

# ERN survey Analysis

## June 2024

## Content summary

<b>Objective</b> .....	2
<b>Methodology</b> .....	2
<b>Results</b> .....	3
<b>Annex 1. 6 Mandatory Questions</b> .....	11
<b>Annex 2 Answers received by country</b> .....	12

---

### Objective

A Survey to evaluate the current situation of ORPHAcodes usage by health care providers (HCPs) linked to ERNs in countries participating in OD4RD2 has been carried out, in order to be able to identify ORPHAcoding practices and barriers in implementation to be able to fine tune both National Actions but also transversal actions at the Coordinating team level within OD4RD & JARDIN.

### Methodology

A survey was sent to email contacts from each of the National ERN centres listed the Orphanet Data Base of expert resources (each Hub could add additional contacts, representing 1523 units registered in Orphanet for the 16 countries having launched the survey in their country). The survey consisted in a Minimum data set of questions (each Hub could add questions if needed) [See Annex 1 for list of mandatory questions].

IN.B: the Netherlands (NL) conducted in depth interviews with the policy advisor for expert centers and ERNs and the coordinator for ERN registries in the Radboudumc (RUMC) instead of sending a survey. Results presented here for the NL, therefore specifically apply to Radboudumc units.

Countries where the survey was launched: Austria, Belgium, Bulgaria, Germany, Spain, Ireland, Italy, Finland, Estonia, Lithuania, Latvia, the Netherlands (only Radboudumc) , Norway, Poland, Portugal and Sweden. Each Hub has provided a report which describes in detail the methodology used and the preliminary results are presented

Data analysis was performed by compiling 16 survey reports even though some information was missing in some reports.

Analysis by ERN was performed by 11 National Hubs: by pooling data: Austria, Belgium, Germany, Italy, Finland, the Netherlands, Norway, Portugal, Poland, Spain, Sweden

