

# AWARENESS OF RARE DISEASES AMONG YOUTH IN GUJARAT STATE

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## ABSTRACT

*Across the globe, the lives of individuals suffering from rare diseases are seen to profoundly impact their physical, emotional, social, and financial situations. It is therefore crucial to provide support networks and advocate for improved medical care and resources to improve their quality of life. Well before such attempts, it is quite important to check the level of awareness among people about rare diseases and their symptoms. The present study aims to assess the level of awareness among the non-medical young population of Gujarat state regarding rare diseases and the associated challenges. Through the use of a mixed-methods approach including binary, descriptive, and multiple-choice questions, the researchers gathered data from a diverse sample of respondents. The findings revealed a mixture of responses highlighting the complexity and nuances surrounding the understanding and awareness of rare diseases in the youth of Gujarat.*

**Keywords:** rare disease, WHO, awareness

## INTRODUCTION

The definition of rare diseases varies across countries, with the total number of affected individuals, prevalence, and lack of treatment availability being key elements. A formal definition aids in identifying diseases that require financial support for drug development and research. The World Health Organization suggests a frequency of less than 6.5-10 per 10,000 people as rare, while the USA defines it as affecting fewer than 200,000 individuals. In China, rare disorders are those affecting less than 1/500,000 people or with neonatal morbidity of less than 1/10,000. India lacks a standard definition, but the Organization for Rare Diseases India proposes a threshold of 1 in 5,000 people or less. Lack of a definition and diagnostic infrastructure hampers accurate statistics on rare disease incidence and prevalence in India.

Approximately 80% of rare diseases have a genetic basis, often monogenic, and around half manifest in early childhood. Conservative estimates suggest 70-100 million people in India may be affected by rare diseases, with many undiagnosed. The lack of approved treatments for 95% of rare diseases, the high cost of available treatments, and delayed diagnosis pose significant challenges. Rare diseases have a larger global impact than cancer and AIDS combined. The orphan drugs market is projected to reach around \$400 billion USD by 2025, yet only approximately 500 approved orphan drugs exist, with over a hundred in clinical trials. (About RD <https://ordindia.in/>, Evaluate Pharma® Orphan Drug Report 2017 4th Edition – February 2017).

The scarcity of epidemiological data on rare diseases in India hinders understanding the burden and establishing an accurate definition. The economic burden of most rare diseases remains unknown, and limited data from tertiary care hospitals show diseases like primary immunodeficiency disorders, lysosomal storage disorders, inborn errors of metabolism, cystic fibrosis, osteogenesis imperfecta, certain muscular dystrophies, and spinal muscular atrophy among the commonly reported conditions. Efforts such as the US National Institutes of Health's Undiagnosed Diseases Network aim to accelerate diagnosis and address these challenges.

Awareness about rare diseases in Gujarat, a state in India, is gradually increasing as evident from literature. However, significant efforts are still needed to improve understanding and support for rare disease patients. While progress has been made, continued efforts are required to strengthen rare disease awareness in Gujarat and ensure that affected individuals receive timely and appropriate support and care.

The lives of individuals suffering from rare diseases are profoundly impacted by the physical, emotional, social, and financial challenges they face. It is crucial to raise awareness, provide support networks, and advocate for improved medical care and resources to improve their quality of life.

Raising awareness of rare diseases is crucial for reducing stigma, improving access to medical care, and encouraging research and development of new treatments. Individuals can contribute by sharing information

on social media, participating in awareness-raising events, joining patient organizations, advocating for research and funding, and educating themselves and others about rare diseases. Educating oneself is particularly important when seeking rare disease health insurance to ensure adequate coverage. By taking these steps, individuals can make a meaningful difference in raising awareness and supporting individuals with rare diseases.

### **OBJECTIVE**

The present study aims to assess the general knowledge and understanding of rare diseases among the young population of Gujarat, especially among the college/university going students having no medical background.

### **METHODOLOGY**

The research methodology employed for this study involves the use of questionnaires as the primary data collection tool to gather information on the level of awareness about rare diseases in the target population of Gujarat. The questionnaires were designed to elicit responses related to general knowledge of rare diseases, specific disease awareness. A structured questionnaire format was utilized, consisting of multiple-choice questions, Likert-scale items, and open-ended questions, to ensure comprehensive data collection and facilitate quantitative and qualitative analysis of the responses.

