ISSN: 2643-4571

Agrawal et al. Int J Rare Dis Disord 2019, 2:008
DOI: 10.23937/2643-4571/1710008
Volume 2 | Issue 1
Open Access

International Journal of
Rare Diseases & Disorders

ORIGINAL RESEARCH ARTICLE

Baseline Knowledge of Rare Diseases in India - A Survey

Agrawal RK[1], Amaresh Rao M[2], Brian M[3], Chowdary GKB[4], Gayatri K[5], Krishnaji Rao M[6], Sambasiva Rao P[7], Namineni S[8], Srinivasa Rao N[9], Sikri BR[10], Sanahullah S[11], Gawron S[12], Agarwal DR[13], Lakshmi Rao K[14] and Ramaiah Muthyala[15]*

[1]*Nakoda Chemicals Ltd, Jeedimetla, Hyderabad 500055, India*

[2]*Nizam Institute Medical Sciences, Punjagutta, Hyderabad 500082, India*

[3]*Paediatrics, University of Minnesota, Minnesota 55455, USA,*

[4]*Nyaya Seva, Guttala Begumpet, Hyderabad 500033, India*

[5]*Tapadia Diagnostic Center, Banjara Hills, Hyderabad 500034, India*

[6]*Lavender Tower, Gachibowli, Telecom Nagar, Hyderabad 500032, India*

[7]*Zen Securities, Begumpet, Hyderabad, 500033, India*

[8]*Pediatric Dentistry, Rainbow Children Hospitals, Banjara Hills, Hyderabad 500034, India*

[9]*Delphis-Center for Complementary Medicine & Therapies, Hyderabad 500001, India*

[10]*ABS Mercantiles PVT. Ltd Haryana 122022, India*

[11]*Akbarbagh, New Malakpet, Hyderabad 500036, India*

[12]*Minnesota Lawyers, Mendota Heights, Minnesota 55118, USA*

[13]*ITAGBS, Kolkata, West Bengal 700010, India*

[14]*CSIR- Center for Cellular and Molecular Biology, Hyderabad 500007, India*

[15]*Experimental Clinical Pharmacology, University of Minnesota, USA*

***Corresponding author:** *Ramaiah Muthyala, Experimental Clinical Pharmacology, University of Minnesota, USA; Indian Organization for Rare Diseases, Plot #397 Road No: 22b, Jubilee Hills, Hyderabad, Telangana, 500033, India, Tel: +9189-7890-0033; +1612-2026-168*

Abstract

Recently, rare diseases have received worldwide attention. The developing countries have fallen seriously behind in regards to awareness, drug development, diagnosis, and social services. India, which has one-third of the world's rare disease population, has neither accepted definition for RD nor an accurate assessment of the problem. Due to the exorbitant cost of orphan drugs, difficulties in diagnosis and treatments, the Indian government is often in a dilemma as to how to effectively, and efficiently formulate a National Health Policy with a 1.45% GDP healthcare budget for 1.3 billion people.

Indian policymakers want to find out the number of RD and the extent of the population suffering from them. It is not possible to give an adequate answer without understanding the scope to which RD occurs within the general population.

A survey was conducted to know the basic knowledge of RD in the general population. The sample survey was obtained from a subset of the population with the criteria arbitrarily set as people with above-average literacy who could be easily reached and expected to understand the proposed study.

A questionnaire was designed to collect information specific to awareness of RDs and ODs. The 6005 people were contacted via emails; 599 responses were received. The results show that the health care professionals appear to have some awareness as compared with non-healthcare professionals, but even among health care professionals, only one third had a rudimentary understanding of RD and OD, whereas three-fourths have virtually no knowledge of RD.

Forty-three percent of health professionals had not seen rare disease patients, and a large percent of practicing physicians had not seen even one rare disease patient in their

Citation: Agrawal RK, Amaresh RM, Brian M, Chowdary GKB, Gayatri K, et al. (2019) Baseline Knowledge of Rare Diseases in India - A Survey. Int J Rare Dis Disord 2:008. doi.org/10.23937/2643-4571/1710008

Accepted: November 04, 2019; **Published:** November 06, 2019

entire professional practice. It becomes clear from this survey that the most important issues are awareness and diagnosis. This survey is the first of this kind ever conducted and is a tentative systematic first step towards understanding the basic knowledge of rare diseases in India. The results serve as a preamble to complete understanding. Much more must be done to gauge the magnitude of the problems to draft the targeted and effective national health care policy for rare diseases.

