

## Early MR Imaging features of Wilson disease

Published on 12.03.2017

**DOI:** 10.1594/EURORAD/CASE.14530

**ISSN:** 1563-4086

**Section:** Neuroradiology

**Area of Interest:** Neuroradiology brain

**Procedure:** Imaging sequences

**Imaging Technique:** MR

**Special Focus:** Metabolic disorders Case Type: Clinical

Cases

**Authors:** Dr. Ashim Kumar Lahiri, Dr. Balraj Dhesi

**Patient:** 21 years, male

### Clinical History:

A young patient presented with acute onset of expressive and receptive dysphasia with associated personality changes. There was no relevant past clinical history, no infective features, no known metabolic or hereditary diseases and no history of any toxic exposure like carbon monoxide or methanol poisoning.

### Imaging Findings:

MRI brain revealed bilateral symmetrical T2 hyperintensity (Fig. 1 and 2) and T1 hypointensity (Fig. 3) of the putamen, globus pallidus and caudate nuclei. Gradient-echo images (T2\* GRE sequence) demonstrated low signal foci within the basal ganglia, suggesting iron or copper deposition (Fig. 4). The T2-fluid attenuated inversion recovery (Flair) images showed central hypointensity, suggesting presence of fluid content (Fig. 5). Thalamus showed very subtle T2-hyperintensity at the ventrolateral aspect (Fig. 1). The diffusion-weighted imaging, post-contrast sequences and MR venogram did not reveal any abnormalities. The mesencephalon, pons, cerebellar hemispheres and cerebral cortex were not involved. Considering the clinical history and MR findings with dominant finding of symmetrical bilateral basal ganglia involvement, the most likely possibility of metabolic disorder was considered. Other differentials which were felt less likely included: hypoxia, ischaemia, infection and primary CNS lymphoma. The diagnosis of Wilson's disease was subsequently confirmed after biochemical investigations.

### Discussion:

Wilson's disease is a rare genetic disorder characterized by deficiency of ceruloplasmin affecting copper metabolism, leading to excessive systemic copper deposition, particularly in liver and brain. The most frequent abnormalities reported in brain imaging are bilateral symmetrical T2-hyperintensity of the putamen and globus pallidus, followed by caudate nuclei, thalami, pons, midbrain, cerebellum and cerebral subcortical regions respectively [1-4]. The T2-weighted high signal changes represent oedema, gliosis, necrosis or spongiform cystic degeneration [2, 3]. Reversal of T2 signal changes can occur following copper chelating treatment [2]. Deposition of copper or iron ferritin can contribute to reduced signal on T2-weighted images. Kim et al reported that T1-weighted hyperintensity of the basal ganglia was the most common MR imaging finding in children which reflect hepatic involvement of the disease [2]. Diffusion restriction is generally seen in the early stages of the disease [1]. MR imaging can reveal a characteristic appearance of the mid brain called "face of the giant panda sign" due to high T2-signal in the tegmentum and normal signal of red nuclei [2, 3]. A similar sign is seen in the pons, albeit slightly smaller, aptly named the "face of the miniature panda" sign [3].

Bilateral symmetrical T2 hyperintensity of the basal ganglia in Wilson disease is most frequently associated with

signal changes within the thalami, followed by the brain stem [1-4], however, our case demonstrated dominant symmetric involvement of the basal ganglia only. This type of presentation with bilateral symmetrical hyperintensity of basal ganglia most likely suggests: neurotoxicity due to carbon monoxide, methanol or cyanide poisoning, hypoglycaemia or hyperglycaemia, extrapontine myelinolysis due to rapid correction of hyponatraemia, Wilson disease and other liver diseases, lymphoma, ischaemia and other rare genetic causes like mitochondria diseases, Leigh disease [1-4]. The association of symmetric thalamic involvement widens the differentials with conditions like stroke, primary CNS lymphoma and infections. Addition of T2\* GRE sequence can help in limiting the differentials with demonstration of hypointensities in basal ganglia which suggest deposition of iron or copper.

To conclude, the MRI features of bilateral T2 hyperintensity of basal ganglia as initial acute presentation of Wilson disease may not be typically diagnostic due to wide differentials. However, in the absence of toxic exposure or blood sugar disorder, no history of rapid electrolyte correction and presence of susceptibility changes at T2\* GRE MR imaging, raises the likelihood of Wilson disease.

