IAMJ

INTERNATIONAL AYURVEDIC MEDICAL JOURNAL

Observational Study ISSN: 2320-5091 Impact Factor: 6.719

AYURVEDIC MANAGEMENT OF MORQUIO SYNDROME IN A PEDIATRIC PATIENT – A CASE STUDY

Amisha Bavarva

Assistant professor, Department of Kaumarbhritya, Ayurveda college and Hospital, RK University, Rajkot

Corresponding Author: amishabavarva@gmail.com

https://doi.org/10.46607/iamj2212092024

(Published Online: September 2024)

Open Access

Article Received: 08/07/2024 - **Peer Reviewed:** 29/07/2024 - **Accepted for Publication:** 14/08/2024.

Check for updates

ABSTRACT

Mucopolysaccharidosis type IV, also known as Morquio syndrome, a rare hereditary disease of autosomal recessive storage that is a progressive disease that affects the skeleton. The affected person develops various skeletal abnormalities, including height, protruding chest, scoliosis, deformed knees, and curvature of both lower limbs, which may be in *Kubjata*. *Kubjata* is one of the *Vataja Nanatmaja Vikara*; this comes under *Janmabala Pravrutta Vyadhi*. In this case *Abhyanga*, *Swedana*, *Mridu Virechana* and *Basti* for 25 days along with a combination of oral medicines such as *Trayodashanga guggulu*-500 mg, *Dashmool kwatha* 20 ml, *Shiva gutika* 500 mg twice per day have been showing that Ayurvedic herbs along with *Panchakarma*, can play a major role in the management of Morquio syndrome to improve the quality of life and prognosis of the disease.

Keywords: *Janmabala Pravrutta Vyadhi*, *Kubjata*, Morquio syndrome, MPS IV A, Mucopolysaccharidosis.

INTRODUCTION

Mucopolysaccharidosis type IV, also known as Morquio syndrome, is a rare autosomal inherited lysosomal storage disease characterized by the accumulation of mucopolysaccharides (Glycosaminoglycans) in various body tissues. Long-chained sugars called mucopolysaccharide, or glycosaminoglycans (GAGs), are essential for the development of bone, cartilage, tendons, corneas, skin, and connective tissue.[1] A progressive condition that primarily affects the skeleton, the lack of lysosomal enzymes necessary for the

metabolism of the different mucopolysaccharides causes the corresponding substrate to accumulate in the cells, causing cellular damage and clinical changes. Affected individuals acquire a variety of skeletal anomalies, similar to those seen in *Kubjata*, such as small height, protuberant chest, scoliosis, malformed knees, and bowing of both lower extremities. Under the *Janmabala Pravrutta Vyadhi's* auspices, *Kubjata* is one of the *Vataja Nanatmaja Vikara*.

The precise global incidence of Morquio syndrome is unclear, however estimates range from 1 in 200,000 in British Columbia, 1 in 75,000 in Northern Ireland, and 1 in 640,000 live births in Western Australia. Twenty-two percent of all lysosomal storage diseases (LSD) in India are caused by MPS. Out of the 85 MPS instances that were found in India, MPS IV is the most prevalent, accounting for 26% of the cases, according to a 2017 study by Khan et al.[2]A case report of a Mucopolysaccharidosis syndrome, precisely Morquio syndrome or MPS IV A with typical radiological manifestations is detailed in this article.

Chief Complain

A 14-year-old female child, born out of a non-consanguineous marriage with was having medical history of inability to stand with and without support; and unable to sit on floor. She was apparently normal at birth till the age of 3 years then complain is gradually progressed. Now since last 4 years patient is also having complaint of severe pain during movement of lower extremities. Upward movement of upper extremities is also restricted, and patient is unable to do her daily routine activity by herself. There are not any significant findings in birth history. A positive family history was discovered for a bone malformation and pain in the right lower limb in the other two siblings, an 8-year-old brother and a 10-year-old sister.

Observation of all available medical documents and examination details of the patient are tabulated as below.

Examinations

Physical examinations

The physical features presented short stature (Height: 131 cm), protuberant chest, scoliosis, deformed knees and bowing of both lower extremities.

Anthropometry

Height: 131 cm, Weight: 37 kg

Head Circumference: 52 cm Chest Circumference: 86 cm

Mid Arm Circumference: Right - 24 cm, Left - 24 cm

Mid-Thigh Circumference: Right – 37 cm, Left – 36.5 cm

General Examination:

Pallor: Eye- Conjunctiva normal, No pallor, No corneal haziness

Palm- Normal, Tongue- Coated

Icterus: Not present

Clubbing: Not present

Cynosis: Not present

Lymphadenopathy: Lymph node enlargement at popliteal region

Systemic Examination:

CNS: Conscious, Alert and Oriented with time, place and person

memory and intelligence - Normal

CVS: S1, S2 Heard Normal

RS: Chest clear, AEBE

GIT: P/A – Soft, No hepatomegaly, no splenomegaly

Upon medical assessment, all major systems revealed no signs of dysfunction.

