Implementing Genomics in EHR's – present and future

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Ian Green, Customer Relations Lead, Europe, SNOMED International

HL7 International

Global Alliance for Genomics & Health

SNOMED International

Conflict of Interest

Ian Green, MSc

Has no real or apparent conflicts of interest to report.

Agenda

- Current situation and requirements
- Use cases
- Working together – GA4GH/HL7/SNOMED International
- Bringing together clinical and genomics data
- Next steps
- The future view

Learning Objectives

- Explore the current challenges facing the development of Genomics, and how this impacts on EHR's
- Describe the joint working of standards bodies within the realm of Genomics and clinical practice
- Assess the work currently underway to address the challenges of bringing together clinical (EHR) data and Genomics research
- Evaluate the affect of current developments related to genomics on the future ways of working within EHR's

- GA4GH / HL7 / SNOMED International

"Implementing Genomics in EHR's – present and future "

Current situation

- Lack of interoperability between EHR and Genomics researchers
- Genomics terminologies only utilized within the Genomics research world
- Clinical terminologies not used by the Genomics research world
- Genomic researchers reliant on summary classification data
- Useful clinical information is available with EHR's, that cannot be Accessed by Genomics researchers
- No agreed exchange format for clinical Genomic information, that has unique requirements
- Dispersed global Genomics research community

Current landscape

- Genomic data refers to the genome and DNA data of an organism.
- Genomic research brings together Genomic data with clinical data, in the form of phenotypes
- A "phenotype" refers to the observable physical properties of an individual
- Current developments rely on taking the enormous amount of genomic data, and analyzing it together with phenotypic data to inform future clinical practice, in the form of precision (personalized) medicine
- Precision (personalized) medicine will allow individuals to receive the optimal treatment for their condition

Current standards

Electronic Health Records (EHR)

- SNOMED CT
- LOINC
- HL7
- ICD-10
- ICD-10CM
- MedDRA
- RxNorm

. . . and numerous others

Genomic research

- Human Phenotype Ontology (HPO)
- Online Mendelian Inheritance in Man (OMIM)
- Orphanet
- Clinical Data Interchange Standards Consortium (CDISC)
- VCF (Variant Call File)

. . . and numerous others

Use case .1.

Supporting the genomics research community's need to access clinical information from the EHR

- Genomics research requires granular details of clinical diagnoses and findings from EHR, to assist in research activities
- An agreed exchange format is required that supports the level of granularity required of the data, the need for consistency, and the need to share data across the global genomics research community
- Genomics researchers use specific terminologies which have been developed to meet their needs, these are not in use within EHR's, nor would they meet the requirements of EHR systems

Use case .2.

