Records of Health Standards of

Revolutionizing healthcare through independent lifetime health records

Towards a universal health information language

Paradigm shifts in health informatics needed for leveraging FAIR data environments*

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Based on a keynote speech at MedInfo 2015 under the title:
"*Translational & Interoperable Health Infostructure - The Servant of Three Masters*"

Agenda

INFORMATICS

ANALYICS

POLICY

Glad to be with you -

- Second time in NETTAB
- Second time in Genova
- And now: my FAIR Genova… ☺

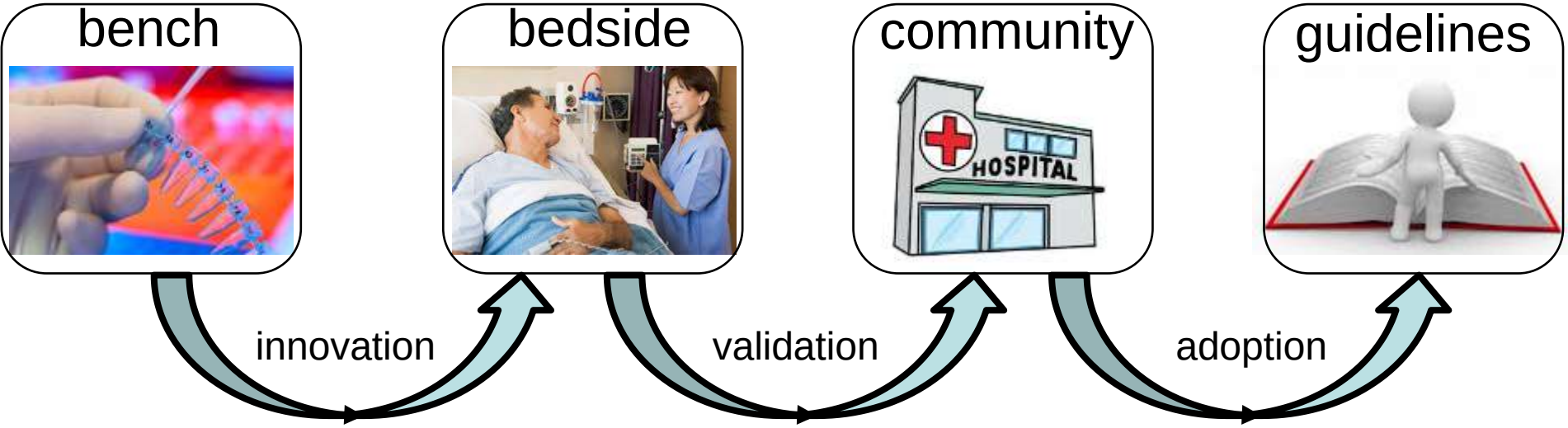
FAIR… and then what?

- **FAIR is important!**
  - **Findable…**
  - **Accessible…**
  - **Interoperable…**
  - **Reusable…**

  - **And then what?**

- **This talk is about how to leverage FAIR and move forward in bridging between science and healthcare**

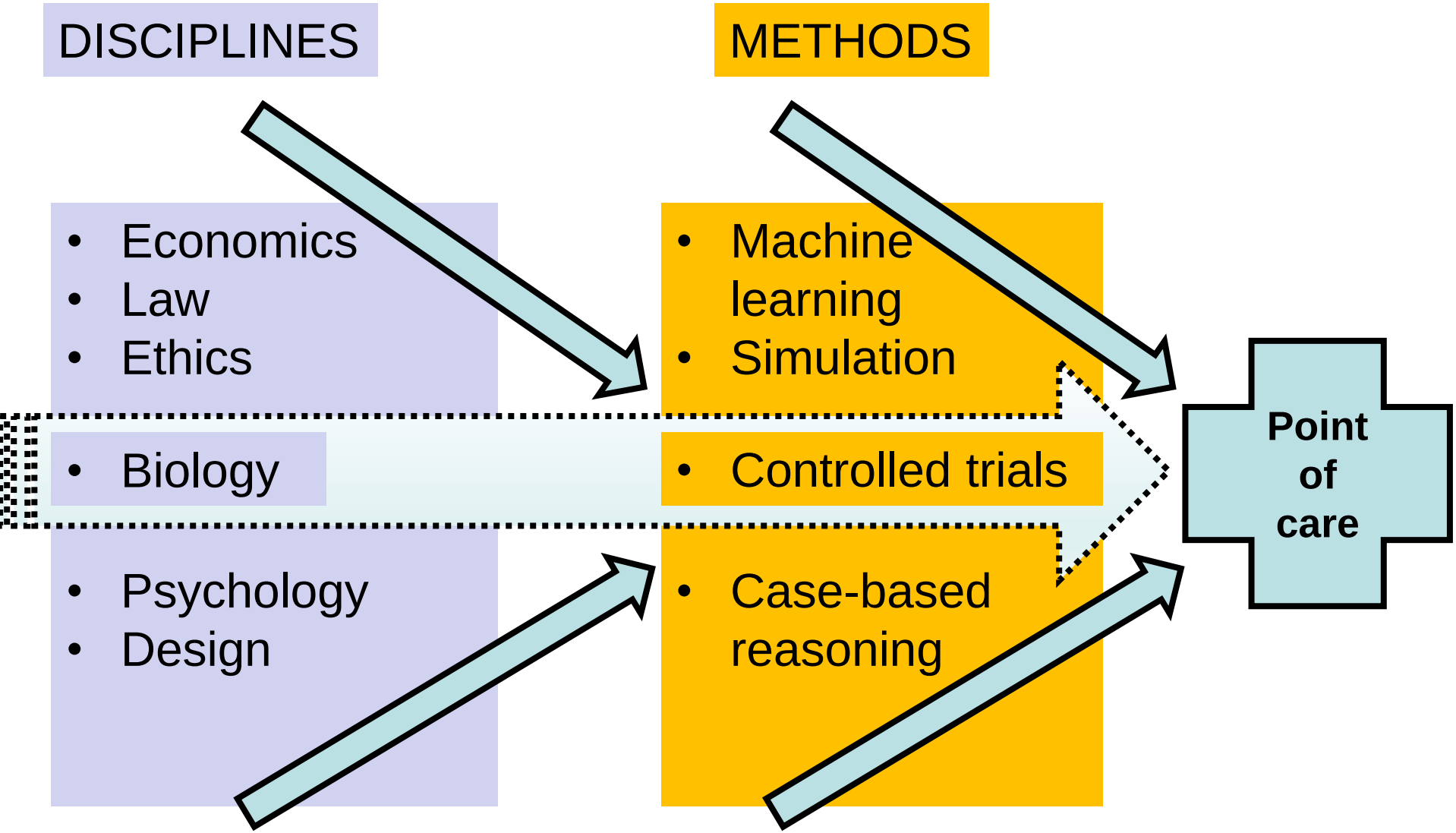
Translational Medicine – Main Barriers



- **The reality**
  - **Some successful bedside interventions do not scale out to community**
  - **Many interventions do not end up in medical societies' guidelines**

