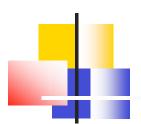
An Advanced Clinical Ontology

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Three-year project: April 2007- March 2010
A couple of thousands of diseases with
necessary/sufficient types to characterize them



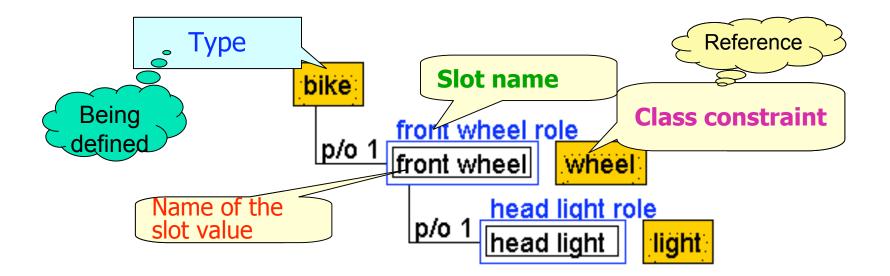
Issues discussed in our ontology

- is-a/part-of issue
 - <finger *part-of* hand>
 - <Injury of a finger *is-a* injury of a hand>
 - Introduction of "p-" operator
- Commonality vs. specificity
 - Introduction of generic structural/disorder components
- Three-layer quality description
 - Exploitation of Hozo's role representation
- Multiple perspectives for classification
 - On-demand is-a hierarchy generation
- Multiple perspectives for an entity
 - Introduction of viewpoint-specific definition function (to be implemented soon)

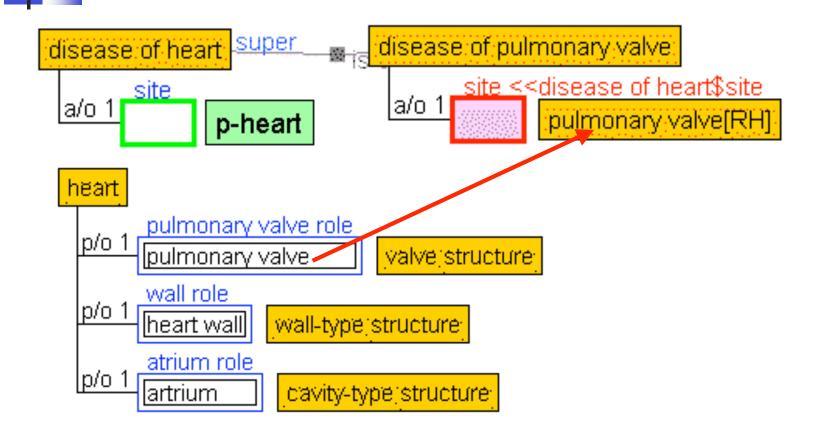


Type definition in Hozo

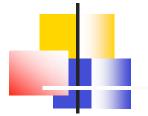
- Mutual dependency between the whole and its parts
- Any part has a role in the context of the whole
- Any part must be defined in the context of the whole
- Tight relationship between theories of roles and parts



Is-a & part-of issue Inheritance: a special case



What is "p-" operator?



Any part of X is-a p-X

p-X = a generic concept representing all parts of X

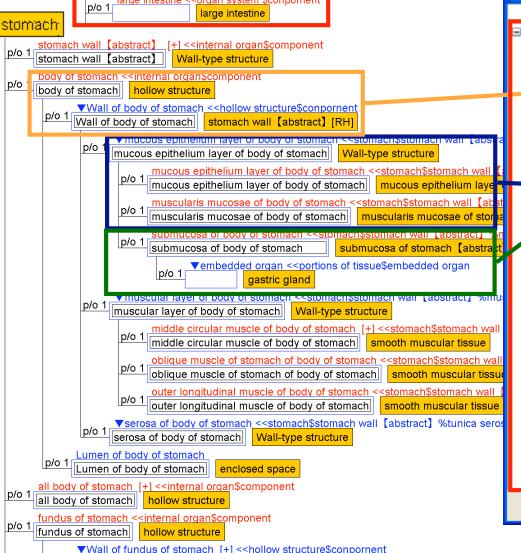
```
left atrium role
p/o 1 left atrium role
p/o 1 right atrium role
p/o 1 right atrium role
cavity-type structrure
cavity-type structrure
cavity-type structrure
cavity-type structrure
cavity-type structrure
cavity-type structrure
p/o 1
p-atrium
```

