Enabling the disabled: Call for intercepting disability surge in Pakistan

Nadia Naseem¹, Usman Jawad²

Departments of ¹Morbid Anatomy and Histopathology and ²Family Medicine, University of Health Sciences, Lahore, Pakistan

ABSTRACT

Muscular dystrophies have always been marginalized culturally and socially, particularly in underdeveloped and developing countries. Pakistan is facing significant dearth of “appropriately” trained neurologists and neuromuscular pathologists. A careful and methodical link between clinical, pathological and molecular analysis must be established before making a diagnosis in such cases. Increased collaboration between local and international neurology societies may add value to collaborative neuromuscular research and education in Pakistan, both in community and health sectors. These goals are all achievable but require persistence, dedicated workforce, and positive efforts to meet them.

Keywords: Disability, health policy, muscle dystrophy

Introduction

The occurrence of muscular dystrophy (MD) is worldwide, and there is no country or zone on Earth where people are not affected by some type of MD. It is regarded that MD, in general, has an incidence of close to 1 in every 7250 males aged 5–24 years. The prevalence of Duchenne MD (DMD) is reported to be three times higher than the prevalence of Becker MD (BMD).¹ DMD prevalence (per 10,000 male individuals) suggest estimates of 0.1 in South Africa to 0.5–1.0 in Asian countries.²

MD has always been marginalized culturally and socially in most countries, primarily in those that are underdeveloped as rightly, the main focus of pediatric medicine in developing societies is on the treatment of infectious diseases and conditions related to poor nutrition.³ Increasingly, as the health status of communities in these countries improves, the focus is shifted to improving the quality of care and life for patients with chronic disorders or genetic conditions.

Pakistan is a developing country rich in ethnic and cultural diversity in its four provinces. It is listed as a country with the sixth highest burden of disease by the World Health Organization (WHO). The present national infrastructure of health facilities is equipped with 945 hospitals, 4755 dispensaries, 562 rural health centers, and 5349 basic health units. There are 1452 patients for every available bed, which is far behind the country requirements.⁴ Even though a steep rise in the number of registered doctors has been seen over decades, there is still only one doctor and one nurse available for every 1254 and 2671 patients, respectively. In the field of neuromuscular diseases, a significant dearth of “appropriately” trained neurologists and limited resources has further worsened the condition. There are only 120 qualified neurologists in Pakistan for the population of 180 million hence the ratio comes to 1:1.5 million people. As the bulk of patients show up at government-based hospitals that already are underequipped and technologically limited, both the physicians and patients are facing huge hindrances in appropriate diagnosis and management of these patients.⁵,⁶

As reported by the WHO, as compared to $34 USD per capita, only $18 USD per capita is available for offering health-care delivery in Pakistan that is seriously compromising the quality of life of individuals.⁷

Address for correspondence: Dr. Nadia Naseem, Department of Morbid Anatomy and Histopathology, University of Health Sciences, Lahore 54000, Pakistan. E-mail: drnadiaf@hotmail.com

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Subsequently, the major difficulties faced by patients with DMD in Pakistan are:

- Lack of information and access to guidance to recognize DMD as a disease
- Scarcity of trained health practitioners
- Inaccessibility of diagnosis; genetic tests are almost unreachable in terms of availability and price, the cost of a polymerase chain reaction test for 22 exons of the DMD gene is about $333.00 USD. Thus, the diagnosis of this disorder is almost unaffordable for families generating per capita income of only $1513 per annum in Pakistan; the citizens of a country fighting its economy in the league of low- and middle-income countries of this world[9]
- Besides, genetic testing, other expenses that family should cover are physiotherapy sessions, steroid treatment, surgery and special education among others. Some of these services are currently provided by nonprofit organizations because public institutions lack optimal management programs[9]
- The information available on the disease is insufficient, as well as the diffusion of this information. For this reason, majority of the people affected by MD, living in developing countries, are not aware of the differences among dystrophy types, its development, evolution, life expectancies, or how it is inherited, as well as how to manage and care for it
- Many of the people it has affected have seen their quality of life diminished and suffer from complications that can lead to death due to ignorance and lack of information, which is very widespread at both the public and medical level. On the other hand, they do not have access to advanced tests of diagnosis, which would allow them to know the specific type of MD they have, and with it to be able to know what the future will bring and the risk of passing the disease on to their children.

