CASE REPORT

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Fahr's Syndrome – A Rare Manifestation of Hypoparathyroidism

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Abstract

Background: Fahr's syndrome is a rare (prevalence <1/1,000,000) neurological disorder characterised by abnormal deposition of calcium in basal ganglia, dentate nuclei and cerebral cortex leading to various neurological manifestations. Distinguishing Fahr's syndrome from Fahr's disease is important because of differences in their aetiology, location of lesions, prognosis, and therapy.

Methods: A 36-year-old lady presented with generalised tonic clonic seizures and a past history of thyroidectomy. Her serum calcium and PTH levels were markedly decreased and her serum magnesium levels were just below normal. Her NCCT head and MRI brain revealed calcifications in white matter of bilateral fronto-parietal lobes, bilateral basal ganglia and bilateral cerebellar hemispheres (dentate nucleus).

Results: A diagnosis of secondary Fahr's syndrome post-thyroidectomy was made. The patient gradually responded to calcium infusions, anti-convulsants and supportive treatment. She was discharged in a satisfactory condition after 2 weeks on oral calcium supplementations, anti-convulsants and thyroxine sodium.

Conclusion: This case highlights the importance of measuring calcium levels and parathyroid hormone levels in patients who present with seizures, especially post-thyroidectomy, which can be life saving.

Key words: Hypoparathyroidism, hypocalcaemia, Fahr's syndrome.

Introduction

Fahr's syndrome is a rare neurological disease with a prevalence of <1/1,000,000 resulting from abnormal intracranial calcific deposits in the basal ganglia, dentate nucleus, and cerebral cortex. This rare condition was first described by German neurologist Karl Theodor Fahr in 1930. Fahr's disease arises from a primary hereditary condition while Fahr's syndrome results from on underlying secondary causes[1]. It is important to differentiate between them because Fahr's syndrome has a specific treatment related to the underlying cause with symptomatic therapy while effective treatment for Fahr's disease is currently unavailable. This case report presents Fahr's syndrome secondary to hypoparathyroidism (HP), which developed as a complication of thyroidectomy.

Case Report

A 36-year-old lady was admitted to the Medicine department with complaints of abnormal body movements for 9 days. There was a history of generalised convulsions with uprolling of eyeballs and urinary incontinence. There was no history of fever, cough, bowel problems or trauma. The patient had similar problems in the form of generalised tonic clonic seizures (GTCS), and muscle spasms for 8 years, for which she used to take medications, but was not relieved. There was a history of thyroidectomy (sub total thyroidectomy) 15 years ago and again 8 years back (total thyroidectomy). Following the surgery, after 1 year, she had the above complaints and was started on anti-convulsants. On examination, the patient was drowsy but oriented to time, place and person. The thyroidectomy scar mark was present on the neck (Fig. 1). The rest of the general physical examination was unremarkable. Chvostek' s sign was negative. Trousseau' sign was positive. Her systemic

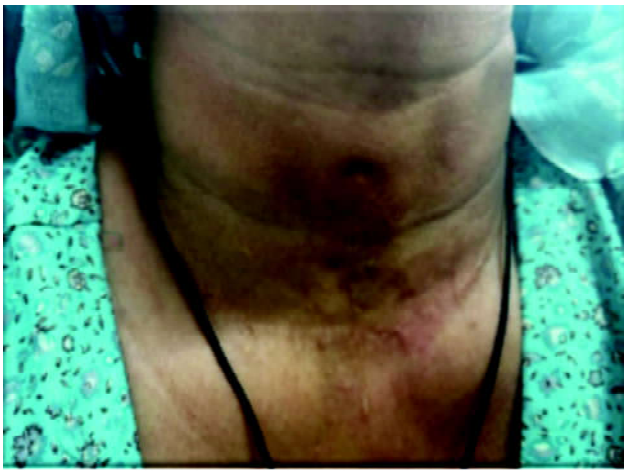

Fig. 1: *Thyroidectomy scar mark.*

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examination was within normal limits. There was no focal neurological deficit and no sign of meningeal irritation.

