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Case Study

PANCHAKARMA BASED THERAPEUTIC INTERVENTIONS IN GNE MYOPATHY- INSIGHTS THROUGH A CASE STUDY

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ABSTRACT

GNE myopathy is a rare autosomal recessive muscular disorder characterized by progressive skeletal muscle atrophy, primarily due to mutations in the GNE gene. In *Ayurvedic* perspective, this condition is categorized as an *Adibala Pravrutta Vyadhi*, where *Dushti* in *Beejabhaga Avayava* is considered the underlying pathogenic factor. This case study aims to evaluate the efficacy of a comprehensive *Panchakarma* protocol in improving muscle strength, functional mobility, and overall quality of life in a patient with GNE myopathy. Methods: A 29-year-old female patient, diagnosed with GNE myopathy, presented with bilateral lower limb weakness, difficulty in walking, and impaired ability to rise from a squatting position. The patient underwent an integrative *Panchakarma* management protocol, which included *Sarvanga Abhyanga, Dashamoola Kashaya Seka, Shashtika Shali Pinda Sweda, Mustadi Yapana Basti, Anuvasana Basti* with *Brihat Chagaladi Ghrita.* The therapeutic focus was on *Amaharana, Srotoshodhana* and *Brimhana,* aiming to restore systemic balance and muscle function. Results: Post-intervention, the patient demonstrated significant clinical improvement. Objective assessments revealed enhanced lower limb muscle strength, improved ability to walk independently, increased confidence in climbing stairs, and a notable improvement in the GNE Myopathy Functional Activity Score (GNEM-FAS). Subjective reports indicated better overall well-being and functional capacity. Discussion: This case highlights the potential role of *Panchakarma* therapies in the multidisciplinary management of GNE myopathy. The combined approach of *Srotoshodhana* and *Brimhana* may contribute to symptomatic relief and functional enhancement by improving microcirculation, reducing muscle wasting, and promoting systemic nourishment. Conclusion: Therefore, this case study highlights the potential of *Panchakarma* in improving the quality of life in such rare genetic disorders.

INTRODUCTION

GNE Myopathy, also known as Hereditary Inclusion Body Myopathy (HIBM) or Nonaka Myopathy, is a rare autosomal recessive neuromuscular disorder characterized by progressive skeletal muscle weakness and atrophy. It typically manifests in early adulthood, often between the ages of 20 and 40, and progresses slowly over time, predominantly affecting distal muscles, particularly in the lower limbs, while sparing the quadriceps until the later stages of the disease.

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The condition is caused by mutations in the GNE gene, which encodes a bifunctional enzyme- UDP-N-acetylglucosamine 2-epimerase/N-acetylmanno samine kinase- that plays a critical role in the biosynthesis of sialic acid. Sialic acid is a vital monosaccharide required for proper glycosylation of proteins and lipids, contributing to the stability and function of muscle cell membranes.

From the Ayurveda perspective, GNE myopathy can be considered an *Adibala Pravritta Vyadhi*, as it is primarily caused by inherent genetic defects. In this context, the disease arises due to *Dushti* of *Shukra* and

Shonita, which leads to improper formation and function of body tissues, manifesting as progressive muscle weakness and atrophy. This interpretation aligns with the Ayurvedic concept that congenital or hereditary disorders (classified under *Adibala Pravritta Vyadhi*) result from the inherent derangement of the body's essential tissues (*Dhatus*), especially *Shukra* and *Shonita*, which play key roles in tissue formation and nourishment.

According to *Dalhana's* commentary on *Susruta Samhita* (*Su. Su.* 24th Chapter), the term *Shukra Shonita Doshānuvaya* indicates that the *Vatadi Doshas* (*Vata*, *Pitta*, *Kapha*) are vitiated in the tissues of *Shukra* and *Shonita*, which contributes to the pathogenesis of disease.

In GNE myopathy, the accumulation of autophagic vacuoles and inclusion bodies can be correlated with *Ama* (*mala sanchaya*) in Ayurvedic terms, indicating the presence of undigested metabolic toxins. Therefore, the initial line of management should focus on *Amahara Chikitsa* (treatment aimed at eliminating *Ama* and improving digestion and metabolism).

Furthermore, due to the inhibition of the enzyme responsible for sialic acid synthesis, there is resultant structural instability and degeneration of muscle fibers. In this context, *Brihmana Chikitsa* (nourishing therapy) becomes the prime therapeutic approach, aiming to strengthen and rebuild the *Dhatus*, especially *Mamsa Dhatu*, and restore structural integrity. These therapies help nourish the affected *Dhatus* and enhance the patient's functional capacity.

