2025

KARNATAKA RADIOLOGY EDUCATION PROGRAM

CASE PRESENTATION:

FAHR'S DISEASE.

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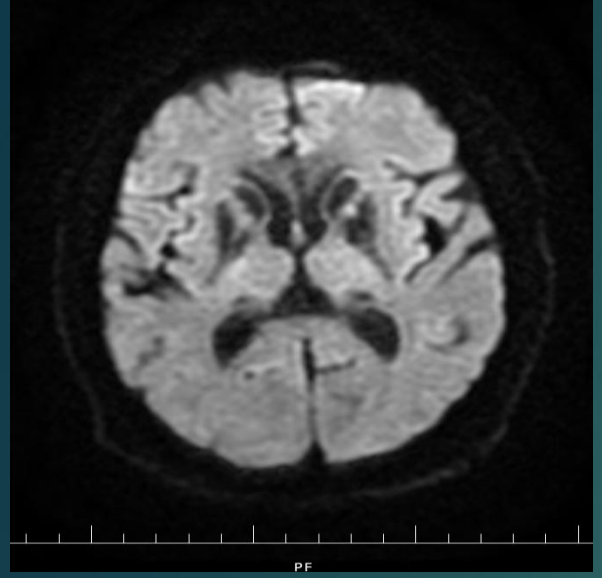
INTRODUCTION

- Fahr disease is named after Karl Theodor Fahr, a German neurologist who first reported the disorder in 1930. It is a rare neurological condition characterized by abnormal idiopathic calcification of basal ganglia and commonly has an autosomal dominant inheritance.
- For primary basal ganglia calcifications, with no known etiology, the term Fahr disease should be used.
- For secondary causes of basal ganglia calcifications, with known underlying causes, the term Fahr syndrome should be used.

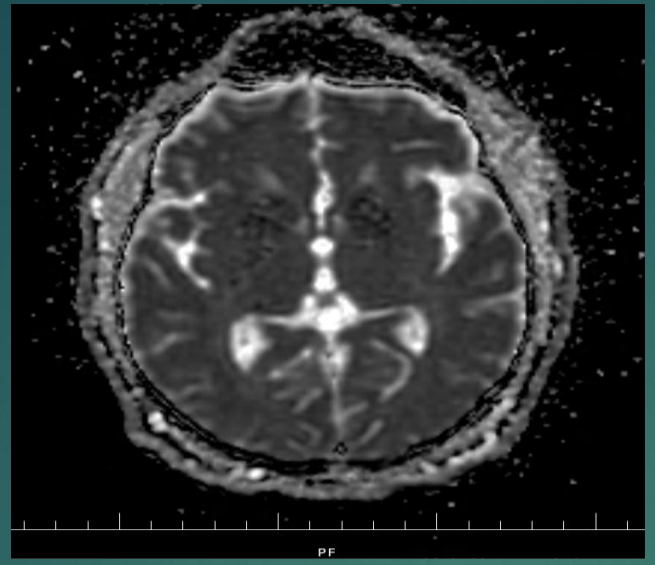
HISTORY

- 51 yr old male presents with

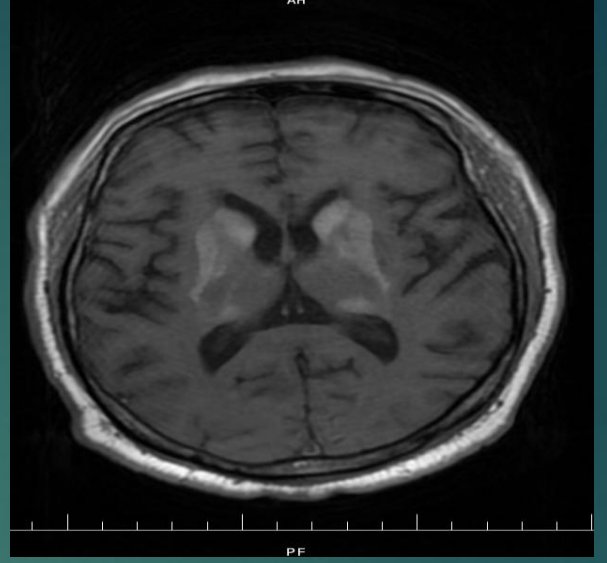
H/o irritability, abnormal movements and disorientation.



