Work Package 5 – Survey #2

Implementation of rare disease patient coding across member states

This survey is an extension of deliverable 5.2 of the RD-ACTION, it is part of the project / joint action '677024 / RD-ACTION' which has received funding from the European Union's Health Program (2014-2020). It has been produced by the co-leaders of the Work Package 5. This survey aims at analyzing the level of implementation of rare disease patient coding across member states. The survey was conducted through an online questionnaire sent to the RD ACTION contact list. This document presents the results collected between April, 26 and August, 18 2017.

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

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Total responses taken into account: 21*

Austria, Belgium, Bulgaria, Cyprus, Czech Republic, England, Estonia, France, Germany, Hungary, Ireland, Italy, Latvia, Lithuania, Norway, Portugal, Spain, Sweden and the Netherlands

** We collected 23 responses and two member states provided two responses (only the first of the two responses was considered for the survey analysis in the following document). The MS information was not available for two responses.*

Highlights

- There are **very few implemented strategies to produce statistics on rare diseases** at national level. Nevertheless, the production of statistics on rare diseases is among the strategies included in many national plans/strategies, most of them being "in design" at the moment.

- **RD patients are mostly coded in health information systems in the framework of the use of general coding system for morbidity/mortality, namely ICD-10**.

- **Multiple terminologies** are used for coding diseases and for phenotypes.

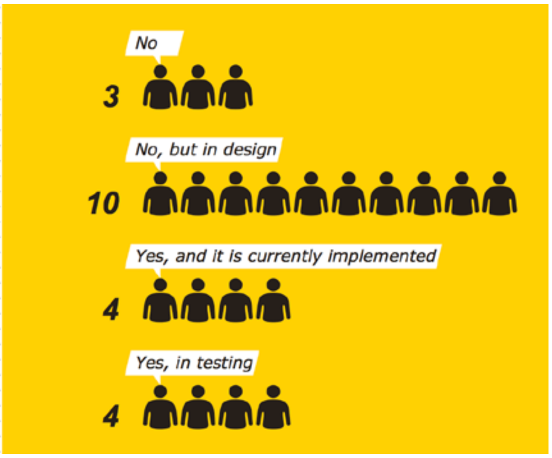
- **Orphacodes** emerge as the main coding system dedicated to RD in both inpatient and outpatient settings.

- **In the majority of MS answering the survey (n=15) a unique and stable patient identifier is in use.** This is an important prerequisite for evaluating RD impact at population level and on the health care systems if orphacodes are adopted, as it allows the tracing of RD patients across different care settings and thus health information systems

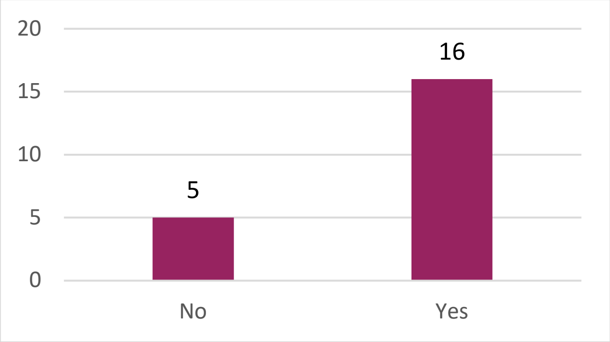
- The RD coding evolving and articulated situation at EU level is a strong base for the adoption and implementation of D5.2 guidelines: **Each of the 6 guidelines are up to at least 60% "planned" or "ongoing" across MS, and 12% were deemed "done" at the time of this survey.**

Rare Disease coding policy in your country

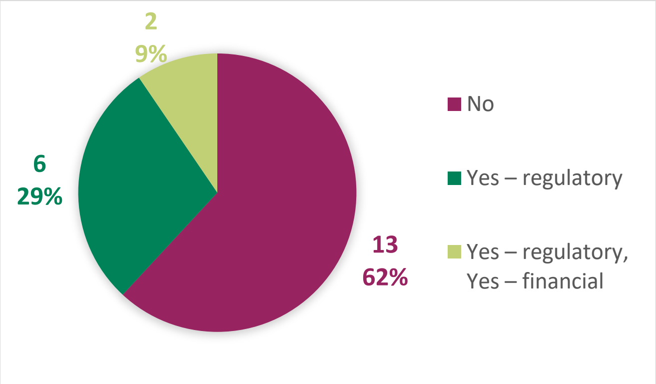
1. **Does a national strategy to produce statistics on rare diseases exist in your country?** (only one possible answer) (21 responses)



