Clinical Image

Apathy as a Major Symptom of CADASIL and MRI Findings

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Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) is one of the inherited cerebrovascular disease which is autosomal dominant and it is clinically characterized by variable manifestations of migraine like headache, recurrent transient ischemic attack or lacunar strokes, and cognitive decline [1] and apathy is associated with cognitive impairment, global functional disability and various neuropsychiatric symptoms which are shown in the course of the disease.

A 46 year-old man, right handed, admitted to our emergency unit with complaints of sudden onset of forgetfulness, meaningless behavior and difficulty in finding words, decrease in speech and

decrease in interest to environment. After evaluating of clinic, examination, MR imaging findings and family history, it was thought the pre-diagnosis of CADASIL and NOTCH3 gene mutation analysis was sent and analyses revealed heterozygous mutation in p.Arg90Cys in 3 exon(Figure 1).

Diagnosis of patients with CADASIL relies mainly on MRI findings. White matter hyper intensities (WMHs) end to be symmetrical and bilateral, distribute in the periventricular and deep white matter. The anterior temporal lobe which is higher specificity-sensitivity and external capsules are the predilection sites for WMHs. The other lacunar infarcts are in the semi oval center, basal ganglia, thalamus and pons. In addition, presence of WMHs probably associated with lacunar infarction while cerebral micro bleeds may be present in both symptomatic and asymptomatic [2].

References

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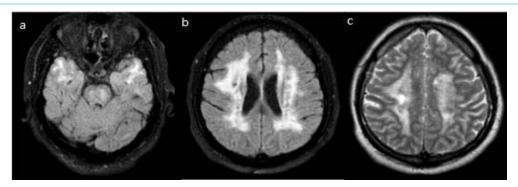


Figure 1: a) MRI axial FLAIR image shows anterior temporal poles hyper intensities b) MRI axial FLAIR image shows periventricular hyper intensities c) MRI axial T2-weighted image shows hyper intensities in the deep white matter.