



International Conference on Biomedical Ontology 2015

Lisbon, 27-30 July

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ICBO 2015

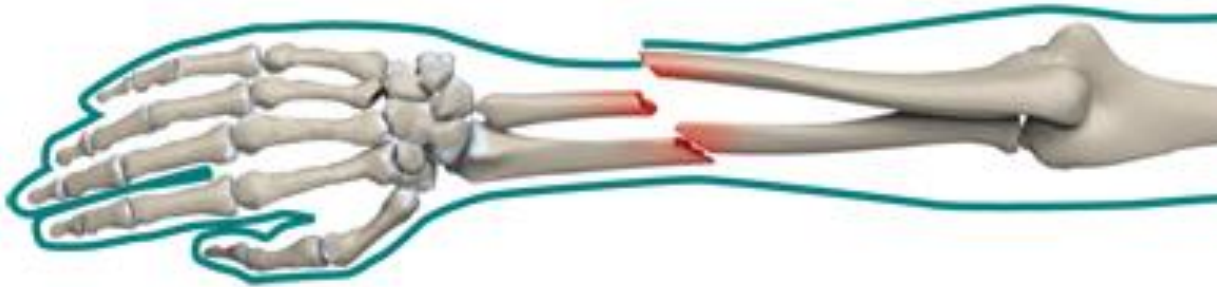
Lisbon, Portugal, July 27-30, 2015

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IHTSDO group „Event, Condition, Episode“

**Formal
representation of
disorder
associations in
SNOMED CT**

Disorder association example



SCTID: 75857000

Fracture of radius **AND** ulna (disorder)

	'after'	'and'	'caused by'	'due to'	'with'	'without'	Σ	Σ_{abs}
<i>Body Struct.</i>	.0	2.5	.0	.0	.6	.0	3.1	951
<i>Clin. Finding</i>	.1	3.0	.1	2.7	4.7	1.3	11.9	11,974
<i>Event</i>	.6	8.1	12.0	1.9	9.6	3.1	44.3	1,627
<i>Obs. Entity</i>	.3	2.3	.0	.0	.5	.0	3.1	257
<i>Product</i>	.0	1.5	.0	.0	.0	.0	1.5	259
<i>Phys. Object</i>	.0	2.3	.0	.0	5.4	1.2	9.0	408
<i>Procedure</i>	.1	5.8	.0	.0	5.8	.3	12.1	6,497
<i>Qual. Value</i>	.2	1.1	.0	.0	.4	.0	1.8	162
<i>Situation</i>	.3	1.8	.0	.2	3.9	.4	6.6	243
<i>Substance</i>	.0	1.1	.0	.0	.4	.0	1.5	353
<i>Others</i>	.0	1.2	.0	.0	.1	.0	1.3	584
ALL	.0	2.9	.2	1.0	3.0	.5	7.7	23,039

Co-ordinating expressions: no clear patterns

Diabetic retinopathy subclassOf
'**associated with**' some *Diabetes mellitus*

Paraneoplastic neuropathy subclassOf
'**due to**' some *Neoplastic disease* using

