

RD-CODE Final Workshop

COLLABORATING PROJECTS: X-eHEALTH

Giorgio Cangioli
Technical Lead – HL7 Europe
X-eHealth WP6 leader



X-eHealth

Exchanging Electronic Health Records
in a common framework

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant. Agreement N° 951938



The slide features two large, semi-transparent orange shapes on the left and right sides. These shapes contain stylized molecular diagrams with red and yellow nodes connected by lines. A smaller, similar diagram is located at the bottom left, and another is at the bottom right.

PS FOR RARE DISEASES

HOW IT SERVES RD/ NEW CONTENT FIELDS, DATA MODEL



Co-funded by the European
Union's Health Programme
(2014-2020)



X-eHealth

Exchanging Electronic Health Records
in a common framework

PROJECT SCOPE

Laboratory Results



Medical Imaging



Rare Diseases



Discharge Letters



Use Cases

UC5.3.1	Laboratory results report
UC5.3.2	Laboratory test order from healthcare provider
UC5.3.3	Querying of lab results
UC5.3.4	Querying of lab orders
<..>	<...>



Co-funded by the European Union's Health Programme (2014-2020)



X-eHealth

Exchanging Electronic Health Records in a common framework

Unplanned healthcare



☐ including emergency/surgery

Planned healthcare

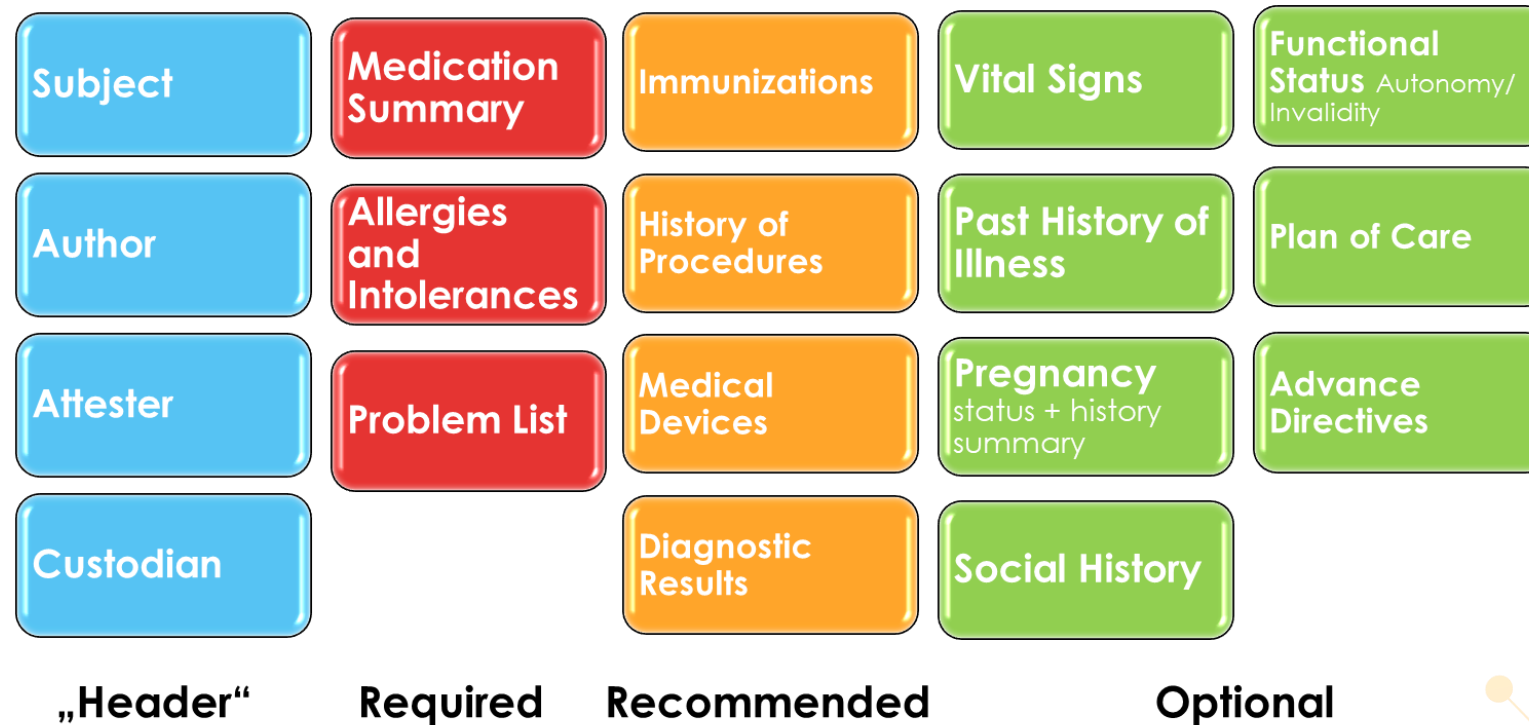


- ☐ Second opinion/multidisciplinary (MDT) consultations
- ☐ Internal consultations
- ☐ Consultations about availability of ongoing clinical studies/eligibility check
- ☐ Concise summary for patients

