Journal of Neonatal Surgery
ISSN(Online): 2226-0439
Vol. 14, Issue 8s (2025)
https://www.jneonatalsurg.com

OPEN ACCESS

A Rare Case of Fahr's Disease with Epileptic Seizures with Severe Hypocalcemia and Calcifications

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Cite this paper as: Dr. Karthik D, Dr. Renuka Jadhav, Dr. Shailaja Mane, Dr. Priyanka Mitra, Dr. Nidhish Raval, (2025) A Rare Case of Fahr's Disease with Epileptic Seizures with Severe Hypocalcemia and Calcifications. *Journal of Neonatal Surgery*, 14 (8s), 494-497.

ABSTRACT

Fahr’s syndrome is a condition defined as bilateral striato-pallido-dentate calcinosis, a neurodegenerative disease with radiological findings of symmetrical and bilateral idiopathic calcifications of the cerebellum, periventricular white matter, and basal ganglia. Clinical correlation with radiological and a calcium metabolism panel is crucial in differentiating between Fahr’s disease and Fahr’s syndrome. In this case, the patient presented with clinical features of hypocalcemic tetany with global developmental delay and had an incidental radiological finding of Fahr syndrome.

I. INTRODUCTION

Fahr’s disease was originally described by Karl Theodor Fahr in 1930 as a rare familial (autosomal dominant) disorder that presents with idiopathic abnormal bilateral basal ganglia calcium deposits, with a higher number of cases presenting between the age of 40-50 years [1]. Common symptoms include both motor and psychiatric disorders. Calcifications are classically symmetrical, most frequently located in the basal ganglia. However, they are also seen in the dentate nuclei, thalamus, brainstem, centrum semiovale, and subcortical white matter [2]. Computerized tomography (CT) scan of the brain is the gold standard for detecting and localizing intracranial calcifications [3].

2. CASE PRESENTATION

A 10-year-old male child, third by birth order in a third degree consanguineous marriage with a past medical history of recurrent generalized tonic clonic seizures since 8 months of age, with each episode lasting for an average time period of 15 to 30 minutes, presented with bilateral Trousseau sign (Image 1) and associated abdomen pain. He was previously on syrup valparoate, in spite of which he had one or two episodes of seizures every month, for which he was started on Levetiracetam syrup at 20 mg/kg/day.

His birth history was normal, with complete immunisation, no significant family history, language delay, delayed dentition and hyperactivity with occasional violent behaviour. On examination, he had Chovestek and Trousseau sign positive with an esotropic squint, a flat nasal bridge, multiple hypopigmented patches on the face suggestive of atopic dermatitis (Pityriasis alba). (Image 2)

Investigations done on admission suggested anemia (Hb 10.8) normal hb (12-14), low PTH level (0.10) normal(15-65pg/ml), normal vit d levels (20.2), with severe hypocalcemia (<6mg/dl) normal calcium (8.8-10.8), hypomagnesemia (1.70), hyperphosphatemia (>9), low ionic calcium (0.37), reduced serum proteins (4.20), hypoalbuminemia (2.5), hypoglobulinemia (1.70).

The patient was started on parenteral calcium gluconate and later, shifted onto oral calcium with vitamin D3.

CT scan of the brain revealed symmetrical calcifications in bilateral gangliocapsular region, thalami, white matter of bilateral fronto-parietal regions and centrum semiovale. These findings were suggestive of Fahr's syndrome and possibly, Fahr's disease (Image 3).

Other investigations, such as 2D echo, ultrasound of the abdomen and pelvis and ECG were normal.

The patient's family declined genetic testing for the child. They were counselled that the focus of care was then on symptomatic control and psychiatric management.

Image 1

Image 2

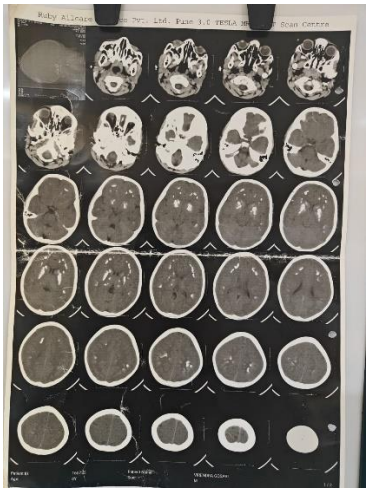

Image 3

3. DISCUSSION

The term "Fahr's disease" refers to the primary form, which is based on genetic alterations. On the other hand, the term "Fahr's syndrome" has been suggested when a secondary, and potentially treatable, cause is found. Various disorders can cause intracranial calcifications, such as metabolic, infectious, congenital, vascular, and neoplastic conditions.