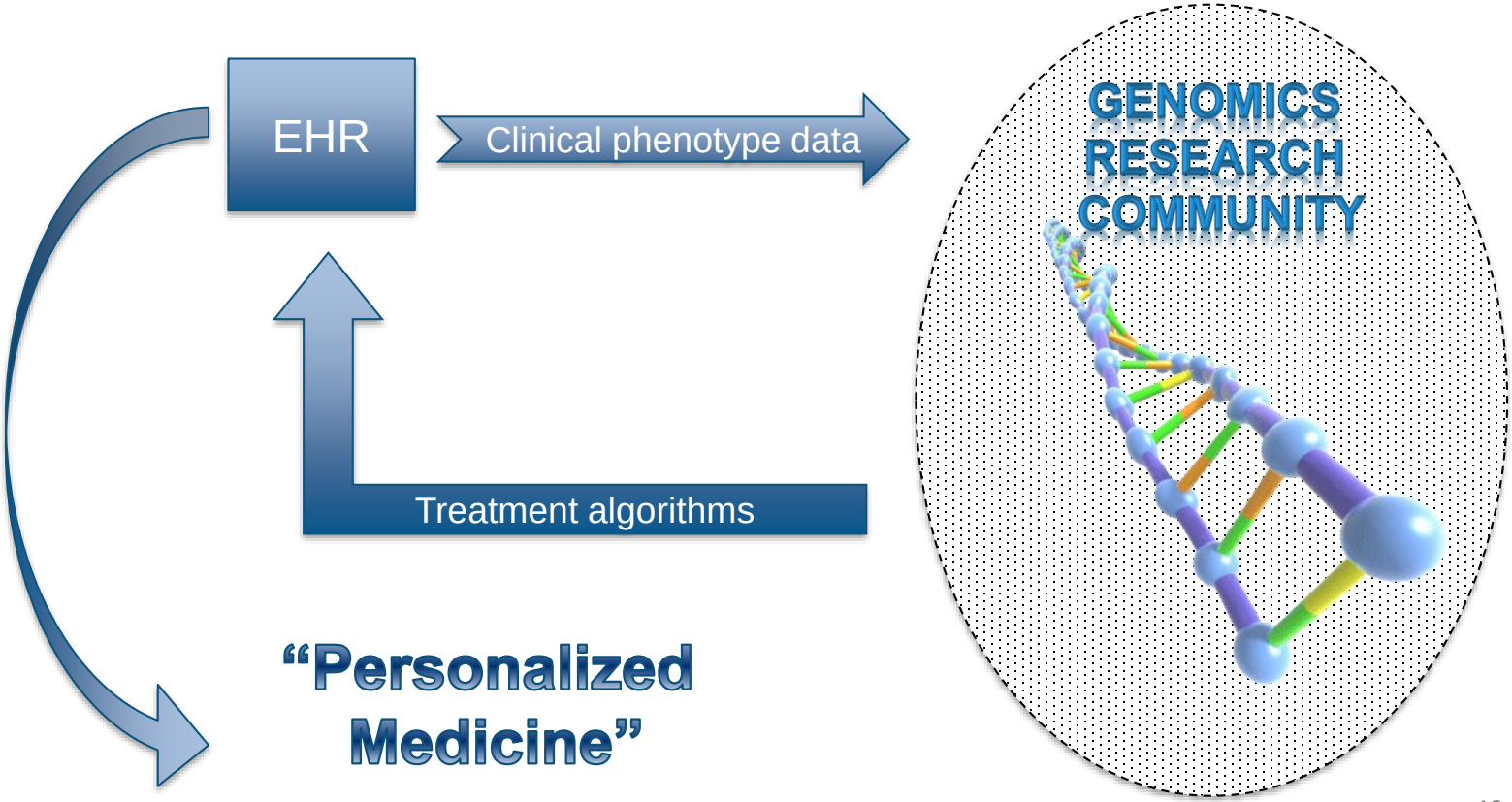
Precision medicine

- To deliver precision medicine into clinical practice requires the genomics research community to specify pathways of care that can be implemented within EHR systems
- Pathways will need to be implemented in an automated fashion to support uptake and accuracy of usage
- Pathways will need to be implemented in an automated manner using clinical decision support systems, that are driven by international terminology standards

Required solutions

- Must support interoperability across the EHR/genomic research boundary
- Must support the sharing of data within global Genomics research community
- Must provide specification of information standards that support new use cases
- Multiple standards must be specified in a way that supports the integration of standards within existing structures and processes, in both the EHR and Genomic communities
- Standards bundle that leverage the strengths of individual existing standards, and package them for ease of implementation

A vision of future process flows



"Personalized Medicine"

#HIMSS19

Personalized medicine

- Understanding each individual's genome – foundation of precision medicine
- Precision medicine uses individuals genetic data to identify optimum treatments
- Medicine is not, and should not be a "one-size" fits all
- Using genomic data, with other medical data, can provide a individual "personalized" view of treatment options
- The goal is to identify the optimum treatment of an individual's illness.

Global Alliance for Genomics and Health (GA4GH)

GA4GH Aims

- The Global Alliance for Genomics and Health (GA4GH) is an international, nonprofit 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. All of our work builds upon the Framework for Responsible Sharing of Genomic and Health-Related Data.

