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Terminologies & Ontologies?

What are they for? What would it mean to QA an ontology (specifically in health care?)

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NIBHI

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Terminology and ontologies in Healthcare: What for? What is meant by Quality?

- A Talk in two parts
- Part 1
  - A review of a bit of history of clinical terminology and ontologies
  - Some fundamental problems
- Part 2
  - Focus on Quality Assurance
  - Quality for what?
  - Three dimensions of quality
- Summary

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Medical Terminology: A bit of history

- It all started with public health, vital statistics and epidemiology…

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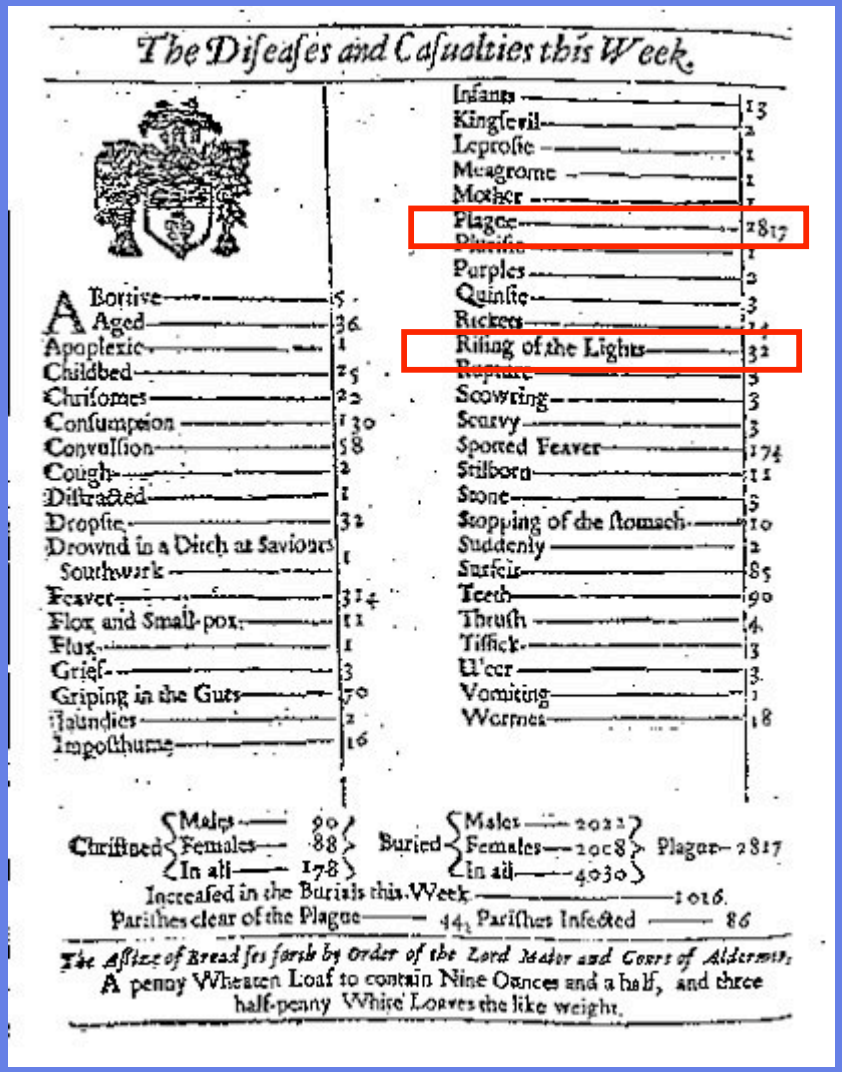
London Bills of Mortality

every Thursday from 1603 until the 1830s



The Diſeaſes and Caſualties this Week.

Abortive	5	Infants	13
Aged	36	Kingſevil	2
Apoplexie	1	Leproſie	1
Childbed	25	Meagrome	1
Chriſomes	22	Mother	1
Conſumption	130	Plague	2817
Convulſion	58	Purples	2
Cough	2	Quinſie	3
Diſtracted	1	Rickets	14
Dropſie	32	Riſing of the Lights	32
Drowned in a Ditch at Saviours Southwark	1	Scowring	3
Feaver	314	Scurvy	3
Flox and Small-pox	11	Spotted Feaver	174
Flux	1	Stillborn	11
Grief	3	Stone	5
Griping in the Guts	70	Stopping of the ſtomach	10
Jaundies	2	Suddenly	2
Impoſthume	16	Surfeit	85
		Teeth	90
		Thruſh	4
		Tiſſick	3
		Ulcer	3
		Vomiting	1
		Wormes	18

Chriſtned { Males 90, Females 88, In all 178 } Buried { Males 2021, Females 2008, In all 4030 } Plague 2817

Increaſed in the Burials this Week 1016.

Pariſhes clear of the Plague 44 Pariſhes Infected 86

The Aſſize of Bread ſet forth by Order of the Lord Maior and Court of Aldermen:
A penny Wheaten Loaf to contain Nine Ounces and a half, and three half-penny White Loaves the like weight.

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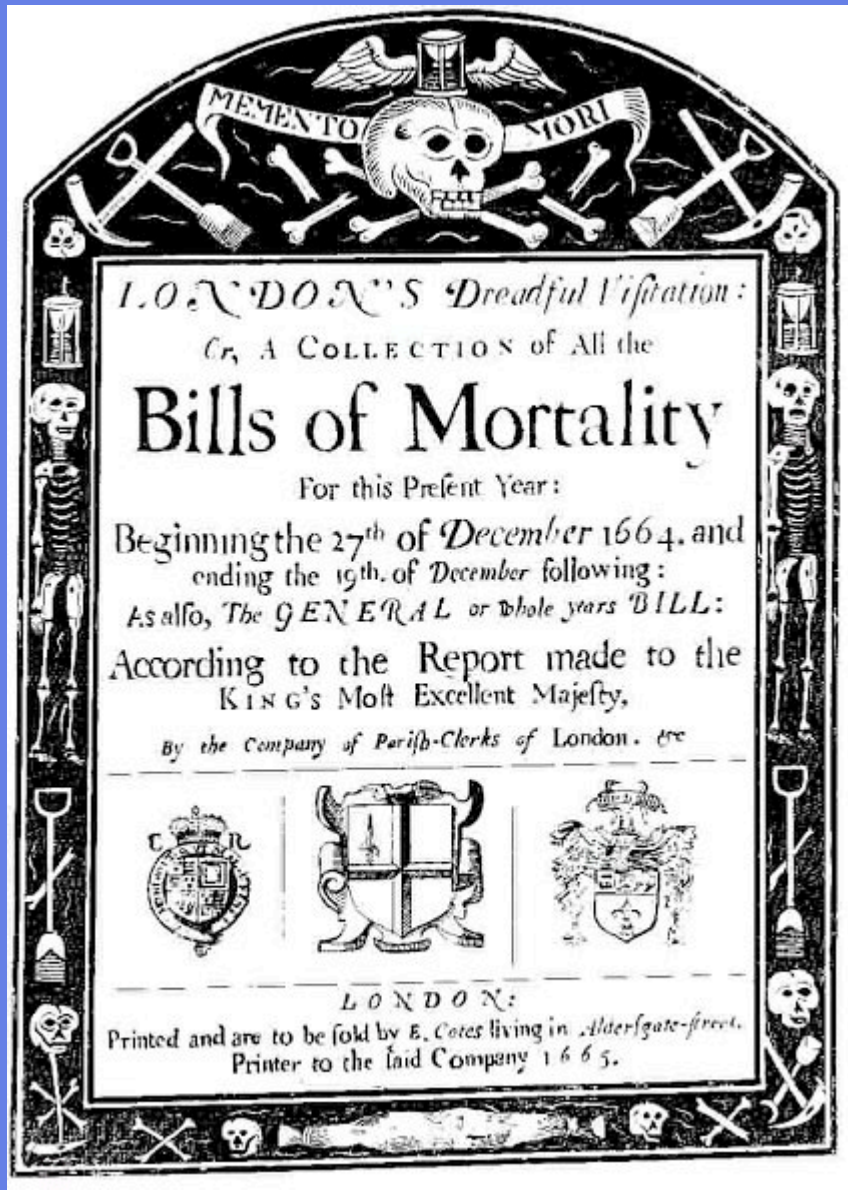
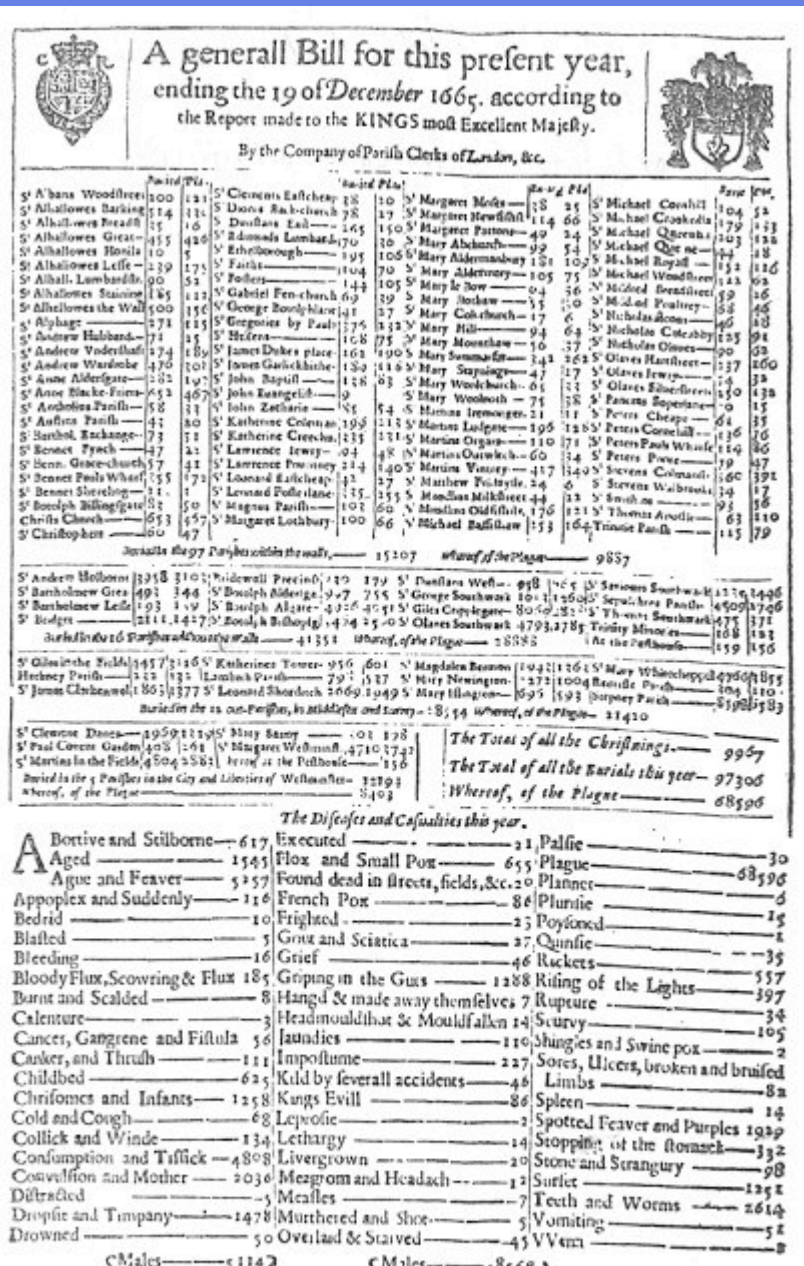
Aggregated Statistics 1665





