# SNOMED CT and Genomic Medicine

## Working together towards a common strategy

Delivering SNOMED CT
The global language of healthcare

# Overview

## Implementing Genomic Medicine

1. Vision
2. Scope
3. Purpose
4. Background
5. SNOMED CT Strategy (what & how)
   - Strategic objectives
6. Next steps (what next)

# Scope

Genomics promises to change the way that medical care is delivered across the globe. These changes will be delivered through the implementation of precision/ personalised medicine. Discoveries within Genomics requires detailed health information to be made available to researchers, and in future the genomics community will supply actionable genomics based guidance to be made available within healthcare institutions, and specifically within EHR systems.

The scope of the following strategy is to focus future developments of SNOMED International to ensure that the requirements for the global Genomics community are supported by SNOMED CT.

# Purpose

The purpose of the following SNOMED International strategy, is to ensure that moving forwards, SNOMED International and SNOMED CT can support the needs of the global Genomics community, and to ensure that SNOMED CT can fully support the future global implementation of Precision Medicine.

# Vision

SNOMED CT: The global clinical terminology to support Genomic and Precision Medicine implementation.

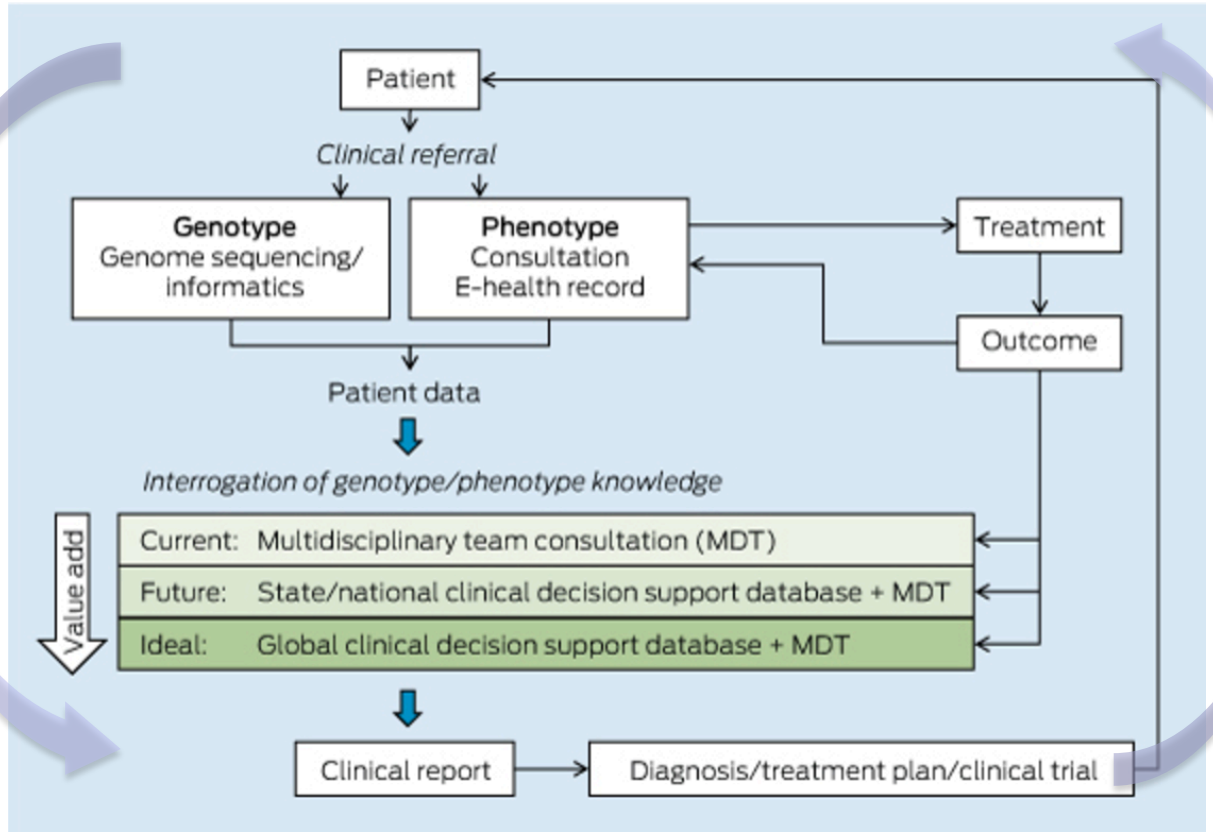
(Supporting clinical information to the Genomics community and detailed genomics information to the clinical community)

