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Muscular dystrophy disorders' prevalence in southern Libya: early observations and challenges as first new experience

Aaedah A. A. Senussi

Department of Medicine, Faculty of medicine, Sebha University, Libya

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ABSTRACT

A series of hereditary illnesses known as muscular dystrophy result in progressive muscle weakening and wasting. Muscular dystrophy can take many various forms, and the incidence rates vary depending on the type and the demographic being researched. The study's objective was to determine the prevalence of muscular dystrophy disorders in Libya's southern area, This study also aims to explain the difficulties and challenges faced by the Scientific Committee and its branches in other southern regions as a first experience with muscular dystrophy patients in terms of sorting and diagnosing these cases. 85 patients had highly suspected hereditary muscular dystrophy, 78 of the 85 patients had positive genetic results.

مدى انتشار امراض الضمور العضلي في جنوب ليبيا: الملاحظات والتحديات المبكرة كتجربة جديدة أولية

عايدة عبد الرحمن علي السنوسي

قسم الباطنة، كلية الطب البشري، جامعة سبها، ليبيا

الكلمات المفتاحية:
التحاليل الجينية
الضمور العضلي
الانتشار
سبها
مناطق الجنوب

الملخص

تؤدي مجموعة من الأمراض الوراثية المعروفة باسم ضمور العضلات الوراثي إلى إضعاف العضلات وضمورها تدريجياً. تلك الامراض العضلية الوراثية لها أنواعا مختلفة عديدة، وتتفاوت معدلات حدوثها تبعاً للتوزيع الجغرافي . وكان الهدف من الدراسة هو تحديد مدى انتشار امراض الضمور العضلي الوراثي في المنطقة الجنوبية من ليبيا، وتهدف الدراسة أيضاً إلى توضيح الصعوبات والتحديات التي تواجهها اللجنة العلمية وفروعها في مناطق أخرى من الجنوب كتجربة أولى مع مرضى الضمور العضلي الوراثي من حيث فرز هذه الحالات وتشخيصها. وأثبتت هذه الدراسة ان 85 مريضا مشتبه ان لديهم ضمور عضلي وراثي منهم 78 مريضا لديهم نتائج جينية ايجابية، تتنوع هذه النتائج بين امراض الحثل العضلي و ضمور الحزام الطرفي وغيرها.

Introduction

Neuromuscular diseases are a group of disorders that affect the muscles and the nerves that control them. These disorders can cause muscle weakness, wasting, twitching, and spasms, as well as difficulty with movement, breathing, and speaking. Neuromuscular diseases can affect people of all ages, from infants to the elderly [1-4]. Some of these diseases are genetic and run in families, while others may be caused by environmental factors or underlying medical conditions [5].

There are many different types of neuromuscular diseases, including: Muscular dystrophy which are a group of genetic diseases that cause progressive muscle weakness and wasting, common types of muscular dystrophy are:- Duchenne muscular dystrophy (DMD) :This is the most common type of muscular dystrophy in children, affecting approximately 1 in every 5,000 male births worldwide , Becker muscular dystrophy (BMD): is less common than DMD, affecting approximately 1 in every 18,000 male births worldwide and Limb-girdle muscular dystrophy (LGMD): The prevalence of LGMD varies depending on the subtype, but it is estimated to affect approximately 1 in every 14,500 people worldwide [6-8], In addition to the conditions mentioned earlier, there are many other types of neuromuscular diseases, including Charcot-Marie-Tooth disease, spinal muscular atrophy, myotonic dystrophy [12-13] , diagnosing neuromuscular diseases can be challenging, as the symptoms can be similar to those of other conditions. So, combination of physical exams, lab tests, imaging studies, and nerve and muscle function tests to make a diagnosis that confirmed by genetic tests [9-11].

It's important to note that these prevalence rates are estimates and can vary depending on the study and population being studied [13]. It's also worth noting that there are many other types of muscular dystrophy that are rarer and may have different prevalence rates[12].

*Corresponding author:
E-mail addresses: aae.senussi@sebhau.edu.ly

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Additionally, some types of MD may be underdiagnosed or misdiagnosed, which can make it difficult to accurately estimate their prevalence [9-10].

