**CASE REPORT**

**Porphyria cutanea tarda - A case report**

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**Introduction**

Porphyria was first described in literature by Waldenstrom in 1937. It is a metabolic disorder that results due to decrease in uroporphyrinogen decarboxylase (URDO) activity, clinically characterized by blisters, photosensitivity, and hyperpigmented macule in sun-exposed area.[1]

There are seven main types of porphyria; they are broadly classified according to clinical features into neuropsychiatric, dermatological, and mixed forms. Acute intermittent porphyria and plumboporphyria are predominantly neuropsychiatric; congenital erythropoietic porphyria, porphyria cutanea tarda (PCT), and erythropoietic protoporphyria have predominantly cutaneous manifestations, and variegate porphyria is classified as mixed as they have both cutaneous and neuropsychiatric features.

**Case Report**

A 64-year-old male patient reported to the department of oral medicine and radiology with a chief complaint of ulcer in the lower lip for a week. The patient gave a history of fluid-filled vesicle which spontaneously ruptured leaving an ulcer in the lower labial mucosa of lip.

History revealed that he was apparently normal 7 years back when he developed blood-filled blisters and peeling of skin from hands on doing outdoor activities predominantly involving the sun-exposed area. The blisters were not associated with pain and used to rupture spontaneously a day later and leave erosions which used to heal with depigmentation [Figure 1]. The patient also gave a history of similar lesions over the bodies wherever he received minor trauma.

Medical history revealed that the patient had visited private practitioner for pain in abdomen 5 years ago, where the patient was subjected for routine blood investigations which showed increased SGPT (127IU/L) and alkaline phosphatase was (258IU/L) and ultrasound revealed increased bilateral renal cortical echogenicity with mild hepatomegaly for which patient was on treatment for few months, but later, he discontinued the treatment, the record of which could not be completely traceable. The patient was a chronic smoker, smokes 10–12 beedies for 25 years.

Extraoral examination revealed long thick eyebrows [Figures 2 and 3], senile comedones were noted over temples and cheek region. Purpuric lesions with petechiae and ecchymosis were present in the malar region; nails were long with onychomadesis.

Abstract

Porphyria is a group of metabolic disorder that results due to decrease in uroporphyrinogen decarboxylase activity. A 64-year-old male patient presented to our department with a chief complaint of ulcer in lower lip for a week. Intraoral examination revealed a healing ulcer in lower lip with petechiae on the palate; general physical examination revealed multiple purpuric lesions over the upper and lower extremities predominantly involving the sun-exposed areas. The clinical features, histological findings with immunofluorescence test, and urine analysis confirmed the diagnosis of porphyria cutanea tarda. The details of which are discussed in this paper. Various primary cutaneous diseases involve the mucous membrane throughout the body, including oral mucosa. Hence, dentist must have sound knowledge to recognize and establish the diagnosis of the dermatosis exhibiting concomitant lesion of the oral mucosa.
Intraoral examination revealed a healing ulcer in the lower lip with multiple petechiae over the palate [Figure 4], there was no new lesion noted intraorally. Further, the patient was subjected to routine blood investigations which were under the normal limits. The patient was referred to the department of dermatology where skin biopsy was obtained and subjected for histopathology. The section study showed skin with thinned out epidermis with subepithelial bullae. There was homogenous eosinophilic material deposited in the papillary dermis and around the capillaries. Congo red stain was negative for amyloidosis. The histological features are suspicious of porphyria [Figure 4]. Further, direct immunofluorescence test was conducted which was positive for immunoglobulin (Ig) G antibodies and pattern was dermal papillae and dermal vessels were observed. Examination of the urine with a woods lamp revealed coral pink fluorescence which confirmed the diagnosis of PCT.

Discussion

PCT is the most common form of porphyria worldwide, with an incidence of 1 out of 10,000–25,000 people. Due to abnormalities in the heme biosynthesis pathway leading to the accumulation of porphyrins, which leads to cutaneous lesion predominantly in sun-exposed areas. It has slight male predilection and affects the patient in the fourth or fifth decades; similarly, our reported case was in male and was on the sixth decade.

