www.od4rd.eu   pmt.orphanet@inserm.fr   @orphacodes

https://github.com/OD4RD/Main-Help-Desk @ [GitHub country alias]

www.orphadata.com/ orphanet-nomenclature-for-coding
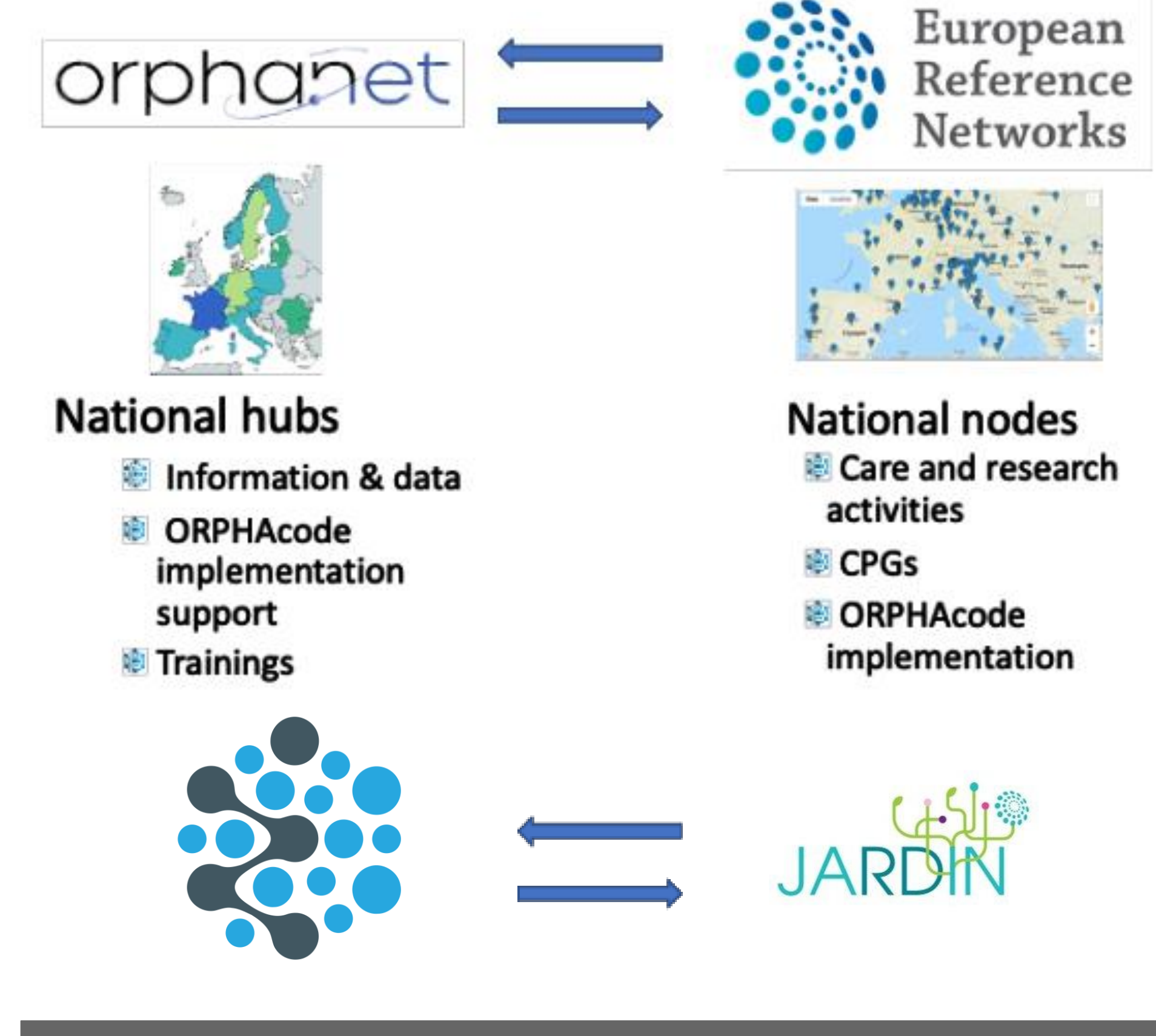
OD4RD
Orphanet Data For Rare Diseases