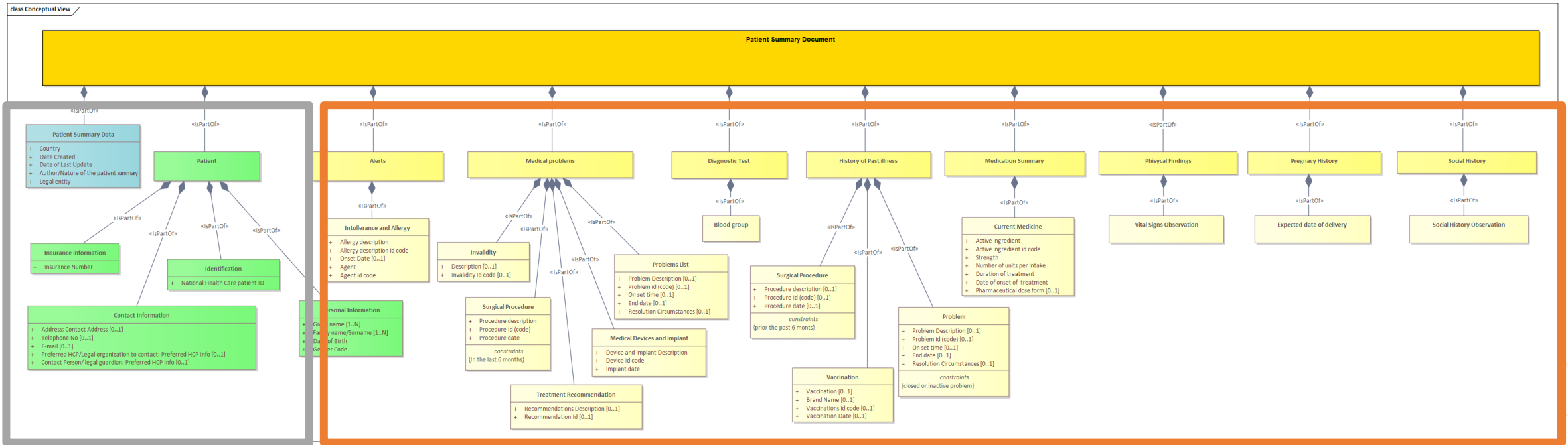




The International Patient Summary



The European Patient Summary data set



Context

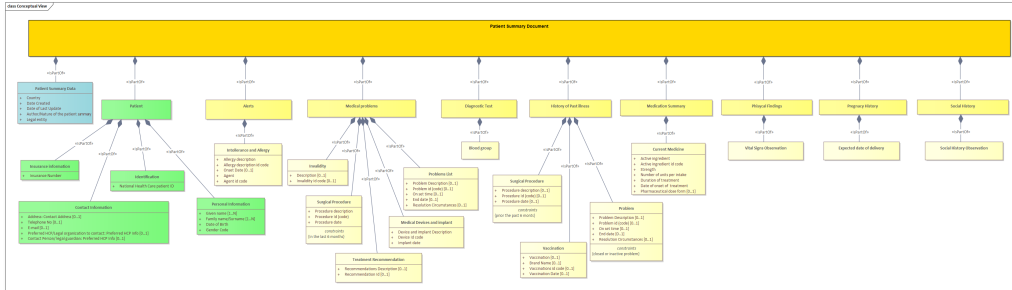
Clinical Content

[https://webgate.ec.europa.eu/fpfis/wikis/display/EHDSI/05.01.+Create+the+eHDSI+Patient+Summary+content#id-05.01.CreatetheeHDSIPatientSummarycontent-3.PATIENTSUMMARYDATA\(InformationaboutthePatientSummaryitself\)](https://webgate.ec.europa.eu/fpfis/wikis/display/EHDSI/05.01.+Create+the+eHDSI+Patient+Summary+content#id-05.01.CreatetheeHDSIPatientSummarycontent-3.PATIENTSUMMARYDATA(InformationaboutthePatientSummaryitself))

Implementation Independent



Co-funded by the European Union's Health Programme (2014-2020)



	Use case:					
	Unplanned care (incl. emergency/surgery)	ERN consultation	MDT Board consultation, second opinion, including internal hospital consultations	GP	Clinical study availability	Summary for patients
RD diagnosis	X	X	X	X	X	X
Disease name	X	X	X	X	X	X
ORPHAcode	X	X	X	X	X	X
Diagnosis assertion status	X	X	X	X	X	X
Method for diagnosis confirmation		X	X	X	X	X
Causative gene (if relevant)		X	X	X	X	X
Gene variant (if relevant)		X	X	X	X	X
Disease history		X	X	X	X	X
Age of onset		X	X	X	X	X
Age at diagnosis		X	X	X	X	X
Heredity (if relevant)		X	X	X	X	X
Clinical manifestations		X	X	X	X	X