A generall Bill for this present year, ending the 19 of December 1665. according to the Report made to the KINGS most Excellent Majesty.

By the Company of Parish Clerks of London, &c.

The Totes of all the Christnings — 9967
The Total of all the Burials this year — 97306
Whereof, of the Plague — 68596

The Diseases and Casualties this year.

Abortive and Stillborne	617	Executed	21
Aged	1545	Flox and Small Pox	655
Ague and Feaver	5257	Found dead in streets, fields, &c.	20
Appoplex and Suddenly	116	French Pox	86
Bedrid	10	Frighted	23
Blasted	5	Gout and Sciatica	27
Bleeding	16	Grief	46
Bloody Flux, Scowring & Flux	185	Griping in the Guts	1288
Burnt and Scalded	8	Hanged & made away themselves	7
Calenture	3	Headmouldshot & Mouldfalken	14
Cancer, Gangrene and Fistula	56	Jaundies	110
Canker, and Thrush	111	Imposthume	227
Childbed	625	Kild by severall accidents	46
Chrisomes and Infants	1258	Kings Evill	86
Cold and Cough	68	Leprosie	2
Collick and Winde	134	Lethargy	14
Consumption and Tissick	4808	Livergrown	20
Convulsion and Mother	2036	Meagrom and Headach	12
Distracted	5	Measles	7
Dropsie and Timpany	1478	Murthered and Shot	5
Drowned	50	Overlaid & Starved	45
		Palsie	30
		Plague	68596
		Planet	6
		Plurisie	15
		Poysoned	1
		Quinsie	35
		Rickets	557
		Riling of the Lights	397
		Rupture	34
		Scurvy	105
		Shingles and Swine pox	2
		Sores, Ulcers, broken and bruised Limbs	82
		Spleen	14
		Spotted Feaver and Purples	1929
		Stoppage in the Stomach	332
		Stone and Strangury	98
		Surfet	1251
		Teeth and Worms	2614
		Vomiting	52
		Wenn	8

Christned: Males 5114, Females 4853, In all 9967
Buried: Males 48569, Females 48737, In all 97306
Of the Plague 68596

Increased in the Burials in the 130 Parishes and at the Pest-house this year — 79003
Increased of the Plague in the 130 Parishes and at the Pest-house this year — 68550

Manchester Mercury

January 1st 1754

List of diseases & casualties this year

19276 burials

15444 christenings

Deaths by centile

Aged 1456
Consumption 3915
Convulsion 5977
Dropsy 794
Fevers 2292
Smallpox 774
Teeth 961
Bit by mad dogs 3
Broken Limbs 5
Bruised 5
Burnt 9
Drowned 86
Excessive Drinking 15

Executed 18
Found Dead 34
Frighted 2
Kill'd by falls and other accidents 55
Kill'd themselves 36
Murdered 3
Overlaid 40
Poisoned 1
Scalded 5
Smothered 1
Stabbed 1
Starved 7
Suffocated 5

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Origins of modern terminologies

100 years of epidemiology

- ICD - Farr in 1860s to ICD9 in 1979
  - International reporting of morbidity/mortality
- ICPC - 1980s
  - Clinically validated epidemiology in primary care
  - Now expanded for use in Dutch GP software

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… then took on new tasks .

Organising Care

- Librarianship
  - MeSH - NLM from around 1900 - Index Medicus & Medline
  - EMTree - from Elsevier in 1950s - EMBase
- Remuneration
  - ICD9-CM (Clinical Modification) 1980
    - 10 x larger than ICD; aimed at US insurance reimbursement
  - CPT, …
- Pathology indexing
  - SNOMED 1970s to 1990 (SNOMED International)
    - First faceted or combinatorial system
    - Topology, morphology, aetiology, function
- Specialty Systems
  - Mostly similar hierarchical systems
    - ACRNEMA/SDM - Radiology
    - NANDA, ICNP… - Nursing

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… and then with computers
Documenting/Reporting Care

- Early computer systems
  - Aimed at saving space on early computers
  - 1-5 Mbyte / 10,000 patients
- Read (1987 - 1995)
  - Hierarchical modelled on ICD9
    - Detailed signs and symptoms for primary care
    - Purchased by UK government in 1990
    - Single use
- Medical Entities Dictionary (MED)
  - Jim Cimino, Hospital support, Columbia, USA
- OXMIS
  - READ competitor
  - Flat list of codes
  - Derived from empirical data
  - Defunct circa 1999
- ICPC
  - Epidemiologically tested, Dutch
- LOINC
  - For laboratory data
- DICOM (sdm)
  - For images
- MEDDRA
  - Adverse Reactions

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Unified Medical Language System

- US National Library of Medicine
- *De facto* common registry for vocabularies
- Metathesaurus
  - 1.8 million concepts
  - categorised by semantic net types
- Semantic Net
  - 135 Types
  - 54 Links
- Specialist Lexicon

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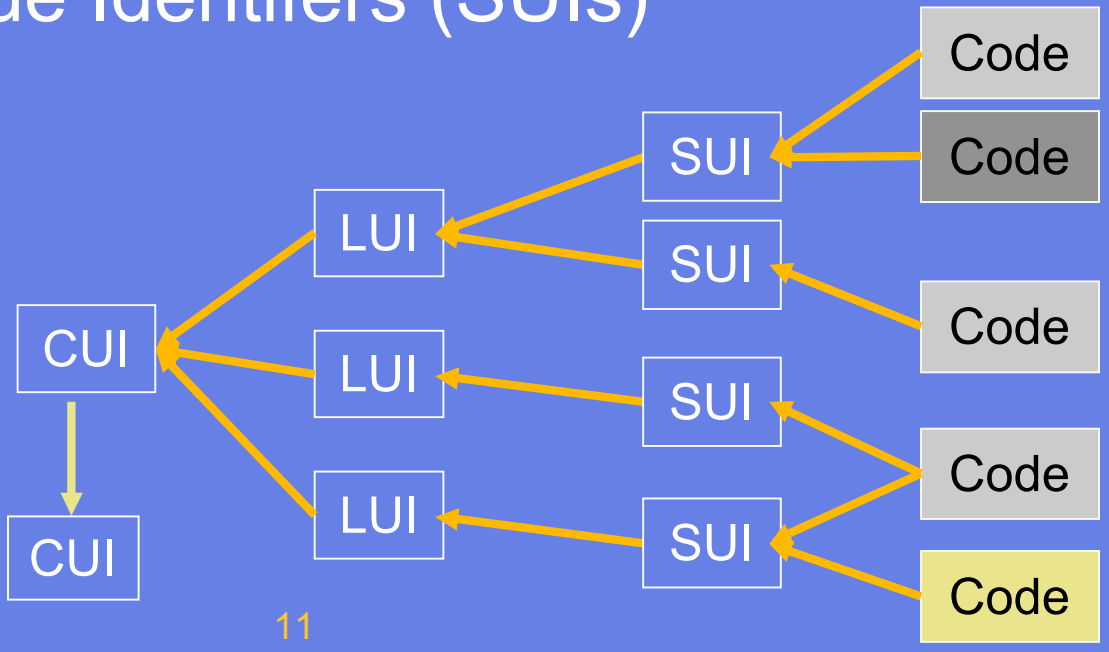
Unified Medical Language System

