

# Health Terminologies and Classifications Observation on SNOMED and ICD



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Chair, International Classification of Disease, WHO

James Read Memorial Lecture  
SNOMED CT Implementation Showcase  
Crystal City, VA 11 Oct 2013

# Health Care Is An Information Intensive Industry

- Control of Health Care Costs ...
- Improved Quality of Care ...
- Improved Health Outcomes ...
- Appropriate Use of Health Technology...
- Compassionate Resource Management...
-  ... depend upon information
-  ... Ultimately Patient Data

# Medical Concepts

## Events, Observations, Interventions

- How should we represent it? Language:
  - Nuance, detail, unfettered combination
  - Timely, current, never obsolete
  - Natural, friendly, established
  - [Ambiguous, imprecise, unpredictable]
- Codes:
  - Concise, precise
  - Structured, consistent, well formed
  - Analyzable, manipulable
  - [Rigid, tedious, high maintenance]

# Will Big Data Save Us?



Genetics  
inMedicine

REVIEW

Genet Med 15: 802-809;  
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Oct, 2013

## Some experiences and opportunities for big data in translational research

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Health care has become increasingly information intensive. The advent of genomic data, integrated into patient care, significantly accelerates the complexity and amount of clinical data. Translational research in the present day increasingly embraces new biomedical discovery in this data-intensive world, thus entering the domain of “big data.” The Electronic Medical Records and Genomics consortium has taught us many lessons, while simultaneously advances in commodity computing methods enable the academic community to affordably manage and process big data. Although great promise can emerge from the adoption of big data methods and philosophy, the heterogeneity and complexity of clinical data, in particular, pose additional challenges

for big data inferencing and clinical application. However, the ultimate comparability and consistency of heterogeneous clinical information sources can be enhanced by existing and emerging data standards, which promise to bring order to clinical data chaos. Meaningful Use data standards in particular have already simplified the task of identifying clinical phenotyping patterns in electronic health records.


*Genet Med* advance online publication 5 September 2013

**Key Words:** clinical data representation; big data; genomics; health information technology standards

# Origins of Big Science

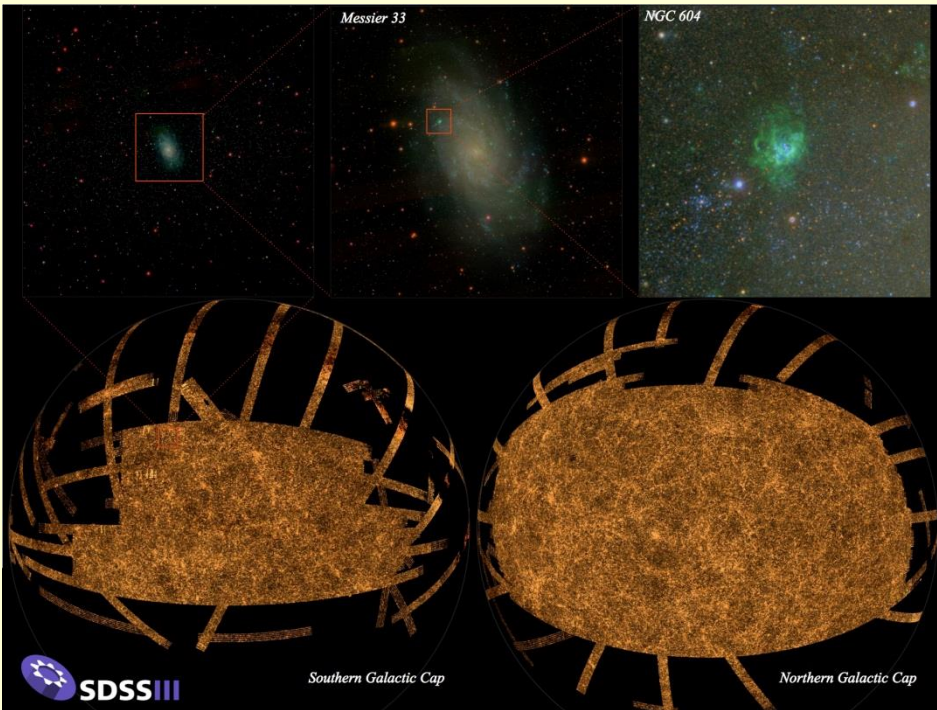
## Astronomy

*Sc. no. Principi.*  
 Galileo Galilei, Humilis. Servus della Ser. V. inuigilata.  
 Io aspirando et di ogni spirito di potere ho solam satisfare  
 alvario che nome della littera di Mathematici nelle Scu.  
 di Padova,  
 Inuere d'auere determinato di presentare al Ser. Principe  
 l'Orchile et il p. essere di giuramento inestimabile di ogni  
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 ste nuove artificio nel maggior segreto et obsequio a disposizione  
 di V. Ser. L'Orchile sanato dalle piu uide speculazioni di  
 prospettiva in l'uantaggio di scoprire l'ogni et Sole dell' inuisibile  
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 il numero et la qualita dei Vesselli giudiare la sue forze  
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 nella campagna aperta uilere et particolarly distinguere ogni suo  
 moto et propriamento.  
 Ad. 7. di gennaio  
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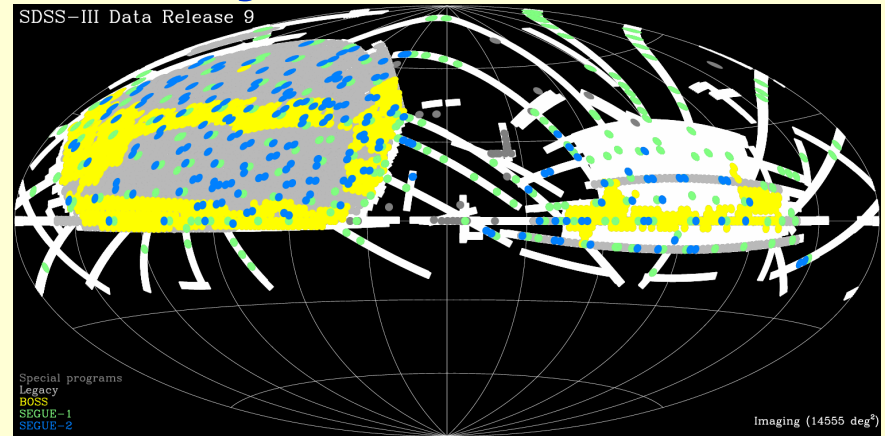





# Sloan Digital Sky Survey III – DR9

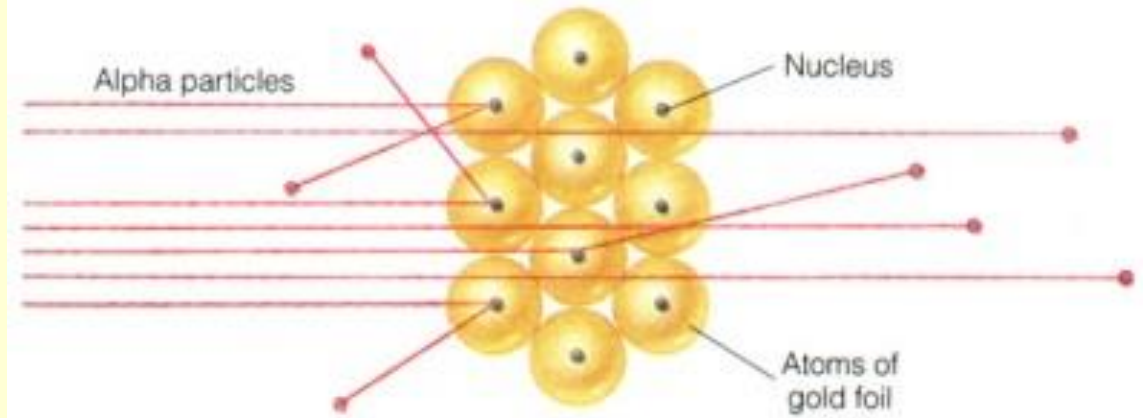
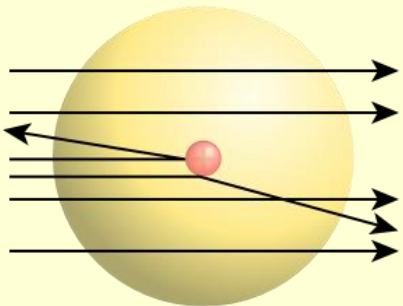
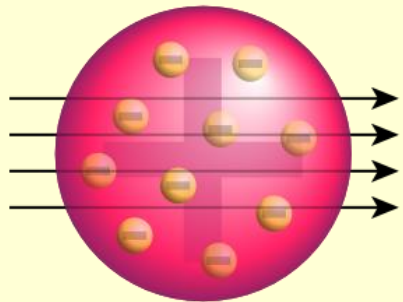
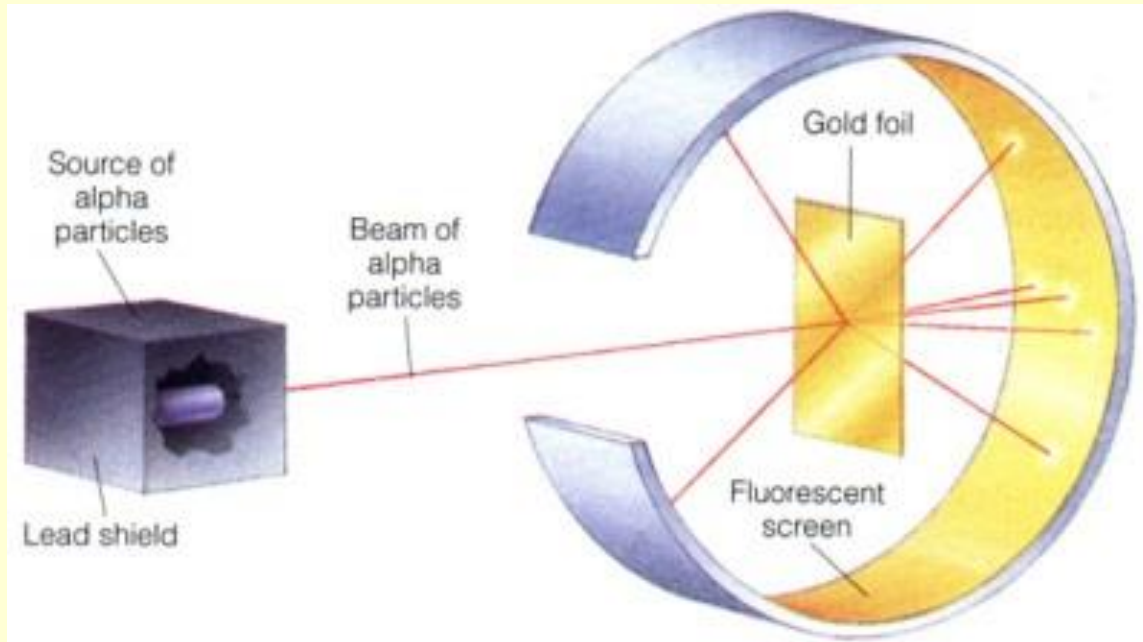


