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THERAPEUTIC EFFECT OF PAÑCAKARMA IN DYSFERLINOPATHY PRESENTING AS LIMB-GIRDLE MUSCULAR DYSTROPHY TYPE 2B: A CASE REPORT

Dr. D. Priyadarshini*[1], Dr. V. Lakshmana Prasad[2], Dr. K. Harshavardhana Appaji[3]

[1]PG Scholar, Department of Panchakarma, S.V. Ayurvedic College, Tirupathi, Andhra Pradesh.

[2]P.G Professor, Department of Panchakarma, S.V. Ayurvedic College, Tirupathi, Andhra Pradesh.

[3]HOD and Professor, Department of Panchakarma, S.V. Ayurvedic College, Tirupathi, Andhra Pradesh.

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***Corresponding Author**
Dr. D. Priyadarshini
PG Scholar, Department of Panchakarma, S.V. Ayurvedic College, Tirupathi, Andhra Pradesh.

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ABSTRACT

Dysferlinopathy is a rare autosomal recessive muscular dystrophy caused by mutations in the dysferlin gene, with a global prevalence of approximately 1–5 cases per million population. Its incidence is estimated to range from 1 in 200,000 to 1 in 1,000,000 live births; however, it is often underdiagnosed due to clinical variability and limited awareness. Clinically, it presents as limb-girdle muscular dystrophy type 2B and is characterized by progressive muscle weakness, difficulty in climbing stairs, calf muscle hypertrophy, elevated serum creatine phosphokinase levels, and a positive Gower's sign. Currently, there is no definitive cure in conventional medicine, and management remains largely supportive. In Āyurvedic perspective, this may be correlated with Māṁsa-medogata vāta. Pañcakarma therapies play a significant role in improving muscle function in both qualitative and quantitative perspectives. The intervention demonstrated notable improvement in muscle strength, functional mobility, and overall quality of life.

KEYWORDS: Dysferlinopathy, Limb-girdle muscular dystrophy, Pañcakarma, Māṁsa-medogata vāta.

INTRODUCTION

Muscular dystrophies are a heterogeneous group of inherited muscular disorders characterized by progressive muscle weakness, degeneration of skeletal muscle fibers, and gradual loss of functional capacity. Among these disorders, dysferlinopathy represents a subgroup of autosomal recessive muscular dystrophies caused by mutations in the DYSF gene, which encodes the dysferlin protein involved in muscle membrane repair. Deficiency or dysfunction of dysferlin leads to progressive muscle fiber damage and impaired regeneration, ultimately resulting in muscle weakness and disability. Dysferlinopathy clinically manifests as different phenotypes, most commonly Miyoshi Myopathy and Limb-Girdle Muscular Dystrophy Type 2B, depending on the pattern of muscle involvement. Miyoshi Myopathy typically presents in late adolescence or early adulthood with distal muscle weakness, particularly affecting the gastrocnemius and soleus muscles of the calf. Patients commonly experience difficulty standing on tiptoes, running, and climbing stairs, and laboratory investigations often reveal markedly elevated serum creatine phosphokinase (CPK) levels. As the disease progresses, weakness may spread to proximal muscles of the hip and shoulder girdle, presenting clinically similar to Limb-Girdle Muscular Dystrophy Type 2B. Despite advances in genetic diagnosis, there is currently no definitive curative therapy in modern medicine, and management mainly focuses on supportive care, physiotherapy, and symptomatic treatment.[1]

From an Ayurvedic perspective, progressive muscular degeneration seen in dysferlinopathy can be correlated with Māṁsa-medogata vāta. The progressive muscle wasting resembles Māṃsa Dhātu Kṣaya, characterized by depletion of muscle tissue and reduced strength.[2] Since the disorder arises due to genetic mutation, it can be conceptually considered under Ādibala Pravṛtta Vyādhi, which originates from defects in bīja (genetic factors). The predominance of degenerative changes indicates a Vāta-pradhāna Tridoṣaja Vyādhi. Additionally, the pathological involvement of muscle tissue reflects Māṃsavaha Srotoduṣṭi, leading to symptoms such as māṃsa śoṣa (muscle wasting) and daurbalya (weakness). Therefore, therapies aimed at pacifying Vāta, nourishing Māṁsa–Meda dhātu, and correcting Māṃsavaha and medovaha srotas are considered beneficial. Panchakarma Procedures such as

snehana (oleation), svedana (sudation) basti (medicated enema) could effectively serve this purpose.