### **DATA COLLECTION**

Questionnaires were utilized as the primary data collection tool in this research study to gather information from a large sample of participants efficiently and systematically. The questionnaires were designed to capture both quantitative and qualitative data, enabling a comprehensive understanding of the research topic.

A structured questionnaire format was employed, consisting of closed-ended questions with predefined response options, such as multiple-choice or Likert scale items, to facilitate data analysis and comparison. Open-ended questions were also included to allow participants to provide detailed and nuanced responses, providing deeper insights into their perspectives and experiences.

The questionnaires were carefully constructed taking into consideration the research objectives and ensuring clarity and relevance of the questions to elicit accurate and meaningful responses.

Three types of questions were used in the questionnaires - binary questions, descriptive questions and multiple-choice questions. Binary questions provide a straightforward and efficient way to collect data and can be useful in obtaining quick insights and comparisons within a given population. Descriptive or definitional questions are designed to elicit information or descriptions that provide clarity, understanding, or definitions of concepts, terms, or ideas. They aim to obtain specific and precise explanations from respondents in order to gather detailed information or to ensure a common understanding of the subject matter. Multiple-choice questions provide respondents with a set of predefined response options from which they can choose the most appropriate answer. Each option represents a different numerical value, and respondents select the option that they believe is the closest or most accurate estimate.

The questionnaires were distributed to participants through various methods, such as online platforms, email and in-person administration depending on the logistics and preferences of the participants. Clear instructions and guidance were provided to participants on how to complete the questionnaires, including any specific formatting or response requirements.

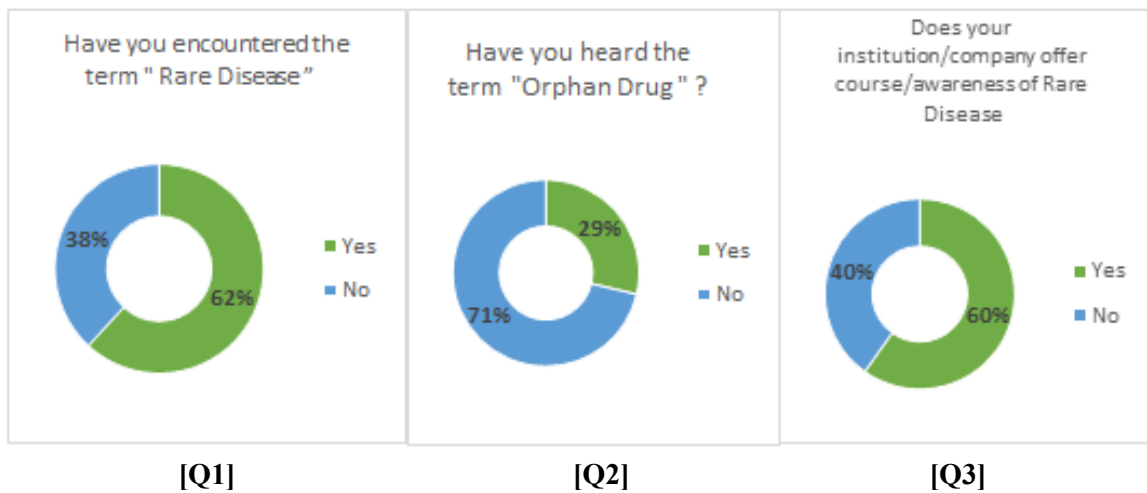
Efforts were made to ensure the anonymity and confidentiality of the participants' responses, assuring them that their data would be used solely for research purposes and treated with strict confidentiality. The data collected through the questionnaires served as a valuable resource for analysis, interpretation, and drawing conclusions in relation to the research objectives, contributing to the overall findings of the study.