- Concept Unique Identifiers (CUIs)
- Lexical Unique Identifiers (LUIs)
- String Unique Identifers (SUIs)



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…but …The Coding of Chocolate

An international conversion guide

Term	CTV3	
Bounty bar	UbOVv	
Crème egg	UbOW2	
Kit Kat	UbOW3	
Mars Bar	UbOW4	
Milky Way	UbOW5	
Smarties	UbOW6	
Twix	UbOW7	
Snicker	Ub1pT	

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…but …The Coding of Chocolate

An international conversion guide

	SNOMED-CT	Term	CTV3	
 ?	C-F0811	Bounty bar	UbOVv	
	C-F0816	Crème egg	UbOW2	
	C-F0817	Kit Kat	UbOW3	
	C-F0819	Mars Bar	UbOW4	
	C-F081A	Milky Way	UbOW5	
	C-F081B	Smarties	UbOW6	
	C-F081C	Twix	UbOW7	
	C-F0058	Snicker	Ub1pT	

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…but …The Coding of Chocolate

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C-F081B	Smarties	UbOW6	
C-F081C	Twix	UbOW7	
C-F0058	Snicker	Ub1pT	

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Origins of modern terminologies

Beyond recording

- Electronic patient records (EPRs)
  - Weed's Problem Oriented Medical Record
  - Direct entry by health care professionals
- Decision support
  - Ted Shortliffe (MYCIN), Clem McDonald (Computer based reminders), Perry Miller (Critiquing), Musen (Protégé)
- Re-use
  - Patient centred information

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Origins of modern terminologies

1990s: a Paradigm Shift

- Human-Human and Human-Machine to Machine-Machine
- From *paper* to *software*
- From *single use* to multiple *re-use*
- From *coding clerks* to direct entry by *clinicians*
- From *pre-defined reporting* to *decision support*

From Books to Software

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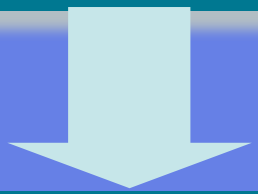
Software

Machine Processing

requires

Machine Readable Information



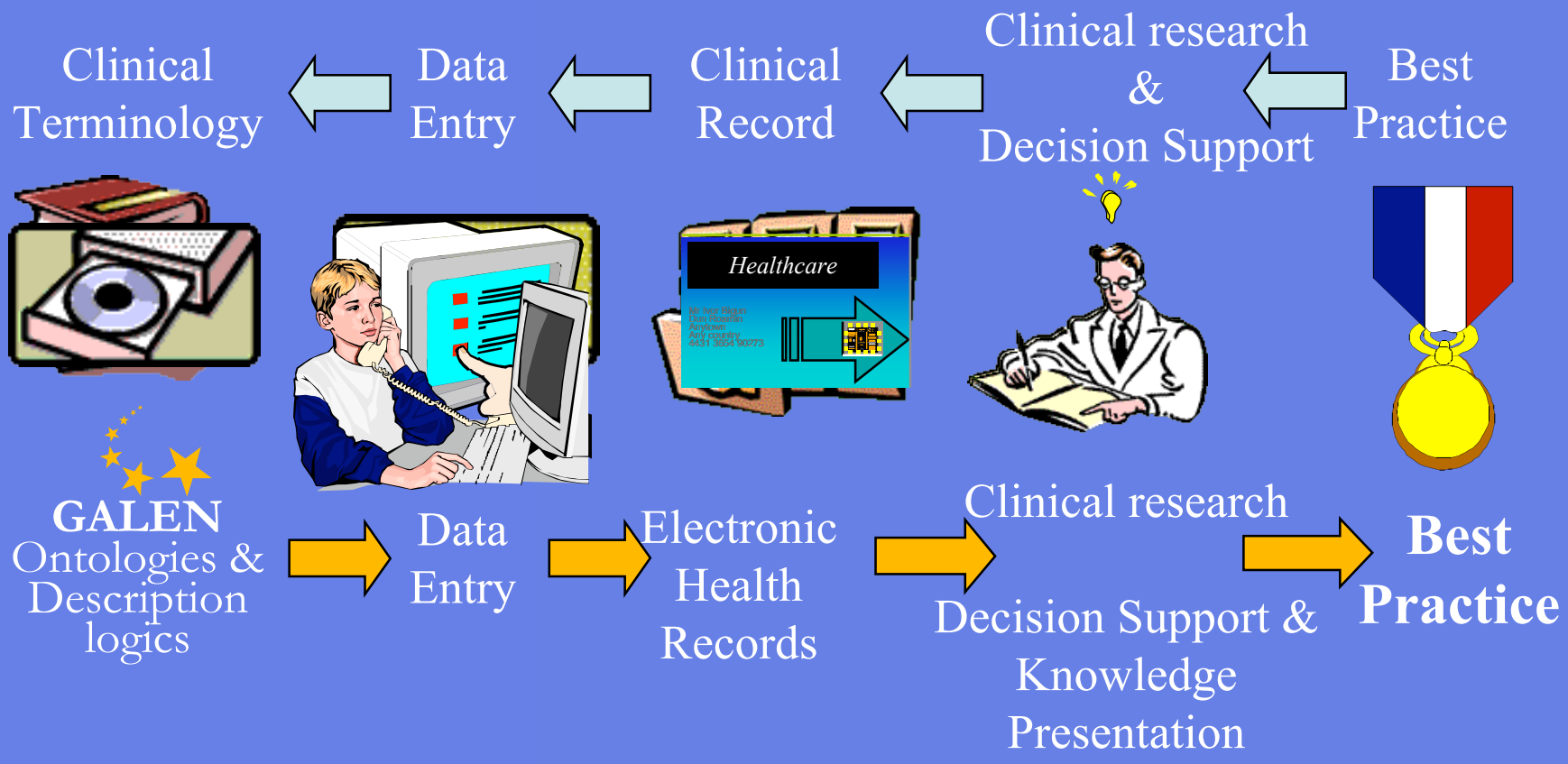
Need shared, multi-use, multi-purpose

computable

Clinical terminology

Compositional logic-based Termiologies

Where I come from



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Fundamental problems:

Enumeration doesn't scale

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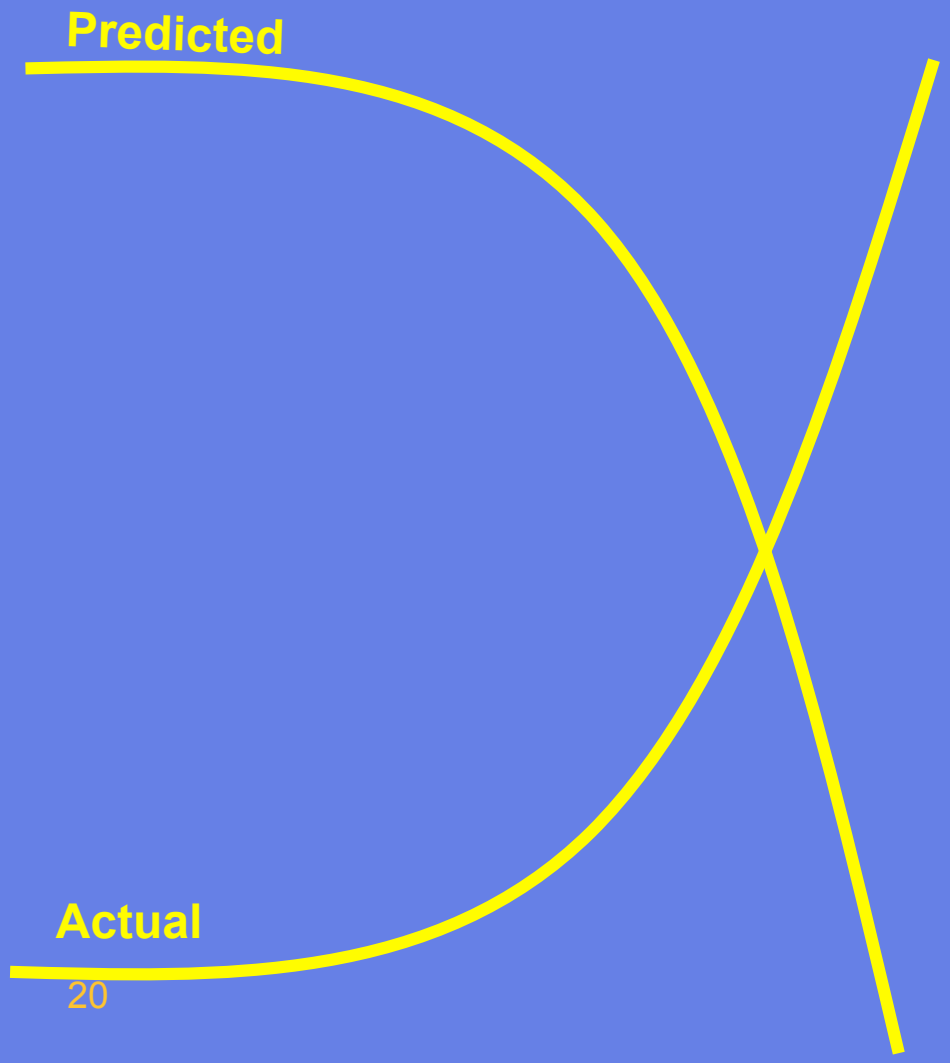
The scaling problem: The combinatorial explosion