Keywords

India, Health policy, Rare diseases, Orphan drugs, Survey, Awareness

Abbreviations

RD: Rare Disease; OD: Orphan Drugs; RDI: Rare Diseases International; IRDIRC: International Rare Diseases Research Consortium; NRI: Non-resident Indian

Introduction

Rare diseases - diseases that affect a small number of people compared to general populations. Very recently, the phrase “rare diseases” got the attention of the public and scientific community in India [1] (Figure 1). Quite often, RDs are mistaken for neglected or tropical diseases such as TB and Malaria. Many RDs are genetic, and in some cases, the cause is unknown. Currently, there are more than 7,000 known rare diseases [2], which affect an estimated 8-10% of the world population. Unlike neglected diseases, which are localized in certain countries, the RDs have no geographical boundaries.

In the early 80s, the rare disease is arbitrarily defined in the USA as a condition, which affects less than 200,000 of the population [3] while, other countries, for example, Europe defined 1 in 2000, Japan defined 1 in 2,500 - a population-based definition. The rarity heightens the isolation and offers limited treatment options. The total RD population could be in hundreds of millions; however, each disease may represent hundreds or thousands of patients, worldwide. Due to the lack of profitability, pharmaceutical companies stopped developing drugs to treat rare diseases patients. In 1983, to provide incentives to pharma companies, the Orphan Drug Act (ODA) was established in the USA [4]. ODA became a guiding light for many countries in the world. By the end of 2017, more than 450 products, for 668 orphan indications were approved by the FDA [5]; while before the ODA, the orphan drugs were fewer than ten. These orphan drugs are not available in India due to import restrictions, unaffordable prices, etc. Further, there is neither such ODA nor any other provisions available to patients in India. As a result, 80-90 million RD patients are suffering.

In India, awareness of RDs lacks, not only the general population but health care professionals, as well as policymakers. Expertise and specialized institutions

