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## Oral Rehabilitation of a Patient with Congenital Erythropoietic Porphyrria: A Rare Case Report

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## **CASE REPORT**

# **Oral Rehabilitation of a Patient with Congenital Erythropoietic Porphyria: A Rare Case Report**

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## **ABSTRACT**

Congenital erythropoietic porphyria (CEP) is an extremely rare disorder involved in chronic porphyrias. One of the recommended evaluations to determine disease severity and needs of a person with CEP is dental assessment. **Objective:** To present a case of CEP and its oral rehabilitation. **Case Report:** A 32-year-old female patient was admitted to the clinic with hyperkeratinization, skin fragility, fissures, and wounds in her perioral region and hands. Intraoral gingival bleeding, mucogingival stress, and missing teeth were recorded. After medical consultation, phase I and II periodontal therapies were performed. The patient expressed satisfaction of the results of the procedures in terms of the parameters evaluated. The rare occurrence of the disease may delay the diagnosis. An unmet need still exists for multidisciplinary orientation of patients with CEP. **Conclusion:** Dentists should have and follow guidelines for treating patients with CEP.

**Key words:** congenital erythropoietic porphyria, oral health promotion, porphyria, rehabilitation

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## **INTRODUCTION**

Porphyrias form a group of hereditary metabolic disorders caused by defects in the biosynthesis of heme and are categorized as hepatic or erythropoietic. Congenital erythropoietic porphyria (CEP) is an extremely rare disorder involving erythropoietic porphyrias and results from the insufficient activity of the fourth enzyme that belongs to the heme biosynthetic pathway, which is coded by the uroporphyrinogen III synthase gene.<sup>1-3</sup> Its symptoms include photosensitivity owing to massive porphyrin accumulation in the skin (subepidermal blistering with inflammatory infiltration)<sup>4</sup>; mild to severe anemia (resulting from induced osmotic hemolysis with the extreme accumulation of porphyrin in erythroid precursors, reticulocytes, and erythrocytes),<sup>5</sup> ocular complications (corneal scarring, ulcerative keratitis, and conjunctivitis),<sup>6-8</sup> hyperpigmentation,<sup>9</sup> facial scars, reddish-brown discoloration in the teeth, and sclerotic and osteolytic round lesions in the skull and jaws.<sup>10</sup>

CEP demonstrates unique dental-craniofacial symptoms in the early phase of the disease. Therefore, dentists may be the first healthcare providers to define this disease at an early stage. To achieve this, dentists should have sufficient knowledge of the relevant clinical features of CEP.<sup>10</sup>

This report presents the clinical, dermatological, and laboratory findings as well as the oral rehabilitation of a patient with CEP having periodontal problems and missing teeth.

## **CASE REPORT**

A 32-year-old female patient was admitted to the Department of Periodontology, Faculty of Dentistry, Necmettin Erbakan University because of gingival bleeding and missing teeth on consultation to another dentist. The patient's medical history revealed her previous diagnosis of CEP. The patient did not take any medication. Porphyria was not reported in the



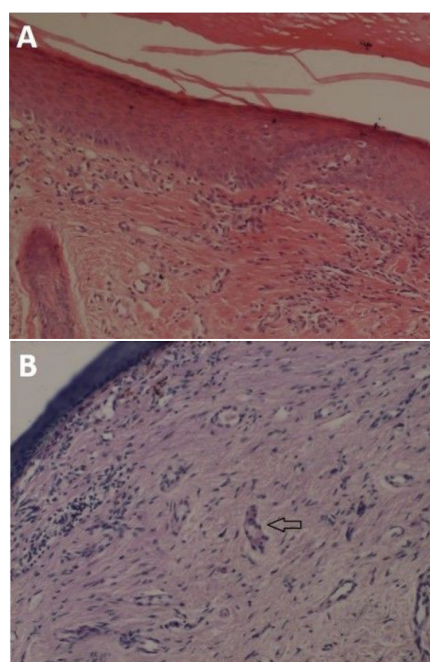
**Figure 1.** Dermatological signs of porphyria were observed in the patient.

patient's family and first-degree relatives. Moreover, the patient did not have regular dental examinations; thus, consultation was requested.