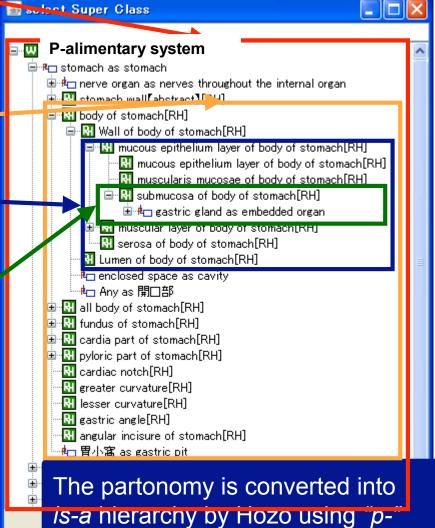
atrium = an entity composed of right and left atriums
 p-atrium = a generic concept representing all parts of atrium
 Left atrium is-a p-atrium; Left atrium part-of atrium
 Right atrium is-a p-atrium; Right atrium part-of atrium



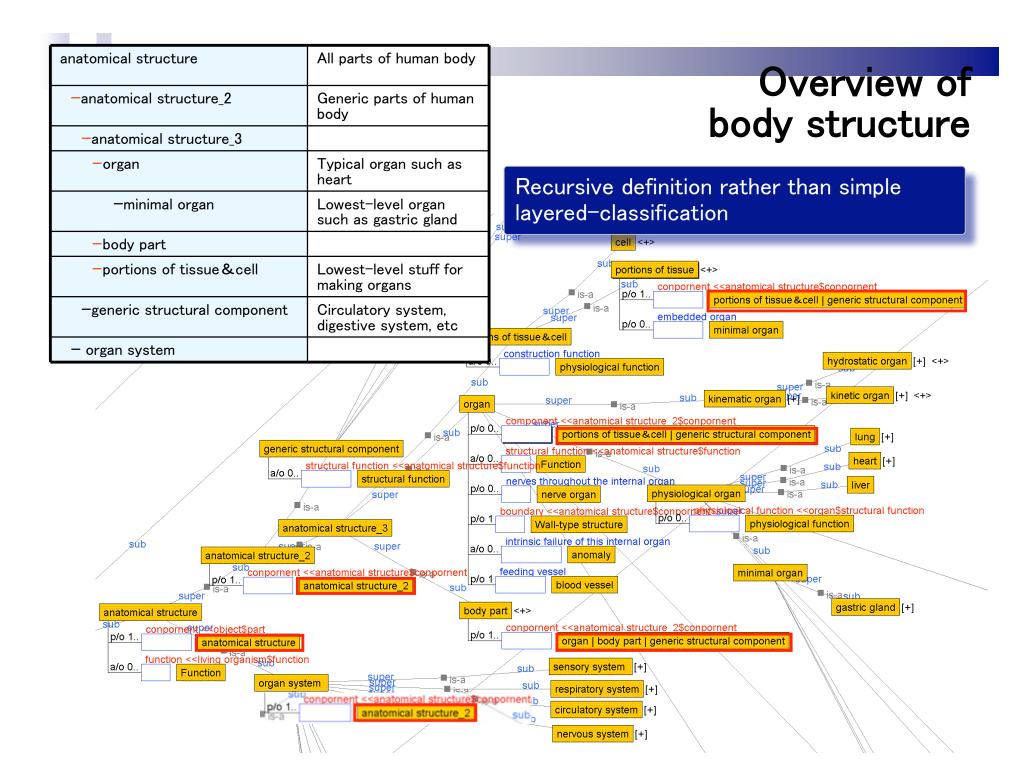
alimentary system p/o 1 stomach < organ system \$conpornent stomach p/o 1 esophagus < organ system \$conpornent esophagus p/o 1 small intestine < organ system \$conpornent small intestine p/o 1 large intestine < organ system \$conpornent large intestine p/o 1 large intestine