Material and methods

The southern region in Libya contains several regions that are located at an estimated distance of 30 kilometers to 800 kilometers or more from sabha . The capital of the south is the city of Sabha, which has a single central hospital (Sabha Medical Center), and it is considered the only center in the region to which all transfer cases in the regions of the south are transferred. The first experience and the beginning of assessing and categorizing cases of muscular dystrophy in the area were overseen by a substantial specialized scientific committee in the city of Sabha beginning in May 2022, together with subcommittees in other southern regions. From May 10, 2022, to May 30, 2023, there were 300 cases, of which 78 were presumed to have inherited muscular dystrophy, and more than 200 were rejected as showing in (Fig.1). Blood samples were submitted to the Biocentia laboratory in Tripoli, the capital of Libya, for genetic analysis to determine the kind of inherited muscular dystrophy condition for a definitive diagnosis.

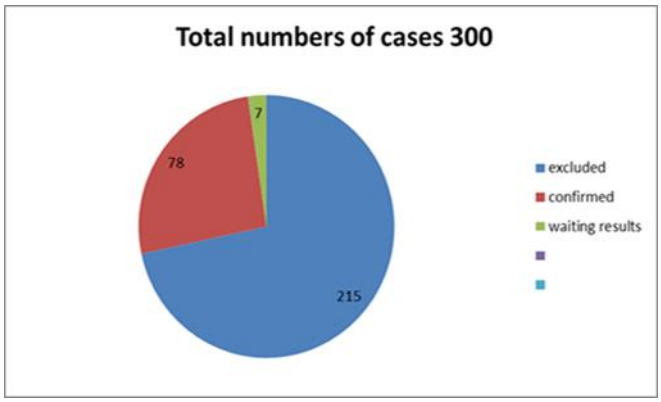

Fig. 1: Total number of cases

Results

85 cases have been reported thus far from various parts of the south (42 from the city of Sabha and the rest from areas outside of Sabha), including 45 cases involving children (age equal to or less than 15 years). 40 cases involved adults (aged over 15), with 16 male and 24 female cases. Of the children, 31 were male and 14 were female. (Fig.2).

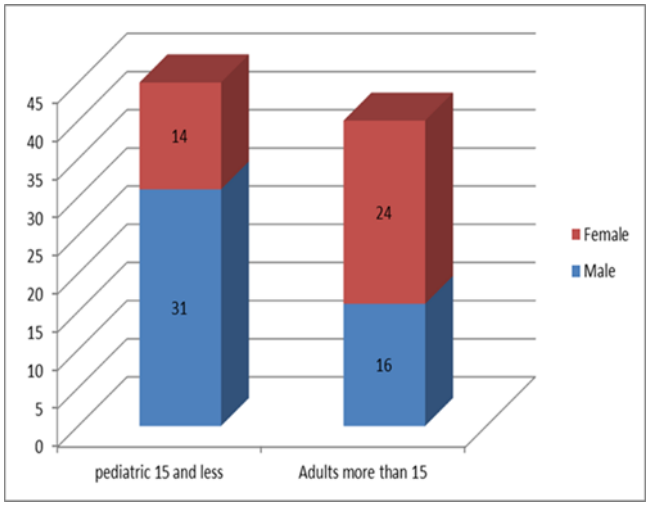

Fig. 2: The number of genders

The results of the genetic analysis of 78 cases are shown in Table No. 1. 7 cases have Duchenne muscular dystrophy, 18 cases have limb girdle muscular dystrophy, 2 cases have Beckers muscular dystrophy, 7 cases have congenital muscular dystrophy, 12 cases have spastic paraplegia, 5 cases have Charcot-Marie Tooth, and 2 cases have very rare diseases, and they are considered the first cases in Libya, namely, Chopra Amiel Gorgon and Floating Harbor Syndromes, as showing in table 1 and fig 3.