Tackling the invisibility of RD in European Member States: the OD4RD project contribution

Sylvie Maiella 1, Marie-Cécile Gaillard 1, Kurt Kirch 2, Carina Thomas 2, Julie Tarahoui 1, Marta Fructuoso 1, Caterina Lucano 1, Elsa Ekblom 3, Charlotte Rodwell 1, Valérie Serrière-Lanneau 1, Marc Hanauer & Stefanie Weber 2 , Rula Zain 3 & Ana Rath 1
& the Orphanet Nomenclature National Hubs

[1] Inserm US14 – Orphanet, France [2] BfArM, Germany; [3] Karolinska Institute, Sweden

Introduction & Rationale

The OD4RD/OD4RD2 projects tackle the invisibility of rare diseases in European member states' health systems, promote harmonisation of practice and facilitate generation of standardised interoperable data around RD, thus contributing to meet the ambitions set by RARE2030 concerning data, with:
- three main general objectives
- a wide geographical coverage (see map)
- by creating cross-links with the JARDIN joint action to further facilitate the implementation of ORPHAcodes which is instrumental for RD visibility in Health Information Systems.
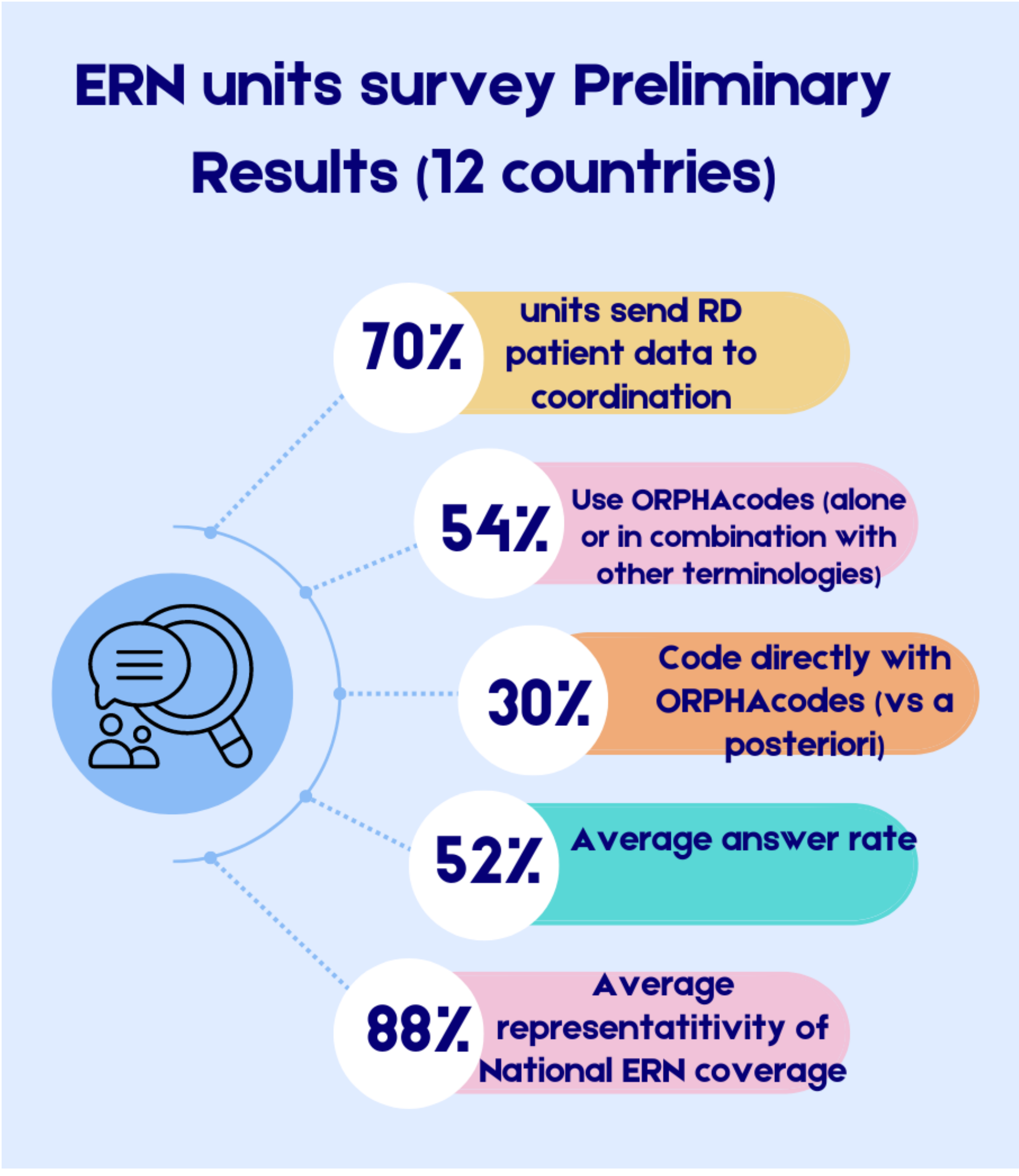
Orphanet ↔ **European Reference Networks**