**Differential Diagnosis List:** Wilson disease, Carbon monoxide poisoning, Extrapontine myelinolysis

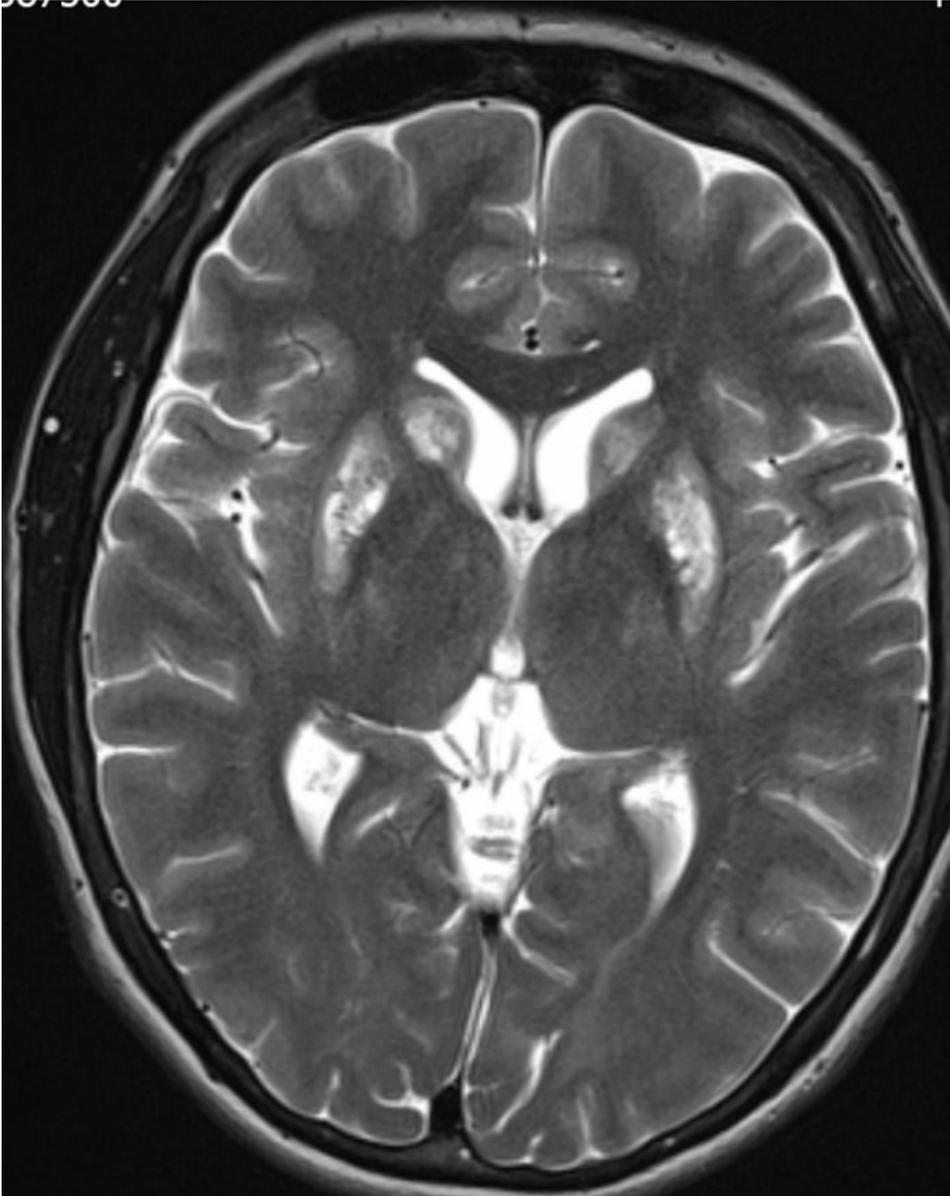
**Final Diagnosis:** Wilson disease

#### **References:**

- Beltz EE, Mullins ME (2010) Radiological Reasoning: Hyperintensity of the Basal Ganglia and Cortex on Flair and diffusion-Weighted imaging. AJR 195:S1-S8 (PMID: [20729404](#))
- Kim TJ, Kim IO, Kim WS et al (2006) MR Imaging of the Brain in Wilson Disease of Childhood: Findings Before and After Treatment with Clinical Correlation. AJNR 27: 1373-78 (PMID: [16775300](#))
- Singh P, Ahluwalia Am Saggar K et al (2011) Wilson disease: MRI features. J Pediatr Neurosci 6(1): 27–28. (PMID: [21977083](#))
- Hegde AN, Mohan S, Lath N et al (2011) Differential diagnosis for bilateral abnormalities of the basal ganglia and thalamus. Radiographics 31 (1): 5-30. (PMID: [21257930](#))