CASE REPORT

A 21-year-old male patient, presented with complaints of generalized muscle weakness for the past two years. The weakness was gradually progressive and predominantly involved the proximal muscles of the lower limbs. The patient reported difficulty in climbing stairs, weakness in the waist region, difficulty in lifting weights, and difficulty in bending down. He was born of a consanguineous marriage and had a strong family history of muscular dystrophy involving his mother and grandparents. Due to the progressive nature of his symptoms, he initially visited an allopathic hospital where he underwent detailed clinical and laboratory investigations. Based on the findings, including genetic evaluation, he was diagnosed with dysferlinopathy associated with limb-girdle muscular dystrophy type 2B. Seeking better management and supportive care, the patient subsequently approached the S.V. Ayurvedic College and Hospital Tirupati.

General Examination

1. Aṣṭa Sthāna Pariksha[3]

1. *Nādi*	78 beats/min
2. *Mūtra*	3 to 4 times a day
3. *Mala*	once a day
4. *Jīhva*	*Alipitha*
5. *Śabda*	*Prakrutha*
6. *Sparśa*	*Prakrutha*
7. *Drk*	*Prakrutha*
8. *Ākrti*	*Madhyama*

2. Daśavidha Parīkṣā[4]

1. Prakṛti	Vata-pitta
2. *Vikṛti*	Māṁsa–medo dhātu Vikṛti
3. Sāra	*Madhyama*
4. *Saṃhanana*	*Madhyama*
5. *Pramāṇa*	*Madhyama*
6. *Sātmya*	*Madhyama*
7. *Sattva*	*Madhyama*
8. *Āhāraśakti*	*Madhyama*
9. *Vyāyāmaśakti*	*Avara*
10. *Vayaḥ*	*Madhyama*

Blood pressure: 130/90 mm of Hg

Respiratory rate: 14 breaths /min

Height: 172cm

Weight: 55kg

Oedema: Absent

Anaemia: Absent

Clubbing: Absent

Cyanosis: Absent

Family history: Present

Personal History

- Diet: Mixed
- Appetite: Moderate
- Sleep: Adequate
- Bowel habits: Regular

Ayurvedic Assessment

- Prakriti: Vata-Pitta
- Vikriti: Māṁsa–Medo Dhātu Vikṛti
- Dushya: Mamsa, medas
- Srotas involved: Mamsavaha, Medovaha
- Rogamarga: Bahyaroga marga
- Rogabala: Yapya
- Rogibala: Madhyama bala

Systemic Examination

Central nervous system	➢ Conscious and well oriented ➢ Higher mental functions- intact ➢ Cranial nerves- intact ➢ Involuntary movements- Absent
Cardiovascular system	S1S2 heard, no murmurs
Respiratory system	Normal vesicular breath sounds heard
P/A	No tenderness, soft, no organomegaly
Musculoskeletal system	➢ Facial muscle weakness ➢ Proximal muscle weakness in lower limbs ➢ Calf muscle hypertrophy and Ankle contractures ➢ Positive Gower's sign ➢ Difficulty in standing from sitting position and climbing stairs

Sensory Examination: All are Intact

Motor Examination

Motor	Right- upperlimb	Left- upperlimb	Right- lowerlimb	Left-lowerlimb
1. Power	5/5	5/5	-4/5	3/5
2. Tone	Normal	Normal	Normal	Normal
3. Bulk	Mid-arm - 30cm	Mid-arm - 30cm	Mid-calf - 40cm	Mid-calf - 41cm

Reflexes: Normal

CRITERIA FOR ASSESSMENT

The North Star Ambulatory Assessment (NSAA) is a functional scale used to evaluate ambulatory ability and motor performance in patients with muscular dystrophies.[5]

Investigations

Genetic Evaluation: To confirm the underlying etiology, Whole Exome Sequencing (WES) was performed.

MedGenome Labs Ltd.
3rd Floor, Narayana Nethralaya Building, Narayana Health City,
#258/A, Bommasandra, Hosur Road, Bangalore - 560 099, India.
Tel : +91 (0)80 67154989 / 990, Web: www.medgenome.com

MEDGENOME

DNA TEST REPORT - MEDGENOME LABS

Full Name / Ref No:		Order ID/Sample ID:	**675621/8016538**
Gender:		Sample Type:	Blood
Date of Birth / Age:		Date of Sample Collection:	NA
Referring Clinician:	Dr. Sireesha Yareeda, Nizam's Institute of Medical Sciences, Hyderabad	Date of Sample Receipt:	13th June 2023
		Date of Order Booking:	14th June 2023
		Date of Report:	4th July 2023
Test Requested:	**Whole Exome Sequencing**		