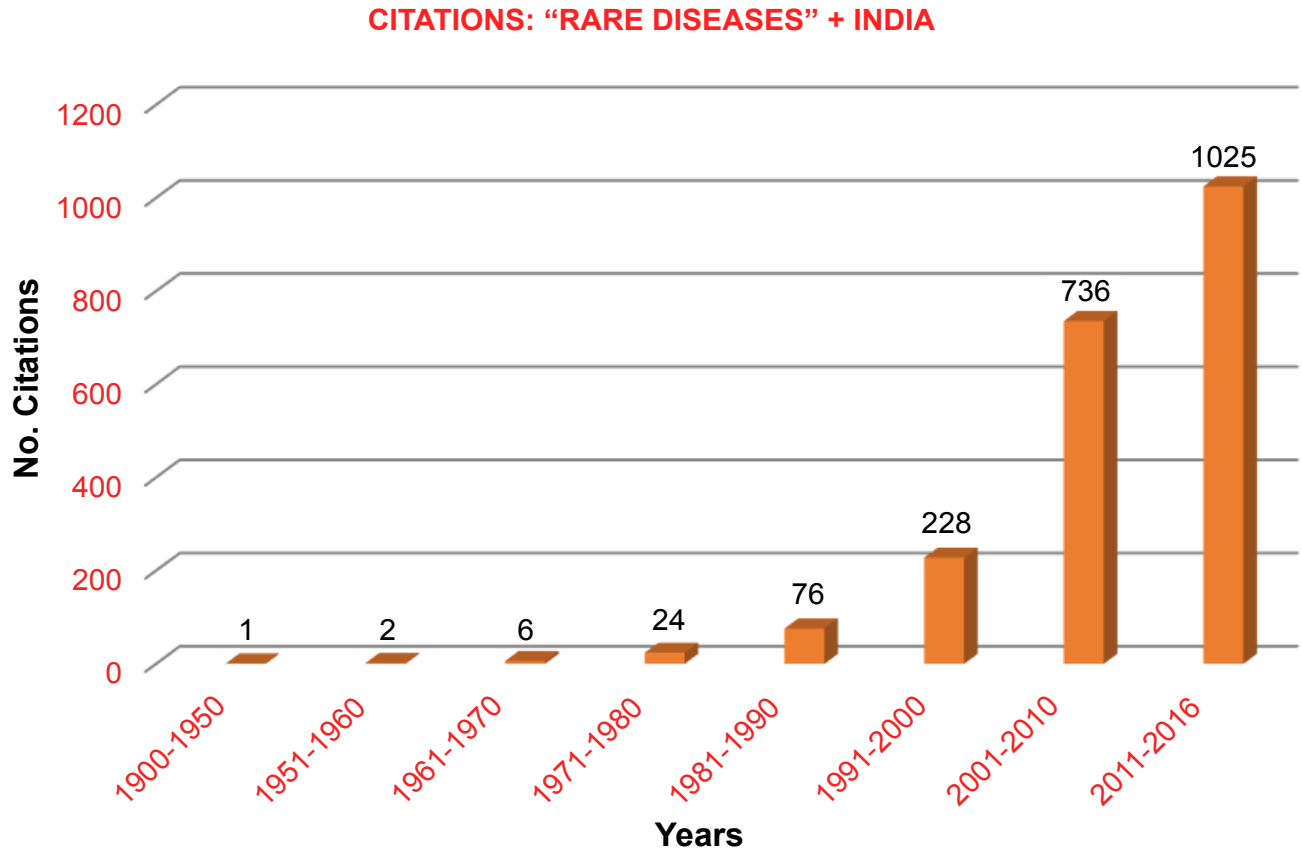


Figure 1: Citations of [Rare Diseases + India] from the PubMed.

are scarce. Significant barriers to access diagnostics and care facilities exist. Documentation of rare diseases within the healthcare system is insufficient. Due to the lack of a centralized disease-specific registry and data collection system, there is no valid information on the epidemiology of rare diseases in the country.

Indian Organization for Rare Diseases (IORD) [6] is an umbrella patient organization representing all rare diseases and rare disease patients in India. Its mission [7] is to raise awareness, advocate public policy, encourage pharma companies [8] to develop drugs and biotech companies, academic institutions to developing diagnostic tools [9]. During the last fourteen years of her service to the RD community in India, IORD recognizes that RD not only affects the patients but also affects the entire community (remaining 92% of non-orphan) - physically, emotionally, and financially.

In India, often a common question arises among policymakers regarding the number of rare diseases and the number of patients suffering from RDs. As India does not have the definition of rare diseases, it is hard to estimate accurately the number of patients suffering from rare diseases. Therefore, it is challenging for policymakers to draft the health care policy that adequately addresses the needs of patients, provides incentives to the pharma industry to develop drugs [10], develop tertiary infrastructure to treat RD patients. In 2017 the Government of India released a policy document [11], and while implementation, it faced hurdles [12].

Over the years, several surveys were conducted in developed countries. However, they are often focused on specific issues such as access to orphan drugs [13], the role of RD patient groups in research [14], challenges faced by patients [15], issues related to rare disease caregivers [16], the social media platform to contact patients [17], patients' survey [18]; balancing care and life [19]. Rare Barometer Voices [20] conducts surveys very frequently on a variety of topics.

IORD initiated a 'first-of-its-kind' survey to obtain a systematic and baseline knowledge of RD in the country. No such survey has ever been conducted by any government or any agency in any country. It is not practical or doable to survey 1.3 billion people in a country with multiple states, languages, economic and social inequalities, and a host of other reasons. We expected a well-represented sample survey to give meaningful information, relatively quickly and inexpensively. Further, the purpose of this survey was to provide a snapshot, the baseline understanding of rare diseases in the country. The information from this survey will be aggregated and compiled for a report and is freely available to anyone. It was expected that the study would give impetus to the national conversation on care and support for rare diseases' families, and provide greater awareness of how RDs impact communities.

There are significant resources for educational materials available to most people in developed nations. However, millions of individuals and families in the developing nations around the world do not have access to most of these resources except to those who can access these services [21] through the internet. The target audience for this survey were those who access to the World Wide Web and would understand the purpose.

Methodology

Due to the large rural population, low literacy, the survey was limited to the population who could be reached through the internet. The sample survey was obtained from a subset of the population with the criteria arbitrarily set as people with above-average literacy such as college degree, which could easily be reached and expected to understand the purpose. For a broader survey, staff, funding, and time are the limitations. The survey was conducted by selecting units from a population, which comprised medical, and non-medical fraternity that could be meaningfully thought of as defining a population to be studied. The participants should be residing in India, or non-resident Indians (NRIs) [22] living in the United States. Non-sampling errors (excluding certain population), may have reduced precision in our estimates. It is difficult to quantify coverage error without special studies of the un-sampled portion of the population. Further, practical issues arose because of the complexities of gathering information on many real issues, for example, knowledge of the known rare diseases, diagnosis, and treatments, etc. The survey instrument involves a questionnaire that provides a script for a standard set of questions and response options (Supplementary Material and Survey Questionnaire).