**Figure 1**

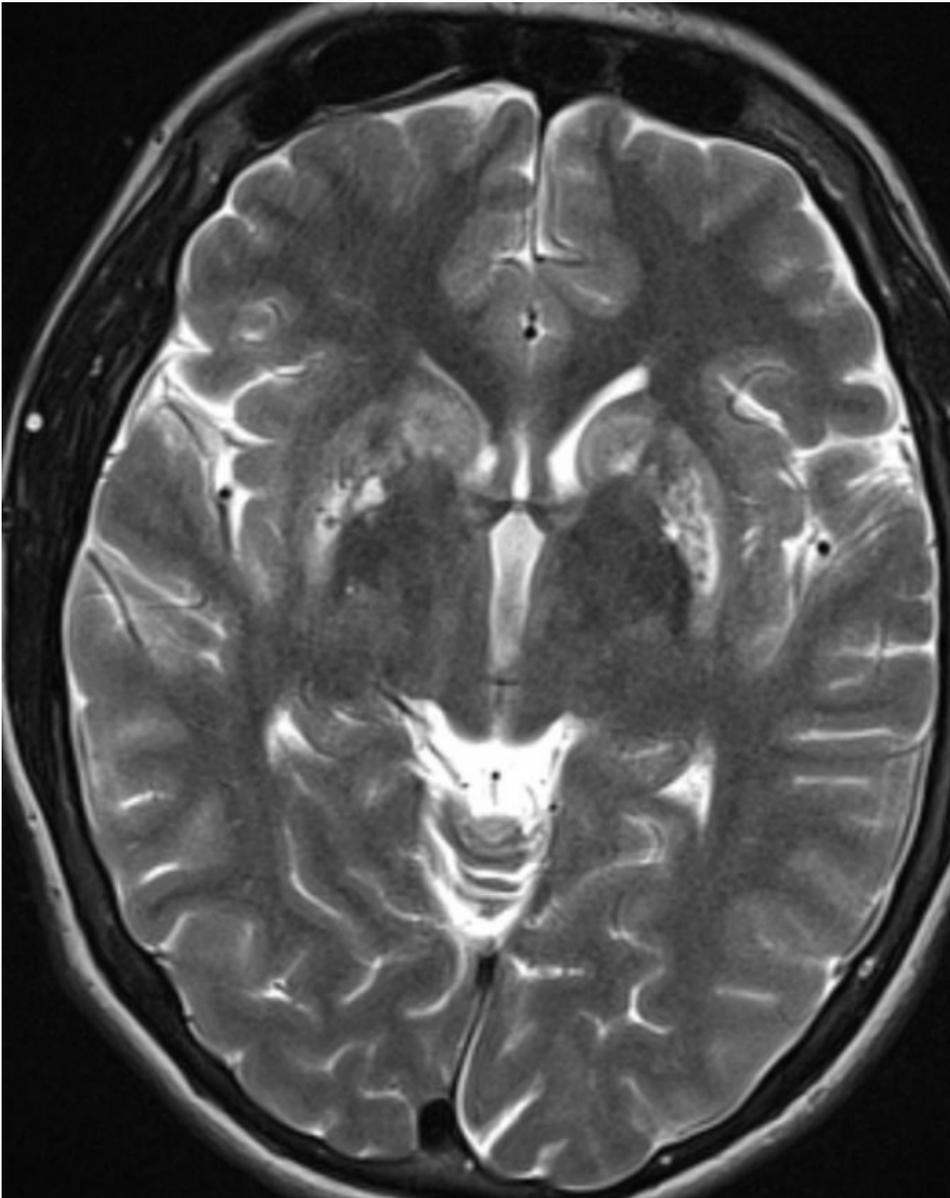
a  
507500



**Description:** T2-weighted axial images demonstrate bilateral symmetrical dominant hyperintensity of lentiform and caudate nuclei. Note of minimal signal changes in thalamus. **Origin:** Lahiri AK, Dhesi B; Department of Radiology, Worcestershire Royal hospital, Worcester, UK

