Service Unit Terminological Services

Onboarding presentation

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Agenda

1. Team and project introduction
2. FHIR Terminology Module overview
3. FHIR Terminology Services overview
4. Technical Details
5. Administrative details
6. Literature recommendations

Team and project introduction

- MII “2b” project in the consolidation and extension phase of the MII: *advancing the harmonization of health data and IT solutions at the university hospital sites in cooperation with the NUM*
- Project funded from 2023 through MII and NUM starting 2027
- Three partner sites:
  - University of Lübeck
  - University of Cologne
  - Hanover Medical School
- **Goal**: provide and support central terminology services to DICs and the MII/NUM in general, to support semantic interoperability
  - Support for CDS designers in the MII for the development of the CDS
  - Support for DICs to provide a reference server for terminology used in MII/NUM
  - Support for FDPG in (search) ontology generation

Semantic interoperability through terminology (services)

- Data from primary systems (generally) is not interoperable by default, even if accessible in standardized formats and aligned to a defined data model
- Coded data often captured in *local* coding systems
  - Laboratory data: order number codes only for hospital (network)
  - eCRFs in network studies: not always captured using standard codes - but great opportunities for improvement!
- Utilization of standard terminology/-ies is one of the most pressing challenges for achieving semantic interoperability
- Only semantically interoperable data is usable by others.
- **Terminology services** are a piece of the infrastructural puzzle to achieve semantic interoperability in defined contexts
  - Terminology services are more than just a terminology server!

Terminology for terminologies

- Not all terminological resources are, strictly speaking, *terminologies*
- We are going to say *terminologies* anyways.

Controlled vocabularies	HL7v2 Coding Tables	Flat list of values
(Statistical) classifications	ICD-10	Monoaxial and monohierarchical system of (disjunct) classes
Systematic Nomenclatures ***Terminologies*** Thesauri	SNOMED II LOINC MeSH	***Terminologies***: multiaxial, polyhierarchical
Formal ***Terminologies***	SNOMED CT	***Terminologies***: multiaxial, polyhierarchical, based on formal definitions of meaning

Common *terminologies* in use in Germany

- **ICD-10-GM** and **ICD-10-WHO**: statistical classifications used mandatorily for coding of morbidities (-GM) and mortalities (-WHO)—derived from international standard
- **OPS**: statistical classification used mandatorily for coding of procedures performed
- **Alpha-ID-SE, ORPHAcode**: used for coding rare diseases
- **SNOMED CT**: formal terminology with rich relationships between 360'000 concepts
- **LOINC**: Terminology for representing order identifiers, mainly used in lab settings
- **UCUM**: grammar for representing interpretable units of measure
- **ATC**: classification for pharmaceutical agents
- **PZN**: controlled (??) vocabulary for identifying concrete pharmaceutical products approved for use in Germany
- **Artifacts defined by HL7** for use in HL7 v2, HL7 v3, CDA and FHIR: generally controlled vocabularies or classifications
- Artifacts **internal to hospitals or projects**: often simple controlled vocabularies ("catalogs"), sometimes classifications (e.g. lab order numbers); including data dictionaries for research

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FHIR Terminology Module overview

