



DIPARTIMENTO DI INGEGNERIA E SCIENZA DELL'INFORMAZIONE

- KnowDive Group -

# Fast Healthcare Interoperability Resources

Document Data: Reference Persons:

- date -

Shaun McNaughton, Jacopo Mocellin, Sander Martins Gonçalves, Zuhairia Ibnat

© 2020 University of Trento Trento, Italy

KnowDive (internal) reports are for internal only use within the KnowDive Group. They describe preliminary or instrumental work which should not be disclosed outside the group. KnowDive reports cannot be mentioned or cited by documents which are not KnowDive reports. KnowDive reports are the result of the collaborative work of members of the KnowDive group. The people whose names are in this page cannot be taken to be the authors of this report, but only the people who can better provide detailed information about its contents. Official, citable material produced by the KnowDive group may take any of the official Academic forms, for instance: Master and PhD theses, DISI technical reports, papers in conferences and journals, or books.

# Contents

1	Kno	owledge Graph Codebook	1
	1.1	Knowledge Graph general description	1
	1.2	Data level	1
		1.2.1 Datasets general details	1
		1.2.2 Datasets metadata documentation	1
	1.3	Ontology level	1
		1.3.1 Ontology general details	1
		1.3.2 Ontology metadata documentation	1
	1.4	Knowledge Graph Evaluation	2
2	Kno	owledge Graph Development Process	3
	2.1	Scope Definition	3
	2.2	Personas	4
	2.3	Scenarios	5
	2.4	Inception	6
		2.4.1 CQs definition	6
		2.4.2 Initial Datasets description	8
		2.4.3 Datasets generation method	8
		2.4.4 Datasets metadata documentation	6
		2.4.5 Datasets collection process	22
		2.4.6 Inception level evaluation	22
	2.5	Informal Modeling	24
		2.5.1 Schema level	24
		2.5.2 Data level	24
		2.5.3 Informal Modeling Evaluation	24
	2.6	Formal Modeling	25
		2.6.1 Schema level	25
		2.6.2 Data level	25
			25
	2.7		25
			26
		•	26

# Revision History:

Revision	Date	Author	Description of Changes
1.0	19/10/2020	Jacopo	Scope definition section written
1.1	21/10/2020	Jacopo	Inception section written
1.2	21/10/2020	Sander	Datasets generation method
1.3	21/10/2020	Shaun	General layout and editing
1.4	22/10/2020	Jacopo	Scope and inception Edits
1.5	22/10/2020	Zuhairia	Datasets Metadata Documentation written
1.6	22/10/2020	Shaun	Initial competency queries and scenarios added
1.7	23/10/2020	Sander	Metadata Synthea and Smart
1.8	23/10/2020	Zuhairia	Query edits

Page i / 26		
Page i / 26		
Page i / 26		
Page i / 26		

## 1 Knowledge Graph Codebook

The first of the two sections, in the current document, contains the codebook of the whole KG (Knowledge Graph), including the description of all the data and information that it contains.

## 1.1 Knowledge Graph general description

This sub section aims to give a general description of the KG, reporting:

- the context/domain in which the KG lives and works;
- The Problem the KG aims to solve;
- How the KG can solve The Problem

## 1.2 Data level

The data level section aims to describe in details the (final version of) datasets collected and managed by the KG, with a description of each variable involved.

## 1.2.1 Datasets general details

In this section are reported the metadata at datasets level, so the metadata regarding the sources, the authors, the collection methods, and so on.

#### 1.2.2 Datasets metadata documentation

In this section are reported the metadata at dataset attribute level, through a description of each variable involved in the datasets collected, specifying the variable types, meanings, value-set (possible values), and every other meaningful variable information.

## 1.3 Ontology level

The ontology level section aims to describe the underlying KG ontology, through the description of its elements at each level, reporting so the language, conceptual and schema resources used within it.

## 1.3.1 Ontology general details

This first sub section of the ontology level description, report the general details such as authors, sources and the description of external ontology eventually adopted to generate the final one.

## 1.3.2 Ontology metadata documentation

In this section instead, are reported the more specific metadata describing the single elements of the ontology (terms, concepts, ETypes and relations).