Investigations (type and results)		X	X	X	X	X
Disabilities		X	X	X	X	X
Antenatal/perinatal information (if relevant)		X	X	X	X	X
Birth date		X	X	X	X	X
Gender at birth		X	X	X	X	X
Malformation detected antenatally		X	X	X	X	X
Medically assisted procreation (yes/no)		X	X	X	X	X
Gestational age at birth (weeks)		X	X	X	X	X
Weight at birth		X	X	X	X	X
Height at birth		X	X	X	X	X
Head circumference at birth		X	X	X	X	X
Family antecedents in relation with the RD		X	X	X	X	X
Affected member (propositus) yes/no		X	X	X	X	X
Parental relationship with the affected member		X	X	X	X	X
Consanguinity: yes/no/unknown		X	X	X	X	X
Other relevant familial antecedents (i.e. cancer)		X	X	X	X	X
Treatments	X	X	X	X	X	X
Treatment specific for the RD: yes/no	X	X	X	X	X	X
Name and posology of the ongoing treatment(s) (RD specific; others)	X	X	X	X	X	X
Care pathway	X	X	X	X	X	X
Name of the physician in charge of the patient:	X	X	X	X	X	X
Rare disease centre following the patient: name of the centre/hospital/ location/phone number (incl. emergency phone number)	X	X	X	X	X	X
Is the RD centre part of an ERN (yes/no)	X	X	X	X	X	X
ERN name	X	X	X	X	X	X
Useful information	X	X	X	X	X	X
url leading to information on the disease, including CPGs, emergency and anesthesia guidelines	X	X	X	X	X	X
Main alert messages	X	X	X	X	X	X