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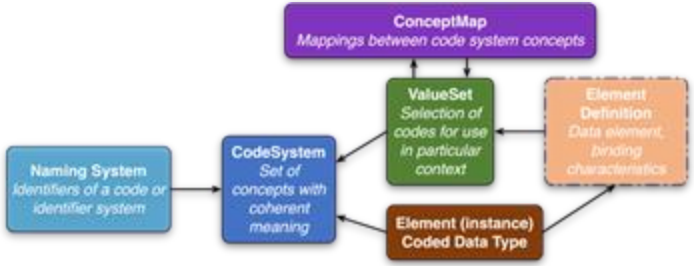
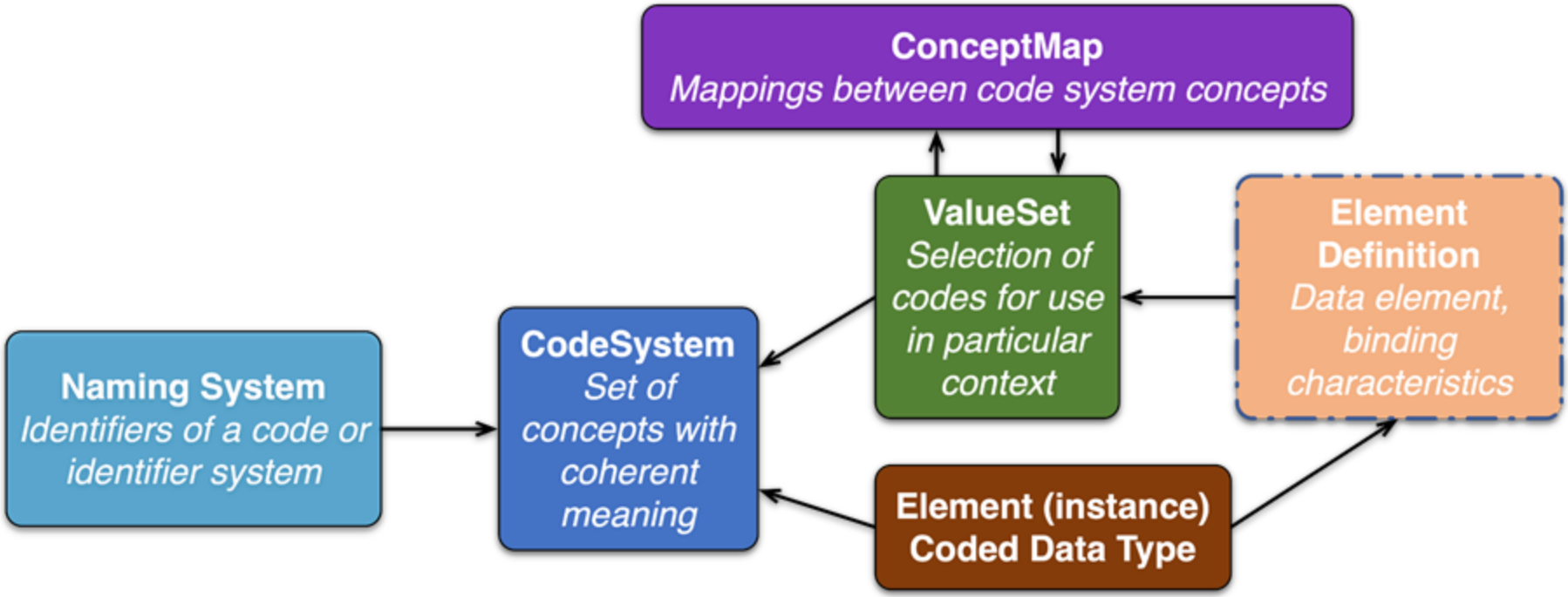
FHIR Terminology Module

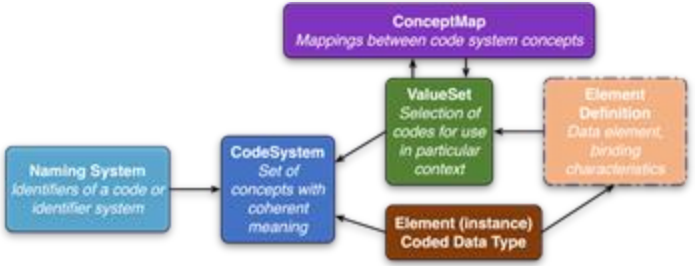


- Simple conceptual model for representing terminological artifacts
- Based on three (four) resource types: *CodeSystem*, *ValueSet*, *ConceptMap*, (*NamingSystem*)
- Used throughout the entirety of the HL7 FHIR Model
  - Both for coded data elements in further knowledge artifacts (e.g. profiles)
  - And in instances for representing e.g. a patients' administrative gender



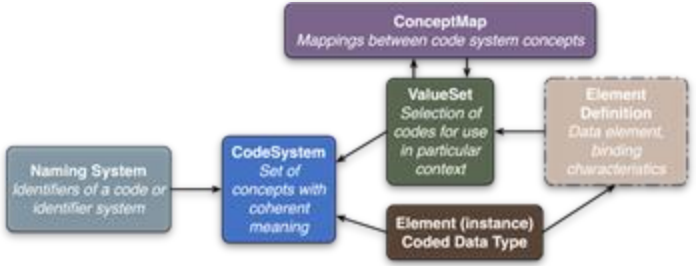
Simplified from: https://www.hl7.org/fhir/terminology-module.html

