Clinical and Dermoscopic Findings in Goltz Syndrome: Case Report

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SUMMARY Focal dermal hypoplasia or Goltz syndrome is a rare X-linked genodermatosis characterized by cutaneous and musculoskeletal defects. Dermoscopy is a noninvasive auxiliary method for the diagnosis of lesions, whether melanocytic or not. Its widespread use in dermatology is resulting in the description of new patterns and characterization of lesions not reported before its use. A typical case of Goltz syndrome presenting multiple malformations was observed and submitted to dermoscopy. Dermoscopy findings of the papillomas in raspberry form in the perioral and ocular regions, revealed a unique vascular pattern, different from viral warts; dermoscopy of some brownish maculas resembling lentigo in the periphery of skin atrophic areas are described as lentigo-like lesions, an uncommon pattern of melanocytic lesions, but without criteria suggestive of malignancy.

KEY WORDS: focal dermal hypoplasia, Goltz syndrome, lentigo, papilloma, dermoscopy

INTRODUCTION

Goltz syndrome is a rare genodermatosis, affecting all races, with 200 to 300 cases reported worldwide (1). The majority of cases do not present a family history but are transmitted by dominant heritage connected to the X chromosome, with mutation in chromosome Xp11.23 (2). It is more common in women (90% of cases) and usually lethal intra-uterus for male sex (1). Alterations are present at birth and can affect the skin, mucosa, cutaneous appendages, bones, teeth, eyes, heart and blood vessels, lungs, kidneys and central nervous system (3,4).

Dermoscopic examination under polarized light was carried out in two groups of typical lesions of the syndrome: perioral papillomas and lentigo-like lesions.

CASE REPORT

The patient was a 19-year-old female of mixed race, without family history of comorbidities, and with consanguineous maternal grandparents. She presented with hyperchromic macules in linear disposition, following Blaschko's lines all over the body and with hypochromic and atrophic plaques on the trunk, hips and lower limbs, present from birth. She also showed microcephaly, acro-osteolysis of the 3rd and 5th fingers, clinodactyly, shortening of the distal phalanx of the 1st fingers and syndactyly of the 2nd and 3rd left fingers and toes, and scarce, fragile and opaque hair.

Ophthalmologic examination revealed coloboma of the lower iris in the left eye. Odontologic examina-

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tion showed hypodontia, malformations of teeth and defects in tooth enamel. No herniation of fat tissue or deformation in the form of lobster claw was observed. Based on clinical criteria, the patient was diagnosed with Goltz syndrome early in childhood.

The patient already presented several papular and vegetating lesions, isolated or confluent, in the perioral (Fig. 1), conjunctival and inguinal regions. All recurrent papillomas were confirmed by histopathology and treated by shaving and electrocoagulation, excision and suture, and CO_2 laser. During adolescence, progressive occurrence of multiple brownish macules in hypochromic areas in the periphery of atrophic lesions began (Fig. 2).

Dermoscopic examination using polarized light (magnification 10X) of papular perioral lesions revealed a vascular pattern of short tortuous vessels, although symmetrical and diffuse in each flesh-colored lobule (visualization with little pressure or without polarized contact light from the dermatoscope) (Fig. 3) or the same tortuous vessels in the periphery of each lobule (visualization with polarized contact light) (Fig. 4).

Examination of the lentigo-like lesions demonstrated a nonspecific pattern with atypical network, streak-like structures (Fig. 5), with absence of globules or other dermoscopic structures. As no changes were recorded at short- and long-term follow up examinations, biopsy was not performed.

DISCUSSION

Dermoscopy is a recognized auxiliary method in the early diagnosis of melanoma and in the follow up of benign melanocytic lesions. Its use is increasing in dermatology, promoting improvement in the knowledge of the already studied patterns (5), and above

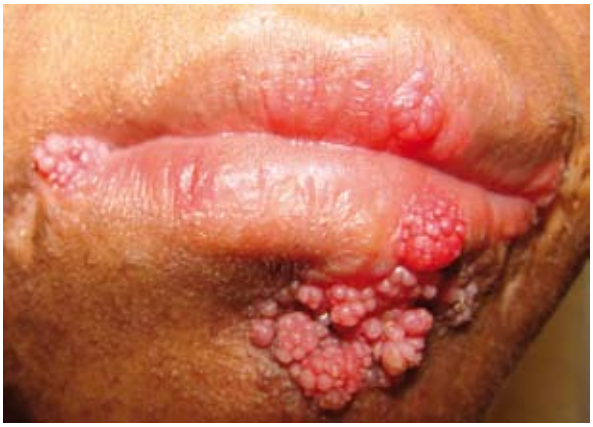

Figure 1. Perioral papillomas.