Musculo-skeletal Examination:

Power: Right upper limb was 4/5 and left upper limbs was 3/5.

The left lower limb was 3/5 and left lower limb was 2/5.

Tone: Hypertonia

Swelling: On B/L elbow, B/L knee, B/L hip and B/L ankle joint

Pain: During touch & movement in B/L knee, B/L Hip and B/L Ankle joint.

Investigation

The radiographic evaluation indicated Mucopolysaccharide i.e.

February 2016, MRI study showed suggestive multiple (4 in number) well defined oval shaped altered signal intensity lesions (hypointense on T2W1, T1W1 and heterogeneously hypointense with peripheral hyperintense rim on PDFS) is noted involving subchondral epiphyseal location of both femoral condyles,

both tibial platue, largest measuring (9×3) mm in medial femoral condyle. Bone marrow oedema (hyperintense on PDFS) is noted surrounding above mentioned lesions and medial femoral epiphyses. Overlying cartilage appears unremarkable. Plica was Absent. Minimal increased signal intensity is noted involving tibial attachment of anterior cruciate ligament. Very minimal joint effusion is noted. Few subcentimeter sized lymph nodes are noted involving popliteal region. Changes of grade -1 osteochondritis dissecans involving both femoral condyles and both tibial platue.

Then, November 2017 MRI of Dorso-Lumber Spine showed bullet shape multiple thoraco-lumber vertebra with superior endplate irregularity and central and beaking involving multiple thoraco-lumber vertebra. Spinal cord appears normal. No evidence of posterior element spinal vertebral defect. Findings are in favour of possibility of skeletal dysplasia. Possibility of multiple epiphyseal dysplasia or mucopolysaccharidoses (MPS-IV: Morquio syndrome) should be ruled out.

On February 2023, X-ray showed B/L hip joint show severe decreased joint space with angulation deformity, both wrists show decreased joint space with soft tissue swelling. L-S Spine show significant osteoporosis with moderate degenerative changes. Multiple level disc compression &vertebral body compression. Anterior central beaking seen in lower dorsal and upper lumber vertebrae.

B/L knee joint also show significant decreased joint space. (R>L) and significant osteoporosis (R>L). There was bowing seen in right femur-lower end and soft tissue swelling (R>L).

Root of the X-ray not recommended due to excessive osteoporosis

Changes are increased as compared to 2016.

Known case of Mucopolysaccharide/skeletal dysplasia.

31/10/2017: Hb-13.10 gm/dl, Polymorphs-43.00%, Lyphocytes-45.00%.

APTT test value- 27.5 (Biol. Ref. Range: 30-40.5), APTT control- 29.80 (Nephelometry), PT (Prothrombin time) test – 11.3 sec (Bio.Ref. Range: 8.1-14.5), PT Control – 11.80 sec (Nephelometry), INR – 0.95 (Nephelometry)

03/02/2023: 25-HYDROXY-VITAMIN D (D3+D2): 11.70 ng/ml (Deficiency: <10; Insufficiency: 10-30)

Hb: 12.8 gm%; Total RBC: 4.64 mill/c.mm; Total WBC: 8800 /cu mm; Platelet Count: 359000, Differential count: N-32.6, L-49.1, E-12.3, M-5.5, B-0.5, Absolute Eosinophil Count: 1082.4 /ul, Blood Indices: PCV-39.3%, MCV-84.7fl, MCH-27.6pg, MCHC-32.6%, RDW-14.4%, ESR– 42

Treatment:

As such, no specific treatment is mentioned separately in the classics. The treatment was planned to delay the disease's prognosis and improve a patient's quality of life. Following the conclusion of Panchakarma operations, the clinical outcome was evaluated. List of medications given on admission [Table 1] List of Panchakarma procedures during admission [Table 2]

Result & Observation [Table 3]

Improvement:

Kyphosis, scoliosis, and discomfort were moderately reduced following a 25-day course of therapy. The patient improved in standing, walking, and joint range of movement.

