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RESEARCH ARTICLE

A RARE CASE OF FAHR'S SYNDROME

***Dr. Puneet S. Shirbur and Dr. Vishal S. Nimbal**

Department of Radiology, Shri B.M Patil Medical College, Hospital and Research Centre, Bijapur

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ABSTRACT

Fahr's syndrome is a rare condition presenting with symmetrical bilateral intracranial calcifications. A 62 year old female presented with psychosis and generalized tonic-clonic seizures. Her neurological examination was normal. Computerized Tomography of brain revealed extensive symmetrical calcifications in bilateral basal ganglia, thalami, bilateral centrum semiovale subcortical white matter, pons and dentate nuclei.

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INTRODUCTION

Fahr's syndrome is a genetically dominant, degenerative disorder characterized by calcifications in brain parenchyma with subsequent neuronal loss. Presence of bilateral and symmetrical intracerebral calcifications in the basal ganglia, thalamus and centrum semiovale region due to unknown aetiology is referred to as Fahr's disease. Though the disease can present in childhood or adolescence. The usual age of presentation is 4th–5th decade with basal ganglia and dentate nucleus being the most common site of involvement. About 40% of patients with basal ganglia calcification initially present with psychiatric features like cognitive, psychotic, and mood disorders are common (Avrahami *et al*., 1994). More extensive calcification and subarachnoid space dilatation are known to correlate with the presence of psychiatric manifestations (Cummings *et al*., 1983).

MATERIALS AND METHODS

The scan was performed using Philips Brilliance 6 slice Computerized Tomography machine.

****Corresponding author: Dr. Puneet S. Shirbur***
Department of Radiology, Shri B.M Patil Medical College, Hospital and Research Centre, Bijapur

DISCUSSION

Fahr's disease is a condition characterized by symmetrical intracranial calcification with a predilection for the basal ganglia and dentate nuclei. The condition that has been closely described with it is primary hypopara-thyroidism (Cummings, 1985 and Konig, 1989). Other causes include lupus, tuberous sclerosis, Alzheimer's disease, myotonic muscular dystrophy and mitochondrial encephalopathies (Lauterbach *et al*., 1998). When there is no explainable cause for striopallidodentate calcinosis, the condition is termed as Fahr's disease. Genetic studies have revealed an autosomal dominant inheritance in the familial cases (Manyam *et al*., 2001). One multigenerational family with linkage to the IBGC1 of chromosome 14 has been identified but the causal gene is still unknown. Genetic studies on other families did not replicate this result. Symptoms can include features of psychiatric disorders, epileptic seizures and dementia (Modrego *et al*., 2005). But other presentations like syncope and pseudohypoparathyroidism have also been reported (Ones *et al*., 2008). Paranoid and psychotic features often present between the ages of 20 and 40 in Fahr's disease (Preusser *et al*., 2007). Two patterns of psychotic presentation in FD are known, including early onset (mean age 30.7 years) with minimal movement disorder and late onset (mean age 49.4 years) attended by dementia and movement disorder.

