



CASE REPORT

Effect of ayurvedic management on Duchenne Muscular Dystrophy - A case report

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Abstract

Duchenne Muscular Dystrophy (DMD) is the most prevalent X-linked recessive neuromuscular disorder, affecting approximately 1 in 3,500 male infants. It arises from a deficiency in dystrophin-a protein encoded by the human genome's largest gene that is essential for stabilizing the muscle sarcolemma. In the absence of dystrophin, the muscle membrane becomes fragile, leading to progressive fiber necrosis and profound muscular weakness, though the nervous system remains intact. While corticosteroids are the primary pharmacological option for modifying the disease course, their use is often complicated by side effects such as osteoporosis, cataracts, and significant weight gain. Clinically, this case involves a 12-year-old male presenting with a long-standing history of proximal weakness, characterized by frequent falls and difficulty navigating stairs or rising from a squatting position. Diagnostic investigations revealed a marked elevation in creatine phosphokinase (CPK) levels, and molecular analysis identified a hemizygous variant of uncertain significance (VUS) within the *DMD* gene. Here the child has been treated in the outpatient department of the tertiary Government Ayurveda Hospital, and medications were administered for one month, followed by inpatient management for one month by considering the concept of *dhatvagni mandya* (poor tissue metabolism), *Ama* (metabolic toxins), etc. There was a significant improvement in the clinical findings, which was assessed by the Vignos scale of lower extremity and quality of life by the Barthel index scale. Significant improvement in this short period of management substantiates the benefit of Ayurvedic medicines in the management of DMD.

Introduction

The muscular dystrophies represent a collection of genetic conditions characterized by a continuous and irreversible decline in muscular strength and volume. Among these, Duchenne Muscular Dystrophy (DMD) is the most prevalent, manifesting as a progressive myopathic disease inherited through an X-linked recessive pattern. Males are main sufferers, and females are carriers but very rarely suffer with a mild course of the disease. The incidence is 1:3500 live male infants, characterized by progressive weakness of a selected group of muscles without the involvement of the nervous system. ^[1]The *DMD* gene is composed of 79 exons and utilizes multiple tissue-specific promoters to encode at least seven dystrophin isoforms. The absence of this protein compromises the structural integrity of the sarcolemma, leading to chronic muscle fiber necrosis and a failure of the body's natural regenerative capacity. Consequently, muscle tissue is gradually replaced by fibrous and fatty deposits. Symptoms generally become apparent at age five, with proximal weakness manifesting as difficulty in climbing stairs or jumping. The disease progresses without remission, typically resulting in wheelchair dependency by the early teenage years. At

the time of presentation, proximal weakness of muscle is evident as difficulty in climbing stairs, hopping, etc. Gowers manoeuvre is seen when trying to arise from the floor. Contraction of the Achilles tendon makes the children walk on toes. Mild lordosis may be seen. [2]The clinical course is often complicated by respiratory distress, driven by the deterioration of the intercostal muscles and the diaphragm alongside skeletal changes. These complications, together with cardiac involvement (cardiomyopathy), represent the leading causes of death. Intellectual disability and impaired gastric motility are the other features. [3]It is also associated with elevation in serum creatinine kinase (CK) (>10 times of normal), Pathogenesis is characterized by a significant rise in serum creatine kinase (CK) levels-often exceeding ten times the normal range-which is widely attributed to the heightened permeability of the muscle sarcolemma. For molecular identification of underlying mutations, Multiplex PCR and the more refined Multiplex Ligation-dependent Probe Amplification (MLPA) remain the diagnostic gold standards. Muscle biopsy may be required in mutation-negative cases and to differentiate between BMD and DMD. [4]

Thus, Duchenne Muscular Dystrophy cannot be equated to one specific disease entity in Ayurveda. Instead, it is understood as part of a group of neuromuscular ailments primarily driven by the vitiation of *Vata*. This disorder can be considered as *Adibalapravritta vyadhi* due to *beeja dushti*. [5] The concepts of *dhatvagni mandya* (poor tissue metabolism), *Ama* (metabolic toxins) formation, *kapha vriddhi* (aggravation of *kapha*), and *vata dushti* (vitiation of *vata*) were present. *Mamsa dhatu dushti* and *Srotorodha* are important in the *samprapti* of this roga. *Srotorodha* produces hypertrophy of a particular muscle, and Over time, these complex biological pathways precipitate the chronic degeneration and irreversible necrosis of the impacted muscle tissues.

