RD-CODE Dissemination Plan
Deliverable D2.1

1) General objectives of the project

The objective of this project is to support Member States in improving gathering information on rare diseases by implementation of ORPHA code (rare diseases specific codification system). The implementation process will be guided by the "Standard procedure and guide for the coding with Orphacodes" and the "Specification and implementation manual of the Master file" both developed in the frame of the former RD-ACTION Joint Action. The aim of the RD-CODE project is to promote the use of the Orphanet nomenclature for implementation into routine coding systems. This enables a standardised and consistent level of information to be shared at European level. Starting with countries that have no systematic implementation of the ORPHA codification yet, but that are actively committed already in doing so, this project will provide a sufficient real-world implementation experience to be captured by other countries in the future.

The work and resources developed by the Workpackage 5 of the RD-ACTION supports and harmonise the implementation process and the process of sharing data at European level. With the knowledge that will be gained in the implementation process, additional or modified rules and guidelines for the use of ORPHA codes and for sharing data at European level can be defined if the findings indicate the requirement to do so. The coordination and cooperation between other Member states or projects, as well as with European Reference Networks, will assure the long term success of the project.

2) Expected outcomes

1. Development of comprehensive rules of use and meta-data documentation for ORPHA codes use in clinical health information and other applications based on previous work of the RD-Action and and that are now rigorously tested in health care systems from EC countries of varying sizes, health information systems, and languages.
2. An electronic repository will be created to house this information(www.rd-code.eu), linking to Orphadata, to include: guidance for use of ORPHA codes; teaching and training documents used in health care systems; tools for export of ORPHA codes to federated data exploitation at the EC level (master file for statistical reporting and maybe additional formats if testing shows requirement).
3. Through collaboration of RD-CODE partners and invited collaboration with key stakeholders at workshops, effective implementation will be achieved at implementing countries as well as momentum will be developed in other jurisdictions to implement ORPHA codes in widespread use to accurately measure the impact of rare diseases in the EC.

3) Impact

Giving visibility to RD patients in routine healthcare pathways will allow them to access the better expertise locally and/or through European Reference Networks. In fact, lack of identification of these

patients results in inequalities accessing the best standard of care between European citizens. RD-specific codification at the national level complements the added value of ERNs and helps monitoring the impact of ERNs at the national level.

The RD-CODE project will directly link in to the ERN development as it is foreseen that ERNs will use ORPHA codes in their clinical patient management system (CPMS) , and refinement of ORPHA codes utilization in the RD-CODE project will smooth the way for their use in the ERN CPMS. RD-CODE's objective 6 in WP4 delineates the development of a ORPHA code information service, which will be a direct benefit to ERNs in delivering ORPHA code implementation, and promoting more efficient ORPHA codes implementation in countries participating to ERNs. ERN members are planned as collaborators in this project, with their attendance at workshops.

RD-CODE will promote effective implementation of ORPHA codes in participant countries, including education measures to increase awareness thus contributing to early diagnosis of RD. It will allow better care, prevention of complications, anticipation of functional consequences, and therefore more efficient use of health resources.

Implementation teams of this project are looking at ways of electronic health care record implementation of ORPHA codes, which is directly related to eHealth. Proper identification of patients also allows for improved recruitment for clinical trials and drug development. It also allows to decision making in accessibility and pricing issues related to orphan drugs.

4) Audience

Through the development of guidelines, all rare disease data collectors as well as data users will be guided in how to collect data in a standardized way thereby allowing a more reliable interpretation of the collected data. Patient groups, decision makers as well as politics, including European expert groups (ERNs board of Member States and the Steering Group on Promotion and Prevention, or SGPP) will benefit as they will be able to compare more reliable data and identify patients better once the standardized way of coding is used. Investigators in clinical research will benefit from a reliable identification of RD patients in health information systems and will be able to capture data from the clinical setting.