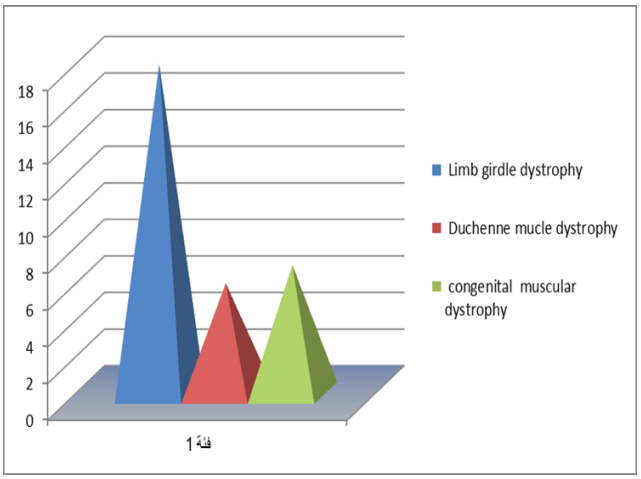

Fig. 3: The most common muscular dystrophy

Table 1: Number of Cases and Type of Muscular Dystrophy

NO-	DISEASES TYPES	NUMBER OF THE CASES	NOTES
1	DUCHENNE MUSCULAR DYSTROPHY (DMD)	7 CASES	3 CASES ELLIGIBE FOR TREATMENT , 3 CASES NON-ELLIGIBLE, DETAILS IN SEPARATE TABLE
2	LIMB GIRDLE MUSCULAR DYSTROPHY	18 CASES	
3	BECKER MUSCULAR DYSTROPHY	2 CASES	
4	CONGENITAL MUSCULAR DYSTROPHY	7 CASES	ONE CASE ULLRICH CMD
5	SPASTIC PARAPLEGIA	12 CASES	
6	Charcot-Marie-Tooth	5 CASES	
7	VERY RARE MUSCULAR ATROPHY	2 CASES	CHOPRA AMIEL GORDON SYNDROME, and FLOATING HARBOR SYNDROME
8	OTHERS DISEASES (METABOLIC)	4 CASES	
9	SMA (SPINAL MUSCULAR ATOPHY) NEGATIVE	29 CASES	REQUEST ANOTHER TEST

Table 2: List of Duchenne Muscular Dystrophy

PATIENTS	AGE	MUTATIONS TYPE	ELLIGIBILITY	DRUGS
A	9 YEARS	Exon deletion 53	Non	-------
B	16 YEARS	Exon deletion 52	ELLIGIBLE	Exondys51
C	17 YREARS	Exon deletion 4 - 52	ELLIGIBLE	Vyondys
D	12 YEARS	Exon deletion 45 -52	ELLIGIBLE	Vyondys
E		Deletion of exons 49-52	ELLIGIBLE	Vyondys
F	15 YEARS	Hemizygous deletion of bp 27.563	NON	----------
G	10 YEARS	Hemizygous deletion of bp 27.563	NON	----------

Other genetic tests were sent, and the results are pending, even though 29 cases were thought to be negative for spinal muscular atrophy. In four cases, a hereditary metabolic illness was evident. As demonstrated in Table 2, Duchenne muscular dystrophy impacted seven patients. In contrast to the three cases of mutations that are now ineligible, four cases of mutations are eligible for therapy.

Discussion

Neuromuscular diseases can affect people of all ages, from infants to the elderly. While some of these illnesses are inherited and run in families, others may be brought on by a person's environment or an underlying medical condition [18-20].

Muscle twitching, cramping, and spasms are typical signs of neuromuscular disorders. It may be challenging to carry out daily activities, including walking, standing, or lifting objects, as a result of these symptoms [14-17].

The first-line confirmatory test for NMD patients' medical care has been purposefully altered during the past ten years to use DNA diagnostics [8-9]. Once the most important clinical, electrophysiologic, and metabolic tests have been completed [12-13] . Several localities in southern Libya are spread out over an estimated radius of 30 kilometers to 800 kilometers or more. Sebha , the capital of the south, is home to the Sabha Medical Center, which is thought to be the only center in the area and where all transfer cases from the southern regions are sent.