FHIR Terminology Module



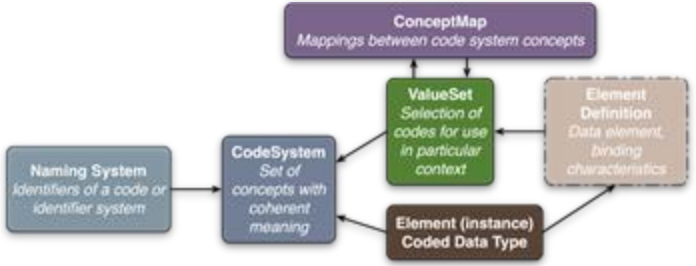
- Common to all knowledge artifacts:
  - Use of a canonical URI—does not change over lifetime of the resource; is globally unique; should be a resolvable URL, but doesn't need to be
  - Further identifiers possible (e.g. OIDs)
  - Business `version` element for managing lifecycle of the resource
  - Metadata elements like `name`, `title`, `publisher`, `copyright`, `status`, `description`, …
- Codes are only defined within a CodeSystem, and can't stand alone
  - What does the code "C04" represent? Is it ICD-10-GM, or ICD-O, or ATC, or …

CodeSystem (CS)



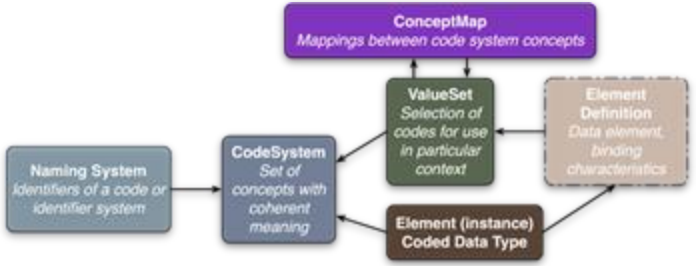
- "The *CodeSystem* resource is used to **declare the existence of and describe** a code system [...] and its **key properties**, and optionally define a part or all of its **content**" (FHIR CodeSystem)
- Defines the existence of a concept
  - Concepts are assigned unique (within the CS) codes
  - Concepts can declare optional properties (`parent`, `child`, and user-defined further properties)
  - Concepts have a display, and can have optional designations (e.g. translations)
- CS supports different values for `hierarchyMeaning`:
  - `grouped-by`
  - `is-a`
  - `part-of`
  - `classified-with`
- Some CodeSystem resources do not contain the concepts they define
  - SNOMED CT, LOINC: not representable as a FHIR CodeSystem, too complex for performant & comprehensive implementation

ValueSet (VS)



- "A *ValueSet* resource instance specifies **a set of codes** drawn from **one or more code systems**, intended for **use in a particular context**. Value sets link between *CodeSystem* definitions and their **use in coded elements**." (FHIR ValueSet)
- 2 aspects:
  - `compose`: "A definition of which codes are **intended** to be in the value set"
  - `expansion`: "The **list** of codes that are actually in the value set under a given set of conditions"
- 2 styles of defining the `compose`:
  - *Extensional:* Explicit list of codes to include
  - *Intensional*: Use of rules
    - *Include all codes that are children of "X"*
    - *Include all SNOMED CT concepts that match an ECL constraint*
- Inclusions as well as exclusions supported
- Optional: pin *compose* statements to a CS version
  - Implications for the expansion of a resource if not stated

ConceptMap (CM)

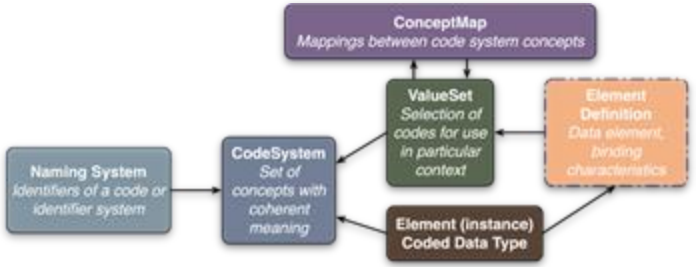


- "A **statement of relationships** from one **set of concepts** to one or more **other concepts**—either concepts in code systems, or data element/data element concepts, or classes in class models." (FHIR ConceptMap)
- "Mappings between code system concepts are **only intended to be defined in the context of a particular business usage**."
  - Scope of a CM (a mapping from CS to CS) defined in terms of a source and target VS
  - Usage context changes mappings, e.g. for "mapping from [SNOMED CT] to [ICD-10] for either data analysis or billing"
- All map elements have a `equivalence` (R4) / `relationship` (R5)

R4	`relatedto`	`equivalent`	`wider`	`narrower`	`unmatched`	...
R5	`related-to`	`equivalent`	`source-is-narrower-than-target`	`source-is-broader-than-target`	`not-related-to`	

- Substantial changes between FHIR R4 and R5 in definition of CM resource
  - MII uses R4!

