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CASE REPORT

IMPROVEMENT IN HURLER SYNDROME WITH AYURVEDIC MANAGEMENT: A CASE REPORT

Dr Lekshmi M K[1]*, Dr Anupama S Kumar[2], Dr Parvathy Jayan[2]** & Dr Sangeetha P Sankar[2]****

[1]* Associate Professor, Department of Kaumarabhritya, Government Ayurveda College, Thiruvananthapuram, Kerala, India
[2]**PG Scholar, Department of Kaumarabhritya, Government Ayurveda College, Thiruvananthapuram, Kerala, India

*Email: lekshmimk@gmail.com

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Abstract

Hurler syndrome is a rare lysosomal storage disorder occurring due to the deficiency of the lysosomal enzyme alpha L Iduronidase and has an autosomal recessive inheritance [1]. Ayurveda explains this condition as *aavaranajanya vatavyadhi* (obstructive pathology) [2]. This is the case of Hurler Syndrome of a female child of 13yrs with a relevant family history (her sister had the same syndrome). The patient had dysmorphic facies, pain and stiffness over multiple joints, dysmorphic facies, difficulty in walking, and mild speech impairment. She visited the outpatient department of our tertiary Ayurvedic health centre three times. She took OP and IP management along with dietary recommendations for about two months and ten days, and significant improvement was noted such as a reduction in contractures, ability to walk independently, self-feeding and the ability for normal schooling. The assessment was done with Cerebral Palsy Quality of Life (CPQOL). The values changed from 330 to 445 before and after treatment, respectively and gradings of spasticity as per the Modified Ashworth scale changed from 2+ to 1+ in both upper limbs and from 3+ to 2+ in both lower limbs.

Introduction

Mucopolysaccharidosis (MPS) is a group of hereditary conditions accompanied by the storage of acid mucopolysaccharides in the tissues [3]. It is of seven types and 13 subgroups which are characterized by an inherent deficiency of the enzymes responsible for the degradation of glycosaminoglycans (GAGs)[4]. Biochemical classification of MPS disorders include Hurler syndrome or Scheie syndrome (Type 1), Hunter's syndrome (Type II), Sanfilippo syndrome (Type III), Morquio syndrome (Type IV), Maroteaux lamy syndrome (Type VI) , Sly syndrome (Type VII), Natowicz syndrome (Type IX). Hurler syndrome is a rare lysosomal storage disorder caused by a deficiency of lysosomal enzyme alpha l iduronidase. Children with Hurler syndrome appear nearly normal at birth, and the most common early presenting features are the nonspecific symptoms of rhinitis and inguinal hernia[5]. Patients develop symptoms shortly after birth and progress rapidly. The symptoms include developmental delay, cognitive decline, characteristic coarse facial features, joint stiffness and contractures, short stature, and

cardiac and hepatic disease[6]. Untreated children do not survive into the second decade[7]. It has an autosomal recessive inheritance resulting in progressive glycosaminoglycans (GAG) accumulation within lysosomes. The overall incidence of MPS I is 0.99-1.99/100,000 live births[8]. Coarse facial features, corneal clouding, hepatosplenomegaly, kyphosis/scoliosis, and cardiac valve disease are the most common symptoms in Hurler syndrome, occurring in 86.4%, 70.9%, 70.0%, 70.0%, and 48.9% of the patients, respectively [9]. Recurrent middle ear infections and hearing loss are common complications in Hurler syndrome[10]. In Ayurveda, these genetic diseases come under *adhibalapravrutta vyadhi* (genetically predisposed disease). Here, due to the *Sahaja nidana* (genetic causes) *beeja* and *beejabhaga dushti* (chromosomal and genetic abnormalities) occurs which lead to *vikrutha garbha* (fetal anomalies). It causes *vatha pradhana tridosha dushti* one among the tridosha) impairing the *agni* (digestive fire). This results in blockage of *vatha* mainly affecting the *asthi* and *medo dhathu* resulting in *asthi vakratha* (deformed joints and bones).

Patient information

A 13-year-old female child reported to the outpatient department with complaints of pain and stiffness over multiple joints, difficulty rising from sitting posture, difficulty walking (toe walking), and joint contractures. Blackish thickened lesions over multiple joints with itching, oozing, and speech impairment for ten years. She had frequent respiratory problems and ear infections. She was the second child of consanguineous parents, whose mother had been suffering from gestational hypothyroidism from the third month. She was born through full-term normal vaginal delivery and had a birth weight of 2.8 kg. She cried soon after birth, but feeding was improper due to breast milk insufficiency. The neonatal period was uneventful, and all developmental milestones were attained at the proper age. Then mother noticed the above complaints, which were progressive. Genetic analysis was performed and diagnosed as Hurler syndrome. Her sister was also a known case of Hurler syndrome, which expired one year back .