#### **Dermatological and laboratory findings**

The following signs were observed during dermatological examination: diffuse alopecia and minimal cicatricial alopecia; facial discoloration, increased telangiectasias, and erythema; cheilitis, erosion, and rhagades of the lips; increased erythema and pigmentation on the neck and body, xerosis, and skin thinning (dermatrophy); erythema hyperpigmentation, xerosis, and dermatrophy on hands and feet; rhagades, atrophy, and keratosis (hardening) in the palms; and nail discoloration, onychodystrophy, and xerophthalmia. Dermatological signs are shown in Figure 1.

In the laboratory examination, ANA and anti-ds DNA were negative, and mild hemolytic anemia, increased IgM level (2.66 g/L [0.4–2.3 g/L]), and mild hemolytic anemic peripheral spreading were determined. In the patient's urine, levels of creatinine, porphobilinogen, uroporphyrins 1 and 3, coproporphyrins 1 and 3, heptacarboxyl porphyrin, hexa-carboxyl porphyrin, and total porphyrin amounts were normal. In radiological examination, no pathology has been detected in the spleen and viscera.



**Figure 2.** (A) Thickening of the stratum corneum layer, increased collagen tissue in the dermis, and proliferation of the vessels (hematoxylin and eosin staining, 100×). (B) PAS-positive reaction in the vessel wall (arrow) (PAS, 100×).

In the biopsy for distinction from scleroderma, thick hyperkeratosis on the epidermis surface, pigment incontinence in the superficial dermis, capillary proliferation, prominence in some vessel walls, perivascular rare lymphocytes, and increase in collagen fibers have been detected in histological sections (Figure 2A). Compared to scleroderma, the collagen had looser texture. In the histochemical examination, although not conspicuous, PAS-positive deposition in some vessel walls has been detected (Figure 2B), and this was considered compatible with porphyria rather than scleroderma.

#### **Oral examination**

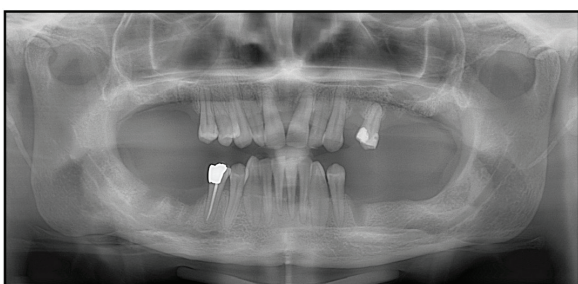
The clinical examination revealed hyperkeratinization, skin fragility, fissures, and wounds in the perioral region. In addition, the edges of the patient's lips tended to rupture. The patient was found to be sensitive to sunlight. She used a moisturizer to minimize this sensitivity.

The patient could not open her mouth widely, and she had 18 permanent teeth as seen during intraoral examination. Each quadrant was terminating edentulous, and no vestibule sulcus depth was seen in the posterior region. In addition, edematous gingiva, hyperemia, bleeding on probing, and plaque accumulation were observed (Figure 3); she was then diagnosed with periodontitis.<sup>11</sup> The patient did not feel any pain in the gingiva during probing. Mucogingival





**Figure 3.** Preoperative intraoral view of the patient showed edematous gingiva, hyperemia, bleeding on probing, and plaque accumulation.



**Figure 4.** Radiographic view of the patient showed alveolar bone loss around the remaining teeth and reduced bone height in the posterior edentulous sites; no periapical pathology.

stress was observed in the anterior region of the maxilla and premolars in the mandible due to shallow vestibular depth and inadequate keratinized tissue band. Moreover, gingival recession of the left central tooth in the maxilla was observed.

Radiographic evaluation showed alveolar bone loss around the remaining teeth and reduced bone height in the posterior edentulous sites; no periapical pathology was observed (Figure 4). The patient was consulted for prosthetic treatment due to missing teeth. To obtain functional and aesthetic result, planning was done with the prosthodontist before treatment.

#### Treatment

The patient was informed about the procedures to be performed, and she provided informed consent. The patient first consulted a physician for the procedures to be performed in the clinic. After consultation, each quadrant was debrided periodontally with ultrasonic and Gracey instruments (scaling and root planing). Then, the patient was informed about oral hygiene. Comfortable dental treatment was not possible owing to the fragility of the rigid lips, but the patient indicated that the postoperative pain was tolerable. The oral hygiene was strengthened during examinations.