- Images
- Spectra
- Object catalog
- Metadata



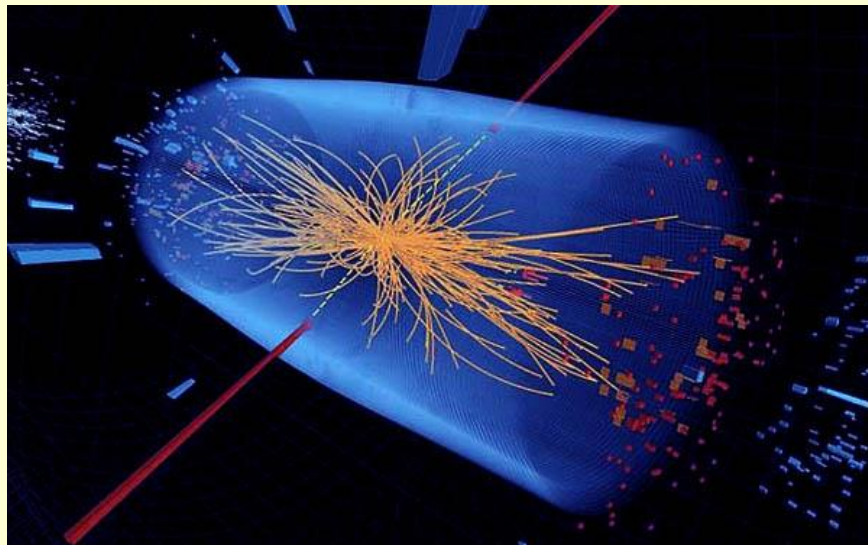
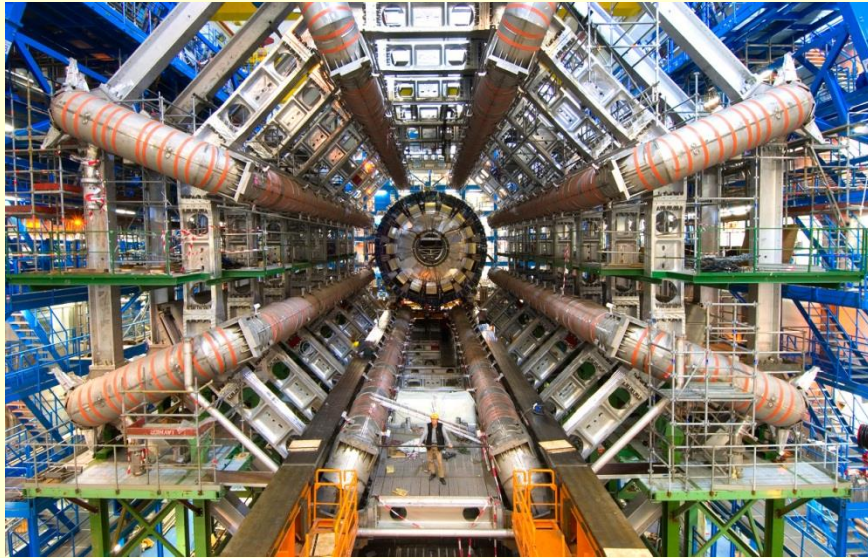
|  |                                     |
|--|-------------------------------------|
| Total area of imaging                    | 31,637 square degrees               |
| Image field size                         | 1361x2048 pixels                    |
| Number fields                            | 938,046 (excluding supernovae runs) |
| Catalog objects                          | 1,231,051,050                       |
| Number of <u>unique, primary</u> sources |                                     |
| Total                                    | 469,053,874                         |
| Stars                                    | 260,562,744                         |
| Galaxies                                 | 208,478,448                         |
| Unknown                                  | 12,682 <sup>6</sup>                 |

# Rutherford "Table-top" Experiment





# That Higgs Boson



- 600 institutions
- 10,000 scientists
  - 2 Nobel prizes
- 800 trillion collisions
- 200PB of data =
  - $2 \times 10^{17}$  bytes of data

Boarding on an astronomical number in its own right!

- $\$13.25 \cdot 10^9$  USD



# Dimensionality of Higgs “Big Data”

- Mass
  - Direction
  - Energy
- 
- **Medicine is more complicated than that**

# Dimensionality of Big Data

- Broad
  - Small amounts of data; Huge number observations
  - National Claims data
- Deep
  - Large amounts of data; Few observation
  - NGS Complete Genome
- Rich
  - Broad and Deep
  - Clinical Phenotyping data (EMRs)
    - Labs, Vitals, Exam, Waveform, Images, Omics, ...
    - Social, environmental, diet,

# Does Big Data Change Criteria for Scientific Evidence?

that “society will need to shed some of its obsession for causality in exchange for simple correlations: not knowing *why* by only *what*. This overturns centuries of established practices and challenges our most basic understanding of how to make decisions and comprehend reality.”

Mayer-Schönberger, V. and K. Cukier, *Big data : a revolution that will transform how we live, work, and think*. 2013, Boston: Houghton Mifflin Harcourt. 242 p.



# Actionable Knowledge: More than just Google Search

- Well known “spurious association” problem
  - Reproducibility
- Power Issues [Have I seen a “patient like that”]
  - Single drug vs. single side effect
  - Stratify across cells by:
    - Age and Sex
    - Co-morbidity
    - Lab values (nomal vs non-normal)
    - Image findings
    - Waveform findings
    - Genetic profile (and other “omics”)

# Counterfactual: Mining Gold from Dirty Big Data

Brief communication



Mar  
2013

## Web-scale pharmacovigilance: listening to signals from the crowd

Ryen W White,<sup>1</sup> Nicholas P Tatonetti,<sup>2</sup> Nigam H Shah,<sup>3</sup> Russ B Altman,<sup>4</sup> Eric Horvitz<sup>1</sup>

► Additional material is published online only. To view please visit the journal online (<http://dx.doi.org/10.1136/amiajnl-2012-001482>).

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### ABSTRACT

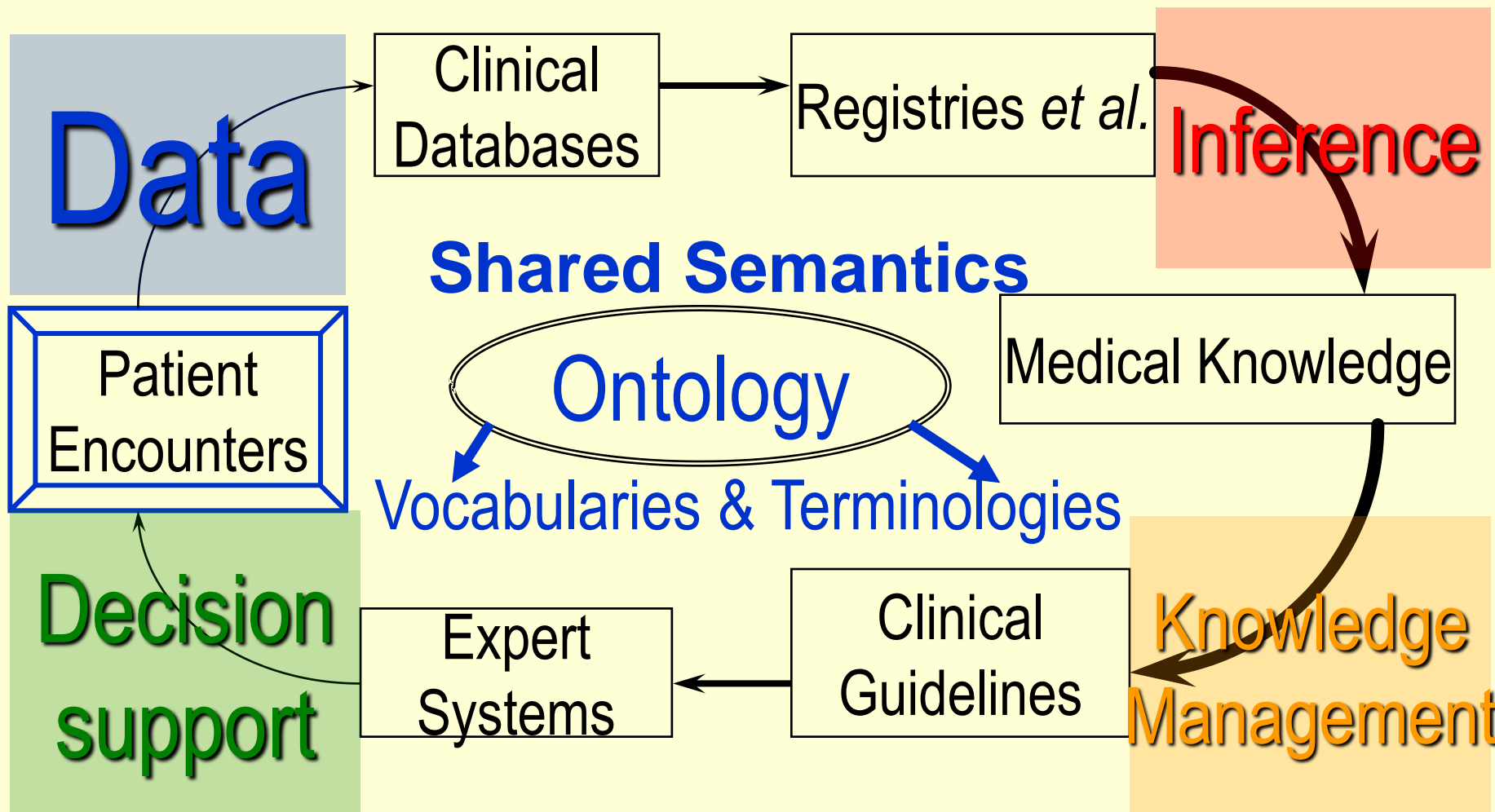
Adverse drug events cause substantial morbidity and mortality and are often discovered after a drug comes to market. We hypothesized that Internet users may provide early clues about adverse drug events via their online information-seeking. We conducted a large-scale study of Web search log data gathered during 2010. We pay particular attention to the specific drug pairing of paroxetine and pravastatin, whose interaction was reported to cause hyperglycemia *after* the time period of the online logs used in the analysis. We also examine sets of drug pairs known to be associated with hyperglycemia and those not associated with hyperglycemia. We find that anonymized signals on drug interactions can be mined from search logs. Compared to analyses of other sources such as electronic health records (EHR), logs are inexpensive to collect and mine. The results demonstrate that logs of the search activities of populations of computer users can contribute to drug safety surveillance.

case an interaction between paroxetine (an anti-depressant) and pravastatin (a cholesterol-lowering drug), which was recently reported to create hyperglycemia.<sup>13 14</sup> This association was extracted from the US Food and Drug Administration adverse event reporting system (AERS) using a data-mining algorithm that aggregates reports to identify drug–drug interactions.<sup>13</sup> The finding was confirmed in a retrospective analysis of the electronic health records of three regionally distinct medical institutions and confirmed in a mouse model.<sup>14</sup> We hypothesized that patients taking these two drugs might experience symptoms of hyperglycemia and may have conducted internet searches on these symptoms and concerns related to hyperglycemia before the association was reported in 2011.