The first experience and the beginning of examining and sorting cases of muscular dystrophy in the south region was at the beginning of May-2022 under the supervision of a major specialized scientific committee in the city of Sebha and sub-committees in other regions of the south.

From May 10, 2022, to May 30, 2023, there have been a reported 30 cases of inherited muscular atrophy. More than 200 cases—or 78 out of around 300 cases—were excluded. neuromuscular diseases can be difficult to diagnose since their symptoms often resemble those of other illnesses[10-13]. To make a diagnosis, doctors may combine physical examinations with laboratory tests, imaging studies, and tests of nerve and muscle function [15-17].

The majority of neuromuscular illnesses do not yet have a cure; however, treatment can help control symptoms and enhance quality of life [20-21]. Immunosuppressants or corticosteroids may occasionally be administered to lessen inflammation and slow the development of the illness [21]. Exercise and physical therapy can preserve muscle strength and increase mobility [16]. In some circumstances, breathing assistance such as a ventilator may be necessary to help with breathing [18-20].

A multidisciplinary strategy for treating neuromuscular illnesses may be used, involving neurologists, physical therapists, occupational therapists, speech therapists, and respiratory therapists [15-17]. To cope with the emotional and social effects of the disease, patients may also find aid in support groups and counseling [12-15] .

Investigations into novel therapies for neuromuscular illnesses are still ongoing and include immunomodulatory medicines, stem cell therapy, and gene therapy [14]. Clinical trials are being conducted to examine the efficacy and safety of these medicines in humans [15-17].

Numerous difficulties, including physical restrictions, social isolation, and financial constraints, can be faced by patients with neuromuscular illnesses [15] . In overcoming these difficulties, assistance from loved ones, close friends, and medical professionals can be very helpful [14].

With regard to muscular dystrophy, which is symbolized by the difficulty of clinical diagnosis due to the similarity of symptoms with other neurological diseases and the significance of genetic analysis for the diagnosis of these diseases[18-20], this resulted in many challenges and difficulties during the first experience of examining and sorting cases due to the small number of medical staff with insufficient experience[16].

The Sabha Medical Center is the only place in the southern region that deals with cases of dystrophy in terms of examination and genetic analyses, and the distance to the genetic analysis laboratory to conduct the analysis also makes it difficult to transfer and delays the results. Another issue is the high cost and scarcity of muscular dystrophy medications that alleviate symptoms, the expert scientific group is therefore working diligently to establish physiotherapy facilities for patients with muscular dystrophy in Libya and to localize treatment and genetic testing there.

Recommendations

1. Establishing a dedicated section of the Sabha Medical Centre with accommodations, follow-up care, and physical therapy services for patients with inherited muscular dystrophy

2. Training the medical staff including (internal medicine, neurologist (adult and pediatric respiratory, orthopaedics, and physical therapist)

3. Due to the difficulty of travel for this group of patients, the Scientific Committee for Muscular Dystrophy makes field visits to far-off southern cities to inspect and sort cases for genetic study

4. Activating scientific workshops on genetic counselling and increasing community knowledge of inherited muscular dystrophy disorders

5. Establishing an internal targeted therapeutic system

Conclusion

Background and purpose numerous hereditary diseases, including neuromuscular disorders (NMDs), can now be validated thanks to genetic testing, which has become a crucial component of modern healthcare. In Libya in general and the southern region in particular, genetic investigations constitute a relatively new occurrence. Genetic testing has found more than 70 cases of patients with muscular dystrophy this year, and it is projected that this number will climb in the years to come.

Acknowledgment

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ETHICAL APPROVAL AND INFORMED CONSENT

This study used information from the Sebha Medical Center's muscular dystrophy clinic's triage department, which accepted suspected inherited muscular dystrophy cases and requested routine clinical examinations. Since the primary objective of this study is the findings of genetic analysis, patient consent is not necessary. All patient IDs and data are also handled with the utmost secrecy. This investigation has received approval from both the Sebha Medical Center's Scientific Committee and the Muscular Dystrophy Scientific Committee, where the study was done

Conflicts of Interest

The authors declare no conflict of interest.

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