Use in Profiles

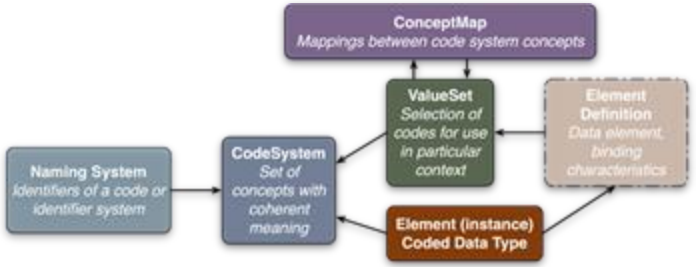


- Profiles/StructureDefinitions **bind** codeable data elements to value domains—i.e. *ValueSets*
- Bindings have different **binding strengths**

`required`	To be conformant, the concept in this element **SHALL** be from the specified value set.
`extensible`	To be conformant, the concept in this element **SHALL** be from the specified value set **if** any of the codes within the value set can apply to the concept being communicated. If the value set **does not cover** the concept (based on human review), alternate codings (or, data type allowing, text) may be included instead.
`preferred`	Instances are **encouraged** to draw from the specified codes for interoperability purposes but are **not required** to do so to be considered conformant.
`example`	Instances are not expected or even encouraged to draw from the specified value set. The value set merely provides **examples of the types of concepts** intended to be included.

Table from: https://hl7.org/fhir/terminologies.html

Use in FHIR instances



- Several data types with different semantics

`code`	The instance represents the code only. The system is implicit - it is defined as part of the definition of the element, and not carried in the instance.	`Patient.administrativeGender` `Observation.status`
`coding`	A datatype that has a code and a system element that identifies where the definition of the code comes from	*Generally within a* `CodeableConcept`
`CodeableConcept`	A type that represents a concept by plain text and/or one or more coding elements	`Observation.code`
`CodeableReference`	A type that can have either a reference to another resource, or a to a concept using a CodeableConcept	Rarely used

- Resources don't use *ValueSets*, but concrete codes from *CodeSystems*

Table from: https://hl7.org/fhir/terminologies.html

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FHIR Terminology Services

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FHIR Terminology Services

- Operations to interact with and query the terminology resources
- Using HTTP requests
  - GET – {server_url_for_specific_resource_type}/${operation}?{parameter}
  - POST – {server_url_for_specific_resource_type}/${operation} + JSON/XML body
- Parameters defined in the operation definition

In Parameters:						
Name	**Scope**	**Cardinality**	**Type**	**Binding**	**Profile**	**Documentation**
code		0..1	code			The code that is to be located. If a code is provided, a system must be provided
system	type	0..1	uri			The system for the code that is to be located
version	type	0..1	string			The version of the system, if one was provided in the source data
coding		0..1	Coding			A coding to look up

CodeSystem	ValueSet	ConceptMap
$lookup $subsumes $validate-code *$find-matches*	$validate-code $expand	$translate *$closure*

HTTP Requests: GET versus POST

- GET: {{server_url}}/{{resource_type}}/${{operation}}?{{parameters}}
- POST: {{server_url}}/{{resource_type}}/${{operation}} + Body als JSON/XML

Example:

Return all concepts that are a subconcept of "E11" and contain "Nicht als entgleist bezeichnet" in their name/display.

Body as JSON

```
{ "resourceType": "Parameters",
  "parameter": [
    { "name": "filter", "valueString": "Nicht als entgleist bezeichnet" },
    { "name": "valueSet",
      "resource": {
        "resourceType": "ValueSet",
        "compose": { "include": [
                      { "system": "http://fhir.de/CodeSystem/bfarm/icd-10-gm",
                        "filter": [{ "property": "concept",
                        "op": "descendent-of",
                        "value": "E11" }
]}]}}}]}
```

