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Case Study

AYURVEDIC INSIGHTS INTO THE FORME FRUSTE OF MARFAN SYNDROME

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ABSTRACT
Marfan Syndrome, an autosomal dominant, generalized disorder affecting the connective tissue, caused by mutations of FBN 1 gene which leads to deficiency of fibrillin 1 leading to reduced microfibril formation and disrupts the mechanical integrity of connective tissue, presents a complex clinical picture affecting multiple systems, poses significant challenges to conventional management. We present an 18-year-old female with Marfan Syndrome, highlighting her clinical features such as thin and slender body with disproportionately long arms and legs as compared with the trunk, bilateral Ectopia lentis, Arachnodactyly, positive Walker Murdoch Wrist sign and positive Steinberg Thumb sign, bilateral Pes planus, asymmetrical chest with mild Pectus carinatum deformity and moderate degree of Kyphoscoliosis along with winging of scapula. This article explores the Ayurvedic concept of *Beeja dosha* and highlights the value of integrating Ayurvedic insights into the management of Marfan Syndrome, offering a holistic and personalized approach to patient care to improve the quality of life and aims to deepen understanding of Marfan Syndrome from an Ayurvedic perspective, laying the groundwork for future research into Ayurvedic management strategies.

INTRODUCTION
Marfan Syndrome is an a rare (0.02% of population) inherited autosomal dominant disorder of connective tissue, predominantly caused by mutations of FBN 1 gene which leads to deficiency of fibrillin 1 leading to reduced microfibril formation. This disrupts the mechanical integrity of connective tissue, giving rise to a wide range of clinical features affecting multiple systems. The incidence of MFS is among the highest of any heritable disorder, 1 in 3000-5000 births in most racial and ethnic groups[1]. Marfan Syndrome exhibits an autosomal dominant inheritance pattern in 75% of cases, while 25% result from de novo mutations. Historically, MFS was diagnosed based on a triad of distinctive features-

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Skeletal Features
- Patients with MFS typically display a marfanoid habitus with tall stature with long limbs.
- The ratio of upper segment (top of head to top of pubic ramus) to the lower segment (top of pubic ramus to floor) disproportional for age, race, and sex.
- The fingers and hand are long and slender and have a spider like appearance (Arachnodactyly).
- Scoliosis/Kyphoscoliosis.
- Anterior chest deformities, including pectus excavatum, pectus carinatum or asymmetry (Increased risk of Pneumothorax).
- Pes planus and Dural ectasia and high arched palate with crowded teeth[1].

Cardio vascular features
- Cardiovascular abnormalities are the major source of morbidity and mortality in MFS.
- Patients often have mitral valve prolapse that develops early in life and that progresses to mitral valve regurgitation of increasing severity in about one-quarter of patients.

- Dilation of root of aorta is characteristic and ominous feature of MFS that can develop at any age. The rate of dilatation is unpredictable but it can lead to aortic regurgitation, dissection of aorta and rupture. Dilation is probably accelerated by physical and emotional stress as well as pregnancy[1].

Ocular features

- Myopia is the most common ocular feature of MFS and often presents in early childhood.
- Displacement of the lens from the center of pupil (Ectopia lentis) occur in 60% of MFS patients.
- Retinal detachment, early cataract formation and glaucoma can occur[1].

Marfan Syndrome diagnosis requires a comprehensive clinical evaluation supported by genetic testing. Imaging by chest X-ray may reveal evidence of aortic dilation but 2D Echo is more sensitive and can also demonstrate valvular disease, if present. Slit-lamp examination[4]. And, through Major and Minor Ghent Criteria[1].

In Ayurveda, heriditary disorders are reffered to as '*Adibala pravrutta vyadhi or Sahaja vyadhi*'. If any part of the *Beeja, beejabhaaga, beejabhaagaavayava,* which is responsible for the formation of particular organ is vitiated, this will result in vitiation of that respective organ (*yasya yasya hyangavayavasya beeje beejabhaaga upatapto bhavathi, tasya tasyangavayavasya vikrutirupajayate)* [2]

Marfan Syndrome patients' life expectancy is mainly tied to cardiovascular severity, but improved treatments have led to notable gains in survival rates. Early detection and appropriate treatment is critical for patients with Marfans Syndrome, as they are at an increased risk of life threatening complications of cardiovascular system such as aortic dissection and rupture.

