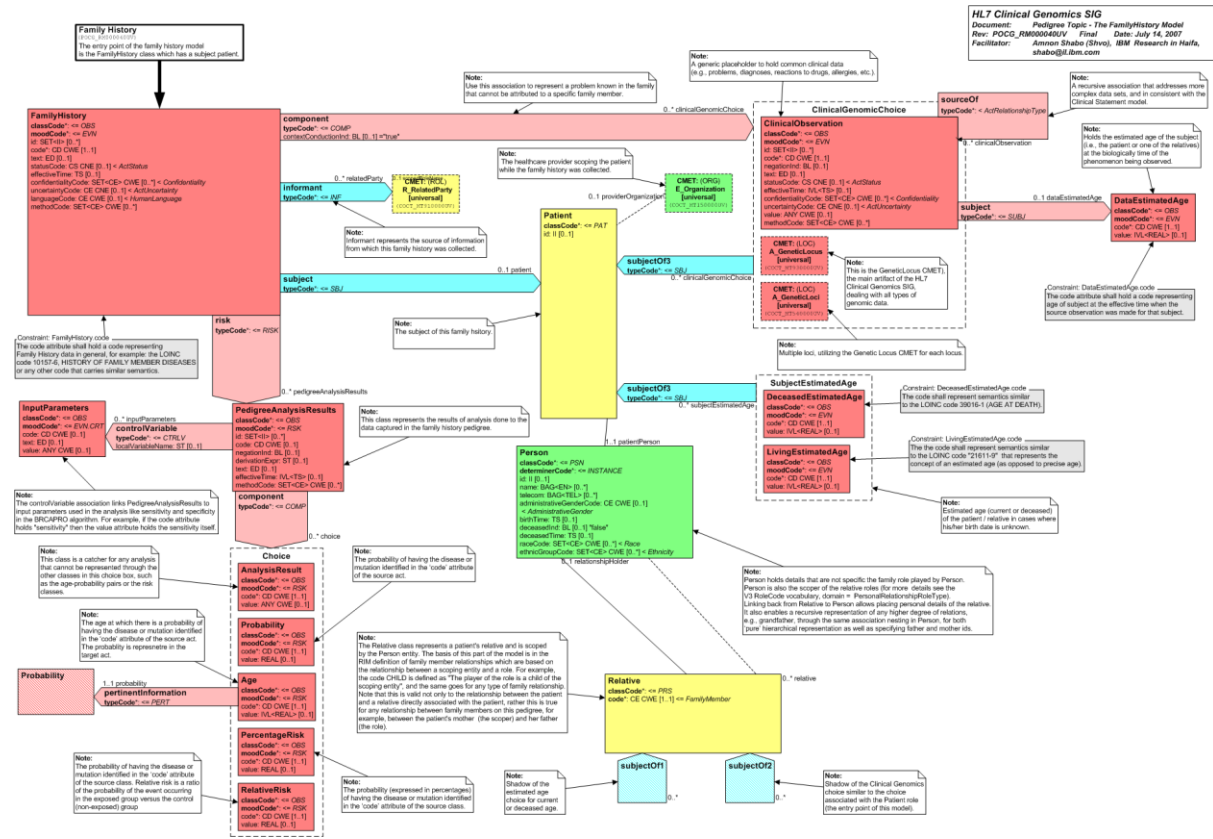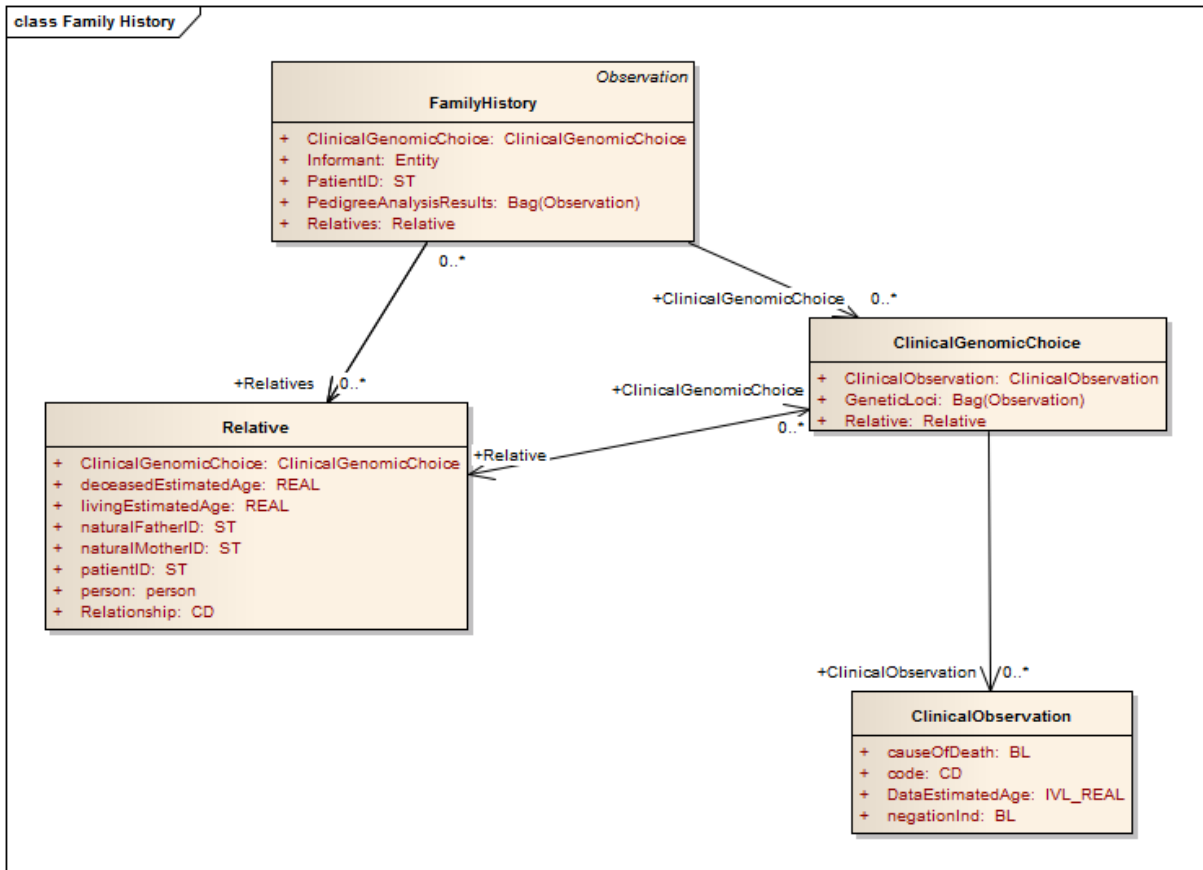vMR Family History Model – based on Genomics RMIM

