eHealth week

Organised by:

In collaboration with:

SNOMED CT in the Age of Precision Medicine

Charles Gutteridge, Clinical Engagement Lead EMEA, SNOMED International

@ gutteridgec

10–12 May 2017 MALTA

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HOW DO HUMANS ACT?

31414400

230063004

16064300

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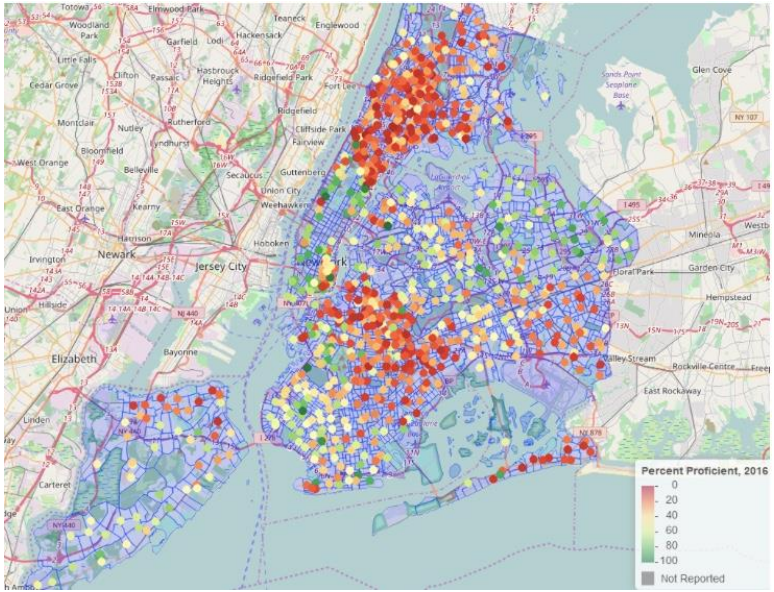
MAPPING VARIATION AND LEARNING



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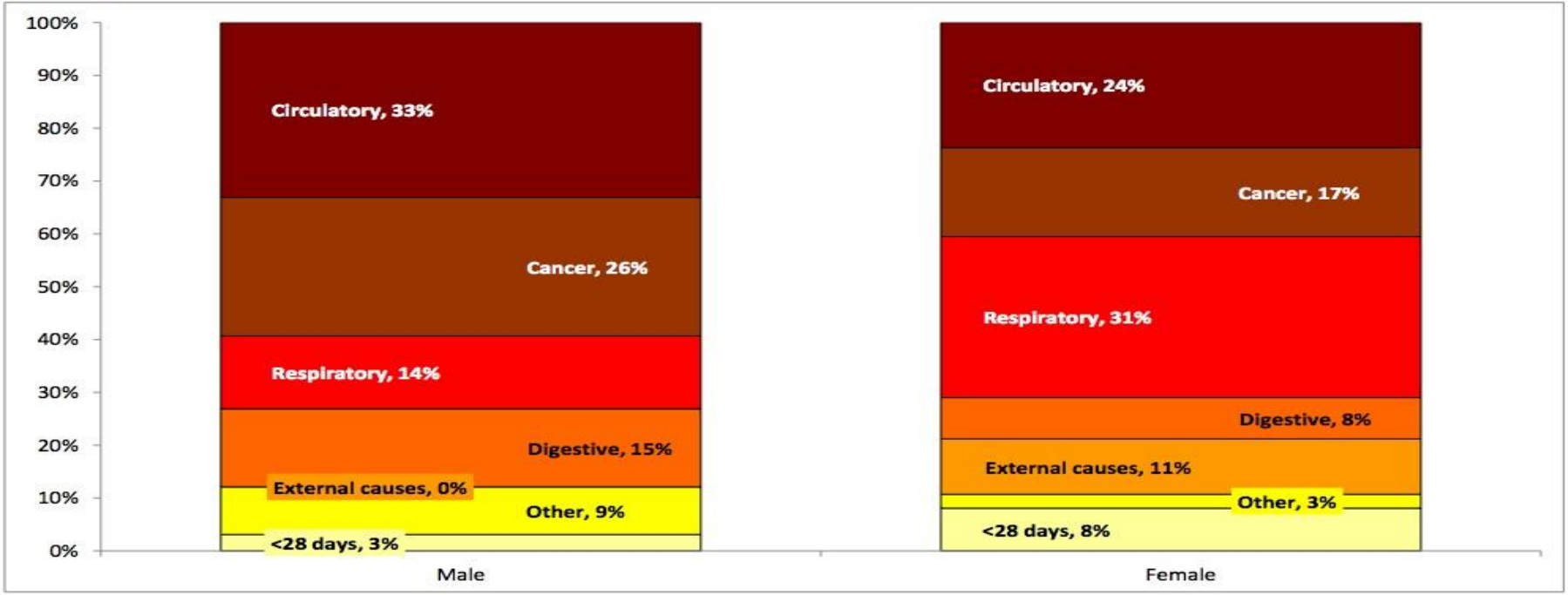
THE SEGMENT TOOL

