Berdeaux DH, McGlasson DL, Marlar RA. Familial Association of Antiphospholipid Antibodies in Index Patients with Primary Antiphospholipid Syndrome. Clin Exp Rheumatology, Vol 10/6, pg. 655, Nov/Dec 1992.

Genetic factors play an important role in the development of autoantibodies and autoimmune diseases. Autoimmune diseases arise from a combination of genetic predisposition and environmental triggers that disrupt the immune system's ability to ignore a person's own tissue and cells. In rare cases, an autoimmune disease is monogenic, caused by mutations in a single gene. Although antiphospholipid antibodies [APA] are in sera of many patients with autoimmune rheumatic diseases, there are few reports of familial association. We have evaluated multiple generations in three unrelated families in which multiple members have abnormal serum APA, and at least one member had the antiphospholipid syndrome [APS]. Serum from 5 of 15 members of family #1 [4 generations] had abnormally elevated APA assays. The proband was a 48-year-old man with positive APA results who had repeated episodes of venous and arterial thromboses including bilateral retinal vein thromboses, two thrombotic cerebrovascular events, a pulmonary embolus and a deep vein thrombophlebitis. His sister had multiple cerebrovascular thromboses starting at age 38 and his father died of a myocardial infarction at age 49. The proband and sister were APA-positive. Sera from 4 of 10 members of family #2 [3 generations] had positive APAs. The proband was a 26-year-old man who died of a dysrhythmia after 7 myocardial infarcts and a pulmonary embolus. Sera from 5 of 8 members of family #3 [2 generations] were APA- positive. The proband was a 26-year-old woman who presented with thrombocytopenia, migraines, visual symptoms, and 3 miscarriages. Her mother had migraines, transient ischemic attacks and 2 miscarriages. The inheritance pattern appears to be autosomal dominant. In conclusion, these familial associations suggest that genetic factors, which are known to regulate other autoantibodies, may also contribute to the regulation of APA.