- It keeps happening!
  - "Simple" brute force solutions do not scale up!
- Conditions x sites x modifiers x activity x context→
  - *Huge number of terms to author*
  - *Software CHAOS*



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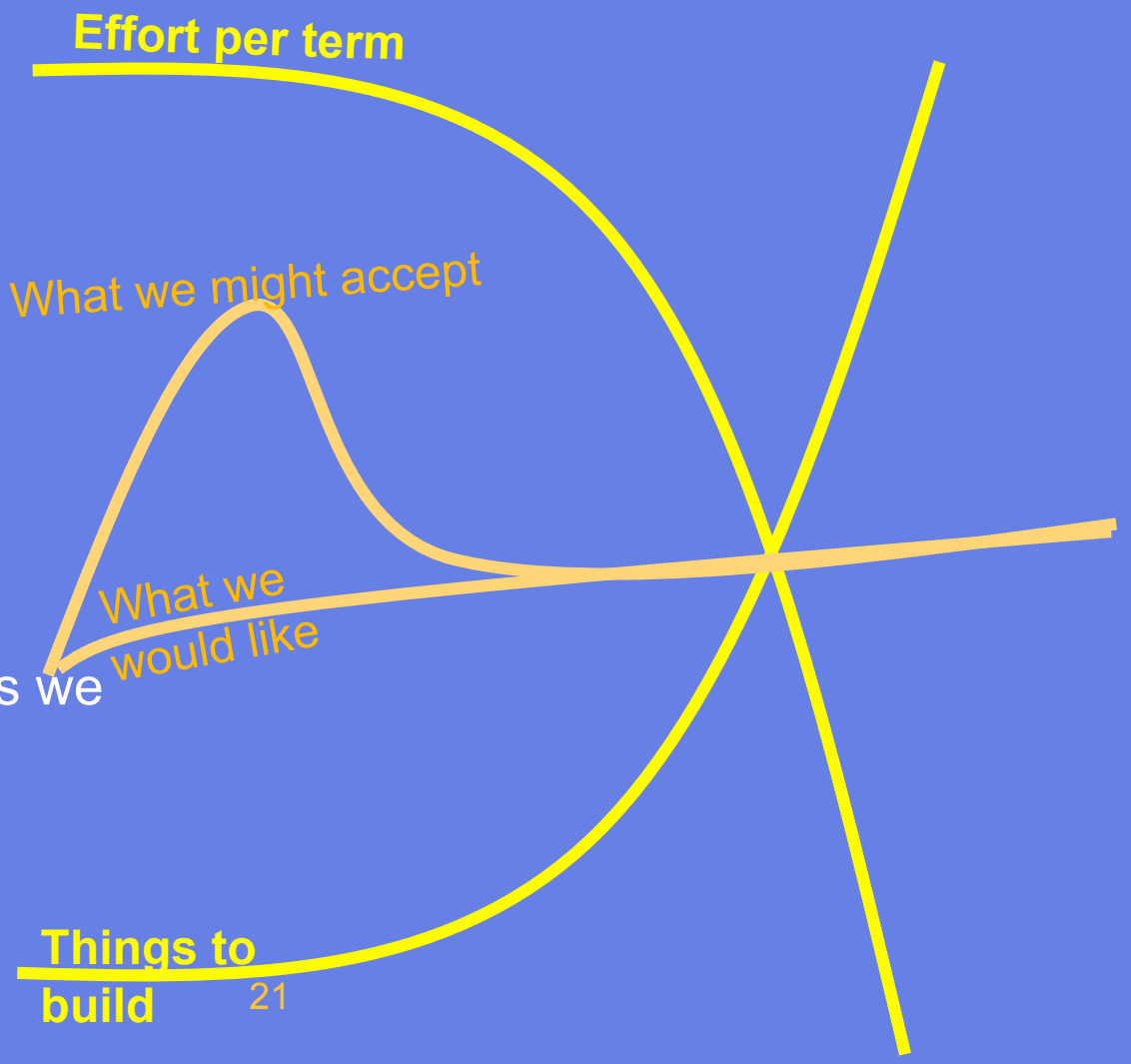
Combination of things to be done & time to do each thing

- Terms and forms needed
  - Increases exponentially
- Effort per term or form
  - Must decrease to compensate
- To give the effectiveness we want
  - Or might accept



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The exploding bicycle

- 1972 ICD-9 (E826) 8
- READ-2 (T30..) 81
- READ-3 87
- 1999 ICD-10 ……

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1999 ICD10: 587 codes

- V31.22 Occupant of three-wheeled motor vehicle injured in collision with pedal cycle, person on outside of vehicle, nontraffic accident, while working for income
- W65.40 Drowning and submersion while in bath-tub, street and highway, while engaged in sports activity
- X35.44 Victim of volcanic eruption, street and highway, while resting, sleeping, eating or engaging in other vital activities

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Defusing the exploding bicycle: 500 codes in pieces

- 10 things to hit…
  - Pedestrian / cycle / motorbike / car / HGV / train / unpowered vehicle / a tree / other
- 5 roles for the injured…
  - Driving / passenger / cyclist / getting in / other
- 5 activities when injured…
  - resting / at work / sporting / at leisure / other
- 2 contexts…
  - In traffic / not in traffic

V12.24 Pedal cyclist injured in collision with two- or three-wheeled motor vehicle, unspecified pedal cyclist, nontraffic accident, while resting, sleeping, eating or engaging in other vital activities