In such cases, most of the diagnostic, therapeutic and rehabilitative services are being offered at only through collaboration with nongovernmental organizations and charity works. It is needless to reiterate that we are poor not only in financial term but also in behaviors in dealing with and supporting to the disabled people of any sort that live in our surroundings. We as a part of the first-ever comprehensive project dealing with not only the detailed clinical but also histological and molecular profile of MD patients in Pakistan considered DMD a model disease to study this changing pattern of emphasis.

Our Project: Enabling the Disabled (Phase I)

Over a period of 4 years (January 2011–2015), we studied 343 patients (2.5–32 years of age) suspected clinically of having MDs [Figure 1]. We devised an algorithm, taking immunohistochemistry (IHC) as a gold standard for diagnosis and classification of muscle disorders with special emphasis on its applicability in the developing countries with economic constraints. This algorithm was more meaningful for the diagnosis of female patients (100%) where molecular analysis fails to yield any conclusive results in most of the cases based on amplification of all target exons in case of carriers or presence of very small point mutations that are detected by laborious and costly molecular methods.

Therefore, studying patients with a wide range of biopsy findings and clinical phenotypes in the present study, aided not only in approaching the pathological diagnosis of abnormalities of all dystrophin-associated glycoprotein complex proteins by IHC but also helped to determine the indications for employing a complete immunohistochemical panel and gene mutation analysis before making a final diagnosis in cases where molecular characterization is also unable to classify the mutation pattern. Furthermore, studying and relating the sociocultural aspects of this disorder, we came across several eye-opening facts kept under covers of ignorance by the community, health-care providers, and the government as a whole.

Limitations

- The delay in diagnosis turned out to be a significant problem that could be avoided if DMD is considered a possibility in patients presenting with developmental delay and muscle weakness in the initial stages
- In addition, there are limited financial and/or infrastructural facilities available and need to give priority to other more prevalent medical problems by the stakeholders of health-care delivery in our set up, makes our efforts further restrained
- The limited opportunities for comprehensive investigations and active management in our patients might have been due to the perception among some families and medical professionals that DMD/BMD is a terminal disorder. Hence, there was a noteworthy reluctance to subject these patients to further tests or procedures
- As most of the patients belonged to the poor socioeconomic class and reside in the rural areas of the province, hence, the role of general health practitioners in the guidance of families is of paramount importance to:
  - Help families to understand and accept the disease
  - Obtain a health managing plan for patients
  - Referring them to specialist labs for confirmatory diagnosis
  - Guide and refer them to geneticists for planning the family and/or prenatal diagnosis
- These issues are often addressed by genetic counselors; nevertheless, access to genetic counseling and multidisciplinary management are not frequently fulfilled
in developing countries, and they are also difficult to accomplish in developed ones.

**Future Goals**

The future of muscle dystrophy is not so dusky today. These recommendations spell out to assess and monitor the actual status of dystrophies in the local population and to promote dissemination activities and trainings for health practitioners that, in turn, will lead to technology transfer for maintaining the implemented services working in a sustainable manner; subsequently, helping children with DMD/BMD to live into adulthood. The researchers believe that adequate treatments will soon be developed and made available to the poorer of the poor with the help of modern science and technology that are well marketed in the industrialized countries.

**Call for Phase II**

The researchers in this project have built a consensus among them for the dire need of creating awareness among people, parents, families, health-care practitioners, geneticists, pathologists, and laboratory workers about what actually the MD is?

- We propose “employing a complete immunohistochemical panel and/or gene mutation analysis (cost-effective technique) before making a final diagnosis on routine basis”
- Besides making an earlier diagnosis, this study also indicates the essential requisite for
  - Genetic counseling and family support
  - Prenatal diagnosis
  - Assistance of the disabled and their families in their daily life movements
  - Surveillance for complications for patients with DMD/other dystrophies and their families; the already underprivileged stratum of population
  - Priority and Focus of the stakeholders of health-care delivery in our set up.
- Finally, there is a colossal requirement to establish a “State of the Art” Screening and Diagnostic Laboratory for Myologic Pathology in Punjab Pakistan offering services at both basic and advanced levels. This can only be possible when overseas neurologists can contribute through their visits and educational programs in Pakistan. Increased collaboration between local and international neurology societies may add value to collaborative neuromuscular research and education. These challenges cannot be met without workforce and funds. The government must contribute by increasing the average health expenditure, which is currently too low. These goals are all achievable but with persistence, dedicated workforce, and positive effort to meet them.

The profile of this disease cannot be raised enough without public awareness and advocacy.
- We cannot raise funds without awareness
- Without funds, there is no research
- Without research, there are no treatment options
- Without options, there is no cure
- Without cure, there is no more hope for life.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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**Conflicts of interest**

There are no conflicts of interest.

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