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Case Study

AYURVEDIC EMPHASIS IN DUCHENNE MUSCULAR DYSTROPHY: A CASE STUDY

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ABSTRACT

Duchenne muscular dystrophy is one of the most common dystrophinopathies known. It is the most common hereditary neuromuscular disorder and is inherited in an X-linked recessive manner. Incidence is 1:3500 live male infants, characterised by progressive weakness of a selective group of muscles without involvement of nervous system. Age of onset being 3-10 years, many children unable to walk before 18 months of age. The patient usually dies by 18-20 years of age. 80% carries have high CPK values with female being the one. Dystrophin gene is the largest human gene with 79 exons, codes for protein dystrophin required for stabilisation of protein complex at sarcolemma, the abnormal DMD gene is on X chromosome at Xp21 locus. Dystrophin deficiency thus, leads to destruction of muscle fibres and progressive muscular weakness. Corticosteroids are the only medications that have shown to alter the course of DMD but have side effects like weight gain, decreased appetite, increase changes of cataract and osteoporosis. The present study is about management 8 years old male child with B/L lower limb weakness and calf muscle hypertrophy.etc, so according to Ayurvedic management with *Panchkarma* procedures and internal medicines given the case was managed. Successful improvement in CPK values along with the signs and symptoms was observed. As per Ayurvedic *Siddhant* and *Samprapti* application considering *Adibalapravrittavyadhi* and the *Beejabhaga avayava dushti* the management done. There is no treatment in any system of medicine and prognosis being unpreventable, Ayurveda instills a regenerative mechanism in neuromuscular disorders with special concern of *Panchkarma, Rasayanas, Rasa aushadhi,* etc. By this the deterioration can effectively be prolonged and quality of life improved.

INTRODUCTION

The muscular dystrophies are diseases of muscle membrane or supporting proteins characterized by pathological evidence of ongoing muscle degeneration and regeneration. Diagnosis of these disorders is based on clinical presentation, genetic testing, muscle biopsy and muscle imaging.

Dystrophinopathies are a group of disorders resulting from mutation in the dystrophin gene (located on short arm of X chromosome, Xp21). Duchenne muscular dystrophy is the most common dystrophinopathy with an incidence of 1 in 3500 live male births.[1]

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Over 4700 mutations are reported in the Leiden Duchenne dystrophy database. 65% of the pathogenic changes are large partial deletions. Mutations in the dystrophin gene can cause Duchenne muscular dystrophy or Becker muscular dystrophy. The phenotypic variation is explained by the reading frame hypothesis. In >90% of cases, mutations that disrupt the reading frame (frame shift) lead to dystrophin deficiency and cause DMD.

Children with DMD become symptomatic before age of 5 years and may have history of delayed walking. Gait difference become apparent at 3-4 years of age. Waddling gait, Grower sign and Calf muscle pseudohyertrophy are classical findings.[2] Weakness of neck flexors is early. Other muscles that show hypertrophy includes vastus lateralis, infraspinatous, deltoid, gluteus maximus, triceps and masseter. The progression of weakness may plateau between 3 abd 6 years of age, followed by increasing gait difficulty, development of contractures and lumbar lordosis. The

age at loss of independent ambulation in untreated patient is between 8.8 to 10.5 years.[3]

Weakness of intercostal and diaphragmatic muscles with spinal deformity affects respiratory function. Dropping of vital capacity <20% of normal leads to nocturnal hypo ventilation. Cardiomyopathy and arrhythmias are major cardiac manifestation. Children with deletion of axons 48 to 53 are especially prone to cardiac complications. The cause of death is the combination of respiratory insufficiency and cardiomyopathy. Other features include intellectual disability and impaired gastric motility.[4]

Serum creatinine kinase levels are highly elevated (>10 times of normal), but do not correlate with severity of the disease or response to treatment. Multiplex PCR and the more sensitive multiplex ligation dependent probe amplification are used for detection of mutations. Muscle biopsy may be required in mutation negative cases and to differentiate between DMD and BMD. Biopsy shows necrosis and attempted regeneration of individual muscle fibers, variable muscle fiber diameter with both hypertrophic and small fibers, and central nuclei. Later, almost the entire muscle is replaced by fibrofatty tissue. On immunohistrochemistry, absence of dystrophin (1, 2, 3) staining is seen in DMD, dystrophy staining is reduced and patchy in BMD.[5]

Around 10% female carriers show variable degree of weakness with elevated levels of creatine kinase, calf hypertrophy, myalgia and cramp. Very rarely, a full DMD phenotype is present in girls with complete inactivation of normal X chromosome. Patients here survive upto 20 years of age and they mainly die because of respiratory and cardiac failure, 96% of them progress to cardiomyopathies.