# Clarity of vision

- SNOMED CT will not look to include content from all Genomic terminologies, but rather to align to those with a clear link to clinical practice through the EHR
- SNOMED CT will look to leverage existing linkages between terminologies/classifications (both clinical and genomic) to enhance its usability
- SNOMED CT will look to develop relationships with terminologies and classifications that will support the implementation of Genomics within SNOMED CT
- SNOMED CT will look to adopt new clinical language and clinical definitions arising from Genomics, and reflect these changes within the descriptions included within the International release, when there are clear use case requirements to do so

**Background**

# Patient journey in Genomic Medicine




- Individual genomic data
- Accurate clinical data
- Alignment to the existing body of knowledge

Mattick JS, et al. *The impact of genomics on the future of medicine and health*. Med J Aust. 2014 Jul 7;201(1):17-20.

# Definitions and what will be delivered

- **Genomic** medicine = precision medicine
  - Genomics refers to the individual / personalization factors within medicine
  - Genomic data provides the patterns of genetic variation associated with phenotypes and disease states
- Precision medicine = personalized health care
  - Improving diagnosis by provision of a greater understanding of rare disorders, through to efficient and individualized diagnosis of common diseases
  - Provision of individualized treatments / therapies to benefit patients
  - Risk reduction through personalized screening
  - Prevention through population-scale screening

# Motivation: General context

- Currently, Genomics has:
  - Fragmented / specialised communities
  - Individual focus – e.g. genes, phenotypes, rare diseases, etc . .
  - Incomplete cross-referencing
  - Inconsistent and low levels of access to EHR data – different levels of specificity and variety of terminologies

- Genomic medicine:
  - Requires a standardized and coherent approach to express:
    - Genomic knowledge
    - Phenotypic knowledge across all types of disorders (rare & complex)
    - Exposures to enviromental factors
    - Clinical data (EHR/secondary usage data)

Background

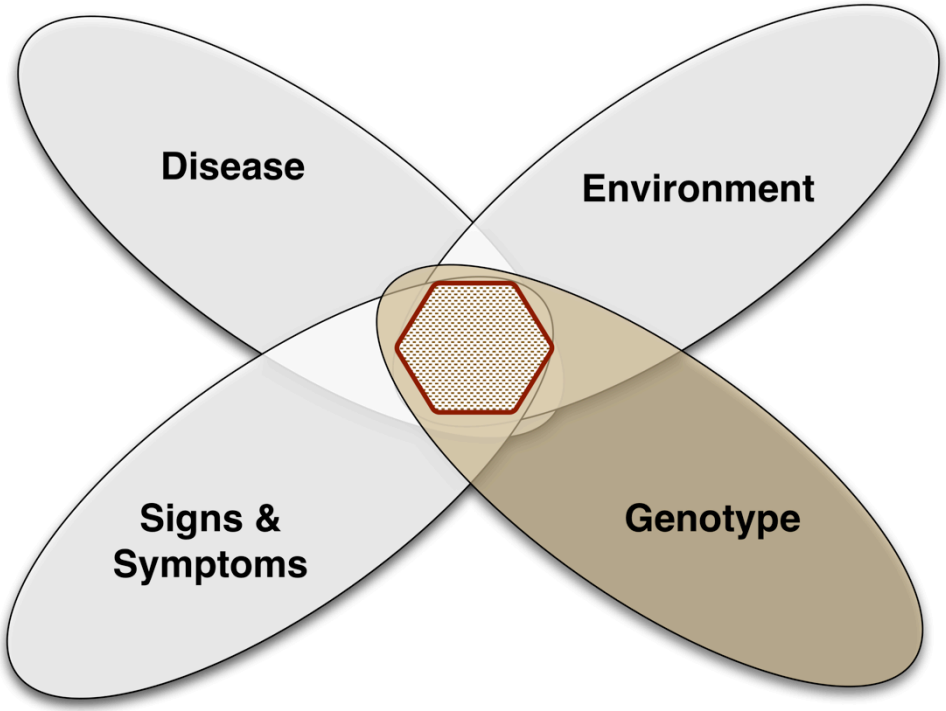
# Motivation: The SNOMED CT context

- **Currently, SNOMED CT has:**
  - Coverage of all clinically-relevant perspectives. Specifically:
    - Clinical findings
    - Phenotypes
    - Exposures/Environmental factors
    - Investigations
    - Anatomy

