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CASE REPORT

A Case Report on Dyschromatosis Universalis Hereditaria with Palmar Involvement

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Abstract: Dyschromatosis universalis hereditaria (DUH) is a rare genetic disorder characterized by hyper- and hypopigmented macules forming a reticulate pattern on the skin. This report presents an unusual case of a 14-year-old male patient with DUH involving the palms, a relatively uncommon manifestation of the condition. The patient presented with hyperpigmented lesions on the palms, which had developed over the previous eight years. Family history revealed similar pigmentary changes, suggesting a hereditary pattern. Clinical examination revealed no history of chemical or drug exposure, photosensitivity, or photophobia. Systemic evaluation was unremarkable, except for a low cortisol level of 1.12 mcg/dl (normal range: 6.2 to 19.4 mcg/dl) on the ACTH stimulation test. Skin biopsy from the left palm revealed diffuse hypermelanosis confined to the basal cell layer and occasionally extending to the lower third of the epidermis. Additionally, increased collagenization was observed in the deep dermis. Based on these findings, a diagnosis of DUH was established. The patient was initiated on long-term hydrocortisone therapy for cortisol hormone replacement. This case highlights the rare occurrence of palmar involvement in DUH and underscores the importance of thorough clinical evaluation and histopathological examination in establishing an accurate diagnosis. DUH is an uncommon genodermatosis, and reports of palmar involvement are scarce in the literature. This case contributes to the existing knowledge about the varied clinical presentations of DUH and emphasizes the need for increased awareness among clinicians.

Keywords: Dyschromatosis universalis hereditaria; Hyperpigmentation; Hypopigmentation; Palmar involvement; Genodermatosis

1. Introduction

Dyschromatosis universalis hereditaria (DUH) is a rare genodermatosis characterized by the presence of hyper- and hypopigmented macules that form a reticulate pattern on the skin. This condition is predominantly reported in individuals of Asian descent, with the majority of cases originating from Japan. DUH is a hereditary disorder that can follow either an autosomal dominant or autosomal recessive pattern of inheritance, although the autosomal dominant form is more common. [1,2] The exact etiology of DUH remains elusive, and the underlying mechanisms leading to the distinctive pigmentary changes are not fully understood. However, it is believed that the disorder is linked to abnormalities in melanin production and distribution within the epidermal melanocytes. The clinical manifestations of DUH typically appear during infancy or early childhood, with the development of hyper- and hypopigmented macules distributed across various body areas, including the trunk, extremities, and occasionally the face.

While the lesions are generally asymptomatic, they can have a significant cosmetic impact, affecting the patient's self-esteem and quality of life. The involvement of other structures, such as hair, nails, teeth, palms, and soles, has also been reported in some cases, highlighting the diversity of clinical presentations associated with DUH. Histopathological examination plays a crucial role in the diagnosis of DUH, revealing characteristic findings such as localized increases or decreases in melanin content within the basal layer of the epidermis. [3,4] Additionally, pigmentary incontinence, characterized by the presence of melanin pigment in the dermis, may also be observed in some cases. The management of DUH is primarily focused on addressing the cosmetic concerns and improving the patient's self-confidence. Various treatment modalities have been explored, including topical agents, phototherapy, and laser therapies, with varying degrees of success. However, it is important to note that the effectiveness of these treatments may be limited, and recurrence of the lesions is common. While DUH is generally considered a benign condition, its rarity and unique clinical presentation make it an intriguing subject for further research and investigation. Understanding the underlying genetic and molecular mechanisms involved in DUH could potentially provide insights into the intricate processes governing pigmentation and contribute to the development of more effective therapeutic strategies. [5, 6]

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2. Case presentation

We present the case of a 14-year-old male patient who sought medical attention for hyperpigmented lesions on his palms, which had been progressively worsening over the past eight years. The patient reported no history of trauma, chemical exposure, or drug intake that could account for the pigmentary changes. However, a family history of similar skin manifestations was noted, suggesting a hereditary pattern. Upon physical examination, the patient exhibited well-defined, reticulated hyperpigmented macules on the palms of both hands. The lesions were asymptomatic and did not cause any functional impairment. No other cutaneous abnormalities were observed on the rest of the body, and the patient denied experiencing any photosensitivity or photophobia.

3. Diagnostic investigations

In the pursuit of establishing an accurate diagnosis for the patient's condition, a comprehensive diagnostic workup was undertaken. This involved a combination of laboratory investigations and histopathological examination of the skin lesions. [7,8]

3.1. Laboratory findings

To assess the patient's overall health status and rule out potential underlying systemic or endocrine disorders contributing to the pigmentary changes, a panel of laboratory tests was performed. These tests included:

3.1.1. Complete Blood Count (CBC)

This test evaluates various blood cell types, including red blood cells, white blood cells, and platelets. The results were within normal limits, ruling out any hematological abnormalities.

3.1.2. Comprehensive Metabolic Panel (CMP)

This panel assesses various metabolic parameters, including liver and kidney function, electrolyte levels, and blood glucose levels. The CMP results were unremarkable, indicating normal metabolic function.