The questionnaire was designed to collect information specific to RD and OD in such a way that the answers are either 'yes' or 'no' type. 'Multiple-choice questions' and some others require open-ended responses - what orphan diseases and orphan drugs are, number of people suffering from RD, causes of RD, cost of medicine, affected age group, the time taken to diagnosis, which group of people affect most, etc. A separate section was included to the practicing physicians who are asked specific questions, - e.g., how often RD patients are seen in their practice and their available resources for diagnosis and treatment, etc. At the end of the survey, the respondents have the opportunity to leave a comment. The survey re-

Table 1: Male and female Respondents.

	# of Respondents	Response %
Total Responses	6005/599	10.0
Male	351	58.6
Female	248	41.4

Table 2: Age groups, medical and nonmedical fraternity respondents.

Age Groups	# of Respondents	Response %
18-20 Years	46	7.7
21-23 Years	111	18.6
24-28 Years	89	14.9
29-30 Years	46	7.6
31-40 Years	132	22.0
41-50 Years	127	21.2
> 50 Years	48	8.0
Category of Respondents		
Non-Medical Fraternity	406	67.7
Medical Fraternity (including Physicians, Nursing & Pharmacists)	193	32.3

Table 3: Awareness levels of respondents on RD & OD.

Unawareness Levels among Respondents on RD and OD		
	# of Respondents	Response %
Never heard of Rare Diseases	142	23.7
Institution NOT offering awareness on RD	180	30.0
Never heard of Orphan Drugs	255	42.6
Unaware of Commercial availability of OD	395	66.0
Unaware of the Definition of RDs	225	37.6
Unaware of # of RDs	290	48.4
Unaware of the # of RD Patients population in India	343	57.2
Unaware of the cause of RD	122	22.3
A common cause of RD is Genetic	249	41.6
RD occurs by birth	84	14.0
RD can occur any age	291	48.5
Unaware of the onset age of RD	116	19.3
1%-2% population will get affected by RD	217	36.3
Unaware of the % of affected RD population	181	30.2
Poor Countries Suffer	105	17.6
Rich Countries Suffer	48	8.0
No RD discrimination in Rich & Poor Countries	446	74.4
RD affects all countries	406	67.7
The time taken to diagnose RD	Seven days to 5 Years	

sponses are compiled in (Table 1, Table 2, Table 3, Table 4 and Table 5).

The average time the respondents would take to complete the survey is expected to be twelve minutes. The survey takers were advised not to refer to Google or other materials and not to consult others for answers; as such attempts would skew the results and defeat the purpose. The survey takers are informed that the objectives of this survey are to determine baseline knowledge of RD and OD in India.

The Survey Questionnaire was sent randomly to people in India, and NRIs in the USA via their e-mails. The participants come from major cosmopolitan cities in India and are limited to urban areas [23]. The survey takers come from randomly selected States in India and some Western, Eastern, and Midwestern States in the United States. The emails of survey takers are obtained from the public, the professional associations, regional non-governmental associations, pharmacy, medical, nursing educational institutes/schools, practicing physicians, industry personnel. The minimum qualification of participants is a college degree in any area of disciplines such as arts, science, and professional degrees in information technology, medical, pharmacy, and nursing, etc. The data was collected between the periods of December 2017 to October 2018.

