RD-Connect WP2 UPDATE: Main achievements and challenges

Domenica Taruscio and WP2 Members

Task 2.1 Map existing registry infrastructure and evaluate databases/registries against standards and best practice.

Lead: ISS. Participants: ISCIII, NIH-ORDR, OPHG, MURDOCH, EURORDIS, UPD-7, AMU, UU.

MAPPING:

The integration of new registries into the ID-Card catalogue is made by

inviting registries at appropriate opportunities (e.g. scientific meetings)

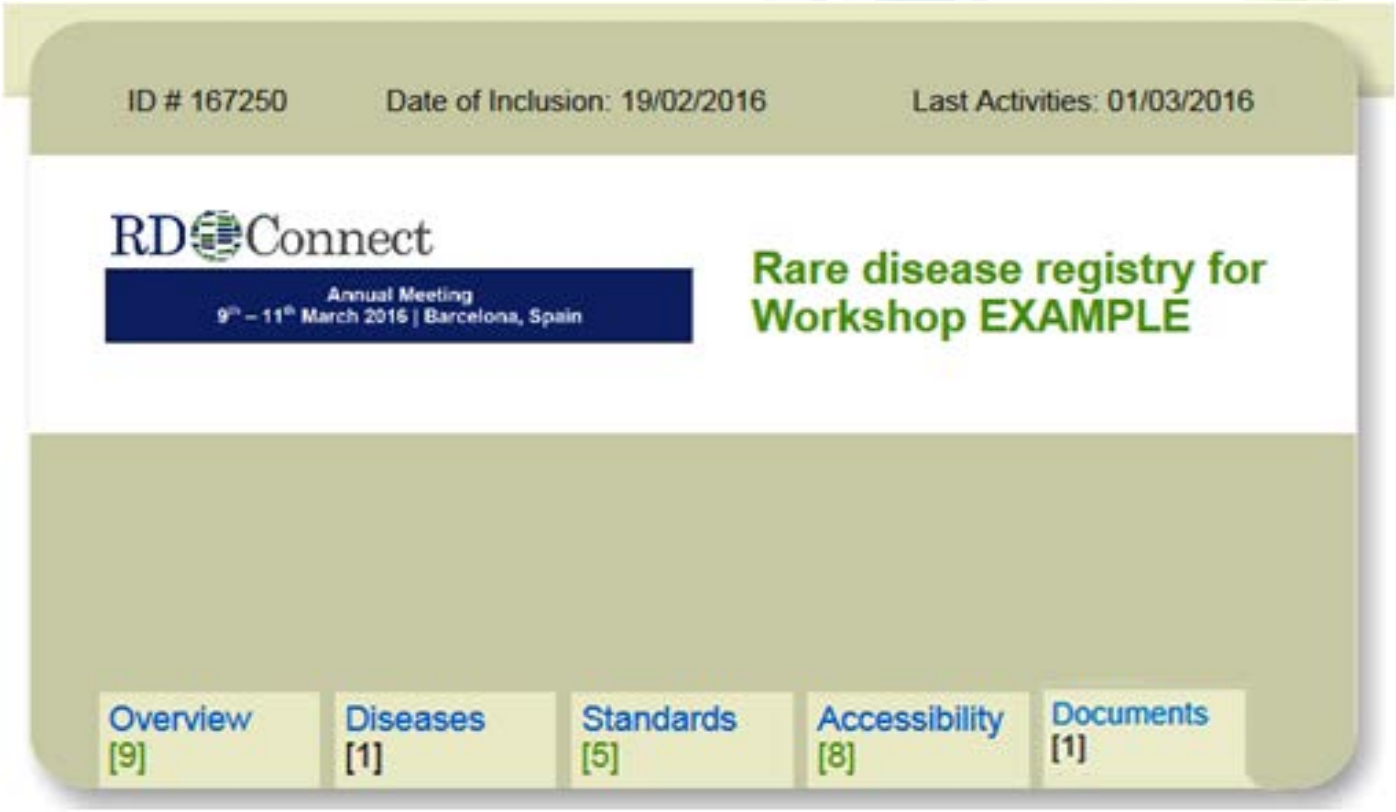
checking the literature through alert systems, Orphanet's report on RD-registries and the main RD registry networks (e.g. TREAT-NMD, GRDR, PatientCrossroads).

