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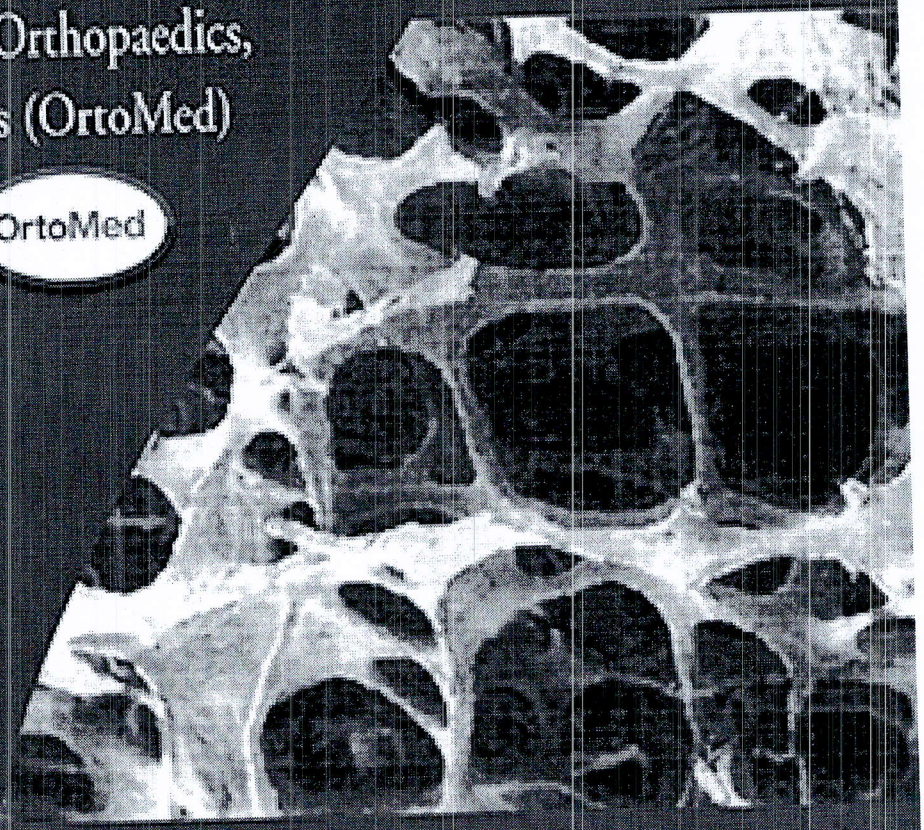

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Femoral neck's fracture in Fahr's Syndrome: case report

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Summary

Fahr's syndrome, also known as "Bilateral Striopallidodentate Calcinosis" (BSPDC) primitive, is a rare neurological disease characterized by the presence of idiopathic, bilateral, symmetrical and abnormal deposition of calcium in areas of the brain that control movements including the basal ganglia, dentate nuclei of the cerebellum, nuclei of thalamus and semi-oval center. We describe a case of a 76-year-old male patient underwent reduction and fixation of a subtrochanteric fracture with intramedullary nail. During post-operative rehabilitation therapists's patient management was difficult due to obvious extrapyramidal symptoms characterized by dysarthria, rigidity, bradykinesia, postural instability. A CT scan, performed for the onset of stiffness and confusion before the operation, showed: IV ventricle eumorphic and in axis; expansion in atrophic sense of supratentorial ventricular system; bilateral, diffuse and coarse calcifications of the basal ganglia in the cerebellar and occipital cortex, elements compatible with Fahr's syndrome. The patient presented repeated postural instability episodes in the upright position, with loss balance tendency and recurrent falls. Fahr's syndrome patient is a "weak" patient, which requires a multidisciplinary approach in order to prevent the mobility reduction, to improve the condition of postural instability, thus reducing the risk of fractures using preventive measures in domestic environment.

KEY WORDS: pseudohypoparathyroidism; fracture; nail; rehabilitation.