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GA4GH

Aims of collaboration activities

- To provide interoperable complete solutions for implementers
- To provide cross standards expertise to advise on developments
- To provide standards specifications that are aligned, and packaged to support implementation
- To engage early with standards communities, to inform the development of standards specifications

Global Alliance
for Genomics & Health

GA4GH working groups

GA4GH Foundational and Technical Work Streams develop standards and tools that are designed to overcome technical and regulatory hurdles to international genomic data-sharing

- Data Security
- Regulatory & Ethics
- **Clinical & Phenotypic Data Capture**
- Cloud
- Data Use & Researcher Identities (DURI)
- Discovery
- **Genomic Knowledge Standards**
- Large Scale Genomics

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GA4GH Clinical & Phenotypic Data Capture & Exchange

Sub-group 2: Data Exchange / Interoperability

Original developments undertaken by Melissa Haendel, Peter Robinson, Chris Mungall

Work Stream Leads:

- Melissa Haendel (Monarch)
- David Hansen (CSIRO / AGHA)

Sub-group leads:

- Michael Baudis (University of Zurich)
- Grant Wood (Intermountain Health Care Clinical Genetics Institute)
- Alejandro Metke (CSIRO / AGHA)
- Jules Jacobsen (QMUL / Monarch)

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GA4GH Genomic Knowledge Standards (GKS)

Goal: Develop a framework of standards-based components that lower barriers for the exchange of genomic information and its translation into clinical practice

Work Stream Leads:

- Andy Yates (EBI)
- Robert Freimuth (Mayo Clinic)

Variant Representation subgroup:

- Larry Babb (Broad)
- Michael Baudis (Univ Zurich)

Variant Annotation subgroup:

- Matt Brush (OHSU)
- Javier Lopez (Genomics England)

Global Alliance for Genomics & Health

HL7

- Founded in 1987, Health Level Seven International (HL7) is a not-for-profit, ANSI-accredited standards developing organization dedicated to providing a comprehensive framework and related standards for the exchange, integration, sharing and retrieval of electronic health information that supports clinical practice and the management, delivery and evaluation of health services.
- Produce a set of international standards for transfer of clinical and administrative data between software applications used by various healthcare providers

HL7 International

HL7 Clinical Genomics (CG)

Goal: Develop standards for the exchange of clinical and translational information related to an individual's genomic data and family health history, and (its linkage to) relevant clinical information

- Bob Milius (National Marrow Donor Program)
- Kevin Power (Cerner)
- Robert Freimuth (Mayo Clinic)
- Gil Alterovitz (Boston Children's Hospital)

HL7 International

SNOMED International

Is a not-for-profit organization that owns, administers and develops SNOMED CT, the world's most comprehensive clinical terminology

SNOMED CT:

- Is the most comprehensive, multilingual clinical healthcare terminology in the world, built on ontological principles
- Is a resource with comprehensive, scientifically validated clinical content
- Enables consistent representation of clinical content in electronic health records
- Is mapped to other international standards
- Is in use in more than fifty countries

SNOMED International

Genomics and Precision Medicine Clinical Reference Group (CRG)

- Established to ensure that the requirements of Genomics and Precision Medicine are incorporated into the content of SNOMED CT International Release by providing a forum for discussion, and also to support the development of derivative products that will support implementations focused on using SNOMED CT for Genomics and Precision Medicine purposes.
- SNOMED International has published its commitment to moving forwards with Genomics and Precision Medicine through its "SNOMED CT and Genomics Medicine" by publication of a strategy for “SNOMED CT and genomics”
- Main commitments within the strategy are:
  - Build links between SNOMED CT and genomic terminologies
  - Develop pilot sites to explore SNOMED CT usage and provide recommendations for future developments

Terminology developments

- Review of exisiting terminologies to assess the strengths and weaknesses of each one
- Acceptance that a single terminology solution is neither desirable or realistic
- Formal collaboration between terminology standards across the Genomics and EHR boundary
- Development of subsets of content that can be used in information exchange specifications to support consistent data representation

Terminology interoperability

- “Acceptance that a single terminology solution is neither desirable or realistic”
- Requirement to view data in a format dictated by the users, dependent on their individual requirements
- The ability to easily transfer between different views of the data using different terminology specifications
- Provide computable and quality assured linkages between terminology standards