CodeSystem – $lookup

- Lookup information for a specific code
- Required parameters
  - system – url of *CodeSystem*
  - code – relevant code
  - [version] – version of *CodeSystem*
- Example (GET):
  - {{server_url}}/CodeSystem/$lookup?system=http://fhir.de/CodeSystem/bfarm/icd-10-gm&code=E11.9

Response:

```
{ "resourceType": "Parameters",
  "parameter": [
    { "name": "code",
      "valueCode": "E11.9“},
    { "name": "display",
      "valueString": "Diabetes mellitus,
                      Typ 2: Ohne Komplikationen"},
    { "name": "name",
      "valueString": "Icd10Gm”},
    { "name": "system",
      "valueUri": "http://fhir.de/CodeSystem/bfarm/icd-10-gm"},
    { "name": "version","valueString": "2025“},
    { "name": "property", "part": […]}
  ]
}
```

See: https://www.hl7.org/fhir/codesystem-operation-lookup.html

CodeSystem – $subsumes

- Query the hierarchical relationship between two concepts
- Results:

codeA	***equivalent***	codeB	E11.9 and E11.9
codeA	***not-subsumed***	codeB	E11.9 and A00
codeA	***subsumes***	codeB	E11 and E11.9
codeA	***subsumed-by***	codeB	E11.9 und E11

- Required parameters
  - system – url of CodeSystem
  - codeA – relevant code
  - codeB – relevant code
- Example (GET):

Response:

```
{{server_url}}/CodeSystem/$subsumes?
system=http://fhir.de/CodeSystem/bfarm/icd-10-gm&
codeA=E11&
codeB=E11.9
```

```
{ "resourceType": "Parameters",
  "parameter": [
    {
      "name": "outcome",
      "valueCode": "subsumes"
    }
  ]
}
```

See: https://www.hl7.org/fhir/codesystem-operation-subsumes.html

CodeSystem – $validate-code

- Check a code for correctness
- Required parameters
  - `url` – url of CodeSystem
  - `code` – relevant code
- Example (GET):
  - {{server_url}}/CodeSystem/$validate-code?url=http://fhir.de/CodeSystem/bfarm/icd-10-gm&code=XXX

Response:


```
{ "resourceType": "Parameters",
  "parameter": [
                { "name": "result", "valueBoolean":
false },
    { "name": "issues",
      "resource": {
        "resourceType": "OperationOutcome",
        "issue": [
          { "severity": "error",
            "code": "code-invalid",
            "details": {
              "text": "The provided code \"XXX\"
                                is not known to belong
to
                          the provided code system
                          \"http://fhir.de/CodeSystem
                                  /bfarm/ icd-10-gm\""
            },
        "location": ["code"]
        }
    ]
}, …]}
```


See: https://www.hl7.org/fhir/codesystem-operation-validate-code.html

ValueSet – $validate-code

- Check a code for correctness
- Required parameters
  - `url` – url of *ValueSet*
  - `code` – relevant code
  - `system` – url of *CodeSystem* for the code
  - `[ version ]` – the version of the *ValueSet*
  - `[ systemVersion ]` – the version of the *CodeSystem*
- Example (GET):
  - {{server_url}}/CodeSystem/$validate-code?url=https://www.netzwerk-universitaetsmedizin.de/fhir/ValueSet/chronic-liver-diseases-icd&code=A01.1&system=http://fhir.de/CodeSystem/bfarm/icd-10-gm

Response:


```
{ "resourceType": "Parameters",
  "parameter": [
    { "name": "result", "valueBoolean": false },
    { "name": "issues",
      "resource": {
        "resourceType": "OperationOutcome",
        "issue": [
          { "severity": "error",
            "code": "code-invalid",
            "details": {
              "text": "The provided code
'http://fhir.de/CodeSystem/bfarm/icd-10-gm#A01.1' was
not found in the value set 'https://www.netzwerk-
universitaetsmedizin.de/fhir/
ValueSet/chronic-liver-diseases-icd|1.0.5'"
            },
        "location": ["code"]
        }
      ]
},…]}
```


See: https://build.fhir.org/valueset-operation-validate-code.html

ValueSet – $expand

- Returns the set of concepts included in a *ValueSet*
- Also allows for efficient code search
- Various options

See: http://www.hl7.org/fhir/valueset-operation-expand.html

ValueSet – $expand

- Returns the set of concepts included in a *ValueSet*
- Also allows for efficient code search
- Various options