## 1.4 Knowledge Graph Evaluation

In the final section of this first chapter, the KG Evaluation is reported. It aims to describe, through specific metrics, the quality of the overall KG on different aspect, like domain coverage, usability, domain representation, and other meaningful aspects.

## 2 Knowledge Graph Development Process

The second chapter of this document aims to describe, in a detailed way, the KG development process. The sections below describe each phase of the KG building project, reporting for each phase, the description of the datasets and their evolution respect the previous phases, the schema construction which will generate the KG ontology in the end, as well as the description of the procedures adopted to manage the data and finally achieve those results. Moreover for each phase is reported an evaluation section, which aims to evaluate the quality of the results achieved at the end of each phase.

## 2.1 Scope Definition

In this part of the project report, we provide the foundational specifications for the further development of our data integration project. In particular, we are going to outline what is the problem our solution is going to address, why we chose that problem, what is the context in which the problem exists in, and why the iTelos methodology is the most suitable technical approach to solving the problem.

EHRs (Electronic Health Records) are systems that allow to store all medical data concerning a patient in a digital format. While innovation could allow increased portability across different health organizations, current EHR implementations offer limited transferability to other systems because of several layers of obstacles. This problem is formalized as a problem of data interoperability.

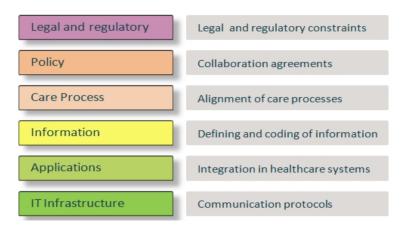


Figure 1: (from eHN's Refined eHealth European Interoperability Framework)

While several layers of complexity need to be addressed in order to achieve complete interoperability, we are going to focus our efforts to implementing a solution that aligns semantic differences across different EHR systems, and therefore our intervention can be ascribed to the "Information" layer of this scheme. This layer is comprised of all aspects of the data model, including coding terminology and the formatting of the medium for transportation of the information.

The problem of incompatible data representations is particularly taxing in Europe, where, on one hand, free circulation of EU citizens across the 27 member states is fostered by a friendly regulatory scenario, but, on the other hand, seamless circulation of citizen's health data has been hindered by the lack of complete interoperability.

In the current European medical context, the eHDSI (e-Health Digital Service Infrastructure) connects eHealth national contact points allowing them to exchange two sets of health data: patient summaries and ePrescriptions. This system serves as reference in the field of interoperable EHRs.

Recently, the European Commission produced and adopted a recommendation on a framework for the further development of a European EHR exchange format that integrates what already accomplished by the eHDSI project. This consists of a set of common technical specifications and principles that will enable citizens to securely access and exchange their health data across borders in the EU.

In the annex to the Commission Recommendation on a European Electronic Health Record exchange format, a total of five types of data are defined as target of a European cross-border exchange solution:

- Patient Summaries
- ePrescriptions/eDispensations
- Laboratory reports
- Medical images and reports
- Hospital discharge reports

In line with this proposed framework, while selecting the types of data that our data integration system should be able to interpret, we opted for restricting our focus to those mentioned above in order to adhere with the requirements of a realistic EHR solution existing in the current European context described.

In this problem space, the iTelos methodology proves to be the most appropriate because it allows the alignment of information that in our case, coming from different source datasets, is scattered and represented in a diversified manner.

## 2.2 Personas

As a top level view, a solution that could help foster data interoperability could benefit several stakeholders in the healthcare panorama:

- Healthcare Facilities: could reduce costs and inefficiencies by reducing the amount of testing needed
- Healthcare professionals: could save time by not having to enter information by hand in a system. Could become able to interpret information in different languages. Could see the history of a patient: reaction to medications, previous operations, allergies, and other intolerances
- Patients: enables to consult a specialist or receive emergency treatment in another EU country
- Researchers: Could enable to recruit a diverse population for clinical studies while being able to access accurate retrospective reporting information about the patients.

Among these, we chose to focus on developing a solution that could fit primarily the needs of patients and health-care professionals taken as a unique dyad of subjects cooperating to foster better outcomes in the therapeutic process

We will now illustrate some proposed personas and relative hypothetical use cases.

- Giannantonio, aged 35, is an Italian sales representative for an important clothing brand who is asked to travel frequently from one country to the other for work. His agenda is made up by appointments fixed with very little advance, which doesn't allow him to have much time to prepare luggage for his business trips or to bring extra paperwork along with him. He has good understanding of English, but not in a medical domain.
- Rita is an Italian woman, aged 73, who lives in a rural town near Caltanissetta. Once a year she travels to the Netherlands to visit her son and his family who emigrated there. She has no knowledge of foreign languages.
- Pedro is a Spanish man aged 45 who was recently diagnosed with Parkinson disease. He is travelling to Germany constantly in order to receive life-saving treatments. However, his clinical situation is complex since he suffers several co-occurring chronic conditions.
- Jean is a 52 year old, General Practitioner from France. He practices in a small clinic near Rennes, but is also working as a Forensic Medical Officer for the French police. While he loves both of his jobs, the additional stress from COVID-19 has been pressuring him to focus on a single job.

## 2.3 Scenarios

As the exchange of health information is an inherently complex and has widely varying needs, we use the personas and expand on them by creating a hypothetical scenarios to which having integrated health systems could be used.

Giannantonio: During one of his frequent trips to Greece, he is struck down by a heart attack. Not having any form of medical documentation with him, the medical team helping him needs to retrieve information elsewhere in order to know which treatments he might not respond well to. [Access to patient history]

Rita: During one of her visits to the Netherlands, Rita falls to the ground and breaks an arm. Even though her condition is not severe, the physician recommends close supervision by a colleague upon hospital discharge in order to avoid incorrect calcification of her fragile bones. Once back home, Rita is aware of the need to consult with her trusted physician in order to avoid complications, but would not be able to report to him any of the indications given by the Dutch doctor. On the other hand, her trusted doctor is unable to understand languages other than Italian. [Access to discharge reports data]

**Pedro**: Due to his complex medical conditions, the different medical teams who treat him in the two different countries need to be constantly updated on prior medications administered in order to inform their future clinical decisions. [Access to patient history]

Jean:Before seeing a new patient, Jean spends time reviewing that patient's medical history. He does this to better understand the patient at both a physical and emotional level. This in turn, allows him to mentally prepare for the consultation and to act in a professional manner. In doing so, he wishes to review at least any basic health information on the patient, for example age, blood type, and past medical history before seeing them. Now, he

wishes to access medical records from an Italian hospital to help diagnose a patient who has recently arrived in France and has a history of hospitalisations. [Access to patient history]

## 2.4 Inception

This section is dedicated to the Inception phase description. Here are reported the initial definitions for CQs (Competency Queries), initial datasets collected and the relative metadata. For each of those elements the procedures and the tools adopted to achieve the results, have to be reported in the sections below.

## 2.4.1 CQs definition

In order to create the competency queries, we use the scenarios and persona descriptions provided in the previous section, to come up with possible queries that a person could want from the system.

The scenarios provide a set of actions for which a competency query can be modelled from. These actions and the mapping to the relevant competence query for each scenario is given below.

In future iterations, we might consider shrinking our focus to a narrower set of pathologies (and therefore queries) since our current dataset provide a data about patients with a broad and diverse set of conditions that could prove to be too extensive to represent in the final DKG.

Scenario	Question	Action	Description
Giannantonio	1.1	Access to patient	Access to known
		history	allergies
			documentation.
			Access to history of
			previous circulatory
			diseases. Access to
			history of
			circulatory related
			interventions
			(previous
			surgeries). Access
			to medications
			taken with timeline
Rita	2.1	Access to discharge	Access to diagnostic
		reports	reports generated
			by the hospital
			upon receiving the
			patient. Access to
			physician's written
			evaluations. Access
			to Imaging
			Laboratory reports.
Pedro	3.1	Access to patient	Access to previous
		history	conditions. Access
			to previously
			administered
			medications, as
			reported by the
			physician in charge
Jean	4.1	Access to patient	Access to
		history	diagnostic reports
			as written by prior
			physicians. Access
			to allergies
			documentation.
			Access to treatment
			and travelling
			timeline.
			timeline.

## 2.4.2 Initial Datasets description

In this section are reported the metadata at datasets level involved in the Inception phase, so those metadata regarding the sources, the authors, the collection methods, and other meaningful information.

