

AYURVEDIC MANAGEMENT OF SIALIDOSIS TYPE 1 – A CASE STUDY

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ABSTRACT

Background: Sialidosis is an autosomal recessive disorder caused by a mutation in the neuraminidase gene (NEU-1), leading to a deficiency of neuraminidase and resulting in abnormal intracellular accumulation of sialyloligosaccharides. There are two types of Sialidosis: Type 1 (milder) and Type 2 (severe). Sialidosis Type 1, also known as normomorphic or cherry-red spot myoclonus syndrome, usually appears in the second or third decade of life. It is a rare condition with an overall prevalence of about one in every 5 to 10 million. The exact pathophysiology of this disorder is not fully understood, but in Ayurveda, it can be seen as a vata vyadhi. Samprathi and smaprapthi ghatakas can be understood, and it can be managed as anuktha vyadhi focusing on smaprapthi vighatana to help control the progression of the disease. **Case report:** A 26- year- old male patient presented with intentional tremor and action myoclonus, accompanied by postural

instability. He sought care at an allopathic hospital three years ago, where further investigation confirmed a diagnosis of Sialidosis type 1, and he has been undergoing treatment. For additional management, he approached our hospital. Diagnosed as vatavyadi, he received treatment with basti and shamana chikitsa; improvement was observed after basti administration. **Conclusion:** Sialidosis can be classified as vatavyadhi, resulting from beeja dosha and margavarodha, with sanga and vimargamana of vata leading to chala guna vriddhi. Therefore, the treatment should aim to clear the margavarodha while simultaneously reducing

the chala guna of vata. Ayurveda can be beneficial in managing these chronic cases to control disease progression and minimize dependency on medication.

KEYWORDS: Sialidosis, Action myoclonus, Vatavyadhi, Basti, Samprapthi.

INTRODUCTION

Sialidosis is an autosomal recessive inherited disorder characterized by a mutation in the neuraminidase gene (NEU-1) located on 6p21.33. This mutation results in a deficiency of the enzyme neuraminidase, which impairs the degradation of sialylated glycoproteins and oligosaccharides, resulting in the accumulation of sialyloligosaccharides within lysosomes in the cytoplasm, often in neurons.^[1] There are two types of Sialidosis based on age of onset, disease progression, and clinical features: Sialidosis Type 1, known as normomorphic or cherry-red spot myoclonus syndrome, typically manifests in the second or third decade of life. Patients may exhibit gait abnormalities, reduced visual acuity, or both, associated with action myoclonus, intentional tremor, cerebellar ataxia, and hyperreflexia. Sialidosis type 2: has an earlier onset, usually in the first decade, and the symptoms are more severe, encompassing intellectual debility, hepatosplenomegaly, and spinal deformities, to name a few. It is a rare condition with a combined prevalence of one in every 5 to 10 million.^[2] The pathophysiology and treatment protocol of this disorder are not completely clear, but it involves progressive accumulation of metabolites leading to oxidative stress and neuronal degeneration.

In Ayurveda, although sialidosis is not mentioned as a specific disease. Through its presentation, especially myoclonus, ataxia, tremor, and visual abnormalities, it can be understood on the lines of anukta vata vyadi and treated through samprapti vigatana. Lakshanas, due to an increase in vata gunas like cala, ruksha, and lagu, largely contribute to the manifestation of tremors, muscle wasting, and progressive neurological debility.^[3]

In this case study, there is an attempt to highlight the role of Ayurvedic understanding and management of rare and progressive neurological diseases, which are not mentioned in the Samhitas but can be understood by the framework provided in them.

CASE DESCRIPTION

A 26-year-old male patient approached the OPD of SJIM, Government Ayurveda Medical College, Bengaluru, with complaints of

- Intention Tremor of bilateral upper and lower limbs.
- Action myoclonus
- Postural instability

The patient had first visited an allopathic hospital 3 years ago and was diagnosed with Sialidosis type 1 and was under medication. He does not have a history of hypertension, diabetes mellitus, or any other metabolic dysfunctions. He is a farmer with no addictions.

MEDICATION HISTORY

- Betacap 20 mg TID
- Ampanel 2 mg TID
- Clonam - IR 0.5 mg TID

DIAGNOSIS

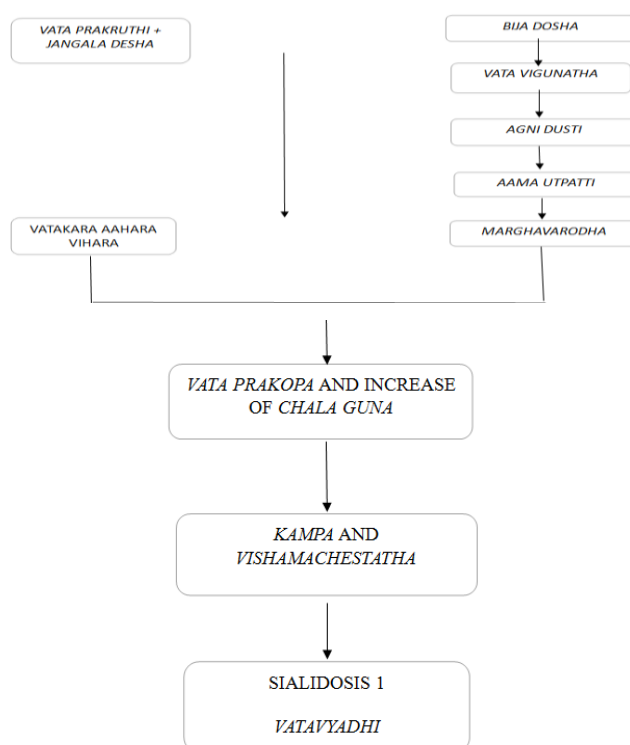
HETU

Bija Dusti: Parents had a consanguineous marriage, and his younger brother also suffers from a similar condition.