- **SNOMED CT requires:**
  - improved representation of genomic knowledge
    - Reflecting genetic findings, a mixture of:
      - –Biological processes, Structural information, Mutation types
    - Providing linkages between SNOMED CT and other Genomics terminologies/classifications:
      - –for example - HPO, Orphanet, OMIM

Background

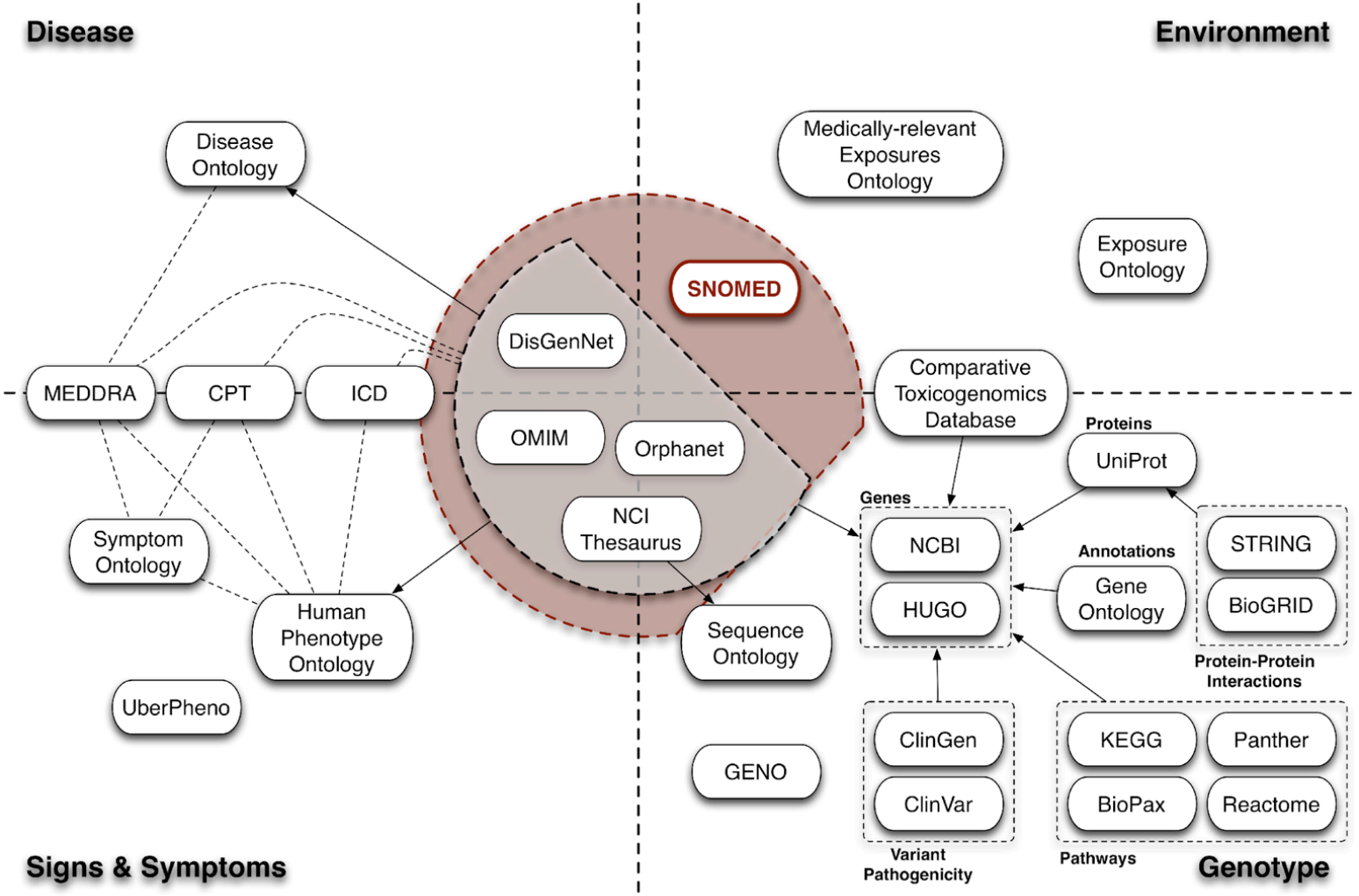
# Opportunity for SNOMED CT: Genomic Medicine




