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A case of dyschromatosis symmetrica hereditaria with an associated eyelid hemangioma

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ABSTRACT

INTRODUCTION AND IMPORTANCE: Dyschromatosis symmetrica hereditaria (DSH) are rare autosomal dominant pigmentary genodermatosis characterized by reticular hyper- and hypopigmented skin macules on the dorsal aspect of the extremities and freckle-like spots on the face, sparing the palms and soles. Cutaneous hemangiomas were not reported in the literature with DSH. We describe for the first time to the best of our knowledge a case of DSH with histopathologically confirmed eyelid hemangioma.
CASE PRESENTATION: A 25-year-old female was diagnosed with DSH in her childhood by a dermatologist then later developed cutaneous lupus erythematosus (CLE). Four years later she presented to our clinic with right lower eyelid painless mass. The histopathological examination showed inflamed epidermis overlying a mixed capillary and cavernous hemangioma. The patient had complete healing of the skin post-operatively with excellent cosmetic result.
DISCUSSION: DSH is usually isolated, however, acral hypertrophy, psoriasis, dental anomalies, aortic valve sclerosis, dystonia and intracranial hemangiomas have been reported in association with the disease. The types of the hemangiomas reported were not specified with lack of tissue diagnosis. Our case is unique because of the late occurrence of this eyelid skin hemangioma, the concomitant CLE, the history of hyperthyroidism, and the positive family history of consanguinity.
CONCLUSION: The pathogenesis of DSH is not well understood, however the previously reported intracranial hemangiomas and the currently reported skin vascular lesion would raise the role of inheritance and variable expression of such an association especially with concomitant CLE. This may warrant further studies on the etiology of DSH.

1. Introduction

Dyschromatosis symmetrica hereditaria (DSH) is a rare pigmentary genodermatosis, which is autosomal dominant with an onset during infancy. The characteristic skin manifestations are freckle-like pigmented macules on the face, and hyper- and hypopigmented similar macules on the dorsal aspect of the extremities, however, the palms and soles are usually not involved [1,2]. Although DSH is reported as an isolated disorder, other conditions have been seen in association with it such as psoriasis, aortic valve sclerosis, dental anomalies, and neurological disorders like dystonia and intracranial hemangioma [1]. To the best of our knowledge, cutaneous hemangiomas were not previously reported in patients with DSH in the English-written literature, but intracranial hemangioma has been described without histopathological confirmation [3]. Here, we report a 25-year-old female who was a known case of DSH, and history of cutaneous lupus erythematosus prior to her presentation to the ophthalmic practice at our institution with an eyelid hemangioma that was also confirmed by tissue diagnosis. This case report has been prepared and reported in accordance with the SCARE 2020 criteria [4].

2. Case presentation

A 25-year-old female, known to have dyschromatosis symmetrica hereditaria, presented to our clinic with right lower eyelid painless mass that was stable in size over 2 weeks. There was no associated tearing or discharge. Her past medical history revealed mild joint pain, skin photosensitivity, and discoid form of cutaneous lupus erythematosus (CLE). Her past surgical history revealed his-

Abbreviations: DSH, dyschromatosis symmetrica hereditaria; CLE, cutaneous lupus erythematosus.

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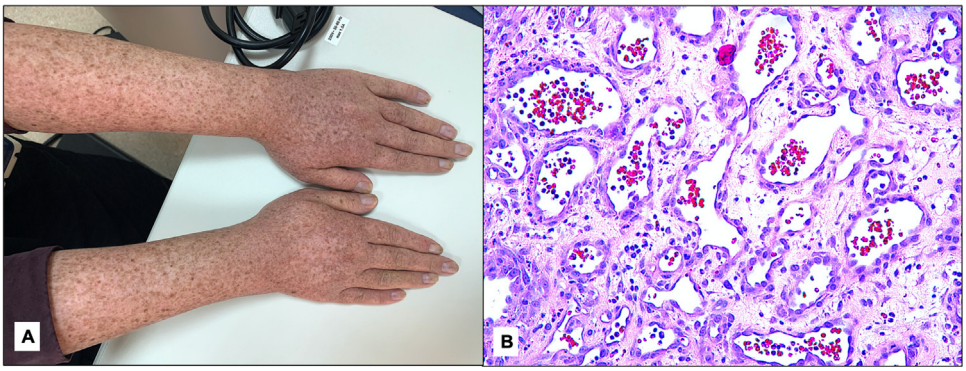

Fig. 1. A: The clinical appearance of the hypopigmented and hyperpigmented macules on the skin of both arms and the dorsum of both hands. B: The histopathologic appearance of the vascular eyelid lesion with numerous capillaries and few dilated endothelial vascular spaces (Original magnification ×200 Hematoxylin and eosin).

tory of total thyroidectomy because of nodular goiter. She was on thyroxine replacement therapy since then. Her family history revealed consanguinity and similar skin pigmentary disorder involving a far-related cousin.

