Antiphospholipid Syndrome

Alternative Names
Antiphospholipid Syndrome, Familial
APS
Lupus Anticoagulant, Familial
Hughes’s Syndrome

WHO International Classification of Diseases
Diseases of the blood and blood-forming organs and certain disorders involving the immune mechanism

OMIM Number
107320

Mode of Inheritance
Autosomal dominant

Description
Antiphospholipid syndrome (APS) is a clinical disorder characterized by recurrent arterial and venous thrombotic events, fetal losses, thrombocytopения, neurological symptoms, livedo reticularis, and hemolytic anemia. Affected individuals show persistently elevated levels of antibodies directed against cellular phospholipid components (aPL). APS can occur in patients with or without evidence of any definable associated disease, in which case it is referred to as primary APS. It may also occur in association with another rheumatic or autoimmune disorder, such as Systemic Lupus Erythematosus (SLE, hence Secondary APS).

Incidence of antiphospholipid syndrome remains unknown. However, the reported prevalence of antiphospholipid antibodies in the general population is low (1-4.5%) and increases with age. The antiphospholipid antibodies have been detected in approximately one-third of the patients with SLE. High anticardiolipin antibodies titers, lupus anticoagulant (LAC) and especially anti-beta2GPI antibodies are important predictors of APS clinical manifestations in SLE patients. A rare form of this disease is the catastrophic APS, in which there is rapid organ dysfunction and failure. The management of thrombosis includes long-term, high-intensity warfarin therapy. For pregnancy morbidity, the recommended therapy is low-dose aspirin (80 mg/day) plus subcutaneous unfractionated heparin or low-molecular-weight heparin.

Molecular Genetics
The aPL antibodies belong to the large family of antibodies that react with negatively charged phospholipids, such as cardiolipin, phosphatidylglycerol, phosphatidylinositol, as well as beta-2 glycoprotein, prothrombin, annexin V, Protein S, and high molecular weight kininogen. The predisposition to clotting characterized in this disease is predominantly due to these antibodies interfering with the antithrombotic role of phospholipids such as cardiolipin, causing platelet aggregation and subsequent vascular occlusion. APS, therefore, is associated with thrombotic, rather than hemorrhagic complications. Familial occurrence of elevated levels of aPL antibodies, as well as an association with HLAs DR4, DR7, DQ7, and DR53 have been reported.

Epidemiology in the Arab World

Bahrain
Ebrahim et al. (2005) undertook a study on 22 Bahraini patients with APS over a period of 16 years. Among these patients, the frequency of primary APS was 45.8%, while secondary was 54.5%. Female: male ratio was 10:1. The percentage of pregnancies was 71.4%, and 57.1% of these ended in miscarriages (80% in primary and 50% in secondary APS). Anticardiolipin IgG and IgM were assayed by ELISA. Ebrahim et al. (2005) found that the aCL IgG was higher in primary APS (80%) as compared to secondary APS (66.7%). However, aCL IgM was higher in secondary (75%)
compared to primary APS (60%). Earlier reports have suggested that patients with IgG aCL antibodies are at higher risk than those with IgM or IgA antibodies. Antinuclear antibodies were present in all of the secondary APS, and in 20% of the primary APS patients. Treatment given to the patients was either in the form of aspirin (33.3%), steroid (76.2%), heparin (28.6%) or warfarin (28.6%). Ebrahim et al. (2005) concluded that APS is not a common problem among hospitalized patients in Bahrain.

**Saudi Arabia**

Owaidah et al. (2003) conducted a single center review of clinicopathological characterization in patients with lupus anticoagulant (LAC) antibodies. Screening tests included activated partial thromboplastin time (aPTT), diluted Russel’s viper venom time, and Kaolin clotting time. Anticardiolipin IgG and IgM were assayed by ELISA. Indirect fluorescence procedure for antinuclear antibodies and noncompetitive EIA for ds-DNA antibodies were also used. The most frequent clinical findings were arterial and venous thrombosis, and obstetric complications. Owaidah et al. (2003) found a sizable number of patients with incidental APS. Also, in a majority of the cases, the activated partial thromboplastin time (aPTT) was elevated and was not corrected by mixing with normal plasma. The prothrombin time (PT), however, was normal in most of the patients.

**References**


**Contributors**

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