

Update zu den Aktivitäten von HL7 Clinical Genomics

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Saal: Berlin



Über mich

- Patrick Werner, M.Sc. Medizininformatik
- Prozesse & Interoperabilität @MOLIT Institut
- Geschäftsführer echinos GmbH
- Co-Chair: Genomics Workgroup, HL7 Int.
- Stellvertretender Leiter: TC Terminologie HL7 Deutschland
- hapi & FHIR core contributor

HL7 Clinical Genomics

- Updates des Genomic Reporting Implementationguide
 - Operations
 - MolecularConsequence
 - HGNC/ HPO CodeSystem
 - Implications
 - Biomarker
- NHS: FHIR Genomics Implementation Guide
- Ausblick: Entwicklung des Genomic Reporting Implementationguide

Operations





















- kein Ersatz für REST Interaktionen, sondern Ergänzung
- “advanced Topic”: Operationlogik muss von jedem Server händisch implementiert werden
- Gedacht für Forschung und Genomic Repositories
 - Normalisierung während der Verarbeitung der Anfrage

Find Subject Diagnostic Implications	Retrieves genetic diagnostic implications for variants.
Find Subject Haplotypes	Retrieve haplotypes/genotypes for specified genes.
Find Subject Molecular Consequences	Retrieves molecular consequences of a DNA variant.
Find Subject Specific Haplotypes	See if specified haplotypes/genotypes are present.
Find Subject Specific Variants	Determine if specified simple variants are present.
Find Subject Structural Intersecting Variants	Determine if structural variants are present that overlap range(s).
Find Subject Structural Subsuming Variants	Determine if structural variants are present that fully subsume a range.
Find Subject Treatment Implications	Retrieves genetic therapeutic implications for variants/haplotypes/genotypes.
Find Subject Variants	Determine if simple variants are present that overlap range(s).

MolecularConsequence

- Repräsentiert die beobachtet, aber meist berechnete Auswirkung einer genetischen Variante
- Ziel: saubere Trennung der Variante, der Auswirkung und Implikationen

MolecularConsequence

Name	Flags	Card.	Type	Description & Constraints
 Observation		0..*	GenomicImplication	Molecular Consequence
 code		1..1	CodeableConcept	molecular-consequence Required Pattern: At least the following
 coding		1..*	Coding	Code defined by a terminology system Fixed Value: (complex)
 system		1..1	uri	Identity of the terminology system Fixed Value: http://hl7.org/fhir/uv/genomics-reporting/CodeSystem/tbd-codes-cs
 code		1..1	code	Symbol in syntax defined by the system Fixed Value: molecular-consequence
  component:coding-hgvs		0..1	BackboneElement	DNA change (c.HGVS)
  component:transcript-ref-seq		0..1	BackboneElement	Reference Transcript
  component:protein-hgvs		0..1	BackboneElement	Protein (Amino Acid) Change - pHGVS
  component:protein-ref-seq		0..1	BackboneElement	Protein Reference Sequence
  component:feature-consequence		0..*	BackboneElement	Feature Consequence
  component:functional-effect		0..1	BackboneElement	Functional Effect

HGNC/ HPO CodeSystem

- Update der zu nutzenden CodeSystem URIs:
- **HGNC**
 - GeneNames: <http://www.genenames.org>
 - GeneGroups: <http://www.genenames.org/genegroup>
- **HumanPhenotypOntology**
 - Alt: <http://hpo.jax.org/app/>
 - Neu: <http://human-phenotype-ontology.org>


















Implications

- TherapeuticImplication
 - Verwendung des gleichen Codes für Observation.code &
 - Observation.component[TherapeuticImplication].code

MolecularBiomarker

- Neues Profile als Basis für (genetische) Biomarker
- Beispiele: TMB, MSI, HER2 Staining

MolecularBiomarker

Name	Flags	Card.	Type	Description & Constraints
Observation			Observation	
 Slices for category		2..*	CodeableConcept	Classification of type of observation Slice: Unordered, Open by pattern:coding
 category:labCategory		1..1	CodeableConcept	Classification of type of observation
 coding		1..*	Coding	Code defined by a terminology system Required Pattern: At least the following
 system		1..1	uri	Identity of the terminology system Fixed Value: http://terminology.hl7.org/CodeSystem/observation-category
 code		1..1	code	Symbol in syntax defined by the system Fixed Value: laboratory
 category:mbCategory		1..1	CodeableConcept	Classification of type of observation
 coding		1..*	Coding	Code defined by a terminology system Required Pattern: At least the following
 system		1..1	uri	Identity of the terminology system Fixed Value: http://hl7.org/fhir/uv/genomics-reporting/CodeSystem/tbd-codes-cs
 code		1..1	code	Symbol in syntax defined by the system Fixed Value: biomarker-category
 category:geCategory		0..1	CodeableConcept	Classification of type of observation
 coding		1..*	Coding	Code defined by a terminology system Required Pattern: At least the following
 system		1..1	uri	Identity of the terminology system Fixed Value: http://terminology.hl7.org/CodeSystem/v2-0074
 code		1..1	code	Symbol in syntax defined by the system Fixed Value: GE
 code		1..1	CodeableConcept	Code for the biomarker Binding: Molecular Biomarker Codes (example)
 Slices for component		0..*	BackboneElement	Component results Slice: Unordered, Open by pattern:code
 component:gene-studied		0..*	BackboneElement	Gene Studied
 component:biomarker-category		0..*	BackboneElement	Biomarker Category

status: final

category: Laboratory ([Observation Category Codes#laboratory](#)), A characterization of a given biomarker observation. ([To Be Determined Codes#biomarker-category](#))

