

Terminologies and Ontologies in Healthcare: a snapshot for the early 21st century

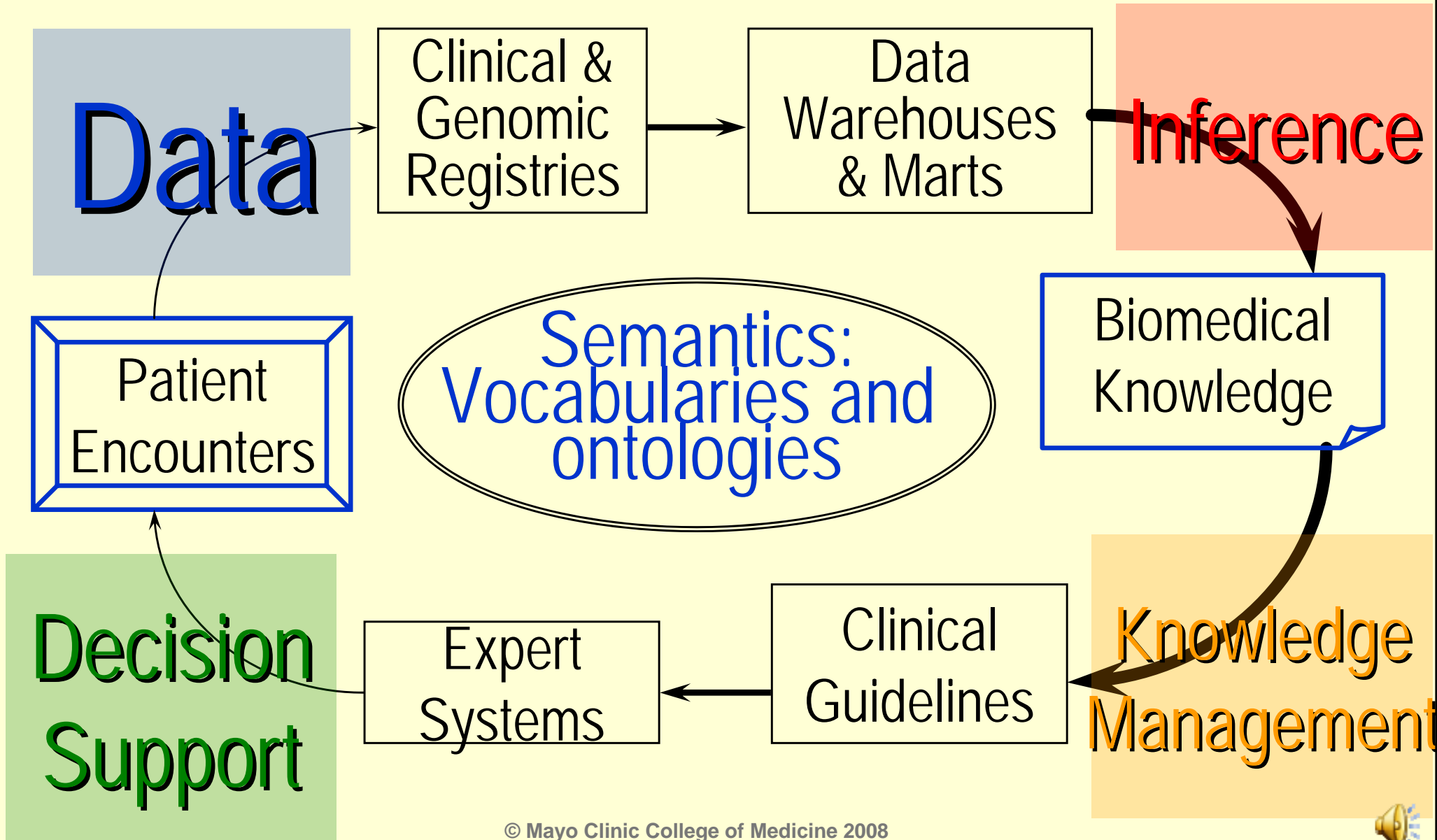
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Mayo Clinic College of Medicine

Congreso Regional de Informacin en
Ciencias de la Salud

Rio de Janeiro, 19 September 2008



From Practice-based Evidence to Evidence-based Practice



Value Proposition

“Those with more detailed, reliable and comparable data for cost and outcome studies, identification of best practices, guidelines development, and management will be more successful in the marketplace.”

SP Cohn; Kaiser Permanente



Disease Understanding Constrained by Knowledge

- Carolus Linnaeus
Carl von Linné
 - *Genera Morborum* (1763)
- Underscored Content Difficulty
 - Pathophysiology vs. Manifestation
e.g., Rabies as psychiatric disease



