Why do it the hard way?
The case for Expressive Ontological Schemas for SNOMED

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The Issues

- SNOMED's schemas were designed to fit DL & hardware technologies from the late 1980s / early 1990s
  - Many constraints are now (probably) outdated
  - "End stage evolutionary development"
- Impose major costs in complexity and development effort
  - Typical "query" is at least 1 page long
  - Huge effort on "subsets" that are difficult to build and will be much more difficult to maintain
  - Great difficulty in binding to EHRs and Messages
    - Length and complexity of the Terminfo document
- Are better schemas possible?
  - If so, what do they require of the implementation language?

The Opportunity

- SNOMED is just the beginning of deployment
  - Very few real records are recorded with complex SNOMED expressions
  - Little software uses post-coordination or interacts with the schema in significant ways
  - The organisational changes following the formation of the IHTSDO give a major oppotunity
- Change easier now than later
  - Before building massive “pregacy”
  - Before software starts to use the DL structure seriously
  - Before an even more massive combinatorial expansion of terms
    - Do we enumerate all the noun phases in medical English?
  - Although change is always hard
    - At least a major feasibility study is warranted

Goals for improved semantics

- **Easier more consistent development**
  - **More effective Quality Assurance**
  - **Easier / more effective specification of use**
    - **Subsetting**
    - **Querying**
    - **EHR-Terminology - and other application - binding**
    - **Effective post-coordination**

- **A simple test for improved semantics:**
  - ***Do the specs / documents / queries / subset mechansims get shorter?***
    - **Measure improvement by the length of SQL/TQL required by applications**
      - ***By the length of the documents***

Basic proposals

- Represent all “recordable codes” uniformly as “Situations”
  - Eliminate the separate classification of “Situations with explicit context”
    - Distinguish “kernel codes/concepts” from “recordable codes/concepts”
  - Rationalise the context model
    - Including separating negation from the rest of context
- To represent “observables” and “findings” in such a way that the equivalences and implications can be inferred uniformly.
  - Recognise that “finding” and “observable” are meta concepts
- To be explicit about whether each site refers to the whole or the whole or its parts
- To build SNOMED as a series of modules
- And make it possible to load / use only those modles needed for a particular application
  - Deveopment strategy: Control the dependencies between modules

The starting point: What does a code in an EHR mean?

- Entering a code in a record makes a statement about a patient -
  - typically that a patient *has* something
    - (as observed by an observer at a time & place)

- Codes represent classes of precates
  - Entering a code for a “Recordable entities” is saying that a patient - as understood by a an observer at a time and place - belongs to the coresponding class
    - ... but patients ARE NOT diseases or procedures
      - *Patients HAVE diseases and procedures*

Therefore ...

- **Divide codes & entities into**
  - **Kernel entities / codes -**
    - **the diseases & acts themselves**
      (and the codes that stand for them)
  - **Recordable entities/codes -**
    - **that the patient *has* a kernel entities**
      (and the codes that stand for having them)
      - ***as observed by an observer at a given time and place***

Therefore:

- **"Recordable concepts" are about the patient as observed by an observer at a time and place**
  - **We label the units of information "Patient Situations" or "Situations" for short**
    - **Proxies for *"the patient state as understood by an observer at a given time and place"***
      - ***e.g. "I state here and now that 'the patient has X'" or "I state here and now that 'the patient does not have X'"***
- **Recordable codes represent classes of "situations"**
  - ***Situation THAT includes SOME Condition***
    - ***e.g. Situation THAT includes SOME Diabetes***
  - ***Situation THAT NOT includes***
    - ***e.g. Situation THAT NOT includes SOME Diabetes***

Situations & Context

- Situations *encapsulate* context and *include* kernel concepts
  - May contain any boolean combination of kernel concepts
- Provide a uniform approach to context
  - Not a new idea
    - GALEN and PEN&PAN both used them
      - *Allow conjunctions and disjunctions of kernel concepts as well as negation*
      - *But particularly useful given an expressive DL with negation*
- All contents included in a “situation” are relative to a single root subject -
  - the subject of that EHR instance
    - i.e. seen from the point of view of care of one patient
      - *Even if actually about some other subject*

Rationalising the Context Model

- **The current SNOMED context model conflates**
  - **"Modalities"**
    - ***The patient is at risk of cardiovascular disease***
    - ***The patient has a family history of type 2 diabetes***
  - **Subject of care**
    - ***The fetus has a depressed heart rate***
  - **(Relative) temporal relative markers**
    - ***The patient has a history of myocardial infarction***
  - **Negation**
    - ***The patient does not have diabetes***

