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WEBINAR JARDIN

**“SURVEY RESULTS.
RARE DISEASE DATA
MANAGEMENT:
BARRIERS AND
EXISTING
SOLUTIONS”**

Join us!



4th

**MARCH
2025**



15:00 - 16:30 H





Agenda

- 1. Welcome & Introduction**
- 2. Objectives**
- 3. Survey method & participation**
- 4. Primary data capture**
- 5. Data Flow**
- 6. Registries**
- 7. Data Sharing & FAIRness**
- 8. Discussion**



Agenda

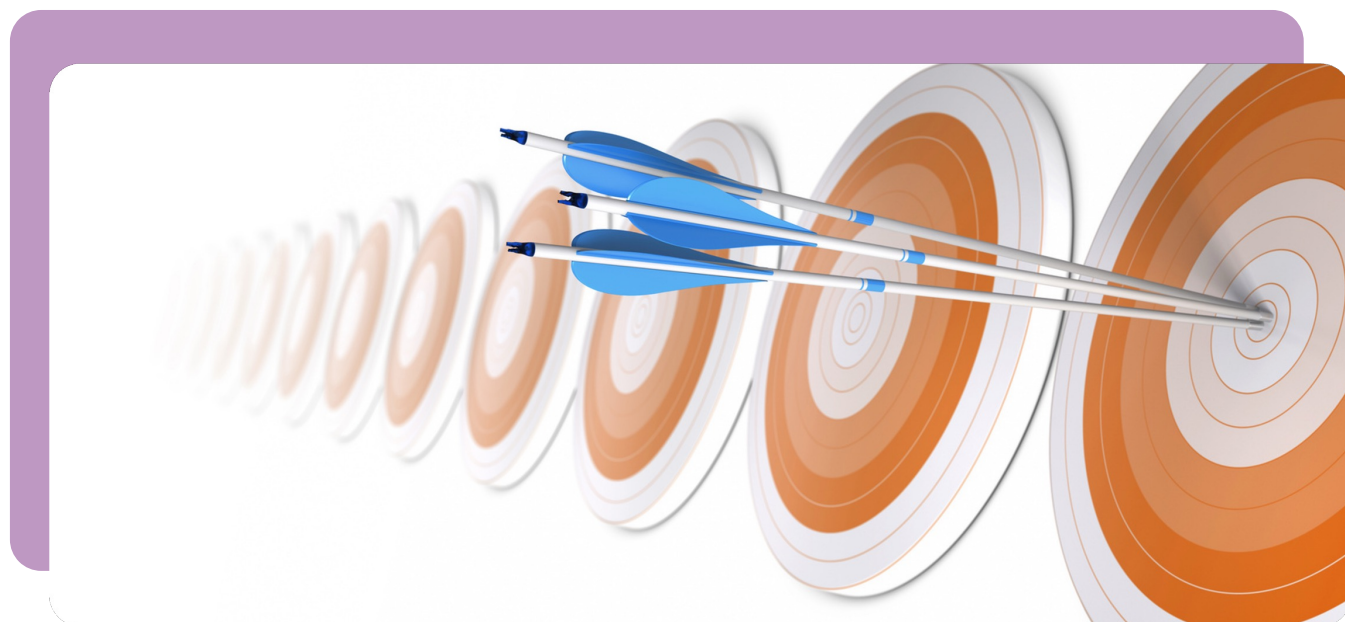
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2. Objectives of the webinar



- Inform you about the outcomes of the surveys to the hospital level
- Gather your feedback on the results
- Collect additional information using mentimeter

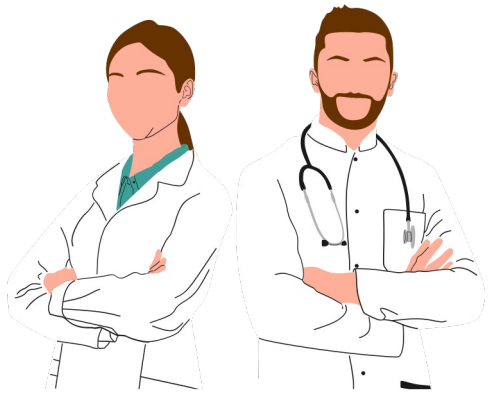


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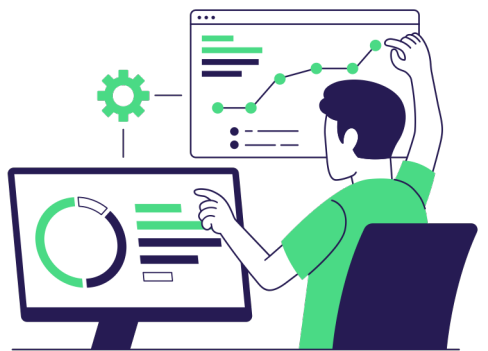
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3. Survey method

- Aim: to find out about current data management practices
- Questionnaires were developed cooperatively with stakeholders involved in the JARDIN Data Management work packages (WP8)
- for the hospital level two target specific questionnaires were sent out:



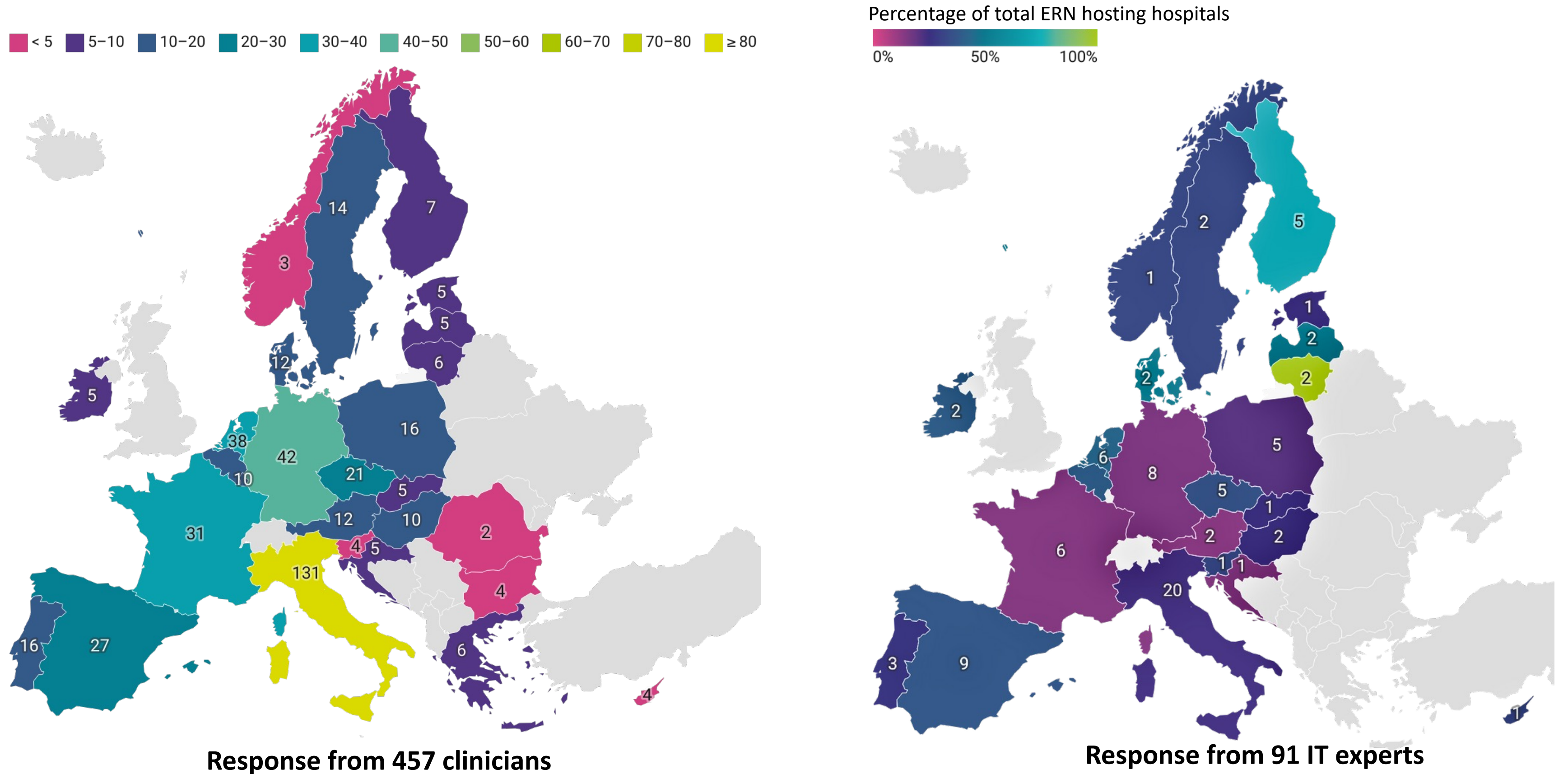
1. ERN unit leads (Clinicians leading an ERN center)