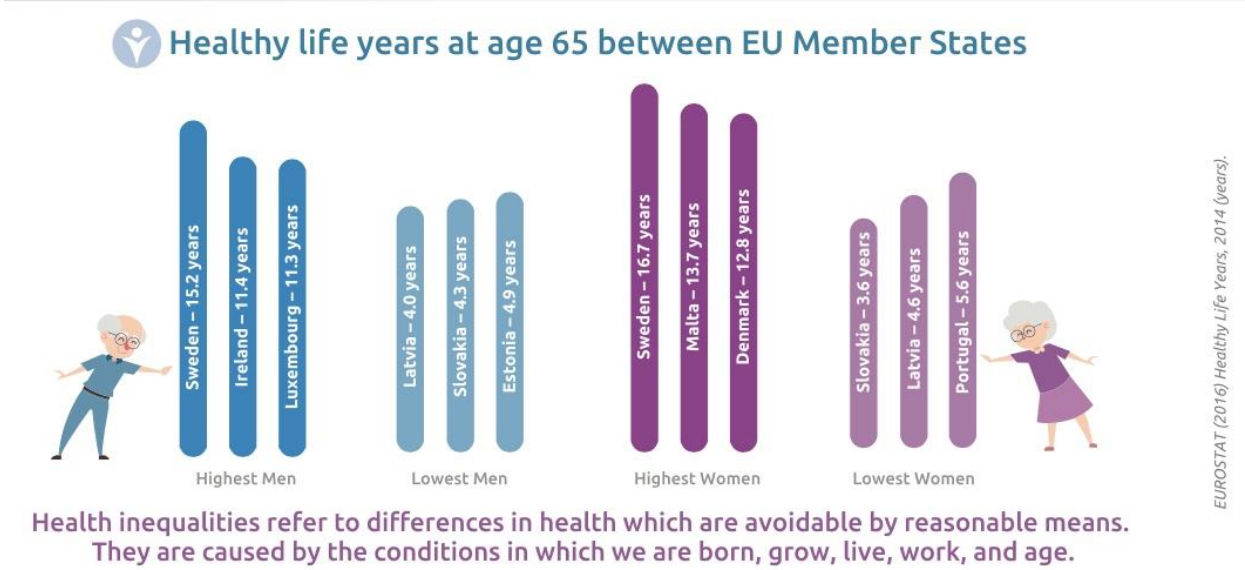
SEGMENTING LIFE EXPECTANCY GAPS BY CAUSE OF DEATH

Public Health England

Chart 1: Scarf chart showing the breakdown of the life expectancy gap between Tower Hamlets as a whole and England as a whole, by cause of death, 2009-2011