Clinical findings

On general examination, vital signs appear normal. On head -to-foot examination, some relevant findings are dysmorphic facies, frontal bossing, cloudy corneas, periorbital swelling, and ptosis. She had a short neck, flattened nasal bridge, and bulbous nose. She possessed macroglossia and malocclusion of teeth. She had an umbilical hernia and abdominal distention. Kyphosis was present. Systemic examination revealed recurrent upper respiratory tract infections associated with otitis media. She had mild breathing difficulty while lying in the supine position. She had a history of hepatosplenomegaly and

Table 1: OP Management Timeline

Date/day of visit	Presentation during visit	Interventions
1/2/2022 – 14/2/2022	First visit to the outpatient department • Pain and stiffness of multiple joints • Difficulty in raising from sitting position • Difficulty in walking • Blackish thickened lesions over multiple joints with oozing and itching • Impaired speech clarity • Indigestion	• *Abhayarishtam* 15ml twice daily after food (9am and 8pm). • 10g *Rajanyadi churna* twice daily (10am and 9pm) after food[11] • *Dhanwantharam gulika* twice daily with arishta (9am and 8pm) • *Gandharvahastadi kashayam* 30 ml twice daily before food (8am and 7pm) [12] • *Sudhadurvadi tailam* for external application
Suggested review after 2 weeks		
15/2/2022-28 /2/2022	• Digestive power improved • Pain of multiple joints reduced but stiffness persist • Respiratory tract infections.	• *Dasamoolahareethaki lehyam* 1tsp at night before bed. • *Sudarshanm gulika* twice daily after food (9.30am and 8.30pm)
Suggested review after 2 weeks.		• Continue the above medications.
1/03/2022 – 6/03/2022	• Symptoms persist with mild relief Advised inpatient admission	• Continued the above medications.

Table 2: IP Management Timeline

Date of admission	Complaints / conditions of patient	Intervention
8/3/2022 – 9/3/2022	Pain and stiffness of multiple joints Difficulty in raising from sitting posture Difficulty in walking Blackish thickened lesions over multiple joints with oozing and itching Impaired speech clarity	10 g *Rajanyadi churna* twice daily (9 am and 7 pm) after food 10 g *Sukumara rasayanam* at night (9 pm)after food[13] *Marmavattu gulika* for external application on both knee joints[14] *Shaddharanam gulika* (500 mg) at night 9 pm after food[15] *Kashayadhara* with *Rasnasaptakam kashayam* for 45 minutes for two days *Pichu* on both elbow and knee joints with *Karpooradi tailam*
10/3/2022 – 12/3/2022	Sneezing present in morning hours Complaints persist	*Abhyanga* with *Karpooradi tailam* for 45 minutes for 7 days *Vyoshadi vatakam* 5 grams twice daily (9 AM
13/3/2022 – 19/3/2022	Burning sensation of skin	*Abhyanga* with *Ksheerabala tailam* for 45 minutes for 7 days[17]
20/3/2022	Stiffness of both elbow joints reduced Itching on ankles	Treatment was restricted due to menstruation but continued internal medication
29/3/2022 – 4/4/2022	Able to walk without experiencing much pain	*Pizhinjuthadaval* with *Karpasasthyadi tailam* for 30 minutes for 7 days[18]
5/4/2022 – 11/4/2022	Blackish lesions on joints reduced Able to raise from sitting posture without expe-	*Takradhara* with *mustha* and *yashti* head and body for 30 minutes for 7 days
12/4/2022 – 18/4/2022	Contractures of joints reduced	*Patrapotala swedam* with *shatahwadi tailam* for 30 minutes for 7 days
18/4/2022	Can walk independently without pain	Patient was discharged

constipation. The skin over both the elbow and medial malleolus of both lower limbs showed blackish-thickened lesions. On nervous system examination, all the cranial nerves were intact except the 3rd, 4th and 6th nerve, as mild ptosis was present. She was able to perform the coordination tests, and the higher mental functions appeared to be normal.

Therapeutic interventions

After a detailed examination, the patient was admitted to the inpatient department for a month and ten days. Initially, the patient was given *Rajanyadi churna* for *agni deepana* (improving the digestive fire) and *Sukumara rasayana* for proper *anulomana* (proper functioning of the *Apana Vayu*). *Marmavattu gulika* was given for external application over both knees to relieve stiffness. *Shaddharanam gulika* (500 mg) at night for *pachana* (one which aids metabolism). *Kashayadhara* (pouring herbal decoction over the body) was done with *Rasnasaptakam kashayam*. It was followed by *abhyanga* (oil massage) with *Karpooradi tailam* for two days, *Ksheerabala tailam* for seven days, and internal medicines. Then *pizhinjuthadaval* (a kind of sudation therapy, pouring and massaging with oil continuously, helps in reducing the stiffness of joints and provides muscle strengthening) was done with *karpasasthyadi tailam* for seven days, followed by *takradhara* (pouring a continuous stream of medicated buttermilk over forehead or whole body to enhance blood circulation to the brain thereby revitalizing the central nervous system) for seven days, and then *patrapotalaswedam* (fomentation with herbal drugs or leaves of medicinal plants to help in reducing the stiffness of joints a strength to the joints, muscles and soft tissues) for seven days.