Results

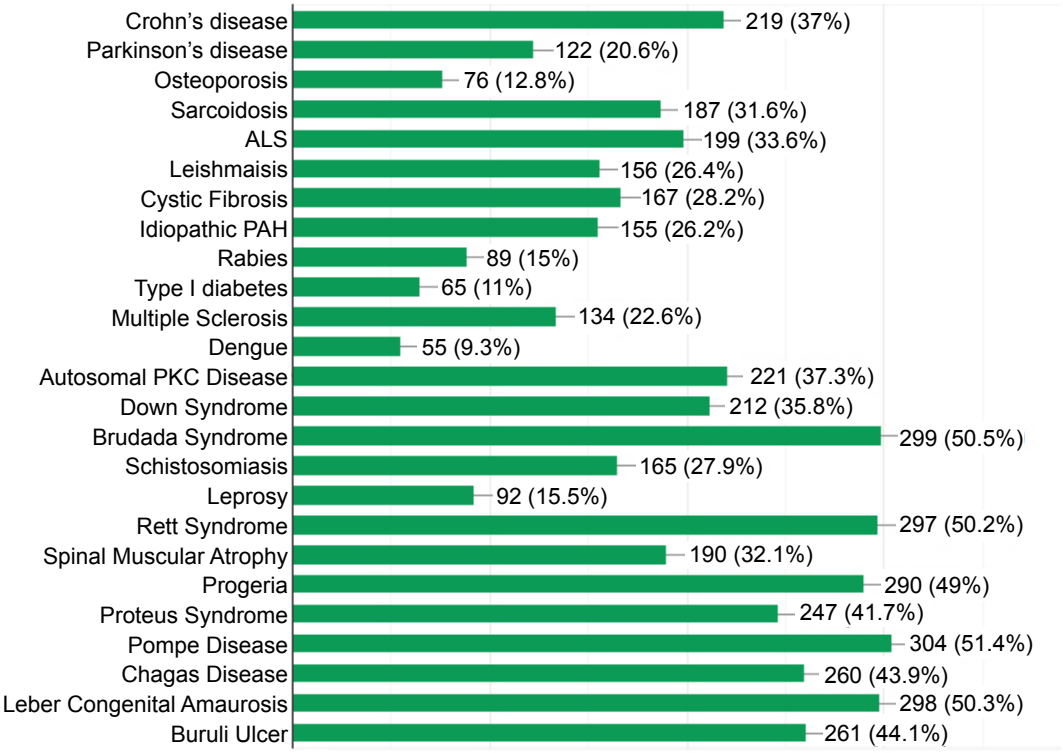
A Survey Questionnaire was e-mailed to 6005 people, and 599 responses were received. The ratio of females (248) to male (351) was 0.69 [24] (Table 1). Among the respondents, 80% were from India, and

the rest of them were from abroad. Responses were evenly spread across all age groups. The responses of the age groups, > 50, 18-20, and 29-30 are the lowest at 8%, 7.7%, and 7.6% respectively and the responses were highest with age groups, 41-40, 21-23, were 21.2% and 18.6% respectively. The next age group 24-28 was 14.9%. The nonmedical respondents had the highest participation at 67.7%, whereas the practicing physicians [25] was at 5.7% (Table 2). 32.3% of respondents of the survey were with medical, pharmacy, or nursing background with a minimum of an undergraduate degree; among them, 36.3% of people had one or more scientific publications, whereas 58.9% of respondents had a membership in professional societies. 70% of respondents had not participated in any voluntary organizations. 30% of respondents indicated that their institutions did not offer rare disease awareness programs. 23.7%, 42.6% of respondents never heard the term "rare diseases" and "orphan drugs", respectively. 17.6% of respondents thought poor countries and 8% of respondents felt that the rich countries suffer most while 74.4% felt that RDs do not discriminate rich or poor (Table 3). 37.6% of respondents did not know the definition of a rare disease. Similarly, 48.4%, 57.2% respondents unaware of the number of RDs and patient population in the country respectively, while 66% of respondents did not know the number of commercially available ODs. 41.6% of respondents think the common cause of rare diseases was genetic, but 20.3% of people were unaware of the cause for rare diseases. Only 14% of respondents think that rare diseases occur by birth, while 48.5% thought they occur at any age. However, 19.3% of respondents did not know the age of onset. Some respondents think RDs effect India more than China, but 67.7% thought rare diseases affect all countries. An approximate time took to diagnosis a rare disease was equally spread from one week to five years. 36.2% of respondents thought that rare diseases affect only 1-2% of the population, while 30.3% of people did not know the affected population. Practicing medical professionals included were medical students, pharmacy, nursing, and practicing doctors. They represented 47.1% (247

Table 4: Awareness/non-availability of resources among healthcare professional.

Unawareness/Non-availability of resources among healthcare professionals (247)		
Key indicators	# of Respondents	Response %
Not seen any patient in their lifetime	83	42.8
No Resources to Diagnose RDs	150	77.6
No Resources to Treat RD Patients	155	80.1
No continuing education program in RDs in their Institutions	147	76.0
> 1 Scientific Publication to their name	217/599	36.3
Membership in a Professional Society	353/599	58.9

Table 5: Number of people identified rare diseases when given along with common and neglected diseases.



respondents) in the survey. From these healthcare professionals, 42.8% had not seen any rare disease patients. Thirteen rare diseases from the National Organization for Rare Disorders (NORD) website [26] and twelve common diseases and four tropical diseases were listed to identify the rare diseases (Table 5). 77.6% had no resources to diagnose, while 80.1% had no resources to treat rare disease patients, and 76% had no continuing education programs in their institutions (Table 4) (Supplementary Material: Graphic Representation of the responses).

