RD-Action WP5 survey

Deliverable 5.1 – part 1:
Review existing technical implementations for RD coding

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This report is part of the project / joint action ‘677024 / RD-ACTION’ which has received funding from the European Union’s Health Programme (2014-2020).
It has been produced by the co-leaders of the Work Package 5 and is part of the Task 5.1: To define and set the necessary strategy and tools to implement the Orpha codes in the European countries (Task Leader: Remy Choquet [BNDMR, APHP, France] - Contributors: All WP5 contributors). It constitutes the first part of Deliverable 5.1: Review existing technical implementations for RD coding.

The RD-ACTION Joint Action was launched in June 2015 for a 36 months period.
More information on the activities of the RD-ACTION Joint Action can be found at www.rd-action.eu

Context

Some European Union (EU) Member States (MS) have already started the work of introducing the Orpha code in their registries or health information systems, and others have expressed their interest adopting them. Different approaches have already been implemented and start producing results, raising problems and bringing solutions that are of interest for all MS. As stated in the document of work (DOW) of the RD-ACTION joint action, a coding nomenclature alone is not enough to guarantee that the patient data will be comparable from a member state to the other. Along with the quality assessed nomenclature of rare diseases (Orphanet), it is required to provide the coders with the right instructions and clear objectives of coding but also to set the necessary regulatory and/or financial incentive. Also, given the nature of the rare diseases patients and the celerity of new discoveries, it is required to handle uncertainty in diagnoses and frequent updates of the nomenclature.
All MS use morbidity and mortality recording systems. Morbidity recording systems utilize, for the generality of diseases and for the majority of countries, ICD classification. Only in a few countries other systems like SNOMED CT are used. The Orpha code classification is specifically dedicated to rare diseases and is used only in few countries. Taking into account these ongoing experiences, the contexts, the prerequisites, the methods to implement specific monitoring systems of RD patients will be defined.

Objectives

This survey was designed in order to update our knowledge about medical coding and to get a more complete view of the coding systems in all participating countries to facilitate the first year's work for the RD-ACTION Joint Action WP5.
It was also the opportunity to better identify which parts of the work program participating countries would like to participate in, and to which level of implication for the years to come in order to build a European roadmap for RD coding.

Methodology

French APHP and German DIMDI teams worked together to identify key questions that needed to be answered regarding the coding systems in participating countries. It was divided in three main parts: morbidity, mortality and registries. A last section was about the contribution to the WP.
The survey was created on an online platform (Zoho Survey) that allowed several reviewers to access the answers. The link to this online survey was sent before the kick of meeting of the RD-ACTION joint action to all the subscribers to the WP5 session. Countries that did not answer previously were contacted individually to ask them to participate in the survey.
Results were extracted from the online tool and analysed by the co-leading teams of the WP5.

Results

1. Participants

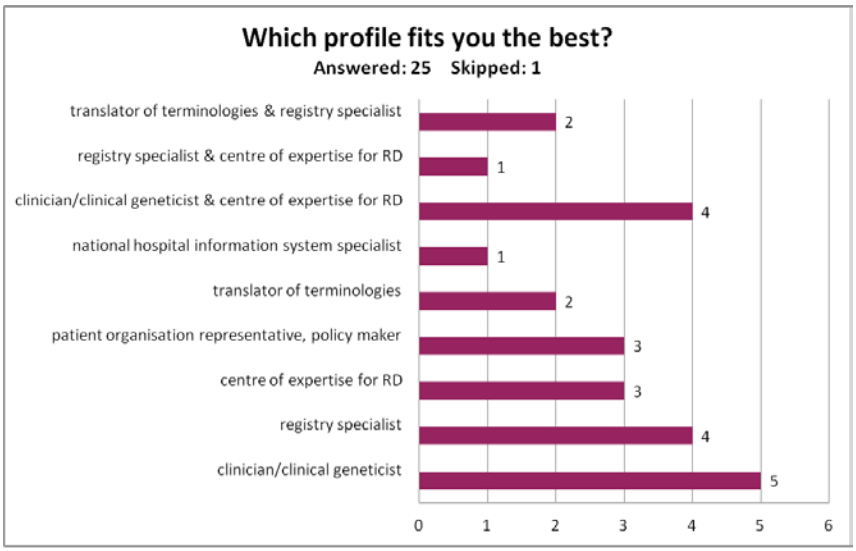
Participants/usable answers

Country	Counted respondents
Australia	*1*
Austria	1
Belgium	1
Cyprus	1
Czech Republic	1
United Kingdom (including England)	3
Estonia	1
Finland	1
France	1
Georgia	*1*
Germany	2
Ireland	1
Italy	1
Latvia	1
Lithuania	2
Netherlands	1
Norway	*2*
Spain	2
Switzerland	*1*
Tunisia	*1*
Total	**26**

The survey has been answered 54 times in total with 26 complete responses and 28 partial responses. Some organizations and countries answered the survey multiple times, so that the data needed cleaning. Responses from the same country were compared and the most complete was kept. When answers differed for a same country, all of them were kept (e.g. United Kingdom).

After data cleansing, 26 datasets could be used for the interpretation. For this interpretation, the answers given by respondent and not the answers per country were counted. An analysis for answers per country will follow.