  → **Expand of VS**
  - `{{server_url}}/ValueSet/$expand?url=http://fhir.de/ValueSet/bfarm/icd-10-gm`
  - Result: entire VS with all 17'065 ICD-10 codes

See: http://www.hl7.org/fhir/valueset-operation-expand.html

ValueSet – $expand

- Returns the set of concepts included in a *ValueSet*
- Also allows for efficient code search
- Various options

➔ **Expand of VS**

- `{{server_url}}/ValueSet/$expand?url=http://fhir.de/ValueSet/bfarm/icd-10-gm`
- Result: entire VS with all 17'065 ICD-10 codes

➔ **Using string filter**

- `{{server_url}}/ValueSet/$expand?url=http://fhir.de/ValueSet/bfarm/icd-10-gm&filter=diabetes`
- Result: 173 codes containing "diabetes" in the display name

See: http://www.hl7.org/fhir/valueset-operation-expand.html

ValueSet – $expand

- Returns the set of concepts included in a *ValueSet*
- Also allows for efficient code search
- Various options

→ Expand of VS

- {{server_url}}/ValueSet/$expand?url=http://fhir.de/ValueSet/bfarm/icd-10-gm
- Result: entire VS with all 17'065 ICD-10 codes

→ Using string filter

- {{server_url}}/ValueSet/$expand?url=http://fhir.de/ValueSet/bfarm/icd-10-gm&filter=diabetes
- Result: 173 codes containing "diabetes" in the display name

→ Using filter operator (+ SNOMED CT ECL)

- POST request – *descendent-of* filter

```
{ "resourceType": "Parameters",
  "parameter": [
    { "name": "valueSet",
      "resource": {
      "resourceType": "ValueSet",
      "compose": {
      "include": [
        { "system": "http://fhir.de/CodeSystem/bfarm/icd-10-gm",
          "filter": [
            { "property": "concept",
              "op": "descendent-of",
              "value": "E11.9“ }
          ]}
]}}}]}
```

- Result: 2 codes that are subnodes of E11.9

See: http://www.hl7.org/fhir/valueset-operation-expand.html

ConceptMap – $translate

- Mapping code from the scope of one *ValueSet* into a code of another *ValueSet*

- Required parameters
  - (url) – url of *ConceptMap*
  - system – url of *ValueSet* of source code
  - (target) – url of *ValueSet* of target code
  - code – relevant code

- Example (GET):
  - {{server_url}}/ConceptMap/$translate?
    url=https://mii-termserv.de/fhir/ConceptMap/
    ICD-O-Topography_to_SNOMED&
    system=http://hl7.org/fhir/sid/icd-o-3&
    target=http://snomed.info/sct&
    code=C80.9

Response:

```
{ "resourceType": "Parameters",
  "parameter": [
     { "name": "result","valueBoolean": true},
     { "name": "match",
       "part": [
              { "name": "equivalence",
          "valueCode": "equivalent"},
         { "name": "concept",
              "valueCoding":
              { "code": "87100004",
                "display": "Topography unknown“,
               "system": "http://snomed.info/sct"}
             },
             { "name": "source",
               "valueString": "https://imi.uni-
               luebeck.de/fhir/ConceptMap/
               icd-o-topo-sct"
             }
]}]}
```

See: http://www.hl7.org/fhir/conceptmap-operation-translate.html

Technical details

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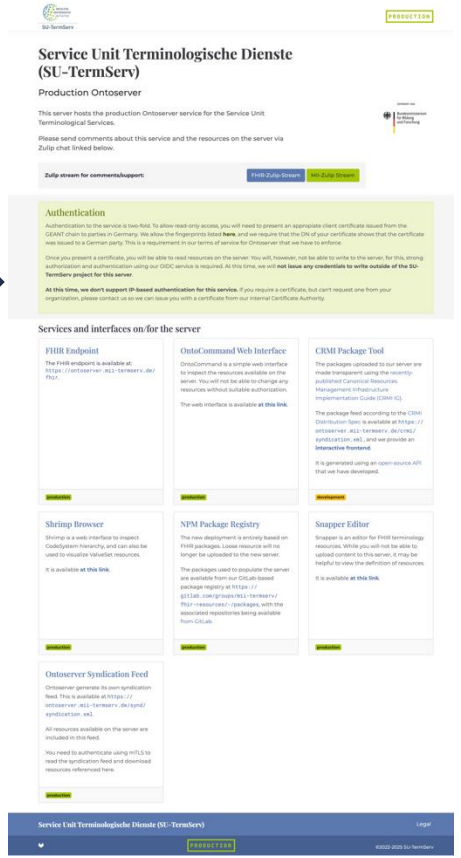
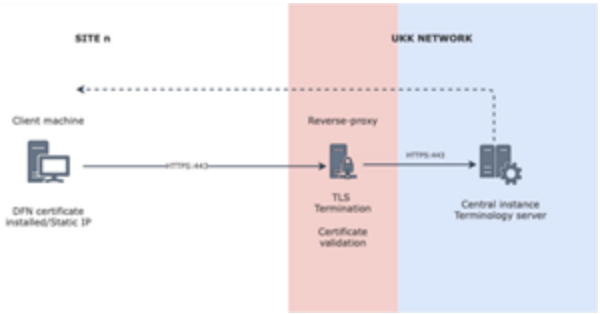
System setup