	Implemented case (incl. emergency/urgent)	Core variables	Other variables (selected genes, including relevant clinical manifestations)	ER	Clinical data availability	Summary for patients
RD diagnosis	X	X	X	X	X	X
Disease name	X	X	X	X	X	X
ORPHAcode	X	X	X	X	X	X
Diagnosis assertion status	X	X	X	X	X	X
Method for diagnosis confirmation	X	X	X	X	X	X
Causative gene (if relevant)	X	X	X	X	X	X
Gene variant (if relevant)	X	X	X	X	X	X
Disease history	X	X	X	X	X	X
Age of onset	X	X	X	X	X	X
Age at diagnosis	X	X	X	X	X	X
Hereditary (if relevant)	X	X	X	X	X	X
Clinical manifestations	X	X	X	X	X	X
Investigations (type and results)	X	X	X	X	X	X
Disabilities	X	X	X	X	X	X
Antenatal/perinatal information (if relevant)	X	X	X	X	X	X
Birth date	X	X	X	X	X	X
Gender at birth	X	X	X	X	X	X
Malformation detected antenatally	X	X	X	X	X	X
Medically assisted procreation (yes/no)	X	X	X	X	X	X
Gestational age at birth (weeks)	X	X	X	X	X	X
Weight at birth	X	X	X	X	X	X
Height at birth	X	X	X	X	X	X
Head circumference at birth	X	X	X	X	X	X
Head circumference at birth	X	X	X	X	X	X
Family antecedents in relation with the RD	X	X	X	X	X	X
Affected member (propositus) yes/no	X	X	X	X	X	X
Parental relationship with the affected member	X	X	X	X	X	X
Consanguinity: yes/no/unknown	X	X	X	X	X	X
Other relevant familial antecedents (i.e. cancer)	X	X	X	X	X	X
Treatments	X	X	X	X	X	X
Treatment specific for the RD: yes/no	X	X	X	X	X	X
Name and posology of the ongoing treatment(s) (RD specific; others)	X	X	X	X	X	X
Care pathway	X	X	X	X	X	X
Name of the physician in charge of the patient	X	X	X	X	X	X
Rare disease centre following the patient: name of the centre/hospital/ location/phone number (incl. emergency phone number)	X	X	X	X	X	X
Is the RD centre part of an ERN (yes/no)	X	X	X	X	X	X
ERN name	X	X	X	X	X	X
Useful information	X	X	X	X	X	X
url leading to information on the disease, including CPGs, emergency and anesthesia guidelines	X	X	X	X	X	X
Main alert messages	X	X	X	X	X	X



Co-funded by the European Union's Health Programme (2014-2020)



X-eHealth

Exchanging Electronic Health Records in a common framework



Implementation Independent

RD diagnosis
Disease name
ORPHAcode
Diagnosis assertion status
Method for diagnosis confirmation
Causative gene (if relevant)
Gene variant (if relevant)
Disease history
Age of onset
Age at diagnosis
Hereditary (if relevant)
Clinical manifestations
Investigations (type and results)
Disabilities
Antenatal/perinatal information (if relevant)
Birth date
Gender at birth
Malformation detected antenatally
Medically assisted procreation (yes/no)
Gestational age at birth (weeks)
Weight at birth
Height at birth
Head circumference at birth

Family antecedents in relation with the RD
Affected member (propositus) yes/no
Parental relationship with the affected member
Consanguinity: yes/no/unknown
Other relevant familial antecedents (i.e. cancer)
Treatments
Treatment specific for the RD: yes/no
Name and posology of the ongoing treatment(s) (RD specific; others)
Care pathway
Name of the physician in charge of the patient:
Rare disease centre following the patient: name of the centre/hospital/ location/phone number (incl. emergency phone number)
Is the RD centre part of an ERN (yes/no)
ERN name
Useful information
url leading to information on the disease, including CPGs, emergency and anesthesia guidelines
Main alert messages



Co-funded by the European Union's Health Programme (2014-2020)



X-eHealth
Exchanging Electronic Health Records
in a common framework

Implementation Independent

New Items	Rare diseases	Rare cancers	Both
ERN to which the rare disease/rare cancer expert centre information belongs			X
Healthcare alert description in relation to the rare disease	X		
Diagnosis assertion	X		
Heredity	X		
Family history	X		
Pathological cancer diagnosis, or clinical cancer diagnosis		X	
Cancer grading		X	
Relevant molecular profiling		X	
Risk stratification or stage		X	



Implementable Specification

9.3.1.1 Formal Views of Profile Content

Description of Profiles, Differentials, Snapshots and how the different presentations work.