MATERIALS AND METHODS

Case Report

Chief complaints

- Patient complains of upper back and chest pain since 2 years and aggravated since 2 months.

Associated complaints

- Improper body alignment (i.e. right shoulder is higher than left shoulder and breast asymmetry) since 2 years.

History of present illness

- An 18-year-old female who was born full term as first child of healthy non consanguineous parents via normal vaginal delivery after an uneventful pregnancy and delivery with attainment of all the developmental milestones without evidence of cognitive deficiency was apparently asymptomatic 10 years ago, she initially presented at 8 years age to an Ophthalmologist with high progressive myopia, for which she is wearing corrective lenses and being followed by Ophthalmologist.
- Later at the age of 17, she gradually developed back pain localised to mid thoracic region, which was non radiating, aggravates during vigorous activities (especially following sports activities like basketball) and disappears after taking rest.
- Also, she was feeling unhappy with her body alignment and has shoulder imbalance (i.e. she started feeling right shoulder is higher than the left shoulder) and breast asymmetry.
- The above symptoms were neglected by the family members thinking that, these symptoms were due to carrying heavy bag pack to the school.
- In the last 2 months, back pain was severe and along with that she developed atypical chest pain, which occurred in the early hours of morning, lasting for approximately 1 min. There were no associated palpitations and autonomic symptoms, pre syncope or syncope and the pain was not related to the respiration or food intake.
- For previously mentioned complaints, she approached Jain Hospital, Vasanth Nagar, Bengaluru and advised to take 2D ECHO and MRI-Whole spine. 2D ECHO was within normal limits and MRI-Whole spine suggested Kyphoscoliotic deformity of Dorso-Lumbar spine with convexity to the right in Mid-dorsal region.
- The patient was advised to undergo Surgery (i.e. Deformity correction using pedicle screw instrumentation and fusion) i/v/o Adolescent Idiopathic Scoliosis, due to the long-term negative impact of signs and symptoms upon her health.
- Her family members refused to undergo surgery after knowing its complications and hence approached to SJGAUH Bengaluru for further management.

Past history: N/K/C/O-DM/HTN/Thyroid Dysfunction K/C/O - Bilateral Ectopia lentis with High Progressive Myopia (for which she is wearing corrective lenses)

Family history: Nothing contributory.

Gynecological history: Age of menarche-14 years.
M/H: 3-5 days of menstrual flow with 28-30 days cycle.

Personal history

Table 1: Showing subject's personal history

Diet: Mixed diet with less quantity	Emotional status: Worried and tensed
Appetite: Good	Addiction: None
Bowel: Regular	Height: 176cm
Micturition: Regular	Weight: 38 kg
Sleep: Disturbed	BMI: 13.9kg/m^2

Clinical Findings

General appearance and built

- The patient appeared too thin and slender with her body weight less than average for her age and sex, hinting at an aesthetic body type and she had notable discrepancy between the length of her limbs and trunk. (Fig 1)
- The girl presented with positive Walker Murdoch Wrist Sign (overlapping of complete distal phalanx of thumb and little finger when wrapped around the opposite wrist) (Fig 2), a positive Steinberg Thumb Sign (extension of the distal phalanx of the thumb beyond the ulnar border of the hand when opposed across the palm) (Fig 3) and Arachnodactyly (long and slender fingers) (Fig 4) with more flexible joints. (Fig 5)
- An examination of the feet revealed that she had bilateral Pes planus (flat foot) with mild pronation along with elongated toes) (Fig 6)
- She had moderate degree (approx. 40°cob's angle) of Kyphoscoliosis along with winging of scapula. (Fig 7 and 8)