Introduction

Fahr's syndrome, also known as "Bilateral Striopallidodentate Calcinosis" (BSPDC) primitive, is a rare neurological disease characterized by the presence of idiopathic, bilateral, symmetrical and abnormal deposition of calcium in areas of the brain that control movements including the basal ganglia, dentate nuclei of the cerebellum, nuclei of thalamus and semi-oval center. It was first described by German neurologist Karl Theodor Fahr in 1930. Usually the syndrome is asymptomatic or with few symptoms, like alterations of motor functions (55% of cases) with rigidity, hypokinesia, tremor, dystonia and ataxia, associated with neuropsychiatric disorders and dementia (1-3). Pathological features, being similar in both adults and infants, are not affected by age. At the molecular level, calcification generally develops within the vessel wall and in the perivascular space, ultimately extending to the neuron. Due to defective iron transport and free radical production, tissue damage occurs which leads to the beginning of calcification. It occurs secondarily around a nidus composed of mucopolysaccharide and related substances. Progressive basal ganglia mineralization tends to compress the vessel lumen, thus initiating a cycle of impaired blood flow, neural tissue injury and mineral deposition. Calcifications commonly occur in basal ganglia, thalamus, dentate nucleus, cerebral cortex, cerebellum subcortical white matter, and hippocampus. Deposits are composed of mineral compounds like calcium phosphate and carbonate. Fahr's syndrome is associated, in most cases, with a calcium and phosphorus metabolism alteration resulting in parathyroid glands dysfunction and shaping itself into a framework of pseudohypoparathyroidism secondary. These alterations depend only partly by dysmetabolic condition, being probably involved also genetic factors like a locus on the long arm of chromosome 20. Endocrine disorders, particularly parathyroid disturbances are most commonly associated with Fahr's syndrome. These abnormalities include idiopathic hypoparathyroidism, secondary hypoparathyroidism, pseudohypoparathyroidism, and hyperparathyroidism.

The etiological manifestationts are reported on Table 1.

The diagnostic criteria of Fahr's syndrome has been modified and derived from Moskowitz (4), Ellie (3), Manyam (5) and it can be stated as follows:

- Bilateral calcification of the basal ganglia visualized on neuroimaging. Other brain regions may also be observed.
- Progressive neurologic dysfunction, which generally includes a movement disorder and/or neuropsychiatric manifestations. Age of onset is typically in the fourth or fifth decade, although this dysfunction may also present in childhood.
- Absence of biochemical abnormalities and somatic features suggestive of a mitochondrial or metabolic disease or other systemic disorder.
- Absence of an infectious, toxic, or traumatic cause.
- Family history consistent with autosomal dominant inheritance.

Case report

A patient of 76-year-old male was admitted to the Orthopae-

Table 1 - Etiological manifestations of Fahr's Syndrome.

S. No	Etiologies	
1	**Endocrine disorders**	• Idiopathic hypoparathyrodism • Secondary hypoparathyrodism • Pseudohypoparathyroidism • Hyperparathyroidism
2	**Adult onset neurodegenerative conditions**	• Neurodegeneration with Brain Iron • Accumulation Disease • Neuroferritinopathy • Polycystic Lipomembranous • Osteodysplasia with Sclerosing • Leukoencephalopathy
3	**Infectious disease**	• Intrauterine and Perinatal infections • Cockayne Syndrome Type 1 • Cockayne Syndrome Type 2
4	**Inherited or early onset syndrome**	• Aicardi-Gouteres Syndrome • Tuberous Sclerosis Complex • Brucellosis • Coat's disease

Table 2 - Patient's data at admission.

Diagnosis	Subtrochanteric fracture left femur
Sex	Male
Height	171 cm
Weight	63 kg
Blood tests	WBC 8.32 10^3/mL, RBC 2.78 10^6/mL, HGB 8.4 g/dL, GLYCEMIA 98 mg/dL, CREATININE 1.84 mg/dL, AST 15 U/L, ALT13 U/L, GGT 20 U/L

dics and Traumatologic Department University of Palermo for subtrochanteric fracture of the left femur following accidental fall at home (Table 2). On admission physical examination was normal; the left leg appeared shorter and external rotated (Figure 1), with complete loss of function and severe pain (VAS 8).

