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Sle as a cause of secondry fahr disease

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Abstract
Fahr disease is a movement disorder characterized by calcium deposition in different parts of brain. It could be familial or secondary to a number of metabolic or infective causes. The age of presentation is bimodal and is according to the etiology. Here we present a case of 21 year old Pakistani female. She presented with sudden left sided weakness and dysphasia. On examination there was generalized rigidity of both upper and lower limbs. Power was unilaterally decreased on left side and her speech was dysrthric. After detailed history, examination and investigations she was diagnosed as a case of Fahr syndrome secondary to Systemic lupus erythematous
Fahr syndrome has very heterogeneous clinical presentations and etiologies. Therefore, it's very imporatant to diagnose it early so that it can be properly treated.

Keywords: characterized, Familial, examination, investigations

Introduction
Fahr's disease has a very low prevalence (<1 per million population) but is considered under-reported [1]. The disease is characterized by bilateral symmetrical calcium deposition in areas of the brain associated with movement control like the basal ganglia and adjacent parenchyma such as the dentate nuclei, putamen, thalami, cerebral cortex, subcortical white matter, hippocampus and cerebellum [2, 3].
It can be either primary (usually autosomal dominant) or secondary to a large number of underlying illnesses or metabolic disturbances. Primary fahr syndrome is equivalent to familial cerebral ferrocalcinosis or primary familial brain calcification (now the preferred term), with no underlying other cause whereas secondary is due to an underlying metabolic, infective or other cause. Secondary cause in Fahr syndrome is typically diagnosed in younger individuals with appropriate intracranial imaging features [4]. This disease can present in childhood or adolescence the usual age of presentation is 4th–5th decade [5]

Case Presentation
22 years female resident of Pakistan, Southern Punjab having siblings. She was alright upto the age of 12 years. she had normal growth and achieved milestones at proper time. Although she did not get any formal education due to socioeconomic status but patient was normal according to her mother and she was able to do all her household work. At the age of 12 years she developed polyarthralgia starting from the small joints of the hands, mainly involving the wrists, metacarpo pharayngeal joints, proximal interpharyngeal joints. Although there was pain in joints but patient did not report any kind of swelling in joints, there was early morning stiffness. After involvement of small joints of hands, patient complained of pain in foot joints and large joints of the lower limbs as well 2 to 3 month after.
Having polyarthralgia patient started having oral ulcers mostly on the buccal muscoa and the tongue. Oral ulcers were recurrent, mostly painless and healed without scarring in 2 to 4 weeks. Meanwhile she started having loss of hair. There was off and on low grade fever documented upto 100 mostly in the night with no sweating and rigors or chills. Her workup was not done at that time. She took medication from the general practioner and she improved for the time being. Then her limited workup was done that showed leukopenia, and thrombocytopenia and borderline hemoglobin. Her liver and renal functions were normal. And there were no proteinuria in urine detailed examination. Her ANA by ELISA was positive on that occasion. She was started on prednisolone and azathioprine at that time. Her symptoms improved and she took medicine on and off. She left treatment when symptoms improved. After 6 years, at the age of 18 she started having problems with speaking with low volume speech, sometimes dysarthria. She took some medicines from local GP. But symptoms continued and there was some slowing of the movements of the body reported by the patient and her mother.
Now after 4 years at the age of 22 she came to HolyFamily Hospital with complaints of sudden weakness of left half of the body with dysphasia. On examination there was no joint swelling, no malar rash, no alopecia. A normal looking girl with normal vital signs. On CNS examination she has generalized rigidity of both upper and lower limbs. Higher mental functions were normal. Speech was dysrthric. Cerebellar signs were absent and cranial nerves were intact. Power was 1/5 in both upper and lower limb of left side. Right side power was normal.

Table 1: Her workup was done that showed

Lab	Result
WBC	4.5
Platelets	156,000
Hemoglobin	11
Liver Function Tests	Normal
Urea	Normal
Creatinine	Normal
Urine RE	No Proteinuria
Calcium	9
Phosphorous	4
Parathyroid harmone	45

Table 2: Specific investigations

Lab	Result
ANA by Immunoflorecence	Positive
AntiDsDNA	Negative
Anti Smith Ab	Negative
Anti RNP	Negative
C3 levels	Low
C4 levels	Normal
Antiphospholipid workup	Negative

CTscan brain (Fig 1 & 2) was done that showed bilateral symmetrical basal ganglia calcification noted. Subtle calcification is also noted in frontal lobes. Hypodense lesion seen in right lentiform nucleus. Findings are suggestive of Fahr disease which is also known as bilateral striatopallidodentate calcinos