For our data integration process, we had to rely completely on synthetic data. The collection of genuine, real-world data is almost impossible because of the special privacy measures that are in place to protect this kind of personal data. This means that real-world data is often protected and needs some kind of authentication to be accessed. Nonetheless, we were able to find satisfying patient data generators by looking into the solutions developed to test other existing EHR systems. Specifically, we are going to provide a list of found datasets and their metadata here.

- EMRBOTS dataset (http://www.emrbots.org): The dataset was created by Uri Kartoun, PhD, in 2015. We are using the 100000-patients dataset, which comprises in total data about 100.000 patients, 361.760 admissions, and 107.535.387 lab observation. The dataset was generated according to the literature on patient population studies in order to create a realistic set of observations. More details about the data can be found on table 1 bellow.
- Synthea patient generator DBs (https://github.com/synthetichealth/synthea): This tool was developed by the MITRE corporation and released freely under an Apache license. The datasets generated with this tool can include data about over 90 different conditions thanks to its modular structure. Data generated includes: Conditions, Allergies, Medications, Vaccinations, Observations/Vitals, Labs, Procedures, CarePlans, Primary Care Encounters, Emergency Room Encounters, and Symptom-Driven Encounters. This data is matched with Configuration-based statistics and demographics. The data can be formatted in unstructured formats or structured (JSON) ones. The latter also allows for formatting according to HL7 standards. The generator can be queried thanks to a Java-based interface.
- Smart on FHIR data generator (https://github.com/smart-on-fhir/sample-patients): This tools is provided as a test suite for SMART on FHIR applications by SMART itself. We are going to exploit it to generate structured data that fits the FHIR specifications about a population of patients.

[Ultimately, our goal is to coherently aggregate this data in order to form the population of patients that is going to be represented in our DKG.

The possibilities are either to keep the datasets separate or to build a single dataset by merging the ones mentioned above, assigning to single patients from the EMRBOTS dataset (or a subset of it) data from the other datasets. We are aware that these datasets currently do not refer to the same population, this we are going to focus on retaining validity in the case of performing this task. In this case, the final result is going to be a single dataset containing very diverse and overlapping data on a set of patients. This data is going to be in different formats, therefore constituting the ground for usage of a DKG to align diversity.

## 2.4.3 Datasets generation method

## **EMRBOTS**

EMRBOTS provided the method used for the creation of the dataset. The process starts by setting the desired

configuration of the population. This means that is possible to set the percentage of gender, marital status, major language, ethnicity, date of birth and income level.

Once this pre configuration is made the next step is the generation of the individuals. For each generated patient, will be created a record that will be its lifetime electronic medical record (EMR). This record is created by randomly assigning the number of admissions, the length of each admission, the result of laboratory exams and the code for the main symptoms of the patient according to the list of International Classification of Diseases (ICD-10-CM). Laboratory results means a list with the measurements of levels of sodium, creatinine, or platelet counts, for example.

## Synthea

Synthea is based on the PADARSER framework. This framework assumes that access to EHR real data is not available and therefore relies on public data to compose the population to be used for the generation of synthetic EHR. These public data are composed by aggregation of health incident statistics, clinical practice guidelines (used to develop caremaps) and medical coding dictionaries. These public will then feed the information and knowledge model in order to generate synthetic data that is composed by patient profiles, diseases and their prevalences, caremaps and coded narratives. Synthea uses Health Level-7 (HL7) the main codification system.

#### 2.4.4 Datasets metadata documentation

In this section are reported the new metadata at attribute level, describing the types, meanings, value-set, validity, and so on, of each variable within the initial dataset collected. The dataset created by EMBROTS has the following attributes:

- Patient ID: A unique number representing a particular patient
- Gender: The sex of a patient; either male or female
- Birth: The date of the birth of the patient
- Race: This describes the ethnicity of the patient. The categories are white, Asian African-American, unknown
- Marital Status: The categories of marital status are married, single, divorced, separated, widowed, and unknown
- Language: The preferable language of the patients are mentioned here
- Population percentage below poverty: This category is represented in percentage
- Admission ID: An admission ID for the patient
- Admission start date: The date the patient was admitted
- Admission end date: The date the patient was released
- Primary diagnosis code: This is an ICD10 code for the primary disease diagnosis
- Primary diagnosis description: The description of the diagnosis is mentioned

