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IJAPC

e ISSN 2350 0204

VOLUME 12 ISSUE 1 2020

GREENTREE GROUP PUBLISHERS (GGP)



Hartnup's Disease - A Case Report

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ABSTRACT

Hartnup's disease is a rare disease due to abnormal absorption and excretion of tryptophan and other aminoacids. It presents with bilaterally symmetrical erythema at the sites of solar exposure. The dermatitis begins in the form of an erythema with acute or intermittent onset gradually changing to an exudative eruption on the dorsa of the hand, face, neck, and chest with pruritus and burning. The condition is correlated to *Vatarakta* based upon its presentations. The case presented here is managed by *Rasayana* (Rejuvenating) medicines which rectified the pathology of *Vatarakta* and relieved the symptoms.

KEYWORDS

Hartnup's disease, Vatarakta, Yashtimadhu, Rasayana



Greentree Group Publishers

Received 27/11/19 Accepted 28/12/19 Published 10/01/2020



INTRODUCTION

The disorders produced by the conjugation of vitiated *vata* and *rakta* generate multiple health ailments which are referred to as *Vatarakta*. The etio-pathogenesis of an illness through *avarana* is unique to *Vatavyadhi*. It is a well observed fact that many diseases are understood and recognized by the site of origin, *dosha* involved and the *srotas* afflicted. The two fold *samprapti* of *margavarana* leading to *avarana* of *vata* characterizes the illness *Vatarakta*. The basis of its *samprapti* includes the *margavarana* of *vayu* by *rakta* or by *kapha* and *medas*¹. In parlance to conventional medicine, *Vatarakta* incorporates multiple disorders including arterial as well as venous diseases collectively referred to as peripheral vascular diseases. Obstructive arterial diseases are characterized by symptoms like pain, numbness, coldness and pallor of the limbs.

Hartnup's disease is caused by a mutation in the sodium dependent neutral amino acid transporter gene. It is inherited as an autosomal recessive trait. Diagnosis is done by showing the characteristic amino acid excretion in the urine. It presents with pellagra like dermatitis, diarrhea, and dementia. Weakness, skin lesion aggravated by exposure to sunlight, fever,

consanguinity between parents supports the diagnosis. Acute dermatitis of pellagra resembles sunburn in the first stages, sometimes with vesicles and bullae. The gastro-intestinal disturbances are: anorexia, nausea, epigastric discomfort and chronic or recurrent diarrhea. Anorexia and malabsorbtive diarrhea lead to a state of malnutrition and cachexia. Neuropsychologic manifestation includes photophobia, asthenia, depression, hallucinations, confusions, memory loss and psychosis. As pellagra advances, patient becomes disoriented, confused and delirious².

CASE REPORT

A male child aged 12yrs not a K/C/O DM, initially noticed reddish rashes over the sole of left foot, which was resembling a mosquito bite, which subsided by its own. He used to experience similar kind of symptoms from mid-September to the month of January and it used to subside by its own. Later he developed painful lesions with blackish discolouration in hand and foot. After one year he developed pustules in foot and was on medicines. Later patient's parent noticed that patients physical growth is inappropriate to his age and the complaint was persisting as it is. Later the lesions were noticed in toes,



fingers, knee joints, gluteal region, cheeks, eye lids and ear lobes. Patient also complained of 3-4 episodes of watery diarrhea for a day in the gap of 15 days. Later reddish black lesions on the face, elbow, knee, shoulders, eye lids and ear lobes presented with pain and watery discharge.

The onset of the condition was gradual, nature of progression was distal to proximal part of limbs. Aggravating factor were exposure to cold, humidity, sunlight and relieving factor was summer season. Patient's parents had a consanguineous marriage. None of the others in family have similar kind of illness.

Examination:

Color of the skin: Erythematous rashes on buttocks & cheeks, blackish discoloration on toes & tip of fingers.