DISCUSSION

Morquio syndrome of MPS IV might occur due to mutations in galactosamine-6-sulfatase genes or due to beta-galactosidase deficiency[3,] which can be considered as *Adibalpravruttavyadhi or Janmabalapravruttavyadhi*. Musculoskeletal features are the most common presenting features of MPS IV. The early skeletal symptoms that the patient reported included scoliosis, kyphosis/gibbus, irregular gait, cervical instability and small stature (131 cm). In MPS IVA, Gibbus is frequently seen as the first indicator and spondylus epiphysis metaphyseal dysplasia is generally diagnosed in the second year of life when the child starts walking. Based on clinical and radiological studies, MPS IV was diagnosed, which can be compared with *Kubjata*. *Kubjata* is one of the *Vataja Nanatmaja Vikara*; this comes under the umbrella of *Janmabala Pravrutta Vyadhi*. The main features of this disease include skeletal abnormalities, which include excessive osteoporosis with moderate degenerative changes, bowing of the lower end of the right femur, and skeletal dysplasia,

which may be due to *Vikruta Vata* (vitiated *Vata*). Vata is vitiated by several etiological variables, including Dhatukshaya (depletion of bodily tissue) and *Margavarana* (obstruction in the natural course of Vata such as normal distribution, synthesis of tissue elements, etc.). In a vicious cycle, the vitiated Vata also causes *Margavarana* and *Dhatukshaya*, which in turn causes multiple epiphyseal dysplasia to emerge. So, the designed Basti can disrupt the pathophysiology of *Vatavyadhi* by eliminating *Margavarodha* through channel cleansing and *Dhatukshaya* through its *Brihmana* characteristic.

Samahana of the body depends on *Dhatu*. Irregularity in the body shape mainly because of the malformation of *Asthi Dhatu* and body stature indicates the malformation of *Mamsa Dhatu*. These two *Dhatu* are essential for physical activities like walking, jumping, running, etc. Since there is no evidence of appropriate aetiology, a suspicion of *Janmabalapravruttavyadhi* is made. *Kubjata* is one amongst *Janmabalapravruttavyadhi* and *Vatajananatmajavikara*.[4] According to Acharya Madhavakara, *Kubjata* refers to an elevated chest or back.[5] Acharya Vagbhata explained it as *Avanamai*. e. bowing of the body.[6] Considering the subtypes of this condition is diagnosed as *Bahirayama Kubjata*.[7] With due consideration of *Mamsa* and *Asthi Dhatu* involvement in this disease the treatment is planned for 25 days. *Abhyanga*, *Swedana*, *Mridu Virechana* and *Basti* are indicated for *Avrita Vata* and chronic *Vatavyadhi*.[8] *Basti* can break pathogenesis of *Vatavyadhi* by removing *Margavarodha* by purification of channels and *Dhatukshaya* by its *Brihana*(nourishing) property. *Matra basti* is indicated for increasing vigour, strength and semen. It is useful in *Katishoola* (backache), pain in thigh and calf region, headache and *Vatarakta* (various diseases of rheumatic spectrum). It also has Rasayana property. Most of *Majja* and *Asthi pradoshaj vikar* (disorders of bone and bone marrow) can be managed with *Basti*.[9] *Trayodashanga Guggulu* is useful in *Snayugatavata* (various tendon and ligament disorders), *Asthigatavata* (disorders of bone), *Majjagatavata* (disorders of bone-marrow), *Khanjavata* (limping disorders) and various *Vatic* disorders (neurological, rheumatic and musculoskeletal diseases).[10] *Dashamula Kwatha* is having *Tridoshaghana* property.[11] *Shiva gutika* is a *Rasayana* and helpful in *Vatarakta*, *Shiroroga*, *Mukharoga*, *Swasroga* (dyspnoea), *Netraroga* etc.[12] Thus these combinations are useful to manage the manifestation of disease in this case. The case study shows that such a Morquio syndrome can be managed with Ayurvedic treatment which improves the quality of life of patient and reduced the complication of disease. It may be possible to give better results in the manifestation of disease if the patient approaches the early stages of disease.

CONCLUSION

There is no specific treatment for Morquio syndrome, hence it demands an effective approach. After 25 days of Soshana and Shaman treatment, kyphosis, scoliosis and pain were moderately reduced. Patients got improvement in standing, walking and range of movement of joints. Clinical experience of this case indicates that Ayurvedic herbs along with Panchakarma can play a major role in the management of Morquio syndrome which is helpful to delay prognosis of the disease and improve quality of life of patient. The observations and experiences of Ayurveda management of morquio syndrome may be useful for its treatment and research.

Patient consent:

Written permission for publication of this case study had been obtained from the patient

Acknowledgments

The author is grateful to the mother of the child for giving us the consent for this paper to be published.

Conflicts of Interest

The authors have no conflict of interest to declare.

Ethical Approval

A written informed consent was obtained from the patient for this case to be reported for publication. The patient's mother was assured of absolute confidentiality and anonymity. Ethical clearance was not required for this study.

