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Case Report Open Access

Dyskeratosis congenita: A report of two cases

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Abstract

Dyskeratosis congenita (DC) is the first genetic syndrome identified among telomeropathies. Its classical phenotype is characterized by mucocutaneous abnormalities, atrophy and pigmentation of the skin, oral leukoplakia, and nail dystrophy. Bone marrow failure and susceptibility to malignancy are rare, and DC is mostly an X-linked recessive, serious, multisystemic disease. This study aims to increase dentists' awareness of this genetic disease, accompanied by oral findings. Two cases of DC are reported in this paper, along with a short review of the literature. Oral findings of the disease include dental caries, gingival recession, short roots, gingival bleeding, severe mobility of the teeth, and alveolar bone loss. DC is clinically seen mostly in males aged 5 to 12. This case report examines dermatological and oral findings consistent with the disease of two brothers diagnosed with DC who applied to the Gülhane Health Sciences University, Faculty of Dentistry, Pediatric Dentistry Department for dental caries complaints. DC is a rare disease, and dentists should pay attention to its systemic and oral symptoms during their examinations. Despite a positive prognosis of this disease, it is important to take into account any unexpected changes in hematological values and mucocutaneous malignant alterations.

Keywords: Dyskeratosis congenita, oral findings, oral health, leukoplakia, keratosis

Introduction

Dyskeratosis congenita (DC) is a rare inherited disorder that affects less than one individual per million (1). The most prevalent mode of inheritance is the X-linked recessive variant, which primarily affects males and is caused by mutation of the DKC1 gene at the Xq28 site, even though the mode of inheritance is unclear (2). Dyskeratosis congenita (DC) is the first genetic syndrome identified among telomeropathies. The classic phenotype is characterized by mucocutaneous abnormalities, atrophy and pigmentation of the skin, nail dystrophy, and oral leukoplakia. Bone marrow failure is rare. DC is mostly an X-linked recessive, serious, multisystemic disease. Patients with DC have a high risk of developing oropharyngeal squamous cell carcinoma, and the cause of early mortality in these patients is often severe pancytopenia (3). The age of onset, symptoms, and severity of symptoms vary in patients with DC. Even if patients have the same gene mutation, the time and form of the disease are variable, so it is sometimes difficult to make an accurate diagnosis (4).

Oral and dental symptoms of DC include increased and extensive dental caries, short roots, gingival recession, gingival bleeding and inflammation with oedema, aggressive periodontitis, alveolar bone loss resembling juvenile periodontitis, smooth atrophic tongue mucosa, leukoplakia and lichen planus, hypodontia, severe mobility in the teeth, early tooth loss, thin enamel, hypocalcification, intraoral brown pigmentation, taurodontism, and blunted roots (1, 5, 6). DC is clinically seen mostly in 5–12-year-old males (7).

Gastrointestinal involvement is seen in 8% of patients. In addition, esophageal diverticulum or stricture, gastroduodenitis, duodenal ulcer, and chronic diarrhea have been reported in relation to the gastrointestinal tract (8, 9).

The oral and dental results of two DC cases are shown in this paper. This study aims to increase dentists' awareness of this genetic disease, accompanied by oral findings.

Case Reports

Case 1

The patient was a 15-year-old boy who applied to Health Sciences University, Gülhane Faculty of Dentistry, Department of Pediatric Dentistry, due to dental caries. The patient was previously diagnosed with DC. The first systemic complaints of the patient appeared on his nails at the age of 4–5 years. The patient's parents noticed that the child's development was insufficient at the age of 6–7 and applied to the medical doctor. A definitive diagnosis was made with blood tests, genetic tests, and systemic findings (complete blood count, complete urine test, liver and kidney function tests, ANA, Anti-DNA, Anti-Scl-70 and Anti-Centromere, urine immunoelectrophoresis, plasma and metabolic porphyrin crops, hematological examination, etc.).

In the head-neck and hand examination, various lesions characterized by partially telangiectatic areas and red poikilodermic pigmentation were localized around the mouth, under the eyes, and on the hands (Fig. 1). According to the anamnesis taken from the parent, the same lesions were on his feet. Longitudinal pitting, cleft formation, and subungual hyperkeratosis were observed in the patient's nails (Fig. 2).

