

One Medicine One Pathology: 2nd annual CASIMIR Symposium
on Human and Mouse Disease Informatics

UMLS and phenotype coding

Anita Burgun, Fleur Mouglin, Olivier Bodenreider

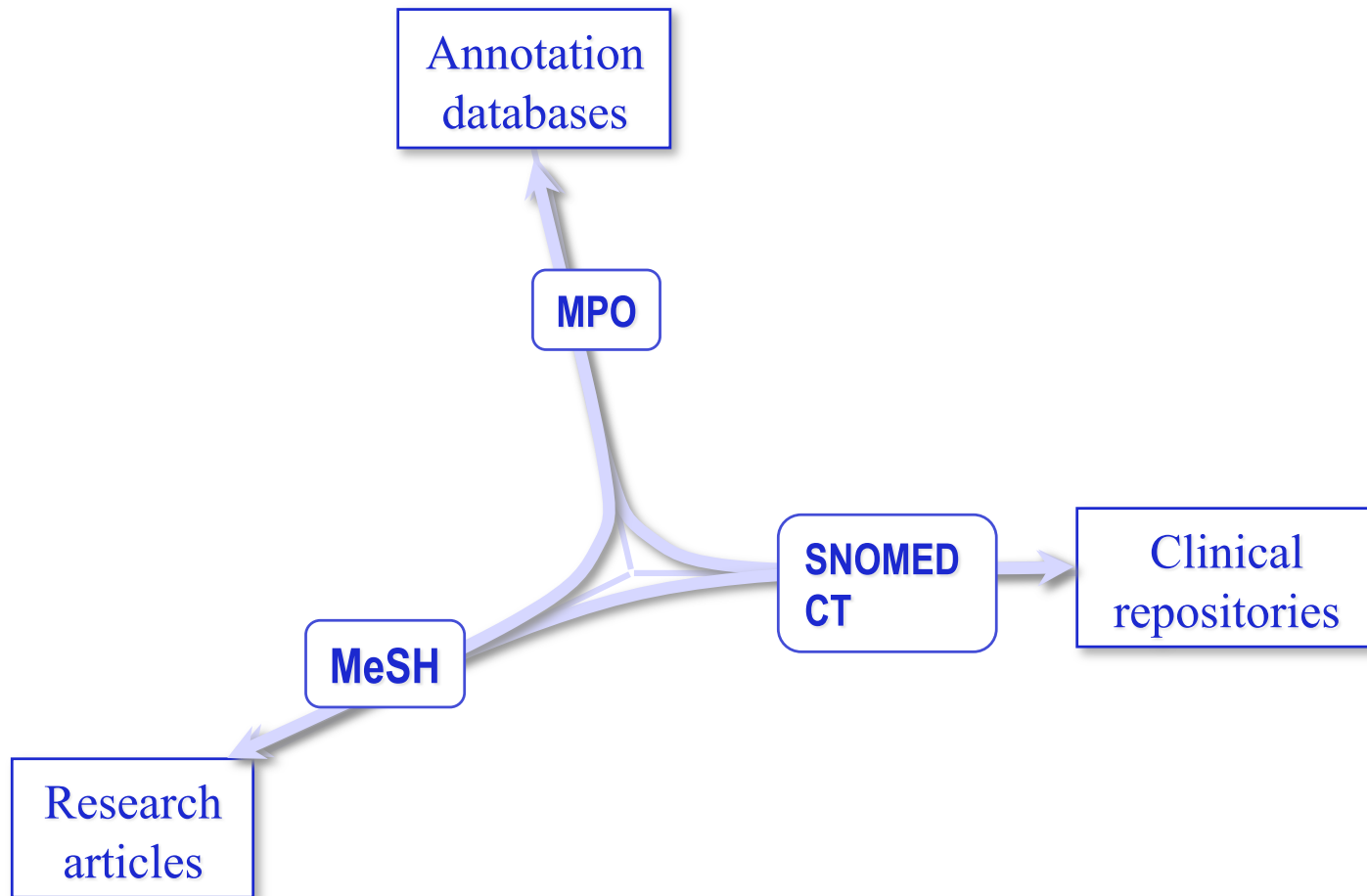
INSERM U936, EA 3888- Faculté de Médecine, Univ. Rennes1

ISPED, Univ. Bordeaux, France

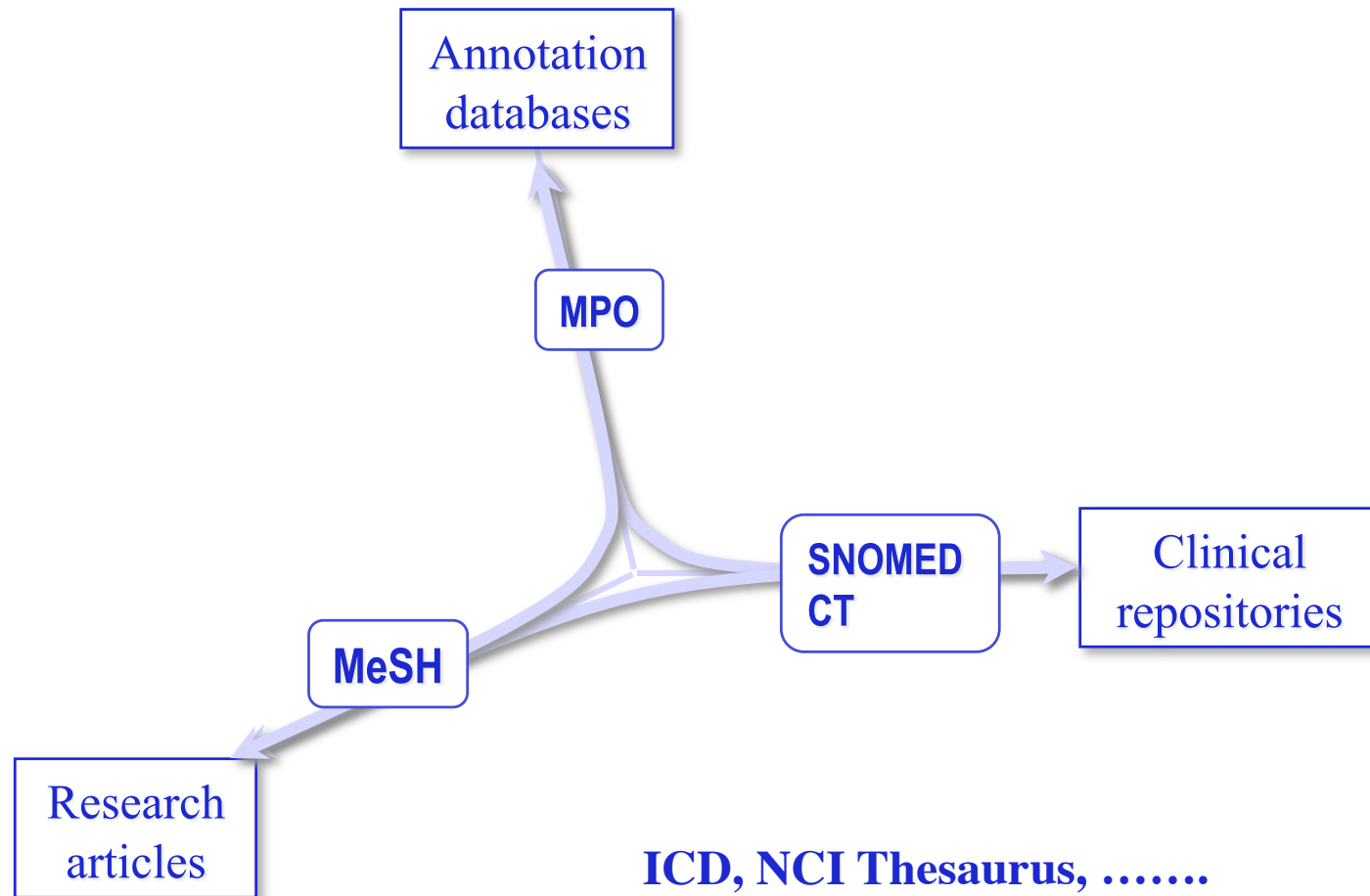
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December 2, 2008