HUMAN CONSEQUENCES



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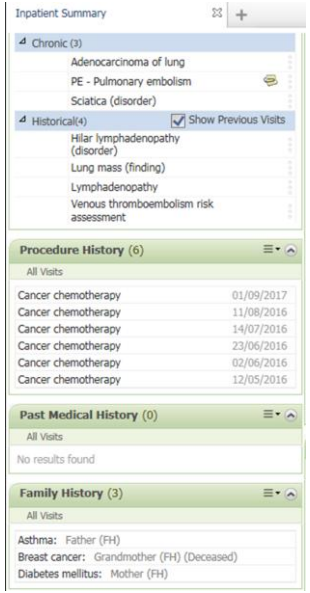
POINT OF CARE DATA FOR COMPUTATION



- Immediate access for care
- Always on
- Activity data recorded
- Reports real time
- Report flexibility
- Clinically driven

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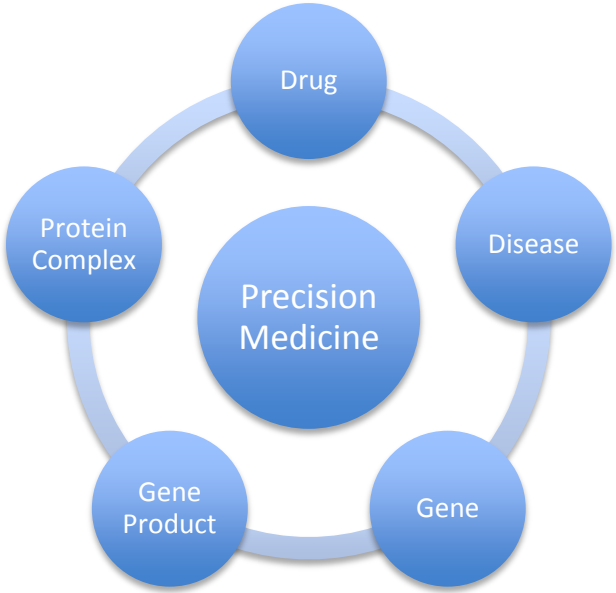
PATIENT AND CLINICAL UTILITY



Show me all the GENES with somatic mutations in **BIOBANK** samples with the SNOMED Clinical Term for LUNG CANCER and reference with PHARMA database to show me all COMPOUNDS known to modulate those GENES

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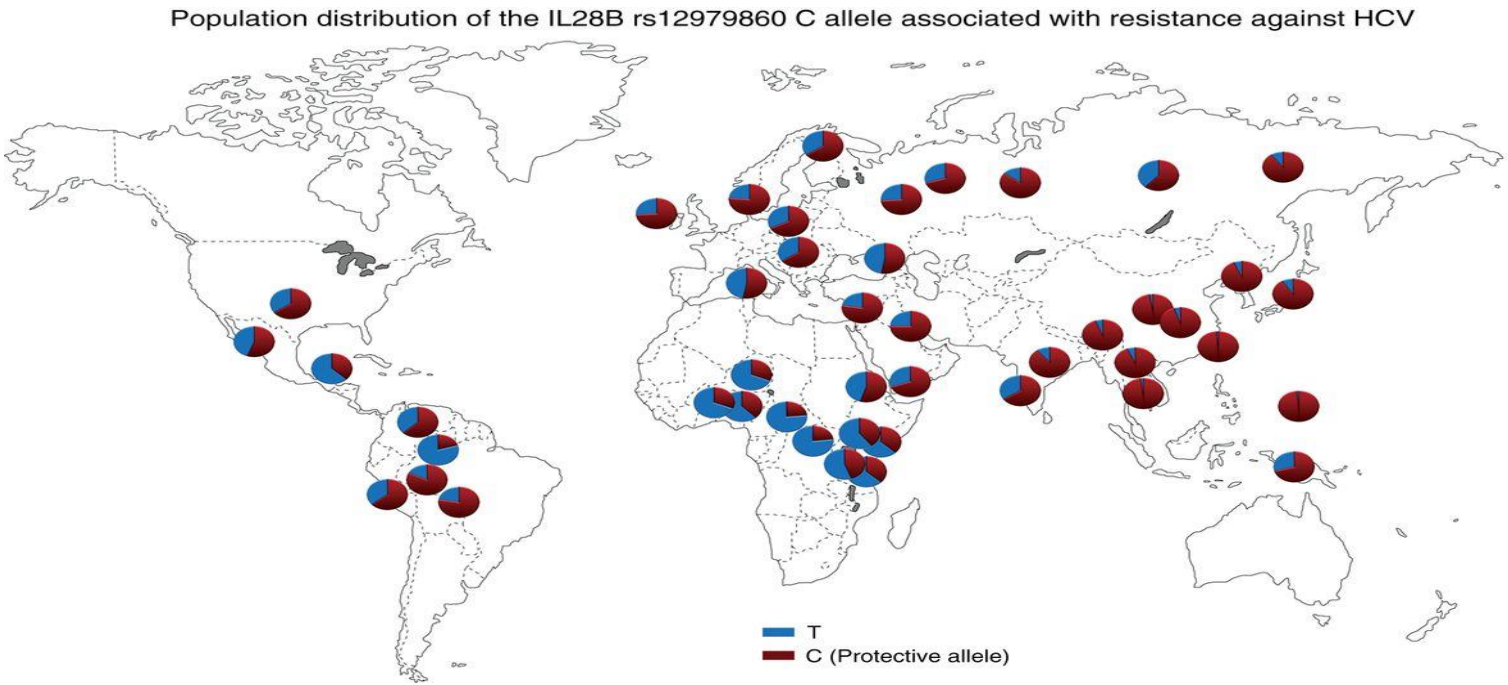
Worldwide distribution of the IL28B SNP rs12979860 allele variation.



