Case report

Antiphospholipid syndrome – little to no attention in the Ethiopian clinical setting

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Abstract

Epidemiologically prevailing antiphospholipid syndrome (APS) is common among the younger population. APS is characterized by recurrent arterial and/or venous thrombosis and miscarriage. Although common among the youth; it has gained little or no consideration in its diagnosis in the Ethiopian clinical setup. We identified a woman with a movement disorder called chorea and recurrent pregnancy loss (abortions). She stayed eight years to be diagnosed with APS. As a devastating disease commonly seen in young patients with stroke, deep vein thrombosis and abortion, physician vigilance to detect APS is invaluable. [*Ethiop. J. Health Dev.* 2018;32(2):123-125]

Key Words: Antiphospholipid Syndrome, Chorea, Anticardiolipin antibody, Lupus Anticoagulant.

Introduction

Antiphospholipid syndrome (APS) is a disease first described 34 years back. This is known to cause mainly clotting disorder and miscarriage (1).

APS is the commonest cause of acquired thrombophilia, which can affect any organ in the body (lung, liver, kidneys heart, skin, brain and others).It is an auto-immune disorder characterized by venous and/or arterial thrombosis, recurrent abortions in the presence of antiphospholipid antibodies (aPL) i.e. Lupus anticoagulant (LA), Anticardiolipinanti bodies (aCL), on two or more occasion at a minimum of 12weeks interval (2).Though its exact prevalence is unknown it is estimated to affect 1-4% of the population (3), with an estimated prevalence of 40-50 per 100,000 people (4).It has no specific geographic selection in the world and affects all races. Women are affected five times more than men, and common among those in the age group 30-40 years. Though it doesn't have a cure, if diagnosed early, it can be treated successfully.

Current estimates show that APS is responsible for one in every six cases of DVT, Pulmonary embolism, stroke and myocardial infarction among people under the age of 50 years (5).In a population based study a PLs were positive in 13% of stroke patients, 11% of myocardial infarction,9.5% of DVT and 6% of pregnancy morbidity(4).As a disease with multiple organ involvement, it can present itself to a diverse group of medical specialties- gangrene to the surgeon, stroke and chorea as in our patient to the neurologist, recurrent fetal loss to the gynecologist and differently to others (1).

Despite its critical impact, APS is under diagnosed and misdiagnosed in many parts of the world (6).To the best of our knowledge and observation much remains unknown about APS in Ethiopia, and its diagnosis is made rarely even in tertiary referral centers. We assume this is partly due to preferential lack of awareness about it and partly due to lack of the lab tests locally, which necessitates samples to be sent abroad and are expensive.

APS is known to affect the central nervous system and ischemic stroke via cerebral arterial thrombosis is the commonest neurologic manifestation. Movement disorders occur rarely in APS and chorea is the commonest, while less common ones are hemibalismus, Parkinsonism, hemidystonia. The etiology of movement disorders is not well understood but it is hypothesized that a direct neurotoxic effect of a PL or autoimmune injury to the basal ganglia are the possible causes.

Patient History: A 32-year- old married women was presented at the neurologic follow up clinic at Tikur Anbessa Specialized Hospital (TASH) exhibiting irregular, involuntary movement involving her head, arm and legs since nine years. This symptom started all over the body (hands, legs and head) and was milder in severity. On additional history she had pregnancy loss four times with no known reasons given or interventions done, despite having multiple physician visits over eight years .The pregnancy loses in the first to fourth pregnancy occurred at 9,3,4 and 6 months of gestation respectively. By the time she visited TASH she had two still births and two miscarriages. She had no family history of abnormal body movement and no history of recurrent sore throat or chest discomfort. Generally she was healthy looking, having records of systolic and diastolic hypertension, with no fever. Mini Mental Status examination was normal (30/30). Recent laboratory tests of CBC, lipid profile, VDRL all were normal. On urinalysis initially she had had proteinuria (+1) and hematuria (+3), with a creatinine of 1.1 and urea 31, which all subsequently either cleared or returned to the normal reference range. On Cardiac evaluation had holosystolic murmur at apex and transthoracic echocardiography showed moderate mitral regurgitation, Anti Nuclear Antibody ANA was positive, but double stranded DNA (Ds DNA) was negative.

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Brain MRI showed left basal ganglia and peri-ventricular infarction and mild cerebral atrophy. With it a diagnosis of ischemic stroke was made [figure 1]. Serum Lupus Anticoagulant LA and Anticardiolipin Antibody (aCL) were positive with high titers in two serial tests done 12 weeks apart fulfilling the Sydney criteria for definite APS(7). Finally after eight long years, with a diagnosis of chorea plus recurrent pregnancy loss and ischemic stroke secondary to primary APS; She was put on clonazepam for the Chorea and showed significant improvement and needed no additional medication, also she is on warfarin for the past nine months and fairly well off with no new complaint and no report of pregnancy happening so far.

