Case Presentation

**S**
- 25 year old Caucasian female, presented with a 4-week history of fatigue, bilateral leg swelling, shortness of breath associated with haemoptosis, epistaxis and prolonged vaginal bleeding

**B**
- No significant past medical history. Recently completed a course of amoxicillin started by GP
- Commenced COCP 4 weeks prior to presentation

**A**
- **Observations and examination:** BP 174/119, HR 120, RR 24, SpO2 97% on air and T 37.2. Bilateral crepitations to the mid-zones & bilateral pitting oedema
- **Bloods:** Cr 1511 umol/L, Ur 33.2 mmol/L, Na 123 mmol/L, K 4.4 mmol/L, Hb 74 g/L, Plt 165 x 10^9/L, WCC 10.2 x 10^9/L, CRP of 82 mg/L. Liver function and clotting normal.
- Urine dip: +++ blood, +++ protein. Chest XFR: bilateral patchy opacification, left sided pleural effusion
- US KUB: showed hyper-echogenic kidneys with comparatively ill-defined cortico-medullary junctions, suggestive of glomerulonephritis or nephrotic syndrome.

**R**
- Acute kidney injury requiring haemodialysis? Cause
- Urgent renal biopsy
- Started on high dose steroids for suspected rapidly progressive glomerulonephritis secondary to a vasculitis

Further Investigations:
- **Autoimmune screen:** Anti-GBM antibody, MPO ANCA, PR3 ANCA, Rhf, CFD screen, Anticardiolipin antibodies = all negative. Complement C3 and C4 were normal.
- **Renal biopsy:** thrombotic microangiopathy
- **ADAMTS13:** normal activity
- **Genetic testing:** heterozygous mutation to complement factor H c.3616C>Tp.(Arg1206Cys)

Diagnosis: Atypical Haemolytic Uraemic Syndrome

Management:
- Initially started on plasma exchange, with poor response.
- Commenced Eculizumab 900 mg infusion every 2 weeks.
- Renal recovery and dialysis independent within 6 weeks: Na 143, K 4, Ur 5, Cr 106, Hb 132, Plt 251

Discussion:
- aHUS classically presents as a triad of MAHA, thrombocytopenia and renal impairment.
- In this case, thrombocytopenia was not present based on laboratory guidelines however, based on bloods after recovery there was evidence of a >25% drop in platelet count. There are case reports of aHUS in the absence of thrombocytopenia.
- aHUS is difficult to distinguish clinically from other causes of TMA.
- Historically, treatments for aHUS were limited and ineffective; with frequent relapses leading to end stage renal failure and long-term dialysis.
- Impaired complement regulation, associated with genetic mutations, is the pathophysiological process underlying aHUS.
- This understanding has led to successful implementation of treatment with eculizumab.

Key Learning Points:
- Renal biopsy remains the gold standard for investigating AKI
- aHUS can present in the absence of thrombocytopenia
- Causes of TMA can be difficult to distinguish clinically.
- Eculizumab is an effective treatment option for aHUS.

References: