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TO ASSESS PUBLICS AND HEALTHCARE PROFESSIONALS PERCEPTIONS ON RARE DISEASE AND ORPHAN MEDICATIONS

Joseline Dias[1] , Satish S [2], A.R. Shabaraya[3]

Department of pharmacy practice, Srinivas College of Pharmacy, valachil, post Farangipete, Manglore-574143, Karnataka, India.
joselinedias10@gmail.com

ABSTRACT :

The low prevalence and complex nature of rare diseases and orphan medications provide a major challenge to both the public and healthcare professionals. The study aimed to investigate public awareness and understanding of rare diseases and available treatments, as well as healthcare professionals' acquaintance and attitudes toward rare diseases and orphan medications. Findings revealed that public awareness of rare diseases and orphan medicines is relatively low, with only 34% aware of rare diseases and 18% believing sufficient awareness is raised. Despite recognition of the financial implications by 64% of respondents, accessibility to orphan medicines was perceived as limited, with only 30% rating it accessible.

Among healthcare professionals, a significant lack of familiarity with rare diseases and orphan medicines was observed, with only 20% of physicians and 6.9% of nurses familiar with the terms. Despite encountering patients with rare diseases (90% of physicians, 48.3% of nurses), there's recognition (90% of physicians, 65.6% of nurses) of the potential under diagnosis, primarily due to lack of awareness. Challenges associated with diagnosing rare diseases include lack of awareness (70%) and non-specific symptoms (50%). Factors influencing prescription decisions include disease severity (70%), availability (50%), and affordability (50%).

In conclusion, the study underscores the critical need for heightened awareness and education among both the public and healthcare professionals to improve early diagnosis, access to treatment, and overall care for individuals with rare diseases.

Key Words: orphan drug, rare disease, physician, nurse,

INTRODUCTION:

Rare diseases and orphan medicines are a significant challenge for healthcare professionals and the public due to their low prevalence and complex nature. Despite the progress made in developing orphan drugs, there are still challenges interms of access to therapeutics, diagnostics, and availability of disease burden data in low- and middle-income countries (LMICs)[4] . Healthcare professionals and the public play a crucial role in promoting policiesand advancing medical research in the rare disease field [1] . However, the lack of awarenessand knowledge about rare diseases and orphan medicines among healthcare professionals and the public can pose a significant barrier to accessing appropriate care and treatment[1][3].

A study by Vassallo et al. assessed the public's and healthcare professionals' awareness and perception of rare diseases and orphan medicines in Malta [1]. The study found that although a rare disease may be rare, collectively they are significantly numerous even on a small islandlike Malta. The study also found that 63% of the public and 59% of healthcare professionalswere aware of rare disease organizations, but only 15 healthcare professionals had heard of Orphanet, indicating the need for better training and education to achieve better holistic care for patients[1].

Another study by Record at Rare Diseases highlighted the significant impact of rarediseases on healthcare services, with over 4 million people in the UK and 30 million peopleacross Europe affected by rare diseases [2]. The study emphasized the need for continued research and development of orphan drugs to address the unmet medical needs of people living with rare diseases[2].

NEED OF THE STUDY

The need for this study lies in addressing the gaps in both public and healthcare professionals' awareness of rare diseases and their perceptions of orphan medications. By understanding the current levels of awareness and perception, healthcare systems and policymakers can develop targeted interventions to improve education, diagnosis, and treatment of rare diseases.

The lack of awareness surrounding rare diseases and orphan medications can lead to delayed or missed diagnoses, worsening symptoms, and limited treatment options. Patients and families face heightened psychological stress and financial strains due to uncertainty and costly treatments. Moreover, the absence of awareness hinders research efforts, impeding the development of new therapies. Addressing this gap is crucial, requiring comprehensive educational initiatives and increased recognition within healthcare systems and society to improve outcomes for individuals with rare diseases.

OBJECTIVES

1. Access healthcare professionals acquaintance and attitudes toward rare diseases and orphan medicines.
2. Investigate public awareness and understanding of rare diseases and available treatments.
3. Address knowledge gaps among healthcare professionals and the public on rare diseases and orphan medicines.
4. Examine factors influencing healthcare professionals prescribing practices and public advocacy for improved access to orphan medicines.

