

ERICA GENERAL ASSEMBLY USE CASE ERN REGISTRIES

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ERN-RND Registry



1. ERN Registries
2. Use case: RD patients without (molecularly) confirmed diagnosis
3. Next steps

Main goal: **Improving healthcare by using registry data**

- Call 2023: This action will ensure continuity for ERNs's coordination and operation for the period 2023- 2027 and will support the provision of specialised healthcare for rare diseases [...] and **will support ERNs towards their integration into national health systems and future sustainability.**

➤ **Call 2023:** all ERNs have applied and have a WP on ERN registries

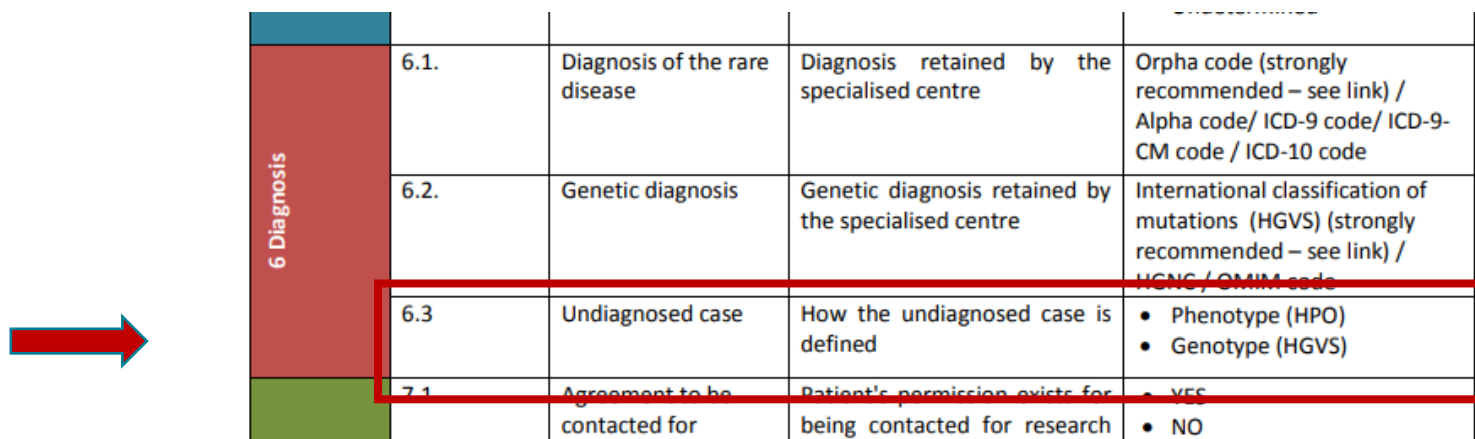
2. Objectives, themes and priorities:

- **Expand the use** of registries
 - The health data collected in the ERN registries could be **made available** - in an aggregated manner and to improve for example knowledge on the prevalence of specific rare diseases - to researchers, public authorities, industry and other stakeholders
- ➔ This should result in the ERNs reaching a new stage of development as the next four years must propel forward and consolidate the development of key technical areas of work, including:...**full deployment and development of registries, making them research-ready...**

USE CASE: RD PATIENTS WITHOUT (MOLECULARLY) CONFIRMED DIAGNOSIS

➤ Expectations regarding data collection and analysis capabilities:

- **Expectation 1:** all ERN registries **collect data** of all patients seen at the ERN members
- **Expectation 2:** ERN registry **data collection includes patients without confirmed diagnosis** that are seen at the ERN members and that fall into the respective phenotypic spectrum



6 Diagnosis	6.1.	Diagnosis of the rare disease	Diagnosis retained by the specialised centre	Orpha code (strongly recommended – see link) / Alpha code/ ICD-9 code/ ICD-9-CM code / ICD-10 code
	6.2.	Genetic diagnosis	Genetic diagnosis retained by the specialised centre	International classification of mutations (HGVS) (strongly recommended – see link) / HGNC / OMIM code
	6.3	Undiagnosed case	How the undiagnosed case is defined	<ul style="list-style-type: none"> • Phenotype (HPO) • Genotype (HGVS)
	7.1	Agreement to be contacted for	Patient's permission exists for being contacted for research	<ul style="list-style-type: none"> • YES • NO

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- **Expectation 4:** all ERN registries are **able to share these analysis results**

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Goal: Description of European cohort of RD patients without (confirmed) diagnosis

Instrument: Establishing a use case

- **Use case:** each ERN registry contributes aggregated data once per calendar year (so for example: 1 April 2024 for 2023)

- **European cohort** of RD patients without (molecularly) confirmed diagnosis

- **Analysis** of the aggregated data
 - number of patient entries per year
 - number of patients w/o confirmed diagnosis
 - Total
 - Break downs: Registry, Disease groups/phenotypes (HPO), Countries



- ✓ **gives an overview** of numbers per registry and disease groups (potentially broken down to countries)

- ✓ **Helps to analyse** differences in patient care and **to develop** further action points

- ✓ **Might serve as outcome indicator for describing the status of RD diagnosis**