CLINICAL DIAGNOSIS / SYMPTOMS / HISTORY

Mr. *C. Satish Babu*, born of a consanguineous marriage, presented with clinical indications of facial muscle weakness, proximal muscle weakness in lower limbs, ankle contractures, calf muscle hypertrophy, positive Gower's sign, difficulty climbing stairs, high grade fever, cough and scanty sputum. His laboratory investigations showed normocytic normochromic RBCs, lymphocytopenia and critically elevated levels of CPK. There is a strong family history of muscular dystrophy in mother and grandparents. Mr. *C. Satish Babu* is suspected to be affected with limb girdle muscular dystrophy and has been evaluated for pathogenic variations.

RESULTS

LIKELY PATHOGENIC VARIANTS CAUSATIVE OF THE REPORTED PHENOTYPE WERE DETECTED

Gene# (Transcript)	Location	Variant	Zygosity	Disease (OMIM)	Inheritance	Classification$
***DYSF* (+)** (ENST00000410020.8)	Exon 26	c.2831G>A **(p.Trp944Ter)**	Homozygous	Miyoshi muscular dystrophy 1 (OMIM#254130); Limb-girdle muscular dystrophy-2 (OMIM#253601); Distal myopathy with anterior tibial onset (OMIM#606768)	Autosomal recessive	Likely Pathogenic **(PVS1, PM2)**
***GNE* (-)** (ENST00000642385.2)	Exon 12	c.2086G>A **(p.Val696Met)**	Heterozygous**	Nonaka myopathy (OMIM#605820)	Autosomal recessive**	Likely Pathogenic **(PM1,PM2,PP3,PP5)**

****This is an autosomal recessive disorder caused by bi-allelic (homozygous or compound heterozygous) pathogenic/likely pathogenic variants in the *GNE* gene. The assay has detected a single heterozygous Likely Pathogenic in *GNE* gene mentioned in the table above. No other clinically relevant variant is detected in the coding region and exon-intron boundaries of these genes. Kindly correlate clinically.**

Copy Number Variants CNV(s)

No significant CNVs for the given clinical indications that warrants to be reported was detected.

The genetic analysis revealed

1. Homozygous nonsense mutation in the DYSF gene

- Variant: c.2831G>A (p.Trp944Ter)
- Inheritance pattern: Autosomal recessive

This mutation associated with dysferlinopathy includes:

- Miyoshi Muscular Dystrophy
- Limb-Girdle Muscular Dystrophy Type 2B

Laboratory Investigations

- Normocytic normochromic red blood cells
- Lymphocytopenia
- Critically elevated CPK indicating muscle fiber damage.

Diagnosis: Mamsa-medogata vata.

Dysferlinopathy (Miyoshi Myopathy)

Treatment Protocol

Procedure	Ingedients	Duration
1. Abhyaṅga.[6] and Nāḍī Sveda.[7]	Nirguṇḍī Taila Nirguṇḍī Patra	3 days
2. Śaṣṭikaśālī Piṇḍa Sveda.[8]	Śaṣṭikaśālī Go Kṣīra Bala Mūla Kvātha	7 days
3. Mātrā Vasti.[9]	Bṛhat Saindhavādi Tailam	7 days
4. Vaitaraṇa Kṣīra Vasti.[10]	Guḍa – ½ Pala Saindhava Lavaṇa – 1 Karṣa Tila Taila – 60 ml Ciñcā Kalka – 1 Pala Go-kṣīra – 1 Prasrutha	Yoga vasti format (8days)

Matra vasti with Bṛhat Saindhavādi Tailam

Day of treatment	Date	Vasti retention time
1st day	10 - 12 - 2025	3 hours
2nd day	11 - 12 - 2025	3 hours
3rd day	12 - 12 - 2025	3 hours
4th day	13 - 12 - 2025	5 hours
5th day	14 - 12 - 2025	6 hours
6^{th} day	15 - 12 - 2025	3 hours
7^{th} day	16 - 10 - 2025	3 hours

Vaitaraṇa Kṣīra Vasti in yoga vasti format

Day of treatment	Date	Vasti retention time
1st day (Anuvāsana Vasti)	17- 12 - 2025	3 hours
2nd day (Āsthāpana Vasti)	18 - 12 - 2025	1 hour
3rd day (Anuvāsana Vasti)	19 - 12 - 2025	5 hours
4th day (Āsthāpana Vasti)	20 - 12 - 2025	1 hour
5th day (Anuvāsana Vasti)	21 - 12 - 2025	5 hours
6th day (Āsthāpana Vasti)	22 - 12 - 2025	30 min
7th day (Anuvāsana Vasti)	23 - 12 - 2025	2 hours
8th day (Anuvāsana Vasti)	24 - 12 - 2025	5 hours

