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# A Case Study on Duchenne Muscular Dystrophy

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## ABSTRACT

Duchenne muscular dystrophy (DMD) is the most common hereditary neuromuscular disease affecting all races and ethnic groups. The incidence is 1 in 3,600 live born infant boys. This is an X-linked recessive disease caused by mutation in the gene for the protein dystrophin which is located on the short arm of X chromosome in the Xp21 region. Its characteristic clinical features are progressive muscle weakness, delay in motor development, intellectual impairment, hypertrophy of the calves, and proliferation of connective tissue in muscles. Patient becomes unable to ambulate independently by age 10, wheel chair dependent by age 15 and die of cardio respiratory problems by age 25-30. In Ayurveda it can be classified under *Adibalapravrittavyadhi* and the pathogenesis occurs due to the *Beejbhaga Avayavadushti* which leads to *Vataprakopa* takes *Sthansamshraya* in *Medomamsadhatu* and depletes them. There is no specific treatment in any system of medicine and disease prognosis is unpreventable. Ayurveda instills a regenerative mechanism in neuromuscular disorders with special concern to *Panchakarma*, *Rasayana*, *Rasa Oushadhi* etc. These do not proclaim to be curative as DMD is *Asadhyaanuvamshika Vyadhi*, but can provide a floor for better quality of life with a longer survival. The present case study is about treating a 4 years male child with DMD through Ayurveda. The reduction in symptomatology was seen after three sittings of *Panchakarma* procedures and internal medications with interval of 20 days.

**Key words:** *Dystrophin, Adibalapravritta, Panchakarma, Rasayana, Rasa Oushadhi.*

## INTRODUCTION

Duchenne muscular dystrophy is a genetically determined, progressive disease of skeletal muscles. It is the most common muscular dystrophy affecting 1 in 3600 boys born worldwide. DMD is inherited in an X-linked recessive pattern (defect at Xp21 locus). Females will typically be carriers for the disease while males will be affected. The son of a carrier mother has a 50% chance of inheriting the defective gene from his

mother. The disorder is caused by a mutation in the dystrophin gene, which codes for the protein dystrophin. Dystrophin is responsible for connecting the cytoskeleton of each muscle fibre to the underlying basal lamina. Without dystrophin, muscles are susceptible to mechanical injury and undergo repeated cycles of necrosis and regeneration. The regenerated muscle is damaged due to lack of dystrophin. The continuous cycle of damage and repair and eventually replacement of muscle with fibro-fatty tissue is responsible for progressive muscle wasting that begins at the age of 3-5 years, delay in motor development and eventually wheel chair confinement followed by premature death at about 30 years from cardiac or respiratory complications. In our classical texts, the direct correlation of DMD with any single disease is not available. Since almost all major neuromuscular disorders are identified with *Vata Dosha*, this condition can be considered as *Adibala-Pravritta Mamsa Vata Kshaya* due to *Srotorodha*. *Aatmakarmaja* and *Beejadosh* bring

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*Khavaigunya* at *Mamsadhatu* levels leading to vitiation of *Vata* which causes *Mamsadhatvagni* impairment. The depleted *Dhatvagni* forms *Aama* instead of proper *Mamsadhatu*. It is followed by vitiation of *Kaphadosha*. While *Srotorodha* produces hypertrophy in particular region, it also manifests as first *Prakopa* then depletion of *vata* ailment. This complex pathogenesis is responsible for progressive wasting and necrosis of the affected muscle fibres.

## CASE REPORT

A 4 yr male child was brought to Dept. of Kaumarbhritya presented with the complaints of;

- Frequent falls while walking,
- Difficulty in running and climbing stairs,
- Difficulty in getting up from lying down or sitting position.
- Weakness in both upper and lower limb

Patient has been suffering from above complaints since 1 year. According to child's mother, he was her first child delivered through NSVD having insignificant antenatal, natal history. Child attained all developmental milestones as per chronological age. He was diagnosed with DMD one year back by modern paediatrician on the basis of genetic testing and serum CPK levels.

## Examination

On general physical examination, the child had thin appearance, difficulty in getting up from sitting position, proximal weakness, calf hypertrophy and positive Gower's sign.