### METHODS

We analyzed the search logs of millions of consenting web users who opted to share search activities

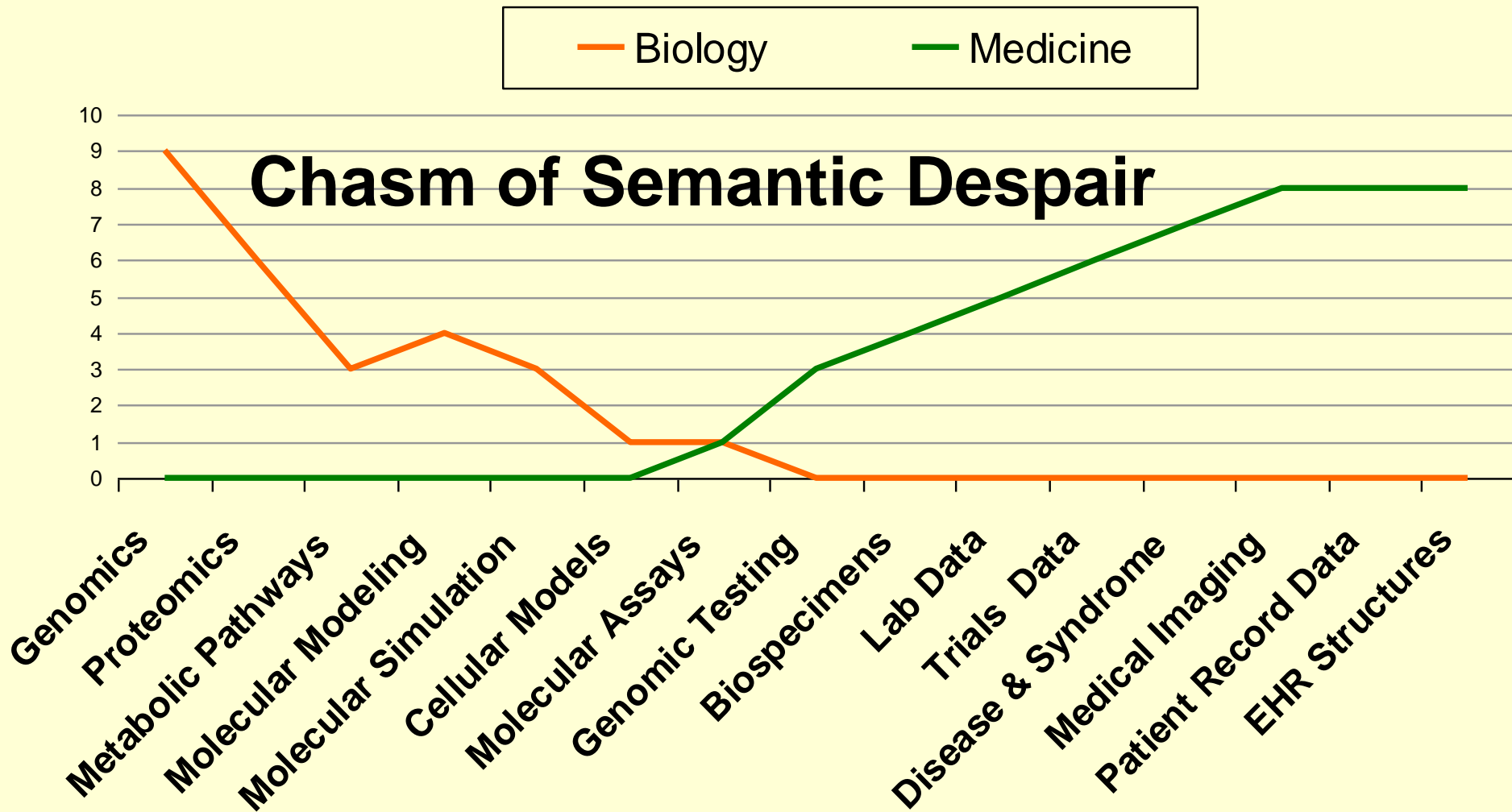
# From Practice-based Evidence to Evidence-based Practice





# The Continuum Of Biomedical Informatics

## Bioinformatics meets Medical Informatics



## Blois, 1988

### Medicine and the nature of vertical reasoning

- Molecular: receptors, enzymes, vitamins, drugs
- Genes, SNPs, gene regulation
- Physiologic pathways, regulatory changes
- Cellular metabolism, interaction, meiosis,...
- Tissue function, integrity
- Organ function, pathology
- Organism (Human), disease
- Sociology, environment, nutrition, mental health...

# Terminology as Crucial Requirement

*Without Terminology Standards...*

- Health Data is *non-comparable*
- Health Systems *cannot* Interchange “Data”
- Secondary Uses (Research) are *not* practical
- Big Data methodologies cannot leverage epidemiologic principles for observational data
  - Adjustment for confounding
    - Stratification
    - Multivariate models
    - Machine learning features



*Natural and Political*  
**OBSERVATIONS**

Mentioned in a following INDEX,  
and made upon the  
Bills of Mortality.

1910  
1665  
245.

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BY  
Capt. **JOHN GRANT**,  
Fellow of the *Royal Society*.

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With reference to the *Government, Reli-  
gion, Trade, Growth, Air, Diseases*, and the  
several Changes of the said **CITY**.

— *Non, me ut miretur Turba, laboro,  
Contentus paucis Lectoribus.* —

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The Third EDITION,  
much Enlarged.

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LONDON,  
Printed by *John Martyn*, and *James Allestry*,  
Printers to the *Royal Society*, and are to be sold at the  
sign of the *Bell* in *St. Pauls Church-yard*.  
MDC LX V.



# The Table of CASUALTIES.

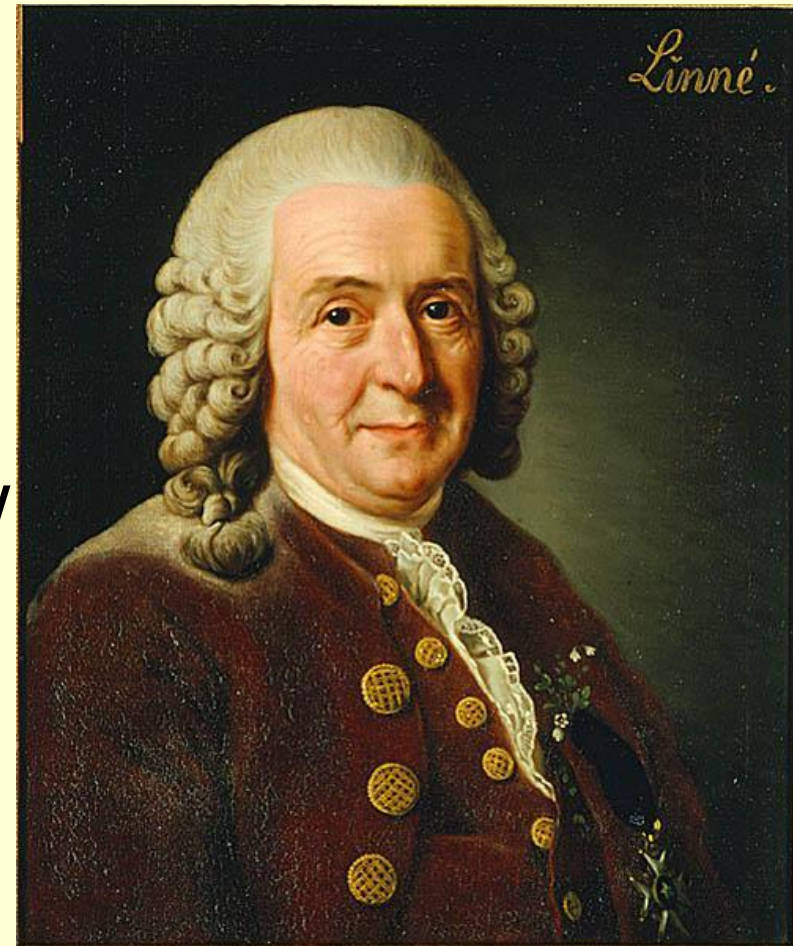
The Years of our Lord

|                                | 1647 | 1648 | 1649 | 1650 | 1651 | 1652 | 1653 | 1654 | 1655 | 1656 | 1657 | 1658 | 1659 | 1660 | 1661 | 1662 | 1663 | 1664 |
|--------------------------------|------|------|------|------|------|------|------|------|------|------|------|------|------|------|------|------|------|------|
| Abortive and Stil-born         | 335  | 329  | 327  | 351  | 389  | 381  | 384  | 433  | 483  | 419  | 463  | 467  | 421  | 544  | 499  | 439  | 410  | 410  |
| Aged                           | 916  | 835  | 889  | 696  | 780  | 834  | 864  | 974  | 743  | 892  | 869  | 1176 | 909  | 1095 | 579  | 712  | 661  | 661  |
| Ague and Fever                 | 1260 | 884  | 751  | 970  | 1038 | 1212 | 282  | 1371 | 689  | 875  | 999  | 1800 | 2303 | 2148 | 956  | 1091 | 1115 | 1115 |
| Apoplex and Suddenly           | 68   | 74   | 64   | 74   | 106  | 111  | 118  | 86   | 92   | 102  | 113  | 138  | 91   | 67   | 22   | 36   |      |      |
| Bleach                         |      |      | 1    | 3    | 7    | 2    |      |      |      | 1    |      |      |      |      |      |      |      |      |
| Blasted                        | 4    | 1    |      | 6    | 6    |      |      | 4    |      | 5    | 5    | 3    | 8    | 13   | 8    | 10   |      |      |
| Bleeding                       | 3    | 2    | 5    | 1    | 3    | 4    | 3    | 2    | 7    | 3    | 5    | 4    | 7    | 2    | 5    | 2    | 5    |      |
| Bloody Flux, Scouring and Flux | 155  | 176  | 802  | 289  | 833  | 762  | 200  | 386  | 168  | 368  | 362  | 233  | 346  | 251  | 449  | 438  | 352  | 352  |
| Burnt and Scalded              | 3    | 6    | 10   | 5    | 11   | 8    | 5    | 7    | 10   | 5    | 7    | 4    | 6    | 6    | 3    | 10   | 7    |      |
| Calenture                      | 1    |      |      | 1    | 2    | 1    | 1    |      |      |      | 3    |      |      |      |      |      |      |      |
| Cancer, Gangrene and Fistula   | 26   | 29   | 31   | 19   | 31   | 53   | 36   | 37   | 73   | 31   | 24   | 35   | 63   | 52   | 20   | 14   | 23   |      |
| Wolf                           |      |      |      | 8    |      |      |      |      |      |      |      |      |      |      |      |      |      |      |
| Canker, Sore-mouth and Thrush  | 66   | 28   | 54   | 42   | 68   | 51   | 53   | 72   | 44   | 81   | 19   | 27   | 73   | 68   | 6    | 4    | 4    |      |
| Child-bed                      | 161  | 106  | 114  | 117  | 206  | 213  | 158  | 192  | 177  | 201  | 236  | 225  | 226  | 194  | 150  | 157  | 112  | 112  |
| Chrisoms and Infants           | 1369 | 1254 | 1065 | 990  | 1237 | 1280 | 1050 | 1343 | 1089 | 1393 | 1162 | 1144 | 858  | 1123 | 2596 | 2378 | 2035 | 222  |
| Colick and Wind                | 103  | 71   | 85   | 82   | 76   | 102  | 80   | 101  | 85   | 120  | 113  | 179  | 116  | 167  | 48   | 57   |      |      |
| Cold and Cough                 |      |      |      |      |      |      | 41   | 36   | 21   | 58   | 30   | 31   | 33   | 24   | 10   | 58   | 51   |      |
| Consumption and Cough          | 2423 | 2200 | 2388 | 1988 | 2350 | 2410 | 2286 | 2868 | 2606 | 3184 | 2757 | 3610 | 2982 | 3414 | 1827 | 1910 | 1713 | 1713 |
| Convulsion                     | 684  | 491  | 530  | 493  | 569  | 653  | 600  | 828  | 702  | 1027 | 807  | 841  | 742  | 1031 | 52   | 87   | 18   | 2    |
| Cramp                          |      |      | 1    |      |      |      |      |      |      |      |      |      |      |      |      |      |      | 1    |
| Cut of the Stone               |      | 2    | 1    | 3    |      | 1    | 1    | 2    | 4    | 1    | 3    | 5    | 6    | 4    |      |      |      |      |
| Dropsie and Tympany            | 185  | 434  | 421  | 508  | 444  | 556  | 617  | 704  | 660  | 706  | 631  | 911  | 646  | 872  | 235  | 252  | 279  | 279  |
| Drowned                        | 47   | 40   | 30   | 27   | 49   | 50   | 53   | 30   | 43   | 49   | 63   | 60   | 57   | 48   | 43   | 33   | 29   |      |
| Excessive drinking             |      |      | 2    |      |      |      |      |      |      |      |      |      |      |      |      |      |      |      |
| Executed                       | 8    | 17   | 29   | 43   | 24   | 12   | 19   | 21   | 19   | 22   | 20   | 18   | 7    | 18   | 19   | 13   | 12   |      |
| Fainted in a Bath              |      |      |      |      | 1    |      |      |      |      |      |      |      |      |      |      |      |      |      |
| Falling-Sickness               | 3    | 2    | 2    | 3    |      | 3    | 4    | 1    | 4    | 3    | 1    |      | 4    | 5    | 3    | 10   | 7    |      |
| Flox and small Pox             | 139  | 400  | 1190 | 184  | 525  | 1279 | 139  | 812  | 1294 | 823  | 835  | 409  | 1523 | 354  | 72   | 40   | 58   | 58   |
| Found dead in the Streets      | 6    | 6    | 9    | 8    | 7    | 9    | 14   | 4    | 3    | 4    | 9    | 11   | 2    | 6    | 18   | 33   | 20   |      |
| French-Pox                     | 18   | 29   | 15   | 18   | 21   | 20   | 20   | 20   | 29   | 23   | 25   | 53   | 51   | 31   | 17   | 12   | 12   |      |
| Frighted                       | 4    | 4    | 1    |      | 3    |      | 2    |      | 1    | 1    |      |      |      | 9    | 1    |      |      |      |



