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Case Study

POTENTIAL ROLE OF AYURVEDA IN THE MANAGEMENT OF MAPLE SYRUP URINE DISEASE

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ABSTRACT

Maple syrup urine disease (MSUD) is a genetically inherited metabolic condition that follows an autosomal recessive pattern. It results from malfunction in the branched-chain alpha-ketoacid dehydrogenase (BCKAD) complex, essential for the breakdown of branched-chain amino acids (BCAAs) such as leucine, isoleucine and valine. A defective BCKAD complex impairs the normal degradation of BCCAs, leading to their accumulation. This condition is marked by neurological and developmental issues, feeding difficulties, a distinctive maple syrup smell in the urine, including encephalopathy. Elevated blood levels of BCAAs and increased urinary excretion of their corresponding ketoacids are hallmark feature. When treatment begins early, individuals often experience positive clinical outcomes. **Material and Methods-** In this case, a 22-year-old male patient who a known case of hypertension, was brought by his parents with concerns of recurrent seizures occurring two to three times per week for the past 8 years, global developmental delay noted since infancy along with recurrent upper respiratory tract infections, salivary dribbling, excessive eating habit, irritable behaviour– since childhood. Associated with reduced sleep since childhood and masturbatory behaviour since 15 years of age. Patient was treated with *Shamanaushadhi* as a part of the management protocol. **Results-** This intervention led to substantial improvement in multiple aspects of the patient's clinical presentation. **Conclusion-** The case indicates potential benefits of Ayurveda treatment in improving clinical outcomes and quality of life in an MSUD patient.

INTRODUCTION

Maple syrup urine disease (MSUD) occurs due to reduced or absent function branched-chain alpha-ketoacid dehydrogenase (BCKAD) complex, impairing BCAA catabolism. This enzyme complex is essential for breaking down the branched-chain amino acids (BCAAs): leucine, isoleucine and valine; initial metabolic step involves converting these amino acids into corresponding alpha-ketoacids via branched-chain aminotransferases within mitochondria. Unlike most amino acid pathways, this conversion primarily takes place in skeletal muscle rather than in the liver. BCAAs are vital for muscle growth and energy metabolism of the mTOR signalling cascade.

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In the brain, support neurotransmitter production, protein synthesis and cellular energy. When the BCKAD complex is impaired, it accumulates to toxic levels, disrupting cellular homeostasis. Accumulated BCAAs and their ketoacid by-products appear in the blood, brain and urine along with the presence of alloisoleucine- a key diagnostic marker[1].

Clinical Presentation

MSUD manifests in five recognized clinical types; classic, intermediate, intermittent, thiamine responsive and E3 deficient, though there is no clear link between genetic mutations and specific phenotypes[2]. The different forms are often classified by the age at which symptoms appear, how severe they are, whether the patient responds to thiamine therapy and biochemical test results. The classic and E3-deficient types generally emerge in newborns. In contrast, other forms such as intermittent, intermediate and thiamine-responsive MSUD can develop at any point in life, often triggered by stress or

illness. Typically, newborns present with symptoms such as poor feeding, developmental delays and an acute encephalopathy with a characteristic maple syrup odour in earwax and urine. Without timely intervention, it rapidly progresses to neurological damage, seizures, metabolic decompensation and fatal outcomes in infancy. The way MSUD presents is closely tied to how much residual enzyme activity is present, with clinical classification also influenced by how well the body processes leucine and handles metabolic stress.

Medical Management

Newborn Screening (NBS)

MSUD has a global incidence of approximately 1 in 1,85,000 live births. However, it is significantly more prevalent in certain founder populations, such as the Old Order Mennonites of Pennsylvania, where the frequency can be as high as 1 in 200 live births. Since MSUD responds well to dietary restriction of BCAAs (or thiamine supplementation in thiamine-responsive forms) and outcomes are generally favorable when treatment starts early, routine newborn screening is recommended.

