

Global Insight into Rare Diseases and Orphan Drugs Definitions: A Systematic Literature Review



Ghada Mohammed Abozaid,^{1,2} Katie Kerr,² Amy McKnight,² Hussain A Al-Omar^{3,4,5}

¹Department of Pharmacy Practice, Princess Nourah bint Abdulrahman University, Riyadh, Saudi Arabia. ²Institute of Clinical Sciences B, Royal Victoria Hospital, Queen's University Belfast School of Medicine, Dentistry and Biomedical Sciences, Centre for Public Health, Belfast, UK. ³Department of Clinical Pharmacy, College of Pharmacy, King Saud University, Riyadh, Saudi Arabia. ⁴Center of Health Technology Assessment, Ministry of Health, Riyadh, Saudi Arabia. ⁵Health Technology Assessment Unit (HTAU), College of Pharmacy, King Saud University, Riyadh, Saudi Arabia

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.

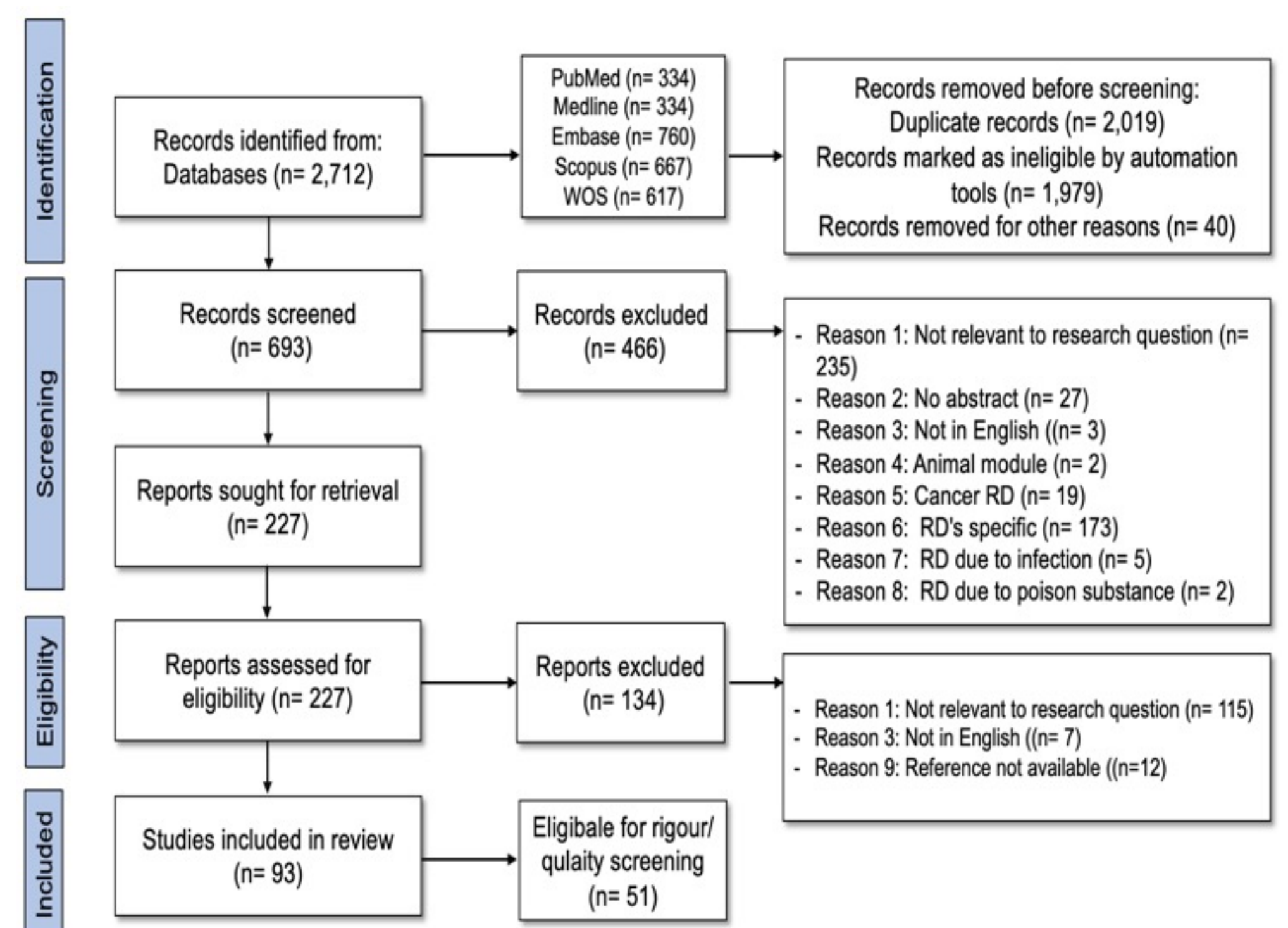
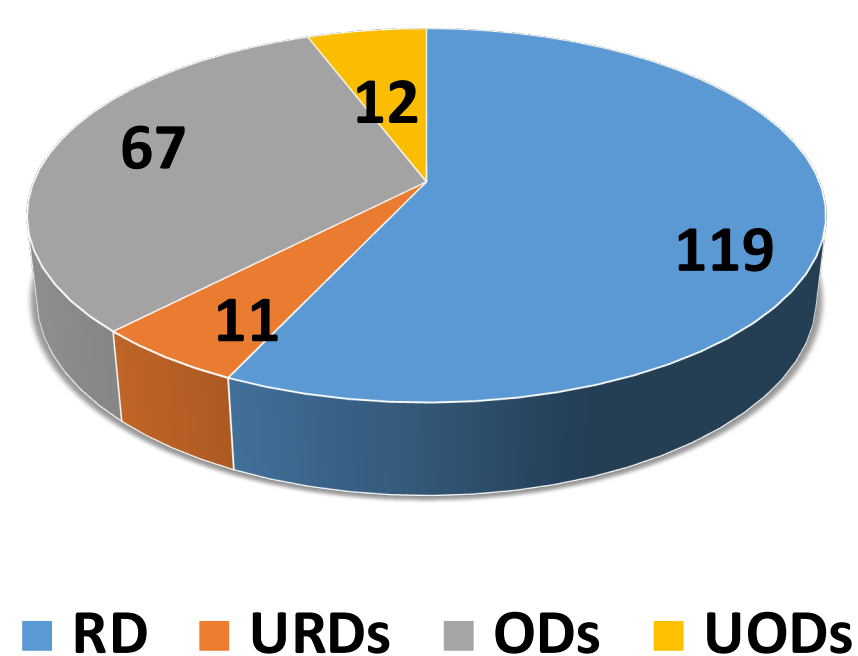
PROSPERO registration number CRD42021252701

RESULTS

• A total of 2,712 publications were identified. Of them, 93 contained relevant information about ODs and RDs. (figure 1)

• A 209 distinct definitions extracted with 209 distinct definitions extracted pertained to RDs and ODs. (figure 2)

figure 2: Frequency of Repeat definitions Retrieved from 93 articles



• In total, 35 **qualitative criteria** for RD, 37 for ODs, 7 for Ultra- rare diseases (URDs), and 11 criteria for Ultra-orphan drugs (UODs) respectively.

• The other hand, 10 criteria for RDs and 5 for ODs, 4 for URDs and 3 for UODs were identified as part of **quantitative criteria**.

CONCLUSION

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

REFERENCE

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