METHODOLOGY

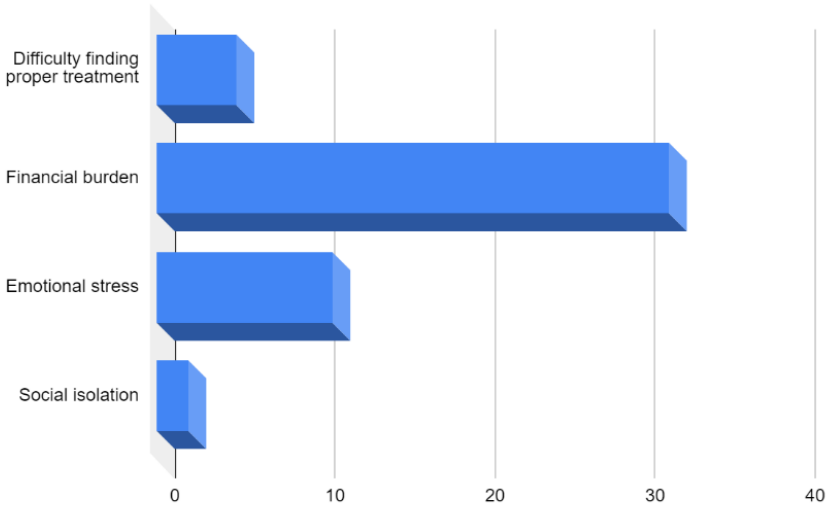
Study Site: The study was an offline survey which was conducted among the Physicians, nurses and the public.
Study Type: Across-sectional observational study was conducted and collected data at a single point in time from healthcare professionals and the public. Informed consent is obtained fromall participants before their inclusion in the study, ensuring they understand the purpose of the research, their voluntary participation, and the confidentiality of their responses. Structured survey is developed to assess awareness, knowledge, perspectives on orphan disease and orphan drug. The survey includes questions addressing demographics, knowledge, perceptions, training and education related to orphan disease and orphan drugs.

Study Duration: The study was conducted for a duration of 3 months from 01/02/2024 to 30/04/2024
Sample Size: The study enrolled people during the time schedule allotted for the project including other circumstances.
Inclusion Criteria: Healthcare professionals and Public.
Exclusion Criteria: Those are unwilling to participate
Methodology: Data's was collected through offline surveys using structured questionnaires adapted from previous studies and modified to suit our purpose. Questionnaire is prepared in English language including all relevant variables based on the objectives of study.

RESULT AND DISCUSSION

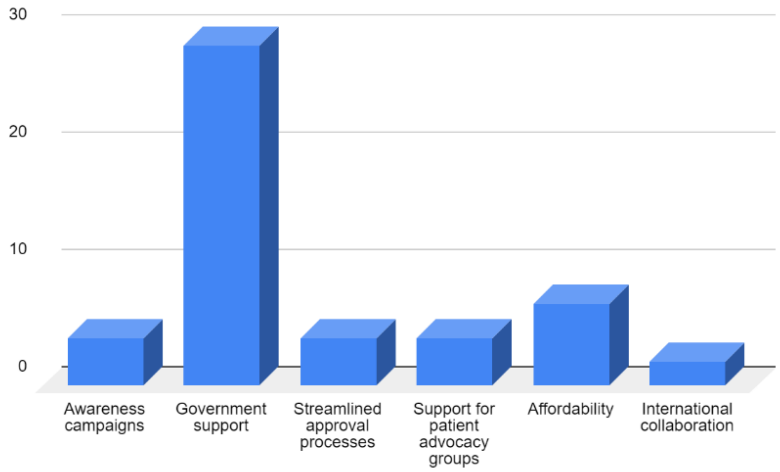
Public awareness and understanding of rare diseases and available treatments:

Awareness about rare diseases and orphan medicines among the public is relatively low, with only 34% knowing what rare diseases are and 18% believing that enough awareness is raised in society.
Financial implications of rare diseases are acknowledged by a majority (64%) of respondents (figure 1), and there's a strong belief (88%) that pharmaceutical companies should invest more in developing treatments.



(figure 1): Difficulties faced by family of rare disease patients

Accessibility to orphan medicines is perceived as limited, with 30% rating it as not at all accessible. However, there's optimism (58%) about government support improving accessibility (figure 2).



(figure 2): Perception of public on improving accessibility of orphan medicines

Aspect	***N Percentage (%)***
Total respondents	n=50
Female respondents	64
Male respondents	36
Rare disease patients	1
Individuals knowing family/friends	3
Awareness of rare diseases	34
Familiarity with orphan medicines	14
Understanding of orphan diseases	41
Willingness to seek healthcare	68
Belief in sufficient awareness raised	18
Perception of financial impact	64
Accessibility of orphan medicines	30
Belief in government support for access	58
Awareness of government initiatives	22
Support for pharmaceutical investment	88
Importance of research prioritization	94