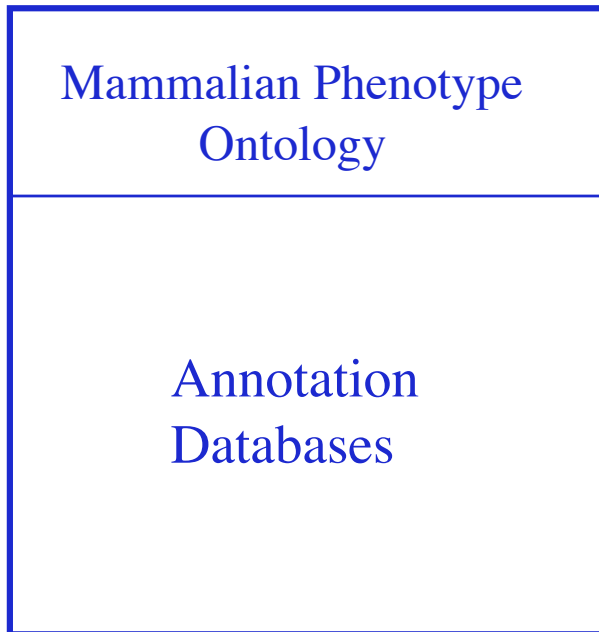
Phenotype coding



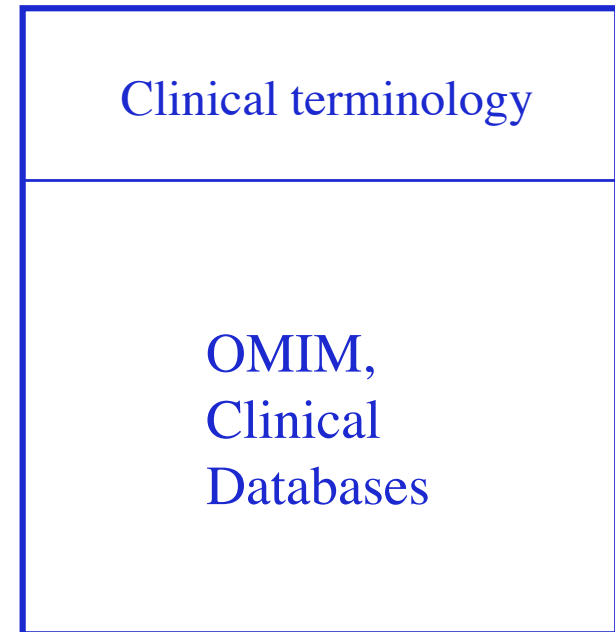
Phenotype coding



Phenotype coding: role of the UMLS

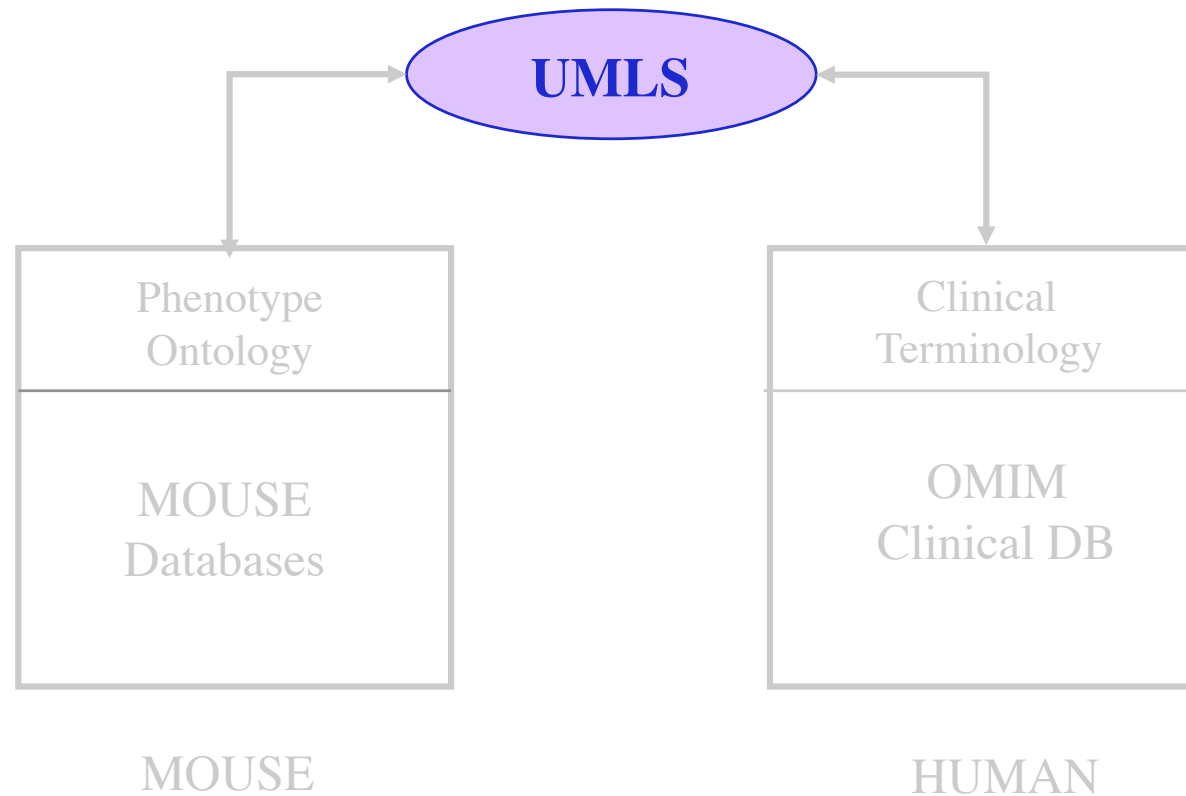


MOUSE



HUMAN

Phenotype coding: role of the UMLS



Unified Medical Language System

- Addresses heterogeneity issues
 - More than 100 source vocabularies
 - Unification
- Clusters terms into concepts
 - Metathesaurus: CUIs
- Organizes hierarchies
 - Metathesaurus: relations
- Categorizes concepts
 - Semantic Network: Semantic Types

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UMLS Metathesaurus

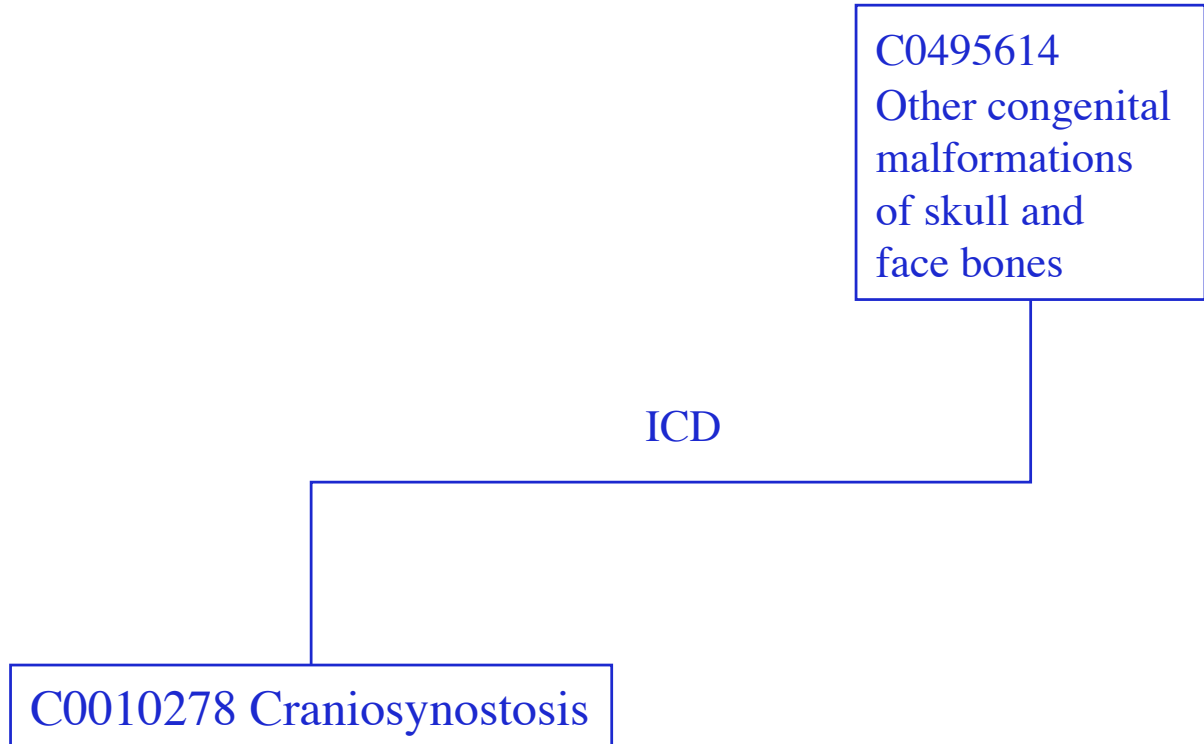
- Craniosynostosis in UMLS Release 2008AA
- Source vocabularies
 - ICD-10
 - ICPC
 - MedDRA
 - MeSH
 - OMIM
 - Read Codes
 - SNOMED CT
 -
- Definition (MeSH) : Premature closure of one or more sutures of the skull