2. **Is it included in a national plan for rare diseases?** (only one possible answer) (21 responses)

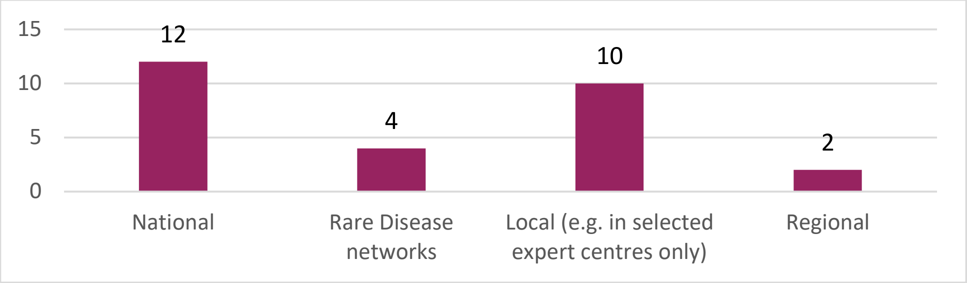


3. **Did you set incentives for using Orphacodes to improve statistics on rare diseases in your country?** (multiple answers possible) (21 responses)



Rare Disease coding implementation in your country

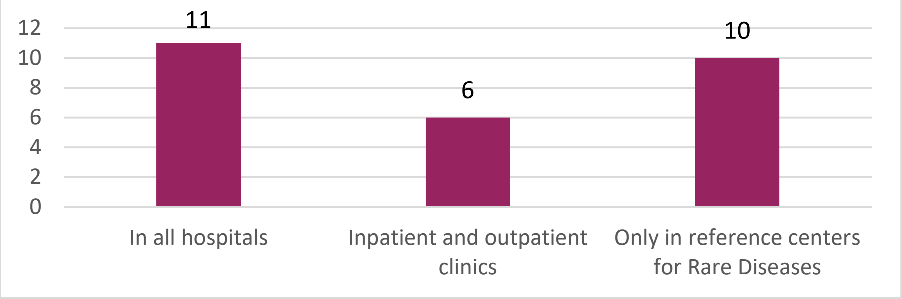
4. Level of implementation (multiple answers were possible) (20 responses + 1 not answered)



Detailed responses (multiple answers were possible)

Local (e.g. in selected expert centres only)	**5**
Local (e.g. in selected expert centres only), National	**2**
Local (e.g. in selected expert centres only), Regional, Rare Disease networks	**1**
National	**7**
Rare Disease networks, National	**2**
Regional, National	**1**
Local (e.g. in selected expert centres only), Regional	**1**
Local (e.g. in selected expert centres only), Rare Disease networks	**1**

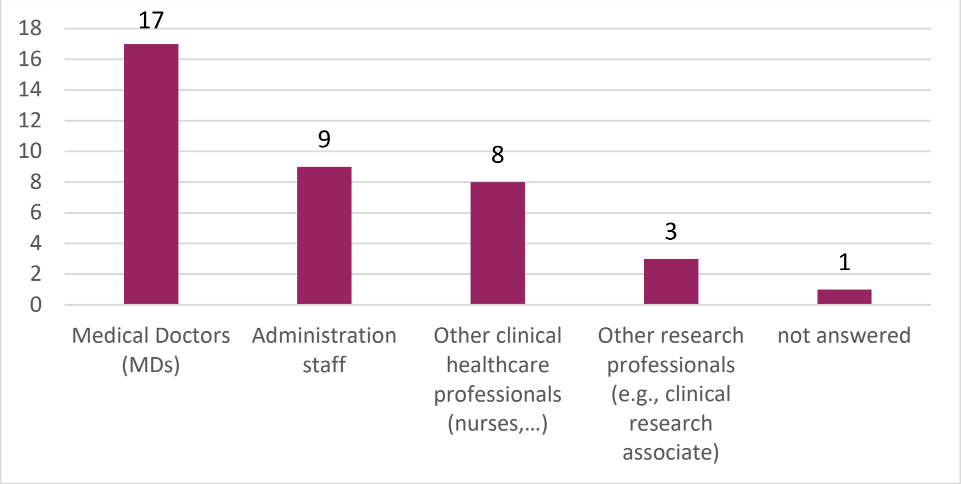
5. Where is the codification performed? (multiple answers were possible) (21 responses)