Figure 1

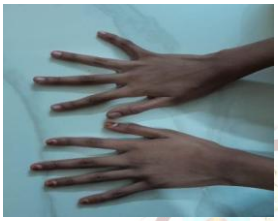


Figure 2

Figure 3

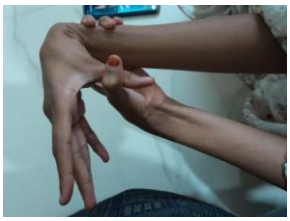


Figure 4

Figure 5

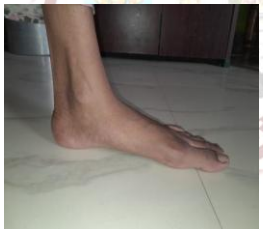


Figure 6

Figure 7

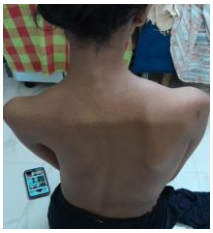


Figure 8

Systemic Examination

A. Central nervous system

- ✓ **HMF**
  - Conscious and well oriented to time, place and person.
  - Intelligence
  - Memory
  - Speech

  } NAD
- ✓ **Cranial Nerves:** Intact
- ✓ **Motor system:** NAD
- ✓ **Sensory system:** NAD
- ✓ Normal Gait and Coordination.

B. Respiratory system

- ✓ **On Inspection**
  - No deviation in the position of trachea.
  - B/L asymmetrical chest with mild Pectus carinatum deformity.
  - Kyphoscoliotic deformity of dorsolumbar spine with convexity to the right was observed at Mid dorsal region.
  - Breathing pattern was thoraco-abdominal.
- ✓ **On Palpation:** Chest expansion and Breathing movements were B/L equal.
- ✓ **On Percussion:** B/L normal percussion noted.
- ✓ **On Auscultation:** B/L NVBS heard, no added sounds were noted.

C. Cardiovascular System

- ✓ **On Inspection:**
  - There were no visible pulsations were observed.
  - Mild Pectus carinatum deformity seen.
  - Kyphoscoliotic deformity of dorsolumbar spine with convexity to the right was observed.
- ✓ **On Palpation:** Apical impulse felt at 1 cm medial to midclavicular line at the left 5th intercoastal space.

- ✓ **On Percussion:** Cardiac dullness was noted.
- ✓ **On Auscultation:** S_1 and S_2 heard, no added sounds were noted.

D. Gastrointestinal System

- ✓ **On Inspection**
  - Shape of the abdomen was scaphoid with centrally placed umbilicus.
  - No surgical marks, striae, scratch, bruising and dilated veins.
- ✓ **On Auscultation:** Low pitched bowel sounds were heard (2-3/min).
- ✓ **On Palpation:** Soft and non-tender on superficial and deep palpation.
- ✓ **On Percussion:** Tympanic all over the abdomen, NAD.

E. Musculoskeletal System

GALS screening

- ✓ **G**ait: Normal with slit tilt in pelvis with normal stance.
- ✓ **A**rm/ UL: Arachnodactyly.
  Positive Walker Murdoch Wrist Sign.
  Positive Steinberg Thumb Sign.
- ✓ **L**eg/LL: Pes planus (flat foot) with mild pronation along with elongated toes.
- ✓ **S**pine:

- Khyphoscoliotic deformity of Dorso-Lumbar spine with convexity to the right in Mid-dorsal region.
- No Tenderness on palpation.
- All other tests were negative.

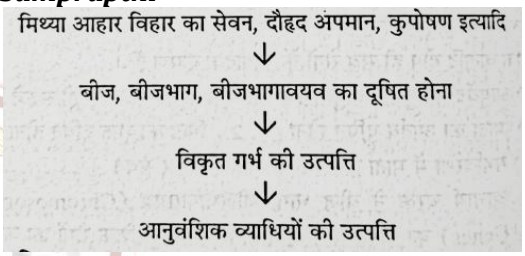
Dashavidha Pareeksha

1. *Prakruti – Vaata pittaja*
2. *Vikruti – Beeja bhaaga and Beeja bhaagaavayava dusti*
3. *Sara- Heena saara*
4. *Samhanana - Asamhata gaatra*
5. *Satmya – Sarvarasa satmya*
6. *Satva - Madhyama*
7. *Pramana* – Height-176cm and weight-38kg
8. *Ahara shakti –Abhyavarana shakti* and *Jarana shakti-Avara*
9. *Vyayama shakti - Avara*
10. *Vaya* - 18 years