Thus, according to Ayurveda, DMD cannot be directly co-related with any single disease in Ayurveda. All most all major neuromuscular disorders are identified with *Vata Dosha*. This disorder can be considered under *Vata vikar* due to *Beeja Dushti* because of *Adibalapravritta*.[6] *Samprapti* taking place under *Strotorodha* and *Beejadushti. Ayurvedic Acharyas* carefully consider this condition as *Adibala-Pravrit Mamsa Vata Kshaya* due to *Srotorodha.*[7,8,9] *Srotorodha* produces hypertrophy in particular region, it also manifests as first *Prakopa* then depletion of *Vata* aliment. This complex pathogenesis is responsible for progressive wasting and necrosis of the affected muscle fibers Also *Samprapti* can be sort out as it is result of *Mamsa-Vata Kshaya* due to *Beeja Dosha* which leads to *Vata Vaishamya* of *Mamsa Dhatu*. This Vitiated *Vayu* causes improper formation of *Mamsa Dhatu* by its influence on the *Dhatvagni* of *Mamsa*.[10] It is genetic predisposition (*Beejadosha*) that convert physiological *Vata* element in to pathological morbidity. The *Srotodushti* is responsible for the *Mamsa Dhatu Kshaya.*

AIM

To study the effect of Ayurvedic management and improving the quality of life in Duchenne muscular dystrophy children.

OBJECTIVES

- To study the etiopathogenesis of this challenging condition of DMD.
- To evaluate the efficacy of Ayurveda drugs in the management of DMD.
- To help improve the quality of life of the child.

MATERIALS AND METHODS

A 8 year old male child reported to Kaumarbhritya OPD of GAC & H dated 02/10/2021,

With the complaints of:

- B/L lower limb pain and weakness.
- B/L calf muscle weakness mostly on exertion. Since 5 months

Patient was asymptomatic till the age of 6 years, then he came across progressive slowness of movements of legs, generalised pain, and muscular weakness particularly.

Patient was a diagnosed case of DMD via Cpk values dated 14/08/2021 at private hospital and genetic study dated 01/09/2021.

Gradually patient started with these symptoms since 5 months increasing day by day.

Birth History

Antenatal: No any significant history, inj. TT 2 doses received at specified period, tablet iron and folic acid taken.

Perinatal: Full term normal vaginal delivery, Birth wt-2.5 kg

Postnatal: No NICU stay, No any significant history noted.

History of past illness – No any

Family history – No any

Dietary history – Mixed diet

Personal history – Appetite: Poor

Bowel: irregular

Urine: normal, 5-6/ day

Sleep: Disturbed

Immunisation History: Received all vaccines upto age, BCG make visible.

Developmental History: Appropriate for the age.

On Examination

- Thin appearance
- Proximal weakness
- B/L Calf muscle hypertrophy
- B/L Deltoid muscle hypertrophy
- Grower's sign: positive
- Valley's sign: negative
- Gait: partially waddling
- Muscle tone: lower limbs hypertonic

- Muscle bulk: wasting of muscle
- CNS examination: normal, conscious with time, place and person. Speech normal.

General Examination

- Eyes: pallor
- Skin: *Anushna*, normal
- Tongue: uncoated
- Nails: *Prakrut*, no clubbing
- Pulse: 78/min
- RR: 20/min
- Temperature: *Prakrut*, afebrile

Investigations

- Sr. Cpk: >20000 U/L
- Sr. Calcium: 10.43mg/dl
- Sr. Phosphorous: 260.9 U/L
- Vit. D: 73.74 ng/ml
- LFT – SGOT: 264.09, SGPT: 270.93, Alk ph.: 463.60
- MLPA test: Hemizygous deletion of Exon 45 to 53 in the Dystrophin gene.

Treatment Plan

4 sittings of *Panchkarma* procedures were planned each with 30 days interval.

- ***Bhayya Chikitsa***

1. *Sarvanga snehana* with *Nimba, Patola, Triphala, Mrudvika, Musta, Vatsak, Kirattikta, Amruta, Chandan, Vishwabheshaj*. (DMD Formula)
2. *Sarvanga Shastika shali pindaswedana* with *Vidarikand churna*.
3. *Karma Basti – Anuvasana* 30ml with DMD Formula *Sidhha tail*.
   Niruha (*Lekhana basti*)[11]= *Madhu* 5ml + *Saindhava* 2gm + *Tila tail* 10ml + *Triphala kalka* and *Lekhana dravya kwatha* (*Musta, Kushta, Haridra, Kutki, Daruharidra, Triphala*) 120ml *Avashistha* with 500ml *Jala* + *Gomutra* 10ml.

- ***Abhyantar Chikitsa***

1. *Medhya Rasayana – Yogendra ras* 5 tablets + *Vishtinduk vati* 15 tablets + DMD FORMULA *Dravyas* 80gm = 1 gm BD with honey for 30 days.
2. *Kanchnar gugullu* 1 BD for 30 days.