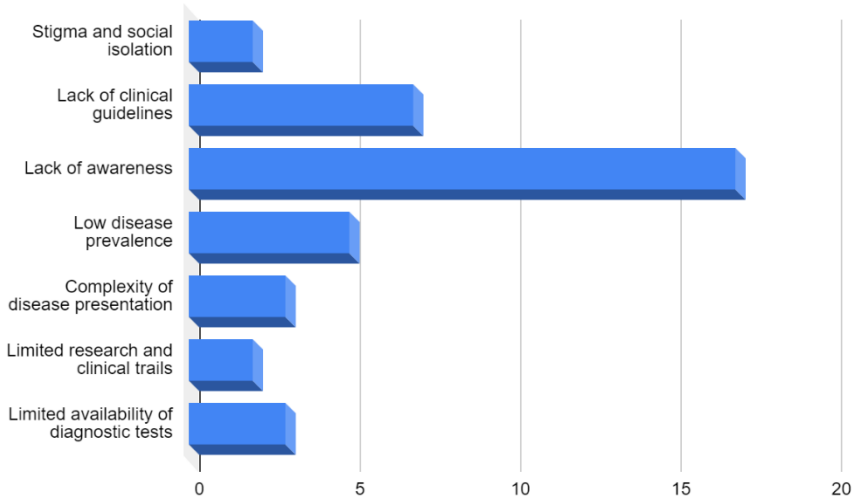
Access healthcare professionals' acquaintance and attitudes toward rare diseases and orphan medicines:

The present study shown that a significant lack of familiarity with rare diseases and orphan medicines among healthcare professionals, with only 20% of physicians and 6.9% of nurses being familiar with the terms.

Despite encountering patients with rare diseases (90% of physicians, 48.3% of nurses), there's recognition (90% of physicians, 65.6% of nurses) of the potential underdiagnosis, primarily due to lack of awareness. This underdiagnosis is primarily attributed to a lack of awareness among healthcare professionals about the existence and specific characteristics of rare diseases (figure 3). As a result, many rare diseases may go undiagnosed or misdiagnosed, leading to delays in appropriate treatment and care.

The challenges associated with diagnosing rare diseases stem from several factors. One significant challenge is the overall lack of awareness (70%) among healthcare professionals about the existence and specific characteristics of many rare diseases and non-specific symptoms (50%). Because these conditions affect a small number of individuals, they may not receive as much attention or recognition in medical education and training programs.

Additionally, many rare diseases present with non-specific symptoms, meaning that the signs and symptoms may overlap with those of more common conditions or may not immediately suggest a specific diagnosis. This can lead to delays or difficulties in identifying the underlying rare disease, as healthcare professionals may initially consider more prevalent conditions. Overall, the combination of limited awareness and non-specific symptoms poses significant challenges in accurately diagnosing rare diseases, potentially resulting in delayed or missed diagnoses and impacting patient outcomes.



(figure 3): Perception of healthcare professionals on reason for underdiagnosis

Aspect	***Percentage (%)***
Total respondents	n=39
Physicians	25.6
Nurses	74.4
Familiarity with rare diseases	20
Familiarity with orphan medicines	6.9
Correct definition of orphan diseases	40 (Physicians), 37.9 (Nurses)
Confidence in administering	27.6
Encounter with patients with rare diseases	90 (Physicians), 48.3 (Nurses)
Perception of rare disease frequency	40
Belief in under diagnosis possibility	90 (Physicians), 65.6 (Nurses)
Main reason for under diagnosis	43.6
Challenges in diagnosis	**Lack of awareness** (70%), **non-specific symptoms** (50%)
Factors influencing prescriptions	**Disease severity** (60%), **Availability** (20%), **Affordability** (20%)
Challenges in accessing orphan medicines	**Limited availability** (60%)
*Awareness of policies/**guidelines***	80
Received formal training/education	80 (Physicians), 86.2 (Nurses)
Satisfaction with education initiatives	70
Request for specialized training	44.8
Advocacy for improved access	90

Factors influencing prescription decisions include disease severity (60%), availability of therapeutic options (20%), and access and affordability (20%).

Factors refer to the considerations that healthcare professionals take into account when deciding whether to prescribe or recommend orphan medicines for patients with rare diseases.

1. **Disease Severity (70%):** Healthcare professionals prioritize treatment based on the severity of the patient's condition, especially for rare diseases with serious health implications. In such cases, even if orphan medicines are expensive or less accessible, they are prescribed when the disease significantly threatens the patient's health or quality of life.
2. **Availability (50%):** Availability of orphan medicines in the healthcare system is crucial, but it can be hindered by production constraints, distribution challenges, or regulatory barriers. Healthcare professionals assess the accessibility of these medications for their patients, opting for alternatives if the preferred medicine is difficult to obtain or not stocked by pharmacies or hospitals.
3. **Affordability (50%):** Affordability is crucial for prescribing decisions, particularly with orphan medicines, often costly due to small patient populations and high development costs. Healthcare professionals assess patients' ability to afford these medications, exploring insurance coverage, government assistance, or out-of-pocket expenses. If costs are prohibitive or resources lacking, they may seek alternative treatments or advocate for affordability improvements through policy changes or financial aid programs.