Detailed responses (multiple answers were possible)

all hospitals and clinical treatment centers codify diagnoses, but they do so for reimbursement purposes. There is no specific codification especially for rare diseases	**1**
In genetic centres	**1**
In all hospitals	**2**
In all hospitals, Inpatient and outpatient clinics	**4**
Only in reference centers for Rare Diseases	**7**
Only in reference centers for Rare Diseases, Childrens University Hospital, Medical Genetics and Prenatal Diagnoses clinic (coding for all cases Congenital anomalies and inherited diseases)	**1**
Only in reference centers for Rare Diseases, In all hospitals	**1**
In all hospitals, Inpatient and outpatient clinics, Specific registries	**1**
National Rare Diseases Office, limited, Information Line only	**1**
In all hospitals, but can be poor quality	**1**

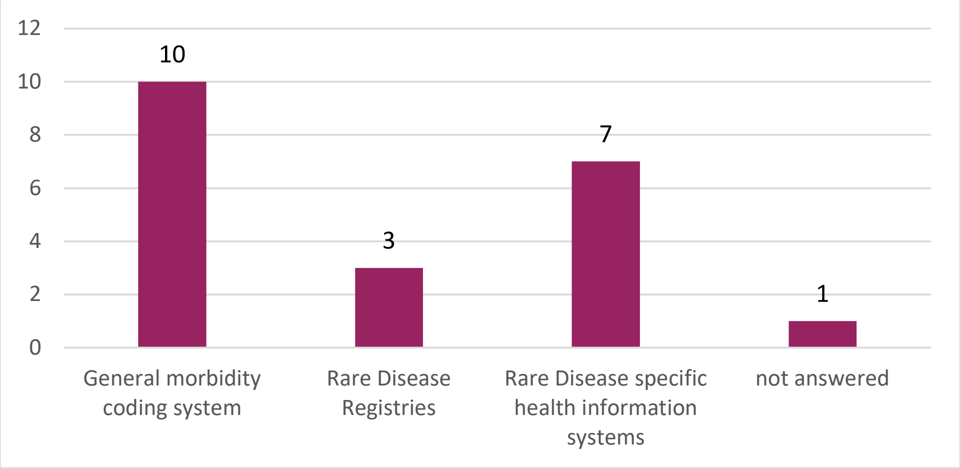
6. **Who is in charge of the codification?** (multiple answers were possible) (20 responses + 1 not answered)



Detailed responses (multiple answers were possible)

Administration staff	**3**
Medical Doctors (MDs)	**5**
Medical Doctors (MDs), Administration staff	**3**
Medical Doctors (MDs), Other clinical healthcare professionals (nurses, …)	**4**
Medical Doctors (MDs), Other clinical healthcare professionals (nurses, …), Administration staff	**1**
Medical Doctors (MDs), Other research professionals (e.g., clinical research associate), Administration staff	**1**
Medical Doctors (MDs), Other research professionals (e.g., clinical research associate)	**2**

7. **Which data stream is used to collect data from the codification system?** (only one possible answer) (20 responses + 1 not answered)