In italic: non EU countries



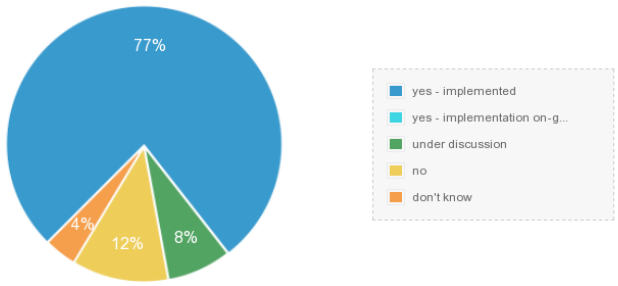
This question allowed multiple answers. Mostly clinicians / clinical geneticist and professionals from centres of expertise participated in the survey. Seven responders fitted in more than one of the named profiles.

The participants of this survey were mainly clinicians/clinical geneticists. Few were experts of the survey subjects.

2. Morbidity

Is there a national regulatory system to record morbidity in your country?

Answered: 26 Skipped: 0

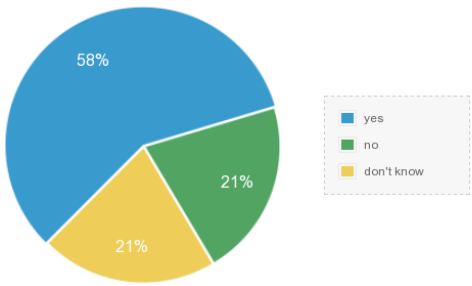


	Response Percent	Response Count
yes - implemented	76.92%	20
yes - implementation on-going	0.0%	0
under discussion	7.69%	2
no	11.54%	3
don't know	3.85%	1

More than 3/4th (20) of the participants declare having a national regulatory system to record morbidity in their countries. In 8% (2) of the countries, the implementation of such a system is under discussion. Only 12% (3) of the countries do not have a system to record morbidity.

Is it connected with the reimbursement system / monetary compensation of hospital activity / patient allocation or insurance?

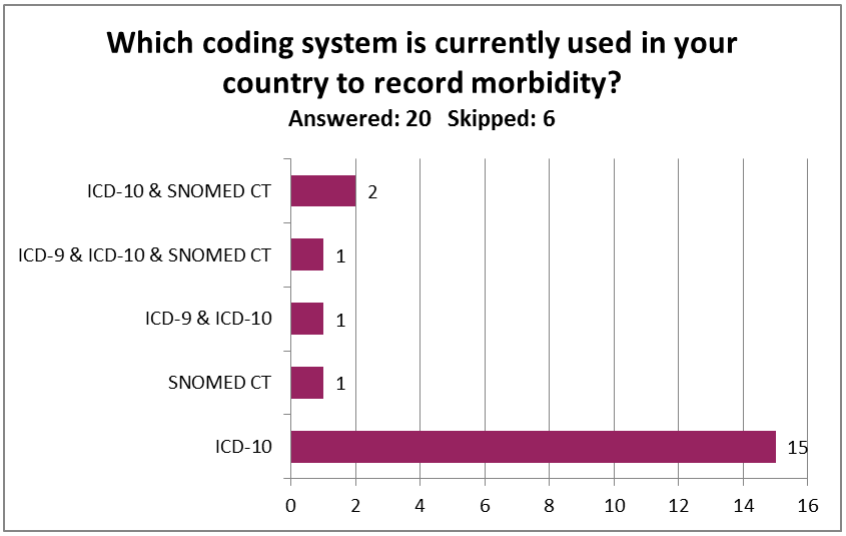
Answered: 19 Skipped: 7



	Response Percent	Response Count
yes	57.89%	11
no	21.05%	4
don't know	21.05%	4

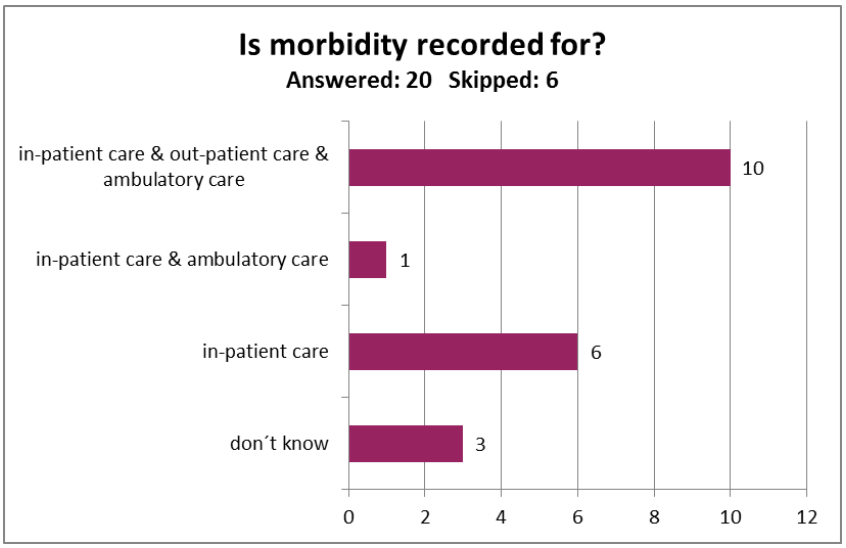
Out of the 3/4th of countries with a regulatory system for recording morbidity, in 58% (11) it is connected to a financial aspect. 21% (4) do not have such a connection between the systems and 21% (4) do not know whether a connection exists or not.

