Genomics and SNOMED CT (the way ahead)

Ian Green
Customer Relations Lead, Europe and Clinical Engagement
Business Manager
The changing face of medicine
The Genomics standards landscape
Accessing clinical data
Provision of genomics data within EHR’s
Where SNOMED CT fits?
Engaging the community
Future SNOMED CT developments
Precision medicine is "an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person."

*Precision Medicine Initiative*
Traditional to precision medicine

- Working diagnosis – treatment based on acknowledged “best practice” based on research trials
  - Treatment based on trial and error at an individual patient level
  - Evidence-based medicine
    - Clinical guidelines
    - Treatment pathways

- Future emphasis will move to the individual patient
  - Genetic profile – sequencing data
  - Personalized treatment based on the individual genetic profile
  - Leverages traditional medical knowledge, with additional genetic insight
  - Requires a robust system to support information exchange, underpinned by clinical terminology
Genomics landscape

Classifications/Terminologies
- Orphanet
- HPO
- OMIM
- Disease Ontology

Specialist Organisations
- NCI
- IARC
- Sangar Institute
- National Human Genome Research Institute (NHGRI)

National Initiatives
- Genomics England
- Australian Genomics Initiatives
- Genome Canada
- European Alliance for Personalised Medicine (EAPM)
Genomics community - Accessing clinical data

- Currently limited to national commissioning datasets (classification data, e.g. ICD-10)
  - Limited to diagnosis and procedures
- Requirement is for detailed knowledge, to include clinical, social and environmental aspects – essential to deliver precision medicine in the future
- Links to pharmacogenomics
  - Access to SNOMED CT International Release (provide internationally shareable data), and specific national drug extensions linked to SNOMED CT (specific national drug availability)
  - Robust linkages between the SNOMED CT International Release and national drug extensions
  - Linkages to substance and product hierarchies within SNOMED CT
- Requirement to “follow-up” cases longitudinally, to view changes in treatment and outcomes (support for ongoing clinical research activities
- Requirement for agreed record standards to ensure the contextual meaning of the patient record is not lost
Fundamental requirement for Precision Medicine

Requirement for new “genomic” SNOMED CT content

SNOMED CT must align with new genomics definitions of disease (definitions and terming)

Links to existing genomics classifications and terminologies need to be provided and preserved

Requirement for the implementation of international shareable treatment protocols (requires a standard clinical terminology)
  - Must link to pharmacogenomics developments and product availability

Genomics requirements are global in nature, and to support these in SNOMED CT requires a collaborative effort
Where does SNOMED CT fit?

- Provides terminology to support Genomics with the EHR
- Provides the linkage between the EHR and Genomics requirements
- Updated SNOMED CT content to reflect Genomics evolving requirements
- Derivative product development to provide computable linkages between SNOMED CT and Genomic classifications/terminologies
- Provides detailed clinical information from EHR to inform Genomics research. This requirement will review the semantics within and HER system to ensure that the context is not lost
- Provides Genomics content to support Precision Medicine initiatives at a global/national level
Engaging with the clinical community

- SME Genomics group – representatives from the international genomics community, national Genomics initiatives and large global Genomics organisations
- Genomics Clinical Reference Group – to engage the SNOMED CT community and provide a focus for Genomics discussions
- Individual engagement activities linked to specific Genomics product types/organisations, driven by use case requirements e.g. HPO and OMIM
- Development of SNOMED CT/Genomics pilots (early adoption sites) to explore/showcase functionality and to refine requirements
- SNOMED International must be open to dialogue with the Genomics community and be willing to work with them to ensure that SNOMED CT remains fit for purpose
Future SNOMED CT developments

- Fundamental – SNOMED CT will not look to incorporate all Genomic content from existing genomic classification/terminologies. The requirements will be driven by discussions with the Genomics community.
- Focus of developments based on Orphanet, HPO, OMIM and Disease Ontology.
- Review of existing SNOMED CT content, and authoring/updating of content.
- Expansion of SNOMED CT concept model to support Genomics content.
- Development of derivative products (maps) where there is a clear requirement to do so.
- Working with HL7 to ensure SNOMED CT is represented within messaging protocols (HL7 FHIR).