Rare Disease coding resources

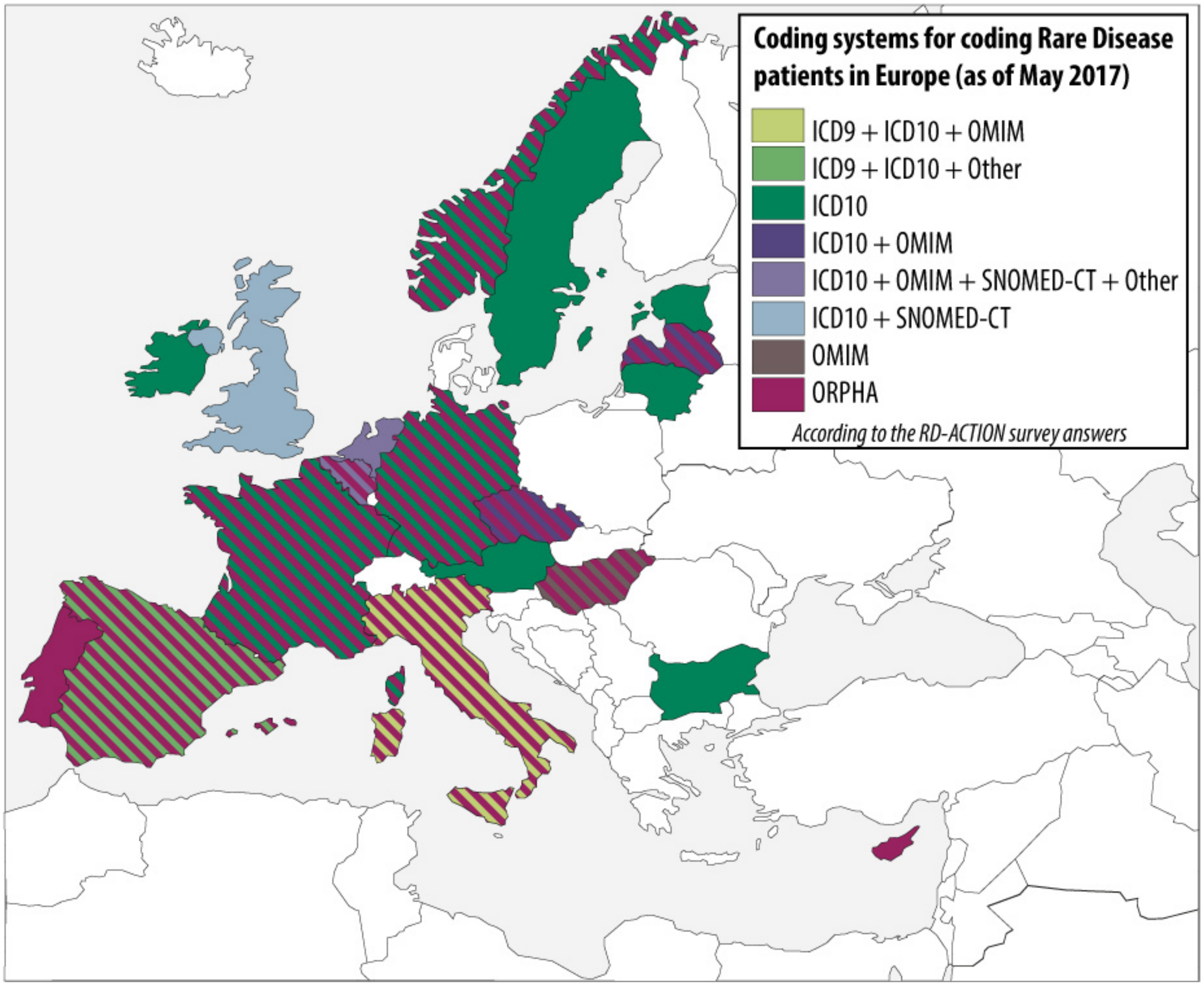
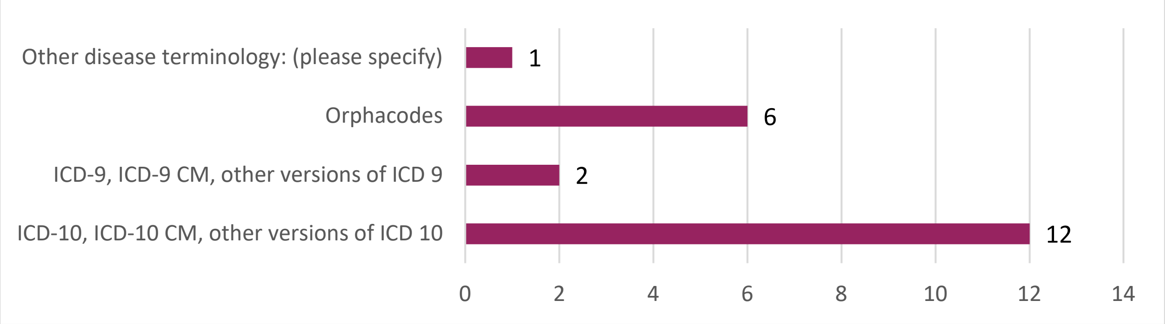


Figure 1. Overview of Rare Disease coding terminologies in Europe

In Figure 1, we present a map of coding systems that are used in Europe for Rare Disease coding. This map is derived from our survey and detailed results (for rare diseases, genes and phenotypes coding systems) are presented in the following sections (8-11).

