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AN *AYURVEDIC* APPROACH IN THE MANAGEMENT OF HYPOMELANOSIS OF ITO

A CASE REPORT

[1]Dr Lekshmi M K, [2]Dr Anupama S Kumar
[1]Associate Professor, [2]Assistant Professor
[1,2]Department of Kaumarabhritya
[1]Government Ayurveda College Hospital for Women & Children, Poojappura, Thiruvananthapuram, Kerala
[2]Sree Narayana Institute of Ayurvedic Studies and Research, Puthur, Kollam, Kerala

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Abstract

Background: Hypomelanosis of Ito (HOI) is a neurocutaneous disorder characterized by bizarre, bilateral and irregularly shaped leukoderma affecting the trunk and extremities, often associated with neurological and musculoskeletal abnormalities. It is also called incontinentia pigmenti achromians and is the third most common neurocutaneous disorder after Neurofibromatosis and Tuberous sclerosis. Considering the symptomatology, this condition can be correlated as *Sahaja Sarvangavatam* (diseases due to *vata* affecting the whole body) with *Vyanavrutha Vatha* (subtype of *vata*) and skin involvement is predominantly visible.

Objective: The article highlights the scope of an integrated approach in treating neurocutaneous disorders, combining Ayurvedic intervention along with physiotherapy.

Methodology: This is a case study of a 4-year-old child presenting with hypomelanotic areas on extensor part of right arm by birth, seizure disorder, mild hearing loss, inability to sit without support and mild spasticity of bilateral lower limbs. He visited the outpatient department of our tertiary Ayurvedic Health Centre and took outpatient management for about two weeks with the administration of *Ashwagandharishtam, Rajanyadi churnam, Kalyanavaleha churnam, Priyalamajjadi modakam* and *Mahanarayana tailam*. IP management was also taken along with dietary recommendations for about one month and twelve days with the line of management of *Vatavyadhi* (diseases caused by *Vata*) *Chikitsa (*treatment) along with *Samana oushadhas* (palliative medicines). The outcome was assessed using Ages and Stages Questionnaire before and after the treatment.

Result: Head holding was improved from Grade 4 to Grade 5 with reduction in the spasticity of bilateral lower limbs. Crawling was attained with reduction in drooling and colour of the hypopigmented patches got mildly regressed along with initiation of new sounds and words with no further episodes of seizures. The Ages and Stages Questionnaire improved from 195 to 245, providing valuable insights into management strategies.

Conclusion: An integrated approach involving Ayurveda and physiotherapy provides a sustainable solution in improving the quality of life of the child.

Keywords: *Ayurveda*, Hypomelanosis of Ito, *Sahaja Sarvangavatam with Apasmaram*, *Vyanavrutha vata*, Ages and Stages Questionnaire.

Introduction

Hypomelanosis of Ito (HI), or pigmentary mosaicism, was previously named incontinentia pigmenti achromians. The disease is currently named Hypomelanosis of Ito to distinguish this condition from incontinentia pigmenti[1]. HI was first described by Ito in 1952. It represents the third most frequent neurocutaneous disease, after Neurofibromatosis type 1 and Tuberous sclerosis[2].

It is a rare neuroectodermal disorder often associated with mental retardation and epilepsy. It is characterised by skin abnormalities in the form of unilateral or bilateral cutaneous macular hypopigmented

whorls, streaks and patches, corresponding to Blaschko's lines. Various chromosomal anomalies have been identified in some patients and the current consensus is that the phenotype of hyperpigmentation or hypopigmentation following Blaschko's lines occurs due to cutaneous mosaicism, either for a monogenic or a chromosomal disorder[3]. The nervous system is the most commonly affected system in the form of intellectual disability (70%), seizures (40%), microcephaly (25%) and muscular hypotonia (15%). The musculoskeletal system is the second most frequently involved system, affected by scoliosis and thoracic and limb deformities. Twenty-five per cent of patients have minor ophthalmologic defects (strabismus, nystagmus) and 10% have cardiac defects. In Ayurveda, these genetic diseases come under *adhibalapravruthavyadhi*[4] (~genetically predisposed disease). Here, this condition can be correlated to *Sahaja Sarvangavata* with *Apasmara* (~seizure) along with the involvement of *Vyanavrutha vata* (subtype of *vata*).

