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Case Report

Charcot-Marie-Tooth

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Management of Charcot-Marie-Tooth Disease through Ayurveda Principles - A Case Report Highlighting Beejadushti with Mamsagata Vata

Meghana V[1*], Girish Kumar SV[2], Meghana SK[3]

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[1*] Meghana V, Final Year Post Graduate Scholar, Department of Kaumarabhritya, Shri Dharmasthala Manjunatheshwara Institute of Ayurveda and Hospital, Bengaluru, Karnataka, India.

[2] Girish Kumar SV, Associate Professor, Department of Kaumarabhritya, Shri Dharmasthala Manjunatheshwara Institute of Ayurveda and Hospital, Bengaluru, Karnataka, India.

[3] Meghana SK, Final Year Post Graduate Scholar, Department of Kaumarabhritya, Shri Dharmasthala Manjunatheshwara Institute of Ayurveda and Hospital, Bengaluru, Karnataka, India.

Charcot-Marie-Tooth (CMT) disease is the most common inherited neuromuscular disorder affecting peripheral nerves, leading to progressive muscle weakness, sensory loss, and foot deformities. It occurs in approximately 1 in 2,500 individuals and is primarily caused by genetic mutations affecting the myelin sheath or axons. Prognosis varies by type, and conventional management includes surgical interventions, orthotic support, and pharmacologic therapy such as NSAIDs and tricyclic antidepressants. In Ayurveda, symptoms such as Mamsa Kshaya (muscle wasting), Sparshahani (sensory loss) and Balakshaya (weakness) suggest vitiation of Vata and Kapha Doshas, correlating with Vatavyadhi and Mamsagata Vata. CMT, being a genetically predisposed condition (Beejadushti), may be considered Asadhya Vyadhi. However, Ayurvedic interventions like Panchakarma and Shamanaushadhi offer symptomatic relief. A 4-year-old female diagnosed with CMT presented with bilateral foot drop, claw hands, and muscle weakness. She was admitted to the Kaumarabhritya department of SDM Institute of Ayurveda, Bengaluru, and treated with Panchakarma therapies including Sarvanga Abhyanga and Matra Basti. The patient showed significant clinical improvement on post-treatment. While CMT remains incurable due to its genetic basis, Ayurvedic management can provide symptomatic relief and improve quality of life.

Keywords: Mamsagatavata Vyadhi, Beejadushti, Charcot-Marie-Tooth disease, Panchakarma procedures, Case Report

Corresponding Author

Meghana V, Final Year Post Graduate Scholar, Department of Kaumarabhritya, Shri Dharmasthala Manjunatheshwara Institute of Ayurveda and Hospital, Bengaluru, Karnataka, India.

Email: meghanavshetty98@gmail.com

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Introduction

Charcot-Marie-Tooth disease (CMT) is the most common inherited neuromuscular disorder that mainly affects the peripheral nerves, leading to muscle weakness, sensory loss and foot deformities. It is a progressive condition which affects about 1in 2,500 people,[1] caused by genetic mutations, primarily affecting the myelin sheath or axons of peripheral nerves. The common presentation includes muscle weakness especially in the feet-lower legs-hands and forearms; foot demormities either high arches or flat feet; decreased reflexes; numbness or reduced sensation in the affected areas; difficulty with balance and walking; possible hand dexterity challenges.[2] Most individuals with CMT exhibit symptoms in the first or second decade of life, with an insidious onset of weakness that begins in the lower extremities and later involves the upper extremities.[3,4] The prognosis and treatment depends on its type. There are no medical therapies for CMT, but physical and occupational therapy can be beneficial as can bracing (ex-ankle foot orthotics for foot drop) and other orthotic devices.[5] Though there is no complete cure for CMT but treatments focus on managing symptoms and improving quality of life. Charcot-Marie-Tooth (CMT) disease is a genetically predisposed condition where both *Vata* and *Kapha Dosha* along with the *Mamsa Dhatu* (muscle tissue) become vitiated, ultimately leading to the manifestation of symptoms like *Mamsa Shosha* (tissue depletion), *Balakshaya* (weakness), *Gaurava* (heaviness), *Stambha* (Stiffness), *Supti* (numbness), *Cheshtahani*.[6] There is no direct correlation for this condition, but it can be comparable with the conditions like *Vatavyadhi* and *Mamsagata Vata*.[7] The present clinical study can be diagnosed as *Beejadushti* with *Mamsagata Vata*. Though the disease is incurable due to its pathogenesis w.s.r to *Beejadushti*. However, symptomatic management and enhancement of quality of life is possible through *Ayurvedic Panchakarma* line of management.[8]

