# An Atypical HUStory: A Rare Cause of Renal Failure with an Effective Treatment

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## Introduction

Atypical hemolytic-uremic syndrome (aHUS) is a rare disease (incidence between 0.23 and 1.9 per million population annually and prevalence between two to ten per million population)[7] characterized by hemolytic anemia, thrombocytopenia, and acute kidney injury. Unlike shiga toxin producing Escherichia coli HUS (STEC-HUS), it is more difficult to recognize and diagnose. This often leads in a delay in diagnosis and subsequent end-organ damage, most commonly renal failure. [1, 4]

We report a case of a 33-year-old man who presented with this triad and was eventually diagnosed with aHUS.

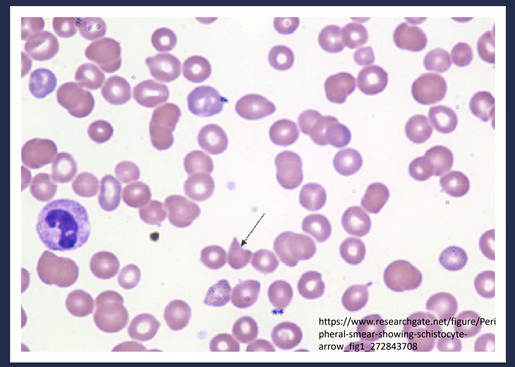
## Case Presentation

33-year-old man with hypertension presented with abdominal pain. He was found to have significantly elevated blood pressures, acute kidney injury (AKI), and thrombotic microangiopathy (TMA). He was recently admitted after a similar presentation, which was presumed due to hypertensive emergency after an unremarkable work-up. At that time, he was discharged on hemodialysis (HD) due to renal failure.
The clinical picture now was complicated by a recent bout of bloody diarrhea, concerning for STEC-HUS. Hematology was consulted upon admission.
Physical exam was largely unremarkable outside of hyperactive bowel sounds.

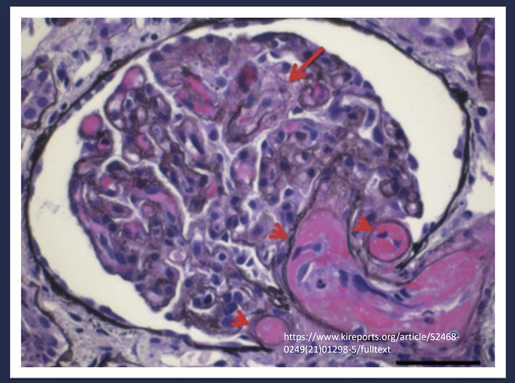
Pertinent results:
H/H 10.5/28, Plt 46
Cr 8.6, BUN 39
Tbili 2.9, haptoglobin undetectable, LDH 852
Peripheral smear: Microcytic anemia, frequent red blood cell fragments. Thrombocytopenia.
Vitamin B12 193
GI PCR panel and *C. difficile* testing negative
ADAMTS13 normal
CT abdomen/pelvis with evidence of diffuse enteritis

Given increasing concern for aHUS, genetic panel collected and high dose vitamin B12 started, as deficiency can manifest as a pseudo-TMA.

After vitamin B12 supplementation, hemolysis labs and clinical picture improved. Given improvement, C5 inhibitor therapy was held. Patient was discharged in stable condition but continued to require HD. aHUS genetic panel resulted with a variant linked to aHUS and Hematology initiated ravulizumab, a C5 inhibitor. Patient has been on therapy for almost 1 month and has continued to improve, though it is too soon to determine if he will experience substantial renal recovery.




Schistocytes (arrow)




Several glomerular thrombi (arrow)

## Discussion

aHUS is a rare condition causing a triad of hemolytic anemia, thrombocytopenia, and renal failure due to uncontrolled activation of the complement system. Oftentimes, aHUS requires an environmental "trigger," though not always necessary. Furthermore, the disease can present alongside severe hypertension, mimicking hypertensive emergency and leading to delayed diagnosis, much like happened in our case. [2, 3, 4, 6]

Given the high likelihood of progression to renal failure, timely diagnosis, urgent referral to Hematology, and initiation of one of the highly effective C5 inhibitor therapies is essential to saving the kidneys- and the patient. [5]

## Conclusions

- aHUS is rare but should remain on the differential of those presenting with the classic triad of thrombocytopenia, hemolytic anemia, and renal failure, as it is an important cause of morbidity and mortality if left untreated.
- Think twice before diagnosing with "just" hypertensive emergency if there is evidence of TMA.
- aHUS should be in the back of your mind!
- Get Hematology involved early if any concern for aHUS
- We have a treatment option that is effective (C5 inhibitor therapy) and relatively safe – we just need to diagnose.

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