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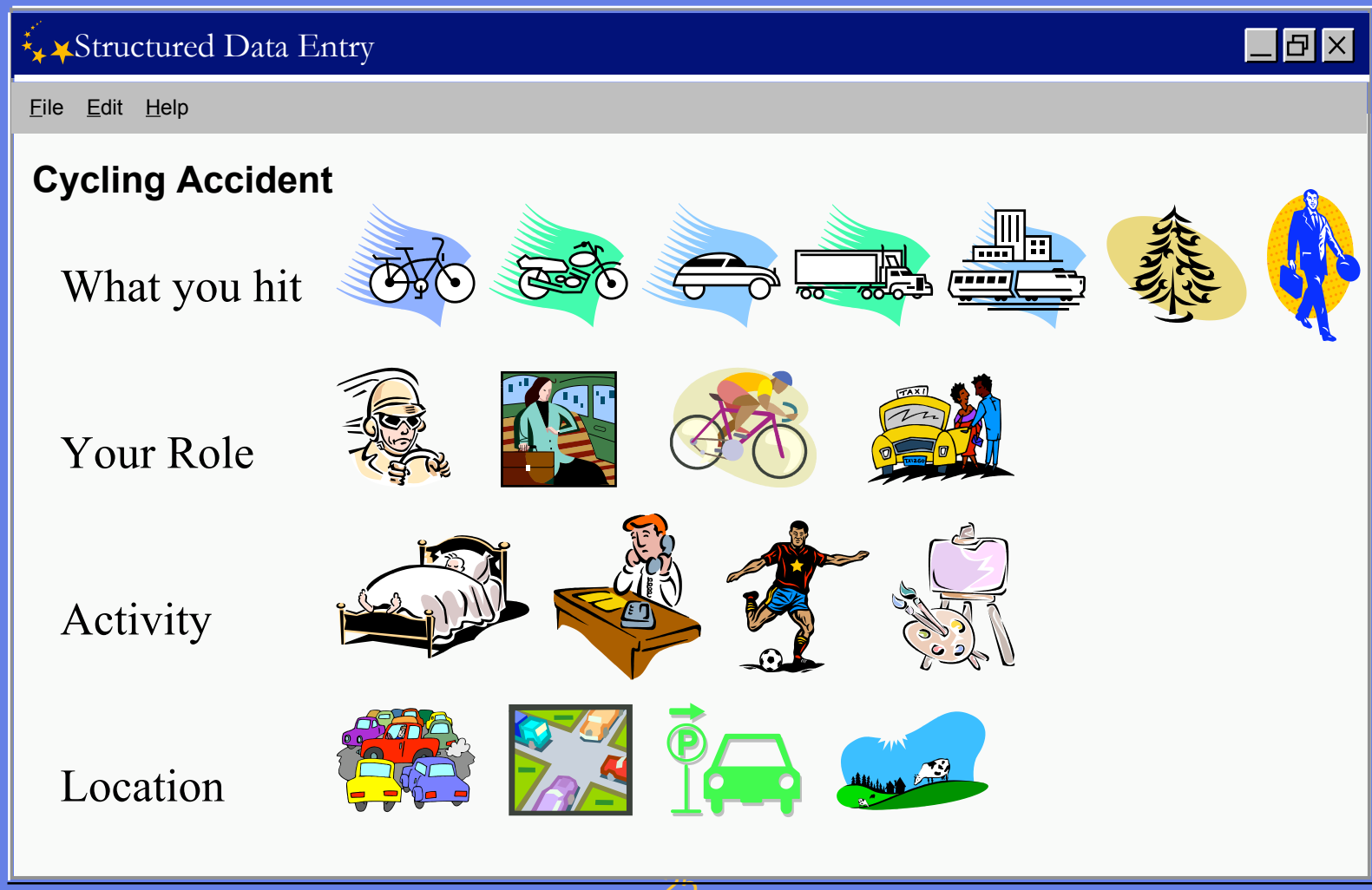
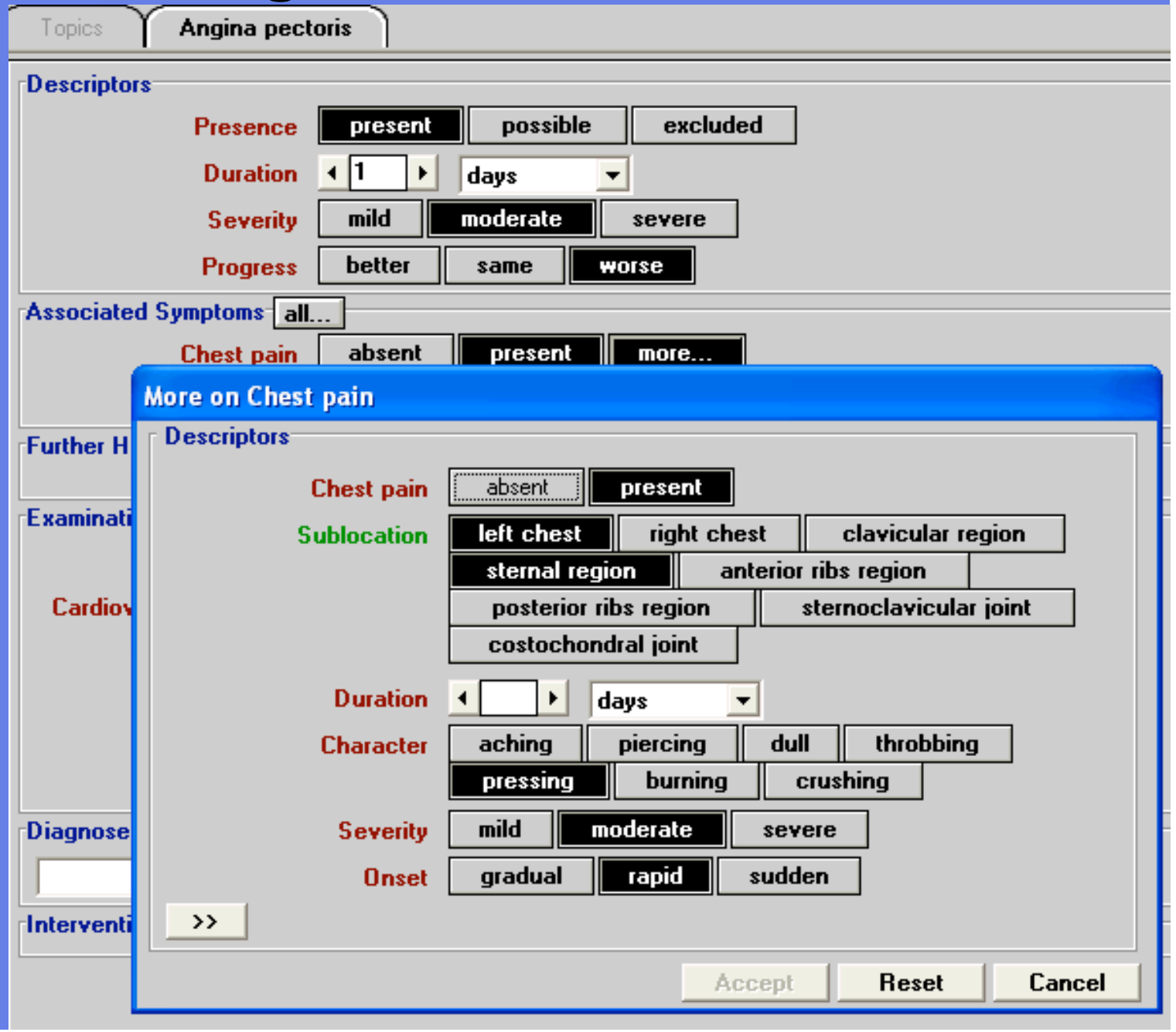
Conceptual Lego… it could be...

Goodbye to picking lists…



Intelligent Forms



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And generate it in language

Summary

Moderately severe angina pectoris for 1 day, getting worse
Rapid onset, moderately severe, pressing pain in left chest and sternal region present

On Examination
Cardiovascular system -
  Slightly raised JVP
  1st and 2nd heart sounds normal
  No added heart sounds
Pulse rate 104 per minute
Blood pressure 138/90 mm Hg

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Logic as the clips for "Conceptual Lego"

hand

extremity

body

chronic

acute

abnormal

normal

ischaemic

deletion

polymorphism

mucus

gene

protein

polysacharide

cell

expression

Lung

infection

inflammation

bacterium

virus

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Logic as the clips for "Conceptual Lego"

"***SNPolymorphism*** of ***CFTRGene*** causing ***Defect in MembraneTransport*** of ***Chloride Ion*** causing ***Increase*** in ***Viscosity*** of ***Mucus*** in ***CysticFibrosis***…"

"***Hand*** which is ***anatomically normal***"