Case Report

Patient information: A 4-year-old female child with k/c/o CMT, came with h/o difficulty and slapping of bilateral foot while walking, b/l claw hands, b/l foot drop and muscle weakness since the age of 3 years.

First child of non-consanguineous parents, was a term baby and extracted through LSCS, cried immediately after birth, weighing 3.5kg. No external congenital anomalies found and no H/O of Birth Asphyxia/ Neonatal Jaundice/ Rh incompatibility, NICU stay.

The child was exclusively breast fed for 8 months after which top feeding with semisolid foods. There was noticeable delay in Gross motor developmental milestones like Crawling (1.2 years), Walking with support (1.7 years), Walking without support (2 years) and fine motor developmental milestones like Palmar grasp (1 year), Pincer grasp (Not attained).

At 3.9 years of age, child was taken to National Institute of Mental Health and Neuro Sciences hospital and consulted paediatric neurologist, where the condition is diagnosed as Charcot Marie- tooth disease, based upon genetic analysis.

Further the child was suggested with Physiotheraphy and B/L AFO solid braces. Later at the age of 4 yrs child was brought to our hospital for the better management of same.

Table 1: Developmental milestones of the child

Gross motor	Normal age	Attained age
Neck control	3rd month	3rd month
Sitting with support	6th month	6th month
Sitting without support	8th month	10th month
Crawling	9th month	1.2 years
Walking with support	1 year	1.7 years
Walking without support	1.3 year	2 years

Fine motor	Normal age	Attained
Unidextrous grasp	6th month	9th month
Palmar grasp	10th month	1 year
Pincer grasp	1 year	Not attained

Personal and Social motor	Normal age	Attained
Social smile	2nd month	2nd month
Recognizing mother	3rd month	3rd month
Smiles at mirror image	6th month	6th month
Waves bye bye	9th month	9th month

Speech and Language	Normal age	Attained
Turns head to sound	1st month	1st month
Cooing	3rd month	3rd month
Laugh loud	4th month	4th month
Monosyllable	6th month	6th month
Bisyllable	9th month	9th month

Clinical Findings

On Physical examination

HMF - conscious, alert, oriented to person-place-time, memory-intact, speech-intact.

Muscle bulk - B/L LL wasting **+ B/L Champagne bottle legs +**

Muscle tone - B/L hypotonia

Muscle power - UL- Rt- 4/5, Lt- 4/5

LL- Rt- 3/5, Lt- 3/5

ROM -

UL- Extension of B/L hands - Partial range of movement

LL- Dorsi and plantar flexion - Partial range of movement

DTR - B/L Areflexia

Tightness - TA (Tendon achelis muscle) tightness+,

No secondary impairment

Balance

Static - standing
Dynamic - fair
Romberg sign - Positive

Gait - Steppage gait

Clinical Diagnosis

Genetic test - Gene# (Transcription)- GDAP1(+), Location- Exon 6, Heterozygous

Disease - **Axonal Charcot-Marie - Tooth disease type 2k**

Inheritance - Autosomal dominant; Autosomal recessive

Therapeutic Interventions

1st Sitting of Treatment [From 1-08-2024 To 10-08-2024; 10 DAYS]

Figure 1: Bilateral Champagne bottle legs

Figure 2: Bilateral flat foot

Table 2: *Panchakarma* Procedures and Internal Medications adopted during the admission