- **Possible explanation**
  - **Disciplines are limited to biology**
  - **Methods are limited to controlled trials**

Broadening & Converging in Translational Informatics

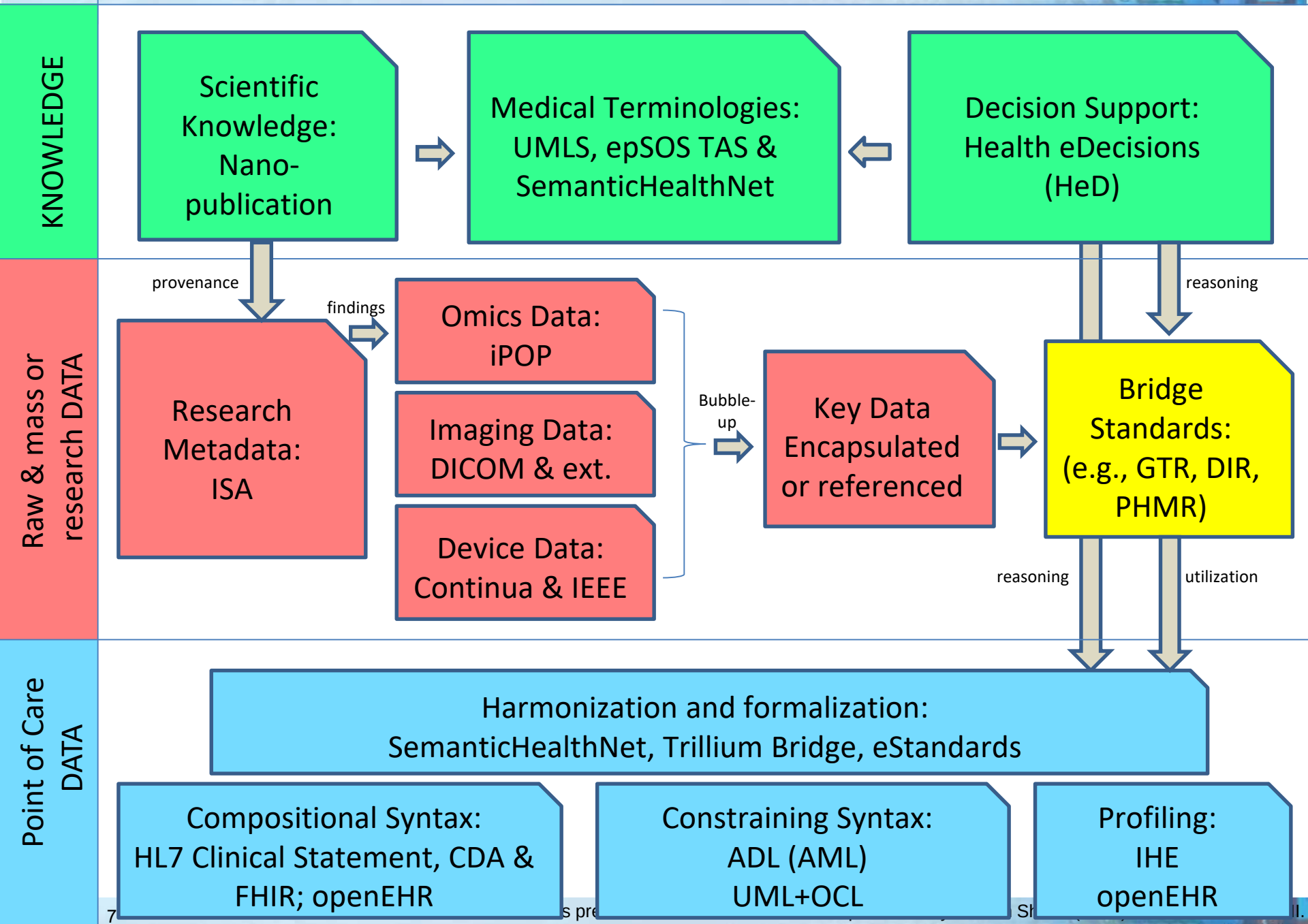


The Translational Informatics Challenge

- **Each discipline has its own informatics!**
- **Each method has its own informatics!**
- **How all of these could be converged??**
- **Even in the biomedical world,**
  informatics is constantly changing…
  so we need touch point to streamline the flow of data

What are some example formats and their possible touch-points?

Biomedical Information Formats Landscape and Touch Points

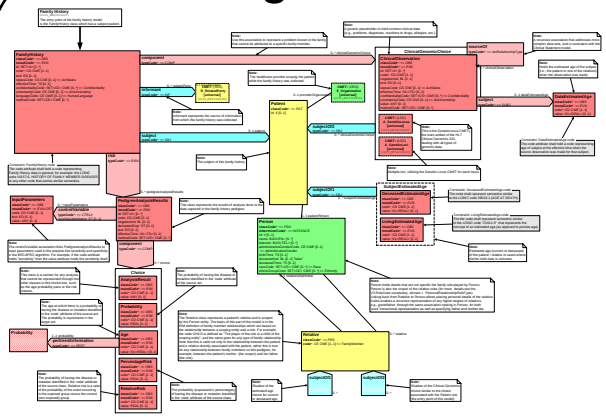


Despite Touch Points, Current Representations have Issues…

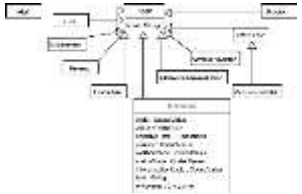
- **Often, current formats /schemas / standards are ‘silos’ because they are:**
  - oriented towards specific sub-domains or scoped-down usages
  - limited in their expressivity with rigid structures
  - inconsistent with each other, regarding core semantics
  - have overlaps in scope

- **For example, formats of family health history (FHH):**
  - Developed by HL7:

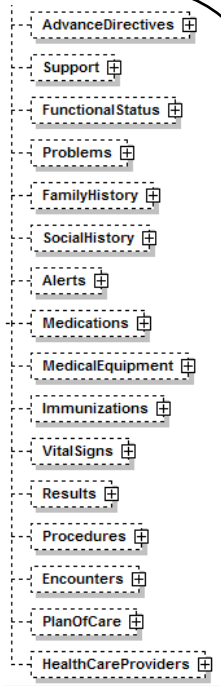
v3 Pedigree



CDS vMR FHH



CCD/CCDA
FHH Section



FHIR Resource

FHIR FH
Genetic Profile

Towards a Health Information Language

- **PCAST reports called for a "universal exchange language"…**

- **…but we need a more generic language, that is:**
  - Any type of information representation – not only exchange
  - Compositional / synthetic – creating expressive compositions
  - Translational – representing various disciplines & methods

Discussed in EFMI THI WG

- **We need a Translational Health Information Language (THIL)**
  - Such a language is closer to a natural language
  - Parsing / processing is harder, but…

  …if NLP algorithms can understand natural languages – couldn't similar algorithms understand a more structured language like THIL?!