- **Phenome** (signs and symptoms) – individualized expression of variation
- **Diseases** – individual associations of variation and phenotypic expression
- **Environment** – influencing factor (via exposures); alters the expression of all other blocks
- **Genome** – foundational block; defining the individual externalization / expression of the phenome and diseases

**Background**

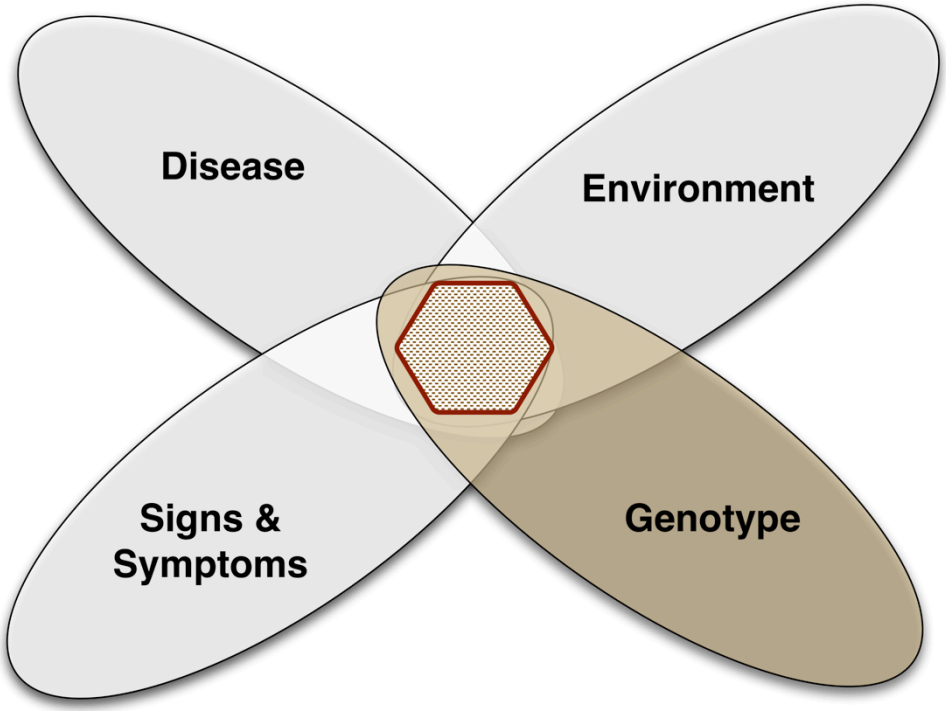
# Where does SNOMED CT fit within the existing Genomics terminology landscape ?




SNOMED CT is well positioned to provide content in each of the four areas, BUT each terminology will have its own strengths and will support specific use cases. SNOMED CT must therefore look to collaborate with existing terminologies.

