



# Wilson Disease in a US Navy Pilot: Delayed Diagnosis with Drastic Career Implications

Zachary M Johnston, MD; Ryan Kwok, MD

Gastroenterology Service, Department of Internal Medicine, Walter Reed National Military Medical Center, Bethesda, MD



## BACKGROUND

Wilson disease is an autosomal recessive genetic disorder of the intracellular copper transport *ATP7B* leading to impaired copper excretion and subsequent accumulation in the liver, brain and cornea. Index symptoms vary widely and include neurologic and psychiatric abnormalities.

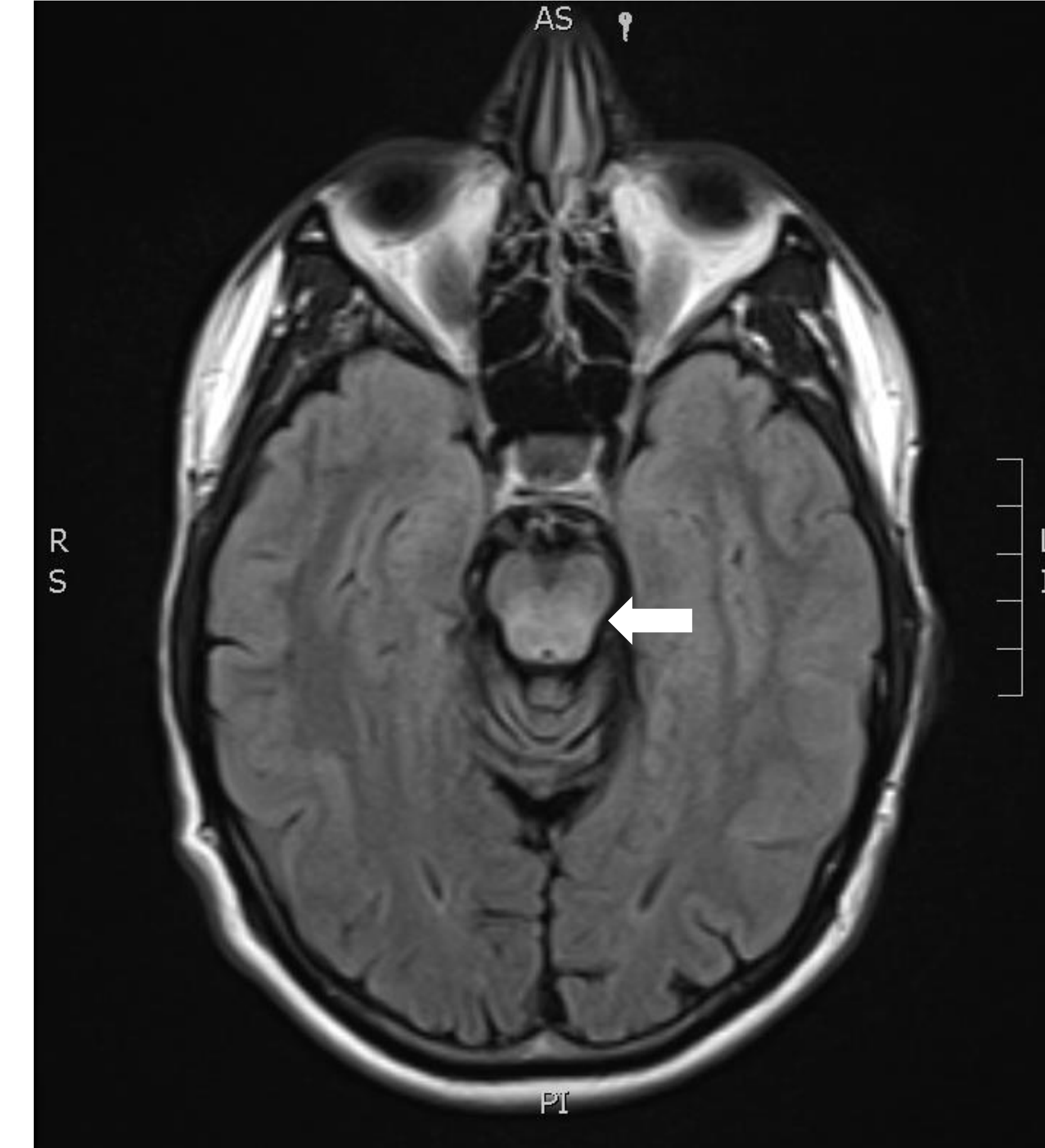
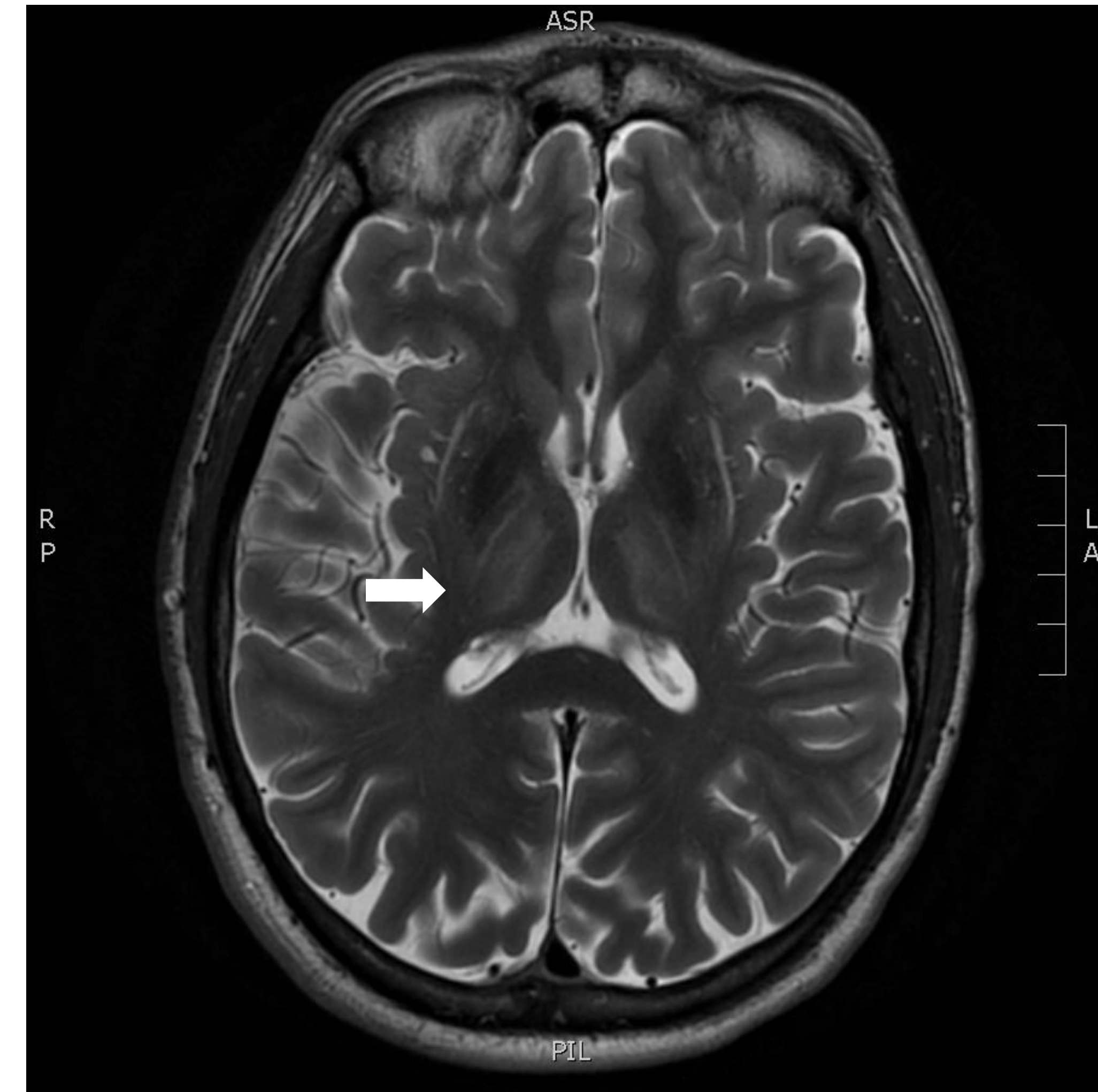
The wide array of subtle symptoms often lead to significant delays in diagnosis and thus prognostic implications for patients diagnosed with this rare disease.<sup>1</sup> In the Armed Forces, the diagnosis and clinical sequelae can have drastic career implications especially when neurologic sequelae are resistant to treatment.