In the intraoral examination of the patient, gingival hyperemia, primary tooth root residue, dental caries, and closure disorder were detected. In addition, the patient's oral hygiene was found to be inadequate (Fig. 3).

Figure 1. Frontal and lateral view of Case 1

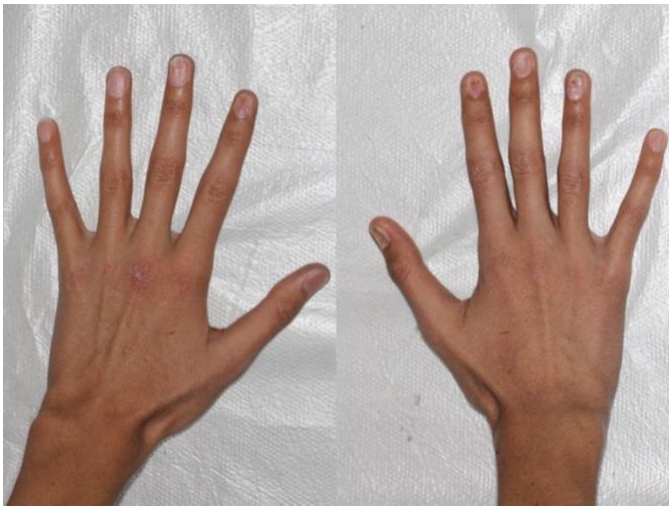

Figure 2. Physical appearance of Case 1 patient's left and right hands

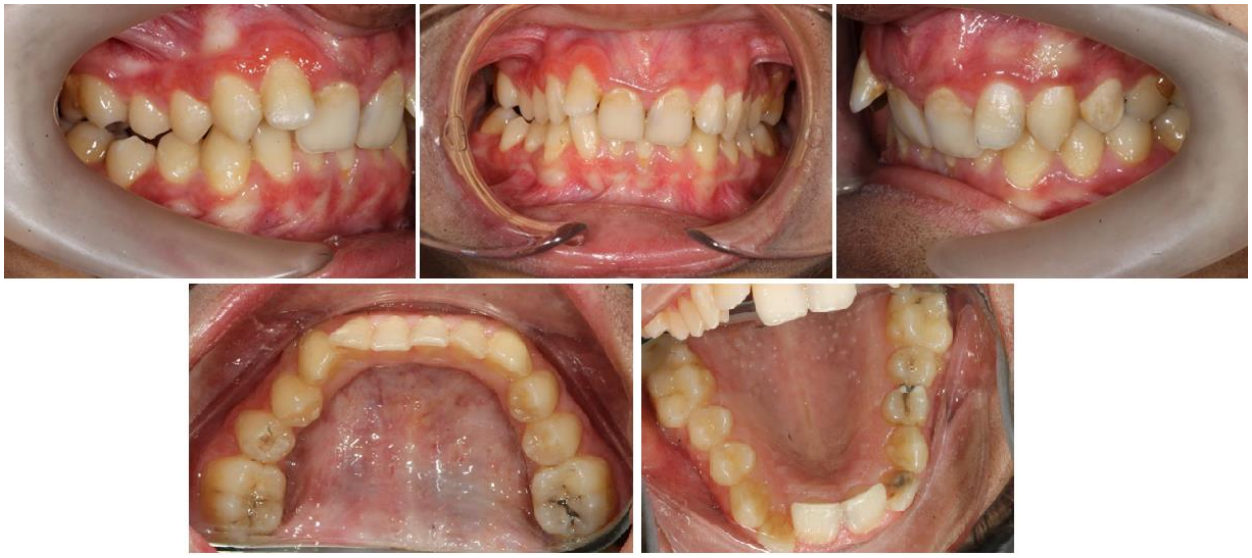

Figure 3. Intraoral view of Case 1

Case 2

The patient was a 10-year-old boy who applied to Health Sciences University, Gülhane Faculty of Dentistry, Department of Pediatric Dentistry, due to dental caries. The patient was previously diagnosed with DC. Since he is the brother of the previous patient, his diagnosis was made at the age of 4–5 years with systemic findings, blood tests, and necessary genetic tests (complete blood count, complete urine test, liver and kidney function tests, ANA, Anti-DNA, Anti-Scl-70, Anti-Centromere, urine immunoelectrophoresis, plasma and metabolic porphyrin crops, hematological examination, etc.).