Date	Treatment	Internal medications
1/08/2024 To 2/08/2024	▪ Udwarthana with Kolakulathadi Choorna and Triphala Choorna f/b Dashamoola Kwatha and Dhanyamla Parisheka – 2 days	▪ Mashabaladi Kashaya & Astavarga Kashaya 10ml each with 20ml of warm water. ▪ Kumarakalyanaka Rasa with gold ½ -0- ½ B/F
3/08/2024 To 10/08/2024	▪ Ashwagandha Bala Lakshadi Taila (ABL) and Mahamasha Taila Abhyanga f/b Shashtika Shali Pinda Sweda - 8 days ▪ Masha Godhumadi Upanaha for B/L LL - 8 days ▪ Matra Basti (Ashwagandha Ghrita and Dhanvantara Taila - 20 ml each) - 8 days ▪ Vastraveshtana with Ashwagandha Bala Lakshadi Taila (ABL) and Mahamasha Taila	

	Discharge advice for 20 days: ▪ § Mahakukkutamamsa Taila - Sarvanga Abhyanga f/b Snana ▪ § Kumarakalyanaka Rasa with gold ½ -0- ½ with Ashwagandha Ghrita 5g-0-5g, B/F ▪ § Mashabaladi Kashaya & Astavarga Kashaya - 10ml each with 20ml of warm water, BD, A/F.

2nd Sitting of Treatment [From 1-09-2024 To 10-09-2024; 10 DAYS]

Table 3: *Panchakarma* Procedures and Internal Medications adopted during the admission

Date	Treatment	Internal medications
1/09/2024 To 2/09/2024 3/09/2024 To 10/09/2024	▪ Udwarthana with Kolakulathadi Choorna and Triphala Choorna f/b Dashamoola Kwatha Parisheka - 2 day ▪ Ashwagandha Bala Lakshadi and Dhanwanthara Taila Abhyanga f/b Mamsa Pinda Sweda - 8 days ▪ Masha Godhumadi Upanaha for B/L LL - 8 days ▪ Matra Basti (Ashwagandha Ghrita and Dhanvantara Taila-20 ml each) - 8 days ▪ Vastraveshtana with Ashwagandha Bala Lakshadi Taila (ABL) and Mahamasha Taila	▪ Nuro XT 1/2-0-1/2 B/F - 10 days ▪ Mashabaladi Kashaya & Astavarga Kashaya 10ml each with 20ml of warm water ▪ Kumarakalyanaka Rasa with gold ½ -0- ½ B/F
	Discharge advice for 20 days : ▪ Mahavishagarbha Taila+Sahacharadi Taila - Sarvanga Abhyanga f/b Dashamoola Dhara ▪ Makaradwaja 0-0-1 B/F ▪ Nuro XT 1-0-0 B/F Both above drugs taken with Ashwagandha Ghrita ½ tsp BD ▪ Mashabaladi Kwatha ▪ Ashtavarga Kashaya 10ml each with 20ml of warm water BD, A/F	

3rd Sitting of Treatment [From 11-10-2024 To 20-10-2024; 10 DAYS]

Table 4: *Panchakarma* Procedures and Internal Medications adopted during the admission

Date	Treatment	Internal medications
11/10/2024 To 12/10/2024	▪ Udwarthana with Kolakulathadi Choorna and Triphala Choorna f/b Dashamoola Kwatha Parisheka – 2 day	▪ Nuro XT 1/2-0-1/2 B/F– 10 days ▪ Mashabaladi Kashaya &Astavarga Kashaya 2tsp each with 4tsp of warm water ▪ Makaradwaja 0-0-1 B/F
13/10/24 To 20/10/24	▪ Sarvanga Abhyanga with Mahavishagarbha Taila and Sahacharadi Taila f/b Mamsa Pinda Sweda - 8 days ▪ Masha Godhumadi Upanaha for B/L LL - 8 days ▪ Matra Basti (Ashwagandha Ghrita and Dhanvantara Taila-20 ml each) - 8 days ▪ Vastraveshtana with Ashwagandha Bala Lakshadi Taila (ABL) and Mahamasha Taila	
	Discharge advice for 20 days: ▪ Mahavishagarbha Taila+Sahacharadi Taila – Sarvanga Abhyanga f/b Dashamoola Dhara ▪ Makaradwaja 0-0-1 B/F ▪ Nuro XT 1-0-0 B/F Both above drugs taken with Ashwagandha Ghrita ½ tsp BD ▪ Mashabaladi Kwatha ▪ Ashtavarga Kashaya 10ml each with 20ml of warm water BD, A/F	

