

Title: “Rare Diseases: From a Pharmacist’s Perspective”

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Abstract:

Background: Rare diseases affect an exceedingly small number of patient population worldwide. Often these are undiagnosed or there is a late diagnosis because of lack of suspicion or awareness about these diseases. Pharmacologists in India form a crucial crux in determining different treatment modalities and it is essential they are made aware of rare diseases which can contribute to meagre treatment modalities.

Objectives: To determine awareness and perspective of pharmacist’s regarding rare diseases in India.

Materials and methods: A questionnaire survey were circulated amongst pharmacologists based in India that contained baseline demographic data, awareness, knowledge and opinion for including study of rare diseases in curriculum. The survey form was prepared using google services and circulated through commonly used social media that included Facebook, WhatsApp, instant messaging amongst others.

Results: A total of 35 responses were recorded from different pharmacologists. The median age of participants was 35 years. 51.43% of participants had experience as pharmacologists between 3 to 5 years. 45.7% of participants had very good or good knowledge about rare diseases. 34.3% of participants felt confident in caring for patients with rare diseases and a similar percentage had studied rare diseases as students. 94.3% of participants felt a need to broaden their knowledge regarding rare diseases. 80% felt need to include rare diseases in medical curriculum.

Conclusion: There is urgent need to include rare diseases in medical curriculum.

Key words: Rare diseases, Pharmacologists

Introduction:

India consists of culturally diverse ethnic groups with high instance of multiple endogamous marriages leading specific marriage patterns. With a huge population, rare diseases translate to a huge disease burden for these rare diseases [1]. Technological advancements such as stem cell therapies, organs-on-chips, etc appear to be promising treatment modalities in management of rare diseases [2]. Drugs are granted an orphan designation if they are for the treatment of rare diseases that are life-threatening or seriously debilitating [3]. An individual rare disease can affect less than 5 to 7 individuals in 10,000 but can collectively affect approximately 6-8% of the population worldwide [4]. Since 2000, New medical entities with orphan designation encompass a significant portion of approved drugs and constitute about 80% of the approved drugs that have established novel human genome-encoded products in recent years [5]. In India, the Central Drugs Standard Control Organization (CDSCO)'s definition is considered as the national standard benchmark. In the new clinical trial guidelines, rare disease is one that afflicts fewer than 500,000 patients in the country [6].

Rare diseases effect an exceedingly small number of patient population worldwide. Often these are undiagnosed or there is a late diagnosis because of lack of suspicion or awareness about these diseases. Pharmacologists in India form a crucial crux in determining different treatment modalities and it is essential they are made aware of rare diseases which can contribute to meagre treatment modalities. With this intention the current questionnaire-based study was carried out to assess the pharmacologists based out of India.

Materials and methods:

A questionnaire survey was circulated amongst pharmacologists based in India that contained baseline demographic data, awareness, knowledge, and opinion for including study of rare diseases in curriculum. The survey form was prepared using google services and circulated through commonly used social media that included Facebook, WhatsApp, instant messaging amongst others. Tables and charts were prepared for depiction of data. The data was expressed in terms of fractions and percentages as applicable. Pie charts and bar diagrams were predominately used.

Response was received from 35 participants from all over India. The survey was anonymous, with a voluntary participation. The participants were explained the need for study when the questionnaire was shared for the survey. All 35 participants replied to all the questions. The data expressed in the results corresponds to all the participants.

The questionnaire was referred from a study by Ahmed NJ et al. [7] (2021) wherein a similar study was conducted to evaluate self-assessment of knowledge of rare diseases.

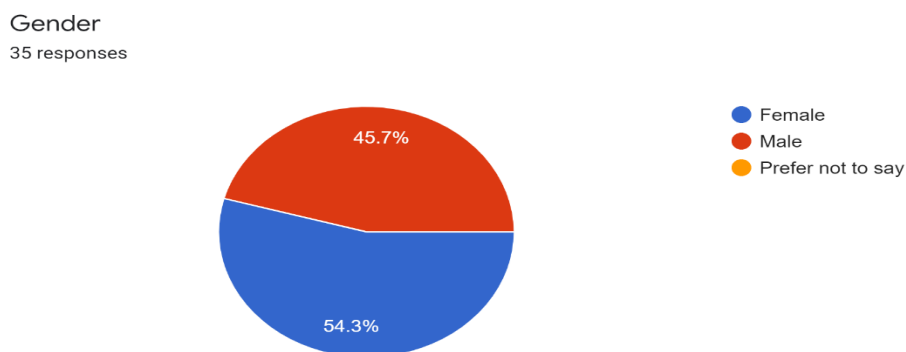
Results:

Table 1: Age parameters of the participants

Mean age	36.14 years
SD	6.96
Age range	27 to 60 years
Median age	35 years

The median age of participants was 35 years with an age range of 27 to 60 years.

