

Dyskeratosis Congenita: Report of a New Case and Review of the Turkish Literature[¶]

DİSKERATOZİS KONJENİTA: BİR VAKA SUNUMU VE TÜRK LİTERATÜRÜNÜN İNCELENMESİ

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Summary

Purpose: Dyskeratosis Congenita is a rare inherited disorder with abnormalities of the skin, mucous membranes and bone marrow. In this case, a 20 years old male patient with Dyskeratosis Congenita was presented and oral findings were discussed.

Case Report: Following the clinical and radiological examination of the patient who has dermatological findings associated with Dyskeratosis Congenita, atrophic tongue mucosa, inflamed gingiva, short-blunted lower incisor teeth are determined.

Conclusion: Dyskeratosis Congenita is a rare disease, systemic and oral symptoms of this disease should be paid attention in dentistry. Although, the prognosis is good in this disease, the unexpected changes in hematological values and mucocutaneous malignant changes should be kept in the mind.

Key Words: Dyskeratosis Congenita, Oral findings

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Özet

Amaç: Diskeratozis Konjenita deri, müköz membran ve kemik iliğinde anormalliklerle seyreden, genetik geçişli, seyrek görülen bir hastalıktır. Bu makalede Diskeratozis Konjenitalı 20 yaşında erkek hasta sunulmuş ve ağız bulguları tartışılmıştır.

Vaka Sunumu: Diskeratozis Konjenita hastalığı ile uyumlu dermatolojik bulguları olan hastada ağızın klinik ve radyolojik muayenesi sonucunda atrofik dil mukozası, enflamasyonlu dişetleri, kısa-künt köklü alt kesici dişler tespit edildi.

Sonuç: Diskeratozis Konjenita nadir görülen bir hastalık olmasına karşın, dişhekimliğinde bu hastalığın genel ve oral bulguları göz önünde bulundurulmalıdır. Prognozu iyi hastalarda bile her an hematolojik olarak kötüye gidişler ve mukokütanöz malign değişikliklerin olabileceği bilinmelidir.

Anahtar Kelimeler: Diskeratozis Konjenita, Oral bulgular

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Dyskeratosis Congenita (DC) is a rare congenital and familial disorder characterized by the triad of reticular pigmentation of the skin, dystrophic nails and leukoplakia of the mucous membranes and it is often associated with severe pancytopenia. The mode of genetic transmission is unknown, but it mainly affects males (1-5).

Although the principal clinical findings of

these conditions are described as a triad, there is a high association of hematopoietic abnormalities with this syndrome. The earliest sign may be either anemia or thrombocytopenia, but ultimate progression to pancytopenia is usual. The skin may become atrophic and telangiectatic and the face appear red. The palms and soles may be hypertrophic (3,5-7).

There are several studies about oral and dental findings of DC. Destruction of alveolar margins, gingival inflammation, severe alveolar bone loss resembling juvenile periodontitis, hypo mineralized teeth, taurodontizm, short-blunted roots, thin enamel structure, tendency to decay, brown pigmentation, tooth loss and gingival bleeding were reported in these studies (5,6,8).

DC usually starts between the ages of 5-12 years with dermatologic symptoms. Pigmentation of the skin and dystrophy and atrophy of the nails are the most frequent manifestations. Leukoplakia may occur on various mucous membranes and become malignant (3,7,8).

Patients with DC usually die by reason of pancytopenia or opportunistic infections or co-etaneous and mucosal carcinomas, which develop, in young affected adults. Most patients usually die between age of 10 and 30. However, some patients have only nail dystrophy and pigmentation otherwise these patients survive as normal (1,4,9).

Case Report

A 20 years old man diagnosed as Dyskeratosis Congenita (DC) before was followed and treated by Department of Dermatology of Gülhane Military Medical Academy, was admitted by reason of edema on left side of his face and toothache to Department of Oral Diagnosis and Radiology. The first systemic complaint of the patient was determined on his nail when he was in 4 ages. Later, other findings were appeared. His mother and his father are relative, but nobody was determined as a carrier in the family tree.

Partly atrophic white and telangeactasic areas and several lesions, which are characterized with red-brown poikilodermic pigmentation, are localized on the neck, on the body and on the dorsum, have been established in dermatological examination. The skin of the patient was thin, dry and wrinkle in this areas. Patient had slightly palmar-plantar hyperkeratosis and syndactylia on his toe,



Figure 1. Slightly hyperkeratosis and syndactylia, partial nail loss on the fingers.



Figure 2. Partial and total nail loss, evident syndactylia on the toes.

there was slightly syndactylia on his fingers (Figure 1,2). It was not established fingerprint on overall finger of his hands. It was observed that patient have longitudinal pitted with split formation on his fingernails and on his toenails, subungual hyperkeratosis together with rudimentary nail and totally nail loss on his toe.

It was observed that gingival recession and gingival hyperemia in oral examination of the patient (Figure 3). It was not fixed leukoplakia in oral cavity of the patient. Oral hygiene was found

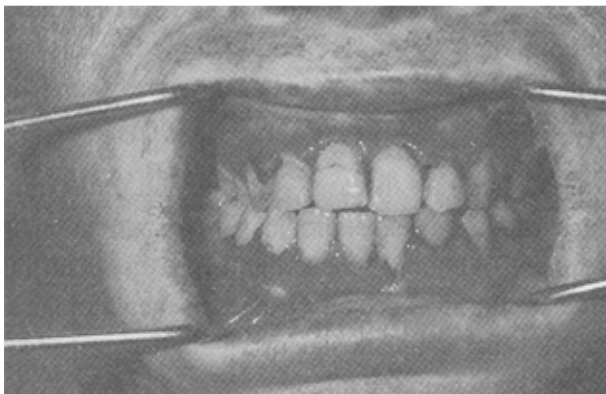


Figure 3. Appearance of tooth and gingival.