Follow-up and outcome

The patient was assessed through various clinical examinations. Assessment of the patient was done before and after treatment by CPQOL and Modified Ashworth scale. Quality of life refers to an individual's perception of their well-being across various domains of life. CPQOL value before the treatment was 330, and after the treatment

changed to 445. The quality of life and self-esteem segments of CPQOL improved. Modified Ashworth scale grading changed from 3+ to 2+ in both lower limbs and from 2+ to 1+ in both upper limbs.

The colour and tone of the skin improved with a significant reduction in the blackish discolouration, the stiffness of joints was reduced considerably (assessed with the help of the Ashworth scale), and the patient could walk independently. Kyphosis was reduced, she could raise from sitting posture easily, and contractures were reduced. Speech clarity also improved. Pain reduced considerably, and muscle endurance and hand and arm use improved. Joints became more mobile, and overall fatigue was reduced. Other parameters, including activities of daily living, social life, and self-esteem, were restored after the treatment. The assessment was also done with the help of CPQOL; the scores obtained before and after treatment are 330 and 445 respectively.

Table 2: IP Management Timeline

MODIFIED ASHWORTH SCALE				
	BEFORE		AFTER	
	RIGHT	LEFT	RIGHT	LEFT
UPPER LIMB	++	++	+	+
LOWER LIMB	+++	+++	++	++

Discussion

The patient visited our hospital with a diagnosed case of Hurler's syndrome. In Ayurveda, this condition can be correlated with *avarana janya vatavyadhi*. Even though it is not possible to cure a genetic disease, Ayurveda can address a patient's quality of life by analyzing the *Doshas*(body humor), *Dushyas*(tissues), *Agni*(digestive fire), and *Srotas* (channels of circulation). In *Samprapti* (pathogenesis), mainly *Vata, pitta*, and *Kapha* are involved. In this case, the functions of *udana vayu*(a type of Vata) were affected, like reduced clarity of speech, restricted activities, and decreased body strength. The restricted range of movements of both the upper and lower limbs depicted derangement of *vyana vayu*. *Prana vayu* is also affected since she had recurrent episodes of respiratory tract infections. Here, *beejadushti* (defective sperm and ovum) itself leads to *vata pradhana tridosha* vitiation, which resulted in *vikrutha garbha* (genetically abnormal fetus). Later, *Kapha avarana* (obstruction to channels of circulation by Kapha) accompanied, which in turn vitiates the *vata*. Simultaneously, *Srotorodha*(obstructive pathway) occurred, and all three *rogamargas* (pathway of diseases) got affected. Since the first line of treatment always points towards *agni* (digestive fire), the treatment started with *deepana* (appetizers) and *pachana*(enhances digestion) such as *Rajanyadi churnam* and *Shaddharana tablet*. Initially, *Kashayadhara* was done over the body with *Rasnasaptakam kashaya*. The drugs in the *kashaya* are *vatahara* and possess *vedanasamaka* property. *Abhyanga* with *Karpooradi taila* was done to address respiratory health due to its *vata kaphahara* property. Then *abhyanga* with *Ksheerabala taila* was used as it helps in pain relief. Then *Pizhinjuthadaval*, a sudation therapy, was done with *Karpasasthyadi taila* which provides an oleation effect and helps reduce joints' stiffness. *Karpooradi taila* was also used as *Pichu* to relax muscle pain and cramps. The internal and external medications improved the patient's overall condition considerably.

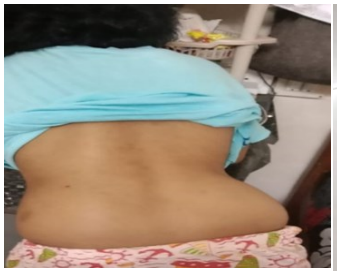

Fig: 1

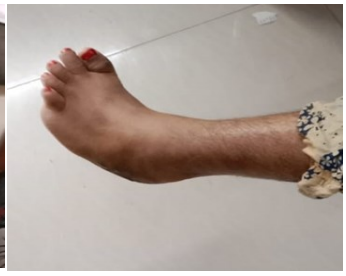

Fig: 2

Conclusion

None of the medical systems provides a complete cure for Hurler syndrome. Clinically significant improvements noted, such as reduction in contractures, ability to walk independently, self-feeding, and the ability for normal schooling, gives a new ray of hope in managing rare disorders through the systematic and holistic approach of Ayurveda. From this case, it is evident that quality of life can be improved within a short period of management for about two months and ten days. Therefore, long-term treatment can have greater results in this rare genetic disorder.