Follow-Up and Outcomes

Table 5: Improvements observed after each sitting, are as follows:

After 1st Sitting	After 2nd Sitting	After 3rd Sitting
▪ Improvements seen in the increased muscle strength of B/L LL. ▪ Mild decrease in the stiffness and pain of B/L LL while walking	▪ Improvements seen in the increased muscle power of B/L LL (4/5) ▪ Slight Pincer grasp attained ▪ Able to walk for some distance with minimal difficulties.	▪ Improvement seen in the muscle bulk of B/L LL. ▪ Able to walk for minimal distance without difficulties ▪ Able to hold the pen/pencil (pincer grasp) for few minutes and able to write a word ▪ Reduction seen in the slapping of B/L feet while walking (gait improved)

Results

Table 6: Effect of overall treatment accessed by MMT[9] (Manual muscle testing) grade

MMT grade	Left limb				Right limb			
	BT	At the end of 1st Sitting of treatment	At the end of 2nd Sitting of treatment	At the end of 3rd Sitting of treatment	BT	At the end of 1st Sitting of treatment	At the end of 2nd Sitting of treatment	At the end of 3rd Sitting of treatment
Hip	3/5	3/5	4/5	4/5	3/5	3/5	4/5	4/5
Knee	3/5	3/5	4/5	4/5	3/5	3/5	4/5	4/5
Ankle	3/5	3/5	2/5	2/5	3/5	3/5	2/5	2/5

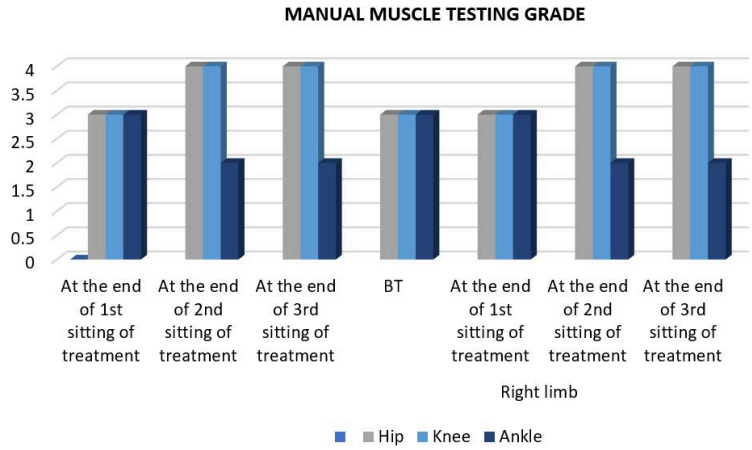

Figure 3: Effect of overall treatment accessed by MMT grade

Table 7: Effect of overall treatment accessed by ONLS[10] (Overall neuropathy limitations) scale

Parameters	Scores			
	BT	At the end of 1st Sitting of treatment	At the end of 2nd Sitting of treatment	At the end of 3rd Sitting of treatment
Arm scale score	4/5	4/5	4/5	3/5
Leg scale score	4/7	4/7	3/7	3/7
Total ONLS score	8/12	8/12	7/12	6/12

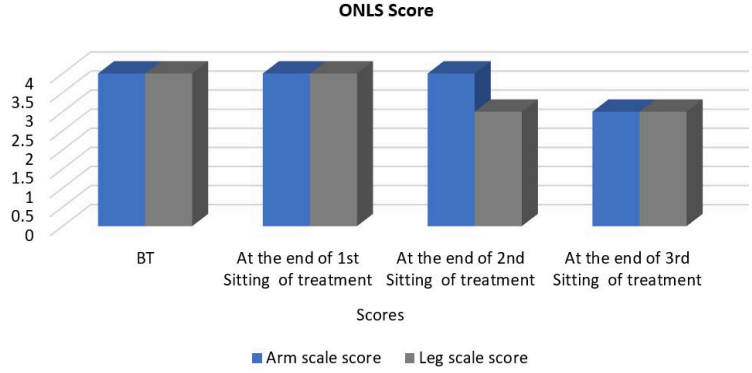

Figure 4: Effect of overall treatment accessed by ONLS

Table 8: Effect of overall treatment accessed by CMTNS[11] (CMT Neuropathy score in Children)