On examination, the lesion was pedunculated, red in color with cauliflower shape, freely mobile, firm in consistency, measuring 3 × 6 mm, and involving the medial aspect of the right lower lid margin. No skin or eyelash destruction was noted. The skin over her face, neck and dorsal extremities was observed to show a mottled appearance with hypo- and hyper- pigmented areas owing to her diagnosed condition (Fig. 1a). The rest of the ophthalmic examination was unremarkable.

The systemic condition was further explained to the patient and she was consented for surgical removal of her eyelid lesion for both diagnostic as well as cosmetic purposes. The patient agreed on the surgical removal of the lesion and underwent successful excisional biopsy in the minor treatment room of our tertiary eye care hospital. The procedure was done by an experienced oculoplastic surgeon under local anesthesia and was well tolerated by the patient with no complications encountered. Postoperatively, the patient was discharged on topical antibiotics with a 2-weeks follow up and the specimen was sent for histopathology. The histopathological examination revealed a skin round lesion showing papillomatosis with an ulcerated and markedly inflamed epidermis overlying a mixed capillary and cavernous hemangioma consisting of predominant hamartomatous capillary proliferation and lobular pattern (Fig. 1b). The patient was instructed to continue her 6-months follow up with the internist for her thyroid and CLE medical issues at a local hospital. For her recent ophthalmic presentation, she was seen in her follow up 2-weeks appointment with complete wound healing at the excision site and excellent cosmetic result. She was informed about the benign nature of the excised skin lesion and was expected to be discharged from our ophthalmic care after her next visit. An informed consent was obtained from the patient for the anonymous use of data.

3. Discussion

Dyschromatosis symmetrica hereditaria (DSH) is a rare pigmentary genodermatosis condition with autosomal dominant inheritance. It is also known as reticulate acro-pigmentation of Dohi. The condition starts in early childhood and stops in adolescence and is manifested by mixed reticular hyper- and hypopigmented macules on the extremities and freckle-like pigmented macules on the face, palms and soles [1,2,5]. Geographically, several cases from various parts of the world but mostly from China and Japan have been reported [1]. Histologically, the hyperpigmented macules reveals abundant melanin deposits in the basal layer and decreased melanin in the hypopigmented areas [6].

DSH is usually an isolated disorder, though other conditions or complications have been associated with the disease such as acral hypertrophy, psoriasis, dental anomalies, aortic valve sclerosis, and neurological manifestations like dystonia [1,7,8]. Eyelid hemangiomas were not reported in the literature in association with DSH. Nevertheless, hemorrhage due to intracranial hemangiomas has been described based on radiological findings only [1,3]. Our patient had also another significant association, which was CLE 4 years prior to her recent ophthalmic presentation, thus she was reported at the age of 21 years by A-Saif et al. [9] DSH can be sporadic and can be familial with an autosomal dominant inheritance [10]. Our patient did not have any genetic testing, but her family history was positive for similar condition in one of the far relatives.

4. Conclusion

Our patient was diagnosed with DSH in her childhood and did not present with hemangiomas elsewhere or remarkable ophthalmic disorders. While the pathogenesis of DSH is still not well understood, such associations of autoimmune cutaneous disease (CLE), her thyroid disorder, and finally the subcutaneous vascular lesion might stimulate further studies to investigate the etiology and to explain the associated pathologies with this disease.

Declaration of competing interest

None.

Funding

None.

Ethical approval

IRB is not required for case reports. However, information was obtained and reported in a manner that was compliant with the standards set forth by the Health Insurance Portability and Accountability Act, and the Declaration of Helsinki as amended in 2013.

Consent

General informed written consent was obtained from the patient including permission for anonymous use of photos and for reporting.

Author's contribution

KMA: data collection, literature review, drafting the manuscript. **HMA**: histopathological images, final approval of the version to be submitted and corresponding author. **ACA:** histopathological diagnosis of the case, critical review of the manuscript. **YAH:** clinical diagnosis and clinical images of the case, critical review of the manuscript.

Registration of research studies

Not applicable.

Guarantor

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Provenance and peer review

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