is-a hierarchy generation from *part-of* information

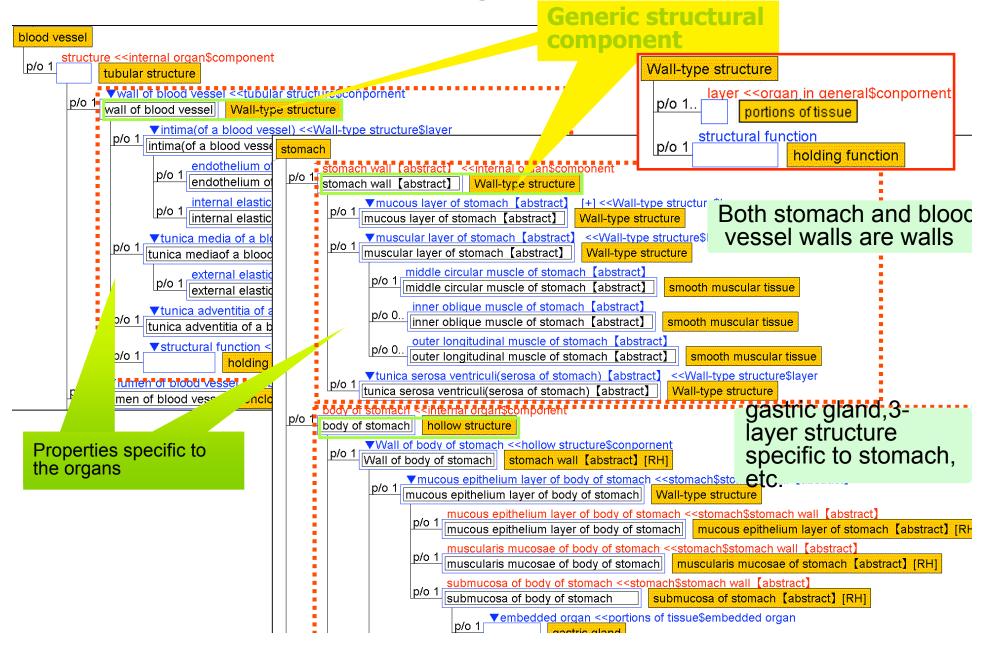




operator technology.



Introduction of generic structural



Concrete example

p/o 1

- Stomach is composed of body of stomach, cardia, etc.
- Defined using hollow structure
- It has cavity and wall
- The wall is composed of mucous, muscular and serosa layers. It has functions to secrete pepsinogen, etc.
- Gastric wall is further specialized to submucosa and cells which has gastric gland.

```
stomach wall【abstract】
                                                         Wall-type structure
 body of stomach <<internal organ$component
body of stomach
                                        hollow structure
                  ▼Wall of body of stomach <<hollow structure$conpornent</p>
                Wall of body of stomach
                                                                          stomach wall [abstract] [RH]
                                                                                                                                      <<stomach$stomach wall [abstract] %muco</p>
                                 mucous epithelium layer of body of stomach
                                                   mucous epithelium layer of body of stomach <<stomach$stomach wall [abstract] %
                                                  mucous epithelium layer of body of stomach
                                                                                                                                                    mucous epithelium laver of stomach
                                                  muscularis mucosae of body of stomach
                                                                                                                                             muscularis mucosae of stomach [abstraction of the control of the c
                                                  submucosa of body of stomach <<stomach$stomach wall [abstract] %mucous laye
                                                  submucosa of body of stomach
                                                                                                                                    submucosa of stomach [abstract] [RH]
                                                                                                                                    of tissue$embedded organ
                                 ▼muscular layer or body or st macri <<stomach$stomach wall [abstract] %muscular layer
                                  muscular layer of body of stomach | Vvall-type st cture
                                                                                                                                                +] <<stomach$stomach wall [abstract]
                                                  middle circular muscle of body of stomach
                                                                                                                                                  smooth muscular tissue
                                                  oblique muscle of stomach of body of sto
                                                                                                                                                ch <<stomach$stomach wall 【abstract】
                                                 gastric gland
                                    p/o
                                                                  component <<organ 2$component
                                                    p/o 1
                                                                                             glandular structure
                                                                                    ▼lumen of gastric gland <<glandular structure$wall of gland
                                                                                   lumen of gastric gland
                Lumen of bo
                                                                                                         ▼component <<tissue$conpornent</p>
                                                                                       p/o 1..
                                                                                                                                  chief cell of stomach (gastric chief cell)
 all body of stomach
all body of stomach
                                                                                                                          ▼function of chief cell of stomach <<chief cell of</p>
                                                                                                                                                    pepsinogen-secretory function [RH
                wall of stoma
                                                                                                         ▼component <<tissue$conpornent
                                                                                      p/o 1.
                                                                                                                                   parietal cell of stomach (gastric parietal cell)
                    p/o 0..
                                                                                                         ▼component <<tissue$conpornent</p>
                                                                                     p/o 1
                                                                                                                                   mucous neck cell
                   p/o 0...
                                                                  function
                                                   a/o 1
                                                                                             secretion function
                    p/o 0.
```

