Case Report

A rare case of Fahr's disease presenting with seizure disorder

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Fahr's disease also known as striopallidodentate calcinosis is a rare neurodegenerative disorder characterised by bilateral and symmetrical calcium deposits in different areas of the brain, particularly in the basal ganglia. We report an unusual case of Fahr's disease in a middle aged male who presented with seizure disorder which is again a rare presentation of Fahr's disease. The patient was diagnosed as a case of idiopathic calcification of Basal ganglia or Fahr's disease based on clinical and radiological criteria in the absence of biochemical abnormalities.

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Fahr's disease is a rare clinical entity consisting of certain metabolic, biochemical, neuroradiologic and neuropsychologic phenomenon characterized by symmetric calcifications of white matter of brain. The globus pallidus within the basal ganglia is the most frequent site of calcification, but deposits may be present in putamen, caudate nucleus, internal capsule, dentate nucleus, thalamus, cerebellum and also cerebral white matter. Fahr's disease may be sporadic or familial and may lead to neurological, psychiatric and cognitive abnormalities. Familial Fahr's disease may have autosomal recessive or autosomal dominant transmission. The expressivity within the family is variable and the age of the onset is decreasing in the family in case of the autosomal dominant inheritance.

Fahr's disease has been described for the first time by Fahr (German neurologist) in 1930 of a man with seizures and diffuse calcifications of the brain vessels and vessel ganglia[1]. Fahr's disease should be differentiated from Fahr's syndrome which is defined as symmetric and bilateral calcification of the basal ganglia associated with neuropsychiatric manifestations that preferentially occur in patients with parathyroid disorder, especially hypoparathyroidsm. Headache, vertigo, movement disorders, paresis, stroke like events, cognitive impairment, psychiatric disorders, pyramidal signals and seizures are the most common manifestations[2,3]. The other causes of basal ganglia calcification are infections (tuberculosis, AIDS, EB virus), hypoparathyroidism, hyperparathyroidism, tuberous sclerosis, lupus and motor neurone disease.

Here, we report a rare case of Fahr's disease who is a known hypertensive presented with seizure disorder of recent onset.

Case Report

A 50 years old male attended the hospital with complaints of three episodes of sudden onset of tonic clonic generalized seizure, at his residence the previous day with momentary loss of consciousness during all the three attacks. He did not give any history of fever, headache, vomiting nor any history of head injury in the past. The seizure was not associated with motor deficit nor any behavioral change. He gave a past history of acute gastroenteritis and generalized weakness associated with mild hypokalemia about seven months back. None in the family including his 7 siblings suffer from seizure disorder.

Examinations — Patient was conscious and well oriented. General physical examination revealed no abnormality. Central nervous system examination was within normal limits. There was no motor or sensory deficit, no sign of cerebellar dysfunction, nor any movement disorder. There was no sign of meningeal irritation. Fundoscopy was normal . The score of Mini-mental state examination was 24.

Blood haemogram was normal, serum glucose were fasting 82 mg/dl and postprandial 100 mg/dl, S. creatinine 1mg/dl and urea 24mg/dl, S electrolytes (sodium139 mmol/L, potassium 4.2 mmol/L, calcium 4.85mg%, phosphate 3.7 mg%, magnesium 2.1 meq/L) were within normal limits. Serum liver function tests and lipid profile showed no abnormality. Serum thyroid stimulating hormone was 3.41miu/ml, serum parathormone was 32.5 pg/ml and HIV I&II, ANA, and TB PCR were all negative. Urine routine examination was normal. CT scan of brain revealed multiple calcifications of the basal ganglia, dentate nuclei of the cerebellum, caudate nuclei, globus pallidum and thalamus bilaterally (Fig 1) which is consistent with Fahr's disease. EEG showed nonspecific changes of seizure disorder. In the absence of biochemical abnormalities, infectious, toxic or traumatic causes of multiple calcifications of the brain the patient was diagnosed as Fahr's disease.

Discussion

Fahr's disease has a broad spectrum of clinical neurological presentation. Patients can present with movement disorders, executive and cognitive impairment, seizures, pyramidal symptoms, cerebellar signs and other different presentation depending on the part of the brain affected[4]. Our patient presented with generalized tonic clonic seizure without any known etiology. Cases presenting with seizure disorders in Fahr's disease has been reported in the literature[5-7].

The diagnostic value of EEG has been evaluated in patients with Fahr's disease. The EEG results yielded no discernible diagnostic value and all kinds of alterations of central electrophysiological activities were possible, but no characteristic EEG pattern asso-

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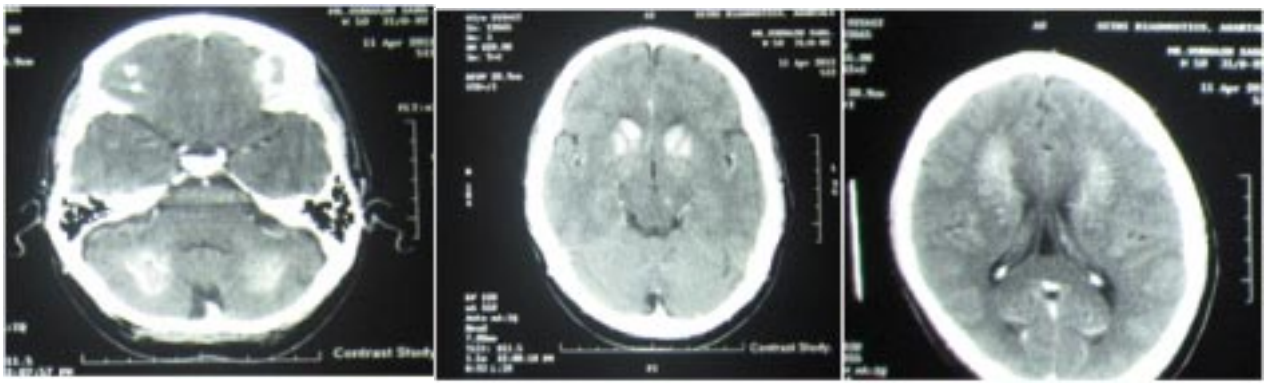

Fig 1 — CT Scan Brain showing bilateral symmetrical calcifications in cerebellar hemisphere, caudate nucleus and periventricular areas of brain

ciated with Fahr's disease was seen[6]. In this present case a non specific pattern of seizure disorder was seen in EEG

Fahr's disease may have familial aggregation, with autosomal recessive or autosomal dominant transmission – Geschwind and his colleagues[8] have described a locus IBGC1 on chromosome 14q involved in the idiopathic basal ganglia calcification. Fahrs disease is diagnosed by exclusion of other conditions with intracerebral calcifications by appropriate investigations. These conditions include endocrinopathies (hypothyroidism, hypogonadotrophic hypogonadism), systemic diseases (systemic scleroderma, systemic lupus erythematosus), infections (toxoplasmosis, neurocysticercosis, German measles, neurobrucellosis, HIV), primary or secondary calcified brain tumors, and various diseases such as tuberous sclerosis, mitochondrial encephalopathy, myotonic muscle dystrophy, measles and smallpox encephalitis, post-anoxia disorders, phacomatosis, Cockayne syndrome, neonatal anoxia, idiopathic hemochromatosis, heavy metal and carbon monoxide intoxication, treatment with methotrexate, and radiotherapy[9].

Conclusion

Fahr's disease is a very rare clinical entity and it is mostly associated with movement disorder and neuropsychiatric manifestations. A rare case of Fahr's disease with seizure disorder is reported.

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