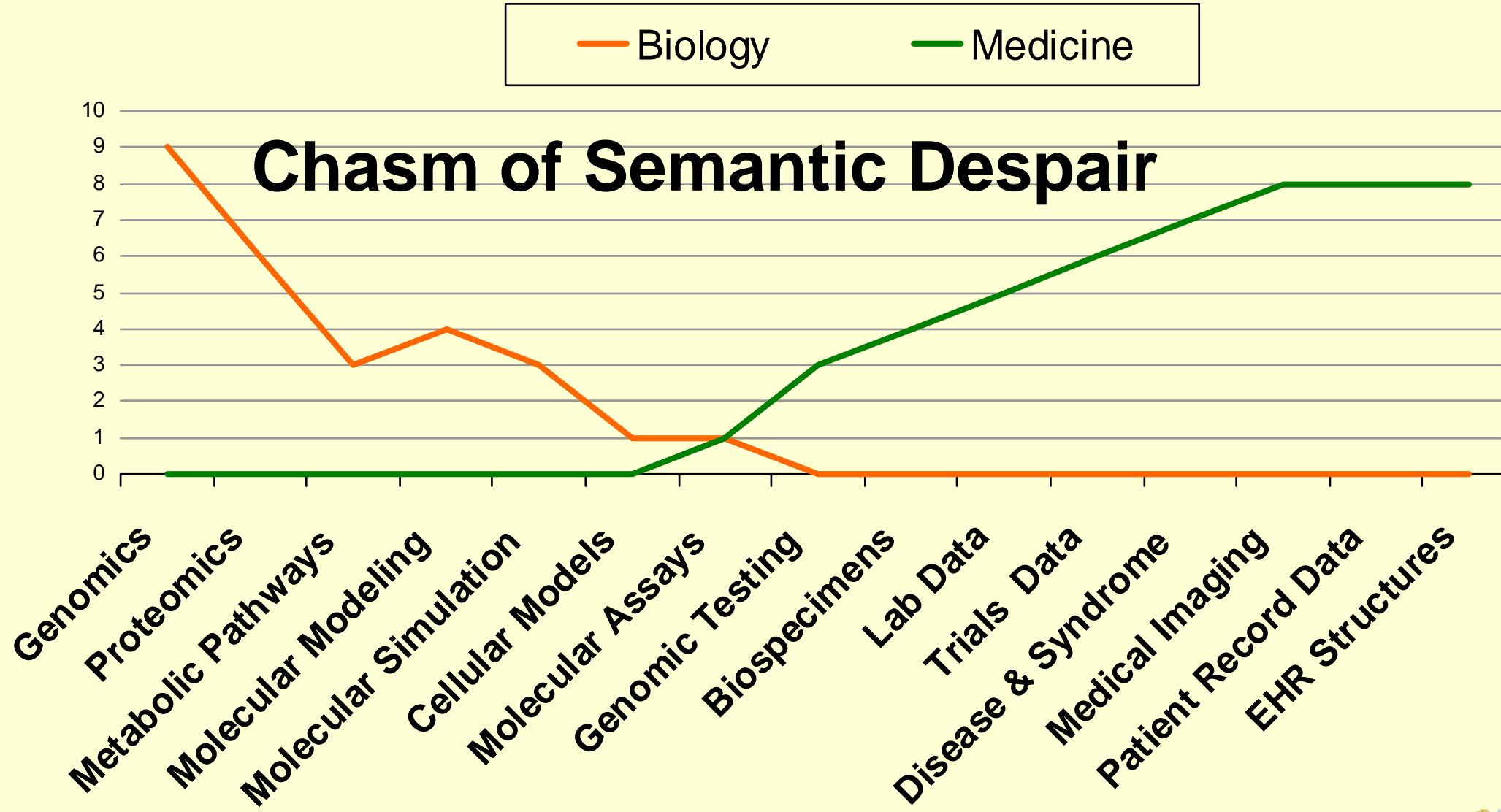
The Genomic Era

- 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



The Continuum Of Health Classification

Biology meets Clinical Medicine



The Impact of Digital Science: An Enabling Aspect of Big Science?

- Physics and Astronomy focus on datasets
- Biology and Medicine are aggregating larger datasets and electronic knowledge resources
- Mechanisms for analyzing huge (terabyte) level datasets are now commonplace
- Distributed computing and Grids:
 - Networked data – data Grids
 - Networked computational power – computing Grids



Issues and Challenges: System Design and Common Models

- If biomedicine is becoming “big” and digital...
- Methods, data, and information workflow must scale across the enterprise
- Ad hoc solutions must fit into larger informatics architecture
- Data and results must be “interoperable”
- If Translational Research is to succeed, we must bridge data and knowledge from research to practice



Blois, 1988

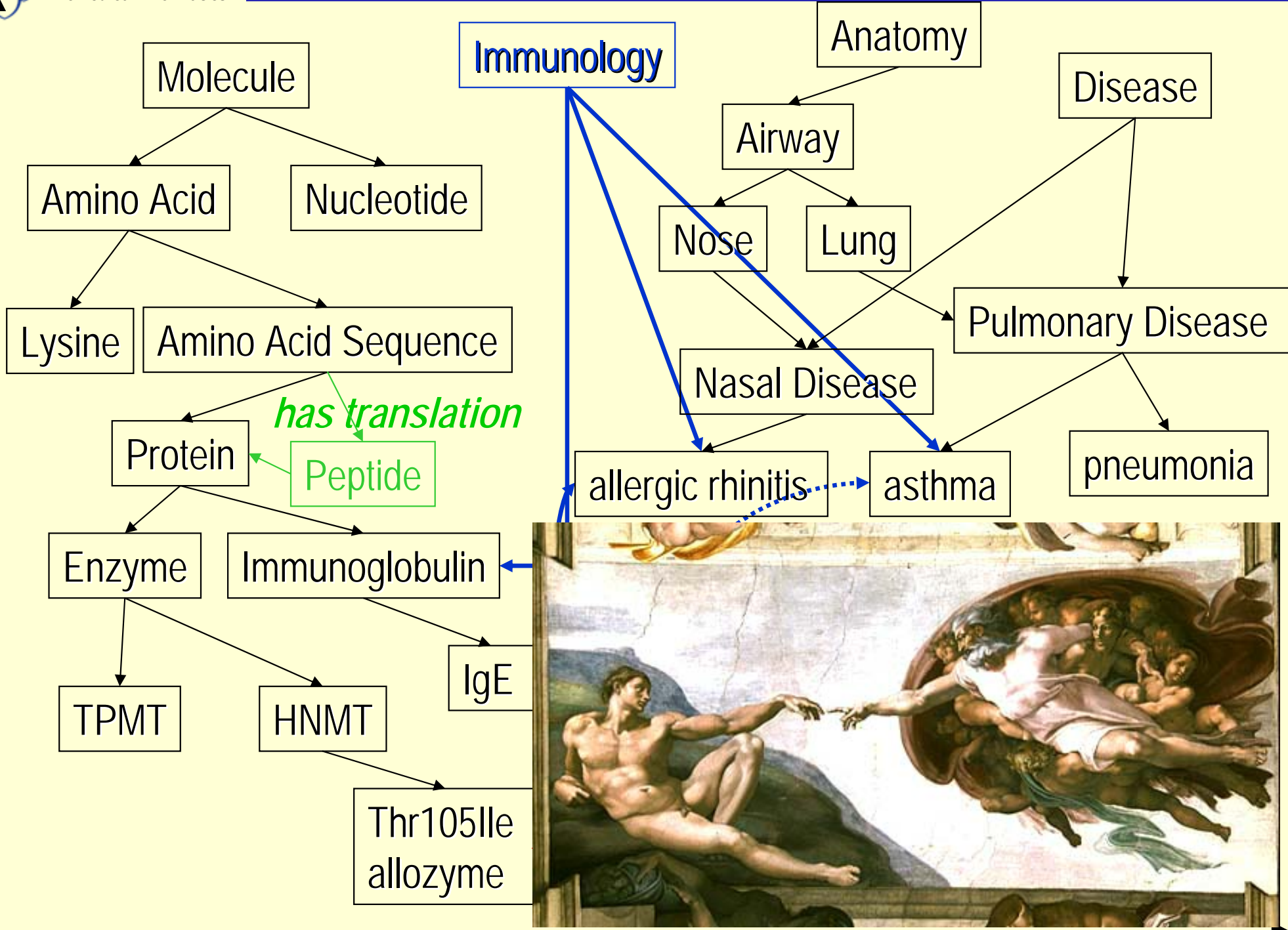
Medicine and the nature of vertical reasoning

- 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...