2. IT experts/Data stewards at the ERN hosting hospitals

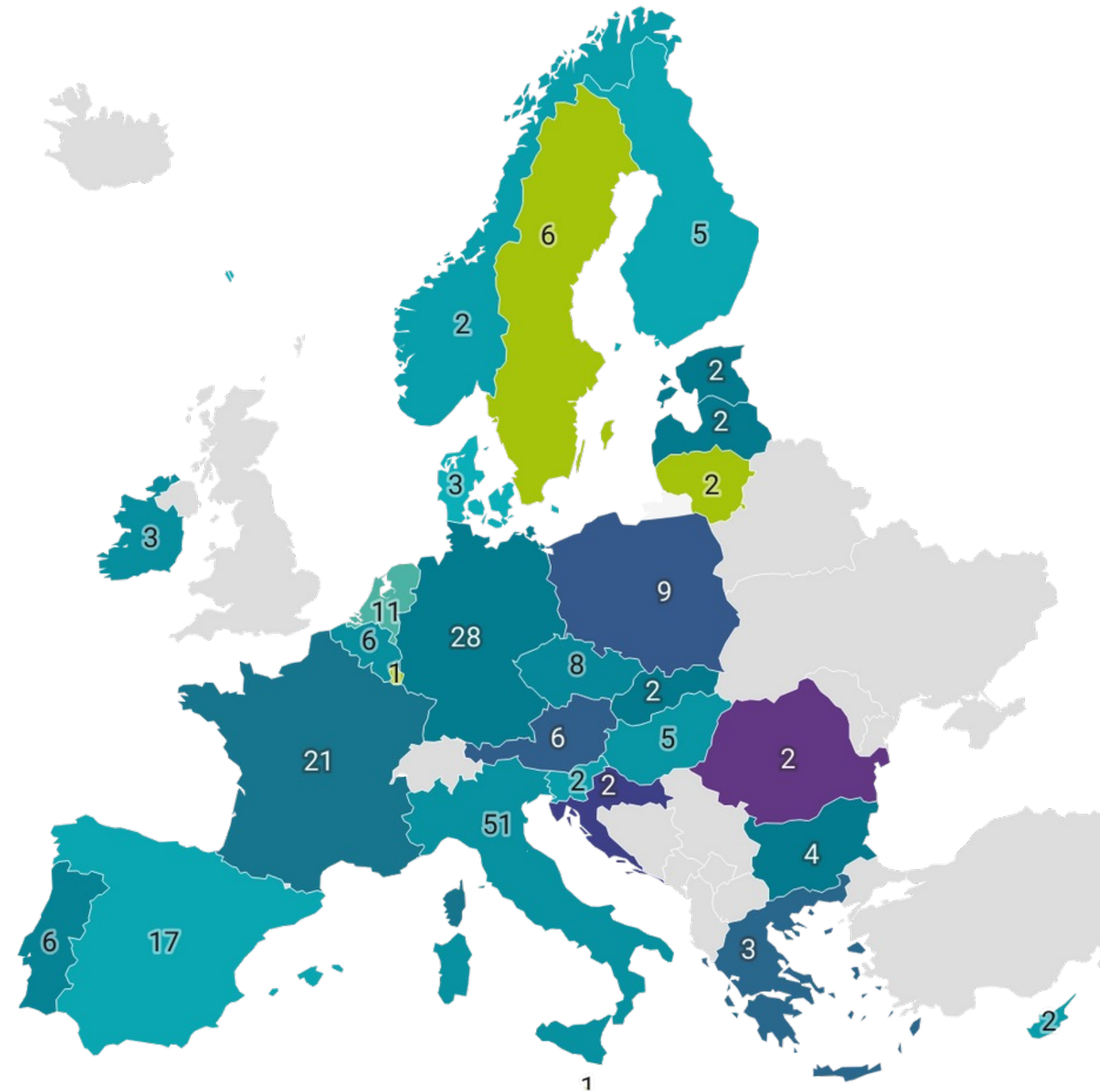


3. Participation per questionnaire

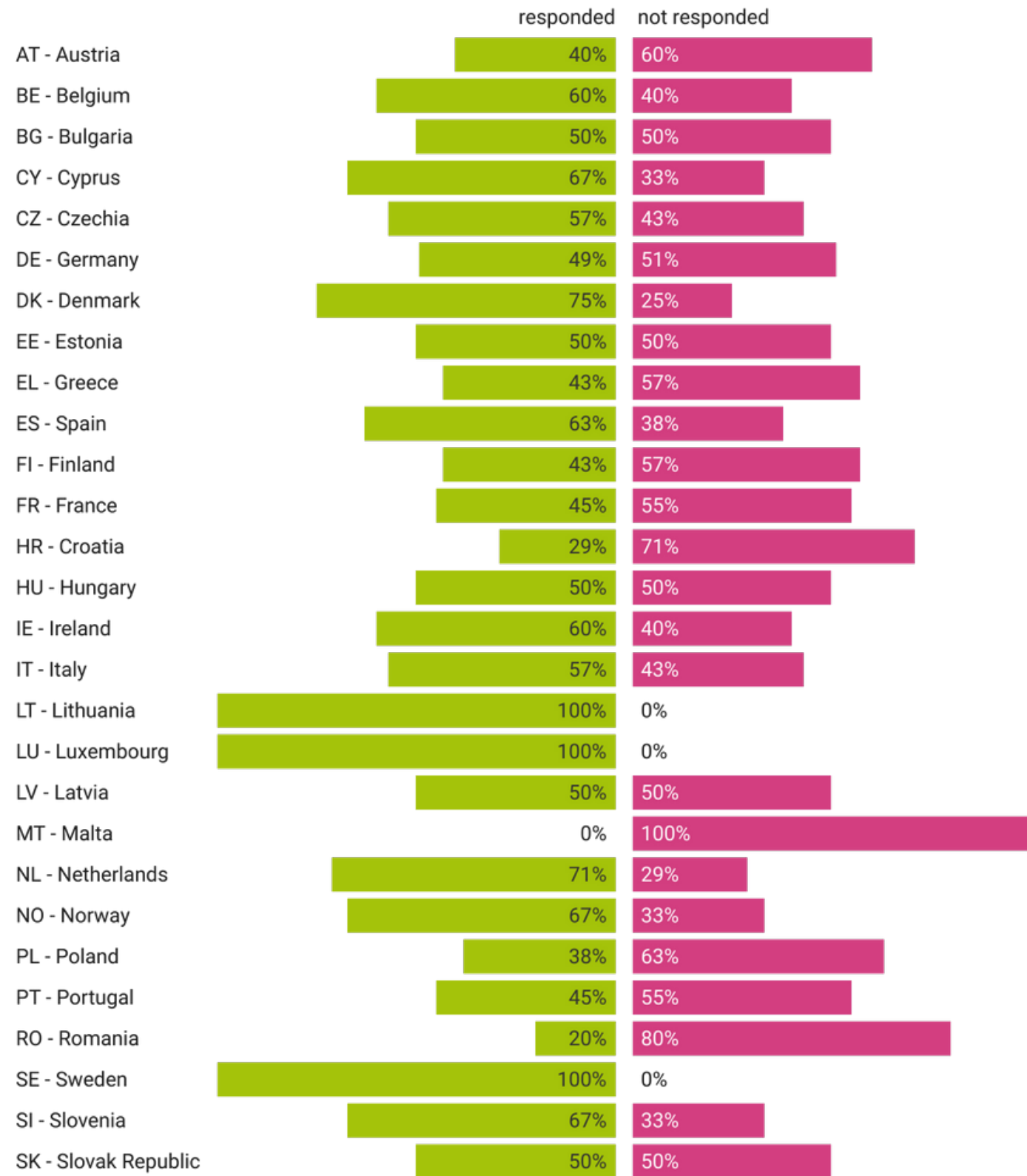


3. Participation

Responsdence of ERN hosting sites



■ responded ■ not responded



3. Participation

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Who is out there?



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4. Primary Capture of RD Patient Data



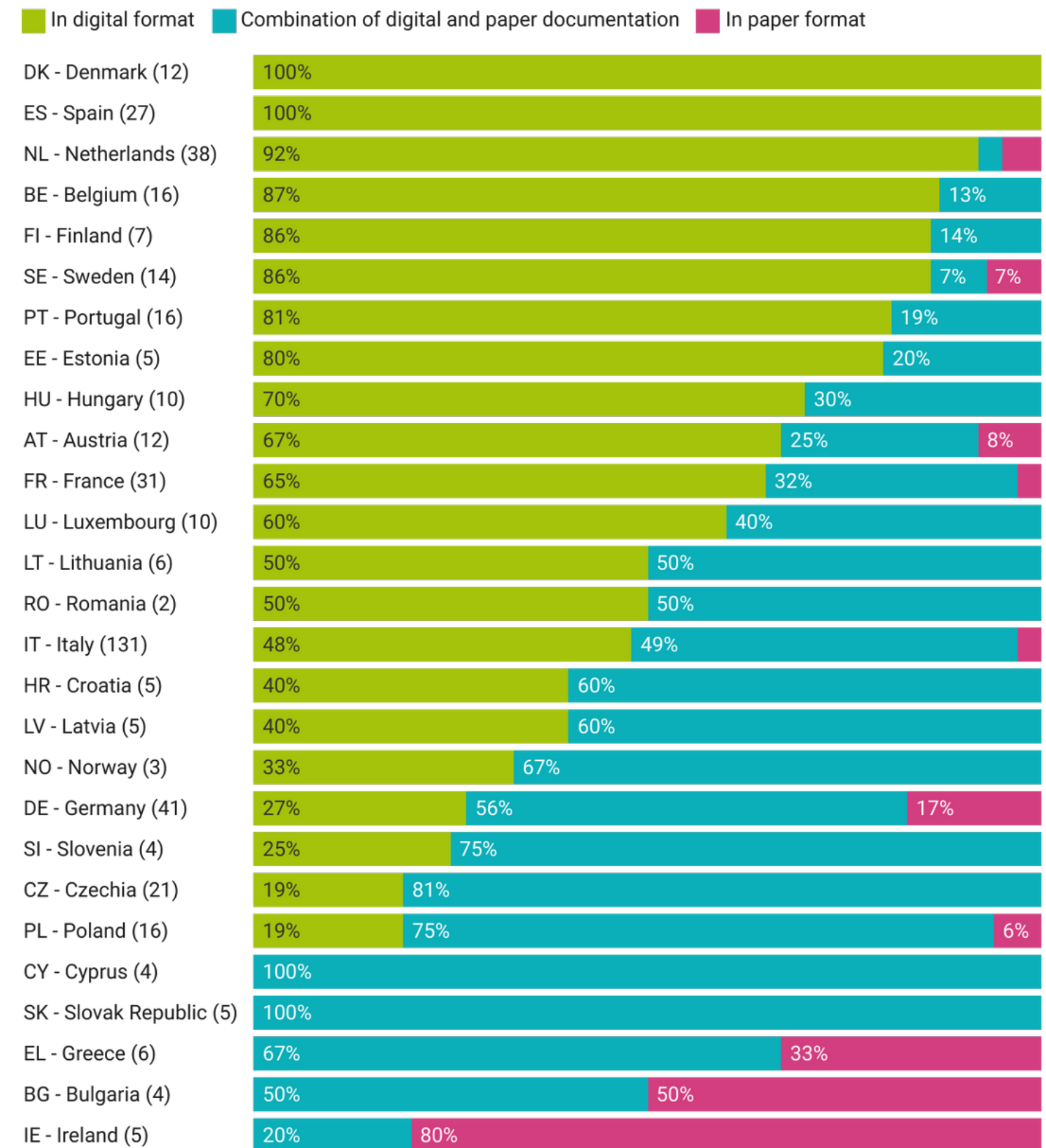
- Methods used to initially capture patient information within your institution
- Electronic health record (EHR) systems
- Coding standards employed for data description and structure
- The challenges faced in data entry and integration for RD patient data

4. Primary data capture

How is the original patient information first captured in your institution?

full digitalization not yet achieved in many countries

→ clinicians often employ a combination of digital and paper-based documentation methods.

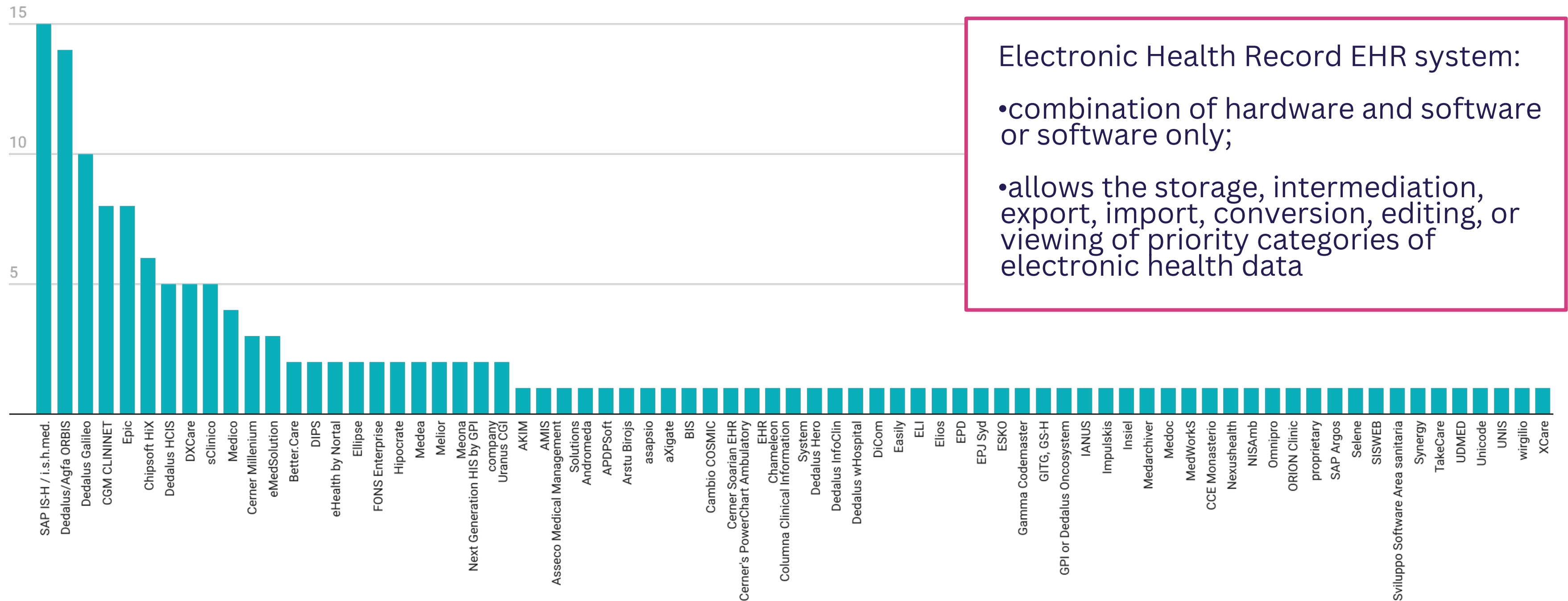


4. Primary data capture

Which Electronic Health Record systems are used?