### **DATA ANALYSIS**

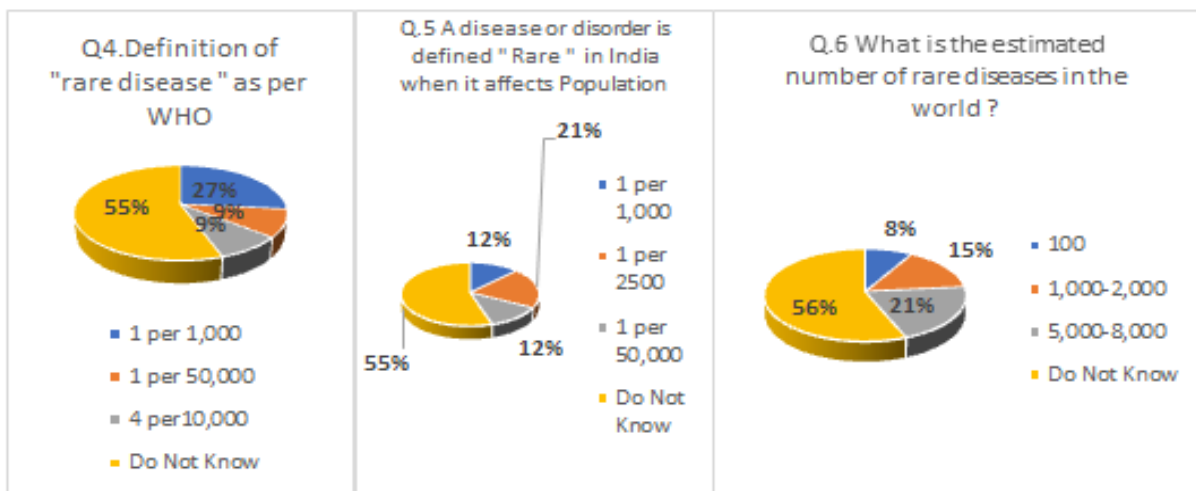
In the present study, Microsoft Excel has been used for data analysis as it is an extensively used tool in research for several reasons. It allows researchers to organize and manage large datasets efficiently, enabling easy data entry and storage. Secondly, Excel offers a wide range of built-in functions and formulas that facilitate data calculations, statistical analysis, and visualization providing researchers with essential analytical capabilities. Thirdly, Excel's flexibility and user-friendly interface make it accessible to researchers with varying levels of technical expertise, allowing them to perform basic to advanced data analyses without

the need for specialized software. Fourthly, Excel's compatibility with other research tools and software enhances data integration and collaboration among researchers. Lastly, Excel's ability to generate charts, graphs, and tables aids in presenting research findings in a visually compelling and understandable manner.

In the first part of analysis, the binary questions have been considered. The three binary questions which were asked to respondents were about whether they were aware of the terms rare disease and orphan drug, and if their institute or company provides any knowledge about rare disease. Majority of the respondents were found to have heard the term “rare disease”. However, most of them opined that they did not know the term “orphan drug” which is a related term with the term rare disease. When asked about whether their institute/company makes efforts to give them awareness about rare disease, majority again said “yes”.