Janmastana: Yadgir, Karnataka – Jangala pradesha with vata pradanyata

Aharaja: Intake of ruksha lagu vatakara bojana, katu tikta rasa pradana bojana

SAMPRAPTI



SAMPRAPTHI GHATAKA

Dosha	Vata
Dushya	Sira
Srotas	Vatavaha
Srotodushti prakara	Atipravrutti, Vimargagamana
Agni	Jataragni mandya
Ama	Jataragni janya
Udbhavasthana	Pakwashaya
Sancharasthana	Sarvasharira
Vyaktasthana	Shakha
Rogamarga	Madhyama

TREATMENT GIVEN

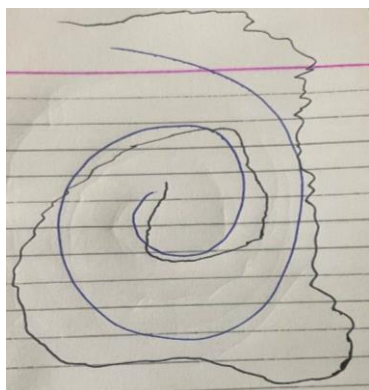
- Kosta shodana with Gandarvahastadi eranda taila – 60 ml for 3 days
- Basti Chikitsa – Kala Basti
 - Anuvasana – Mahamasha taila
 - Niruha – Mustadi yapana basti
 - Makshika -120ml
 - Lavana – 9 gm
 - Sneha – Ashwaganda ghrutha – 200ml
 - Kalka – Rasna + kapikachu + shatapushpa -50 gm
 - Kwatha – mustadi yapana basti Kashaya -500 ml: Total = 870 ml
- Abyanga with mahamasha taila
- Shamana Aushadhi – Agnitundi vati

TREATMENT OUTCOME

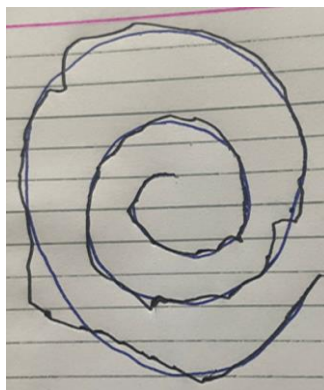
	Before treatment	After treatment
Tremor	Intentional tremor present	Reduction in tremor
Postural Instability	Romberg's Positive	Romberg's Negative
Dependence on Conventional medication	TID	OD
Appetite	Reduced	Improved

Archimedes Spiral

Before treatment



After treatment



DISCUSSION

Sialidosis 1 is a rare lysosomal storage disorder that has neurological, visual, and motor system involvement with no definitive cure in conventional medicine, and provides only symptomatic treatment. It is a progressive disorder causing progressive loss of vision, ataxia, tremors manifested in the whole body, and hearing loss, leading to a significant decrease in the quality of life of the patient since an early age. Therefore, Ayurveda enhances the understanding and management of such diseases by interpreting the lakshanas based on dosha vishamya and implementing the classical chikitsa based on samprapti vighatana in mitigating the progression of the disease.

Even though this condition is not mentioned by name in the Samhitas, its signs and symptoms are mentioned in the Vataprakopa lakshana, and it can be considered as Anukta vatavyadi and treated accordingly.^[3] The nidanas in this case, like bijadosha, jangala desha, ruksha laghu, and katu tikta pradana ahara sevana, lead to gunathaha and karmathaha vruddhi of cala guna of vata. If the nidana parivarjana is not done at the initial stage, then all the gunas of vata, along with other doshas, take place and give rise to further accumulation of the metabolites and worsening of the disease outcome.

The treatment principle of samprapti vighatana was adopted in these cases. At first, kostashodana with gandharvashatadi eranda taila was given, followed by basti treatment. Kala basti, i.e., the basti course of 16 days with 10 anuvasana basti with Mahamasha taila and 6 niruha with mustadi yapana basti was given. Mahamasha taila was used for both anuvasana and abhyanga as it is indicated in hasta kampa, and it is the best taila for urdhvajatru gata roga.^[4] Mustadi yapana basti has properties of both shodhana, which removes the

accumulated ama (intercellular cytoplasmic accumulation), and bhrumhana, which further arrests the vriddi of vata.^[5]

Clinical improvements were seen post treatment such as marked reduction in tremor along with postural instability. The appetite of the patient increased and his dependency on existing medication was cut down to single dose per day.

CONCLUSION

This case report highlights the use of Ayurvedic principles in the management of rare diseases that are not mentioned in the classics. Sialidosis, when understood and treated on the lines of Anuktha vata vyadi, gives a far more robust approach for its treatment.

Correct and precise interventions of basti chikitsa, especially mustadi yapana basti have proven to be effective in many neurological disorders due to its vata prashamana, srotoshodana and brimhana properties.^[6]

However, further clinical studies and documentation are required to establish the samprapti and treatment of Sialidosis type 1. This case study emphasizes on the ability of Ayurveda in the management of rare neurodegenerative disorders due to its pratipurusha sidhanta i.e. individualised treatment based on the patient's constitution and pathophysiology involved.

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