A CT scan, performed for the onset of stiffness and confusion before the operation, showed: IV ventricle eumorphic and in axis; expansion in atrophic sense of supratentorial ventricular system; bilateral, diffuse and coarse calcifications of the basal ganglia in the cerebellar and occipital cortex, elements compatible with Fahr's syndrome (Figure 2).

To confirm the suspected diagnosis we studied calcium and phosphorus metabolism and laboratory test showed hypocalcaemia (7.2 mg / dl), increased serum phosphate (4.6 mg / dl), urinary calcium in the normal range (177 mg / 24 h), while the rates of serum PTH current assets were higher than the normal range (74 pg / ml); the dosage of vitamin D was 25.3 ng/ml. With these data it was possible to diagnose a condition of pseudohypoparathyroidism, closely related to the Fahr's syndrome. The patient underwent reduction and fixation of the fracture with

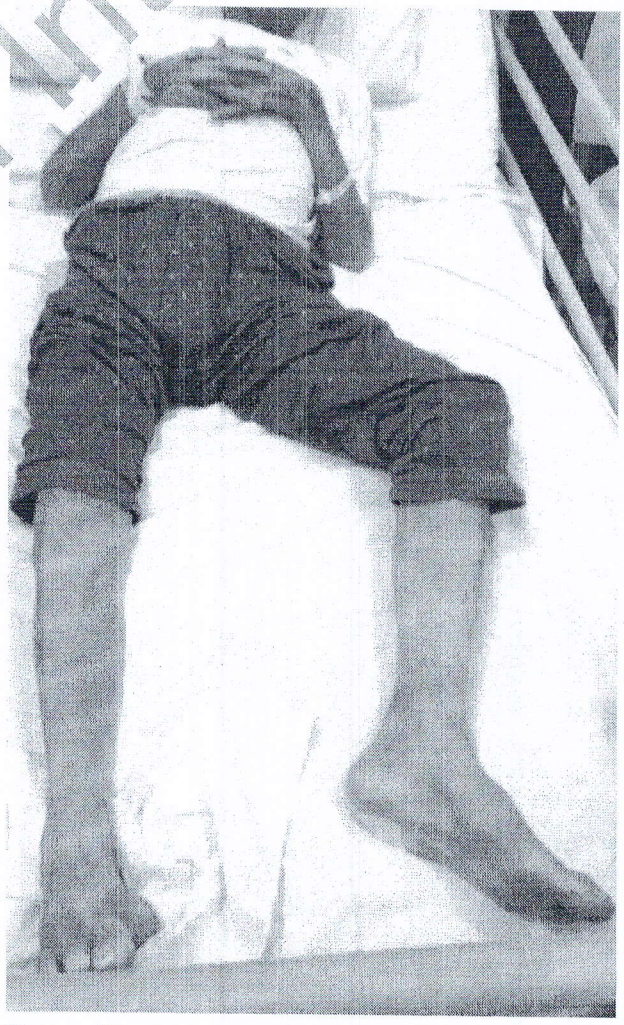

Figure 1 - Shorter and external rotated left leg.