→ 158 hospitals reported over 70 systems

→ partly different systems are used for in and out patients



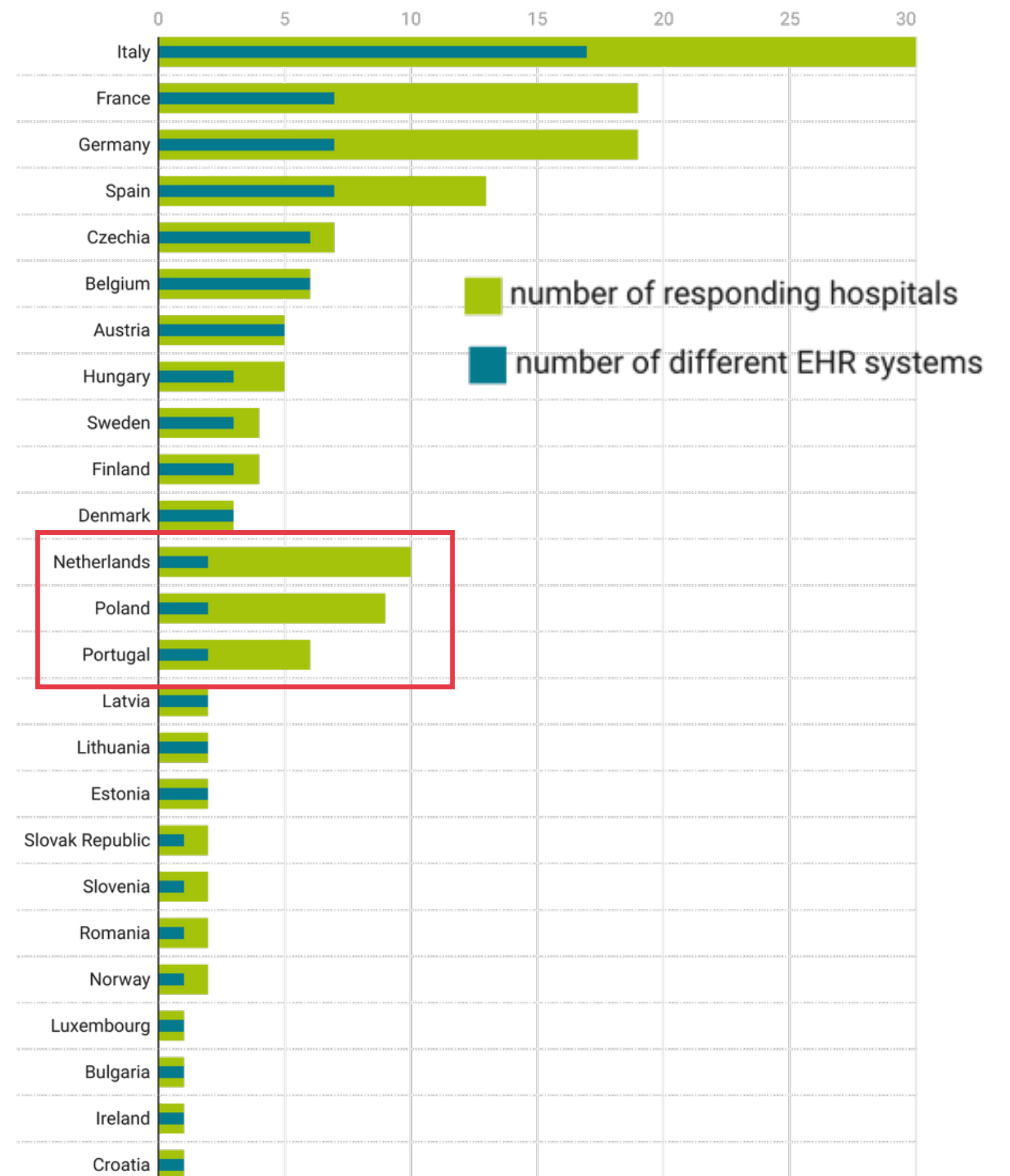
4. Primary data capture

Which Electronic Health Record systems are used?

→ Fragmentation of EHR systems across ERN hosting hospitals

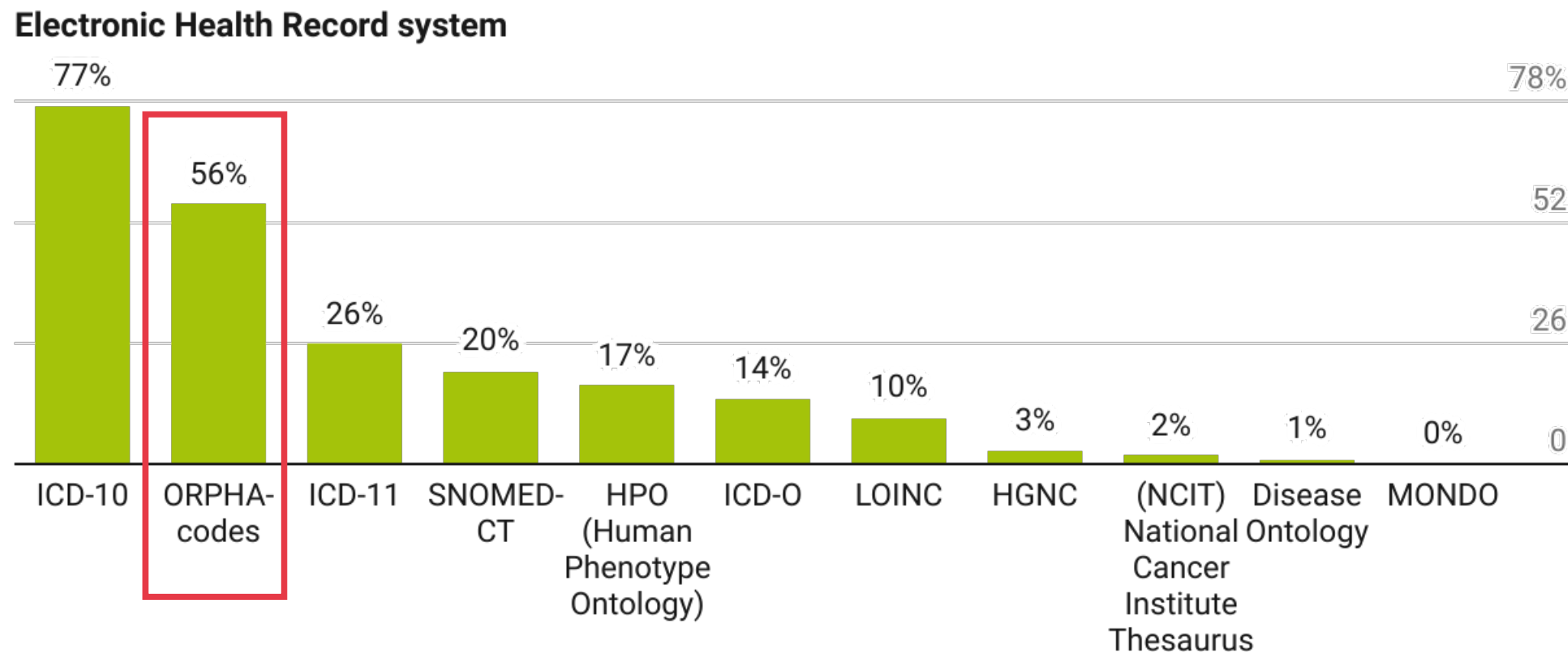
Barriers

- Lack of digitalization
- Fragmentation of EHR systems highlighting the need for system independent solutions



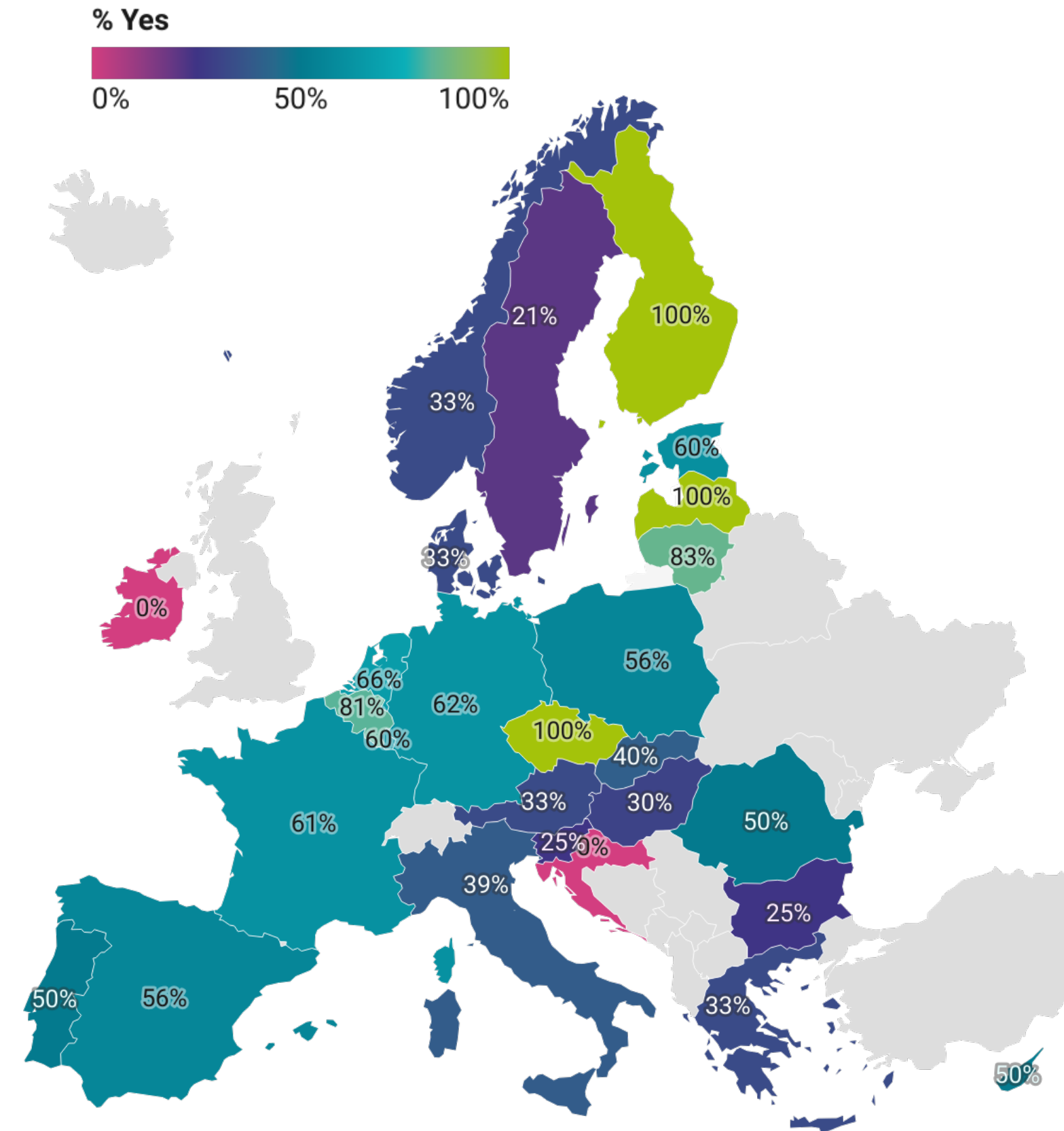
4. Primary data capture

Which coding standards do you use in your EHR systems applicable in your hospital for clinical data, diagnostic, genomics, and lab results?



