Clinical findings, dental treatment, and improvement in quality of life for a child with Rothmund-Thomson syndrome

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Abstract

The purpose of this study was to report the clinical findings, dental treatment, and improvement in quality of life for a child with Rothmund-Thomson syndrome. The patient had alopecia, delayed speech, low weight and height, cholestasis, and iron deficiency anemia. Furthermore, there were carious lesions and darkened spots on all primary molars. Microdontia of a premolar was observed at the radiographic examination. The patient and family had no commitment to her oral health and dental treatment at first appointments. Oral hygiene instructions, composite restorations, endodontic treatments, teeth extractions, and stainless steel crown installations were performed. The patient was followed up for 7 years through the present due to other possible future clinical findings associated with the syndrome. An improvement in social aspects was observed after removal of toothache and improved esthetics. Such patients need continuous periodic services, which contributes to improving the quality of life in both buccal and general aspects.

Keywords: Behavior difficulties, dental treatment, quality of life, Rothmund-Thomson syndrome

Introduction

The Rothmund-Thomson syndrome (RTS) is an autosomal recessive disorder that manifests in childhood, usually with early skin lesions in the 1st year of life and can also be characterized by skin atrophy telangiectasia, hyper- and hypo-pigmentation, skeletal birth defects, short stature, saddle nose, premature aging, juvenile cataracts, and predisposition to carcinomas and osteogenic sarcomas.[1,2]

RTS is assigned to a mutation on the RecQL4 (8q24) gene, which codifies the RecQ DNA helicase protein, associated with mutation of chromosomic stability.[1] RTS is included in the National Organization for Rare Diseases database as one of the 1200 rarest diseases, so its exact prevalence is unknown, but there are approximately 300 cases described in the literature.[3]

There are a few reports in the literature of oral characteristics of RTS to date, but none regarding the impact of dental treatment on the life of patients with this syndrome.[4,5] The aim of this study was to report the clinical findings, dental treatment, and improvement in quality of life for a child with RTS.

Case Report

The patient began the dental treatment when she was 2-year-old (2009). In her clinical hospital records, we found cholestasis, iron deficiency anemia, spongiotic superficial dermatitis with secondary infection, alopecia, and saddle nose. Poikiloderma, speech delay, and low weight and height were also noticed. The patient is under treatment by genetic, dermatology, gastroenterology, urology, and speech therapy specialists.

From oral examination, it was possible to detect high caries risk and activity, with white spots and carious lesions in

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several teeth, as well as coronary darkened staining in all primary molars [Figure 1]. Furthermore, at late radiographic evaluation, microdontia of the maxillary left second premolar germ was observed.

Because of the diagnosis of immunosuppression, there was the need for antibiotic prophylaxis with amoxicillin 50 mg/kg 1 h before the clinical procedures, recommended in writing by a physician. The initial treatment plan included composite restorations and oral hygiene/cariogenic diet control instructions. In addition, four sessions of rubber cup/pumice prophylaxis were performed, plus 0.12% chlorhexidine mouthwashes and topical fluoride application once a week, to control and reverse the caries risk and activity.

Unfortunately, there was no commitment observed in the patient or her parents in contributing to the success of the dental treatment since they did not regularly attend the scheduled appointments and did not contribute to biofilm reduction at home. Thus, the oral hygiene of the patient was never satisfactory, which resulted in changes of the treatment plans several times during the follow-ups. Teeth extractions and repairs of the composite restorations were required [Figure 2].

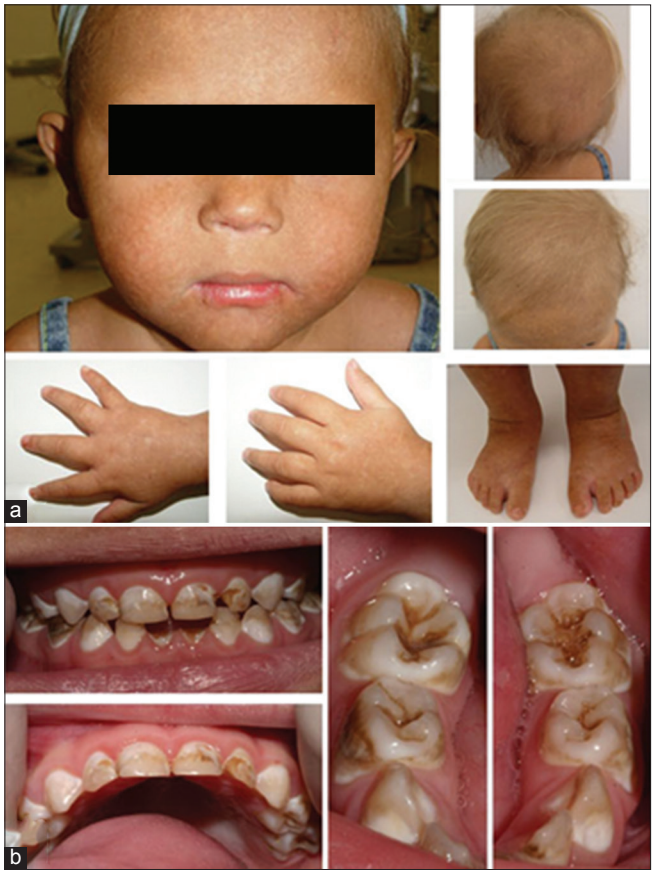

Figure 1: (a) Intra- and extra-oral findings at the first dental visit. Poikiloderma on the face, hands and feet skin; sparse hair (b) carious lesions in the primary maxillary incisors. White spot lesions and coronary darkened staining in all primary molars

