

WP6: Ontologies and Terminologies *Background, Findings & Recommendations*

Alan Rector, U Manchester

with

Jean Marie Rodrigues, U Jean Monnet, Saint-Etienne

Anand Kumar, U Jean Monnet, Saint-Etienne

Dipak Kalra, UCL

Bedirhan Ustun, WHO

Pieter Zanstra

Sounds Easy but has proved Hard

- 150 years of effort has not produced a solution
 - 20 years of intensive work in IT has not provided a solution
 - 10 years work on SNOMED provides at most a start on a solution
 - May even have be an “anti-solution” to parts of the problem
 - » Certainly resulting in building serious “pregacy” - pre-built legacy ... effort & expertise in circumventing flaws and problems of its own creation
- Too many requirements ... so priorities unclear
 - Intimately intertwined with
 - EHRs, Public health, Decision support, Clinical care...
 - “What’s it for?”
- Temptation to do more than is possible
 - The best is the enemy of the good
- Those who must pay do not benefit;
those who benefit will not pay
 - The benefit is to the common; the cost is to the private

Basic constraints & Limitations on the Possible

- **Scaling and the combinatorial explosion**
 - You can't provide a phrase book big enough to write a novel
 - Or an EHR
- **Real world variability**
 - Some things are just different
 - Standards will just mislead
- **Human variability**
 - People can't always interoperate
 - Machines will never interoperate better than the people that use them
- **Poor match of problem space & solution space**
 - Poor definition of purpose
 - "What's it for?"
- **Lack of Quality assurance**

Vocabulary - Things sometimes called “Terminologies”

- Controlled vocabulary with identifiers (codes)
 - List of terms for some entities plus “codes” to identify them
 - Code - meaningless identifier independent of language
- Lexicon
 - The collection of linguistic entities - attached to a given controlled vocabulary, codes, or ontology
 - May include grammatical & other information; may be multilingual
- Classification
 - An organisation of entities into classes *for a specific purpose*, e.g. ICD and DRGs
- Thesaurus
 - A collection of entities/terms arranged for *human* navigation via broader-than/narrower-than and associative relations
- Ontology (sensu informatics)
 - A *logical* model of the *meanings of the* entities about which information is to be expressed for use in computers
- Knowledge Representation System
 - A *model* of the background knowledge assumed, expressed so as to be used in computer systems - including but not limited to the ontology

Findings



State of Play - Facts on the Ground

- **Most data will be collected using local codes**
 - SNOMED unlikely to supplant local codes in EU, US or elsewhere except
 - UK & ?Australia & ?Canada Infoway
 - Maps to SNOMED may be important
- **HL7 v2 + LOINC will dominate messaging standards for labs**
 - except in UK which will use V3 + SNOMED + local schemes
 - except in primary care which will use Clinical Terms (Read) & Nordic countries
- **ICD will continue to be the main form of international recording**
 - ICD 11 might convergence with SNOMED
- **Subsets of SNOMED will be used in many areas as a controlled vocabulary**
 - Without major investments other aspects will remain valueless or worse
 - And developing subsets is proving costly and rarely re-usable
 - But an alternative international effort is unlikely
- **Lack of tools and people will be major constraints**
- **Bio and Translational Medicine Terminologies will increase**
 - e.g. Gene Ontology, NCI Thesaurus
- **Web based initiatives will happen**
 - Web 2.0 & Google-type approaches will be increasingly important