Jérémy Manry, and Lluis Quintana-Murci Cold Spring Harb Perspect Med 2013;3:a012450

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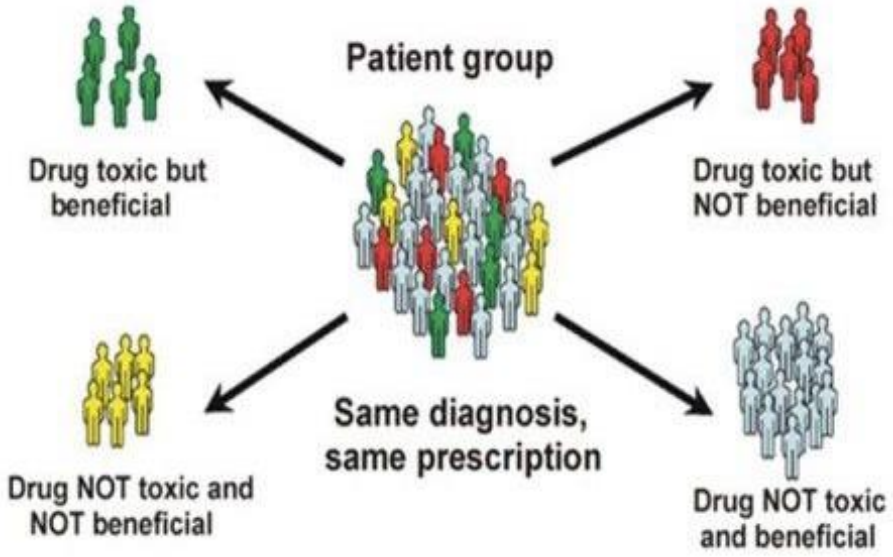
DRUG EFFECTS AND VARIATION



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USING DATA FROM ELECTRONIC HEALTH RECORDS FOR DISCOVERY

PLoS One. 2017; 12(2): e0171745.
Published online 2017 Feb 21. doi: 10.1371/journal.pone.0171745
PMCID: PMC5319785

Genome-wide study of resistant hypertension identified from electronic health records