Target stakeholders have been identified that either currently have parallel work ongoing in cross-border rare disease classification; or who will have future projects in rare disease codification. These stakeholders have been identified as not only requiring the information and expertise generated by this project, but also as groups who, by their participation, can help ensure the generalizability of the results generated. The following groups have been identified as important users of ORPHA code who will also provide feedback during RD-CODE by participating in the face-to-face workshops:

- **EMRaDi -** The Interreg financed EMRaDi - (RDs in the Euregio Meus-Rhine) project, has undertaken comparative mapping of the situation for RD patients by collecting prevalence and cost data regarding 60 selected RDs for the regions using claims (insurance) and hospital data. This allows description of the differences in routine rare disease data collection and processing in DE, NL and BE.

- **ERNs -** The development of 24 thematic European Reference Networks for Rare and Complex Disease management (ERNs) is a major EU development to tackle the issue of cross-border healthcare in the field of rare diseases. The ERN IT Platform (CPMS) was launched at the end of 2017. A cooperation with the European Commission's division in charge of the ERN IT platform as well as with the 24 ERNs will have the aim of addressing the issues of clinical management as well as RD data collection, under the scope of the ongoing and forthcoming ERN RD registries.
- **HL7** - Health Level Seven International (HL7) is a not-for-profit, ANSI-accredited standards developing organization dedicated to providing a comprehensive framework and related standards for the exchange, integration, sharing, and retrieval of electronic health information. HL7 Europe have been involved in most of the European cross-border eHealth services, and promotes the development and adoption of ehealth standards locally. HL7 may have a role to play in EHR and eHealth standards and incorporation of ORPHA code into an International Patient Summary.
- **JRC -**The European Platform on Rare Diseases Registration (ERDRI), being developed by the JRC in collaboration with DG SANTE, aims to cope with the enormous fragmentation of data. It will provide EU-level solutions for data collection and data sharing by creating a pan-European, anonymised federated rare disease statistics. The JRC has included ORPHA code as a required data element in its publication entitled Common Data Elements for Rare Disease registration
- **Patient Engagement -** Although the proposed project is a highly technical one, there is still much to be gained from patient involvement. Implementation of national data collection must be done with the awareness of the citizens involved, and it is important that dissemination information is accessible and understandable to the public. Patient-experts will be sought via the EURORDIS partner in each country organizing a workshop, and will preferably have undergone ePAG training with EURORDIS or have a pertinent background so that they have a minimum knowledge of rare disease and ORPHA code projects.

A target stakeholder has been identified where RD-CODE partners participate in committee work and are positioned to share RD-CODE work and results with the stakeholder.

- **International Classification of Diseases -** Parallel to the further refinement of the "Standard procedure and guide for the coding with Orphacodes" and "Specification and implementation manual of the Master file" the development of the main disease classification ICD in its 11th revision will be ongoing. Through collaboration with WHO through partners in this grant (DIMDI and INSERM) and member states using the classification, the further alignment of these two systems will be pursued and best ways of harmonization of the two coding systems for countries should be addressed. E.g. the identification of best practices on how the "Specification and implementation manual of the Master file" shall be handled in the realm of a possible use of ICD-11 in the future and shall be further specified together with WHO or its EU Collaborating Centres working in this field.

During the course of RD-CODE, the team will try to establish a connection with groups as possible future partners. In many EU projects as well as in other international projects a process of agreeing

on semantic standards is under way. For example the eHealth Digital Service Infrastructure (eHealth DSI) is the initial deployment and operation of services for cross-border health data exchange under the Connecting Europe Facility (CEF). eHDSI sets up and starts deploying the core and generic services, as defined in the CEF, for Patient Summary and ePrescription. In this ORPHA code have not been fully addressed and the Master Value Catalogue does not make use of the ORPHA code yet.

5) Resources

- Responsibility for delivery of dissemination plan:

Orphanet US14: International coordinator.