HL7 FHIR

Name	Flags	Card.	Type	Description & Constraints
Condition	I	0..*	Condition	Documentation of a health problem of the patient
Observation	I	1..1	Condition	Concept - reference to a terminology or just text
VerificationStatus		0..1	Condition	Concept - reference to a terminology or just text
Category		0..*	Condition	Concept - reference to a terminology or just text
Severity		0..1	Condition	Concept - reference to a terminology or just text

	Unplanned care (incl. emergency/surgery)	Level 1 consultation	Use case: second opinion, including	GP	Clinical study availability	Summary for patients
RD diagnosis	X	X	X	X	X	X
Disease name	X	X	X	X	X	X
OPPM/Module	X	X	X	X	X	X
Diagnosis assertion status	X	X	X	X	X	X
Method for diagnosis confirmation	X	X	X	X	X	X
Causative gene (if relevant)	X	X	X	X	X	X
Gene variant (if relevant)	X	X	X	X	X	X
Disease history	X	X	X	X	X	X
Age of onset	X	X	X	X	X	X
Age at diagnosis	X	X	X	X	X	X
Heredity (if relevant)	X	X	X	X	X	X
Clinical manifestations	X	X	X	X	X	X
Investigations (type and results)	X	X	X	X	X	X
Disabilities	X	X	X	X	X	X
Antenatal/perinatal information (if relevant)	X	X	X	X	X	X
Birth date	X	X	X	X	X	X
Gender at birth	X	X	X	X	X	X
Malformation detected intrauterine	X	X	X	X	X	X
Medically assisted procreation (yes/no)	X	X	X	X	X	X
Gestational age at birth (weeks)	X	X	X	X	X	X
Weight at birth	X	X	X	X	X	X
Height at birth	X	X	X	X	X	X
Head circumference at birth	X	X	X	X	X	X
Family antecedents in relation with the RD	X	X	X	X	X	X
Affected member (autosomal recessive)	X	X	X	X	X	X
Parental relationship with the affected member	X	X	X	X	X	X
Consanguinity: yes/no/unknown	X	X	X	X	X	X
Other relevant familial antecedents (i.e. cancer)	X	X	X	X	X	X
Treatments	X	X	X	X	X	X
Treatment specific for the RD: yes/no	X	X	X	X	X	X
Name and posology of the ongoing treatment(s) (RD specific: others)	X	X	X	X	X	X
Care pathway	X	X	X	X	X	X
Name of the physician in charge of the patient	X	X	X	X	X	X
Rare disease centre following the patient: name of the centre/hospital/ location/phone number (incl. emergency phone number)	X	X	X	X	X	X
Is the RD centre part of an ERN (yes/no)	X	X	X	X	X	X
ERN name	X	X	X	X	X	X
Useful information (all leading to information on the disease, including CPGs, emergency and anaesthesia guidelines)	X	X	X	X	X	X
Max alert messages	X	X	X	X	X	X



Unplanned healthcare



including emergency/surgery

Planned healthcare



- Second opinion/multidisciplinary consultations
- Internal consultations
- Consultations about availability of ongoing clinical studies/eligibility check
- Concise summary for patients

[illegible]

Co-funded by the European
Union's Health Programme
(2014-2020)

	Unplanned care (incl. emergency/surgery)
RD diagnosis	X
Disease name	X
ORPHAcode	X
Diagnosis assertion status	X
Treatments	X
Treatment specific for the RD: yes/no	X
Name and posology of the ongoing treatment(s) (RD specific; others)	X
Care pathway	X
Name of the physician in charge of the patient:	X
Rare disease centre following the patient: name of the centre/hospital/ location/phone number (incl. emergency phone number)	X
Is the RD centre part of an ERN (yes/no)	X
ERN name	X
Useful information	X
url leading to information on the disease, including CPGs, emergency and anesthesia guidelines	X
Main alert messages	X



Co-funded by the European
Union's Health Programme
(2014-2020)



X-eHealth

Exchanging Electronic Health Records
in a common framework

ERN name

Useful information

un leading to information on the disease, including CPGs, emergency and anesthesia guidelines