Background

# SNOMED CT in the context of Genomic Medicine

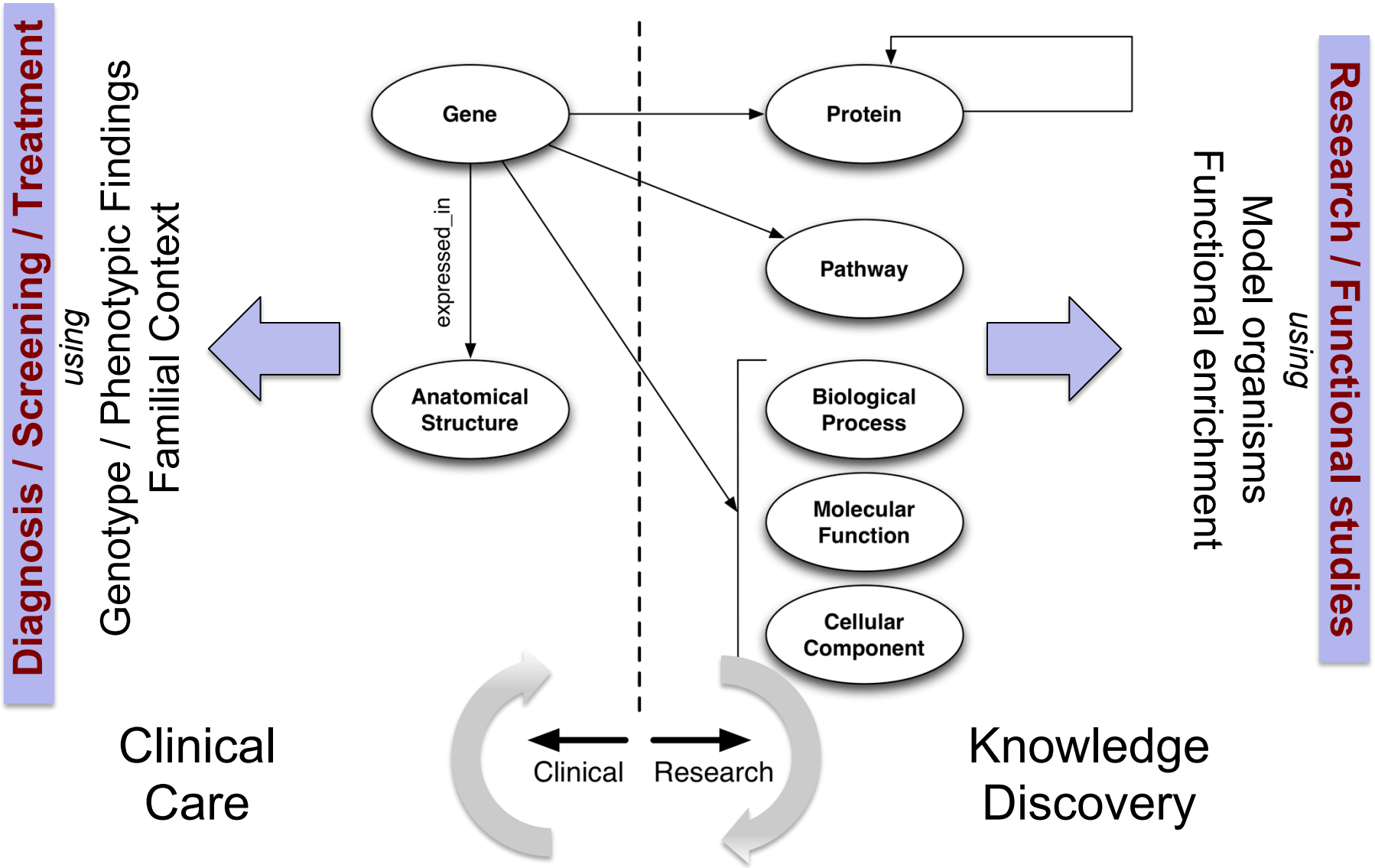



- SNOMED CT is ideally placed to support the vision of Precision / Genomic medicine, focusing on clinical utility by providing:
  - Diagnosis
  - Family history
  - Phenotypes
  - Environmental factors
  - Social context

  from the EHR, whilst providing a link to existing genotypes within the EHR

Background

# Delineating *Clinical Utility* from *Research*



**Background**

# Technical Desiderata - Integration of Genomic Data into EHR's

- Used to advise on the direction of coverage of genomic data within SNOMED CT
  - Maintain separation of primary molecular observations from the clinical interpretations of those data
  - Support lossless data compression from primary molecular observations to clinically manageable subsets
  - Maintain linkage of molecular observations to the laboratory methods used to generate them
  - Support compact representation of clinically actionable subsets for optimal performance
  - Simultaneously support human-viewable formats and machine-readable formats in order to facilitate implementation of decision support rules
  - Anticipate fundamental changes in the understanding of human molecular variation
  - Support both individual clinical care and discovery science

The Technical Desiderata provides a framework for addressing the needs of Genomic data within EHR's. Whilst we are are not bound to them, we will look to shape future developments within this context

*Technical Desiderata for the Integration of Genomic Data into Electronic Health Records*. Biomed Inform . 2012 June ; 45(3): 419–422. doi:10.1016/j.jbi.2011.12.005.

Background

# Restructure Domain knowledge - initial approach

- SNOMED CT content areas
  - Clinical findings, Observables, Procedures, Situations, Organisms, Anatomy
- Content focus, based on genomics
  - Completeness of content (Do we have the necessary concepts?)
  - Text formation (Do we have the correct wording, based on genomics and future precision medicine requirements)
  - Modeling (Do we have the necessary attributes available to meet the requirements of the genomics community)
- Derivative products
  - Maps/linkages (Do we have the necessary products to meet the stated requirements of the Genomics community)
  - Subsets (Do we have the required subsets of content, and a process to generate necessary subsets in the future?)