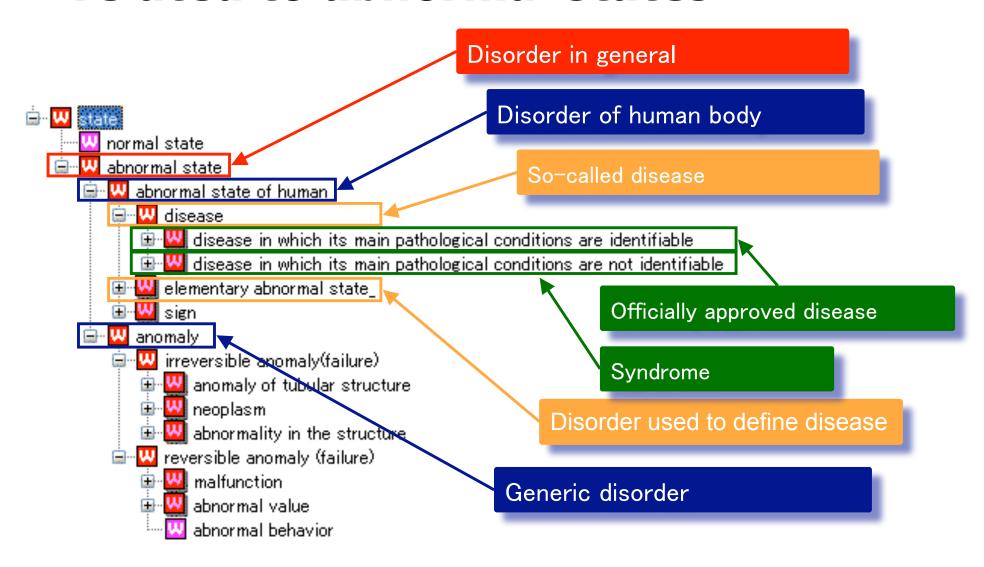


Disease modeling

- Issues and decisions -

- Various states: pathological state, disease, disorder, dysfunction, failure
 - All are kinds of disorder of human body
 - Introduction of generic disorder to use for defining organ- and diseasespecific disorder
 - The generic disorder is composed of reversible and irreversible states
- Various viewpoints of diseases
 - Necessity of common and fundamental viewpoint
 - A fundamental classification as an infra, and on-demand generation of other classifications according to the viewpoint specified
- Three-layer quality description
 - Exploitation of Hozo's role representation
- History of transition of disease names
 - Names associated with time slot
- Exclusion of transition of disease state (future work)