code: HER2 Ag [Presence] in Tissue by Immune stain ([LOINC#18474-7](#))

subject: Patient/CGPatientExample01 "EVERYMAN"

value: Present ([LOINC#LA9633-4](#))

component

code: Gene studied [ID] ([LOINC#48018-6](#))

value: ERBB2 ([geneId#HGNC:3430](#))

component

code: A characterization of a given biomarker observation. ([To Be Determined Codes#biomarker-category](#))

value: immune stain category ([Molecular Biomarker Ontology Codes#immuneStain](#))

component

code: A characterization of a given biomarker observation. ([To Be Determined Codes#biomarker-category](#))

value: cell receptor category ([Molecular Biomarker Ontology Codes#cellReceptor](#))

component

code: A characterization of a given biomarker observation. ([To Be Determined Codes#biomarker-category](#))

value: protein category ([Molecular Biomarker Ontology Codes#protein](#))

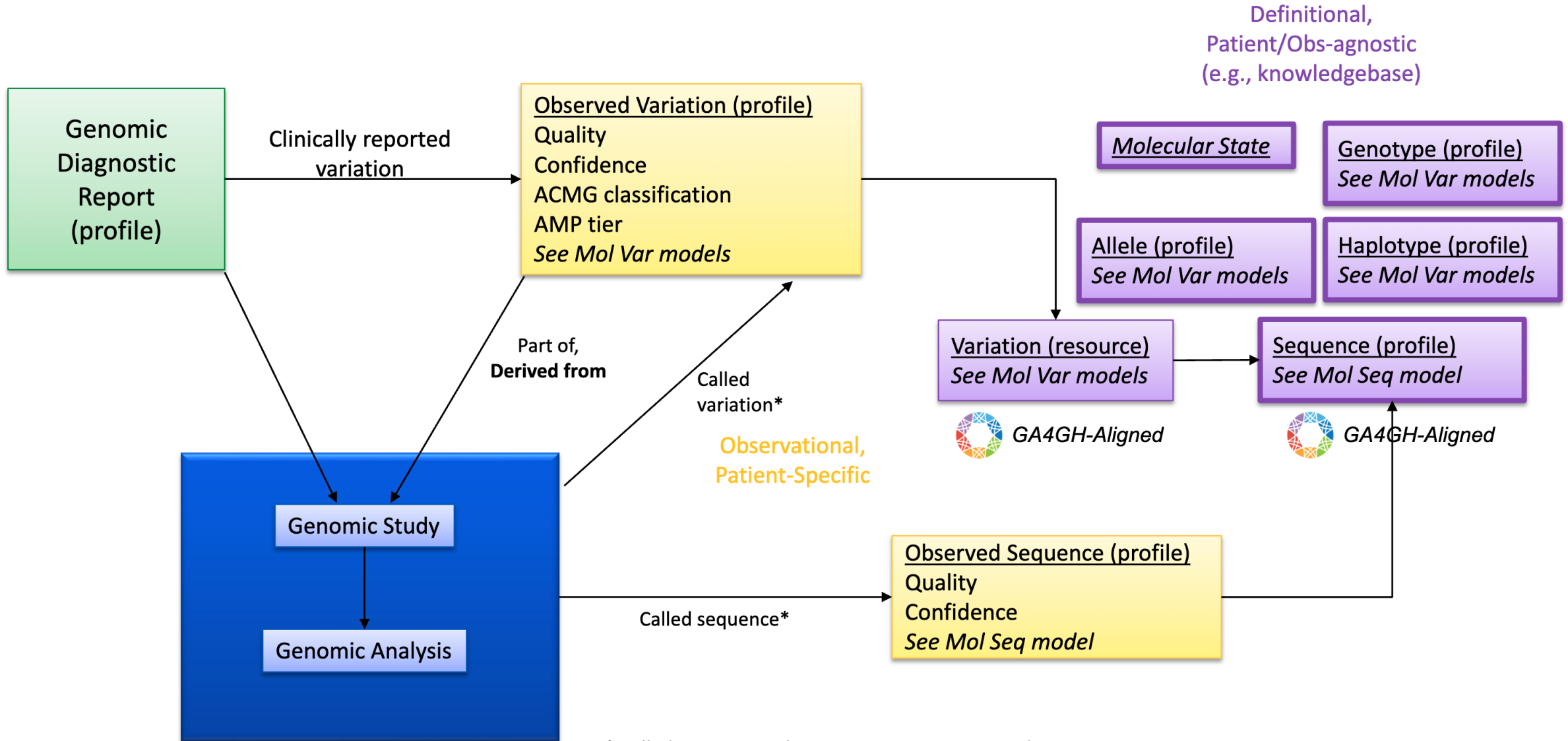
NHS: FHIR Genomics Implementation Guide

- Das NHS bitte um Feedback für die erste Version ihres **FHIR Genomics Implementation Guide**
- Schwerpunkt: OrderEntry Management
- Befunde aktuell noch vor allem PDF Dokumente
- IG: [NHS Genomics IG](#)
- Zulip Diskussion: [NHS Genomics IG Zulip Thread](#)

Ausblick Genomics IG

- Neue Ressourcen in R5:
 - GenomicStudy (Metadaten einer genetischen Untersuchung, wird es auch als R4 Backport geben)
 - MolecularSequence (Ressource wurde aufgeräumt, da Überschneidungen mit Genomics IG vorhanden waren)

HL7 FHIR GENOMICS (FUTURE STATE)



shown: implication, interpretation, etc

* Called variation and sequence are not restricted to clinical testing and reporting (e.g., research studies)

Danke



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