Hypoparathyroidism is the most common endocrine disorder associated with intracranial disorders. Toxoplasmosis, rubella, cytomegalovirus, HIV, tuberculosis, and syphilis are some of the most common infectious diseases that cause intracranial calcifications. An autoimmune condition such as systemic lupus erythematosus (SLE) may also cause intracranial calcification [4](table 1).

	Fahr's Syndrome	**Fahr's Disease**
Age of Onset	30 to 40 years	40 to 60 years
Genetic Traits	None	Autosomal dominant or Autosomal recessive
Radiological Findings	Symmetrical and bilateral intracranial calcifications.	Coarse, progressive, bilateral and symmetrical striato-pallido-dentate calcifications.
Associated Conditions	Endocrinopathies: Idiopathic hypoparathyroidism secondary hypoparathyroidism, pseudo-hypothyroidism, hyperparathyroidism, or presence of any of the following conditions: Brucellosis infection (intrauterine or perinatal), neuroferritinopathy, tuberous sclerosis, mitochondrial myopathy, lipoid proteinosis	None
Treatment	Treatment directed to specific aetiology and adjunctive symptomatic treatment.	No specific remediation, only symptomatic treatment.

The disease usually manifests after the age of 30 [5]. It progresses slowly with a male preponderance. Based on the previous studies, the incidence of intracranial calcifications in the basal ganglia detected by CT head indicates that Fahr's syndrome is approximately between 0.24% and 1.64% [6].

Fahr's disease is characterized by bilateral symmetrical calcification of the basal ganglia. The globus pallidus is the most involved basal ganglion. Calcium deposits can also be found in dentate nuclei, thalami, and other deep cortical structures [7]. It is also characterized by movement disorders, cognitive impairments, neuropsychiatric symptoms (such as hallucinations, delusions, anxiety, irritability, and aggression), mood disorders (depression and bipolar disorder), and personality disorders [8].

Baseline biochemical investigations are indicated to rule out other possible diagnosis

Types of Laboratory Investigations	**Indication**
Serum calcium, magnesium, phosphate, serum parathyroid hormones	To exclude hypocalcaemia, hypomagnesaemia, hyper- or hypoparathyroidism
Serum Vitamin D and calcitonin	To exclude vitamin D deficiency and secondary hypoparathyroidism.
Ellsworth-Howard test	To assess for hypoparathyroidism
Blood and urinary heavy metals level	To exclude heavy metal toxicity
CSF evaluation	To exclude infection and autoimmune aetiology

Other complications or clinical presentations include stroke, orthostatic hypotension, and syncope.

Molecular genetic testing is indicated if there is a strong family history of autosomal dominant inheritance. SLC20A2 sequencing is the first test to be performed. If no identifiable mutation or deletion of SLC20A2, one must consider PDGFRB sequence analysis

The exact aetiology of Fahr's syndrome is still unclear. Genetic alteration at chromosome 14 has been suggested as a cause of this condition. It is thought to be autosomal dominant in transmission. The 14q chromosome is most commonly affected in Fahr's syndrome.

Diagnostic criteria for Fahr's syndrome/disease

Diagnostic Criteria
Neuroimaging characterized by bilateral basal ganglia calcifications
Progressive neurological dysfunction that constitutes a variety of manifestations from motor disorder to neuropsychiatric presentation
The typical age of onset is thought to be around the fourth or fifth decades of life
In the absence of biochemical abnormalities or somatic features, another diagnosis has to be considered. For instance, mitochondrial disorders or metabolic conditions have to be excluded
Diagnosis of exclusion after evaluation for infectious, toxic, or traumatic causes
Presence of autosomal dominant familial inheritance disorder

4. CONCLUSIONS

Fahr's disease and Fahr's syndrome have a widespread clinical presentation with radiological findings of bilateral symmetrical basal ganglia and dentate nuclei calcifications. Therefore, it's essentially a diagnosis of exclusion after ruling out metabolic disorders. Treatment is tailored to symptom control for Fahr's disease and correction of underlying metabolic abnormalities.

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