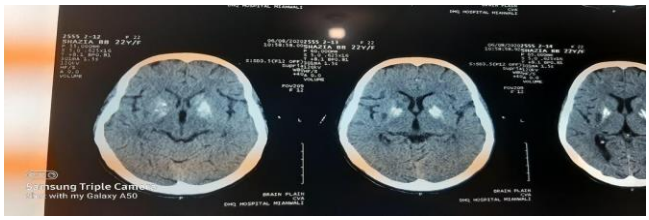

Fig 1

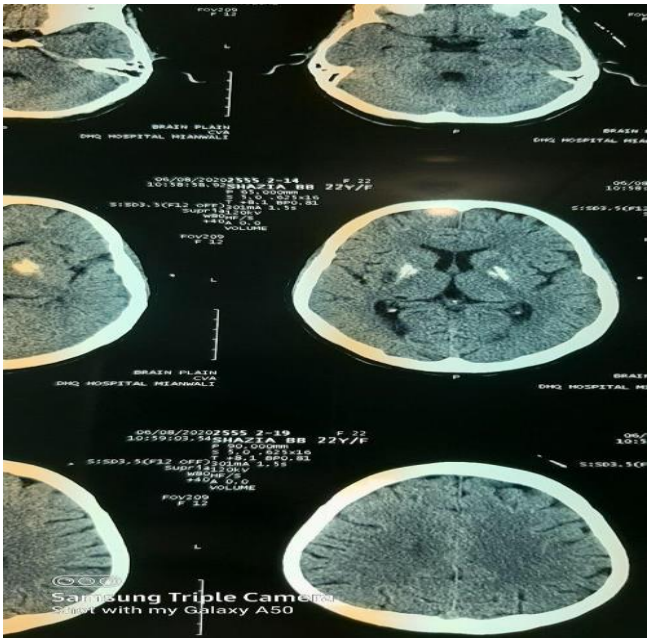

Fig 2

Based upon these findings the patient has Secondry Fahr Disease with underlying Systemic lupus erythromatosis. Which is a rare diagnosis to make. Symptoms of SLE along with parkinsonism symptoms were consistent with Fahr disease. She was started on Hydroxycloroquine, Prednisolone, Antiplatelets, and Statins.
Plan is to control the underlying disease.

Discusssion

Although the exact prevalence of Fahr's syndrome is not known yet intracranial calcifications can be detected incidentally in up to 0.3–1.2% of NCCT examinations of brain [6]. Laboratory examinations should include tests for blood calcium and parathormone which in addition to the other routine blood tests will help in differentiating idiopathic Fahr's syndrome (unremarkable laboratory test results) from secondary cases especially due to hypopara-thyroidism. NCCT brain will demonstrate the presence and extent of parenchymal calcification [5]. When brain imaging is positive for bilateral striopallidodentate calcification, obtaining serum calcium and parathormone levels should help differentiating Fahr's disease from hypoparathiroidism, which is the major differential diagnosis [7, 8]. The diagnosis of Fahr's disease is complex and requires both clinical and radiological evidence. The criteria include bilateral calcification of the basal ganglia with neuropsychiatric and/or extrapyramidal features associated with normal calcium and phosphate metabolism [9]. These calcifications which are usually idiopathic occur most commonly at the basal ganglia, but other structures may also be affected [10]. Patients exhibit progressive neurological symptoms such as seizures, rigidity, and dementia with classical bilateral basal ganglia calcification shown on CT imaging. CT imaging is said to be more sensitive in identifying the basal ganglia calcification in comparison to magnetic resonance imaging (MRI) [11]. The basal ganglia calcification may occur as a consequence of several other known genetic, infectious, and metabolic conditions [12]. Endemic infectious agents such as cytomegalovirus (CMV), HIV, toxoplasmosis, and neurocysticercosis were eliminated as a cause, as were secondary hypoparathyroidism, hypothyroidism, neurofibromatosis, tuberculosis, tuberous sclerosis, cerebral hemorrhage, and vascular disease. Other potential causes to be considered include systemic lupus, hypervitaminosis D, lead poisoning, and radiotherapy. In limited resource settings, CT scans are becoming increasingly available and it remains a very useful modality for diagnosis of Fahr's disease. However, subarachnoid hemorrhage can easily be confused with Fahr's disease on the noncontrast brain CT scan in emergency departments [13].

Table 3: Etiological classification of basal ganglia calcifications [14]

Primary forms	
Gene	Chromosome position
SLC20A2	8p11.21
PDGFRB	5q32
PDGFB	22q13.1
XPR1	1q25.3
MYORG	9p13.3
Secondary forms	
	Calcium/phosphorus abnormalities
	- Idiopathic or secondary hypoparathyroidism
	Infections (brucellosis, AIDS, toxoplasmosis, TORCH complex)
	Toxic exposure (lead, carbon monoxide)
	Disimmunopathies (SLE)
Other conditions	
	Pseudohypoparathyroidism
	Cockayne syndrome I and II
	Aicardi-Goutières syndrome
	Mitochondrial diseases (MELAS, MERRF)
	Coat's syndrome
	Neuroferritinopathy, NBIA

Fahr disease is usually a rare disorder having primary and secondary types. Also known as Fahr syndrome. Primary disease usually presents around the age of 40 years. Most of the patients do not have any symptoms. But patients who have symptoms include bradykinesia, tremors, rigidity, movement disorder, dyarthria, seizures, headache, athethosis, chorea. It is a genetic disease but exact pattern of inheritance is unkown or autosomal dominant. Secondry Fahr disease is seen in younger age group. There is an underlying disease like hypoparathyrodism, pseudohypoparathyrodism, seudopseudohypoparathyrodism, vascuilitis like SLE, mitochondrial disorders, radiation, chemotherapy, carbonmomoxide poisoinig and some inherited disorders like cockayne syndrome and neuroferrinopathy.

There were usually deposition of calcium around atreas like basal ganglia and cereberal cortex. Globus pallidus is the first to be effected. Other investigations that can also be done include MRI and FDG-PET. There is no underlying treatment of Primary Fahr disease. But for the treatment of secondry Fahr disease underlying cause should be treated aggressively.

Conclusion

Fahr Disease is associated with a variety of neuropsychiatric signs and symptoms and could be unnoticed for years

Patient presented with speech problems, movement disorders, decreased tone and left sided weakness. Follow up was done after one and 3 months. Power was increased from 1/5 to 3/5 of left side. Tone became normal and rest of the symptoms remained static and no worsening was seen.

New treatment modalities need to be discovered and employed to minimize loss of functionality associated with the disease. Screening asymptomatic individuals has not been able to demonstrate immediate medical benefits in adults or young individuals. It is of significant use only in adults as it can help them in decision making in their personal life regarding matters of finance, marriage and career. Screening of young individuals is considered unnecessary as it can result in psychological harm without being medically beneficial.

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