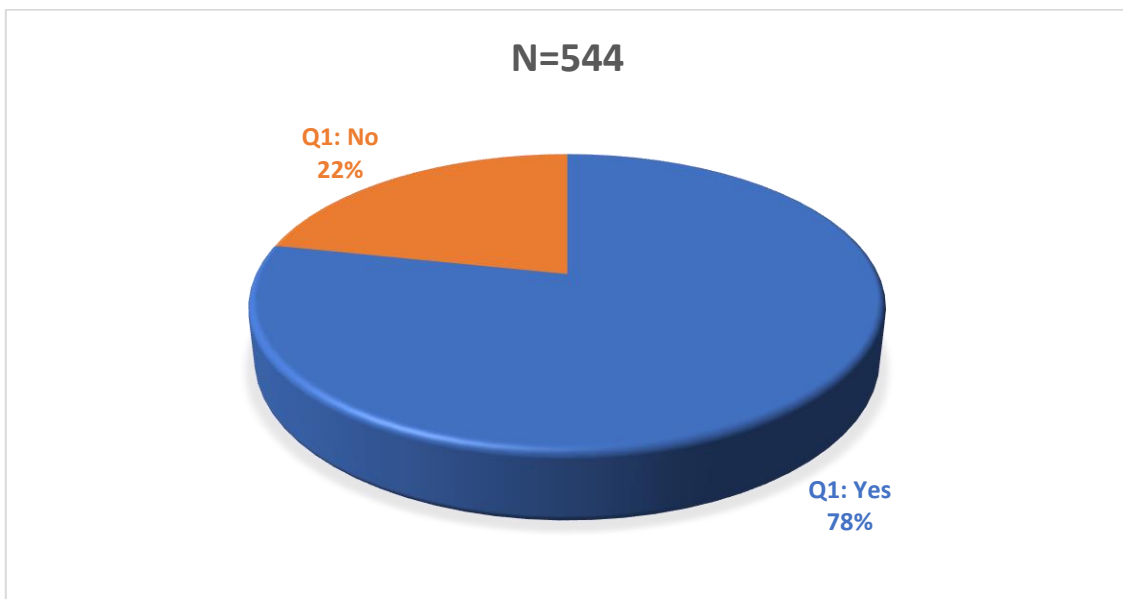
## Results

### A. Compiled data

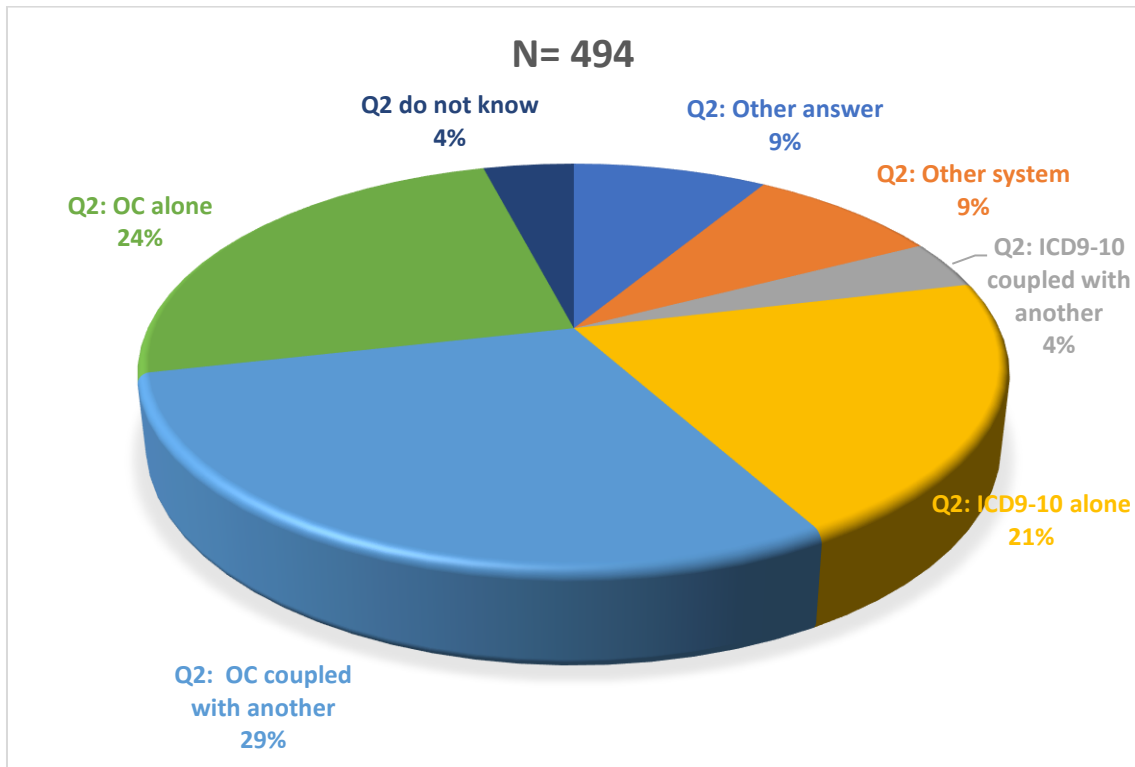
Info	Countries having provided this info	Total	
Reports received	16		
# ERN units contacted	15	1447	Ranging from 629 to 8 according to the country*
Unit Answers received	16	544	
Answer rate		58%	Ranging from 88% to 61% according to the country
Mean coverage of country ERN (at least 1 unit per ERN and per country)	16	85%	Ranging from 100% to 61% according to the country

\*In the Orphanet Knowledge base 1523 units are registered overall for these countries. It must be noted that 1 country has not provided information of the numbers of units contacted and that the Netherlands decided to contact only the units present at RUMC.