- UMLS Metathesaurus
- CUI C0010278 Craniosynostosis (Preferred Term)
 - Craniostenosis (ICD, ICPC, OMIM, SNOMED CT)
 - Craniosynostosis syndrome (SNOMED CT)
 - Synostosis (cranial) (CRISP)
 - *Word phrases*
 - Premature closure of cranial sutures (MedDRA, SNCT)
 - Congenital ossification of cranial sutures
 - Congenital ossification of sutures
 - Congenital ossification of sutures of skull
 - Premature cranial suture closure (SNOMED)
 - *Abbreviations*
 - CRS, CSO, CRS1 (OMIM)
 - *More specific terms*
 - Craniosynostosis, type 1 (OMIM)
- Possible synonyms and related
 - Hurst syndrome (C0014077)
 - Christian syndrome 1 (C0795794)
 - SCARF (skeletal abnormalities, cutis laxa, craniostenosis, psychomotor retardation, facial abnormalities) syndrome (C0796146).....

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ICD-10



MedDRA

C0852332
Musculo
skeletal
and
connective
tissue
deformities
of skull,
face
and buccal
cavity

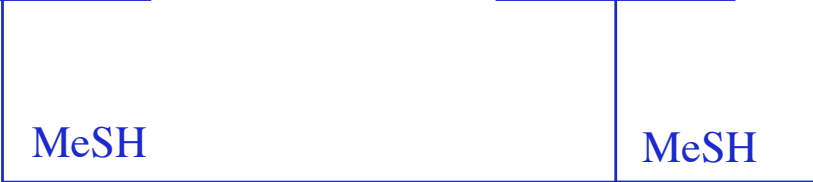
MedDRA