Main alert messages

[-] section:AlertSection	S	0..1
[-] title	S	1..1
[-] code	S	1..1
[-] coding		1..1
[-] system		1..1
[-] code		1..1
[-] text	S	1..1
[-] Slices for entry		0..*
[-] entry:flags	S	0..1

2021-11-30 RD-CODE Final Workshop

XeH Alerts Section –

History (1)

Description

Context

Parent nodes of template element with id 2.16.840.1.113883.10.12.201 CD

Classification

CDA Section Level Template

Open/Closed

Open (other than defined elements are allowed)

Used by / Uses

Used by 0 transactions and 1 template, Uses 1 tem

Relationship

Adaptation: template [2.16.840.1.113883.10.20.22.2.58](#)

Adaptation: template [2.16.840.1.113883.10.12.201 CD](#)

Expand All

Collapse All

Search by name



Co-funded by the European
Union's Health Programme
(2014-2020)



X-eHealth
Exchanging Electronic Health Records
in a common framework

Where the on-develop EHRxF specs are ?

- **HL7 CDA Templates**

- <https://art-decor.org/art-decor/decor-project--eehrxf->

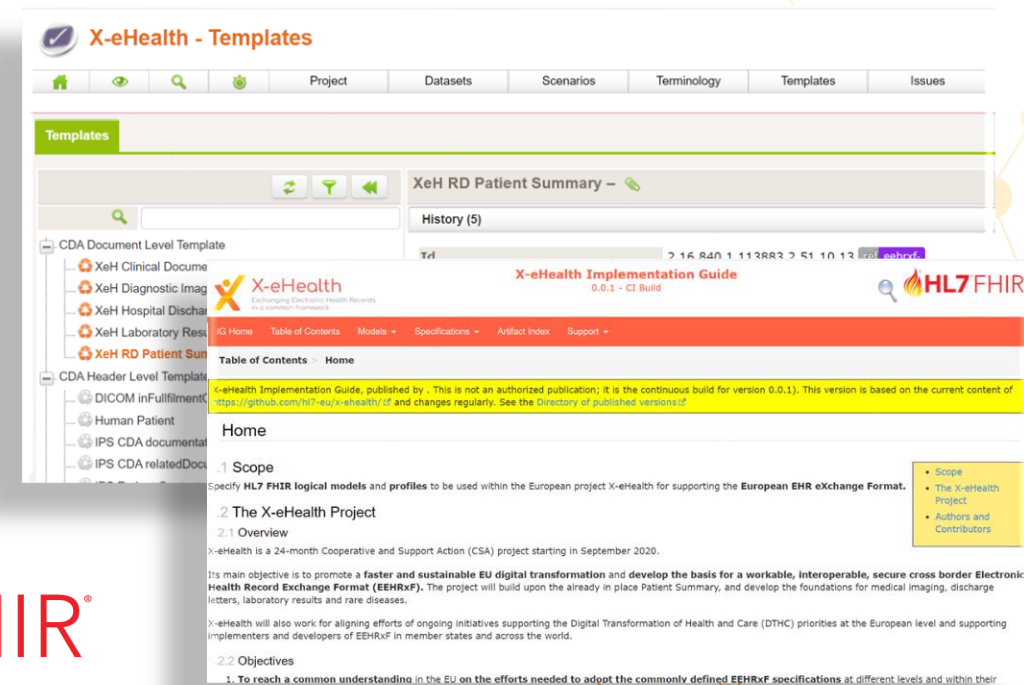


- **HL7 FHIR profiles**

- <https://build.fhir.org/ig/hl7-eu/x-ehealth/>

- **GitHub repos**

- <https://github.com/hl7-eu/x-ehealth>





X-eHealth

Exchanging Electronic Health Records
in a common framework

Interested in diving in ?



HL7[®]CDA[®]



HL7[®]FHIR[®]



THANK YOU & FOLLOW US!

Social Media:



@x_ehealth / #XeHealth

Website:

<https://www.x-ehealth.eu/>