Dermatomycosis associated with AIDS subClassOf
'**associated with**' some AIDS

AIDS with dermatomycosis (disorder) **subclassOf** AIDS



C#1: Chips and Fish

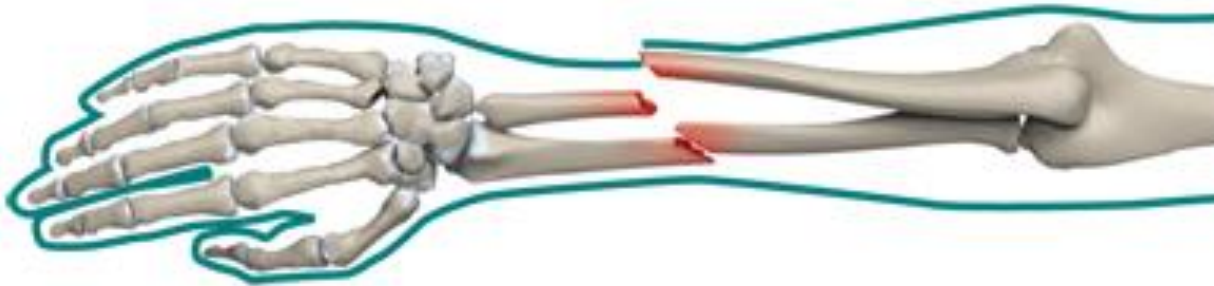


C#2: Fish and Chips

Methods

- Random selection of SNOMED CT concepts labelled by co-ordinated expressions
- Collaborative identification of recurring patterns done by the IHTSDO group Event, Condition, Episode
- Using BioTopLite2 as domain upper ontology
- Scope limited to of the investigation to the SNOMED CT hierarchy *Clinical Finding / Disorder*
- Interpreting SNOMED CT disorders and findings as Clinical Life Phases (aka Clinical Situations)

“A and B” : Current SNOMED pattern



Parents

- ☰ Fracture of radius (disorder)
- ☰ Fracture of ulna (disorder)

☰ Fracture of radius AND ulna (disorder)



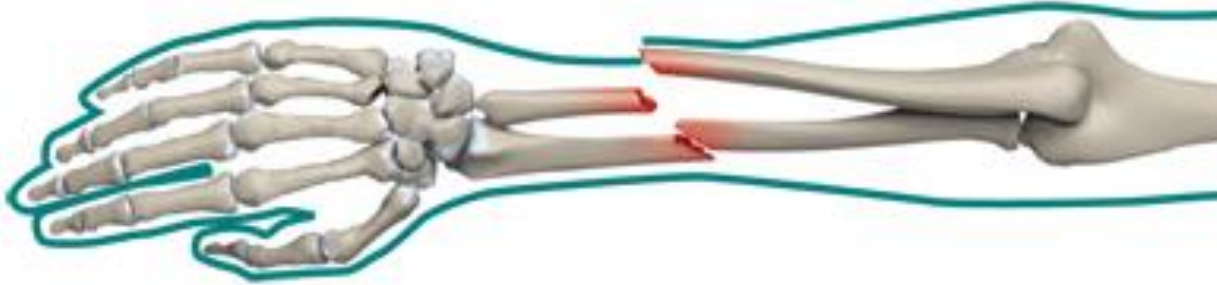
SCTID: 75857000

Fracture of radius AND ulna (disorder)
Fracture of radius AND ulna

Finding site → Bone structure of radius
Associated morphology → Fracture

Finding site → Bone structure of ulna
Associated morphology → Fracture

“A and B” : Current SNOMED pattern



'Fracture of Radius and Ulna' equivalentTo

RoleGroup some

('Finding site' some '*Bone structure of ulna*' and

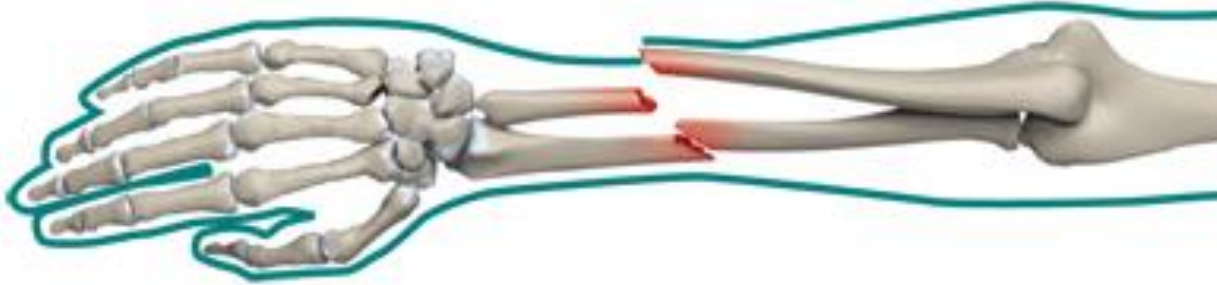
'Associated morphology' some *Fracture*) and