- Vitals - Normal
- CNS examination - Normal
- Motor System

Fasciculation and irritability - Absent

Muscle tone - Normal

Muscle bulk - Wasting of muscle

B/L Pseudohypertrophy of calf muscle

Power grade >3/5=both upper limb

grade>3/5 - both lower limb

Gait - Short step gait

Gower's sign - present

Reflexes - Deep tendon reflex = normal

Plantar reflex = decreased

## Dashvidha Pariksha

*Prakriti* : *VataKapha*

*Vikriti* : *Vatapradhanatridoshaja*

*Sara* : *Twak*

*Samhanana* : *Madhyama*

*Desha* : *Jangala*

*Satmya* : *Sarva Rasa*

*Satva* : *Madhyama*

*Ahara Shakti*: *Madhyama*

*Vyayama Shakti* : *Avara*

*Vayah* : *Kumara*

## Investigation

- Hb = 12g/dl
- ESR = 20mm in the first hour
- TLC = 5500 cells/cu mm
- Sr. CPK = 11,000
- MLPA test = Hemizygous depletion of axon 49 & 50

## Treatment Plan

Three sittings of *Panchakarma* procedures were planned each at 20 days interval

SN	Procedure	Drugs used	Duration
1.	<i>Aama Pachana</i>	<i>Panchkola Choorna</i> (1 gm twice a day)	3 days
2.	<i>Udvardana</i>	<i>Yavkulatha Choorna</i>	7 days
3.	<i>Abhyanga</i>	<i>Ksheerbala Taila</i>	7 days
4.	<i>Shastishali</i>	<i>Shali rice + Go-dugdh +</i>	7 days

	<i>Pinda Sweda</i>	<i>Bala Kwatha</i>	
5.	<i>Mamsa Rasa Basti</i>	<ul style="list-style-type: none"> <li>▪ <i>Kalka - Draksha, Kharjur</i></li> <li>▪ <i>Kwath - Shatavari, Yastimadhu, Guduchi, Bidarigandha</i></li> <li>▪ <i>Go-ghrita</i></li> <li>▪ <i>Mamsarasa</i></li> </ul>	7 days

Internal medications used were;

SN	Name of Drug	Dose	Duration	Anupana (vehicle)
1.	<i>Balchaturbhadra Churna</i> <i>Pravalpishti</i> <i>Kukkutandatwak Bhasma</i>	500 mg 100 mg 250 mg	Thrice a day	<i>Madhu</i> (honey)
2.	<i>Kumarkalyan Rasa</i>	125mg	Once a day	<i>Madhu</i> (honey)
3.	<i>Ashwagandharishta</i>	7.5 ml	Twice a day	Water
4.	<i>Balarishta</i>	7.5 ml	Twice a day	Water
5.	<i>Kalyanakghrita</i>	5 ml	Twice a day	Milk

### Effect of therapy

- Serum CPK level decreased to 9500 IU after 1<sup>st</sup> sitting, 8000 IU in 2<sup>nd</sup> sitting and after final sitting the level came down to 7000 IU.
- Frequency of falls while walking decreased.
- Decrease in calf muscle hypertrophy was observed.

- Patient was able to sit from lying down position.
- Muscle bulk was increased and power grade was >4/5 in both upper and lower limb.
- Gower sign = persists

### DISCUSSION

Breaking down the pathogenesis of the disease, means removing the *Srotorodha* and pacifying *Vatadosha*, was the main aim of this treatment. *Amapachana* and *Udvartana* helps in removing the *Srotorodha*. *Abhyanga* nourishes the tissue and increases strength. *Swedana* makes the body supple, removes stiffness, cleanses the *Srotas* and improves blood circulation. *Basti* therapy is considered as '*Ardhachikitsa*' and some physician praised it as complete treatment due to its curative, preventive and promotive aspects. It is the best choice of treatment for *Vata* disorders. *Mamsarasa Basti* used in this case is a type of *Brimhana Basti* which nourishes and rejuvenates the degenerative *Mamsadhatu*.

Both *Ashwagandharishta* and *Balarishta* cure *Vata* induced disorders, increase digestive power, nourishes and strengthen the body. *Ghrita* is *Vata-Pitta Shamaka* and also overcomes *Kaphadosha* due to *Samskaranuvaritana Guna*, *Rasayana* (immune modulator), *Balya* (enhance strength, immunity or bulk of the body), *Brimhana* (nourishment of the body), *Yogavahi* (bioavailability enhancer) and *Ojavaradhaka* (immune enhancer). *Kalyanakghrita* is *Ayu-Varna-Balaprada*, used in *Mandagni*, *Sosha*, *Kshaya*. *Kukkutanda Twak Bhasma* and *Pravala Pishti*, both are good sources of calcium, and are used effectively in the prevention of calcium deficient spinal contractures and bone deformities. *Kumarkalyan Rasa* improves immunity.

### CONCLUSION

There is no cure for DMD in any system of medicine. All can be done to improve the quality of life in DMD patients. By observing the case study it can be safely concluded that specific *Panchakarma* procedures

along with internal medications can improve function, ambulation and thus increase life expectancy of the diseased child.

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