Variable and Category	Patients (n=100,000)
Mean age as of $1/1/2015$ , years (SD)	57.8 (17.3)
Gender (%)	
Female	52.0
Ethnicity (%)	
White	49.0
Asian	23.0
African American	15.0
Unknown	13.0
Mean number of admissions per patient,	
days (SD)	3.6 (1.5)
Mean length of stay (SD)	11.0 (5.2)
% Population with length of follow-up (years)	
0 - 9	13.1
10 - 15	9.3
> 15	77.6
Population below poverty (%)	21.6
Comorbidities; Prevalence (%)	
Malignant neoplasm	41.4
Rheumatoid arthritis	25.6
Diabetes (type I or II)	24.4
Renal complications	17.0
Coronary artery disease	7.0
Laboratory values (Mean; SD)	
Blood urea nitrogen (mg/dL)	17.5; 7.2
Platelets (k/cumm)	284.9; 95.3
Creatinine (mg/dL)	0.9; 0.2
Albumin (gm/dL)	4.2; 1.0
Lymphocytes (%)	25; 5.8

Table 1: Data setting - size 100.000 - EMRBOTS

- Lab name: This category includes the tests of the patients. The CBC tests are: White blood cell count, Red blood cell count, Haemoglobin, Hematocrit, Mean corposcular volume, MCH, MCHC, RDW, Platelet count, Absolute Neutrophils, Absolute lymphocytes, Neutrophils, Lymphocytes, Monocytes, Eosonophils, Basophils. The metabolic tests are: Sodium, Potassium, Chloride, Carbon dioxide, Anion gap, glucose, BUN, creatinine, total protein, Albumin, Calcium, Bili total, AST/SGOT, ALT/SGPT, ALK PHOS. The urinalysis tests include: pH, specific gravity, red blood cells, white blood cells
- Lab value: This represents the numeric value of the tests
- Lab units: This attribute shows the unit of the lab tests
- Lab date: The date of the lab tests is shown in this attribute

The dataset created by Synthea has the following attributes: Information related to the patient:

- Patient ID: A unique number representing a particular patient
- Name: Corresponding name of the patient

- Gender: The sex of a patient; either male or female
- Birth: The date of the birth of the patient
- Race: This describes the ethnicity of the patient. The categories are white, Asian African-American, unknown
- Marital Status: The categories of marital status are married, single, divorced, separated, widowed, and unknown
- Address: Information related to the residence of the person. It is composed by Country and City
- Language: The primary language spoken by the patient
- : Education level: Categorical variable indicating the education level of the patient
- Identification: Documents of the patient, could be passport, driver's id or other document
- Allergy: Information if the patient has any kind of allergy of food or medication
- Diagnostic: Information related to the identified disease with its related code (SNOMED CT format)
- Treatment: The treatment given to the patient for treating its condition
- Dosage: Information of the quantity of medicine was prescribed
- Period: Information related to the beginning and the ending of the admissions
- Immunizations: The code of the vaccine applied or NA in case it was not prescribed

Other variables related to the diagnostics or other information:

Age	Age months	Alzheimer's	BMI	Breast Cancer
		Medication		
CF	Cystic Fibrosis	DALY	ER PR	ER medication
Complication			medication	
Early	G155D	Glucose	HER2	Hypoxemia
menopause due	Mutation		medication	
to treatment				
Left ventricular	Lung cancer	Lung cancer	Lung cancer	MDD care plan
Ejection		condition	type	
fraction				
Memory Loss	Mucus	OTC	Oxygen	QALY
		Bronchitis Med	Saturation	
QOLS	RH NEG	SSRI	Septic Shock	Sinusitis
				Condition

- AGE
- AGE MONTHS
- Alzheimer's Medication

- $\bullet$  BMI
- Breast Cancer
- CF Complication
- Cystic Fibrosis
- DALY
- ER PR medication
- ER medication
- Early menopause due to treatment
- $\bullet$  G155D Mutation
- Glucose
- HER2 medication
- Hypoxemia
- Left ventricular Ejection fraction
- Lung Cancer
- Lung Cancer Condition
- Lung Cancer Type
- MDD care plan
- Memory Loss
- Mucus
- OTC Bronchitis Med
- Oxygen Saturation
- QALY
- $\bullet$  QOLS
- RH NEG
- SSRI
- $\bullet\,$  Septic Shock
- Sinusitis Condition
- Sore Throat Antibiotic