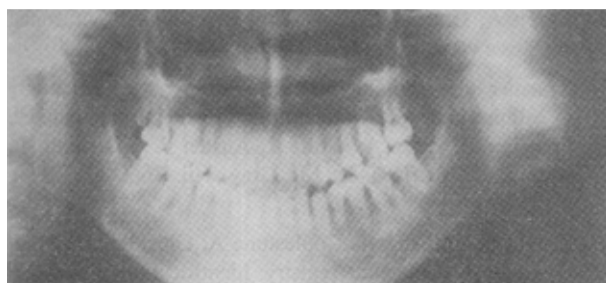


Figure 4. Radiographic appearance short-blunted roots of mandibular incisors.

insufficient. Detected plaque and calculus accumulation were abundant on the teeth. There was acute apical parodontitis because of the profound caries in maxillary first molar. Generally, inflammation and bleeding were observed in the maxillary and mandible gingiva. The short and blunted roots of mandible incisors were determined in panoramic radiography (Figure 4). Patient couldn't completely close on his lips, due to atrophic formation on his skin. There was ectropion on his low eyelid (Figure 5).

Medical investigation includes complete blood count, complete urine test, liver and kidney function tests, ANA, Anti DNA, Anti Scl 70, and

Anti Sentromer were found negative. Other medical investigation includes immunoelectrophoresis, plasma and metabolic porfirin crops in urine; abdominal ultrasonography, esophagiagastroduodenoscopy and rectosigmoidoscopy were with in normal range. Wrist radiography was in accordance with 16 age, but radiography of the hand and the toe has different degree bone deformity. No pathology detected in hematological examination.

It was established that atrophy on the epidermis, smooth in the retia, telangiectatic vascular formation in the papiller dermis taking from extensor and flexor areas of forearm was observed. This appearance has found to be associated with the characteristically findings of poikiloderma.

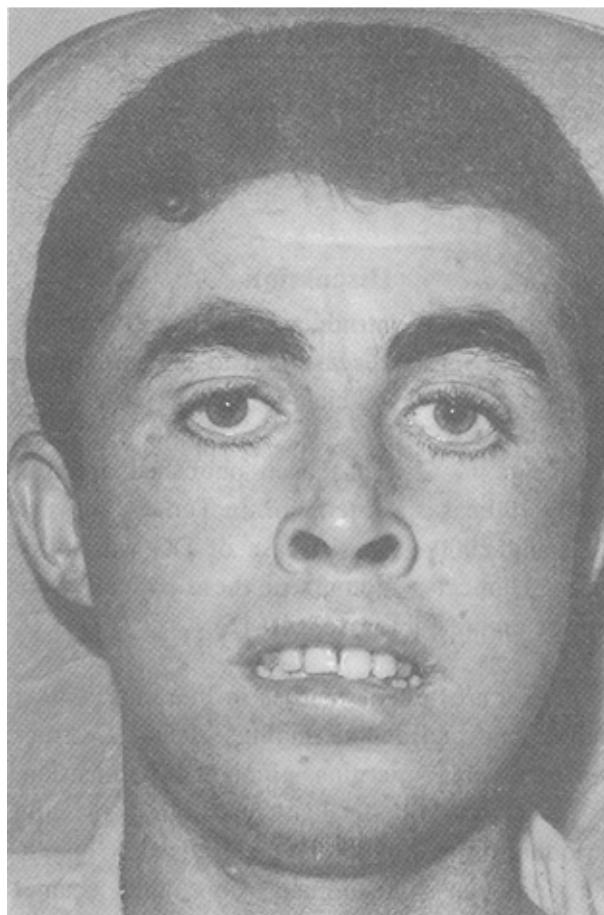


Figure 5. Ectropion on the eyelid and dryness on the lips.

Table 1. Clinical and laboratory findings in literature cases

Skin Findings	Poikilodermia Palma-plantar hiperhidrosis Palma-plantar keratodermia
Oral Findings	Leukoplakia Gingival recessions Dental caries Gingival bleeding Periodontitis
Nail Findings	Dystrophic nail Longitudinal lines Subungual hyperkeratosis Discoloration Rudimentary nail
Other Findings	Splenomegaly Ectropion Narrowness in meatus of penis
Laboratory Findings	Anemia Leukopenia Increases in immunoglobulins

Discussion

This case is introduced because DC which seem to be rarely disease and to put on display both oral and gingival changes.

Up to now, it has been published approximately 100 cases in literature. In Turkey, between 1982 and 2000 years 10 case of DC was introduced. Clinical symptoms of these patients were showed on the Table 1 (6,8,10-16).

There are several studies in literature, which include oral and dental findings of DC. Destruction of alveolar margins, gingival inflammation, severe alveolar bone loss resembling juvenile periodontitis, hypo mineralized teeth, taurodontism, short-blunted roots, thin enamel structure, lichenoid lesion and leukoplakia on tongue and buccal mucosa were reported (3,6-8). Even though oral leukoplakia was not found in

our case, the mucosa of the tongue was atrophic, smooth and shining. Short-blunted roots of mandible incisors in our case were found in accordance with the literature.

DC is a rare disease, systemic and oral symptoms of this disease should be paid attention in dentistry. Although, the prognosis is good in this disease, the unexpected changes in hematological values and mucocutaneous malignant changes should be kept in the mind.

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