Chapter

Rare Disease Advocacy Groups and Their Significance in Diagnosis, Management, Treatment, and Prevention of Rare Diseases

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Abstract

Rare diseases are those diseases that are not seen frequently in a population. There are about 7000 rare diseases that have been identified worldwide, and 80% of them are caused by genetic changes. Since a small number of individuals are affected with rare diseases, most clinicians are not aware of such diseases, and thus, they remain undiagnosed and untreated. Awareness regarding such diseases is essential to train clinicians to diagnose individuals affected with these disorders and to develop National/International Registries, which will serve to give information about the disease prevalence, its natural course, treatment, and management options available, to the medical fraternity. Patient advocacy groups play a remarkable and unique role in forming the collective voice of individuals living with rare diseases. They help in the identification, diagnosis, management, treatment, and prevention of such diseases. Advocacy Groups form collaborative partnerships with scientists studying such rare diseases, clinicians managing these diseases, pharmaceutical companies developing drugs, and Government officials overseeing and policy makers implementing medical regulatory processes. Thus, advocacy groups play a key role in helping patients and families with rare diseases.

Keywords: rare disease, patient advocacy group, rare disease registry, diagnosis, management, treatment

1. Introduction

A rare disease is so called because its frequency in any given population is very low [1]. There are about 7000 rare diseases that have been discovered, and more are being described in medical literature. Rare diseases have different causes, and about 80% of them have a genetic basis that could be chromosomal or genomic [2]. Rare diseases also include certain rare infections, cancers, and even autoimmune disorders. A rare disease is defined differently in individual countries and is based on the following parameters:

- The total number of people affected by the disease in that country
- Prevalence of the disease in that country
- Non-availability of treatment for the disease in that country

The USA defines a rare disease as a condition that affects less than 200,000 people. The definition of rare diseases as is defined in the USA was coined in the Congress during the Orphan Drug Act of 1983. Such diseases also came to be known as Orphan Diseases as drug companies were indifferent to adopting the research and manufacture of novel drugs for their treatment [2, 3]. The World Health Organization defines a rare disease as a disease with a frequency of less than 6.5–10 per 10,000 people. In Europe, it is defined as a disease seen in less than 5 of every 10,000 people, while in Australia, it is taken as one in 10,000 individuals, and in India, it is taken as one in 5000 individuals [4, 5].

A rare disease in isolation may affect a small population because of which clinicians are not aware of the disease and their symptoms, hence such individuals remain undiagnosed and untreated [1, 6, 7]. Although individually these disorders are rare, when taken together the people affected with rare diseases constitute a large population of the country. Such diseases are debilitating and without a proper diagnosis may cause gross morbidity and mortality, thereby posing a challenge to the healthcare system of the country [8]. A rare disease has an adverse impact on the everyday lives of the whole family and their care givers [9–12]. The cost of treatment/management is high and causes considerable financial burden to the individuals and their families [8, 11, 12], as there is a lack of Government policies regarding this aspect, hindering their treatment.

2. Advocacy groups

Since several rare diseases are being diagnosed and brought into light, it is required that more time and effort should go into research for understanding and preventing such diseases. A remarkable and unique aspect of rare disease treatment and management is the evolving role of advocacy groups and their collaborative partnerships with scientists studying such diseases, pharmaceutical companies developing drugs, and Government officials and policy makers overseeing medical research and health care [3, 6]. Rare disease advocacy groups have played a vital role over the years in the adoption of public policies, relocation of available research funding, and other factors affecting the research for rare diseases [1, 7]. In most settings, the rare disease advocacy groups are created by the family members of the affected individuals. They are the ones who look into the formation of public policies, help fast-track treatment approvals by regulatory bodies, and facilitate the welfare of individuals and their care givers.

The National Organization for Rare Disorders (NORD) in the USA was one of the first advocacy groups to be formed, followed by Rare Diseases International, which is a global alliance of patients with rare diseases across various nationalities and is dedicated for supporting treatment and formulating policies for rare disorders. Apart from these, patient advocacy groups have been formed all over the world, which individually or in alliance help to alleviate the various problems faced by individuals with rare diseases and to pressurize companies and countries to provide life-saving drugs and at a reasonable cost.

3. Patient advocacy groups in India

There are many patient advocacy groups currently active in India. The Pompe Foundation started by Mr. Prasanna Shirol in 2008 and caters to individuals and families affected with Lysosomal Storage Disorders. The Metabolic Errors and Rare Diseases Organization of India (MERD), founded by Mr. Vikas Bhat, promotes awareness regarding Inborn Errors of Metabolism and newborn screening. Both these organizations have been founded by parents having an affected child. There are a few other advocacy groups for Spinal Muscular Atrophy (SMA), Spino-Cerebellar Ataxia (SCA), Duchenne Muscular Dystrophy (DMD), and Osteogenesis Imperfecta. Twenty-five such organizations together joined hands to form the Organization for Rare Diseases India (ORDI), which is actively involved in helping patients and their families through the involvement of NGOs. These advocacy groups, however, need to be better organized, so that they can obtain and disseminate information about diseases, diagnostics, and treatment avenues to the affected families.