Discussions

All the information from this survey is completely anonymous and does not collect personal or identifiable information about the respondents or institutions, and there is little or no potential for invasion of privacy or breach of confidentiality. This information is essential to assure the survey takers voluntarily complete the study. The respondents were informed before taking the survey that the information derived from the study would be reported as a public report or a journal publication. Also, the study may be made available to India's Ministry of Health and Family Welfare, and others who create programs and services for parents and caregivers. It is expected that the sample study will help to move the national conversation on care and support for rare disease patients and families and provide greater awareness of how rare diseases affect communities. Health budgets are insufficient to provide care for all. Allocating resources to treat a person with a rare and expensive disorder over a person with a more common, less expensive disorder is a problem for cautious policymakers who are concerned about justice and fairness in allocating their community health resources. Therefore, establishing the baseline knowledge of RDs in the country is essential.

Among all the respondents, one third have one or more scientific publications and have professional society membership. This number of professionals is very close to the respondents who indicated that they are medical professionals (students, physicians, etc.). This fact is considered as an indication that a subset of the respondents has some understanding of RD.

Among the respondents, the oldest and youngest generation appears to be less familiar than other age groups. NRIs who are generally expected to have a higher standard of living appears to have the same awareness as people living in less privileged places in India. The nonmedical respondents have the highest participation, whereas practicing physicians are fewer. This information is in line with the ratio of medical and non-medical participants. These results are in parallel to the familiarity of the awareness of RD among the respondents who are health care professionals and those participants with voluntary and, professional membership and publications. One-fourth of the participants have not heard the terminology of RD and OD. It may be that there are no programs in their medical and other educational curricula. Again, it appears that there is a correlation between the knowledge of rare diseases and awareness programs.

The definition of rare diseases varies from country to country. The three-fourths of respondents are not familiar with a definition for RD. The knowledge of the cause of rare diseases is variable; one-fourth people have no idea what causes the RD. More than half of the respondents are unaware of the number of RD and the number of people suffering from RD. Half of the people think the RD can occur at any age. The time to diagnose the RD is evenly spread from one week to five years.

The question relating to rare diseases population in the country, half of the respondents indicated that they either do not know the definition for a rare disease or do not know what Rare Diseases are. Similarly, half of the respondents do not know the number of rare diseases and the number of orphan drugs. Since 1983, many global companies started developing orphan drugs after the Orphan Drug Act implementation. There is none at this time in India, although in 2017, the Drugs & Cosmetic Act 1945 has been amended [10] to include "rare diseases and orphan drugs". Half of the practicing medical professional group has not come across the term RD, and three-fourths of them have neither facility to diagnose nor treatments for the rare diseases (Table 4). From the list containing rare diseases, common diseases, and neglected diseases provided to the survey takers, only 50% of people have correctly identified six of thirteen RDs (Table 5). Unlike the USA and other developed countries, there are not many patient organizations in India [27], and respondents indicated that their institutions do not offer rare disease awareness programs. This response most likely comes from medical professionals.

Out of 599 respondents, only 243 have some connection to healthcare. The lack of information on rare diseases or general awareness of its issues is not unique to India. The world's most populous country, China has the world's largest number of rare disease. However, these countries have no system of registering cases of rare diseases. Therefore, there is very little documented information on the epidemiology of those diseases in the most populous countries. Although developed countries have well-established policies, they are now recognizing the importance of developing countries [28]. India's exposure to rare diseases is limited, particularly international exposure. Policymakers in India are catching up to determine the status of rare diseases in the country. Patients and patient organizations do not have a voice. Therefore, they are not part of discussions for creat-

ing National Health Policy or in the review process of existing policy documents [29].

At the end of the survey, the respondents were asked to leave comments or suggestions. A good majority of the respondents have opted for awareness programs as the primary focus. It is said awareness brings cures, help policymakers to realize the importance of RD in the healthcare system. Many felt the awareness of RD is the first step for everything-for the definition, health care policy, diagnosis, treatment. Through education at all levels - college or professional education - the awareness could be brought through the public health. Funds could be collected for research from various sources-investors, companies, government, and philanthropy- suggesting private-public-partnerships is vital. The government should support the discovery and development of Orphan drugs [30]. Many respondents suggested including rare diseases in public policy as a priority issue. Interestingly some suggested involving the United Nations to make RD a global priority [31]. Diagnostics has taken the next priority after awareness programs. New programs are to be created to include rare diseases in the medical college curriculum. Many suggested that having seminars, workshops, and RD health camps in rural areas are important. Practicing physicians expressed concerns about not having practical experience. Medical and paramedical students expressed that they did not have much information about the rare disease. Some others, suggested incorporating distinguished lecture series in the university and a chapter in medical textbooks is mandatory. Some respondents suggested to creating databases on RD and OD. It is suggested to have specialized hospitals.

