Genomics and Precision Medicine Clinical Reference Group (CRG)

Clinical Reference Group (CRG) Scope

The scope of the CRG initially is intentionally broad, to reflect the broad range of subspecialties included within Genomics and Precision Medicine. Over time, as the groups understanding of how SNOMED CT can support these areas, the group may choose to create subgroups focused on specific areas of interest. The group will focus on the usability of SNOMED CT for the representation of clinical data to support both clinical care and research in the application of genomics.

SNOMED International strategy for Genomics

Ian Green
Customer Relations Lead, Europe &

Clinical Engagement Business Manager

SNOMED CT EXPO 2020
Virtual Conference October 8-9

SNOMED International

SNOMED CT and Genomic Medicine

"Working together towards a common strategy"

Aim:

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)

Strategy scope and purpose

Genomics promises to change the way that medical care is delivered across the globe. These changes will be delivered through the implementation of precision/personalized 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.

SCOPE

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.

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
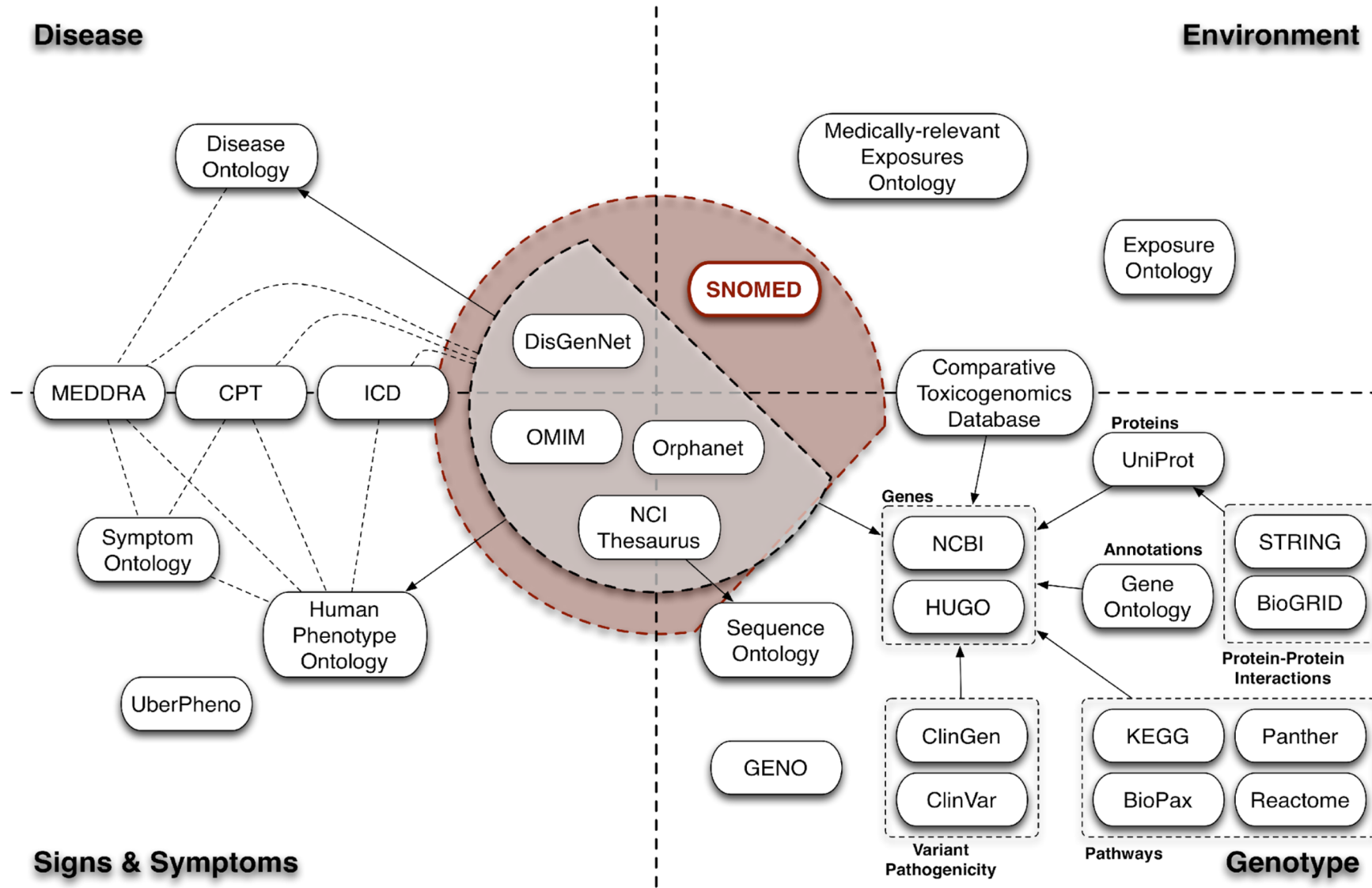
Patient journey in Genomic Medicine
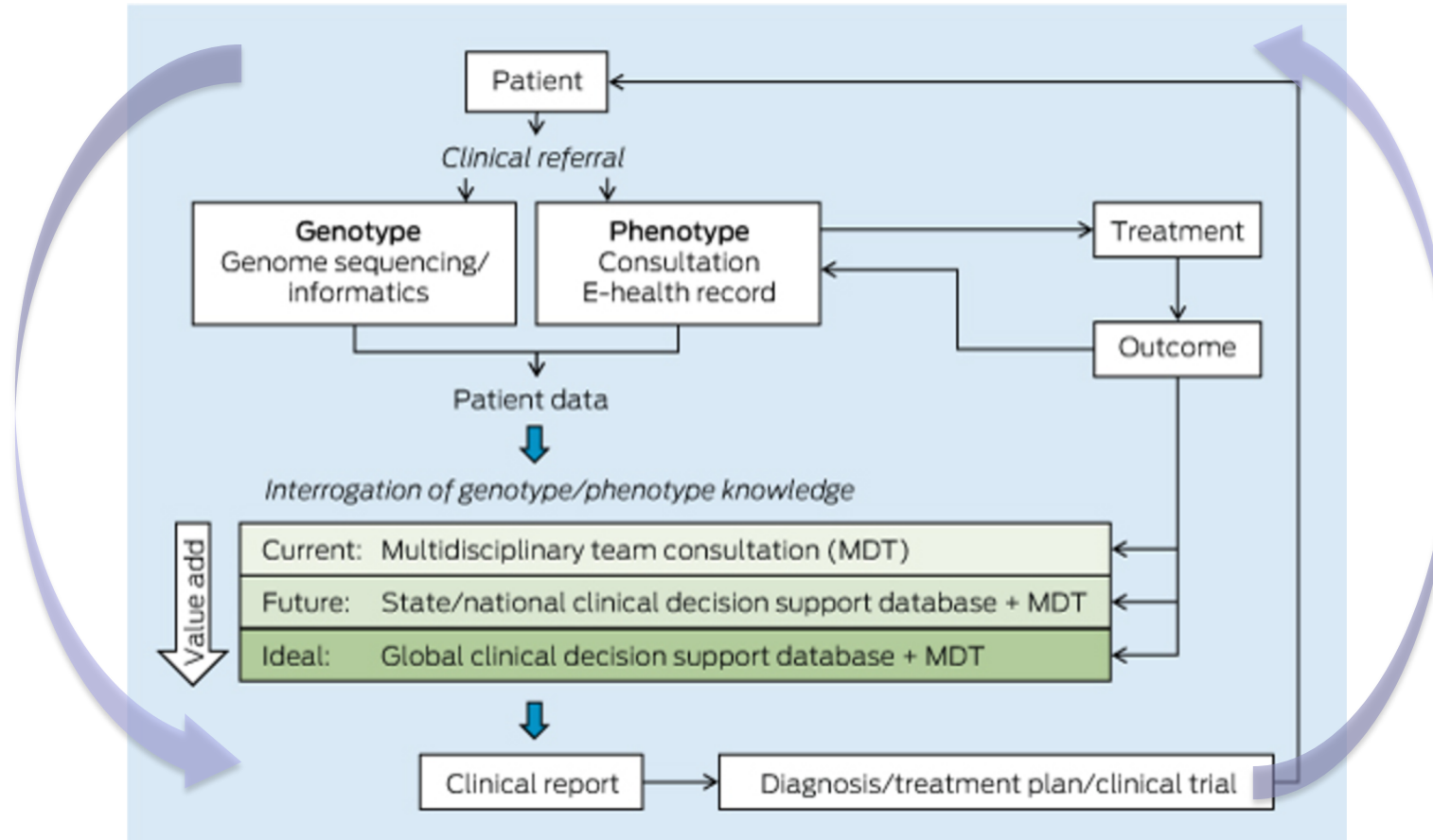
- Individual genomic data
- Accurate clinical data
- Alignment to the existing body of knowledge

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.

Where does SNOMED CT fit into Genomic Medicine

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.