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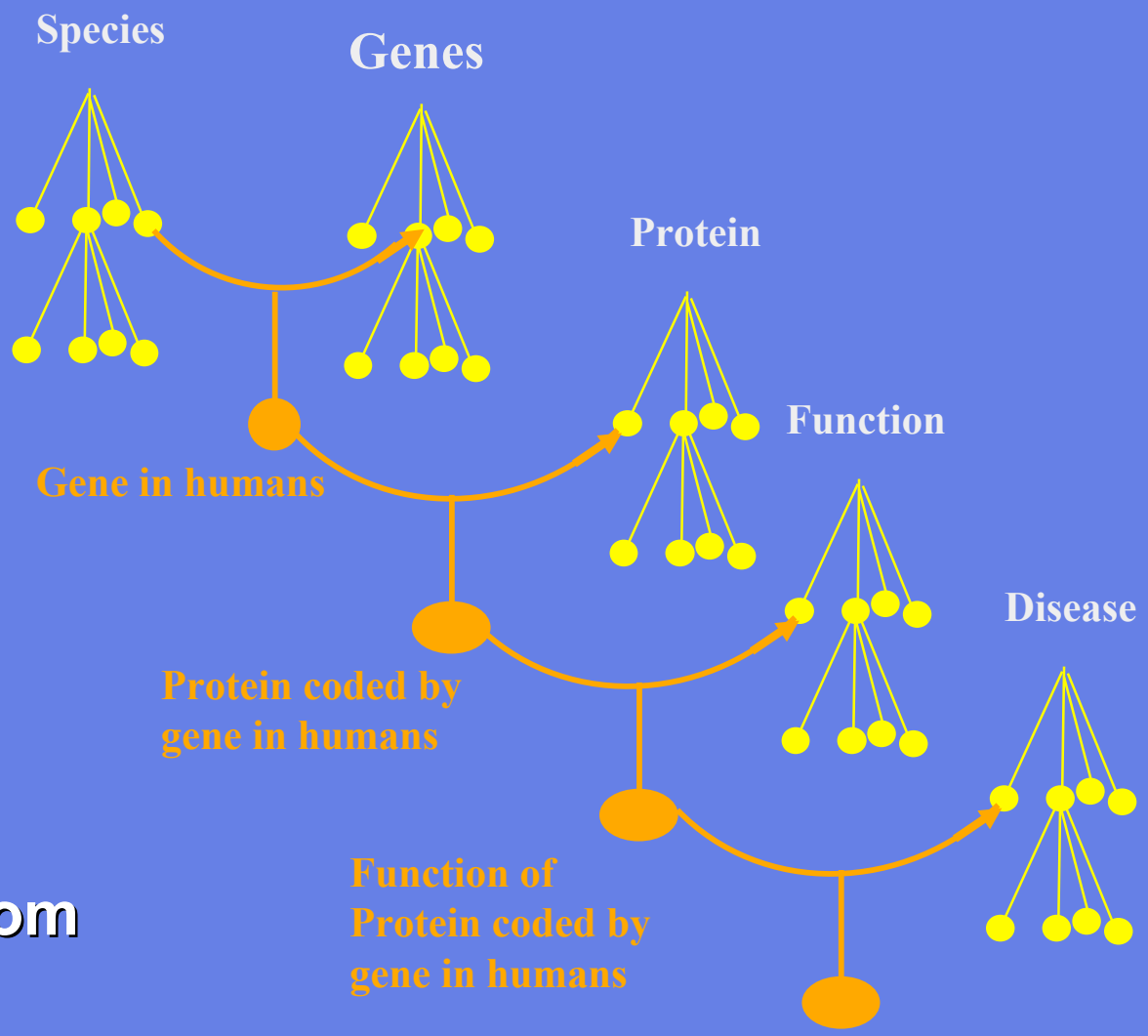
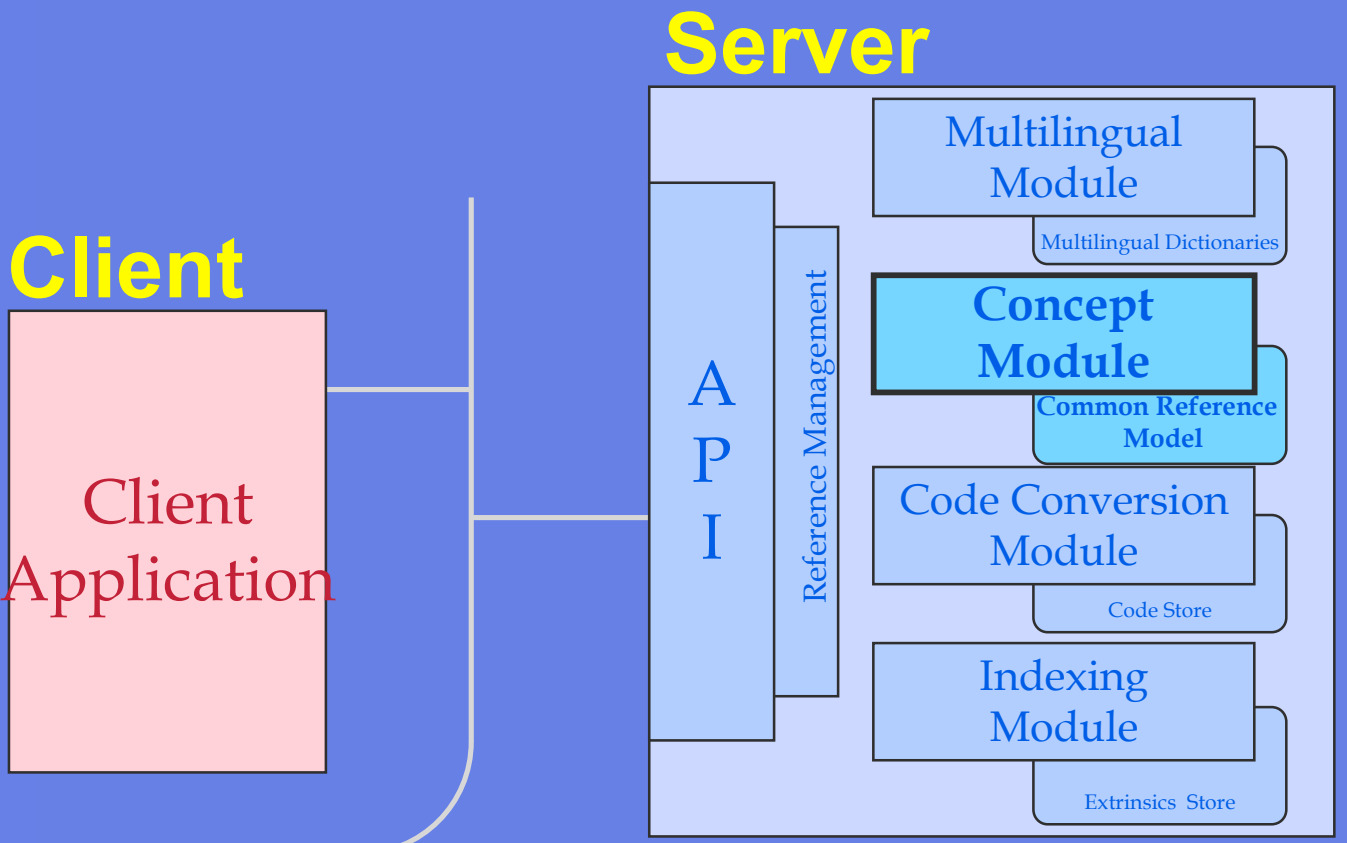


Build complex representations from modularised primitives

But of course the logic is not all you need
Modules in the GALEN Server



A single point of access for language, classification, code conversion, and indexing - well separated internally

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Problem:

System may be perfect …but Users still fallible

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User Problems

Inter-rater variability

ART & ARCHITECTURE THESAURUS (AAT)

Domain: art, architecture, decorative arts, material culture

Content: 125,000 terms

Structure: 7 facets, 33 polyhierarchies

Associated concepts *(beauty, freedom, socialism)*
Physical attributes *(red, round, waterlogged)*
Style/Period *(French, impressionist, surrealist)*
Agents: *(printmaker, architect, jockey)*
Activities: *(analysing, running, painting)*
Materials *(iron, clay, emulsifier)*
Objects: *(gun, house, painting, statue, arm)*

Synonyms

Links to 'associated' terms

Access: lexical string match;
hierarchical view

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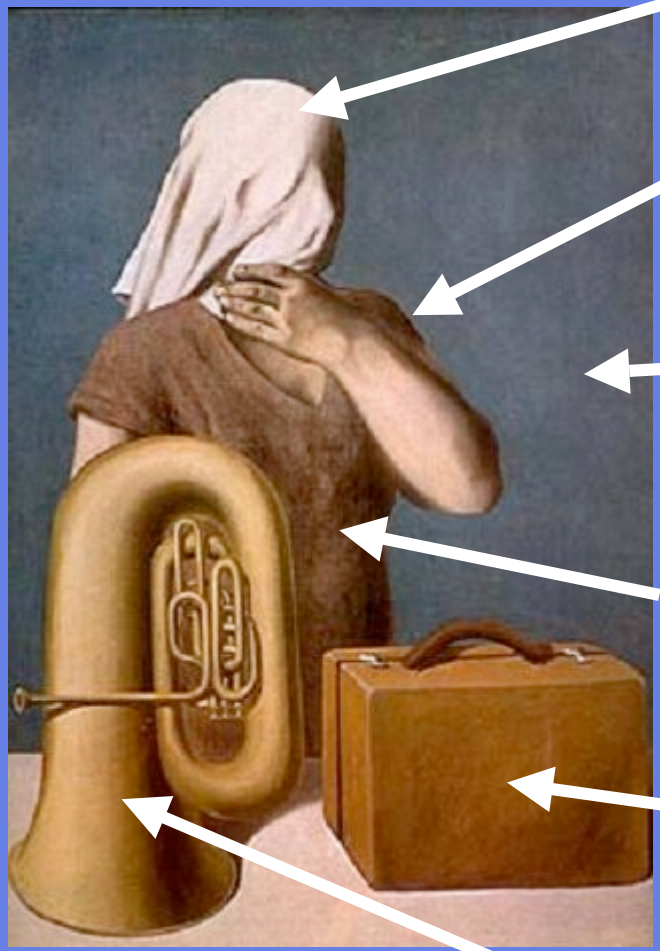
User Problems

Inter-rater variability



Headcloth				X	X				
Cloth	X	X					X		
Scarf						X			
Model Person		X		X		X			
Woman	X			X	X		X	X	
Adults			X						
Standing				X	X	X	X	X	
Background			X	X					
Brown	X	X	X	X	X				
Blue		X	X	X					
Chemise				X					
Dress					X	X	X	X	
Tunics			X						
Clothes		X							
Suitcase	X	X		X		X			
Luggage							X		
Attache case			X						
Brass Instrument				X	X		X		
French Horn		X							
Horn						X			
Tuba	X		X						

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User Problems

Inter-rater variability

New codes added per Dr per year

READ CODE	*Practice A*	*Practice B*
Sore Throat Symptom	0.6	117
Visual Acuity	0.4	644
ECG General	2.2	300
Ovary/Broad Ligament Op	7.8	809
Specific Viral Infections	1.4	556
Alcohol Consumption	0	106
H/O Resp Disease	0	26
Full Blood Count	0	838

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Repeatability
Inter-rater reliability

- Only ICPC has taken seriously
  - Originally less than 2000 well tested rubrics with proven inter-rater reliability across five languages
    - As it has been put into wider use, has grown and is less tested
- Includes the delivery software
  - Confounding, but we can't ignore it

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Where next?

