

# 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/ $\mu$ l  
WBC 8000/ $\mu$ L
- Blood smear : No schizocytosis, no blasts
- Direct Coombs negative
- Bilirubin total 2 mg/dl, bilirubin 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
- ❑ Eculizumab should be introduced rapidly

# Welcome to Rabat!