Figure1: Gender



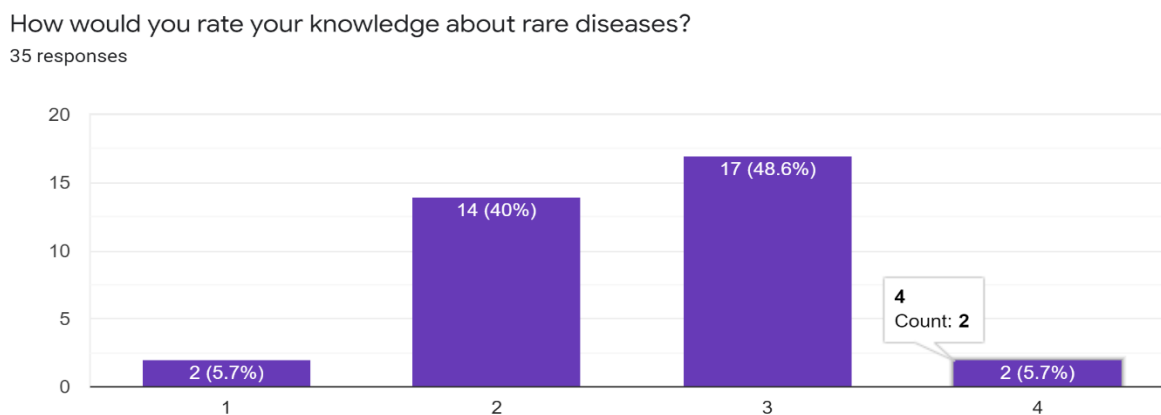
54.3% of the participants were females whereas males constituted 45.7%.

Table 2: Duration of experience

Duration of experience	No of Participants	Percentage
< 3years	6	17.14%
3 to 5 years	18	51.43%
6 to 10 years	8	22.86%
Above 10 years	3	8.57%
Total	35	100%

The majority of the participants had an experience of 3 to 5 years at 51.43%, followed by those between 6 to 10 years of experience at 22.86%. Those with experience of less than 3 years constituted 17.14% with those above 10 years at 8.57%.

Figure2: Knowledge about rare diseases



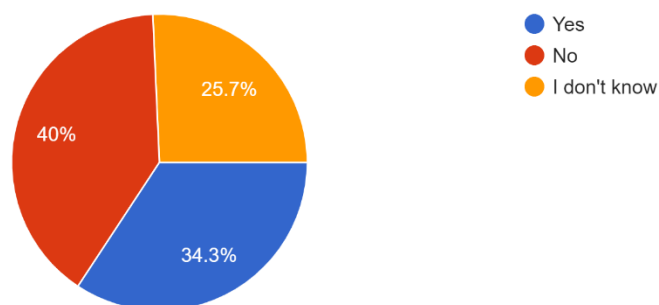
Legend: 1: Very good 2: Good 3: Average 4: Poor

48.6% had average knowledge regarding rare disease, those with good knowledge included 40%. The extremes of very good and poor knowledge included 5.7% each of the participants.

Figure 3: Preparation as a pharmacologist for caring for a patient with rare disease

Do you feel prepared for caring for a patient with a rare disease?

35 responses

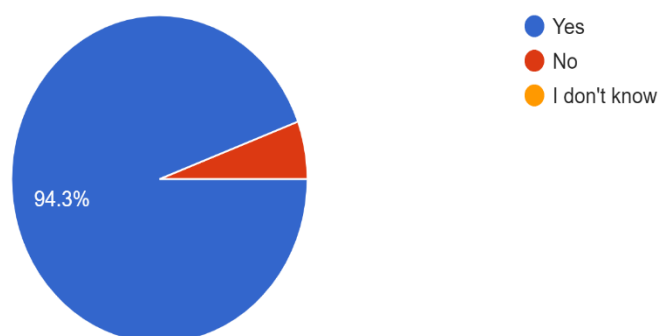


34.3% felt prepared for caring a patient with rare disease whereas 40% were unprepared for caring patients with rare disease.

Figure4: Learning about rare diseases

Would you like to broaden your knowledge about rare diseases?

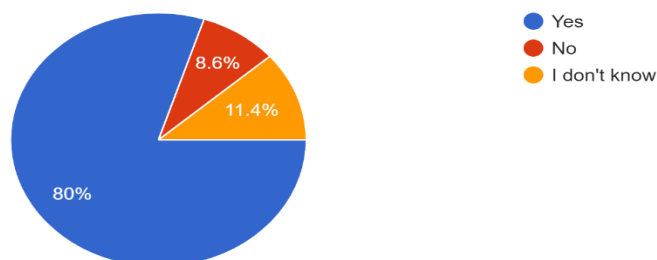
35 responses



An overwhelming 94.3% of the participants were interested in broadening their knowledge about rare diseases.