Attention will be paid to other initiatives in patient registration in the public health field, in particular the EU registration system led by the Joint Research Centre in Ispra.

EVALUATION:

The completion of all the sections in the ID-card, including the standards and the accessibility sections, is an important tool for **the self-evaluation of registries**

ID-Card


ID # 167250
Date of Inclusion: 19/02/2016
Last Activities: 01/03/2016
RD Connect
Annual Meeting
9th – 11th March 2016 | Barcelona, Spain
Rare disease registry for
Workshop EXAMPLE
Overview
[9]
Diseases
[1]
Standards
[5]
Accessibility
[8]
Documents
[1]

Task 2.2 Establish a Databases and Registries Core Implementation Group.

Lead: AMU, UPD-7.

Participants: EHDN, CUNI, PC, NIH-ORDR, OPHG, ISS, ISCIII, EURenOmics, Neuromics.

Established at month 18; GIC members involved in call conferences and meetings

Task 2.3: Create online searchable catalogue of databases and registries.

Lead: ISS.
Participants: ISCIII, CNAG, NIH-ORDR, PC, OPHG, MURDOCH, UPD-7.

D2.10: Annual update of online catalogue of database/registries, M36

ID-Cards created for 300 RD registries
A detailed user guide made available on the catalogue interface
M24-36 further updates to the software and additional registries invited to join the system.

More than 400 invitations sent through the catalogue system and in some cases tailored messages to the PI.

90 registries replied to the invitation and updated their ID Card and disease matrix within the catalogue.

Simplify and automate the disease matrix

Disease Name	Number of Patients, Donors	Gene	ORPHA Code	ICD 10	OMIM	Synonym(s)	Modified Date

Manual entry	Excel import	Push service	Federation
-Add and edit in the form -Auto completion with orphanet support -Prefilling form	-Import new diseaseMatrix from excel -Import updated excel file into ID-Card -Export the existing diseaseMatrix to excel	-Import data via API -Update also possible for network -Cooperation with PatientCrossroads -Multiupload of entries	-Aggregate data from molgenis sample catalogue -Planning: Aggregate Data from scaleus linked data registry data

Task 2.4: Define registry common data elements and implement standardised coding systems, ontologies, classifications, and standard operating procedures for collecting, storing and retrieving data (M1-M36)

Lead: IIER-ISCIII, CNMR-ISS, ORDR-NIH
Participants: PC, AMU, OPHG, CCG, UU, LUMC

Objectives of Task 2.4

- Definition of registry common data elements.
- Implementation of standardized coding systems, ontologies and classifications.
- Implementation of a global unique patient identifier (GUID).
- Development of standard operating procedures for collecting, storing and retrieving data.

Task 2.4

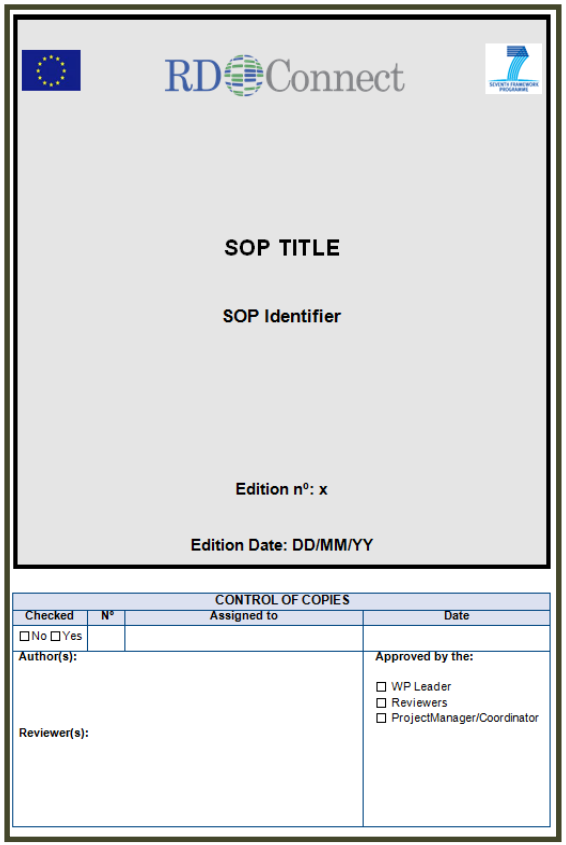
- **D 2.11 SOPs for patient registry data collection, storage and retrieval M36**