Case Report

A 29-year-old female patient was apparently asymptomatic until five years ago. In 2020, her relatives noticed that she was crossing her legs while walking, although she herself did not perceive any abnormality at the time. She initially ignored the symptoms for about six months, after which she began experiencing reduced strength while walking associated with heaviness in bilateral lower-limb.

She consulted a neurosurgeon, who prescribed calcium supplements. However, she took them for one month without any noticeable improvement. Over time, she began having difficulty climbing stairs, particularly favoring the right leg over the left, and started using a railing for support. Descending stairs remained manageable initially.

Over the next 3-4 months, she experienced increasing difficulty in rising from a squatting position, requiring hand support. Over the following year, her condition progressed with difficulty standing on tiptoes to reach high shelves, and frequent falls.

She also tried Ayurvedic treatment during this period, but it did not lead to any improvement.

Eventually, genetic testing confirmed a diagnosis of Nonaka myopathy. She has now presented for further management upon recommendation.

No h/o upper limbs symptoms

No h/o bowel bladder symptoms

Past History

N/K/C/O DM, HTN

K/C/O Hypothyroidism since 5 years – left medicine 1 year back

Family History

Her mother was having similar complaints and died naturally in the year 2021

Her Uncle is having similar complaints

Table 1: Personal History

Ahara	Mixed diet
Rasa pradhana	*Sarva rasa*
Vihara	Nothing specific
Vyasana	None
Agni	*Madhyama*
Kostha	*Madhyama*
Nidra	Timing- Irregular Quality- Sound Quantity- Good
Emotional status	Normal

Rogi Pareeksha

Table 2: General Examination

General appearance: Healthy	Pallor- Absent
Built: Moderately built	Icterus – Absent
Height: 5.3 feet	Cyanosis – Absent
Weight: 68 kgs	Clubbing – Absent
Pulse rate: 74bpm	Lymphedenopathy– Absent
Blood pressure: 120/80 mm of Hg	Edema – Absent
BMI- 26.06 kg/m2	

Table 3: *Asta Sthana Pareeksha*

Nadi -74 Bpm	*Shabda- Prakrita*
Mutra - Prakrita	*Sparsha- Prakrita*
Mala- Prakrita	*Drik- Prakrita*
Jihwa – Lipta	*Akriti-Madyama*

Table 4: *Dasha Vidha Pareeksha*

Prakruti- Vata kapha	*Ahara shakti- Abhyavarana- Pravara* *Jarana- Pravara*
Vikruti – Vata	*Vyayama shakti- Madhyama*
Sara – Madhyama	*Pramana- Madhyama*
Samhanana – Madhyama	*Vaya – Madhyama*
Satva- Madhyama	
Satmya- Sarva rasa satmya	

Nidana Panchaka

Nidana: Beejabhaga avayava dushti

Poorvaroopa: Nothing specific

Roopa: Weakness in bilateral lower limb, difficulty in walking.

Upadrava: Nothing specific

Upashaya-Anupashaya: None

Table 5: *Samprapti Ghataka*

Dosha: Vata	*Sanchara sthana: Sarva shareera*
Dushya: Mamsa	*Vyakta sthana: Sarva shareera*
Agni: Mamsadhatvagnijanya	*Roga marga: Abhyantara*
Ama: Mamsadhatvagni mandyajanya	*Swabhava: Chirakari*
Udbhava sthana: Bheejabhaga avayava	*Sadhyasadhyata: Yapya*

Systemic Examination

Respiratory System: NAD

Cardiovascular System: NAD

Gastro Intestinal System: NAD

Gait: Myopathic Gait

Central Nervous System Examination

Higher Mental Functions: Intact

Cranial Nerves: Intact

Motor System Examination

Table 6: Muscle Bulk

	Right	Left
Mid arm	27 cm	27 cm
Mid forearm	22 cm	22 cm
Mid-thigh	54 cm	54 cm
Mid-calf	32 cm	32 cm

Table 7: Muscle Tone

	Right	Left
Upper Limb	Normotonia	Normotonia
Lower Limb	Normotonia	Normotonia

Table 8a: Muscle Power of Hip Muscles

Hip flexor- Iliopsoas	4/5	4/5
Hip extensor – Gluteus maximus	4/5	4/5
Hip abductor- Gluteus medius and minimus	5/5	5/5
Hip adductors-Adductor Magnus	3/5	3/5