Investigations

Her complete blood count, renal functions, electrolytes, blood glucose and urine examination were normal. Her serum calcium level was markedly decreased – 6 mg/dL, ionised calcium level was low – 0.89 mmol/L, serum magnesium level was just below the normal – 1.60 mg/dL and serum phosphorus level raised – 5.4 mg/dL. Her PTH level was decreased – 3.5 pg/mL. The ECG showed a prolonged QT interval, T-wave inversions. NCCT head and MRI brain showed calcifications in white matter of bilateral fronto-parietal lobes, bilateral basal ganglia and bilateral cerebellar hemispheres (dentate nucleus) (Figs. 2, 3). A diagnosis of secondary Fahr's syndrome post-thyroidectomy was made and patient was started on Intravenous calcium

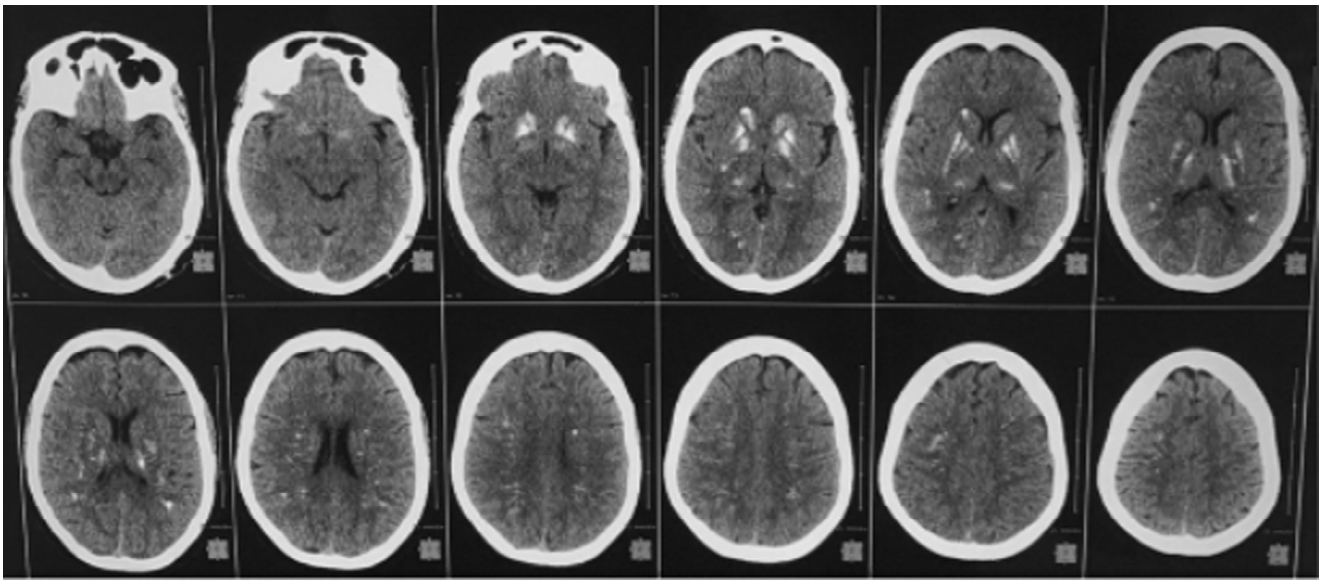

Fig. 2: *NCCT brain showing calcifications in white matter of bilateral fronto-parietal lobes, and bilateral basal ganglia.*