RoleGroup some

('Finding site' some '*Bone structure of radius*' and

'Associated morphology' some *Fracture*)

Re-interpretation as “Clinical Life Phase”



'Fracture of Radius and Ulna' equivalentTo

'**has Condition**' some

('Finding site' some '*Bone structure of ulna*' and

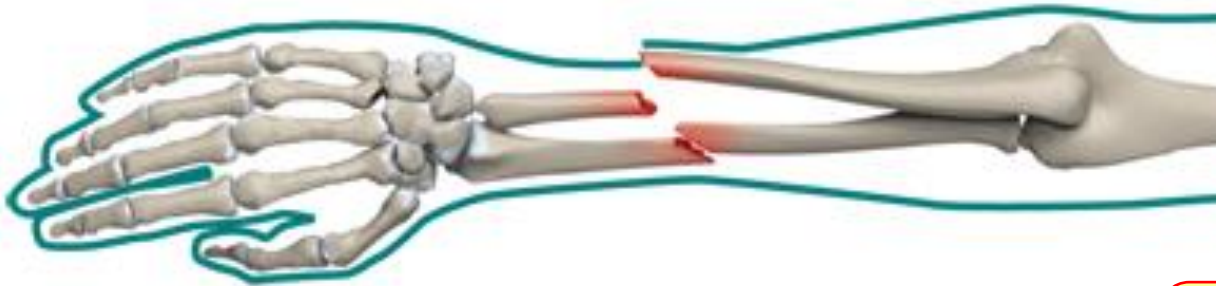
'Associated morphology' some *Fracture*) and

'**has Condition**' some

('Finding site' some '*Bone structure of radius*' and

'Associated morphology' some *Fracture*)

Re-interpretation as “Clinical Life Phase”



Clinical
Condition

'Fracture of Radius and Ulna' equivalentTo

'**has Condition**' some

('Finding site' some '*Bone structure of ulna*' and

'Associated morphology' some *Fracture*)

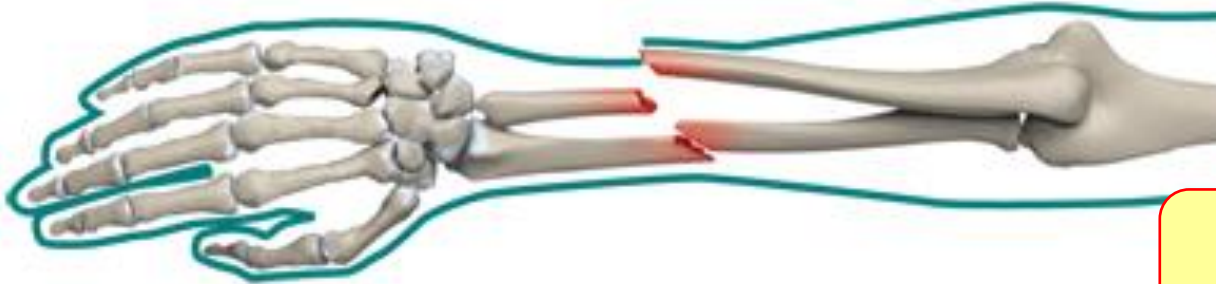
and

'**has Condition**' some

('Finding site' some '*Bone structure of radius*' and

'Associated morphology' some *Fracture*)

Re-interpretation as “Clinical Life Phase”



'Fracture of Radius and Ulna' equivalentTo

'**has Condition**' some

('Finding site' some '*Bone structure of ulna*' and

'Associated morphology' some *Fracture*)

and

'**has Condition**' some

('Finding site' some '*Bone structure of radius*' and

'Associated morphology' some *Fracture*)