Background

# SNOMED International Strategic directions

- **SD1. Product:** Enable continuous development of the product to meet customer requirements
- **SD2. Adoption:** Provide scalable products and services that remove barriers to drive adoption of SNOMED CT
- **SD3. Innovation:** Leverage new trends and technologies that benefit SNOMED CT and its stakeholders

# Genomics Strategy objectives

- **Objective 1:** Identify key stakeholders with appetite for trial/ adoption of SNOMED CT for Genomic Medicine
- **Objective 2:** Identify and gather use cases to support and validate SNOMED CT in the context of Genomic Medicine
- **Objective 3:** Restructure domain knowledge around clinical genomics in SNOMED CT
- **Objective 4:** Demonstrate added value provided by SNOMED CT in the context of Genomic Medicine
- **Objective 5:** Raise awareness around SNOMED CT globally, and reinforce its suitability for supporting Precision Medicine

Objective 1

# Identification of key stakeholders

- Deliverable:
  - Identified group of key genomics stakeholders/collaborators
- Action plan:
  - Select (or expand and then select) a group of key collaborators through direct contact
  - Form a SNOMED CT subject matter expert group to provide continuous input into developments
  - Develop and deliver engagement sessions focused on SNOMED CT
  - Scope out and deliver a mid-to-long term engagement plan with the stakeholders forming the SNOMED CT stakeholders consultancy group
- Measure of success:
  - List of key genomics stakeholders/collaborators actively involved in SNOMED International developments
  - Agreed mid-to-long term engagement plan
  - (longer term) Proactive participation of stakeholders in showcasing events (see Objective 5)

Objective 2

# Gathering of use cases

- Deliverable:
  - Set of use cases (in a standardised format) to drive both the development, as well as the showcasing of SNOMED CT in Genomic Medicine
- Action plan:
  - Deliver use case gathering workshops with the members of the SNOMED CT subject matter expert group, and document specified use cases
  - Iteratively refine use cases based on findings emerging from O3
  - Publish and disseminate use cases on the website
  - Disseminate use cases at key academic / industry clinical genomics events
- Measure of success:
  - Number of documented use cases
  - Coverage of the domain captured by the use cases

Objective 3

# Restructure domain knowledge around clinical genomics in SNOMED CT

- Deliverable
  - Implemented and documented Genomics components in SNOMED CT
- Approach
  - Community building
  - Practical development
  - Outreach
- Measure of success:
  - Number of use cases successfully validated based on the new model

Objective 3

# Restructure domain knowledge around clinical genomics in SNOMED CT – Step 1

- Community building:
  - Select and interact with a group of key experts through direct contact
  - Form a SNOMED CT subject matter expert group consisting of both external, as well as internal SNOMED experts / editors
  - Develop a quality assurance process through expert feedback loops and iterative knowledge enrichment

Objective 3

# Restructure domain knowledge around clinical genomics in SNOMED CT – Step 2

- Practical development:
  - All content change will be driven by clearly documented use cases, reviewing the clinical and genomics perspectives
  - Incrementally 'build' the Genomics component of SNOMED CT:
    - Restructure 'Genetic finding'
    - Assess and fill-in gaps in the clinical space (familial context, report, genetic test)
    - Align phenotype modeling approach to current research and development methodologies
    - Create a subset of clinically-relevant exposures
    - Provide support for annotations (disease-phenotypes; genomic data - exposures; genomic data – phenotypes)

Objective 3

# Restructure domain knowledge around clinical genomics in SNOMED CT – Step 3

- Outreach:
  - Liaise with prospective external knowledge sources (e.g., GO, NextProt) cross-reference mechanisms
  - Create a mechanism for integration of community feed-back, provenance and micro-attribution
  - Develop long-term sustainability and maintenance plan