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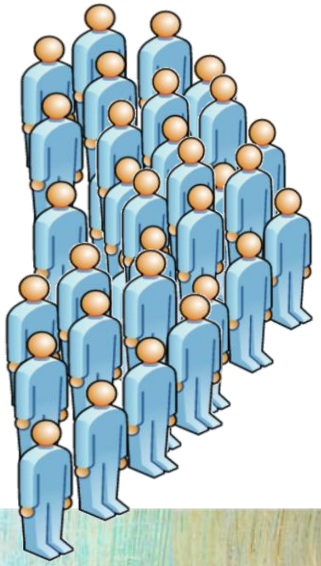
PRECISION MEDICINE AND CLINICAL PHENOTYPING

Big Data Analytics



- Epidemiology
- Population Health Needs Assessment
- Clinical Audit
- Clinical Research
- Large Clinical Trials
- Gathering Evidence for Best Practice

Small Data Analytics

- Patient Record Summary
- Point of Care Reporting
- Knowledge Linkage
- Applying Best Practice Guidelines
- Adverse Reaction Reduction
- Decision Support

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BARRIERS TO PRECISION MEDICINE: INCONSISTENT REPRESENTATION OF CLINICAL CONTEXT

- Past history of appendicitis
- Appendectomy done in 2007
- Abdominal pain
  - Present, absent, resolved
- Diagnosis
  - Differential, provisional, suspected, possible, confirmed, excluded …

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PRECISION MEDICINE AND CLINICAL TERMINOLOGY

- Consistent approaches to representation of clinical ideas and context are required to address barriers to clinical analytics
- A well designed clinical terminology should enable data captured in different ways to be represented in semantically compatible and comparable forms
- But what constitutes a well-designed clinical terminology?

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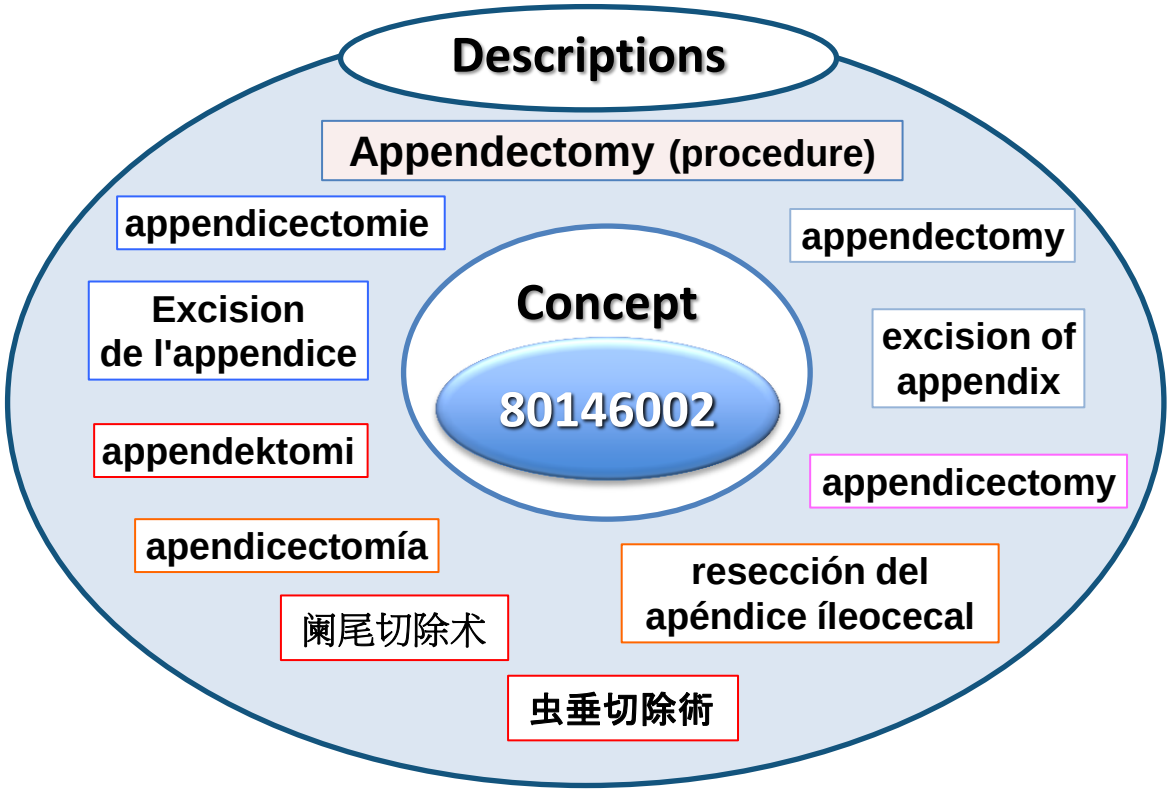
Concept Orientation