# Flawed Information Model

- Carolus Linnaeus  
Carl von Linné
  - *Genera Morborum* (1763)
- Underscored Content Difficulty
  - Pathophysiology vs Manifestation  
e.g. Rabies as psychiatric disease
- Lacked the Germ Theory of Disease
  - Was not incorporated into an information model





# The Genomic Era

- The genomic transformation of medicine far exceeds the introduction of antibiotics and aseptic surgery
- The binding of genomic biology and clinical medicine will accelerate
- The implications for shared semantics across the basic science and clinical communities are unprecedented

## VIEWPOINT

**JAMA**

April 10, 2013, Vol 309, No. 14

# Genomic Medicine, Health Information Technology, and Patient Care

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Christopher G. Chute, MD, DrPH

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Isaac S. Kohane, MD, PhD

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**C**ELEBRATING THE TENTH ANNIVERSARY OF COMPLETING the draft human genome sequence in 2011, authors from the National Human Genome Research Institute of the US National Institutes of Health outlined the influence of genomic understanding across 5 domains: structure, the biology of the genome, the biology of disease, medicine, and improvements in health care.<sup>1</sup> The authors assert that this is the era of enhanced genomic understanding of medicine, which is expected to usher in improvements in health care effectiveness by the end of this decade. It is thus fitting to explore how health information technology will contribute to or hamper the promise of genomic medicine.

pital and medical practice in the country. Given the accelerating pace of genomic discovery, this is neither efficient nor scalable. Any expectation that a clinician can or should “know” the vast permutation of emerging genomic influences on disease risk, treatment, or prognosis, as well as the interactions of these influences with drugs or other diseases or, most confusingly, their co-occurrence with other genomic or environmental factors, is unrealistic.

The state of the art for academic medical centers in 2013 is determining a small number of relatively high-profile genomic variants from some or all of their patients at risk for specific drug treatments and integrating these findings into the electronic health records (EHRs) of those patients. Then, if a drug such as warfarin, clopidogrel, mercaptopurine, or codeine is ordered and a clinically significant drug-gene interaction is known, an alert to the physician or pharmacist is made, and in some settings an alternative recommended

# Naming System Universally Needed

“The distinction of the genera of diseases, the distinction of the species of each, and often even that of the varieties, I hold to be a necessary foundation of every plan of physic, whether dogmatical or empirical.”

- William Cullen, Edinburgh, 1785  
*Synopsis Nosologie Methodicae*

# Weights and Measures

## William Farr

“The nomenclature is of as much importance in this department of inquiry, as weights and measures in the physical sciences, and should be settled without delay.”

- First Annual Report of the Registrar-General of Births, Deaths, and Marriages in England. London: 1839 p. 99.



## What was it that James Read (and others) was trying to do?

- Use computers in General Practice?
  - Create a coding system?
  - Manage clinical documentation?
- 
- Support secondary use – analytics
  - Discover what helps and what hurts – effectiveness
  - Computer aided management – decision support



## Then What Makes for A Good Coding System?

- Cimino desiderata – ~~NEC~~ ~~NOS~~
- Terminology vs. Classification
- Whither Ontology?
- What kind of coding system are we talking about?
  - Atomic reference terminology?
  - Problem list coding scheme?
  - Human entry terms?
  - NLP mapping space?
  - Foundation for analytics and secondary use?
  - Clinical decision support triggers, authoring?
  - Reimbursement?

# Monolith vs Federation

- Domains in Healthcare proliferate
  - Diseases to biological pathways to genes
- Should all concepts be represented in a single terminological framework?
- Is the alternative of a suite of coordinated, interlocking, non-overlapping, nomenclatures preferable
- How should terminologies relate to classifications?

# Whither SNOMED?

- Terminology vs. Classification issue is gracefully evolving!
- Reference terminology vs. Entry terms must be reconciled
  - Target for NLP processing
  - Ultimately target for speech recognition parsing
- Integration of genomic disease characteristics
- Should domain-specific terminologies derive?
  - Moral equivalent of Linearizations for SNOMED?
  - Are such derivatives entry terminologies?

# Celebrate Collaboration Achievements

- WHO – ICD “The Common Ontology”
- GMDNA – Device nomenclature
- LOINC – Peace and happiness finally
- Drugs –coordination clearly emerging

Clear

cellulitis rt foot with osteomyelitis of the

Add to Working List

Add to Personal List

P1: Sir William Osler, MD 10:30 AM

Cellulitis: M-41650

    Right foot: T-D9710

Osteomyelitis: D1-60210

    Third metatarsal bone: T-12870

Without: G-C009

    Lymphangitis: D3-A0040

#### Suggested Terms

Right foot: T-D9710

Cellulitis: M-41650

Osteomyelitis: D1-60210

Rel

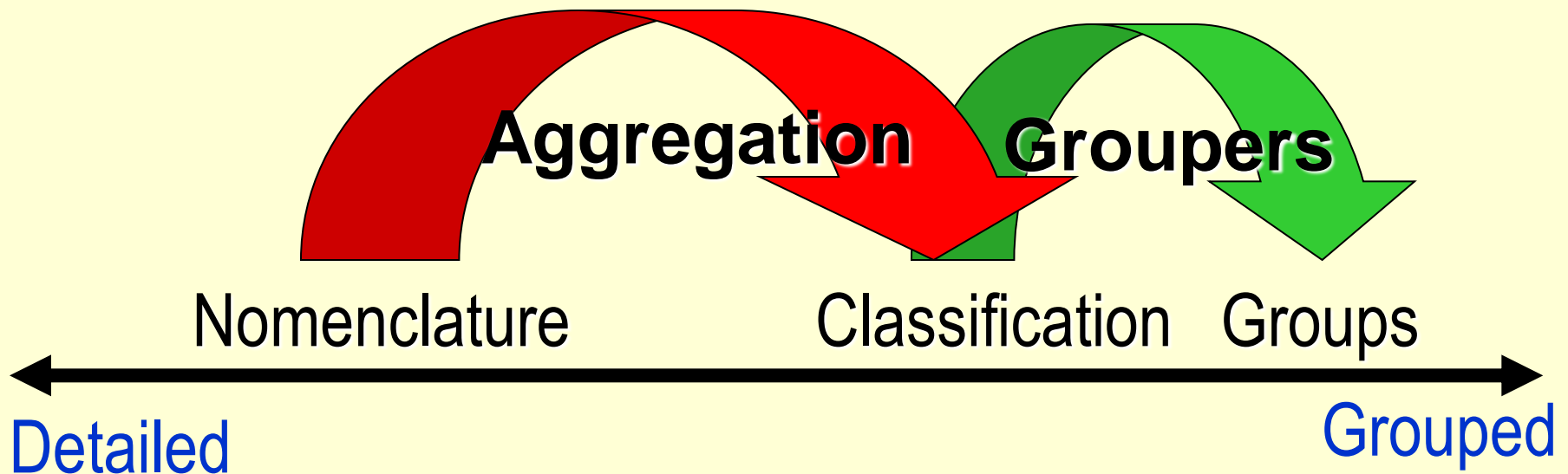
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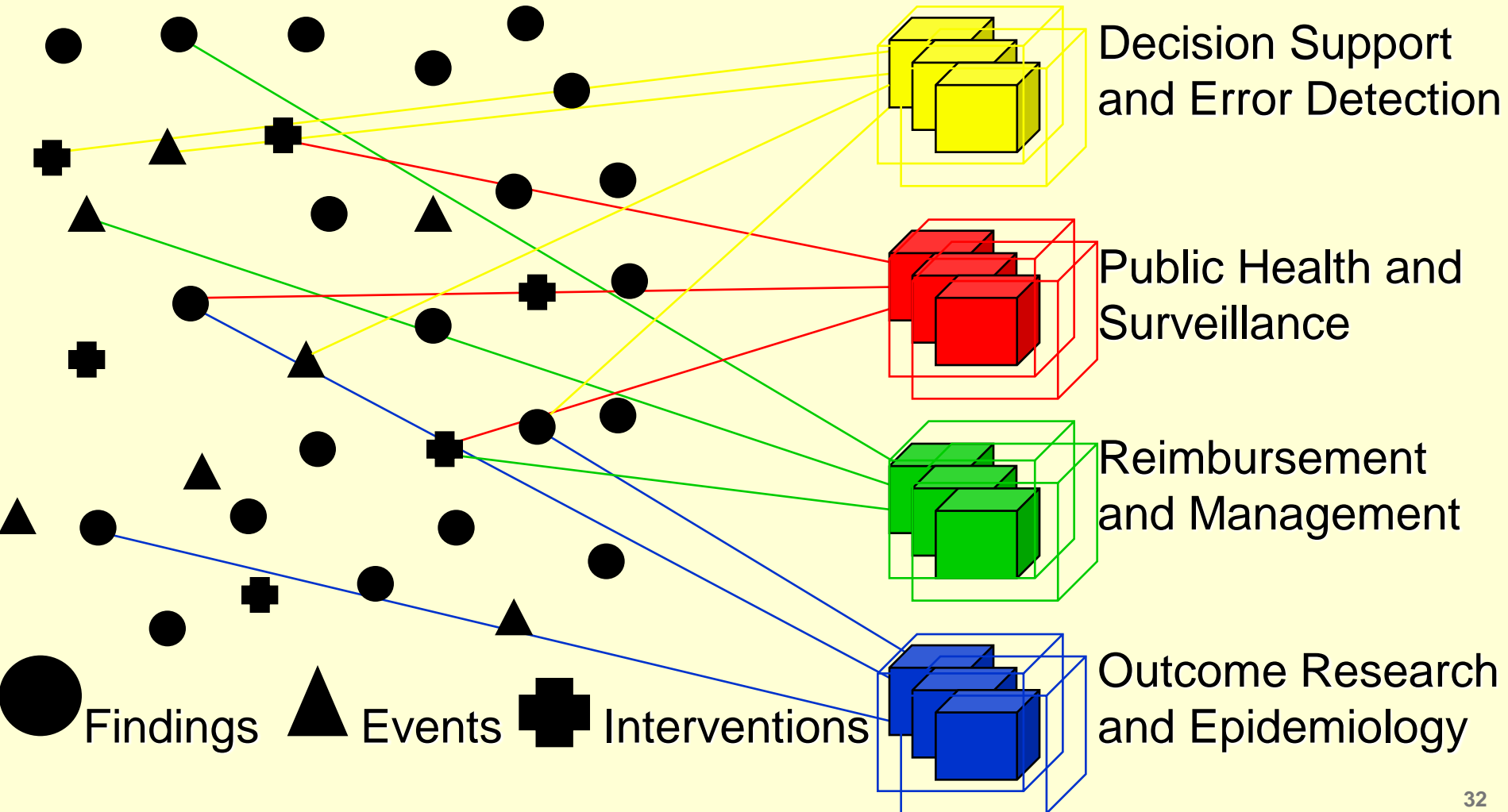
# Familiar Points Along Continuum Modern Health Vocabularies