Patient information

A 4-year-old male child, was brought to the OPD presenting with hypopigmented patches over the right extensor and flexor aspect of right upper limb and over the right upper shoulder region, mild spasticity of bilateral lower limbs with early infantile epileptiform encephalopathy, mild conductive hearing loss of left ear and inability to sit without support. From a non consanguinous parentage, he was born through LSCS due to premature rupture of membranes with a birth weight of 3.4kg with good sucking. Child developed polymorphic tonic clonic myoclonic seizure on 2nd day of delivery. 2nd episode of seizure was developed after 2 weeks of birth. Child was shifted to Tertiary Pediatric Neurological Centre and ventilatory support was provided for 1 month. After that sucking was poor and tubal feeding was done for the next 4 months. Since then the child started taking allopathic medication. All the developmental milestones were delayed with multiple episodes of seizure lasting for about 1 – 2 seconds. At 7 months of age, he started taking physiotherapy and continued till 2 years of age with left hand preference noted. At 2.5 years, he took treatment from a tertiary private Ayurvedic Hospital, but no satisfactory improvement was obtained. Four months back, he developed seizure with jerky movements with up rolling of eyes. They also started taking Homeopathic medications for 1 year. Father's cousin brother and mother's sister had seizure history.

Figure 1. Hypopigmented patches over the right shoulder region

Figure 2. Hypopigmented patches over the extensor and flexor aspect of right upper limb

Clinical findings

On examination, the child was found to have hypopigmentation over the extensor and flexor aspect of right upper limb, convergent squint, dental caries and mild spasticity of bilateral lower limbs. Mild wheeze noted during the morning hours with mild conductive hearing loss over the left ear. After detailed systemic examination, the treatment protocol was planned based on *Agni* (~digestive fire), *dosha* , *dushya* (bodily tissues) and *srotas (*the channels).

Timeline

The timeline of the patient's treatment and outcomes are enlisted in the table.

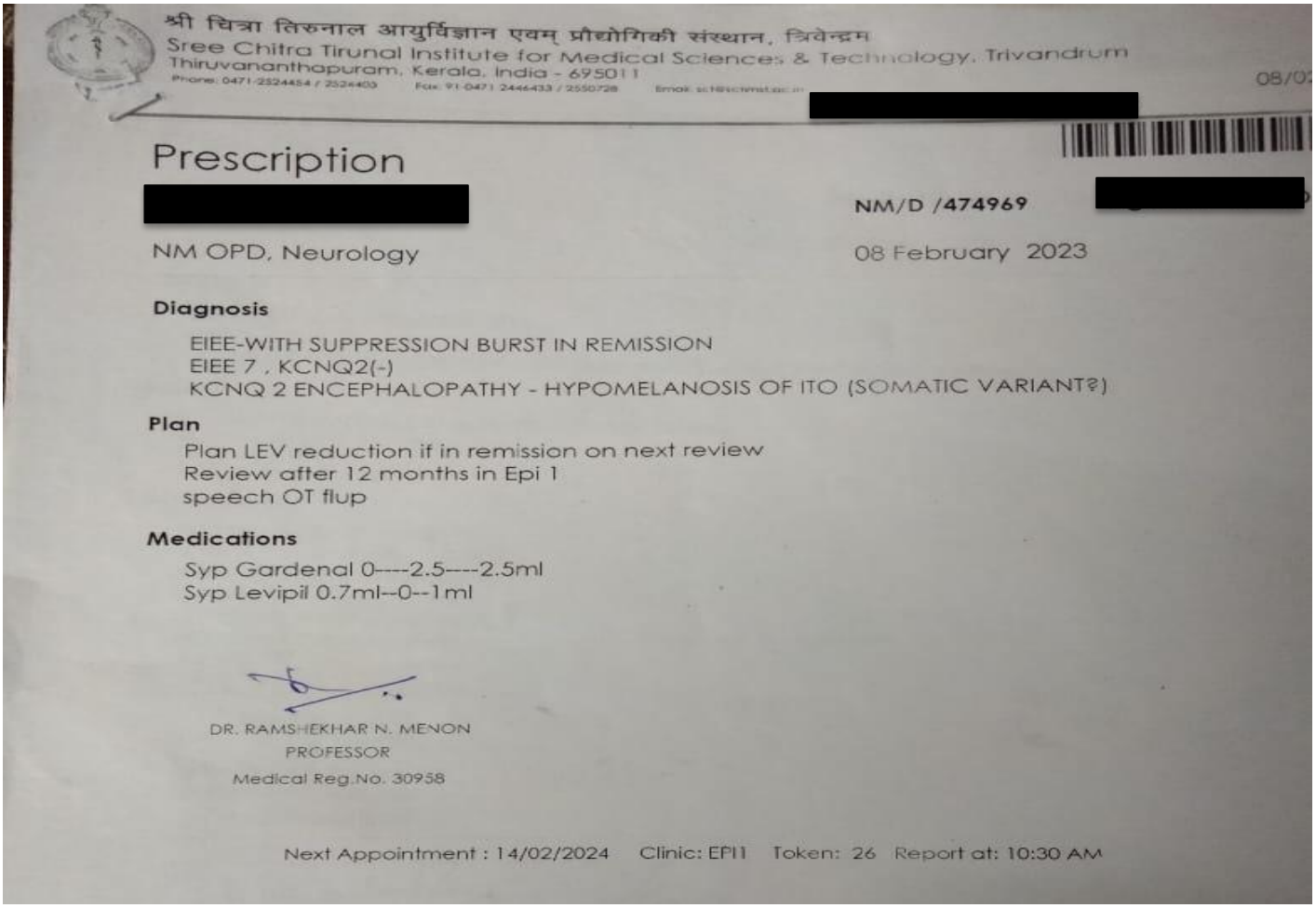
Table 1. Timeline showing treatment and improvement

