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A Case of Oculocutaneous Albinism in a Patient with Hashimoto's Thyroiditis

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What Is Known about This Topic?

- Oculocutaneous albinism is a heterogeneous autosomal recessive disorder characterized by reduced or absent melanin synthesis. To our knowledge, the coexistence of albinism and autoimmune thyroid diseases has not been described prior to the present case report.

What Does This Case Report Add?

- The association between albinism and Hashimoto's thyroiditis in two brothers suggests a possible pathogenetic link between the two conditions.

Keywords
Albinism · Hashimoto's thyroiditis · Thyroid · Autoimmunity

Abstract

Background: Oculocutaneous albinism (OCA) is a heterogeneous autosomal recessive disorder characterized by reduced or absent melanin synthesis. Its prevalence is approximately one in 17,000 individuals worldwide. OCA causes a complete or partial absence of pigment in the skin, hair, and eyes. Reduction of melanin in the eyes results in reduced visual acuity, photophobia, and nystagmus. To our knowledge, the coexistence of albinism and thyroid autoimmune diseases has not been reported. ***Case Report:*** A 37-year-old male with OCA came to our observation for a familial history of autoimmune thyroiditis. He complained with mild asthenia. His brother was affected by OCA and autoimmune thyroiditis. Physical examination revealed the presence of fair skin, platinum white hair, and hypopigmentation of the iris. Laboratory data revealed the presence of subclinical hypothyroidism with positive serum anti-thyroid antibodies. Neck ultrasonography showed a markedly heterogeneously hypoechoic pattern of the thyroid, with a honeycomb-like appearance. ***Discussion:*** In the majority of cases, OCA in Caucasians is caused by mutations in the tyrosinase gene. The tyrosinase peptide is normally expressed on the surface of melanocytes, although recent studies have shown its presence in thyroid follicular epithelial cells of patients with Hashimoto's thyroiditis. Therefore, although the mechanism is unclear, the present case report may suggest a pathogenetic link between OCA and Hashimoto's thyroiditis.

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Introduction

Oculocutaneous albinism (OCA) is a heterogeneous autosomal recessive disorder characterized by reduced or absent melanin synthesis, with a prevalence of approximately one in 17,000 individuals worldwide [1]. OCA causes a complete or partial absence of pigment in the skin, hair, and eyes. Reduction of melanin in the eyes results in reduced visual acuity, photophobia, and nystagmus [2]. To our knowledge, the coexistence of albinism and autoimmune thyroid diseases has not been described. Here we report a case of albinism in a patient with Hashimoto's thyroiditis (HT), whose brother also has a similar association.

Case Report

In July 2017, a 37-year-old male with OCA came to our observation in our outpatient endocrinological clinic for a familial history of autoimmune thyroiditis. He complained of mild asthenia. His brother was affected by OCA and autoimmune thyroiditis. The physical examination revealed the presence of fair skin, platinum white hair, hypopigmentation of the iris, and an enlarged thyroid gland of increased consistency and irregular surface. Laboratory data revealed a subclinical hypothyroidism, with a serum TSH concentration of 5.88 μIU/mL (normal range 0.4–4 μIU/mL), an FT3 of 4.08 pg/mL (normal range 2.7–5.7 pg/mL), and an FT4 of 0.96 ng/dL (normal range 0.7–1.7 ng/dL). The concentration of anti-thyroperoxidase antibody (TPOAb) was >1,000 IU/mL (normal range ≤10 IU/mL) and that of anti-thyroglobulin antibody (TgAb) was 755 IU/mL (normal value ≤30 IU/mL). As shown in Figure 1, ultrasonography of the neck revealed a markedly heterogeneously hypoechoic pattern of the thyroid, with a honeycomb-like appearance and without nodules, as is typical of autoimmune thyroid diseases [3].

Discussion

The coexistence of OCA and HT in our patient as well as in his brother suggests a possible pathophysiological link between the two conditions.

