

Clinical Profiles: Computable Knowledge for Translational Research

William J. Howe, Maya M. Lapinski, Christopher G. Chute

Johns Hopkins Institute for Clinical and Translational Research Johns Hopkins University



INTRODUCTION

- FHIR (Fast Healthcare Interoperability Resources) provides a standard and schema for clinical data exchange focused on interoperability across healthcare applications and use cases
- EBM (Evidence-Based Medicine) is the process of translating knowledge into a machine-readable form in order to analyze, synthesize, and implement this knowledge into clinical care
- Clinical profiles provide population-level knowledge of disease behavior by aggregating statistical information derived from patient records

This project aims to take basic science knowledge and make it available to clinical practice.

IMPLEMENTATION

FHIR Resources Modular data components comprised of an identity, metadata, implicit rules, and a language

Evidence-Based Medicine

Category of resources describing knowledge (evidence, assertion, recommendation)

Clinical Profiles

Registry of compositions of data about clinical data, readable by machines and divided into cohorts



Observations

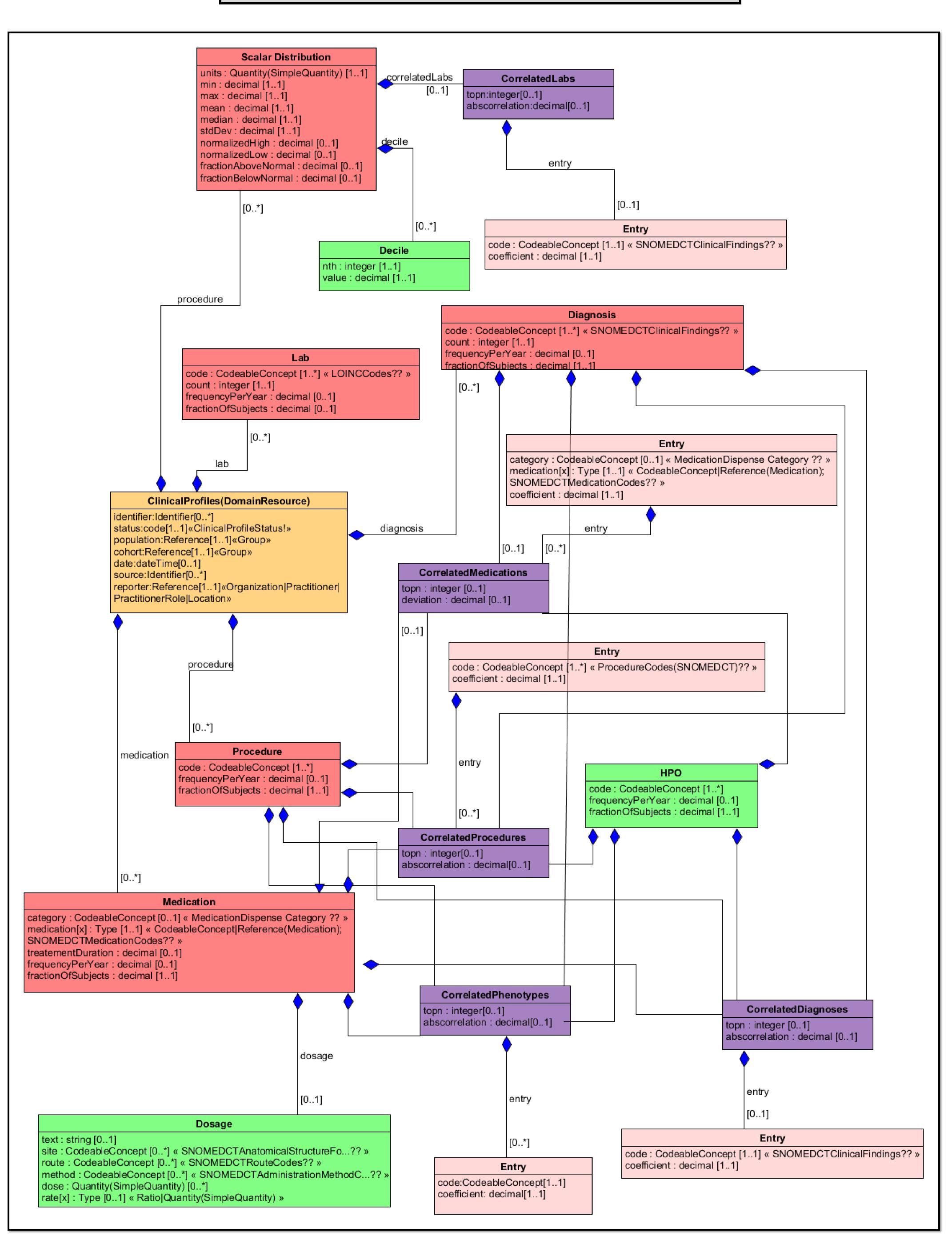
Core data composing clinical profile, such as medication type and dosage, demographic information, and procedures