SNOMED CT: Current Assessment

- **Purpose**
 - *remains ill defined ... but being used in UK and elsewhere for controlled vocabulary*
- **Controlled vocabulary and identifiers**
 - *Well managed but very slow response (months .. years)*
- **Scale**
 - Overgrown - victim of combinatorial explosion and Zipf's law
 - Too big to QA, manage and fix, find terms, use reproducibly
 - But still often $\leq 25\%$ coverage for specific applications
- **Reproducibility**
 - *Poorly studied... often poor*
- **Hierarchies and relations**
 - *Unusable*
 - Too unreliable to depend on to behave as documented. Not QAed.
 - Systematically flawed in principle; Limited by State of Art circa 1990
- **Multilingual / cross-cultural support**
 - *Minimal* - fundamentally an anglophone organisation
 - Neither understood nor a priority of the IHTSDO
 - Spanish and Canadian French versions might appear
 - Separation of language and concepts still problematic; tools absent
- **Openness and accessibility for QA, contribution, & social computing**
 - *Unusable* - remains effectively closed
 - Not generally available on the Web
 - Opportunity cost of participation prohibitive; Influencing policy difficult
 - Remains the province of a small self-reinforcing clique
- **Could be fixed at modest cost relative to total cost of health it interoperability**
 - Priority is a feasibility study on ~20K concepts

Interaction of Terminology, EHRs & DSSs

- **HL7-Terminfo provides a base**
 - But no tools (yet) & proving difficult to implement consistently
- **Archetype experiments provide a start**
 - Terminology Query Languages promising
 - Common formalism a major challenge
- **Better technologies exist and have been demonstrated experimentally**
 - Using logical tools, OWL, UML2, Model Driven Architectures...
 - But so far under-developed
 - Efforts at tools are under way in the UK
 - » Outcome remains uncertain
 - *More development urgently needed*

Recommended Principles: Technical

- **Separate Language and Concepts**
 - Lexicon and Coding System/Ontology - more radically than in SNOMED today
 - Otherwise endless confusion
 - Develop multilingual cross-cultural systems
- **Make it easy to participate -**
 - Hide complexity if not needed - separate levels
 - End users /Author-experts & configuration staff / Terminology experts
- **Develop binding to EHR and Decision support**
 - Requires new techniques and tools
- **Leverage modern tools for development, QA & deployment, especially from the Web community**
 - Web 2.0
 - Web Ontology Language OWL and modern logic formalisms
 - ... but also what the EC has already paid for
 - See SNOB browser for SNOMED developed from GALEN tools
- **Build small core ontologies**
 - Use logic for post-coordination or provide just-in-time services for what people need as determined empirically
- **Develop QA methodologies**
 - and use them
- **Human factors and reproducibility matter as much as technical structure**
 - Success is seeing it get simpler - GALEN reduced training time from 3 mo to 3 days

Recommended Principles: Organisational

- Process as well as product
 - All actions must aim at long term institutions that can be sustained
- Involve healthcare providers and systems vendors
 - They are who must interoperate
 - Provide incentives; mitigate costs
 - Interoperability may be a *disbenefit* to them otherwise
- The terminologies must be owned by their key end users
 - Be responsive, cooperative and open
 - The Web gives us the tools - use it!
IP worries are the enemy of interoperability
- Terminology development must coordinate with EHR and Decision support Development
 - All terminologies must have purposes
 - And be shown to be fit for purpose
- Think global; act local
 - Be multilingual and cross-cultural
 - Will only happen if EC intervenes

Key Recommendations for Terminologies

Short Term

- Support WHO open collaborative development of ICD-11
Try to open SNOMED to open collaborative development
 - Develop generic Web 2.0 / social computing
 - Seek mechanisms for opening the SNOMED process to social computing
- Develop open terminology tools that scale up to ICD & SNOMED
 - A European network of Terminology Servers & Web 2.0 Terminology sites
 - Tools based on SNOB, Protégé OWL, others
 - Cultivate open source communities - Empower users and specialist groups
 - Develop QA tools and techniques
- Support feasibility study of reformulation of SNOMED on a small scale
 - 25K terms max
 - Show that SNOMED need not be a tax on medicine
 - ... or accept that it will be

Key Recommendations for Terminology Research Medium & Long Term

- Develop language technologies
 - *Text generation* to present and QA
 - *Text extraction* to build and encode
- Support development of methodologies and tools for binding terminologies, EHRs and Decision support
 - Immediate investment in tools and medium term research
- Support research into methods & metrics for Quality assurance
- Support research on foundations of terminologies
 - Representation and meaning
 - Practical issues in representation, scaling, and use in applications
 - Terminologies & software engineering
- Support studies of human factors and reproducibility -
 - Demand QA
- Support training & human capacity