- Sore Throat Condition
- Suspected Lung Cancer
- Systolic Blood Pressure
- Triglycerides
- Type of Alzheimer's
- Unusual Vaginal Bleeding
- Ventilator Reason
- active emergency encounter
- $\bullet\,$  active urgent care encounter
- ullet active wellness encounter
- $\bullet$  address
- adhd medication
- $\bullet$  adherence
- adherence probability
- adv drug
- alcoholic
- alcoholism
- allergic rhinitis
- anemia
- anemia medication
- anemia pregnancy
- $\bullet$  antibiotic prescription
- ullet arthritis careplan
- $\bullet$  assessment done
- $\bullet\,$ asthma careplan
- asthma condition
- $\bullet$  asthma type
- $\bullet$  atopic

- atrial fibrillation
- atrial fibrillation risk
- birth city
- birth country
- birth state
- birth type
- birthdate
- birthplace
- blindness
- blood pressure controlled
- bmi percentile
- breast cancer ER
- breast cancer HER2
- breast cancer Location
- breast cancer M
- breast cancer N
- breast cancer PR
- breast cancer T
- breast cancer aromatase inhibitors
- breast cancer chemoCount
- breast cancer condition
- breast cancer init followup counter
- breast cancer neoCount
- breast cancer neoMed
- breast cancer survival
- breast cancer tamoxifen
- breast cancer treat count
- breast cancer triple negative

- breast cancer years after treatment
- broken bone
- bronchitis careplan
- burn injury
- cardio risk
- cardiovascular disease med changes
- cardiovascular procedures
- cause of death
- $\bullet$  chemoMed
- chemotherapy1
- $\bullet$  chemotherapy2
- chf
- chf admissions
- ullet chf emergency admissions
- $\bullet\,$  chf icu admissions
- chf inpatient admissions
- chf med check
- chf med step
- chf stage d
- chf type
- ckd
- ullet colorectal adenoma
- colorectal cancer
- colorectal cancer stage
- concussion injury
- $\bullet$  contraceptive
- ullet contraceptive type
- copd variant

- coronary heart disease
- covid19
- $\bullet\,$  covid 19 admission days
- covid19 all labs
- covid19 bacterial infection
- covid19 c reactive protein
- covid19 cardiac troponin
- covid19 careplan
- covid19 clinical trial
- covid19 clinical trial duration
- covid19 clot
- covid19 creatine kinase
- covid19 ddimer
- covid19 death
- covid19 diagnosed clot
- ullet covid19 extubation recovery days
- covid19 ferritin
- covid19 hospitalization days
- covid19 icu days
- covid19 inr
- covid19 interleukin6
- covid19 lactate dehydrogenase
- $\bullet\,$ covid 19 laryngoscope
- covid19 lymphocytes
- $\bullet$  covid19 pao2
- covid19 procalcitonin
- $\bullet$  covid19 pt
- $\bullet$  covid19 risk

- covid19 severity
- covid19 ventilator
- ullet cr chemo count
- current weight length percentile
- dermatitis
- diabetes
- diabetes amputation necessary
- diabetes severity
- diabetes stage
- diabetic eye damage
- diabetic nerve damage
- dialysis reason
- $\bullet$  ecg result
- education level
- $\bullet\,$ end stage renal disease
- $\bullet$  ethnicity
- fatal pregnancy complication
- fibromyalgia careplan
- fibromyalgia prescription
- $\bullet\,$  first language
- first name
- food allergy
- ullet food allergy careplan
- gender
- $\bullet$  gout careplan
- gout nsaid
- $\bullet$  gunshot wound
- homeless

- ullet homelessness category
- $\bullet\,$ hyperglycemia
- ullet hyperlipidemia
- hypertension
- hypertension dx
- hypertension medication
- ullet hypertension medication 2
- ullet hypertension medication 3
- ullet hypertension medication step
- hypertension severe
- hypertriglyceridemia
- hypothyroidism
- $\bullet$  id
- identifier drivers
- $\bullet\,$ identifier passport
- identifier ssn
- immunizations
- $\bullet$  immunotherapy counter
- immunotherapy status
- $\bullet$  income
- infertile
- injury careplan
- inpatient-los
- instances of homelessness
- $\bullet$  joint replacement
- knee injury
- $\bullet$  laceration injury
- $\bullet$  last name