Note:
The connection between the coding process of morbidity and financial aspects may influence the accuracy and quality of the coding. This is a potential risk factor that should be taken into account in the future works and evaluation of this work package.

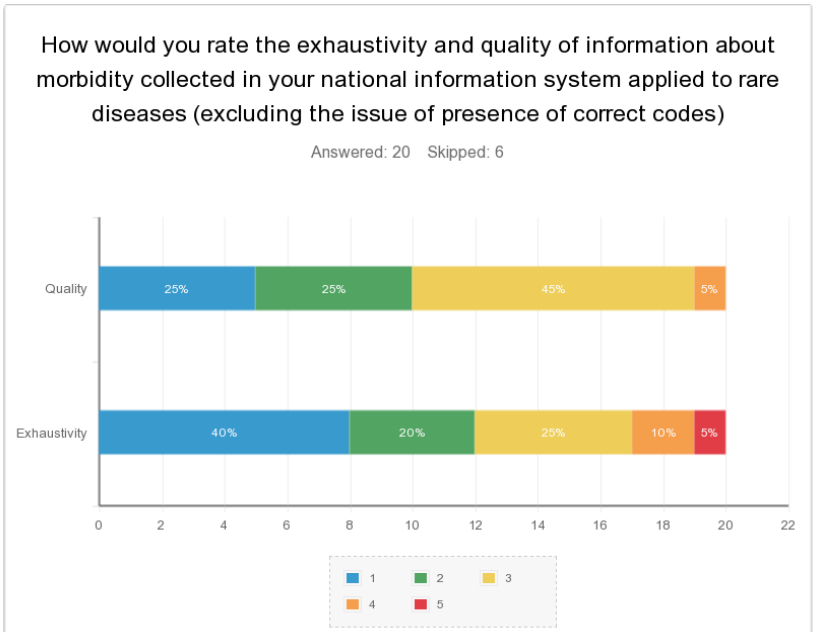


This question allowed multiple answers. The coding system that is mostly used for recording morbidity is the ICD-10 classification. It is used in different versions and adaptions. In addition, ICD9, ICD-10 and SNOMED CT use can be combined.

Note:
Because it is used in most cases, the master file should concentrate on coding with ICD-10.



This question allowed multiple answers. In half of responses, morbidity is recorded in the in-patient care, the ambulatory care and the out-patient care. In 6 out of 19 answers, the records are only for the in-patient care. One country records morbidity both in inpatient care and ambulatory care.

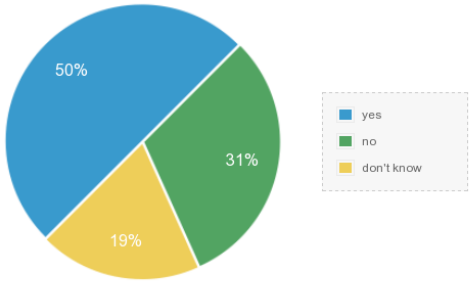


	1 ★	2 ★	3 ★	4 ★	5 ★	Rating Average	Response Count
Exhaustivity	40.0% (8)	20.0% (4)	25.0% (5)	10.0% (2)	5.0% (1)	2.2	20
Quality	25.0% (5)	25.0% (5)	45.0% (9)	5.0% (1)	0.0% (0)	2.3	20

The chart shows the subjective view of respondents. Rating was possible from 1 star (the worst) to 5 stars (the best). Both the exhaustivity and the quality of information about morbidity collected in the national information systems applied to RD were rated as insufficient. The average ratings are of 2.2 and 2.3 respectively. The quality was rated slightly better than the exhaustivity, but one country seems to have a perfect exhaustivity.

Is there a centralised management of the coding system for morbidity in your country?

Answered: 26 Skipped: 0

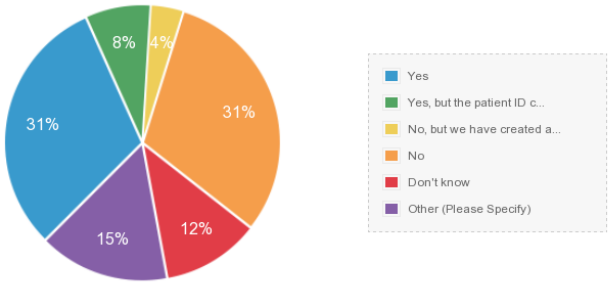


	Response Percent	Response Count
yes	50.0%	13
no	30.77%	8
don't know	19.23%	5

In half of the answers, the coding system for morbidity is managed centrally, whereas 30% (8) is not.

Is your country able to make record linkage based on a patient identifier for rare disease patients ?

