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MUSCULAR DYSTROPHIES AND IT'S APPROACH IN CHILDREN: A BRIEF REVIEW

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Abstract: Muscular Dystrophies are a group of inherited disorders characterized by progressive muscle weakness and wasting caused by mutations in genes, usually involved in making muscle proteins like dystrophin. Dystrophin is present in normal individuals from fetal life onwards in all skeletal, heart and smooth muscle and in some neuronal cell types. It works to strengthen the muscle fibers and protect them from injury as the muscles contract and relax. When dystrophin is absent or dysfunctional, healthy muscle tissue is progressively replaced by fibrous tissue and fat, leading to a loss of muscle function and reduced ability to generate force. DMD/BMD affects 1 in 5,000 individuals globally, mostly diagnosed in childhood. Dystrophin-associated muscular dystrophies can be understood in *Ayurveda* as *Adibalapravritta Vyadhi* (inherited disorders) caused by *Shukra-Shonita Dosha* (defects in gametes) and *Beejabhagavayava Dushti* (genetic abnormalities). These defects lead to degeneration of *Mamsa Dhatu* (muscle tissue), which manifests as muscular weakness and impaired function. In conditions such as gene mutation-related dystrophies are considered *Yapya* or *Asadhya Vyadhi* in *Ayurveda*, complete cure is not feasible. However, *Ayurveda* interventions including *Panchakarma*, *Pitta shaman chikitsa* and *Rasayana Chikitsa* can aid in symptom management and improve quality of life to a certain extent. In such disorders, only *Lakshanik Chikitsa* (symptomatic management) can be administered, owing to the irreversible nature of the pathology.

Index Terms - Muscular Dystrophy, Beejabhagavayava Dushti, Rasayana Chikitsa* and *Yoga

I.INTRODUCTION

Muscular Dystrophies are a heterogeneous group of inherited neuromuscular disorders characterized by progressive muscle weakness and wasting. [(1)] The condition arises due to genetic mutations, most commonly in genes responsible for structural and functional muscle proteins such as dystrophin. The absence or dysfunction of dystrophin leads to gradual replacement of healthy muscle fibers with fibrous and fatty tissue, resulting in functional impairment and disability. Muscular dystrophies are a group of genetically distinct disorders with variable onset and severity, ranging from congenital to adult forms.

1. Epidemiology

Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD) are the most prevalent forms affecting children, with a combined global incidence of approximately 1 in 5,000 individuals. These disorders are progressive and life-limiting, requiring multidisciplinary and integrative management strategies. *Ayurveda*, though not describing MD in direct terms, offers conceptual frameworks that help in understanding and addressing such hereditary muscle-wasting conditions.

2. Etiology

Dystrophin, a large cytoskeletal protein, is expressed from fetal life onwards in skeletal, cardiac, and smooth muscle, as well as in certain neuronal cell types. The DMD gene, the largest in the human genome on the X chromosome, is highly prone to mutations and follows an X-linked recessive inheritance, affecting mostly males while females are usually carriers. Males (XY) express the disease phenotype if their single X chromosome, inherited from their mother, carries the mutation**.** In rare cases, daughters may also be affected if they inherit defective alleles from both parents. Dystrophin mutations determine clinical severity: out-of-frame mutations cause almost complete loss of dystrophin, leading to severe Duchenne, while in-frame mutations allow partial dystrophin, resulting in milder Becker disease.

3. Pathophysiology

The sarcolemma, which is the specialized cell membrane surrounding muscle fibers, plays a crucial role in maintaining their shape and providing mechanical stability during contraction and relaxation. It also regulates the exchange of ions and molecules between the intracellular and extracellular environments. Mutations in the dystrophin gene (DMD) or in extracellular matrix proteins such as

LAMA2, COL6A, and FKRP lead to Sarcolemmal instability, which allows uncontrolled influx of calcium ions into the muscle cells. In addition, mechanical stress causes overactivation of stretch-activated calcium channels, further increasing intracellular calcium levels. This excess calcium enters the mitochondria, where it disrupts ATP production and triggers the opening of the mitochondrial permeability transition pore (mPTP), ultimately leading to apoptosis. Elevated intracellular calcium also activates calcium-dependent proteases, particularly calpains, which degrade essential structural proteins and accelerate muscle degeneration. This muscle fiber damage subsequently triggers inflammation, with immune cell infiltration and cytokine release that further worsen degeneration. Cycle repeats leading to Progressive Muscle Degeneration, Atrophy& Fibrosis ultimately manifesting DMD. [2]

4. Classifictaion of Muscular Dystrophies

Muscular dystrophies are classified based on the age of onset. Childhood-onset types include Duchenne muscular dystrophy, Becker muscular dystrophy, limb girdle muscular dystrophy, Emery Dreifuss muscular dystrophy, and congenital muscular dystrophies. In contrast, adolescence or adulthood-onset types comprise facioscapulohumeral muscular dystrophy, distal muscular dystrophy, myotonic muscular dystrophy, and oculopharyngeal muscular dystrophy. Among the various forms of muscular dystrophies, Duchenne muscular dystrophy (DMD) represents the most common and severe childhood-onset type. The clinical features along with the typical age of symptom onset and stages of ambulation of DMD are presented below.