C0010278 Craniosynostosis

MeSH

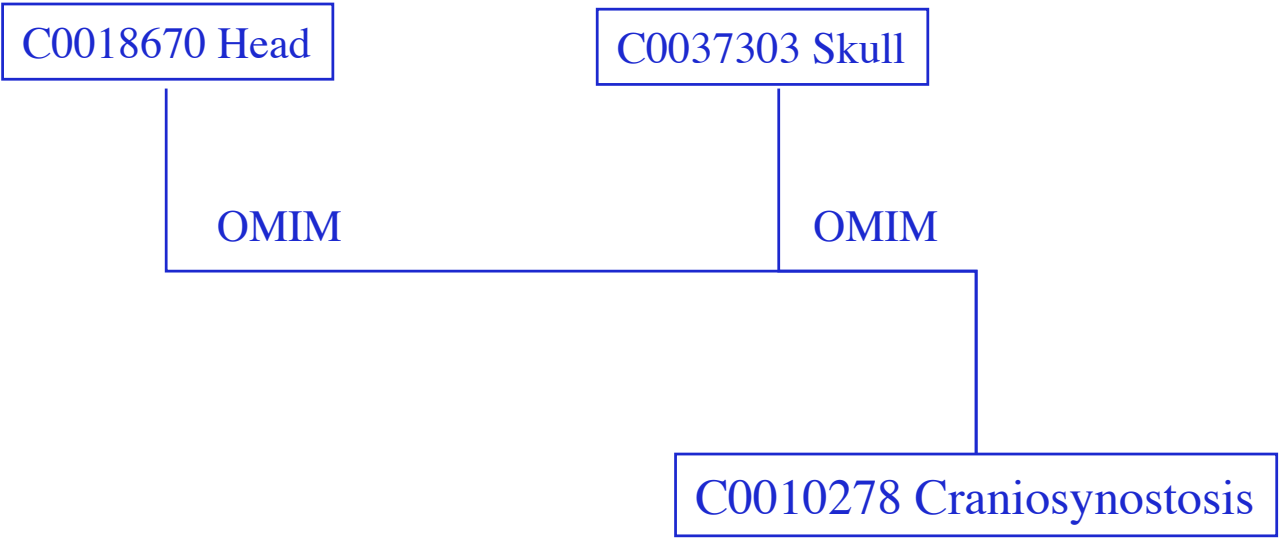
C0376634
Craniofacial
Abnormalities

C0039093
Congenital
abnormal
synostosis

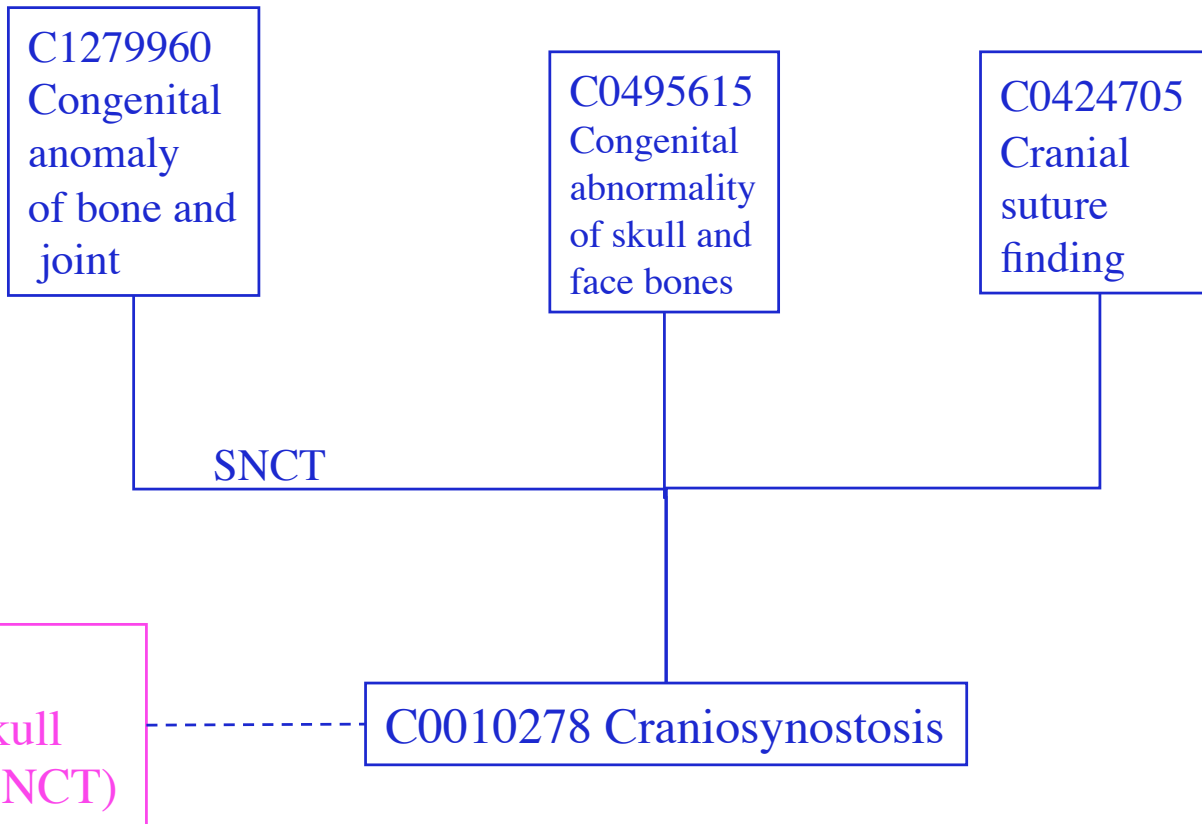


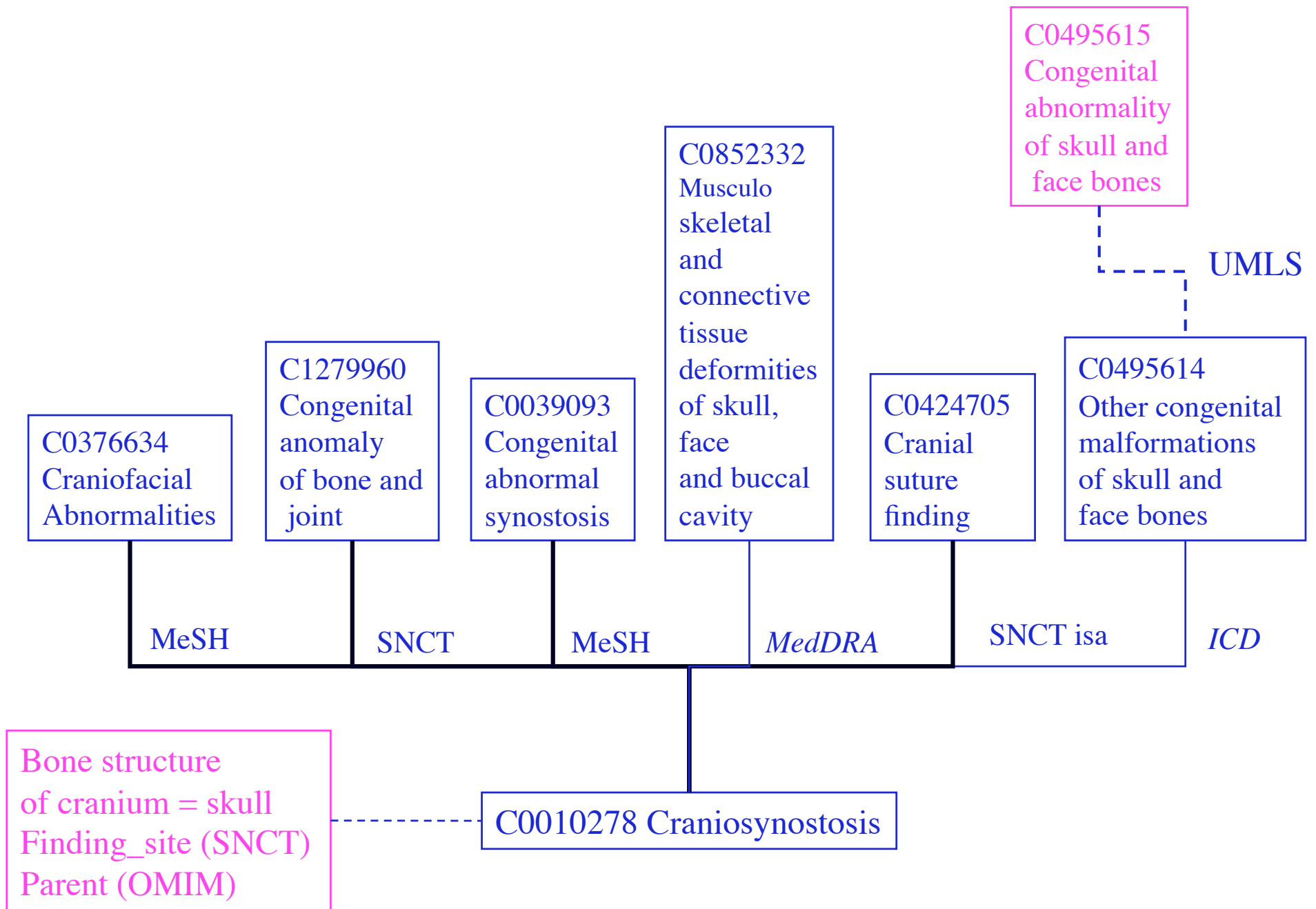
C0010278 Craniosynostosis

OMIM



SNOMED CT

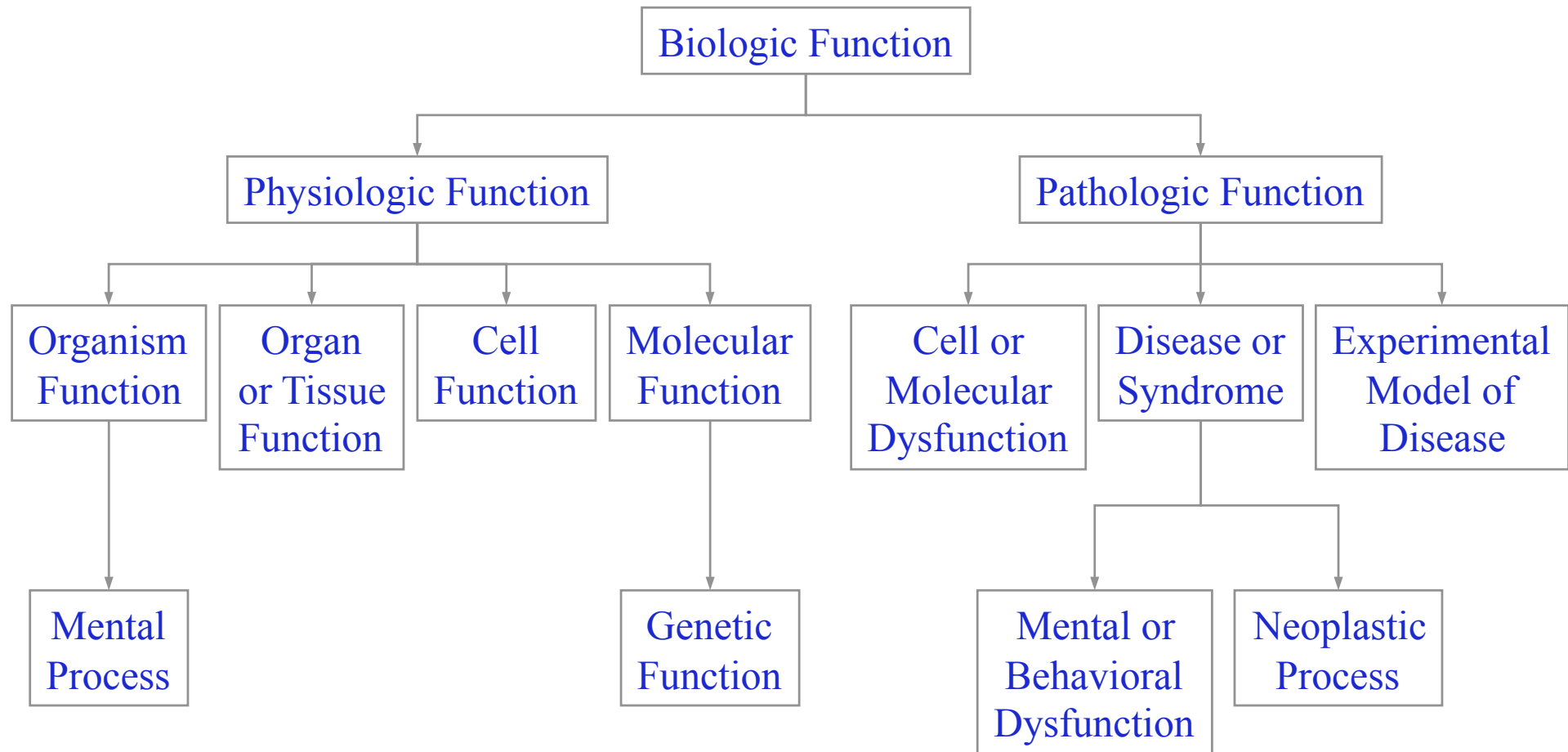




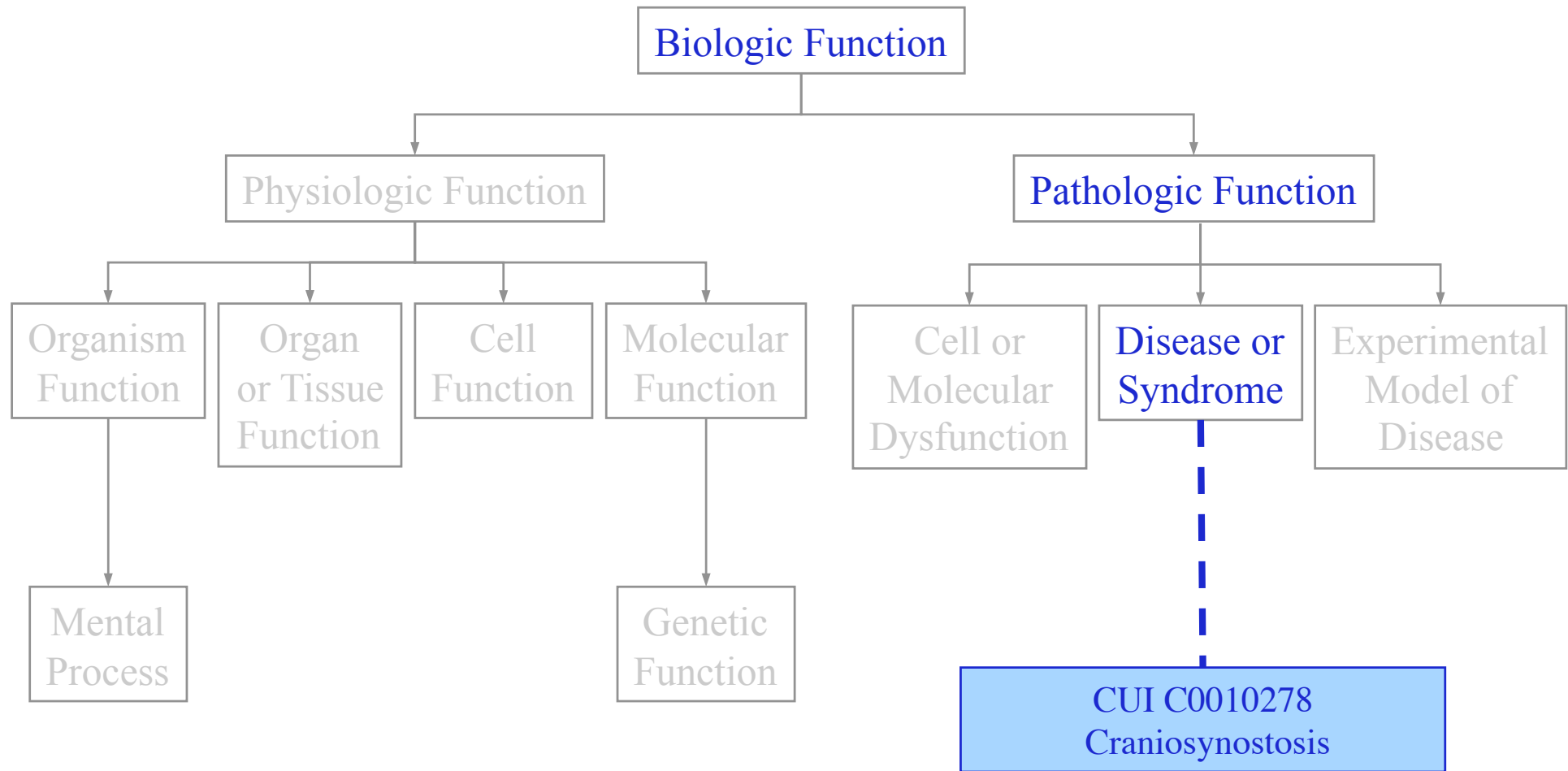
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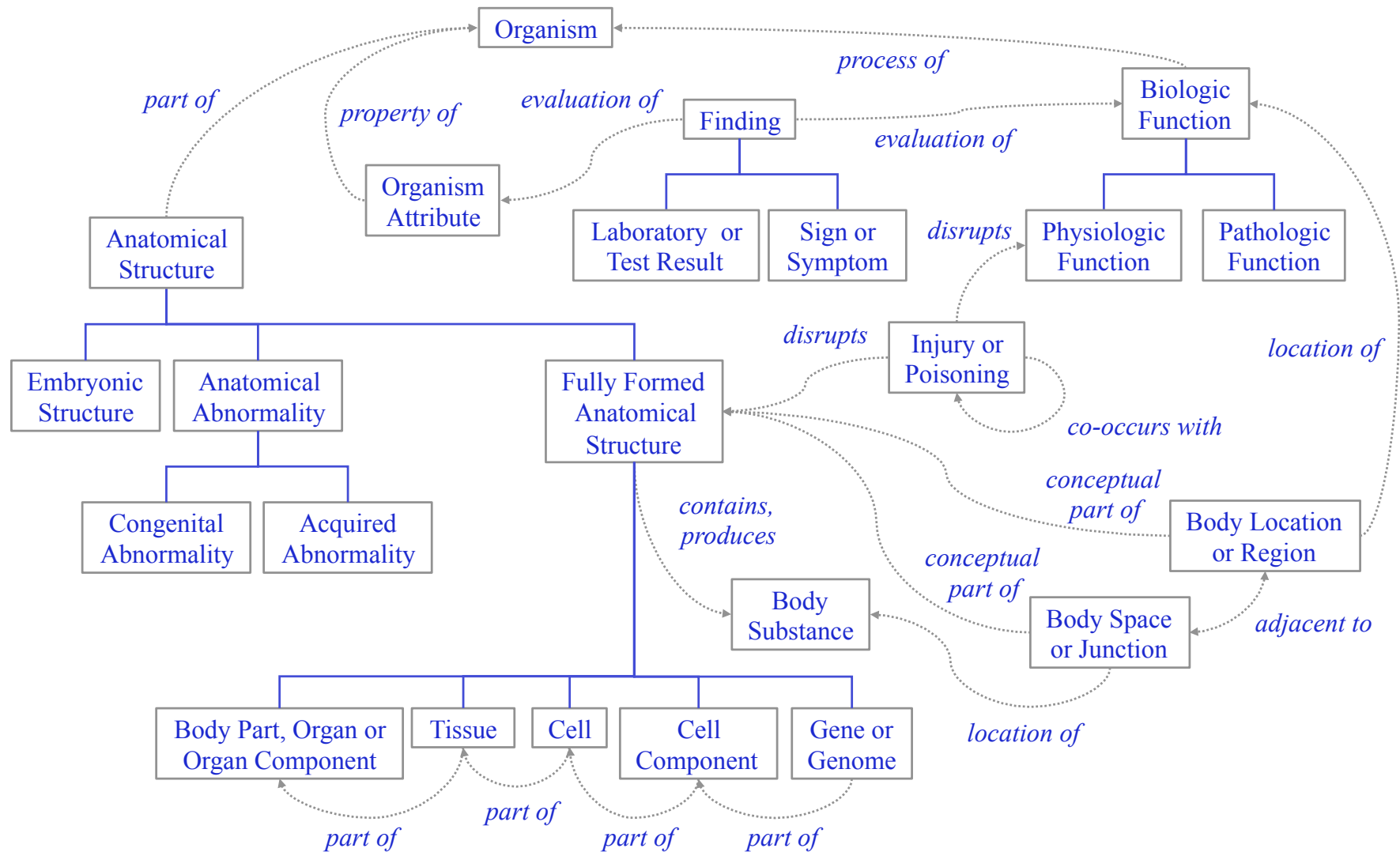
“Biologic Function” hierarchy (isa)