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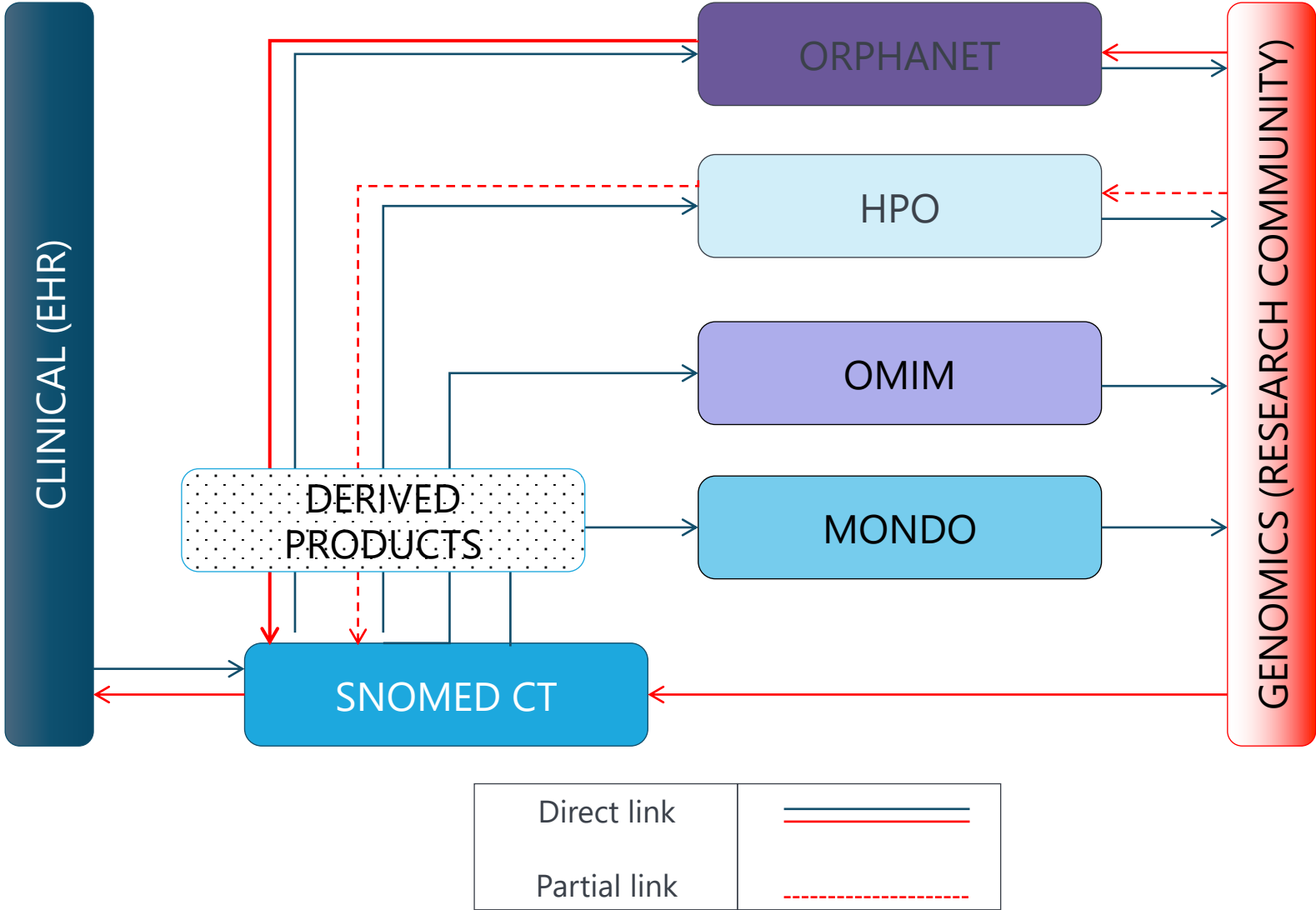
Technical Desiderata for the Integration of Genomic Data into Electronic Health Records.
Biomed Inform . 2012 June ; 45(3): 419–422. doi:10.1016/j.jbi.2011.12.005.

Future SNOMED CT developments
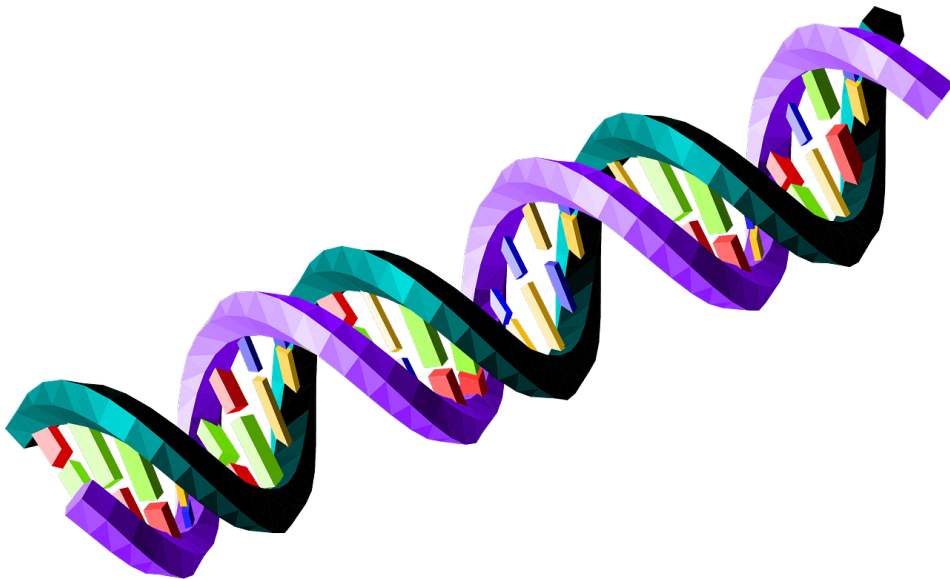
- Fundamental – SNOMED CT **will not** look to incorporate all Genomic content from existing genomic classification/terminologies. The requirements will be driven by discussions with the Genomics community