An appointment for periodontal surgery was made for shallow vestibular sites where the patient could not



**Figure 5.** Vestibuloplasty procedures in the (A) maxilla and (B) mandible. The periodontal status of the patient was reevaluated, and with a diode laser, vestibuloplasty of the maxilla and mandible was performed under local anesthesia



**Figure 6.** Clinical view two weeks after dental treatment.

provide oral hygiene because she could not place the brush properly. On the day of surgery, the periodontal status of the patient was reevaluated, and with a diode laser, vestibuloplasty of the maxilla and mandible was performed under local anesthesia (Figure 5). The involved areas were closed with a eugenol-free periodontal dressing to ensure postoperative comfort. The patient was informed about postoperative care and prescribed flurbiprofen (100 mg tb, S.2X1) and chlorhexidine mouthwash. An appointment was made after 1 week to monitor healing. On the appointment day, the periodontal dressing was gently removed. The healing was uneventful, and the patient was asked to brush the operated areas gently.

At four weeks after the operation, the construction of a removable denture was started, and oral rehabilitation was completed two weeks later (Figure 6). Postoperative comfort, function, and aesthetic satisfaction were evaluated in control examinations, and the patient expressed satisfaction of the results of the procedures.

## **DISCUSSION**

Dermatological diseases and syndromes are often diagnosed by determining systemic signs and symptoms. Dental pathology is observed in a number of dermatological disorders, but it is often overlooked. A number of dermatological skin diseases and syndromes are associated with dental signs and periodontal pathology.<sup>12</sup> Assessment of specific dental signs related to CEP may be helpful in diagnosis.<sup>13</sup> Although CEP is an extremely rare metabolic disorder, it is still encountered clinically. We believed that the present report may be useful for clinicians owing to the few studies available in the literature on dental assessment of patients with CEP.

To provide optimal treatment in patients with porphyria, dentists should be in dialogue with the patient's physician. As management of patients with porphyrias, pharmacological concerns have been considered important. Drugs have been categorized according to their porphyrinogenic potential: safe or likely safe, unsafe or likely unsafe, and unclear.<sup>14</sup> In the present case, planned operations were discussed with the physician. As a result of the consultation, the relevance of the planned procedures was confirmed, and no further recommendations were made by the physician.

After phase I of periodontal therapy, the patient had good oral hygiene, and vestibuloplasty under local anesthesia (using epinephrine) was performed uneventfully. In addition, the prescribed drug showed no adverse effect during the postoperative period. Antibiotics were not prescribed. Moreover, cobalt-chrome alloy used for the partial denture did not have an adverse effect within one year of follow-up.

Although the patient uses moisturizer, comfortable treatment could not be achieved due to limited mouth opening (insufficient visual area) and fragile and rigid lips with a tendency to rupture. Elimination of the shallow vestibular depth, which is a problem in ensuring oral hygiene, provided comfort to the patient. Moreover, gingival recession due to mucogingival stress, which caused a non-aesthetic view, was eliminated to some extent. Given the limited mouth opening and the formation of a second surgical site, the patient preferred vestibuloplasty procedure rather than other periodontal plastic surgeries. As a result, regions that served as reservoirs for plaque accumulation during the preoperative period were eliminated.

During the operation, her lips were bleeding because of limited mouth opening. This condition made the treatment difficult for both the dentist and the patient.

Insufficient visual area may cause mild pain and discomfort inevitably, and the quality of life of the patient is adversely affected during the postoperative period. CEP is a multisystemic disorder. In addition to hematologic complications, cutaneous, ocular, oral, and skeletal findings contribute to determining the severity of disease and health-related quality of life.<sup>7</sup> The influence on the quality of life also affects the outcome and success of treatment.

## **CONCLUSION**

In conclusion, we recommend the following to achieve optimal success in the dental treatment of a patient with CEP. Firstly, it is a necessity of a multidisciplinary cooperation between family physicians, dermatologists, and porphyria specialists is essential for the diagnosis and management of porphyria,<sup>15</sup> the patient should consult a physician to perform optimal treatment.<sup>16</sup> Secondly, cooperation with the patient is important, the patient must be informed about the planned operations, and the expectations must be answered. Thirdly, if the mouth width is limited, using a soft child toothbrush or an electric toothbrush may help provide the best possible oral hygiene,<sup>17</sup> and dental visits at least two times a year are a must to maintain a healthy oral status.

## **CONFLICT OF INTEREST**

The authors declare that there were no conflicts of interest related to this case report

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