The genome / 'omics explosion

- Open Biolological Ontologies (OBO)
  - Gene Ontology, Gene expression ontology (MGED), Pathway ontology (BioPAX), …
    - 400+ bio databases and growing
- National Cancer Institute Thesaurus
- CDISC/BRIDG - Clinical Trials
- HL7 genomics model…
- …

Coming to an Electronic Healthcare Record near you!

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Enter the ‘O’ word the ‘M’ word and the ‘S’ word

- “Ontologies” - claimed by philsophers, computer scientists, …
  - Logically, computationally solid skeletons
- “Metadata”
  - Applications that know what they need and resources that can say what they are about
- “Service Oriented Architectures”
  - Loosely coupled computing based on discovery
    - The GRID

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Key issue 1: Creating an open community

- Terminologies have succeeded for three reasons
  - Coercion - use them or don't get paid
    - ICD-CM, CPT, MEDDRA, Read 2
  - They belonged to the community and were useful or key to software
    - LOINC, HL7v2, Gene Ontology, Read 1 …
  - They gave access to a key resource
    - MeSH, BNF, …

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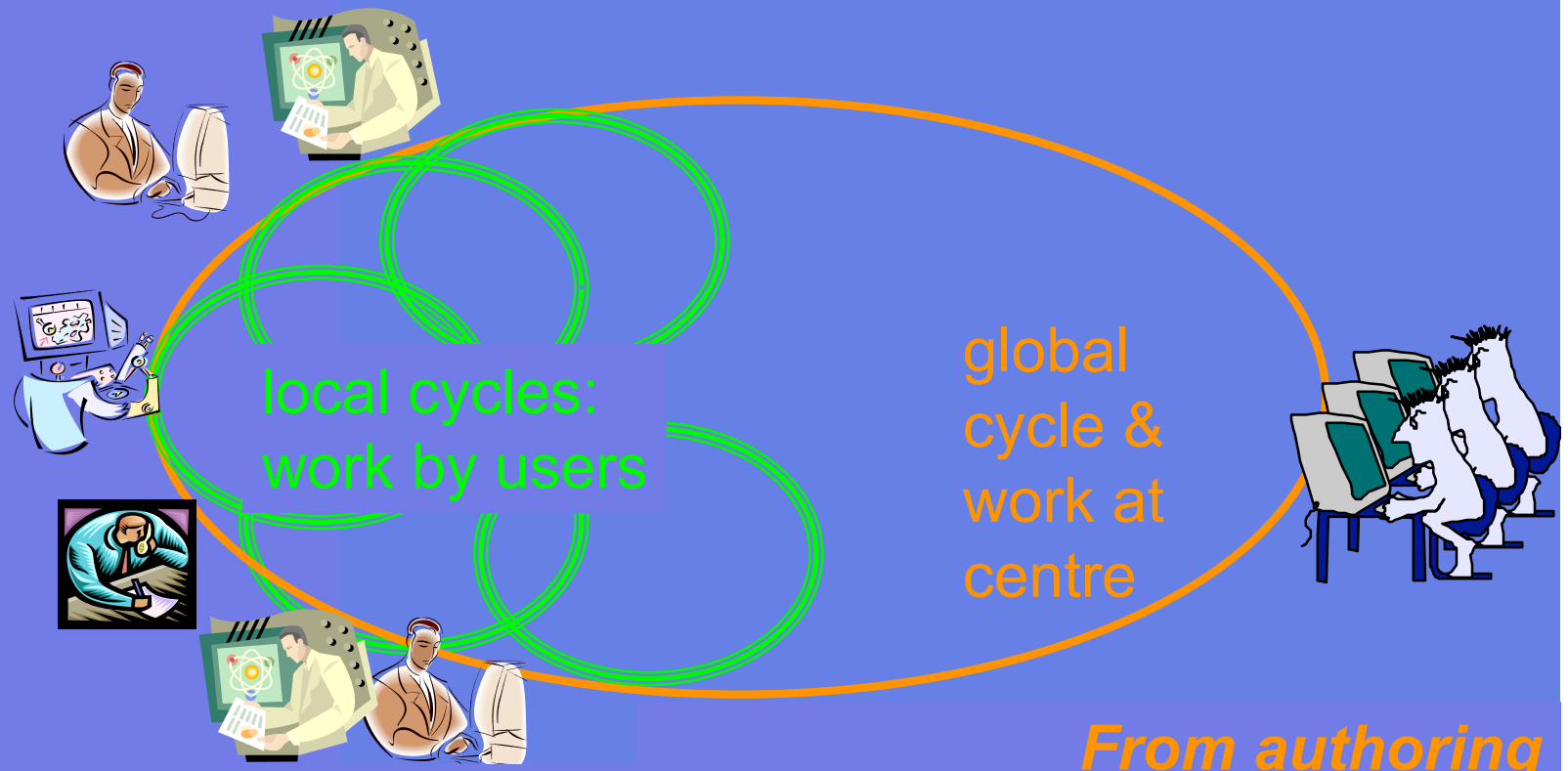
Logic + Web liberates users Open 'Just-in-time Terminology'

- If you can test the consequences then you can give users the freedom to develop
  - New compositions
  - New additions to established lists
- Hide the complexity
  - "Close to user forms"
    - GALEN's "Intermediate Representation"
      - *Training time down from 3 months to 3 days!*
        - *The logic is the assembly language*
- Move the development to the community
  - Look at OpenDirectory, Wikipedia, FLKR, etc.
    - Social computing
- Requires more and better tools
  - Requires a different style of curation

Supports Loosely coupled distributed ontology development



From 80% central/global effort
to 10% central/global effort

From authoring
to meta-authoring

User effort cut by 75% compared with manual methods
Mostly in reduced committee meetings & arguments

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Key issue II: Applications centric development

- If it is built for everything it will be fit for nothing!
  - Must have a way to see if it works
  - If it is built for just one thing it will not be fit even for that
    - Change is the only constant
- Cannot predict which abstractions needed in advance
  - Even very large ontologies tend to be missing 50% or more of terms in practice
    - Compose them when you need them and share
- Is there a optimal '90-10' point?
  - You can only tell against a specific application

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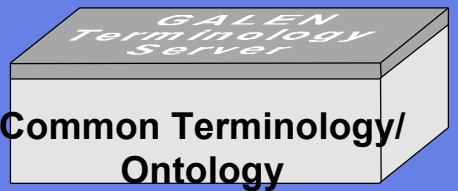
Applications centric Development

Meta-authoring

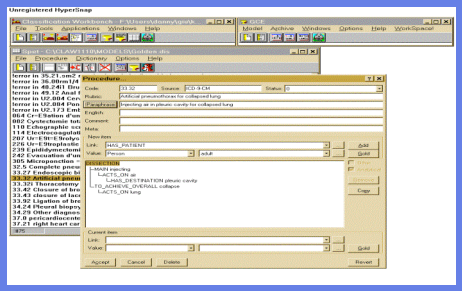
templates/ views



Common Terminology/ Ontology

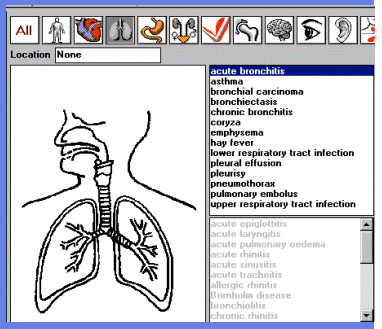
templates/ views

Meta-authoring



authoring environments Intermediate Representations

clinical applications



clinicians / Applications builders
Empowered Authors

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Key issue III: Binding to Applications & the EHR

- HL7 v3 + SNOMED = Chaos
  - Unless we can formalise the mutual constraints
    - The documentation is beyond human capacity
      - *To write or to understand*
- Templates/Archetypes + SNOMED = Missed opportunities
  - Unless we avoid trivialising terminology

… or chaos if we attempt to use the terminology

- Requires new tools
  - Formalisms probably adequate

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Key issue IV: Decision support

- Meaningful decision support is still rare
  - Terminology is not the only problem
    - But it is a barrier
  - Ontology should be the scaffolding
    - But requires the terminology to be computable
      - *SNOMED still too idiosyncratic to use easily*
  - Inter-rater reliability crucial
    - Can we afford GIGO for patient management?
  - Semantics of combined EHR+Terminology must be well defined

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Key issue V: Avoiding "Pregacy"

- Prebuilt legacy
  - Errors built in from the beginning
- ≤ .01% of SNOMED coded data to be held in 10 years time has been collected
  - Fixes now will be less expensive than fixes later
  - Rigorous schemas rigorously adhered to
  - Conformance and Regression testing
    - Cannot depend on people to do it right
      - *Must be formally verifiable*
  - It's software - Let's have some basic software engineering!

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Key issue VI: Empirical data

- Need empirical data on
  - What's worth doing - what's esssential
    - Language used by doctors
    - Terms used
  - What works
    - Reliability of terms used - errors made
    - Effect on Decision Support and other applications
  - What scales
    - What are the consequences of design decisions
  - Effort required to develop software
    - Usability of development tools
  - Effort required by users
    - Usability of interfaces and clinical systems
- Where is the science base for our work?

