

# Collection and codification of patients without determined diagnosis in ERN registries

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## Cross-ERN survey mapping the current situation

The principal aim of the European Reference Networks (ERNs) is to improve healthcare for rare disease patients, including that more people obtain a diagnosis. To improve diagnostic approaches and care pathways, it is of critical importance to know which people have not yet been diagnosed so that targeted strategies can be developed.

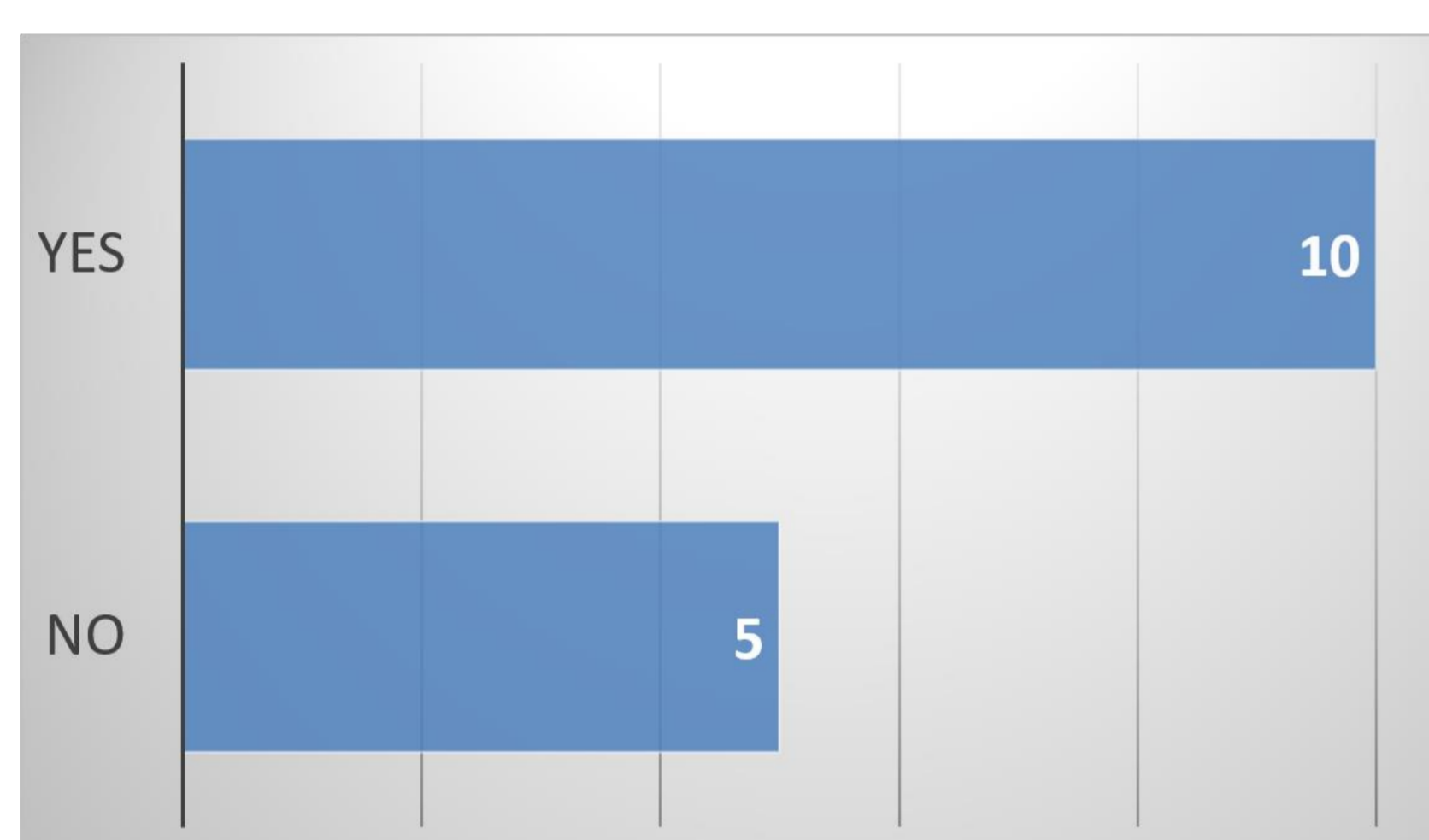
All 24 ERNs have to collect rare disease patient data in ERN registries. This might include patients without a determined diagnosis which means that the diagnosis is neither clinically nor molecularly confirmed. These patients face specific problems, for example not being able to access disease specific self-help groups and social support nor enrolling in clinical studies (Berger et al. 2021 Orphanet Journal of rare diseases volume 16). A new orphacode 616874 was introduced to codify for rare disorder without a determined diagnosis after full investigation (<https://www.orpha.net/en/disease/detail/616874>).

