

INTRODUCTION

Rare diseases (RDs) are often chronic and progressive life-threatening medical conditions that affect a low percentage of the population compared with other diseases. RDs affect approximately 6% of the worldwide population.¹ Many patients with RDs experience difficulties accessing appropriate treatment options. Globally, less than one-tenth of patients with RDs receive treatment, i.e., orphan drugs (ODs).²

Unfortunately, there is no universal definition of RDs or ODs. The varied terminology and inconsistent definitions of RDs & ODs are considered major challenges in treatment accessibility.

AIM

The aim of this study is to identify the criteria used to define RDs and ODs from both qualitative and quantitative perspectives and explore the rationale behind these criteria.

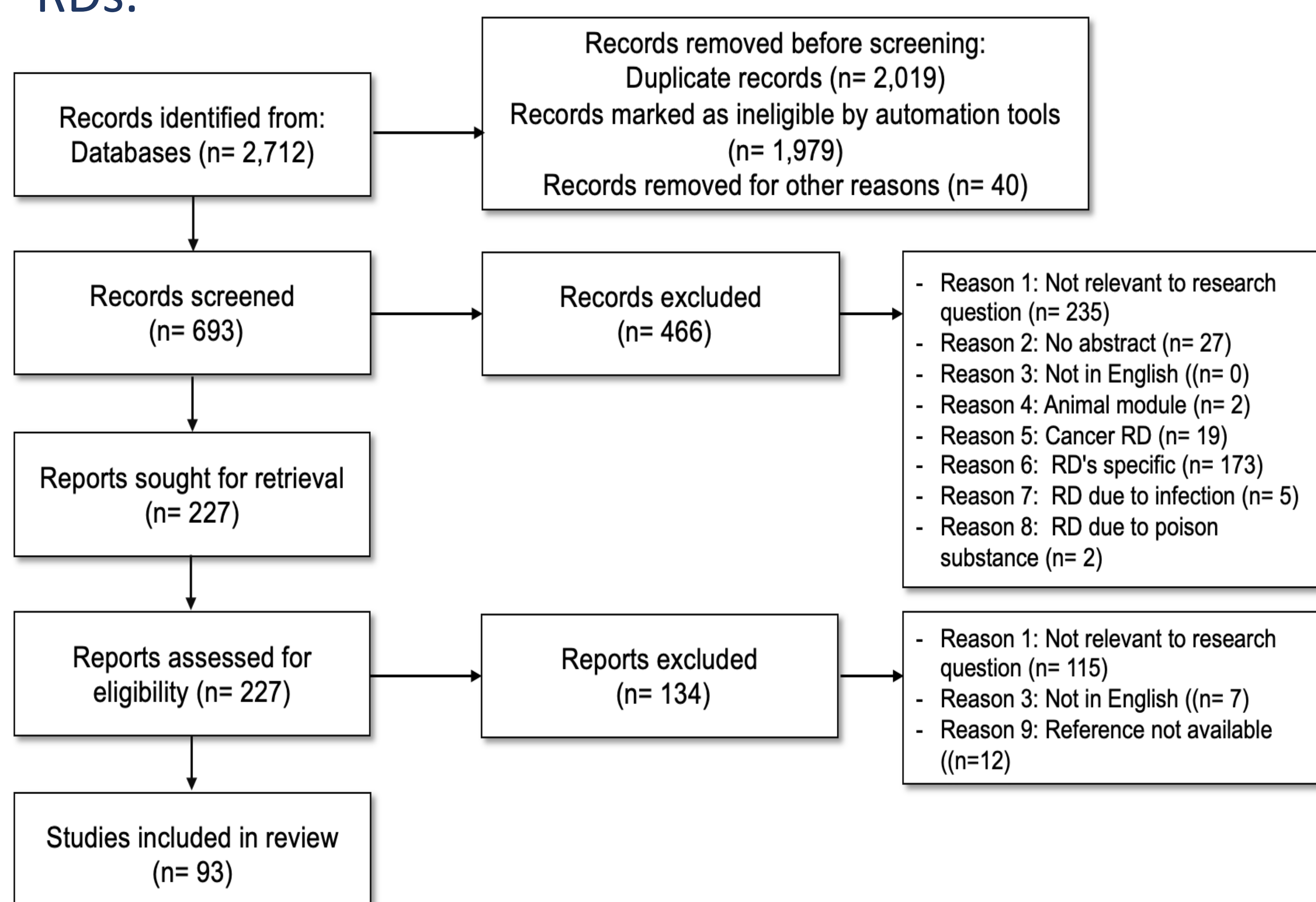
METHOD

A systematic literature review was performed in following databases: PubMed, MEDLINE, EMBASE, Scopus, Web of Science. Eligible publications were selected based on predetermined inclusion criteria. Extracted data were analysed using thematic and content analyses for qualitative descriptors, whereas quantitative data were analysed descriptively.