Analysis

Information relating observations, such as correlations and statistical summaries

UML



CLINICAL PROFILES

- Patient-level data from 1.9 million patients
 - Labs
 - Medication
 - Diagnosis
 - Procedure
 - HPO (human phenotype ontology)
- Reduced to population-level using
 - Statistical summaries
 - Frequencies
 - Distributions
 - Relative Counts
 - Correlations and covariance
- Provides summary and features of population without personal or identifiable health information, allowing comprehensive access for translational use
- Aggregated into phenotypically relevant cohorts based on condition/demographics
- Machine-readable and parsed in standard renditions

XML

```
<ClinicalProfile xmlns="http://hl7.org/fhir">
<!-- from Resource: id, meta, implicitRules, and language -->
<!-- from DomainResource: text, contained, extension, and modifierExtension -->
<identifier><!-- 0..* Identifier Additional identifier for the ClinicalProfile --></identifier>
<<status value="[code]"/><!-- 1..1 complete | draft | error -->
cpopulation><!-- 1..1 Reference(Group) The base population against which this profile was generated -->/cpopulation>
```

JSON

```
"resourceType" : "ClinicalProfile",

// from Resource: id, meta, implicitRules, and language

// from DomainResource: text, contained, extension, and modifierExtension

"identifier" : [{ Identifier }], // Additional identifier for the ClinicalProfile

"status" : "<code>", // R! complete | draft | error

"population" : { Reference(Group) }, // R! The base population against which this profile was generated
```

Turtle

```
a fhir:ClinicalProfile;

fhir:nodeRole fhir:treeRoot; # if this is the parser root

# from Resource: .id, .meta, .implicitRules, and .language

# from DomainResource: .text, .contained, .extension, and .modifierExtension

fhir:ClinicalProfile.identifier [ Identifier ], ...; # 0..* Additional identifier for the ClinicalProfile

fhir:ClinicalProfile.status [ code ]; # 1..1 complete | draft | error

fhir:ClinicalProfile.population [ Reference(Group) ]; # 1..1 The base population against which this profile was generated
```

fhir:ClinicalProfile.cohort [Reference(Group)]; # 1..1 The cohort within the population that this profile represents

CONCLUSIONS

- Quantitative knowledge can be rendered in a computable format
- This knowledge is relevant to clinical practices and questions
- The data science of making computable evidence available to clinical practice is rapidly emerging

ACKNOWLEDGEMENTS

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The authors would like to thank Tricia Francis, Toni Cheeks Shaw, Xiaohan Zhang, Joseph L. Bondi, Jeremy A. Epstein, Jordan K. Matelsky, Kenneth D. Roe, Harold R. Solbrig, Richard L. Zhu, and Casey Overby Taylor for their guidance and support.