Table 8b: Muscle Power of Lower-limb Muscles

	Right	Left
Plantar Flexion– Gastrocnemius and Soleus	5/5	5/5
Dorsiflexion– Anterior tibialis muscle	3/5	3/5
Extension of knee– Quadriceps femoris	5/5	5/5
Flexion of knee - Hamstrings	4/5	4/5
Extensor of toes – EDL, EDB, EHL	4/5	4/5
Flexor of toes – FD, HL, HB	4/5	4/5

Reflexes

Superficial Reflex

Jaw jerk: Normal

Abdominal reflex: Normal

Corneal reflex: Normal

Ankle jerk: Flexor

Deep Reflexes: Bilaterally Normal reflex

Sensory System Examination: Intact

Investigation

Creatinine kinase: 403 U/L (17/6/23) raised

Nerve conduction study report (17/6/23):

Mildly reduced EMR amplitude in lower limb motor neurons

MRI of lumbosacral spine (15/12/21): Atrophy of B/L paraspinal muscles in lumbar region

Muscle MRI (12/1/24): Atrophy of anterior tibial muscle, hamstrings, gastronimeus and soleus and sparing of quadriceps.

Genetic Testing Report (11/1/24): Nονακα Myopathy

Table 9: Treatment Protocol Adopted

1	*Sarvanga Abhyanga with Ksheerabala taila* followed by *Dashamoola Kashaya Seka*	3 days	26/4/25- 28/4/25
2	*Sarvanga Abhyanga* with *Ksheerabala taila* followed by *Shashtika Shali Pinda Sweda*	7 days	29/4/25- 5/5/25
3	*Mustadi Yapana Basti* *Anuvasana basti* with *Brihat Chagaladi Gritha*	10 days	6/5/25-15/5/25

Table 10: Ingredients of *Basti*

Ingredients	
Honey	60ml
Saindhava lavana	10g
Brihat Chagaladi Ghrita	70ml
Aswagandha kalka	20g
Mustadi Yapana Basti Kashaya	250ml
Mamsa Rasa	100ml
Total	510 ml

Anuvasana with *Brihat Chagaladi Gritha* - 60ml

Table 11: *Basti* Plan

6/5	7/5	8/5	9/5	10/5	11/5	12/5	13/5	14/5	15/5
A	N	N	N	N	N	N	A	A	A
	A	A	A	A	A	A			

Table 12: Assessment Parameters

	BT	**AT**
GNEM-FAS score	85	92
Muscle power Dorsiflexion (anterior tibialis muscle) Hip adductors (Adductor Magnus)	 3/5 3/5	 4/5 4/5
6- minute walk test	200-250 metres	300-400 metres

OBSERVATION AND RESULTS

Table 14: Observation and Results

	Treatment	**Result**
26/4/25-28/5/25	*Sarvanga Abhyanga* with *Ksheerabala taila* followed by *Dashamoola Kashaya Seka*	Mild reduction in heaviness of body.
29/4/25-5/5/25	*Sarvanga Abhyanga* with *Ksheerabala taila* followed by *Shashtika Shali Pinda Sweda*	Mild improvement in balance while walking and confidence while walking.
6/5/25-15/5/25	*Mustadi Yapana Basti*	Was able to climb the steps without the support of railing, able to walk for longer distance, was able to reposition herself in the bed.

DISCUSSION

GNE myopathy, also known as Hereditary Inclusion Body Myopathy (HIBM) or Nonaka Myopathy, is a rare autosomal recessive neuromuscular disorder characterized by progressive muscle weakness and atrophy. It is caused by mutations in the GNE gene, which encodes the bifunctional enzyme UDP-N-acetylglucosamine 2-epimerase/N-acetylmannosamine kinase. This enzyme plays a critical role in the biosynthesis of sialic acid, a vital component in glycoprotein and glycolipid structures of the cell membrane.

The disease typically manifests in early adulthood, with patients presenting weakness initially in distal muscles, particularly the anterior tibialis, leading to foot drop, followed by gradual involvement of proximal muscle groups. Despite the progressive nature of muscle wasting, respiratory and cardiac functions are usually preserved until late stages. The hallmark pathological features include the presence of rimmed vacuoles and inclusion bodies in muscle biopsy samples. GNE myopathy is an exceptionally rare neuromuscular disorder, with global prevalence estimates ranging from 1 to 9 cases per 1 million individuals.