Currently, NBS for MSUD is part of standard screening programs in the United States, five Canadian provinces, 22 European countries, two Latin American countries (Costa Rica and Uruguay) and eight countries in the Asia-Pacific region. Newborn screening for MSUD began in 1964 with the introduction of the bacterial inhibition assay for leucine on dried blood spots (Guthrie specimens). Modern NBS for BCAA metabolism disorders now commonly uses tandem mass spectrometry for quantitative plasma amino acid profiling. If elevated BCAA levels are found, further tests may include urine organic acid analysis by gas chromatography-mass spectrometry (GC-MS), the dinitrophenylhydrazine (DNPH) test and quantitative plasma amino acid testing using liquid chromatography-mass spectrometry (LC-MS), along with confirmatory molecular testing in suspected cases. Diagnosis of MSUD involves detecting high plasma levels of BCAAs and alloisoleucine, along with branched-chain alpha-hydroxy acids in urine. MS analysis also measures the leucine-to-isoleucine ratio, along with other amino acids like alanine, glutamine, tryptophan, methionine and histidine. Early diagnosis and prompt treatment can greatly improve outcomes. Treatment requires lifelong limitation of dietary BCAAs and consisting biochemical monitoring. Even with treatment, patients can experience metabolic crises requiring emergency care[3].

Case Report

Chief complaints

Parental concerns regarding recurrent seizures (twice or thrice in a week) – in the past 8 years.

Global developmental delay noted since infancy.

Recurrent upper respiratory tract infections, salivary drooling, excessive eating habit and irritable behaviour – since early childhood.

Associated complaints

Disturbed sleep pattern reported since childhood.

Masturbatory behaviour since the age of 15 years.

History of present illness

A male patient of age 22 years, the second-born of consanguineous parents, presented with recurrent seizures in the past 8 years with global developmental delay, frequent upper respiratory tract infections, salivary drooling, excessive eating habit and irritable behaviour since early childhood. Also associated with disturbed sleep since childhood and masturbatory behaviour since the age of 15 years. He was born at term via normal vaginal delivery with a normal cry at birth and birth weight of 3.5kg. The antenatal and perinatal periods were uneventful. However, feeding difficulties and persistent crying were noted in early infancy. Initial evaluations at that time revealed no abnormalities. He had global developmental delay, with both mental and motor milestones lagging behind age norms. At 8 months, he experienced his first focal motor seizure (right-sided) and was started on phenytoin (5mg/kg/day). Despite regular follow-up with a neurologist, he had a recurrence of generalized tonic-clonic seizures at the age of 1 year. At around one year of age, parents noticed a distinct foul odour in the urine. At that time, examination revealed an alert child with adequate physical growth, but no eye contact or response to verbal stimuli, along with motor stereotypies, repetitive head nodding, generalized hypertonia and brisk deep tendon reflexes. Ocular movements and fundi were normal. Routine blood tests were normal; however, arterial blood gas analysis showed metabolic acidosis. A positive Dinitrophenylhydrazine (DNPH) test and elevated BCAAs on HPLC confirmed the diagnosis of intermediate MSUD. MRI brain revealed diffuse white matter hyperintensity with involvement of the globus pallidus. He was started on thiamine, syrup carnitine (Caritor) and a protein-restricted diet, after which a noticeable improvement in both mental and motor development was observed. He was continued on a specialized MSUD diet and under regular metabolic and neurological follow-up. However, at the age of 15 years, he developed masturbatory behaviour, for which he was started on behaviour-modifying medications (Risdone-1 BD). Following this, he began

experiencing recurrent seizures (2–3 episodes per week). Despite increasing the dosage of antiepileptic medications, there was no significant improvement in seizure control. Due to the worsening neurological symptoms and inadequate response to ongoing treatment, the parents have now approached the OPD of Panchakarma, Government Ayurveda Medical College and Hospital, Bengaluru for further evaluation and management.

Past Medical History

N/K/C/O Diabetes Mellitus

K/C/O/MSUD under regular medication since infancy.

Tab. Carnisure 500 BD, Tab. Lacosam 100mg OD and150mg HS, Tab.Tiam 100 3BD, Risdone-1 BD, Tab. Clobium-10 OD, Tab. Petril 0.75 OD

And K/C/O/Hypertension since 9 years, on medication-Tab. Inderal 10 TID

Physiotherapy (from 1 year of age to 10 years) and speech therapy (at 3 years of age for 1 year) was given.

Birth and Developmental History

Mode of delivery- Full-term normal delivery

Birth weight- 3650grm

No any birth asphyxia or delayed crying.

Developmental milestones: Motor, language, social – delayed.

Table 1: Milestone achievement

Domains of development	Milestones	Age of attainment	Expected age of attainment
Gross motor	Head control, sitting without support	2 years 3 years	3 months 8 months
Fine motor	Immature pincer grasp	2 years	9 months
Personal and social	Ask for food when hungry	3 years	2 years
Speech	Monosyllables	2 years	6 months

Immunization History

Child was immunised as per the national immunization schedule appropriate for age.