In the head-neck and hand examination performed on our patient, various lesions characterized by partially telangiectatic areas and red poikilodermic pigmentation were localized around the mouth, under the eyes, and on the hands (Fig. 4). According to the anamnesis taken from the parent, the same lesions were on his feet. Longitudinal pitting, cleft formation, and subungual hyperkeratosis were observed in the patient's nails, as with his elder brother (Fig. 5).

In the intraoral examination of the patient, intense plaque accumulation, deep dentin caries in more than one permanent tooth, hyperemia in the gums, edema, and closure disorder were detected (Fig. 6).

According to the anamnesis we received from their parents, chronic diarrhea is observed in both boys, and they use medication to prevent diarrhea.

The necessary dental treatments of the patients continued, and they are under our follow-up.

The same disease was diagnosed in the other 5-year-old patient as his elder brother.

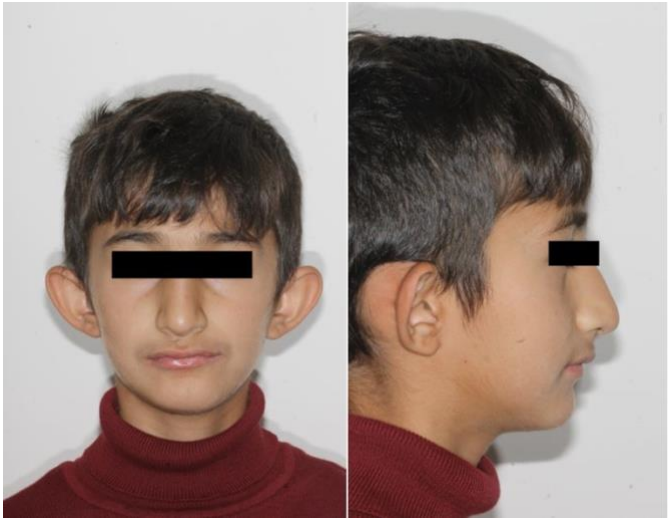

Figure 4. Frontal and lateral view of Case 2

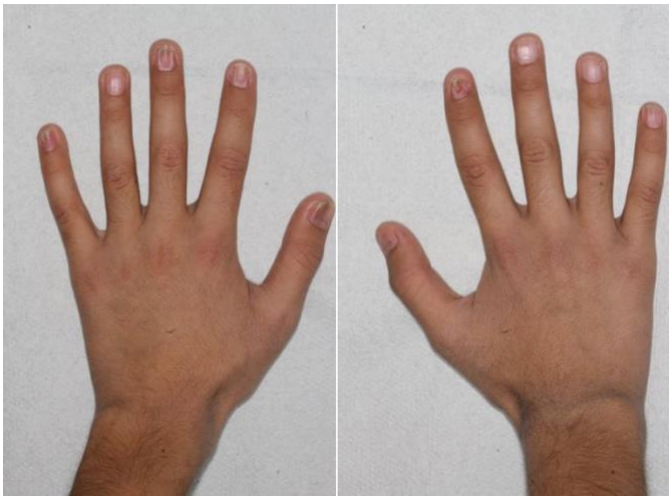

Figure 5. Physical appearance of Case 2 patient’s left and right hands

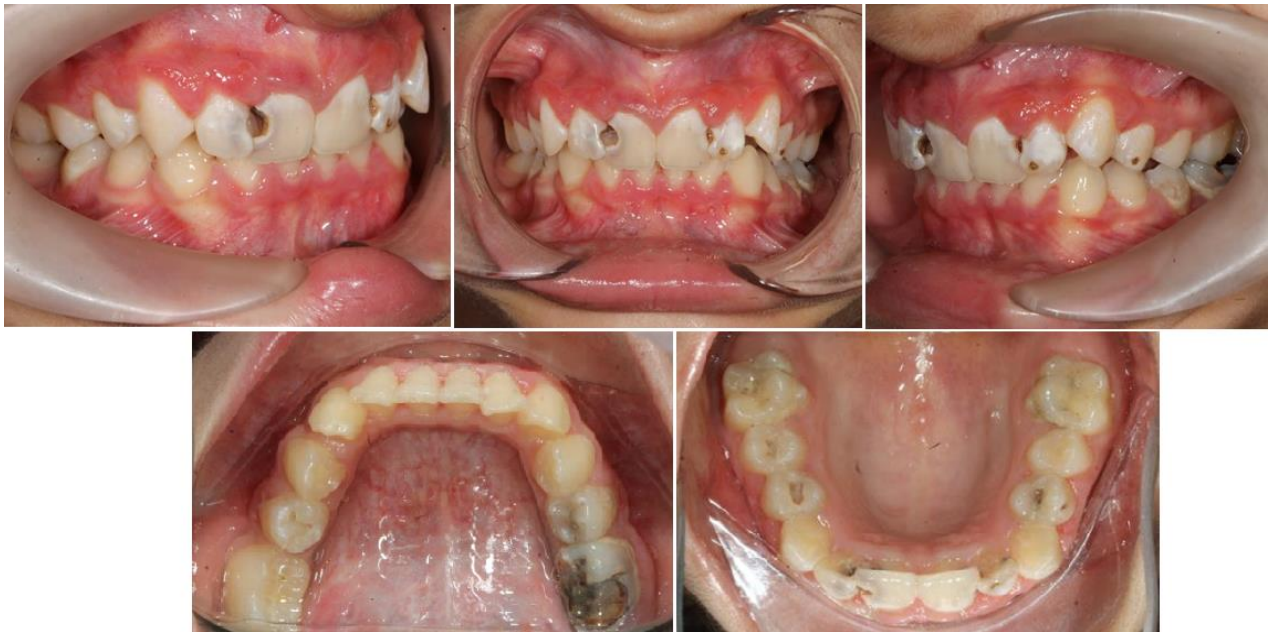

Figure 6. Intraoral view of Case 2

Discussion

Zinsser reported first description of Dyskeratosis congenita (DC) in 1906. Mucosal leukoplakia, nail dystrophy, and aberrant skin pigmentation are the hallmarks of the uncommon genetic disorder dyskeratosis congenita, also referred to as Zinsser-Engman-Cole syndrome (10, 11). The disease DC is lethal. In the keratotic white patches, the majority of individuals experience malignant transformation and aplastic anemia (12). In our cases, we determined longitudinal pitting, cleft development, and subungual hyperkeratosis in the patients' nails.

Similar dental characteristics, including hypodontia, delayed tooth eruption, short, blunt roots, increased tooth decay, gingival issues and bleeding, loss of alveolar bone, leukoplakia of the buccal mucosa, and irregular ulcers, were noted in Baran et al.'s study (13). A case of DC with several dental abnormalities, including tooth mobility and decay, gingival recession and bleeding, short-blunted roots, severe alveolar bone loss, and a leukokeratosis lesion on the buccal area, was also reported by Yavuzyılmaz et al. (5). Extensive caries lesions, deep dentin caries and plaque accumulation were determined in our cases.