As it is not known whether the ERN registries collect data on patients without a determined diagnosis falling in the respective disease groups covered by an ERN how the different ERNs collect and codify these patients we have conducted a survey across all 24 ERN registries.

## Results

15 ERNs (63%) responded to the survey and furthermore provided additional explanatory information.

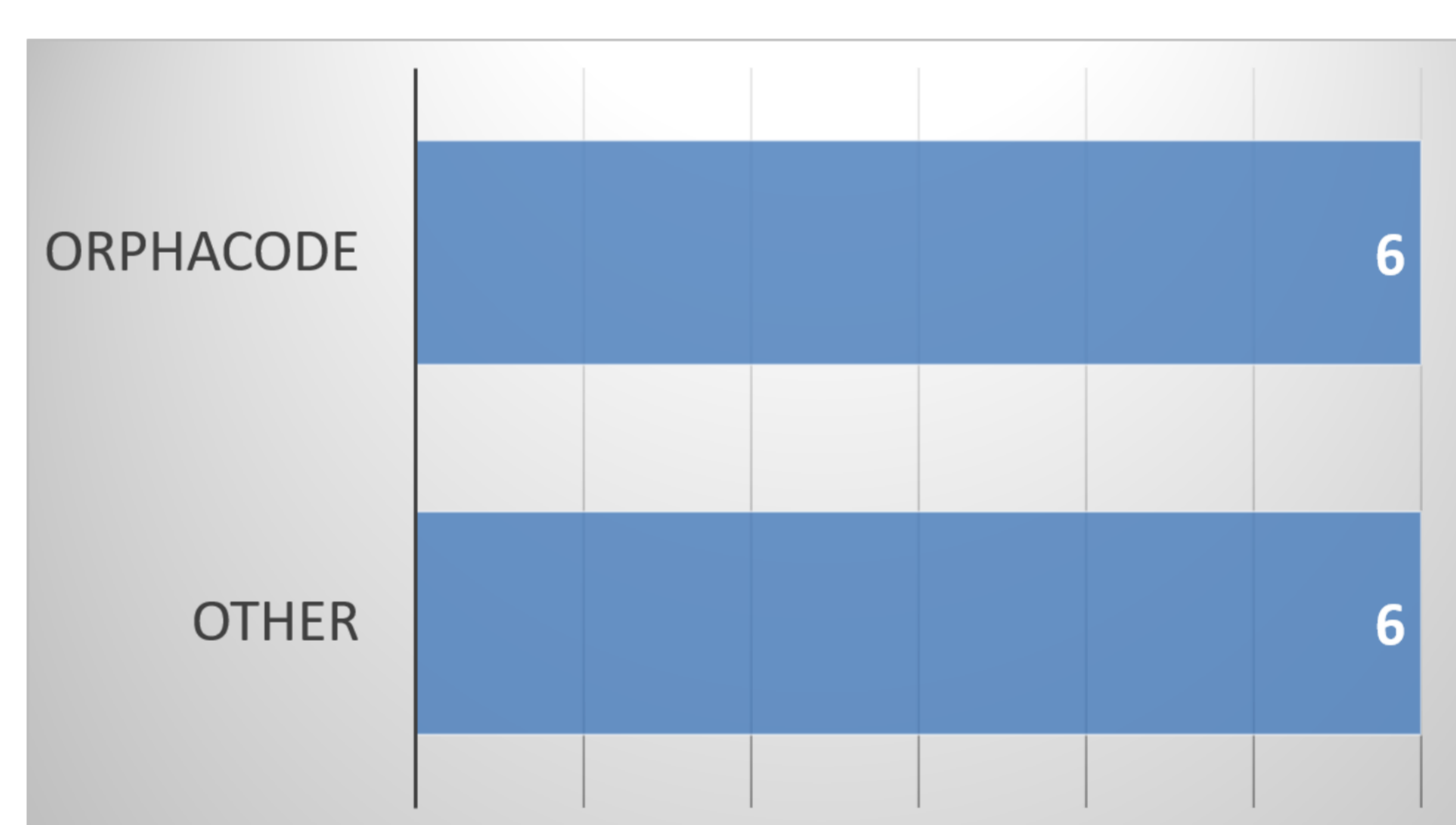
### 1. Does your ERN registry include patients without determined diagnosis?