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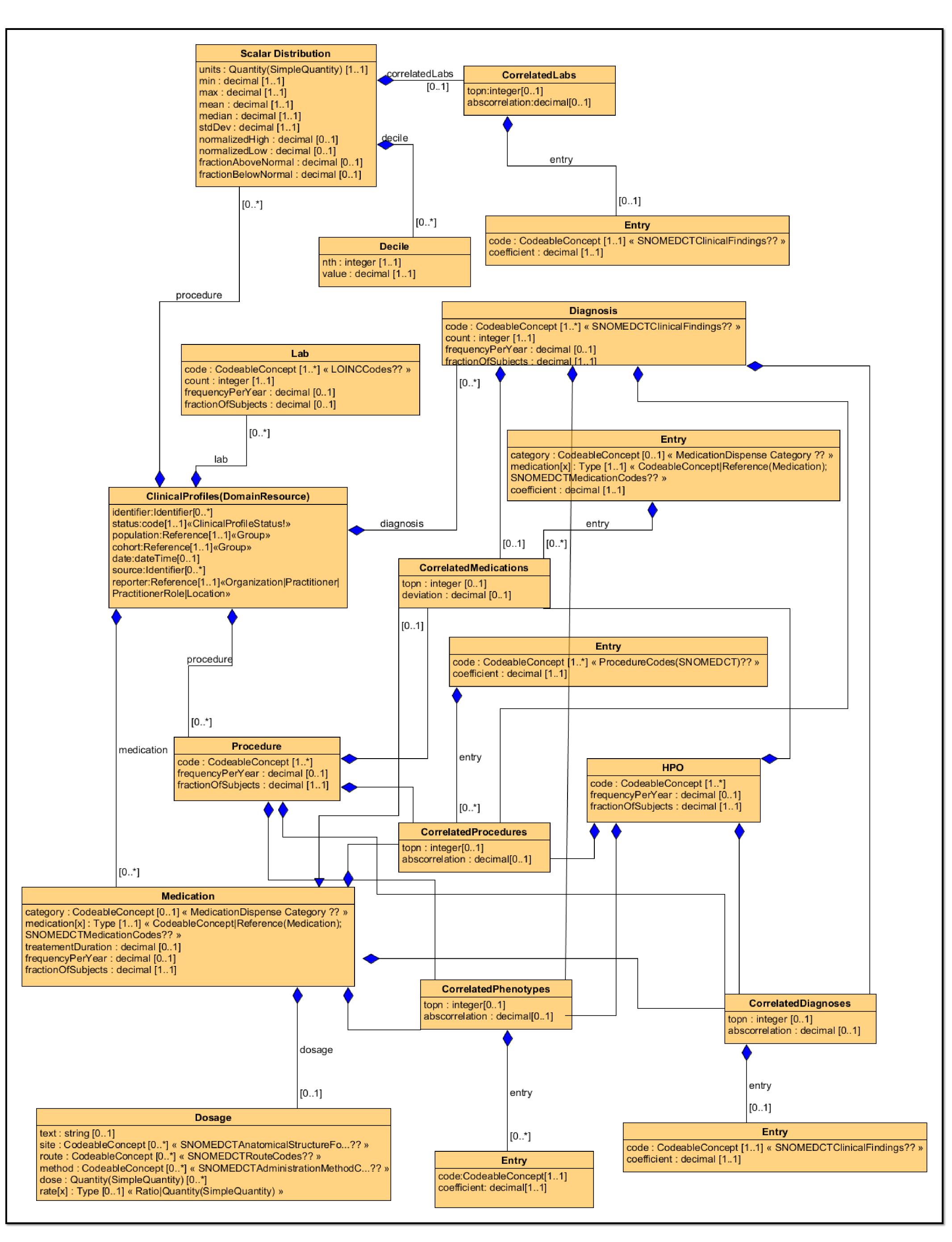
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// from DomainResource: text, contained, extension, and modifierExtension

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