Separating out the cases - 0

- **Simple conditions**
  - **e.g.**
    - **The paient has cardiovascular disease**
    - **The patient has type 1 diabetes**
  - **Represent as simple inclusion in situations**
    - ***Situation THAT includes SOME Cardiovascular_disorder***
    - ***Situation THAT includes SOME Diabetes_type_1***

Separating out the cases - 1

- **"Modalities"**
  - **e.g.**
    - **The patient is at risk of cardiovascular disease**
    - **The patient has a family history of type 2 diabetes**

  - ***"Risk of CVD" is not a kind of cardiovascular disorder***
    "Family history of diabetes" is not a kind of diabetes

  - **Represent as concepts derived from kernel concepts**
    - ***Situation THAT includes SOME (Risk THAT is_of SOME Cardio_vascular_disorder)***

    - ***Situation THAT includes SOME (Family_history THAT is_of SOME Diabetes_type_2)***

Separating out the cases - 2

- **Subject of care**
  - **e.g.**
    - **“The fetus has a depressed heart rate”**
    - **“The mother is depressed”**
  - **Represent as a nested situation within a situation**
    - ***Situation THAT has_subsituation SOME***
      (Situation THAT
      has_subject SOME Fetus AND
      includes SOME Depressed_heart_rate)
    - ***Situation THAT has_subsituation SOME***
      (Situation THAT
      has_subject SOME Mother AND
      includes SOME Depression)

Separating out the cases - 3

- Relative temporal markers
  - e.g.
    - History of Myocardial infarct
- Two alternatives
  - Simpler alternative:
    Represent by analogy with modalities
    - *Situation THAT includes SOME*
      (History THAT refers_to SOME Myocardial_infarct)
  - More expressive alternative:
    Represent by analogy with subject of care
    - *Situation THAT has_subsitutation SOME*
      (Situation THAT
      has_temperal_marker SOME History AND
      includes SOME Myocardial_infarct)
- We have no use case requiring more expressive alternative.
  - *Do you have any?*

Separating ou the cases 4

- Negation
  - Entities not included in the situation
  - e.g.The patient does not have cardiovascular disease
  - Represent as “NOT includes SOME X”
    - Situation THAT NOT includes SOME Cardiovascular_disease

What we organise with logic are classes of situations, negation inverts the hierarchy e.g.

- **Positive**

  "situations that include Metabolic Disorder"

  "situations that include Diabetes"

  "situations that include Type 1 Diabetes"

- **Negation**

  "situation that do not include Type 1 Diabetes"

  "situation that does not include Diabetes"

  "situation that does not include Metabolic Disease"