Answered: 26 Skipped: 0



	Response Percent	Response Count
Yes	30.77%	8
Yes, but the patient ID cannot be used (limited to administrative / allocation of resources)	7.69%	2
No, but we have created a specific RD identifier	3.85%	1
No	30.77%	8
Don't know	11.54%	3
Other (Please Specify)	15.38%	4

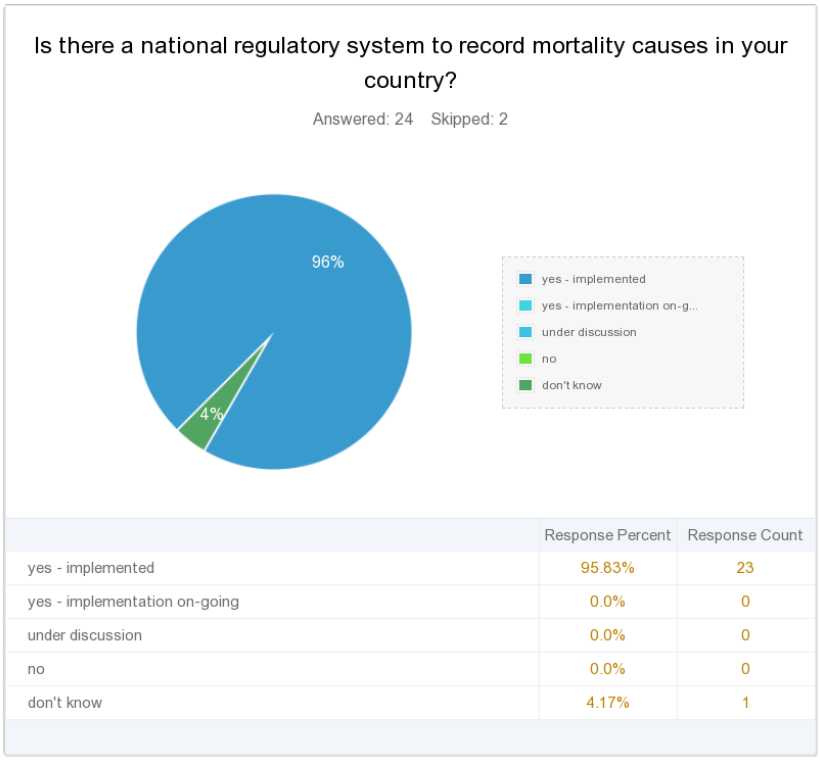
A patient identifier (ID) for RD patients that enables record linkage is implemented in 31% (8) of the countries, whereas the same amount does not have a patient ID at all. One country has created a specific ID for RD patients, even though it does not enable record linkage. In two countries the record linkage is possible but the patient ID cannot be used.

Other answers were:

- that a patient ID for RD patients is at the first step of development
- that the legislation passed for Unique Patient ID to be implemented in the near future
- that there is a patient ID but it cannot be used for identifying RD patients
- that the Patient ID is only usable in accordance with the Law of Personal Data Protection.

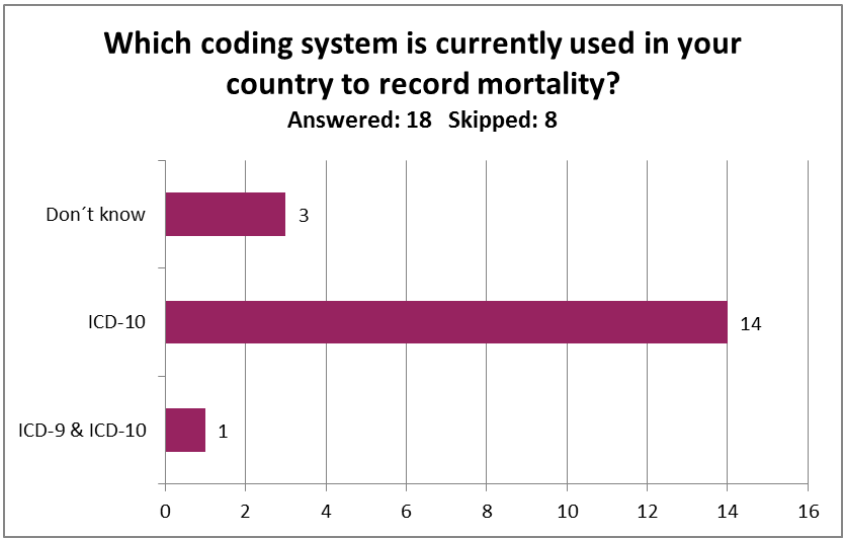
Most of the participating countries have a national regulatory system to record morbidity, which is often linked to the reimbursement system and mainly use ICD-10 codes. It is mostly recorded for in-patient care, but half of the respondents record it for all kinds of care (in-patient, out-patient and ambulatory care). The quality and exhaustivity of information about morbidity collected in the national information system (for RD) are poorly rated. In half of the participating countries there is a centralised management of the coding system for morbidity. But it is not always possible to link records based on a patient identifier for RD patients.

3. Mortality



	Response Percent	Response Count
yes - implemented	95.83%	23
yes - implementation on-going	0.0%	0
under discussion	0.0%	0
no	0.0%	0
don't know	4.17%	1

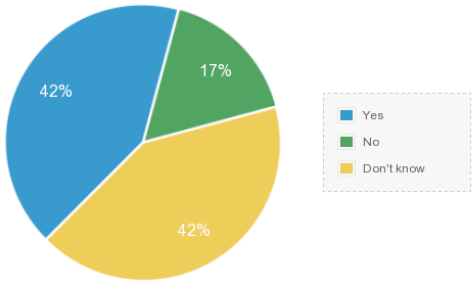
A national regulatory system to record mortality is implemented in 23 out of 24 respondent countries. In general, mortality is better documented/coded than morbidity.