Reasons not to include patients without determined diagnosis:

- only diagnosed patients are in the scope of the registry
- diagnose necessary to name the disease
- not yet decided in the ERN to do so

### 2. How is a patient without a determined diagnosis codified/marked in your registry?\*

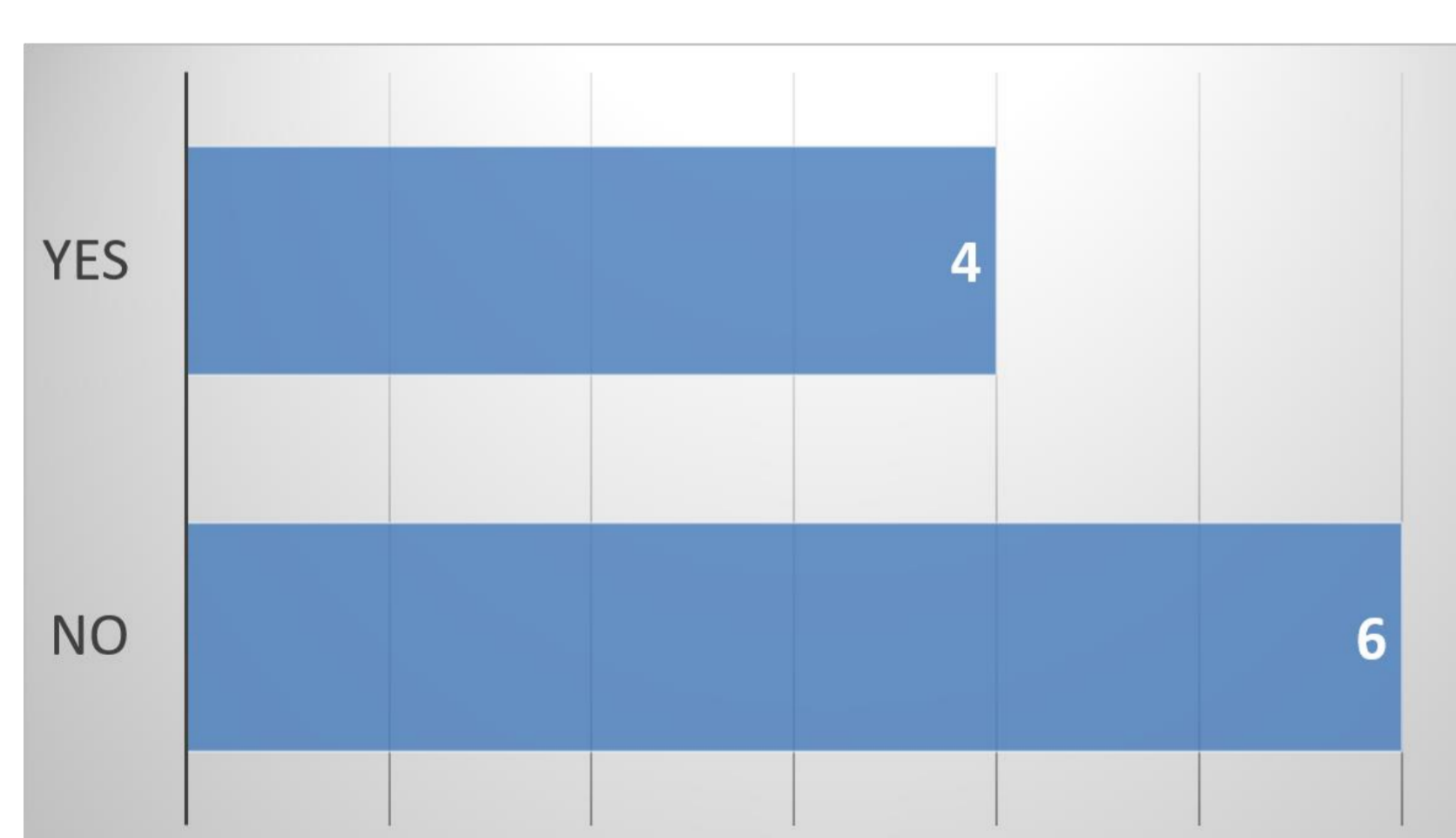


OTHER codifying/markings:

- term „unknown“ used
- to name the disease group
- report a genetic diagnosis with clinVar/dbSNP/HGVS-notation
- HPO/ICD10
- OMIM
- open text

\*referring to „yes“ in question 1; more than one answer possible, orphacode in general (not only 616874)