CMTNS Scores	BT	At the end of 1st Sitting of treatment	At the end of 2nd Sitting of treatment	At the end of 3rd Sitting of treatment
Sensory symptoms	2/4	2/4	1/4	1/4
Motor symptoms legs	2/4	2/4	2/4	1/4
Motor symptoms arms	3/4	2/4	1/4	1/4
Pin sensibility	2/4	2/4	1/4	1/4
Vibration	2/4	2/4	1/4	0/4
Strength legs	3/4	3/4	2/4	2/4
Strength arms	3/4	2/4	2/4	2/4
Ulnar CMAP (Median)	1/4	1/4	1/4	1/4
Ulnar SNAP (Median)	1/4	1/4	1/4	1/4
Total score	19/36 (Moderate Neuropathy)	17/36 (Moderate Neuropathy)	12/36 (Mild Neuropathy)	10/36 (Mild Neuropathy)

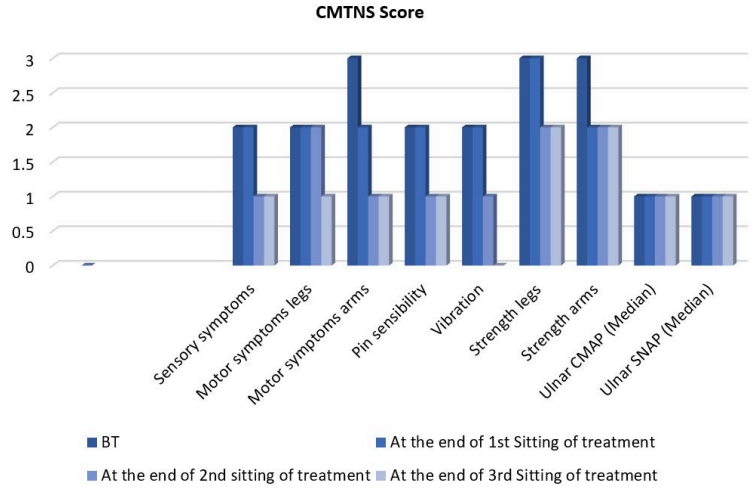

Figure 5: Effect of overall treatment accessed by CMTNS.

Discussion

Charcot-Marie-Tooth disease, an inherited neurological disorder that affects the peripheral nerves, which connect the brain and spinal cord to muscles and sensory organs. This condition primarily leads to progressive muscle weakness,**[12]** atrophy and sensory loss, especially in the limbs. In *Ayurveda*, by considering the genetic influence, this condition can be considered as *Beejadushtigata Vikara*, where the *Dushita Beeja* which is responsible for the formation of *Mamsa* is affected. Due to which the *Prakupita Vata* take *Sthana Samsharaya* in *Mamsa Dhatu* at the same time in addition to *Beeja Dushti* certain other factors like *Garbhopaghatakara Bhavas*, *Vatakara Ahara-Vihara* during pregnancy,**[13]** once again causes vitiation of *Vayu* which is already taken shelter in *Mamsa Dhatu.* Ultimately the *Prakupita Vata* causes further deterioration of *Mamsa Dhatu* which finally leads to the clinical manifestation in the form of *Mamsa Shosha* (tissue depletion)*, Mamsa Daurbalya* (muscle weakness), *Balakshaya*, pain (*Shoola*), *Gaurava* (heaviness)*, Supti* (numbness),*Cheshtahani* (restricted movements). Though the disease is incurable due to its pathogenesis w.s.r. to *Beejadushti,* but considering the predominance of *Dosha* especially *Vata Dosha*, symptomatically the condition can be managed through *Rasayana* therapies,**[14]** *Shamanoushadhis* and various *Panchakarma* therapies.