SOPs template

List of SOPs prepared for subsequent development in three scenarios:

1. Online ID-Card Catalogue
2. Federated Patient Catalogue (not defined yet)
3. RD-Connect Platform Genomics Interface

Task 2.4: SOPs template



RD-Connect

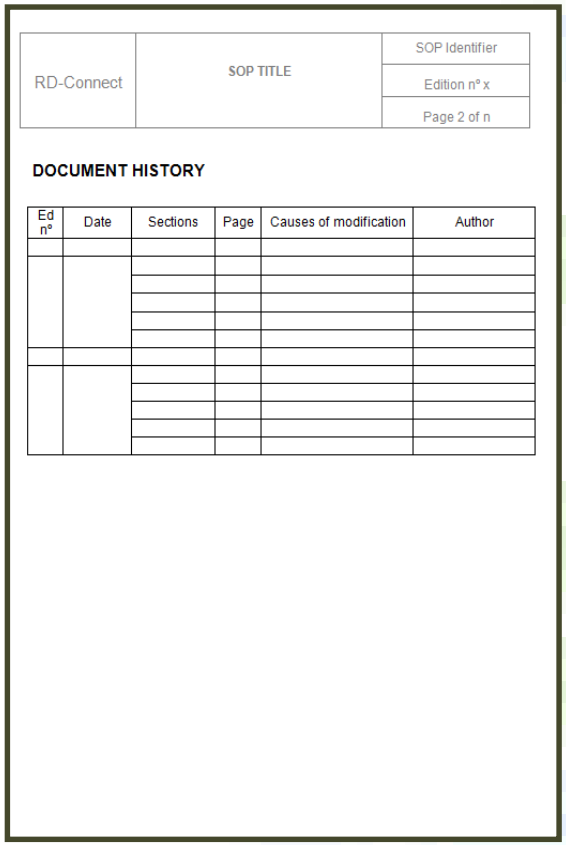
SOP TITLE

SOP Identifier

Edition n°: x

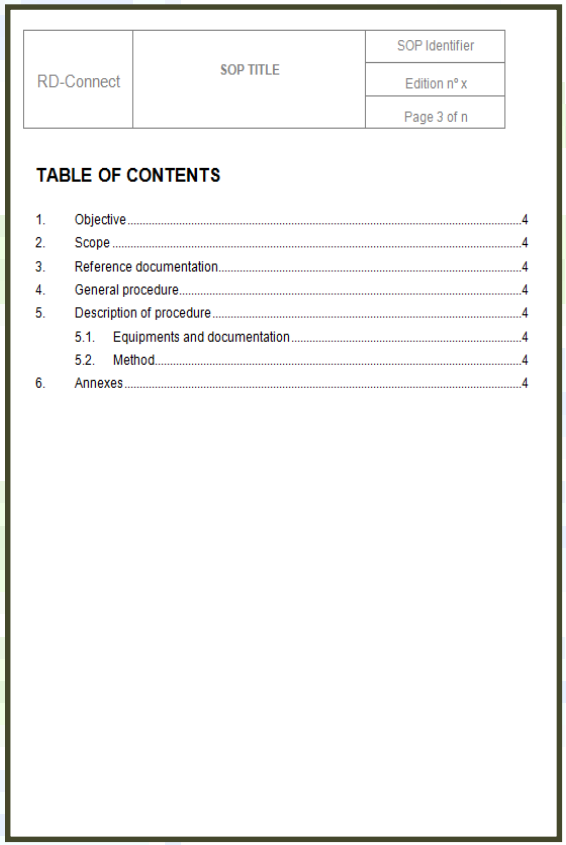
Edition Date: DD/MM/YY

CONTROL OF COPIES			
Checked	N°	Assigned to	Date
☐No ☐Yes			
Author(s):			**Approved by the:** ☐ WP Leader ☐ Reviewers ☐ ProjectManager/Coordinator
Reviewer(s):			