- Nomenclature – Highly Detailed Descriptions (SNOMED)
- Classification – Organized Aggregation of Descriptions into a Rubric (ICDs)
- Groupings – High Level Categories of Rubrics (DRGs)



# Aggregation Logics by domain

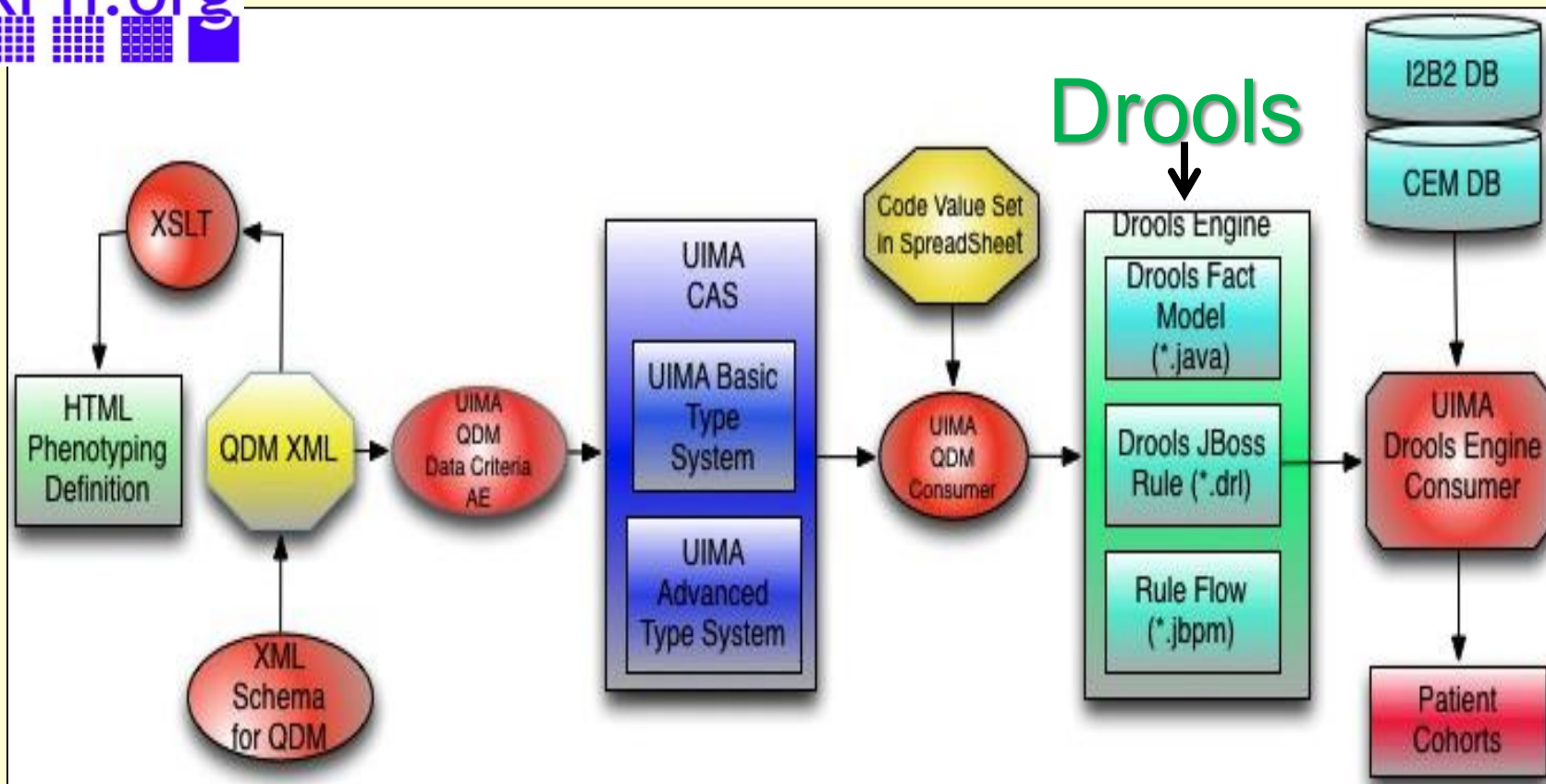
## rule-based aggregations



# Modeling and Executing Electronic Health Records Driven Phenotyping Algorithms using the NQF Quality Data Model and JBoss® Drools Engine

Dingcheng Li, PhD<sup>1</sup>, Gopu Shrestha, MS<sup>1</sup>, Sahana Murthy, MS<sup>1</sup> Davide Sottara, PhD<sup>2</sup>  
Stanley M. Huff, MD<sup>3</sup> Christopher G. Chute, MD, DrPH<sup>1</sup> Jyotishman Pathak, PhD<sup>1</sup>  
<sup>1</sup>Mayo Clinic, Rochester, MN <sup>2</sup>University of Bologna, Italy <sup>3</sup>Intermountain Healthcare, Salt Lake City, UT

SHARPN.org



# Initial Premises for ICD-11 development 2007

- Rubrics defined by Aggregation Logics from terminologies [Clinical Criteria phenotypes]
- Human language definitions will be explicit ✓
- “core” representation will be in description logic based ontology [Common Ontology, queries]
- A linear serialization will be derived as a view to maintain longitudinal consistency
  - May require corresponding “rules” for practical use
  - [sanctioning rules, also post-coordination]

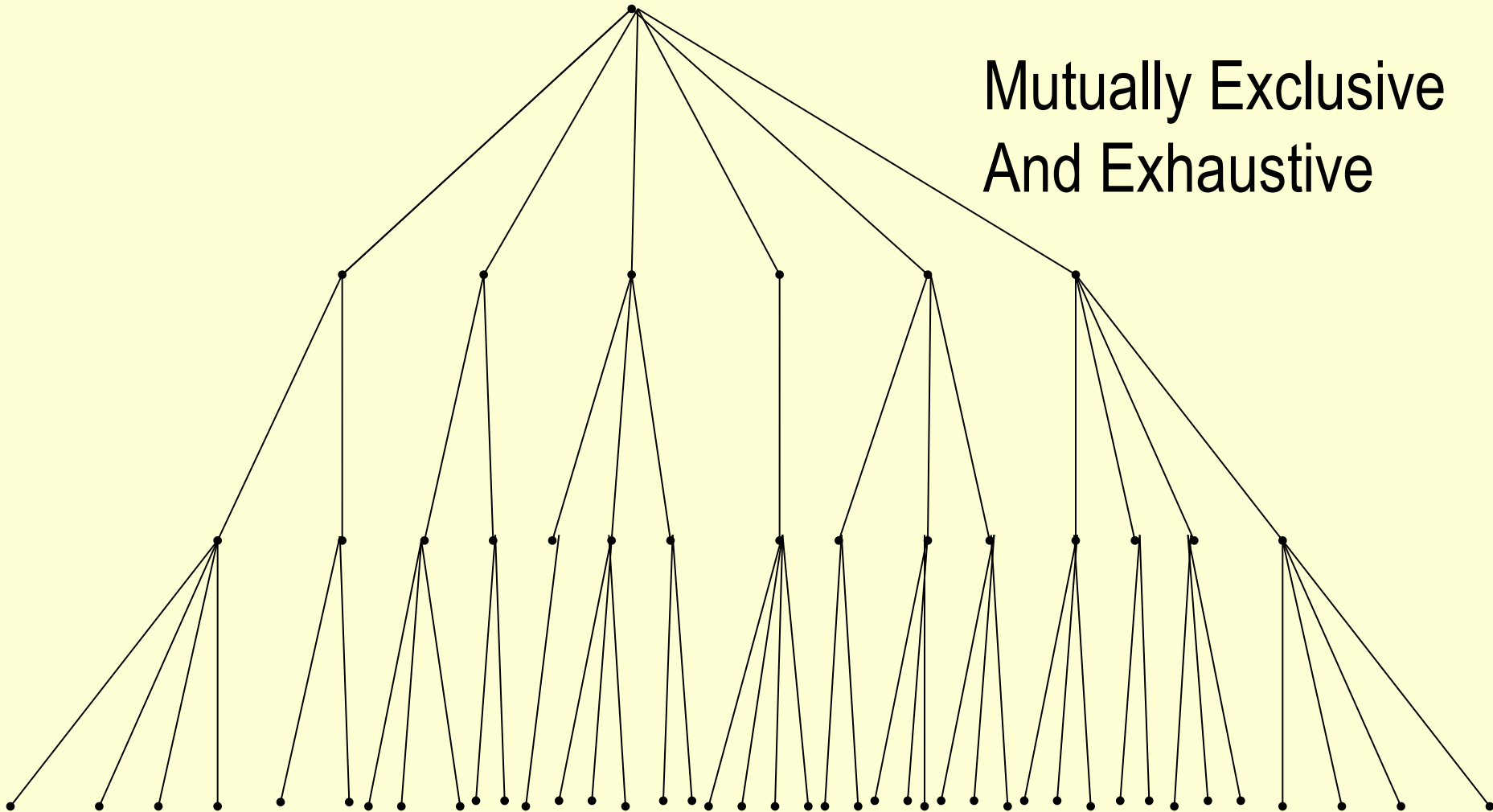
# ICD11 Use Cases

- Scientific consensus of clinical phenotype
- Public Health Surveillance
  - Mortality
  - Public Health Morbidity
- Clinical data aggregation
  - Metrics of clinical activity
  - Quality management
    - Patient Safety
  - Financial administration
    - Case mix
    - Resource allocation