- One unique code (identifier) for each distinct clinical meaning (concept)
- Terms with the same meaning are represented as descriptions linked to the same concept
- A description linked to a concept may contain a term in any language or dialect

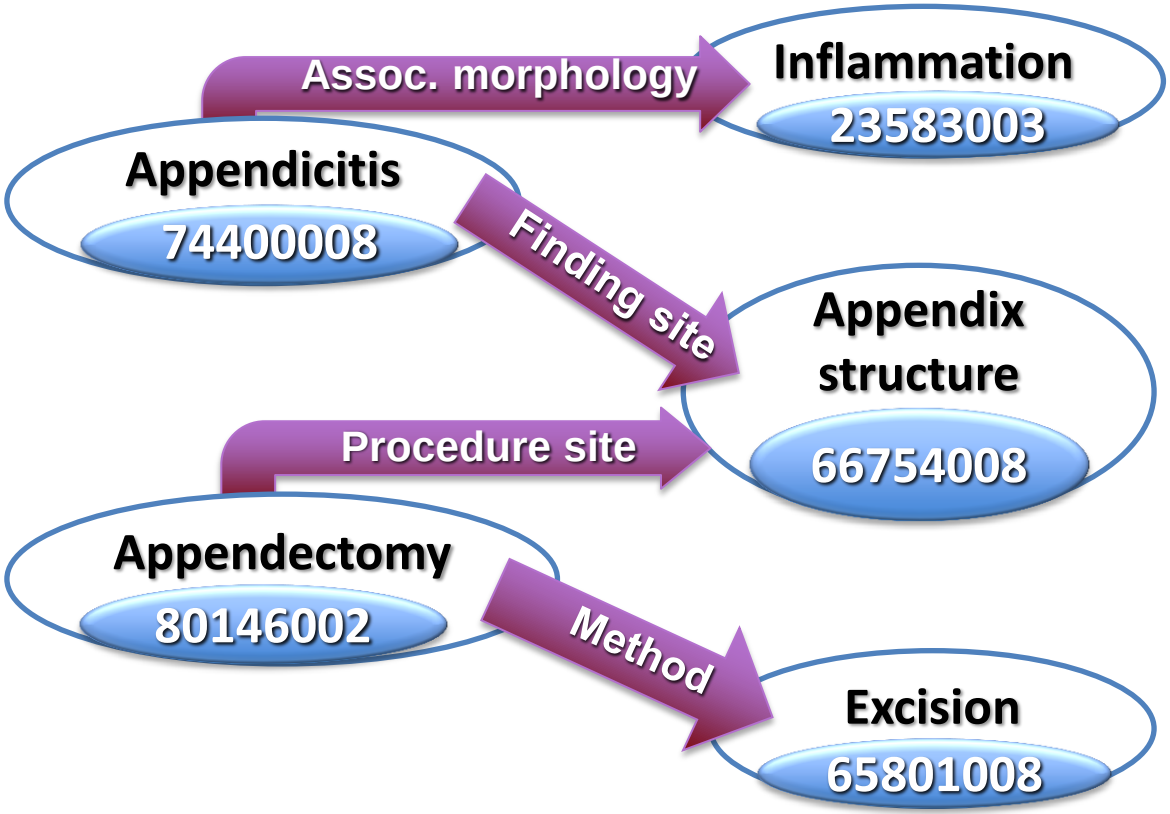