Highly Aggregated

Fine Detail



Molecular

Clinical



Whither Phenotype?

Spans spectrum from enzymes to disease

- Pharmacogenomics – enzyme functionality
- Physiologist – cellular function
- Systems biologist – pathway circuit flow
- Sub-specialist – organ functioning
- Patient/Clinician – disease manifestation
- Public Health – population characteristics

Highly specific to use-case context



Cancer Phenotype

- Increasingly dependent on genomic characteristics
 - Blend with pathology, imaging, laboratory
 - Extent of disease measures are crucial
 - Comparable and consistent data ultimately depends upon common ontologies
- | | | |
|-------------|---------------------|----------------|
| • BiomedGT | • HUGO | • Drugs Rx |
| • SNOMED CT | • GO | • Radiation RT |
| • ICD-O | • Adverse
Events | • Surgery Px |





A Synthesis of Modern Terminologies The WHO ICD-11 Project



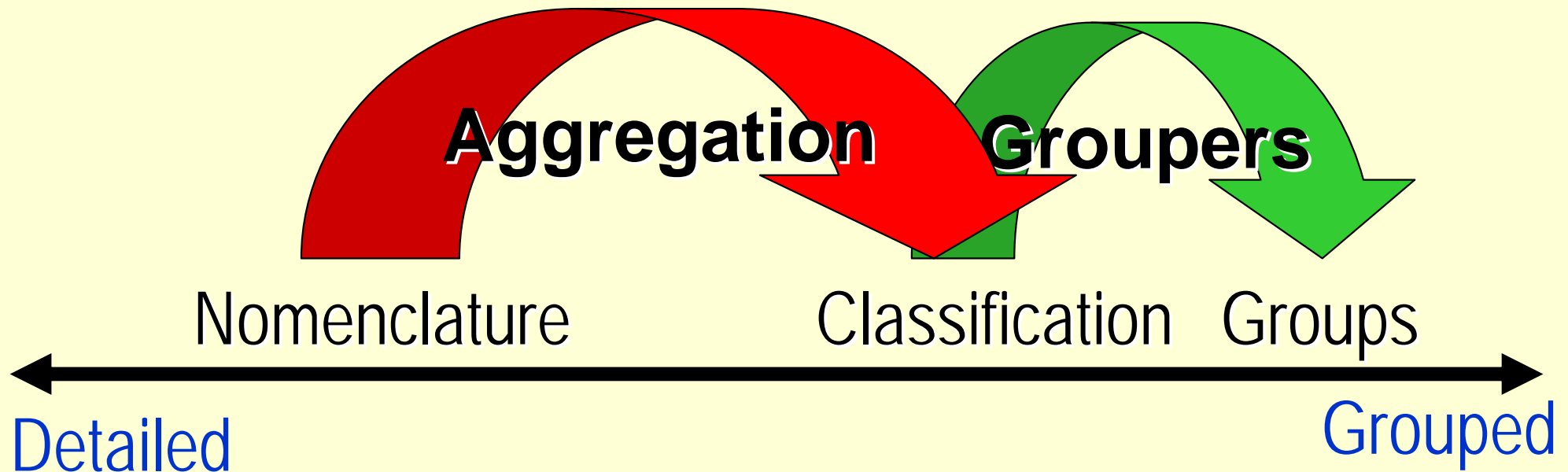
Some Premises for ICD-11 development

- Rubrics defined by Aggregation Logics from terminologies
- Human language definitions will be explicit
- “core” representation will be in description logic based ontology
- A linear serialization will be derived as a view to maintain longitudinal consistency
 - May require corresponding “rules” for practical use



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)