Furthermore, the patient had behavior problems during the dental visits, probably due to her self-perception of sparse hair, stained skin, and decayed tooth appearance. For behavior management, nonpharmacologic methods were used such as voice control, nonverbal communication, tell-show-do, positive reinforcement, and distraction. Protective stabilization with a restrictive device performed by parents was also used.

After the conclusion of dental treatment and several follow-up visits, it was possible to detect fractures of some of the composite restorations, so endodontic treatments were performed and steel crowns were installed. Therefore, strict oral hygiene instructions were consistently applied.

An improvement in patient behavior during the dental visits, as well as in her self-esteem and sociability was noticed once she began to use a wig [Figure 3]. In addition, the change in her dental aspect and the elimination of toothache contributed to reaching a successful dental treatment with a better commitment observed by the improved home care hygiene and diet habits. After 7 years of follow-up, the dental treatment remained satisfactory.

Discussion

Rudimentary teeth, faulty root formation, microdontia, and higher susceptibility for dental caries[5,6] characterize the dental alterations of RTS, when present. Moreover, delays in tooth eruption, rhomboid glossitis, and hyperkeratotic tongue were also reported.[4]

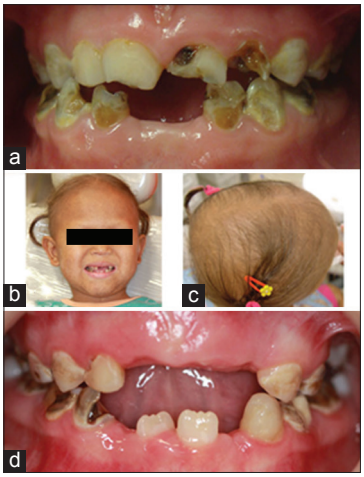

Figure 2: Findings during the dental treatment (patient was 4-year-old). (a) Intraoral view. (b) View of the face. (c) Sparse hair (d) patient at 5-year-old, still proving difficult in regards to commitment to oral health

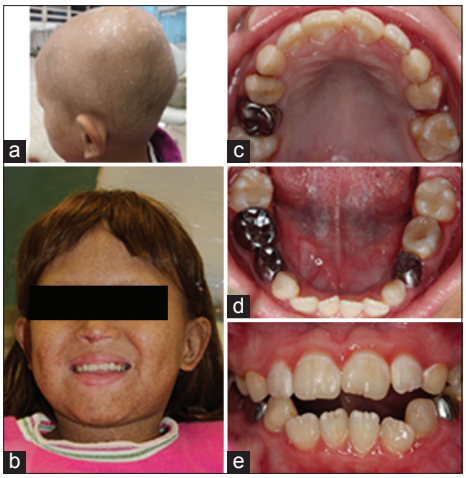

Figure 3: Patient at 8-year-old. (a) Lack of hair (b) extraoral view showing the use of a wig (c) conclusion of the treatment with steel crown and composite restorations of the maxillary (d) and mandibular arch (e) intraoral front view

Our patient presented microdontia, and general characteristics in agreement with other reports in the literature such as skin poikiloderma, sparse hair, alopecia of eyelashes and eyebrows, photosensitivity, short stature, and normal intellectual development.[1,2,5]

Tjon A Ten and Houwen (2007) reported the case of a patient with neonatal hyperbilirubinemia, primary central incisors with a greenish color (green teeth) and hypoplastic enamel. Later, she presented with the permanent molars also with a greenish color. This was due to bilirubin, which is incorporated into the teeth during the enamel matrix formation.[7] The patient reported in the present study had bile duct obstruction (cholestasis) and primary molars with color changes. The presence of cholestasis is not a common finding in the literature in patients with RTS; however, this was a relevant finding in our patient.

At odds with other findings, our patient showed no malignant neoplasms of skin and bones[8] or ocular alterations.[5] In addition, she did not present periodontitis (despite mild gingivitis) which may be due to young age.

Genetic disorders that alter the immune response is also etiological factors of periodontal diseases.[9] Structural defects of the epithelium and of connective tissue can also act as risk factors of periodontal destruction.[10] Our patient presented hyperemic and edematous gingiva.[4] Therefore, since early-onset periodontitis is a common finding in the literature,[5] it is important to control the gingivitis so that it does not evolve into periodontitis.

The dental care of patients with special needs is of great importance because it provides better quality of life by restoring oral health, which is often neglected. It contributes to the motivation in addition to having a positive influence on the general health of the patients. Contributions in the area of this rare syndrome help to demystify and increase knowledge of the peculiarities of the disease to promote appropriate and safe dental treatment for these patients.

The patient remains in assistance for orthodontic treatment, as well as for follow-ups of clinical findings associated with the syndrome.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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