ORIGINAL RESEARCH PAPER

Neurology

CLINICAL AND DIAGNOSTIC PROFILE OF MYOPATHY IN A TERTIARY CARE CENTRE

KEY WORDS:

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ABSTRACT

Myopathies are a diverse group of disorders that primarily affect the structure, metabolism, or channel function of skeletal muscles. They typically exhibit muscular weakness that interferes with daily activities. Motor dysfunction without any sensory complaints is a common feature of myopathies.

INTRODUCTION :

Myopathies are disorders that impair the skeletal muscle's channels, structure, or metabolism[1]. They can be acquired or inherited. They include congenital myopathy, muscular dystrophy, mitochondrial, inflammatory, metabolic, toxic and drug induced[1]. Myopathies have distinctive clinical and laboratory features that can distinguish them from other disorders of the motor unit, including the neuromuscular junction, peripheral nerve, or motor neuron. The most prevalent muscular dystrophies are limb girdle muscular dystrophy (LGMD) in adults and Duchenne muscular dystrophy (DMD) in children[2].

Early research was conducted by Abraham et al. and Desai et al. in 1960 on muscular dystrophy, revealing the clinical characteristics of Myopathies[3][4]. At the KEM Hospital in Mumbai, Mondkar and Bhabha studied the clinical characteristics of 126 cases of muscular dystrophies in 1984[5]. But very limited studies are available describing the clinical profile of Myopathies. The present study highlights the clinical profile of myopathies in patients who have visited a tertiary care hospital in Madurai.

Aims And Objectives:

To study the clinical and diagnostic profile of myopathy in a tertiary care center.

Study Design:

Hospital based prospective cross-sectional study.

Place Of Research:

Department of neurology, Madurai medical college, Madurai.

Study Period: March 2021-February 2022.

Sample Size: 50

Inclusion Criteria:

All patients with probable myopathy were included.

Exclusion Criteria:

Patients with concurrent Neuropathy, Radiculopathy, Stroke, Myaesthenia Gravis, Motor neuron disease were excluded from the study.

METHODOLGY:

Evaluation of patients suspected of having myopathy was done which includes thorough history and clinical examination based on symptoms and progression of the disease with family history. Diagnostic tests are ordered to add laboratory evidence in support or against the clinical suspicion. Electro diagnostic studies were done as an extension of the physical examination.

RESULTS:

A total of 50 patients with suspected cases of myopathies were included, of whom 38 (76%) were males and 12(24%) patients were females with male to female ratio of 3:1. The age group with the highest prevalence of myopathy in our study was 1–10 years old with 28 cases (56%) with 36 (72%) Patients had a positive family history.

Table 1: Age wise Distribution

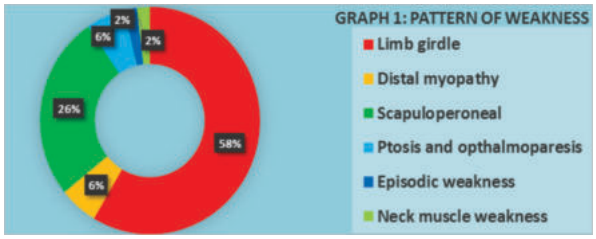
AGE	NO OF PATIENTS
1-10 YEARS	28
10-20 YEARS	18
20-30 YEARS	3
>30 YEARS	1

In our study, autosomal recessive (AR) inheritance was the most common mode of inheritance in 16 cases (32%), followed by sporadic inheritance in 14 cases (28%).

Table 2: Inheritence Pattern

INHERITENCE	NO OF PATIENTS
AD	9
AR	16
X LINKED	11
SPORADIC	14

Out of 50 patients Limb girdle pattern of weakness was most common pattern of weakness followed by scapuloperoneal.



Out of 50 cases 15 cases were dystrophinopathies, 11 cases were calpainopathy, 6 cases were dysferlinopathy,5cases were sarcoglycanopathy, 3 cases were FSHD, 3 cases were myotonic dystrophy, 2 cases were Emery Dreifuss muscular dystrophy(EDMD), 1 case was dermatomyositis,1 case was statin induced myopathy, 1 case was myotonia congenita, 1 case was oculopharyngeal muscular dystrophy(OPMD), 1 case was hypothyroid myopathy. Cases of myotonic dystrophy and FSHD presented with facial weakness. OPMD patient had dysphagia with monotonous speech. True muscle

hypertrophy was seen in myotonia congenital where as pseudohypertophy of calf was seen in Dystrophinopathies and few LGMD. Muscle atrophy was seen in dysferlinopathy variant of LGMD, FSHD, myotonic dystrophy. Early contractures were seen in EDMD(figure 1 &2) and dystrophinopathies.