Upper types related to abnormal states

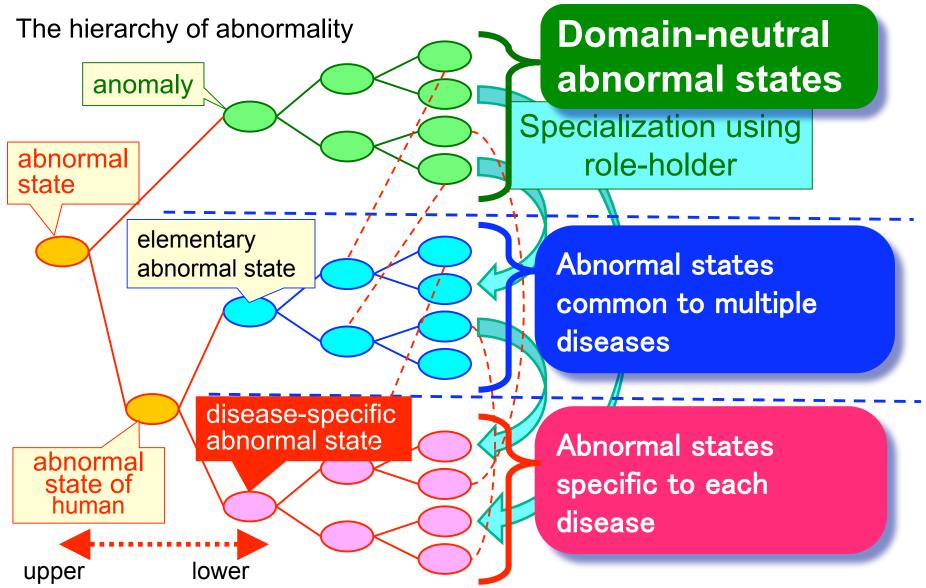






Three-layer description of abnormal states

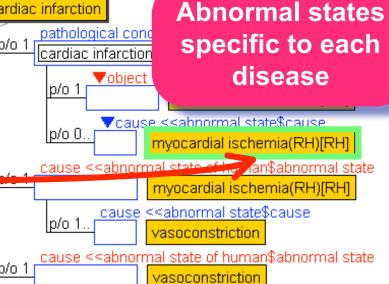




An example of specialization using the idea of role holder in Hozo

state of ischemia a/o 1 abnormal state <<ahnormal state of human\$abnormal a/ó 1 ischemia deficient quantity per unit time ▼lackings <<deficient quantity per unit time:</p> a/o 11 oxygen nt necessary <<deficient quan la/o 1 **Abnormal states** quantity per unit time common to multiple provided <<deficient quantit la/o 1 quantity per unit time diseases cardiac infarction myocardial ischemia p/o 1 pathological cond cardiac infarction abnormal state <<abnormal state \text{human\$abnormal} p/o 1.. myocardial ischemia(RH) ▼object |p/o 1 | objed |p/o 1 r cardiac muscle tissue[RH] |p/o 0...| deficient object **deficient quantity per unit tin la/o 1 oxygen ent <<deficient quantity per un Abnormal states quantity per unit time |p/o 1...**[** common to multiple supplied <<deficient quantity p diseases p/o 1 quantity per unit time

```
| lackings << lack | Domain-neutral | abnormal states | amount | a/o 1 | amount | a/o 1 | amount | amount | provided << lacking state$lake | a/o 1 | alo 1 | amount | amount | provided << lacking state$lake | quantity per unit time | a/o 1 | physicale | physicale | a/o 1 | physicale | a/o 1 | a
```