Date of Admission	Complaints/ conditions of the patient	Intervention
20/07/23 – 21/07/23	Inability to sit without support Hypopigmented patches over the extensor and flexor aspects of right upper limb and over the right upper shoulder region Mild conductive hearing loss of left ear Stand on toes with support Developmental delay Speech delay Convergent squint	2.5gm of *Rajanyadi churna*[4] once daily after food 5 drops of *Dhanwantharam Avarthi* once daily before food.
21/07/23 – 23/07/23	Complaints persist	*Kashayadhara* with *Dasamoola Kashaya churnam* for 3 days for 45minutes
24/07/23 – 28/07/23	Complaints persist Child was irritable during the procedure Reduced appetite at morning hours and good appetite during afternoon	*Udwarthanam* with *Kolakulathadi Churna* for 5 days 5gm of *Kalyanavaleha churna* along with honey applied over the tongue 5gm of *Priyalamajjadi modakam* given twice daily

29/07/23 – 4/08/23	Appetite improved Drooling reduced slightly. Other complaints persist.	*Abhyangam* with *Karpooradi tailam* and *Karpasastyadi tailam* for 7days *Thalam* with *Dhanwantharam tailam* and *rasnadi churnam* for 7 days. *Spine pichu* with *Mahamasha tailam* – Inverted T shaped *pichu* for 45minutes
5/08/23 – 11/08/23	<u>Improvements</u> Drooling reduced Sleep increased – started taking nap for 2 -3 hours during afternoon <u>Complaints</u> Appetite slightly reduced in the morning hours Less urine output comparatively	*Manjal kizhi* done for 7 days 10 drops of Caspa Drops given twice daily after food. 5gm of *Dadimashtaka churna* given along with honey twice daily after food.
	Urine Analysis Pus cells – 1-3/hpf Epitehlial cells – 1-2/hpf Albumin, sugar – Nil	Advised *Chandraprabha* tab – 1 tab twice daily after food for 5 days.
12/08/23 – 14/08/23	Complaining of persistent crying, reluctant to food intake and lack of sleep and bowel not passed Motion passed once with normal stool consistency after *matravasthi*	*Ksheera vasthi* with *Dasamoola ksheera kashayam* for 2 days *Tailam – Dhanwantharam mezhuku pakam* *Ghritha – kalyanaka ghritha* *Ksheera vasthi* restricted on 12/08/23 due to above complaints. Advised, *Sukumara ghritha* – 15ml to be given twice daily before food *Swedanam* done with *Erandatailam* and *Karpooradi tailam.* *Matravasthi* done with *pippalyadi anuvasana tailam* 30ml
13/08/ - 14/08/23	Symptoms reduced	*Ksheera vasthi* done for 2 days
15/08/23	Reduced appetite Urine output – Less with yellowish discolouration during the morning hours Tenderness over the lower abdomen with persistent crying lasting for about 30 minutes	Took rest for 1 day Took homeopathic medication
16/08/23 – 18/08/23	Stability and body strength improved with reduced spasticity. Colour of the hypopigmented patches faded.	*Shastika lepam* done for 3 days with *Kolakulathadi churna and Shashtika*
19/08/23 – 25/08/23	Starts making new sounds and tries speaking new words.	*Sirodhara with Mahanarayana tailam*
26/08/23 – 2/09/23	Tries to speak ‘amma’ occasionally. Duration of sitting with support increased. No further seizure episodes	*Sirolepam* with *musta takram* done for 7 days.

