Delivering
SNOMED CT
The global language of healthcare

SNOMED International
Leading healthcare terminology, worldwide

Genomics Update and Genomics Pilot(s)

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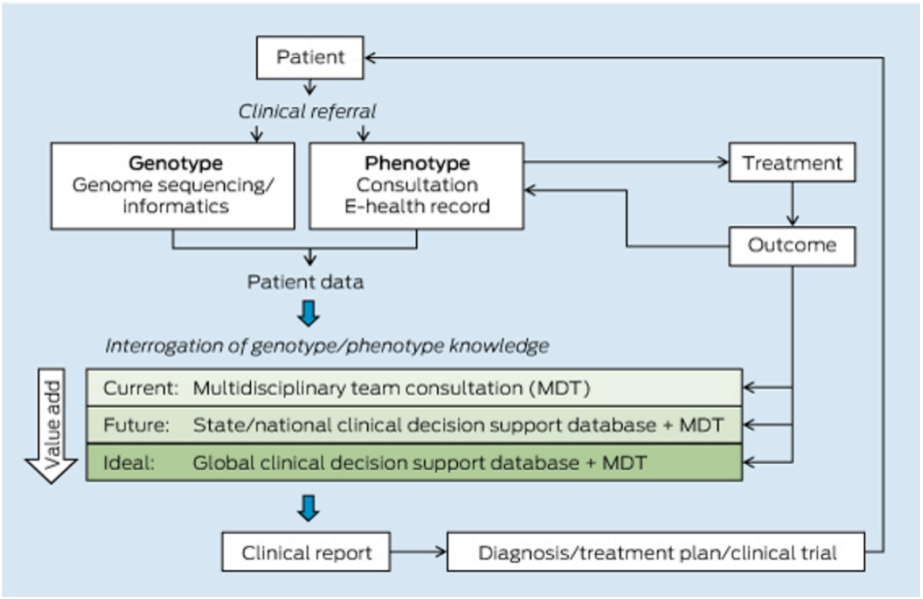
30th, October 2019

Genomics Strategy

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)



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.

Clarity of vision

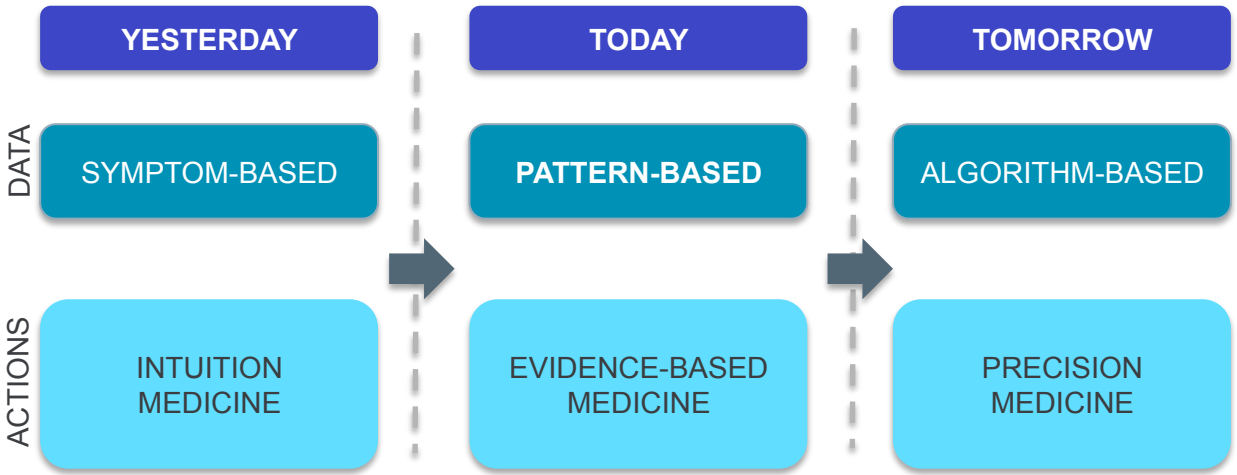
- SNOMED CT will not look to include content from all Genomic terminologies, but rather to align to those with a clear link to clinical practice through the EHR

- 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

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

Genomic Pilots

- Strategy driven – proof of concept
- Focus on the use of SNOMED CT phenotype data within EHR systems
- Two pilot sites (both based in Australia)
    1. Genetic Kidney Disease (Queensland)
    2. Cardiovascular Disease (Australia)
- Focus – End-to-end, data capture and data analytics
    - ✓ Use and reuse of SNOMED CT data natively