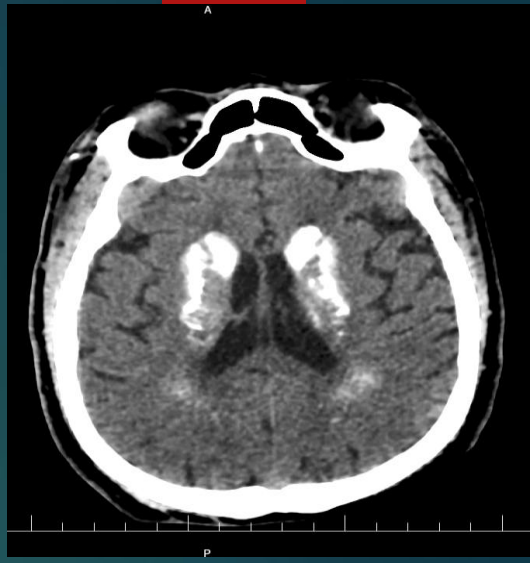
AXIAL DWI



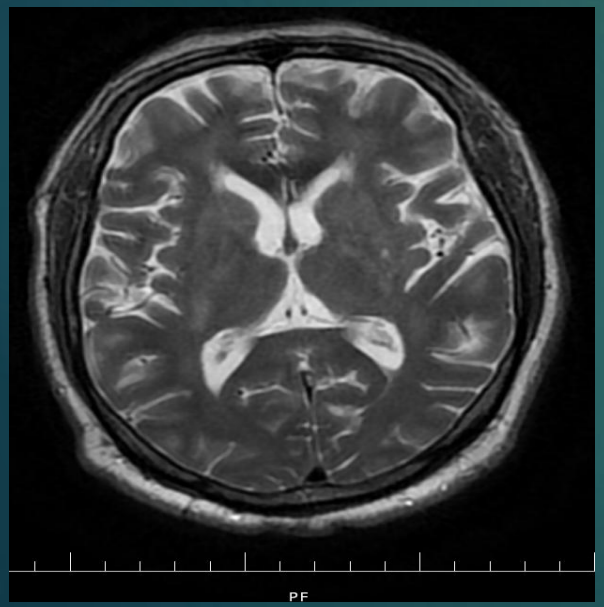
AXIAL ADC



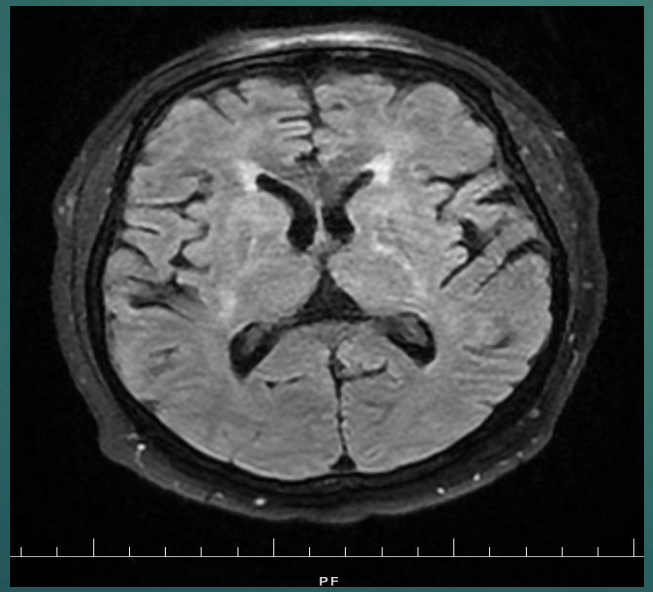
AXIAL T1 WI



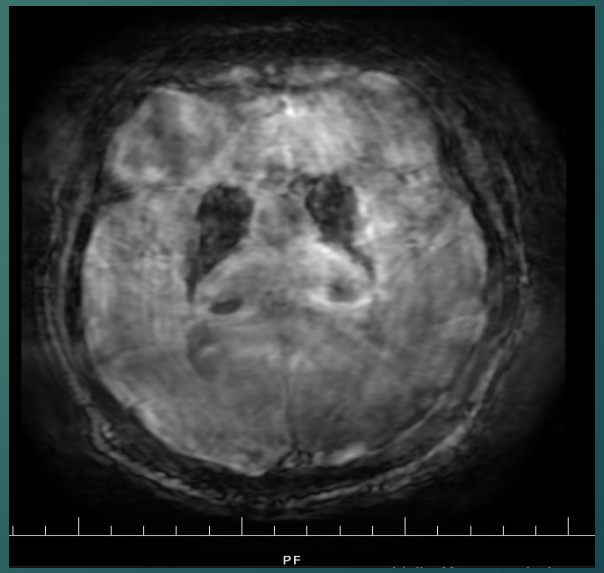
AXIAL CT



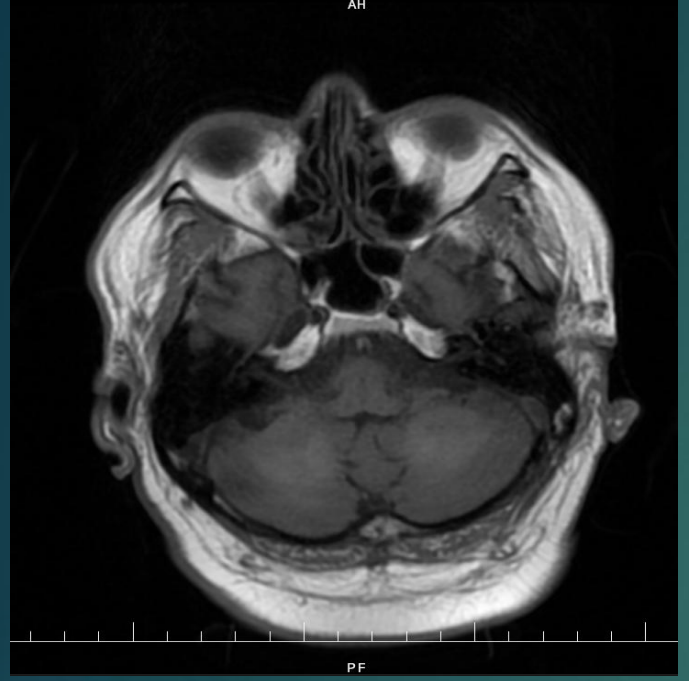
AXIAL T2



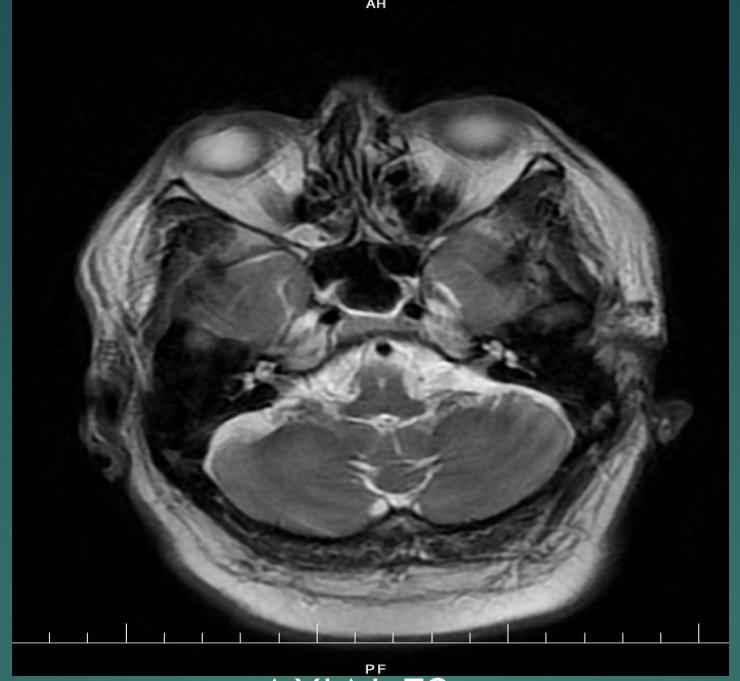
AXIAL T2 FLAIR



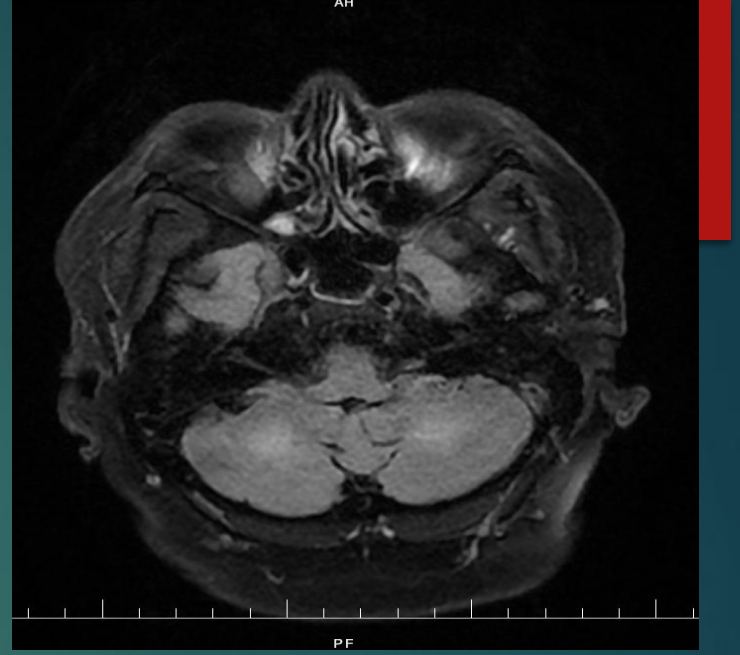
eSWAN



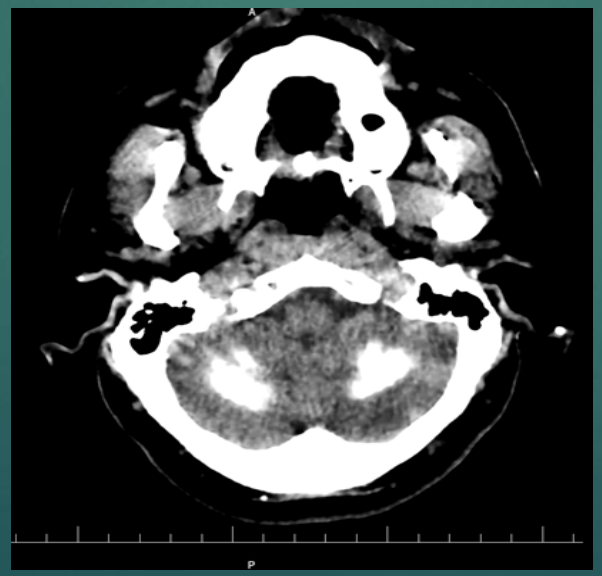
AXIAL T1



AXIAL T2



AXIAL T2 FLAIR



AXIAL CT

FINDINGS

- Bilateral symmetrical signal changes involving the caudate, lentiform nuclei, thalami, dentate nuclei and deep sub cortical white matter. It appears hyperintense on T1 and hypo to isointense signal on T2 with blooming on SWAN. No e/o diffusion restriction.

- CT shows bilateral symmetrical calcification in caudate, lentiform nuclei, thalamus and dentate nuclei.