Nidana Panchaka

1. *Nidana-Beejabhaaga* and *Beejabhaagaavayava dusti, Prajagara, Alpabhojana.*
2. *Poorvaroopa – Avyakta.*
3. *Roopa* - Back pain with Improper body alignment.
4. *Upashaya – Vishrama.*
5. *Anupashaya – Vyayama* and *shrama.*

Samprapthi

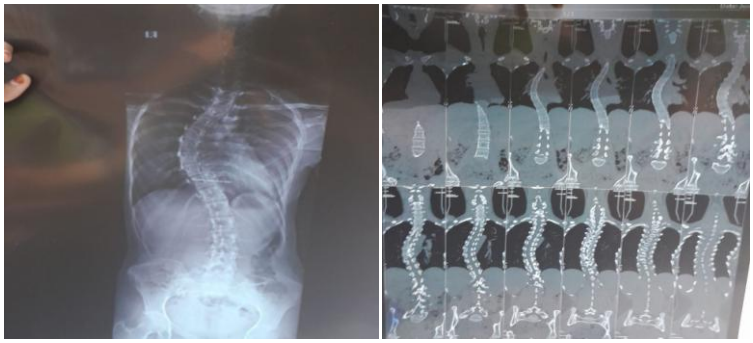


Differential Diagnosis

Disease	Inclusion Criteria	Exclusion Criteria
Adolescent Idiopathic Scoliosis	Kyphoscoliosis	There were extra symptoms other than this.
Loeys-Dietz Syndrome	Tall stature with Arachnodactyly Progressive Myopia Kyphoscoliosis Pes planus Pectus carinatum	No evidence of Club foot, Cleft palate and Bifid uvula.
Marfan Syndrome	Tall stature with Arachnodactyly Progressive Myopia Kyphoscoliosis, Pes planus Wrist sign and Thumb sign Pectus carinatum	-

Diagnostic Assessment

Investigations



DISCUSSION

In Ayurveda, *Sahaja vyadhis* are said to be *Asadhya*, symptomatic and supportive treatment can be given to improve the quality of life. Main Symptom in this case being Kyphoscoliosis, aim of our treatment is to halt the progression of the curve, to strengthen the supporting structures and to increase the flexibility of spine. We can adopt *Vata vyadhi chikitsa, Udwartana, Pizhinjuthadaval, Shashtika Shaali Pinda Sweda* and *Basti* along with *Rasayanas* and Physiotherapy. *Udwartana* given as *strotoshodhana* and also it is *Deepana* and *Pachana* at *Sukshma dhatu* level. *Pizhinjuthadaval* is *Snehana* and *Swedana* at the same time. *Snehana* also has the property of *Dhatu poshana*. The main aim of *Shashtika Shaali Pinda Sweda* is to strengthen the supporting structures. *Basti karma* with *Vatahara dravya* and *Rasayanas* such as *Bhallataka rasayana* also gives better results.

CONCLUSION

Ayurveda's understanding of genetic principles dates back centuries, offering insights into disease prevention and treatment. Today, lifestyle disorders account for a larger share of healthcare spending and research than genetic disorders.

Genetic disorders occur due to alterations, deletions, duplications, or mutations in the DNA sequence, affecting gene function. As per WHO, Genetic disorders and birth defects affect 2-5% of newborns, driving paediatric hospitalizations and accounting for half of childhood deaths in developed nations.

Acharyas (ancient Indian sages) discussed genetic concepts, including:

1. *Beeja* (sperm and ovum)
2. *Beejabhaaga* (chromosomes)
3. *Beejabhaagaavayava* (genes)

They emphasized planning for healthy progeny, starting before marriage. Certain genetic conditions (*Sahaja Vyadhis*) are incurable (*Asadhya*). When treatment options are limited, following the Acharyas' guidelines can help minimize genetic issues in future generations.

Their recommendations focus on:

- Pre-marital planning
- Healthy lifestyle choices
- Genetic awareness

By adopting these principles, society can reduce the incidence of genetic disorders.

Declaration of patient consent:

The authors declare that they have obtained the patient's informed consent, including permission to publish photographs.

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