8. **What is your main coding system for coding RD patients**? (only one possible answer)

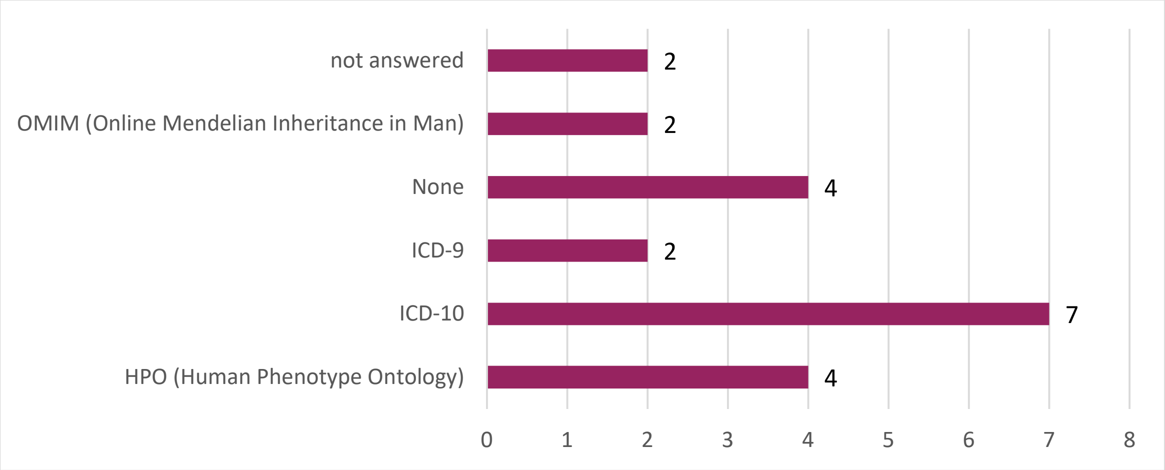
8.1 For diseases (21 responses)



Comments:

SNOMED-CT was not reported as main coding system for diseases. However, the Netherlands use SNOMED-CT which they linked to a diagnosis thesaurus. SNOMED CT is planned to be used in England by 2020.

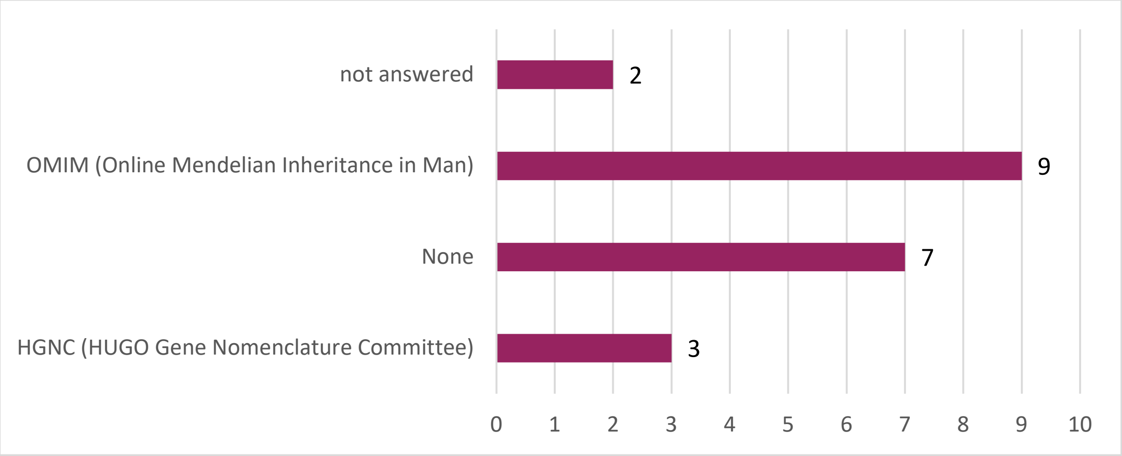
8.2 For phenotypes (19 responses + 2 not answered)



Comments:

no country reported SNOMED-CT or LDM (London Medical Database) as main coding system for phenotypes. Other phenotypes terminologies were reported: HPO is used in Italy, in addition to ICD9; SNOMED CT is planned to be used in addition (or as an alternative) to HPO in England.