- Focus of developments based on Orphanet, HPO, OMIM and MONDO

- 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

CLINICAL (EHR)

ORPHANET

HPO

OMIM

DERIVED PRODUCTS

MONDO

SNOMED CT

GENOMICS (RESEARCH COMMUNITY)

Direct link	———————
Partial link	-------------------

SNOMED CT
EXPO 2020
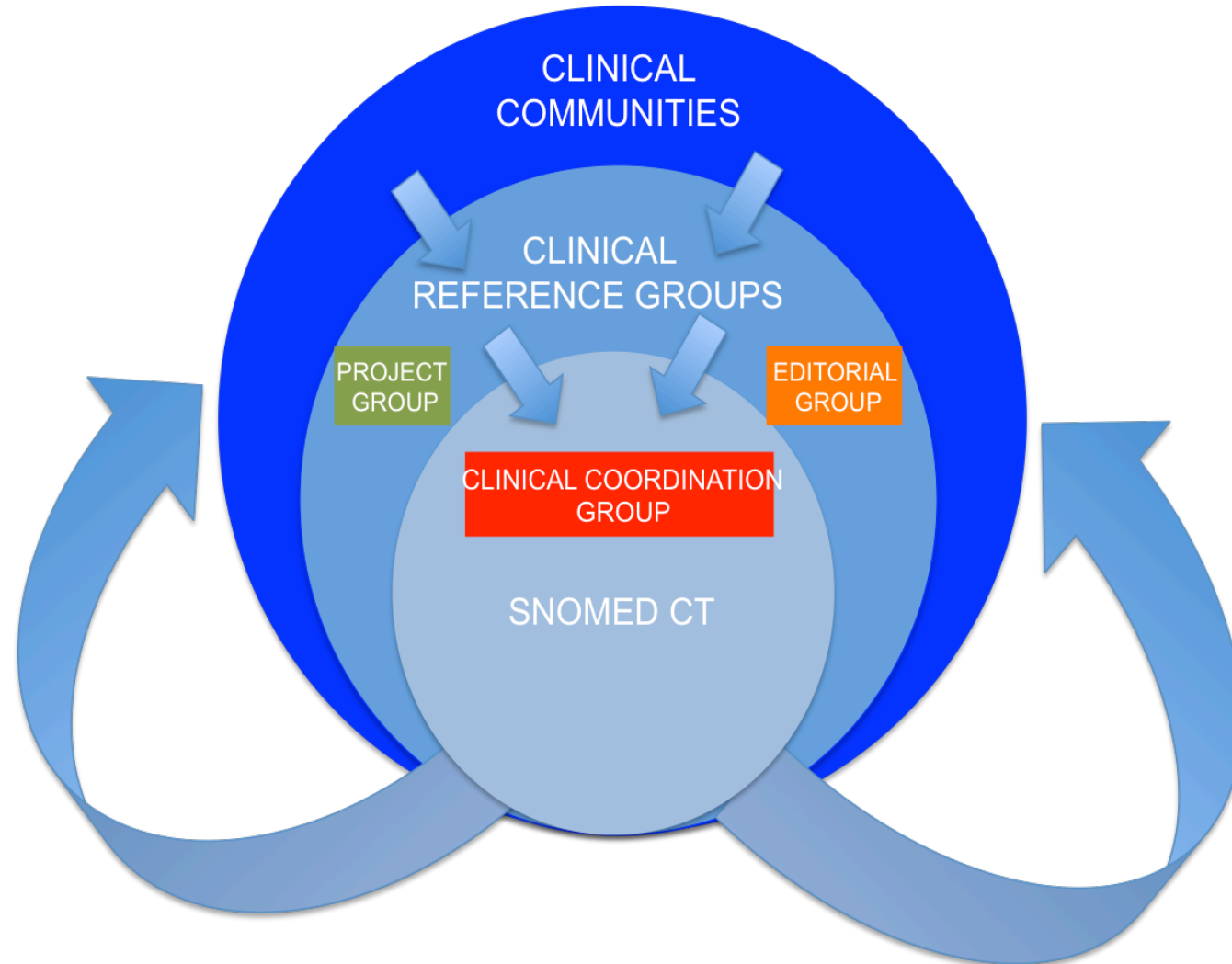Virtual Conference October 8-9

Genomics and Precision Medicine CRG

Jane Millar
Collaboration Lead

Clinical Engagement model

Clinical reference Groups (CRGs)

- Facilitate discussion between clinicians focused on specific clinical specialties or topic areas

- Provide a platform for clinicians to discuss with colleagues any questions, issues and experience relating to SNOMED CT and its implementation

- Dedicated Confluence area and zoom accounts

- Clinical community leadership

- Open to all, input not limited to just clinicians from the clinical specialty

- Input from SNOMED CT author and access to SNOMED CT change request mechanism where appropriate

What is the reality of CRG's?

