

Perception of Rare Diseases and Orphan Medicines

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INTRODUCTION

The European Organisation for Rare Diseases (EURORDIS) defines a rare disease as “a disease that occurs infrequently or rarely in the general population” in 2005. The definition for a rare disease varies between legislation and policies of different countries and to date there is no standard definition for rare diseases worldwide.¹

AIMS

1. To put forward a new definition for rare diseases and orphan medicines
2. To analyse the level of experience and awareness with regards to rare diseases and orphan medicines

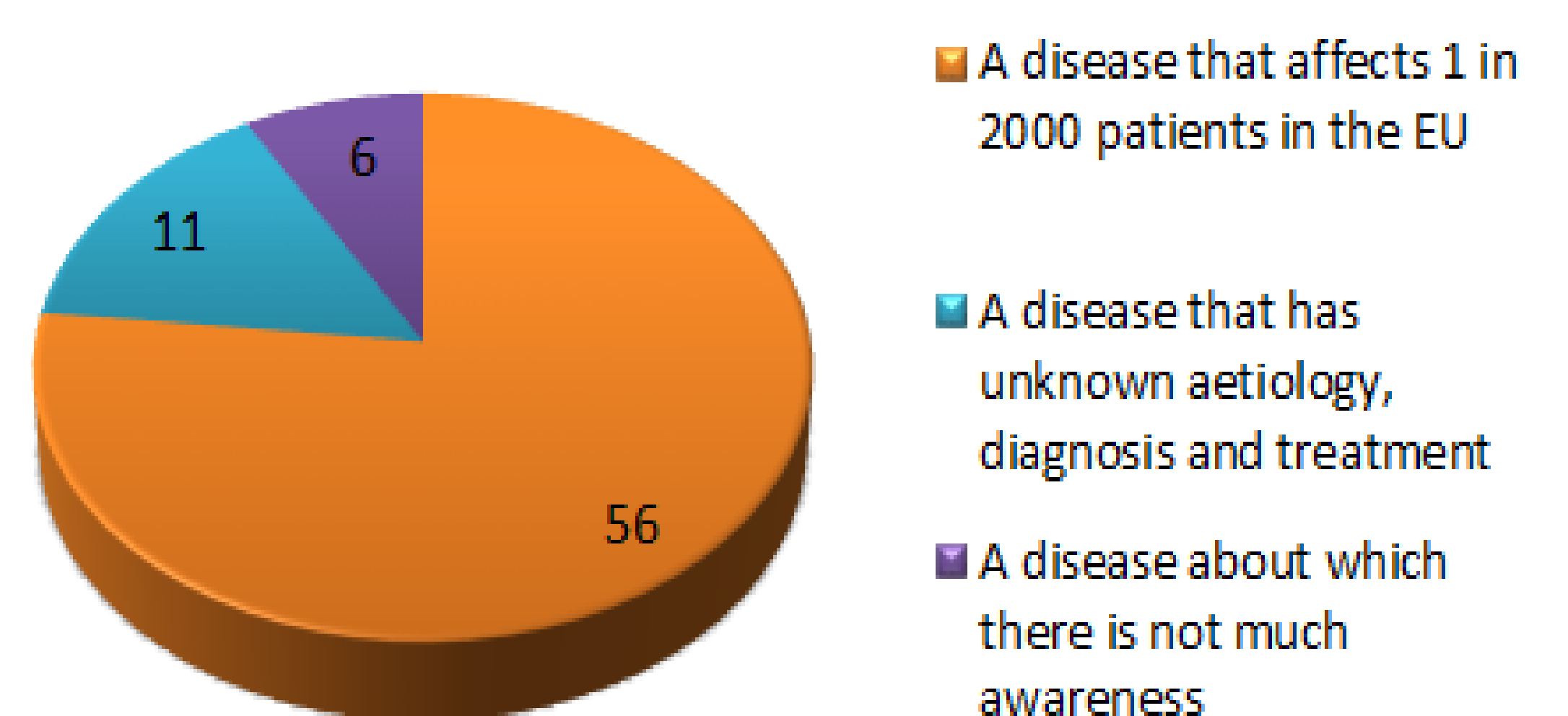
METHOD

- A questionnaire for healthcare professionals was developed and validated by a focus group comprising of two pharmacists; a community pharmacist and a medical representative, one specialist in family medicine, one basic specialist trainee in surgery, one volunteer from the National Alliance for Rare Diseases Support Malta and two lay persons.
- The questionnaire was analysed in terms of content, comprehensiveness, clarity and grammar. Ethics approval was granted prior to dissemination.
- The questionnaire was distributed to different pharmacies and clinics in all districts of Malta and made available online. The level of the professionals' awareness, experience and knowledge about rare diseases and orphan medicines was analysed.
- Common terminology towards the concept of rare diseases and orphan medicines was ranked according to association and relevance using a 5-point Likert scale, 1 being the lowest score and 5 being the highest. A revised definition for rare diseases and orphan medicines was proposed from the highest ranking terminology obtained from the questionnaire.

RESULTS

- 73 HCPs completed the questionnaire, of whom 62 were pharmacists, 8 general practitioners and 3 community nurses. Years of practice varied from 1 to 36 years.
- 39 respondents had diagnosed, encountered or examined a rare disease patient at a point in their career.
- 56 respondents had identified the definition for a rare disease as a disease that affects 1 in 2000 patients in the EU (Figure 1).
- The definitions for an orphan drug obtaining the highest mean rating score from the 73 HCPs were,
 - i. “drugs used for diseases for which there is no possible alternative treatment” with a score of 4.12.
 - ii. “intended for the diagnosis, prevention or treatment of a life-threatening or chronically debilitating condition” with a score of 4.10.
 - iii. “medicinal products with a significant benefit for those affected by the condition” with a score of 4.08.
- The definitions for a rare disease obtaining the highest mean rating score from the 73 HCPs were,
 - i. “presents a major psychological burden” with a score of 4.12.
 - ii. “a disease with an incidence of 0.65-1%” with a score of 3.93.
 - iii. “chronically debilitating” with a score of 3.85.

Figure 1: The definition of a 'rare disease' as defined by healthcare professionals (N=73)



CONCLUSION

The dissemination of existing definitions aims to extrapolate a standard definition which will be fully representative of rare diseases. The analysis of the perception and awareness on rare diseases and orphan medicines can improve healthcare in this sector and provide a more holistic approach to rare diseases.

REFERENCES

¹Yazhou C Jinxiang H. A proposed definition of rare diseases for China: from the perspective of return on investment in new orphan drugs. Orphanet Journal of Rare Diseases. 2015; 10(25); 1-3.