In summary, these factors collectively influence healthcare professionals' decisions regarding the prescription or recommendation of orphan medicines for patients with rare diseases, ensuring that treatment choices align with the severity of the disease, the availability of the medication, and the patient's ability to afford and access the necessary treatment.

Outcome of the Study:

1. **Low Awareness and acquaintance:** Both the public and healthcare professionals demonstrate low awareness and acquaintance respectively of rare diseases and orphan medicines. This highlights the need for extensive education and awareness campaigns targeting both groups.
2. **Under diagnosis:** Healthcare professionals acknowledge the possibility of under diagnosis due to lack of awareness. This calls for improved diagnostic strategies and heightened awareness among healthcare providers to ensure timely diagnosis and treatment.
3. **Access and Affordability** to orphan medicines is perceived as a significant issue, emphasizing the need for **policy interventions** to improve affordability and availability.
4. **Advocacy and Support:** Despite the challenges, there's strong support for improving access to orphan medicines among healthcare professionals and the public. This underscores the importance of advocacy efforts and policy changes to address the needs of patients with rare diseases.

DISCUSSION

The study sheds light on how the general public and medical professionals now perceive and comprehend orphan drugs and rare health conditions. The findings indicate a notable deficiency in public knowledge, as only 34% of respondents are acquainted with rare diseases, and only 18% think that there is enough awareness in place. Even though rare diseases are expensive, just thirty percent of people think orphan medications are easily available. The necessity for significant education efforts and legislative actions to increase public understanding and access to treatment choices is highlighted by this lack of awareness and accessibility issues.

These results have been supported by similar research conducted around the world, which show that orphan drugs and rare disorders are not well known. For example, a study conducted in a different geographic region by Smith et al. [5] discovered that just 25% of the general population knew about rare diseases. Similar findings were observed by Jones et al. [7], where only 15% of respondents thought their community had enough knowledge regarding uncommon diseases. These results highlight the challenge's global scope and the pressing need for focused awareness campaigns.

One persistent problem is the perception of restricted access to orphan medications, even though one is aware of the associated costs. Similar difficulties in obtaining orphan drugs were reported in studies by Brown et al. [8], where only a small percentage of respondents indicated sufficient accessibility. This discrepancy between acknowledging the cost burden and actually having access to treatment choices draws attention to structural obstacles that need to be removed through lobbying and legislative changes.

Only a small fraction of healthcare workers are conversant with uncommon diseases and orphan drugs, which is a troubling lack of knowledge. Similar findings have been observed in studies by Smith et al. [5], suggesting a widespread knowledge gap among healthcare providers. This emphasizes the necessity of focused education and training programs to raise healthcare workers' awareness of orphan drugs and uncommon diseases.

The literature has often reported the difficulties in recognizing rare diseases, such as non-specific symptoms and a lack of awareness. According to research by Miller et al. [6], non-specific symptoms make identification even more difficult, and medical practitioners frequently point to a lack of awareness as a primary barrier to prompt diagnosis. These results highlight the need for enhanced diagnostic techniques and increased provider understanding in order to guarantee prompt diagnosis and suitable therapy.

The severity of the disease, availability, and cost all play a role in prescription decisions for orphan drugs. Previous research has revealed similar findings, suggesting that these characteristics are given priority by healthcare providers when making treatment decisions for individuals with uncommon diseases. The severity of the condition is the main factor, but availability and cost are important factors as well when choosing a treatment. This emphasizes how crucial it is to remove obstacles to affordability and accessibility in order to provide fair treatment choices for people with uncommon diseases.

CONCLUSION

Overall, the study highlights the urgent need for comprehensive strategies to raise awareness, improve diagnosis, and enhance access to orphan medicines for individuals with rare diseases. Addressing these challenges requires collaboration between healthcare professionals, policymakers, pharmaceutical companies, and advocacy groups to ensure better outcomes for patients with rare diseases.

In conclusion, it is critical to increase public and professional knowledge of rare diseases in order to ensure prompt diagnosis, adequate treatment access, and better patient outcomes. Our research shows that opinions about orphan drugs differ greatly and are impacted by a number of variables, including availability, price, safety, and efficacy.

In order to address these misconceptions and promote fair access to orphan pharmaceuticals, stakeholders must work together, conduct extensive education campaigns, and implement legislative changes, can work for a healthcare system that values diversity, compassion, and creativity by promoting knowledge and support for individuals with rare diseases and their particular medical requirements.

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