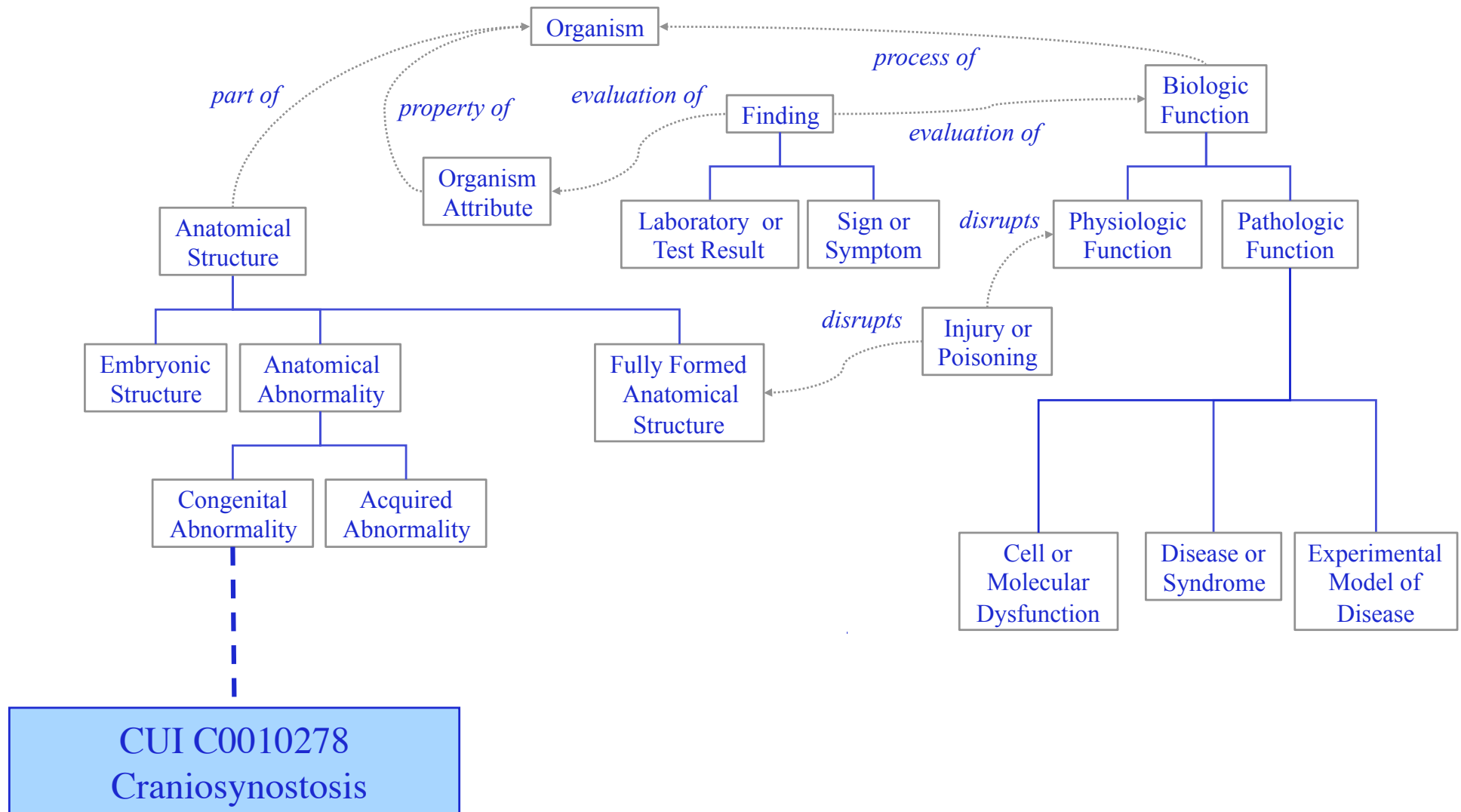
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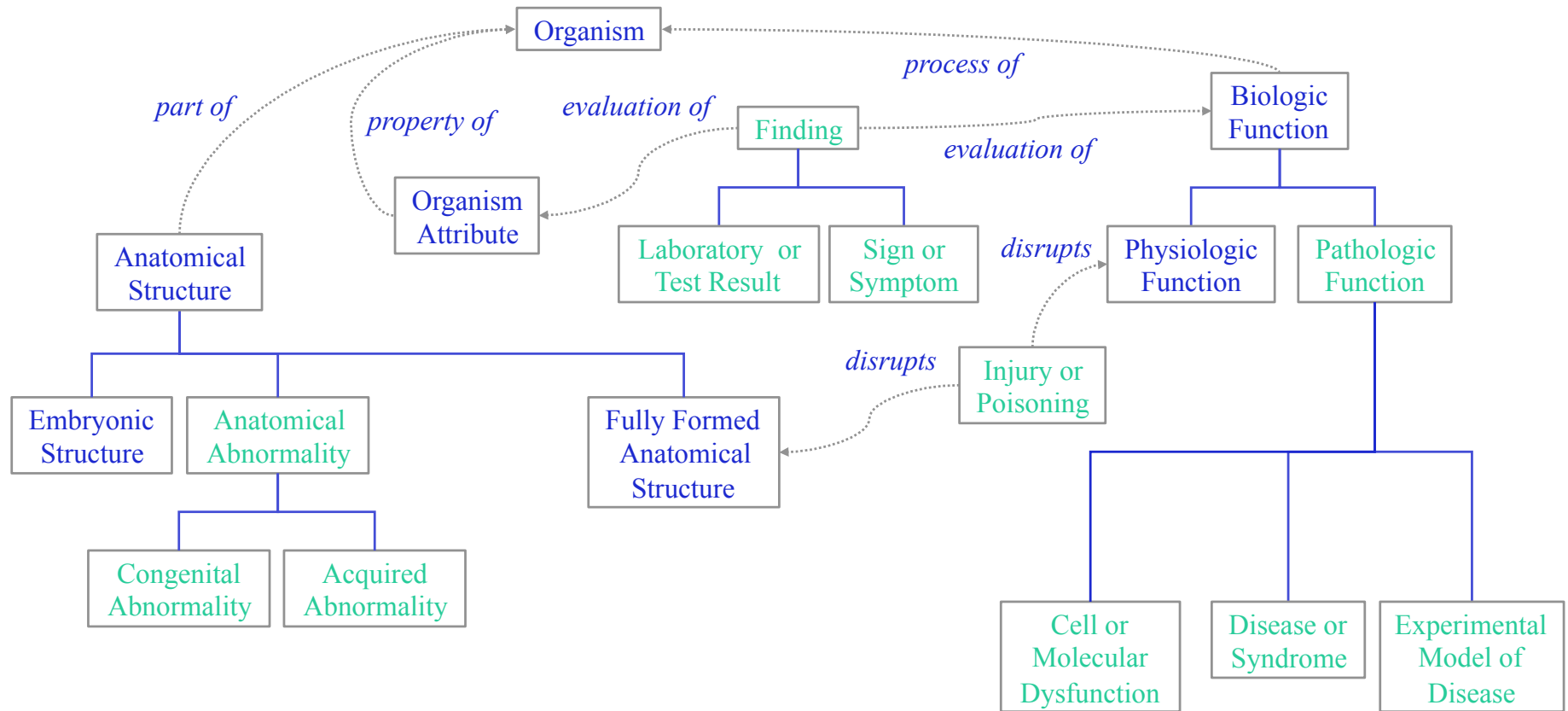
Overview of the Semantic Network



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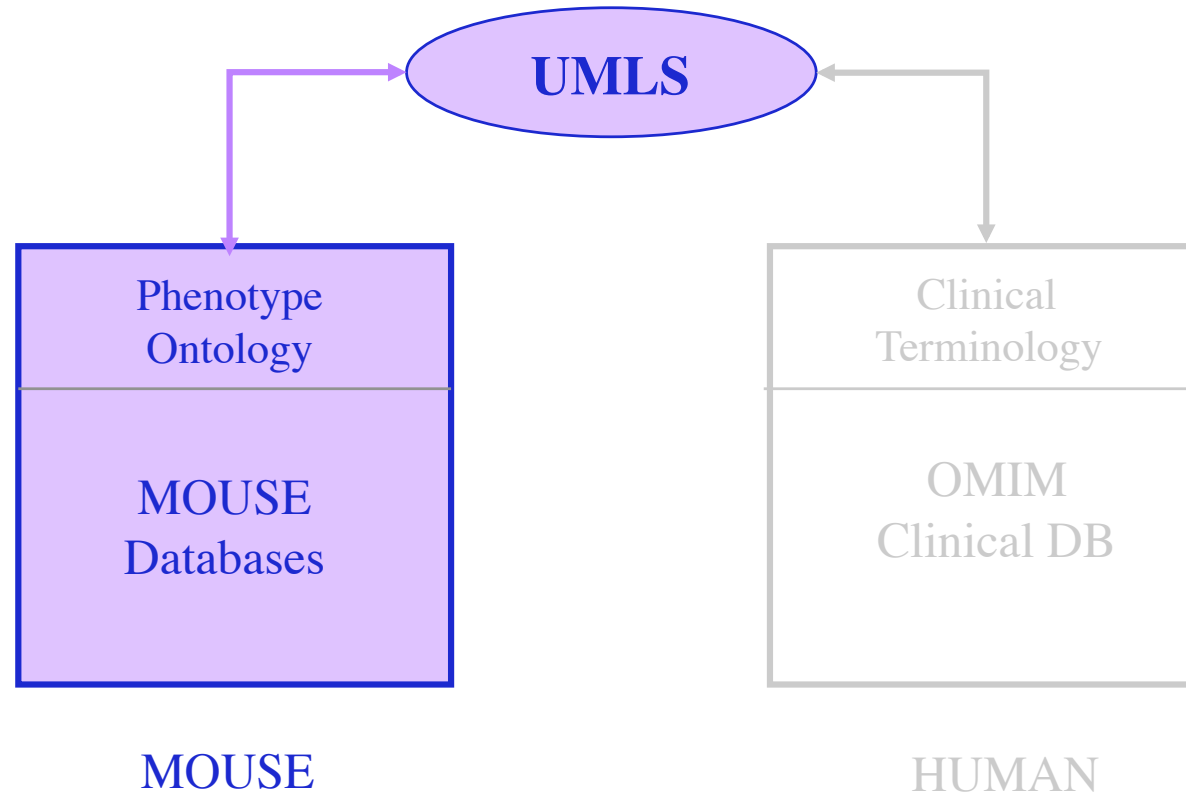


Semantic Groups



Disorders

Phenotype coding: role of the UMLS



Mammalian Phenotype Ontology (MPO)