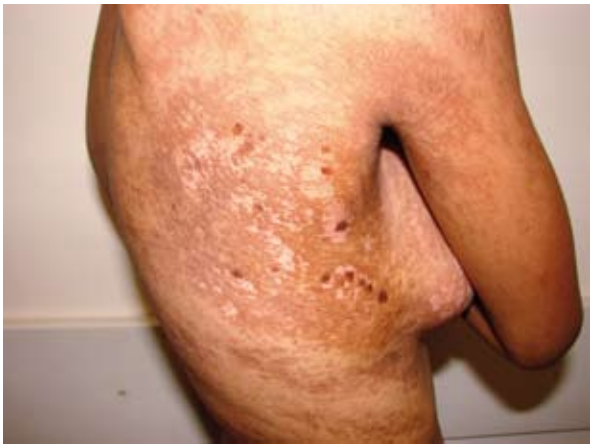

Figure 2. Lentigo-like lesions in the periphery of hypopigmented atrophic lesions.

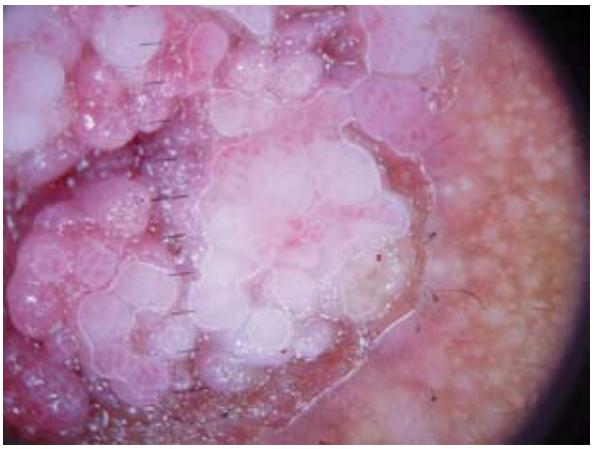

Figure 3. Dermoscopy with little contact pressure of perioral papilloma.

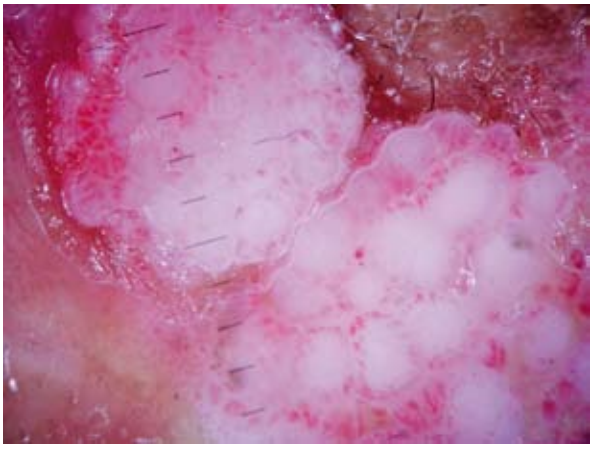

Figure 4. Dermoscopy with contact of perioral papilloma.

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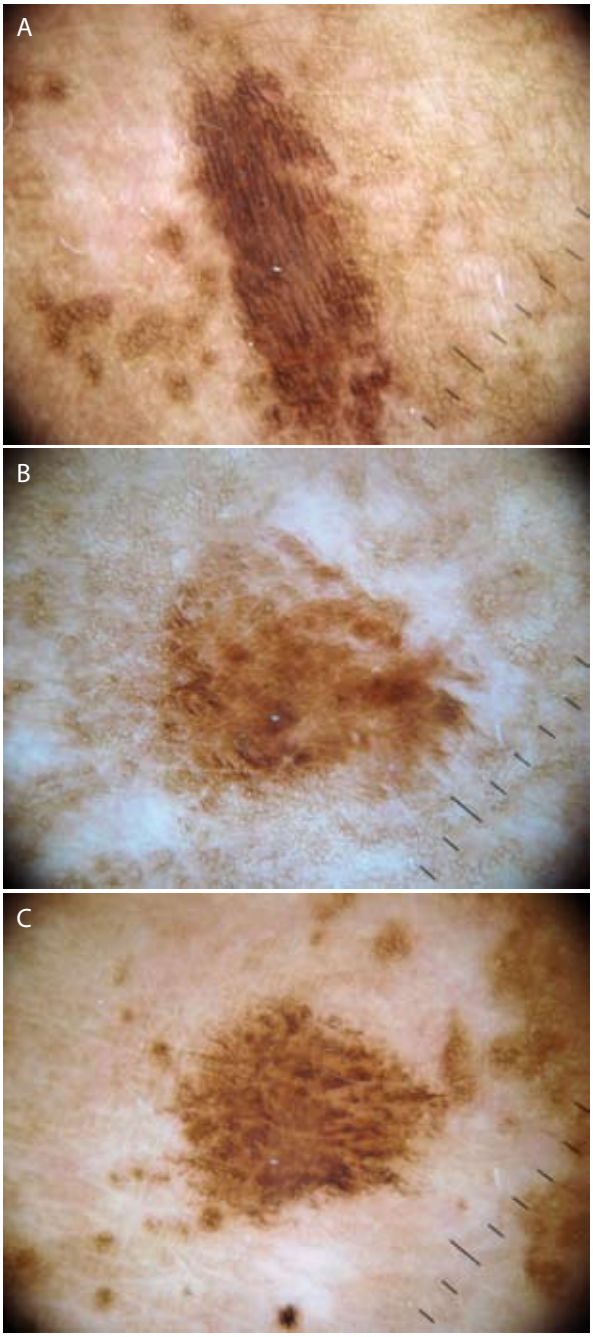