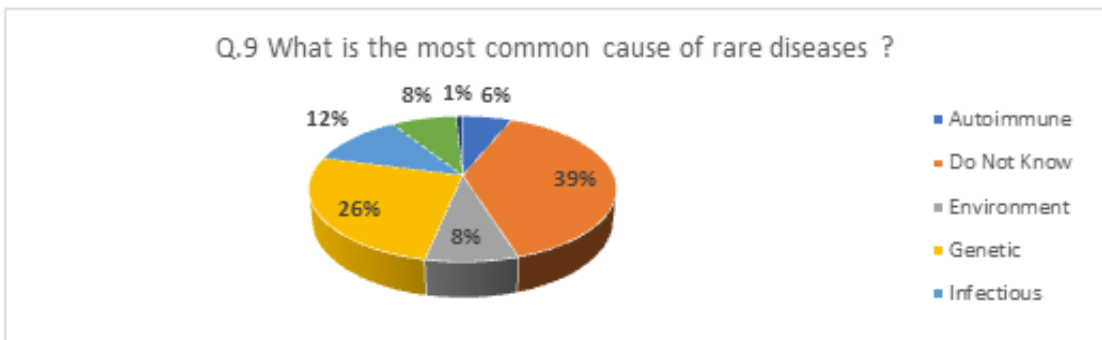
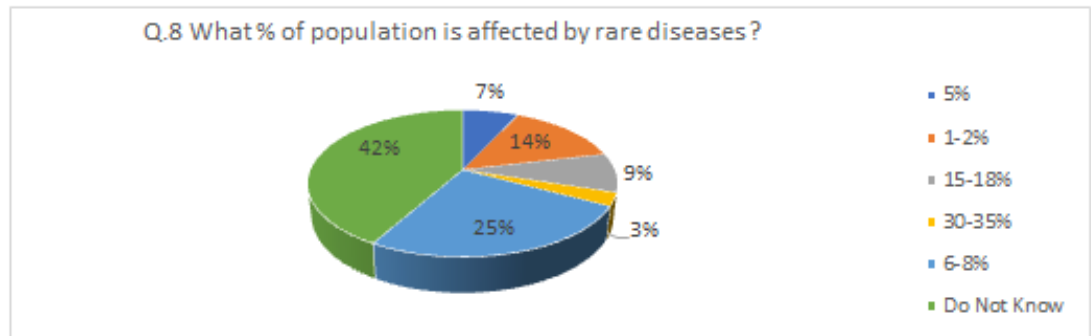
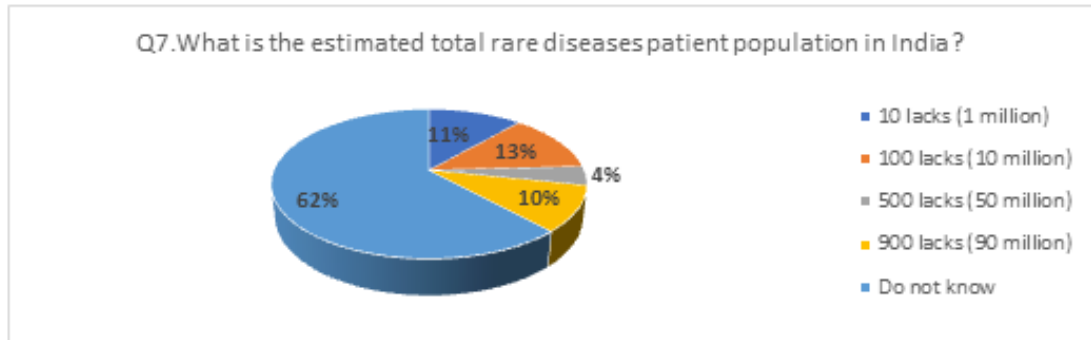
The mix of responses indicates that individuals within the surveyed population hold different viewpoints or exhibit different behaviours in relation to the specific questions asked. This could be due to the factors such as personal preferences, beliefs, or diverse circumstances that influence their decision-making. Analyzing the mixed responses allows researchers to identify the range of perspectives and understand the complexity and diversity within the population being studied. It highlights the need for further investigation and exploration to uncover the underlying factors contributing to the mixed responses and gain a more comprehensive understanding of the topic on hand.



