CASE REPORT

FAHRS DISEASE:
A YOUNG GIRL PRESENTING WITH FITS

A. SABEEN RAHMAN*, SHAISTA AHMED, TANVEER AHMED AND TAHIR HUSAIN
Karachi Medical and Dental College and Abbasi Shaheed Hospital

ABSTRACT

Fahr’s disease refers to a rare syndrome characterized by symmetrical and bilateral intracranial calcification. We describe a 28 year’s old unmarried girl who presented with fits, anxiety, and panic attacks. She had bilateral symmetrical intracranial calcification. She was proved to have hypoparathyroidism and pleural tuberculosis.

Keywords: epilepsy, intracranial calcification.

INTRODUCTION

Fahr’s disease has been described for the first time by Fahr in 1930 (Gulsan et al., 2006 and Malik et al., 2004). It is a rare clinical entity consisting of certain metabolic, biomechanical, neuroradiologic and neuropsychologic phenomena in which extrapyramidal phenomenology clinically dominates (Mustafa et al., 2007). The disease is characterized by symmetric calcifications of dentate nucleus and white matter. In the etiology, metabolic diseases, hypoparathyroidism, hyperparathyroidism, mitochondrial cytopathy, infectious diseases, epstein barr virus infections, tuberculosis and AIDS may play a role (Gulsan et al., 2006). It usually appears between 40-60 years of age. However it may also be rarely seen in children (Gulsan et al., 2006, Malik et al., 2004, Cummings et al., 1983). According to reports in medical literature, Fahr’s disease is often familial. It is believed to have autosomal dominant inheritance but a few cases have been reported to have autosomal recessive inheritance and even sporadic cases have been reported in literature.2 It may present with an array of clinical aspects (Gulsan et al., 2006). The clinical course of the disease has a degenerative component. There may be mental and motor disability which accompanies epileptic syncope, increased neuromuscular excitability and tetany, paresthesia, intracranial calcification and cataract. The mineral deposition may lead to cell loss in the cerebral cortex, basal ganglia, dentate nucleus and subthalamus (Gulsan et al., 2006 and Foley, 1951). Neuropsychiatric features comprise of apathy with intermittent disinhibition, anxiety, irritability, frequent mood changes, ritualistic and antisocial behavior and psychosis (Benke et al., 2004).

Case Report

Our patient is a 28 year’s old unmarried female who was born via spontaneous vaginal delivery to a healthy mother and father as a second child. She had a history of fits for 20 years, and was taking phenobarbitone 60 mg twice a day and carbamazepine 200 mg twice a day. Despite taking these medicines she had one to two episodes of fits per day. Her fits were tonic clonic, with loss of consciousness, tongue bitting and urinary incontinence. She was admitted with increased frequency of fits when her anti-epileptics were changed from carbamazepine to sodium valproate 4-5 days back. Whenever she used to have fits she was given injection diazepam intravenously which used to halt her fits. She also had episodes of...
anxiety, panic attacks and depression and had a history of fever (intermittent and low-grade) for one year with weight loss. There was no associated cough, dyspnea, palpitations or sweating. She had a past history of incision and drainage of an abscess at her right hip one month back and was transfused one pint packed cells. She also had cataract surgery of both eyes at the age of thirteen years.

Her family history did not reveal any similar illness. She had diabetes and hypertension in her first degree relatives. No history of tuberculosis in her family and had no history of addictions or allergies. Her sleep, diet, bowel habits and menstrual cycles were normal and had normal milestones. She was normally studying in school with her other siblings till the age of eight years when she started having fits and was socio-economically poor.

On examination, she looked ill and depressed, well oriented having pallor and ankle edema. Her chest examination revealed findings of left sided pleural effusion (reduced chest movements, reduced vocal fremitis, stony dull percussion, reduced vocal resonance and absent breath sounds) which was consistent with her X-ray chest. Her gait was broad based and ataxic. Her memory, motor and sensory system were intact with normal fundi.

On the basis of her history and physical examination she was started on anti-tuberculous therapy empirically on which she responded and her general health and pleural effusion gradually improved.

Her investigations revealed haemoglobin of 9.9 mg/dl (normocytic normochromic picture) with normal white cell count and platelets, her erythrocytic sedimentation rate was 66, urea, creatinine electrolytes, liver function test, blood sugars and urine detailed report were normal. Her serum calcium was 6.9 mg/dl, serum albumin was 2.7 gm/dl, and serum phosphate was 6.5 mg/dl. Her corrected calcium was found to be 7.94 mg/dl. Her serum parathormone levels were low (less than 3 pg/ml), pleural fluid showed an exudative picture with predominantly lymphocytes. Her pleural biopsy was done which was consistent with pleural tuberculosis. Her electrocardiogram and ultrasound pelvis was normal. Her EEG was reported as normal.

CT scan brain showed diffuse, extensive, bilateral symmetrical calcifications seen in basal ganglia, thalami, cerebellum, white matter and periventricular region.
basal ganglia, thalami, cerebellum, white matter and periventricular region (Figs. 1 and 2).

**DISCUSSION**

Fahr’s syndrome involves calcification of basal ganglia and dentate nuclei of the cerebrum. Clinically it may present with an array of movement disorders, dementia and other behavioural disturbances. Sporadic and familial cases have been reported with or without calcium, phosphorus metabolism (Modrego et al., 2005).

Here we have reported a single case of a 28 years old unmarried female with fits, anxiety and panic attacks. She was proven to be hypoparathyroid and had pleural tuberculosis and had responded to antituberculous therapy. Brain C. T showed Fahr-type calcifications in basal ganglia, cerebellum, thalami and periventricular region. Our case had no family history of similar illness and was of sporadic type.

Clinical expression of Fahr’s syndrome varies greatly. Symptoms include psychiatric disorders, epileptic seizures, and extrapyramidal syndrome and various neurological conditions. Diagnoses require CT brain scan which identifies calcium deposits in the basal ganglia. The main cause is hypoparathyroidism, whether primary or post-operative. Cases due to other causes of dysparathyroidism are rare. The pathophysiology of this condition remains unknown and results of treatment are often unsatisfactory. Since correcting the impaired calcium phosphorus metabolism often leads to considerable improvement, it is essential to systematically search for dysparathyroidism in patients presenting with neuropsychologic manifestations associated with calcifications of the basal ganglia (El-Maghraoui et al., 1995).

Fahrs disease is a rare clinical entity most commonly involving the basal ganglia, and most cases present with extra-pyramidal symptoms. Fahrs disease has been reported with prominent frontal lobe symptoms like uncontrollable bursts of laughter and crying spells, with dysarthric speech and choreoathetoid movements. Imaging has also revealed progressive cerebral atrophy. As frontal lobe symptoms are usually in conspicuous in the early stage, the presence of the symptoms might be overlooked in clinical practice when compared with those suffering from prominent movement disorders (Lam et al., 2207).

The diagnostic value of EEG has been evaluated in patients with fahrs syndrome, patients of a family history with hereditary occurrence of the syndrome and other patients where the characteristic calcifications were discovered by chance or in C.T screening of persons who had undergone thyroidectomy. The comparison of the EEG results yielded no discernable diagnostic value of this investigation. All kinds of alterations of central electrophysiological activity seen possible, but no characteristic EEG pattern associated with Fahrs syndrome was seen. Only a vague correlation of alterations of the age of the patient could be surmised, this being wholly non-specific. Thus psychopathological, neurological data and C.T are the prime techniques for diagnosis of fahrs syndrome (Schmid et al., 1986).

**REFERENCES**