This question allowed multiple answers. ICD-10 is the system that is used the most to record morbidity as well as mortality in the member states. One country reported the use of both ICD-10 and ICD-9.

Do you have a software to assist you in the coding of mortality?

Answered: 24 Skipped: 2

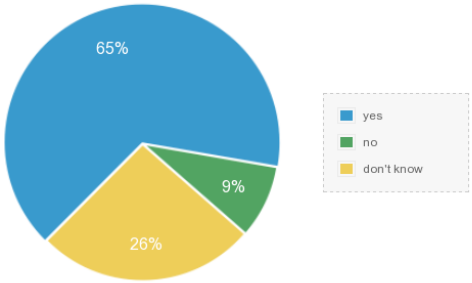


	Response Percent	Response Count
Yes	41.67%	10
No	16.67%	4
Don't know	41.67%	10

42% (10) of the answering countries are using software to assist in mortality coding. In 13% (3) no software is used. The most common software in use is Iris, an ICD-10 browser.

Is there a centralised management of the coding system for mortality in your country?

Answered: 23 Skipped: 2

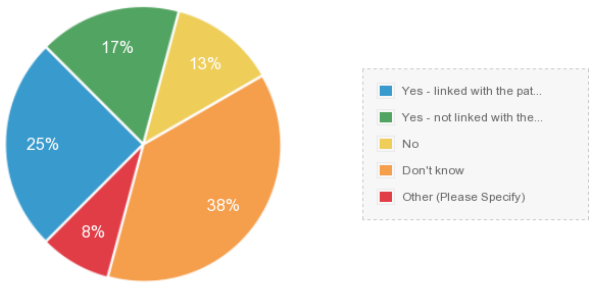


	Response Percent	Response Count
yes	65.22%	15
no	8.7%	2
don't know	26.09%	6

Almost 2/3rd (15) of the respondent countries are having a centralized management of the coding systems for mortality, whereas 9% (1) don´t have a centralized management.

Is there a patient identifier for mortality in your country?

Answered: 24 Skipped: 2



	Response Percent	Response Count
Yes - linked with the patient identifier for morbidity	25.0%	6
Yes - not linked with the patient identifier for morbidity	16.67%	4
No	12.5%	3
Don't know	37.5%	9
Other (Please Specify)	8.33%	2

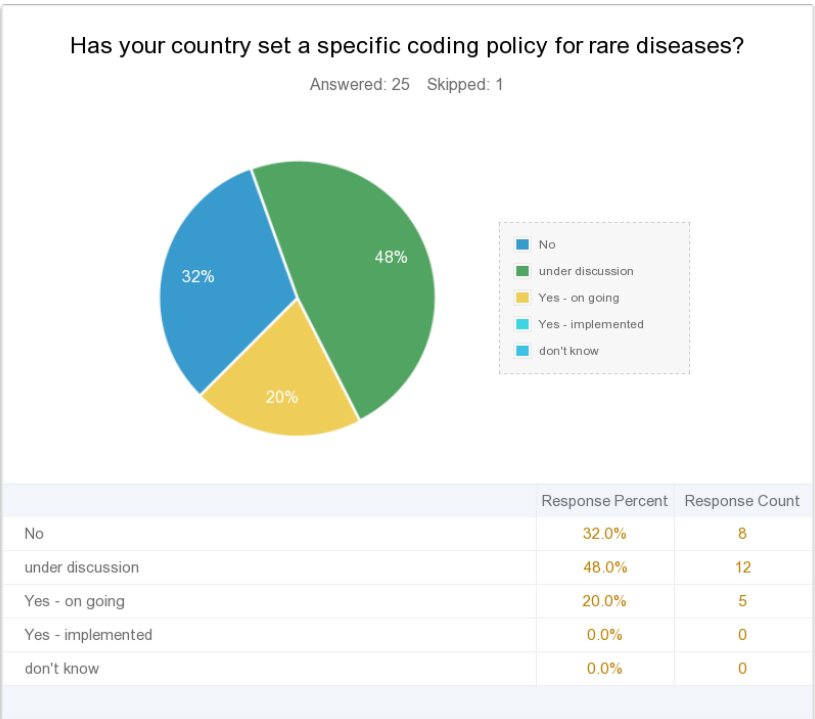
The answers about a patient ID for coding mortality are wide spread. The biggest ratio with 39% (9) shows that participants do not know if there is a patient ID for mortality coding in their countries. 25% (5) said they have an ID that is also linked to the patient ID for mortality coding, when 17% (4) have a patient ID that is not linked with the ID for mortality. 13% (3) of the countries answered that they do not have such an ID.