- **Domain/usage-oriented standards will then be just specific compositions of that language, not set-in-stone constructs!**

- **Could we then phase out the current rigid standards?!**

Five Informatics Imperatives towards THIL

Should be applied to standards & their usage,
which might bring them closer to THIL
(underlying reference models should use ontology developing principles, e.g. BFO)

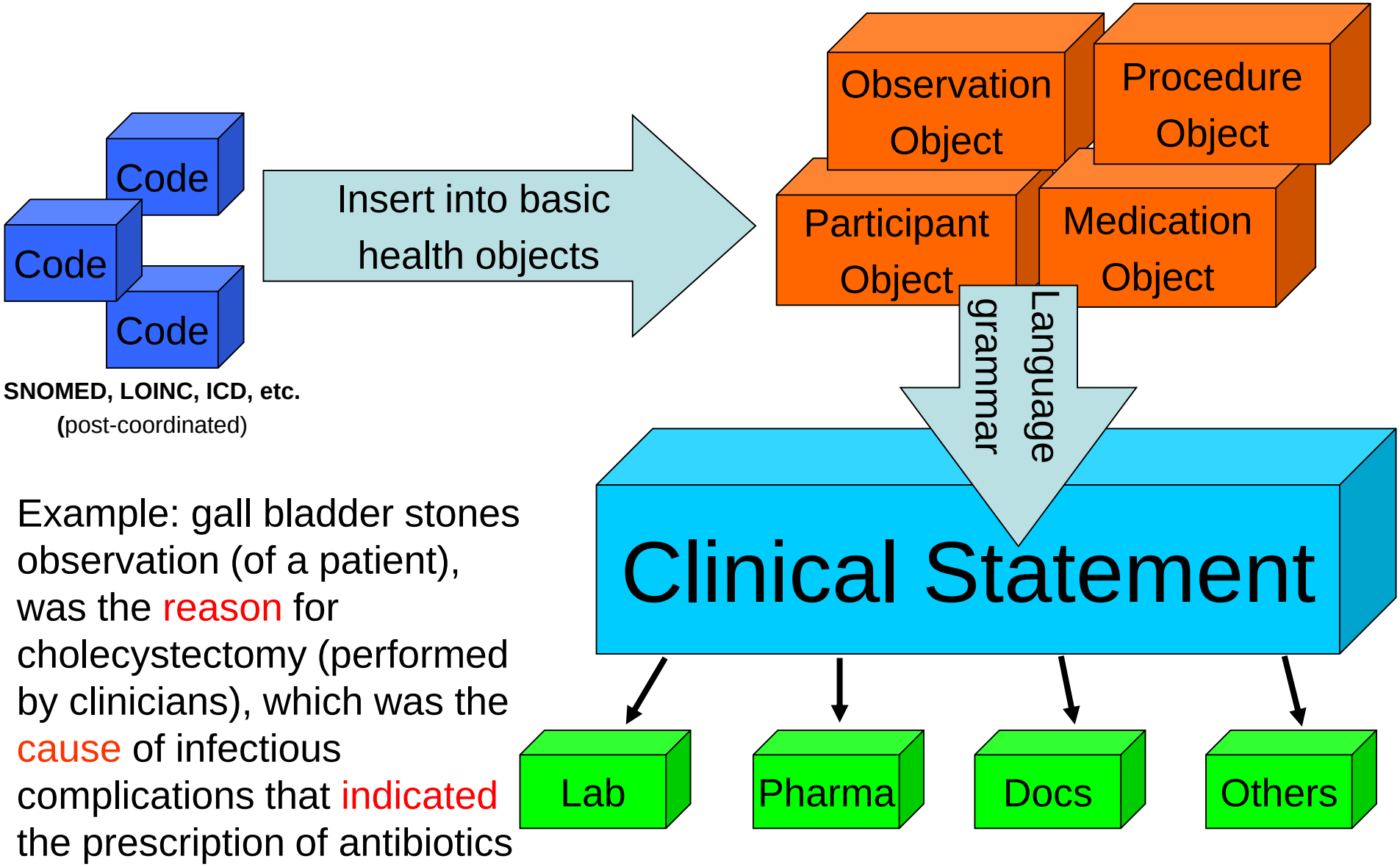
1. **Represent** contextual semantics explicitly
2. **Strike** a balance of narrative-structured data
3. **Encapsulate** key raw data (omics, sensors, images)
4. **Constrain** generic formats by model-driven tools
5. **Organize** all data into an EHR (+family history)

Represent Contextual Semantics Explicitly

Health data semantics and context cannot be faithfully represented using flat structures (e.g., a list of disconnected entries), rather, it requires the association of entries into meaningful statements (while using post-coordinated codes)

