Fabry disease with lenticular degeneration without pulvinar sign

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Background: FD is an X-linked lysosomal storage disorder caused by a GLA gene mutation. The most frequent neuroimaging finding is non-specific T2 HSI in the periventricular white matter while the most specific MRI sign of brain involvement in FD is HSI in the bilateral pulvinar on T1WI. Case Report: A 38-year-old man visited the neurology department with a tingling sensation in both the upper and lower extremities in 2016. He had been on hemodialysis since 2009 due to end-stage renal disease (ESRD). He was diagnosed with Fabry disease (FD) via GLA gene testing [c.902GA (p.Arg301Gln) hemizygote]. The patient had been stable with regular hemodialysis. Neurologic examination showed no abnormal findings except decreased deep tendon reflexes. Brain MRI revealed high signal intensity (HSI) in the bilateral lentiform nuclei on T1-weighted imaging (T1WI), of which the core lesion was iso-intense. The core of the lesion showed low signal intensity (LSI) on T2-weighted imaging (T2WI) and diffusion-weighted imaging. There were no abnormal signal intensities in either thalamus. No other significant findings, such as cerebral atrophy or periventricular white matter changes suggesting cerebral small vessel disorders, were observed. Conclusion: In the present case, lesions were unexpectedly found in the lentiform nuclei with a similar appearance to the pulvinar signs observed in previous studies. The present case described an unusual neuroimaging finding of FD. Further observations are needed to determine whether FD should be included in the differential diagnosis of bilateral T1 hyperintensities in the lentiform nuclei.