LICHEN SPINULOSUS IN A PATIENT WITH WILSON'S DISEASE

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Abbreviations **LS** = lichen spinulosus; **WD** = Wilson's disease.

Case report. A 12-year-old girl born to non-consanguineous parents presented to the dermatology clinic with white, itchy, millimetric follicular papules on her arms and legs, which had persisted for 6 months (Fig. 1). The patient had been diagnosed two years earlier with Wilson's disease – confirmed by genetic analysis which showed the mutation C2363 C>T(p.Thr788Ile) of the gene *ATP7B*, classified as likely pathogenic – and was taking D-penicillamine (20 mg/kg/day, divided into two doses) and a low-copper, high-zinc diet. The patient's family members did not present similar lesions. She had no eczematous lesions or other findings that would support the diagnosis of atopic dermatitis.

Punch biopsy revealed orthokeratosis in the epidermis, dilated hair follicles with keratotic plugs, spongiosis around the hair follicles, dermal vascular proliferation, and perifollicular lymphocytic and histiocytic infiltration (Fig. 2). Clinical and histopathological findings led to the diagnosis of lichen spinulosus (LS). Laboratory findings revealed a decreased ceruloplasmin level – 9 mg/dL (nr 20-40) – and a decreased beta-carotene level – 102 mg/dL (nr 150-1250) – , while other tests, including copper, 24-hour urinary copper excretion, liver enzymes, and allergy tests, were normal. Beta-carotene supplementation of 8 mg once daily for 2 months was started, after which the symptoms resolved. Both gastroenterological and dermatological outpatient follow-ups were performed, and no recurrence was observed in the following 2 years. The patient continues to consume a restricted diet, rich in beta-carotene and zinc, and low in protein and copper.

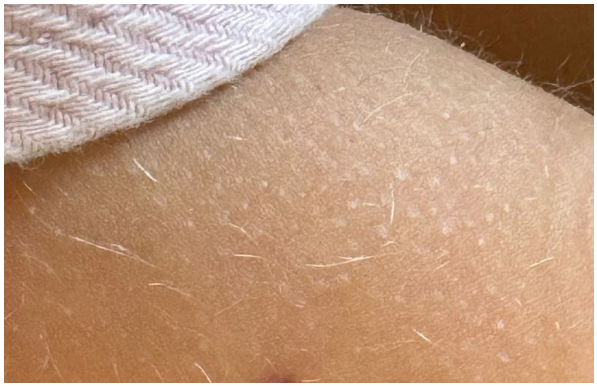

Fig. 1

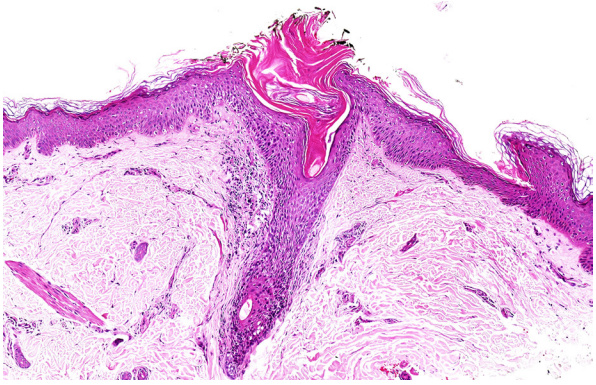

Fig. 2

Fig. 1, 2: Lichen spinulosus: white follicular papules (Fig. 1). Histological examination (H &E, x40) shows dilated follicles with keratotic plugs and perifollicular lymphocytic infiltrate.

Discussion. Lichen spinulosus (LS) is a rare skin disorder that occurs mainly in children and young adults and is characterized by round or oval, skin-colored, folliculocentric papules on the extensor surfaces of the limbs and chest (1). However, its pathogenesis remains unclear (2).

Wilson's disease (WD) is an autosomal recessive disorder caused by a biallelic mutation in the *ATP7B* gene encoding the ATPase-7B protein (adenosine triphosphatase -7B), which prevents the hepatic elimination of copper, disabling the main route of elimination of Cu from the body. Accumulation of copper leads to the formation of superoxides and hydrogen peroxides. In the presence of the superoxide anion radical (O_2^-) or other reducing substances, the copper ion Cu^{2+} can be reduced to Cu^+, which is able to catalyze the formation of hydroxyl radicals (.OH) from hydrogen peroxide (H_2O_2) via the Fenton reaction. Oxidative stress resulting from increased production of free radicals together with deficiencies in antioxidant defense may play a central role in WD (3). Alpha-tocopherol, beta-carotene, ascorbate, and glutathione are members of the nonenzymatic antioxidant defense. Previous studies have reported decreased beta-carotene and ascorbate concentrations and abundance of lipid peroxidation markers in patients with WD (4).

Xerosis and keratosis follicularis are common skin conditions in children, sometimes associated with vitamin A deficiency (5). Because of the similarity between keratosis follicularis and LS, we measured the patient's beta-carotene level and found it to be low. Her symptoms were controlled with supplements rich in beta-carotene and zinc. The latter was prescribed for its antioxidant effects and its competition with copper.

This case illustrates the potential link between LS and WD. Although further studies are needed to confirm this association, it is important to consider the role of vitamin deficiency and oxidative stress in dermatological manifestations related to WD.

Conclusion. The current case was presented to inform the dermatologist and pediatrician about the possible link between Wilson's disease and lichen spinulosus.

Conflicts of interest

The authors declare that they have no conflicts of interest.

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