PCS or CPT procedure code
frequencyPerYear		0..1	decimal	Frequency of procedure per patient per year
fractionOfSubjects	Σ	1..1	decimal	Fraction of patients with this procedure per year
correlatedLabs		0..1	see correlatedLabs	Correlated labs
correlatedDiagnoses		0..1	see correlatedDiagnoses	Correlated diagnosies
correlatedMedications		0..1	see correlatedMedications	Correlated medications
correlatedProcedures		0..1	see correlatedProcedures	Correlated procedures
correlatedPhenotypes		0..1	see correlatedPhenotypes	Correlated phenotypes

Phenotypes



				correlatedPhenotypes	
hpo			0..*	BackboneElement	HPO Profile Entry
code		Σ	1..*	CodeableConcept	HPO code
frequencyPerYear			0..1	decimal	Frequency of this code per patient per year
fractionOfSubjects		Σ	1..1	decimal	Fraction of patients with this procedure per year
correlatedLabs			0..1	see correlatedLabs	Correlated labs
correlatedDiagnoses			0..1	see correlatedDiagnoses	Correlated diagnosies
correlatedMedications			0..1	see correlatedMedications	Correlated medications
correlatedProcedures			0..1	see correlatedProcedures	Correlated procedures
correlatedPhenotypes			0..1	see correlatedPhenotypes	Correlated phenotypes

Documentation for this format

Sample Uses

Binder

Next Steps
Short Term

- Resource Granularity — (re-) partition into separate lab, medication, procedure, phenotype elements

- Queries and access paths — query vs. search on groups and the like

- Content — medications, HPO still need to be (re-) added

- Server — a bit fragile at the moment (due to resource sizes)

Next Steps
Short(er) term

- Knowledge Graph Wrapper
    - Interesting questions about what “facts” are actually represented — “patients with asthma”, “lab result”, “medication”, “distribution”, …

Next Steps
Long(er) term

- Transform model into EBMonFHIR equivalent

- Formalize and extend Group definitions
  - Need to coordinate with CDS and others

- Static —> semi-dynamic —> dynamic(?) queries

The Team

- Christopher Chute
- Casey Overby Taylor
- Richard Zhu
- Jordan Matelsky
- Stephanie Howson
- Amber Zhou
- Andrew Massoud
- Tricia Francis
- …