Exchange standards

- To provide a standard for the transmission of data between clinical and genomic settings, and also to support the transfer of data between genomics organizations
- Specification should provide a standard way of recording Genomics information, specifically phenotypic data, supports transfer and data usage
- Leverage work currently in progress – Genomic Knowledge Standards
  - HL7 Genomics Working Group (HL7 Domain Analysis Model – Clinical Sequencing)
  - GA4GH Clinical & Phenotypic Data Capture working group

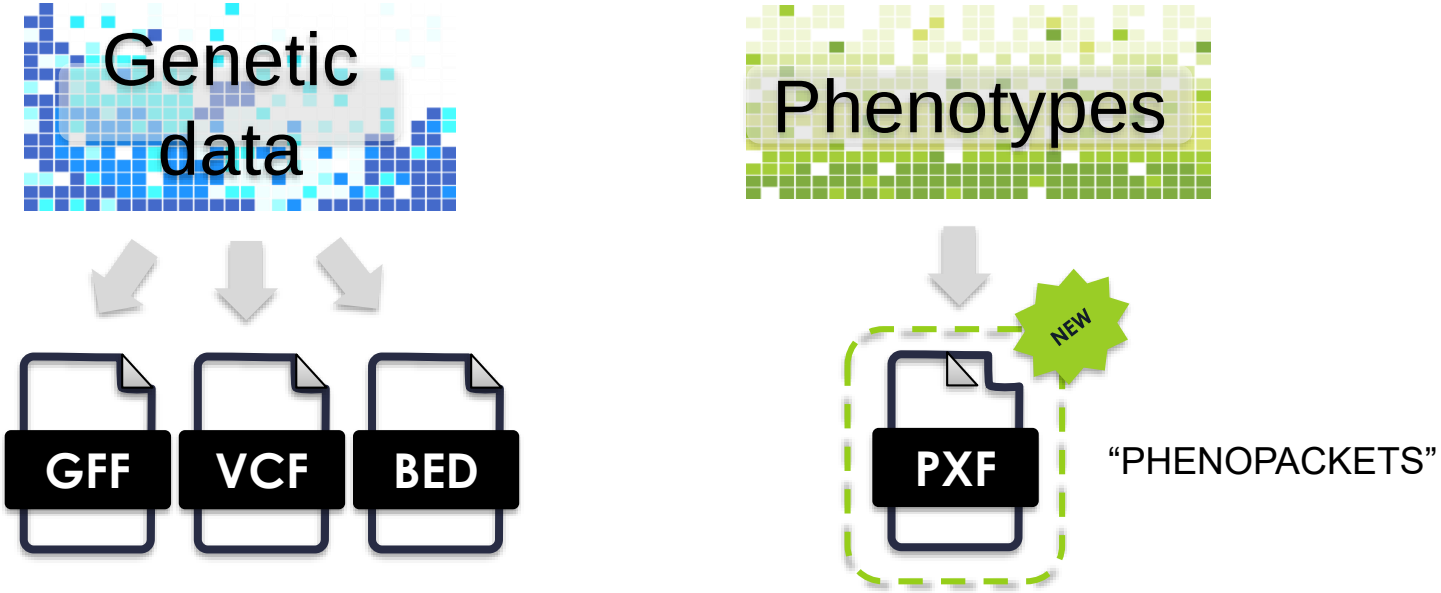
FHIR message development

- While ontologies and terminologies provide the standard data concept definitions for capturing clinical information, an information model is required to successfully exchange that information between clinical information systems and with related information systems
- The standard will provide information models with different levels of complexity to enable high level clinical phenotype information as well as deep clinical phenotype information to be exchanged
- Leveraging the strengths and flexibility of the FHIR specification, to develop FHIR resources specification specific to the requirements of recording and communicating phenotype information
- Development of example specifications based on specified use cases
- Identification of driver projects to validate proposed FHIR specifications to ensure they are fit for purpose