Formal Description Logic Definitions

- Description logic definitions formally represent the meaning of concepts

Examples

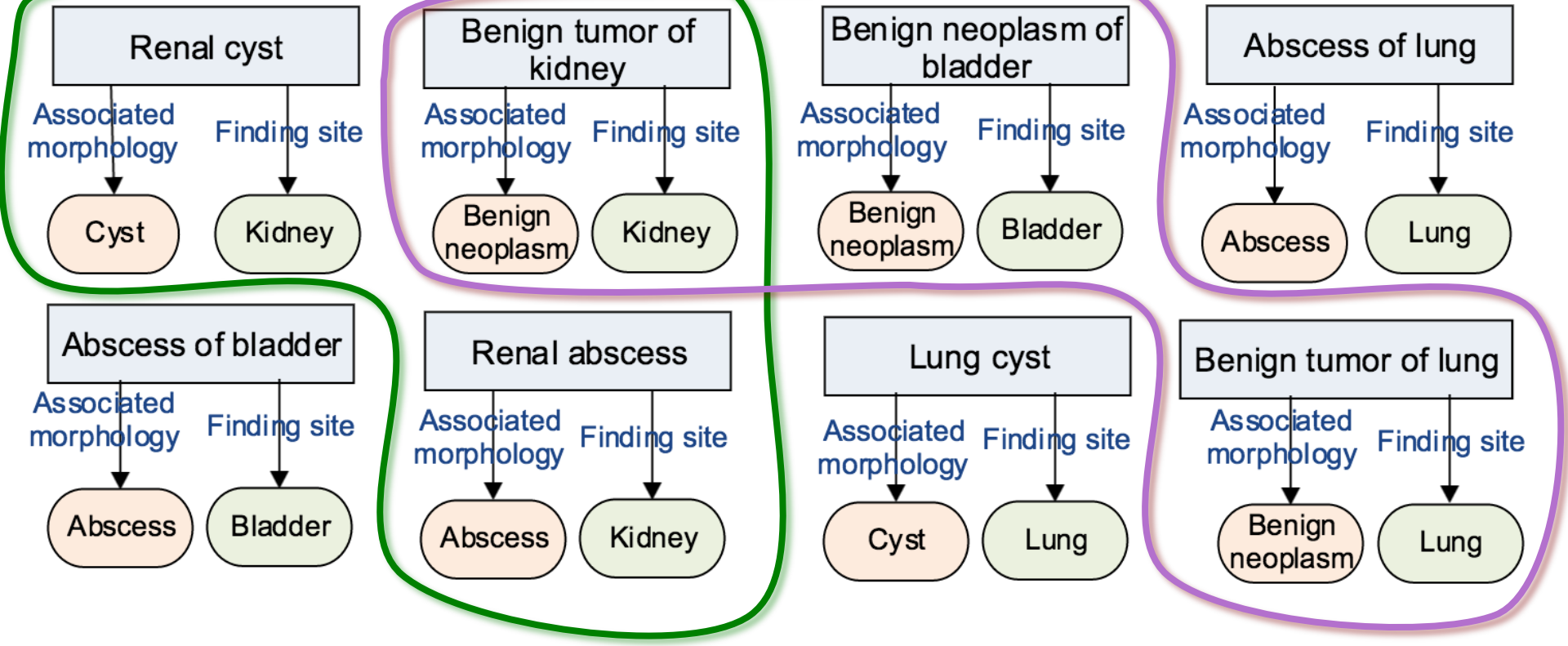
- Appendicitis is defined as
  - Inflammation of the
  - Appendix structure
- Appendectomy is defined as
  - Excision of the
  - Appendix structure