PCT has been classified into three types, all of which reduce the activity of UROD. Type I, the most common, is sporadic form and accounts for 75%–80% of PCT cases. UROD levels are normal, but enzymatic activity in the liver, exclusively, is reduced by at least 50%. Type II is familial inherited as autosomal dominant and accounts for 15%–20% of PCT cases. Type III PCT has been observed in which a genetic predisposition leads to decreased UROD activity in the liver alone.
Various genetic and acquired susceptibility factors have been identified and studied in PCT patients. Most PCT patients have three or more precipitating factors. The most common are cigarette smoking, alcohol or estrogen use, iron overload, hemochromatosis, and viral infections. Cigarette smoking has been found in 81% of patients with PCT. PCT patients who smoke tend to develop cutaneous symptoms at a younger age than non-smoking PCT patients. Smoking induces CYP1A2 synthesis that may, in turn, lead to increased production of UROD inhibitor.[4]

The UV radiation with absorption of the Soret band (400 nm to 410 nm wavelength) by porphyrins that accumulate in blood vessels of the upper dermis causes the photosensitivity found in PCT. Destabilization of porphyrins followed by sun exposure leads to the formation of reactive oxygen species that clinically presents with characteristic bullae and skin fragility on sun-exposed areas such as the dorsal hands, forearms, ears, and face.[5]

The bullae in PCT are filled with clear fluid and are formed by sun exposure or minor trauma. The fragile bullae rupture, creating erosions, and shallow ulcers that heal slowly and lead to scarring, milia, and/or altered pigmentation, similar such presentation was noticed in our case. Facial hypertrichosis can be the first clinical presentation.

A urine sample is often, but not always, grossly discolored with a tea or wine - colored tint examination of the urine with a woods lamp may reveal coral pink fluorescence due to excessive porphyrins. Other investigations include complete blood count to assess hemoglobin levels measurement of iron stores, which may be increased in over 30% of patients.

Histopathology of PCT reveals a subepidermal bulla with little to no inflammatory infiltrate and an undulating, festooning dermal papilla projecting into the bulla. Thickened blood vessel walls in the dermis occur due to deposition of PAS-positive material. Caterpillar bodies, a common, but not diagnostic, characteristic of PCT consist of PAS-positive eosinophilic, elongated, and wavy structures usually found in the epidermis above the bulla. Direct immunofluorescence shows IgG, IgM, fibrinogen, and complement in the basement membrane and around vessels of the upper dermis.

Differential diagnosis includes epidermolysis bullosa acquisita is a rare disease with an incidence of 0.2% cases per million. In mild cases, clinical presentation is similar to PCT, while severe cases are comparable to hereditary recessive dystrophic epidermolysis bullosa. Autoantibodies targeting Type VII collagen, the major component of anchoring fibrils that connect the basement membrane to dermal structures exclusion is based on normal uroporphyrin.[6]

Pemphigus vulgaris is an autoimmune, intraepithelial, blistering disease affecting skin, and mucous membranes. Nikolsky’s sign is positive and usually diagnosed by biopsy.

Management of PCT begins with avoidance of all possible precipitating factors such as alcohol and estrogen. Physical barriers and sun protection, such as clothing and sunscreens containing zinc oxide, and avoiding skin trauma protect against worsening skin disease. This initial approach may be sufficient to treat PCT.

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Conclusion

There is considerable overlap of clinical features of various vesiculobullous lesions. The oral mucosa is thin, easily traumatized causing vesicles and bullae to break rapidly into ulcers resulting in non-specific appearance of the lesions on oral mucosa compared to skin lesions. Hence, dentist must have sound knowledge on clinical features, histological, and immunological data that enables early diagnosis of such chronic dermatological disorders.

References