1. Establish a cross-ERN registry WG

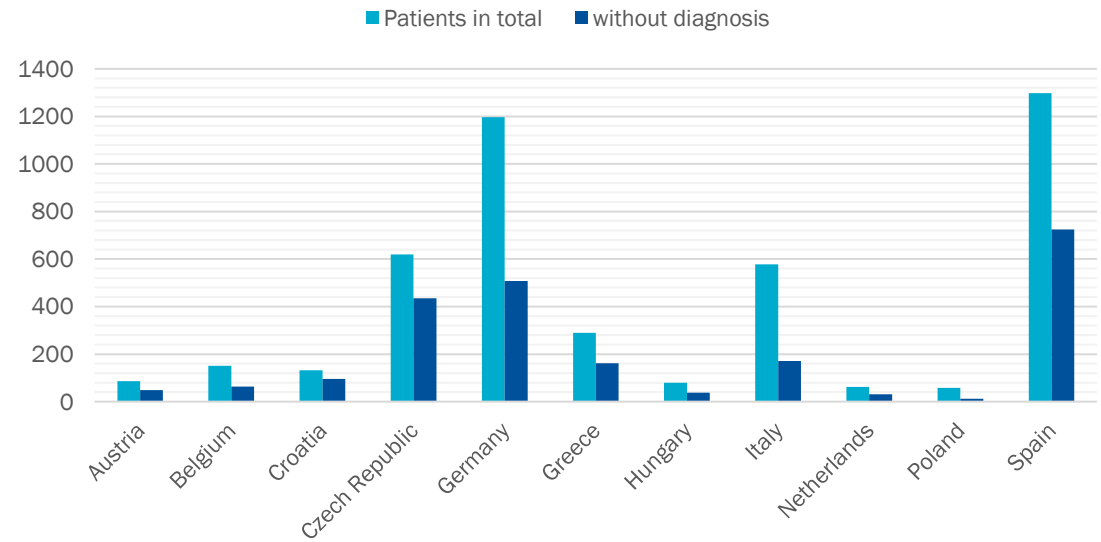
➤ First potential activities

- Map the status of ERN registries regarding the data collection of diagnostically not confirmed RD patients
- Develop an interoperable template for submission of annual aggregated datasets
 - which datapoints shall be submitted?
 - using which tool? (excel, virtual platform?)
- Address administration questions
 - e.g. applications to Data Access Committees

2. Implement a pilot

- Collect and analyse data
 - of a smallish number of registries which already have unconfirmed cases
- Report on pilot results

ERN RND: Patients without diagnosis



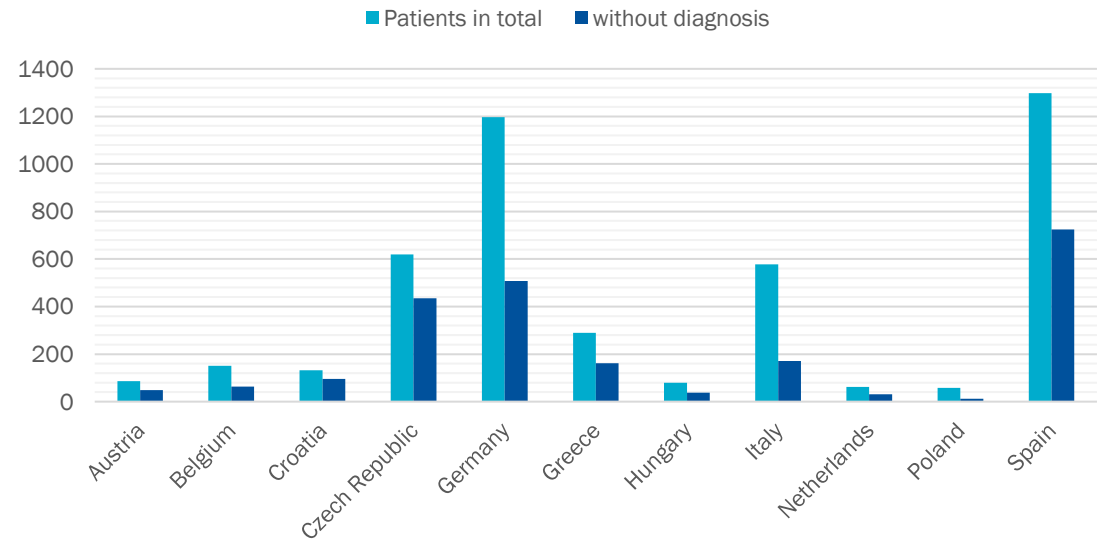
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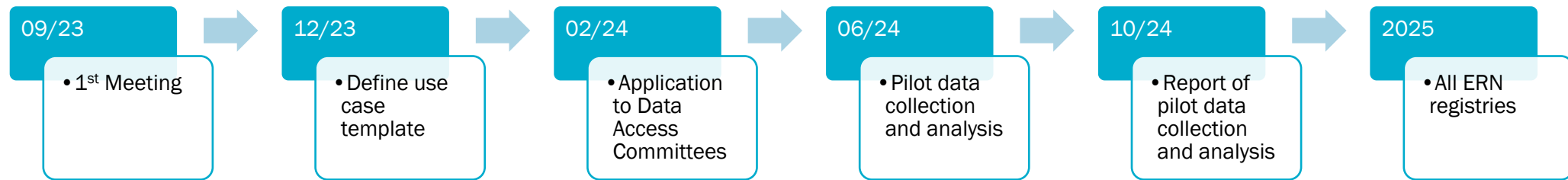
3. Scale use case up to all ERN registries

- Collect and analyse data on annual basis
- Annual report

ERN RND: Patients without diagnosis



Timeline:



NOW: Discussion

After meeting: sign up for working group by contacting me:

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**THANKS FOR YOUR
ATTENTION!**

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