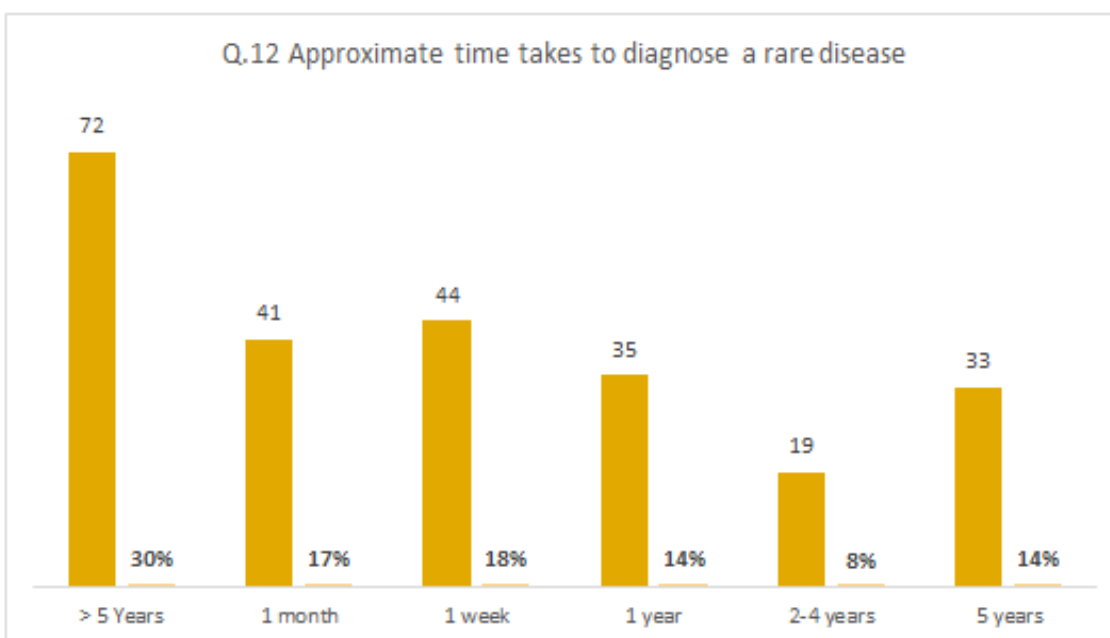
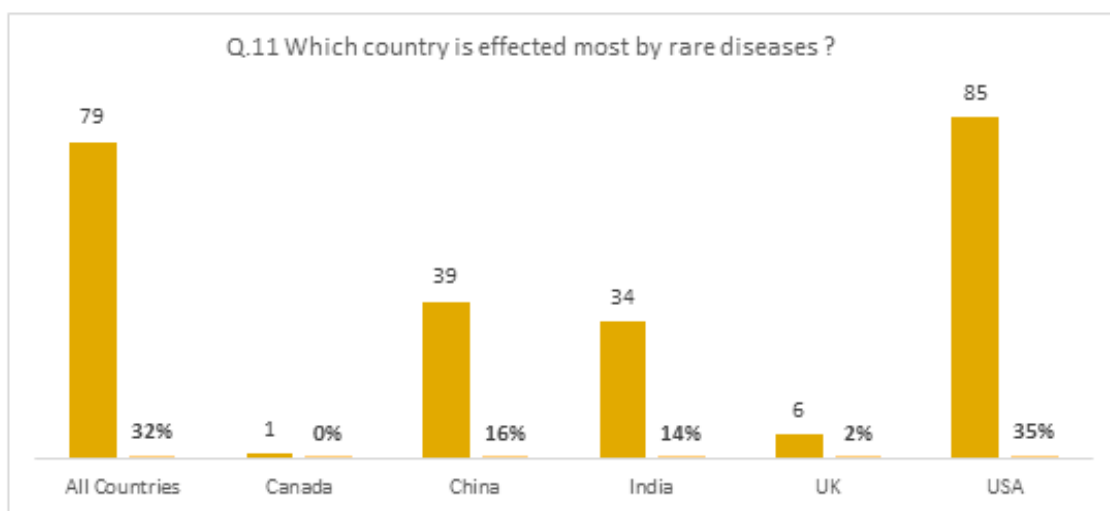
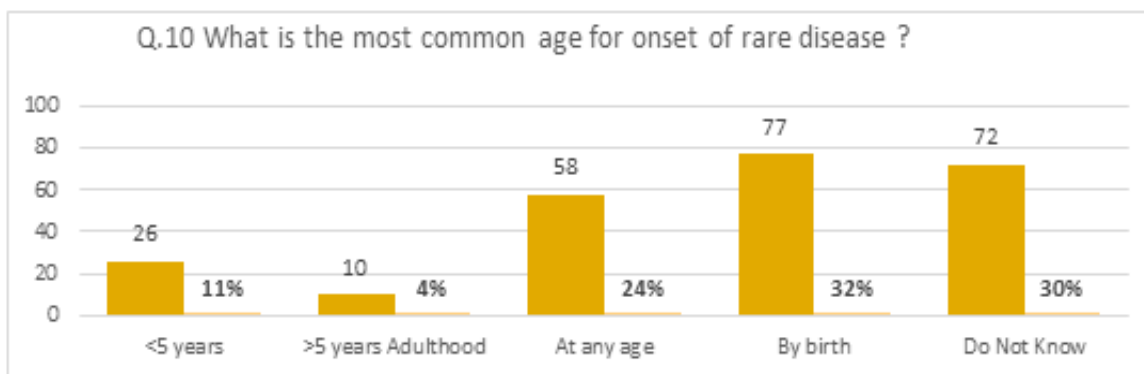
Questions 4, 5 and 6 are multiple choice questions asked to check the general knowledge of the respondents related to rare disease. About the meaning of the term “rare disease” according to WHO 55% of the respondents said that they are unaware. According to WHO a disease is called a rare disease when it affects a certain percentage of population in the country. When the related question was asked for India as a country again 55% respondents were found in the ‘don’t know’ category. A very less number of respondents gave

answer close to the right one. Our survey shows that majority of the respondents also do not know the approximate number of rare diseases prevailing in the world.