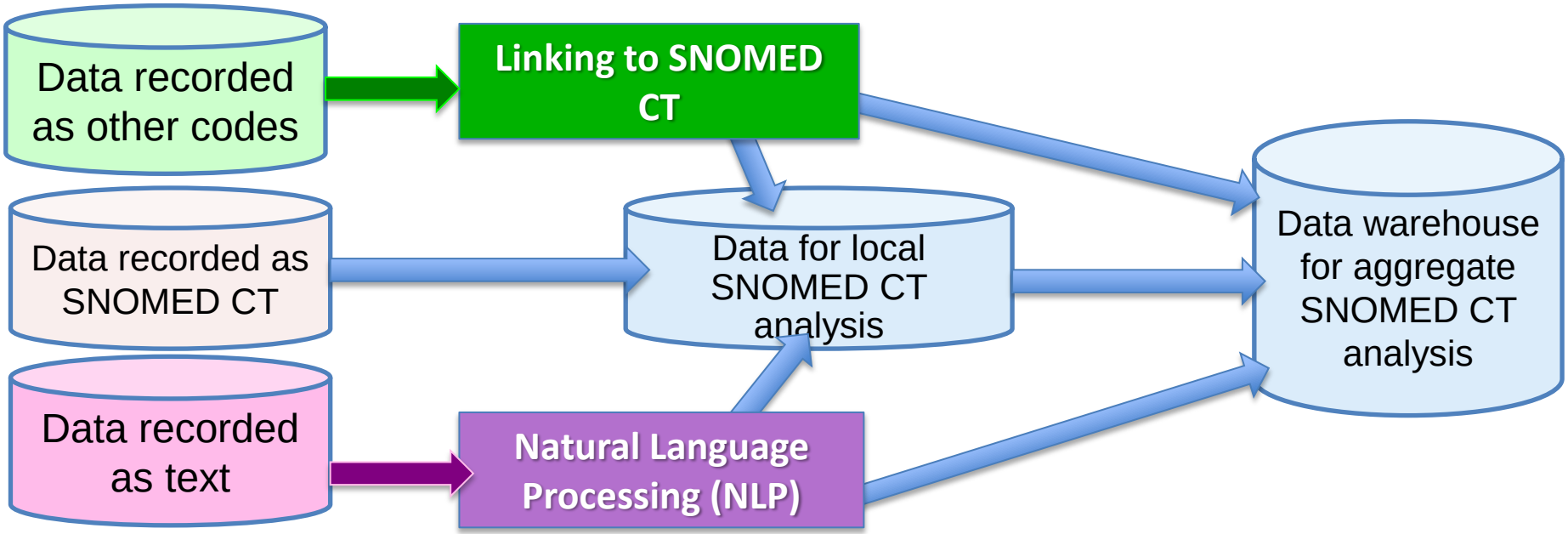
Queries Based on Logic Definitions

Query: Disorders with finding site kidney

Query: Disorders with associated morphology benign neoplasm



Preparing Data for Analytics with SNOMED CT



FUTURE DIRECTION FOR SNOMED CT IN PRECISION MEDICINE

- Publication of additional case studies focusing on clinical decision support and other aspects of data analytics
- Guidance on use of SNOMED CT to enhance decision support
- Publication of guidelines on to use of SNOMED CT in other standards (e.g. HL7 FHIR) to support interoperability and enable analytics over data recorded in different record structures
- Greater use of SNOMED CT's features in more vendor products
- Realization of large scale benefits from more effective clinical analytics

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CONTROLLED VOCABULARY: SNOMED CT

SMART precision cancer medicine: a FHIR-based app to provide genomic information at the point of care

FREE

Jeremy L Warner; Matthew J Rioth; Kenneth D Mandl; Joshua C Mandel; David A Kreda; Isaac S Kohane; Daniel Carbone; Ross Oreto; Lucy Wang; Shilin Zhu; ... Show more

J Am Med Inform Assoc (2016) 23 (4): 701-710. **DOI:** https://doi.org/10.1093/jamia/ocw015
Published: 27 March 2016 **Article history** ▾

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BUILDING THE PLATFORM FOR PRECISION MEDICINE

- Health data is for patients, carers and families
- Information connects people to their carers and providers
- Single systems must be planned to connect
- Interoperability matters
- Connectedness needs security
- Connectedness should be a health outcome measure

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Thank you!

Charles Gutteridge, Clinical Engagement Lead, SNOMED International

@gutteridgec

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