REFERENCES

1. Fauci A.S. Kasper DL, Braunwald E, Hauser SL, Longo DL, Jameson JL et al., Harrison's Principles of Internal medicine, 18th ed, United States of America, McGraw-Hill; 2012.p.1392
2. Saloni Gupta;J Lab Physicians.Morquio Syndrome Presenting with Dural Band Pathology: A Case Report; 2020, Dec; 12(4): 285–288; Published online 2020 Dec 30. doi: 10.1055/s-0040-1722548;Retrieved on dated 15.3.23
3. KamleshunRamphul, Morquio Syndrome: A Case Report; Published online 2018 March 5. Doi:10.7759/cureus.2270; Retrieved on dated 15.3.23
4. Acharya Agnivesha, Charaka samhita. Vaidya Yadavaji Trikamji Acharya, editor Varanasi: Chaukambhaorientalia, reprint,2011 p.113
5. Sharma A. Madhavanidanam with Madhava Vimarshini Commentary. 1st _edition, Varanasi: Chaukambha Sanskrit pratishthan; 2007.p.403
6. Vagbhatacharya, Astangasangraha. Indu, Dr. Shivprasadsharma, editor, Varanasi, chaukambha press, reprint, 2012 p.330
7. Acharya Agnivesha, Charaka samhita. Vaidya YadavajiTrikamji Acharya, editor Varanasi: Chaukhambha orientalia, reprint,2011, p.618.
8. Acharya Agnivesha, Charaka Samhita, Chikitsa Sthana 28, Vaidya Yadavaji Trikamji Acharya, editor Varanasi: Chaukhambha orientalia, reprint, 2011.
9. Acharya Agnivesha, Charaka Samhita, Siddhi sthana, 12/15Vaidya Yadavaji Trikamji Acharya, editor Varanasi: Chaukhambha orientalia, reprint,2011, p.1096
10. Govinda Das Sena, Bhaisajyaratnavali, Vatavyadhirogadhikara, 98-101, ed by Rajeshwar Dutta Shastri, 18th ed. Varanasi: Chaukhamba Prakashana; revised 2007. p.526.
11. Bhavprakash nighantu, guduchyadi Varga 26/41, p.294, Available from: https://niimh.nic.in/ebooks/e-Nighantu/bhabprakashnighantu/?mod=read [Last accessed on 24 February,2024]
12. Govinda Das Sena, Bhaisajyaratnavali, Rasayanadhikara 73/161-169, ed by Rajeshwar Dutta Shastri, 18th ed. Varanasi: Chaukhamba Prakashana; revised 2007. p.1120.

Source of Support: Nil
Conflict of Interest: None Declared

How to cite this URL: Amisha Bavarva: An observational case study of Morquio syndrome in a pediatric patient – a rare disease. International Ayurvedic Medical Journal {online} 2024 {cited September 2024} Available from: http://www.iamj.in/posts/images/upload/1742_1747.pdf

Tables

Table 1: List of medications given on admission

Sr. No.	Name of medicine	Dose
1.	*Trayodashanga guggulu*	500 mg BD
2.	*Shiva gutika*	500 mg BD
3.	*Dasmoola Kwatha*	20 ml BD (Before food)

Table 2: List of Panchakarma procedure during admission

Sarvanga Abhyanga followed by *Nadi Swedana*	with *Bala Taila*	1st to 3rd day [12/01/2023 to 14/01/2023] for 3 days
Mrudu Virechana	Castor oil in the dose of 20 ml with milk	4th day [15/01/2023 for 1 days]
Matrabasti with *Bala taila*	with *Bala Taila*	5th day to 25th day [16/01/2023 to 05/02/2023 for 21 days]

Table 3: Result & Observation

	Symptoms	**Before treatment**	**After treatment**
1.	Short stature	Height: 131 cm	No improvement
2.	Stand without support	Not able to be standing	3 min 8 second
3.	Bending (Hand feet touch)	Not able to bend	2 min 48 second
4.	Half *Paschimotanasana*	Not able to do	2 min 16 second

5.	Butterfly position	Unable for forward bending at all (half *Paschimo-tanasana*)	Not able to do
6.	Vajrasana position	Not able to do at all	4 min 35 second
7.	**Daily activities**		
A) Hair comb	Unable to combing hair by her-self	60% able to comb hair by herself	
B) To wear socks	Unable to wear socks by herself	Able to wear socks by herself	
C) Change dress	Unable to Change dress by herself	Able to Change dress by herself without help of others	

Joint	**BT**	**AT**
Wrist joint	No swelling, No pain	No swelling, No pain
Elbow joint	Swelling in B/L elbow joint, No pain Not able to straight and bend from elbow. Carrying Angle – 60	No swelling, No pain Able to straight & bend elbow joint Carrying Angle – 15
Shoulder joint	Fully straight upward movement of hand not done. (Hand does not touch to ear)	Fully straight upward movement of hand done. (Hand touch to ear)
Knee joint	Swelling & pain during touch & movement in B/L knee	No swelling & no pain during movement and walking
Ankle joint	Swelling & pain during touch & movement in B/L Ankle joint	No swelling & no pain during movement and walking
Hip joint	Swelling & pain during touch & movement in B/L Hip joint	No swelling & no pain during movement and walking