- Deployment via the Uniklinik Köln MeDIC
- Currently deployed at https://ontoserver.mii-termserv.de/fhir
  - Launch page: https://www.ontoserver.mii-termserv.de
- Read-only access enforced
- Access secured via two ways:
  - Mutual TLS via the DFN PKI – **preferred!**
  - Mutual TLS via internal fall-back CA

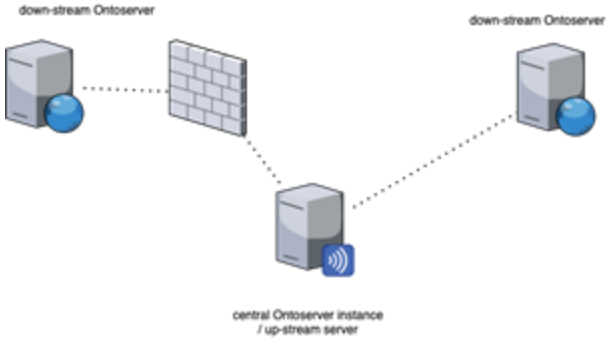


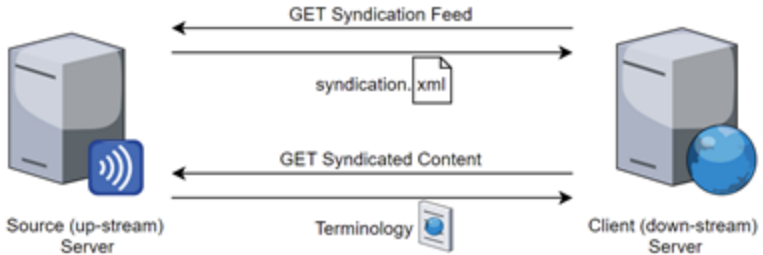


From: https://mii-termserv.de/assets/pdf/slides/workshop2023_june/session3_muhammad-adnan_ontoserver.pdf

Syndication

- Protocol to distribute terminology resources as a feed of resources
- Developed by CSIRO for Ontoserver, but currently being standardized as part of FHIR
- Automatic synchronization **not implemented**, loading resources has to be triggered manually
- Why?
  - Consistency across systems
  - Upstream as a single source of truth
  - Local availability of content - performance, uptime, scalability, security concerns





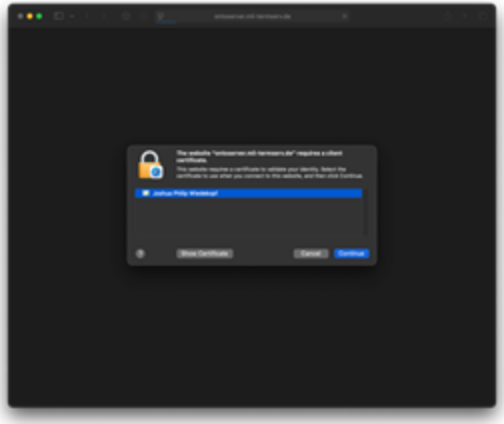
From: https://mii-termserv.de/assets/pdf/slides/workshop2023_june/session2_lukas-emmerich-terminologies_syndication.pdf

Setup of local servers

- Provision via a docker image; `docker-compose.yml` is available
- Ontoserver needs a lot of memory to index SNOMED CT from scratch, and not a lot if only using Syndication for this
- Syndication feed currently at: https://ontoserver.mii-termserv.de/synd/syndication.xml
- Configuration of the software using environment variables in the `docker-compose.yml` file
- Documentation for all variables in the Ontoserver documentation
    - Technical FAQ: https://s.mii-termserv.de/synd-setup