Diagnostic assessment

Hypomelanosis of Ito was diagnosed clinically as child fulfilled the major and minor criterias. The major criteria fulfilled by the child was the presence of hypopigmented patches present over the extensor aspect of right upper limb and right upper shoulder region, seizures and developmental delay. The child was having mild conductive hearing loss of left ear and convergent squint with likely pathogenic variant detected in the genetic study which in turn fulfilled the minor criteria. MRI report taken at the age of 4 months was suggestive of cerebellar hypoplasia simplified gyral pattern with frontal pachygyria. Genetic study revealed likely pathogenic variant – KCNQ2 encephalopathy – Hypomelanosis of Ito (Somatic variant). Hearing evaluation revealed mild conductive hearing loss over the left ear. EEG report was taken at the age of 1year and 9 months revealed focal epileptiform abnormalities over bilateral independent centro parietal and midline centro parietal region.



श्री चित्रा तिरुनाल आयुर्विज्ञान एवम् प्रौद्योगिकी संस्थान, त्रिवेन्द्रम
Sree Chitra Tirunal Institute for Medical Sciences & Technology, Trivandrum
Thiruvananthapuram, Kerala, India - 695011

[redacted]

Prescription

[redacted] NM/D /474969

NM OPD, Neurology 08 February 2023

Diagnosis

EIEE-WITH SUPPRESSION BURST IN REMISSION
EIEE 7 , KCNQ2(-)
KCNQ 2 ENCEPHALOPATHY - HYPOMELANOSIS OF ITO (SOMATIC VARIANT?)

Plan

Plan LEV reduction if in remission on next review
Review after 12 months in Epi 1
speech OT flup

Medications

Syp Gardenal 0----2.5----2.5ml
Syp Levipil 0.7ml--0--1ml

DR. RAMSHEKHAR N. MENON
PROFESSOR
Medical Reg.No. 30958

Next Appointment : 14/02/2024 Clinic: EPI1 Token: 26 Report at: 10:30 AM

Figure.3. Report showing the patient was diagnosed with Hypomelanosis of Ito

Therapeutic intervention

After a thorough examination in the outpatient department for about 2 weeks, the patient was admitted in the inpatient department for about 1month and 12 days. Internal medication along with external therapies with dietary restrictions were implemented. The OP medication included *Rajanyadi churnam* (1/4tsp given once daily along with *kurukk*) for *agni deepana* (improve digestive fire) and *vatanulomana* (helps in normal downward movement of vata). *Aswagandharishtam* (5ml twice daily after food) was given as it deals with epilepsy, promotes attention, memory and was useful in treating neurological diseases by boosting the neurotransmission. *Kalyanavaleha churna* (5gm once daily with honey) was given due to its efficacy in dealing with dysarthria, laryngitis, stiffness of tongue and dysphonia. To provide nourishing properties and to improve the overall strength of the body, the child was given 5gm of *Priyalamajjadi Modaka* twice daily. *Dhanwantharam* 101 A was given to have *rasayana* properties (rejuvenation). The IP treatments started with

*Kashayadhara (*pouring medicated water over the body*)* and *Udwarthanam* (powder massage) for 2 and 5 days respectively, then *Abhyanga* (oil massage) done for 7 days, followed by *Manjal Kizhi (*herbal pottali using turmeric as the main ingredient*)*, a kind of fomentation therapy which contains turmeric powder, dried resin of aloe vera and egg white and done for 7 days. Then as *Shodhana*, (purifactory procedure) *Ksheeravasthi* (medicated milk enema) done for 2 days followed by *Shastika lepa* (for 3 days. Focussing on the *Adhishtana, Sirodhara* was planned with *Mahanarayana tailam* for 7 days and lastly with *Musta takra Sriolepam* for 7 days.

