MAK HILL Publications Research Journal of Medical Sciences

Case Report
doi: 10.36478/makrjms.2024.12.201.205

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**Key Words**
Acropigmentation of dohi, dyschromatosis, nevus spilus

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**Received:** 10 October 2024
**Accepted:** 21 November 2024
**Published:** 23 November 2024

**Citation:** Matta Harika Priya, Metta Arun Kumar, Suggu Sree Ramu, Turpati Narayana Rao, Kethu Preethi Reddy and Kusireddi Trinadh, 2024. Dyschromatosis Symmetric Hereditary with Nevus Spilus: A Rare Association. Res. J. Med. Sci., 18: 201-205, doi: 10.36478/makrjms. 2024.12.201.205



# Dyschromatosis Symmetric Hereditary with Nevus Spilus: A Rare Association

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**ABSTRACT**
Dyschromatosis Symmetric Hereditary (DSH), also referred to as Acropigmentation Symmetric of Dohi or Symmetric Acropigmentation of Dohi, is a rare autosomal dominant genodermatoses with high penetrance. Here, we are presenting 17 years old male with combined hyper pigmented and hypopigmented macules over dorsal aspects of distal extremities and face, scattered dark spots over a faint brownish patch on the trunk . Skin biopsy revealed increased melanin pigment and absence of melanin pigment from hyper pigmented and hypopigmented lesions respectively. It is conservatively managed with counselling and topical sun screens.

## INTRODUCTION

Dyschromatosis Symmetrica Hereditaria ( DSH ), also known as Acropigmentation Symmetrica of Dohi, is a rare pigmentary genodermatoses that is more common in Japan, China and Korea, with a few reported cases in Brazil and India[1]. It is mostly inherited in an autosomal dominant pattern, although sporadic cases and autosomal recessive inheritance are also observed[2]. The onset of lesions occurs in infancy or early childhood and typically stops progressing before adolescence[3]. The condition is characterized by combined hyper pigmentation and hypopigmentation macules on the dorsal aspects of the extremities and the face[4].

**Case Report:** A 16-year-old boy born to non -consanguineous parents presented with asymptomatic mottled hypopigmented with hyper pigmented spots on the dorsal aspects of both hands, feet and hyperpigmented spots on face. These lesions began at the age of 5 years and gradually advanced . At the age of 11 years, his mother noticed scattered dark spots over a faint brown patch over his trunk. His mother had a similar presentation with hyper pigmented macules on the face and less prominent hypopigmentation on the extremities (fig. 3b and 4). On cutaneous examination, multiple hypopigmented macules, ranging in size from 1-3mm, were symmetrically distributed in a discrete and reticulate pattern with a few discrete hyperpigmented macules on the dorsae of both hands and feet (fig. 1 and 2). Multiple freckle-like macules were also present on the face and neck (fig. 3a). Along with this, multiple scattered lentigines of size 0.5 mm over a solitary faint brownish patch of size 5x3 cm distributed over the lower back region of right side of the trunk(fig. 5).The systemic examination did not reveal any notable findings and there were no associated co-morbidities. Routine biochemical investigations including thyroid profile were normal. Chest X-ray and 2D ECHO did not revealed any abnormality. Histopathological examination of a 3 mm punch biopsy from hyper pigmented lesion showed increased melanin pigment whereas from hypopigmented lesion showed absence of melanin pigment in the basal layer ( fig. 6). Based on history, clinical and histopathological features, a diagnosis of dyschromatosis symmetric hereditary with Nevus Spilus was made.

