H.K. Dermatol. Venereol. Bull. (2004) **12**, 106-107

Dermato-venereological Quiz

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This 17-year-old man presented at the age of four years with easy splitting of the nails which subsequently became dystrophic (Figure 1). Skin rash appeared on sun-exposed areas a few years later. On examination, the skin on the chest was atrophic with both hypo- and hyper-pigmentation and telangiectasia (Figure 2). Subsequently, oral lesions also appeared (Figure 3). He also suffered from dental caries, dysphagia and multiple infections (recurrent oral candidiasis, staphylococcal infections). Investigations showed a pancytopaenia with haemoglobin level of 9.0 g/dl, total white cell count $3.0x10^9$/L and platelet count $90x10^9$/L.

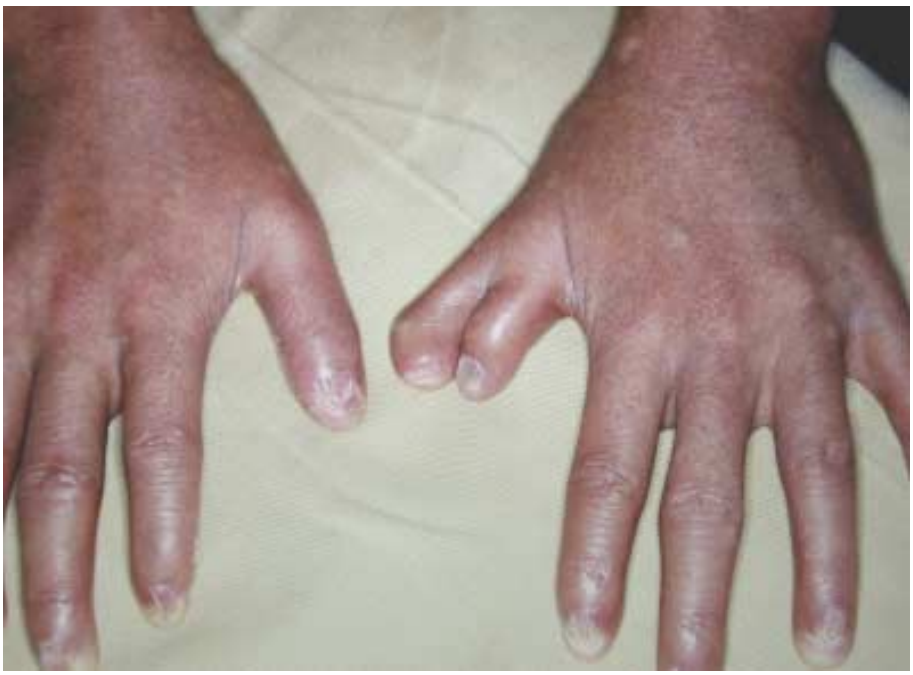

Figure 1.

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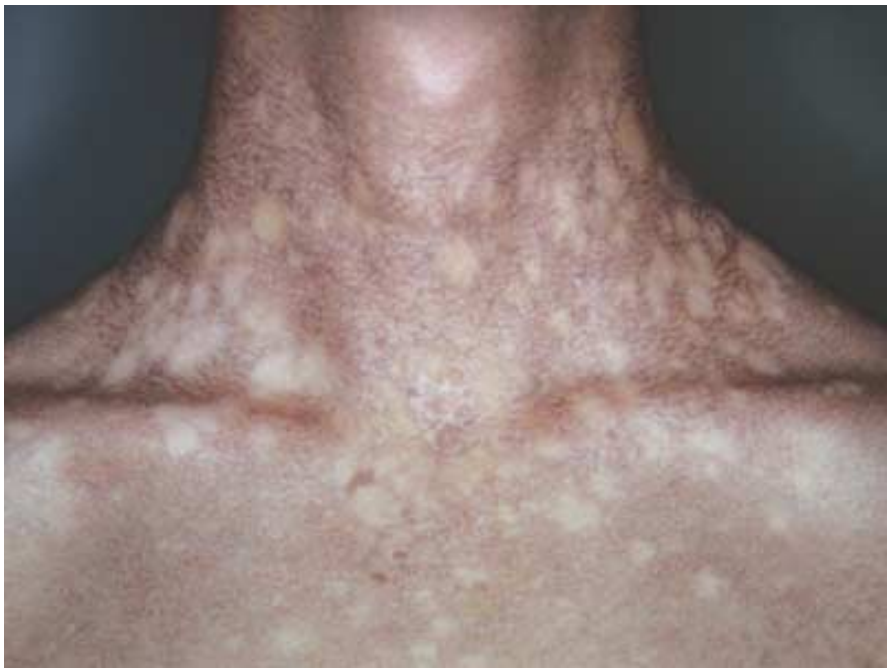

Figure 2.