## CASE PRESENTATION

A 28 year old US Navy pilot male presented to behavioral health clinic with complaint of anhedonia and depressed mood. He was diagnosed with major depressive disorder which was resistant to traditional treatment for over a year. He developed random muscle twitching of his neck and arms which was associated with initiation of an SSRI. He developed a hand tremor along with increased irritability. He had several concerning incidents occur including driving over a median and through several red lights. When he had difficulty landing an aircraft due these progressive symptoms, he was deemed incapable of flight.

Neurology was consulted and examination showed hyperkinetic non-stereotypic movements of his extremities with head and voice tremors. Lab work revealed ALT 52 and AST 48 and later, a ceruloplasmin of 6.7 (normal 16.0-31.0). A slit lamp exam was performed demonstrating Kayser-Fleischer rings. MRI brain showed abnormal T2 signal within the basal ganglia, thalamus and midbrain consistent with copper deposition. MRI liver was consistent with cirrhosis. A liver biopsy confirmed the diagnosis of Wilson disease with cirrhosis. He was started on trientene and zinc for chelation and his mood markedly improved.

However, his neurologic abnormalities have persisted despite ongoing therapy for over 4 years, permanently ending his piloting career.



## DISCUSSION

Our case highlights the importance of careful clinical consideration in patients presenting with psychiatric complaints. We aim to highlight this patient's response to therapy as the range of patient responses can be quite broad.

The majority of patients show significant or even total resolution of symptoms.<sup>3</sup> Our patient had significant improvement in mood, irritability and anxiety after chelation therapy but his neurologic symptoms did not recover.

Navy pilots are held to a very high medical standard. Strict regulations and medical evaluations are required to ensure the safety of pilots, crew and equipment.

Regrettably, the patient's Navy pilot career was brought to an early close given the severity of neurologic symptoms and lack of response and thus, inability to perform as an aviator.

## CONCLUSION

Prompt recognition of Wilson disease is paramount in preventing long term morbidity.

Patient symptom resolution is variable upon initiation of treatment.

A unique population, military pilots are expected to be in peak physical and mental form at all times. A diagnosis which significantly effects both is devastating to a pilot's career.

**"I feel absolutely grateful to have Wilson Disease. Don't get me wrong – it's a waking nightmare – but I have become a stronger, better man for surviving it. I got lucky. Not everyone does. I will never fly again, but I have found a higher calling."**

## REFERENCES

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3. Beinhardt S., Leiss W., Stättermayer A.F., et. al.: Long-term outcomes of patients with Wilson disease in a large Austrian cohort. Clin Gastroenterol Hepatol 2014; 12: pp. 683-689.