ÜBERSICHT relevanter (inter)nationaler VORARBEITEN

1. Datenmodelle der Medizininformatik-Initiative (MI-I)

Diese umfassen den sogenannten „Kerndatensatz“ mit den *Basismodulen* Person, Fall, Diagnose, Prozedur, Laborbefund und Medikation und die *Erweiterungsmodule* (u.a. Onkologie, Consent, Pathologie-Befund, Mol.gen. Befund, Bioprobendaten).

- Link zu den Implementation Guides (IGs) der FHIR-Spezifizierung aller Module auf Simplifier: https://simplifier.net/organization/koordinationsstellemii/~projects

2. Genomics Reporting Implementation Guide

Die Profilierung des Mol.gen. Befundes der MI-I richtet sich nach dem Genomics Reporting IG von HL7 International

- Link zum IG: http://hl7.org/fhir/uv/genomics-reporting/

Der IG setzt sich zusammen aus Komponenten (FHIR Profilen/artifacts): http://hl7.org/fhir/uv/genomics-reporting/artifacts.html

Allgemeine FHIR Profile:

FHIR Profil: *Beschreibung*

- Genomics Report: Defines the overall genomic report
- Specimen: Constraints on Specimen for use with clinical genomics reporting
- Recommended Followup: Task describing the followup that is recommended
- Request for Genomics Test: Lab order or request that triggered execution of genomics test
- Grouper: Organizes information within genomic report
- Overall Interpretation: Provides overall interpretation of genomic results reported
- Haplotype: Assertion of particular haplotype on basis of 1+ variants
- Genotype: Assertion of particular genotype on basis of 1+ variants or haplotypes
- Variant: Details about set of changes in the tested sample compared to reference sequence.
- Sequence Phase Relationship: Indicates whether two entities are in Cis or Trans relationship to each other
- Inherited Disease Pathogenicity: Provides indication of whether there's a pathologic risk associated with particular genotype, haplotype, variant or combination, and if so, what associated pathology is
- Region Studied: Provides a description of the region studied.

Profile zu somatisch genomischen Inhalten:

- Somatic Diagnostic Implication: Finding of whether particular somatic genotype/haplotype/variation or combination supports or opposes diagnosis of particular cancer
- Somatic Prognostic Implication: Finding of whether particular somatic genotype/haplotype/variation or combination-thereof predicts particular outcome for specified cancer - either on its own or in conjunction with 1+ interventions
- Somatic Predictive Implication: Finding of whether particular somatic genotype/haplotype/variation or combination-thereof predicts implication of specified medication or combination of medications

Profile zur Beschreibung genomischer Implikationen für Medikation (Pharmacogenomics):

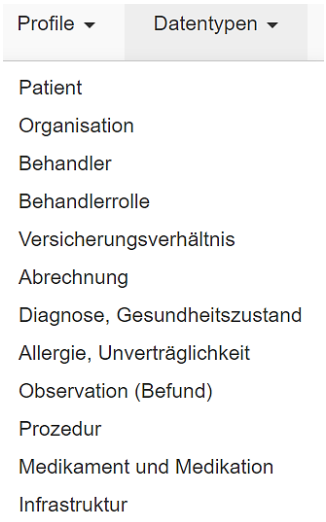
- Medication Metabolism Implication: Assertion of expected implication of particular genotype on the ability of subject to metabolize medications
- Medication Transporter Implication: Assertion of expected implication of particular genotype on ability of subject to actively transport medications
- Medication Efficacy Implication: Assertion of expected implication of particular genotype on efficacy of medications for subject
- High Risk Allele: Assertion of whether the patient has a high-risk allele
- Medication Usage Implication: Task describing what sort of change (if any) should be made in a patient's medication based on an identified genotype
- Current Medication: MedicationStatement describing a med potentially being taken by the patient that may require adjustment

3. Deutsche FHIR Basisprofile

Der Leitfaden (Implementation Guide) der deutschen FHIR Basisprofel soll als Hilfestellung und Richtschnur vor und während der Entwicklung von Software oder abgeleiteten Profilen dienen. Die enthaltenen Profile können aber auch für die Validierung bereits entwickelter Komponenten verwendet werden.