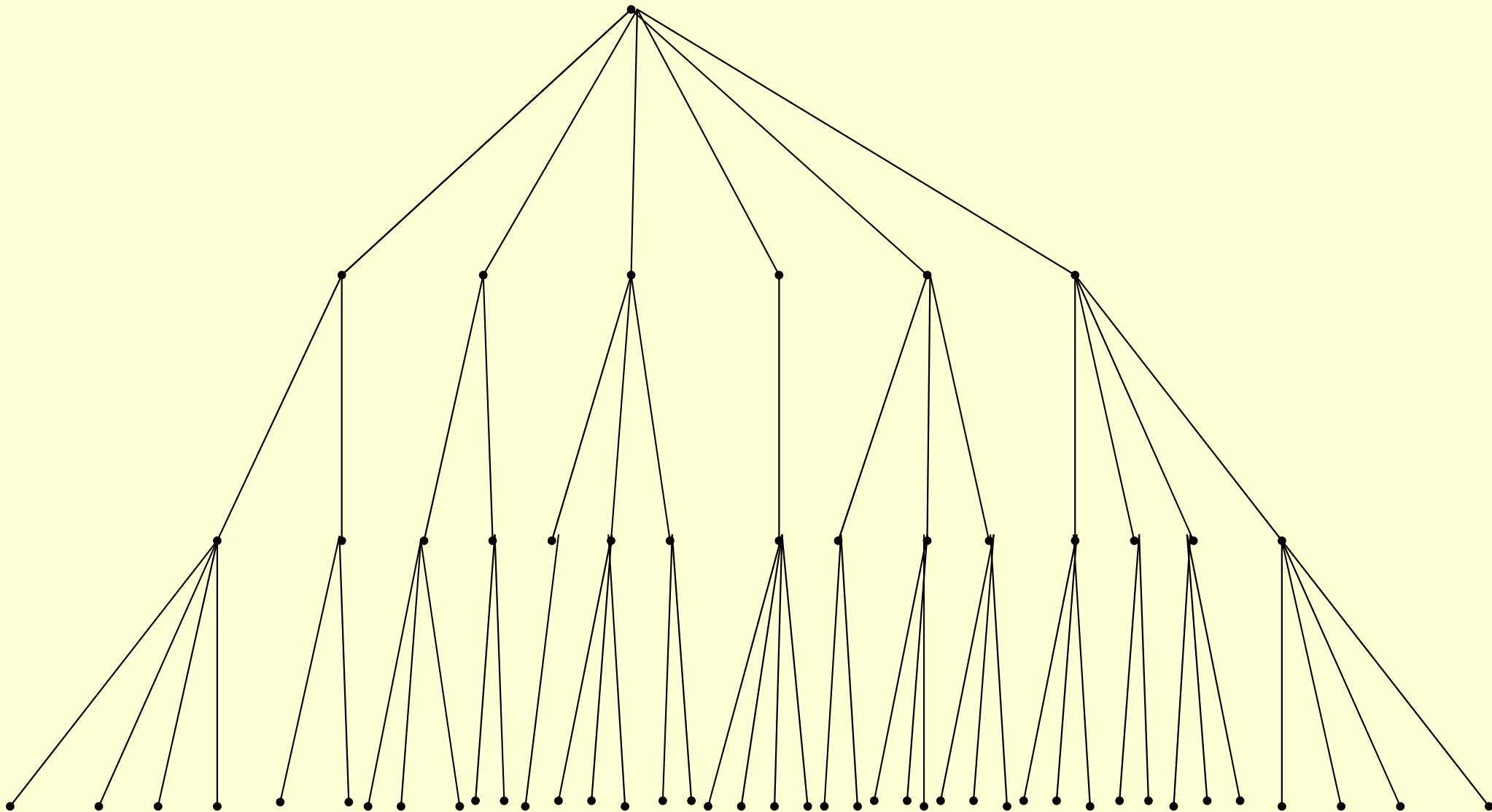


ICD11 Use Cases

- 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



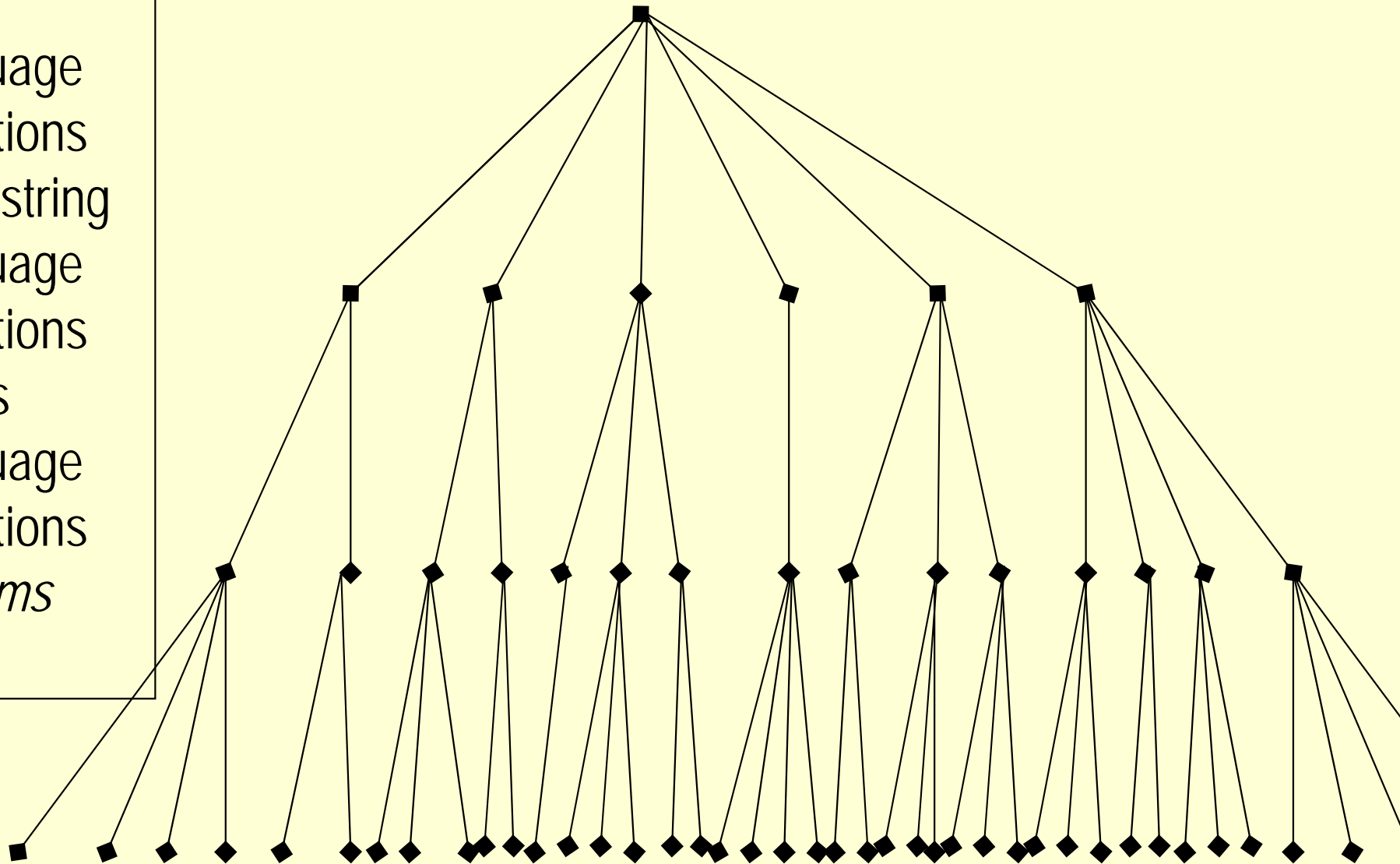
Traditional Hierarchical System ICD-10 and family