- Lack of awareness of coding practices
- Misunderstanding of the question?
- Orphacodes are not sufficiently implemented

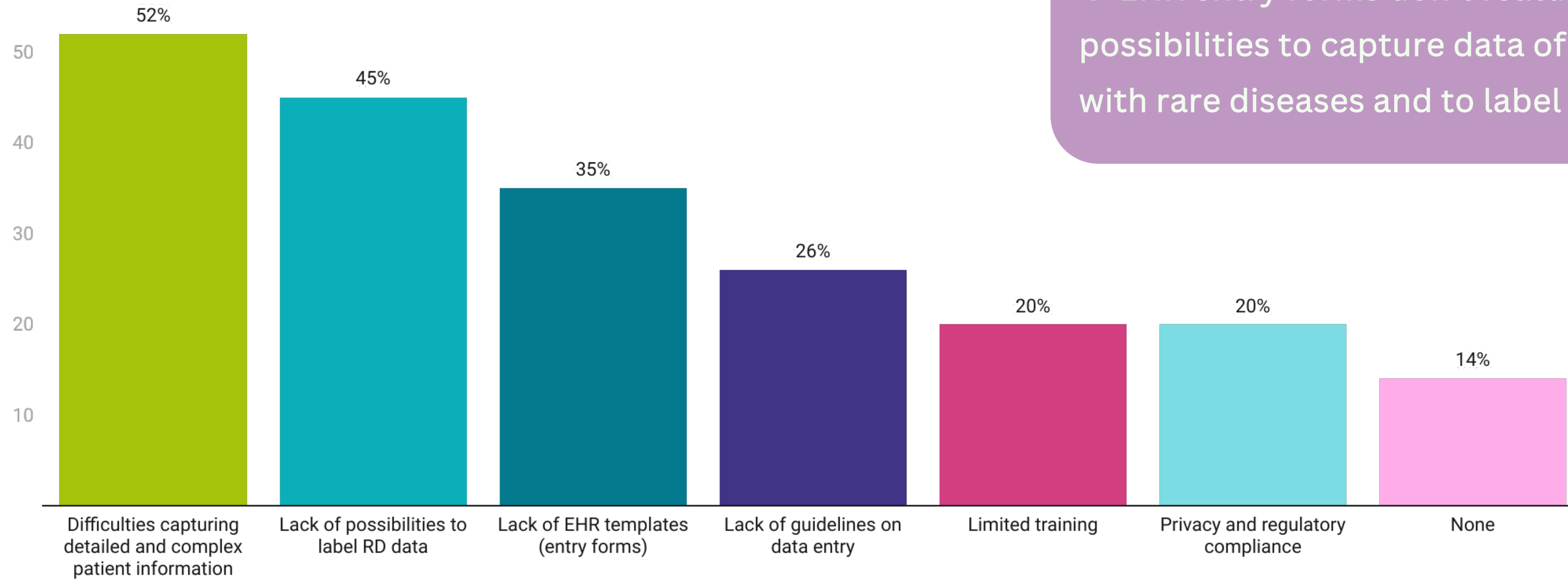
Are you using Orphacodes in your institution?



Response from 457 clinicians

4. Primary data capture

What challenges do you encounter regarding entering RD patient data into EHR systems?



Barrier:

→ EHR entry forms don't feature sufficient possibilities to capture data of patients with rare diseases and to label it as such

Response from 397 clinicians

4. Primary data capture

Barriers

- Lack of digitalization
- Heterogenous systems highlighting the need for system independent solutions
- Lack of awareness of coding practices
- Current EHR systems lack possibilities to report and label RD case sufficiently

Solutions

- Training and raising awareness of coding standards
- Implementation of Orphacodes
- Increase structured data capture
- Long term: The EHDS regulation established requirements for EHR systems and standardize their components
→ requirements for RD relevant data elements needed!

4. Primary data capture

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Your opinion matters!



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5. Data flow



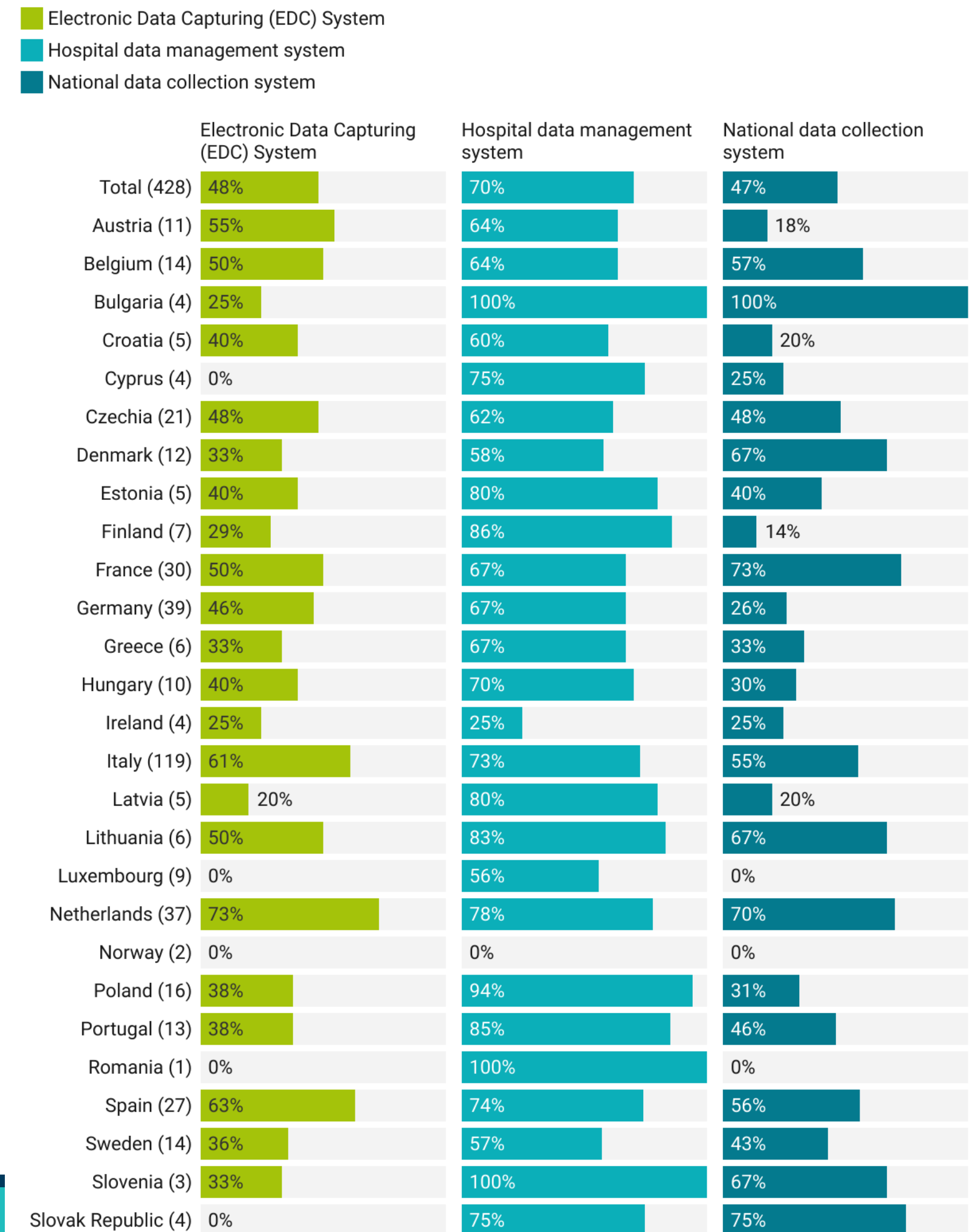
- Existing systems that data flows from and into
- Automation of data transfer across systems
- Usage of vocabularies across different systems

5. Data flow

What data management systems are in place for data to flow in and out until it reaches an ERN registry?

Definitions:

- Electronic Data Capturing system:** system designed to collect clinical data in Electronic format, mainly used for clinical studies/trials (e.g., RedCap)
- Hospital Data Management system:** system enabling hospitals to manage information and data related to all aspects of healthcare – processes, providers, patients, and more (e.g., Hospital data warehouse)
- National Data Collection System:** structured system used to gather, manage, and analyze data on a national level, often involving various data sources to monitor and report on specific issues

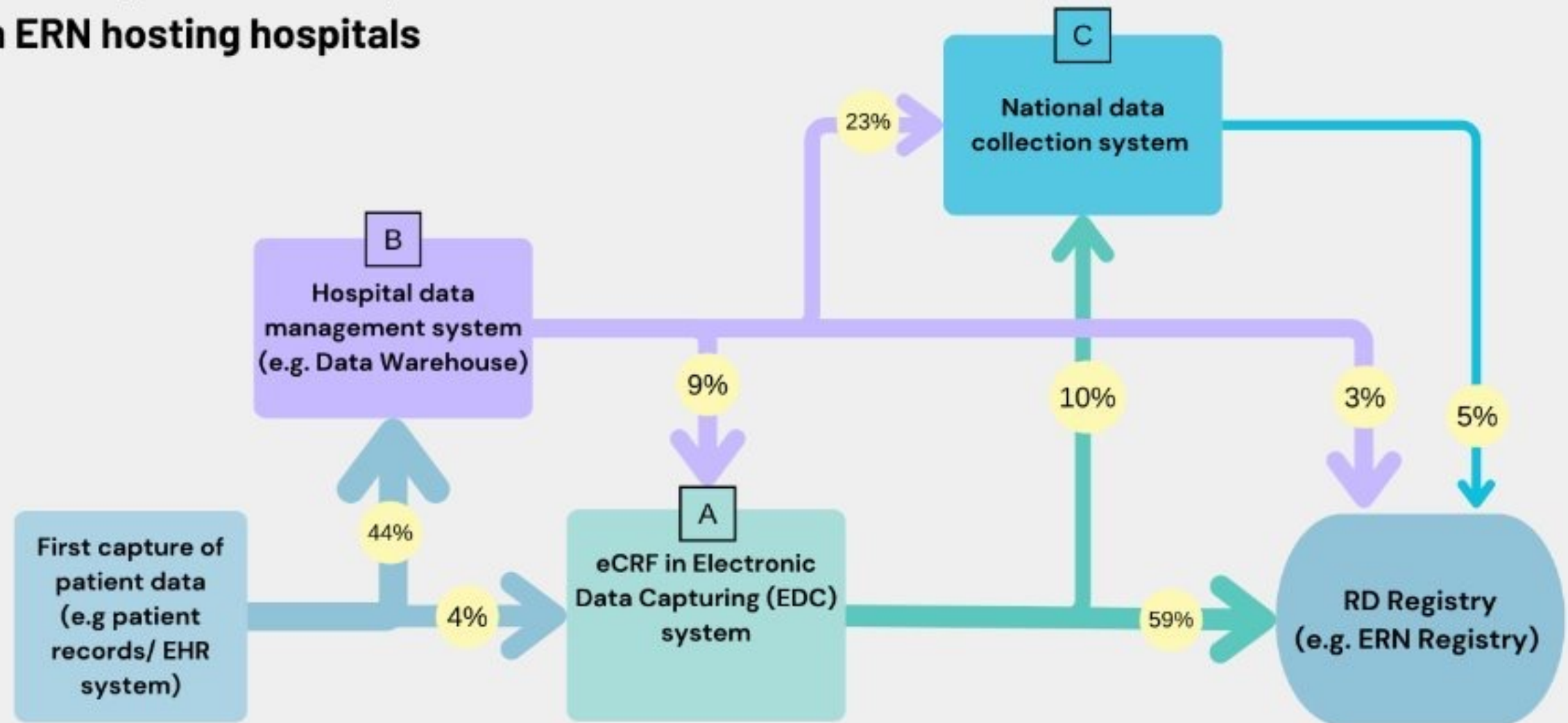


5. Data flow

Is this data transfer automated?