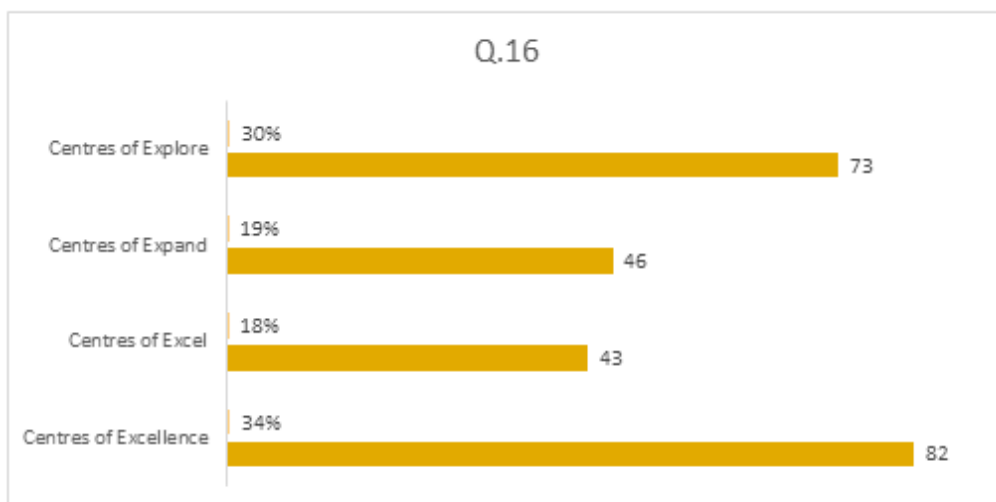
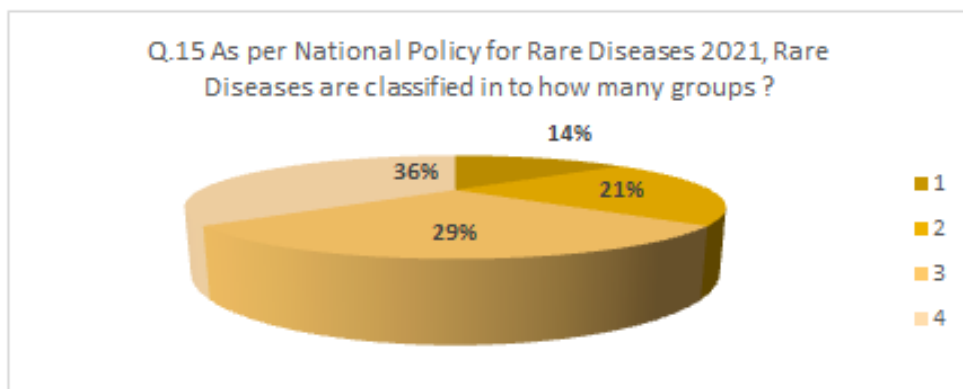
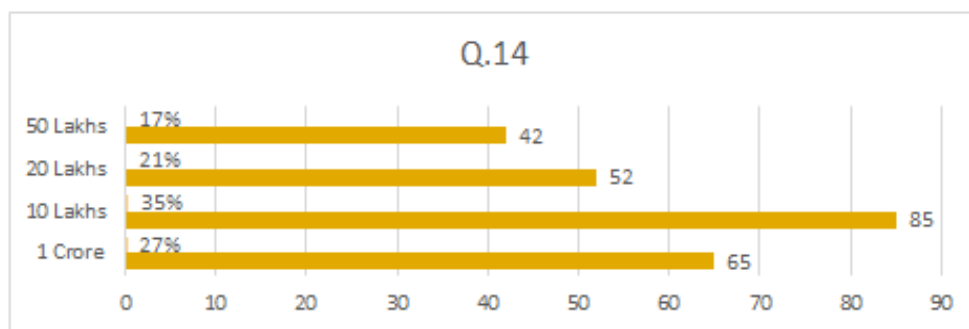
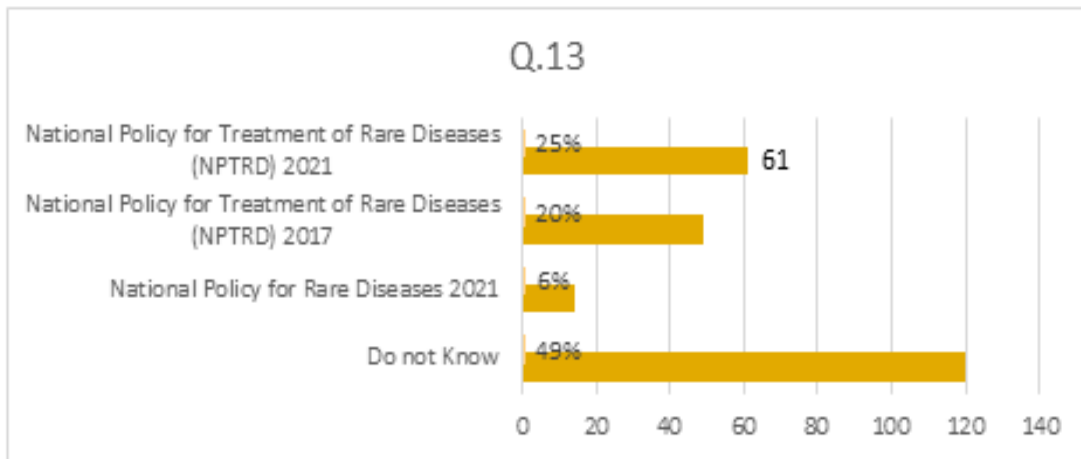
About the estimated total rare disease population in India, majority of respondents (62%) said that they did not have any idea about the same. When being asked about the percentage figure of rare disease affected population of the country the answers were quite skewed which means the respondents were not very sure of the answer. When they were asked about the common reason behind rare disease 26% of respondents believed that it is genetic while 39% said they did not know the answer.