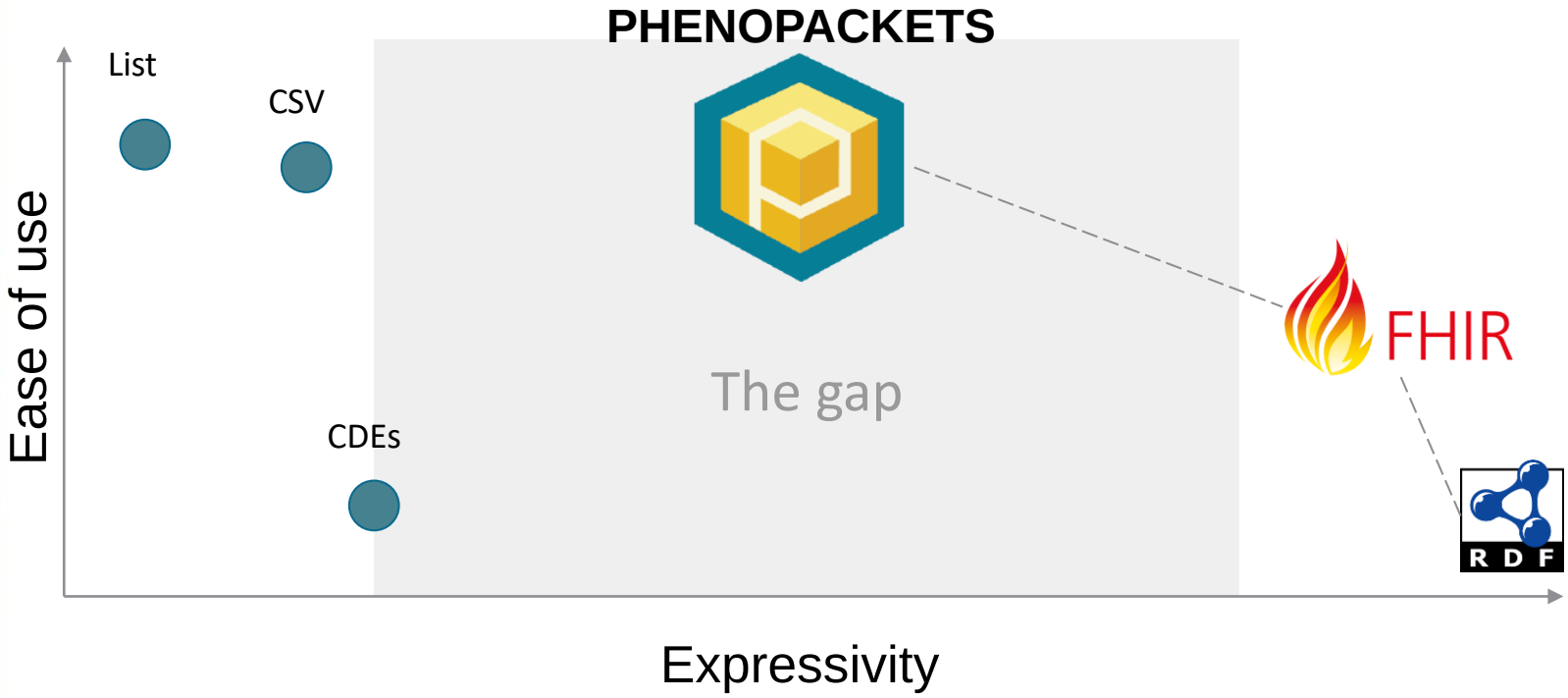
Phenopackets

- “Phenopacket” is a collection of data types focused on the representation of rare disease and cancer samples for both initial capture and analysis
- Identification of use cases to engage in trial the use of “Phenopackets” to exchange clinical information between systems, and also for Genomics researchers to use in the analysis and “Discovery” processes
- Identify and engage with Genomics research community organizations
- Production of implementation guidance to support the implementation of FHIR resources and Phenopackets