CRG's provide the following:

- A framework for multi-disciplinary topic discussion and working across professions

- Global perspective, especially required for SNOMED CT international edition

- Access to expertise from all clinical domains

- A mechanism for different levels of engagement

- Access to discussions on clinical developments by other groups (e.g. implementers), to ensure that products are developed meet clinical requirements.

- Early identification of global changes to healthcare delivery

Clinical Project Groups and Editorial Groups

Clinical project groups

- Task and finish groups to address specific areas of clinical content agreed as part of work plan

- Review of existing content and identification of changes/additions to the international release

- International requirements, led by external SME

- Input from a SNOMED CT author

Editorial groups (where relevant)

- To provide editorial review of derivative products e.g. reference sets and maps

https://confluence.ihtsdotools.org/display/CP/Directory+-+Clinical+Reference+Groups

Directory - Clinical Reference Groups

Created by Ian Green, last modified on 2020-May-07

The clinical resource groups are listed below in alphabetical order, and are listed by specialty. Each group is linked by hyperlink to the specific confluence pages belonging to that group. Please use the links provided to access the clinical reference group required.

> ⓘ **Suggest a new Clinical Reference Group**
> If you have a requirement for a new Clinical Reference Group, please email the details to crg@snomed.org, and mark the query Clinical Reference Groups

> ⓘ **General queries**
> For general queries relating to Clinical Reference Groups or Clinical Engagement, please email crg@snomed.org

ABC

Allergies Hypersensitivity and Intolerance

Anaesthesia

DEF

Dentistry

GHI

Genomics and Precision Medicine

General practice/Family practice

JKL

MNO

Mental and Behavioural Health

Nursing

PQR

Pathology and Laboratory Medicine

STU

VWXYZ

Active Project Groups

Cancer Synoptic Reporting Project Group   Diabetes Project Group   ICNP 2019 Alignment Project Group

Nutrition Care Process Terminology Clinical Project Group

Clinical Expert Project Groups

Epilepsy Clinical Expert Project Group

Supporting SNOMED International Project Groups

Devices Project   Drug Project   Event, Condition and Episode Model Project

Observable and Investigation Model Project Group   Organism & Infectious Disease Model Project   SNOMED CT Languages Project Group

ICNP 2019 Alignment Project Group

Editorial Groups

Dentistry Derivatives Editorial Group   GP/FP Derivatives Editorial Group   Nursing Derivatives Editorial Group

Genomics and Precision Medicine Clinical Reference Group

Scope:

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Key points for discussion

- Leadership of CRG – normally 2 persons from different backgrounds and parts of globe

- Membership of CRG:

  ➤ Organisations

  ➤ Individuals

- Key topics to start group

  ➤ Ways of working:

  ➤ Confluence and zoom
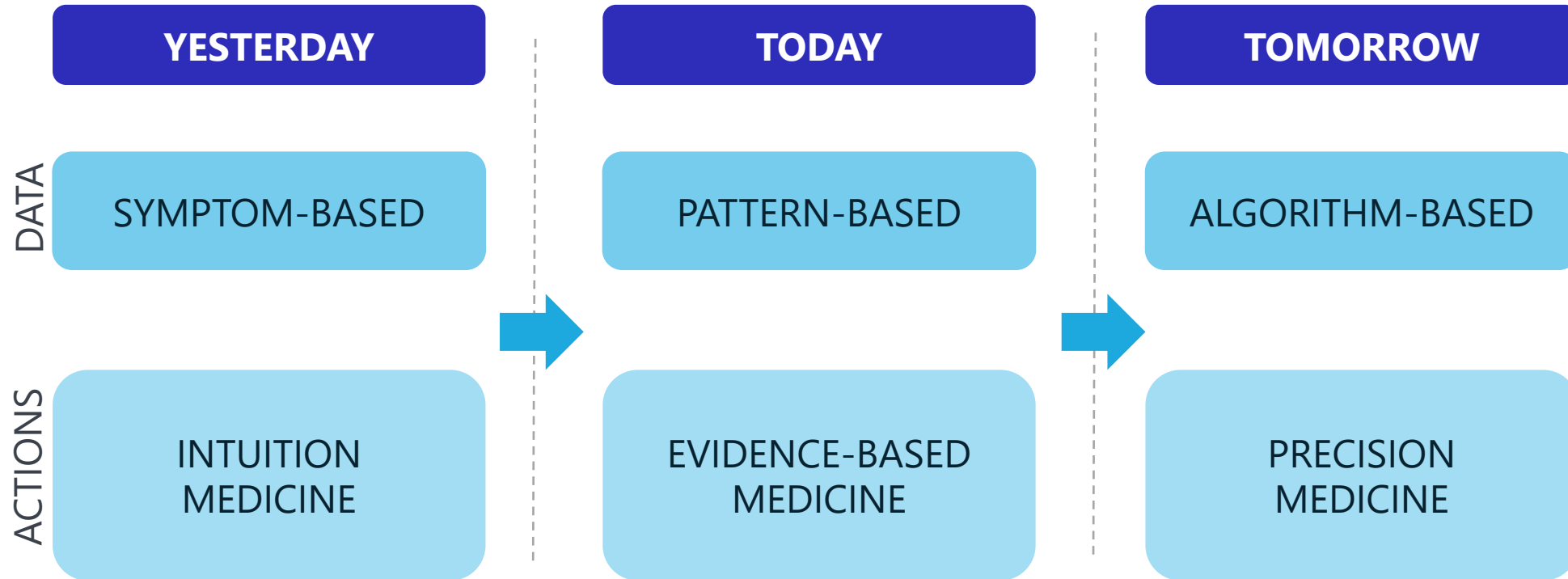
  ➤ Meeting schedule