Example - classic problems just fall out

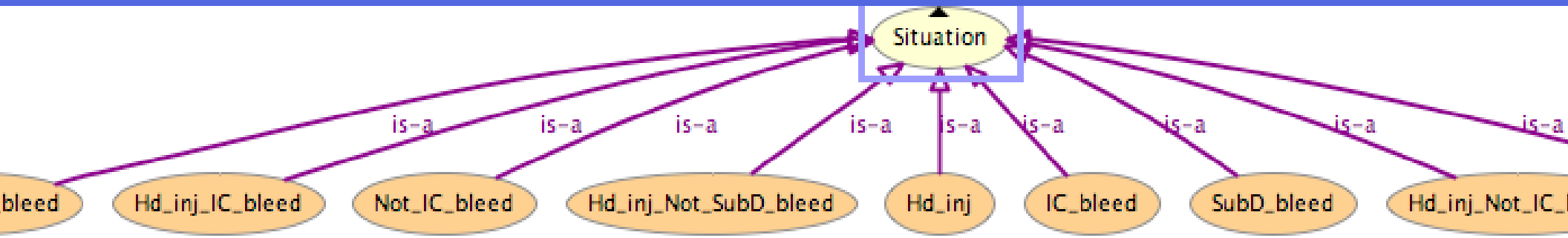
e.g. Head trauma with/without fracture

with/without Intracranial_bleed

subdural_hematoma

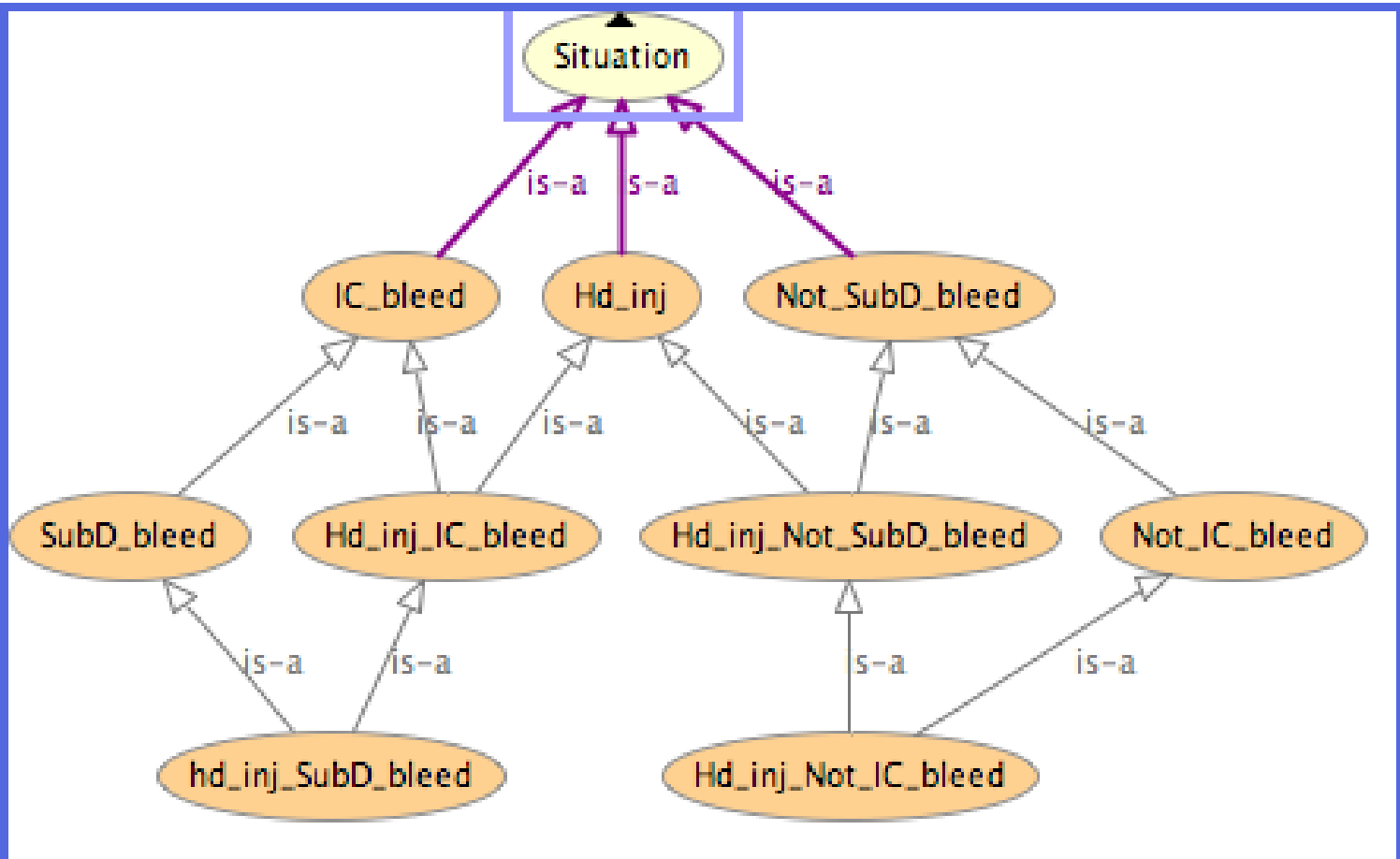
...

Head Injury with and without intracranial bleed, *before classification*



after classifica -tion

Let the classifier do the work



Aside - 1

- Ignores problem of “negative findings”
  - e.g. “Absent pedal pulses”
    - See Medinfo paper for full treatment
      - http://www.cs.man.ac.uk/~rector/papers/Whats-in-a-code/
        - *Also accessible from my ome page*

Aside - 2

- **True negation does require a more expressive DL but...**
  - **Negation has proved hard to ban negation from the terminology**
  - **Current SNOMED/HL7 guidelines state that negation should be included in the code**
    - **But the representation does not capture so that it can be used correctly**
      - ***We are "lying to the logic"***
        - *Even complete logical algorithms will not prevent false conclusions following from false premises.*

Observables and Findings: Metacategories

- **Basic maxim:**
  All statements should convey information
  - **There is no point in stating a tautology -**
    - **Or even something that is redundant given assumed background knowledge**
- **Key distinction**
  - **Some entities are always present - "observables"**
    - **Qualities of the organism or one of its parts or functions**
      - ***e.g. Body temperature, white count, skin colour, etc.***
        - *They must be described or given a value to convey information*
  - **Some entities are variably present - "findings"**
    - **Pathologies or variations in the orgaqnism or its parts or functions**
      - ***e.g. Diabetes, stroke, third heart sound, rales, wheezes, ...***
        - *Saying that they are present does convey information*

