

Background

In the past few years patient registries played an essential resource; for understanding the natural history of many hereditary neuromuscular disorders and the development of clinical trials in many countries worldwide, allowing access for mutation specific therapies.

On the contrary patients in Africa and some middle East countries still lack awareness and knowledge on the multidisciplinary care for such heterogeneous group of disorders.

This report is to highlight the impact and challenges of the “Egyptian neuromuscular registry” as an example in developing awareness in the last seven years.

The Egyptian neuromuscular registry

Egypt is an African country of 100,000,000 population with very high consanguinity rates and big family members, due to cultural background.

The EN registry is the first registry in Egypt, Africa and Arab countries specific to neuromuscular disorders.

The registry was founded in 2012 through voluntary work in collaboration with TREAT-NMD Alliance. The ENR is now hosted by Myo-Care National Foundation, after receiving approval of the ethical committee.

The data base was set for nine different hereditary neuromuscular disorders (DMD/BMD, LGMD, SMA, GNE, FSHD, CMD, DM, CMT and CMS), accepting patients from Egypt, Arab and African countries.

In the last seven years the ENR could establish strong collaboration with International and National organizations which helped in developing the registry role; (1-TREAT-NMD international Alliance, NCNP Japan, LAND Lebanon, Egyptian muscular dystrophy patient organization Egypt, Egypt Air Hospital and other NGOs’ in Egypt.

The Main Objectives of the Egyptian Neuromuscular Dystrophy:

- 1- Establishing structured dataset that includes social-demographic data, clinical features, genotype, and biomaterial data.
- 2- Raise awareness on the clinical features and standards of care for each disorder.
- 3- Incorporating patients in the community.
- 4- Advocating at national health authorities for patients’ rights to access of the new therapies.

Role of networking with national and International organizations

Registry data base

According to the geographical site of Egypt in Africa and Middle East we received patients from other countries in the region.

To date the registry have enrolled 950 patients (850 from Egypt and 100 patients from other African and Arab countries, including the expanded SMA data set since 2018.



Core Dataset for TREAT-NMD SMA Registries

The Egyptian Neuromuscular registry contributed with TREAT-NMD from May 2017 to August 2018, in a project which was carried out to expand the core dataset for SMA Registries affiliated to the TREAT-NMD network. This work was done so that the TREAT-NMD Global SMA Registry can: 1) better inform on the natural history of SMA, 2) provide context to understand the safety and effectiveness of new treatments, 3) support post marketing surveillance (PMS) for those new treatments.

Contribution to international pharmaceutical enquiries

Working with TREAT-NMD the registry Contributed in more than 10 pharmaceutical enquiries, TREAT-NMD meetings as TGDOC member and in the DMD/SMA master classes which allowed better development of the data set and expanding the role of the registry to work on the standards of care awareness nationally.

Advocacy for patients access to new therapies

Working with the Egyptian Muscular dystrophy patient society in 2018 we formed advocacy group for the rights of the patients to benefit from the Egyptian disability law in the Egyptian parliament and pressuring the health authorities on the importance of patients access to new therapies.

In April 2019, we could grant approval from the Egyptian ministry of health for the first Egyptian DMD Patient to receive the new approved therapy “EXONDYS 51” in Egypt.



Working with media

Our work on the awareness included the media Through newspaper articles, YouTube videos and television programs.

<https://www.egypttoday.com/Article/6/40988/Muscular-Dystrophy-Patient-Approved-for-New-Therapy>
Sherif

Contribution to Disease awareness

Since 2014 in collaboration with the Egyptian muscular dystrophy patient society, Egyptian NGOs’ and internationally with the NCNP Japan, And LAND (The Lebanese association for neuromuscular diseases) the registry contributed in 12 medical convoys and awareness meetings in Cairo and remote Egyptian villages (Aswan, Edfo, Menia, Suez) for medical awareness and clinical consultation, genetic diagnosis for muscle disease patients and monthly physiotherapy and respiratory care awareness clinics.

The fact that most of the registry patients first language is Arabic, it was important to provide Arabic version of SMA standards of care family guide. The registry in collaboration with TREAT-NMD started.

International Awareness meetings

The registry actively contributed through invited lectures in number of international meeting highlighting the impact of patient registries on rare disorders in different countries (Kuwait, Alexandria, India and Iran)



Discussion

Although there are still many cultural and economic challenges related to the region as regards the society understanding of the importance of early management and screening of rare disorders specially for non treatable disorders, furthermore working on convincing the governments on the importance of including patients of hereditary neuromuscular disorders in the scope of the health insurance and access to new therapies.

The impact of the Egyptian neuromuscular registry through collaborating with national and international organizations changed the medical providers, patients and the society perspective towards rare neuromuscular disorders with the understanding of the importance of early diagnosis, rehabilitation and implementing the multidisciplinary care according to the international consensus.

In the Future collaboration with more countries in the region with established data base for such disorders can help in better understanding of the nature and genetic features in the region furthermore help research and better care for patients in Africa and the Middle East.

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