#### Q1. Do you provide RD patient data to the ERN coordination?

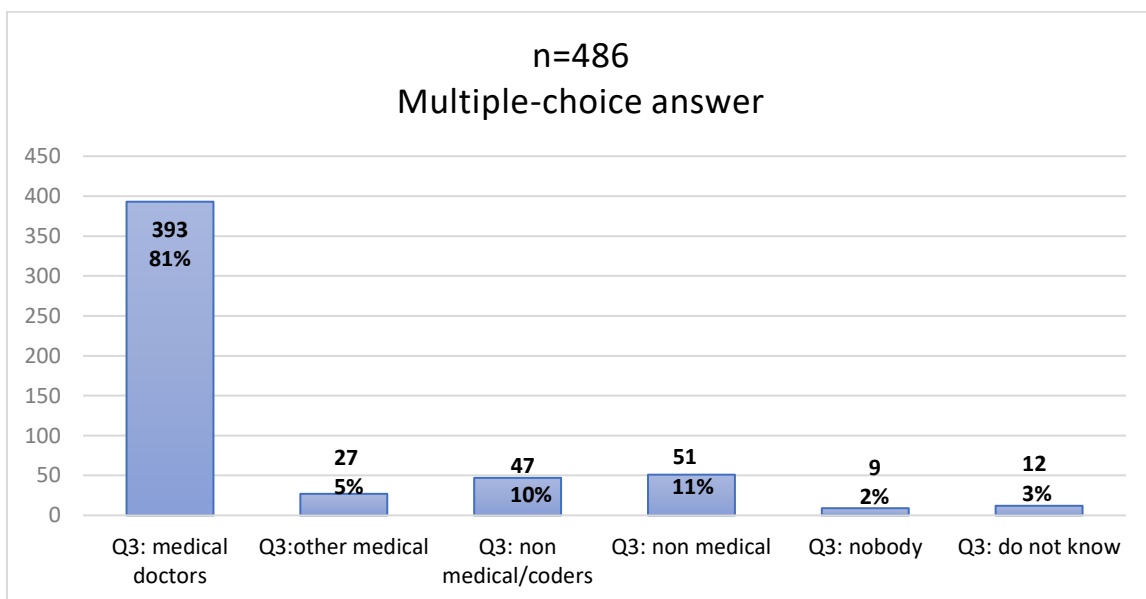


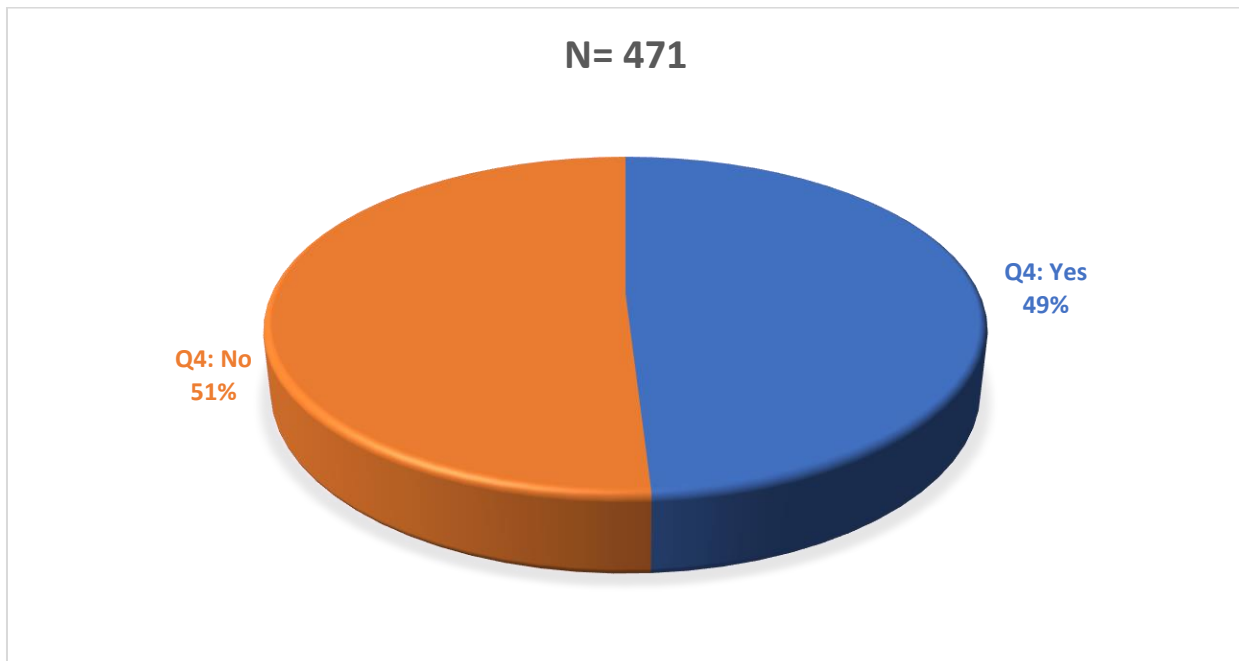
**Q2. What codification system do you use for that purpose?**



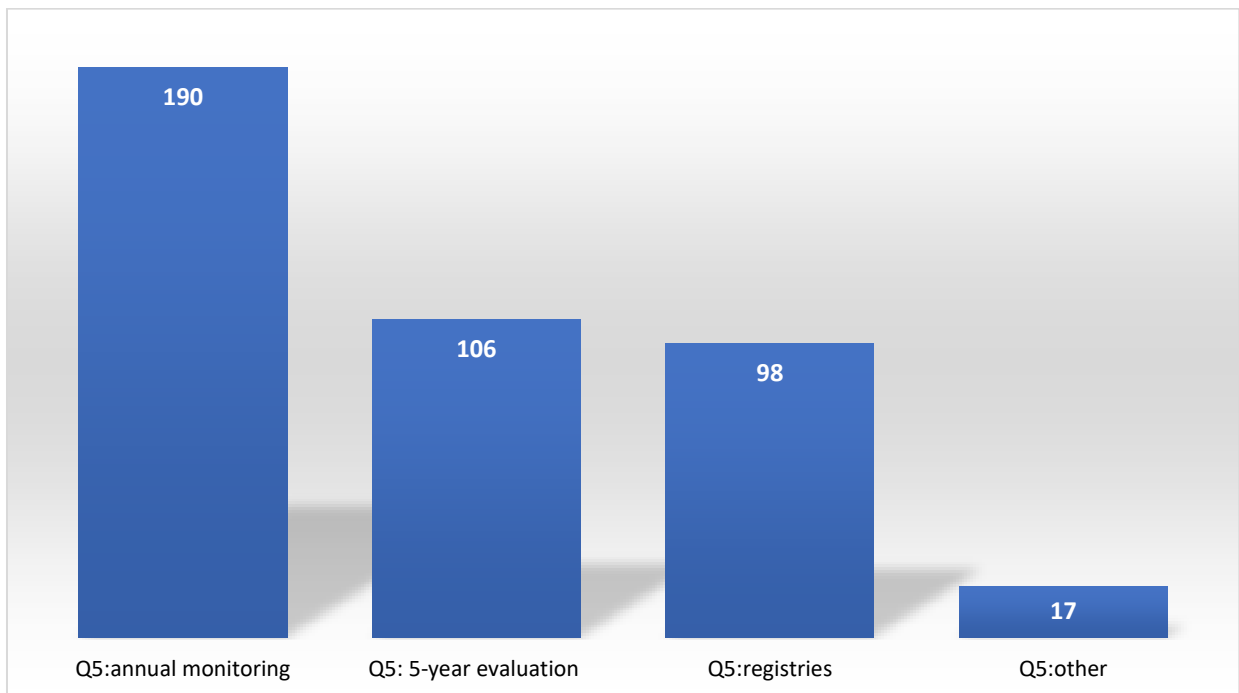
**Q3. What professionals are in charge of codification**