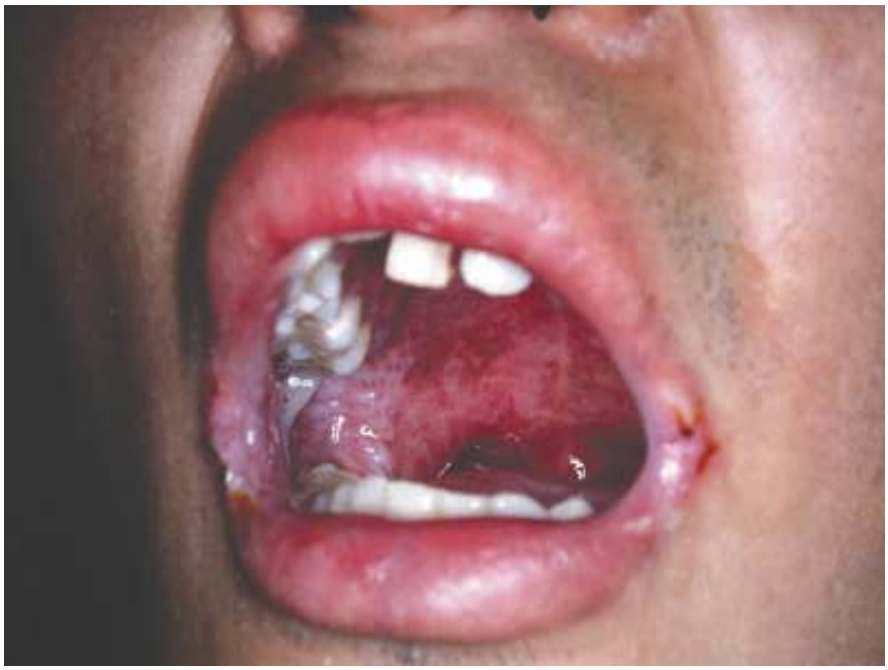

Figure 3.

Questions

1. What is the most likely diagnosis?
2. What are the complications of this condition?
3. What is the pathogenesis of this condition?
4. What treatments are available?

(Answers on page 115)

Answers to Dermato-venereological Quiz on pages 106-107

1. The most likely diagnosis is **dyskeratosis congenita**. This is a rare, inherited multi-system disease, consisting of nail dystrophy, poikiloderma-like rash, leukoplakia, bone-marrow failure and an increased incidence of malignancy. The condition can be inherited in an autosomal dominant, autosomal recessive, or X-linked fashion. Nail dystrophy usually presents at three to five years of age. Pigmentary changes appear later, typically affecting sun-exposed areas, leading to a poikiloderma-like appearance. There may be hyperhidrosis, and hyperkeratosis of the palms and soles. Leukoplakia most commonly affects the oral mucosa, and may also be found in the gastrointestinal tract, urethra, glans penis, rectum and vagina.

2. There is an increased incidence of malignancy, often affecting areas of leukoplakia in the third or fourth decade. Squamous cell carcinoma of the mouth, cervix, oesophagus and vagina are the most commonly reported neoplasms. Other reported neoplasms include adenocarcinoma of the pancreas and Hodgkin's disease. Bone marrow failure with refractory anaemia or pancytopaenia may also occur. In addition, impaired cell-mediated immunity and immunoglobulin deficiency may also be present.

3. Although the exact pathogenesis is unknown, it is known that the DKC1 gene at Xq28 is affected in the X-linked forms and mutations in hTR, the RNA component of telomerase, are present in the autosomal dominant form. The affected gene in the autosomal recessive form is unknown. These predispose cells to chromosomal rearrangements.

4. Bone-marrow transplantation may be suitable for the associated aplastic anaemia. Retinoids have been used to reduce leukoplakia and granulocyte-colony stimulating factor may improve haematological parameters.