In the present clinical study, the condition has been diagnosed as *Beejadushti* with *Mamsagata Vata* where *Panchakarma* therapies like *Abhyanga, Swedana, Matra Basti, Upanaha* etc., has been adopted.

Udvarthana aids in eliminating *Srotorodha* (obstruction of channels), there by *Poshakarasa* of *Mamsadhatu* get nourished. It removes *Kaphavarana* there by address the *Mandaguna* or *Chestahani* caused by *Kapha Dosha*. *Kolakulathadi Choorna* & *Triphala Choorna* due to its *Vata Kapha-Hara* properties,**[15]** *Ushna Virya* and *Srotogamitva* quality, further it enhances the benefit of *Udvarthana*.

Abhyanga **-** basically, *Vatashamaniya* and *Bhrimana* in nature. It stimulates blood circulation and lymphatic flow, enhancing tissue nourishment and detoxification. This supports *Rasa Dhatu* and *Utharothara Dhatus*. However, in this case *Ashwaganda Bala Lakshadi Taila, Dhawanthara Taila, Mahamasha Taila* were used, which helps in addressing the specific vitiated qualities of *Vata Dosha* like *Chestahani, Mamsakshaya, Balahani*.**[16]** *Sahacharadi Taila* is also used, which is specifically indicated in *Adhogata Vata*.**[17]**

Swedana having *Sthamahara, Gouravahara* and *Sheetahara* qualities. In the present case instead of *Nadi Sweda, Bhashpa Sweda* or *Patrapinda Sweda* - *Parisheka Sweda* with *Dashamoola Kashaya***[18]** was used, which has *Vatashamaka* quality. *Parisheka Sweda* is *Mrudu Sweda*, which is highly indicated in paediatric practice, considering the *Soukumaryata, Alpabala* of the *Bala*. At the same time this type of *Swedana* is helpful in removing the *Sthamba* and *Gourava*, which is very much essential in the management of CMT, where the *Balakshaya* and *Cheshtahani* is present. *Dhanyamla Dhara* which is helpful in removing the *Kapha Avarana*. *Upanaha Sweda* - penetrate deeply into the tissues and here *Masha-Godhumadi Upanaha* was used, which is helpful in improving *Mamsa-Bala* and removes *Chestahani.*

Matra Basti is a type of *Anuvasana Basti*, which can be used for longer duration and can be administered at any time and doesn't have any complications. It gives nourishment to *Dhatus*, reduces inflammation and improves mobility. *Ashwagandha Ghrita***[19]** *and Dhanvantara Taila***[20,21]** is used, which is a type of *Sneha* called *Yamaka Sneha,* which has deep tissue penetration quality and enhances overall tissue health and helps to slow the progression of muscle degeneration, improve movement and alleviate discomfort, making it a valuable therapeutic approach in the management of neurological disorders. Orally ***Mashabaladi Kashaya***[22] consists of drug like *Masha, Bala, Yava, Kulatta, Devadaru, Rasna, Eranda Moola,* having the properties like *Vata-Kapha Shamaka, Balya, Srotoshodhana* and *Ama-Pachana* and mainly indicated in *Mamsagata Vatavyadhi* and ***Astavarga Kashaya***[23] consists of drugs like *Bala, Sahachara, Eranda, Shunti, Rasna, Devadaru, Nirgundi, Lasuna* have the properties of *Vatashamaka, Balya, Rasayana* and *Sroto Shodhana*, act as anti-inflammatory, analgesic and potentially slow the progression of the disease. ***Kumarakalyanaka Rasa*** is a compound preparation, having *Swarna Bhasma* as one of the main ingredients which act as *Rasayna, Nadi Balya* and has natural scavenger activities.**[24]**

Conclusion

In *Ayurveda* there is no direct correlation, but it can be compared with *Beejadushti* with *Mamsaghata Vata*. Considering *Beejadushti,* it can't be treatable, it is an incurable disease. But symptomatically we can manage, through various *Panchakarma* procedures and *Shamanoushadhis.* Among them *Swarna* and *Swarna* preparations are highly indicated in this condition which act as *Rasayana*, immuno-modulator, antioxidant, antimicrobial, nervine tonic. Very limited data is available regarding CMT disease, further exploration is needed.

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