Materials And Methods

Case study

Patient information

A 12-year-old boy presented to the outpatient department with a six-year history of progressive mobility issues, specifically characterised by frequent falls and difficulty climbing stairs and difficulty in rising from squatting, sitting, and lying positions and low back pain after walking a few steps from 8 years of age. Pseudohypertrophy of the calf muscle has been present since the last year. The patient was born second into a family with no history of consanguinity. Birth history reveals a full-term delivery via LSCS due to cephalopelvic disproportion. He had a birth weight of 2.5 kg. The baby cried soon after birth with a normal sucking reflex, and the neonatal period was uneventful. The child attained neck holding at 6 months of age and walking with support at 1 year of age. The child was normal till six years of age. Following this period, the family noted a gradual onset of gait instability, characterized by recurrent falls and an increasing difficulty in ascending stairs. But the parents neglected it because they thought that it was normal as he was in the growing age. At the age of 8 years, the parents notice an imbalance in walking and difficulty rising from the squatting position. Then the parents took medical advice, and a genetic study was advised. A hemizygous variant of uncertain significance (VUS) was detected on exon 68 of the DMD gene. Medications were given, and physiotherapy was

advised. As age progressed, the condition worsened, and lumbar lordosis increased, and Gowers sign became evident.

Clinical findings On Examination

A hyper lordotic curve was evident with anterior pelvic tilt while standing. Both Gowers' sign and the Valley sign were present in this case.

CPK was found to be elevated.

Arrhythmia, chest pain, and respiratory complications were absent.

Vital signs and higher mental functions were found to be normal.

After thorough examination, a treatment protocol was planned based on dosha (regulatory functional factors of the body).

Investigations

Serum CPK levels on 30/9/2017 were elevated and were 5825 IU/L, and ALP was 128 IU/L. Genetic analysis for DMD deletion or duplication conducted on 14/2/2018 reported that a hemizygous variant of uncertain significance (VUS) was detected on exon 68 of the DMD gene and diagnosed the condition as X-linked recessive DMD.

TIMELINE

The detailed timeline of the patient's treatment is enlisted in Tables 1 and 2.

THERAPEUTIC INTERVENTION

After thorough examination in the outpatient department, the patient was given internal medicines for one month along with necessary dietary restrictions. Initially the patient was given *Vaiswanara choorna* (6 gm at 11 am before food) for *pachana* (enhancing metabolic fire) and *Gandarvahastadi Kashaya* (50 ml at 7 am and 5 pm before food) as *vataanulomana*. [6], [7], [8] *Gandha taila* (5 drops with milk twice daily) was given after one week onwards as *balya* (enhances strength). [9] Following the attainment of *deepana*, *Gandharvahasthadi kashayam* was transitioned to *Ashtavargam kashayam* to achieve *balya* and *srotoshodhana* effects. After that the patient was managed in the inpatient department of the hospital (Table 2). The child was managed with both internal and external medications, including physiotherapy. Treatments such as *udwartanam* (rubbing of medicated powder on body) with *kolakulathadi choornam*, *Dhanyamladhara* (pouring of medicated liquid from fermented cereal), repeated topical applications over the calf muscles, various *snehana* and *swedana* procedures, *Ksheeravasthi*, etc., have been done. The assessment was done before and after the treatment.

FOLLOWUP AND OUTCOME

The patient has been examined through various clinical examinations. At the end of treatment for 2 months, significant improvements were noted in his clinical features. The Barthel Index assessment was done. [10] After the treatment, the score increased from 64 to 78. The Vignos scale for functional assessment of lower extremities was done. There was a notable improvement in mobility, moving from a Grade 5 (ambulatory but unable to rise from a chair or use stairs) to a Grade 2 (capable of walking and climbing stairs with railing support). Observations infer the scope of Ayurvedic intervention in managing DMD and thereby improving the functional status and quality of life of the patient. No adverse reactions were noted during the treatment.

