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Case Report

Late cutaneous form of porphyria cutanea tarda in a 26-year-old man: a clinical case report

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ABSTRACT

Porphyria cutanea tarda (PCT) is a rare form of enzymatic disorder of heme-biosynthesis pathway. Due to a defect of enzyme uroporphyrinogen decarboxylase (UROD) this disease occurs. Porphyria cutanea tarda can be sporadic and can be familial. Here, we are presenting a late cutaneous form of porphyria cutanea tarda in a 26-year-old male patient without any known triggers. The patient was diagnosed by genetic and laboratory testing and showed typical appearance of PCT. All the details of PCT from presentation of symptoms, complications, involvement of organs and diagnosis is discussed in this article. Furthermore, this article provides thorough discussion about PCT.

Keywords: Porphyria cutanea tarda, Uroporphyrinogen decarboxylase, Familial PCT, HCV, Phlebotomy

INTRODUCTION

Overall porphyria cutanea tarda estimates prevalence ranged from 5 to 10 persons from 100 000 people usually.[1] In Norway, the incidence of PCT is estimated at 1 in 100,000, this includes 50% of familial form of PCT and 50% acquired form of PCT.[2]

In Sweden, a study in the Gothenburg area found approximately 1 patient with PCT per 10,000 inhabitants.[3] It suggests higher prevalence in Scandinavia compared to many other countries. In PCT studies A male predominance of 1.8:1 was found or male predominance in percentage (66.7%) found out. The median age at diagnosis was about 49 years that ranges from (18-71). Family history of PCT was observed in 19.5% of patients. Two or more acquired precipitating factors were present in 42.5% of cases of PCT.[4,5] Porphyria cutanea tarda (PCT) is a type of disease that is associated with defective enzymes of heme-biosynthesis pathway. It is a type of metabolic disorder with defective enzymes that can be inherited or acquired. In PCT the defective enzyme is uroporphyrinogen decarboxylase (UROD).[6] Either reduction or inhibition of this enzyme (UROD) in the liver leads to this disease (PCT). This enzyme (UROD) is encoded by a gene named UROD and it is found on chromosome.[11] In time of liver damage if UROD activity is reduced to less than 20%, intermediates uroporphyrin I/III and heptacarboxyl porphyrin (figure 1) accumulates in liver and when it accumulates more in liver it also starts to appear in plasma and bile. At this stage detection of enzymes by laboratory analysis is possible.[1] Well-known risk factors for PCT are mainly HCV infection after alcohol abuse and in 50% of male patients with PCT showed this pattern simultaneously. Another risk factor includes URO-D gene mutation and estrogen exposure; this pattern is commonly seen in women who were diagnosed with PCT.

Another risk factor includes mutation at HFE locus that has been detected in 63% of PCT patients.[7,8] The Swedish study found that 29% of male patients tested positive for hepatitis C virus antibodies, 45% of tested patients had impaired glucose tolerance and 57% of tested patients had

haemochromatosis gene mutations.[3] These genetic and viral factors appear to play important roles in disease development and may contribute to geographic variation in prevalence.