N.B. This was a multiple-choice answer, therefore percentage and values do not add up to the total of respondents

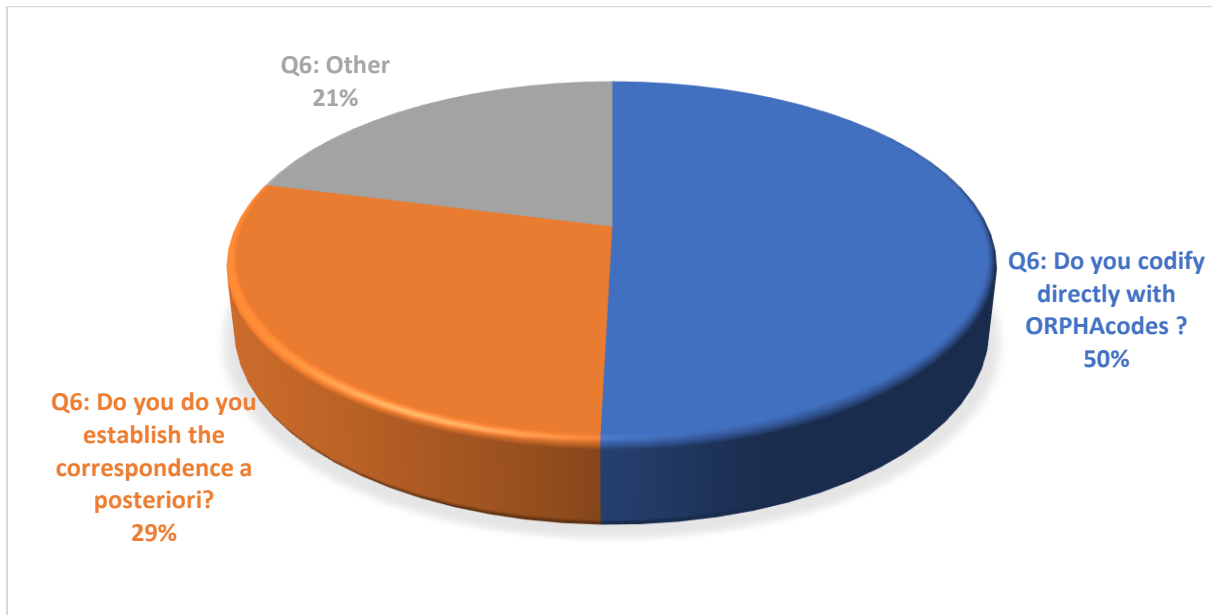


**Q4. Do you use ORPHAcodes to send data to ERN coordination?****Q5. If yes, do you use them for**

N.B. This was a multiple-choice answer; therefore, respondents could choose more than one answer according to their practice



**Q6. Do you codify directly with ORPHAcodes or do you establish the correspondence *a posteriori*?**  
**N=363**



**Q7. Why do you not use ORPHAcodes**

An open question aimed at understating the main reason for not using ORPHAcodes was also present in the survey. Answers for this question were variable according to the users but they have been categorized by theme and the results show that the main reasons for not using ORPHAcodes are (in decreasing importance order):

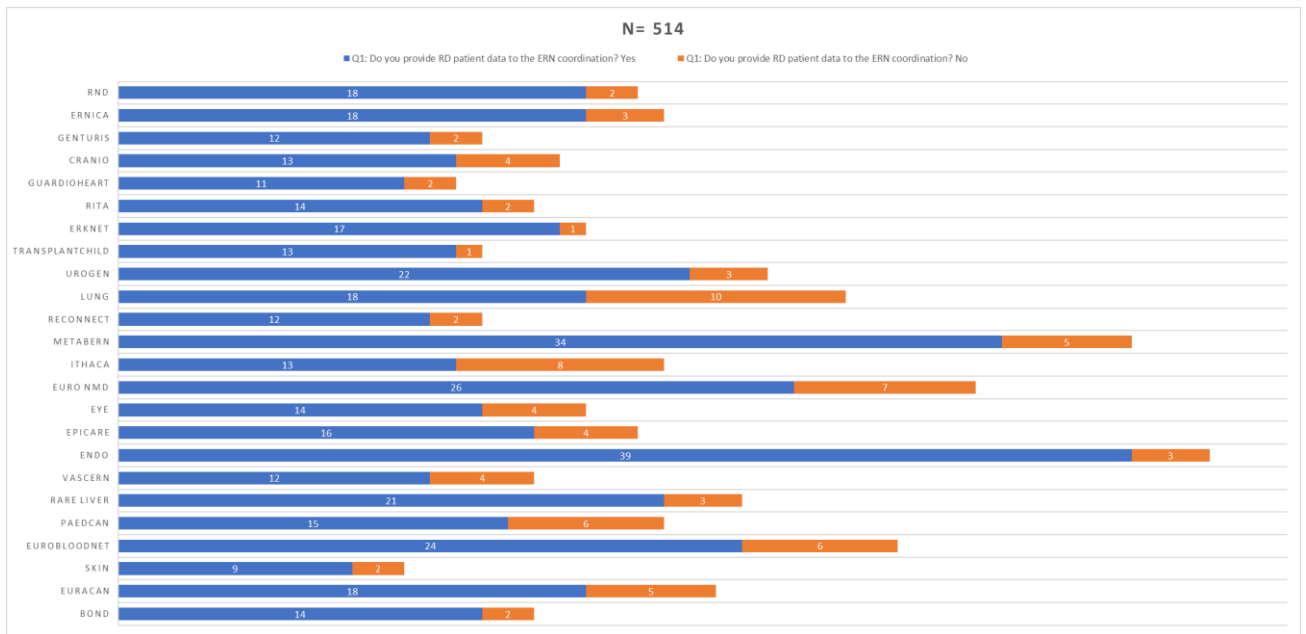
- Unavailability of ORPHAcodes in Health Electronic Records
- ORPHAcodes use not being mandatory
- Lack of time/resources
- ORPHAcodes not suitable
- Lack of knowledge