HT is the most common autoimmune endocrine disorder [4]. It is well known that autoimmune diseases tend to cluster in the same patient and in the family. The most common diseases associated with HT are alopecia, vitiligo, celiac disease, and type 1 diabetes [4, 5]. Certain non-autoimmune, chromosomal conditions, namely Turner's syndrome and Down's syndrome, can also be associated with HT [6]. To our knowledge, the coexistence of HT and albinism has not been described prior to this case report.

As mentioned above, OCA is a genetic disorder characterized by hypopigmentation of the hair, skin, and eyes [1]. The clinical spectrum of OCA is variable, with OCA1A being the most severe type, characterized by a complete lack of melanin production throughout life, while the other forms (OCA1B, OCA2, OCA3, and OCA4) are milder and can show some pigment accumulation over time [7]. Prevalence of the different forms of albinism varies depending on the different mutations in different genes. OCA1 (OCA1A + OCA1B) accounts for about 50% of cases worldwide and is the most common form in Caucasians [2, 8]. OCA1 is caused by mutations in the tyrosinase gene located on chromosome 11q14.3 [9]. The gene consists of 5 exons encoding a protein of 529 amino acids, which is an enzyme catalyzing the first two steps in the melanin biosynthesis pathway, converting tyrosine to L-dihydroxy-phenylalanine (DOPA) and subsequently to DOPAquinone [2, 7]. OCA1A is caused by mutations completely abolishing tyrosinase activity, while other mutations of the gene only reduce enzyme activity, allowing some accumulation of the melanin pigment over

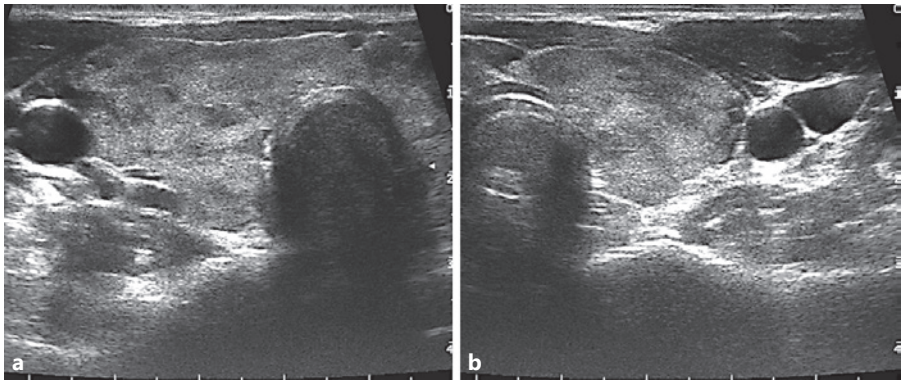

Fig. 1. Ultrasound examination of the thyroid showing an inhomogeneous, hypoechoic, honeycomb-like pattern. **a** Right lobe. **b** Left lobe.

time and causing the OCA1B form. The tyrosinase peptide is normally expressed on the surface of melanocytes, although recent studies have shown its presence also in thyroid follicular epithelial cells of patients with HT [10], suggesting a role of the enzyme in the autoimmune response against the thyroid [11]. Therefore, although the mechanism is unclear, we can speculate a possible link between OCA and HT. In our patient, no genetic analysis was performed, and therefore, it is not known whether and to what extent tyrosinase was involved in the pathogenesis of his albinism. Chromosomal aberrations can be associated with HT [6], but they are unlikely responsible for the coexistence of albinism, in which chromosomal aberrations are not known not play a role.

To our knowledge, this is the first report of a coexistence of albinism and autoimmune thyroiditis. Whether the two conditions share a common pathogenetic mechanism and/or our observation represents something more than a mere, casual event remains to be established.

Statement of Ethics

All diagnostic and therapeutic procedures were in accordance with ethical standards of the institutional and national research committee and with the 1964 Helsinki Declaration and its later amendments or comparable ethical standards. Informed consent was obtained from the individual participant included in the study.

Disclosure Statement

The authors have no conflict of interest.

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