India has a huge diversity in the kind of rare diseases seen in different states, which can be attributed to certain cultural practices such as consanguinity in South India and endogamy in the North [13]. Based on the data from these organizations, a Rare Disease Registry has been initiated. This has helped in re-classifying rare diseases based on their prevalence in different states. Diseases like β-thalassemia are more prevalent in Punjab, Gujarat, West Bengal, Odisha, and Andhra Pradesh but are rare in other states [14, 15]; hence, they cannot be classified under rare diseases in these states. Similarly, house-to-house survey carried out by Molecular Diagnostics, Counseling, Care and Research Center (MDCRC) Coimbatore estimates that the prevalence of Duchenne Muscular Dystrophy in Tamil Nadu is high and cannot come under a rare disorder in that state [16]. Gradually, such data need to be combined, so that advocacy groups can focus their efforts on rare diseases and would help in developing a comprehensive and factual National registry, which would further aid in framing the National Policy for Rare Diseases [17, 18].

4. Multi-specialty hospital-based advocacy group

In this chapter, we would like to highlight our study that was to evaluate the feasibility of initiating an advocacy group for rare diseases in a multi-specialty hospital setting with the support of the Department of Genetics and Molecular Medicine. The genetic counselors were instrumental in liaising between different departments such as pediatrics, nephrology, neurology, orthopedics, and oncology for the identification of patients with suspected rare disease. Around 200 such patients were identified during the period of April 2016 to April 2019. The patient families were encouraged to register with the advocacy group, which would support and follow-up the patient and their families and provide the necessary management and treatment options as required.

Patients evaluated were identified and categorized based on age into the pediatric and the adult age group. About 63% of the patients were in the pediatric age group, and the remaining 37% were in the adult age group.

4.1 Rare disease advocacy group at Kamineni Hospital

To cater to patients affected with such diverse diseases, the first hospital-based advocacy group was created at Kamineni Hospital, a multi-specialty hospital located in the cosmopolitan city of Hyderabad in South India. It was named Maitri,

which originates from the Sanskrit word meaning "friendship." Maitri looks into the collective interests of individuals with rare diseases. The rare disease community is often denied the most basic of rights. Society is ill-equipped to understand the cause and gravity of the diseases. This often leads to a number of psychological problems. A diagnosis is important to understand a disease, its progression, symptoms, possible treatment options, and also for its prevention in future generations. Most individuals can lead a normal life. However, due to the lack of awareness, such individuals are not allowed to do so. People in general lack the sensitivity to accept and work alongside individuals suffering from such diseases. Maitri aims to change this scenario by raising awareness among clinicians, the general public, in schools, and colleges. It also looks into extended family screening and counseling for making informed reproductive decisions.

4.1.1 MAITRI (bond of friendship) – Kamineni Hospital's rare disease advocacy group

The Department of Genetics and Molecular Medicine along with its team of genetic counselors created Maitri to collectively address the various problems faced by individuals suffering from rare diseases.

Objectives of Maitri:

- To raise awareness about rare diseases among healthcare professionals, general public, and policy makers
- To evaluate and diagnose an individual with a suspected rare disease
- To identify individuals with rare diseases at Kamineni Hospital and include them in the Kamineni Rare Disease Registry
- To counsel the patient, parents, and family about the disease and its prognosis, management, and treatment options (if any)
- To identify individuals in the extended family at risk of having the disease
- To create self-help groups of individuals with similar symptoms/problems for discussion and possible management options
- To help cope with psychosocial issues
- To conduct parental/family group sessions about the different rare diseases and on the various psychosocial problems faced by them
- To involve teachers from the pre-school level, school, special schools, and college level in the advocacy group. To create awareness among them and other teaching staff about dealing with an individual affected with rare diseases to integrate individuals into the mainstream schools/colleges and allow them to lead a normal life.

4.2 General workflow of Maitri

Every patient visiting the Department of Genetics and Molecular Medicine was evaluated in two or more sessions as a part of primary evaluation (F**igure 1**).