**B. Analysis by ERN.**

Answers from all countries were pooled by ERN (for this only one answer per unit was considered and in case of discording answers from the same unit, both answers were discarded as we are not able to assess which one is correct). 11 countries performed this analysis: Austria, Belgium, Germany, Italy, Finland, the Netherlands, Norway, Portugal, Poland, Spain, Sweden. In the table below are presented the number of units per the above mentioned countries as per Orphanet database registration vs the number of answers received.

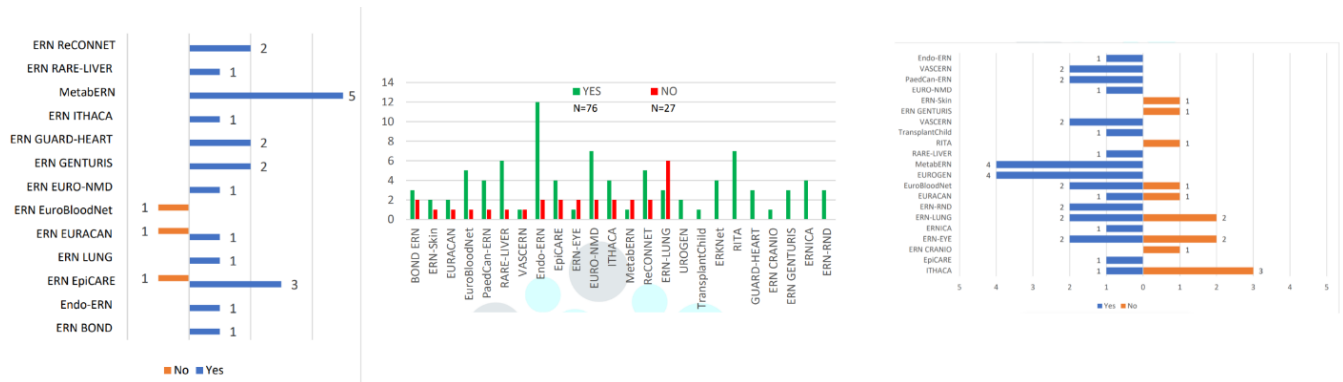
ERN	Total Units registered in the DB for the 11 Countries	Total Answers received
Endo-ERN: European Reference Network on Rare Endocrine Conditions	91	42
EpiCARE: European Reference Network for Rare and Complex Epilepsies	30	20
ERKNet: European Rare Kidney Diseases Reference Network	67	18
ERN CRANIO: European Reference Network on Rare craniofacial anomalies and ENT disorders	30	17
ERN GENTURIS: European Reference Network on GENetic TUmour Risk Syndromes	40	14
ERN GUARD-HEART : Gateway to Uncommon And Rare Diseases of the HEART	37	13
ERN PaedCan: European Reference Network for Paediatric Cancer (haemato-oncology)	80	21
ERN RARE-LIVER: European Reference Network on Rare Hepatological Diseases	52	24
ERN ReCONNET: European Reference Network on Rare and Complex Connective Tissue and Musculoskeletal Diseases	56	14
ERN RITA: Rare Immunodeficiency, Autoinflammatory and Autoimmune Diseases Network	57	16
ERN TRANSPLANT-CHILD: European Reference Network on Transplantation in Children (incl. HSCT, heart, kidney, liver, intestinal, lung and multiorgan)	39	14
ERN-BOND: European Reference Network on Rare Bone Disorders	40	16
ERN-EYE: European Reference Network on Rare Eye Diseases	44	18
ERNICA: European Reference Network for rare Inherited and Congenital Anomalies	37	21
ERN-LUNG: European Reference Network on rare respiratory diseases	93	28
ERN-RND : European Reference Network on Rare Neurological Diseases	70	20
ERN-Skin: European Reference Network on Rare and Undiagnosed Skin Disorders	55	11
EURACAN: European Reference Network on Rare Adult Cancers (solid tumors)	95	23
EuroBloodNet: European Reference Network on Rare Hematological Diseases	90	30
eUROGEN: European Reference Network on urogenital diseases and conditions	54	25
EURO-NMD: European Reference Network for Rare Neuromuscular Diseases	71	33
ITHACA: European Reference Network for Rare Malformation Syndromes, Intellectual and Other Neurodevelopmental Disorders	63	21
MetabERN: European Reference Network for Rare Hereditary Metabolic Disorders	79	39
VASCERN: European Reference Network on Rare Multisystemic Vascular Diseases	41	16
Total	1374	514

Table. Answers pooled by country and by ERN vs number of units expected in the 11 countries



Q1. Do you provide RD patient Data to ERN coordination?