Figure 5: Inclusion of rare diseases in medical curriculum

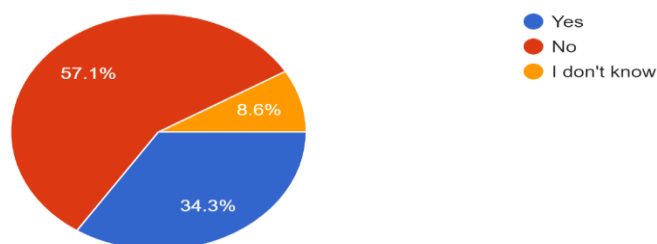
Do you think that there should be a mandatory course on rare diseases in medical curricula?
35 responses



80% of the participants felt the need for inclusion of mandatory course on rare diseases in medical curriculum.

Figure 6: Training as a student about rare diseases

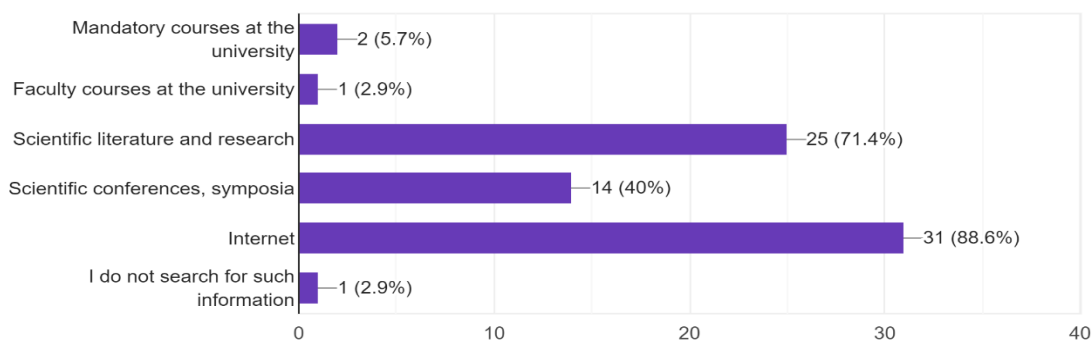
Have you had any classes about rare disease during your studies?
35 responses



A meagre 34.3% had classes on rare diseases during studies whereas 8.6% were unaware and 57.1% mentioning that no classes were held.

Figure 7: Source of knowledge about rare diseases

Where do you get your knowledge about rare disease from?*(can select multiple options)
35 responses



88.6% of the participants dependent on internet to obtain information regarding rare disease. A substantial amount depends on scientific literature and research (71.4%) and scientific conferences and symposia (40%). Most of the participants selected more than one option.

Discussion:

The current study was conducted to determine awareness and perspective of pharmacologist's regarding rare diseases in India. A total of 35 responses were recorded from different pharmacologists. The median age of participants was 35 years. 51.43% of participants had experience as pharmacologists between 3 to 5 years. 45.7% of participants had very good or good knowledge about rare diseases. 34.3% of participants felt confident in caring for patients with rare diseases and a similar percentage had studied rare diseases as students. 94.3% of participants felt a need to broaden their knowledge regarding rare diseases. 80% felt need to include rare diseases in medical curriculum. 88.6% of the participants dependent on internet to obtain information regarding rare disease. A substantial amount depends on scientific literature and research (71.4%) and scientific conferences and symposia (40%).

The findings of the current study are in line with various studies such as Medić B et al. [8], Vandeborne L et al. [9] that emphasized on need for revising academic medical education on rare disease to keep abreast with latest trends in rare diseases.

Groft SC et al. [10] suggested joining mainstream research and treatment based on reliable epidemiological data for rare diseases. Ramalle-Gómara E et al. [11] noted that Primary care physicians and specialists showed low training level in rare diseases.

As observed from our study and global trends; lack of awareness and training for medical professionals and inadequate emphasis on rare diseases in medical education are few of the contributory factors about lack of early diagnosis and standard point of care for patients suffering from rare diseases. In India where pharmacologists are predominately involved in drug research, it is prudent for their involvement in studying rare diseases. This will contribute a higher pool of qualified professionals in drug research and development for patients suffering from rare diseases. It is equally necessary for the government to formulate policy decisions for research on rare diseases and incentivizing orphan drugs.

Conclusion:

The emphasis on rare disease is less than satisfactory in medical curriculum. An overwhelming majority of the pharmacologists finds it useful for broadening knowledge about rare diseases and its inclusion in medical curriculum. These steps will be useful in broadening further research especially on treatment modalities of rare diseases. This will be particularly useful in a country such as India where a high number of cases of rare diseases would be available.

Conflict of interest: None

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