In Ayurvedic terms, the pathogenesis of GNE Myopathy can be correlated with classical concepts of disease development. The mutation in the GNE gene represents a fundamental *Beeja Bhaga Avayava Dushti* (genetic or congenital defect in the tissue-forming elements), which is the root cause of the disease. This genetic defect leads to a disruption in sialic acid synthesis, which can be interpreted as *Sanga* (obstruction) within the metabolic pathways responsible for proper cellular function. As a result of this obstruction, there is impaired glycosylation of structural and functional proteins in muscle cells, leading to the accumulation of autophagic vacuoles, inclusion bodies, and abnormal protein aggregates within muscle fibers- representing the accumulation of *Ama* (undigested metabolic toxins or waste products).

The progressive accumulation of *Ama*, together with impaired clearance mechanisms, eventually leads to *Mamsa Dhatu Kshaya* (depletion and degeneration of muscle tissue), which manifests clinically as muscle weakness, atrophy, and functional impairment.

In this context, the disease manifestation occurs due to *Bheeja Bhaga Dushti* (vitiation of the genetic or reproductive component), which leads to *Kha Vaigunya* (functional defect) in the *Mamsavaha Srotas* (muscle tissue channels). This results in *Sangha* (obstruction) and *Mamsa Dhatu Agni Mandya* (weakness of tissue metabolic fire), ultimately causing the formation of *Ama* (toxic metabolic byproducts) and progressive *Mamsa Dhatu Kshaya* (muscle tissue degeneration). Therefore, the management should focus on initially *Amaharana* to remove the accumulation of autophagic vacuoles followed by therapies aimed at nourishing and strengthening the *Mamsa dhatu.*

Dashamoola Kashaya Seka: Since the patient presented with weakness and heaviness in the bilateral lower limbs, *Seka* was planned initially, considering that *Sanga* (channel obstruction) in the pathogenesis leading to the formation and accumulation of *Ama. Dashamoola Kashaya Seka* was chosen as a form of *Rooksha Swedana,* as *Rookshana* is the primary line of management in *Amaja* conditions. Most of the herbs in *Dashamoola* possess *Usna Veerya* (hot potency), *Katu Vipaka* (pungent post-digestive effect), and *Stambaghna* (anti-stiffness) properties, making the *Kashaya* ideal for reducing heaviness, promoting the digestion and removal of *Ama*, and alleviating *Sanga*. Therefore, it was planned as the initial line of management to address the underlying pathology and improve the patient's condition.

Shashtika Shali Pinda Sweda: Once the *Sanga* is removed the next step focuses on strengthening the *Mamsa dhatu* so *Shashtika Shali Pinda Sweda* has been opted. It is a procedure where a preparation of milk, *Bala Mula Kashaya* and *Shashtika Shali* is massaged over the body. It performs *Swedana*, *Snehana*, and *Brihmana* simultaneously. *Shashtika* possesses *Snigdha*, *Guru*, *Sheeta*, *Sthira*, and *Tridoshaghna* properties. It contains larger starch granules compared to other varieties, which confer high gelatinization temperature and superior thermal stability. Additionally, *Navara* contains Bowman-Birk Inhibitor (BBI), a well-known protease inhibitor primarily found in rice. BBI is reported to protect muscle tissue from proteolytic degradation, especially in conditions involving inflammation or muscle wasting. Its anti-inflammatory properties help in reducing inflammatory cytokine activity, which otherwise triggers excessive protease expression and muscle breakdown. Since the primary goal in this patient is *Brihmana* (nourishment and strengthening of tissues), the use of *Navara* has been adopted as an ideal treatment strategy.

Mustadi Yapana Basti: A specialized form of nourishing enema therapy is scheduled due to its *Vatashamaka*, *Brihmana* and *Rasayana* effect. Since *Vata* vitiation is seen in this disease pathology, this basti acts at root level by pacifying aggravated *Vata* and enhancing nourishment. It may help slow disease progression, improve strength, mobility and promotes functional restoration there by improving quality of life of the patient.

CONCLUSION

This case report of GNE myopathy showed marked improvement in heaviness of the lower limbs, confidence while walking, and overall functional capacity following Ayurvedic management with *Navara Dhara* and *Basti Chikitsa*. In modern medicine, GNE myopathy remains a challenging condition with no definitive cure, and the current management is primarily supportive. From an Ayurvedic perspective, the condition was understood as *Bheeja Bhaga Avayava Dushti.* The treatment initially focused on *Amaharana* using *Dashamoola Kashaya Seka*, followed by *Shashtika Shali Pinda Sweda* to provide a *Brihmana* effect. *Mustadi Yapana Basti* was administered further to pacify *Vata Dosha* and promote functional restoration by acting as both *Brihmana* and *Rasayana* therapy. The outcome suggests that Ayurvedic interventions can offer an effective treatment approach for managing such rare genetic disorders.

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