When respondents select the "don't know" option in the majority of cases for multiple-choice questions, it can indicate several possibilities. Firstly, it could suggest their lack of knowledge or familiarity with the subject matter. Secondly, it might indicate ambiguity or confusion in the question itself, making it difficult for respondents to provide a confident answer. Additionally, the "don't know" option could be selected as a cautious response, where respondents prefer not to guess or provide inaccurate information. Analyzing such patterns of responses can provide insights into areas where further clarification or education may be needed, or where the question design and options may need improvement to elicit more accurate responses from respondents.



There is a mix response from respondents in questions 10 to 12. It highlights the need for further exploration and investigation to better understand the underlying reasons behind the diverse responses and to ensure a comprehensive understanding of the subject matter.





The presence of a mix response in multiple-choice questions serves as a signal that further exploration and investigation are necessary to delve into the underlying reasons behind the diverse responses. It indicates that there may be complex factors at play influencing the respondents' choices, such as individual perspectives, contextual influences, or varying levels of knowledge or understanding. By conducting additional research and analysis, researchers can uncover the underlying motivations or factors contributing to the mix response, allowing for a more comprehensive understanding of the subject matter. This deeper exploration is crucial in order to capture the full range of perspectives, identify any patterns or trends within the responses, and generate more accurate and meaningful insights. Ultimately, it enables researchers to refine their understanding, develop more targeted strategies, and make informed decisions based on a comprehensive understanding of the diverse responses received.

## CONCLUSION

In conclusion, the presence of mix responses in research findings from binary, descriptive, and multiple-choice questions highlight the diversity, complexity, and subjective nature of the surveyed population's opinions, perspectives, and behaviours. This variation in responses underscores the need for further exploration and investigation to better understand the underlying factors contributing to the mix of responses. It emphasizes the importance of considering individual differences, contextual influences, and personal preferences when interpreting research data. Analysing the mix responses allows for a comprehensive understanding of the subject matter, uncovering the range of perspectives and providing insights into the complexities of the topic. This thorough exploration ensures that research findings are robust, reliable, and accurately reflect the diverse perspectives and behaviours present within the surveyed population.

From the survey results it is quite evident that raising awareness of rare diseases in Gujarat is of utmost importance for several compelling reasons. Firstly, increased awareness can help reduce the stigma and misconceptions associated with rare diseases, fostering a more inclusive and supportive environment for individuals and families affected by these conditions.

Secondly, awareness campaigns can improve early detection and diagnosis of rare diseases in Gujarat. Many rare diseases often go undiagnosed or misdiagnosed, leading to delayed treatment and worsening of symptoms. By educating healthcare professionals and the public about the signs and symptoms of rare diseases, individuals can receive timely and accurate diagnoses, enabling them to access appropriate medical interventions and support.