Addition of structured attributes to concepts

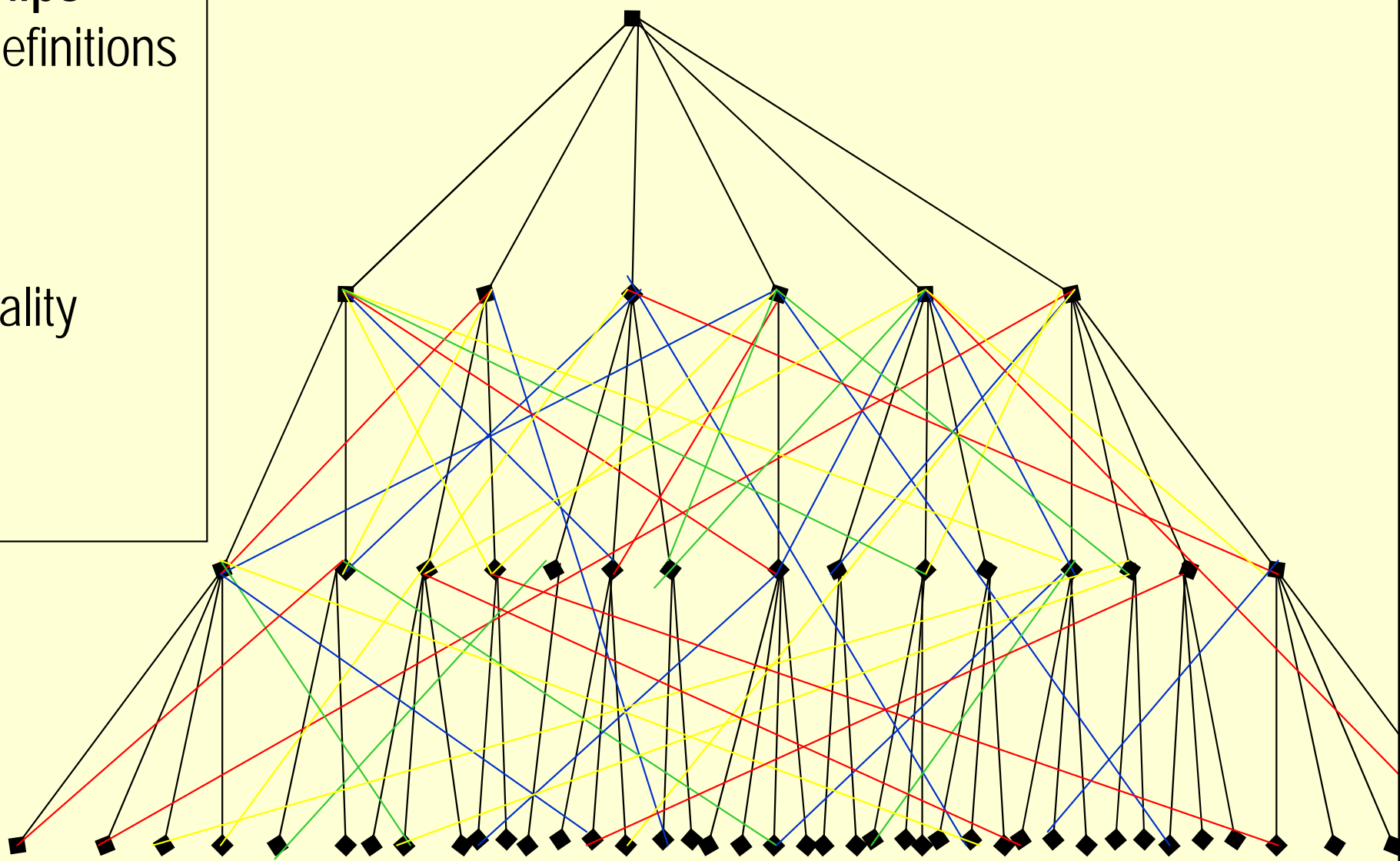
Concept name

- Definition
 - Language translations
- Preferred string
 - Language translations
- Synonyms
 - Language translations
- *Index Terms*

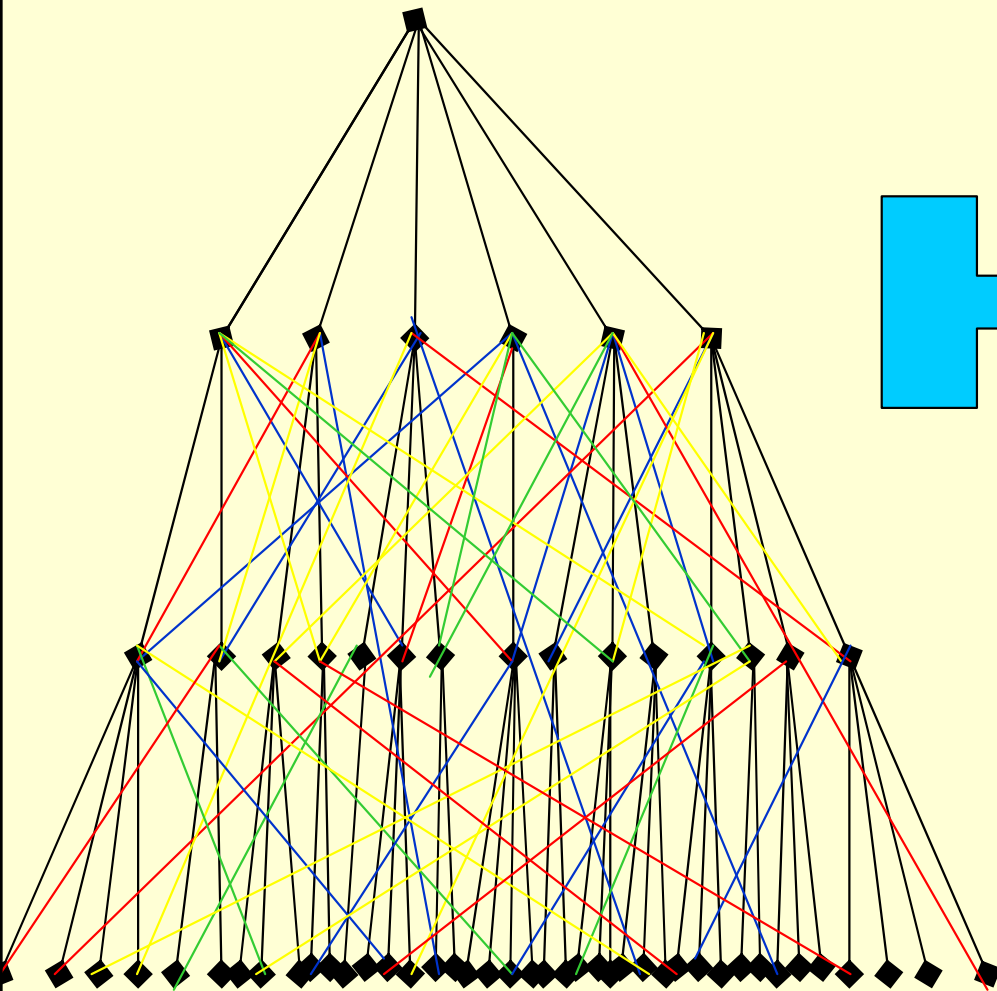


Addition of semantic arcs - Ontology

- Relationships**
- Logical Definitions
 - Etiology
 - Genomic
 - Location
 - Laterality
 - Histology
 - Severity
 - Acuity