Declaration and patient consent

Authors certify that they have obtained a patient consent form, where the caregiver has given consent for reporting the case in the journal. The caregiver understands that his name and initials will not be published, and efforts will be made to cover the identity, but anonymity cannot be guaranteed.

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References

1. Dr. Vaddadi Srinivas, Dr. NAVSK Ravikumar, Dr. V Radha Srinivas, Dr.R Vikram Vardhan;Hurlers Syndrome : A rare Case Report; Vaddadi Srinivas et al ; Sch J Med Case Rep 2015; 3(7): 568-570.
2. Prof. K R Srikantha Murthy , Vagbhata's Ashtanga Hrdayam; 6th edition ; Chowkhamba Press Varanasi; Chowkhamba Krishnadas Academy , Varanasi 2012. p.149.
3. Parthasarathy A , Menon P S , editors , IAP Textbook of Pediatrics, Inborn Errors of Metabolism. 4th ed.New Delhi : Published by Jaypee Brothers Medical Publishers
4. Nagpal R, Goyal RB, Priyadarshini K, Kashyap S, Sharma M, Sinha R, Sharma N. Mucopolysaccharidosis: A broad review. Indian J Ophthalmol. 2022 Jul;70(7):2249-2261. doi: 10.4103/ijo.IJO_425_22. PMID: 35791104; PMCID: PMC9426054.
5. Belmont PJ Jr, Polly DW Jr. Early diagnosis of Hurler syndrome with the aid of the identification of the characteristic gibbus deformity. Mil Med. 1998 Oct;163(10):711-4. PMID: 9795551.
6. R, Bollu PC. Hurler Syndrome. [Updated 2022 Jul 12]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2022 Jan-. Available from: https://www.ncbi.nlm.nih.gov/books/NBK532261/
7. Pillai NR, Ahmed A, Vanyo T, Whitley CB. Early Neonatal Cardiac Phenotype in Hurler Syndrome: Case Report and Literature Review. Genes (Basel). 2022 Jul 22;13(8):1293. doi: 10.3390/genes13081293. PMID: 35893030; PMCID: PMC9331972
8. Leroux S, Muller JB, Boutaric E, Busnel A, Lemouel F, Andro-Garçon M, Neven B, Valayannopoulos V, Vinceslas C. Syndrome de Hurler : diagnostic et prise en charge précoces [Hurler syndrome: early diagnosis and treatment]. Arch Pediatr. 2014 May;21(5):501-6. French. doi: 10.1016/j.arcped.2014.02.013. Epub 2014 Mar 31. PMID: 24698225.
9. Nan H, Park C, Maeng S. Mucopolysaccharidoses I and II: Brief Review of Therapeutic Options and Supportive/Palliative Therapies. Biomed Res Int. 2020 Dec 4;2020:2408402. doi: 10.1155/2020/2408402. PMID: 33344633; PMCID: PMC7732385.
10. Kariya S, Schachern PA, Nishizaki K, Paparella MM, Cureoglu S. Inner ear changes in mucopolysaccharidosis type I/Hurler syndrome. Otol Neurotol. 2012 Oct;33(8):1323-7. doi: 10.1097/MAO.0b013e3182659cc3. PMID: 22918113.
11. Murthy SK, editor. Ashtanga Hridayam of Vagbhata, Uthara Sthana ; 6th edition; Ch.2, Ver 38-40 Varanasi; Chowkhamba Krishnadas Academy , Varanasi 2012. p.20.
12. Krishnan Vaidyan AK, Gopalapillai S, editors. Kashaya Prakarana. Sahasrayogam. 33rd ed. Alappuzha: Vidyarambham Publishers; 2015. p. 78.
13. K.Raghavan Thirumulppad , Ashtanga samgraham ;2nd edition; Vaidyabhushanam Raghavan Thirumulppad ; Chalakkudy ; 1998.p.55
14. M. Narayanan Vaidyar, Sahasrayogam, 1st edition, Ashoka Pharmaceuticals, Kannur; 2001.p.349.
15. P V Sharma, Susruta Samhitha, Chaukhambha Bharati Academy, Varanasi; 2010.p.303.
16. Murthy SK, editor. Ashtanga Hridayam of Vagbhata, Uthara Sthana ; 6th edition; Ch.2, Ver 5-7 Varanasi; Chowkhamba Krishnadas Academy , Varanasi 2012. p.20.
17. M .Narayanan Vaidyar, Sahasrayogam, 1st edition, Ashoka Pharmaceuticals, Kannur; 2001.p.182
18. M.Narayanan Vaidyar, Sahasrayogam, 1st edition, Ashoka Pharmaceuticals, Kannur; 2001.p.184

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