In their study, Atkinson et al. (6) assessed 17 individuals with DC. Oral leukoplakia (found in 65% of the patients overall), a decreased root/crown ratio (75% of patients), and mild taurodontism (57% of patients with radiographs) were the most frequently seen oral abnormalities in their study.

Oral findings are typical with DC, which often appears between the ages of 5 and 12 (5, 11). Most of the cases that have been published involve males. The ratio of male to female is reportedly 13:1 (1). The ages of the patients in our cases were 10 and 15 years old. We have male patients as well.

Early childhood caries is a prevalent occurrence that affects 17% of DC patients (2). A few cases of oral and dental abnormalities have been documented; these include the following: extensive caries, lichen planus, hypocalcification, thin enamel, gingival inflammation with oedema, alveolar bone loss, periodontitis, and extensive caries. In our cases, we observed hyperemia in the gums, edema, gingival hyperemia, primary tooth root residue, dental caries, and closure disorder (2, 13).

Conclusion

The presented DC cases show a disease under control, displayed for demonstrations in both the mouth and gums.

There are various case reports in the literature, and in these cases, gingival inflammation, severe alveolar bone loss resembling juvenile periodontitis, hypomineralized tooth taurodontism, short blunt roots, thin enamel structure, lichenoid lesion, and the presence of leukoplakia in the tongue and buccal mucosa have been reported.

DC is a rare disease, and dentists should pay attention to the systemic and oral symptoms of this disease during their examinations. Although the prognosis is good for this disease, unexpected changes in hematological values and mucocutaneous malignant changes should be considered and not ignored.

Disclosures

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