Other answers (9%; 2) were:

- that they have a unique patient ID that is used for all documents
- that this topic is currently under discussion and for certain areas, the linkage is already possible (e.g. cancer as a pilot)

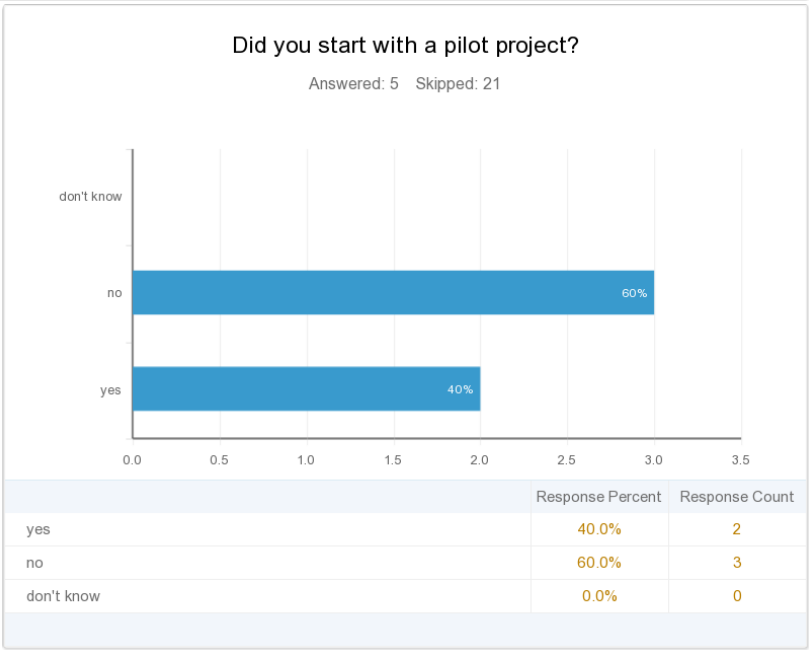
Participating countries usually have a national regulatory system to record mortality, using ICD-10. In 2/3rd of the participating countries there is a centralised management of the coding system for mortality. Less than half of the participants confirmed there is a patient identifier available in their country, which is not always linked to the patient ID for mortality.

4. Policy



	Response Percent	Response Count
No	32.0%	8
under discussion	48.0%	12
Yes - on going	20.0%	5
Yes - implemented	0.0%	0
don't know	0.0%	0

According to the given answers, there is no implemented policy for coding RD in any of the participating countries. 20% (5) of the countries said that they have an on-going process of such an implementation, and about half are discussing it in their countries. A third said they don´t have a specific coding policy.

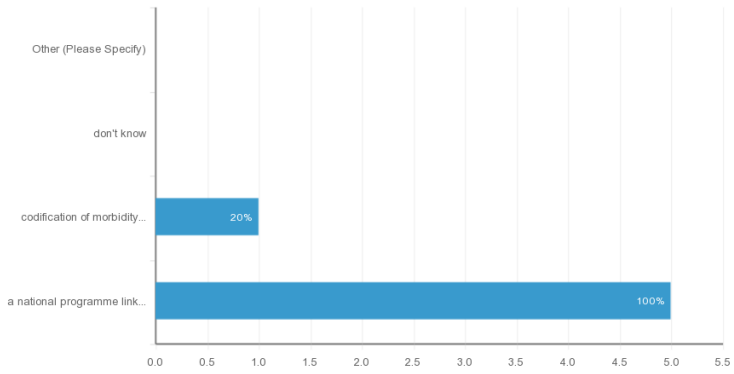


	Response Percent	Response Count
yes	40.0%	2
no	60.0%	3
don't know	0.0%	0

Among the 5 countries for which the implementation of a specific policy for RD coding is on-going, 2 countries have started it with a pilot project.

Was this coding policy created in the framework of:

Answered: 5 Skipped: 0



	Response Percent	Response Count
a national programme linked to rare disease registries or national data repository	100.0%	5
codification of morbidity/mortality in HIS	20.0%	1
don't know	0.0%	0
Other (Please Specify)	0.0%	0

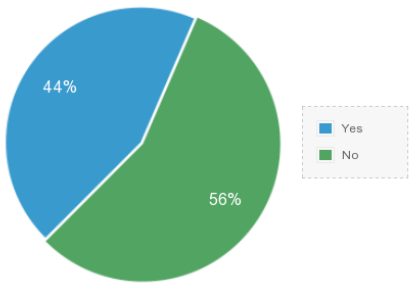
Among the 5 countries for which the implementation of a specific policy for RD coding is on-going, it was always created in the framework of a national programme linked to RD registries or national data repository. One was also created in the framework of codification in HIS.

A specific coding policy for RD has been set up in 5 countries, always in the framework of a national programme linked to RD registries or national data repository and 2 countries started with a pilot project. Half of participants declare a specific coding policy is under discussion.

5. Registries

Do you have a clear knowledge of current RD registries in your country?

Answered: 25 Skipped: 1

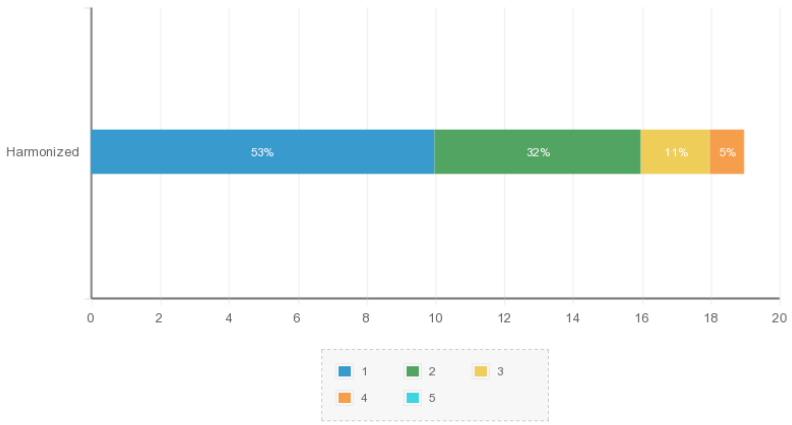


	Response Percent	Response Count
Yes	44.0%	11
No	56.0%	14

More than half of the respondents don't have a clear knowledge of current RD registries in their country.

