

CLINICAL CASE

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Clinical history

- □ 37-year-old moroccan man
- ☐ He is heterozygous for Beta thalassemia
- ☐ Sometimes he had mouth sores
- ☐ No history of smoking or drugs
- ☐ Complains:
 - **≻** Fatigue
 - > Red-colored urine

Physical examination



Differential diagnosis

- ☐ Vascular occlusion?
- ☐ Vasculitis?
- ☐ Buerger disease?
- ☐ Calciphylaxis? Oxalosis?
- ☐ Infection?
- ☐ Toxic?

Physical examination

- ☐ T: 37°C, Bp: 120/70 mmHg, Pulse rate: 80 bpm
- ☐ Urine analysis : normal
- Jaundice
- ☐ No mouth aphtosis, no genital scar
- ☐ No hepatosplenomegaly
- no adenopathy
- ☐ Cardiorespiratory and neurological examination found no abnormalities

Laboratory tests

☐ CRP 195 mg/l, ESR 101 mm ☐ PT, PTT, Fg normal ☐ Hb 9 g/dl MCV 80 fl Plt 33109/µl WBC 8000/μL ☐ Blood smear : No schizocytosis, no blasts ☐ Direct Coombs negative ☐ Bilirubin total 2 mg/dl, billirubin direct 0.5 mg/dl No hemoglobinuria, no hematuria Electrolytes, hepatic enzymes and creatinine normal ☐ HBV, HCV, HIV negative

Laboratory tests

- ☐ Skin biopsy: epidermal necrosis with no sign of vasculitis
- ☐ Prot S, Prot C, FV Leiden, Prothrombin mutation G20210A, Antithrombin , Antiphospholipid negative
- ANA, complement normal
- ☐ ANCA negative, Cryoglobulinemia negative
- ☐ Bone marrow normal

Laboratory tests

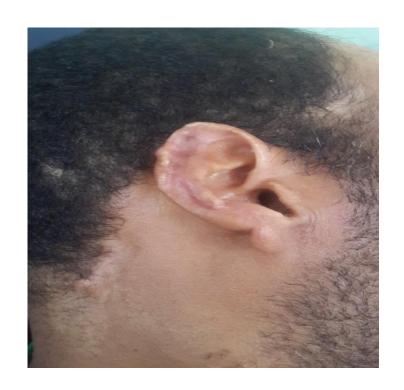
□ Flow cytometry → PNH clone size
Granulocytes 95%
Monocytes 91%



Classical Paroxysmal Nocturnal Hemoglobinuria (PNH)

Treatment

- ☐ Methylprednisolone IV (1g/d) then prednisone 1mg/kg/d
- ☐ Anticoagulant
 Tinzaparin 175 UI/kg/d
- ☐ No allogeneic HSCT
- No Eculizimab



After 1 month

2 months later...

- ☐ Acute abdominal pain + Jaundice + fever
- □ Abdominal CT scan : splanchnic vein thrombosis
- ☐ Death by necrosis of the intestine and liver failure

Take home messages

☐ PNH: rare acquired somatic mutation of the HSC related to PIG-A gene ☐ Sites affected are manifold and often at atypical localization ☐ Nearly three-fourths of PNH patients do not present with hemoglobinuria ☐ Thrombosis: the major life-threatening complication affecting outcome in PNH despite anticoagulation Li Eculizumab should be introduced rapidly

Welcome to Rabat!

