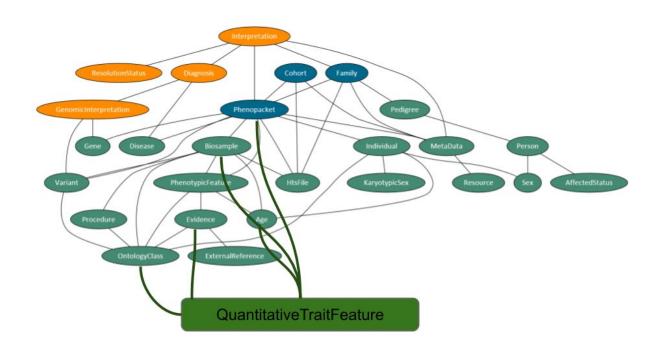
How to incorporate the quantitative trait data model + integration with trait ontologies into the phenopacket model?

As a new phenopacket building blog:



Quantitative_trait_feature

This is a list of quantitative traits as clinical signs and laboratory findings observed in the subject. See below for further information.

QuantitativeTraitFeature

This element is intended to be used to describe a quantitative trait that characterizes the subject of the Phenopacket. For medical use cases the subject will generally be a patient or a proband of a study, and the quantitative traits will be clinical or laboratory measurements described by an ontology such as the Clinical Measurement Ontology. The word quantitative trait mean An individual trait feature, observed as either present or absent (negated), with possible onset, modifiers and frequency.

Data model

Field	Туре	Status	Description
description	string	optional	human-readable verbiage NOT for structured text

type	OntologyClass	required	term denoting the quantitative trait feature
negated	boolean	optional	defaults to false
value	string	required	Measurement value
unit	OntologyClass	required	term denoting the unit of measurement
assay	OntologyClass	optional	Term denoting the assay type
onset	Age OR AgeRange	recommended	Age at which the feature was first observed
biosample	Biosample	recommended	Unit of biological material from which the substrate for analyses are extracted
evidence	Evidence	recommended	the evidence for an assertion of the observation of a type (ECO)

Example

Description

This element represents a free-text description of the quantitative trait.

Type

This element represents the primary ontology class which describes the quantitative trait, e.g. Temperature (CMO:0000015).

Negated

This element is a flag to indicate whether the quantitative trait was measured or not.

Value

This element is a string that describes the magnitude measured of the trait.

Unit

This element is an ontology class that describes the unit label of measurement.

Assay

This element is an ontology class that describes the type of assay.

Onset

This element is recommended to describe the age at which the quantitative trait feature was first noticed.

Biosample

This element is recommended to describe the unit of biological material from which the substrate for analyses are extracted.

Evidence

This element is recommended and contains one or more <u>Evidence</u> elements that specify how the trait was determined.

Data model OBSOLETE

Field	Туре	Status	Description
description	string	optional	human-readable verbiage NOT for structured text
type	OntologyClass	required	term denoting the quantitative trait feature
negated	boolean	optional	defaults to false

quantity*	OntologyClass	required	
severity	OntologyClass	optional	description of the severity of the feature described in type (representing HP:0012824? Or PATO as appropriate)
modifier	List of OntologyClass	optional	representing one or more terms (from HP:0012823? Or PATO as appropriate)
onset	OntologyClass	optional	Age at which the features was first observed, e.g. HP:0011462 ?
evidence	Evidence	recommended	the evidence for an assertion of the observation of a type (ECO?)

*QUDT model: https://qudt.org/

Example

```
"type": {
  "id": "CMO:0000015",
  "label": "Temperature"
},
  "quantity": {
    "id": "qud:Quantity"
    "value": {
        "id": "qud:QuantityValue",
        "magnitude": "40.1"
      },
    "Kind": {
        "id": "qud:QuantityKind",
        "label": "Temperature"
      },
  "unit": {
```

```
"Id": "qud:Unit",
         "label": "Celsius Degrees",
         "system": {
                  "id": "OntologyClass",
                  "label": "metric"
               },
         },
  },
  "severity": {
     id": "OntologyClass",
     "label": "Severe"
  },
  "modifier": {
     id": "OntologyClass",
     "label": "Increased"
  }
}
```

Description

This element represents a free-text description of the quantitative trait.

Type

This element represents the primary ontology class which describes the quantitative trait, e.g. Temperature (CMO:0000015).

Negated

This element is a flag to indicate whether the quantitative trait was measured or not.

Quantity

This element is an ontology class that describes the quantity measured of the trait.

Severity

This element is an ontology class that describes the severity of the condition.

Modifier

This element is a list of ontology class elements that can be empty or contain one or more ontology terms that are intended to provide more expressive or precise descriptions of a quantitative trait feature, including attributes such as positionality and external factors that tend to trigger or ameliorate the feature. Terms can be taken from the hierarchy of Clinical modifier in the HPO (noting that severity should be coded in the severity element).

Onset

This element can be used to describe the age at which the quantitative trait feature was first noticed. For many medical use cases, either the Age sub-element or an ontology class (e.g., from the HPO Onset (HP:0003674) terms) will be used.

Evidence

This element is recommended and contains one or more <u>Evidence</u> elements that specify how the phenotype was determined.