Standard exchange formats exist for genes … but not for phenotypes



Finding the sweet spot between simple and expressive

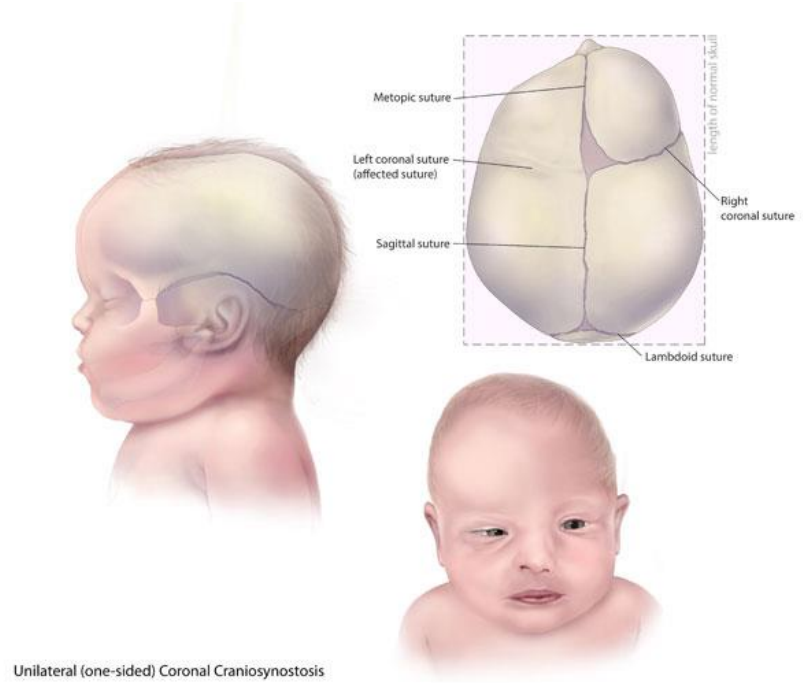


Single term…

Craniosynostosis

HPO:0001363

Craniosynostosis syndrome (disorder)
SCTID: 57219006



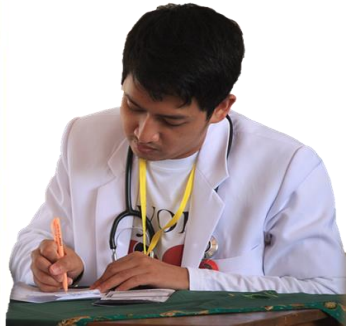
Unilateral (one-sided) Coronal Craniosynostosis

List of terms…

Craniosynostosis	HPO:0001363	Craniosynostosis syndrome SCTID: 57219006
Brachydactyly	HPO:0001156	Brachydactyly SCTID: 43476002
Proptosis	HPO:0000520	Proptosis SCTID: 18265008
Broad thumb	HPO:0011304	Broad thumbs SCTID: 249773003

Can we do better?

- ✓ Craniosynostosis
- ✓ Brachydactyly
- ✓ Proptosis
- ✓ Broad thumb…



We need structure!

```
patient:
  id: "PROBAND#1"
sex:
  id: "PATO:0000384"
  label: "male"
```

```
phenotypes:
- type:
    id: "HP:0000520"
    label: “Proptosis"
  severity:
    id: "HP:0012828"
    label: "Severe"
  classOfOnset:
    id: "HP: 0003577"
    label: “Congenital onset"
```

who?

what?

how?

when?

Phenopacket Implementations

- Reference implementation/spec:
  - Phenopacket-schema GitHub repo (https://github.com/phenopackets/phenopacket-schema)
- Proof of concept applications:
  - Exomiser web (Monarch web-app) (bit.ly/phenopacket-app-beta)
  - FHIR -> Phenopacket service
  - Exomiser service
  - SMART on FHIR app
- In production systems:
  - Biosamples exporting Phenopackets (e.g. https://www.ebi.ac.uk/biosamples/samples/SAMN05324082)

Next steps

- Engaging the community
- Collecting use case requirements
- Development of standards bundles to assist implementation
- Collaboration between SDO’s, and clinical and Genomic bodies

Future view

EHR

- Increased terminology content to support recording of detailed Genomic related information
- Automated clinical pathways supporting the implementation of personalized (precision) medicine
- Data transfer available to other organizations available in an automated manner

Genomics researchers

- Access to granular clinical information
- Automated collection of clinical data from EHR's
- Availability of data, using different terminology standards as required

Questions

Ian Green
Customer Relations Lead Europe and Clinical Engagement Business Manager
SNOMED International

w: www.snomed.org
e: igr@snomed.org