Wilson disease is an inborn copper metabolism disorder with autosomal recessive pattern of inheritance caused by ATP7B mutation leading to abnormal accumulation of copper in various tissues, particularly in the liver and the brain. Presence of magnetic resonance imaging (MRI) abnormalities occur in virtually 100% of patients with neurological dysfunction among the various MRI findings face of giant panda sign, face of miniature panda sign and bright claustrum signs are characteristics. We describe a 16 year old boy presented with limb dystonia, tremor, and cognitive impairment with positive family history of willson’s disease. Besides the other diagnostics marker such as serum copper, ceruloplasmin and urinary copper he had characteristics MRI findings (Figures 1 A, B and C). Figure 1A showing face of giant panda sign i.e high signal intensity of T2W image in the tegmentum sparing the red nucleus (eyes, black thin arrow), preservation of signal intensity of the lateral portion of the substantia nigra (Face of Giant panda sign) (B) T2W image showing thin rim of hyperintensity in claustrum (bright claustrum sign) (C) hypointensity of MLF and central tegmental tract hyperintensity of aqueduct opening into the fourth ventricle (face of miniature panda sign).

Some patients also have characteristics pontine lesion i.e. face of miniature panda sign (Figure 1C) where hypointensity of MLF and central tegmental tract form the eyes, hyperintensity of aqueduct opening into the fourth ventricle form the nose and mouth and the superior cerebellar peduncle form the panda’s cheeks. Inspite of the presence of excess copper within the brain, pathologic findings are limited primarily to the basal ganglia, thalamus, mid brain and pons. Histopathologic studies showed atrophy, spongy softening, cavitation, neuronal loss, increased cellularity, and the presence of Opalski cells. Though the classical neuroimaging findings are found only in few proportions of patient but its presence increases the possibility of diagnostic value and treatment related prognostication.

References