Imperative #1

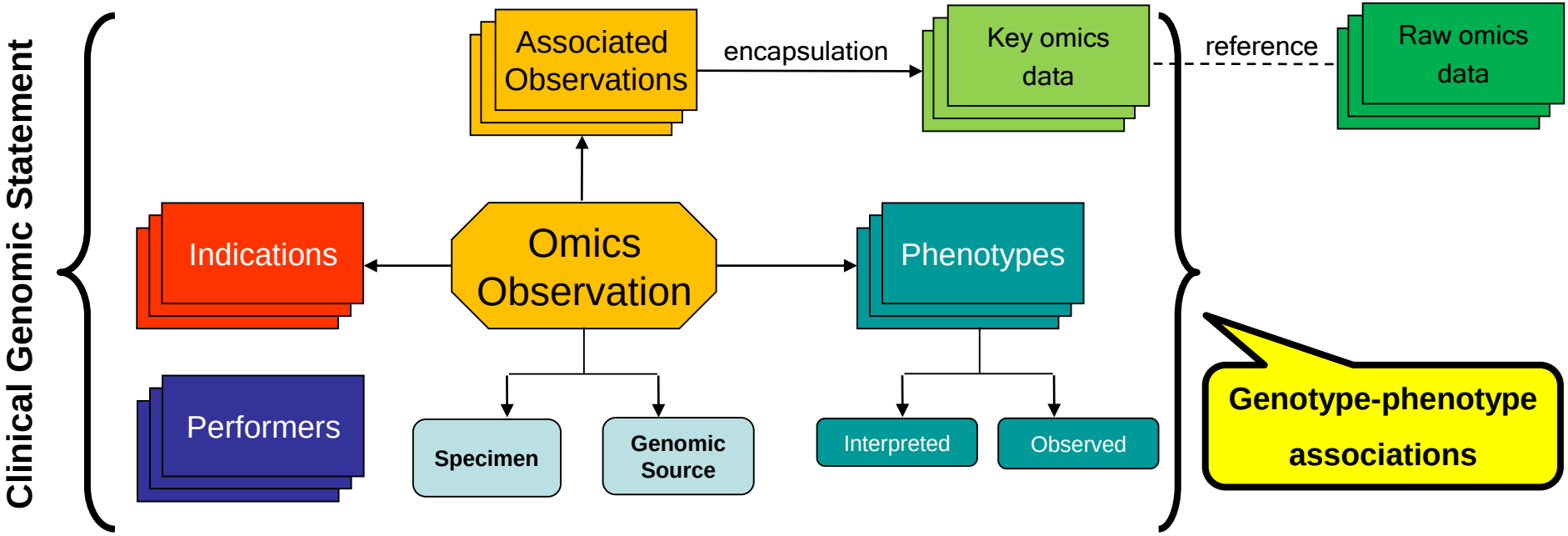
From Codes to Entries to Statements



Example: gall bladder stones observation (of a patient), was the reason for cholecystectomy (performed by clinicians), which was the cause of infectious complications that indicated the prescription of antibiotics

It's already available through the new generation of standards, but not used in practice!

Clinical Genomics Statement Model



- **Specializes the HL7 Clinical Statement model**
- **Aligned with HL7 Clinical Genomics specs**
- **Subset is used by the Genetic Testing Report (GTR)***

Developed by the HL7 Clinical Genomics WG

* GTR was created by constraining the HL7 Clinical Document Architecture (CDA) base standard

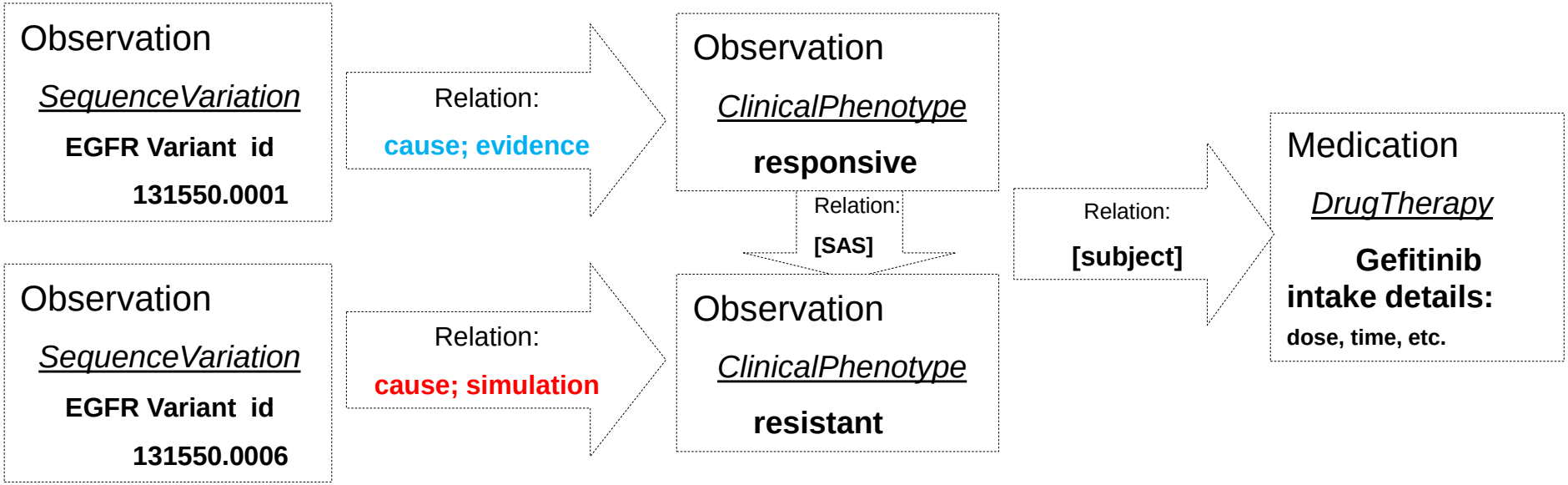
Translational Clinical Genomics Statement

Kobayashi et al. (2005) reported a patient with advanced non-small cell lung cancer in complete remission during treatment with Gefitinib (he was Gefitinib-responsive due to somatic EGFR-mutant). However, after 2 years he got into relapse.

Clinical genomics

Re-sequencing DNA of the EGFR gene in his tumor biopsy specimen at relapse revealed the presence of a second mutation. Structural modeling and biochemical studies showed that this second mutation led to the Gefitinib resistance.

Translational



Strike a Balance of Narrative-Structured Data

Accommodate unstructured data (e.g., clinician's narrative, patient's story or research manuscript), while maintaining interlinks to structured data entries corresponding to contents that have been structured

Imperative #2

HL7/ISO CDA (Clinical Document Architecture)

- Printed
- Bedside
- …

- EMR
- Transcription
- …

Rendered narrative

Rendered narrative

Human-to-Human

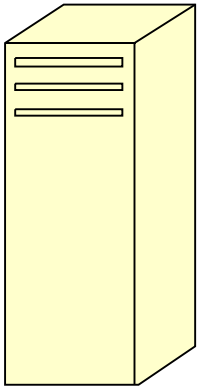
CDA

interlinks

Machine-to-Machine

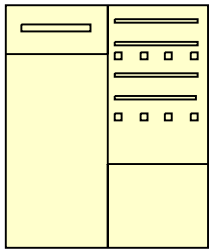
Structured data

Structured data



- Clinical Decision Support
- Patient held-records alerts
- …

- Medical Records
- Transformation
- …



Encapsulate Key Raw Data of Individuals

Key data sets out of raw/mass data should be encapsulated by clinical structures in its native format, and-