Table 1. AMBULATION STAGES AND CLINICAL FEATURES IN DMD

STAGES OF DMD	STAGE	AGE RANGE	SYMPTOMS/PROGRESSION
Ambulatory	Early Symptoms	2–3 years	Walks independently
Early Ambulatory	Progressive Weakness	4–9 years	Calf Pseudohypertrophy Tiptoe walking Can climb stairs Waddling gait Gowers sign
Late Ambulatory	Loss of Ambulation	10–12 years	Cannot arise from the floor Unable to walk
Early non-ambulatory	Respiratory & Cardiac Involvement	9-14	Frequent Falls Scoliosis may develop May use powered mobility like wheelchair
Late non-ambulatory	Non-Ambulatory	14+	Upper limb weakness Completely wheelchair-bound or bedridden. Contractures develop. Death due to cardiac and/or respiratory failure

5. *Ayurveda* Perspective

Although Muscular dystrophies are not explicitly described in the *Ayurveda* texts, they can be interpreted through related pathological principles. These conditions fall under the category of *Adibalapravritta Vyadhi* (hereditary disorders) and *Shukra-Shonita Dosha*, which reflects defects in gametes. *Ayurveda* considered three genetic units in the form of *Beeja* (gametes), *Beejabhaga* (development of organ, system & physiological functions), and *Beejbhagavayava* (genes). The concept of *Beejabhagavayava Dushti* [3] explains genetic abnormalities affecting the development of specific organs and tissues. *Mamsa* is *matruja bhava.* Hence, *Beejabhagavayava Dushti Janya Mamsa Dhatu Kshaya* describes impaired nourishment and degeneration of muscle tissue due to such defects. Thus, from an *Ayurveda* standpoint, DMD may be regarded as a *Mamsa Dhatu Kshaya Janya Vyadhi* primarily involving depletion of *Mamsa Dhatu*, often accompanied by secondary infiltration of fat (*Meda Dhatu)* in the form of pseudohypertrophy.

6. Management

For Muscular Dystrophy, there is no complete cure since it is an *Asadhya vyadhi*, only *Dravyata* and *Karmata* of the *Mamsa dhatu* can be improved to some extent. The treatment aims at *Srotorodhara* and *Amahara* locally, along with *Kapha-hara* measures to relieve obstruction. Correction of *Agni* is essential, wherein *Pittahara* interventions are indicated for managing *Tikshnagni*, while *Vatadoshara* measures address the associated *Vata dosha* predominance. In the management of *Vata-dominant* conditions, In the initial days of treatment, *Udwartana* with *Triphala Choorna* is beneficial due to its *Laghu* and *Ruksha guna*, which helps in *Tridoshahara* and removes *Srotorodha*. Followed by, for *Snehana*, *Mahanarayana Taila*, *Sahacharadi Taila*, and *Kottamchukkadi Taila* are indicated in *Vata rogas*, followed by *Nadi Sweda* with *Dashamoola Kwatha* to promote circulation by *Dosha Vilayana.* Likewise, *Mrudu Sweda* (*Mrudu apatarpana*) like *Shashtik Shali Pinda Sweda* can be beneficial. *Matra Basti* with *Tiktaka Ghrita* is advocated as it is *Vatahara*, *Agnisandeepana* and *Rasayana* effects. In parallel, *Brihatchagalyadi Ghrita* can be used for *Matra Basti*. Moreover, *Yoga Basti* consisting of *Niruha Basti* prepared with *Erandamoola Kwatha Choorna* processed with *Ksheera* has *Vata-Pittahara* effect. *Anuvasana Basti* such as *Lakshadi Taila* or *Mahanarayana Taila* recognized for its *Tridoshahara* properties can alleviate aggravated *doshas*. Collectively, these interventions contribute to comprehensive management.

Rasayana therapy may play a beneficial role in such conditions by delaying disease progression and slowing degeneration, thereby preventing rapid muscle wasting. Its antioxidant, [4] anti-inflammatory and immunomodulatory properties help in mitigating chronic low-grade inflammation in the muscle tissue, as reactive oxygen species [5] play a role in the pathology of Muscular Dystrophy. Furthermore, *Medhya Rasayanas* such as *Panchagavya Ghrita*, *Kalyanaka Ghrita* and *Brahmi Ghrita* when administered post *Panchakarma* therapy will support mental and cognitive functions, which is particularly important, as children affected by DMD may develop depressive tendencies, borderline IQ, and emotional disturbances Supportive care with physiotherapy, stretching exercises and

mobility aids such as braces or wheelchairs helps maintain function, although prolonged immobilization may contribute to joint stiffness. *Yoga* practices like *Bhujangasana*, *Shalabhasana*, *Tadasana* and *Setubandhasana* [6] aid in flexibility, joint mobility and help in reducing the contractures that develop. While breathing techniques such as *Anuloma-Viloma* and *Bhramari Pranayama* help to maintain lung capacity and their regular practice may delay or reduce respiratory difficulties that develop over time in DMD.

II.DISCUSSION

The *Beejabhagavayava Dushti Janya Mamsa Dhatu Kshaya* is a key factor in the pathology of Muscular Dystrophy. Correcting *Tikshna Agni by pitta shamana* is crucial to reduce excess metabolism in *Dhatu paka Avastha. Ayurveda* interventions including *Panchakarma* and *Rasayana Chikitsa* help in symptomatic management to some extent as it is *Yapya /Asadhya vyadhi.* Therefore, in this disease, *Lakshanik Chikitsa* only can be administered as there is no cure for gene mutated dystrophies.

III.CONCLUSION

DMD is a progressive, disabling and life-limiting disorder that significantly affects the lives of patients and their families. A combined approach using *Ayurveda*, Physiotherapy & *Yoga* can help enhance the quality of life of children with DMD. Therefore, by following *Ayurveda* treatment methodologies, quality of life can be improved and increase life span few more years and prolong the ambulatory phase.

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