- Link zum IG auf Simplifier: https://simplifier.net/guide/leitfadenbasisde/home

Darin beschriebene FHIR Profile sind:



4. Informationstechnische Systeme in Krankenhäusern (ISIK) Implementation Guide

Für den Austausch von Gesundheitsdaten erarbeitet die gematik einen verbindlichen Standard über eine standardisierte Schnittstelle für informationstechnische Systeme in Krankenhäusern. In der Spezifikation "ISiK-Basismodul" beschreibt die gematik die für diesen Zweck entwickelten FHIR-Ressourcen, die ein Primärsystem in Form von Datenobjekten über das REST-basierte Application Programming Interface überträgt. Use-case-übergreifende Funktionalitäten, die vom ISiK-Basismodul unterstützt werden: z.B: Suche nach Fallinformationen anhand einer Fallnummer, nach Patienten anhand demografischer Kriterien und Abfrage der Versicherungsinformationen eines Patienten und Diagnosen

- Link zum IG: https://simplifier.net/guide/ImplementierungsleitfadenISiK-Basismodul/Einfuehrung; (FHIR Profile sind unter dem Reiter „Datenobjekte“ gelistet)

5. minimal Common Oncology Data Elements (mCODE) Implementation Guide

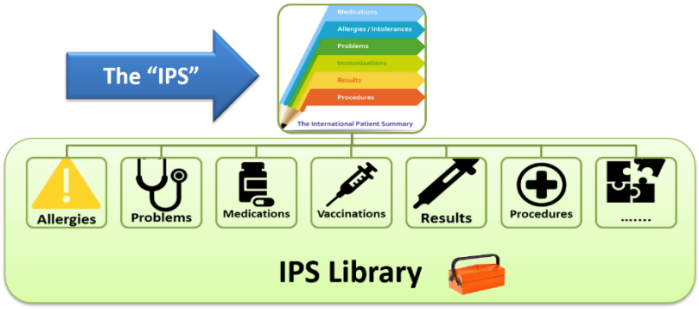
mCODE™ (short for Minimal Common Oncology Data Elements) is an initiative intended to assemble a core set of structured data elements for oncology electronic health records (EHRs). mCODE is a step towards capturing research-quality data from the treatment of all cancer patients.

- Link zum IG: http://hl7.org/fhir/us/mcode/

6. International Patient Summary (IPS) Implementation Guide

An International Patient Summary (IPS) document is an electronic health record (HER) extract containing essential healthcare information about a subject of care. As specified in EN 17269 and ISO/DIS 27269, it is designed for supporting the use case scenario for ‘unplanned, cross border care’, but it is not limited to it. The IPS dataset is minimal and non-exhaustive; specialty-agnostic and condition-independent; but still clinically relevant. The IPS document is composed by a set of robust, well-defined and potentially reusable sets of core data items.

- Link zum IG: http://hl7.org/fhir/uv/ips/



7. Pedigree

Ein FHIR-basierter Pedigree Standard wird im Rahmen der GA4GH Allianz im Moment erarbeitet.
The GA4GH Pedigree Standard allows for the computable exchange of family health history as well as representation of larger, more complex families.

- Link zum github Projekt: https://github.com/GA4GH-Pedigree-Standard/pedigree
- Link zum draft IG: https://ga4gh-pedigree-standard.github.io/pedigree-fhir-ig/index.html

8. Terminologien (Code Systems) in FHIR Profilen

HL7 International provides a list of URIs to terminology systems that may be used in the *"system"* element of the *"Coding"* data type within FHIR resources.

- Link: http://build.fhir.org/terminologies-systems.html

The following is an excerpt of externally defined code systems:

Defined Externally | Defined as part of FHIR

URI	Source	Comment	OID (for non-FHIR systems)
Externally Published code systems			
http://snomed.info/sct	SNOMED CT (IHTSDO)	See Using SNOMED CT with FHIR	2.16.840.1.113883.6.96
http://www.nlm.nih.gov/research/umls/rxnorm	RxNorm (US NLM)	See Using RxNorm with FHIR	2.16.840.1.113883.6.88
http://loinc.org	LOINC (LOINC.org)	See Using LOINC with FHIR	2.16.840.1.113883.6.1
https://fhir.infoway-inforoute.ca/CodeSystem/pCLOCD	pCLOCD (pCLOCD)	See Using pCLOCD with FHIR	2.16.840.1.113883.2.20.5.1
http://unitsofmeasure.org	UCUM: (UnitsOfMeasure.org) Case Sensitive Codes	See Using UCUM with FHIR	2.16.840.1.113883.6.8
http://ncimeta.nci.nih.gov	NCI Metathesaurus	See Using NCI Metathesaurus with FHIR	2.16.840.1.113883.3.26.1.2
http://www.ama-assn.org/go/cpt	AMA CPT codes	See Using CPT with FHIR	2.16.840.1.113883.6.12
http://va.gov/terminology/medrt	MED-RT (Medication Reference Terminology)	See Using MED-RT with FHIR	2.16.840.1.113883.6.345