## RESULTS AND DISCUSSIONS

Dyschromatosis Symmetrica Hereditaria (DSH), also known as Reticulate Acropigmentation of Dohi (RAD), was initially described in Japanese patients by I. Toyama in 1929[5]. Prior to this, the Professor Keizo Dohi studied the case series of patients (1920-1922) which were published as 'Symmetric Acropigmentation of Dohi' by Koyama[1]. DSH is generally inherited in an autosomal dominant manner, although there are also reported cases of autosomal recessive and sporadic occurrences[2]. Miyamura *et al.* discovered that heterogeneous mutations of the gene Adenosine Deaminase acting on RNA1 (ADAR1), located on chromosome 6q24.2-q25.22, are responsible for DSH[3,6].It is characterized by the simultaneous occurence of hyperpigmented and hypopigmented macules on the dorsal aspects of hands and feet along with freckles-like macules over the face[3]. It typically only affects the skin, although a few cases have been linked to extra cutaneous abnormalities[7,8]. Family history is present in 56-77.6% of patients with DSH[7,8]. It is always isolated entity as in our case, however related anomalies are idiopathic torsion dystonia, mental deterioration, brain calcification, depression, aortic root sclerosis[8], Congenital Heart Disease[13], aplasia of dental roots, neurofibromatosis[11], achalasia[12], ß thalassemia major[9], acral hypertrophy[10], psoriasis[10] and polydactyly[9] have been reported. Al-Saif *et al* reported a case linked to cutaneous lupus erythematosus and hyperthyroidism[14]. It's important to distinguish this disorder from dyschromatosis universalis hereditarian (DUH), which involves predominantly trunk and extremities and may include anomalies like short stature, deafness, grand mal epilepsy and solar elastosis, unlike DSH[3,8]. Other potential differential diagnoses are reticulate acropigmentation of Kitamura, Familial Speckled acral hypopigmentation, xeroderma pigmentosum and Dowling-Degos disease[8]. Reticulate Acropigmentation of Kitamura shows palmar pits, macules of atrophy and absence of hypopigmented macules seen[15]. Familial Speckled acral hypopigmentation shows hypopigmented macules in speckled form over the dorsae of hand and feet with absence of hyper pigmented macules as, in contrast with DSH. Dowling-Degos disease shows flexural involvement with comedo like lesions and pitted scars with autosomal dominant inheritance[3,15]. Xeroderma pigmentosa shows erythema, telangiectasia, extensive freckling over sun exposed areas due to defective Deoxyribonucleic Acid (DNA) repair mechanism[3,15]. Nevus spilus is also known as Speckled lentiginosus naevus (SLN), a congenital melanocytic naevus presenting as a lentiginous macule early in life which subsequently developing multiple darkly pigmented macules or papules in a speckled distribution[16].As there is no definitive and effective treatment available, the patient to be counselled which alleviates patient's

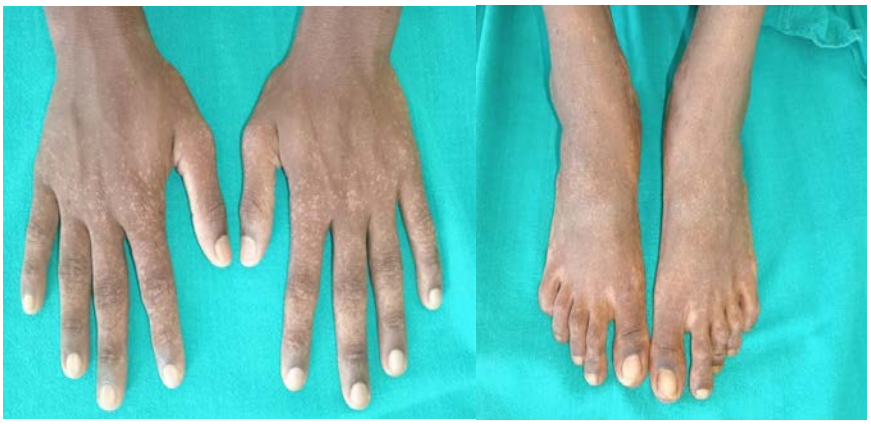

Fig. 1: Reticulated Hypopigmented Macules with a Few Discrete Hyper Pigmented Macules Over the Dorsal Aspect of Both Hands (a) and Feet (b)

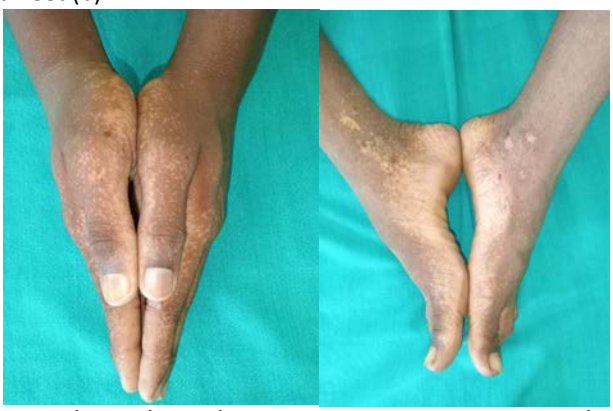

Fig. 2: Reticulated Hypopigmented Macules with a Few Discrete Hyper Pigmented Macules Over Lateral Aspect of Bilateral Hands (a) and Feet (b)