Timeline of outpatient management (Table 1)

Date of admission	Complaints/condition of the patient	Interventions
9/11/2021-	First visit at the outpatient department	6 gm of <i>vaiswanara choornam</i> at 11 am before food
18/12/2021	Difficulty In getting up from the squatting position Difficulty in climbing stairs Calf muscle hypertrophy Frequent falls Decreased appetite	<i>Gandarvahastadi Kashaya</i> (50 ml at 7am and 5pm before food)
18/11/2021-	Appetite improved Other	<i>Ashtavargam kashayam</i> 50 ml at 7am and 5pm (before food) <i>Balaristam</i> 10ml twice daily at 9 am and 8 pm after food
24/11/2021	Complaints persist	<i>Gandha tailam</i> 5 Drops twice daily with milk before food (8am and 7pm)
25/11/2021-	Appetite improved	Above 4 medicines continued
8/12/2021	Other Complaints persist	

Timeline of IP management (Table 2)

Date of admission	Complaints/condition of the patient	Interventions
9/12/2021-18/12/2021	Difficulty In getting up from squatting position Difficulty in climbing stairs Calf muscle hypertrophy Frequent falls Recurrent cough	6 gm of <i>vaiswanara choornam</i> at 11 am before food. <i>Balaristam</i> 10ml twice daily at 9 am and 8 pm after food <i>Shadpala ghrim</i> 5gm twice daily 10 am and 9pm after food <i>Elakanadi Kashaya</i> 30 ml muhumuhu
10/12/2021-17/12/2021	Complaints persist	<i>Dhanyamladhara</i> for 7 days with 45 min duration
18/12/2021-24/12/2021	Cough reduced	<i>Ashtavargam kashayam</i> 50 ml at 7am and 5pm before food) <i>Udwartanam</i> with <i>kolakulathadi choorna</i> for 14 days with 45 min duration.
25/12/2021-31/12/2021	Can do exercise for more duration	Same internal medicines repeated <i>Kadikizhi</i> with <i>kolakulathadi choorna</i> mixed in <i>dhanyamla</i> for 7 days with 45 min duration
1/1/2022		<i>Upanaha</i> with <i>kolakulathadi choorna</i> for 21 days on bilateral calf muscle
2/1/2022-8/1/2022	Frequency of falls reduced	<i>Sarvangaabhyanga</i> with <i>karpooraditaila</i> for 7 days with 45 minutes duration
9/1/2022- 15/1/2022		<i>Patrapottala sweda</i> with <i>eranda,sigru,arka,nirgundi</i> , tamarind leaves fried in <i>tila taila</i> and dipping of <i>pottali</i> in <i>karpooradi taila</i>
16/1/2022- 22/1/2022		<i>Utsadanam</i> with <i>choorna</i> of <i>kulattha</i> and <i>priyangu</i>
23/1/2022- 27/1/2022	Walking stability increased	<i>Kayaseka</i> with <i>karpasastyadi taila</i> for 7 days with 45 min duration
From 28/1/2022		<i>Ksheera vasthi</i> with <i>indukantham gritha</i> and <i>pippalyadianuvasana taila</i> and <i>panchatikthaka Kashaya</i> for 5 days
07/02/2022	Can climb stairs without fall	Patient was discharged