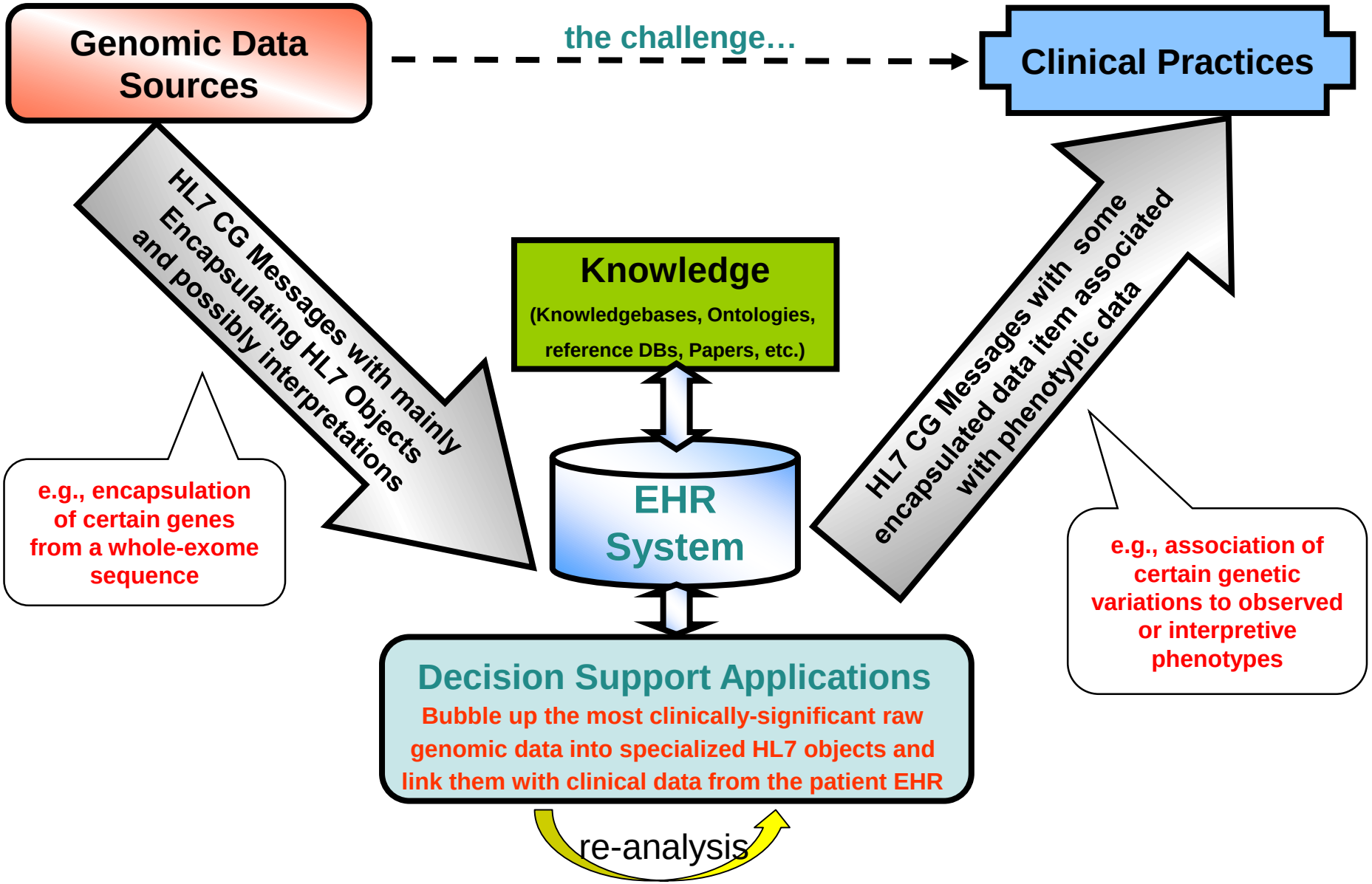
gradual ⇓ distillation

relevant items out of the key data sets should then be associated with phenotypic data (while maintaining traceability)

7

Imperative #3

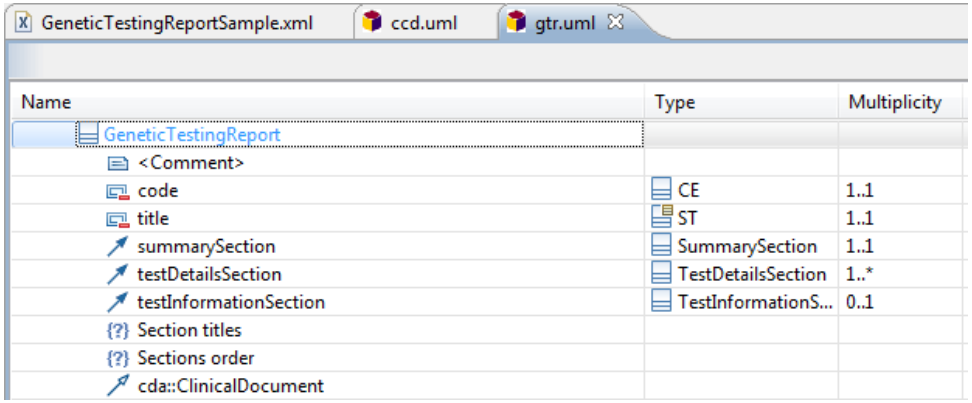
HL7 Clinical Genomics: Encapsulate & Bubble-up



Constrain Generic Constructs by Model-Driven Tools

Often, generic formats need to be constrained, however, derivatives might divert from the base semantics;
Model-driven constraining technologies prevent divergence!