This is the latest Version of the Family History RMIM:

The model below does not include some values from Clinical Genomic Choice related to the patient, but only values related to relatives. ?Are those values recorded about the patient.

The vMR model uses the RMIM to make the relationships explicit and easier to query without losing any data. The relationships in the RMIM are fixed and represented using the RIM whereas the VMR Model uses an explicit object model. The object model allows direct access to a collection of relatives as well as the collection of conditions that the relative suffers from.

Model in use with GELLO as example language:

1. Family history of colitis using SNOMED-CT codes

```
Context SinglePatient

Let IBD: CD = Factory.CodedValue('24526004', 'SNOMED-CT') --IBD
Let Colitis: CD = Factory.CodedValue('64226004', 'SNOMED-CT') --Colitis

FamilyHistory.ClinicalGenomicChoice -> select(
   ClinicalObservation -> select(
      (code.implies(IBD).value or code.implies(Colitis).value) and (not negationInd.value)
                          ).notEmpty()
                                      ).notEmpty()
```

2. Colon cancer at 50 or less in a Natural Parent (NCI codes from example message)

```
Context SinglePatient

Let ColonCancer: CD = Factory.CodedValue('C2955', 'NCI') --Colon Cancer NCI Code

FamilyHistory.ClinicalGenomicChoice -> select(
        (Relative.Relationship.code = 'NMTH' or Relative.Relationship.code = 'NFTH') and
        ClinicalObservation -> select(
                        code.implies(ColonCancer).value and (not negationInd.value) and (DataEstimatedAge.low.value <= 50.0)
                                ).notEmpty()
                                        ).notEmpty()
```

Alternate representation using GELLO class notation:

```
class FamilyHistory extends Observation
   ClinicalGenomicChoice: Sequence(ClinicalGenomicChoice)
   Informant: Entity
   PatientID: ST
   PedigreeAnalysisResults: Sequence(Observation)
   Relatives: Sequence(Relative)

class ClinicalGenomicChoice extends Any
   ClinicalObservation: Sequence(ClinicalObservation)
   GeneticLoci: Sequence(Observation)
   Relative: Relative

class ClinicalObservation extends Any
   causeOfDeath: BL
   code: CD
   DataEstimatedAge: IVL_REAL
   negationInd: BL

class Relative extends Any
   ClinicalGenomicChoice: Sequence(ClinicalGenomicChoice)
   deceasedEstimatedAge: REAL
   livingEstimatedAge: REAL
   naturalFatherID: ST
   naturalMotherID: ST
   patientID: ST
   person: Person
   Relationship: CD
```

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