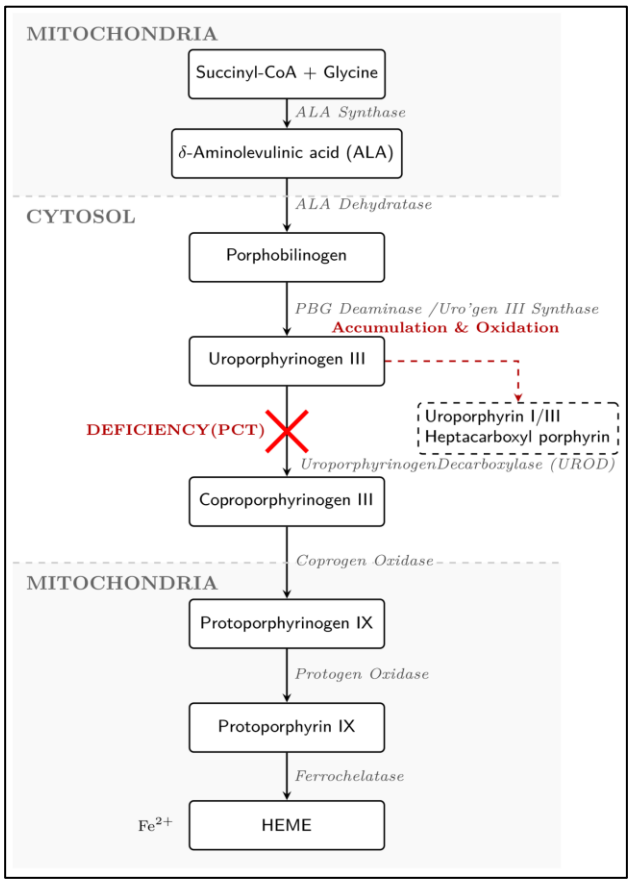

Figure 1: The heme biosynthesis pathway. Deficiency of UROD manifests as PCT with accumulation of porphyrins intermediates (uroporphyrin I/III and heptacarboxyl porphyrin).

CASE REPORT

26-year-old male (patient T.), Sportsman (hockey player), Height 192 cm, Weight 96 kg presented to Grodno hospital with complaints of blisters and wounds on the skin of his hand. He had never used drugs, alcohol or steroids. Only proteins for sports nutrition.

Two years ago, he noticed the appearance of wounds, micro-cuts, and blisters that were only on the skin of his hands (the rest of his body was clear). Those wounds were mildly painful and healed very poorly, leaving red scars at the site. He did not notice any connection with sun exposure.

He consulted a dermatologist in Grodno and took all the recommended treatments but he did not notice any positive effects. After that he consulted another dermatologist, who recommended some tests and an ultrasound. Elevated liver enzymes ALT 174.5 units/l (normal: <41 units/l), AST 85 units/l (normal: <37 units/l) and hepatosplenomegaly were detected. The patient then received treatment for a suspected diagnosis of hepatitis/cirrhosis and initially these drugs were given: ursodeoxycholic acid, Essylimar 100 mg, LIV-52, Hepa-Merz, and Vitamin E.

Ultrasound details of abdominal organs; showed signs of minor diffuse changes in the liver of a chronic nature and splenomegaly.

Liver size was not enlarged, size of liver lobes right sided 157 mm and left sided 81 mm and increased echogenicity. Spleen was enlarged with dimensions of 140×60 mm and homogenous structure. Other organs of the abdominal cavity (kidneys, adrenal glands, gall bladder and bile duct) were normal in size and showed no abnormality on ultrasound examination.

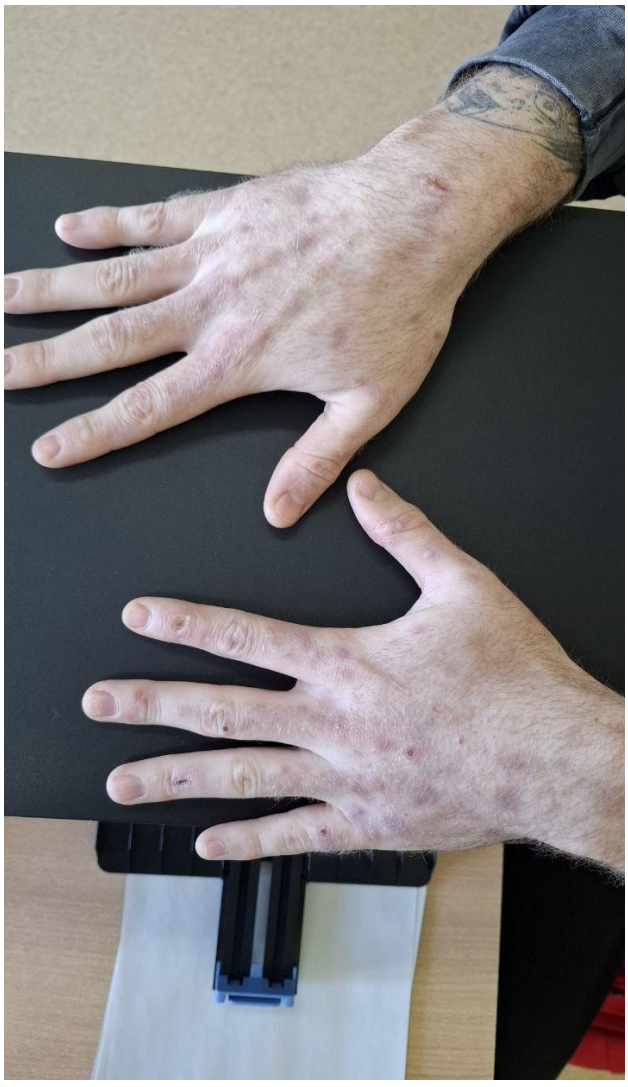

Figure 2: PCT patient T. hand after treatment with multiple courses of phlebotomy and low dose hydroxychloroquine.