Imperative #4



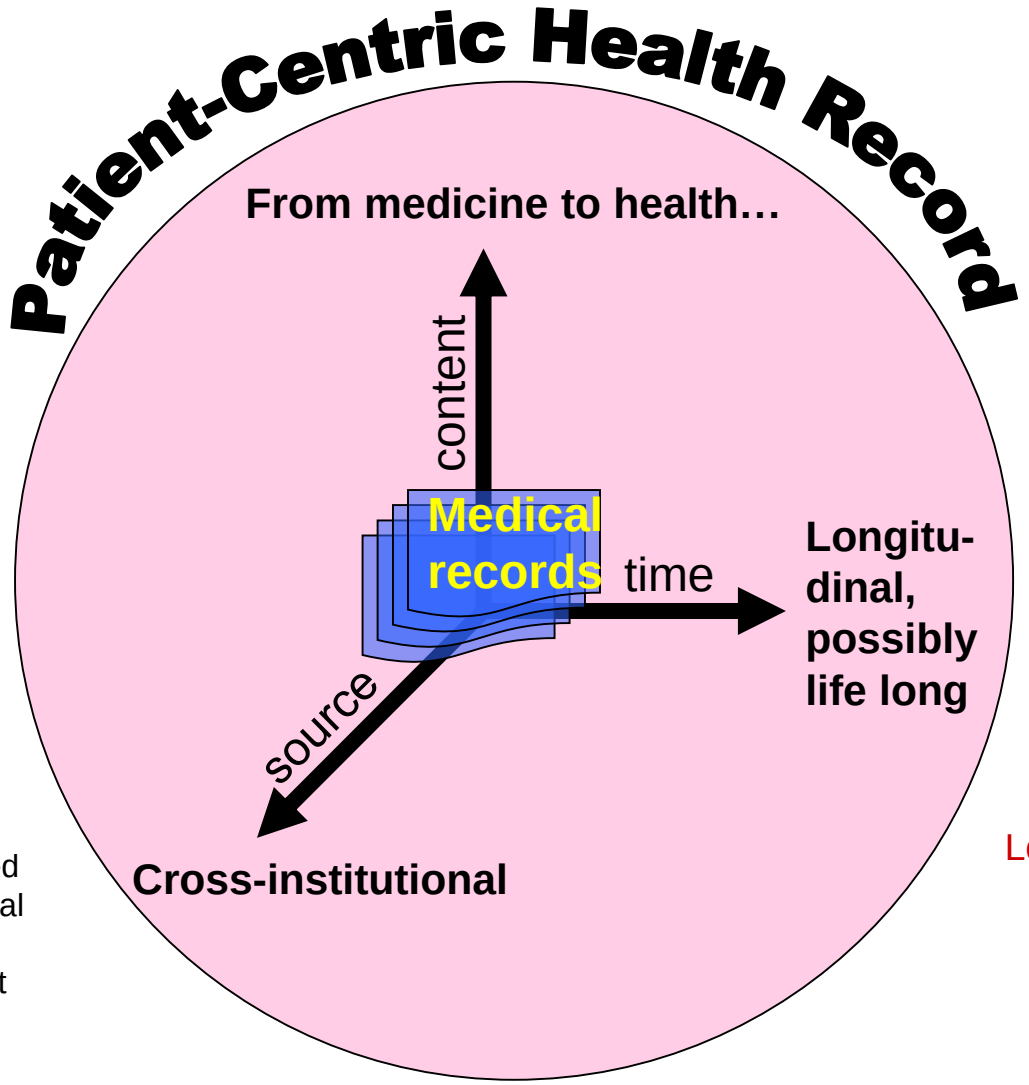
MODEL-DRIVEN HEALTH TOOLS

Organize all Data into an EHR (+Family History)

Data compliant with various biomedical standards should be integrated into a single & coherent information entity, representing the complete health information of an individual – a.k.a - the Electronic Health Record (EHR)

Imperative #5

From Medical Records to the EHR…



Health record

Any data items related to the individual's health (including data such as genetic, self-documentation, preferences, occupational, environmental, life style, nutrition, exercise, risk assessment data, physiologic and biochemical parameter tracking, etc.)

Should also include bio data

Medical record

Every authenticated recording of medical care (e.g., clinical documents, patient chart, lab results, medical imaging, personal genetics, etc.)

Longitudinal (possibly lifetime) EHR

A single computerized entity that continuously aggregates and summarizes the medical and health records of individuals throughout their lifetime

What's Missing? Analytics for Personalization!

Trial & error

Rule & case-based Knowledge (intuition?)

Humans

DATA:
New types of data;
Incomplete history

KNOWLEDGE:
We don't know much more than we know

Decision making
Is hard!

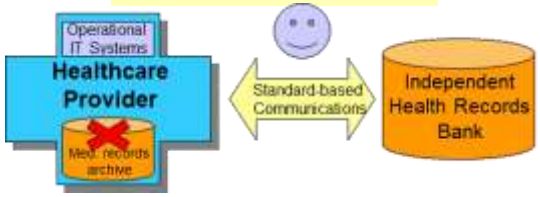
Machines

Add case-based reasoning for personalized care

The case is the lifetime EHR
(including family health history)