- Budget

Available at coordinating level

6) Strategy

- Partners/Influencers to engage
  - All the stakeholders involved see section 4
  - ERNs

- Channels
  - www.rd-code.eu
  - Orphanews
  - RD conferences
  - Partners' newsletter
  - Multistakeholder workshops (see section 8 for further details)

7) Dissemination workplan

Action Items	Channel	Timeframe	Lead
Webpage availability*	www.rd-code.eu	March 2019	Inserm US14
Leaflet distribution*	OrphaNews and www.rd-code.eu,	after validation at the KOM	Inserm US14

	congresses		
Deliverables news*	OrphaNews and www.rd-code.eu,	the month following the delivery	Inserm US14
Deliverables release*	www.rd-code.eu	the month following the delivery	Inserm US14
Leaflet distribution through Orphanet Booth	ECRD 2020	May 2020	Inserm US14
Leaflet distribution through Orphanet Booth	ESHG 2020	May/June 2020	Inserm US14
Leaflet distribution through partners Booth	RD conferences		consortium
Multistakeholder workshop 1 report	OrphaNews and www.rd-code.eu,	July 2020	IHIS
Multistakeholder workshop 2 report	OrphaNews and www.rd-code.eu,	June 2021	CIBER
Final leaflet publication*	OrphaNews and www.rd-code.eu, congresses	June 2021	Inserm US14
Article in peer review journal			

8) Additional info: Multi-stakeholder workshops

These workshops are intended to disseminate project results and to foster collaboration between stakeholders with common goals regarding rare disease coding and cross-border care and codification exploitation.
Workshops attendees will include:

- EMRaDi - Two representatives to participate in all workshops to contribute cross-border experience
- ERNs - representatives of the ERNS will be invited to the multistakeholder workshops
- HL7 - Two representatives will be invited to the final workshop
- JRC - Two representatives to be invited to the closing workshop to discuss alignment of diverse national data with centralized and federated European data collection
- Patient Engagement - Two local ePAG patient-experts to all workshops

- Ad hoc invitations of relevant stakeholders according to the items included in the final agenda, if budget allows it.

To further promote and support the implementation of ORPHA code into national routine coding systems a cooperation with other European projects should be maintained. It will be achieved by inviting interested parties, including collaborating partners, to 2 multistakeholder workshops, in order to capture the exchange of experiences, needs and ideas in a structured way.

In this task, the supporting material for WS will be produced as well as WS reports. Workshops will be organised from a logistic point of view. Supporting material will be proposed to the Steering committee for validation prior to the workshops.

Multistakeholder workshop 1 - May 2020
Venue - Prague
Workshop coordinating team: Czech Republic implementation group
Provisional agenda

- Presentations from implementing countries on implementation progress and problems and opportunities encountered
- Roundtable discussion of implementation barriers and solutions, commonalities and differences between implementation projects, with advice from WP5 team and input from collaborating partners
- Presentation of ORPHA code webpage
- Presentation from WP5 team about documentation produced
- Roundtable on use of documents team's developed resources based on the ongoing implementation
- Roundtable discussion for finalizing documentation on codification of suspected/undiagnosed rare diseases, with SOLVE-RD experts
- Dissemination plan update

Participants: all project teams and HSE team and EMRADI
Guests: SOLVE-RD project experts; 2 EPAG members from Prague, 18-24 ERNs representatives (according to availability and resources), additional guest can be invited if budget allows it (i.e. Czech Republic National alliance representative)

Multistakeholder workshop 2 - April 2021
Venue - Valencia
Workshop coordinating team: Spain implementation group
Provisional agenda

- Presentation of final results by implementing countries
- Roundtable discussion of data exploitation from implementation projects
- Roundtable on use of developed resources to support ORPHA code implementation based on the project experience, with particular focus on whether are they equally applicable to hospital coding systems, EHRs and ERNs. Input from implementing and collaborating partners will be seeked.
- Presentation by JRC on their plan for federated national rare disease population data
- Feedback from ORPHA code webpage team - implementing teams and ERN use of ORPHA code Presentation on use of ORPHA code in ERN registries and in CPMS

Participants: all project teams and HSE team and EMRADI

Guests: 18-24 ERNs representatives (according to availability and resources); 2 ePAG members from Spain, Spanish Regional RD registries' representatives. Additional guest can be invited if budget allows it (i.e. Spain National alliance representative)