RESULTS

The neurological assessment observed before the treatment and after the treatment of the investigations, muscle power, reflexes, muscle tone and Anthorpometry has noted in the Table 1, 2, 3, 4, 5 respectively. These tables show the detailed evaluation Pre assessment and Post assessment. The figure 1 and 2 shows the B/L calf hypertrophy before treatment and slight changes after treatment. And figure 3 and 4 shows the reports of CPK value before treatment and after treatment respectively.

Table 1

Investigations	Before treatment (dated:14/08/2021)	After treatment (dated: 31/05/2022)
Sr. CPK value	>20000 U/L	2000 U/L
LFT : SGOT	264.09 U/L	130.91 U/L
SGPT	270.93 U/L	186.74 U/L
ALK. Ph.	463.60 U/L	147.15 U/L

Table 2

Muscle Power	Before Treatment	After Treatment
Upper limb	Rt – 3/5 Lt – 3/5	Rt – 4/5 Lt – 4/5
Lower limb	Rt – 2/5 Lt – 2/5	Rt – 4/5 Lt – 4/5

Table 3

Relaxes	Before Treatment	After Treatment
Knee	Rt – 3 Lt – 3	Rt – 2 Lt – 3
Biceps	Rt – 3 Lt – 3	Rt – 2 Lt – 2
Triceps	Rt – 3 Lt – 3	Rt – 2 Lt – 2
Ankle	Rt – 3 Lt – 3	Rt – 2 Lt – 3
Planter	Rt – 3 Lt – 3	Rt – 2 Lt – 3

Table 4

Muscle tone	Before treatment	After treatment
Upper limb	B/L Normotonic	B/L Normotonic
Lower limb	B/L hypertonic	Rt – Normotonic Lt – Slight

Table 5

Anthropometry	Before treatment	After treatment
Weight	19.5kg	22kg
Height	118 m	123cm
Head circumference	47.5cm	52cm
Chest circumference	54.5cm	56cm
Mid arm circumference	17cm	17.5cm
Calf muscle circumference	B/L – 27 cm	B/L – 24cm

There was seen improvement in the B/L calf muscle pain and weakness also in B/L lower limb pain. Walking was improved.

DISCUSSION

The DMD FORMULA *Dravyas* used are all best to balance *Vatadosha* and reducing *Stabdhata, Sandhisaithilya,* etc and are *Brumhaniya, Balya, Amahara* and *Stairyakara.* Thus, the use of this formula for *Snehana* and in *Anuvasana* in form of oil was done and best results were obtained as stated.

In DMD *Samprapti* stated according to Ayurveda, it is *Mamsagata vata* where there is predominance of *Vata dosha* with *Sthanasanshraya* in *Mamsa* and *Meda dhatu* also and vitiates and deplete them. According to *Samanya Vishesh sidhdhanta*, if *Brumhana* is provided via *Annavaha strotas* less *Brumhana* is achieved but *Tvak* being a *Mamsadhara Kala*, *Brumhana* to *Mamsa* is best achieved by *Snehana* and *Pindaswedana*. *Abhyanga* pacifies the *Vatadosha*, nourishes the tissue and increases strength.[12] While *Swedana* with specially formed bolus of medicinal drugs like *Shastika shali pindaswedana* provides relaxation to the constricted stiff muscles while providing tonicity to the body,[13] thereby increasing the mobility. It increases the metabolic rate by increasing the blood circulation and oxygen flow in the body also stimulates sweat glands and nerves.[14]

Basti is the *Ardhchikitsa* of *Vata* and is a complete treatment due to curative, preventive and promotive aspects. *Dravyas* of the *Basti* gets absorbed in the colon and show their systemic action. Also excretes Mala which is accountable for the disease process.[15] It reduces the *Vata* and is *Brumahana* and rejuvenates degenerative *Mamsadhatu*.

Yogendra rasa included in internal medicine is a *Suwarna kalp*, pacifies *Vata* and *Kapha*, strengthens the nerves and muscles thus used in treating neuromuscular conditions.[16] *Vishatinduk vati* pacifies *Vata* and *Kapha*, stimulates sense organs, blood vessels, nerves and muscles. Its alkaloids are likely to increase Glutamic Acid (GA) level in the brain. GA excites muscle contractions by stimulation of excitatory nerve impulses. It helps improve muscle tone.[18] The DMD FORMULA content has the *Guna* of *Rasayan*, is *Balya, Brumhaniya, Yogavahi* and *Ojovardhak*. *Kanchanar gugullu* was helpful for the pseudohypertrophy noted of calf muscles.[18]

CONCLUSION

As there is no cure for DMD, Ayurvedic medicines have great potential in management of it without side effects. As Acharya Charak, *Vatavyadhis* are *Kashta Sadhya* and in chronic state they are *Asadhya*. Although complete cure not possible but quality of life of patients improves. Thus, the present study improve the functional and physical capacity of activity, reduce the rate of disability to interrupt further evolution or development of disease and helps to retain the process of ambulation for a longer period of time along with its improved quality of life and activities of daily routine.

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