Myoptonia was seen in myotonic dystrophy and myotonia congenita(figure 7). Early respiratory and cardiac involvement seen in myotonic dystrophy. The respiratory and cardiac manifestations of dystrophinopathies, sarcogly canopathies, calpainopathies, and EDMD were late. Mild mental retardation was found in dystrophinopathy and Myotonic dystrophy patients. Dystrophinopathy patients showed valley sign(figure 6). Dysferlinopathy patients showed diamond on quadriceps sign(figure 3 & 4) and biceps lump. Frontal baldness, hatchet facies, temporal wasting was seen in myotonic dystrophy. Poly hill sign (figure 5) was seen in FSHD patient. Helitrophe rash(figure 8), shawl sign, gottron papules were seen in Dermatomyositis patient.

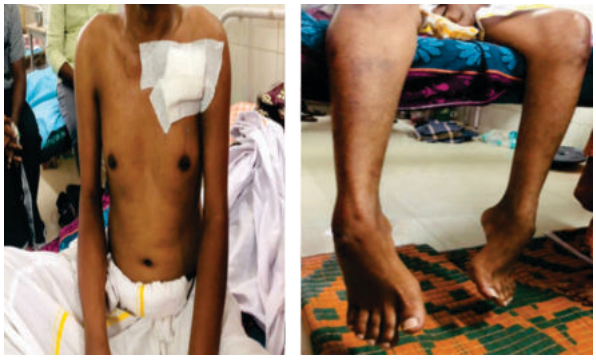


Figure 1 and Figure 2 : Case of Emery Dreifuss Muscular Dystrophy showing scapuloperoneal pattern of weakness with elbow and ankle contractures

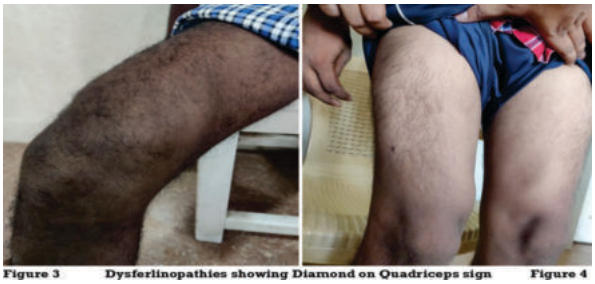


Figure 3 Dysferlinopathies showing Diamond on Quadriceps sign Figure 4

Fig 5: Case of FSHD showing Polyhill sign

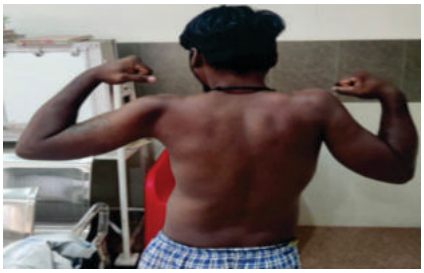


Fig 6: Case of Becker's Muscular Dystrophy showing Valley sign

Fig 7: Case of Myotonic Dystrophy showing percussion Myotonia.

Fig 8: Case of Dermatomyositis showing Heliotrophe rash.

DISCUSSION:

Every community in India experiences DMD[6]. Pradhan's description of the "Valley sign," which consists of distinct muscle wasting and hypertrophy, helped in the diagnosis of dystrophinopathy in our patients[7].The most prevalent muscle condition affecting adults is LGMD[6]. LGMD contains multiple subgroups defined genetically and by protein anomalies[8]. Differential weakness was seen in the form of extensors and abductors of hip, extensors of knees are more commonly involved in dystrophinopathies in our study similar to Jose A et al & Darras BT et al[9][16]. Calf hypertrophy, knee flexors weakness, positive hip abductor sign with winging of scapula were seen in sarcoglycanopathies in our study similar to wicklund MP et al[10]. Knee plantar flexor weakness with diamond on quadriceps sign, hypertrophy of calf were seen in dysferlinopathies in our study similar to Nalini et al[11][12]. Our study showed winging of the scapula, hip extensor weakness, and muscle atrophy in calpainopathies as per Khadilkar et al[13]. There are few cases of facioscapulohumeral dystrophy (FSHD) reported because it is rare. Srinivas[14] found that just 2.3% of patients in a sample of 211 instances of muscular dystrophy had FSHD, while Das[8] reported a 1.3% FSHD phenotype. In Our study 6% of total cases were FSHD. Scapular winging, polyhill sign, Beevor sign were seen in FSHD in our study as per Pradhan et al study[15]. Calf hypertrophy with proximal weakness pseudomyotonia is seen in hypothyroid myopathy. Serum CPK level were normal in thyroid myopathy, mildly elevated in FSHD & Emery(<500), markedly elevated in other patients. Dystrophinopathy, sarcoglycanopathy presented with cardiomyopathy whereas EDMD, myotonic dystrophy presented with arrhythmias. Electrodiagnostic tests showed fibrillation potential with small MUP and early recruitment.

CONCLUSION:

Due to a paucity of knowledge about muscle disorders in India, parents of afflicted children frequently feel unsure of where to turn for assistance. In order to diagnose the type of dystrophy, a detailed history and a thorough clinical examination aided by electrophysiological support are required.

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