Distribution: Symmetrical

Extent: Toes, tip of fingers, elbow, knee joints, gluteal region, cheeks, eye lids and ear lobes

Condition of the skin: Dry, not wrinkled

Elasticity: Normal

Thickness: Normal

Tenderness: Absent

Temperature: Normal

Texture: Smooth

Sensation: Intact

Photosensitivity: Absent

Lesion: Macules, Vesicles, Blisters

Site: Toes, tip of fingers, elbow, knee joints, gluteal region, cheeks, eye lids and ear lobes.

Pattern: Diffuse, Acral distribution

Type: Primary lesion: Macular

Secondary lesion: Scaling, lichenification with crusting

Color: Blackish red, acrocyanosis

Raynaud's Phenomenon: present

Auspitz sign: Negative

Candle grease sign: Negative

Based on the detailed history, examination and investigations patient was diagnosed as *Uthana Vatarakta* and Hartnup's disease

Haematological investigations:

Haemoglobin 10.25%, TC 4700, DC - N 35, L60, E 5, M 0, B 0, ESR 28, LFT, RFT WNL. Urine routine WNL

Intervention:

The principles of management of *Vatarakta* is decoding the *samprapti* of *avarana* of *vata* by *rakta*. This is handled by rectifying the *rakta dushti* and alleviating the *vata dosha*³. As the patient is a child, *Shodhana* treatment could not be administered and the patient was managed by *shamana* (*bahya* and *abhyantara*) and *rasayana*⁴ medicaments. Treatment protocol was as Shown in table no. 1.



Table 1 Showing the treatment details and follow up

Date	Symptom	Medication	Duration
21/1/16	Skin discolouration Lesions	<i>Cap .Guduchi 6 tid</i> <i>Dashamoola khada 3 tsf tid</i> <i>Tab Kaishora guggulu DS 1 tid</i> <i>Yastimadhu taila for Veshtana⁵</i>	2 months
31/3/16	Skin discolouration Lesions	<i>Cap Yastimadhu Rasayana 1tid,2 tid,3 tid,4tid. Cap .Guduchi 6 bd</i> <i>Yastimadhu taila for Veshtana</i>	3 months
27/9/16	Skin discolouration Lesions	<i>Cap lashuna Rasayana 2 tid</i> <i>Cap .Guduchi 6 bd</i> <i>Chitrakasava 3 tsf tid</i>	2 months
26/12/16	Skin discolouration Lesions	<i>Adv admission: Cap Yastimadhu Rasayana 4 tid</i> <i>Yastimadhu taila vestana to all limbs</i> <i>Tab kaisharo guggulu DS 1 tid</i> <i>Chitrakasava 3 tsf tid</i>	1 month
27/1/17	Skin, discolouration, Lesions	<i>Cap .Guduchi 4 tid</i> <i>Lashunadi Vati 1 tid</i> <i>Yastimadhu taila for E/A</i>	3 months
14/10/17	Skin discolouration Lesions	<i>Cap .Guduchi 2 tid</i> <i>Yastimadhu taila for E/A</i>	2 months

RESULTS

As evidenced in the images, the skin manifestations of erythematous rashes, macular lesions, pustules, pain, tenderness, discolouration, scaling, lichenification with crusting have reduced to a maximum extent. Most of the lesions have completely healed and the secretions have completely stopped. Inflammatory lesions have healed as shown in figure 1 to 10





Figure 1-6 showing before treatment



Figure 7-10 showing after Treatment

Thus symptoms of pain and tenderness have subsided. There was no major changes in the haematological parameters except for the reduction in the WBC count and slight improvement in the haemoglobin percentage.

DISCUSSION

Skin lesions manifested at an early age, initiated with rashes, reddish and blackish discolorations especially in the acral areas to begin with. These were later followed by blister like lesions which were painful. Chronic course of the illness with mild remissions and exacerbations is the course of the illness. The patient presents with discolorations, and an increase in winter and cold seasons, reddish-blue lesions of the face and nose with swellings of the fingers (Acral). The condition was correlated to Uttana Vatarakta based on the features.