How would you evaluate the harmonisation of diagnosis coding for rare diseases in existing registries ?

Answered: 19 Skipped: 7



	1 ★	2 ★	3 ★	4 ★	5 ★	Rating Average	Response Count
Harmonized	52.63% (10)	31.58% (6)	10.53% (2)	5.26% (1)	0.0% (0)	1.68	19
Additional Comment							7

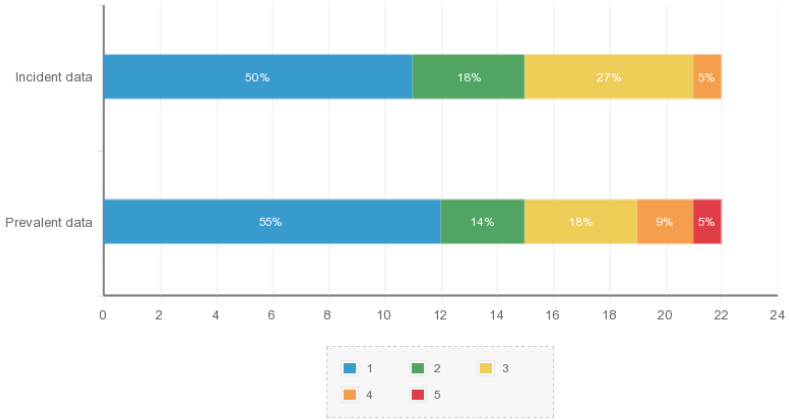
The chart shows the subjective view of respondents.
The harmonization of diagnosis coding RD in existing registries was rated negatively, with an average of 1.68 points. 16 out of 19 respondents gave no more than 2 out of 5 possible points.

Additional comments to this question highlighted the fact that:

- harmonization rate varies a lot across registries,
- few RD have a registry
- based on possibilities/limitations of general coding system there is no intentional/standardized harmonization process so the harmonization rate is not known
- Orpha codes are implemented in Congenital Anomalies registry since September 2014. The Registry is managed centrally
- the National Health Registries on the whole population do not harmonize on RD coding so far, but the existing national registries for RD are on specific diagnoses and diagnostic groups, and they harmonize quite well

Would you judge your country registries a valuable source of data for prevalent or/and incident data for rare diseases ?

Answered: 22 Skipped: 4

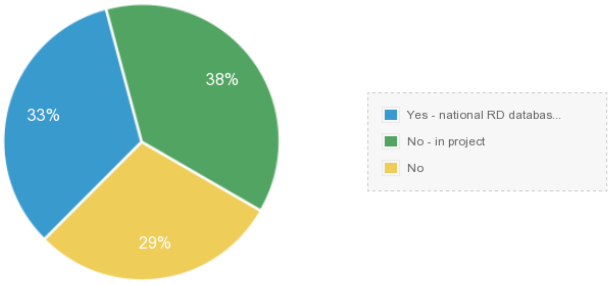


	1 ★	2 ★	3 ★	4 ★	5 ★	Rating Average	Response Count
Prevalent data	54.55% (12)	13.64% (3)	18.18% (4)	9.09% (2)	4.55% (1)	1.95	22
Incident data	50.0% (11)	18.18% (4)	27.27% (6)	4.55% (1)	0.0% (0)	1.86	22

The value of data collected in country registries is as well rated negatively. The prevalent data is rated slightly better than the incident data (respectively 1.95 points (prevalence) and 1.86 points (incidence)) but in both cases 15 respondents gave only 1 or 2 points.

Has your country a national program to integrate registries ?

Answered: 24 Skipped: 2



	Response Percent	Response Count
Yes - national RD database program	33.33%	8
No - in project	37.5%	9
No	29.17%	7

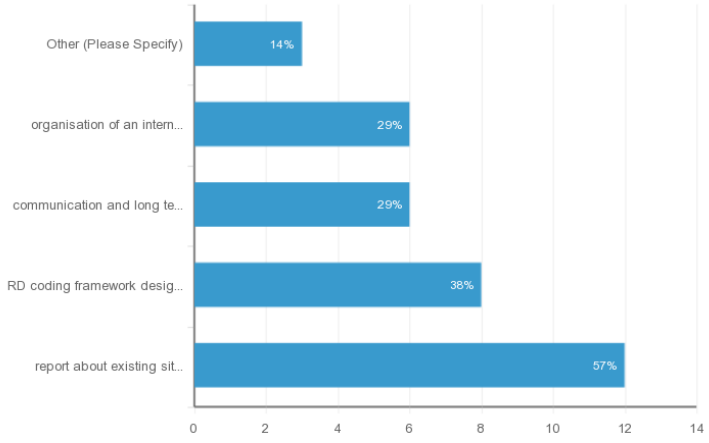
A third (8) of participating countries have a national RD database program, while 29% do not have such a program. Countries with no program but a project to integrate RD databases into registries have the highest ratio with 38% (9).