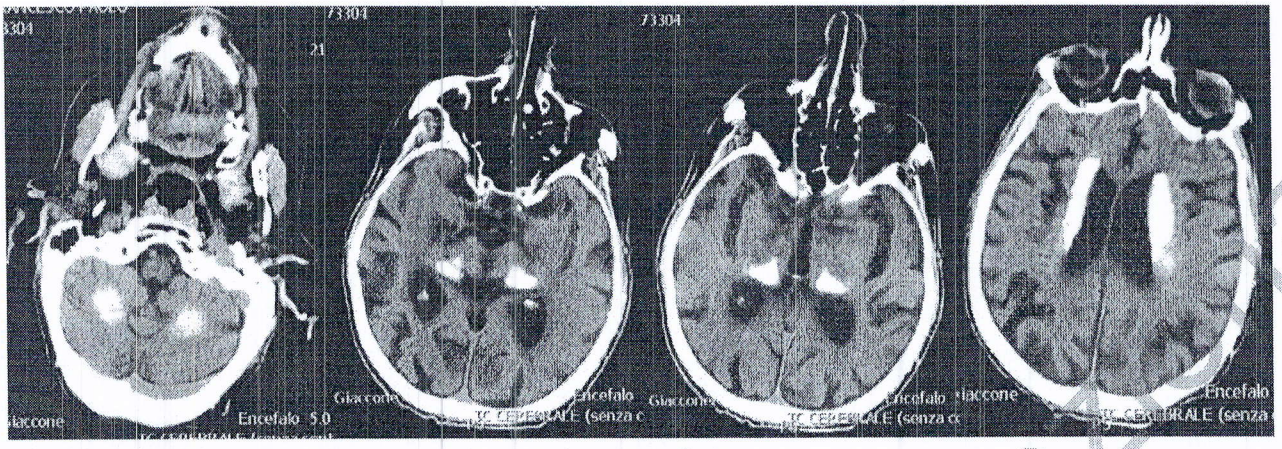

Figure 2 - CT scan of brain showing bilateral cerebellar and basal ganglia calcification.

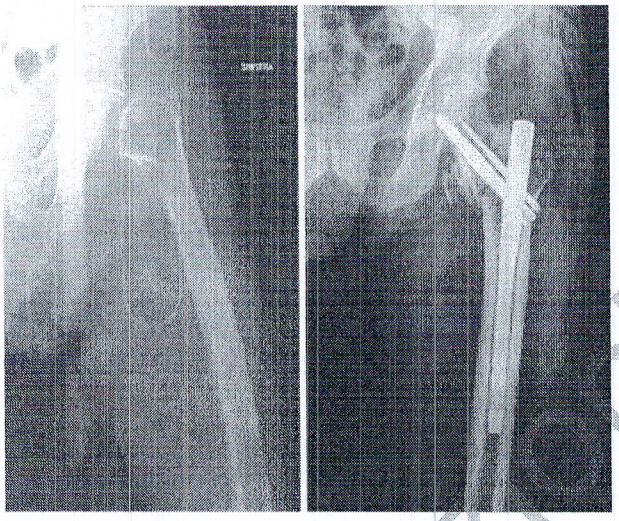

Figure 3 - Pre-operative and post-operative X-rays.

intramedullary nail (Figure 1). In the second postoperative day started the rehabilitation plan (5 days a week for 12 weeks) including: recovery of hip and knee ROM, antigravity muscles tropism and bed's postural changes autonomy as short-term goals and trunk control achievement, postural stability, walking without aids although with supervision as medium-term goals. During post-operative rehabilitation therapists's patient management was difficult due to obvious extrapyramidal symptoms characterized by dysarthria, rigidity, bradykinesia, postural instability.

Discussion

This case is a rare example of Fahr's syndrome onset characterized by clinical extrapyramidal symptoms with initial cognitive impairment and behavioral disorders. The phosphocalcic metabolism alteration in the patient was compatible with a clinical picture of pseudohypoparathyroidism; therefore therapy at discharge included the patient calcium and vitamin D supplementation (1000mg + 880 I.U. daily). The patient presented repeated postural instability episodes in the upright position, with loss balance tendency and recurrent falls; calcium phosphorus metabolism alterations, is a predisposing factor of increased risk fractures. Fahr's syndrome patient is a "weak" patient, which requires a multidisciplinary approach in order to prevent the mobility reduction, to improve the condition of postural instability, thus reducing the risk of fractures using preventive measures in domestic environment.

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