National hubs
- Information & data
- ORPHAcode implementation support
- Trainings

National nodes
- Care and research activities
- CPGs
- ORPHAcode implementation

JARDIN

To contribute to the generation of standardised, interoperable data on RD diagnosis for primary and secondary use, by the maintenance and the support to the implementation of the Orphanet nomenclature of RD

To contribute to the harmonization of data collection amongst settings (health records, registries) **and amongst countries**, by the dissemination of coding good practices at the data source level

To contribute at supporting evidence-based decision-making in the frame of the European strategy around ERNs, by supporting the exploitation of reference corpus of data and information on RD

Methodology and Results
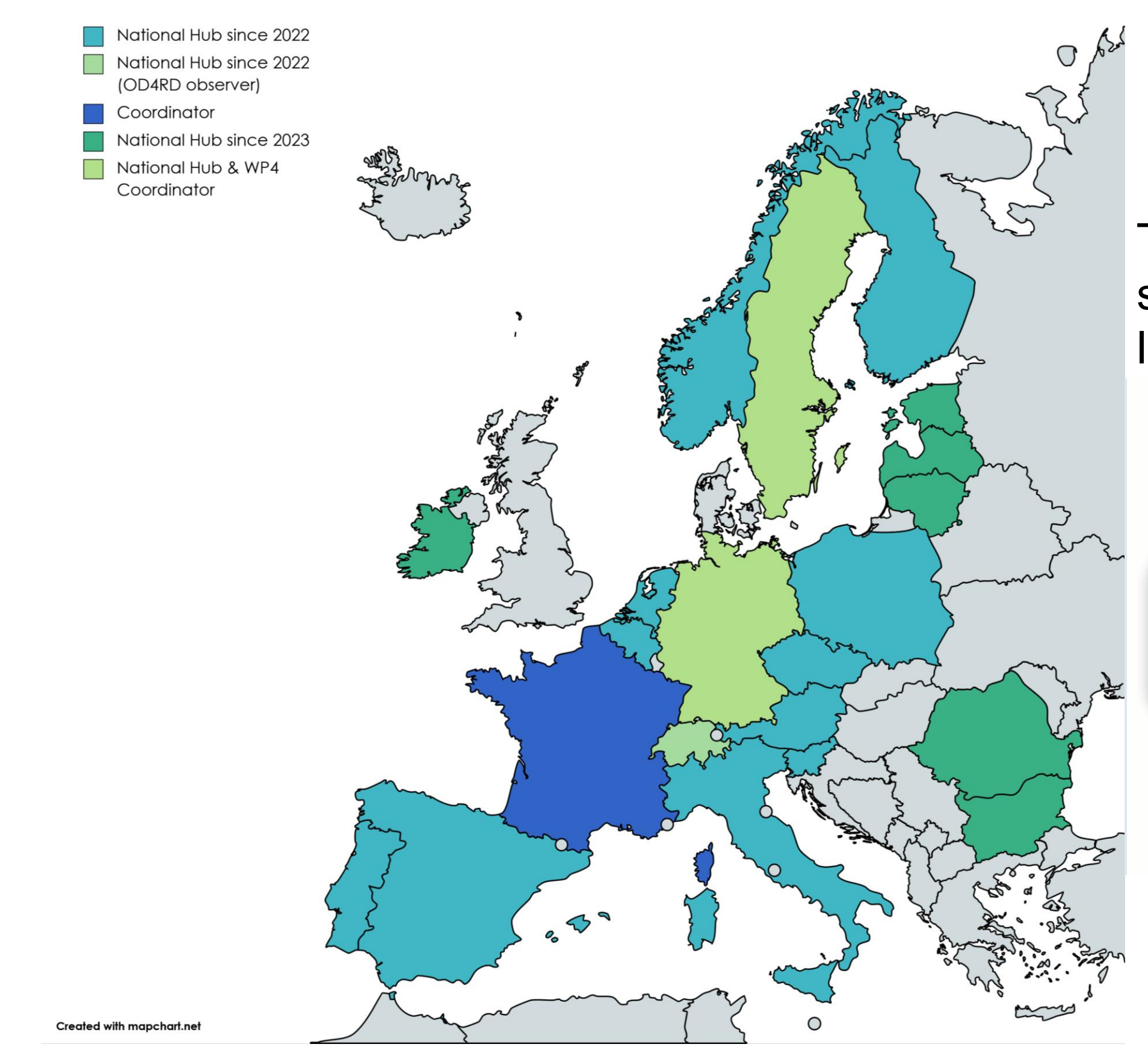
Consolidate and Expand national Orphanet Nomenclature Hubs

The project provides coordinated support for ORPHAcodes implementation in Health Information Systems of 20 member states' hospitals hosting ERNs thanks to the growing Network of National Nomenclature Hubs, so as to reinforce the national level to add European value. (see Fig 1)
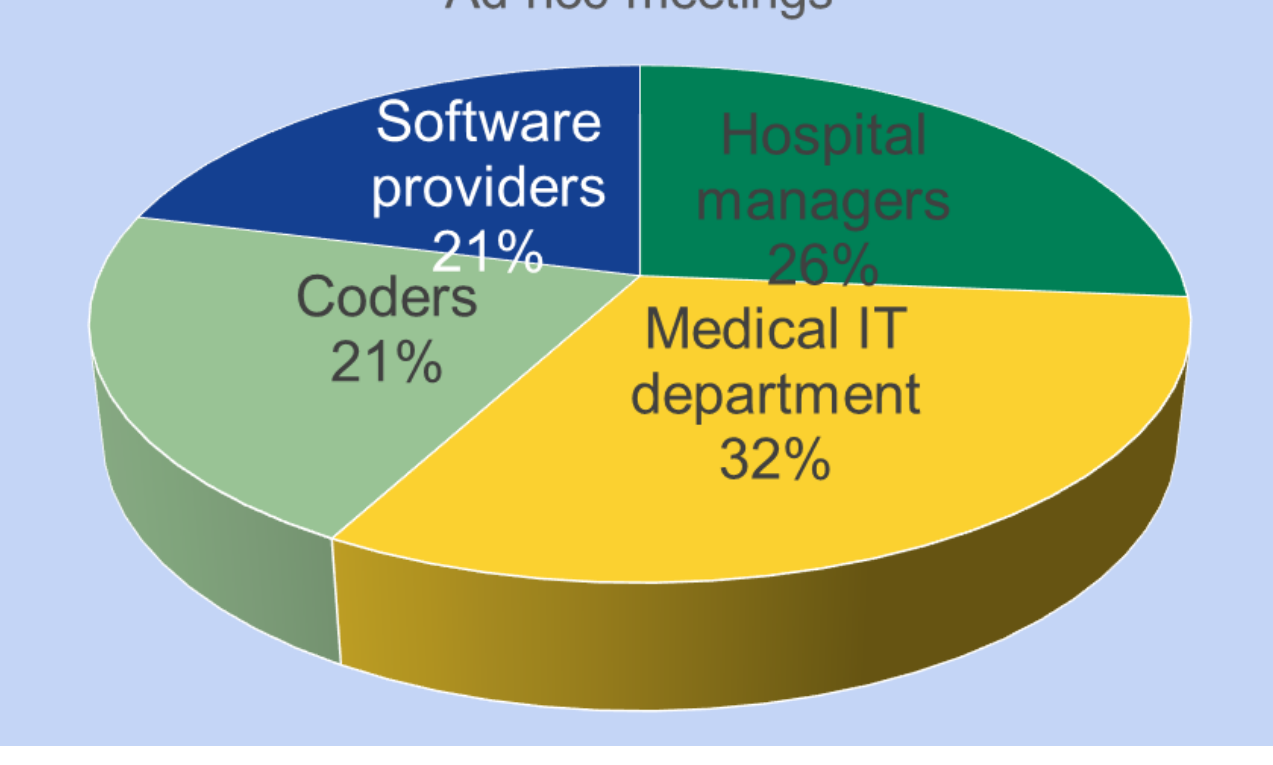
Legend:
- National Hub since 2022
- National Hub since 2022 (OD4RD observer)
- Coordinator
- National Hub since 2023
- National Hub & WP4 Coordinator