- 14,662 terms
- 6,307 concepts
- MP:0003561 Rheumatoid arthritis
- MP:0000218 increased leukocyte cell number
 - increased leukocyte count
 - increased WBC count
 - increased WBC number
 - increased white blood cell number
 - leukocytosis
- MP:0000410 waved hair
 - curly hair
 - waved fur
 - wavy hair

Mapping to UMLS (1/3)

- Step 1 : Exact/normalized match
- Results
 - 2,065 MPO terms mapped successfully(14%)
 - 1,495 MPO concepts mapped successfully (24%)
 - Among them, 1,432 correspond to Disorders in UMLS (SG)
- Examples mapped successfully
 - MP:0000062 increased bone density-> C1141880 Bone density increased (NSI)
 - MP:0000081 craniostosis -> C0010278 Craniosynostosis (syn in MPO, EM)
 - MP:0000061 brittle bones -> C0029434 Osteogenesis Imperfecta (syn in UMLS, EM)
- Examples unmapped
 - MP:0000100 abnormal ethmoidal bone
 - MP:0000101 absent ethmoidal bone
 - MP:0000687 small lymphoid organs
 - MP:0000689 abnormal spleen structure

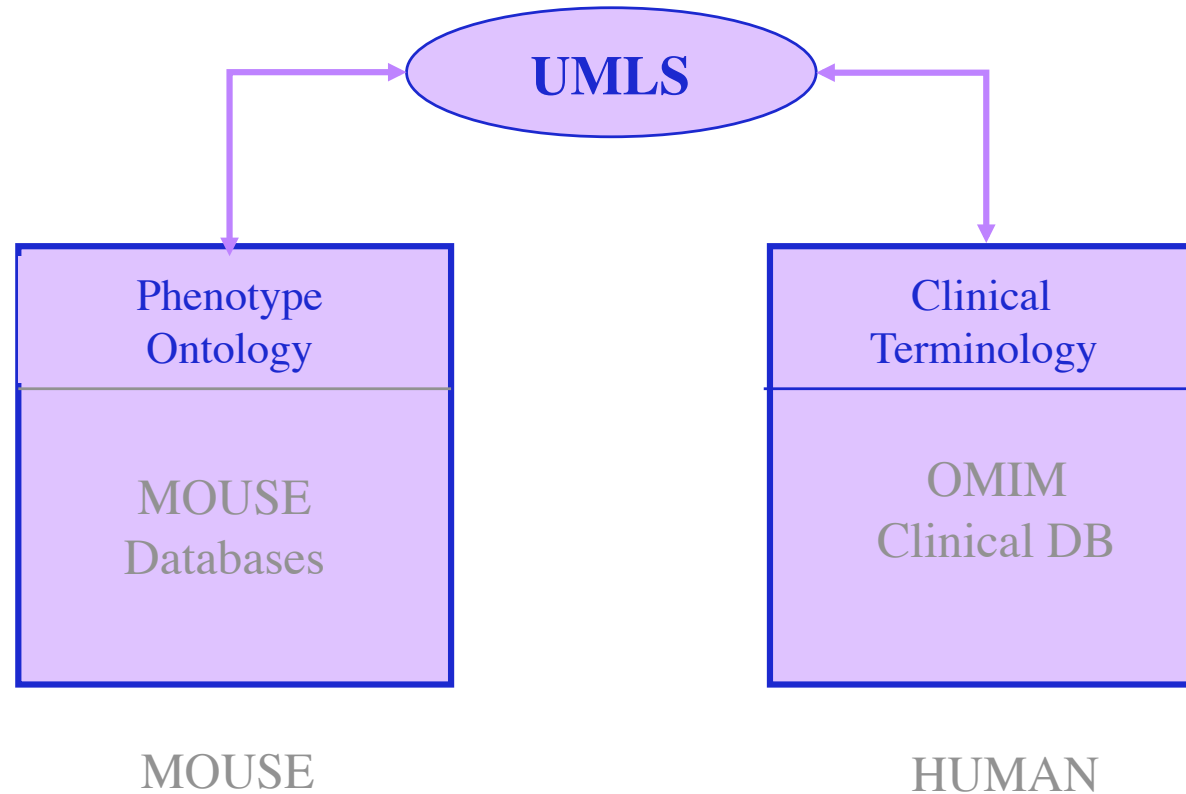
Mapping to UMLS (2/3)

- 11,466 unmapped terms (4,812 concepts)
 - MP:0000100 **abnormal** ethmoidal bone
 - MP:0000101 **absent** ethmoidal bone
 - MP:0000687 **small** lymphoid organs
 - MP:0000689 **abnormal** spleen structure
- Step 2 : demodification
 - 30 modifiers: abnormal, absent, small.....
 - Demodified terms
 - Mapping to UMLS
- Results : demodified terms
 - 9,845 <<modifier> xxx> terms in MPO
 - 7,925 unique terms (after demodification)

Mapping to UMLS (3/3)

- Results : mapping after demodification
 - 2,359 MPO terms mapped successfully after demodification (out of 11,466 , 20%)
 - Unique terms : 1,586 (20%)
 - 1,645 MPO concepts mapped successfully after demodification (out of 4,812, 34%)
- Demodified terms correspond mostly to:
 - Anatomical concepts
 - Semantic Group **Anatomy** in UMLS
 - 1410 terms, e.g., abnormal <anatomical structure>
 - MP:0000005 (increased) brown fat -> C0006298 Brown Fat (Tissue, **ANAT**)
 - Physiology
 - Semantic Group **Physiology** in UMLS
 - 516 terms, e.g., abnormal <physiological process>
 - MP:0000057 (abnormal) osteogenesis -> C0029433 Osteogenesis (Organ or Tissue Function, **PHYS**)

Phenotype coding: role of the UMLS



Discussion

- Compositionality in phenotype terms

< abnormal <anatomical structure > >

30 modifiers

Functional Concept

Qualitative Concept

abnormal
decreased
increased
reduced
small

Anatomy

Physiology

SGs

Class Name: abnormal (PATO:0000460)

Is A: deviation(from_normal)