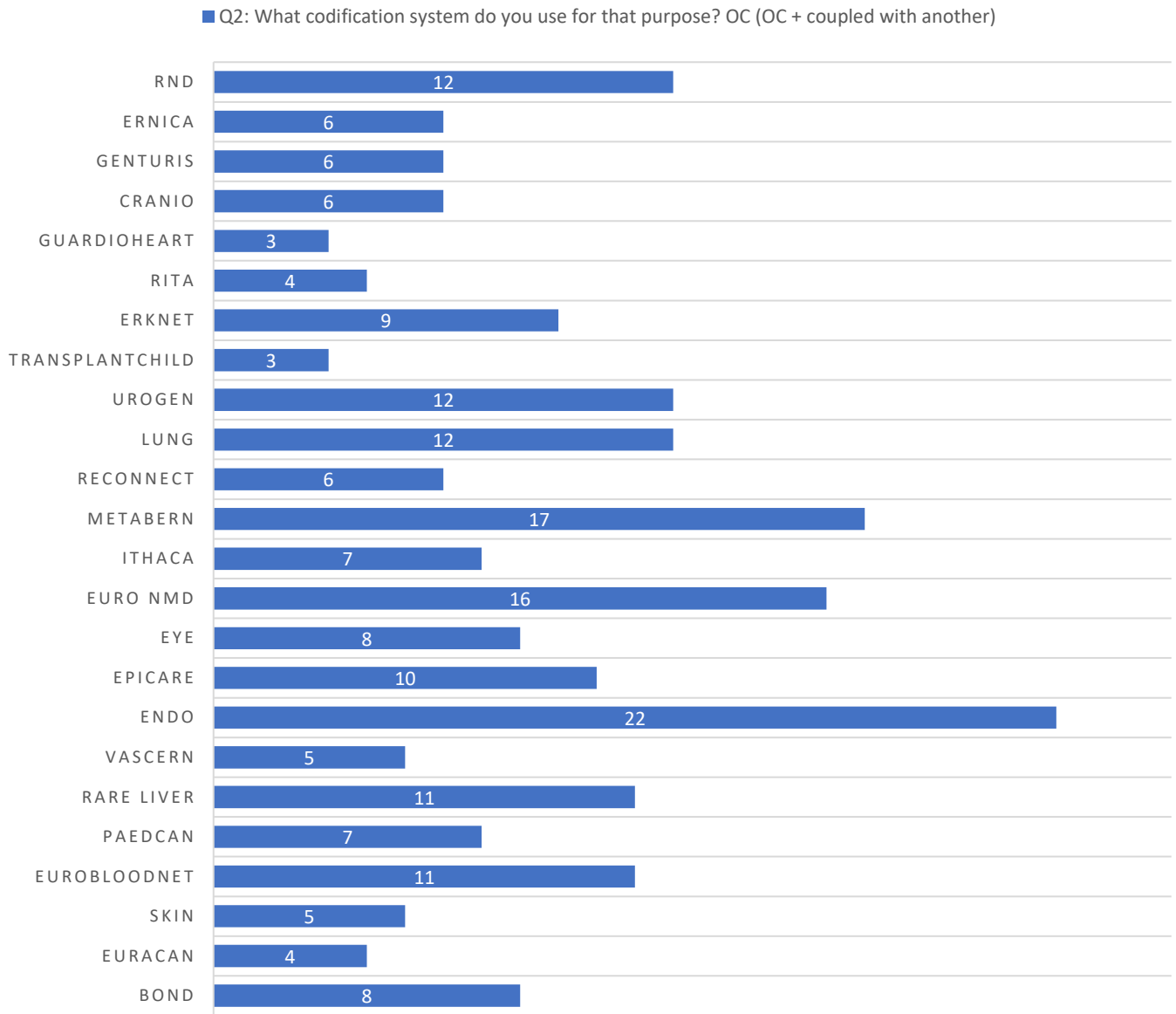
These results show that 82% (423/514) of the units confirm providing RD Patient data to the ERN coordination. But is very heterogeneous with regard to the respective ERNs, indeed for each of the 24 ERNs we observe both “Yes” and “No” answers from the units belonging to each of them. Notably we observe the same heterogeneity from Units within a same ERN within the same country (below as an example results from Spain, Italy and Portugal).