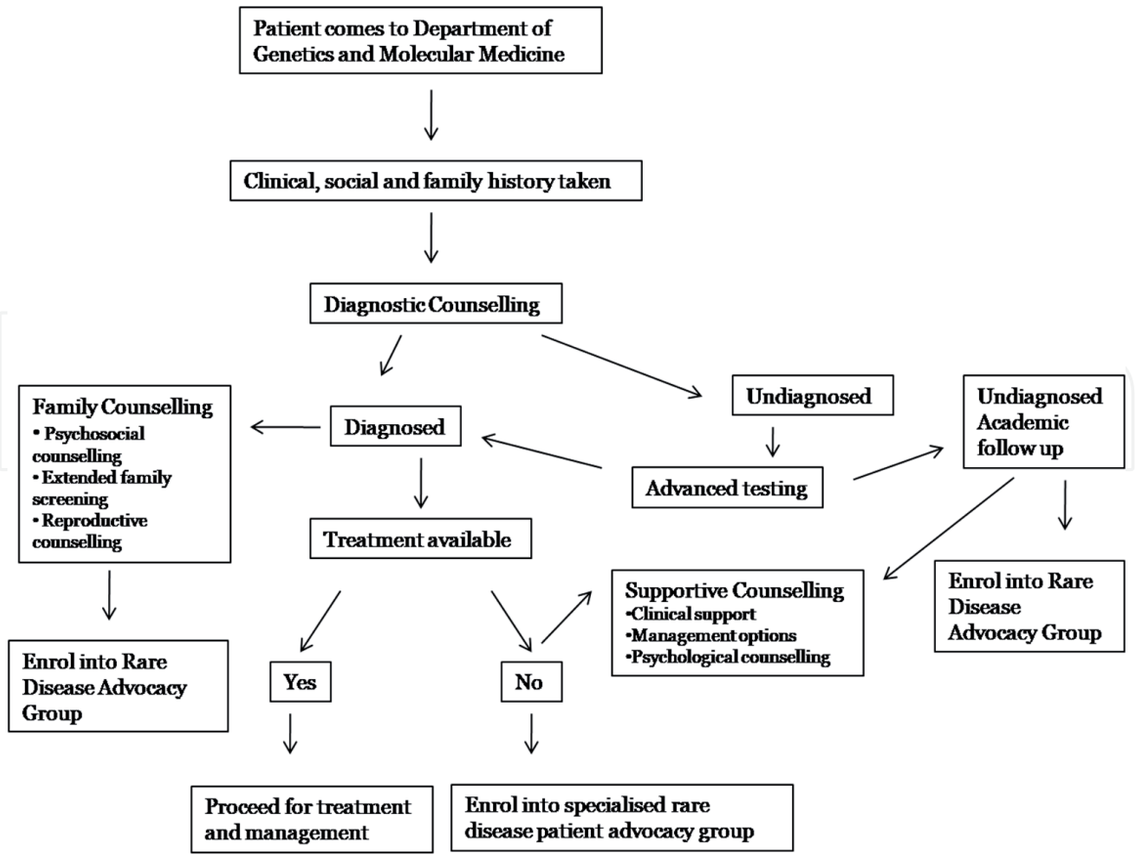

Figure 1.
Flowchart for the working of Maitri.

Patients were segregated based on the following age groups and advised accordingly.

a. Pediatric age group:

Age 0–5 years: Parental discussions on symptom management, addressing various needs of the child. Refer for clinical follow-up and psychological evaluation of the child prior to school admission.

Age 6–15 years: Addressing psychological needs of the patient. Group discussion programs with teachers, parents, and other healthcare professionals. Address issues faced at school and mitigate it.

b. Adult age group:

Age 16–22 years: Patient and parental psychological consultation to address various psychosocial issues. One-on-one patient meetings with geneticist and psychologist to address puberty-related problems. Reproductive counseling is also given.

Age 23–40 years: Pre-marital, pre-pregnancy genetic counseling, group sessions with other patients and their family to address common issues. Refer for clinical follow-up if needed.

Age > 40 years: Group sessions for patient and families for discussion of symptoms, their management, possible treatment options, and psychosocial problems faced due to the condition. Pre-symptomatic genetic testing and subsequent advice for children of individuals affected with rare diseases.

- Individual patients requiring vocational, behavioral, or occupational therapy are referred for the same

The importance of a rare disease advocacy group at the National and International level has been established by many esteemed clinicians, geneticists, and social workers. However, such groups at a hospital setting are important in a country like India, where there are limited electronic medical records, and there is huge literary and financial disparity in the population, such advocacy groups may contribute to maintaining crucial information for providing better healthcare and support to patient families.

5. Conclusion

Hospital-based Rare Disease Advocacy Groups like Maitri are crucial in a number of ways they help in establishing prevalence of rare disorders through the hospital-based registries. They promote awareness, so that every affected individual may be tested for a diagnosis. Such advocacy groups help bring together families with same, similar, or even different rare diseases, so that they can help and support each other. Groups like Maitri also perform a very important role in extended family screening, wherein they identify individuals at risk of developing the disorder and are counseled regarding appropriate testing and preventive measures. It also encourages reproductive genetic counseling for the families with an affected individual, which would help prevent the disorder in the future generations.

Advocacy groups along with policy makers are instrumental in creating public awareness about such diseases. Increase in awareness would make their prevention a public health concern, thus making it mandatory to have definitive screening and preventive strategies in the country. Not only advocacy groups are important for the support of individual families, but they also play a role in mass awareness and prevention of rare diseases.

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Conflict of Interest

There was no conflict of interest for the given project.

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