Technical access via PKI

- Certificates generally granted via the respective partner institution
- Supported client certificate profiles:
  - Personal certificates for users
  - Certificates issued to servers/DNS names (can also be used for HTTPS communication)
- Refer to your local IT department or the DFN for more information.
  - Through our fallback PKI, we can issue certificates ourselves.
- Certificate has to be provided to the OS/client system to access our Ontoserver
- Ontoserver: load into the container, set environment variables for Syndication



Services of the SU-TermServ

Established

- Provision of Ontoserver, a powerful FHIR-based terminology server and comprehensive terminology services
- Provision of “all” terminological resources required by the MII projects, with a focus on the Core Dataset of the MII
  - Continuous updating
- Package-based resource distribution
  - https://gitlab.com/mii-termserv/fhir-resources
- Conversion of non-FHIR terminology resources to FHIR (using own tool, BabelFSH)
- User Support, including advanced SNOMED CT usage

Planned as of early 2025

- Implementation & Provision of a synchronized minimal terminology server for the DICs for local connection to FHIR Validators
- Always: more and powerful tooling, UIs, …

Administrative aspects

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Getting access to the server

- Agreement between requesting institution and SU-TermServ mandatory, available on the website
- Write access is not granted, there is a workflow for requesting uploads

SU-TermServ

Vereinbarung zur Nutzung der Dienste der SU-TermServ

Für die Nutzung der Software Ontoserver (CSIRO), welche im Rahmen des Projektes Service Unit "Terminological Services" (SU-TermServ) der Medizininformatik-Initiative durch das Universitätsklinikum Köln zur Verfügung gestellt wird, versichere ich für den Gebrauch an meiner Institution, dass das Software Licence Agreement (SLA) mit der Firma CSIRO vom 23.01.2020 eingehalten wird. Dies beinhaltet ebenfalls den Abschnitt „Terms of Licence" des SLAs. Ich bestätige hiermit, dass uns das SLA mit der Firma CSIRO zugegangen ist, und ich erkläre mich mit den darin getroffenen Vereinbarungen einverstanden.

Weiterhin bestätige ich, dass die Bestimmungen des Benutzerhandbuchs („Terminology Server Services Concept") berücksichtigt werden.

Darüber hinaus stelle ich sicher, dass der Endgebrauch der Software, inklusive der enthaltenen Terminologien, nur in Deutschland stattfindet, dass meine Institution keinen externen Personen oder Institutionen (Dritten) Zugang zur Ontoserver-Instanz der SU-TermServ verschafft und, dass installierte Terminologien (z.B. SNOMED CT) nur, sofern zutreffend, mit entsprechender eigener Lizenz gemäß der entsprechenden Lizenzbedingungen der Ressourcen genutzt werden.

Die bereitgestellten Terminologien, die über den Synchronisationsmechanismus ("Syndication") bereitgestellt werden, fallen ebenfalls unter diese Vereinbarung. Dritte, die wiederum diese Dienste und Terminologien nutzen wollen, benötigen ebenfalls eine Nutzungsvereinbarung mit der SU-TermServ, um die Dienste und Terminologien nutzen zu können.

Unterschrift der zeichnungsberechtigten Person

Name und Funktion der zeichnungsberechtigten Person in Druckbuchstaben

Name der Institution

Anlagen:

- Nutzungshandbuch
- Service Level Agreement Ontoserver

Contact details

- SU-TermServ Mailing List: team@mail.mii-termserv.de
- MII-Zulip Stream *SU-TermServ*
- Our website mii-termserv.de is our default communication stream

Literature recommendations

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- FHIR
  - FHIR Docs; Terminology Module; Using Codes in Resources
  - Resource documentation for CodeSystem, ValueSet, ConceptMap
  - Benson & Grieve: Principles of Health Interoperability, 4th ed. 2021 (including parts on SNOMED CT and LOINC)
  - Braunstein: Health Informatics on FHIR: How HL7's API is Transforming Healthcare, 2nd ed. 2022

- SNOMED CT
  - SNOMED CT Foundation Course
  - BfArM (National Release Center)
  - Ingenerf & Drenkhahn: Referenzterminologie SNOMED CT, 1st ed. 2024
  - SNOMED International Confluence

- LOINC
  - Learn LOINC