- ullet last visit symptom total
- ullet left ventricular hypertrophy
- $\bullet$  lung cancer
- $\bullet\,$ lung cancer nondiagnosis counter
- $\bullet$  lung transplant
- lupus careplan
- ullet lupus corticosteroid
- $\bullet$  mTBI
- $\bullet\,$  macular edema
- maiden name
- marital status
- $\bullet$  mdd
- $\bullet$  mdd psych
- ullet metabolic syndrome
- $\bullet\,$ microalbuminuria
- miscarriage
- most-recent-daly
- most-recent-qaly
- multiple birth status
- name
- name father
- name mother
- $\bullet$  name prefix
- name suffix
- $\bullet$  nephropathy
- $\bullet$  neuropathy
- non low risk pregnancy
- ullet nonproliferative retinopathy

- number of children
- nyha class
- obesity
- $\bullet$  occupation level
- opioid addiction
- opioid addiction careplan
- opioid prescription
- $\bullet$  osteoarthritis
- osteoporosis
- otc antihistamine
- otc pain reliever
- ullet outgrew food allergies
- $\bullet$  prediabetes
- $\bullet$  pregnancy birth sex
- ullet pregnancy careplan
- ullet pregnancy complication
- pregnant
- probability of fall injury
- probability of lung cancer
- probability of suicide
- proliferative retinopathy
- prostate cancer
- proteinuria
- ptsd
- ullet ptsd careplan
- $\bullet\,$  quit alcoholism age
- quit alcoholism probability
- quit smoking age

- quit smoking probability
- $\bullet\,$ ra careplan
- race
- recurrent colorectal adenoma
- $\bullet$  retinopathy
- seizure
- seizure meds
- sexual orientation
- sexually active
- $\bullet$  smoker
- smoking
- $\bullet$  smoking medication
- ullet socioeconomic category
- $\bullet$  spinal injury
- sprain injury
- ssri
- $\bullet$  statin initial
- statin renewal
- stroke history
- $\bullet$  stroke points
- stroke risk
- suicide
- $\bullet\,$  suicide attempt
- suicide careplan
- suicide risk
- $\bullet$  telecom
- $\bullet$  text
- $\bullet$  uti

- veteran
- veteran population override
- visit allergist
- watchful waiting
- zip

The dataset created by Smart has the following attributes:

- Patient ID: A unique number representing a particular patient
- Name: Corresponding name of the patient
- Birth: The date of the birth of the patient
- Diagnostic: Information related to the identified disease
- Treatment: The treatment given to the patient for treating its condition
- Period: Information related to the beginning and the ending of the admissions

## 2.4.5 Datasets collection process

The collection process section aims to describe the collection methods adopted to obtain the initial datasets in this phase, as well as the tools used.

The collection has proven to be challenging for reasons mentioned earlier: Health data constitutes sensitive information and is therefore always protected with special provisions. Access to real-world health data most of the times is granted only via an accreditation process (As is the case for the MIMIC dataset, including data about real-world intensive care unit stays). After clashing with this bureaucratic obstacles, we started looking into free-to-use, open solutions available on the internet. We were finally able to find resources given by communities active in both academia and industry. The community behind the development of current EMR solutions has implemented testing datasets or dataset-generation tools that we eventually opted for using in our solution.

## 2.4.6 Inception level evaluation

We initially struggled with the initial concept behind what we were required, first to find data sources that were relevant and secondly to create a set of personas for which we could then create queries from. However, we can evaluate our performance of this stage of this phase by considering the following specific KPIs:

- 1. Number of datasets found: The current number of data sources amounts to 3, which is in line with the given indications.
- 2. Diversity of data collected: The number of datasets creates sufficient diversity in the data gathered. In fact, data collected is diverse in format (structured vs unstructured) and in representations (e.g. measurement units)

3.	Coherence of data with purpose: Given the found data sources represent information in the medical domain, we are reassured that it fits coherently with the purpose of integrating medical information.