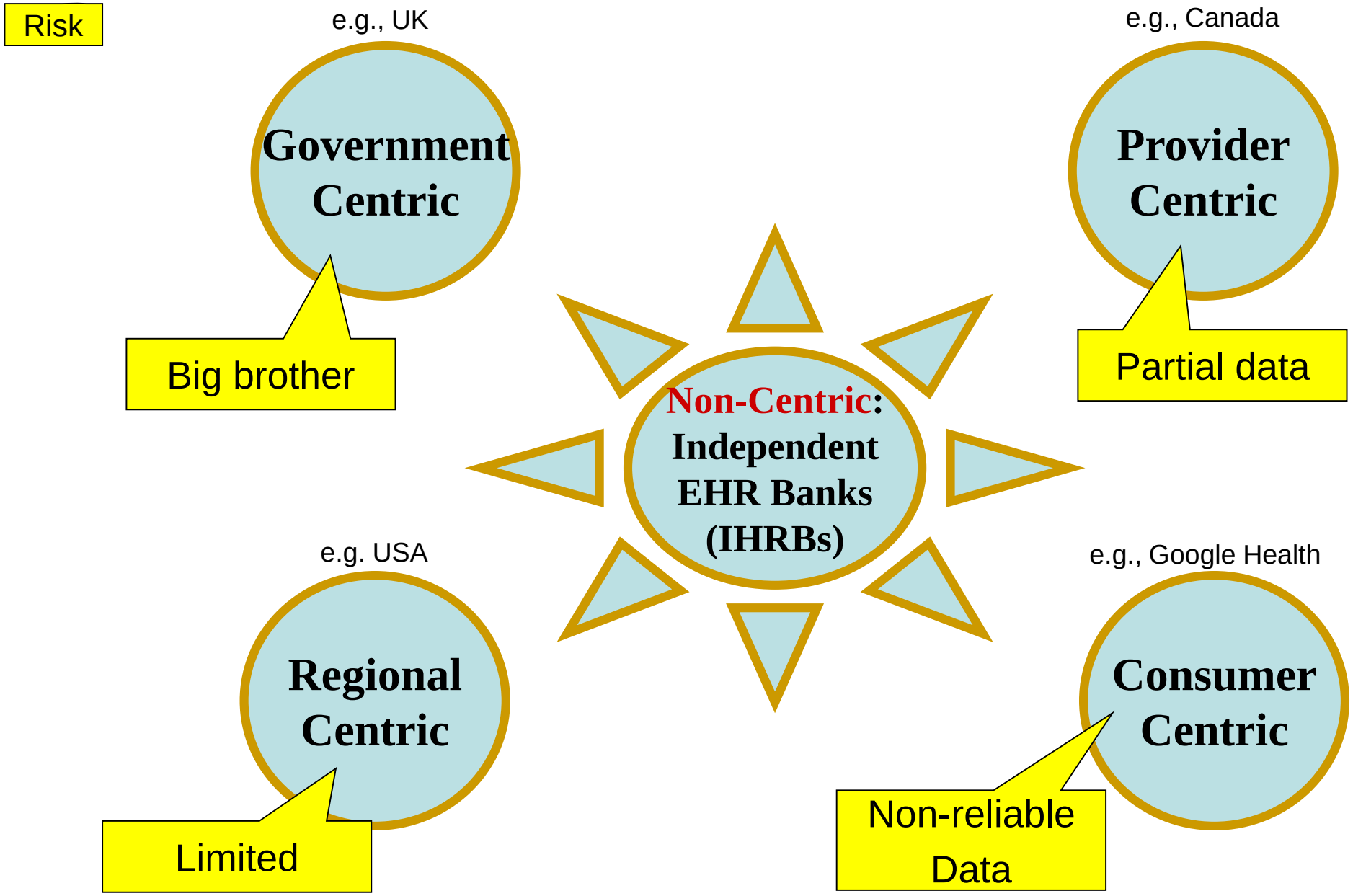
Sustainability

Health Record Banking



This presentation consists of materials published by Amnon Shabo (Shvo) in MedInfo and EFMI.

EHR Sustainability Constellations



EHR Sustainability and IHRB Assertions

- Longitudinal EHRs should not be federated (virtual) because:
  - Sources might not be available (down or out-of-business)
  - True summarization cannot be done "on the fly"

- Main assertion*:

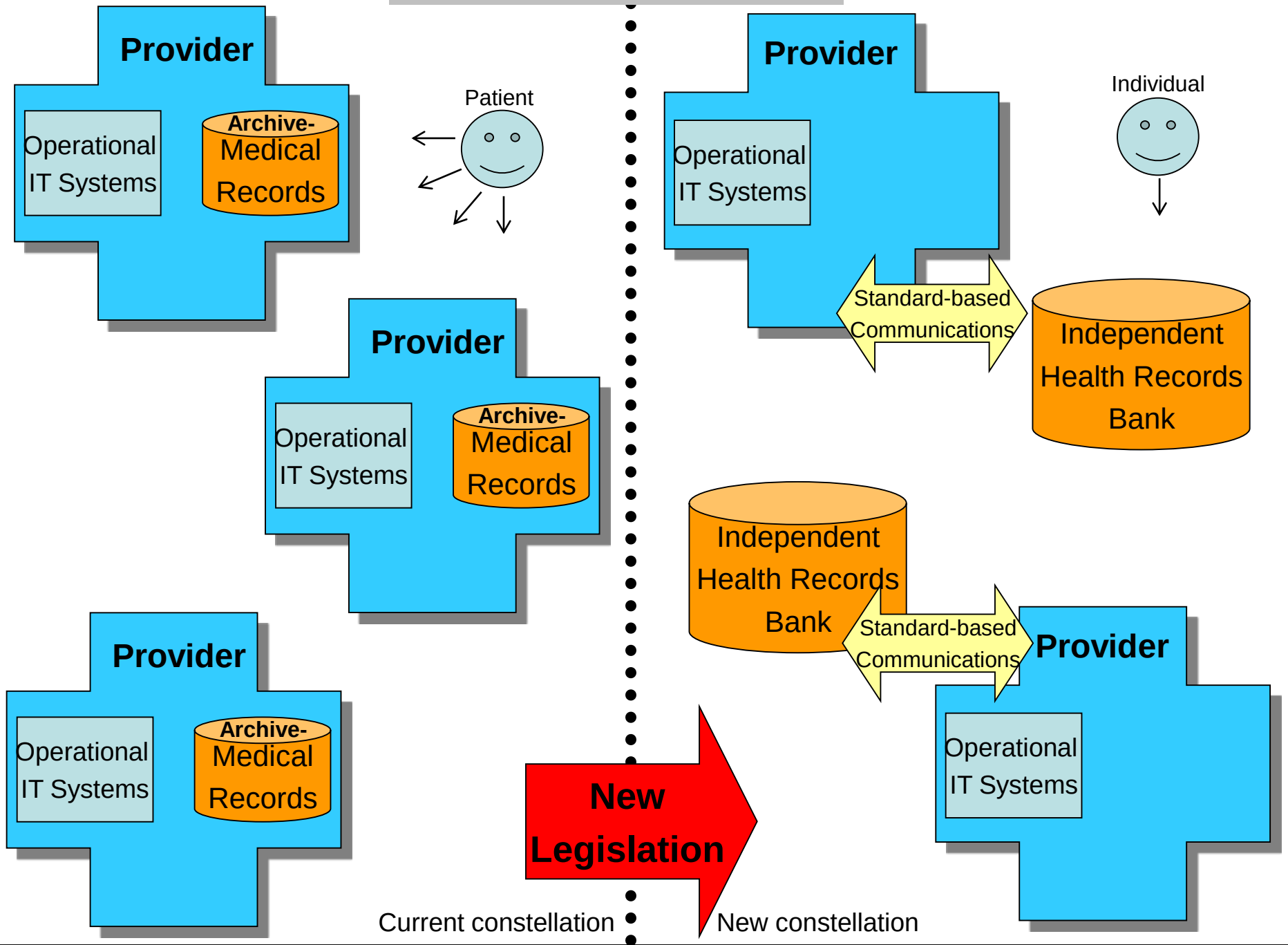
  None of the existing players in the healthcare arena can, or should, sustain aggregated lifetime EHRs

- Rationale:
  - Involves intensive IT computing tasks (archiving, preservation, etc.) which are not the main focus nor expertise of existing players

    Can not
  - If an existing player sustains EHRs, it might lead to ethical conflicts