Objective 3

# Focus on Clinical Utility

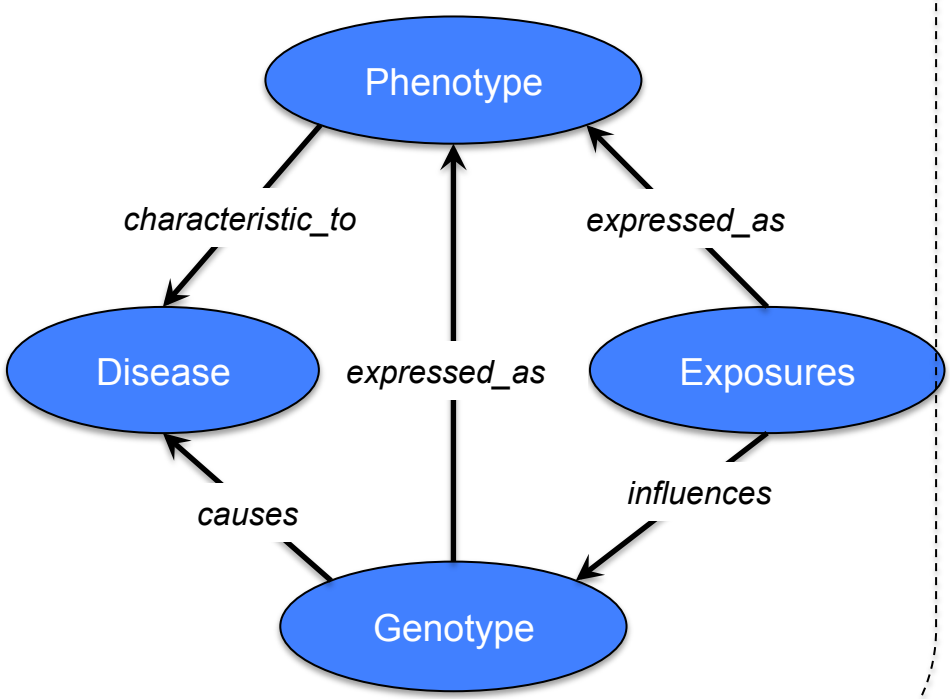
SNOMED International

**Clinical space**

- Familial context
- Diagnosis
- Genetic Testing
- Clinical Trials

**Clinical Utility**

**Knowledge space**



Objective 4

# Showcasing added value

- Deliverable
  - Tool / application that demonstrates the value added by SNOMED CT in Genomic Medicine
- Action plan:
  - Select exemplary use case to be implemented in the application
  - Develop application in collaboration with both the experts and stakeholders groups (e.g., using SMART on FHIR)
  - Validate application with relevant stakeholders use case
  - Use tool for presentations and conference globally
  - Disseminate and make tool freely available to the public
  - Pilot sites
- Measure of success:
  - Application is operational and accessible
  - Chosen use case is validated by the application
  - Number of accesses of the application

Objective 4

# Showcasing added value – Pilot sites

- Focus
  - Use case driven
  - Use of EHR information in Genomics
  - Inter genomics information transfer
  - Support for Rare Disease databases
- Size
  - Pilot size will be based on use case requirements, on a case to case basis
- Funding
  - Explore access to Genomics funding

Objective 5

# Raising awareness

- Deliverable:
  - Web presence (e.g., documentation, forum for discussions, downloadable materials) around the Genomic component of SNOMED CT
  - Calendar identifying key dissemination opportunities
- Action plan:
  - Showcase the capabilities of SNOMED CT via the use cases
  - Develop and deliver showcasing events that feature members of the SNOMED CT stakeholders consultancy group, as well as experts from the SNOMED CT experts group
  - Develop and disseminate materials describing and exemplifying the Genomic component of SNOMED CT
  - Develop and deliver hands-on workshops / tutorials / sessions at key clinical genomics events - e.g., ASHG, ACMG, ESHG
- Measure of success:
  - Number of attendees at the various events
  - Number of users accessing materials

Objective 5

# Enabling partnerships

- Community building
  - Clinical reference group
  - Project groups focused on specific Genomics related deliverables
- Cross pollination to enable knowledge transfer and realization of a common vision
  - Research initiatives
  - Clinical/Research institutes
  - Industry leaders
- Using past and current experience to deliver the future

# Next steps

- Community building
  - Stakeholders
    - Identify stakeholders/stakeholder groups
    - Stakeholder engagement
  - External resources
    - Identify resources that can be leveraged to support the development
  - Internal working groups
    - Subject matter expert group (expert input)
    - Genomics clinical reference group (community engagement)
- Working group / task force creation
  - Goals
  - Action plan
  - Taking ownership of the strategy
- Implementation
  - Development