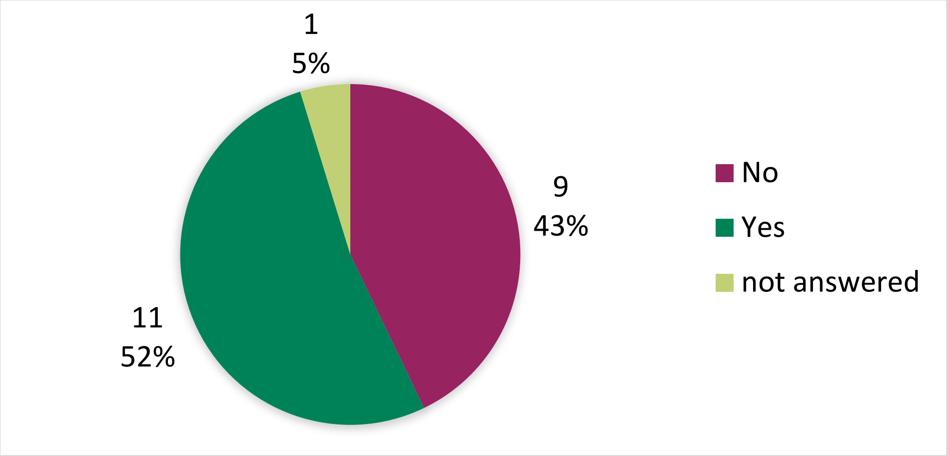
8.3 For genes (19 responses + 2 not answered)



Comments:

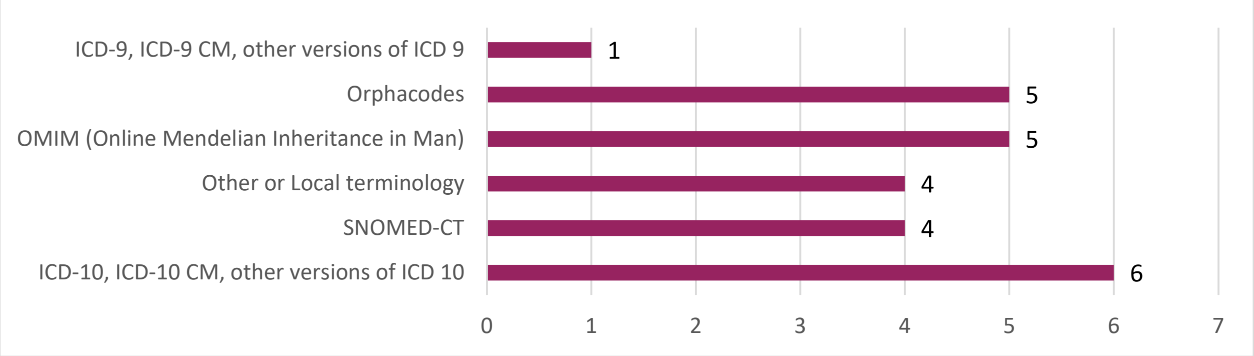
No “other” or “local” terminologies were reported as main coding system for genes.

9. **Do you use additional coding systems?** (20 responses + 2 not answered)



10. If yes (you use additional coding systems), please precise which one(s)?

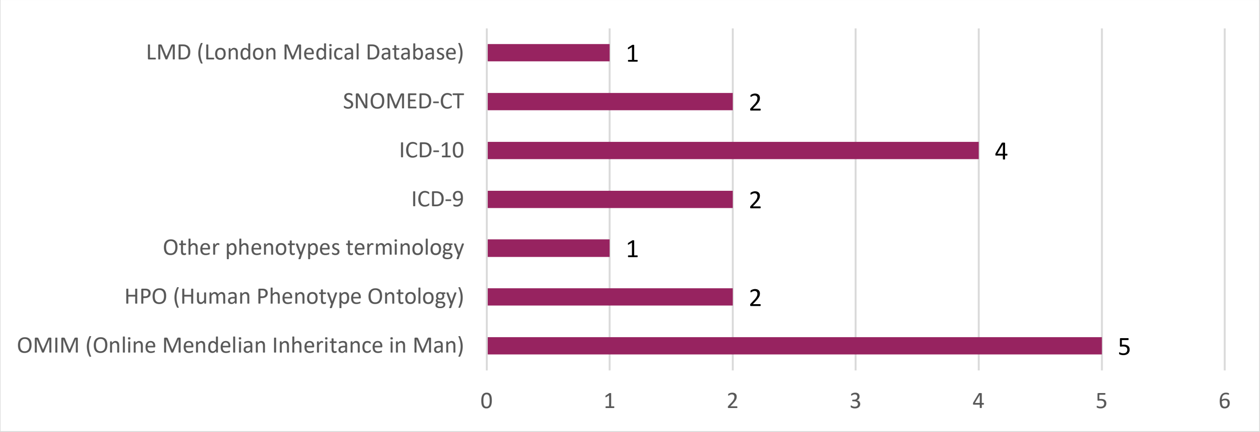
10.1 For diseases: (multiple answers possible) (12 responses)