**Figure 2**

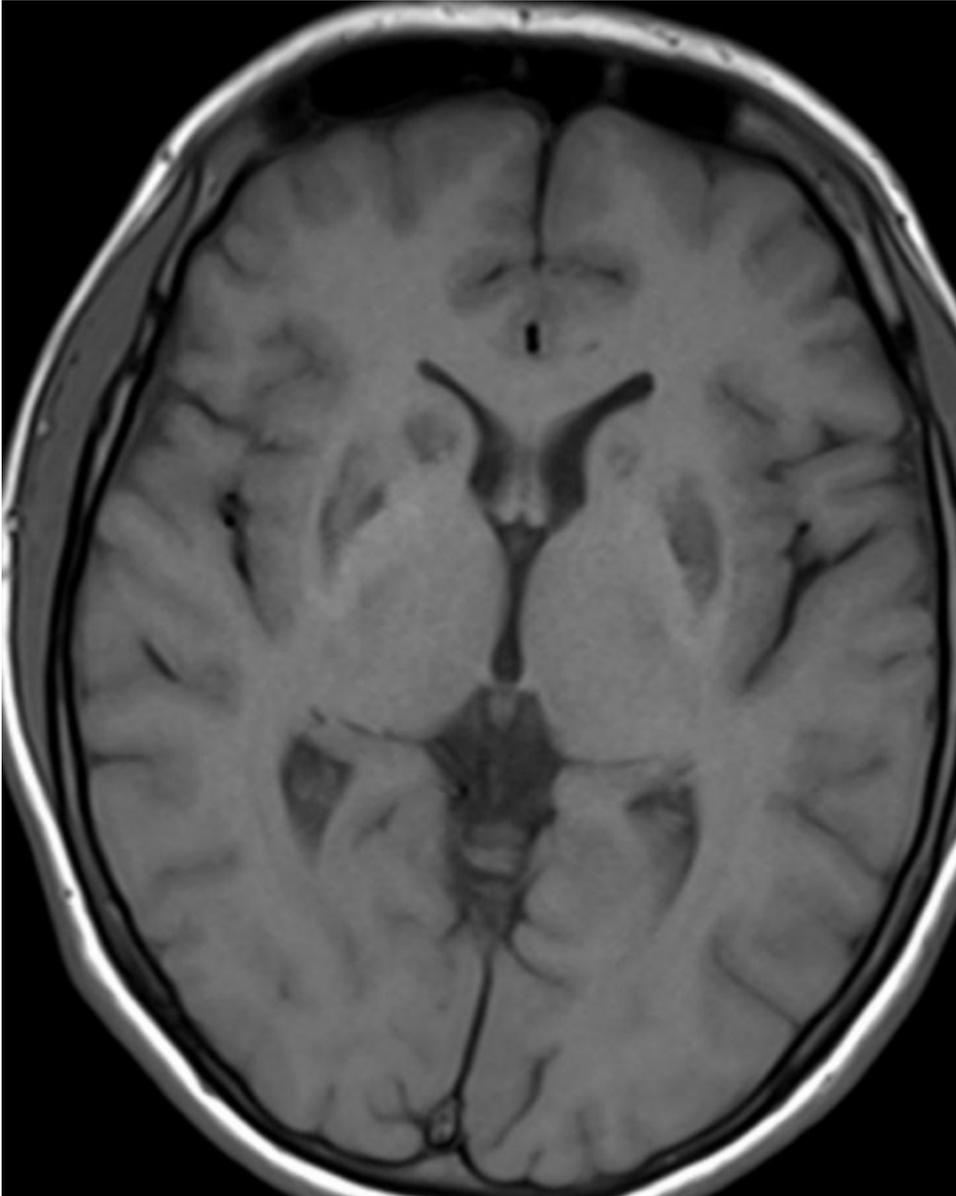
a



**Description:** T2-weighted axial images demonstrate bilateral symmetrical dominant hyperintensity of basal ganglia, mixed with small low signal foci. **Origin:** Lahiri AK, Dhesi B; Department of Radiology, Worcestershire Royal hospital, Worcester, UK

