Stroke in an adolescent girl - combined Primary Anti phospholipid syndrome and Hyperhomocysteinemia - case report
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Abstract : Stroke remains a major cause of mortality and morbidity worldwide. Stroke in young persons can be devastating in terms of productive years lost and impact on a young persons life. The diagnosis of stroke in children and young adults can be difficult and the cause of ischemic stroke often remains unexplained, even after extensive evaluation. Here we present a case of ischemic stroke with right hemiparesis and aphasia in a 17-year adolescent girl. On evaluation of the patient, we concluded that Primary Anti phospholipid syndrome (APS) and Hyperhomocysteinemia accounts for the etiology for our patient. She had been started on anticoagulants and antithrombotics which she responded well to the treatment. She is on further follow up. This case is presented for the combined presentation of Primary Anti phospholipid syndrome and Hyperhomocysteinemia as ischemic stroke in young adolescent girl.

Keyword : APS, lupus anticoagulant, APLA, Hyperhomocysteinemia

Introduction:
Anti phospholipid syndrome is a non inflammatory auto immune disease, and its principal pathologic process is thrombosis. It is characterized by recurrent thrombosis associated with pregnancy complications and the presence of antiphospholipid antibodies (APLA) in serum. APLA is found in 10-46% of young patient with stroke and over all it accounts for 10% of stroke patients. Stroke patient with APLA tend to be younger. Small vessel block is most common than large vessel block in APLA. Overall, incidence rates under the age of 45 ranges from 7 to 15 in 100 000 people/year for all stroke (1, 2). Hyperhomocysteinemia is a new predisposing condition in which inherited and acquired factors interact cumulatively. It is increasingly recognized that single genetic abnormalities are seldom the sole cause of stroke (3). Patients with combined APS and hyperhomocysteinemia are high risk for stroke and its recurrence (4, 5).

Case report:
17 year old female presented with right side hemiparesis with aphasia. CT brain showed left capsuloganglionic infarct. (fig.1) MRI brain showed acute left basal ganglia infarct (fig.2,3,4) and MRA showed Thinned Internal Carotid artery. (fig.5) Her routine complete hemogram, Peripheral smear, RFT, LFT were found to be within the normal range. Chest x-ray, ECG and Echo were normal. Total cholesterol was 122 and triglycerides 65 mg/dl respectively. The aPTT was normal, ANA negative, HIV 1& 2 negative. IgM anticardiolipin antibodies were > 25 U/ml and lupus anticoagulant were present on two occasions 12 weeks apart. IgG anticardiolipin was negative. Her homocysteine levels were also elevated at 20.4 umol/L.

As per the guidelines of Sydney investigational criteria for APS (revised Sapporo criteria) she was diagnosed as APS. She was started on Acitrome and Aspirin. She was under regular training programme by physiotherapist and speech therapist. Her neurological status recovered to normal level. Speech deficit is on recovering phase. The patient is under follow up now.

Discussion:
Antiphospholipid antibodies (aPLs) are a group of antibodies directed against phospholipids or phospholipid protein complexes. These antibodies have been linked to a clinical syndrome consisting of thrombosis, thrombocytopenia, and recurrent fetal loss. Lupus anticoagulant is a major risk factor for arterial thrombotic events in young women and reported that 17% of patients with ischemic stroke as tested positive for the antibody [6]. Lupus anticoagulants are considered to carry a 5 to 16 times higher risk for thrombotic events than anticardiolipin antibodies [7]. Anticardiolipin antibodies and Lupus anticoagulant when present together are associated with a higher stroke risk.

APS can be diagnosed based on an international consensus criteria (Sydney investigational criteria for APS) and its diagnosis is based on clinical criteria of vascular thrombosis or one or more occurrence of pregnancy morbidity, and laboratory criteria of the presence of antiphospholipid antibodies on two or more occasions at least 12 weeks apart [8, 9]. Our patient fulfills the Definite APS which is presence of one clinical and one laboratory criteria.

Our patient has presented at a young age of 17 yrs. The mean age at presentation in APS is 35 to 45 years [10, 11, and 12]. Our patient has elevated homocysteine levels as well which confers increased risk of recurrent episodes of thrombosis. Recent studies have suggested an
association between moderate hyperhomocysteinemia and risk of ischemic stroke, indicating that hyperhomocysteinemia confers at least a 2.5-fold increased risk of stroke (13). Recently, the homocysteine-lowering effects of folate and vitamins B6 and B12 has been explored and vitamin supplementation is a novel therapeutic strategy for vascular thrombotic diseases. Patients with APS and thrombosis should be treated with warfarin for the long-term, and maintained at a therapeutic international normalized ratio (INR) of 2.0 to 3.0. It can also be combined with aspirin. Here we have presented a 17 yr old adolescent female with ischemic stroke due to combined primary APS and hyperhomocysteinemia.

Conclusion:

Stroke in the young requires a different approach to investigation and management than stroke in the elderly given differences in the relative frequencies of possible underlying causes. Extensive evaluation is necessary to clench the diagnosis as well as to reduce mortality and morbidity. APS, Hyperhomocysteinemia due to B12 deficiency are treatable causes which on treatment will prevent recurrent episodes of stroke.

References:

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