QUIZ 1

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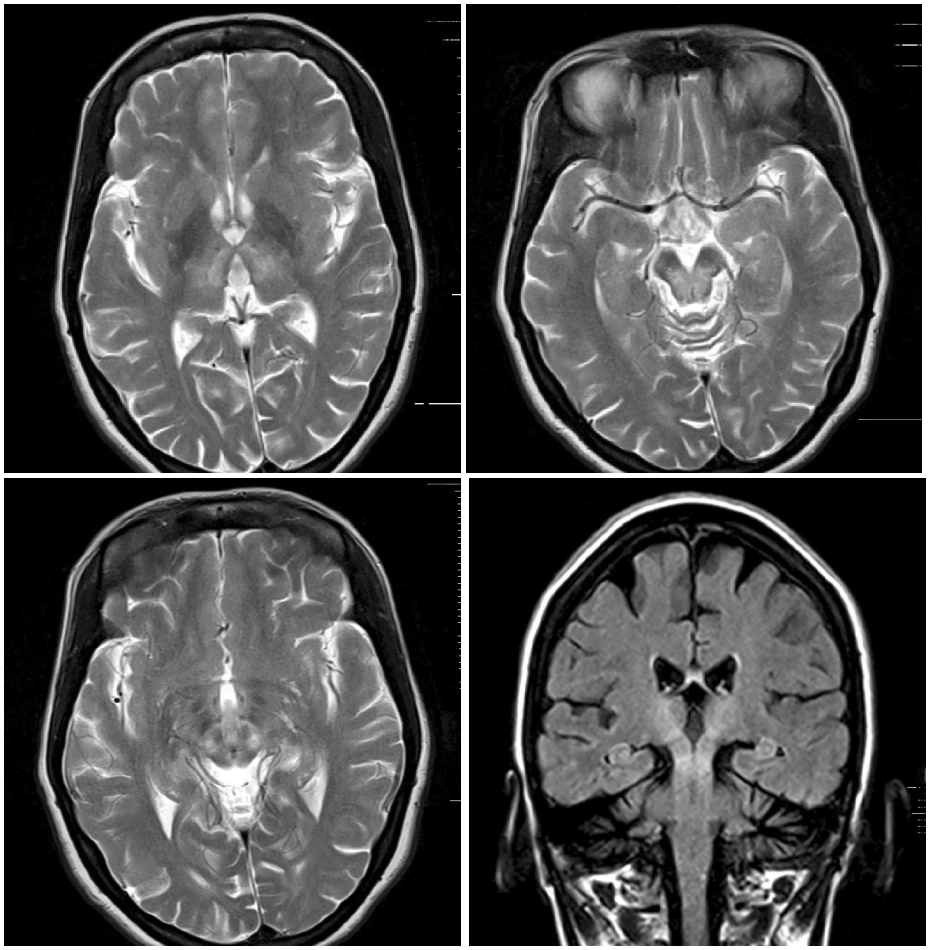
40 year old woman with jaundice and ataxia:



Questions

Q1. What are the findings on the MRI?

Q2. What is the likely diagnosis?

Q3. How is it confirmed clinically?

QUIZ 1

Answers

Answer 1: Hyperintense T2 signal from the thalami and the midbrain involving the red nucleus and the peri aquaductal grey matter. Low signals on T2 weighted image in the lentiform nucleus. The changes represent the deposition of manganese deposition in the basal ganglia due to chronic liver dysfunction and excess copper deposition leading to oedema in the basal ganglia.

Answer 2: Hepato lenticular degeneration (Wilsons's Disease)

Answer 3: Visualization of Kayser-Fleischer ring on the cornea and estimation of urinary copper excretion.

Discussion

This is a case of 40 year old woman who have two years history of memory loss and movement disorders.MRI Brain was done which shows hyperintense signals in the midbrain more marked in periaqueductal region and substantia nigra. Abnormal signals extending into thalamus. There is no diffusion or SWI abnormality and no post contrast enhancement. T2 hypointense signals in the putamen also identified. Based on the clinical and imaging findings, diagnosis of Wilson disease was made.

Wilson disease is autosomal recessive disease having inborn error of copper metabolism. Abnormal signals in the basal ganglia, thalamus and midbrain can be seen due to excessive copper throughout brain. These patients usually presents with asymmetric tremor, ataxia, dyskinesia and rigidity.

References

Wilson's Disease in Diagnostic Imaging: Brain, G Osborn, Salk Lake City USA. (AMIRSYS).