Created with mapchart.net

Assess users'needs at national level

Organise capacity building around the Orphanet Nomenclature & tools

Support & advice on implementation & ORPHAcode use

ERN units survey Preliminary Results (12 countries)

70% units send RD patient data to coordination

54% Use ORPHAcodes (alone or in combination with other terminologies)

30% Code directly with ORPHAcodes (vs a posteriori)

52% Average answer rate

88% Average representatitivity of National ERN coverage

Assess users'needs: A survey to evaluate the current situation of ORPHAcodes usage by health care providers (HCPs) linked to ERNs in countries participating in OD4RD has been carried out, in order to be able to fine tune both hubs' National Action Plans but also transversal actions at the OD4RD Coordination level. In every country the National hub contacted at least each contact point of each ERN unit as per registration in the Orphanet Database and submitted a subset of questions (min. of 7 questions, which could be expanded according to national needs).

Interestingly, the preliminary results, deriving from the analysis of 12 national survey reports (representing >440 Units), indicate that patient data sharing with the ERN coordination is very heterogeneous in relation to the respective ERNs, as well as within a same ERN within the same country.

Main reasons for not using ORPHAcodes is (in decreasing importance order):
- **unavailability of ORPHAcode in HER**
- **ORPHAcode use not being mandatory**
- **lack of time/resources**
- **codes not suitable**
- **lack of knowledge**

Capacity building events organised according to national needs
- **110** Trainings in National Language in **15** countries
- **2850+** participants of which **236** ERN representatives
- **122** Ad Hoc Meetings

Ad hoc meetings
- Software providers 21%
- Hospital managers 26%
- Medical IT department 32%
- Coders 21%

Support & advice on implementation & ORPHAcodes used:
National Helpdesk to address & discuss coders' needs & queries in National Language:
- **12** national Githubs in place with **284** demands received mainly by clinicians (43%) and coders (32%).
- 53% Demands handled in autonomy by each National Hub and 47% handled by the coordinating team & discussed collectively for capacity building and standardisation of practice