- Data transfer for secondary use heavily relies on human intervention;

Roadmap of data flow possibilities in ERN hosting hospitals



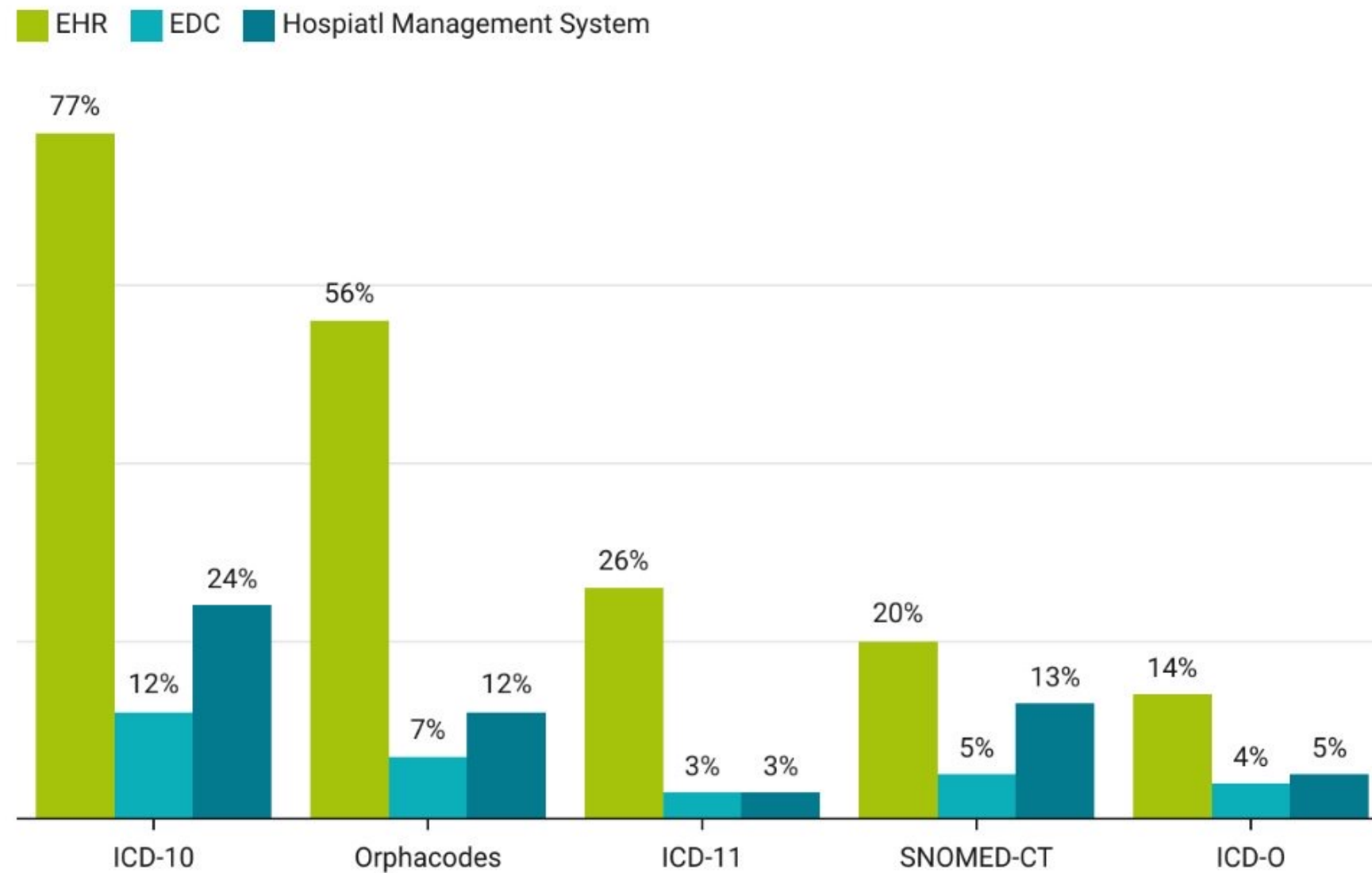
→ **Arrows** represent the likelihood of the pathway's existence (the thicker the arrow, the more people reported its existence).

● **Circles** represent the percentage of participants who report that the pathway between one station and another is automatic.

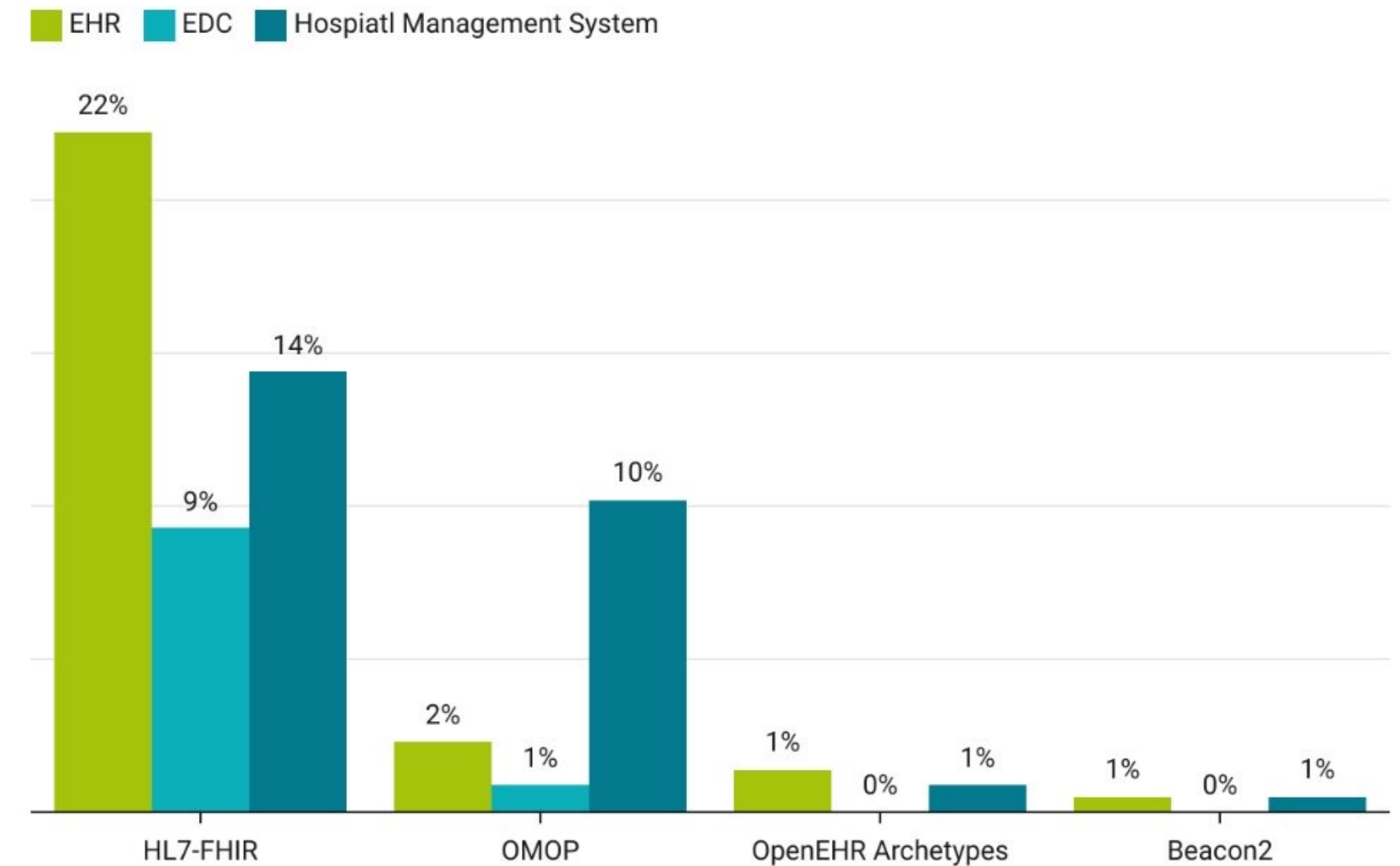
5. Data flow

Which standardized vocabularies are used across different systems?

Coding standards used for clinical data, diagnostics, genomics, and lab results?



Exchange standards used in local Data Management Systems?



5. Data flow

Barriers

- Low automation grade of data transfer across different systems
- Low awareness of data flow/data architecture
- Heterogenous adoption of standardized vocabularies across different systems

Solutions

- XpandH projects already exploring automated extraction of EHR data.
- Policy guidelines are needed to promote harmonizing of data flows, coding standards and data model
- Training health staff on correct procedures for accessing patient data
- Centralized data location for the purposes of data sharing

5. Data flow

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Your opinion matters!



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6. RD Registries

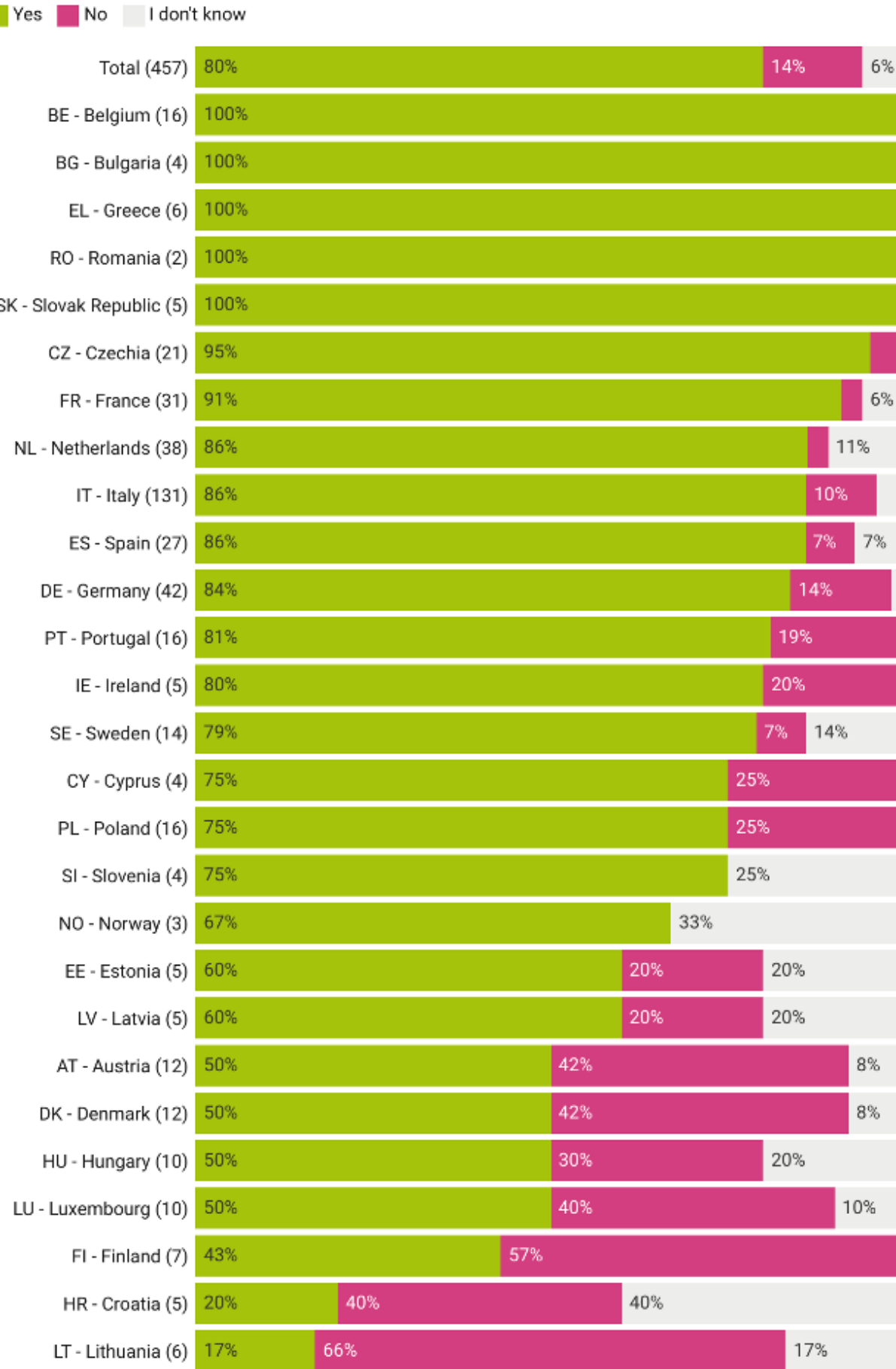


- Registry participation in ERN hosting hospitals
- Challenges
- Workload for registry operations