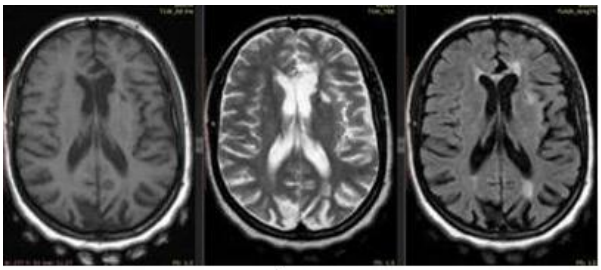

Figure 1:
Brain MRI: T1, T2 and FLAIR (a,b and c respectively) images with left basal ganglia and peri-ventricular Hyper intense lesions on T2WI and FLAIR study, Hypointense in T1WI which shows no mass effect suggestive of infarction, Mild widening of the cerebral sulci is evident suggestive of mild cerebral atrophy.

Discussion: From this particular case it is clear that diagnosis could get delayed for so long resulting in needless personal suffering and family tragedy.

APS also can present with multiple neurologic symptoms of which ischemic stroke is the commonest and rarely with chorea (1.3%) (8).

The patient doesn't have a history suggestive of rheumatic fever, like recurrent sore throat, skin rash of rheumatic fever or arthralgia though she had echocardiographic abnormalities; in light of her fulfilling the criteria for APS the diagnosis of Sydenham's chorea was not considered and APS associated chorea was entertained. Of Huntington's disease in this patient, other than the chorea she doesn't have the cognitive deficit of it and as an autosomal dominant disorder she doesn't have similar family history. When we see chorea gravidarum that occurs during pregnancy and associated with eclampsia and use of OCP and tend to have seizure (which our patient doesn't have). With the above mentioned reasons we considered her to have Chorea associated with APS.

Conclusion:
APS is a fairly prevalent condition, especially among, young patients with DVT, myocardial infarction and stroke. Also it is associated with recurrent pregnancy loss or abortion. Every physician and health facility dealing with clots, gangrene, stroke, myocardial infarction and recurrent abortions is expected to be vigilant and have a high index of suspicion of APS, which is a fatal syndrome in order to avert potential consequences.

In addition, awareness at service provider's level, improving diagnostic capacities by trained professionals, equipment and supplies, subsidization of costs would improve early diagnosis and prevention of fatal consequences of APS. Failure to do so however would inflict its avoidable disability, family tragedy and death.

Abbreviation
APS- Antiphospholipid Syndrome
TASH-TikurAnbessa Specialized Hospital
CBC -Complete blood count
ANA- Anti Nuclear Antibody
Ds DNA-Double stranded DNA
LA- Lupus Anticoagulant
aCL- Anticardiolipin Antibody

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References
1. Graham r V Hughes, Ronalda Asherso, Munther A Khamashta. Antiphospholipid syndrome: Linking many specialties**.** Annals of the Rheumatic Diseases, 1989; 48:355-3566.

2. Miyakis S, Lockshin MD, Atsumi T, Branch DW, Brey RL, Cervera R, et al. International consensus statement on an update of the classification criteria for definite antiphospholipid syndrome (APS). J Thromb Haemost (2006) 4(2):295–306.
3. Petri M. Epidemiology of the antiphospholipid antibody syndrome. J Autoimmune 2000; 15:145-51**.**
4. Ricard Cervera. Antiphospholipid syndrome Department of Autoimmune Disease**s**, Hospital clinic, Barcelona, Catalonia, Spain. Thrombosis Research 151, Suppl. 1 2017:S43–S47.
5. Antiphospholipid syndrome. Fast Facts. 2017 American College of Rheumatology.www. rheumatology.org/i-Am-A/patient-Caregiver/ disease-Conditions/Antiphospholipid-Syndrome. (Retrieved December 27).
6. P. Kirk, MB, CCFP, J.A. Moran, MB, MCLSC, CCFP. Recognizing antiphospholipid syndrome Case report: misdiagnosis delayed treatment. Canadian Family Physician, Le MIdecindefamillecanadien .VOL 42: FEBRUARY, 1996.
7. http://www.rheumatologynetwork.com/fibromyalgia/sydney-classification-criteria-definite-antiphospholipid-syndrome (Retrived June 14, 2018).
8. G. Sanna, M. L. Bertolaccini, M. J. Cuadrado , M. A. Khamashta and G. R. V. Hughes. Central nervous system involvement in the antiphospholipid (Hughes) syndrome. Rheumatology. 2003; 42:200–213.