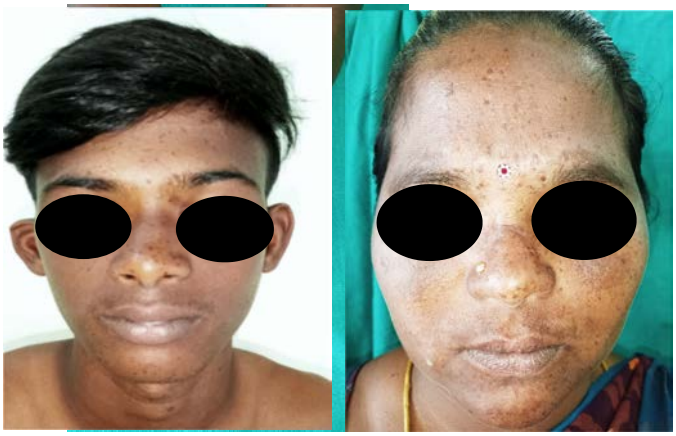

Fig. 3: (a)Freckles-Like Macules Present Over face, (b) Similar Freckles-Like Macules Over the Face in His Mother

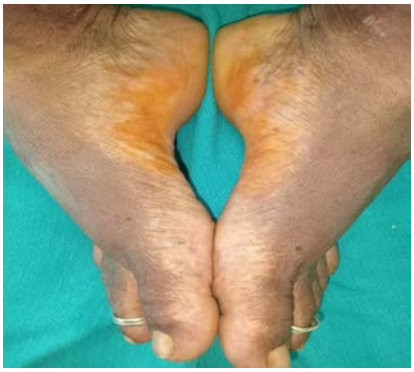

Fig. 4: Less Prominent Hypopigmented Macules( Black Arrow) with Few Discrete Hyper Pigmented Macules (Red Arrow) Over Both Feet in His Mother

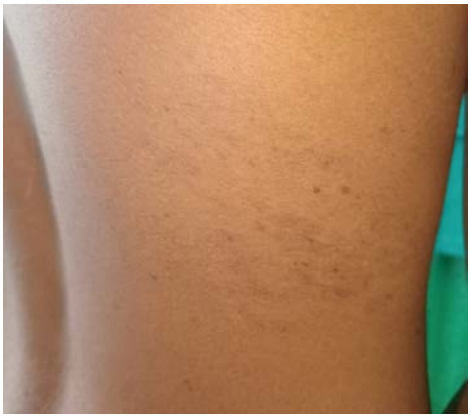

Fig. 5: Multiple Scattered Lentigines Over Brownish Patch Over the Lower Back Region of Trunk

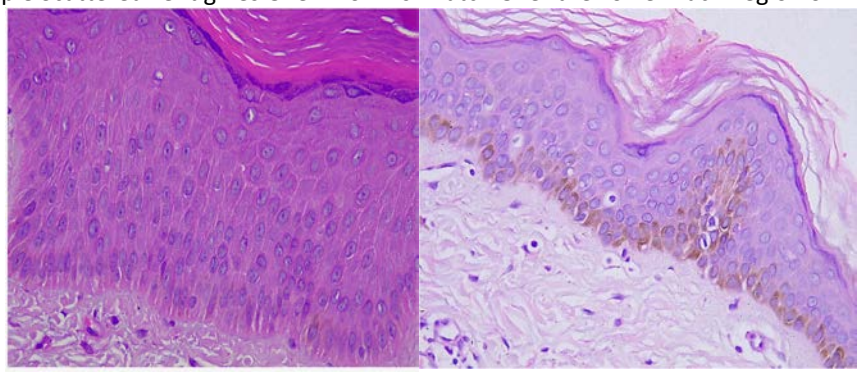

Fig. 6: (a)Absence of Melanin Pigment (Yellow Arrow) and (b) Increased Melanin Pigment (Red Arrow) in the Basal Layer (HandE-High Power)

anxiety . Topical sunscreens can be utilized to reduce the contrast between hypopigmented and hyper pigmented macules[17].

## CONCLUSION

Thorough literature search, revealed only about five cases reported from India. Our case is probably one of its kind DSH associated with Nevus Spilus in Indian context. Most cases are often unreported because of only cutaneous involvement and asymptomatic nature. Hence, good diagnostic acumen and thorough work-up necessitates to rule out the other associations. All these patients require proper counselling regarding the benign nature of the disease.

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