# Traditional Hierarchical System ICD-10 and family

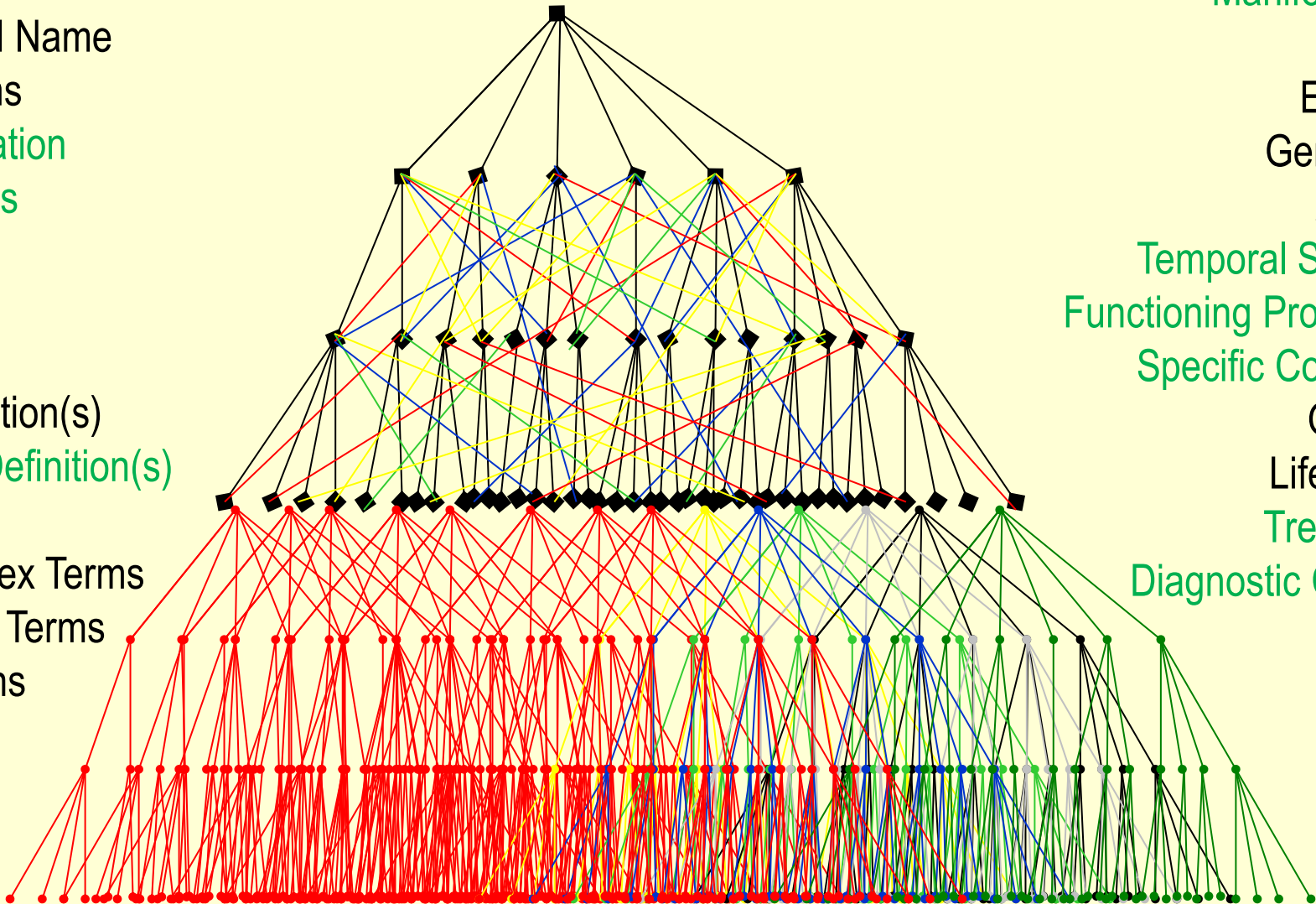
Mutually Exclusive  
And Exhaustive



# The ICD11 Foundation Layer a Semantic Network

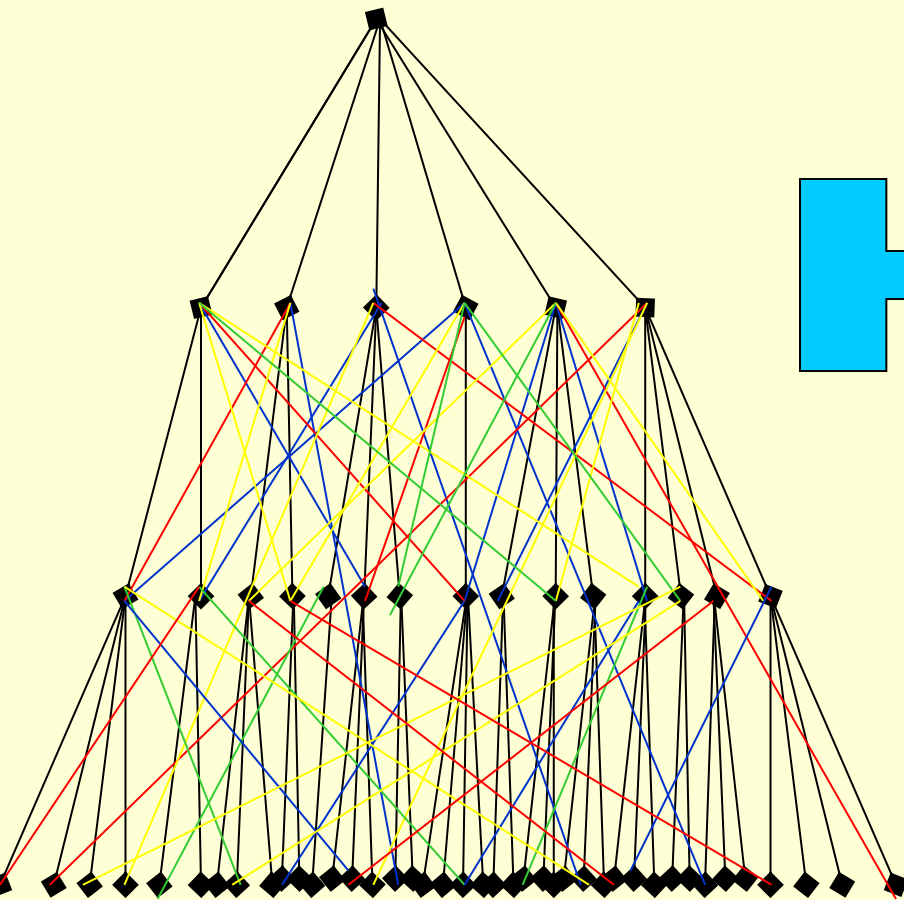
ICD Concept Title  
Fully Specified Name  
Preferred Name  
Synonyms  
Classification  
Properties  
Parents  
Type  
Use and  
Linearization(s)  
Textual Definition(s)  
Terms  
Base Index Terms  
Inclusion Terms  
Exclusions

Body System/Structure  
Manifestation  
Causal  
Etiology  
Genomics  
Agents  
Temporal Severity  
Functioning Properties  
Specific Condition  
Gender  
Life Cycle  
Treatment  
Diagnostic Criteria



# Algorithmic Serialization of the Foundation Component into a *Linearization*

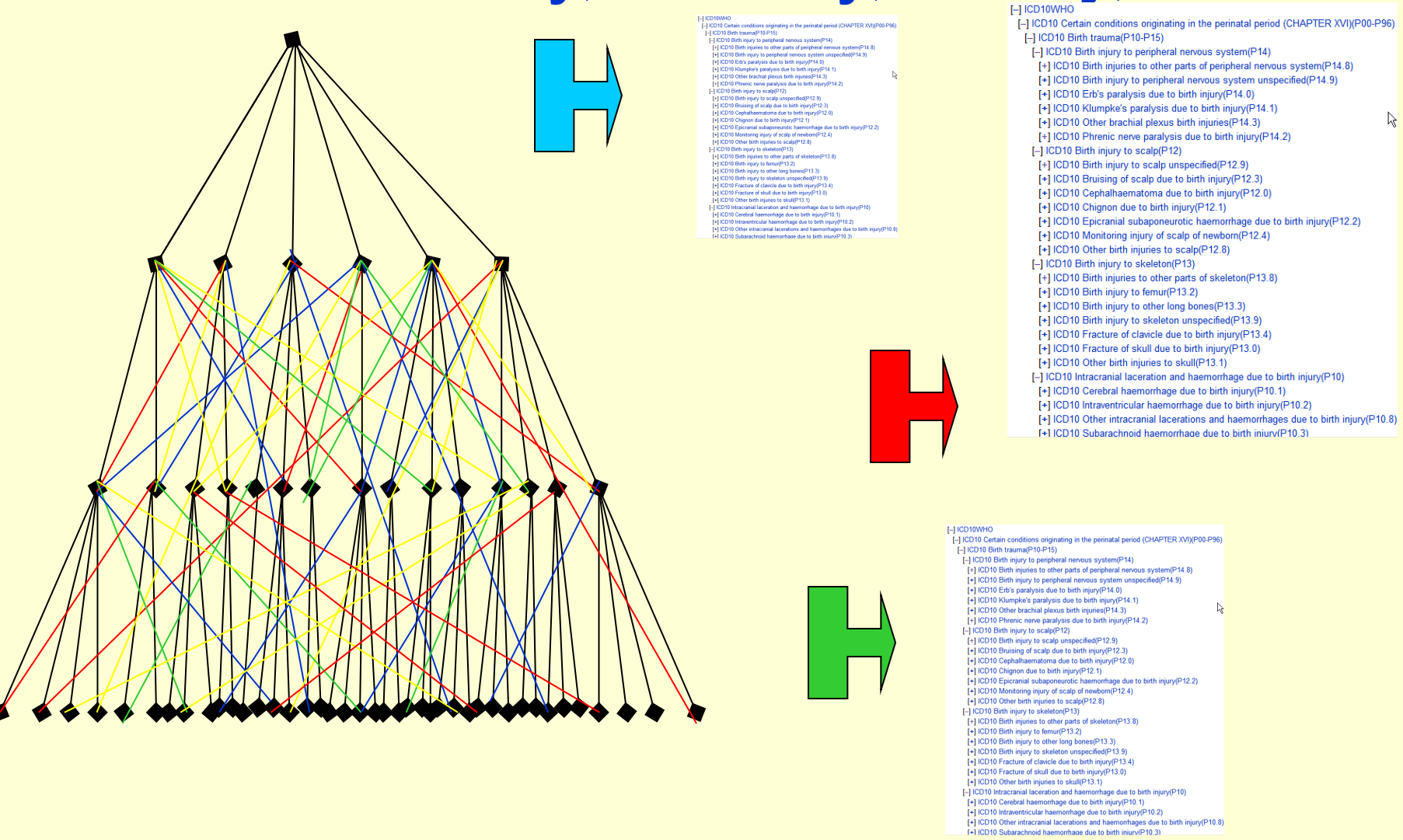
Mutually Exclusive  
And Exhaustive



- [-] ICD10WHO
- [-] ICD10 Certain conditions originating in the perinatal period (CHAPTER XVI)(P00-P96)
  - [-] ICD10 Birth trauma(P10-P15)
    - [-] ICD10 Birth injury to peripheral nervous system(P14)
      - [+] ICD10 Birth injuries to other parts of peripheral nervous system(P14.8)
      - [+] ICD10 Birth injury to peripheral nervous system unspecified(P14.9)
      - [+] ICD10 Erb's paralysis due to birth injury(P14.0)
      - [+] ICD10 Klumpke's paralysis due to birth injury(P14.1)
      - [+] ICD10 Other brachial plexus birth injuries(P14.3)
      - [+] ICD10 Phrenic nerve paralysis due to birth injury(P14.2)
    - [-] ICD10 Birth injury to scalp(P12)
      - [+] ICD10 Birth injury to scalp unspecified(P12.9)
      - [+] ICD10 Bruising of scalp due to birth injury(P12.3)
      - [+] ICD10 Cephalhaematoma due to birth injury(P12.0)
      - [+] ICD10 Chignon due to birth injury(P12.1)
      - [+] ICD10 Epicranial subaponeurotic haemorrhage due to birth injury(P12.2)
      - [+] ICD10 Monitoring injury of scalp of newborn(P12.4)
      - [+] ICD10 Other birth injuries to scalp(P12.8)
    - [-] ICD10 Birth injury to skeleton(P13)
      - [+] ICD10 Birth injuries to other parts of skeleton(P13.8)
      - [+] ICD10 Birth injury to femur(P13.2)
      - [+] ICD10 Birth injury to other long bones(P13.3)
      - [+] ICD10 Birth injury to skeleton unspecified(P13.9)
      - [+] ICD10 Fracture of clavicle due to birth injury(P13.4)
      - [+] ICD10 Fracture of skull due to birth injury(P13.0)
      - [+] ICD10 Other birth injuries to skull(P13.1)
    - [-] ICD10 Intracranial laceration and haemorrhage due to birth injury(P10)
      - [+] ICD10 Cerebral haemorrhage due to birth injury(P10.1)
      - [+] ICD10 Intraventricular haemorrhage due to birth injury(P10.2)
      - [+] ICD10 Other intracranial lacerations and haemorrhages due to birth injury(P10.8)
      - [+] ICD10 Subarachnoid haemorrhage due to birth injury(P10.3)