### 3. Do you include patients that have been codified with the orphacode 616874?\*

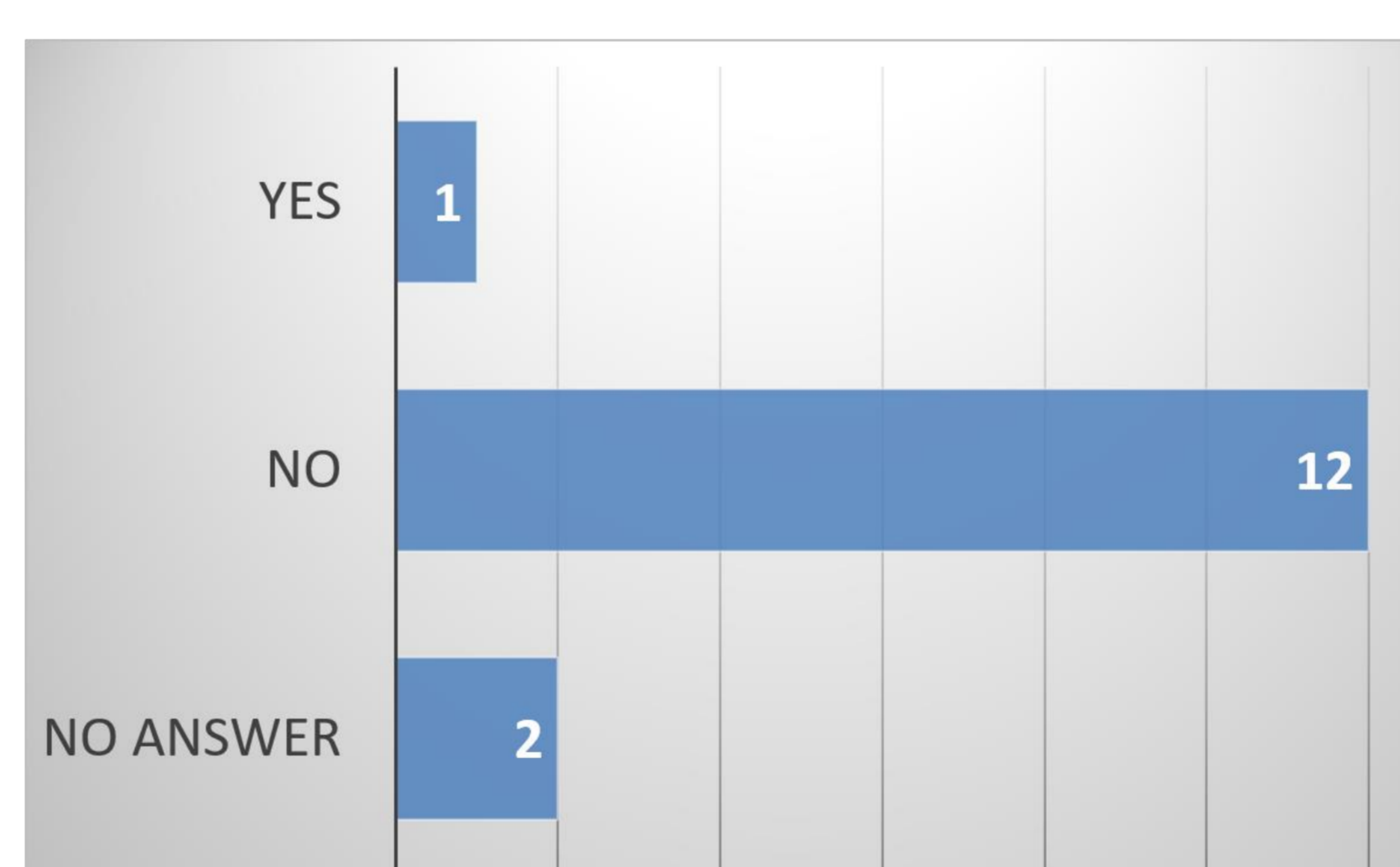


Further comments:

- it is discussed to use the ORPHAcode 616874

\*referring to „yes“ in question 1

### 4. Do you know, if HCPs of your ERN are using the orphacode 616874?



One ERN provided a list of HCPs using this orphacode.

## Conclusions

The results provide an overview of as to whether and how ERN registries include, collect data on and codify patients without a determined diagnosis. Two thirds of the ERN registries that responded include data on these patients.

However, the manner how patients without a determined diagnosis are included is characterized by a high degree of heterogeneity. In particular, codification is handled very heterogeneously. The new established orphacode 616874 for rare disorder without a determined diagnosis after full investigation has hardly been used by ERN registries.

## Next step

An already established cross-ERN working group will use these survey results to try to develop an approach how to collate and analyse data on patients without a determined diagnosis across ERN registries. This will provide the basis to improve and harmonise data collection but also to analyse the data in a longitudinal fashion.

## Outlook

Depending on an improved data collection and the ability to analyse data of patients without a determined diagnosis, this work might evolve into a mean to monitor absolute and relative numbers of these patients in Europe in particular in relation to diagnosed rare disease patients.