# AN ATYPICAL TRIAD

G. ABSIL[1], AF. NIKKELS[1], B. DEZFOULIAN[1]
[1]Department of Dermatology, University Hospital of Liège, B-4000, Belgium

## CASE REPORT

A man in his fifties was referred to the dermatology department for evaluation of skin and nail lesions that had appeared during early childhood. The lesions were completely asymptomatic and had slowly progressed over the years. Familial history was negative for such clinical features. His medical history revealed high blood pressure, hypothyroidism, non-alcoholic steatohepatitis and a 10-year history of severe idiopathic pulmonary fibrosis. Routine laboratory testing, including complete blood count, were within normal ranges.

**Physical examination** showed a lacy, reticulated hyperpigmentation of the trunk, neck and upper extremities (*Fig. 1,2*) associated with bilateral epiphoras, areas of leukoplakia of the dorsal aspect of the tongue (*Fig. 3*) and complete atrophy of the nails (*Fig. 4*).

Based on these findings, a clinical diagnosis of dyskeratosis congenita (DC) was made. **Genetic testing** revealed a DKC1 gene mutation, not previously described, compatible with an X-linked inheritance pattern of DC.

Unfortunately, a few years later, the patient died shortly after diagnosis of a metastatic adenocarcinoma of the stomach.

His asymptomatic daughter carries the same mutation. She refused genetic testing for her 10-year-old asymptomatic son but performed prenatal testing for a second pregnancy.

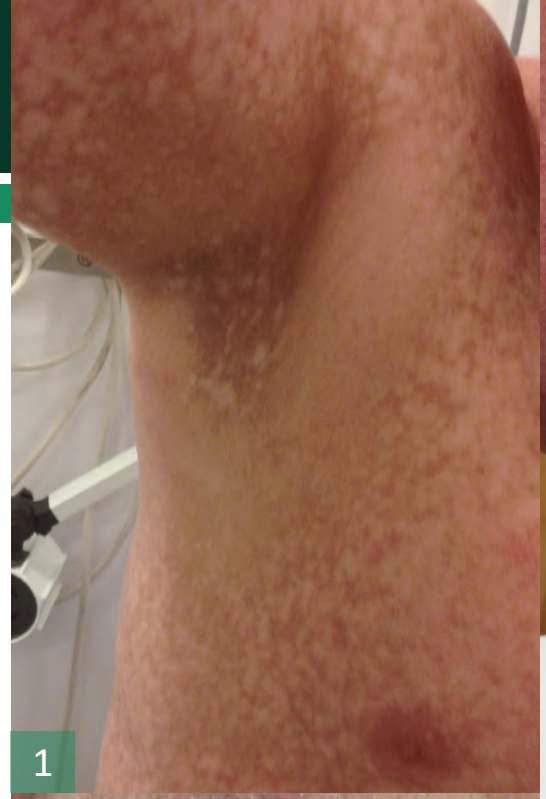
## DISCUSSION

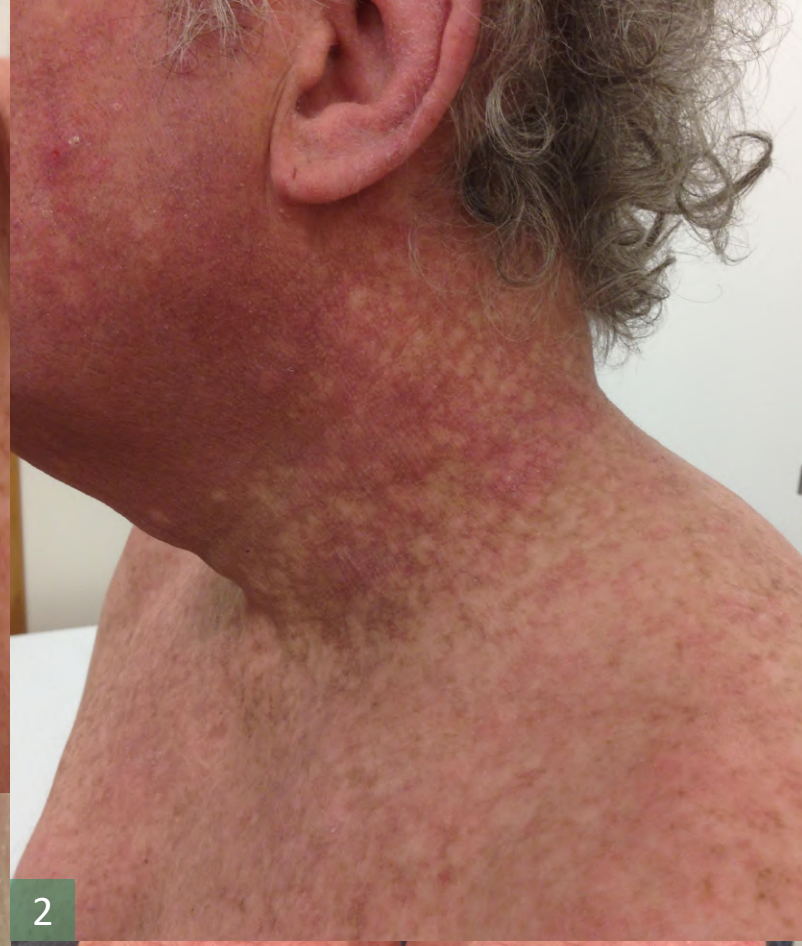
***Dyskeratosis congenita***, first described as Zinsser-Cole-Engman syndrome, is a very rare genodermatosis due to defective telomere maintenance. It is usually inherited in an x-linked pattern with a mutation in *DKC1*, but autosomal dominant and autosomal recessive patterns have also been described.[1]