# Linearizations for multiple use-cases

## Morbidity, Mortality, Quality, ...



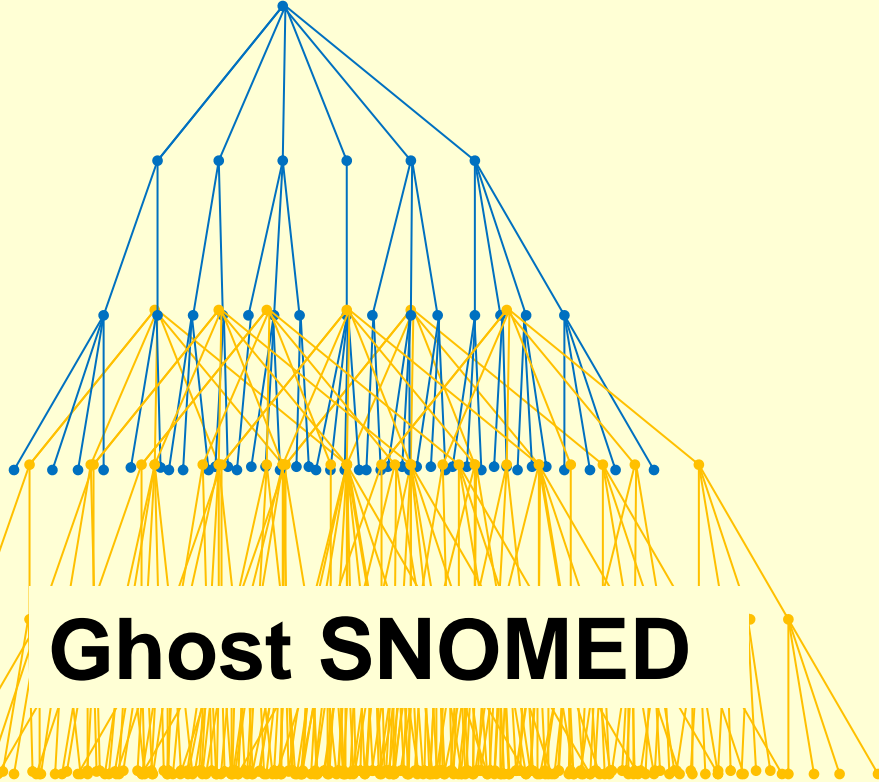


# Relationship with IHTSDO SNOMED content

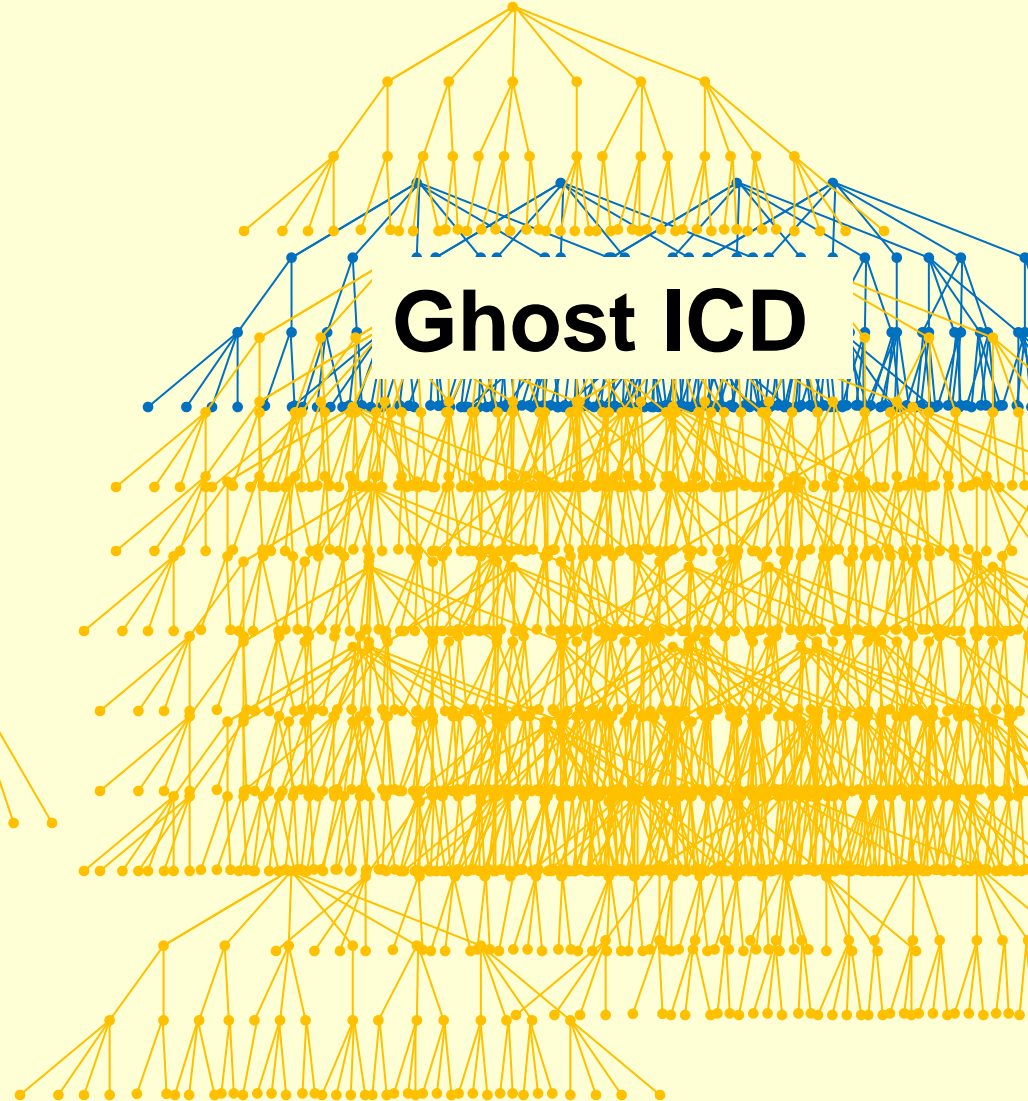
- IHT (SNOMED) will require high-level nodes that aggregate more granular data
  - Use-cases include mutually exclusive, exhaustive,...
  - Sounds a lot like ICD
- ICD-11 will require lower level terminology for value sets which populate content model
  - Detailed terminological underpinning
  - Sounds a lot like SNOMED
- Memorandum of Agreement – July 2010!
  - WHO right to use for authoring and interpretation