Results: Four Patterns identified

	Definition	Example
#1	Both <i>X</i> and <i>Y</i> are co-occurrent, but with no causality or manifestational relationship between <i>X</i> and <i>Y</i>	<i>Hay fever with asthma</i>
#2	<i>X</i> is due to <i>Y</i> , but <i>X</i> and <i>Y</i> are not necessarily co-occurrent	<i>Disorder of optic chiasm due to non-pituitary neoplasm</i>
#3	<i>X</i> temporally follows <i>Y</i> . This does not specify that <i>X</i> is due to <i>Y</i> , although causality is frequently implied	<i>Postvaricella encephalitis</i>
#4	<i>X</i> is due to <i>Y</i> , and both <i>X</i> and <i>Y</i> are co-occurrent	<i>Hernia, with intestinal obstruction</i>

Pattern #1 – "co-occurrent with"

Both X and Y are co-occurrent, but with no causality between X and Y .

X with Y_{CLP} equivalentTo X_{CLP} and Y_{CLP}

X_{CLP} equivalentTo '**has condition**' some X

Y_{CLP} equivalentTo '**has condition**' some Y

X with Y_{CLP} equivalentTo '*Clinical life phase*' and

'**has condition**' some X and

'**has condition**' some Y



Corresponds to current Radius / Ulna pattern

Pattern #2 – "due to"

X is due to Y but X and Y are not necessarily co-occurrent

$X_{\text{causedBy}}Y_{\text{CLP}}$ equivalentTo X_{CLP} and
'**due to**' some Y_{CLP}

Corresponds to current SNOMED CT representations

* $X_{\text{causedBy}}Y_{\text{CLP}}$ equivalentTo '*Clinical life phase*'
and '**has condition**' some
(X and '**is caused by**' some Y)

Preferrable, currently infeasible

Pattern #3 – "follows"

X temporally follows Y. This does not specify that X is due to Y, although causality is frequently implied

$X \text{ follows } Y_{\text{CLP}}$ equivalent to X_{CLP} and
follows some Y_{CLP}

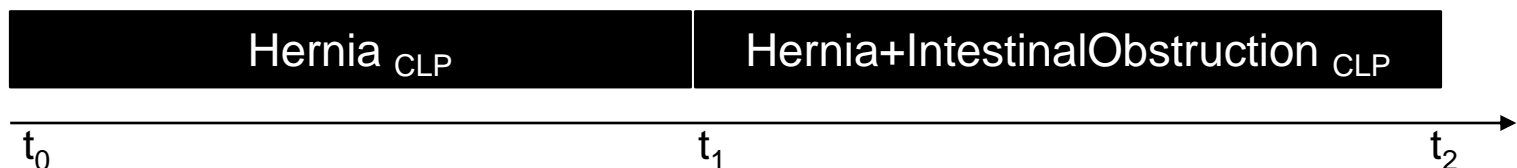
Corresponds to current SNOMED CT representations

Pattern #4 – "co-occurrent and due to"

X temporally follows Y. This does not specify that X is due to Y, although causality is frequently implied

X due To Cooccurring Y_{CLP} equivalent To
 X_{CLP} and Y_{CLP} and 'due to' some Y_{CLP}

Y_{CLP} referenced twice – two different instances



Conclusion / Outlook

- SNOMED CT: Terminology in use, still having significant quality issues due to its legacy
- Therefore, graceful evolution instead of radical redesign:
 - Analysing the implicit meaning of content, e.g. disorders / findings as clinical life phases
 - Formulating design patterns based on recurring phenomena
 - Identifying SNOMED CT content matching these patterns
 - Fitting heterogeneously modelled content to patterns
 - Continuous Validation by modellers and users
- Goal: SNOMED CT as an ontology-based terminology, increasingly incorporating Applied Ontology good practices, together with consistent guidelines for content development



Thank You

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