Comments:

Other and local terminologies reported are: Alpha-ID (Germany), CINEAS (the Netherlands), CRAMP (Belgium) and ERA-EDTA Codes, ICD-10 BPA (Spain).

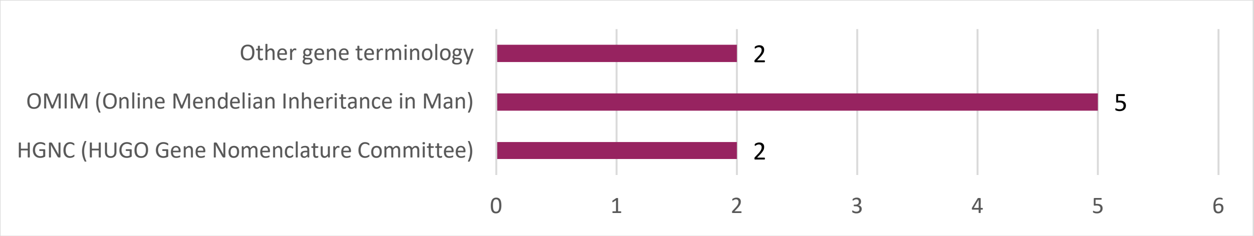
10.2 For phenotypes: (multiple answers possible) (9 responses)



Comments:

One other terminology was reported: CINEAS (the Netherlands).

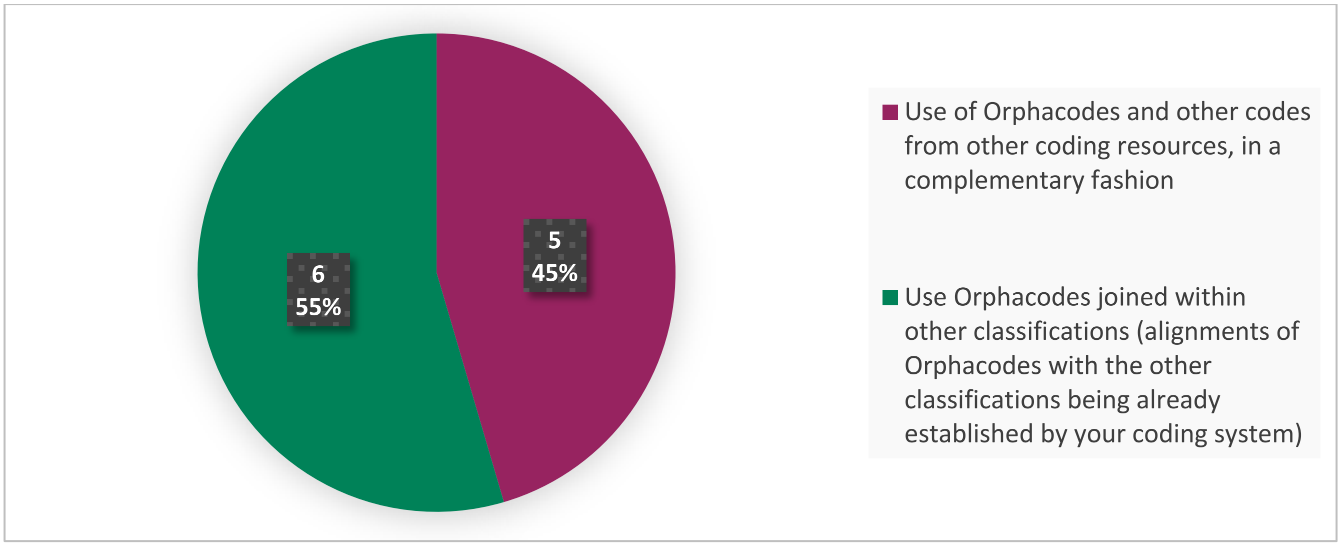
10.3 For genes: (multiple answers possible) (9 responses)



