SNOMED CT and IHTSDO

Large scale ontology development and maintenance

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- IHTSDO and SNOMED CT
- Uses of SNOMED CT in health care
- SNOMED CT as an ontology
- IHTSDO tools
- Maintenance and quality assurance
- Development projects

IHTSDO and SNOMED CT

- International Health Terminology Standards Development Organisation
 - www.ihtsdo.org
 - 2007-
 - 15 countries (SE, DK, US, UK, AU, NZ, NL, ES, ...)
- Systematized Nomenclature of Medicine Clinical Terms
 - Roots in CAP work since 1960ies + NHS work since 1980ies
 - Originally manually maintained

Uses of SNOMED CT

- Clinical coding
 - Clinical findings symptoms, signs, disorders, ...
 - Activities treatments, diagnostics, ...
 - Anatomical structures, Organisms, Substances, Pharmaceutical products, Devices, ...
- Aggregation by subsumption hierarchies

SNOMED CT as an ontology

- EL+ ~ OWL2 EL
- 293 000 classes
- Defined w 830 000
 role instances
- 765 000 English names

Name	Syntax	Semantics
top	Т	$\Delta^{\mathcal{I}}$
bottom	\perp	Ø
nominal	$\{a\}$	$\{a^{\mathcal{I}}\}$
conjunction	$C \sqcap D$	$C^{\mathcal{I}} \cap D^{\mathcal{I}}$
existential restriction	$\exists r.C$	$ \{ x \in \Delta^{\mathcal{I}}_{\mathbb{N}} \mid \exists y \in \Delta^{\mathcal{I}} : \\ (x, y) \in r^{\mathcal{I}} \land y \in C^{\mathcal{I}} \} $
concrete domain	$p(f_1, \dots, f_k)$ for $p \in \mathcal{P}^{\mathcal{D}_j}$	$ \{x \in \Delta^{\mathcal{I}} \mid \exists y_1, \dots, y_k \in \Delta^{\mathcal{D}_j} : \\ f_i^{\mathcal{I}}(x) = y_i \text{ for } 1 \leq i \leq k \land \\ (y_1, \dots, y_k) \in p^{\mathcal{D}_j} \} $
GCI	$C \sqsubseteq D$	$C^{\mathcal{I}} \subseteq D^{\mathcal{I}}$
RI	$r_1 \circ \cdots \circ r_k \sqsubseteq r$	$r_1^{\mathcal{I}} \circ \dots \circ r_k^{\mathcal{I}} \subseteq r^{\mathcal{I}}$

• 279 000 Swedish names

SNOMED CT as an *ontology*

- Ontology-based error detection in SNOMED-CT. W Ceusters, B Smith, A Kumar... Medinfo, 2004
- The Semantics of Procedures and Diseases in SNOMED® CT. S Schulz, S Hanser, U Hahn... -Methods of information in ..., 2006
- Debugging SNOMED CT using axiom pinpointing in the description logic EL+. F Baader... -Proc. of KR-MED, 2008
- Investigating subsumption in SNOMED CT: An exploration into large description logic-based biomedical terminologies. O Bodenreider, B Smith, A Kumar... Artificial intelligence in ..., 2007
- Would SNOMED CT benefit from Realism-Based Ontology Evolution? WM Ceusters, KA Spackman... AMIA Annual Symposium ..., 2007
- SNOMED reaching its adolescence: Ontologists' and logicians' health check. S Schulz, B Suntisrivaraporn, F Baader... International Journal of ..., 2009
- Why do it the hard way? The case for an expressive description logic for SNOMED. AL Rector... - Journal of the American Medical ..., 2008
- Ontological analysis of SNOMED CT. G Héja, G Surján... BMC Medical Informatics and ..., 2008

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IHTSDO tools

- IHTSDO workbench
 - Eclipse- and CollabNet-based infrastructure for ontology and terminology development
 - Open source
- IHTSDO classifier
 - EL+ classifier, based on CEL

Maintenance and Quality assurance

- Organizational structure
 - Four standing committees
- Staff terminologists/ontologists
- Project groups and SIGs
- Internal standards process

Quality assurance

- QA Framework is being implemented
- Content is being check against a set of production rules
- External audits

Development projects

- Current ongoing "ontologization" of SNOMED CT
 - Alignment to e.g. BioTop, BFO, FMA
 - Scalability and Reproducibility!
- Expressivity tests
 - Negation tests, Value restriction
 - Role inclusions, Concrete domains