A congenital Antithrombin III (AT III) deficiency affecting 7 members of 3 families is reported. The first thrombo-embolic accidents were observed between the age of 22 and 35: they were spontaneous or occurred after delivery or oral contraception. In one patient, a deep vein thrombosis was observed during heparin treatment. In 2 cases, recurrent pulmonary embolic episodes required vena cava ligation. No thromboembolic accident was observed during oral anticoagulation.

AT III was measured by an amidolytic method and by the Mancini method on plasma and serum; the antithrombin activity was determined on serum by the von Kaulla method. In 7 patients, a decreased AT III was found by all the methods performed. The AT III level was around 50% in patients treated or not by oral anticoagulants.

One patient was studied during heparin treatment and then under oral anticoagulants: AT III levels were lower under heparin.

Antithrombin III deficiency has been reported to be decreased in the cases of several thrombotic disorders and the decreased Antithrombin III is known to induce the hypercoagulable state. This study was started from the 28-year-old male patient who developed the superior sagital sinus thrombosis after appendectomy and it was followed by deep vein thrombosis of extremities. Antithrombin III level was 19 mg/dl and activity was 68% by the progressive antithrombin assay and other laboratory examinations were within normal range excepts for the elevated serum lipids. Antithrombin III was assayed for his family members in three consecutive generations by single radial immunodiffusion method, coagulation assay, and chromogenic assay. Four out of eight members were confirmed to have low Antithrombin III level and activity ranging from 59%-68% of normal values, although the two of these four members had no history of thrombosis. Mother of this propositus is deceased, but it was suspected of having the defect of Antithrombin III.

History of peptic ulcers were found in all members of this family. The inheritance pattern of Antithrombin III deficiency was characteristic of an autosomal dominant disorder.

Antithrombin activities in 30 severely malnourished children and 40 normal children were estimated in clotting tests by thrombin neutralization, as antiXa and by a heparin antithrombin assay; and by immunodiffusion as α2-globulin and α1-antitrypsin. The patients' mean α2-globulin was severely depressed, and there were less marked depletions in mean values for thrombin neutralization, antiXa and in the heparin antithrombin assay (which showed the flat curve thought to reflect a thrombotic tendency). The α1-antitrypsin values were normal. The findings support the concept of antithrombin as the summation of α2-globulin and α1-antitrypsin (with α2-macroglobulin); and may be related to the high incidence of thromboses reported in childhood malnutrition.