## 2.5 Informal Modeling

This section is dedicated to the Informal Modeling phase description. The Section is divided in Schema and Data level in order to report the details of the elements involved in the generation of the schema, as well as the description of the datasets evolution in this phase. Moreover a specif section, one for each level, reports the difference between the elements defined in this phase and the definitions in the previous phase, analyzing in this way the variance in the different phases.

#### 2.5.1 Schema level

The schema level in this phase report the first informal definition of the ETypes and of the EER model constructed using them.

## 2.5.1.1 ETypes and EER Model definition

This section reports an informal definition of the ETypes involved in the datasets collected in the previous phase. This section includes a list of metadata associated to each of the elements generated.

## 2.5.1.2 Variance respect CQs definition

This section aims to define the variance between the schema elements produced in this phase, and the definition of the CQs reported in the previous phase. This a way to define the quality of the outcomes for the current phase as well as the alignment of the overall project development process.

#### 2.5.2 Data level

The data level section in this phase reports the evolution of the datasets collected previously, reporting the metadata information for each new data, or new version of data, obtained.

### 2.5.2.1 Datasets management process

During the Informal Modeling phase the datasets collected in the previous phase are filtered and managed in order to obtain more suitable sets of data. In this section are described the procedures adopted to obtain that result.

#### 2.5.2.2 Datasets metadata documentation

In this section is reported a list of new metadata in order to describe the modification performed on each datasets and attribute, to achieve the new version of the datasets.

#### 2.5.2.3 Variance respect Inception datasets

This section aims to define the variance between the data elements (datasets and attributes within them) produced in this phase, and the initial datasets collected in the previous phase. This a way to define the quality of the outcomes for the current phase as well as the alignment of the overall project development process.

## 2.5.3 Informal Modeling Evaluation

The last section of the Informal Modeling phase report the evaluation of the outcomes obtained in this phase, through specif evaluation metrics.

## 2.6 Formal Modeling

This section is dedicated to the Formal Modeling phase description. The Section is divided in Schema and Data level in order to report the details regarding both the ontology generated and the datasets version in the current phase.

#### 2.6.1 Schema level

The schema level section in the current phase, reports the detailed description of the ontology generation.

## 2.6.1.1 Ontology definition

This section reports in details how the ontology is generated stating from the informal schema of the previous phase, which tools are used to do that, as well as usage of external ontology resources adopted to obtain the final KG ontology. Moreover a list of metadata is reported in this section, in order to describe all the elements of the ontology defined.

## 2.6.1.2 Variance respect to the EER Model

Once the ontology has been built, this section report the differences, and so the variance, respect the EER model defined in the previous phase. This a way to define the quality of the outcomes for the current phase as well as the alignment of the overall project development process.

#### 2.6.2 Data level

As in the previous phase the data level section here, reports the description of the new version of the datasets, after formatting operations.

#### 2.6.2.1 Formal Modeling datasets management

In this section are reported the operations and the tools adopted to format the dataset collected, in order to align them to the ontology definitions generated at schema level.

#### 2.6.2.2 Datasets metadata documentation

In this section eventually new metadata information are added in order to describe the evolution of the datasets.

## 2.6.2.3 Variance respect Informal Modeling datasets

This section aims to define the variance between the data elements (datasets and attributes within them) produced in this phase, and the initial datasets collected in the previous phase. This a way to define the quality of the outcomes for the current phase as well as the alignment of the overall project development process.

## 2.6.3 Formal Modeling Evaluation

The last section of the Formal Modeling phase report the evaluation of the outcomes obtained in this phase, through specif evaluation metrics.

## 2.7 Data integration

This section is dedicated to the Data Integration phase description.

## 2.7.1 Data integration operations and tool

This section is dedicated to the description of the usage of the data integration tool that allows to map the datasets generated and well formatted in the previous phases, with the final ontology generated. The last datasets adaptation performed using the tool, as well as the mapping operation are here detailed.

## 2.7.2 Variance respect Formal Modeling datasets

The last section of the data integration phase aims to describe the variance, analyzing the differences, between the datasets integrated with the ontology, in the data integration platform which contain the KG, and the datasets collected in the previous phase. This analysis can highlight the results of the operations performed during the final phase of the data integration process.