Confluent and Reticulated Papillomatosis Associated with 15q Tetrasomy Syndrome

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Chromosome 15q tetrasomy is in the heterogeneous group of extra-structurally abnormal chromosomes (1). The syndrome displays distinctive clinical findings represented by developmental delay, intellectual disability, epilepsy and autistic behaviour. Skin pigmentation has been observed in several cases (1). However, no previous reports have described the skin lesions in detail. We report here the first case of a patient with 15q partial tetrasomy syndrome who presented with skin pigmentation that was clinically and histologically consistent with confluent and reticulated papillomatosis (CRP).

CASE REPORT

A 22-year-old man presented to our outpatient clinic with a 1-year history of persistent brownish macules on the trunk. The patient had been diagnosed with 15q partial tetrasomy syndrome at 8 years of age by our hospital's paediatrics department. He has been developmentally delayed since birth and severely mentally retarded, with no speech and little social response. He had experienced frequent generalized seizures since 8 years of age, which were treated with clobazam and carbamazepine. On physical examination, asymptomatic reticulated brownish macules with slight scales were observed on his abdomen and back (Fig. 1a). The lesions did not involve axillae or other intertriginous areas. Pityrosporum was negative in scale by light microscopy using potassium hydroxide. On dermoscopic examination, a diffuse brown background pigment pattern was seen. Histological examination of a skin biopsy taken from the abdomen showed hyperkeratosis, papillomatosis and mild superficial perivascular lymphocytic infiltrate (Fig. 2). Treatments with topical steroid and topical antifungal agents were unsuccessful. Oral minocycline was started at 100 mg twice daily, and the cutaneous lesions disappeared within one month (Fig. 1b). Based on the clinical course and the histological findings, the case was finally diagnosed as CRP.

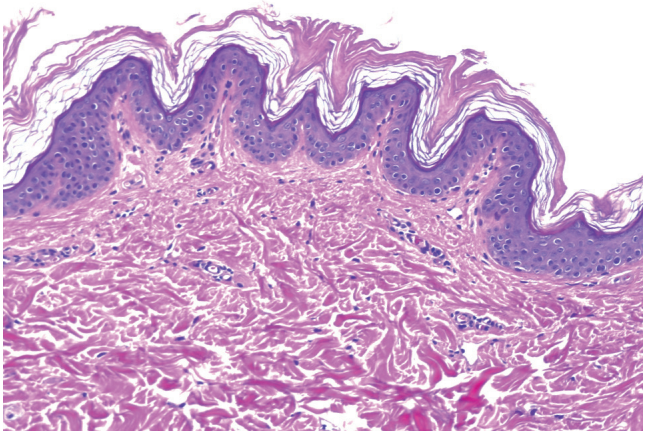

Fig. 2. On histopathological examination, undulating hyperkeratosis, papillomatosis and mild superficial perivascular lymphocytic infiltrate were seen (haematoxylin and eosin (H&E); original magnification ×100).

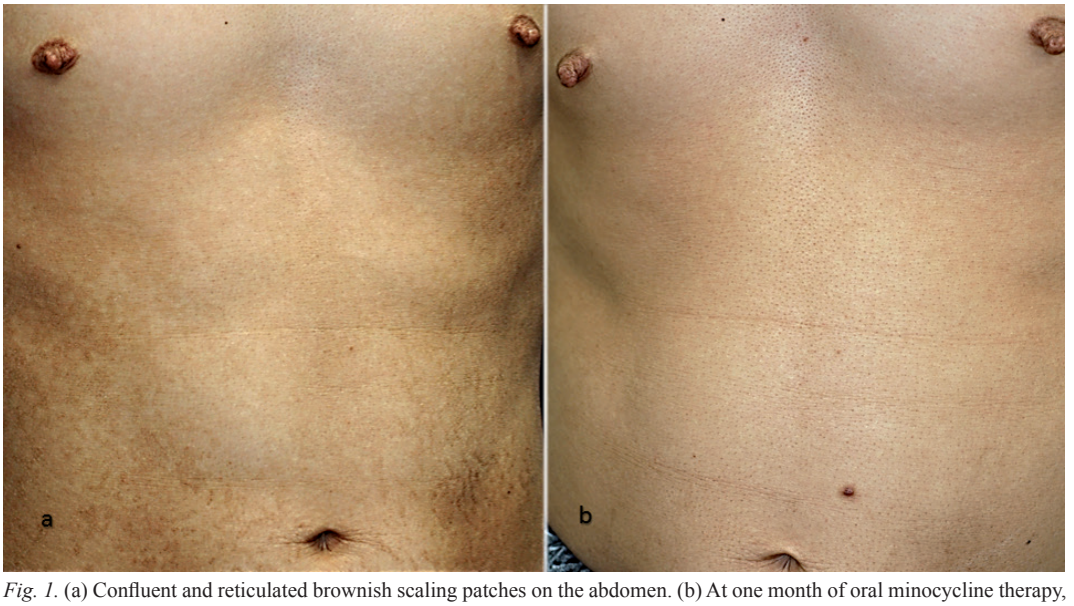

Fig. 1. (a) Confluent and reticulated brownish scaling patches on the abdomen. (b) At one month of oral minocycline therapy, the scaling has almost completely resolved.

Table I. *Review of cases with 15q tetrasomy syndrome with skin involvement*

Age, years/sex	Distribution	Eruption	Source
49/F	Face, trunk	Multiple pigmented naevi	Wisniewski et al., 1979 (2)
12/F	Trunk	Hyperpigmented area	Maraschio et al., 1981 (3)
10/M	Thighs	Patchy pigmentation	Maraschio et al., 1981 (3)
12/M	Back	Increased and reduced pigmentation	Crolla et al., 1995 (4)
15/M	Neck, trunk, legs	Multiple areas of whorl-like hypopigmentation	Battaglia et al., 1997 (5)
8 months/F	Trunk, legs	Diffuse hypo- and hyperpigmentation	Qumsiyeh et al., 2003 (6)
22/M	Trunk, back	Reticulated brownish macules	Present case

DISCUSSION

15q partial tetrasomy syndrome, also called isodicentric 15q or inverted duplication 15, is a rare chromosome abnormality, with only about 160 cases reported (1). A review of the English literature found that only 6 of the patients who were diagnosed with this syndrome had skin involvement (2–6) (Table I). Akahoshi et al. (7) reported a woman with duplication of 15q11.2-q14 who exhibited striking generalized skin hyperpigmentation. They estimated that her pigmentation was associated with duplication of the P gene, encoding a melanosomal protein. According to Jimbow et al. (8), ultrastructurally, the hyperpigmentation of CRP is due to an increased number of melanosomes in the hyperkeratotic horny layer. Detailed histological descriptions of the eruptions of the patients with 15q tetrasomy syndrome were unavailable because a biopsy was not performed on any of the cases. However, some eruptions of the cases involved regions that were consistent with areas predominantly affected in CRP. Davis et al. (9) stated that CRP was not fully recognized by physicians or even by dermatologists: only 39 patients in 32 years at the Mayo Clinic have been precisely evaluated. Thus, there is the possibility that the skin hyperpigmentation associated with 15q tetrasomy syndrome is being diagnosed as CRP. In addition, taking into account the high effectiveness of minocycline therapy in the majority of patients with CRP (9), some eruptions related to the syndrome would be treatable.

This is the first case report to detail the eruptions associated with 15q tetrasomy syndrome. It is important to be aware of CRP as a possible cause of skin pigmentation in 15q tetrasomy syndrome, because CRP is a treatable condition.

The authors declare no conflicts of interest.

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