Comments:

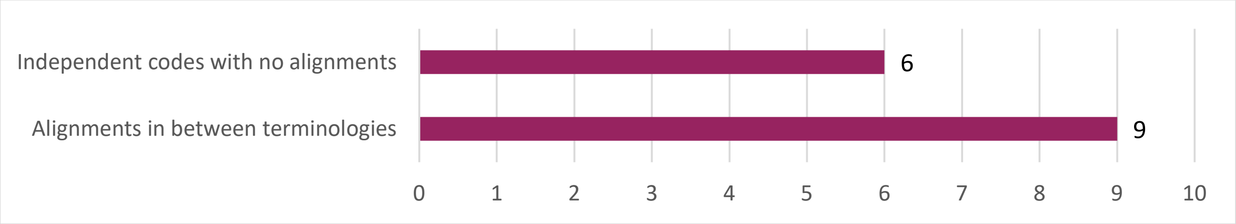
Other and local terminologies reported are: CINEAS (the Netherlands) and LOINC (Belgium).

11. **If you use Orphacodes, then what describes your use of Orphacodes for RD codification the best?** (11 responses)

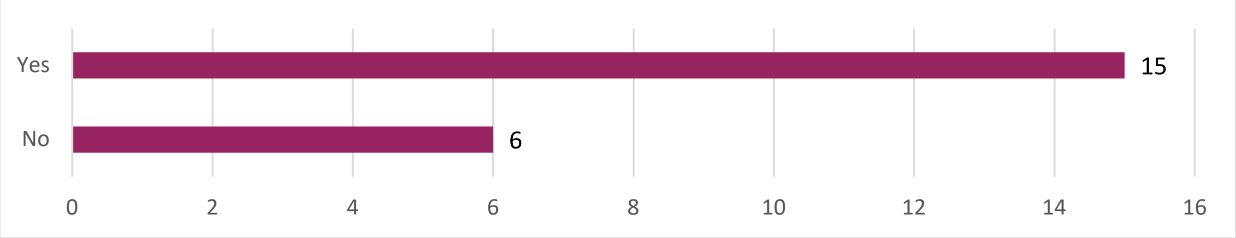


About the coding system in your country

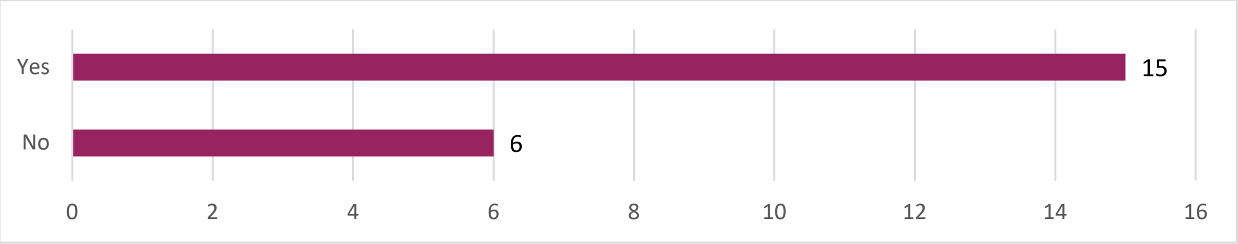
12. **If you use multiple coding system, then what is the best description of the use of multiple coding systems for RD in your country?** (15 responses)



13. **Does your countries national health care system implement a unique and stable patient identifier in your country?** (21 responses)



14. Is the source of the collected health care data trackable i.e. from an expert center? (21 responses)



15. **Is a specific set of guidelines for the coding process implemented in your country?** (19 responses, 2 not answered)

RD-Action guidelines

16. In this incentive, is your country able to follow the RD-Action guidelines?

(20 responses + 1 not answered)

Reminder:

- Guideline #1- In your country, the strategy to produce data or statistics for RD maximize exhaustiveness as well as possible re-use of existing data collections
- Guideline #2- The coding and data collection system of your country allows reaching the international recommended list of Orphacodes
- Guideline #3- The assertion of the diagnosis is captured for all RD cases in your country
- Guideline #4- Your countries health statistics for RD at population level are produced by instruments additional to RD registries (disease, population or patient based)
- Guideline #5- In your country, you update your coding resource according to the internationally agreed cycle
- Guideline #6- If Orphacodes are used together with another national coding system for coding, the two systems are linked in your country in a standardized way