More than half of the respondents do not have a clear knowledge of current RD registries in their country. The harmonization of diagnosis coding RD in existing registries was poorly rated, so did prevalence and incidence data value. A third of the participating countries have a national program to integrate registries.

6. Participation in WP5

How do you wish to contribute in the first year of the Work package 5 of the RD-ACTION?

Answered: 21 Skipped: 5



	Response Percent	Response Count
report about existing situation and needs assessment from countries and European bodies (Eurordis/Eurostat…) (including a first elaboration of roadmap and a first draft of requirements and objectives)	57.14%	12
RD coding framework design (including product options / guidelines and coding procedures development / definition of the master file)	38.1%	8
communication and long term roadmap for sustainability (including the, JRC, EUROSTAT, RD-CONNECT link)	28.57%	6
organisation of an international workshop on RD coding	28.57%	6
Other (Please Specify)	14.29%	3

This question allowed multiple answers. From the 21 responding countries, 8 countries want to participate in more than one of the named options.
It reflects the willingness of participation of the countries.

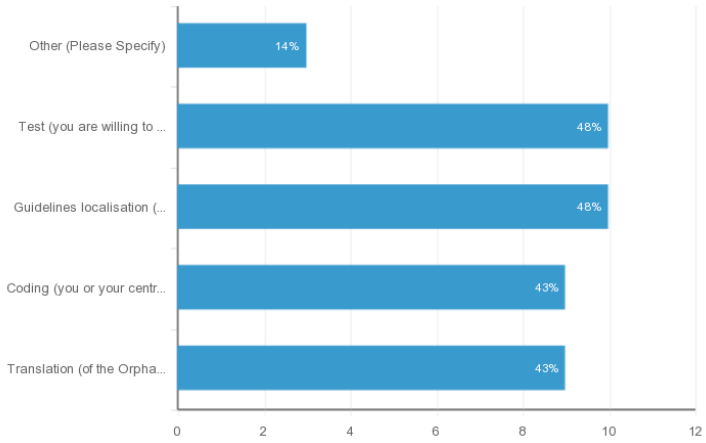
Most of them (12) are willing to report about existing situation and needs assessment from countries and European bodies.

Other answers on how to participate in the first year were:

- to make a link to other policy priorities and
- two countries were not sure how they want to participate yet.

Local implementation (you will be participating to the local implementation of the RD coding system in your country - from year 2)

Answered: 21 Skipped: 5



	Response Percent	Response Count
Translation (of the Orpha terminology)	42.86%	9
Coding (you or your centre is willing to code in real situation)	42.86%	9
Guidelines localisation (you are willing to translate and adapt the coding guidelines)	47.62%	10
Test (you are willing to test the JA solutions)	47.62%	10
Other (Please Specify)	14.29%	3

This question allowed multiple answers. From the 21 responding countries, 10 countries want to participate in more than one of the named options, and 3 of them are willing to participate in all the options. It also reflects the willingness of participation of the countries.
The participations wishes are well equilibrated over the propositions.
Some respondents were not sure of how much they could do and notified it in the “other” category.

7. Other expectations on WP5

The expectations on WP5 were wide spread as well. Given answers were:

- the development of tools for an ontology (knowledge) management platform,
- to have a workshop,
- to learn on what will be the strategies on WP5 at European level,
- a cooperation with relevant developers of a methodology for registration of RD to broaden the expertise and support of the implementation and
- to get knowledge about the involvement of development of integration of RD in ICD-10 and ICD-11

Discussion and conclusion

Coding rare diseases in health information systems or registries is a necessary step to enable wide data analysis across a country, and across Europe. The present situation (not having specific codes or relying on DRG systems) is not satisfactory and is a clear limit to obtain sound statistical data about RD patients. In addition, given the limited number of patients, biases should be avoided.
The introduction of a new coding system may although be not sufficient to succeed. i) not all countries can gather precise morbidity data centrally that could be used to build central data studies (aka. National prevalence of RDs), for those who have a central DRG based payment system for

data collection using ICD-10, it is not always linked to out-patient clinics which represent a large proportion of the RD care activity, ii) few countries have a central registry program, iii) very few countries have a regulatory or financial incentive to facilitate a widespread homogeneous data capture by healthcare professionals, and as a matter of fact, few countries have an expert human network, iv) a national unique patient ID does not even exist in some countries for 'common' diseases with low re-identification risks.

On the other hand, when a codification system for morbidity exists, ICD-10 is the main standard. Many of the respondents are interested in finding solutions for better identifying RD patients in their countries but may not be the necessary national contact point that would be in the better position to implement such a measure. This survey also shows that the current EU situation is very heterogeneous and few countries have started to work on it. Few countries have started a pilot implementation at local level, and very few at national level. As seen in the US by the current implementation of ICD-10 (update from ICD-9), the introduction of a new nomenclature into all hospital information systems is expensive, is very long, requires a national regulation and local expertise to use the codification system. Implementation is then possible (depending on the implementation method) when some pre-requisites are met (technical, legal, expert's network, funding, etc.).

The WP5 task force will now i) refine the list of MS contact points, ii) produce a s MS situation matrix to better grasp opportunities and strategies in implementing Orpha codes and iii) produce a roadmap for implementation across MS.