DOCUMENT HISTORY

Ed n°	Date	Sections	Page	Causes of modification	Author



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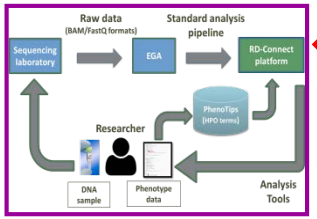
Task 2.4: List of SOPs for collecting, storing & retrieving patient registry data



Scenario	SOPs to be developed	Type of SOP	Currently (February 2016) available SOPs or draft SOPs
Online ID-Card Catalogue	SOPs for **collecting** aggregated information related to each participating patient registry	Developed for external users	User guide to the RD-Connect online catalogue of registries and biobanks (Jan/2016/v1)
	SOPs for **storing** aggregated information related to each participating patient registry	Elaborated for being used by developers	
	SOPs for **retrieving** aggregated information related to each participating patient registry	Developed for external users	User guide to the RD-Connect online catalogue of registries and biobanks (Jan/2016/v1)
Federated Patient Catalogue	SOPs for **collecting** individualized information related to each patient	Developed for external users	
	SOPs for **storing** individualized information related to each patient	Elaborated for being used by developers	
	SOPs for **retrieving** individualized information related to each patient	Developed for external users	
 Not defined yet¡	SOPs for **ethical and legal issues** (ELSI)	Developed for patients or researchers	Informed consent (Deliverable 6.05 "Guidelines for informed consent")
			Material and data transfer agreement (Mascalzoni et al., 2015; doi:10.1038/ejhg.2014.197)
			Adherence agreement for authorized access to data and biospecimens in the RD-Connect platform (Approved at the Executive Management Committee of RD-Connect 2015-06-11)
RD-Connect Platform Genomics Interface	SOPs for **collecting** genomic and phenotypic patient data	Developed for external users	Workflow for submitting omics data to the RD-Connect platform (https://docs.google.com/document/d/1U-5xrincSGjW_3yl4lDxtQ76hR9fNgFgTvupctz4sTo/edit?usp=sharing)
			Instructions given to the data submitter from the RD-Connect platform (e-mailed to the data submitter)
			EGA User Guide (https://www.ebi.ac.uk/ega/about/introduction)
			Phenotips User Guide (https://www.phenotips.org/UserGuide/GettingStarted)
	SOPs for **storing** genomic and phenotypic patient data	Elaborated for being used by developers	
	SOPs for **retrieving** genomic and phenotypic patient data	Developed for external users	
	SOPs for **ethical and legal issues** (ELSI)	Developed for patients or researchers	Informed consent (Deliverable 6.05 "Guidelines for informed consent")
			Material and Data Transfer Agreement (Mascalzoni et al., 2015; doi:10.1038/ejhg.2014.197)
			Adherence agreement for authorized access to data and biospecimens in the RD-Connect platform (Approved at the Executive Management Committee of RD-Connect 2015-06-11)

RD Connect

Task 2.4: Future challenges

CONSENSUS between Developers & Registry Owners

Task 2.5: Develop pilot registry for capturing clinical data on EURenOmics and Neuromics patients without a genetic diagnosis.

Lead: ISS, NIH-ORDR.
Participants: PC, ISCIII, CUNI, OPHG, EURORDIS.

D2.12:
Pilot registry for capturing clinical data on patients without a genetic diagnosis, M36

NeurOmics and EURenOmics are both using the Human Phenotype Ontology for the phenotypic description of undiagnosed cases and each project has developed its own strategy to integrate phenotype and Omics data.

WP2 has made proposals for the translation of standardised phenotype description into ad hoc collection of predefined data fields.

this can be achieved once a significant number of matches are made and a number of phenotype traits may be used to create disease codes (Orphanet and/or OMIM).