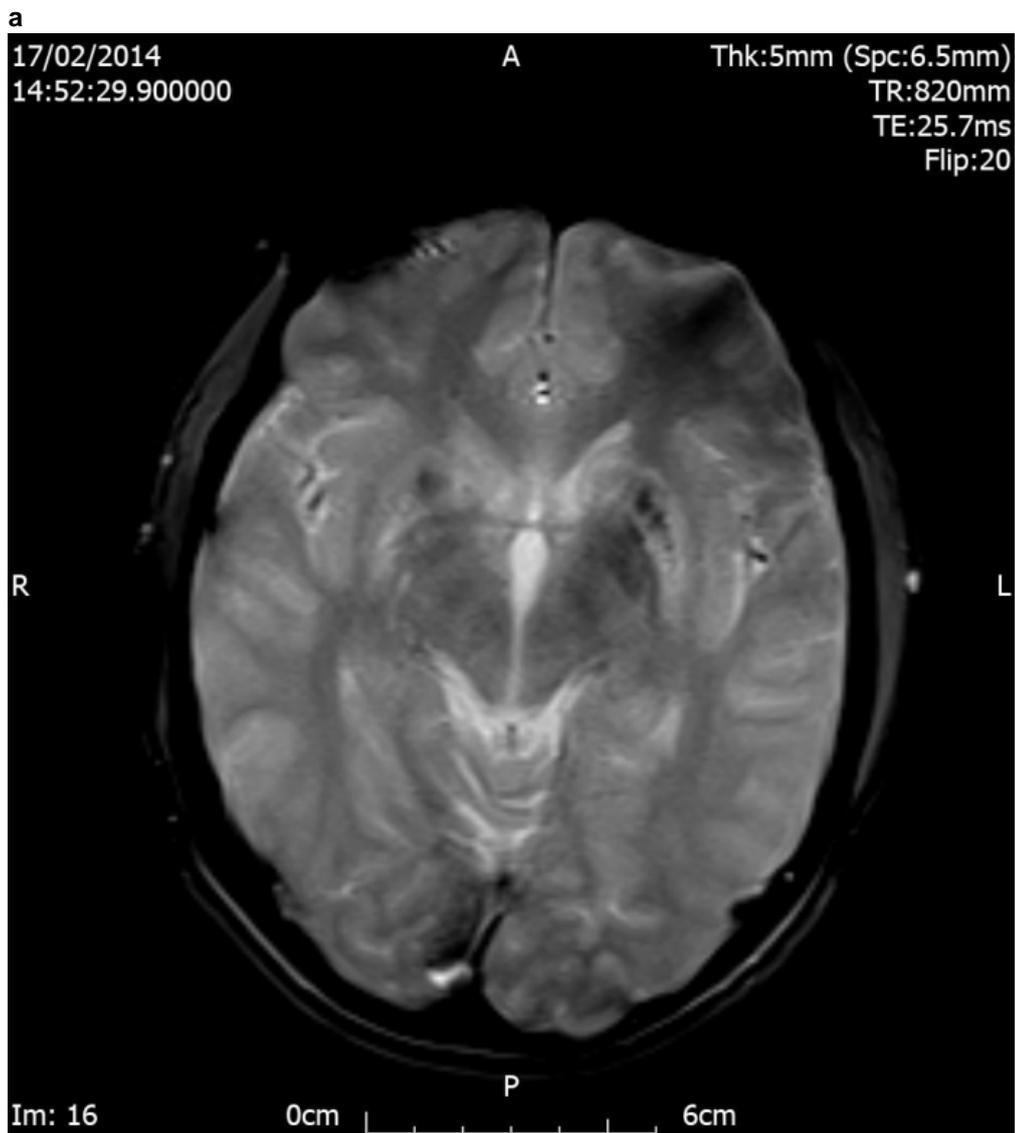
**Figure 3**

a



**Description:** Axial T1-weighted images show symmetrical reduced signal in basal ganglia. **Origin:** Lahiri AK, Dhesi B; Department of Radiology, Worcestershire Royal hospital, Worcester, UK