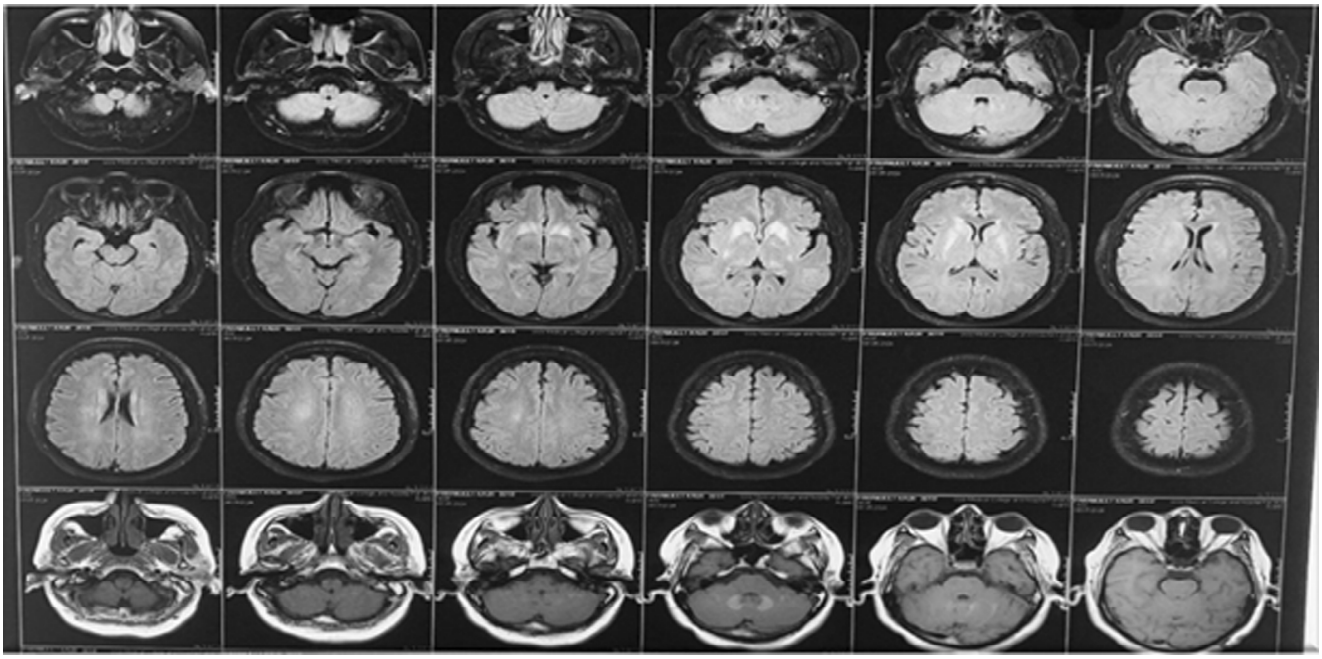

Fig. 3: *MRI brain showing hyperintensities in bilateral basal ganglia, and dentate nuclei.*

infusion, anticonvulsants, vitamin D and tablet thyroxine sodium 50 ug. Her general condition improved and GTCS were controlled with repeat calcium levels - 8.0 mg/dl. Finally, she was started on oral anticonvulsants, calcium supplements,thyroxin sodium 50 ug and was discharged in a satisfactory condition after 2 weeks.

Discussion

Fahr's syndrome is a rare neurological disorder characterised by abnormal deposition of calcium in basal ganglia, dentate nuclei and cerebral cortex leading to various neurological manifestations such as gait disorder, speech dysfunction, cognitive impairment, neuropsychiatric disorders, generalised or partial seizures[1]. The most common endocrine disorder related to Fahr's syndrome is hypoparathyroidism[2]. It is important to differentiate between Fahr's syndrome and Fahr's disease. Fahr's syndrome typically presents in individuals aged 30 - 40 years and is characterised by symmetric bilateral intracranial calcifications and an underlying disorder while Fahr's disease is more commonly observed in individuals aged 40 - 60 years, with progressive symmetric bilateral calcification in the basal ganglia and autosomal dominant or recessive inheritance. Fahr's syndrome has a specific treatment related to the underlying cause with symptomatic therapy while effective treatment for Fahr's disease is currently unavailable[3]. Secondary hypoparathyroidism is a comparatively frequent complication of total or subtotal thyroidectomy with an incidence ranging from 0.9% to 1.6% for permanent hypoparathyroidism[4]. There occurs metabolic dysfunction in hypoparathyroisdism which results in ectopic soft tissue calcifications[5]. A variety of organs can be affected by calcification, more frequently kidneys (as nephrolithiasis or nephrocalcinosis), but also joints, eyes, skin, vasculature and, although rarely seen, intracerebral calcifications[5]. According to Clarke *et al*, anterior neck surgery is the most common cause of acquired HP, and responsible for about 75% of cases; less than 1% - 5% experience permanent HP, even though as many as 50% may develop transient HP[6]. The diagnosis of HP occurs when the iPTH level is normal or inappropriately low in a patient with subnormal total or ionized calcium values, high serum phosphorus or at the high end of the normal range, and after hypomagnesaemia has been ruled-out[6,7]. CT and MRI of brain are the imaging of choice in patients with Fahr's syndrome[8]. Management of Fahr's syndrome involves symptomatic management and treatment of underlying cause. Benzodiazepines are prescribed for dystonia, atypical antipsychotics for neuropsychiatric manifestations, and seizures are managed with antiepileptics[9,10].

Conclusion

Fahr's syndrome is a poorly understood and rare condition. However, it should be kept in mind in all cases of patients with classical clinical manifestations of hypocalcaemia (Carpopedal spasm, Chvostek's sign and Trousseau sign and neuroimaging findings (Intracranial Calcification). Any suspected hypoparathyroidism should be treated immediately, especially in patients who have undergone thyroidectomy, to prevent the formation and progression of brain calcific lesions.

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