



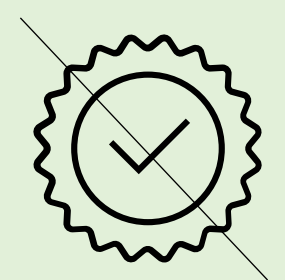
# Definitions of the concept rare diseases: insights from a grey literature review

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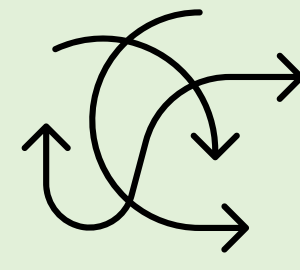
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## Background



**Absence** of commonly accepted **definition** of rare diseases (RD)



**Complicates application** in health policy & decision-making aiming at addressing unmet needs for patients with RD

## Objective

To explore **RD definitions** used in the **European** healthcare policy domain:

Identify **recurring** elements

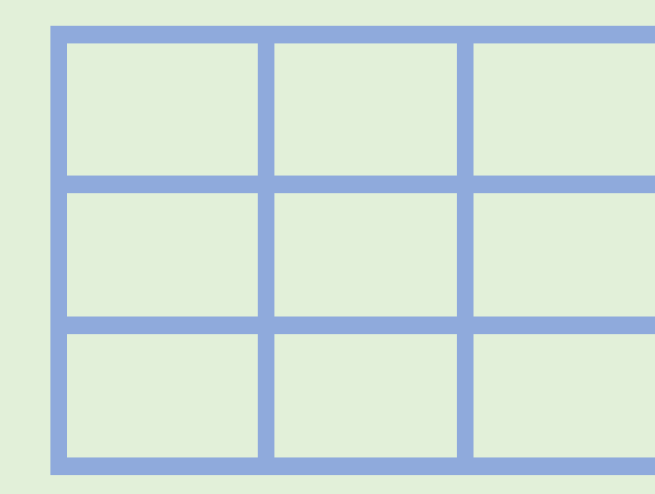
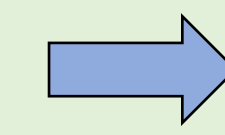
Assess **differences** between relevant stakeholder groups

## Methods

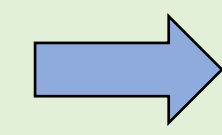
**Grey literature review** (May – August 2023)



Screening of relevant **stakeholders' websites**



**Data extraction** in Excel sheet



**Comparative analysis**

4 **stakeholder** groups, at the **European & national member state** level (n=136):

HTA bodies & payers (n=38)

RD patient organisations (n=32)

Regulatory agencies or government (n=33)

Pharmaceutical industry trade organisations (n=33)

## Recurring elements in RD definitions

Two **main elements** are identified:

### 1. The prevalence of the disease

When prevalence was part of the definition, it corresponded for 93.9% to **≤ 5 in 10 000 people\*** (few exceptions).

### 2. Qualitative aspects

Four categories of criteria could be distinguished:

- **Disease characteristics** (e.g., progressive, life-threatening\*, chronically debilitating\*)
- **Knowledge gaps** (e.g., natural progression unknown, no available treatment known\*)
- **Disease aetiology** (e.g., genetic origin)
- **Burden on patients** (e.g., affecting QoL, severe burden of illness)

Not all stakeholders use both elements in their definition.

RD and orphan disease are often used interchangeably.

\*These aspects are covered by the EC regulation 141/2000 on Orphan medicinal products.

## Proportion of stakeholders reporting RD definition

Stakeholder	Proportion (%)
HTA bodies/payers	18.4
Regulatory agencies or government	30.3
Industry trade organisations	33.3
RD patient organisations	75.0
Total	38.2

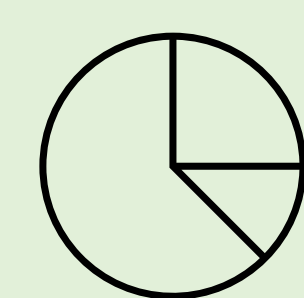
**Patient organisations** were the **most** and **HTA bodies** the **least consistent** in reporting a RD definition.

## Number of stakeholder RD definitions reporting the different recurring elements

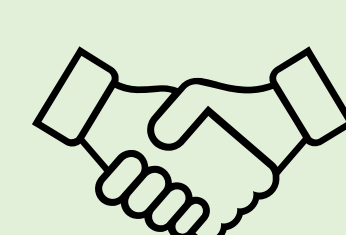
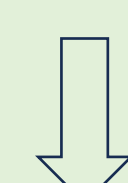
	Prevalence (≤ 5 per 10 000)	Qualitative criteria	Prevalence + other qualitative criteria	In line with Orphan Regulation
HTA bodies/payers	3	1	2	1
Regulatory agencies or government	5	1	1	3
Industry trade organisations	7	1	3	/
RD patient organisations	5	/	7	12

Results show **differences** in RD definitions **across countries** and **within stakeholder** groups.

## Conclusions



A significant portion of stakeholders report on RD, however without providing a clear definition



Attempts are needed to harmonize RD definitions with standardized criteria in the EU

## Acknowledgments

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## Abbreviations

RD = Rare disease, HTA = Health Technology Assessment, EC = European Commission, QoL = Quality Of Life