Synonym: aberrant, atypical, defective

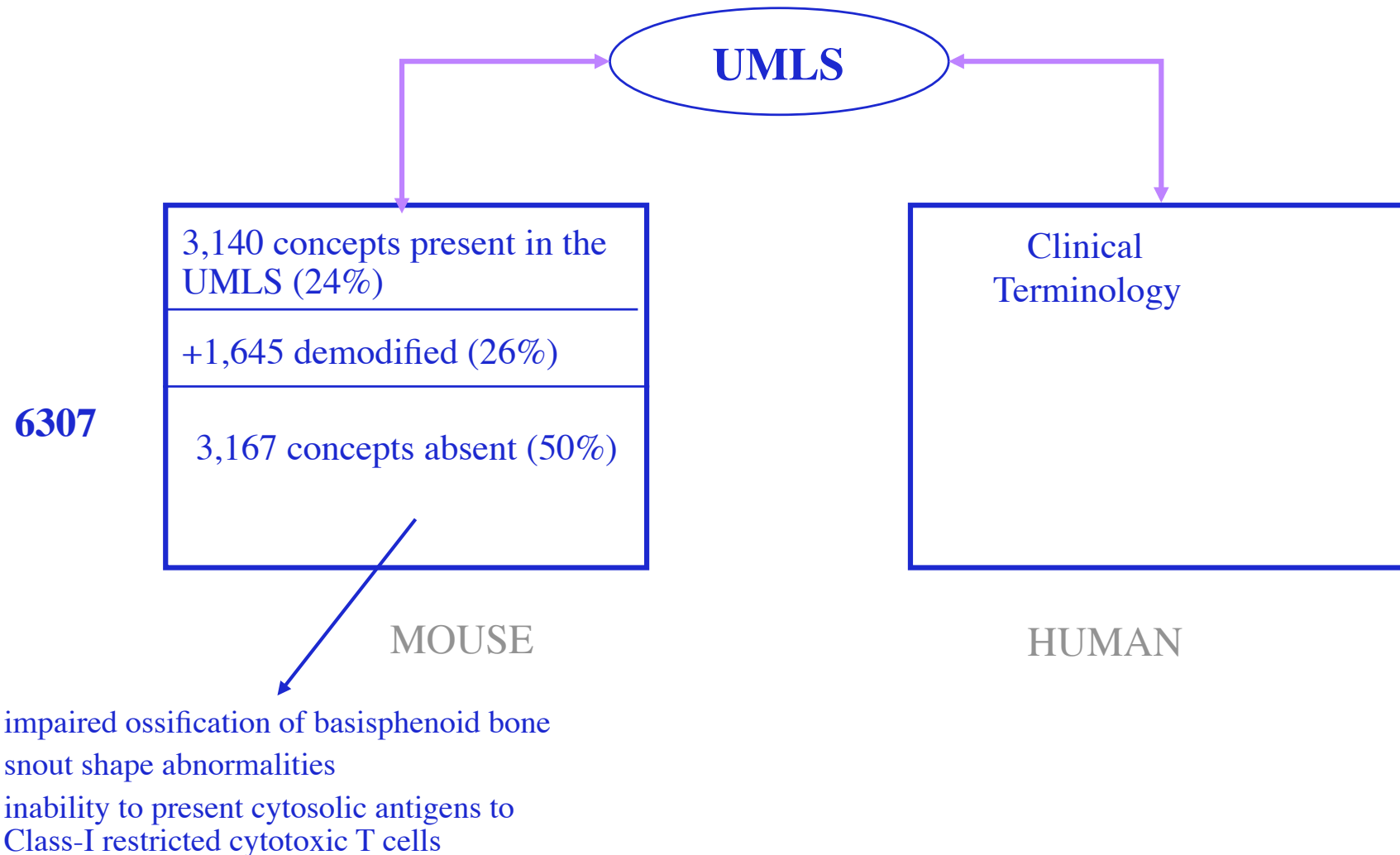
[R]Is A: pathological

entities from any ontology

Phenote : EQ model : combine entities from any ontology
with qualities / traits (such as those in PATO)

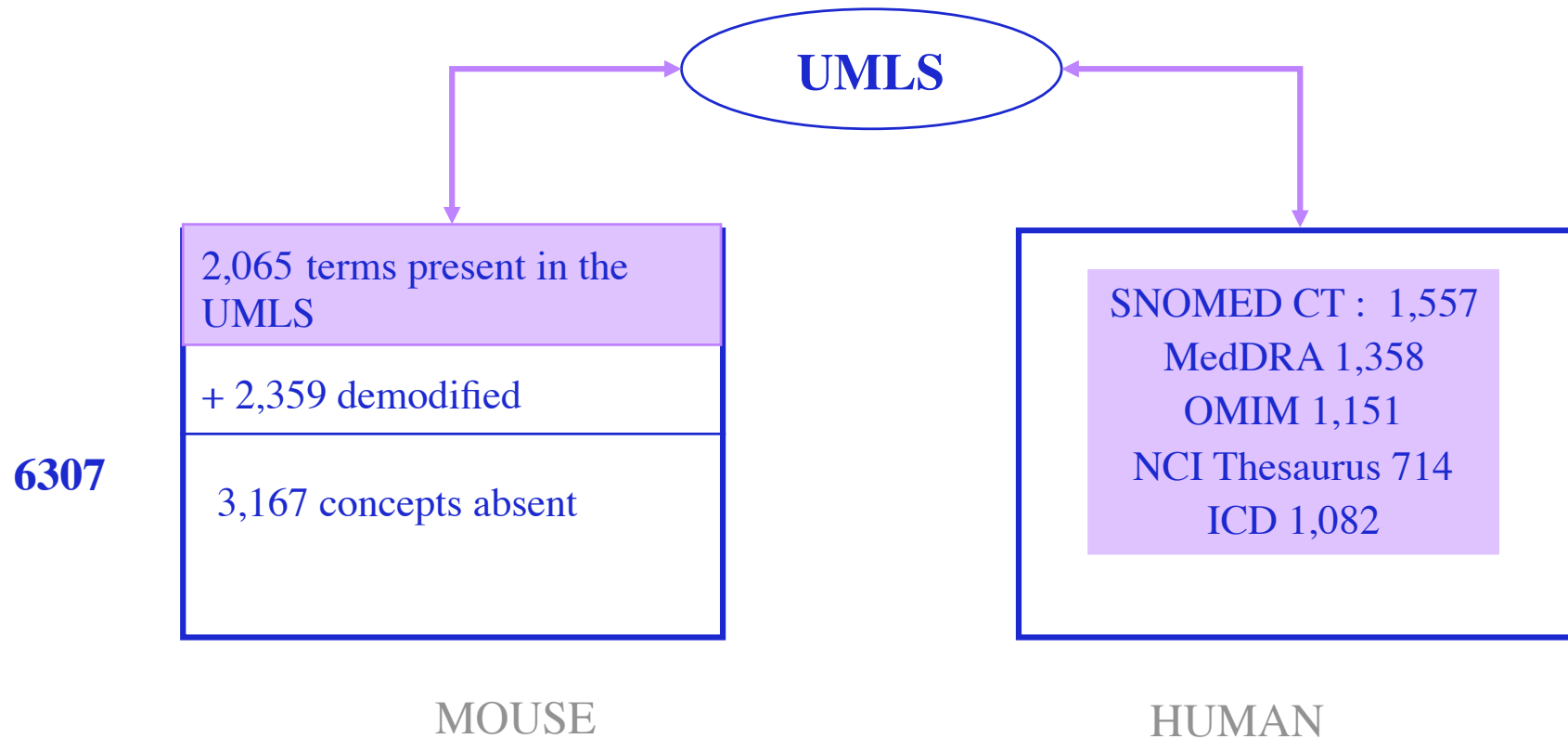
Discussion

- Role of the UMLS in integrating phenotype terminologies



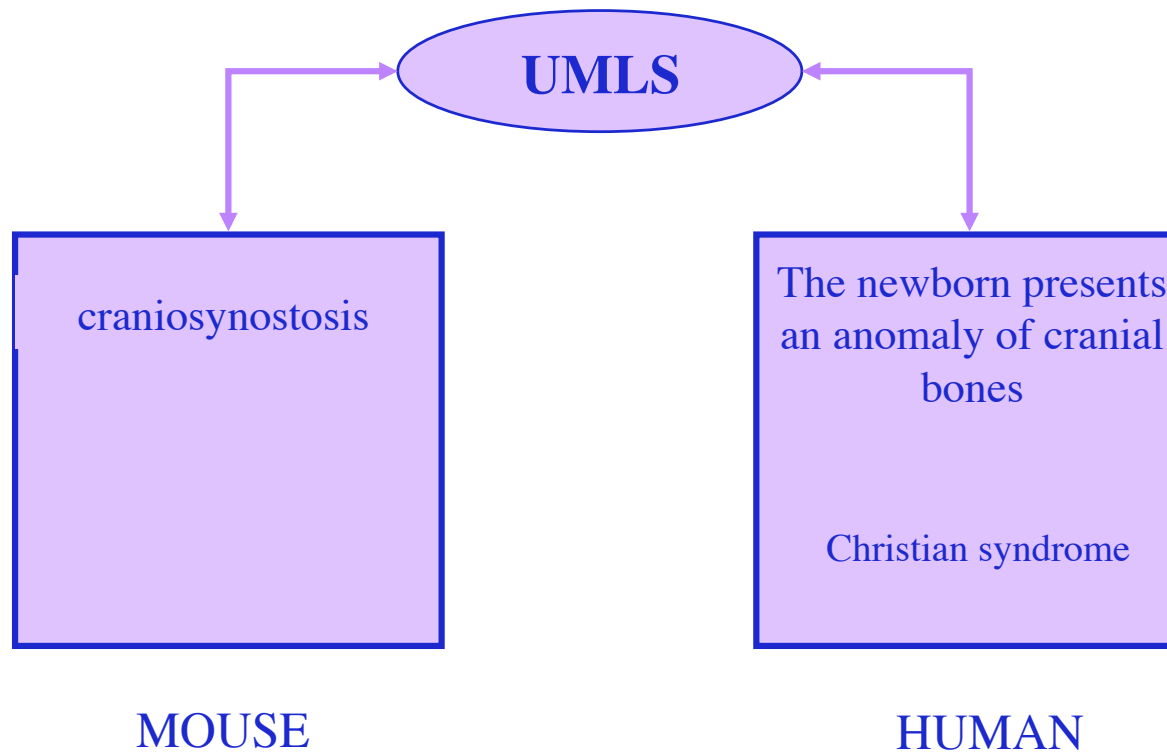
Discussion

- Role of the UMLS in integrating phenotype terminologies



Discussion

- Role of the UMLS in integrating phenotype data
 - OMIM, Clinical DBs, Mouse DBs



Acknowledgements

- Olivier Bodenreider, NLM
- Fleur Mougín, ISPED

- Download/ customize/
browse the UMLS
- Knowledge Source Server

- umlsks.nlm.nih.gov/