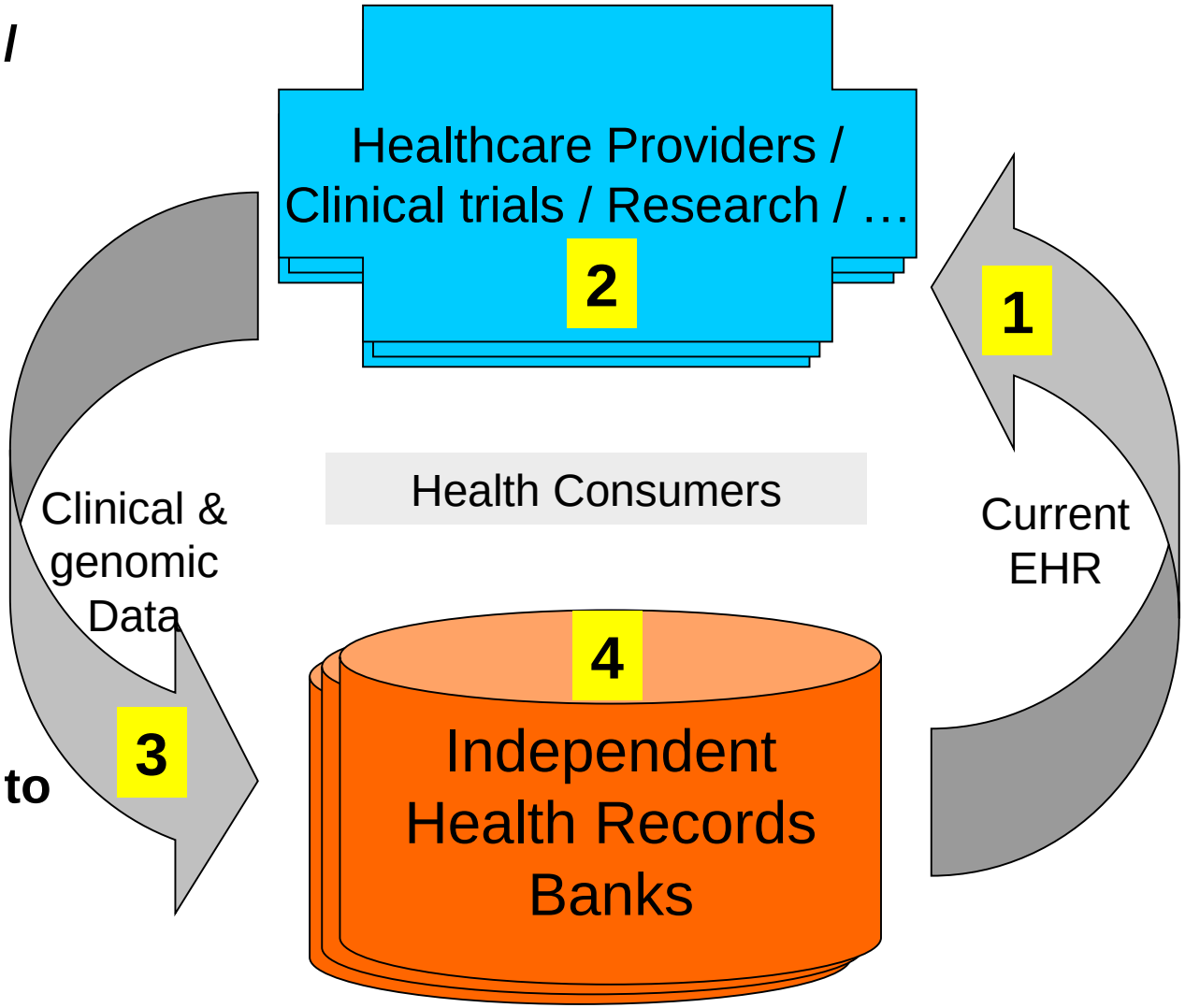
    Should not

The Conceptual Transition



The EHR Continuous "Production Cycle"

1. **Healthcare Provider / Clinical Trials or Research sponsor receives the current EHR from the patient's IHRB**
2. **Provides care / conduct research on the patient**
3. **Sends medical / health records back to the patient's IHRB**
4. **EHR is updated**

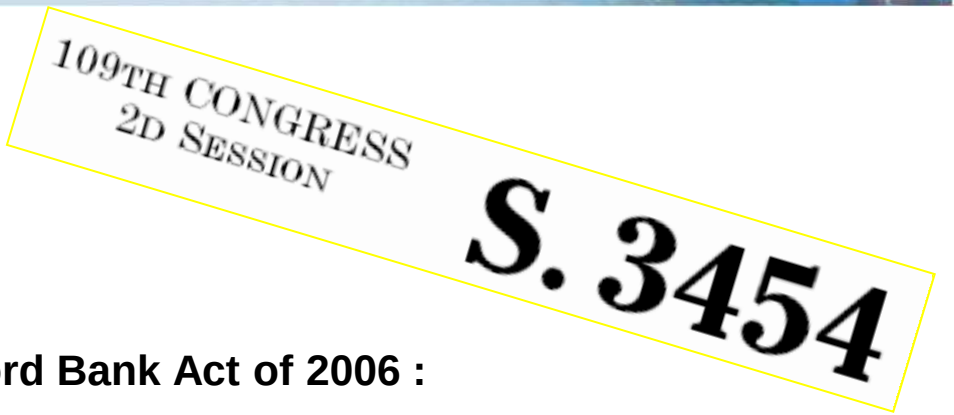


IHRB Legislation - Main Principles

- The medico-legal copy of a medical record resides solely in an IHRB
- An IHRB must be independent of healthcare providers, health insurers, government agencies, or any entity that might present a conflict of interests
- Allow for multiple independent IHRBs, regulated by the authorities, preferably functioning as not-for-profit organizations (cooperatives?)
- A consumer can move from one IHRB to another
- A consumer's EHR is identified by its IHRB account number, so there is no need for unique IDs at any level

IHRB Bills Introduced in the US Congress

Legislators were influenced from my publications, but didn't go all the way as I suggested… ☺



Brownback (R-KS): Independent Health Record Bank Act of 2006 :

- IHRB goals are to save money and lives in the health care system
- Only non-profit entities are permitted to establish IHRBs
- IHRBs function as cooperative entities that operate for the benefit and interests of the membership of the bank as a whole
- Revenue:
  - IHRB's may generate revenue by
    - charging health care entities account holders account fees for use of the bank
    - the sale of non-identifiable and partially identifiable health information contained in the bank for research purposes
  - Revenue will be shared with account holders and may be shared with providers and payers as an incentive to contribute data
  - Revenue generated by an IHRB and received by an account holder, healthcare entity or health care payer will not be considered taxable income

Discussions take place in IMIA HRB (Health Record Banking) WG and HRBA (HRB Alliance)

Summary

- Translational health informatics
- Biomedical information formats landscape
- Moving away from rigid domain/usage standards…
  …towards Translational Health Information Language (THIL)
- Five Informatics Imperatives when moving towards THIL
  - Represent associations between entries explicitly
  - Strike a balance of narrative and unstructured data
  - Encapsulate key items out of raw/mass data
  - Constrain generic info structures using model-driven tools
  - Use the EHR as the main organizer for individual data (incl. FHH)

INFORMATICS

- Refine clinical decision support through case-based reasoning
- The case is the lifetime EHR (the 5 imperatives make it rich)

ANALYTICS

- Healthcare providers should not be the record keepers, rather –
  independent EHR data banks should sustain personal EHRs!

POLICY

The End

- Thanks for your attention!
- Questions?
- Comments: amnon.shvo@gmail.com

Records of

Revolutionizing healthcare through independent lifetime health records

Health

Standards of

Towards a universal health information language