Among the *chikitsa* mentioned for *Vatarakta*, *Shodhana* and *Shamana*, *Shamana* is preferred based on the age and



fitness. *Dosha shamana* medications and *Rasayana* medications are used in this case. *Guduchi* is considered to be the *agrya dravya* for the disease. *Guduchi* is the best *Vyadhihara Rasayana* in chronic lingering disorder. *Guduchi* having *kashaya* (astringent), *tikta* (bitter) *rasa* is effective in conditions of morbid *vata* and *kapha dosha* though it undergoes *madhura vipaka*. It is *laghu* in nature thus gets digested and assimilated easily and is *ushna veerya* and thus is effective in counteracting *kapha vata dosha* which is the pathological basis of *Vatarakta*. Aqueous extract of the *Guduchi* in a dosage of 12 capsules, where each capsules is of 500mg in divided doses in empty stomach was administered. As it is in concentrated form it can be inferred that its action will also be more than the regular drug. To some extent it can be taken as similar to *ghana* administration. *Guduchi* is explained to be *kushtaghna*, *raktashodhaka*, *dahaprashamana* and *rasayana*. *Guduchi* contains alkaloids, diterpenoid lactones, glycosides, steroids, sesquiterpenoid, aliphatic compound and anti-oxidants. Its antibacterial, analgesic, immunostimulant, antioxidant and anti-inflammatory properties are established. The same is expected in the case and achieved⁶. *Lashuna rasayana* contains only freeze dried powder of *Lashuna (Allium sativum)*. *Lashuna* has been told as best for

vatavyadhi. It is explained as an *ahara dravya*, as a *shamana aushadhi* and also as a *vyadhihara rasayana*. The properties of *Lashuna* are antagonistic to morbid *vata* and secondarily to morbidity of *kapha*. The *Lashuna rasayana vidhi* has been detailed in the literature with various *anupana*. *Kshira* has been selected in the study for the pacification of *vata pitta dosha*. During the whole course of the treatment patient was extremely comfortable with no undesirable effects. The administration of the medicine gives symptomatic relief thus owing to the fact that the local inflammation is being cleared. Remission of the pain is also observed. *Lashuna* is said to possess *ushna* and *tikshna* properties which may precipitate morbidity of the *pitta* and *rakta dosha*⁷. Thus in treatment of *avarana* pathology of any disease including *Vatarakta*, *Lashuna* is not indicated in the first phase of the disease. In *Raktavruta vata*, *Vatarakta chikitsa* is used. *Raktapitta anilahara chikitsa*, and *Rasayana* is beneficial⁸. Thus *Lashuna* is used in the second phase of treatment.

Yasthimadhu is used both as a *bahya chikitsa* and *rasayana* medication in this patient. *Yasthimadhu* has *madhura rasa*, *sheeta virya*, *madhura vipaka*. It is *vata-pitta shamaka*. The main chemical constituent of liquorice is glycyrrhizin (about 2-9%), a triterpene saponin with low



haemolytic index. Glycyrrhetic (glycyrrhetic) acid (0.5-0.9%), the aglycon of glycyrrhizin is also present in the root. Other active constituents of liquorice include isoflavonoids, chalcones, coumarins, triterpenoids and sterols, lignans, amino acids, amines, gums and volatile oils. Moreover, studies conducted on modern scientific parameters have proved the wound healing, anti-ulcer, anti-inflammatory antithrombotic effect, hepatoprotective effect, cerebroprotective effect, antidyslipidaemic activity, antioxidant activity and skin regeneration⁹.

CONCLUSION

The medicaments selected on the basis of rakta and vata dushti prevalent in the disease Vatarakta have helped in rectifying the pathogenesis of the disease evidenced by the reduction of signs and symptoms. The benefits of rasayana have been proved.



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