Family and Social History

Consanguinity-second degree consanguineous union.

No family member with similar illness.

Socioeconomic status- Rich

Personal history

Table 2: Subject's personal history

Name: xyz	Bowel: Regular
Age: 22 years	Appetite: Excessive hunger
Sex: Male	Habits: None
Marital status: Unmarried	Height: 174cm
Diet: Vegetarian	Weight: 64kg

Table 3: *Ashta sthana pareeksha*

Nadi	*Prakruta,* 78bpm
Mutra	*Aprakruta* distinct foul smell 4-5times/day, 0-1 times/night
Mala	*Prakruta,* 1 time/day
Jihwa	*Lipta*
Shabda	*Aprakruta*
Sparsha	*Prakruta*
Drik	*Prakruta*
Akriti	*Sthoola*

Table 4: *Dashavidha pareeksha*

Prakriti: Vata pitta	*Satmya: Sarva rasa satmya*
Vikriti: Kapha pradhana tridosha	*Ahara shakti: Pravara*
Sara: Madhyama	*Vyayama shakti: Madhyama*
Samhanana: Pravara	*Vaya: Madhyama* (22 years)
Satva: Avara	*Pramana:* Ht- 174cm Wt- 64kg

Systemic examination

Cardiovascular system: S1 S2 heard, no abnormality detected.

Gastrointestinal system: P/A- soft, non-tender.

Respiratory system: Chest shape- normal, on auscultation- wheeze is heard in middle lung zones bilaterally.

Central nervous system

Higher mental functions

- Conscious but inattentive.
- Orientation to person and place only.
- Speech - Single selective words with slurred and indistinct articulation.
- Memory could not be reliably assessed due to poor cooperation.
- Cognitive function- Impaired

Cranial nerves- Within normal limits

Motor system

- Tone - Hypertonic
- Power – 4/5 in lower limbs and 5/5 in upper limbs bilaterally
- Bulk - Normal

Sensory system – No sensory deficit

Reflexes- All deep tendon reflexes were brisk, with an extensor plantar response bilaterally.

Gait – Spastic gait

Table 5: *Nidana panchaka*

Nidana	*Beejadushti*
Purvaroopa	-
Roopa	Recurrent seizures, global developmental delay, recurrent upper respiratory tract infections, salivary drooling, excessive eating habit, irritable behaviour, disturbed sleep pattern and masturbatory behaviour.
Upashaya	Protein-restricted diet and medications
Anupashaya	Protein rich diet

Table 6: *Samprapti ghataka*

Dosha	*Kapha Pradhana Tridosha*	*Udbhavasthana*	*Beejabhaga Avayava*
Dushya	*Rasa, Rakta, Mamsa, Meda* and *Majja*	*Sancharasthana*	*Sarva Shareera*
Agni	*Jatharagni, Dhatvagni*	*Vyaktasthana*	*Sarva Shareera*
Agnidushti	*Mandagni*	*Adhistana*	*Kosthta* and *Rakta*
Srotas	*Rasa, Rakta, Mamsa, Meda* and *Majjavaha*	*Rogamarga*	*Abhyantara*
Srotodushti	*Sanga, Vimargagamana*	*Sadhyasadhyata*	*Yapya*

Table 7: Treatment protocol adopted

Yoga	**Dose**	***Aushadha sevana kala***	***Anupana***	**Duration**
Ajamodadi churna	5gm BD	Before food	Lukewarm water	7 days
Manasamitra vati	1 BD	After food	Lukewarm water	Initially 1month
Saraswatarishta	10ml BD	After food	Lukewarm water	Initially 1month
Smritisagara rasa	1 BD	After food	Lukewarm water	After one month- given for 2 months with intermittent gap.
Brahmi vati	1 BD	After food	Lukewarm water	Continued until current evaluation

Table 8: Assessment before and after treatment

Assessment criteria	Before treatment	After treatment
Recurrent seizures	Twice or thrice in a week	No episode till last follow up
Recurrent upper respiratory tract infections	+	-
Salivary drooling	+	-
Excessive eating habit	+	-
Irritable behaviour	+	-
Disturbed sleep pattern	+	-
Masturbatory behaviour	+	Reduced
Allopathy medications	Tab. Carnisure 500 BD	Tab. Carnisure 500 BD
	Tab. Lacosam 100mg morning & 150mg HS	Tab. Lacosam 50mg HS
	Tab. Tiam 100 3BD	Tab. Tiam 100 3BD
	Tab. Risdone-1 BD	Tab. Risdone-1 HS
	Tab.Clobium-10 OD	Stopped
	Tab. Petril 0.75 OD	Stopped
	Tab. Inderal 10 TID	Tab. Inderal 10 morning