Q1. Do you provide RD patient Data to ERN coordination? Results for Spain, Italy and Portugal

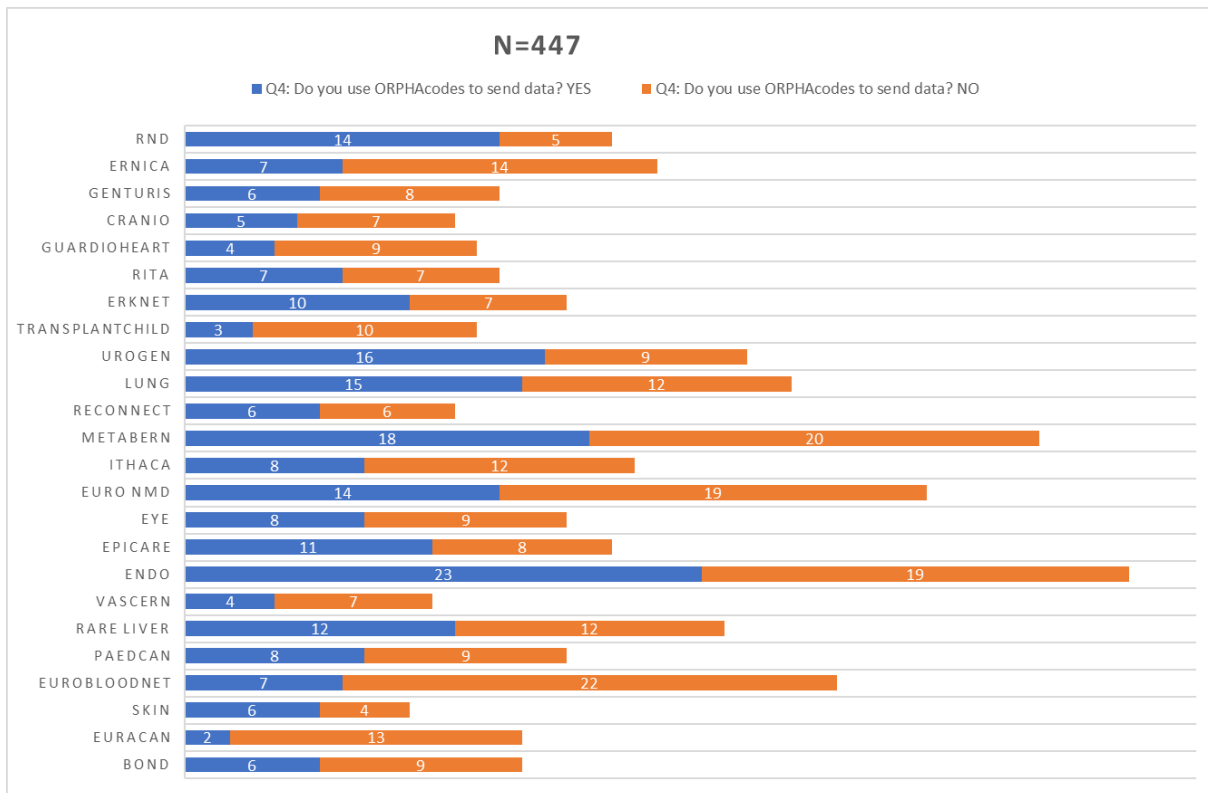


## Q2: WHAT CODIFICATION SYSTEM DO YOU USE FOR THAT PURPOSE? OC (OC + COUPLED WITH ANOTHER) N=210



*Q2. What codification System do you use for what purpose: we pooled the answers selection "ORPHAcodes" and "ORPHAcodes coupled with another"*

210 units confirmed using ORPHAcodes either alone or in combination with another system for the purpose of sending data to the ERN coordination.

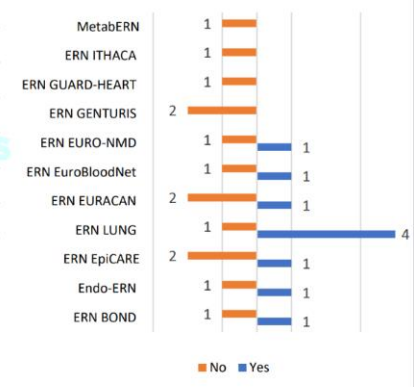
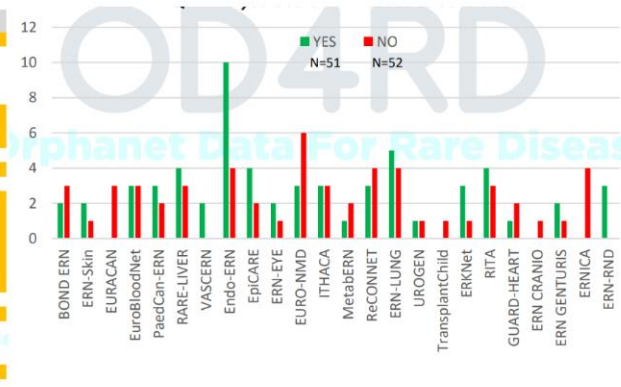


Q4. Do you use ORPHAcodes to send data? Y/N

Here 51% (220/434) confirm using ORPHAcodes to send data. But for this question also, it is very heterogeneous with regard to the respective ERNs, indeed for each the 24 ERNs we observe both “Yes” and “No” answers from the units belonging to each of them. Notably we observe the same heterogeneity from Units within a same ERN within the same country (below as an example results from Spain, Italy and Portugal).

