PRELIMINARY FIELD-WORK OF BEST GUIDANCE DATABASE FOR TURKISH NEUROMUSCULAR PATIENTS

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INTRODUCTION

In Turkey, with a population of about 70 million, it is assumed that there are approximately 100.000 individuals affected by one of nearly 200 different neuromuscular disorders (NMD). The clinical evaluation needs to be confirmed with other parameters such as muscle biopsy, EMG and genetic testing. The major goal is towards the full treatment and prevention of these disorders. However, the prevention of any disease brings on a demand for a collaborative work between medical specialists, parent-patient organizations and the government. The Ministry of Health (MOH) is officially responsible for implementation of health policies and delivering health care services nationwide. Public institutions, non-governmental and private organizations contribute to providing health services. However, the educational guidance of patients and their relatives is as important as getting efficient healthcare services clinically, financially and locally .

Almost all NMD are considered to be among *'rare disorders'* some of which have a defined genetic background. Genetic analysis provides quick and precise diagnoses; thus has the most powerful impact towards both confirmation and prevention of these diseases. The number of genetic centres in Turkey have increased immensely during the last decade, most of which are located in the big cities of Turkey. Due to this rapid development, the major problem seems to be the lack of information both for clinicians working in small cities and for the families who needs to be directed to the nearest and most appropriate hospital or centre.

We have aimed to execute a fieldwork to locate all departments, centres and laboratories giving services for neuromuscular disorders, including physical examination, EMG, muscle biopsy, other tests and genetic services in Turkey.

In this preliminary study, we have concentrated mostly on genetic centres since families with a hereditary NMD require genetic counselling, diagnostic testing, carrier identification and prenatal diagnosis. This data and information will be used to prepare a 'Best Guidance Database' and hopefully this work will lead to a detailed database providing the answers to frequently asked questions starting with Where?-Who?-How?-Why?-When?

APPROACH

The first step was taken by making a list of all the hospitals in Turkey. There are a total number of 1191 hospitals. A questionnaire was prepared to outline whether these hospitals had a neurology department, intensive care unit and if they performed EMG, echo, muscle and nerve biopsy, histopathology, electrocardiogram, PEG, respiration capacity, polysomnography, physiotherapy, hydrotherapy, surgeries.

Second step was to list all genetic centres and this work revealed that there are a total number of 31 genetic centres in Turkey which are recognised by the government. We have prepared a questionnaire limited for those NM disorders which are more commonly recognised in our population and having a defined genetic background. The questions involved Duchenne/Becker muscular dystrophy (DMD/BMD), spinal muscular atrophy (SMA), Friedreich's ataxia (FRDA), myotonic dystrophy type 1 (DM1) and 2 (DM2), facioscapulohumeral muscular dystrophy (FSHD), Charcot-Marie-Tooth disease (CMT), limb girdle muscular dystrophy (LGMD).

Our approach was to find out; 1. Which NMDs are studied by these centres; 2. If they provide a full service for clinical diagnosis, carrier identification and prenatal diagnosis and genetic counselling; 3. Are all these tests supported financially the government?.

RESULTS

Our first findings showed in a total number of 1191 hospitals, 836 are university and government hospitals and 355 are private hospitals. The data also helped to make an estimation that nearly 270 of university and government hospitals have a neurology department and the localization of these hospitals are mostly in big cities: İstanbul (52), Ankara (35), Izmir (26), Bursa (22), Konya (22) and Balıkesir (21). This data was drawn from the first feedbacks.

In a total of 31 genetic centres 10 are built in university hospitals, 3 are within the government hospitals and 18 are private organizations meaning that more than half (58%) of genetic services are provided by private centres (Figure 1). Unfortunately these are not supported financially by the government.

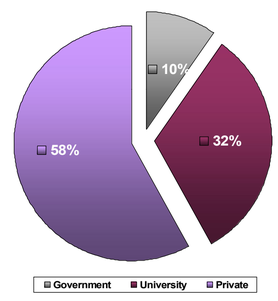

Figure 1. Distribution of 31 genetic centers according to their status

The localization of these centres are mostly in three biggest cities of Turkey ; 13 in Istanbul, 10 in Ankara and 4 in Izmir. Other 4 are in Eskisehir, Kayseri and Bursa (Figure 2).

Figure 1. Distribution of 31 genetic centers according to localization

Only 19 centres perform molecular analysis which is more widely used in diagnosis of NMD compared to cytogenetic testing. The questioner revealed that, DMD/BMD and SMA deletion analysis for postnatal and prenatal testing is routinely performed by 10 laboratories. Mutation analysis or carrier identification which require dosage analysis are only performed by 3 centres and only one lab performs point mutation analysis for DMD/BMD. Other disorders in question, FRDA, DM1, DM2, FSHD, CMT, and LGMD, are either carried out by research centres or by collaborative studies with other centres outside Turkey. All centres which are recognised by the government are giving genetic counselling however the research centres do not have the authorization to inform patients directly.

DISCUSSION

The future aim of this study is to prepare a 'Best Guidance Database' for families affected by NMD. Our preliminary results showed that, although there are research centres within the universities helping to meet the demands of clinicians and families, there is an urgent need for an increase in the number of centres performing genetic analysis for NMDs. The genetic counselling services cannot reach all families living in small cities at present but the government is taking steps towards employing genetic specialists in all governmental hospitals. The policies towards the financial support for all genetic testing are being standardized and hopefully the system will be implemented by the end of this year.

The data presented in this study will be enhanced with the help of all public institutions, governmental and private organizations and we will be able to implement an information network to make knowledge available to improve the quality of life of people affected by neuromuscular disorders.