DISCUSSION

- Fahr disease is reported to commonly affect people in their 40s and 50s. Patients are usually in good health in their youth and tend to develop this progressive neurodegenerative disease later in adulthood.

- Abnormal calcium deposition is hypothesized to be due to either abnormal brain calcium metabolism or metastatic deposition due to locally altered blood-brain barrier. Defective iron transport and free radical production cause tissue damage, which initiates calcification around a nidus composed of mucopolysaccharides and related substances.

- Progressive calcification compresses nearby vessels reducing blood flow and hence continuing the vicious cycle of decreased blood flow, tissue injury, and mineral deposition.

- Primary familial brain calcification (Fahr disease) progresses steadily, and there is no known cure or specific treatment, thus management is supportive depending on the clinical manifestations.

Clinical Features

Movement disorder like features:

- Signs and symptoms resembling parkinsonism, such as bradykinesia, rigidity, tremor, hypophonia, hypomimia, mask-like facies, shuffling gait.
- Clumsiness, Fatigability, Gait dysfunction, Choreoathetosis, Dystonia, Slurred speech, Muscle cramping

Neuropsychiatric features .

- Depression, Apoplexy, Dementia, mostly fronto-executive type resembling subcortical dementia in Wilson disease and Huntington disease

- Causes include :

- Endocrinopathies
- Vasculitis (e.g. systemic lupus erythematosus)
- Mitochondrial disorders (e.g. Kearn-Sayre syndrome)
- Infections
- Other inherited disorders
- Radiation
- Chemotherapy
- Carbon monoxide poisoning
- medications (e.g. bisphosphonates)

THANK YOU