He did not notice any positive effects, after which he was recommended for additional testing and he tested positive

for porphyria. Upon examination of blood, doctors found out increased level of pentacarboxylporphyrins 174.72 nmol/day (normal:<=10 nmol/day) and hexacarboxyporphyrins 17.33 nmol/day (normal:<=8 nmol/day) and heptacarboxyporphyrins 14.17 nmol/day (normal:<=9 nmol/day). The increased level of these intermediates shows presence of PCT. increased iron was 35.02 μmol/l (normal:11.6-31.3 μmol/l) and level of ferritin 383 μg/l (normal 22-275 μg/l) also consistent with PCT. The normal level of intermediates like delta-aminolevulinic acid was 5.14 micromol/l (normal:<15 micromol/l) and porphobilinogen was 2.16 micromol/l (normal:<=2.2 micromol/l) suggests that it is not acute intermittent porphyria. Other intermediates coproporphyrin 1(136.92 nmol/day) (normal:<=168 nmol/day) and coproporphyrin III (23.24 nmol/day) (normal :<= 230 nmol/day) were also within normal ranges and that suggests other enzyme of heme-biosynthesis pathway works normally that is the reason level of these intermediates are within the normal range.

An initial phlebotomy was performed, with recommendations for a monthly blood draw of 450 ml, hydroxychloroquine 200 mg twice daily, and ursodeoxycholic acid 500 mg in the evening for one month.

Initially, three courses of phlebotomy were recommended, followed by a follow-up consultation. Patients denied any history of genetic diseases in families (including PCT). The patient T. also did a genetic test to rule out any possibility of acute intermittent porphyria for that test of HMBS gene done. That test did not detect extended deletions and duplications of the HMBS gene and no pathologic variants in the non-coding region of the gene and he tested negative for HMBS gene test but this single test can not rule out other forms of porphyria that are caused by mutation in other genes.

Hepatitis markers were checked with usage of ELISA and both HBsAG and Anti-HCV results came back negative and that rules out common triggers (Hepatitis E and C infection) for development of PCT. There was a rash on his hands consisting of blisters (left thumb), crusted lesion and hyperpigmentations. Increased skin fragility and hypertrichosis is also visible.

After multiple courses of phlebotomy patients' condition improved (figure 2). Although there is no definitive treatment for PCT, phlebotomy often shows promising prognosis in patients without contraindications of phlebotomy.

Patient T. denied any consumption of alcohol or tobacco smoking and he never had hepatitis or any liver infection that he knew of. He also denied the role of the sun in PCT symptoms and prognosis. He does not recall any chronic or severe disease in the past (including liver infection). Patient T. didn't have any problems until the age of 24 when he started to notice all of the symptoms including blisters and wound of the dorsal surface of both hands. He denied having any infection in childhood and he was a totally healthy child.

Now in his treatment he got 2 courses of phlebotomy once in a month. Each time 500 ml. Both time before and after the phlebotomy laboratory examination was performed and we saw prognosis after phlebotomy. Increased levels of AST(60 units/l) (normal:<37 units/l), ALT(160 units/l) (normal:<41 units/l) and total bilirubin(21.6 micromol/l) (normal:3.4-20.5 micromol/l), cholesterol(5.18 mmol/l) (normal:<5 nmol/l) as well as iron (35.02 micromol/l) (normal:11.6-31.3 micromol/l) and ferritin (383 microgram/l) (normal:22-275 microgram/l) were determined before phlebotomy. After phlebotomy all levels except ALT (50 units/l) (normal<41 units/l) came within the normal ranges. Hematocrit result after phlebotomy was 49.4% (normal:39-49). Tablets of hydroxychloroquine he took for 2 months. He mentioned that he has the same changes of diseases of PCT with or without hydroxychloroquine.