ERN	Total Responses	No	It is optional	It is compulsory
Endo-ERN	4	2		2
ERNet	5		3	2
ERN BOND	2		2	
ERN CRANIO	3	1		2
ERN EpicARE	3		3	
ERN Rare-Liver	3	1	1	1
ERN ReCONNET	3	1		2
ERN Skin	2	1		1
ERN TransplantChild	3	1		2
ERN EYE	3	1		2
ERNICA	3	1		2
ERN LUNG	4	2	1	1
ERN RITA	3	1	1	1
ERN RND	6	2	1	3
EURACAN	3	2	1	
EuroBloodNet	3	1	1	1
EUROGEN	4	1	2	1
EURO-NMD	6	3		3
GENTURIS	2		2	
GUARD-Heart	4	2		2
ITHACA	1		1	
MetabERN	7		5	2
PaedCan-ERN	2		1	1
VASCERN	2	1		1

(Table 1)



Q4. Do you use ORPHAcodes to send data? Y/N. Results for Spain, Italy and Portugal.

## Annex 1. 6 Mandatory Questions

The Six Mandatory Questions are listed below

Q1. Do you provide RD patient data to the ERN coordination?

Y/N

Q2. What codification system do you use for that purpose? 1 choice possible

- Other answer (i.e written diagnosis description...)
- Other system
- ICD9-10 coupled with another (non OC)
- ICD9-10 alone
- OC coupled with another
- OC
- Do not know

Q3. What professionals are in charge of codification? Multiple choice

- medical doctors
- other medical (i.e. nurses...)
- non medical: coders
- non medical (i.e. Administrative staff...)
- nobody
- do not know

Q4. Do you use ORPHAcodes to send data?

Y/N

Q5. If yes, do you use them for:

- Annual monitoring
- 5 Years evaluation
- Registries
- other

Q6. Do you:

- codify directly with ORPHAcodes ?
- establish the correspondence a posteriori?

## Annex 2 Answers received by country

Colonne1	AT	BE	BG	DE	ES	IE	IT	FI	EE	LT	LV	NL	NO	PL	PT	SE
# ERN units contacted	49	94	8	224	103	30	629	32	25		22		33	45	48	68
Unit Answers received	38	62	8	62	77	16	103	28	14	18	15		16	20	24	32
ERN present at national level	24	24	8	24	24	18	24	24	21	18	20	24	17	21	21	21
ERNs represented in the survey (not necessarily by all centres)	22	22	8	21	24	16	24	23	13	16	15	18	13	15	15	17
Q1: Do you provide RD patient data to the ERN coordination? Yes	27	52	11	30	72	12	76	12		16	13	32	9	17	21	26
Q2: What codification system do you use for that purpose? Other answer	18	8		2	1	7	4					0	3		0	
Q2: What codification system do you use for that purpose? Other system	4	11		4	2	1	7			3		0	1	3	7	
Q2: What codification system do you use for that purpose? ICD9-10 coupled with another	0	0		7	1		2					0	1		8	
Q2: What codification system do you use for that purpose? ICD9-10 alone	0	3	11	7	8	3	26	6		3	1	4	2	12	4	12
Q2: What codification system do you use for that purpose? OC coupled with another	4	5	7	8	62	2	16	3		8	13	10	1		7	
Q2: What codification system do you use for that purpose? OC alone	1	22		8	42	1	21	3		2	2		1	10	4	4
Q2 do not know	0	0			0	2				1	1	0	3		0	13
Q3: What professionals are in charge of codification: medical doctors	8	59	17	31	69	8	83	11		14	14	32	8		20	19
Q3: What professionals are in charge of codification: other medical	0	10	2	0	4	1	7					0	1		0	2
Q3: What professionals are in charge of codification: non medical/coders	0	8		17	16	3						0	0		3	
Q3: What professionals are in charge of codification: non medical	1	11	2	6	4	1	6	12		1	0	0	0		0	7
Q3: What professionals are in charge of codification: nobody	0	0		0	0	6	1					0	0		2	
Q3: What professionals are in charge of codification: do not know	0	0		0	0	1	6				1	0	0		0	4
Q4: Do you use ORPHAcodes to send data? YES	5	26	7	17	57	3	51	7		9	13	12	3	10	10	1
Q4: Do you use ORPHAcodes to send data? NO	22	36		21	20	13	52	16		7	1	4	6		14	28
Q4 total answers	27	62		38	77	16	103	23			15	16	9		24	32
Q5: If yes, do you use them for annual monitoring	5	19		17	60	2	38	6		6	12	4	3	6	9	3
Q5: If yes, do you use them for 5-year evaluation	1	12		12	8	2	21	23		3	8	8	1	5	1	1
Q5: If yes, do you use them for registries	2	24		9	16	2	26	2		2	8		1	2	4	
Q5: If yes, do you use them for other		0		6	0					4	0	0	0		0	7
Q6: Do you codify directly with ORPHAcodes ?	1	25	4	10	47	3	14	23		2	7	32	1	10	4	
Q6: Do you do you establish the correspondence a posteriori?	4	11	8	14	12		34			2	4	0	0		9	6
Q6: Other	0	26		5	4		3			7	3	0	2		0	26