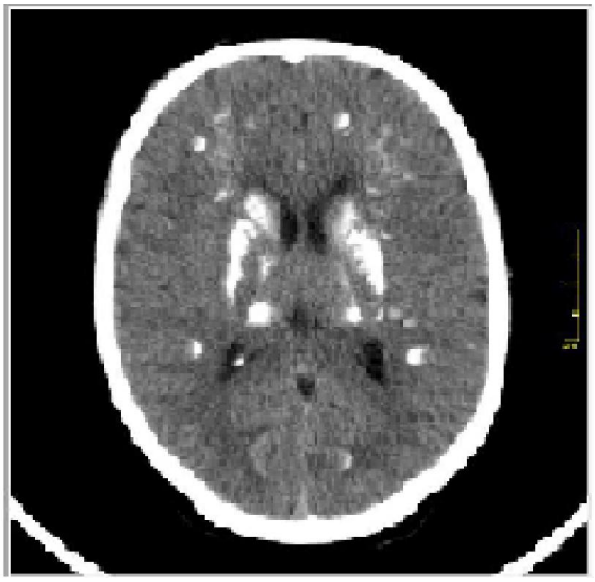

Fig. 1. Calcifications in bilateral basal ganglia and thalami

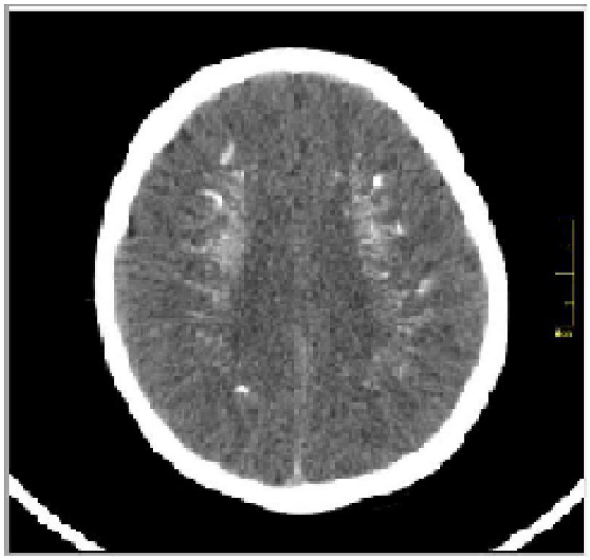

Fig. 2. Calcifications in bilateral centrum semiovale

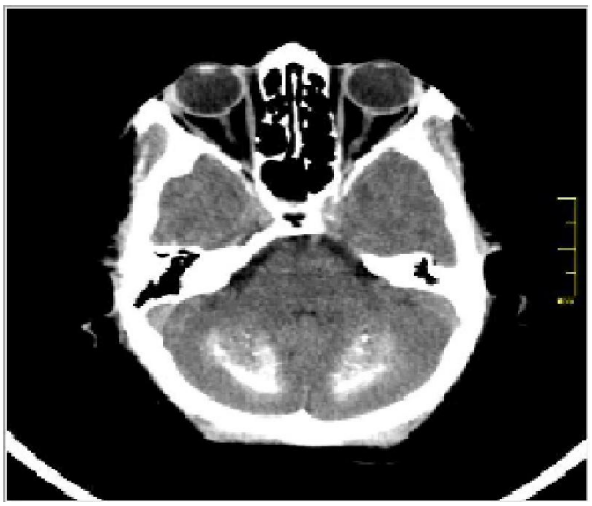

Fig. 3. Calcifications in bilateral cerebellar peduncles and dentate nucleus

Calcifications consist of hydroxyapatite variety, other elements including zinc, iron and magnesium may also be present (Scotti *et al*., 1985). Because of this material composition, they are always hyperdense on CT. On magnetic resonance imaging (MRI), however, their signal is variable. On T1 weighted images, low signal is due to the low proton density of calcium and other mineral ions present in higher concentration. However, they might present hyperintense signal, due to proteins and mucopolysaccharides binding the mineral ions. They might also go undetected on MRI when in an intermediary stage (Simone *et al*., 2008). T2 GRE magnetic resonance images (MRI) sensitively demonstrate areas of calcification as areas of low signal. The usefulness of 99mTc-HMPAO brain perfusion SPECT in deciding clinical approach to Fahr's syndrome has been suggested (Smeyers-Verbeke *et al*., 1975). To summarize, though Fahr's syndrome is a rare entity, it should be suspected in patients with neuropsychiatric disturbances and seizure disorder. Routine biochemical investigations should always be performed to rule out metabolic causes. Conversely, all patients with incidentally detected striopallidodentate calcinosis should be subjected to thorough neuropsychiatric examination and if required, biochemical tests. Knowledge of the associated conditions will not only help to rectify the treatable cause but will also prevent unnecessary treatment in others.

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