Conclusions

Given the current situation of rare diseases in India, knowing where India stands in this regard is an important first step for drafting the rare diseases policy and providing incentives to orphan drug development to improving the quality of patients and host of other things. The purpose of this sample survey is accomplished by obtaining baseline knowledge of rare diseases in the country. The survey is given to people with a minimum of a college education and people living in metropolitan cities and to Indian people living in the USA. The survey obtained information on issues such as definition, population, orphan drugs, education, and policies. The survey categorized two sets of people; one with limited knowledge of health, and others have no connection to health. The results indicate the awareness in all categories falls short. It is found that more than 70% of people in India do not have basic knowledge of rare diseases. Sixty percent of people with little or no formal education live in rural areas. Therefore, the number of people with no knowledge of rare diseases would increase substantially if the rural population is included in the survey.

This survey suggests that The government of India to drafting an RD policy document, defining the RD, and creating the Orphan Drug Act should give careful consideration should. The sample survey results are consistent with the expectation that India needs to do more to raise the awareness of rare diseases, for example, include the RD in the 2021 census.

Acknowledgment

We thank Mary Dunkle, National Organization for Rare Disorders for her suggestions, encouragement from the conception to the finishing of this paper. I sincerely acknowledge William Toscano, and George Maldonado, School of Public Health for their advice, Robert Straka, Experimental Clinical Pharmacology, for reading the manuscript and Danielle Carrigan, for technical assistance, college f pharmacy, University of Minnesota.

Disclosure

The authors have no conflict of financial interest.

References

1. A literature survey was conducted using search terms “Rare diseases + India” on PubMed and Medline (Jan 8, 2017). See Fig 1: The one citation in 1900-1950 is due to diabetes, in a decade, 1991-2000 only 228 citations and are gradually increasing since then.
2. "Orphanet” the worldwide reference database for rare diseases, lists 5,833 different entities. The European Commission estimates a total of up to 8,000 rare disorders in Europe.
3. Abbey Myers, founder of NORD as a representative of patients, and Dr. Marion Finkle, the first director of the FDA Office for Orphan Products Development USA, played a key role in the definition for rare disease. “We (the rare disease support group community) felt the definition should define orphan diseases as conditions affecting fewer than 200,000 Americans”.
4. The US Congress passed the Orphan Drug Act of 1983 to stimulate the development of drugs for rare diseases. The law provides seven-year marketing exclusivity to sponsors of approved orphan products, a tax credit of fifty percent of the cost of conducting human clinical testing, and research grants for clinical trials of new therapies to treat orphan diseases. The Food and Drug Administration administers the Orphan Drug Act and reviews applications for orphan designations. The Office of Orphan Products Development awards designations and administers the small grants program. The Center for Drug Evaluation & Research and the Center for Biologics Evaluation and Research reviews applications for marketing approval. Office of Inspector General, United States Department of Health and Human, Services. For the Orphan Drug Act implementation and impact.
5. Kwon D (2018) How orphan drugs became a highly profitable industry. The Scientist.
6. A registered not-for-profit organization in the USA and India. Its mission is to raise awareness, advocate public policy, promote orphan drug development and diagnosis.
7. Over the last fourteen years, IORD initiated several pro-

grams to raise awareness of RD. The former President of India, Dr. Abdul Kalam initiated RD awareness for the first time meeting in Hyderabad, 23 March 2014.

8. Pharmaceutical Export Promotion Council of India is the authorized agency of the government of India for promotion of pharmaceutical exports from India. It was set up under the provisions of Foreign Trade Policy by the Ministry of Commerce and Industry in May 2004. In 2014, the Pharmexcil organized a meeting on orphan drugs in Hyderabad where Indian government agencies and India pharma industry were brought to one stage and discussed the wants and needs of Orphan Drugs.

9. Overseas marketing Strategies, Opportunities for Orphan Drugs, 25th Sep 2013.

10. In 2017 Drug Controller General of India incorporated "Orphan Drugs" and "Rare Diseases" terminology in the Drugs and Cosmetic Act amendments.

11. National Policy for Treatment of Rare Diseases.

12. Longer wait for patients as the government calls its rare disease policy impractical.

13. Posada M, Ramírez A, Carroquino MJ, Messlich H, Schuster R, et al. E-Rare: Era Net for Research Programmes on Rare Diseases.