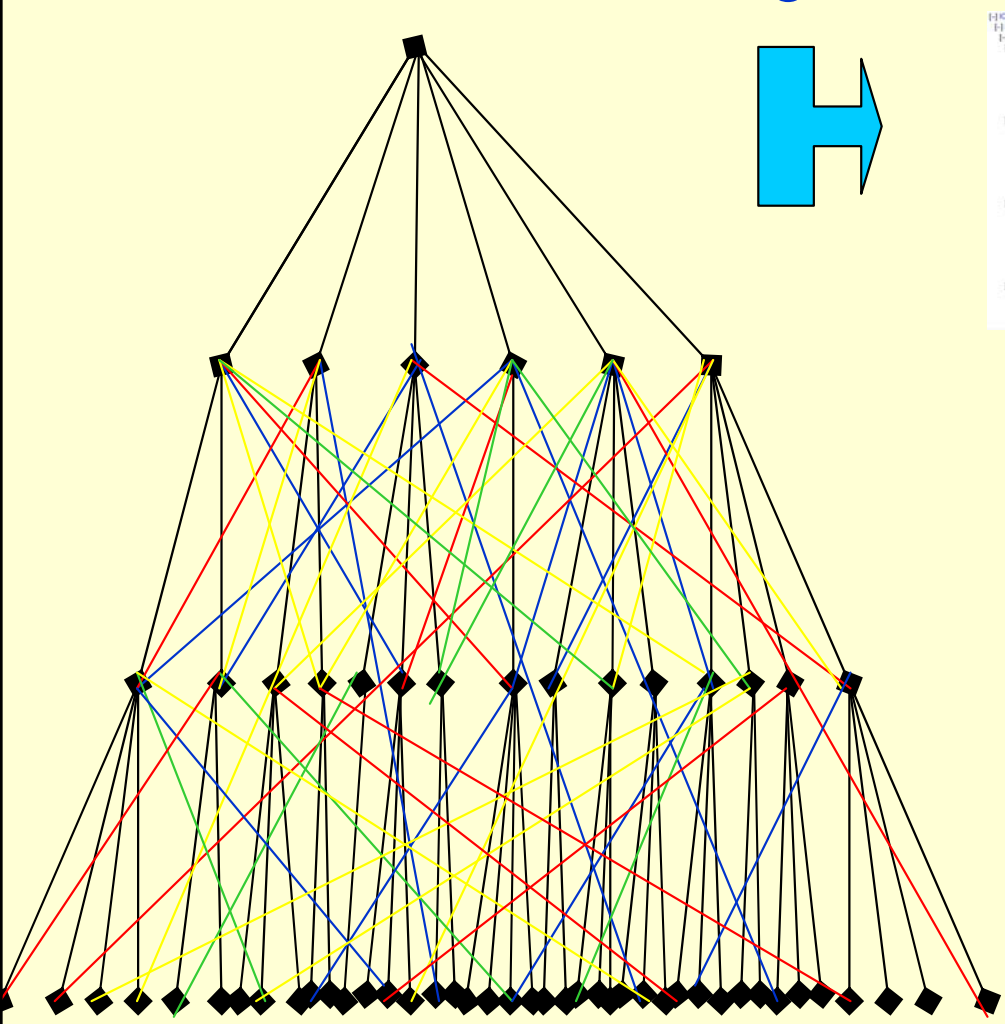
Serialization of "the cloud" Algorithmic Derivation



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Linear views may serve multiple use-cases Morbidity, Mortality, Quality, ...



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LexWiki

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Leave a comment ...

ICD10+ Collaborative Revision Platform

This is a ICD10+ collaborative content development platform based on Semantic MediaWiki.

See [Quick Start Guide - A walk through the wiki and workflow process](#)

ICD10 Classifications

- [ICD10WHO](#) - The Second Edition of the International Statistical Classification of Diseases and Related Health Problems, Tenth Revision (ICD-10).
- [ICD10CM](#) - The 2007 release of International Classification of Diseases, Tenth Revision, Clinical Modification (ICD-10-CM).
- [ICD10AM](#) - The Fifth Edition of International Statistical Classification of Diseases and Related Health Problems, Tenth Revision, Australian Modification (ICD-10-AM).

LexWiki SandBox

Users may play here to get familiar with ICD10 content representation and proposal creation. See [Help Page](#).

[+] [SandBox ICD10 Other leukaemias of specified cell type\(C94\)](#)

User Scenarios and Usability

- [User Scenarios](#) - a page to describe the potential user roles and general user scenarios.
- [ICD Collaborative Platform Integration](#) - a collaborative effort for integration of the LexWiki with the [WHO ICD_Revision Platform](#) .
- [LexWiki Feature Request](#) - a page to add your feature request.

[testGraph](#)

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toolbox

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category

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Category:Orphanet Lesch Nyhan syndrome(PatId197)

Subcategories

[\[show\]](#)

Proposals

[\[show\]](#)

Lexical

Properties

Associations

Factbox

Lexical

Concept Code: PatId197

Preferred Name: Lesch-Nyhan syndrome

Coding Scheme: urn:dns:bmi.mayo.edu:orphanet

Definition: Deficiency of hypoxanthine-guanine phosphoribosyltransferase (HPRT) activity is an inborn error of purine metabolism associated with uric acid overproduction and a continuum spectrum of neurological manifestations depending on the degree of the enzymatic deficiency. The prevalence is estimated at 1/380,000 live births in Canada, and 1/235,000 live births in Spain. Uric acid overproduction is present in all HPRT-deficient patients and is associated with lithiasis and gout. Neurological manifestations include self-harm, action dystonia, choreoathetosis, ballismus, cognitive and attention deficit, and self-injurious behaviour. The most severe forms are known as Lesch-Nyhan syndrome (patients are normal at birth and diagnosis can be accomplished when psychomotor delay becomes apparent). Partial HPRT-deficient patients present these symptoms with a different intensity, and in the least severe forms symptoms may be unapparent. Megaloblastic anaemia is also associated with the disease. Inheritance of HPRT deficiency is X-linked recessive, thus males are generally affected and heterozygous female are carriers (usually asymptomatic). Human HPRT is encoded by a single structural gene on the long arm of the X chromosome at Xq26. To date, more than 300 disease-associated mutations in the *HPRT1* gene have been identified. The diagnosis is based on clinical and biochemical findings (hyperuricemia and hyperuricosuria associated with psychomotor delay).