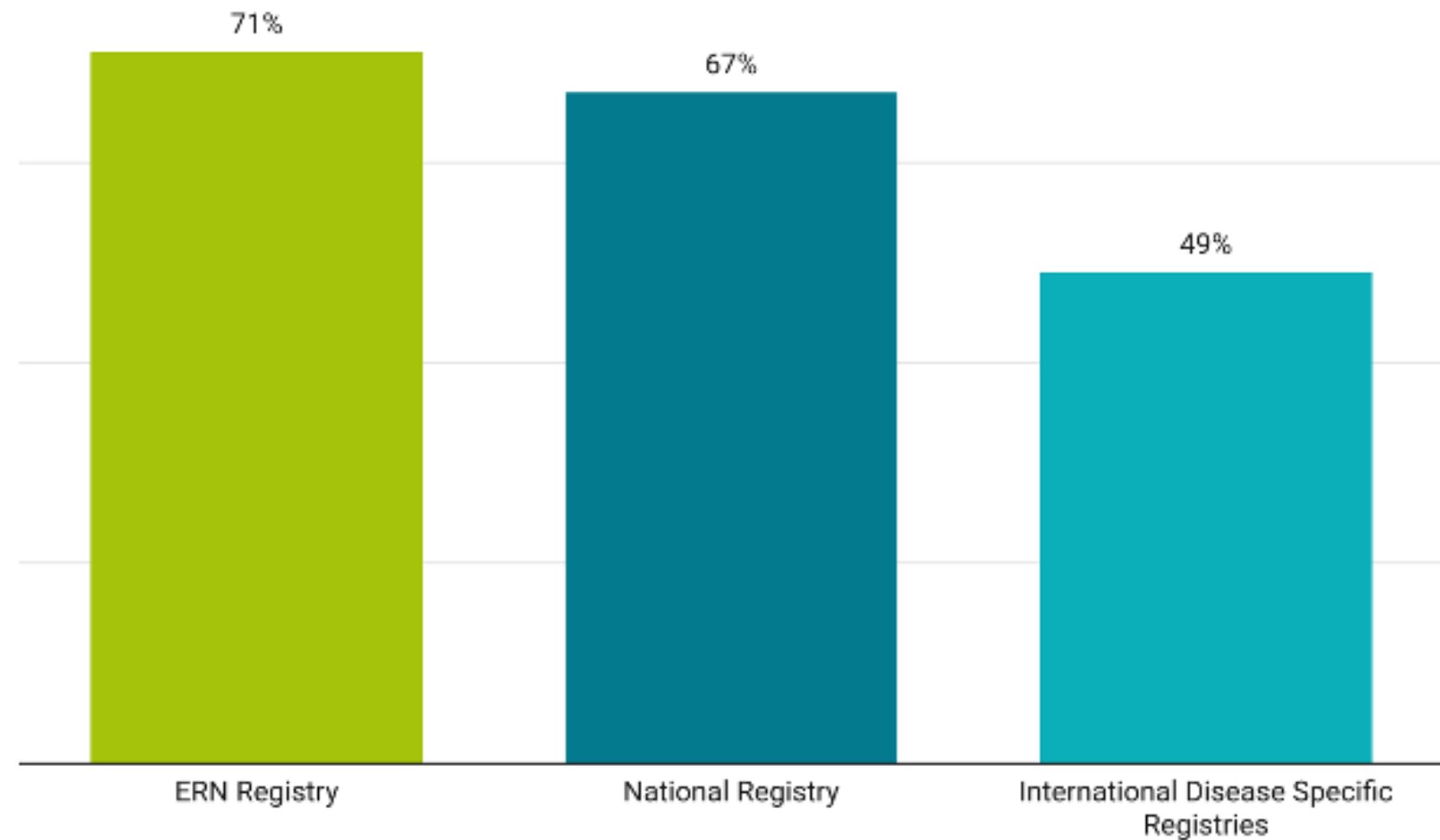
6. Registries

← Does your centre/unit contribute to one or several RD registries?

If yes, specify the type(s) of RD registries your centre/unit contributes to:



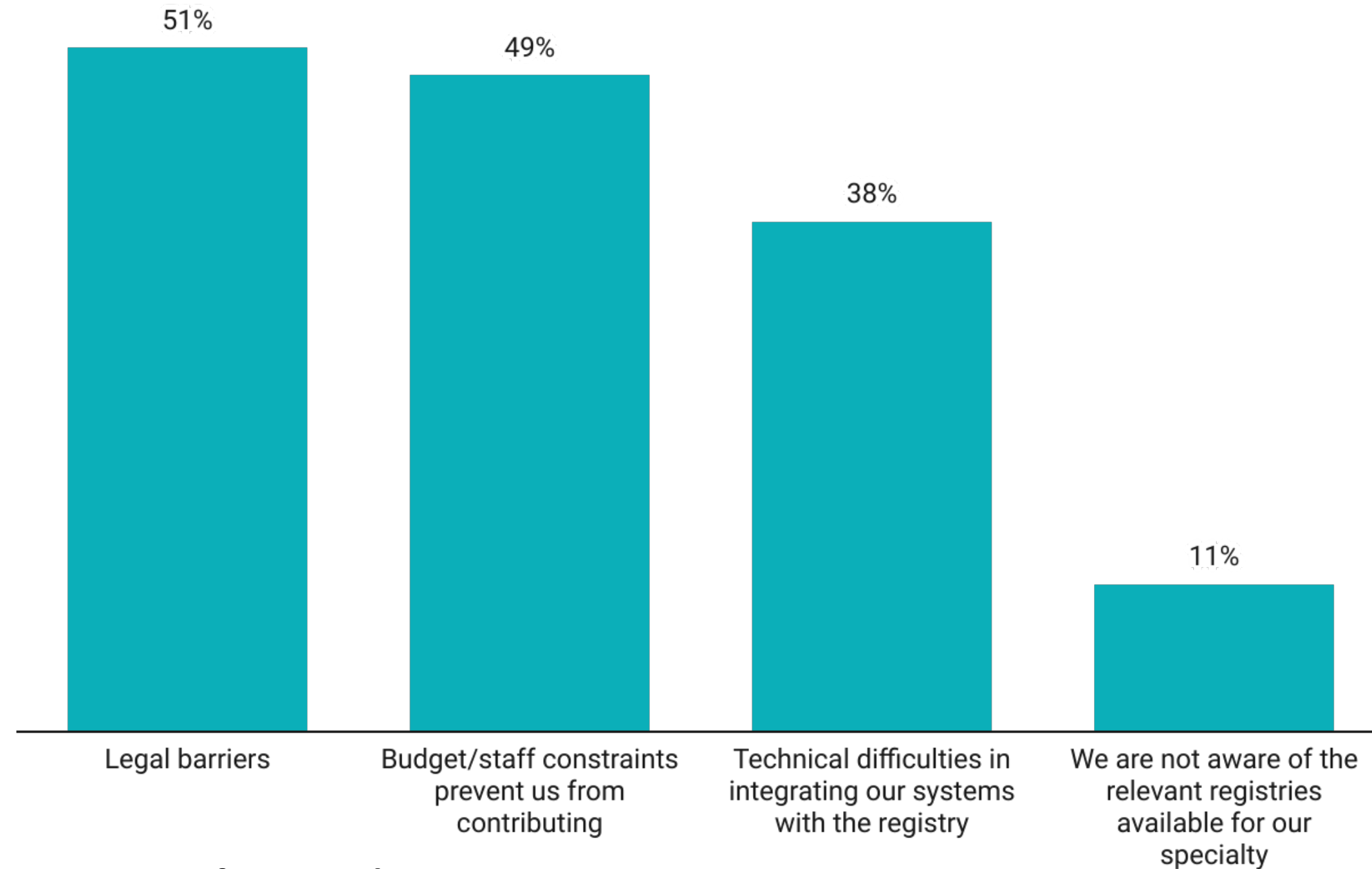
Response from 457 clinicians



Response from 366 clinicians

6. Registries

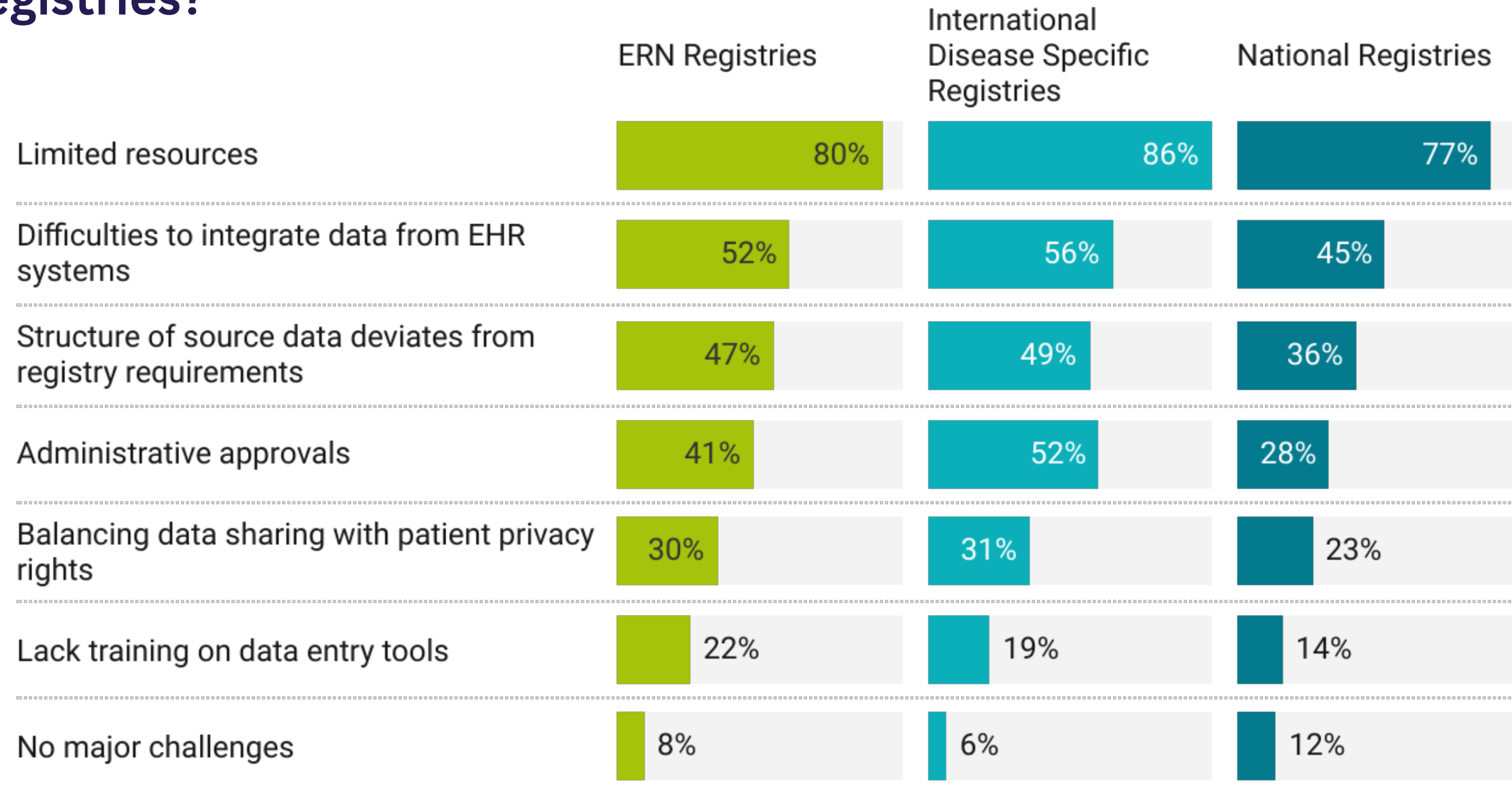
What are the main reason(s) why your hospital/unit does not contribute to a RD registry:



Response from 63 clinicians

6. Registries

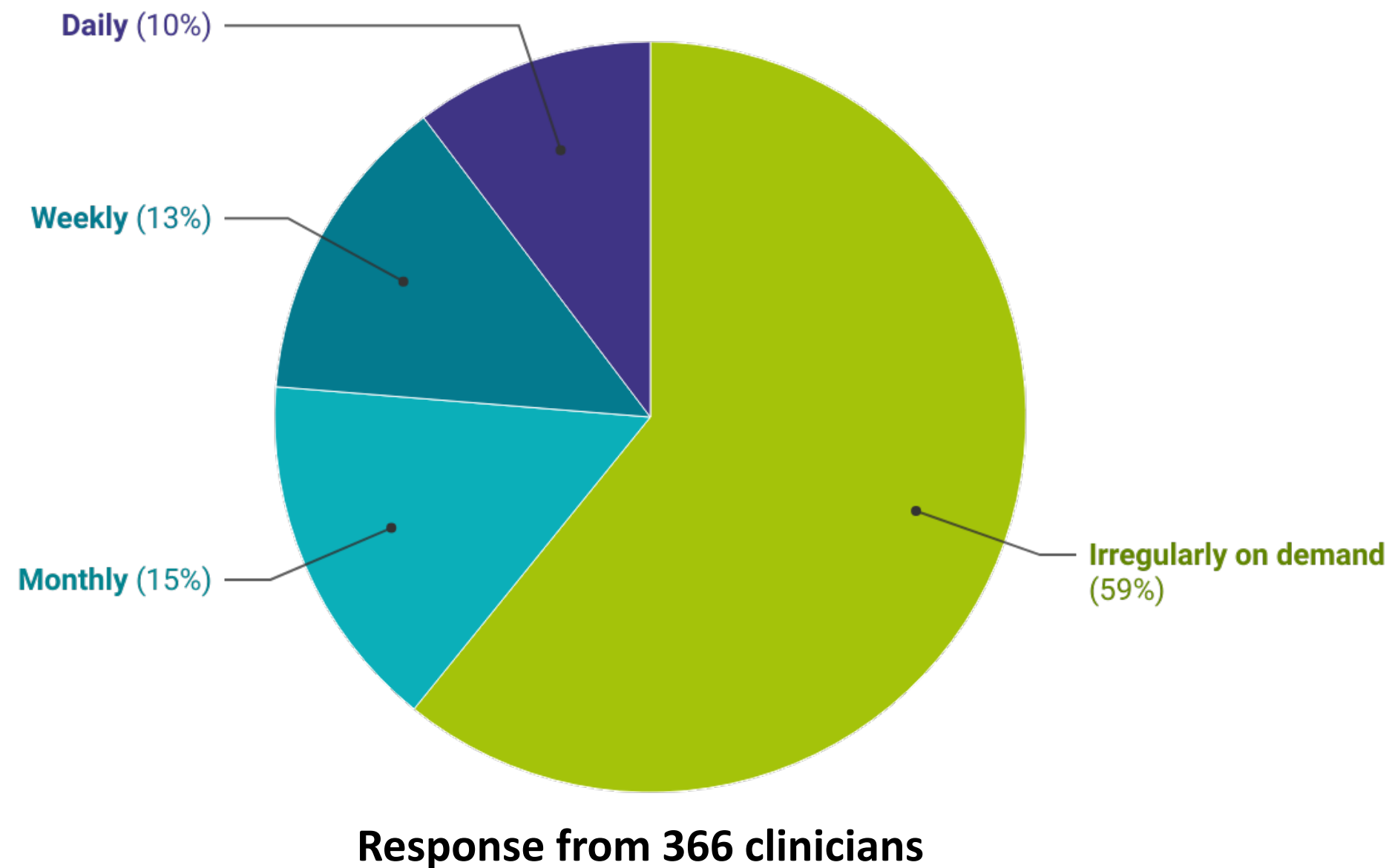
Which of the following challenges do you encounter while entering data into one of the following registries?



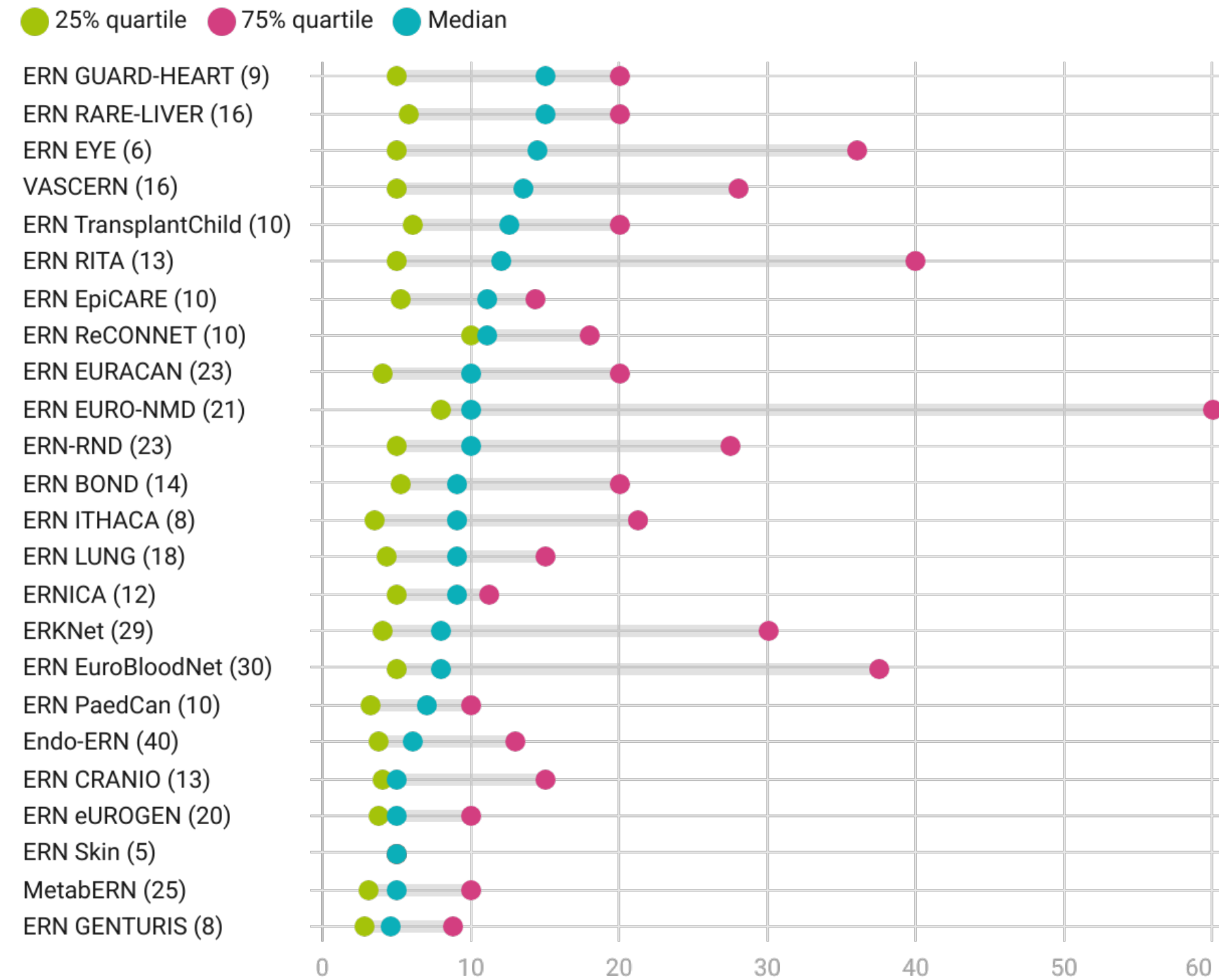
Response from 266 clinicians

6. Registries

How often is data entered into the registry?



How many working hours per month are spent on average for manual data entry?



6. Registries

Barriers

- Legal barriers for registry participation
- Limited resources for registry operations
- Lack of automation

Solutions

- ERN Legal advisor for registry operations
- Option of staff sharing for registry entries
- Moderate staff time required (10 h p. month per registry)
- Central or local support for manual entries
 - by ERNs/national authorities/hospitals?
- Long term: Harmonized digital solutions and automated data transfer
 - Improved digitalization will reduce resource burden

6. Registries

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Your opinion matters!



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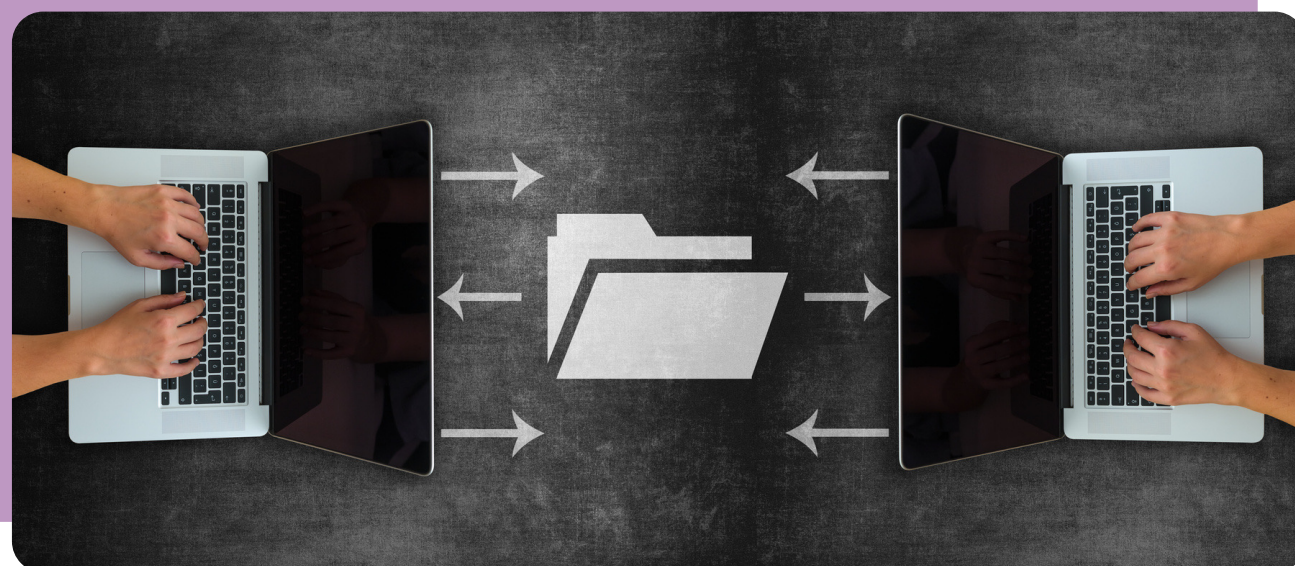