Figure 5. (A) Dermoscopy of a lentigo-like lesion: reticular pattern with atypical network with thickened lines; (B) dermoscopy of a lentigo-like lesion: unspecific pattern with branched lines, and streaks at the periphery; (C) dermoscopy of a lentigo-like lesion: unspecific pattern with thickened branched lines.

all awakening interest in the analysis of new lesions, especially non-melanocytic ones.

In Goltz syndrome, the perioral papular lesions, although clinically very similar to viral warts, are not caused by human papillomavirus. Histopathology of these lesions is usually controversial. It can present as a fibrovascular peduncle of loose connective tissue and numerous swollen blood vessels surrounded by intense mixed inflammatory infiltrate and the epidermis can be normal or show acanthosis, with or without papillomatosis (6-9).

Dermoscopic examination of the papular perioral lesions revealed a unique vascular pattern, with short tortuous vessels, although symmetrical and diffuse, in each flesh-colored lobule or the same tortuous vessels in the periphery of each lobule with the absence of thrombosed vessels, typically found in lesions of viral etiology.

The hyperchromic lesions described in Goltz syndrome have a linear aspect, following Blaschko's lines. Brownish lesions, resembling lentigines, located over hypochromic areas and in the periphery of atrophic areas, without following Blaschko's lines, were not clearly referred to in first publications (10-12). Only two reports of similar lesions were found in the literature, all appearing progressively during adolescence (8,13).

According to Kanitakis *et al*. (8), the histopathology of these brownish lesions reveals club-shaped acanthosis and melanotic hyperpigmentation in the basal layer caused by increased melanocytic activity due to strong expression of the enzyme tyrosinase, demonstrated by the reaction with monoclonal antibody HMB 45. There is a discrete inflammatory infiltrate in the papillary dermis. These findings and their untypical location in the periphery of atrophic areas suggest that epidermal melanocytes are being stimulated, resulting in an increased melanin production. The precise mechanism how this occurs, however, is not well established and possibly the inflammatory dermal infiltrate has some influence (13). In other diseases with hypochromia or atrophy, the pattern of lentigo-like lesion in the periphery was also evidenced, as in achromic segmental nevus, hypomelanosis of Ito, Galli-Galli disease and dyschromatosis universalis hereditaria (14-16). Among some attributed causes, exposure to sunlight, treatment with narrow band UVB and somatic mosaicism should be mentioned (14,17,18).

Dermoscopy of these lentigo-like lesions showed a linear reticular pattern suggestive of benign melanocytic lesion. The presence of structures resembling streaks suggests a similar aspect to recurrent melanocytic lesions over atrophic or cicatricial areas (19,20). However, these are lighter color, broadened and discontinuous structures that do not show the classic honeycomb aspect, but resemble other descriptions of lentigines found in the literature.

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It is, thus, a rare syndrome with cutaneous lesions eligible for dermoscopic evaluation and histopathologic correlation. Additionally, the absence of criteria of malignancy in the pigmented lesions permits the dermoscopy follow up, without the need of surgical removal.

CONCLUSION

No reports of dermoscopy of the specific lesions of Goltz syndrome were found in the literature available. Observation of other cases will be necessary to decide if it is possible to confirm that there is a dermoscopic pattern of perioral papilloma and lentigo-like lesions of focal dermal hypoplasia or Goltz syndrome. So far, although dermoscopy is not a valid tool to improve the diagnosis of this syndrome, it may be useful in the follow up of pigmented lesions.

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