Results And Discussion

The patient had visited the hospital as a diagnosed case of DMD. In Ayurveda this condition cannot be correlated to a specific disease. Although genetic deformity cannot be cured, quality of life of a person can be improved through Ayurvedic interventions by analyzing the *dosha* and *dhatu* involved in the disease. In *samprapthi* (pathogenesis), mainly *vata dosha* and *kapha dosha* are involved. Function of *vyana vata* (a subtype of *vata*), such as *gati apakshepana* (movement) and *sarvakriya sakthi* (ability to do various actions), and that of *udanavayu* (a subtype of *vata*), which is responsible for *bala* (strength), are deranged. Among *kapha doshas*, *sleshaka kapha*, which is responsible for *sandhi samslesha* (integrity of joints), is affected. Among the *dooshyas*, *rasadhatu kshaya lakshanas* is there, which is manifested as *glani* (tiredness), *sosha* (depletion), and *srothorodha* (obstruction of channels). Features of *mamsadhatu kshaya* were also present as pain in joints. *Beejadusthi* itself leads to *vatothara tridoshadushti*, which later affects the *dhatuparinama* process. Hence, in this case, *dhatvagni mandhya* is present at the level of *rasa*, *raktha*, and *mamsa dhatu*, which further produces *dhatugata amavastha*. This *dhatugataama* leads to *srothodushti* of *rasa*, *raktha*, and *mamsavaha srotas* and *kapha prakopa*. The *mamsavaha sroto dushti* further leads to improper formation of *mamsa dhatu*, which causes progressive degeneration of *mamsa dhatu*. The avarana of *vyana vayu* by *kapha* takes place due to the *kaphaprakopa*. The features of *ojovyapat* were also seen in this case. *Ojo vyapat lakshanas*, such as *stabdhaguru gatrada*, *tantra*, and *mamsakshaya lakshanas*, are manifested. Here the treatment was started with *Deepana* (appetizers), *pachana* (digestive), *anulomana*, and *srotasodhana* medicines such as *vaiswanara choorna*, *Gandarvahastadi kashaya*, and *Astavargam Kashaya*. *Udwartanam* with *kolakulathadi choorna* is done for pacifying the *kapha dosha*, gives *anga sthiratha* (integrity of the body), and removes *srothorodha*.^[11]^[12] *Dhanyamladhara* was done over the full body, which is *vata-kapha hara*, as *dhanyamla* has properties like *bhedi* and *sramaklamahara*.

Snigdha kriyas such as *sarvanga abhyanga* are done to pacify the *vata dosha* and enhance the strength of muscles.^[13] *Karpooradi taila* was selected to prevent respiratory infection and reduce the vitiated *vata dosha*.^[14] After that, *swedana* is done with *patra pottala sweda* (medicated leaves are fried in a suitable *taila* and a *pottali* is made) to pacify *vata* and *kapha dosha*. *Utsadanam* with *choorna* of *kulattha* and *priyangu* was done in between for maintaining the *anga sthiradha*. *Kaya seka* was done with *karpasastyadi taila*, which reduces *vata* and prevents further *dhatu kshaya*.^[15] *Ksheera vasthi* was done for *sodhana* (elimination of *doshas*), *brimhana*, and for pacifying *vata dosha*.^[16] *Ksheera vasthi yoga* was prepared with 120 ml *panchatikthaka ksheera Kashaya*, 60 ml honey, 60 ml ghee (*shadpala ghritha*), and 60 ml *taila* (*pippalyadi anuvasana taila*). The dose was calculated according to the age of the patient. *Shadpala ghritha* has *srotasodhana*, *agni deepana*, and *vatahara* properties, which correct the *dhatwagni mandhya*.^[17] *Pippalyadi anuvasana taila* was very effective, as it is effective in weakness of the *uru* (thigh) and *kati* (back or dorsum) region.^[18] *Upaana* was done with *kolakulathadi choorna* and *dhanyamla* over the bilateral calf region, which reduces the pseudohypertrophy of the calf muscle and reduces *kapha avarana*. *Balaaristam* and *Gandha tailam* were included in the internal medicines, which improves *bala* (strength) and *ojus* (essence of all *dhatu*s).^[19]

Mudgayoosha and *mamsarasa* were advised as the diet that gave nourishment to all *dhatu*s.

Both internal and external medicines brought significant improvement in the condition of the patient.

Conclusion

In the case of DMD, no complete cure is available in any of the medical systems. Clinically significant improvement in a twelve-year-old boy with a three-month course of ayurvedic treatment (including outpatient and inpatient departments) gives a ray of hope for further research in this field. Follow-up of this case is needed for further evaluation. Treatment should be provided with a view of improving patients quality of life targeting gastrointestinal, respiratory, cardiac, and orthopaedic problems, as well as physiotherapy. Regular follow-up and evaluation are needed for this disease.

Conflict of Interest

No potential conflict of interest was reported

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