Encapsulation

- Often use qualitative descriptions for qualities
  - e.g. Body_temperature = “elevated”
- Sometimes we encapsulate the whole thing as a single expression
  - “Elevated body temperature”
- The “recordable entity” for both is:
  - *Situation THAT incudes SOME*
    (Body_temperature THAT has_level SOME Elevated_value)
- In general an “Observable + Value” = Finding
  - So the observable / finding distinction cannot be a first order distinction
    - It is not about what things “are” but how they occur
      - *A “meta distinction”*

Testing for Findings vs Observables

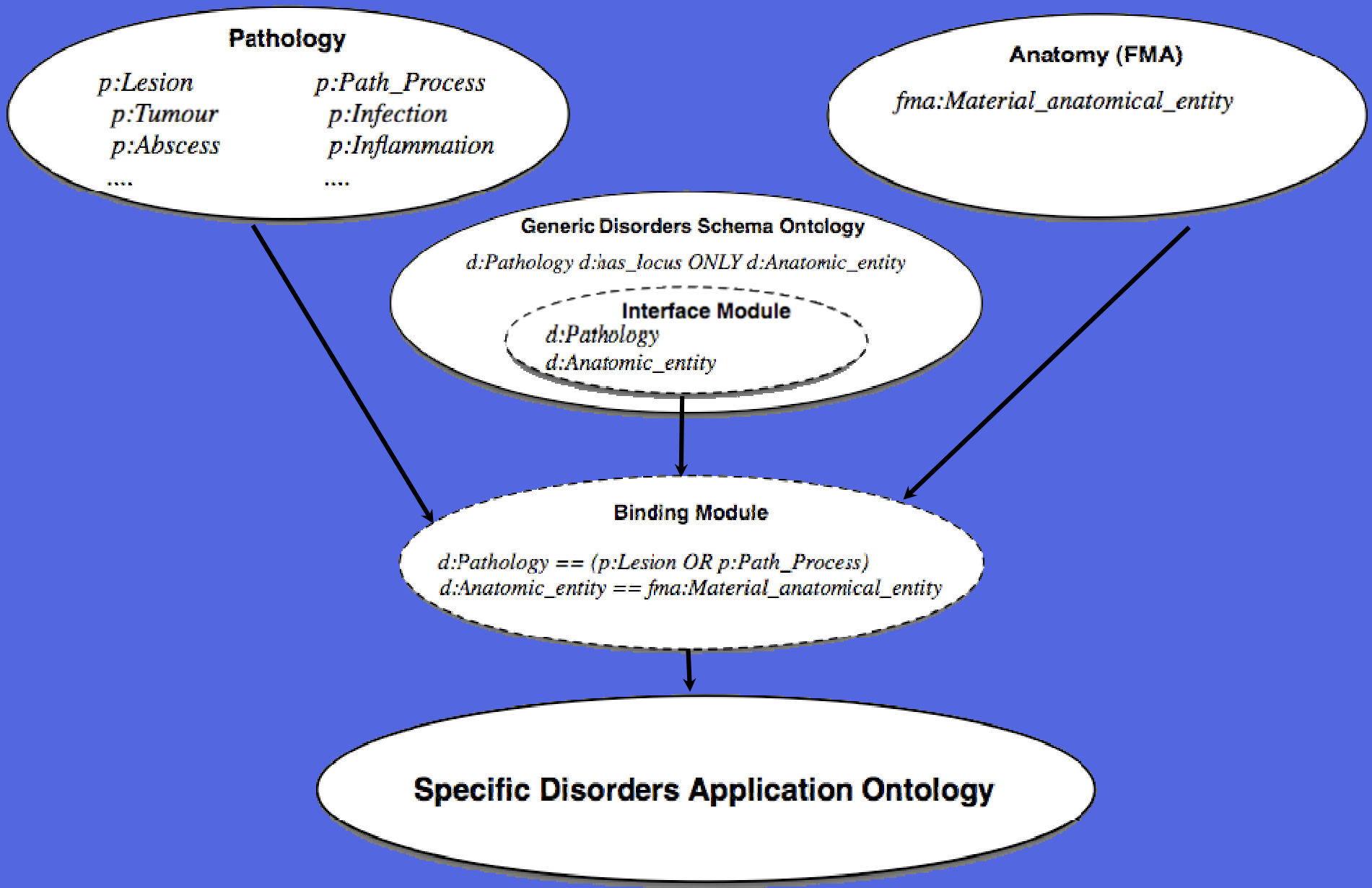
- **The criterion is the meta-test for tautology / redundancy**
  - **IF *Situation includes SOME Kernel_X*** ≣ Situation
    THEN ***Kernel_X* is an "observable"**
    ELSE *Kernel_X* is a "finding"
    - **In a complete ontology that included all axioms of the schema**
      - ***Situation ➔ includes SOME Observable_type_X***
        for all observables