  ➤ Group support

SNOMED International

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Precision Medicine

Ian Green
Customer Relations Lead, Europe &

Clinical Engagement Business Manager

SNOMED International

Precision Medicine

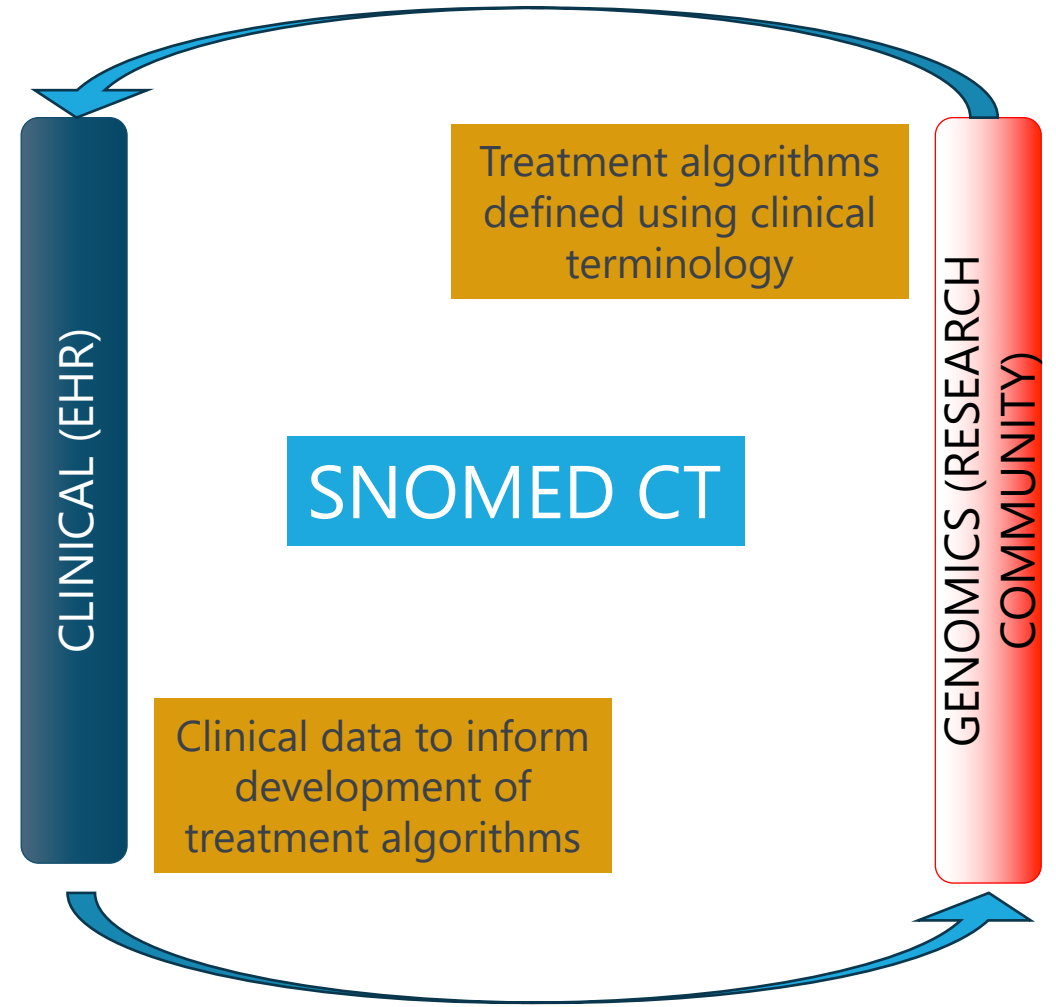|	YESTERDAY	TODAY	TOMORROW
DATA	SYMPTOM-BASED	PATTERN-BASED	ALGORITHM-BASED
ACTIONS	INTUITION MEDICINE	EVIDENCE-BASED MEDICINE	PRECISION MEDICINE

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

Where does SNOMED CT fit with Precision Medicine ?