Task 2.6: Develop an architecture to allow interoperability with NIH ORDR GRDR to share worldwide de-identified patient registry data.

Lead: PC, ISS.
Participants: ISS; NIH-ORDR; OPHG, MURDOCH, CCG, CNND.

D2.13: Report on interoperability measures, M36

- Meeting in Rome (June 30th 2015 to July 2nd 2015) dedicated to registry and biobank integration and to interoperability measures. Co-organised by the National Center for Rare Diseases (ISS), the University of Leiden (LUMC), and the Elixir project.
- Discussion on the development of a common Application Programming Interface (API)
- Discussion on the list of CDEs proposed by D2.05, definition of mandatory and optional DEs
- Glossary started to clarify terms
- Initial version of a recommended Standard Operating Procedure, SOP, for making RD data interoperable at the "source"

Task 2.6: Develop an architecture to allow interoperability with NIH ORDR GRDR to share worldwide de-identified patient registry data.

Lead: PC, ISS.
Participants: ISS; NIH-ORDR; OPHG, MURDOCH, CCG, CNND.

D2.13: Report on interoperability measures, M36

Interoperability measures (IM) implemented by the RD-Connect platform:

- International standards and nomenclature;
- Common data elements;
- Ontologies for standardizing phenotype description in registries;
- linked data;
- interoperable patient identifiers;
- Common APIs, Dictionary;
- Standard operating procedures (SOPs) for collecting, storing and retrieving patient registry data and for making data interoperable at the source.

Task 2.6: Develop an architecture to allow interoperability with NIH ORDR GRDR to share worldwide de-identified patient registry data.

Lead: PC, ISS.
Participants: ISS; NIH-ORDR; OPHG, MURDOCH, CCG, CNND.

D2.13: Report on interoperability measures, M36

Data sharing solutions tested by Patientcrossroads that can be described to fit into one of three broad system descriptions:

1) A local API server with custom data mapping
2) Local web application server with mapped local common data store
3) Central data store

Task 2.7: Develop training materials and an online “registries toolkit” for new databases and registries, and hold training workshops.

Lead: ISS.
Participants: All WP partners.

D2.14: Training material and registry toolkit, M36

3rd EpiRare International Workshop: Rare Disease and Orphan Drug Registries,
http://www.iss.it/cnmr/?lang=2&id=2290&tipo=41
Rome, 24-25 November 2014

1st RD-Connect Bring Your Own Data meeting
Rome, 26-27 November 2014

Follow up BYOD, survey
February 2015

Task 2.7: Develop training materials and an online “registries toolkit” for new databases and registries, and hold training workshops.

Lead: ISS.
Participants: All WP partners.

D2.14: Training material and registry toolkit, M36

RD-Connect Registry and Biobank Meeting, BYOD2

Rome, 30 June to 2 July 2015 (2 july was reserved for a software engineers meeting)

3rd International Summer School on: “Rare Disease and Orphan Drug Registries”

Rome, , 21-23 September 2015

Rd-Connect workshop: data linkage and ontologies

Rome, 24-25 September 2015

Task 2.7: Develop training materials and an online “registries toolkit” for new databases and registries, and hold training workshops.

Lead: ISS.
Participants: All WP partners.

D2.14: Training material and registry toolkit, M36

Training materials developed from M24 to M36

available in folders at the link : http://www.iss.it/cnmr/?lang=1&id=2585&tipo=3.

List of training materials.

- Registry feasibility and sustainability
- Registry scope and adaptation
- Registry interoperability and standardization
- Quality assurance
- Data elaboration essentials
- Patients' needs and strengths regarding registries
- Experiences with building and managing a registry
- From ontologies concepts to Linked Data
- Usefulness of ontologies for RD
- The Experimental Factor Ontology-EFO
- Using EFO with the Human Phenotype Ontology
- Orphanet Rare Disease Ontology, ORDO
- Wikidata a linked data platform
- Data linkage for rare disease registries: data linkage and ontologies (part 1)
- Data linkage for rare disease registries: data linkage and ontologies (part 2)
- Data linkage tutorial or how to get more out of your registry because data is linkable