- **Consequences:**
  All attempts to organise a subsumption hierarchy based on the Finding/Observable distinction are doomed to failure!
    - **Because it is a meta-distinction**
      - ***Requires higher order rather than first order logic***
        - *... although many branches of the subsumption hierarchy consist only of findings or*
          only of observables

Developmental Modularisation and OWL - Plug and Play development

- OWL provides powerful import mechanims
  - Because an OWL ontology is just a collection of axioms, importing just means adding in the axioms
    - Order independent.
- Need to define interfaces beween modules
  - Analogy to API between java packages
    - OWL equivalence and subclass axioms provide a means of defining interfaces between ontologies
      - *Usually by means of a separate module - an Ontology Binding Interface*

Using an Ontology Binding Interface



Technical aside

- **Possible in any DL or other based representation**
  - **Any language in which adding modules is “just adding axioms” and axioms are “monotonic”**
    - **But not exploited until recently**
      - ***OWL has stimulated the interest but***

Parts and Wholes
"Adapted SEP Triples"

- **Requirement**
  - **Distinguish disorders and procedures on the whole from those on the whole or its parts**
  - **Example 1**
    - **Our usual meaning of "Heart disease" is really**
      "Disease of the heart or any of its functional parts"
    - **But "Cardiomyopathy" affects the heart muscle as a whole**
      - ***Usually but not always classes of diseases of the whole include diseases of the parts***
        - *In general faults of a part are faults of the whole*
  - **Example 2**
    - **An "Amputation of a finger" is a "Procedure on the Hand"**
      but not a "Amputation of the hand"
      - ***Removals act on wholes***
        although in general classes of procedures include procedures on parts

Ironic aside

- Property-paths were originally developed to meet this requirement
  - GALEN's refined_along/specialised_by
  - SNOMED's right identities
  - OWL's property paths
- There is a choice to put the burden on the properties or the classes
  - But it seems more intuitive to put it on the classes
    - Lots of "druidisms" go away
      - *Inspired by SEP triples from Schulz and Hahn*
- ... but property paths turn out to be powerful inference mechanisms for other purposes

Typical representation of most broad disease and procedure classes

- **Typical**
  - ***Heart_disorder*** ≣
    Disorder THAT has_locus SOME
    (Heart OR is_clinical_part_of SOME Heart)
- **But**
  - ***Cardiomyopathy*** ≣
    Myopathy THAT has_locus SOME Myocardium
- **Typical**
  - ***Procdure_on_hand*** ≣
    Procedure THAT has_locus SOME
    (Hand OR is_part_of SOME Hand)
- **But**
  - ***Complete_amputation_of_finger*** ≣
    Removal THAT has_locus SOME Finger

Technical Aside

- Does not actually require true negation
  - Can be done with “Psuedo-disjunction” - A B
  - syntactic sugar for creating SEP triples only wen you need them
  - Formally:
    - A lowest primitive concept A B that subsumes both A and B
    - A ➔ (A B) ➔ (A ∨ B)
      B ➔ (A B) ➔ (A ∨ B)
    - but not
      (A ∨ B) (A B)

Summary

- A cleaner set of schemas is possible
  - Situations allow a clean revision of context model
    - Eliminate of separate axis for “Situations with explicit Context”
    - Make clear distinction between “recordable codes” and “kernel codes”
    - Clean separation of:
      - *Negation, modality, temporal markers and subject of care*
        - *A formalism with negation handles classic troublesome cases easily*
  - Adapted SEP triples make part-whole representations clearer
- DLs allows modularisation and defintion of Ontology Binding Interfaces
  - A strategy for building in pieces and “pluggable” extensions
- Only selective use of OWL is needed
  - And only negation requires anything that can’t be encoded in EL+
    - Dilemma of complete inference over theory conaining known falsehoods or incomplete inference over a theory without them
  - Choices require empirical tests at scale - e.g. 25K concepts
    - The time to do it is NOW!