Genomic Pilot (Genetic Kidney Disease)

Scope

Phase 1

Work with Flagship clinicians to replace the current free-text fields used to capture patient diagnosis at different stages with SNOMED CT coded fields, using the new REDCap FHIR Terminology plugin

Migrating existing free text data to the new coded fields and discuss complex cases with Flagship clinicians[1]

Phase 2

Define an analytics use case around the evolution of patients' diagnoses to help understand to what extent the diagnoses are refined at every encounter (e.g., at referral, at the clinic, etc.) and provide insights into how to optimise the current process

Implement the use case in the CSIRO analytics platform to validate its design

Genomic Pilot (Genetic Kidney Disease)

Deliverables

- A FHIR value set of SNOMED CT codes for the KidGen Flagship
- A report describing the process of migrating the REDCap forms to use SNOMED CT to represent the diagnoses of patients in coded form, at different points during the patient journey
    - The report will identify any challenges encountered in this process including complex cases and any gaps or modelling issues identified in SNOMED CT
- A report detailing how SNOMED CT and FHIR can be used as part of an analytics platform and how the KidGen data was used to implement an example around the evolution of patients' diagnoses

Output and lessons from the pilot

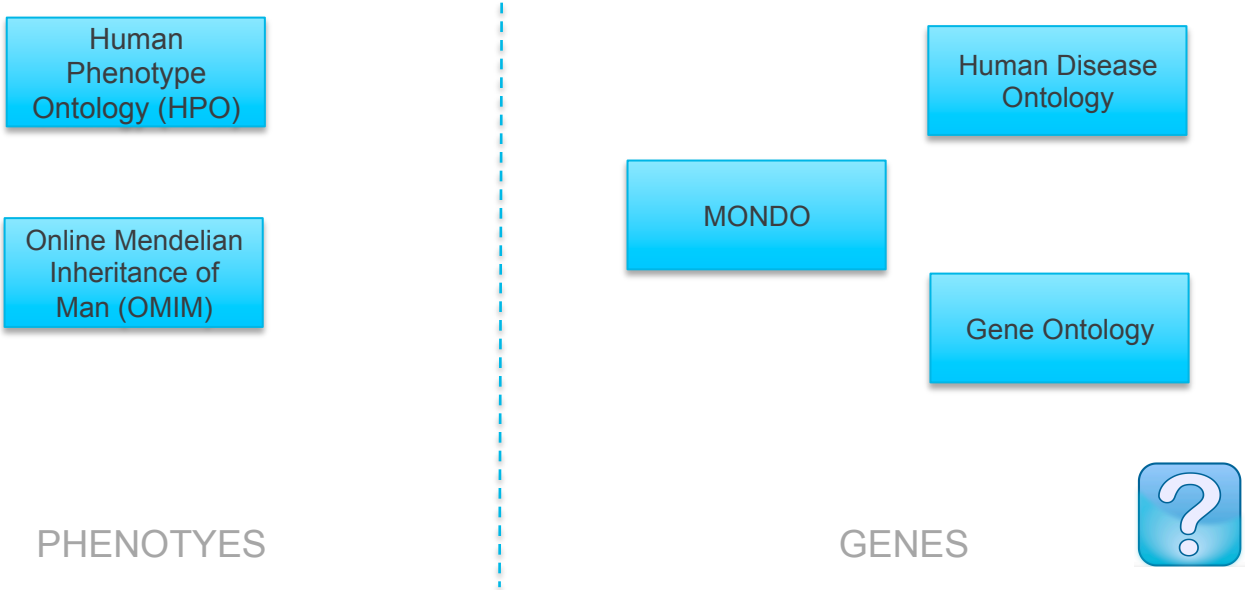
- Can utilise Patient data from EMR systems with SNOMED on-board
- Can serve discrete genomics use cases
- SNOMED CT Expression Constraint Language
- Serves data capture and analytics (inputs and outputs)
- Formal, programmable, re-producible
- Little manual handling
- Documented methods, techniques and tools
- Extensible, maintainable
- Compatible with FHIR
    - FHIR Value set (SNOMED CT phenotypes)

Going forwards

USE CASES

1. Supporting genomics researchers to access of SNOMED EHR data
2. Supporting definition of precision medicine algorithms for use in EHR systems
3. Development of SNOMED disease definitions in line with genomic definitions of disease

Going forwards - approaches



Please contact Ian Green…. igr@snomed.org for further information.