Association Graph

Category:Orphanet_Lesch_Nyhan_syndrome(PatId197)

HAS_CLINICAL_SIGN(Very frequent)

Category:Orphanet_Mental_retardation

HAS_CLINICAL_SIGN(Frequent)

Category:Orphanet_Mental_retardation

PAR

Category:Orphanet_Anemia_megaloblastic

HAS_CLINICAL_SIGN(Very frequent)

Category:Orphanet_Mental_retardation

HAS_CLINICAL_SIGN(Very frequent)

Category:Orphanet_Behavioral_disorders

PAR

Category:Orphanet_Dystonia_associated_with_mental_disorders

PAR

Category:Orphanet_Genetic_disorders

HAS_CLINICAL_SIGN(Frequent)

Category:Orphanet_Mental_retardation

HAS_CLINICAL_SIGN(Very frequent)

Category:Orphanet_Hypertension

HAS_CLINICAL_SIGN(Very frequent)

Category:Orphanet_Mental_retardation

PAR

Category:Orphanet_Intellectual_disorders



Discussions with IHTSDO

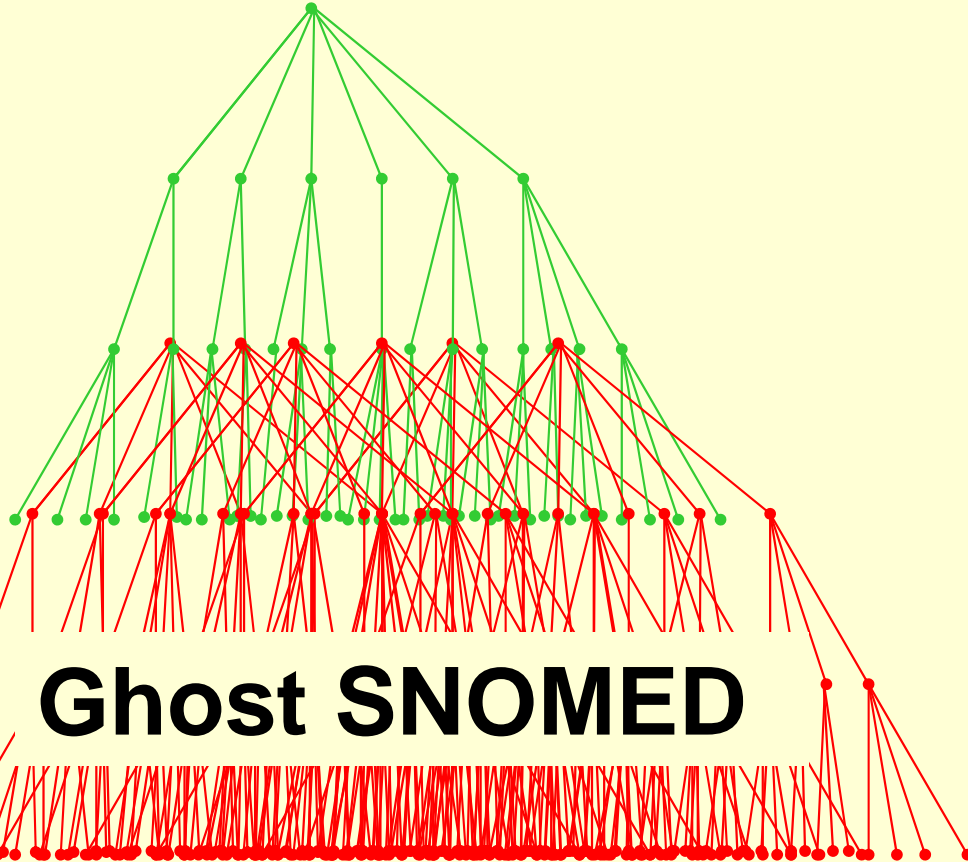
International Health Terminology (IHT)

- 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 aggregation logic definitions
 - Detailed terminological underpinning
 - Sounds a lot like SNOMED



