Fahr’s Disease: A rare neurodegenerative disorder

Fahr’s disease refers to a rare syndrome characterized by symmetrical and bilateral intracranial calcification. While bilateral striopallidodentate calcinosis is commonly referred to as ‘Fahr’s disease’, there are 35 additional names used in the literature for the same condition. Clinically, Parkinsonism or other movement disorders appear to be the most common presentation, followed by cognitive impairment and ataxia. We present a case of striopallidodentate calcinosis presenting with ataxia and fall.

Key Words: ataxia, calcinosis, Fahr’s disease, parkinsonism

Bilateral striopallidodentate calcinosis (BSPDC), commonly known as Fahr’s disease, is a rare syndrome characterized by symmetrical calcification over the basal ganglion and dentate nucleus. The basal ganglia are the most common site of involvement.

Case Report

A 63-year-old gentleman presented to the Emergency room with history of unprovoked fall down the stairs two hours prior to presentation. There was no history of loss of consciousness, seizure or external bleed. On examination he had a GCS of 15, and had ataxia and positive finger nose test and heel-shin test. A plain computed tomography (CT) scan was ordered suspecting intracerebellar bleed. The CT scan revealed bilateral symmetrical basal ganglia and bilateral cerebellar calcifications (Figure 1). He had normal blood levels of parathyroid hormone, thyroid stimulating hormone, calcium, phosphate and iron studies.

Discussion

Fahr’s disease (or bilateral striopallidodentate calcinosis) is a rare neurodegenerative disorder of
unknown cause characterized by symmetrical calcium deposition in the brain. The German pathologist Karl Theodor Fahr first described it in 1930. Pathological studies show that calcium is the major element present and it accounts for the radiological appearance of the disease. Mucopolysaccharides, traces of aluminum, arsenic, cobalt, copper, molybdenum, iron, lead, manganese, magnesium, phosphorus, silver, and zinc are also present. Calcium and other mineral deposits were found in the walls of capillaries, arterioles, and small veins and in perivascular spaces. The most common manifestation of BSPDC is movement disorders like Parkinsonism. In addition, cognitive impairment, psychiatric manifestation, seizures, cerebellar signs and dysarthria can be seen. Laboratory studies are important to rule out underlying calcium metabolism disorders such as hypoparathyroidism. Manyam has proposed a classification based on anatomical sites of calcium deposits.

Radiographic studies show symmetrical calcification in the cerebellum, basal ganglia, thalamus and centrum semiovale. While CT seems to have greater specificity for basal ganglia calcification, MRI findings correlate better with functional impairment. Treatment is supportive with symptomatic management. Disodium etidronate showed symptomatic benefit without reduction in calcification.

References

5. Manyam BV. What is and what is not ‘Fahr’s disease’. Parkinsonism Relat Disord; 11:73–80, 2005