Pictorial CME

Those white Spots in the Brain

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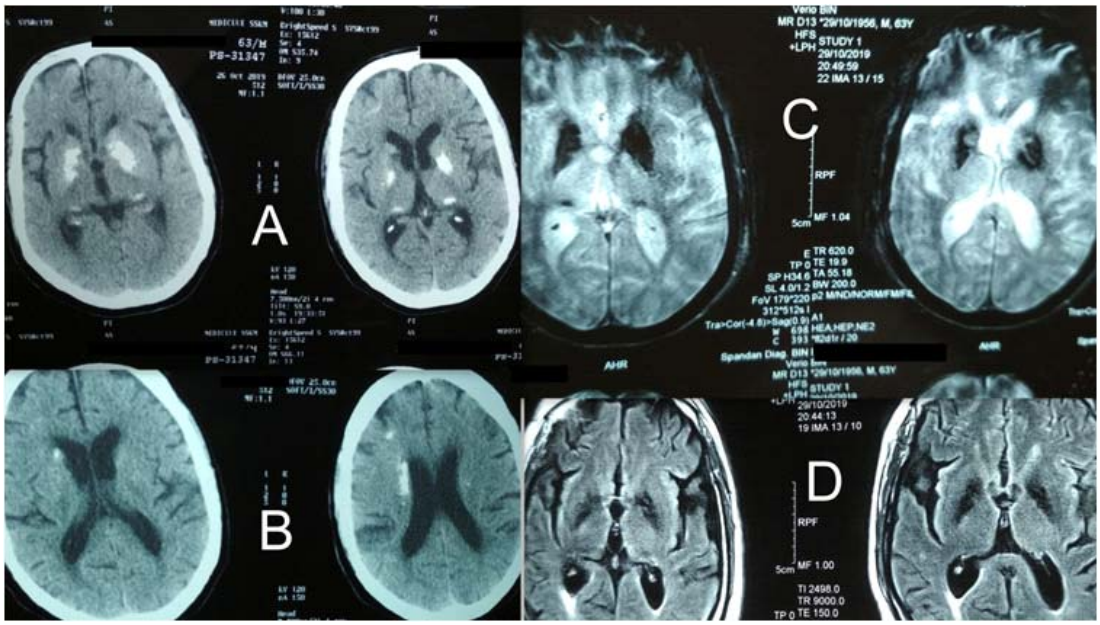

Fig 1 — CT scan of brain (Panels A and B) showing extensive calcification in bilateral basal ganglia and periventricular regions. Panels C and D show the MRI appearances with blooming in GRE

A 63-year-old man was admitted with sudden onset dizziness. At admission, a CT Scan of brain was done. When certain abnormalities were found in that scan, an MRI scan was also done. Both are shown in Fig 1. The patient had since recovered with no neurodeficit. Blood tests done at the time of hospital stay were all normal.

(1) What is the diagnosis?
(2) What are the usual clinical features?
(3) What are the genetic links of this condition?

Answers :

(1) The brain imaging shows areas of bilateral calcification in the brain, especially in and around the basal ganglia. In view of the normal blood tests, this is most probably idiopathic brain calcification or Fahr disease.

(2) As in our case, the patients with limited cerebral calcification may be completely asymptomatic. However, in more severe cases, there may be extrapyramidal features with motor impairment, dementia, dysarthria or movement disorders like athetosis. Neuropsychiatric manifestations may be the dominant presenting symptom in many cases. In addition to the periventricular region (as shown in this Figure), the calcification spots may be located in the cerebellum, hippocampus, thalamus etc. Many patients are asymptomatic in childhood or young adulthood and present with progressive neurodeficits in the 4th and 5th decades of life.

(3) The disease may be sporadic or may be inherited in autosomal dominant manner. The exact gene responsible for the syndrome in unidentified in 50% of the cases. However, some candidate genes which have been proven to be associated with the disease include PDGFB, PDGFRB, SLC20A2 (Chr. 8), XPR1 etc.

References

1 Zhang Y, Guo X, Wu A — Asssociation between a novel mutation in SLC20A2 and familial idiopathic basal ganglia calcification. *PloS one* 2013; **8:** e57060.

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