RESULT

North Star Ambulatory Assessment (NSAA).[11]

Activity	Before treatment	After treatment
1. stand	2	2
2. walk	2	2
3. Stand up from chair	2	2
4. Climb box step Right	2	2
5. Climb box step Left	2	2
6. Gets to sitting	2	2
7. Jump	1	1
8. Run	1	1
9. Stand on one leg - Right	2	2
10. Stand on one leg - Left	2	2
11. Descend box- Right	1	2
12. Descend box- Left	2	2
13. Stand on heels	2	2
14. Rise from floor	1	1
15. Lifts head	2	2
16. Hop- Right	0	0
17. Hop-Left	0	0
TOTAL	26	27

NSAA Total Score: 34/34 (Normal)

The patient's NSAA total score improved from 26 to 27, primarily due to improvement in descending the box step (right side), while other activities remained stable. Subjectively, the patient reported reduced fatigue, improved walking ability, enhanced stair climbing capacity, and increased low back strength. Objective improvements included better lower limb control. Over all, the patient with Dysferlinopathy demonstrated mild functional improvement and better ambulatory performance following treatment.

DISCUSSION

Dysferlinopathy is an autosomal recessive muscular dystrophy caused by DYSF mutations, leading to defective dysferlin and impaired muscle repair. It manifests as either Miyoshi Myopathy or Limb-Girdle Muscular Dystrophy Type 2B, with progressive proximal weakness, calf hypertrophy, difficulty climbing stairs, and elevated CPK, while reflexes are usually normal. In Āyurveda, it corresponds to Māṁsa–Meda Dhātu vāta, where vitiated Vāta affects Māṃsa dhātu, medo dhatu along with Māṃsavaha Srotas, medovaha srotas causing muscle wasting (māṃsa kṣaya) and weakness (daurbalya) with accumulation of adipose tissue (medovriddi). Its hereditary, degenerative nature aligns with Ādibala Pravṛtta Vyādhi and Vāta-pradhāna Tridoṣaja Vyādhi, supporting therapies for Vāta pacification, muscle nourishment, and Srotas restoration.

In this case, a Pañcakarma-based intervention was administered

1. Abhyaṅga with Nirguṇḍī Taila and and Nāḍī Sveda with Nirguṇḍī Patra (3 days) – provided external oleation and sudation, improving microcirculation, joint mobility, and muscle pliability, while the medicinal herb Nirguṇḍī is known for its Vāta- Śamana and anti-inflammatory properties.
2. Śaṣṭikaśālī Piṇḍa Sveda using Śaṣṭikaśālī rice, Kṣīra and Bala Mūla Kvātha (7 days) – delivered heat and nutrient bolus therapy, promoting muscle strength, nourishment, and pain relief, with Bala (Sida cordifolia) supporting muscle regeneration and Vāta pacification.
3. Mātrā Vasti with Bṛhat Saindhavādi Tailam (7 days) – a medicated enema to balance the Vāta, strengthen the core muscles along with improvement in muscle co-ordination. and for the depletion of ama medo dhatu.
4. Vaitaraṇa Kṣīra Vasti (Yoga Vasti format, 8 days) – containing Guḍa, Saindhava Lavaṇa, Tila Taila, Ciñcā Kalka, and Go-kṣīra is designed for nourishing mamsa dhātu, for depleting medo dhatu, for correcting the pathology related to the mamsavaha & medovaha Srotas, and for improving muscular function.

Functional outcomes showed improvement: NSAA score increased from 26 to 27, mainly due to better performance in descending the box step. Subjectively, the patient reported reduced fatigue, improved walking ability, stair climbing, and increased low back strength.

CONCLUSION

The integrative Ayurvedic management of Dysferlinopathy demonstrated a mild but measurable improvement in terms of functional mobility, muscle strength, and quality of life. Objective measures includes NSAA score, showed enhanced ambulatory performance, while subjective reports indicated reduced fatigue, improved stair-climbing ability, increased independence, and better daily activity performance. Panchakarma therapies, targeting Vāta pacification, muscle nourishment, and Srotas correction, may serve as a supportive approach in genetically determined myopathies like Dysferlinopathy. These findings highlight the potential of Ayurvedic interventions as adjunctive care, warranting further studies to validate efficacy and standardize protocols.

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