Ticket type
- Coding 44%
- Nomenclature 34%
- Others 22%
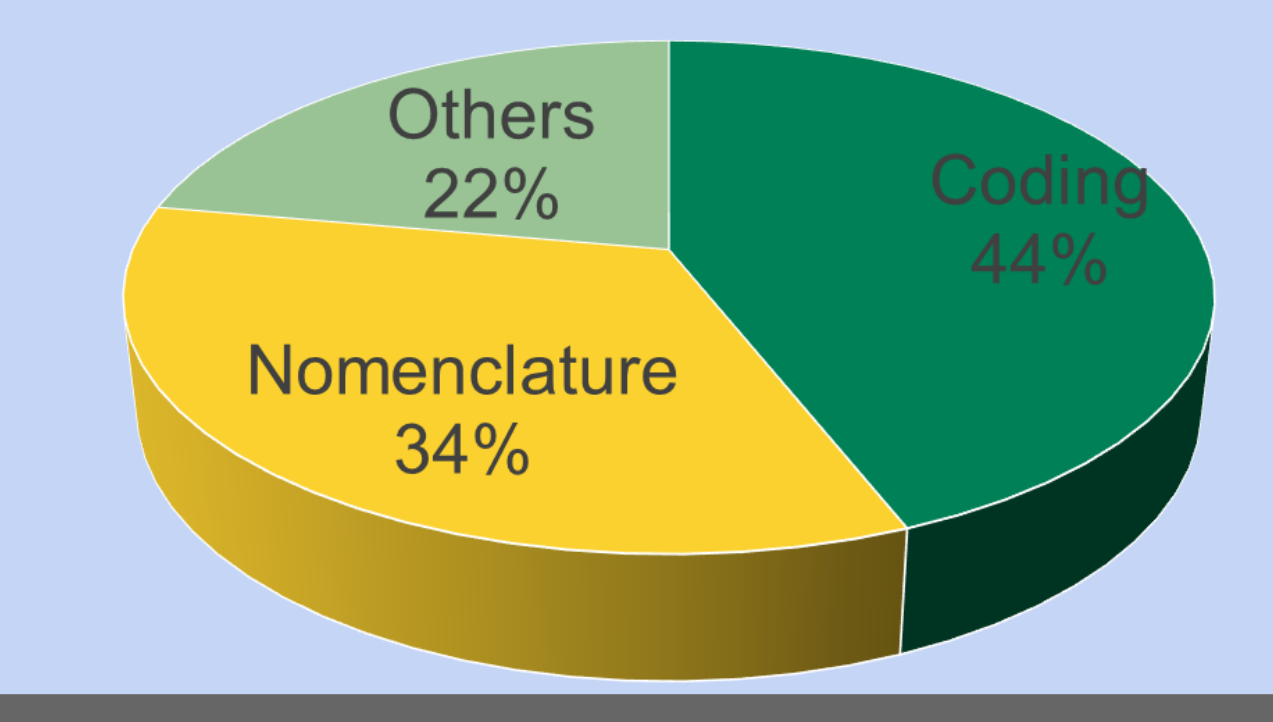
Conclusions

- The operations around organisation of capacity building and support and advice are of satisfaction and match project goals, but the need to increase ERN attendance to trainings seems clear.
- The interesting preliminary survey results will be compared with those that will be obtained in the JARDIN survey, in order to obtain a complete picture of practices across ERN and across countries. Globally these confirm the project strategy is in line with the « field » barriers identified in the survey
- Improvements already in place: increase National hubs capacity-building around the interoperability with other terminologies work carried out at Orphanet headquarters & around the existing studies demonstrating the benefits of ORPHAcoding to trace RD DIAGNOSIS. A General Public Booklet around the benefits of ORPHAcoding has been issued in English.
- What's next: Cross-link and fine-tune advocacy and demonstrator activities with JARDIN, increase numbers of ERN trainings at National level, organise a ERN day and Hospital manager Day to raise awareness about ORPHAcoding benefits and to discuss with ERN coordinators on their strategy regarding data to facilitate harmonisation.
- A white paper on implementation issues of ORPHAcodes in relation to other code systems will be delivered in 2025