# Potential Future States (2007)

## ICD-11



## SNOMED



# Joint ICD-IHTSDO

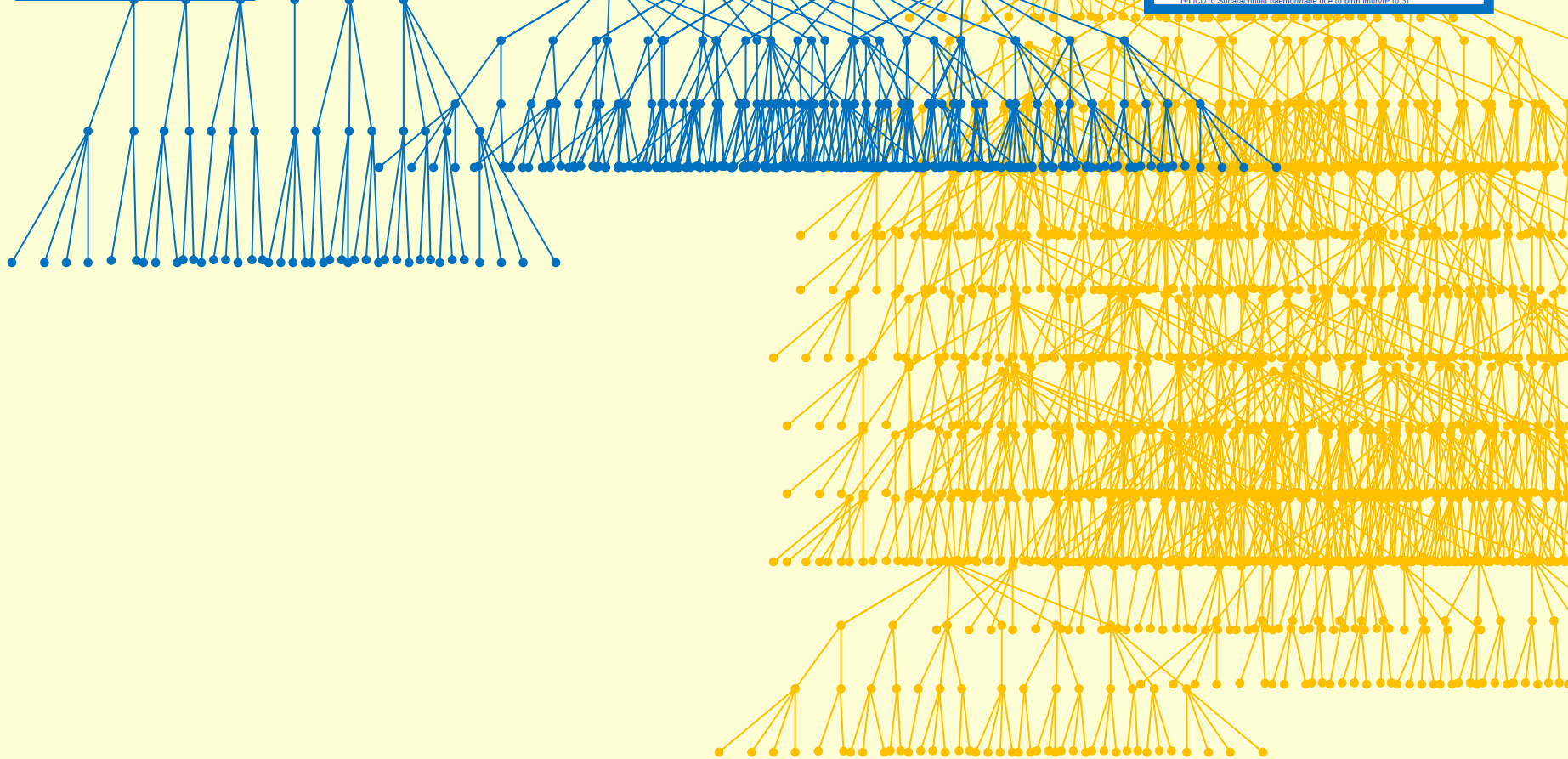
# Effort

# SNO

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-11

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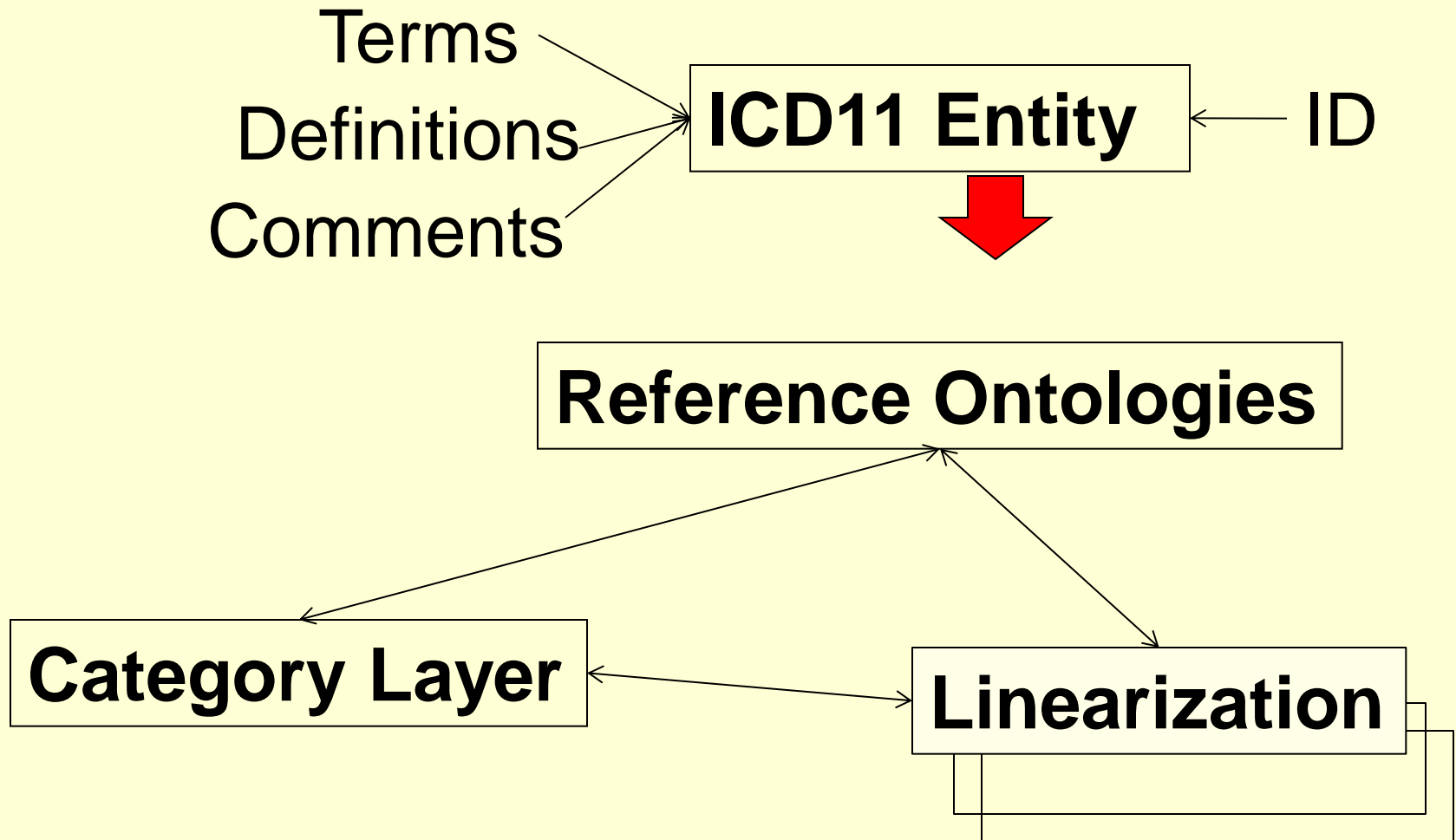


# ICD11/SNOMED “Shared Layer” vs Common Ontology

- Classification  $\leftrightarrow$  Terminology dissonance
  - Focus on higher levels of abstraction
- Thesaurus  $\leftrightarrow$  Description Logic dissonance
  - Pragmatic hierarchies – parent-child
  - Formal logic where all *is-a* are always true
- Common Ontology is:
  - Based on Description Logics and “queries”
  - Provides a shared scaffolding for
    - The Foundation Layer of ICD 11 and SNOMED
    - Ignores residual categories of linearizations



# High Level Structure – Core Model



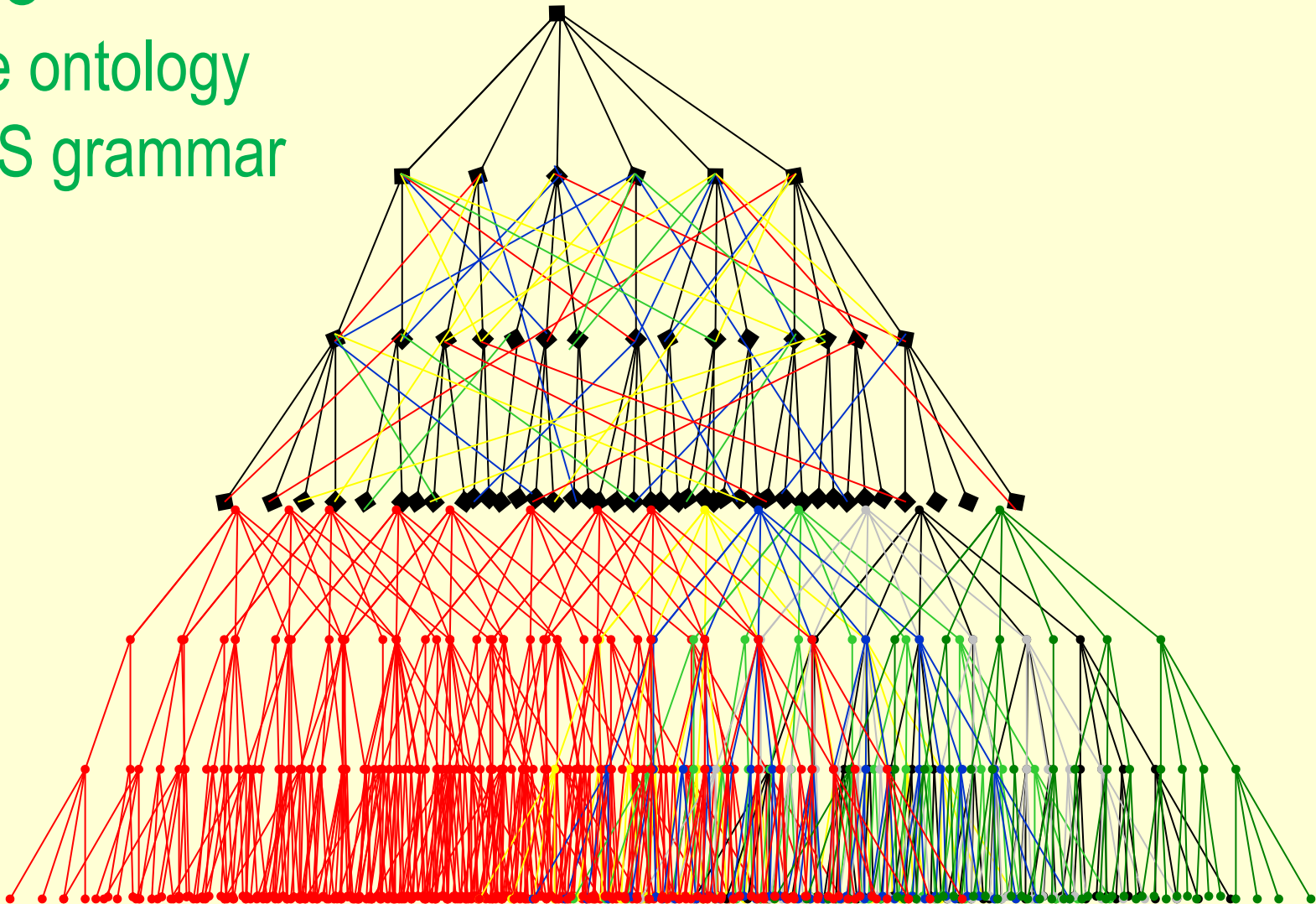
# Permissive Partnerships for Ontologic Extensions

HUGO

Gene ontology

HGVS grammar

.....



# ICD 11 Architecture

SNOMED  
CT

Common Ontology  
(definitions)

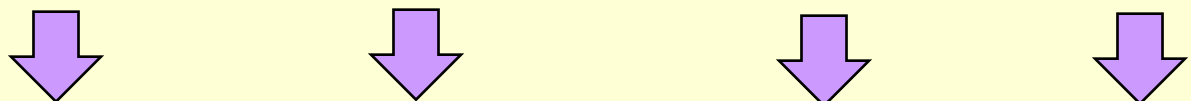
Foundation Layer

Contingent knowledge:

signs, symptoms  
causes, ...,  
linkage entities)

Common Ontology

a subset of SNOMED CT classes and axioms



Mortality

Morbidity

Primary Care

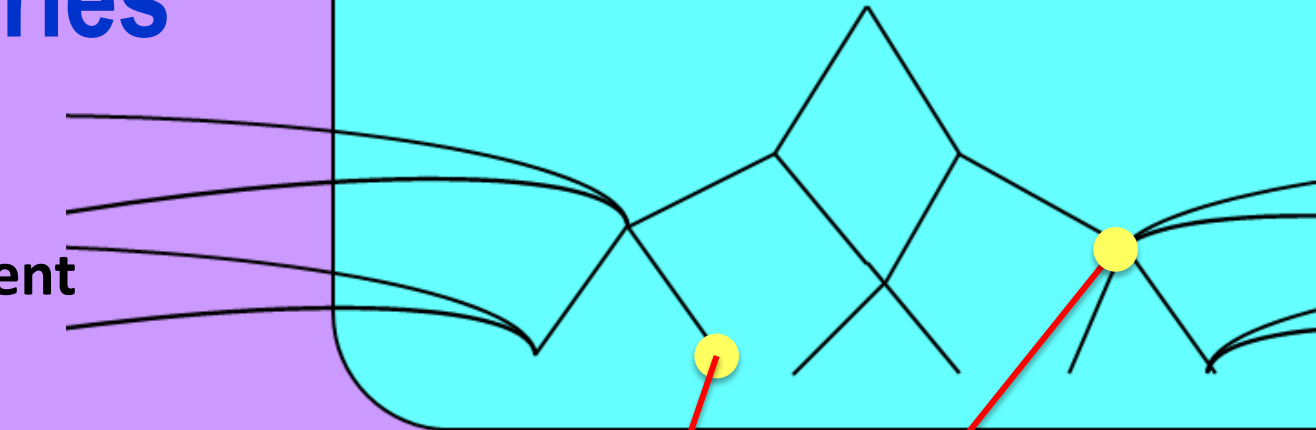
...

# ICD 11 Linkage Queries

Links between  
Foundation Component  
and Linearizations

## Common Ontology

a subset of SNOMED CT



All linearization  
entities are represented as  
queries against  
Common Ontology

### Morbidity Linearization

Residual Categories  
**NEC NOS**

#### Morbidity

“Hypertension  
excluding Pregnancy”

```
SELECT ?CN WHERE  
  (?CN SubclassOf Hypertension)  
  MINUS  
  (?CN SubclassOf  
    Disorders of Pregnancy)
```



# Tough Love for SNOMED

- Clarify a “reference terminology”
  - Underpinning basis for all derivatives
  - Logical target for NLP, speech parsing
- Embrace genomic linkages (extramural?)
- Create “Linearizations” (not RefSets)
- Collaborate on Common Information Model (CIMI)
- Accommodate simple “value sets”
  - Simple enumerated lists, linked to reference
- Publish “usable” formats and REST services
  - Common Terminology Services – CTS2
  - Keep RFX for developers and researchers

## Conclusion

- Terminology Is the Second Most Urgent Issue in Healthcare Information Today
- Problem Underlies Virtually All Machine-assisted Uses of Patient Data – 1° and 2°
- Convergence Toward Collaborating Systems is Critical and Occurring
- SNOMED's Future is Bright
  - Though strategic organization is required