3.1.3. Thyroid Function Tests

These tests evaluate the levels of thyroid hormones, such as thyroid-stimulating hormone (TSH), triiodothyronine (T3), and thyroxine (T4). The results were within normal ranges, excluding thyroid disorders as a potential contributing factor.

3.1.4. ACTH Stimulation Test

This test evaluates the adrenal gland's response to adrenocorticotropic hormone (ACTH) by measuring cortisol levels before and after ACTH administration. Notably, the patient's cortisol level was found to be low at 1.12 mcg/dl (normal range: 6.2 to 19.4 mcg/dl), suggesting a potential adrenal insufficiency.

3.2. Histopathological Examination

To further investigate the nature of the pigmentary changes and support the clinical diagnosis, a skin biopsy was performed on one of the hyperpigmented lesions on the patient's left palm. [9, 10] Histopathological examination of the biopsy specimen revealed distinctive findings characteristic of dyschromatosis universalis hereditaria (DUH):

3.2.1. Diffuse hypermelanosis

The epidermis exhibited an increased concentration of melanin pigment, primarily localized to the basal cell layer, extending occasionally to the lower third of the epidermal thickness.

3.2.2. Basal layer changes

The basal layer of the epidermis displayed an irregular distribution of melanocytes, with areas of increased and decreased melanin content.

3.2.3. Dermal changes

The deep dermis showed evidence of enhanced collagenization, a common finding in DUH.

These histopathological findings, coupled with the clinical presentation and family history, strongly supported the diagnosis of dyschromatosis universalis hereditaria (DUH)

4. Differential diagnosis

While establishing the diagnosis of DUH, it is essential to consider other conditions that may present with similar pigmentary changes. The differential diagnosis for DUH includes:

4.1. Dyschromatosis symmetrica hereditaria (DSH)

Also known as reticulate acropigmentation of Dohi, DSH is a closely related genodermatosis characterized by hyper- and hypopigmented macules in a reticulate pattern, primarily affecting the extremities.

4.2. Xeroderma pigmentosum (XP)

This rare genetic disorder is characterized by an extreme sensitivity to ultraviolet radiation, leading to the development of hyperpigmented and hypopigmented macules, as well as an increased risk of skin cancers.

4.3. Incontinentia pigmenti (IP)

A rare X-linked dominant disorder affecting primarily females, IP presents with characteristic linear and whorled hyperpigmented lesions, often accompanied by other systemic abnormalities.

4.4. Post-inflammatory hyperpigmentation

This condition occurs as a result of inflammatory processes, such as injury, trauma, or certain skin conditions, leading to localized areas of hyperpigmentation.

4.5. Melasma

A common acquired hyperpigmentation disorder, melasma is characterized by irregular, patchy areas of hyperpigmentation, typically affecting the face, and is often associated with hormonal changes or sun exposure.

5. Discussion

Dyschromatosis universalis hereditaria (DUH) is an exceptionally rare genodermatosis, with the majority of reported cases originating from Japan and a relatively small number from other parts of the world. [11, 12] The case presented here highlights an unusual manifestation of DUH involving the palms, which is an infrequent occurrence. This case adds to the limited literature documenting the diverse clinical presentations of this condition. The etiology of DUH remains largely unknown, although it is believed to be linked to abnormalities in melanin production and distribution within the epidermal melanocytes. The presence of a positive family history in this case supports the hereditary nature of the condition, which can follow either an autosomal dominant or autosomal recessive pattern of inheritance. [13, 14] The diagnosis of DUH is primarily based on the characteristic clinical presentation of hyper- and hypopigmented macules arranged in a reticulate pattern, along with histopathological findings of localized increases or decreases in melanin content within the basal layer of the epidermis. In this case, the histopathological examination played a crucial role in confirming the diagnosis, revealing diffuse hypermelanosis confined to the basal cell layer, as well as increased collagenization in the deep dermis. While DUH is generally considered a benign condition, the cosmetic impact of the pigmentary changes can significantly affect the patient's self-esteem and quality of life. [15, 16] Various treatment modalities have been explored, including topical agents, phototherapy, and laser therapies, but their efficacy remains limited, and recurrence of the lesions is common. The finding of a low cortisol level in this patient adds an interesting dimension to the case, as adrenal insufficiency has not been widely reported in association with DUH. This observation warrants further investigation to determine if there is a potential link between DUH and adrenal function, or if it is an incidental finding in this particular case.

6. Conclusion

This case report highlights a rare presentation of dyschromatosis universalis hereditaria (DUH) involving the palms of a 14-year-old male patient. The combination of clinical findings, family history, and histopathological examination enabled an accurate diagnosis of this uncommon genodermatosis. While DUH is generally considered a benign condition, its impact on the patient's cosmetic appearance and self-esteem cannot be overlooked. This case contributes to the existing knowledge about the diverse clinical manifestations of DUH and underscores the importance of thorough evaluation and diagnostic workup in such cases

Compliance with ethical standards

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Conflict of interest statement

The authors declare no conflict of interest.

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Author's short biography

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