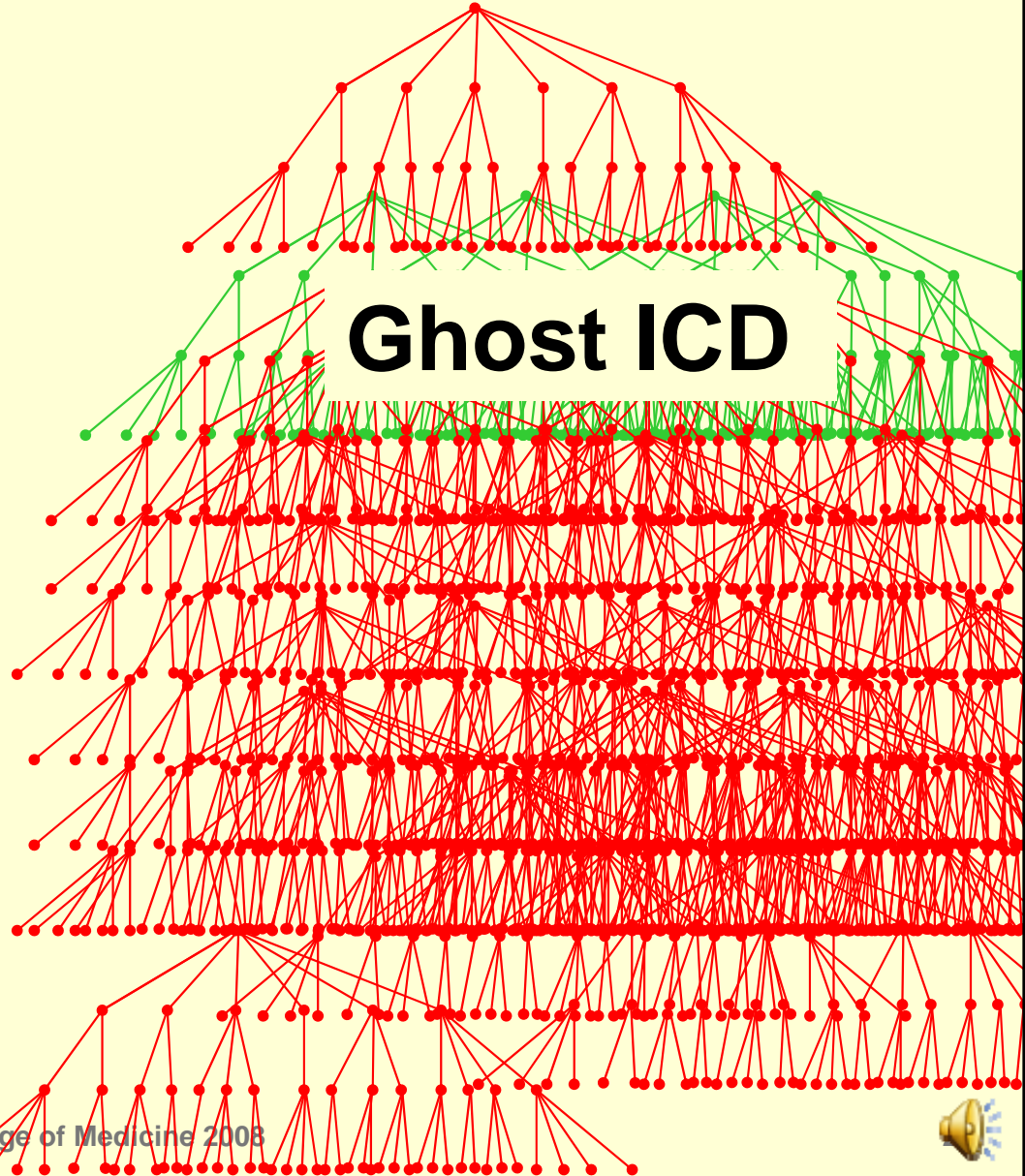
Potential Future States

ICD-11



Ghost SNOMED

SNOMED



Ghost ICD



Joint Alternate Future ICD-IHTSDO

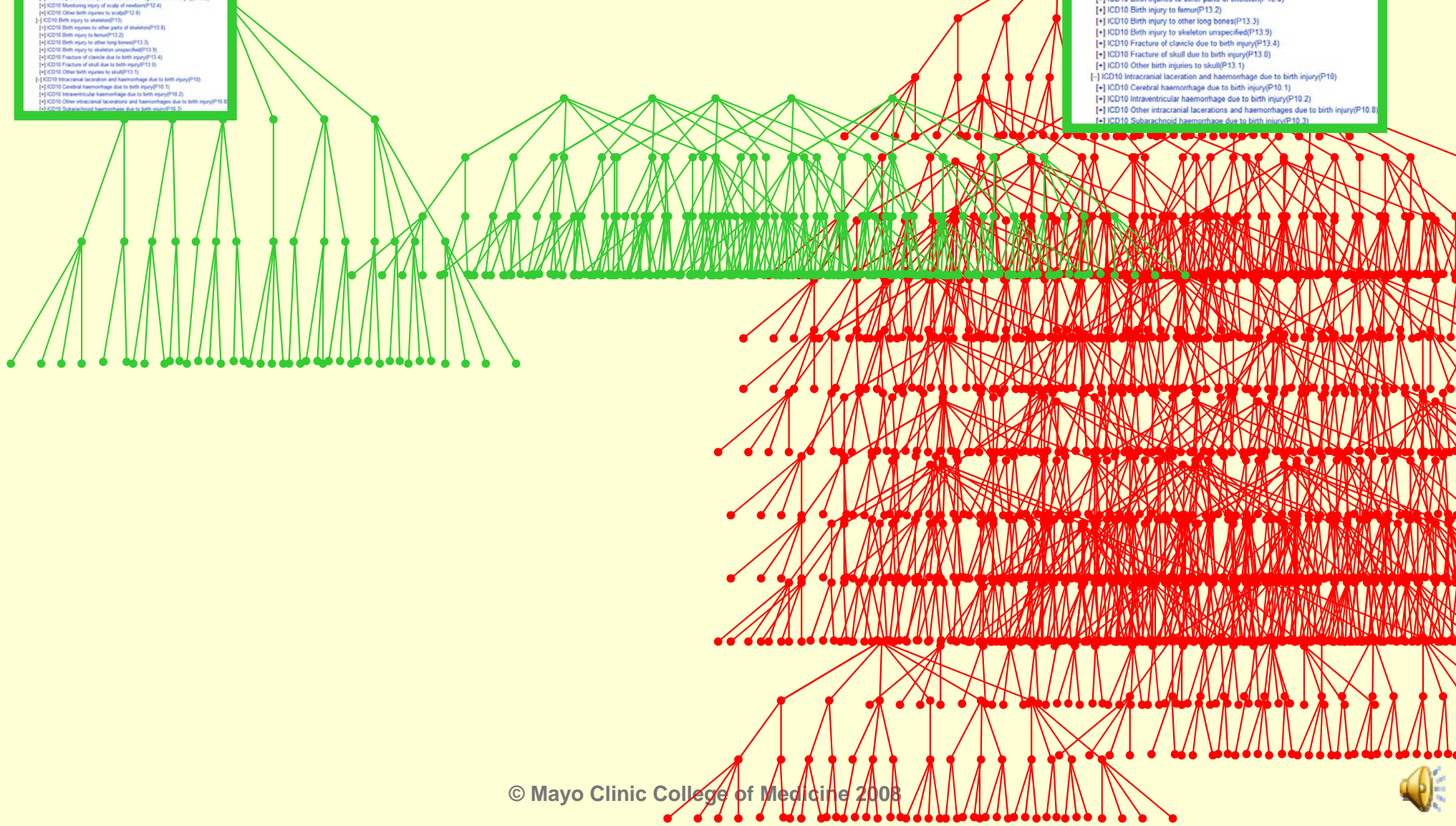
Effort

SNO

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Advantages to Collaboration

- Both organizations avoid “Ghost” emulations
- Both organizations leverage expertise and content
 - More resources brought to the table
- Both organizations retain independent intellectual property and derivatives (e.g. Linear formats of ICD-11)
- Mappings become moot
 - Aggregation of SNOMED is definitional to ICD



Caveat

ICD and IHTSDO

- No agreements have been finalized
- Intellectual property sharing is expected
- Shared tooling is being discussed
- Harmonization Board has been proposed



Conclusion

- Biomedicine concepts have become complex and intertwined
- Research synergies will depend upon established interoperability standards
- Standards are exploring “inter-standard” interoperability
- Vocabularies and ontologies will continue to increase in importance for intelligent health care

