Author’s response to reviews

Title: An example of dark side of the liver diseases; PORPHYRIA CUTANEA TARDA: a case report

Authors:

Hanife Usta Atmaca (hanifeusta@yahoo.com)
Feray Akbas (atlibatur@yahoo.com)

Version: 1 Date: 04 Oct 2018

Author’s response to reviews:

Thank you very much for the kind efforts of the reviewers. There is so much information to guide us and enhance quality of our work in their detailed comments. We appreciate the kind help.

Revision notes about reviewer reports:

Reviewer #1:

Some specific comments:

1. The diagnosis pattern is described.

Porphyrin (24 hour urine): 832 ug/24h (N ≤ 100), Porphobilinogen (24 hour urine): 1.65 mg/24 h (N: ≤ 1.65). We could not fractionate the urine porphyrins, as we don’t have the necessary equipment to measure carboxylate porphyrins (uroporphyrin and hepta-, hexa-, and pentacarboxyl porphyrin) in our hospital. The patient was diagnosed with PCT according to his medical history, typical skin lesions and supportive laboratory findings.

2. Response of the patient to phlebotomies is mentioned.

Phlebotomy was started as treatment regimen (450 cc/every 2 weeks). After the 6th phlebotomy, his symptoms regressed and declared that he felt better. His laboratory analysis also showed improvement (AST 30U/L ALT 38U/L ferritin: 43ng/ml. A follow-up was started and an appointment to see him in 3 months was made.

3. Exclusion of porphyrias other than PCT is described.
As commonly seen in PCT patients, our patient also had atrophic and hyperpigmented skin lesions. There were no typical clinical signs or autoimmune laboratory findings for scleroderma. Thus we eliminated scleroderma in differential diagnosis. Lack of abdominal pain and neuropsychiatric findings of acute intermittent porphyria, discarded variegate porphyria and hereditary coproporphyria.

4. The action mechanism of chloroquine and hydroxychloroquine is redescribed.

In some cases, patients can be treated with low-dose antimalarials, such as hydroxychloroquine or chloroquine, drugs that act as mobilizers of porphyrins from the liver, by transforming hepatocyte porphyrins into water-soluble complexes which excrete in the urine.

5. We could not obtain new photographs, the present one can be removed if it’s not appropriate.

All manuscript is checked for grammar and spelling mistakes.

Reviewer #2:

1. Do you believe the case report is authentic? Yes

2. Do you have any ethical concerns?

Comments: No ethical concerns. Informed consent was obtained.

3. The Introduction is rewritten to explain the relevance of the case to the medical literature.

We referred that PCT is "an acquired disease and sometimes genetic" and did divide in sporadic (type 1) or familial (type 2 and 3) and referred epidemiology of different types.

The porphyrias are a group of rare metabolic disorders, inherited or acquired, along the heme biosynthetic pathway, which could manifest with neurovisceral and/or cutaneous symptoms, depending on the defective enzyme.

4. Does the article report the following information?

-Demographic information (age, gender, ethnicity) and main symptoms are identified.
59-year-old Turkish male patient presented with fatigue, loss of energy and dark urine color. When asked, he declared that hyperpigmentation occurred in his hands and face after exposure to sun since last year and sometimes those skin parts blistered and healed leaving a scar.

- Medical, family and psychosocial history are mentioned.

He used to consume alcohol socially but since last year started to take alcohol on daily basis. His medical history and family heredity history were both unremarkable. He was a butcher and he consumed over 300 gr of meat on most days. He declared that his complaints exaggerated after consuming large amounts of meat.

- Reference to HCV or HIV status was made.

Hepatitis C, hepatitis B, and HIV testings were negative.

- Types and mechanism of intervention is explained. A summary of the clinical course of all follow-up visits is added.

Phlebotomy was started as treatment regimen (450 cc/every 2 weeks). After the 6th phlebotomy, his symptoms regressed and declared that he felt better. His laboratory analysis also showed improvement (AST 30U/L ALT 38U/L ferritin:43ng/ml. A follow-up was started and an appointment to see him in 3 months was made.

5. Is the interpretation (discussion and conclusion) well balanced and supported by the case presented?

New parts are added to discussion, especially to describe the relationship between the disease and alcohol intake and his occupation as susceptibility factors.

Skin lesions are redescribed.

New references are added to describe the diagnosis pattern and the cost-effective approach.

6. Is the anonymity of the patient protected? Yes

7. Is the Abstract representative of the case presented?

All sections (introduction, discussion, conclusion) are rewritten. The complete name of the uroporphyrinogenedecarboxylase (UroD) enzyme is written instead of "urrod enzyme" once it is the first reference.
8. Does the case represent a useful contribution to the medical literature?

Treatment and follow-up are described in more detail.

9. Additional comments for the author(s)?

Whole text is revised for grammar and spelling mistakes.