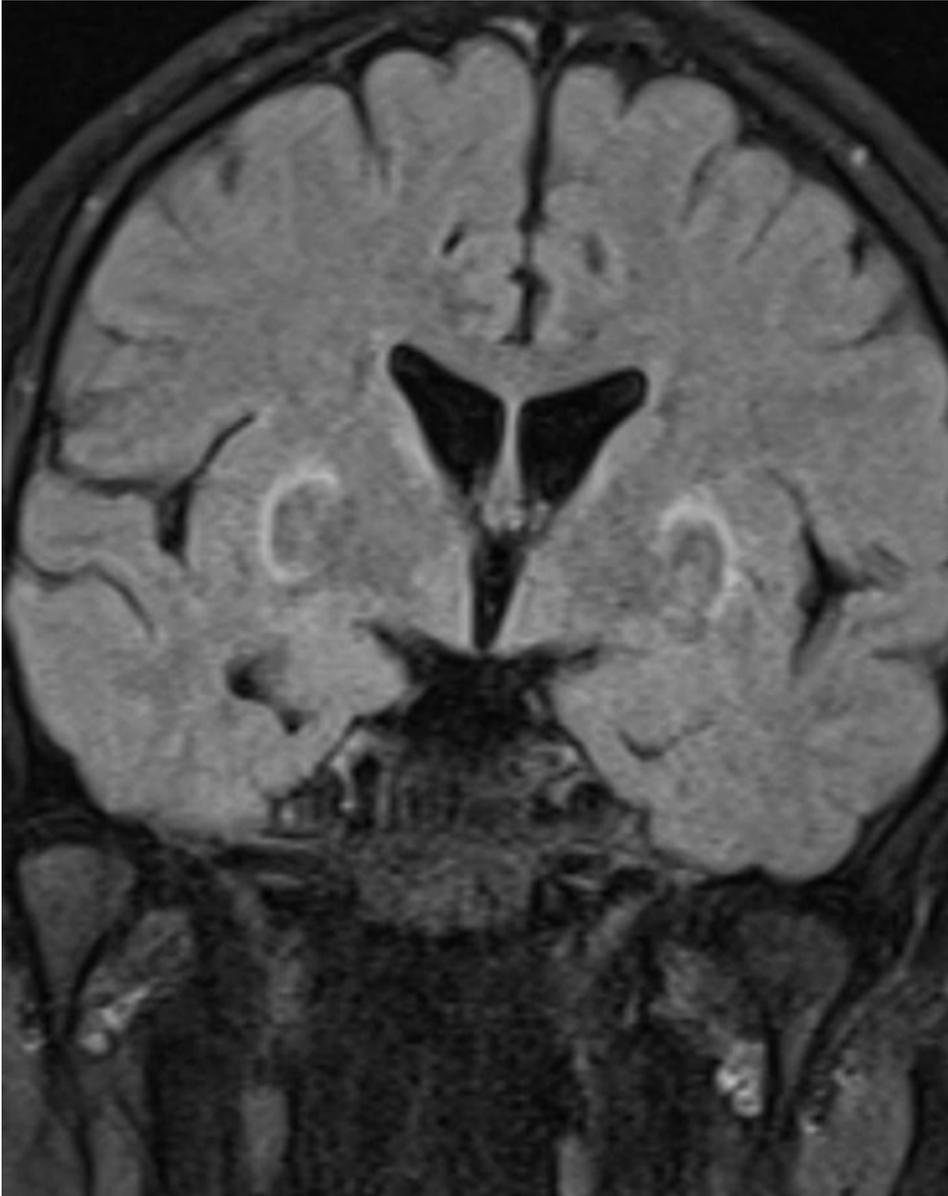
**Figure 4**



**Description:** Axial T2\* GRE images demonstrate susceptibility changes with focal hypointensities in putamen. **Origin:** Lahiri AK, Dhesi B; Department of Radiology, Worcestershire Royal hospital, Worcester, UK

## Figure 5

a



**Description:** T2-fluid attenuated inversion recovery (Flair) coronal images reveal central reduced signal with peripheral hypointense rim, suggesting oedema/fluid contents. **Origin:** Lahiri AK, Dhesi B; Department of Radiology, Worcestershire Royal hospital, Worcester, UK