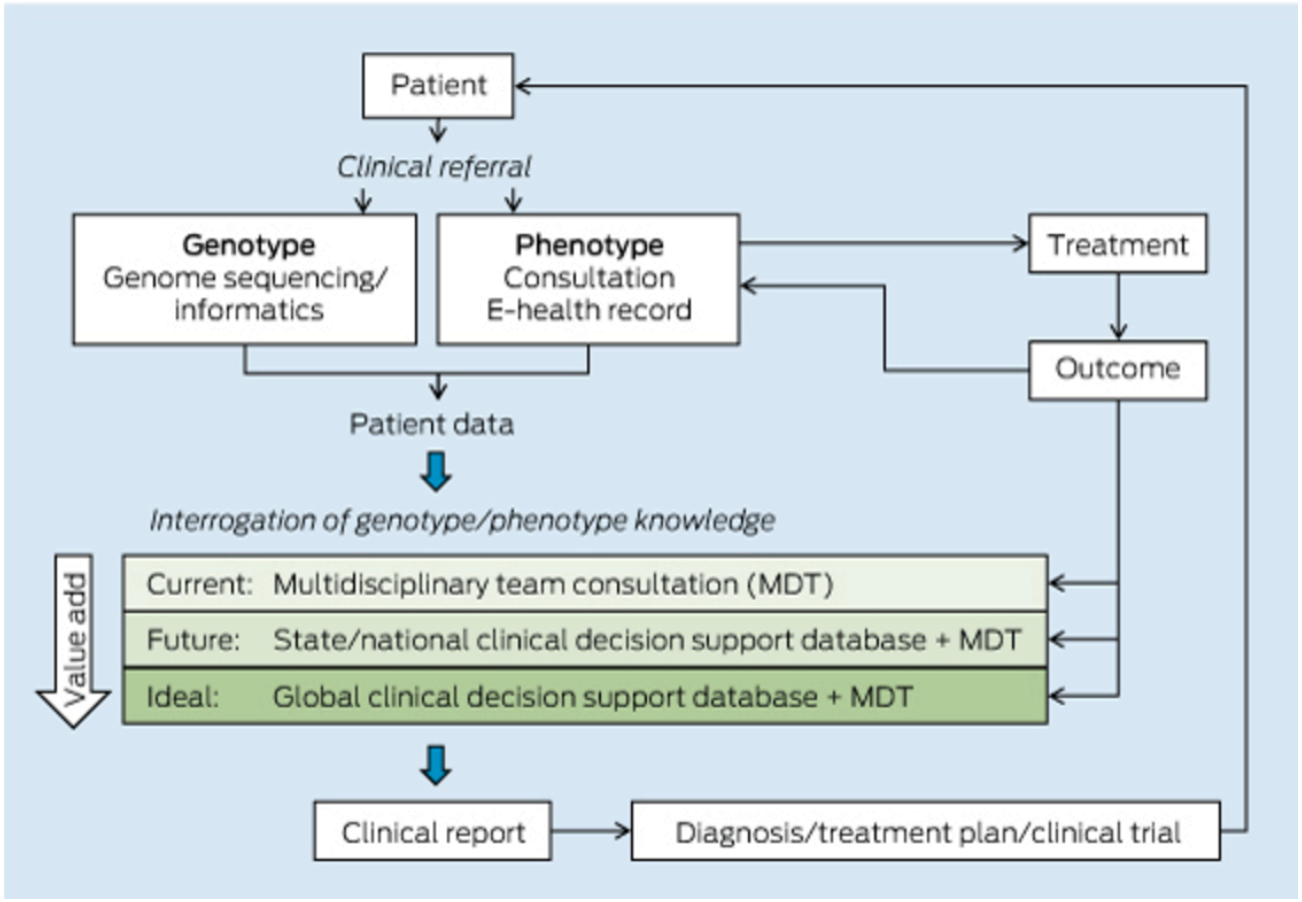
# Next steps - 2017 deliverables

- Agreed strategy in place
- Agreed work plan in place
  - Detailed work plan for 2017
  - Work plan for 1-3 years
- Engagement activities
  - Genomics SME group
  - Genomics Clinical Reference Group
- Pilot site - identify and establish
- Detailed requirements for the representation of phenotypic data within SNOMED CT
- Detailed requirements for the linkage of SNOMED CT to external global phenotype terminologies
- White paper on SNOMED CT and Precision Medicine

# The future - Developing Genomic Medicine to support Precision Medicine

- White paper on SNOMED CT and Precision medicine
  - Delivery Q3 2017
- Review requirements for Genomics information to feed into EHR's
  - Completeness of concepts within SNOMED CT
  - Review/updating of terming in line with Genomics led changes
  - Analytics requirements
  - Decision support requirements based on the implementation of precision medicine
- Access to emerging knowledge
  - Links to national Precision Medicine programmes, e.g. US, UK

# Summary: Imagine the future




**Standardized / integrated terminology**