14. Yvonne Zurynski, Aranzazu Gonzalez, Marie Deverell, Amy Phu, Helen Leonard, et al. (2017) Rare Disease: A national survey of pediatricians' experiences and needs. BMJ Paediatrics Open 1: e000172.

15. (2005) Rare Diseases: Understanding this Public Health Priority. EURORDIS.

16. Rare diseases not only affect the patients but affect the entire family. The caregivers of people with rare and chronic conditions make physical, emotional, and financial sacrifices. The important role that caregivers have in the management of rare disease patients are surveyed.

17. Social media continues to be a useful tool for researchers looking to connect with rare disease patients for clinical studies.

18. Researchers turn to Facebook to survey rare disease patients for study.

19. Juggling care and daily life: The balancing act of the rare disease community.

20. "Rare Barometer Voices" is a EURORDIS initiative that aims to make the voice of rare disease patients stronger. It collects the views of the rare disease community and communicates as a unified voice to bring about change. Patient organizations can use their surveys to raise awareness among policy makers in their countries.

21. National Library of medicine includes activities in support of interoperable health records, development of a robust knowledge base for personalized health care and more.

22. NRI: Non-resident Indians who are migrants from India. They are, generally, educated with a minimum of college degree who have professional jobs with high earnings. The NRIs in the USA is one of the largest among the groups of immigrants with an estimated population of about 3.2 million, or ~1.0% of the US population.

23. Sixty percent of the Indian population live in rural areas and do not have access to email services. Although the rural population is not included in the survey by visiting a few villages, we observed that the burden of rare diseases is much more prevalent due to lack of education, poverty, consanguine marriages, etc.

24. In the Population Census of 2011, it was revealed that the population ratio in India is 940 females per 1000 of males.

25. India has doctors and patient ratio 1/921, which is way ahead of the suggested doctor population ratio 1:1000, by the WHO. The number of doctor population in India includes Ayurveda, Homeopathy, and Unani practitioners along with Allopathy doctors.

26. NORD's Rare Disease Database provides brief introductions for patients and caregivers of specific rare diseases.

27. Very few patient specific support groups are present in India. (Thanks to Steve Groft, Director NIH ORDR kindly provided this information) All most all of them are functioning in bigger metropolitan cities. Some of them have websites but may not have official NGO status, less than 0.2% of disease-specific NGO is registered).

28. For the first time, in 2015, Rare Diseases International (RDI), was created to include developing countries, in advocating for rare diseases to be an international public health priority. RDI essentially, turning the rare disease patient movement into an international one. It started with 60 patient organizations, including IORD and 30 countries, including India, in 2014. Its main objectives are to raise public awareness, influencing policy making and enhance the capacities of members to improve the lives of those living with and affected by a rare disease through information exchange, networking, mutual support, and joint actions and represents members and people living with rare diseases.

29. To draft rare diseases policy, the government of India appointed a committee consists of high-powered senior most people from prestigious institutions in India, but there is no representative from the patients even though there are estimated nine crores of rare diseases patients. Further, approximate costs of enzymes for enzyme replacement therapy – lysosomal storage disorders have been included. These enzymes are most expensive; none of the cheaper drugs from the approved ~ 450 orphan drugs for other rare diseases have mentioned.

30. To encourage drug development collaboration or collaborating with international organizations is a win-win situation for India. IRDiRC was created in 2011 to have a concerted effort to organize the rare disease research community and funding with the collaboration of NIH and EC. ZIts broad vision is to have an accurate diagnosis, care, and available therapy within one year of coming to medical attention. The consortium extends the membership to many funding organizations, investing more than 10 million USD over five years in rare disease research; and umbrella patient advocacy organizations (non-funding) representing broad patients' interests for all rare diseases in a country. The international rare diseases research community is eager to share knowledge and experience and work collaboratively across borders to bring diagnoses and therapies to patients. IORD became a member of IRDIRC in April 2017.

31. As rare diseases do not recognize country boarders, international rare diseases community is taking global action through the formation of the NGO Committee for rare diseases. It is a substantive committee established under the umbrella of the Conference of NGOs in Consultative relationship with the United Nations. Its goal is to promote rare diseases as a priority in global health research and social and medical care as part of the UN 2030 Agenda - the sustainable development goals. Its mission is to bring visibility and understanding about rare diseases to the UN, act as an advocacy platform for the issue of rare diseases creating a space for collaboration; collect and share accurate information about rare diseases and associated challenges.