CLINICAL (EHR)

Treatment algorithms defined using clinical terminology

SNOMED CT

GENOMICS (RESEARCH COMMUNITY)

Clinical data to inform development of treatment algorithms

Next steps

- SNOMED International is looking to document and publish, where and how SNOMED CT can be used to support the implementation of precision medicine

- Forming a SME group to advise on the development of the document

- Timescale for group formation is by end of year

- Target date for publication – April 2021

Possible sections to be included in the document

- Provision of clinical data from genomic researchers

- Extension of clinical data in SNOMED CT where required

- Provision of metadata to support Genomic workflows within SNOMED CT

- Use of "mappings" between SNOMED CT and genomic terminologies

- Support for genomic researchers to define treatment algorithms using terminology/ontology principles

- Specification of treatment algorithms using SNOMED CT

- Incorporation of treatment algorithms in decision support systems

ORPHANET project

Jane Millar
Collaboration Lead

Project summary

- Collaboration between SNOMED International and INSERM (Institut national de la santé et de la recherche médicale - French National Institute of Health and Medical Research

- Started in 2015.

- Focus – a map between SNOMED CT and a <u>priority set</u> of Rare Diseases in Orphanet

- Creation of content in SNOMED CT where a gap identified and subsequent map

- Requirement for validated definitions for Orphanet content – essential for hierarchy positioning in SNOMED CT and therefore analytics.

Where are we now?

- Completed agreed content work

  ➢ 3,533 new Rare Diseases added

- Map produced and quality assured

  ➢ 5,690 maps

  ➢ Exact matches semantically

- Alpha release of SNOMED CT to Orphanet map, May 2020 based on January 2020 releases

  ➢ SNOMED CT RF2 format

  ➢ Spreadsheet format

  ▪ Feedback on Alpha release invited to end November 2020.

Next steps

- Collate feedback on quality issues related to the map

- Agree work to be done to update to July 2021 release of SNOMED CT and Orphanet (same scope of content as Alpha release), including management of any inactivations

- Production release of SNOMED CT to Orphanet map – October 2021

- Create maintenance and updating processes to produce an annual release of the maps, including how to prioritise the addition of content/maps not included in the released map

Further work with INSERM

Annual release of SNOMED CT to Orphanet map for Rare Diseases

➢ Updates based on changes in underlying terminologies

➢ Requirements from users

Proposal (for discussion by group):