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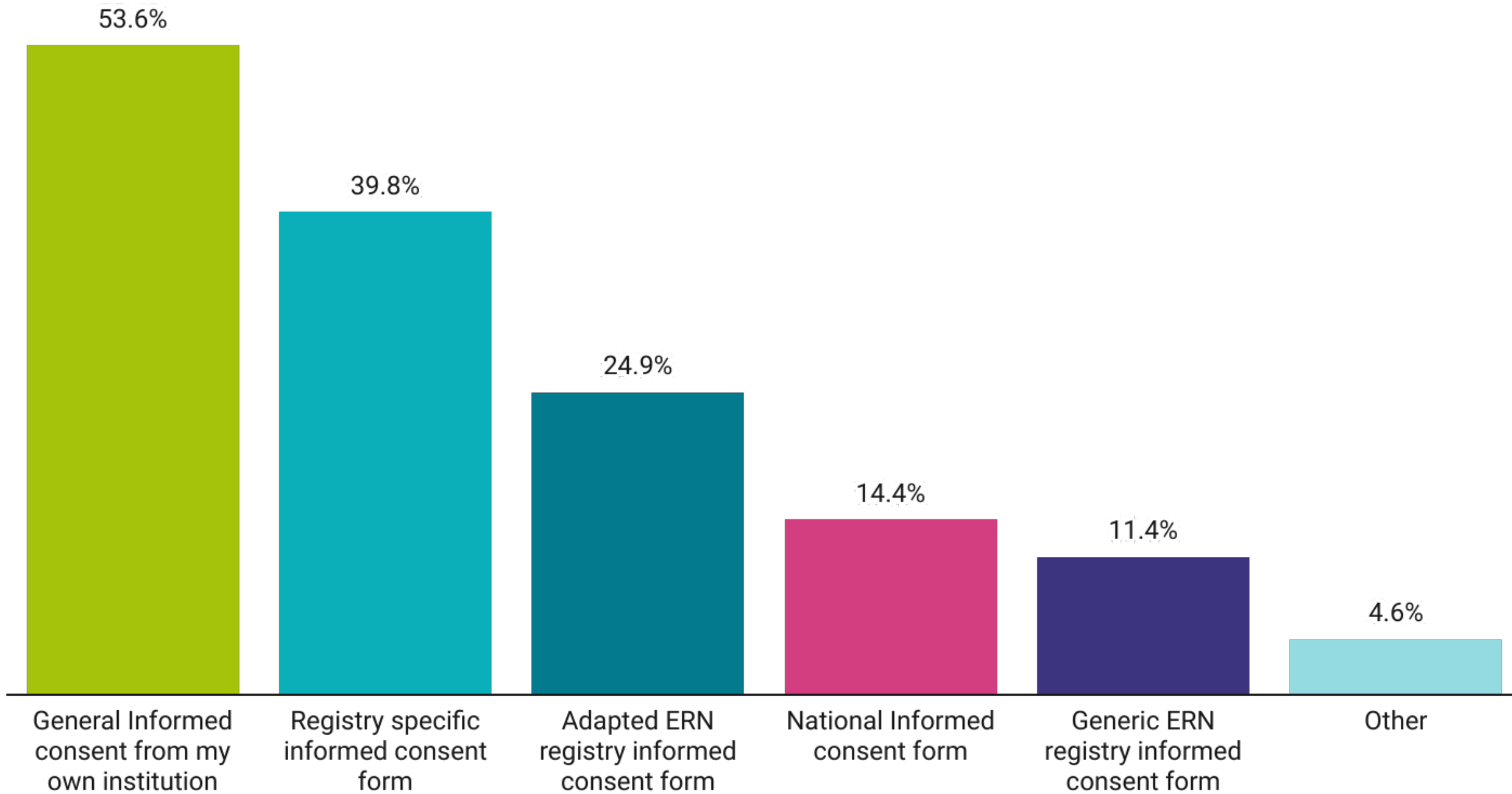
7. Data sharing & FAIR Principles



- Informed consent forms
- Digitalization of informed consent
- Awareness of data FAIR principles

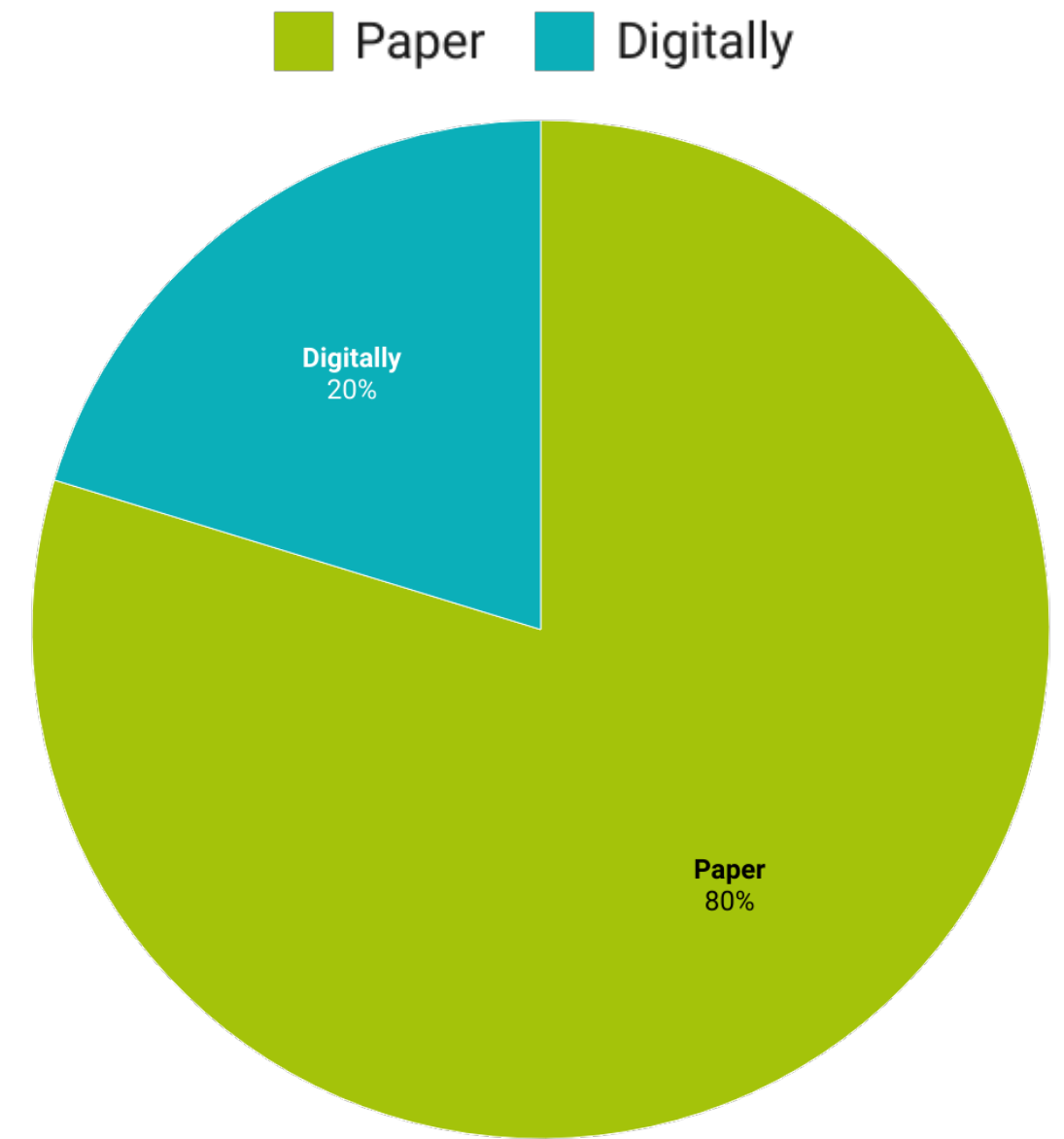
7. Data Sharing

Which of the following templates do you use to capture informed consent?



Response of 393 clinicians

How is informed consent captured?

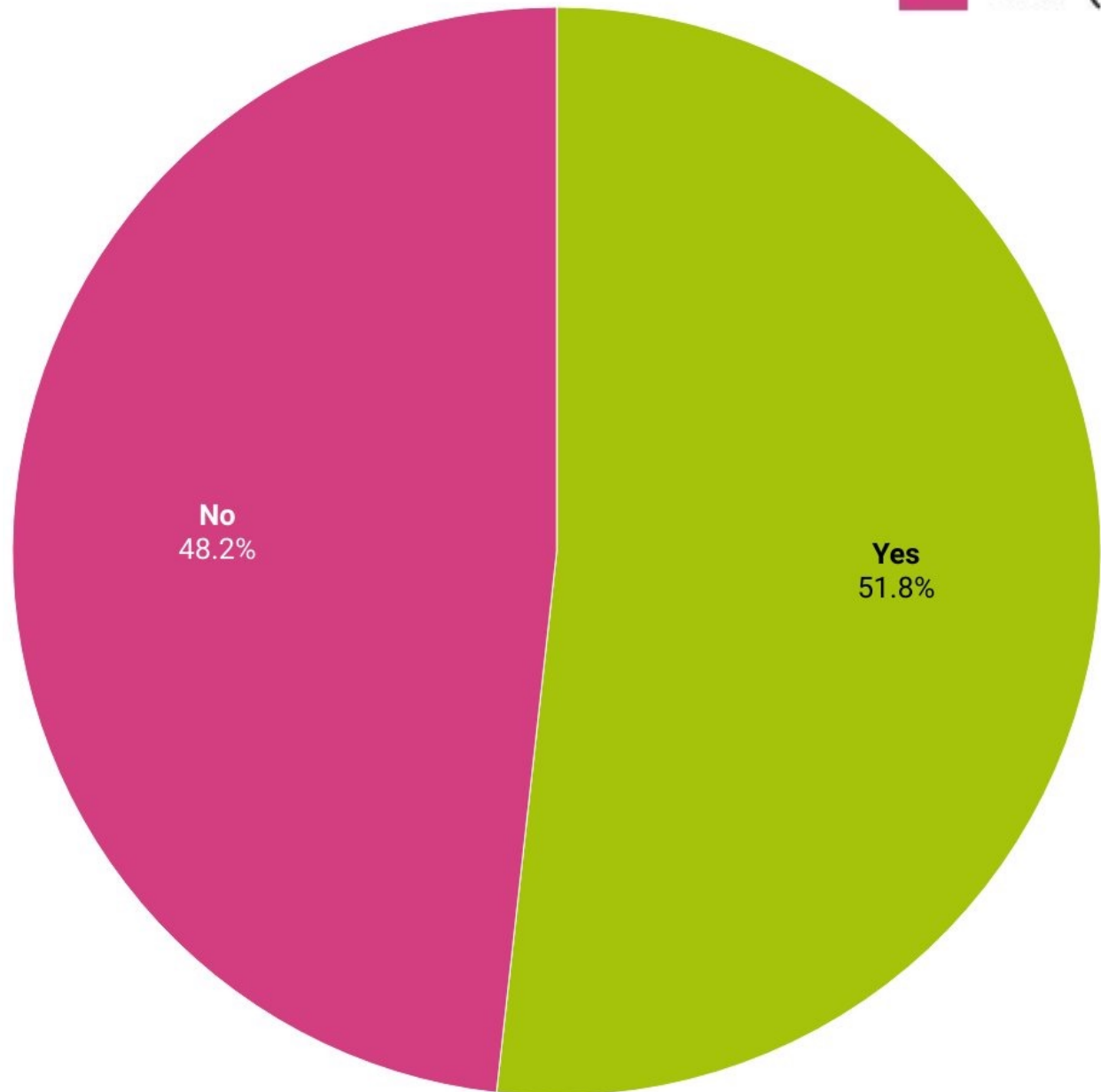


Response of 400 clinicians

7. Data Sharing

Is there a section in the EHR that displays whether a patient has been consented or not?

■ Yes (51.8%)
■ No (48.2%)



Barriers:

- Many templates used simultaneously



- Often consent is captured in paper format (80% of respondents)



- It's not always guaranteed that the EHR record includes information about whether the patient consented or to which purposes can their data be reused

6. Data sharing

Barriers

- Many templates used simultaneously
- Often consent is captured in paper format
- EHR records may not always indicate patient consent or data reuse permissions

Solutions

- EHDS will develop a HealthData@EU infrastructure to facilitate cross-border data access
- Manage Informed consent via patient portals
- Promote implementation of FAIR principles for sharing data more efficiently

7. Data Sharing

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THANK YOU!