DISCUSSION

PCT can be type 1 and type 2. Type 1 of PCT is an acquired disorder and it is triggered by iron overload or viral infection. Type 2 is autosomal dominant.[9] Triggers for familial(genetic) form of PCT can be usage of alcohol, smoking and disease involving level hepatitis C because UROD enzyme is synthesized mostly in hepatocytes and UROD enzyme is defective in PCT.[10]

Primary Genetic Tests for PCT includes UROD Gene Testing the UROD gene is the most important genetic marker for PCT. Genetic testing for UROD mutations is essential to distinguish between familial PCT (Type 2) and sporadic PCT (Type 1).[11] Type 2 PCT, which accounts for about 25% of PCT cases, involves an inherited partial defect in UROD activity that can be identified through genetic sequencing.[12] However, Type 1 PCT, the more common sporadic form, typically does not involve UROD mutations but rather acquired enzyme deficiency in the liver.

HFE Gene Testing The hemochromatosis gene (HFE) is frequently tested because iron overload is a major risk factor for PCT manifestation.[12] The two most common HFE mutations tested are C282Y and H63D Approximately 50% of PCT patients have HFE gene mutations and these mutations significantly influence iron metabolism, which can trigger PCT development in genetically predisposed individuals.[13]

Diagnosis of PCT is depend on detailed history of patient, clinically and characteristics appearance of wounds and blister on the skin of back of the hand but main diagnosis still depends on laboratory evaluation (blood test, urine test and skin biopsy), increased level of porphyrins intermediates (uroporphyrin I/III and heptacarboxyl

porphyrin) is helpful for definitive diagnosis. Other methods include gene testing in case of familial PCT.[6]

Treatment regimen of PCT includes low-dose hydroxychloroquine, repeated phlebotomies or both as well as removing triggers (offending agent). Before the course of phlebotomies laboratory evaluation is usually performed.[14] In this case study, the patient was recommended for a monthly blood draw of 450 ml, hydroxychloroquine 200 mg twice daily, and ursodeoxycholic acid 500 mg in the evening for one month. Elevated level of ferritin is the main precipitating factor of PCT so in order to overcome that phlebotomy is performed.[15] Some studies found out that relapse of PCT with low or high dose hydroxychloroquine is the same but it is lower with phlebotomy.[16] So nowadays phlebotomy is one of the most popular treatment options for PCT.

Differential diagnosis of PCT can include vesiculobullous disorders, mainly on the skin of the sun exposed areas, pseudoporphyria that can be idiopathic or drug induced (especially with non-steroidal anti-inflammatory drugs or non-inflammatory type of epidermolysis bullous acquista.[17] It is very important to distinguish PCT from other porphyria and it is generally difficult to do that.[18]

Uncommon Manifestations, while dermatological involvement is primary, discuss potential extradermatological complications; Ocular involvement: Corneal-scleral thinning can occur and in untreated cases, may lead to sight-threatening progressive involvement.[19]

In this case study our patients' rash and appearance of symptoms was typical but PCT found out without any involvement of triggers. Our patient denied any form of trigger (alcohol, smoking, etc). Exposure to the sun was also not a factor of development of PCT in our patient.

It is very important for physicians to think about PCT when they see blisters and erosion with Mila and hypertrichosis on the skin of sun-exposed areas. Through history of family and genetic analysis distinguishing between porphyria is possible and diagnosis can be possible according to that.

CONCLUSION

Porphyria cutanea tarda, though rare in young males and often linked to identifiable hepatic risk factors, should remain in the differential diagnosis of photosensitive blistering disorders across age groups. In this case of a 26-year-old man without a clear trigger, targeted phlebotomy to reduce hepatic iron burden combined with low-dose hydroxychloroquine produced rapid and durable clinical improvement and normalization of porphyrin parameters. This experience underscores the value of a proactive, multidisciplinary approach and individualized therapy in atypical PCT presentations. Primary healthcare and as well as (dermatologists and hematologists) clinicians should maintain vigilance for PCT in younger patients with compatible cutaneous phenotypes, pursue a comprehensive diagnostic workup and implement a coordinated management plan that addresses potential triggers, hepatic health and recurrence risk.

The diagnosis can usually be made with high levels of intermediates uroporphyrin I/III and heptacarboxyl porphyrin that present in plasma and urine in the setting of PCT. Patients with PCT management are done by low-dose of hydroxychloroquine and removal of triggering factors as well as phlebotomy.

Finally, this case study highlights gaps in definitive treatment of porphyria disorders including PCT and lack of awareness of PCT initially in terms of diagnosis by clinicians because PCT is easy to be misdiagnosed. This study points out an abnormal form of PCT without any known trigger.

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