➢ Sub group of Genomics and Precision Medicine CRG

➢ Identify gaps in map based on usage

➢ Include other stakeholders such as those responsible for registries

➢ Develop ways of collating requirements in conjunction with INSERM

Working with the Genomics community

Ian Green
Customer Relations Lead,
Europe &

Clinical Engagement
Business Manager
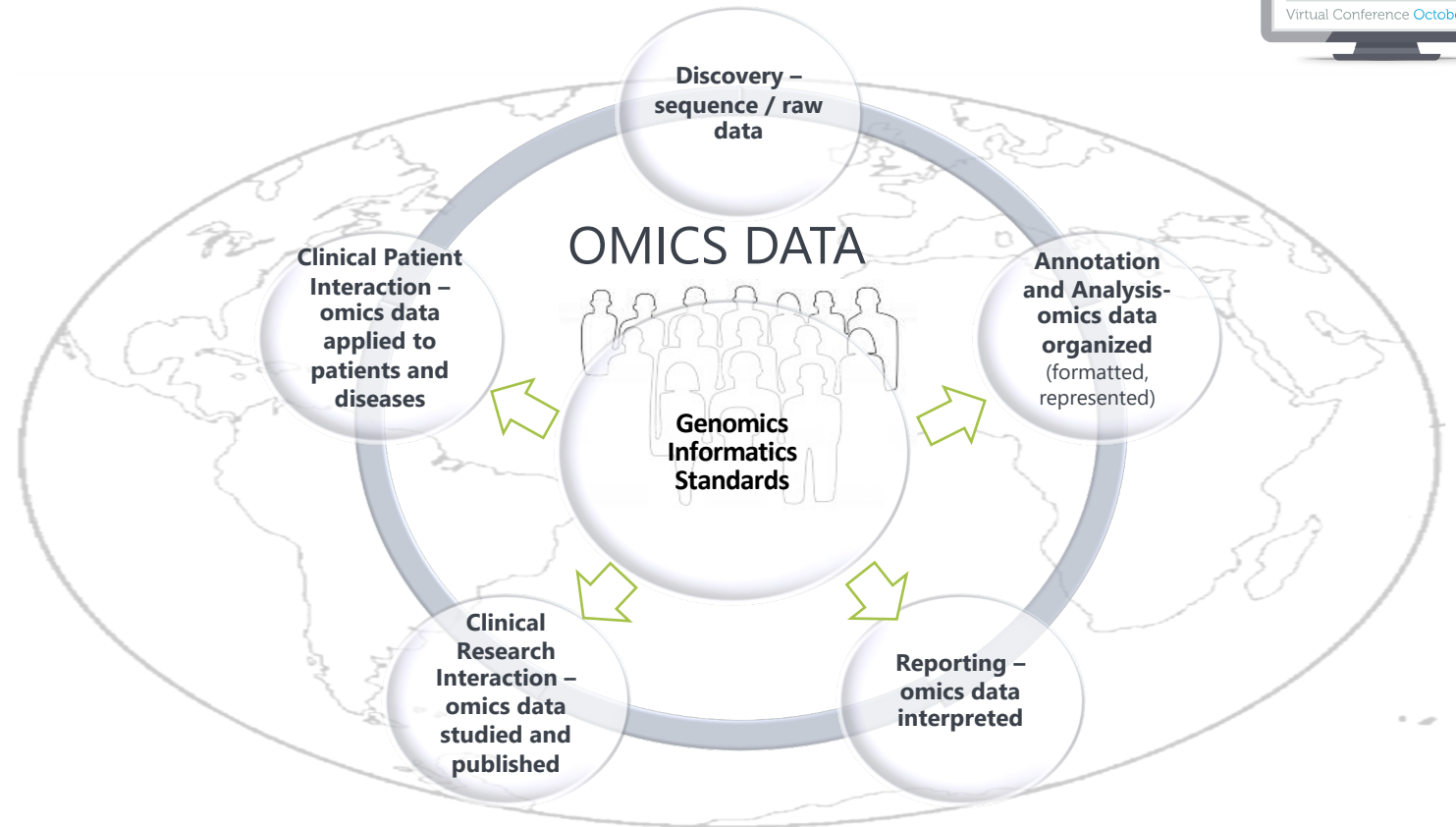
GA4GH
(Global Alliance for Genomics and Health)

The **Global Alliance for Genomics and Health** (GA4GH) is an international, non-profit alliance formed in 2013 to accelerate the potential of research and medicine to advance human health. Bringing together 500+ leading organizations working in healthcare, research, patient advocacy, life science, and information technology, the GA4GH community is working together to create frameworks and standards to enable the responsible, voluntary, and secure sharing of genomic and health-related data.

- Working directly with the clinical and phenotype workstream to ensure new global standards are included into SNOMED CT, and discussion and developments internationally include a SNOMED CT perspective

ISO TC215 SC1

OMICS DATA

Discovery – sequence / raw data

Annotation and Analysis- omics data organized (formatted, represented)

Clinical Patient Interaction – omics data applied to patients and diseases

Genomics Informatics Standards

Reporting – omics data interpreted

Clinical Research Interaction – omics data studied and published

Standardization of computable data, information, and knowledge, including their representation and metadata, for the application of omics, including but not limited to genomics, phenomics and proteomics to support human health and clinical research

Genomic terminologies

HPO (Human Phenotype Ontology)

- Working to provide additional SNOMED CT concepts and development of a SNOMED CT to HPO map

OMIM (Online Mendelian Inheritance of Man)

- Working to provide additional SNOMED CT concepts and development of a SNOMED CT to OMIM map

MONDO

- Use of diagnosis definition to inform the enhancement of SNOMED CT disorder contents (concept model development)

Gene ontology

- Provision of linkages to assist genomic focused logical definition of concepts within SNOMED CT

Phenotype authoring project

Authoring of clinical phenotypes

- Project due to complete in February 2021

- Authoring is being undertaken by external contractor

- 4,000 concepts

- All modelled and aligned to the July 2020 release

- Quality assurance is being undertaken by SNOMED International authoring team

- Published in the community area, with its own moduleId

- Will deliver a considerable addition of content which is focused on specific clinical specialist areas

Future phenotype content developments

- 4,000 concepts will align with future work with HPO and OMIM

- Will be included in the scope of any future mapping work with genomic terminologies

- Publishing in the community area will provide a mechanism for community review and feedback on the content

- Future content development will build on the experience of this initial project, and may well include a larger group of genomic/phenotype related content based on use case requirements

Getting involved

Discussion

"Getting involved"

- What are the next steps ?

- Formation of the CRG ?

- Identification of a chair for the group ?

- Meeting schedule ?

- Initial topics for discussion