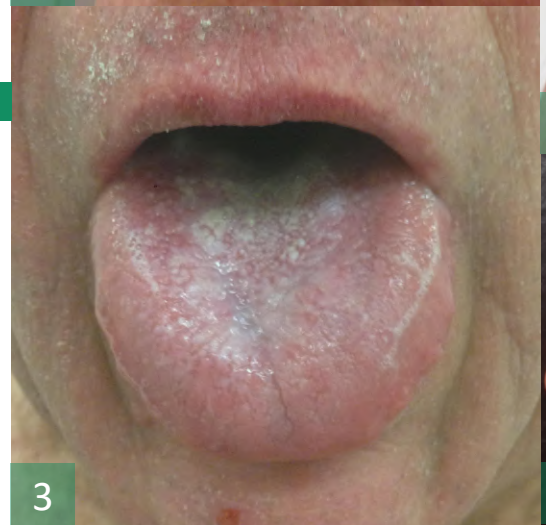
Symptoms usually first appear during early childhood with a ***triad*** of reticulated hyperpigmentation of the neck, trunk and extremities, nail dystrophy and leukoplakia of the oral mucosa. Epiphora, or excessive tearing, is another common feature due to a blockage of the tear ducts. ***Less common skin findings*** including hypopigmentation, poikiloderma, premature hair greying, patches of alopecia and palmoplantar hyperkeratosis may also be found.[2]

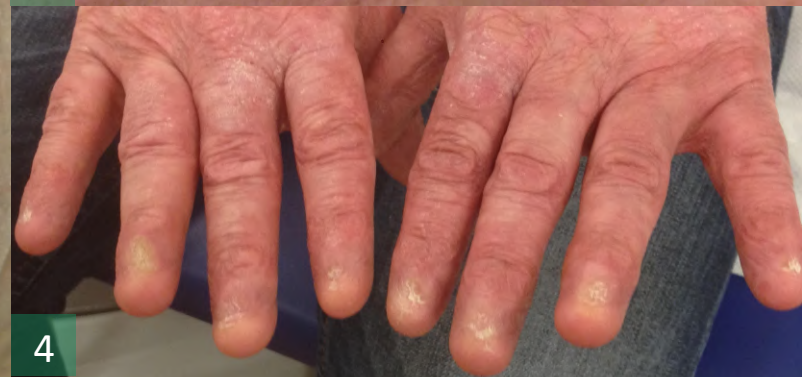
DC patients carry a ***high risk for systemic diseases***, particularly bone marrow failure (BMF) with pancytopenia, pulmonary fibrosis, hepatic complications (mainly cirrhosis) and ***various types of cancers*** such as squamous cell carcinomas of the tongue, Hodgkin's lymphoma, or adenocarcinomas of the gastrointestinal tract.[3]

There is ***no treatment for the skin manifestations***. Close monitoring is mandatory to diagnose complications as soon as possible and to allow early treatment.


1


2


3


4

References
1. Mason PJ, Bessler M. The genetics of dyskeratosis congenita: a literature review. *J Dtsch Dermatol Ges*. 2020;18(9):943-967.
2. Alsabbagh MM. Dyskeratosis congenita: a literature review. *J Dtsch Dermatol Ges.* 2020;18(9):943-967.
3. Garcia MSF, Teruya-Feldstein J. The diagnosis of dyskeratosis congenita: a review. *J Blood Med*. 2014;21(5):157-67.