Thirdly, raising awareness can facilitate better access to specialized healthcare services and resources. Due to the rarity of these diseases, individuals and families may face challenges in finding appropriate medical experts and support networks. By promoting awareness, Gujarat can establish networks of healthcare providers, support organizations, and resources dedicated to rare diseases, ensuring that affected individuals receive the care and support they need.

Moreover, raising awareness can spur research and innovation in the field of rare diseases. By highlighting the prevalence and impact of these conditions, it encourages scientific and medical communities in Gujarat to focus their efforts on advancing knowledge, developing new treatments, and finding potential cures for rare diseases.

Additionally, raising awareness can aid in the development and implementation of public policies and initiatives that address the specific needs of individuals with rare diseases. Through advocacy and awareness campaigns, Gujarat can prioritize the allocation of resources, funding, and support systems for rare disease research, treatment, and patient care.

Furthermore, increased awareness can empower individuals and families affected by rare diseases, giving them a voice and a platform to share their experiences, challenges, and needs. It creates a sense of community and solidarity, allowing individuals to connect with others facing similar circumstances, fostering support networks, and promoting mutual understanding and empathy.

Raising awareness also encourages the general public to become more educated and empathetic about rare diseases, fostering a compassionate society that values and supports individuals with these conditions.

Additionally, increased awareness can attract attention and support from philanthropic organizations, corporations, and government agencies, leading to increased funding and resources dedicated to rare diseases in Gujarat.

Furthermore, by raising awareness of rare diseases, Gujarat can contribute to the global efforts in understanding and addressing these conditions. Sharing knowledge, research, and experiences can contribute to international collaborations, advancements in medical science, and improved outcomes for individuals with rare diseases worldwide.

Ultimately, raising awareness of rare diseases in Gujarat can make a significant difference in the lives of affected individuals, promoting early diagnosis, better access to care, increased research efforts, and a more compassionate and supportive society.

### RECOMMENDATIONS:

1. The establishment of a Centre of Excellence dedicated to the treatment of rare diseases is a pressing need in Gujarat State, especially when compared to other states. The government's active involvement in this endeavour is crucial due to the lack of awareness surrounding rare diseases in the State. A dedicated Centre of Excellence would serve as a hub for specialized care, research, and awareness programs, providing much-needed support to individuals affected by rare diseases and their families. By centralizing resources and expertise, the government can ensure that patients receive timely and accurate diagnoses, access to innovative treatments, and comprehensive support systems. Such a centre would not only address the medical needs of individuals with rare diseases but also play a significant role in creating awareness, education, and advocacy, ultimately improving the overall quality of care for this vulnerable population.
2. Share personal stories and experiences of loved ones who have rare diseases, one can provide first-hand insights into the challenges and impact of these conditions. This can help raise awareness and foster empathy and understanding among others.
3. Support advocacy and awareness campaigns: People should get involved in local, national, or international campaigns focused on raising awareness of rare diseases. This can include participating in walks, fundraisers, or online initiatives that aim to educate and engage the public.
4. Utilize social media platforms: social media is a powerful tool for spreading awareness. We all should use platforms like Facebook, Twitter, and Instagram to share information, facts, and stories about rare diseases. Engage with relevant hashtags and connect with communities and organizations working in this field.
5. Educate yourself and others: We all should take time to learn about different rare diseases, their symptoms, available treatments, and the challenges faced by individuals living with these conditions. Share this knowledge with others, whether through conversations, presentations, or online resources.
6. Support patient organizations: There is a great need of research and connect with patient organizations focused on specific rare diseases or rare disease communities. These organizations provide valuable resources, support networks, and platforms for advocacy. Consider donating, volunteering, or participating in their events to contribute to the cause of raising awareness.

The present study which aimed to check the level of awareness among the youth of Gujarat about rare diseases has come up with the conclusion that the level of awareness about rare diseases in the state of Gujarat is quite low or negligible. Huge efforts are required in educating the people, especially the young folk, regarding the causes and effects of rare diseases. Sharing personal stories and experiences. Efforts like support advocacy and awareness campaign, utilization of social media platforms, educating self and other, and role of support organisations are some of the effective methods which must be used in order to level up awareness about rare diseases at least among the youth which is the biggest wing of the society.

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