Key issue VII:
Human Factors-
Helping with a humanly impossible task

- Language technology will help
  - But will always have limitations
- Tailored forms will help
  - But we must beet the combinatorial explosion
- …but the key issues are organisational, social & clinical
- …and needs empirical data

Requires **serious** investment and Commitment

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Part II:Quality and Quality Assurance: What's it For?

Quality can only be assured against purpose!

Fit for what?

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Purposes of Terminology in Healthcare

- A controlled vocabulary
  - Lexicon of "Terms"
  - Management of identifiers
    - "Nonsemantic identifiers"
      - *Most Healthcare application use meaningless alphanumerics as the primary identifier*
        - *Google: Cimino Desiderata*
  - "Coverage" / "Sensitivity"
- A browsable index and finding
  - "Specificity"
- Classification/retrieval for epidemiology
- Formal representation for inference
  - Subsumption
  - Partonomy
  - Additional relations

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Quality Assurance

- Consequences
  - Inferences
  - Results in applications
- Content
  - Coverage, Precision, appropriateness
- Human factors
  - Reliability, usability
- Context: specification and binding to applications
  - Rigour and standards in context
- Process
  - Evolution, change management, responsiveness, provenance, metadata
  - Openness, transparency
  - Quality assurance procedures
  - Linkage to other resources
- Humility
  - Test against scope

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Points of testing

- On basis of documentation and publilc information
  - Inevitably makes many trivial errors over implicit assumptions
    - But what is undeniably there
- With collaboration of developers
  - Avoids trivia
    - But must make the implicit assumptions explicit if to be of value

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Consequences: Inferences and Engineering

- Ontologies are mathematical theories
  - They are tested by whether the ‘correct’ inferences follow from them
    - Within scope and for purpose
- The test of the formalism/schema is the results
  - If they give the wrong / inadequate inferences, they are inadequate
  - If they give the correct answers within scope, need strong reasons to reject
  - If two give the same inferences, then there is little to choose between them
- Criteria for “correctness”
  - Observation of the world
  - Consensus of authorities
  - Linguistic usage
- Criteria for engineering
  - Robustness
  - Change
  - *Scaling!!!*

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Content

- A Priori Coverage just a matter of size
  - Test against what purposes
  - Are the constructs there? Are the building blocks there?
    - Every application needs different abstractions
      - *Leeds to 25% - 50% raw coverage in clinical systems*
- Entitites / Concepts
  - Can all meanings be represented
- Lexicons / language
  - Are they said in the right way?
    - Use of language technology
      - *To mine for terms*
      - *To generate output*

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Human factors

- Inter-rater reliability
  - Of localisation/configuration staff
  - Of end users
  - Language
- Ease of use
  - Too big - too hard to find things
  - Too small - inadequate to say things
  - Too complex - distinctions without a difference
  - Too far from common usage - too hard to express things

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Context: Specification of use

- Rigour of specification of use
  - Binding of terminology to application
    - For medical records a particular problem
- (We'll come back to this later)

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Product and Process

- Ontologies are living artefacts
  - Must evaluate the process as well as the product
  - Updates, tracking, provenance, metadata
  - Sustainability, authority, openness, …
- The test of process is change
  - What is required to make a change
  - How long does it take
  - Test designs for change before use

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Some examples of problems in clinical terminologies

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Meaning & Use

- Nesting of Terminology and Medical record
  - Nesting of terminology in statements
    - Nesting of statements in Archetypes
  - Nesting of Archetypes in Templates
  - Nesting of templates in records
- Querying of the result
  - How do I ask if the patient has
    - Had a elevated diastolic blood pressure?
    - Has had their left ureter removed?

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Example ontology nested in the EHR

the ehr (hl7 rim)

[moodCode="Event"
subject="Relative"
code={ diabetes (subject person_in_family) }]

the ontology (snomed-ct)

→

<family_hx (assoc_find Diabetes)>

the combined meaning

What's it really mean?
What is legal? Required?
Mandatory?…

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Problems….
Negation & context Terms

- Very unlikely to be exhaustively in static terminology
  - Because too numerous
- Must not be detached from ‘kernel term’
  - Patient with ‘no heart disease’ must never be mistaken for patient with ‘heart disease’
- Terminological phenomenon
  - But places particular constraint on how the information model, and queries on it, must work
- Despite this…
  - Legacy terminologies pre-coordinated negated terms…**but only a subset**
  - Legacy information systems must therefore allow negation to cover e.g. negations not present in terminology

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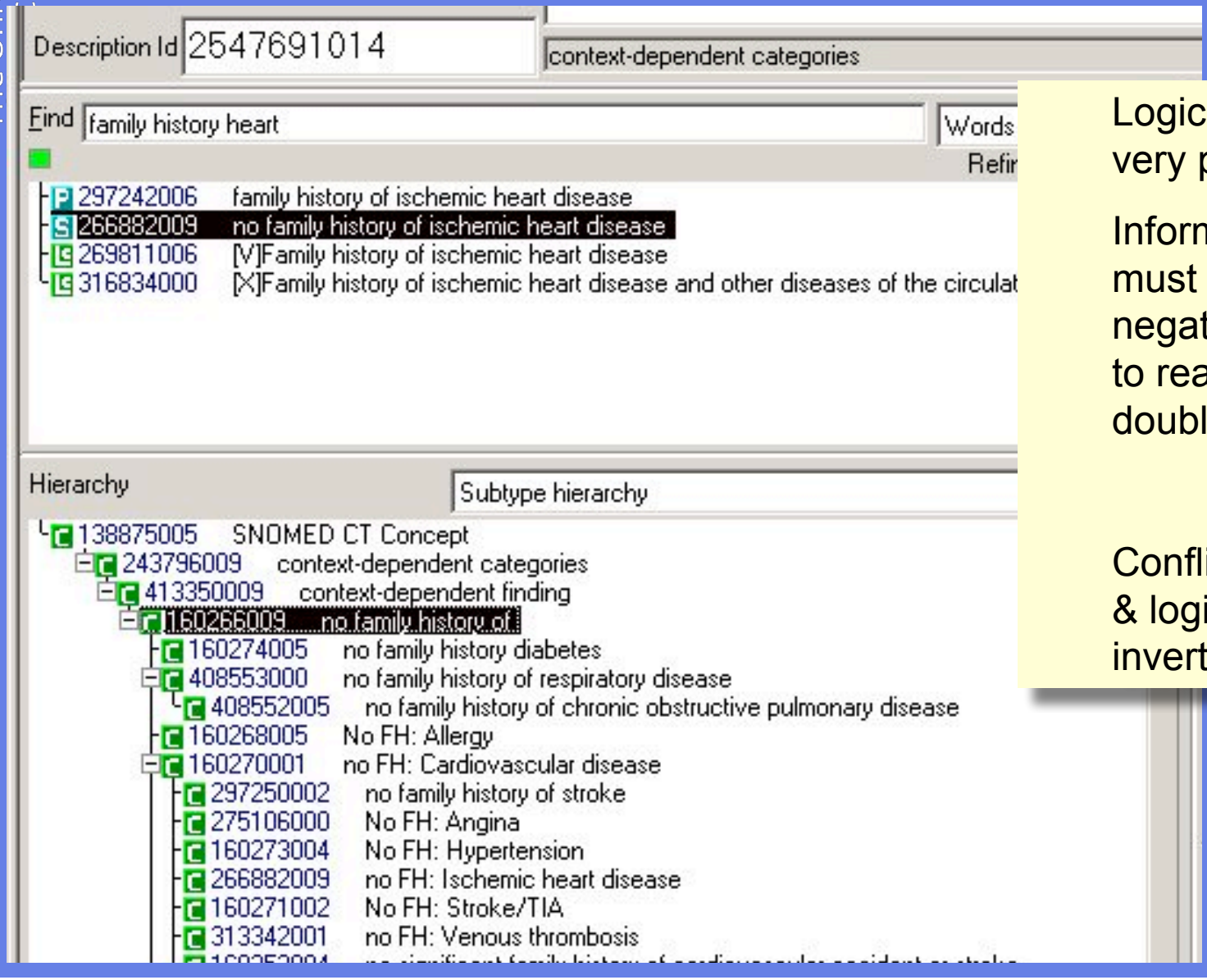
Problems….
Negation & context Terms



Logical negation very problematic

Information model must support negation…but how to reason across double negatives?

Conflict of intuition & logic - hierarchies inverted

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Summary: Lessons & Directions for terminology

- Understand scaling and the combinatorial explosion
  - All lists are too big and too small
  - Too many niches to cope with one by one
- Focus on applications: *"What's it for?"*
- *Quality assurance*
  - Consequences: Gather empirical data; Change and scaling critical
  - Content: Appropriateness and precision as well as coverage
  - Context: Rigorous specification of binding to applications
  - Process: Evolution, Openess, sustainability, linkage
  - Implicit information: Consult with developers, avoid critiquing known trivia
  - Humility: It is only good for what it's good for
    - "It won't make the coffee"
- ***Human factors!***