# Genomics and SNOMED CT (the way ahead)

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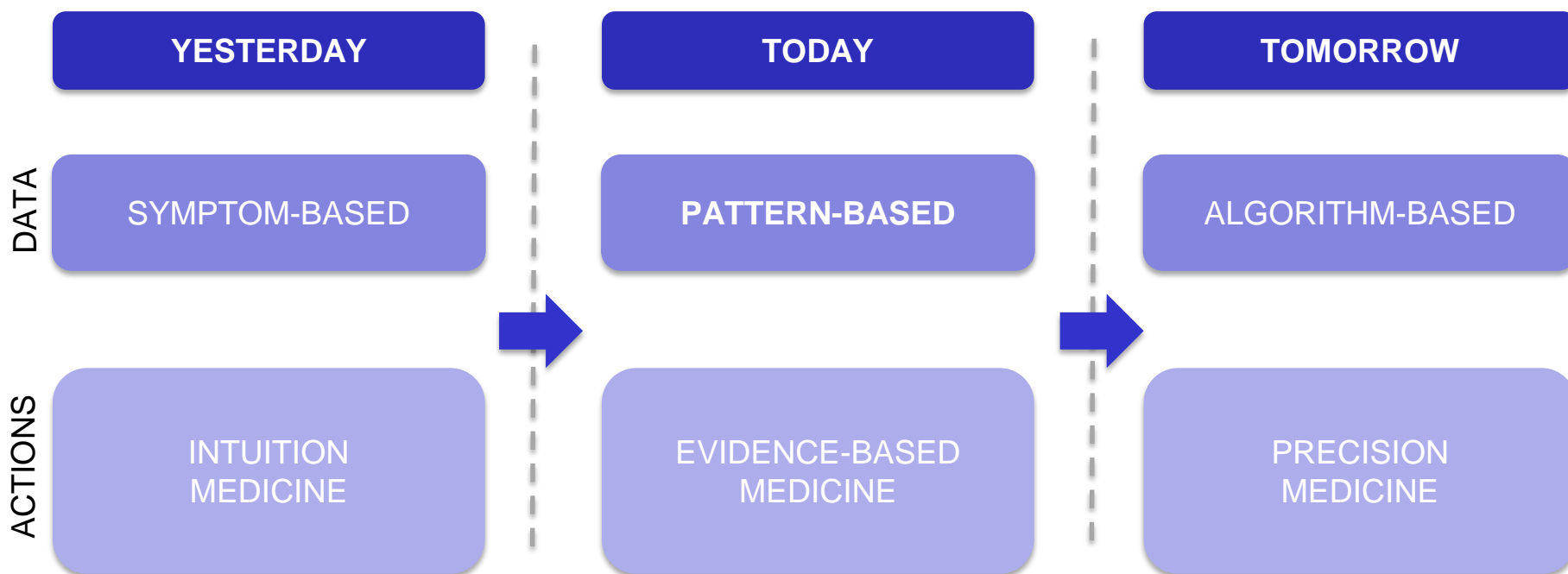Customer Relations Lead, Europe and Clinical Engagement Business Manager

Delivering
**SNOMED CT**
The global language of healthcare

# Agenda

- 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



| | YESTERDAY | TODAY | TOMORROW |
|---|---|---|---|
| **DATA** | SYMPTOM-BASED | **PATTERN-BASED** | ALGORITHM-BASED |
| **ACTIONS** | INTUITION MEDICINE | EVIDENCE-BASED MEDICINE | PRECISION MEDICINE |

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

Global Alliance For Genomics and Health (GA4GH)

MONARCH Initiative

European Bioinformatics Institute

## 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

# Provision of genomics data within EHRs

- 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 exisiting 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
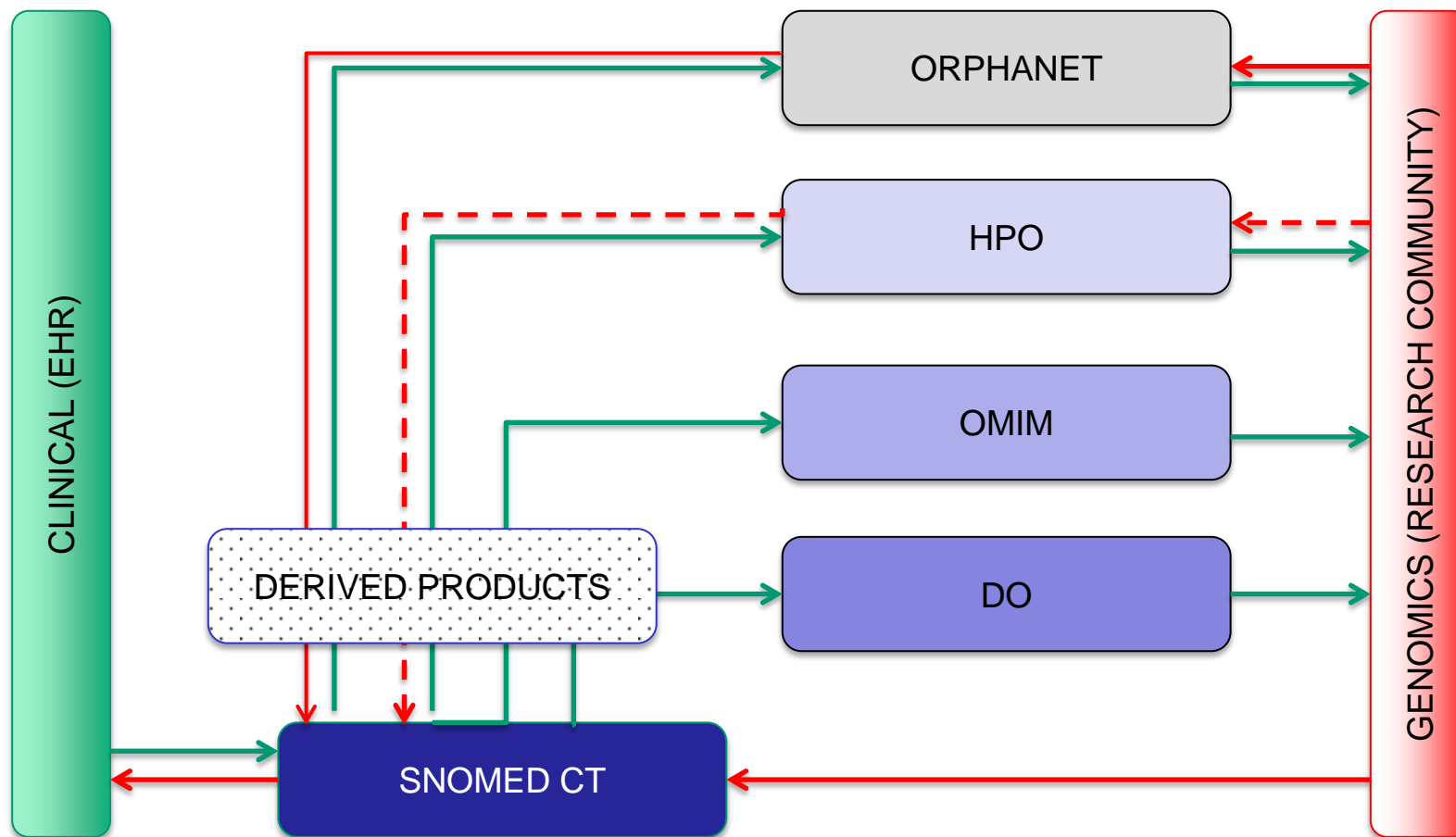
# Engaging with the clinical community

- 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)

# Possible future state

# Questions