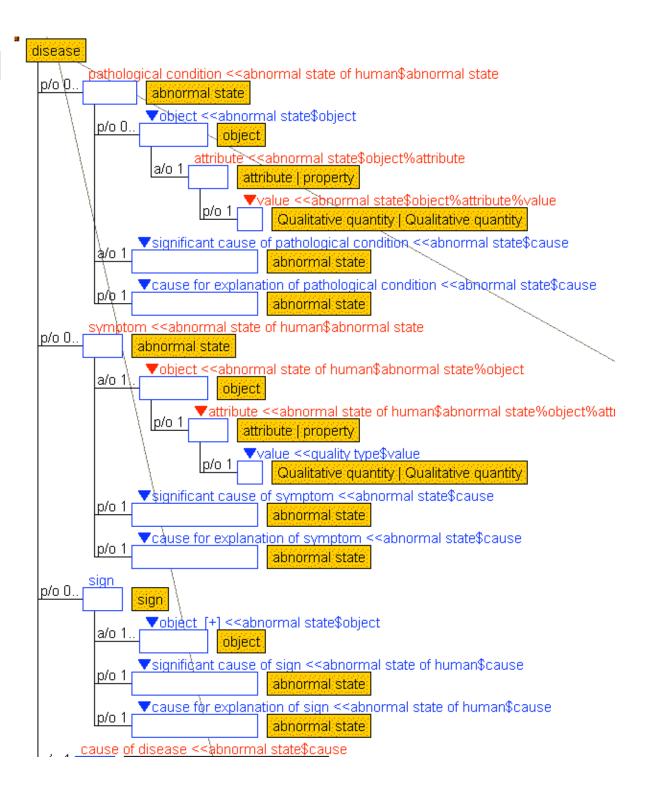


abnormal state p/o 0.. al normal state Disease model a/o 1.. a)(o 1 attribute | property √value << quality type\$ value </p> type:1/diabetes disease name <<diabetes\$disease name p/o 1 disease name A disorder is composed of several ▼ICD-10.id <<diabetes\$disease name%ICD-10.id</p> a/o 1 E10 finer-grained disorders ▼disease named system <<disease name\$disease named system disease pathological condition <<diabetes\$pathological condition type 1 diabetes hyperglycemia chronic hyperglycemic condition[RH] p/o 0.. [abnormal state **Object** cause <<diabetes\$pathological condition%cause ▼object <<abnormal state\$object lower serum insulin level of type 1 diabetes[RH] b/o 0.. **Parameter** thogenesis of disease <<diabetes pathogenesis of disease linkage attribute | property normal state\$objec Abnormal Type lower serum insulin level of type 1 diabetes causal chain p/o 1.. educed amount of insulin that is produced All kinds of disorders are captured by reduced amount of insulin that is produced[RH <E, A, V> formalism condition of chronic hyperglycemic condition chronic hyperglycemic condition <<elementary abnormal state of chronic hyperglycemic condition abnor Object Disorders referred to by many are Vobject <<abnormal attribute value\$obi a/o 1 defined as basic types blood **Parameter** attribute <<abnormal attrib a/o 1 blood suga Abnormal Type Causal chains are modeled as a value <<abnormal at the value sobjects high sequence of disorder states



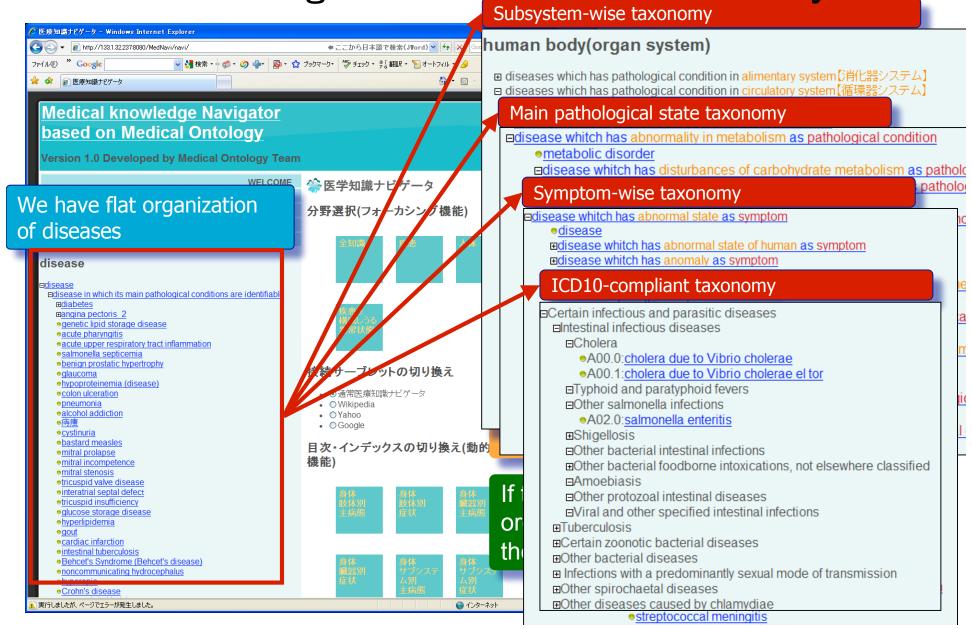
How to dynamically generate *is-a* hierarchy

- Diseases are organized based on states in rather flat structure
- By choosing the slot the user is interested in, then diseases are organized according to the is-a hierarchy of its class constraint.





On-demand generation of is-a hierarchy





Concluding remarks

- Achievements
 - P-Operator for is-a/part-of issues
 - Commonality vs. specificity
 - Introduction of generic structural/disorder components
 - Three-layer quality description
 - Exploitation of Hozo's role representation
 - Multiple perspectives for classification
 - On-demand is-a hierarchy generation
- Future work
 - Increasing the number of diseases
 - Alignment with the existing ontologies