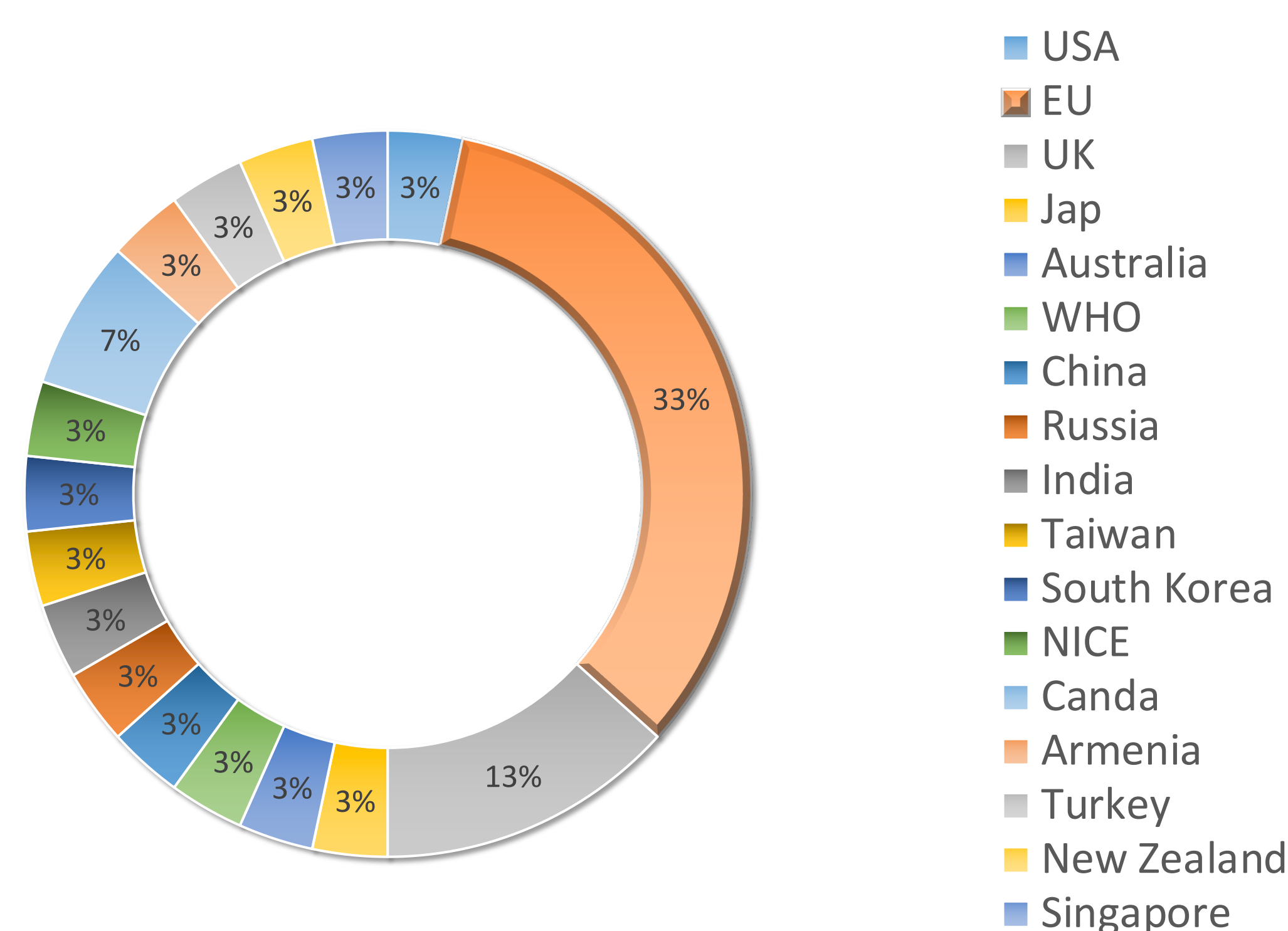
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RESULTS

- A total of 2,712 publications were identified. Of them, 93 contained relevant information about ODs and RDs.



- Only 63 (68 %) publications included at least one non repeated definition for either RDs, ultra – rare disease (URD), ODs, and ultra- orphan drugs (UOD) alone or in combination of two or more terms (*figure 1*)



- Thirteen countries were reported to have one definition for RDs and ODs, while 3 countries reported two or more definitions for RDs and ODs owing to jurisdictional variation.
- In total, 24 descriptors for RDs and 14 descriptors for ODs were identified as part of qualitative criteria.
- The other hand, 5 descriptors for RDs and 6 descriptors for ODs were identified as part of quantitative criteria.

CONCLUSION

Overall, we couldn't identify a single unified globally accepted definition for either RDs and ODs. Moreover, there were no scientific bases for all published RDs and ODs definitions. In addition, there were no consensus on the definition on different qualitative descriptors.

These facts address the important of having a widely accepted definition with scientifically sounded criteria. Since these can impact drug registration, prices for market entry and reimbursement recommendations which can affect patient access to breakthrough innovative medications.

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