Follow up and outcome

The patient was assessed through various clinical examinations. The colour of the hypopigmented patches regressed, with reduction in the spasticity of the bilateral lower limbs. The child showed initiation of speech as he tries to make new sounds and tries to speak, occasionally speaks 'amma'. His overall body strength and stability improved. Drooling got reduced and his appetite improved. He was able to crawl and head holding was improved from Grade 4 to Grade 5. No further episodes of seizure was noticed during and after the treatment period. He was assessed before and after with the help of Ages and Stages Questionnaires and the value changed from 195 to 245.

Discussion

The patient visited the OPD as a diagnosed case of Hypomelanosis of Ito. There is no one to one correlation involved in the Ayurveda classics with HI, but there are conditions and some causative factors linked for such type of diseases. Here, as it is a genetic condition, beeja dushti is one among the *nidana* (causative factor) and involves *Vatapradhana tridosha dushti*. Hence, this condition can be correlated to *Sahaja Sarvangavata with Apasmaram*[5]. Even though a complete cure cannot be assured in genetic disorders, atleast an overall improvement can be made in the Quality of life of the child by analysing the *dosha, dushya and srotas*. In *Samprapthi* (etiopathogenesis), mainly the three *doshas* are involved *Vata, Pitha and Kapha*. Among the *Panchavayu*, *Prana* (*indriya and chithadruk, annaprav*esha affected), *Udana (vakpravruthi, urja , bala* affected) , *Vyana (gati, apakshepana, utkshepana*) and *Samana vayu (anna grahana and pachana*) are being involved. In case of *Pitha, Pachaka (pakadi karma), Bhrajaka (twak* affected) , *Alochaka pitha (drik* affected; convergent squint) are involved. Among the *Panchakapha, Sleshaka* (functioning of indriyas affected) and *Tarpaka Kapha* (*sandhi samslesha*) are involved. The main *dhathus* involved are *rasa, rakta, mamsa, medas, asthi and majja*. The child was treated based on the principle of *Vatavyadhi chikitsa* with various Ayurvedic medicines given internally and appropriate *Panchakarma* procedures along with Physiotherapy and Speech therapy. The external procedures started with *Kashayadhara* (pouring herbal decoction over the body) with *Dasamoola Kashaya churnam* as it prepares the body for the successive procedures. Then *Udwarthanam* (massaging with medicated powder) was done with *Kolakulathadi churna* as it helps in augmenting the fire of the skin, improves blood and lymphatic circulation. *Abhyangam* (oil massage) with *Karpooradi* and *Karpasasthyadi tailam* was done as it helps in *vatanulomana* (helps in the normal movement *of Vata*) , nourishes the body and provides better physical stability. Then *Manjal Kizhi* (a type of fomentation technique in which turmeric is the main content along with other drugs) was done as this procedure helps in reducing the spasticity of bilateral lower limbs, helps to open the channels and remove the *doshas*. Also helps in kindling the digestive fire and softens the body. *Ksheeravasthi* with *Dasamoolaksheera kashayam, Dhanwantharam mezhuku pakam, Kalyanaka Ghritham* and honey was done for 2 days as *vasthi* acts directly on Vata to bring it back to normalcy. Here, *Ksheera vasthi*, a kind of *Brimhana vasthi* helps in purifying the body by removing the toxins carrying out *mridu shodhana* and provides nourishment to the body. *Shastika lepam* (rejuvenating technique) was done for 3 days as it induces nerve stimulation and muscle's relaxation and increase the efficiency of muscle action. Finally, *Sirodhara with Mahanarayana tailam and Sirolepam* were done as they deeply relaxes the nervous system, lowers metabolism and improves the cognition. *Mahanarayana taila*m in turn helps in relieving the *Sakhasritha vata* added on to the efficacy. Both the internal and external therapies along with the implementation of Physiotherapy brought considerable improvement in the patient's overall condition.

Conclusion

There is no complete cure for neurocutaneous syndromes and only symptomatic management can be provided. Panchakarma therapies along with internal medications helps in attainment of certain gross and fine

motor development, no seizure episodes noted and speech also improved. The Quality of Life of patient can be improved and this holistic approach of Ayurveda gives a new ray of hope in the management of these rare disorders.

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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