DISCUSSION

MSUD is a genetic metabolic condition resulting from reduced activity of the BCKAD enzyme complex. Mutations in both copies of the genes that encode components of this complex impair its function, leading to the buildup of branched-chain amino acids (BCAAs), which can be toxic to skeletal muscle and brain tissue. While MSUD lacks a direct reference in classical Ayurveda literature, treatment approaches can be customized on symptomatology. In this case, treatment focused on balancing metabolic disturbances by correcting *Agni* and supporting neurological function using *Shamanaushadhi* and diet, allopathy medications were tailored according to the patient's symptoms. Over time, notable improvement was observed in seizure frequency, sleep, behavioral symptoms and general health.

Ajamodadi churna

The patient was initially prescribed *Ajamodadi Churna* for *Ama pachana* and *Agni Deepana*, as he presented with symptoms like *Adhyashana, Lalasrava*, *Jihva liptata* and *Pratishyaya,* indicative of *Ama lakshana*. *Ajamodadi Churna* contains ingredients such as *Ajamoda, Vidanga, Saindhava, Chitraka, Pippali* etc which possess *Katu* and *Tikta rasa*, *Ushna veerya* and known as *Kaphavatamayan jayet*[4]. It improves the digestion and metabolism by enhancing the *Agni* to support efficient protein metabolism and their byproducts. Reduces *Ama* (metabolic toxins) by assisting the body in breaking down and clearing the unprocessed metabolic waste.

Manasamitra vati

It contains numerous herbs that enhance *Buddhi, Medha* and *Vacha*; and is indicated in *Manovikara*[5] due its *Medhya* (nootropic) property. Helps in improving the cognitive, sleep and behavioural symptoms by regulating the impaired brain functions through its neuroprotective action.

Saraswatarishta

This formulation known as *Amritasama,* contains ingredients like *Brahmi, Shatavari, Vidari, Haritaki, Ushira, Ardraka,* and *Shatapushpa*. Promotes *Ayu, Smriti, Medha, Bala, Kanti* and it is an excellent *Rasayana*[6]. *Arishta* preparations are known to cross the blood-brain barrier, provide immediate action and possess *Kaphahara* properties thereby aiding in the reduction of neurotoxic accumulation and alleviating neurological stress. As the patient exhibited predominantly *Kaphaja* and *Sama lakshana, Saraswatarishta* was administered for one month, which showed improvement in seizure episodes and irritable behavior.

Smritisagara rasa

It has ingredients such as *Shuddha Parada, Gandhaka, Haratala, Manahshila, Tamra* each 1 parts, are subjected to 21 *bhavana* with each of *Vacha kwatha*, *Brahmi swarasa* and *Jyotishmati bija taila*. It possesses *Apasmara nashana* property[7], is widely used in seizure conditions. It prevents neuronal hyperexcitability as well as seizure-induced neuronal damage. This formulation contains *Rasaushadhis,*

which provide a rapid therapeutic response even in chronic condition. Its nanoparticles act on the nervous system within a short span of time. Clinical research supports its use in improving cognitive abilities by reducing the oxidative stress in brain.

Brahmi vati

It contains *Brahmi, Shankhpushpi, Vacha, Maricha, Swarna makshika* and *Rasa sindhura* which is well proven as *Medhya rasayana*[8]. This formulation is used to improve memory, retention, mental fatigue, stress, depression and sleep disorders. The main ingredient, *Brahmi* contains active compounds called bacosides, which helps in enhancing learning, memory and cognitive function by supporting nerve cell health and improving brain signalling.

CONCLUSION

Maple Syrup Urine Disease, though a rare metabolic disorder, presents significant challenges in diagnosis and long-term management. While modern medicine emphasizes dietary restrictions and metabolic control, Ayurveda interventions offer a supportive role in improving neurodevelopmental outcomes, digestion and metabolic balance. Ayurveda formulations such as *Deepana, Panchana* and *Medhya rasayana,* along with individualized dietary and lifestyle modifications, may help in reducing the frequency and severity of symptoms and complications. Though limited, clinical experiences suggest that Ayurveda will contribute to improving quality of life through an individualized, symptom-based interventions. Continued clinical observation and research will strengthen and validate the potential role of Ayurveda in managing MSUD.

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